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A GENETIC AND PSYCHOSOCIAL STUDY OF ALBINISM IN SOUTHERN AFRICA

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A thesis submitted to the Faculty of Medicine, University of the Witwatersrand, in fulfilment of the requirements for the degree of Doctor of Philosophy.

Johannesburg 1985.

DECLARATION

I declare that this thesis is my own unaided work, and that all the assistance I received in its preparation consisted of technical advice detailed in the acknowledgements; that all the field-work was performed by myself and by assistants acting under my direct supervision; and that the conclusions I have reached have resulted from my own project. No part of this thesis has been submitted before for any degree or examination in any other University.



J G R KROMBERG

This study is dedicated to the memory of my father, David Norman Murray, (1909 - 1976), who devoted a life-time to the development of social services for individuals, families and communities, and to my mother, Grace Laura Murray, who stimulated independent thinking, encouraged learning and provided a rich cultural background for her children.

ABSTRACT

Albinism is a recessively inherited condition found in all human populations including populations throughout Africa. People affected by the condition have problems which are due not only to the physical features of albinism, but also to the attitudes of society. The objective of the present study was to investigate: the prevalence of albinism; the types found locally; the skin cancer risks; the life expectancy; the skin colour and fertility in homozygotes and heterozygotes; and the impact of the birth of an albino baby on the black parents.

The subjects for the study were drawn from the urban area of the greater Johannesburg metropolitan region, including Soweto, and from four rural areas in Botswana, Kwazulu, Transkei and Swaziland. The subjects included albinos and their families, as well as matched controls where necessary, and they were investigated by means of interviews, hair-bulb incubation tests, physical examinations, skin reflectance measurements, and observations.

The urban prevalence rate was found to be 1:3 900, but the prevalences for rural populations belonging to four of the major ethnic groups were higher and exceeded 1:2 000 in both Botswana and Swaziland. The rates in these groups were correlated with the consanguineous marriage rate, the Sotho-Tswana groups generally having higher rates and the Nguni lower rates. The albinos were classified into five groups, of which the tyrosinase-positive type was found to be the commonest, followed by the brown and rufous types, while the tyrosinase-negative and yellow mutants were very rare. All the albinos had some form of pigment in their hair and/or eyes. About a quarter of the urban sample had malignant or premalignant skin lesions. The risks for skin cancer increased with age and were associated with ethnic group, the Sotho-Tswana group being most susceptible. Both squamous and basal cell carcinoma were reported and the commonest site was on the head. Life expectancy appears to be reduced, and there was a significantly younger age distribution among albino males than among the general population. Skin colour was lighter in heterozygote females than in matched controls and a lighter skin is considered locally to be attractive, so that there appears to be a selective advantage in being a carrier for the albinism gene which could partly explain the high prevalence rates. Fertility was found to be reduced in albinos, but whether this is due to stigmatization or to physiolo-

gical factors remains to be clarified. Parents and grandparents, however, appeared to have normal fertility rates.

The birth of an albino baby had an impact on the parents which could be described by the mothers and observed in their behaviour. Many mothers were depressed and unhappy about the birth, were initially uncomfortable with close contact with their babies, and were observed to interact less with them than did the mothers in the control group. Maternal attachment appeared to have been delayed in these mothers, but by the time the baby was three months changes had occurred and most mothers showed similar levels of maternal-infant interaction as did the controls, although many still expressed feelings of unhappiness. Nine months after the birth most mothers had accepted their affected infants. Many of the babies, however, were slower to sit and crawl (according to their mothers) than the controls, suggesting that either the partial maternal deprivation, or their poor eye sight, or both these factors, had had an effect on their normal development.

The small co-operative group of fathers who were interviewed and observed only once, when the albino infants were about six months of age, appeared to have accepted them and were not planning to alter their reproductive plans as a result of the birth.

Recommendations arising from the study include the establishment of both a hospital based clinic specifically for the treatment and counselling of albinos, and an association to meet their needs. If this research results in such action the albinos in Southern Africa can be expected to enjoy a better quality of life in future.

PREFACE

The present study was carried out by the writer during her employment as the senior genetic social worker in the Department of Human Genetics, School of Pathology, South African Institute for Medical Research and University of the Witwatersrand, Johannesburg. The study was part of a series of studies on the subject of albinism in the black populations of Southern Africa carried out by the S A Medical Research Council (MRC) Human Ecogenetics Research Unit. The section of the thesis on the urban prevalence of the condition has been published as a journal article (Kromberg J G R and Jenkins T (1982). Prevalence of albinism in the South African Negro. S Afr med J 61: 383 - 386). Although not directly related to this thesis two other studies on albinism in South Africa have already been published by the writer. These are:

Manganyi N C, Kromberg J G R, Jenkins T (1974). Studies in albinism in the South African Negro I. Intellectual maturity and body image differentiation. J Biosoc Sci 6, 107 - 112.

Kromberg JGR, Jenkins T (1984). Albinism in the South African Negro III. Genetic counselling issues. J Biosoc Sci 16, 99 - 108.

Professor Trefor Jenkins, head of the Department of Human Genetics, provided an atmosphere conducive to research, as well as the stimulation, moral support and supervision required for completion of this projeet. Professor J W Mann, head of the Department of Psychology, University of the Witwatersrand, was co-supervisor, and throughout the study he assisted the writer with constructive criticism, wise advice and encouragement, gave generously of his time and expertise, and continually motivated the forward movement of the work.

Without the support of the Director, Professor J Metz, and the considerble resources and helpful staff of the South African Institute for Medical Research, the study would not have been possible. The writer is particularly indebted to Professor T Jenkins, Dr G Nurse and Dr M de Saxe who examined the albinos; to Mrs L Battaglia, the Librarian, and her staff for searching out articles and Journals not frequently consulted by medical researchers; to Mr D Dunn, for assisting with the laboratory work required in making the solutions for the hair-bulb incubation test, to the late Mr M Ulrich, Mr M Anderson and staff of the photographic unit for much of the studio photo-

graphic work, and to Ms S Hall for the art work. Special thanks are due to Miss J Robbie, Mrs C Smit and Mr R Colgate for putting the text into the word processor so willingly and efficiently and to Mrs G Dique, Mrs S Woods and Mrs S Alper who typed parts of earlier drafts.

The staff of the MRC provided invaluable assistance. Their senior nurse/research assistant, Mrs Esther Zwane, carried out most of the interviews for the psychosocial component of the study. Her experience in interviewing, her sympathetic approach with the subjects, and her enthusiastic co-operation was much appreciated. The MRC Institute for Biostatistics staff were also most helpful and advised the writer on the use of statistical tests, as well as carrying out some of the calculations.

The writer is grateful to the West Rand Administration Board who provided permits for purposes of visits and research in Soweto, and to the local education authorities who allowed access to the schools there.

Since the study was multidisciplinary, the expertise and advice of many specialists was sought. Discussions were held with anthropologists, child, clinical and social psychologists, sociologists, social workers, geneticists, medical practitioners, pathologists, child and adult psychiatrists, epidemiologists, medical technologists, biochemists and statisticians, and all gave new insights and contributed to broadening the writer's view of the subject of albinism and its effect on individuals and their communities.

The subjects themselves were friendly and approachable and the writer is most grateful for their co-operation. Alex Lipworth and Son supplied gratis a special anti-actinic cream for the subjects, using a formula compiled by the late Dr Paul Keen of the National Cancer Association, and thanks are due to them for this community service.

Lastly, my warmest thanks go to my children, Jacqueline and Jonathan, who patiently, sacrificially, and with love, indulgence and understanding put up with a mother who worked long hours and was often preoccupied with the project, particularly during the latter stages.

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ABBREVIATIONS

BA	Brown albinism
ca	carcinoma
Con	control
exp	expected
het	heterozygote
mg	milligram
nm	nanometer
OA	ocular albinism
obs	observed
OCA	oculocutaneous albinism
RA	rufous albinism
ty-neg	tyrosinase-negative
ty-pos	tyrosinase-positive
uv	ultraviolet
ym	yellow mutant

CHAPTER 1

INTRODUCTION

1.1 Background

The background to the present study is based on the writer's strongly held belief that the field of human genetics requires more input from the social sciences. Human origin and human experience are two sides of the same coin. Multidisciplinary studies are therefore important for the understanding of the totality of human existence. They can throw new light on inherited disorders and the way in which these disorders affect individuals, who in turn are inextricably intertwined with their family and community. It is vital then to know, for example, not only how frequently a disorder occurs, but what social factors might be causing it to occur so frequently and how others react to affected people.

Childs (1978, p29), in discussing priorities in research, has expressed this emphasis very well:

> `Much of the biochemical and developmental configuration assumed by each human being originates in his social behaviour; medical transactions are social, and the life of a patient with a disease is social. These aspects of health and disease have not been exactly neglected by investigators, but as subjects for research they do not enjoy the attraction not to say the glamour, of other more conventional scientific pursuits. Perhaps molecular biology is queen because the possibility to test hypotheses precisely and clearly is at a maximum, while research into attitudes is beset by problems of definition, quantification and research method. But thinking of the causes of birth defects as originating partly in the ways in which living is organized, that is, in the geographic, economic, and social elements which determine where and how we live, and learning something about how or even whether people perceive the connection between their everyday habits of living and their health; and learning about how to harmonise one's endowment with one's environment and how to accept people whose appearance and behaviour is `different' are all as necessary to the solution of problems posed by birth defects as learning about the molecular mechanisms'.

Research on genetic disorders in the local black population has generally received little attention. If genetic services and counselling are to be offered to this community in the future, data must be collected so that a basis will be available for not only calculating occurrence and recurrence risks, but also for achieving an understanding concerning what dis-

orders mean to this population group with its unique culture, practices, customs, and life-style.

These beliefs motivated the writer to pursue an interest in the lives and problems of people, families and communities affected by a genetic disorder. A researching medical social worker, such as the writer, does not profess to be as expert in such specialities as genetics and psychology as the specialists themselves, but has to be adequately acquainted with several disciplines in order to carry out her research. The disorder chosen for the research was albinism, since it is quite common, very distinct, easily distinguishable and relatively easily identified and diagnosed by a non-medical person, because it is so striking in a black population. The present study represented a continuation of previous work on this population which had covered such aspects as the intellectual maturity of albinos (Manganyi, Kromberg and Jenkins, 1974) the adjustment of albino youth (Kromberg, 1977), the urban prevalence of the condition (which was a report on some of the data from the present study, Kromberg and Jenkins, 1982), some of the genetic counselling issues associated with the albinism (Kromberg and Jenkins, 1984), and in a more general study, the common birth defects in South African blacks (Kromberg and Jenkins, 1982). These projects provided answers to some of the queries concerning albinism but uncovered other issues which merited attention. These issues raised the questions: Is albinism in the black population really as common as it appears to be, and if so why is it so common in Southern Africa? Is the urban rate a true reflection of the situation generally? Do marriage patterns influence the apparently higher rates in some population groups? Is there heterozygote advantage conferred by the gene? What are the types of albinism found in Southern Africa? What are the characteristics, health problems and life-expectancy of local albinos? How does the birth of an albino baby affect a black family? The basic issues that these problems address suggest that despite the small contribution of our previous studies, research in the field is still very sparse. The present study was initiated, therefore, to explore these areas and to collect the data necessary to gain a broad understanding of these questions.

1.2 Aims of the present study

In general the purpose of the study was to obtain information on some selected genetic and psychosocial aspects of albinism in the black population in Southern Africa. Specifically the aim was multifaceted for both these as-

pects, and could be stated in six parts for the genetic study and two parts for the psychosocial study as follows:

1. Genetic studies:

(i) To determine: the prevalence of albinism in urban and rural areas, in the different ethnic groups of the population, and in males and females and the consanguinity rate in the affected families.

(ii) To identify, classify and describe the various types of albinos found during the study.

(iii) To investigate the risk of skin cancer, the types found and sites affected in albinos.

(iv) To assess the life-expectancy of albinos.

(v) To measure skin colour in both heterozygotes and homozygotes for the albinism gene.

(vi) To examine the fertility rates in homozygotes, heterozygote x heterozygote matings (parents of albinos) and heterozygote x presumed normal matings (grandparents of albinos).

2. Psychosocial studies:

To explore the nature of the impact of the birth of an albino baby on the black family, to examine any changes which might occur over time, and specifically to investigate:

(i) The mother: in terms of her medical problems, childhood history, pregnancy, delivery, neonatal problems, care of the infant, feelings about the infant, her view of the paternal response to the infant, of the relationship with the father, and of the grandmother's and siblings' response to the affected baby, her behaviour towards and attachment to the infant, and her feelings of anxiety.

(ii) The father: in terms of his medical history, care of the infant, feelings towards the infant, his view of the maternal response to the infant, the maternal-paternal relationship, and his behaviour towards the infant.

1.3 Setting and scope of the study

To meet the aims of the study both urban and rural settings were required. The urban settings selected included Soweto, the town adjacent to Johannesburg, the city of Johannesburg itself (with a black population of over a million), and eleven other smaller black suburban townships attached to the various towns of the Southern Transvaal. The urban areas were chosen because

of their large black populations consisting of representatives from all ethnic groups and because of their easy accessibility from Johannesburg. The rural areas were selected to represent two with probable high rates (Botswana and Swaziland) and two with probable lower rates (Kwazulu and Transkei) based on the data from the Soweto study. The large village of Mochudi (population 18 300) provided the setting for the Botswana study, because good contact with an anthropologist working there had been established prior to the studies and this facilitated the arrangements and organization for the field-work. The setting in Kwazulu was the district of Nongoma (population 90 000) where one of the original groups of the Zulu people lived with their King, and where there was a hospital which provided a base for the work. The Transkei study covered eleven remote districts with scattered population clusters and health clinics, which were being visited by a team providing care for the blind. It was possible for the writer to accompany this team and to co-ordinate the field-work with their activities. The Hhohho district (population 160 000) of Swaziland was selected as the setting for the rural study of the Swazi, because it was the closest Swazi district to Johannesburg, it had three scattered hospitals where bases could be established and the communications net-work was very efficient.

The urban settings provided the main group of subjects for the study. Many of these subjects had participated in previous studies on albinism and they were generally well-disposed towards research. The prevalence, identification of types, skin cancer, life-expectancy, and fertility studies were carried out with this co-operative group of subjects. Similarly, the study of new-born albino babies and their families was set in the accessible urban areas, where a relatively large number of births could be expected to occur during the study period.

The rural areas were the setting for the four studies of rural prevalence. Life-expectancy and skin colour were also investigated during the field-work in Swaziland, because of the good population age statistics, the normally very dark skin colour of the inhabitants (Seligman, 1966, p122) and because the expertise of Professor D Roberts of Newcastle UK, who brought to South Africa a skin colour measuring instrument and joined the writer in the Swazi research work, was available.

Further details concerning these areas, the cultural customs, social and demographic parameters of the people living there, which are relevant to the aims of the present study, will be presented in the Chapter 3.

1.4 Design of the study

A programme of field-work was designed to accomplish the aims of the study:

1. Genetic studies

First a study of the selected settings and of the literature on albinism was conducted, so that the subjects might be understood against both their environmental (which is important in any ecogenetic study) and their historical background. Then albinos were ascertained by all available means in the various urban and rural settings and compared with the numbers of the general population in each area so that prevalence rates could be calculated. Each subject, and wherever possible other members of the family, were interviewed, for information on the family history so that pedigrees containing at least three generations could be compiled. These interviews produced data useful for the calculations of consanguinity, life-expectancy and fertility rates. Hair-bulbs were collected by the writer when the subjects agreed to co-operate, for incubation tests, a medical examination and clinical description was carried out by a medical practitioner, and photographs were taken wherever possible, so that data would be available for the identification of types. At the medical examination malignant and premalignant lesions were noted to give information on skin cancer. Further data were collected on this problem from the records of the Histopathology Department, School of Pathology, South African Institute for Medical Research and University of the Witwatersrand, in Johannesburg and from the Cancer clinic at the Hillbrow Hospital in Johannesburg. Skin colour was measured in albinos and their mothers in Swaziland. The data were analysed and compared with those collected from either control groups, where necessary, or from the reported general population statistics, and examined for significance by means of statistical tests. Results were obtained and discussed, in relation to findings reported in the literature, and conclusions were drawn.

2. Psychosocial studies

Information concerning births was obtained from the maternity wards of hospitals in and around Johannesburg and the new-born babies with albinism were identified. They, together with their mothers and fathers, were admitted to the study. A longitudinal investigation of these subjects over fifteen months was planned to include six interviews and five observation sessions with the mothers and one interview with one observation session with the fathers. The first four interviews, held at three monthly intervals with

the mothers, were followed by the observation during which the behaviour of the mother with her baby was assessed. After the sixth interview the maternal-infant interaction in a play situation was observed and a scale to measure anxiety completed. The results from the questionnaires administered at these interviews and on the scores on observation were analysed and compared with those for a matched control group. Conclusions were based on statistical analysis of the data, and the impact of the birth of the albino baby on the mother and father was assessed.

In this way the study was designed to collect data relevant to the aims in both the genetic and psychosocial fields.

1.5 Clarification of terms

Various terms were used in the writing of this thesis, sometimes interchangeably, which might cause some confusion for the reader. Definitions are presented and discussed here to clarify the matter.

Albinism: The word albinism is used throughout the study in its limited sense to describe subjects with the generalised form of the condition resulting in hypopigmentation of the eyes, skin and hair. Individuals with the different types of partial albinism (e.g. ocular albinism and piebaldism) were excluded from the study (a detailed definition of albinism and a discussion of the types investigated in the study is presented in Chapter 2).

Genetics: Genetics has been defined as `the scientific study of heredity' (King, 1972). In the present study the word genetics has been used in its widest sense and the genetic studies include such issues as the prevalence and identification of types of albinism, as well as related health issues such as skin cancer risks, life-expectancy and fertility, and the effect of the gene on skin colour.All these individual topics are concerned with a hereditary disease, which has been defined as a `pathological condition caused by a mutant gene' (King, 1972),and therefore they are included under the heading `genetic studies' in the present report.

Psychosocial: `psychosocial' has been defined very broadly as `involving both psychological and social aspects' (Stedman, 1982). More specifically psychosocial factors have been defined as `social situations, relations and pressures that have psychological effects' (Goldenson, 1984). One part of the present study was concerned with the impact of the birth of an albino baby on the family and in this part both psychological factors and

social situations were important. This part of the study endeavoured to throw light on the internal psychological and external social side of the problem of albinism and to assess how parents functioned in their relationship with the affected baby. The term psychosocial therefore seemed appropriate in this context.

Black/Negro: Many terms have been and still are used to describe the various population groups of South Africa. The physical anthropologists and geneticists use terms concerned primarily with the genetic constitution of a population (Jenkins, 1972), and so they designate the `black' people of Soweto, for example as `Negroes'. The social scientists however consider the term Negro to apply to a black person domiciled in the United States of America. To the American black person, on the other hand, the term Negro is connected with slavery, and it is included in a list of derogatory terms drawn up in a UNESCO report and categorically condemned (Baird, 1970).

The alternative terms used to describe the population of Soweto are `Bantu', `African' or `black'. Wilson and Thompson (1969, pxi) abandoned the use of the term `Bantu' (except in connection with linguistics) because `it is disliked by those to whom it is applied'. The term `African' is likewise unacceptable, since South Africans (as well as Zimbabweans, Zambians and people from other African colonized countries) may be of either European or African origin. The word `black' has its disadvantages, particularly in the context of the present study, since describing an albino as a `white black man' is a contradiction in terms. Also designating people as `black' only describes them physically and does not indicate their cultural identity. Nevertheless as Baird (1970) states `the word black is at least on a level of semantic parity with the word white as a description of persons and is therefore much more acceptable than Negro'.

The procedure in the present study is therefore to use the term `black' to designate persons of African or Negroid origin, and the term `white' for those of European or Caucasoid origin. There will be rare instances however when it will be necessary to use the term `Negroid' to contrast with `Caucasoid', usually in conjunction with the word albino. Terms such as `African', `Non-European', and `Bantu' will be used only very occasionally where these were the official terms in a specific document or context under discussion.

Ethnic group/tribe: The word ethnic group has been used during the present study for the cultural sub-groups of the black population. The word tribe, in the past, described these groups (Schapera, 1937), but this word

is no longer generally acceptable, since it is vague and ambiguously defined by laymen and anthropologists and technical definitions vary (Hammond-Tooke, 1974, pxv). Both the noun `tribe' and the adjective `tribal' are also considered to have perjorative overtones of `primitive', `backward' or `inferior' (Hammond-Tooke, 1974, pxvi) and they were therefore avoided. Hammond-Tooke (1974, pxvi) has suggested the word `chiefdom' be used for the political units under chiefs, and `people' for the larger cultural-regional groups. These words did not however seem applicable in the present context and the finer anthropological distinctions seemed unnecessary, since they were not important to the main focus of the study. The more general alternative of `ethnic group' was therefore selected and used for those units of the population which are distinct historically, culturally, geographically and politically, and which form the study of ethnographers. Members of such groups when confronted by aliens tend to classify them on an ethnic basis (Hammond-Tooke, 1974, pxvi) and the concept of an `ethnic group' appears to reflect the popular general classification. For these reasons this term seemed appropriate for the purposes of the present study.

Ascertain/identify: `ascertain' is defined as to `find out (for certain)' or to `get to know' (Fowler and Fowler, 1964). In genetic studies however the word ascertainment is used in more specific sense to imply `the finding and selection of families with an hereditary disorder' (Emery, 1979) and `the result of a search or inquiry' (Roget, 1972). This word has therefore been used interchangeably in the present study with the words identify or find, in connection with the detection of subjects for the study.

1.6 The plan of the thesis

In this thesis the study is broadly motivated, the aims defined, the setting and design of the study described, the terms clarified, and the thesis plan laid out in the introductory chapter. The history and current state of knowledge of albinism, with particular emphasis on African studies, is briefly and selectively reviewed, in terms of the aims of the study in the second chapter. This review is presented in two major parts reflecting the genetic and psychosocial studies. Various aspects of the disorder concerning its definition and classification of types, prevalence, and related problems of skin cancer, life-expectancy, skin colour and fertility, are considered in greater detail in the genetics section and the observations of others on these aspects are reported and critically assessed. In the psychosocial sec-

tion the studies on albinism with a psychological emphasis are described, together with the research on the impact of the birth of a defective baby on the family (since specific reports concerning albino babies appear to be lacking), and on the maternal- and paternal-infant relationships in white and black dyads. This review provides a background of relevant studies in which the present study can be placed and with which its findings can be compared, and serves the purpose of assessing and defining the gaps which might be filled by specific investigations.

The materials and methods used in the study are outlined in the third chapter of the thesis. The urban and rural settings are described in detail, with particular emphasis on the statistics, social factors and cultural practices, which have bearing on the study and are important in achieving an understanding of the circumstances in which the subjects lived. The methods used in the genetic studies and those used in the psychosocial studies are then presented separately according to the sub-divisions of the aims of the study. In general, the selection of subjects, controls, and an interviewer is described, the construction of questionnaires, the forms and the tests used are discussed, and the methods of statistical analysis specified.

The results of the study are reported in the fourth chapter. The findings from the genetics section are presented according to the six sections of the aim i.e. the prevalence, classification, life-expectancy, skin cancer risks, skin colour and fertility. The results of the psychosocial part of the study includes a section on the data collected at the interviews with and observations of the mothers and a second briefer section on the data collected from the fathers.

In the final chapter the discussion of the findings, in comparison with those from the literature, and the conclusions are presented. In addition applications, recommendations, particularly concerning education and service, and suggestions for future research are made.

1.7 Summary

The present study was motivated by the writer's interest in biosocial scientific research and particularly in problems of black individuals and families with the inherited disorder of albinism. The study aimed to investigate some of the genetic and psychosocial aspects of the condition. In the genetic part of the study the aspects selected for attention were prevalence, identification and classification of types, skin cancer risks, life-expect-

ancy, skin colour and fertility. In the psychosocial part the aim was to study the impact of the birth of an albino baby on the family, particularly the mother and father.

The urban settings for the study included Soweto, a black town near Johannesburg, as well as the other black townships attached to smaller towns in the Southern Transvaal Province of South Africa. The rural settings were districts of Botswana, Kwazulu, Transkei and Swaziland. The different sections of the study were carried out in different settings depending on the availability of resources, the feasibility and accessibility in each case.

The design of the study included the reviewing of the literature on albinism, the defining of aims, the planning of field-work, the ascertainment, interviewing, testing and examining of albino subjects as well as controls where necessary, the analysis of the data collected by statistical means, the assessment and discussion of results and the drawing of conclusions relevant to the aims.

Certain terms used in the presentation of the study were clarified since they might be considered ambiguous or vague by different research workers. These terms included albinism, genetics, psychosocial, black/Negro, ethnic group/tribe and ascertain/identify.

The plan of the thesis was then laid out so that a general view of the whole could be achieved.

CHAPTER 2

A REVIEW OF THE LITERATURE

A review of selected sections of the literature on albinism is a necessity in order that the present study may be placed in context against a background of previous studies.

The discussion will be divided into two sections. The object of the first section will be to give an historical and bibliographical account of albinism with special reference to African studies of the condition; to discuss the definitions, description and classification of the different types, the prevalence locally and elsewhere, the cancer risks, life expectancy, skin colour and fertility in albinos and heterozygotes. Where information is lacking on albinism the discussion will be supplemented with relevant literature on other recessive disorders. The second section will cover the studies which are concerned with the psychosocial aspects of albinism. Since no data on the effects of the birth of an albino on the family are available in the literature apart from a few descriptions of individual cases, studies on the impact of the birth of a defective and/or `different' child on the family will be reviewed, together with reports on the parents' reaction, and on maternal-infant and paternal-infant relationships in both affected and normal families.

2.1 Genetic studies

2.1.1 Historical background

According to Sorsby (1958) it is possible that the first written description of an albino appeared in the Pseudoepigrapha in the book of Enoch. The birth of Noah is described to the prophet Enoch (Noah's great grandfather) in this extra-Biblical book of Methuselah (Enoch's son and Noah's grandfather). Methuselah, referring to Noah, stated that: "His colour is whiter than snow, he is redder than a rose, the hair of his head is whiter than wool, his eyes are the rays of the sun and when he opened them he illuminated the whole house". Noah's father, Lamech, according to the story, questioned the faithfulness of his wife after seeing the baby.

Another very early account is that of Pliny who wrote in the first century AD. He quotes Isigoneus of Nicaeca, who related that in Albania there were men whose eyes were of sea-green colour, who had white hair from childhood, and who could see better at night than in the day (Plinius Secundus,

1948).

The term albino is derived from the Latin word albus, meaning white. It was first used in about 1600 by Balthazar Tellez, a Portuguese explorer and historian, to describe the white Negroes he had seen in Africa (Pearson et al, 1911).

de Maupertius, a French naturalist who lived in the eighteenth century, studied a few common traits in man, such as extra digits (polydactyly) and absence of hair and skin pigmentation (albinism) (Emery 1979, p2). From examining pedigrees he first showed that these two conditions were inherited in different ways.

Albinism was one of the first inherited abnormalities to be investigated statistically on the basis of population ascertainment (Froggatt, 1960a). This investigation was undertaken by Raseri in 1879 in Italy and he estimated that the condition occurred in 1 in 29 000 Italians (Pearson et al, 1911). Garrod (1908) suggested that albinism was the result of an inborn error of metabolism due to the lack of an `introcellular enzyme' which synthesized melanin. In 1910 Davenport and Davenport, having analysed many pedigrees of albino families, concluded that the condition was inherited as a simple Mendelian recessive. It appeared in 25 per cent of the off-spring from the marriages of carriers and did not occur in the children of albinos with normally pigmented spouses.

In their noteworthy monograph on albinism Pearson, Nettleship and Usher (1911), presented the results of a world-wide survey into the prevalence of the condition. The monograph also included an extensive historical and bibliographical background and a discussion on the nature and heredity of albinism. The geographical distribution of albinos in the light-skinned, yellow, red and black-skinned races was described with the support of many photographs.

Witkop and his group have in the last two decades contributed much to our understanding of albinism and have defined, delineated and described several sub-groups, as well as clarified the terminology (Witkop et al, 1983).

2.1.2 Studies on albinism in Africa

Because albinism is so striking in heavily pigmented populations, individuals have been described, commented upon, examined and investigated over the centuries by explorers, missionaries, anthropologists, and travellers, as well as by medical practitioners working in Africa.

This review of the literature on albinism in Africa is selective and limited to those papers which have a bearing on the present study, with special emphasis on the Southern African studies. Reports on biochemical studies on albinism have generally been omitted, as has the discussion on the legend of a white native race, which is frequently mentioned in the oldest writings (Pearson et al, (1911), but the existence of which appears to be unlikely (Froggatt, 1960). Each paper is critically reviewed, summarized and presented in chronological order arbitrarily divided into four sections: the early observational and descriptive studies including Pearson et al's (1911) comprehensive monograph which includes many reports from Africa (0 - 1912); the slightly more detailed and scientific studies starting with Stannus' (1913) work (1913 - 1951); the more systematic data collecting research initiated by Barnicot's (1952) study (1952 - 1972); and the more sophisticated studies of the last twelve years (1973 - 1984). Some of these papers were found in obscure, and others in discontinued journals (e.g. The Transvaal Medical Journal which was published from 1905 to 1913). Much information in the first section (0 - 1912) was obtained from Pearson, Nettleship and Usher's (1911) 'A Monograph on Albinism in Man' but some of the references could not be checked in the original sources, because they were in some cases centuries old and unavailable. The early observations and descriptions of albinos were generally not based on scientific study, but on the possibly biased and exaggerated reports of travellers and explorers and their local informers. Also, the studies reported here, with few exceptions (such as Okoro, 1975), have been conducted by European workers visiting Africa and have generally not been as extensive nor in such depth as some of the European studies (e.g. Froggatt's (1960) study in Northern Ireland). Nevertheless, this review presents a broad background, for what it is worth, which is both necessary, to obtain an overall view of of the subject, and relevant to the present study.

0 - 1912 AD

Probably one of the earliest references to a group of possible albinos in Africa was that of Pliny, writing in the first century AD. He referred to Leucaethiopes living in North Africa (Plinius Secundus, referred to by Pearson et al, 1911). In 1666 Vossius described the ancient state of knowledge concerning albinos, and according to an extensive footnote in Latin in the Monograph by Pearson, et al, (1911), he stated that not only were albinos, or Leucaethiopes, found in the lands around the fountains of the Nile, but

also in the midlands of Africa. He described them as being so pale that one could see them from afar and mistake them for Belgians or Germans, with their blue eyes and blonde or red hair. Their colour was cadaverous and similar to that of lepers. He also observed that albinos had black parents and that they avoided people as if their condition was contagious. They disliked the sun, and used ointments, oil and fat to cover their skin and maintain good health. These Leucaethiopes were also called `albini', and although nearly blind were said to be very strong.

In the same century Ludolf (in 1691, according to Pearson et al, 1911) referred to the albinos in the court of the King of Loango and attributed the first use of the word `albino' to the Portuguese explorer and historian Balthazar Tellez who travelled through Africa in the early years of the century. In 1688, Da la Croix stated that although albinos were attendants in the King of Loango's court, yet they were regarded as monsters. In support of this view a century later Equiani, an African, is quoted as stating `I remember while in Africa to have seen three Negro children who were tawny and another quite white, who were universally regarded by myself and the natives in general, as deformed' (Pearson et al, 1911). Bowditch (in 1819, according to Pearson et al 1911) found at the court of the King of Ashanti (in present day Ghana) nearly a hundred, thin, diseased negroes of different colours from red to copper and white.

In the mid-nineteenth century David Livingstone, missionary, explorer and doctor, observed people with the condition during his travels. He reported in detail on a boy brought to him in present-day Botswana and stated that `he was remarkably intelligent for his age. The pupil of the eye was of a pink colour and the eye itself unsteady in vision. The hair, or rather wool, was yellow and the features were those common among the Bechuanas' (Livingstone, 1857, p493). The mother of this boy had clung to him for many years against the father's orders and as a result had had to live apart from the father. She eventually tired of this arrangement and murdered the boy. This appeared to be common practice in Botswana and Angola, and Livingstone commented that the authorities took no action against the woman. He did not see as many albinos further North in Africa, but had read, in accounts published by the Portuguese, that they were held in favour as doctors by certain chiefs. Livingstone also noted that the bodies of the albinos he met were always `blistered' on exposure to the sun and that their skin was `more

tender' than that of the blacks. One woman wished to be made black, so Livingstone gave her nitrate of silver, taken internally, but it did not produce `its usual effect'.

In 1909 observations were made during a study on pigmentation and cancer by Watkins-Pitchford, a pathologist in South Africa, who in 1913 became the first Director of the South African Institute for Medical Research. He commented that the true albino should display the greatest liability of all to cancer, since this liability appeared to increase as pigmentation decreased. He added that rodent ulcers, for example, were rarely seen in the `natives' of Natal and that upon diligent enquiry only one such case was reported and that was in an albino.

Early in the twentieth century Pearson et al (1911, p112-166) attempted to determine the geographical distribution of albinism in Africa and they published their findings in `A Monograph on Albinism in Man'. On the basis of published articles and on reports that they received from numerous co-operating medical practitioners and travellers, they compiled a list of the types of albinism and suggested that these might be more easily observed in the black people of Africa than in lighter skinned people. Their rough classification was as follows: (i) the xanthous negro, (ii) the complete albino, with possible sub-classes, (iii) the spotted, freckled negro, (iv) the yellow-eyed, white-skinned negro, (v) the blue eyed, white skinned negro with photophobia and nystagmus, but not red pupils, (vi) the partial albino or piebald negro. They established that albinos occurred throughout Africa and that they had been mentioned and described by travellers for centuries. Albinos were said, by some, to marry and bear normal children (unless they married another albino in which case all offspring were said to be white), and by others to have no posterity. In the Sudan, according to Raffenel (Pearson et al, 1911), they lived without having to work, and asked for whatever they needed with the certainty that they would not be refused. Among the Foulah people of the Sudan everyone apparently gave freely to the albinos to win favour from heaven. The Foulahs were said to believe that God did not wish the albinos to work like other men, so he gave them the colour of whites. In the Foulah language they were called `danedio' which meant white, not `toulako' which meant white-man. In the western Sudan reports asserted that children born from two albinos were black.

In Guinea in 1803, Dr T. Winterbottom observed that the natives considered albinism as a great deformity and a misfortune to the family, but that

none of them appeared to be intellectually retarded (Pearson et al, 1911). Reports of confusion between mulattos and albinos and an attempt by an albino to pass for white in Sierra Leone were also received. Albinism was noted to occur in Liberia, the Ivory Coast, Angola and Madagascar (present day Malagasy). In Ashanti (now in Ghana) and Dahomey the kings who kept albinos at court occasionally offered the females as wives to passing travellers. Albinos were believed to be under special Divine protection. They were also sent by explorers as exhibits to International Exhibitions in London and elsewhere.

Dr Henry Strachan reported in a letter from Nigeria, to Pearson, Nettleship and Usher (1911) that albinism occurred there with its usual problems and variations. Earliest reports from the Congo stated that the albino children, called `Dondos´, were always presented to the King in Loango and that they were then educated as sorcerers and nobody dared offend them. They were sometimes described as incapable of coitus and, in other reports, as not being permitted to multiply. They were generally regarded as somewhat sacred and inviolate, and no man might strike them. Further north, albinos in Gabon were regarded as unlucky and frequently killed, unless missionaries intervened. In Angola, however, albinos appeared to be treated with great respect and fear. From Southern Africa reports were received on albinos in the Transkei, in Basutoland and in Zululand, where red-skinned people were described; infanticide of albinos was reported to occur quite frequently, and men queried the faithfulness of their wives when the baby was a partial albino. In Basutoland, however, albino women commanded greater bride-wealth (lobola) than normal women (Pearson et al, 1911).

According to Pearson et al (1911), Negroes were said to consider albinos as the product of illicit relations between a Negress and a gorilla or baboon, and although no authority was given for this legend, they wrote that it appeared repeatedly in writings on the subject. A similar legend had been reported in Malaysia with the orangutang as the non-human parent.

Dr G A Turner, medical officer to the Witwatersrand Native Labour Association in South Africa, gave Pearson and Usher much information in letters and in a report which was initially sent to the University of Aberdeen. This information does not appear to have been published by Dr Turner. Pearson et al (1911) state, however, that Turner reported on 7 000 males whom he examined for employment on the mines. Among these he found eleven with xanthism, they all had `tawny´ or reddish skin, brown eyes, generally brown

hair (although several had black hair and one had light brown reddish hair on the temples), and only one had nystagmus. Turner only saw one albino in this series of men and commented that infanticide was still commonly practiced, and that albinos were unlikely to seek work on the mines, if they could exist elsewhere. The family histories he collected on the xanthic individuals suggested that there could be a connection between albinism and xanthism as the two conditions were occasionally found together in members of one family. Turner wrote that he had seen albinos in all the principal tribes of South Africa, except the Bushman (San), Hottentots (Khoikhoi) and Griquas. Affected individuals appeared to get on quite well in their communities, and yet a man would not sell one of his daughters to an albino man, and young men were frightened to take an albino girl.

Sir Harry Johnston, who worked in British Central Africa, reported that albinism was not uncommon there (Pearson et al, 1911). He added that the type where the skin and iris were quite light yellowish-brown was much admired especially in women, for there was a tendency to admire a lighter rather than a darker skin, and the wives of chiefs often had this pale yellow-brown colouring.

Albinos were also reported from Kenya and Uganda, where they were apparently looked upon as curiosities and kept in the establishments of kings and great chiefs. In Somaliland a report on the ethnographic characteristics of the people included a comment that there was a tendency to admire and accordingly to marry into the lighter shades (Paulitschke, referred to by Pearson et al, 1911).

From their review of the statements and descriptions of the 213 albinos reported to them from Africa, Pearson et al (1911) concluded that the proportion of `complete' to `incomplete' albinos was 3:1 and that where albinism was prevalent the normal negro appeared to have a lighter skin colour than other Africans.

1913 - 1951

Pearson, Nettleship and Usher's classification of albinos, however, led to some confusion, since the interpretation of the colour of the eye and the presence of the red reflex appeared to be very subjective, causing a lack of standardization in the reported details. Stannus (1913), who wrote a doctoral thesis on the subject for the University of London, therefore, on the basis of his survey in Nyasaland (now Malawi), attempted to be more precise and suggested an alternative classification for albinos: (i) complete: white

skin, straw coloured hair, liquid blue iris with red reflex through pupil, when examined with artificial light; (ia) Spotted: as in (i) but showing spotty pigmentation of the skin; (ii) Partial incomplete: as in (i) but the iris light yellow or hazel, photophobia and nystagmus not always present; (iii) Xanthism: reddish-brown hair and skin, light brown irides; (iv) Incomplete: skin light brown, not red, hair dirty light yellow, not brown; irides hazel; (v) Partial albinism: including piebalds and spotlings.

Stannus proceeded to describe his subjects in great detail and to classify them. He stated that in all his cases the pupil appeared black in ordinary day-light and that although no pigment was obvious in many cases, yet on microscopic examination some pigment might be demonstrated in hair and eyes. He found that several albinos appeared to be intermediate between two groups. In some families he reported a parent or relative of lighter than average skin colour and in another family he reported the custom of killing the albino at birth. He described the variation of colour in affected siblings and discussed a family where the older child had xanthism, with reddish skin, hazel irides, no nystagmus, light brown hair, while the younger brother had lighter skin, light hazel eyes, nystagmus and light straw-coloured hair. He concluded that: albinism was very common in Nyasaland (although he did not attempt to calculate a prevalence rate); it was a `family affliction' and often directly hereditary; there were varying grades in the same family; the degree might vary with age; pigmented spots were common; lanugo body hair was generally associated with the condition; and maldevelopment of the teeth might be found.

McCrackin (1937), who worked in the French Cameroons, reported two cases of twins, one in which both had albinism and the other in which one twin was an albino and the other normal. He also commented upon local superstition stating that albinism was said to be caused by maternal impression, or by the mother conceiving the baby by a water spirit, or as a result of adultery, or the sins of one or both parents, or the stepping over instead of walking around, the legs of an albino, while pregnant. Heredity was also recognized and said to be the biggest single factor held strongly by local tribes. McCrackin had observed `semi-albinos' with brown hair, reddish skin, pale blue eyes and no photophobia or nystagmus, and added that the apparent mental and physical slowness was probably due to the `psychological effect of the albino being regarded as inferior and ridiculous, from the time he is born, and to his difficulty in using his eyes for gaining knowledge'. He reported

on a case of a father who, as recently as 1924, intended killing his children if they were albinos. This man's first child was an albino, whom he found he could not murder, but the child was apparently poisoned by well-meaning friends and died.

A report on `partial' albinism and nystagmus in two Nigerian families was made in 1944 by Loewenthal. He defined his term `partial albinism' as referring to persons `in whose skin the pigment content is universally reduced though not completely absent'. He stated that Turner (the South African medical practitioner referred to by Pearson et al, 1911) was the originator of the word `Xanthism',and drew a pedigree of one of Turner's affected families (from apparently unpublished information) showing red, yellow, normal and albino negroes in one family. Loewenthal himself had encountered a `red' albino with bright copper-coloured skin, lentigines, ginger hair, reddish-brown irides and nystagmus, and a `yellow' albino, with clay-coloured skin, lentigines, black and gold hairs, hazel irides and nystagmus. He concluded that because of the presence of nystagmus these individuals should be considered as albinos.

Vallois, an anthropologist, visited Cameroon and on hearing that albinism was quite common there, cursorily investigated the matter. He found four albinos in the villages of Badjoui, which contained about 10 000 individuals, and in another area he found a further four among 13 000 people (Vallois, 1950). He observed that males appeared to be affected more frequently than females and that affected people did not have the right to marry. One albino he came across, however,was a sorcerer, who had given himself the right to marry and had had normal children. Vallois also described and measured skin colour in black infants and found that they were very light at birth and only darkened at about three days of age.

1952 - 1972

Barnicot (1952) appears to have been the first person to attempt a study to estimate the frequency of albinism in Africa. He believed that previous studies had been based mainly on casual observation and, although scientists had claimed that albinism was commoner in Negroid than Caucasoid people, the available data were inadequate to prove the point. In Nigeria he found a frequency of one albino in 2 858 school children, which was high in comparison with European rates, which varied, he said, from one in 20 000 to one in 40 000. In a local Nigerian township Barnicot estimated a frequency of one albino per 5 000 people. He noted: that all the albinos he saw could be

classified as incomplete in Pearson's terminology, since all had some pigment in the skin, hair, and especially iris; that their ages were not markedly divergent from those of the general population; and that there was an excess of males over females (although this difference was not statistically significant).

On detailed examination of a sample of his albinos, Barnicot found that twenty-five of twenty-nine had pigmented freckles, that these were commonest on cheeks and forehead and tended to develop with age. On examination of a biopsy of one of these freckles and adjacent skin, melanoblasts were found to be as abundant in the lightly as in the heavily pigmented skin, but there was heightened pigmentary activity in the heavily pigmented areas. Freckles were not a consistent finding in affected family members. The irides of his subjects were found to be on average darker than in European cases, but the darkest was light orange-brown in colour,which was considerably lighter than the lightest eyes in normal Africans. Irides which were green or blue frequently had orange-brown pigment at the pupillary margin or brown flecks. There was a slightly higher frequency of blue eyes in the youngest age group (0-10 years) suggesting that eyes darken with age, but no sex difference was suggested. Nystagmus was obvious in thirty-six of thirty-eight cases, the two unaffected subjects were aged thirty-five and seventy-five years,and it seemed that nystagmus improved with age. No systematic study of strabismus or other visual defects was made. The hair colour ranged from white tinged with yellow to yellowish-brown, but there seemed to be little evidence of a correlation between either hair colour and age or between hair and eye colour.

Barnicot collected hair specimens from a group of albinos and a group of normally pigmented relatives for reflectance spectrophotometric studies. The samples from the two groups differed significantly at all wave-lengths and the difference was greater at the red end of the spectrum. Barnicot also observed that individual albinos could have patchy hair of light and darker colours and on microscopic examination that many (nine of thirteen) had alternate light and dark banding on a few of their hairs.

Information on skin cancer and the susceptibility of albinos was published by Cohen et al (1952) who studied malignant disease and skin cancer in the Transvaal. They found that African albinos accounted for one fifth of all skin cancer in the 'Bantu people', which was grossly disproportionate to the occurrence of albinism, and that younger age groups and, generally,

exposed areas were most often affected. As prophylactic treatment they suggested that adequate headgear, protective clothing, the use of cosmetic ointments, and perhaps the growth of beards, would afford some protection. They also emphasised that early treatment of hyperkeratoses would prevent the development of skin cancer.

During a subsequent study of his albino sample in Nigeria, Barnicot (1953) investigated and described twenty-three xanthous individuals. He found that the relation between hair and skin colour was not always close, and one of his subjects had light yellowish-red hair with normal skin colour, while another had brownish-red hair and copper-coloured skin. Freckling appeared in only three subjects and eye colour ranged from medium brown to `as dark as in the general population'. Nystagmus was present in only one of twenty-three subjects. In a school survey, which he conducted, he found that red hair occurred in about one in 650 children. He commented that the name `xanthism', (which he attributed to Pearson et al, 1911) was not well chosen because in the majority of cases the skin and hair were not yellow. He suggested that further recording of hair and skin colour by spectrophotometry was required.

Further comments were made on the albino skin by Schrire (1958). He mentioned the anecdotal evidence of a surgeon in Johannesburg, who had a large experience with albino `Bantus', and had said that they died early of cancer of the skin. The epitheliomas usually occurred on the shoulders and forearms (areas exposed to trauma and to the sun),especially in the young who were often completely unclothed for many years.

Cancer researchers have played an important role in describing albinism and its associated problems in South Africa. Oettle (1963), in the course of his study on skin cancer in Africa, attempted to collect information on albinos to estimate the frequency of the condition in the rural areas of the Transkei. He ascertained 197 albinos and estimated a minimum rate of 1:3759. The sex ratio in this group was 0.89, however males tended to migrate to the cities in search of employment and there were also fewer males in the general population. He was unable to offer an explanation for the high rate, but proposed that if heterozygotes had a paler skin they would have great selective value, as this was regarded as a mark of beauty and might therefore represent a selective advantage over the normal population.

Burrell, who was engaged in a survey of oesophageal cancer in the Transkei, proceeded to collect further information on albinos in many districts,

but did not publish his findings. Rose (1973) had access to Burrell's register of albinos compiled in 1964, and reported that Burrell had found 458 cases in Transkei. Rose examined seventy-one of these albinos in forty-one families and on clinical examination alone stated that all appeared to be of the tyrosinase-positive type, and most had `splotchy' freckles and blue to yellowish-khaki coloured eyes. People with brown eyes, yellow hair and red skin were brought to Dr Rose while she examined the albinos, and she found that, when young, their skin was sensitive to the sun. Some of these red-skinned people, who are called by the same name as albinos in the Transkei, were reported in Burrell's register of albinos. In a subsequent report, Rose (1974) made an unsubstantiated statement that all the reds in the sample of families she examined, had albino relatives and most of the albinos had a red relative (a similar situation in two families had been described by Loewenthal, 1944). She therefore concluded that the reds had a form of partial albinism. On her retirement in 1980 Dr Rose handed over Burrell's material to the writer.

Burrell's papers proved interesting since they not only provided lists of albinos in many of the districts of the Transkei, but also the first seven pages of Burrell's preliminary report (which was apparently never completed). In these pages he discussed the ages of the subjects, and the nature of the different skin colour and textures in relatives of albinos and others, as distinguished by the local Xhosa people. Burrell ascertained 458 albinos and the population of the area, according to the National census of 1960, was 1 621 200, which gave a prevalence rate of one albino per 3 710 of the population. This figure confirmed Oettle's (1963) original estimate on his smaller sample. From his analysis of the ages of albinos Burrell stated that their population pyramid was similar in pattern to that for the general population, so that there was no indication that albinos died prematurely in the Transkei. He added that only one case of skin cancer was detected, but that many albinos suffered from solar dermatitis, especially in the winter when they sought the sun for warmth. He compiled pedigrees on twenty-eight affected families and found that in seventeen families dusty, light brown skins (which Burrell attributed to Hamitic vestiges) were found in one or more grandparents, parents or siblings. The classical signs of photophobia and nystagmus were recorded in less than half the 458 albinos, but since these observations and records were made by poorly trained field assistants,

probably without any equipment, this estimate might be unreliable. Eye colour was assessed in a similar fashion and 13.2 per cent of his subjects were reported as having pink/red eyes, 40.3 per cent green eyes, 17.6 per cent blue eyes and 28.9 per cent brown eyes. The albinos were often named `wise little person' since they tended to stay in the shadows close to the hut and to overhear conversations. They were certainly not `dull-witted' but the older ones were considered to be recluses. Teachers however, found them above average scholars. There were 248 affected females and 210 affected males in the sample. On further investigation of his ideas on skin colour, Burrell compared the findings on albino relatives with those for their neighbours in whom there was no history of albinism. He classified subjects as having black, light brown (dusty or smooth) and ordinary skin, and found that only 0.3 per cent of relatives of albinos had black skin, whereas 4.6 per cent of their neighbours had black skin. His method of classifying skin colour was rather arbitrary, but nevertheless as an exploratory study, and in view of the reports of light skin in parents of albinos from other studies (Pearson et al, 1911), his findings are worth mentioning here.

Another attempt at estimating prevalence rates of albinism in Negro groups in South Africa was made by Hitzeroth and Hofmeyer (1964). These two research workers collected their data from the passport-type photographs of African females presenting to the Reference Bureau for purposes of registration. They found an average prevalence rate of 1 in 14 833 with figures ranging from 1 in 9060 in the Tswana, 1 in 9 444 in the Swazi, 1 in 15 705 in the Zulu and 1 in 16 858 in the Xhosa, to 1 in 46 991 in the Tonga. Although their method of ascertainment was somewhat inadequate, since women for various reasons do not all present for registration, and judging skin and hair colour from photographs could be quite unreliable, their figures (which represent minimum estimates) are useful for purposes of comparison with those found in the present study. When comparing the frequencies in the different ethnic groups, by means of chi-square analysis, Hitzeroth and Hofmeyer found that there were significant differences between the Zulu/Xhosa and the Tswana, and between the Xhosa and Swazi. They stated that there was no evidence to support the hypothesis that a differential mutation rate between the ethnic groups might explain the varying frequencies. But they added that marriage customs might differ and these could influence the frequency of albinism. In the Sotho (e.g. Tswana) ethnic groups the custom was for a man to marry the daughter of his maternal uncle, whereas in the Nguni groups (e.g.

Zulu, Xhosa, Swazi) exogamous marriages were the rule (Schapera, 1937). They were puzzled however that of the four groups with the highest frequencies (Tswana, South Sotho, Ndebele and Swazi) one (the Swazi) was of Nguni origin. They suggested that pedigrees should be collected to estimate inbreeding and to determine the selective disadvantage under which albinos reproduce, and that the social status of the albino should be investigated to assess his reproductive chances and popularity (if he is considered to be of supernatural origin) or otherwise (if the condition is considered an abormality), as a marriage partner.

1973 - 1984

A small study conducted by Dogliotti in 1973 on the treatment of solar keratoses in albinos included probably the first assessment of the tyrosinase status in nine African subjects. Hair-bulbs were sent to Professor C J Witkop in Minnesota, USA, for this purpose, and the results indicated that three subjects had tyrosinase-negative and six tyrosinase-positive albinism. This was the first attempt at classifying African albinos by biochemical means.

Probably the first systematic psychological evaluation of albinos in Africa was presented by Manganyi, Kromberg and Jenkins (1974). They investigated intellectual maturity (since no locally validated IQ testing instruments were available for intelligence testing) and body image boundary differentiation, in twenty-eight South African albinos and a carefully matched normally pigmented control group. The albinos were found to be slightly more intellectually mature than the controls and to have a more clearly defined, although still rather diffuse, body image boundary. Compared to the controls they appeared to have a negative self-evaluation, which tended to corroborate Burrell's description that many were recluses.

A series of Nigerian albinos was studied with a view to registering them early in life, counselling their families and advising on methods of treatment and prevention of associated problems (Okoro, 1975). Okoro found that skin cancer was a major problem and consequently life expectancy was reduced. There appeared to be a higher prevalence in Southern Nigerian population groups than in the Northern groups. In the South consanguineous marriages were favoured to preserve the assumed superior qualities of the people living there. He also observed that the proportion of married albino women was very low, that albinos were taunted, dropped out of school because of both uncorrected refractory errors and discouragement, and had difficulty in ob-

taining employment. Socially, life was very difficult for them.

Okoro's work was followed by three subsequent Nigerian studies. The first described the skin cancer (particularly squamous cell carcinoma) in Ibo albinos (Witkop et al, 1977). The second investigated chromosome breaks and sister chromatid exchanges in Nigerian albinos, and found that there was no difference in the frequency of these breaks and exchanges in fourteen albinos compared to a smaller sample of normal controls (Cervenka et al, 1979). The third study involved the investigation and classification of seventy-nine Nigerian albinos (King et al, 1980). Tyrosinase-positive albinism was found in fifty-six albinos, and the remaining twenty-three were reported as brown albinos. The latter type of albinism had been described by Barnicot (1952), Pearson et al (1911) (who had presented descriptions of one brown albino, provided by the explorer Sir Richard Burton, and of other brown albinos in a group of subjects with what was then called `Xanthism'), and Stannus (1913) who had classified them as a separate group of `incomplete albinos'. The brown albinos, however, had not been characterised in detail until the study of King et al (1980). It is noteworthy that no tyrosinase-negative individuals were found in this sample of Nigerian albinos. This may have been by chance, since only seventy-nine subjects were examined. Nevertheless, the finding indicates that tyrosinase-negative albinism is either rare in Nigeria, or that the sun's effects (in an equatorial region), and/or their eye problems, are so severe in this type of albinism, that affected individuals do not leave the shelter of their huts nor present themselves for investigations.

The adjustment of thirty-five young South African albinos and thirty-five normally pigmented controls was explored by the present author and attitudes towards albinos were examined (Kromberg, 1977). The two groups showed similar general levels of adjustment. The albino's specific problems were associated with the physical side-effects of their condition, with their difficulty in obtaining employment, and with the community belief that albinos do not die natural deaths. There was much ignorance regarding the cause of albinism, but attitudes to albinos were generally found to be positive, although the word `nkau' (monkey) was frequently used when referring to them.

Albinism in Cameroon was investigated by Aquaron, Giraud and Battaglini in 1978 and again by Aquaron in 1980. The former group of workers reported on three pairs of twins with albinism and two pairs with unialbinism (one twin albino and the other normally pigmented) in the Bamileka tribe, in

which there appeared to be a high rate of consanguineous marriages. Blood groups were studied and zygosity in the twins was determined. Aquaron (1980) examined 216 albinos and calculated the prevalence rate in one area inhabited by the Bamileka tribe as 1 in 3 800. There were more males than females affected, and this difference appeared to be significant when compared with the sex ratio in the general population. Albinos also had a shortened lifespan and the tyrosinase-positive type appeared to predominate. In a subsequent report on this group, a family containing albino twins with a rufous mother and sibling was described (Aquaron, Ronge and Aubert, 1981). This report is worth noting because of the similar reports of these two conditions occurring together in one family in other parts of Africa (Rose 1973, Loewenthal 1944, Pearson et al, 1911). Biochemical studies were carried out on on the Cameroon albinos, and studies on blood groups showed no significant difference between albinos and controls (Mallet and Aquaron, 1983, Aquaron et al, 1984). In investigating the history of the Bamileka tribe Aquaron (1980) found that around the beginning of the century the tribe had had two polygamous albino chiefs who had had many children, so that founder effect for the gene in this population was clearly illustrated.

A small study on skin cancer in albinos, treated between 1973 and 1979 in Tanzania, was reported by Alexander and Henschke (1981). They reviewed ten cases of advanced squamous cell carcinoma and found that radiotherapy could produce acceptable results, but that education of patients, concerning avoidance of sun exposure particularly during peak intensity (10.00 to 15.00 hours), the wearing of protective clothing, and the use of sun-screening agents, would, perhaps, be the most practical method of preventing skin cancer in albinos.

Two recent papers dealt with albinism in South Africa (Kromberg and Jenkins, 1982 and 1984). The first of these reported data from the present study on the prevalence of the condition in South Africa. In the second study the issues to be covered in genetic counselling were investigated. The findings indicated that young albinos needed counselling concerning the cause of albinism, the associated physical problems, and the discrimination against them especially as marriage partners and employees. The implementation of a broadly defined genetic counselling programme, as advocated by Frazer (1974), could assist the albinos in all these areas.

This review of the African studies on albinism indicates that several different aspects have been investigated, some in greater detail than others.

The prevalence has been determined in Nigeria, in the Transkei, in the Cameroon, and in an urban area of South Africa, and rates appear to be similar in these countries (i.e. around 1 in 4 000). Several different types of African albinos have been described for probably at least a century, but recent studies suggest that the tyrosinase-negative type is rare and the tyrosinase-positive and brown albinos much more common. Skin cancer has been described as the albinos' major problem, and is probably associated with a shortened life-span (found in Nigeria and Cameroon, but not in Transkei). Skin colour in the families of albinos has been described in passing in various reports and these suggest that it is lighter than in the general population, but no studies on skin colour using measuring instruments have been conducted. It has also been suggested that a light skin colour has been looked upon as beautiful in some areas of Africa, as far apart as Somaliland and the Transkei, for many decades. Fertility has also only been mentioned in passing and the albino has been reported to be impotent, infertile or normally fertile. Psychosocial reports are rare and only present findings concerning the average intellectual ability, the negative self evaluation, and generally normal level of adjustment of albinos. This review therefore serves to identify the gaps in the African research work, indicates that several questions remain unanswered and that further investigations are required.

2.1.3 Definition, classification, description and clinical problems

In 1971 Witkop used what he termed the relatively imprecise definition of albinism as a hereditary defect in the metabolism of melanin resulting in a decrease or absence of this pigment in skin, mucosa, hair or eyes. However, in 1983, Witkop, Quevedo and Fitzpatrick stated that `the use of the term albinism should be restricted to congenital heritable hypomelanosis that is limited to the eye (ocular albinism, OA) or involves the eye and integument (oculocutaneous albinism, OCA), and in which nystagmus, photophobia and decreased visual acuity are present'. They specified, that the terminology used in classifying the different types of albinism should indicate the extent and nature of the pigmentary involvement, wherever possible. In the heterogenous group of disorders called albinism there are two main categories, i.e. the metabolic defects involving either the entire melanocyte system (OCA) or the melanocytes at a specific site (e.g. OA). The present study is concerned only with the former category.

Ten different types of OCA have been described and these may be differentiated from one another by their clinical, biochemical and genetic characteristics. Witkop, Quevedo and Fitzpatrick (1983) have compared and characterized these ten types and an abbreviated form of their classification, presenting only the factors relevant to the study, appears in Table 2.1 a and b. The Table is in two sections; the first (section (a)) includes the commoner types of OCA with which the present study will be concerned, while the second (section (b)) indicates the remaining types (four of which are associated with specific syndromes) which will not be investigated during the present study.

Subsequent to Witkop et al's (1983) classification King and Olds (1985) have presented a paper wherein they use the following classes: Type IA (tyrosinase-negative) and Type IB (yellow mutant, thought to be allelic to Type IA) with low or no measurable hair-bulb tyrosinase activity, and heterozygotes could be detected with a hair-bulb tyrosinase assay; Type II (tyrosinase-positive) with moderate to high tyrosinase activity, and heteroygotes could not be detected; Type III (minimal pigment), a new type of albinism, phenotypically like Type IA but with a pigment-ring in the irides (as in the Type II B described by King and Olds, 1982), had low tyrosinase activity and heterozygote levels were useful in detecting this type; Type IV (Brown); Type V (Red); Type VI (Hermansky-Pudlak syndrome) with moderate to no measurable activity and heterozygotes could not be detected; and Type VII (autosomal dominant). Witkop's et al's (1983) classification, has been used during the present study with the addition of the Type II B (as described by King and Olds, 1982, and called Type III, minimal pigment, in their 1985 paper).

There is still some difficulty in classifying an individual albino unless all the relevant symptoms are manifest and full information, on the characteristics and the necessary test results, is available. For example, a yellow mutant (ym) albino may appear very similar to one with tyrosinase-positive (ty-pos) albinism and, if no reliable details are available concerning the colour of the hair and skin at birth in the ym, or if the hair-bulb incubation test result is equivocal, it could be impossible to classify the individual correctly. Similarly, there appears to be some overlap between the brown albino (BA) and rufous (RA) classes which, possibly together with the ym group, were, in the past, often termed Xanthous albinos. The rufous or red albinos, for example, have been reported in Papua New Guinea to have

TABLE 2.1(a) Characteristics of 5 commoner recessive types of OCA (after Witkop et al, 1983)

Characteristic	Ty-negative	Ty-positive	Yellow Mutant	Brown	Rufous
Hair colour	White throughout life	White, yellow-tan and darkens with age	White at birth and yellow red by six months	Beige to light brown in Africans	Mahogany red to deep red
Skin colour	Pink to red	Pink-white to cream	White at birth, cream, slight tan on exposed skin	Cream to light tan on exposed skin	Reddish brown
Pigmented, nevi and freckles	Absent	May be present and numerous	Present	May be present	May be present
Susceptibility to skin neoplasia	++++	+++	Unknown	Similar to Caucasians in Africa +	
Eye colour	Grey to blue	Blue, yellow-brown, age and race dependent	Blue in infancy and darkens with age	Hazel to light brown	Reddish brown to brown
Transillumination of iris	No visible pigment	Pigment cartwheel effect	Cartwheel effect in adults	Cartwheel effect	Slight
Red reflex	Present	May be absent in dark-race adults	Present	Present in children, may be absent in adults	Unknown
Fundal pigment	0	0 to + in adults	0 to + in adults	+ to ++ in adults	+ to +++
Nystagmus	++++	++ to +++	+ to +++	+ to ++	0 to ++
Photophobia	++++	++ to +++	+ to ++	+ to ++	0 to ++
Visual acuity	Most legally blind, constant or worse with age 20/200 to 20/400 +	Children, severe defect, adults same or better 20/60 to 20/400+	Same as Ty-negative. 20/90 to 20/400 May improve with age	20/30 to 20/100	Normal to 20/100
Incubation of hair bulb in tyrosine	No pigmentation	Pigmentation	None to questionable increase	Pigmentation	Pigmentation
Other	Heterozygotes have less than half normal tyrosinase activity	Probably a heterogeneous group		To-date seen only in Africans and New Guineans	Seen in Africans and New Guineans

TABLE 2.1(b) Characteristics of 5 rarer types of OCA (after Witkop et al, 1983)

Characteristic	Hermansky-Pudlak syndrome	Chediak-Higashi syndrome	Cross syndrome	Autosomal dominant OCA	Black locks - albinism syndrome
Hair colour	White, red, brown	Blond to dark brown, steel grey tint	White to light blond	White to cream with reddish tint	Snow white with pigmented locks
Skin colour	Cream grey to light normal	Pink to pink white	Pink to pink-white	White to cream	White with melanized macules
Pigmented, nevi and freckles	Present	Present	Present	May be present	May be present in macules
Susceptibility to skin neoplasia	+++	++	Unknown	Unknown	Unobserved but probably ++++
Eye colour	Blue-grey to brown, age and race dependent	Blue to dark brown	Grey-blue	Grey to blue	Grey to blue
Transillumination of iris	None to cartwheel effect	Cartwheel effect to normal	Unknown; cataracts	Translucent to cartwheel effect	No visible pigment
Red reflex	Present in light Caucasians, not in dark	Present, less after five years	Unknown; cataracts	Present in children	Present in children and adults
Fundal pigment	0 to + in adults	+ to +++	Unknown; cataracts	0 to +	0
Nystagmus	+ to +++	0 to ++	+++ to ++++	++ to +++	++++
Photophobia	+ to ++++	0 to ++	Unknown	++ to +++	++++
Visual acuity	20/70 to 20/400	Normal to moderate decrease	Blind	20/70 to 20/200	20/300 to 20/400 +
Incubation of hair bulb in tyrosine	Pigmentation	Pigmentation	Pigmentation	Pigmentation	No pigmentation in white hair, pigment increase in pigmented hair
Other	Platelet defect Ceroid storage	Susceptible to infection, often fatal in childhood	Oligophrenia; microphthalmia; gingival fibromatosis; athetosis; mental retardation		

hair colour ranging from fair to red-brown (Walsh, 1971, Harvey 1971). Classifying an albino who has fair hair and a light reddish-brown skin therefore presents a problem, since the assessment of the hair and skin colour tends to be rather subjective, and the individual could be grouped with BA or rufous subjects. Jay, Witkop and King (1982) also found some difficulty in classifying all of the 201 patients in their series in England, and a definitive diagnosis could not be made in thirty-six patients. Van Dorp et al (1983) suggested that to compensate for the 'wide diversity in albino expression', diagnosis should be based on the results of combinations of tests.

Features of albinism differ in the Caucasoid and Negroid albinos, and in the ty-pos type the intensity of the pigment which accumulates with age is dependent on the race of the subject. Phenotypically the clinical characteristics of the ty-pos albino may overlap with those of the ty-neg subject, and in the African albinos the red reflex may be absent in adults with the ty-pos type (Witkop et al, 1983).

Clinically, the person with any type of albinism has several problems associated with the hypopigmentation of the eyes and skin. Visual problems include nystagmus (the eyes move in a more or less rhythmical manner, from side to side, or in a rotary manner from the original point of fixation, McNalty, 1965), photophobia (abnormal intolerance of, or sensitivity to light, McNalty, 1965), strabismus (squinting) and reduced visual acuity. Surgical correction of the strabismus has been attempted but is invariably unsuccessful in restoring binocular vision, because of the underlying abnormality in the optic pathway (Witkop et al, 1983). Dark glasses and occasionally tinted contact lenses are used to reduce the photophobia. Since near vision is reasonably good in most albinos (except perhaps those with the tyrosinase-negative type) they should be able to cope in ordinary schools with the sympathetic help of the teachers. The majority of albinos (excluding perhaps the brown and rufous types) will, however, not have sufficient vision to enable them to drive a motor-car (Taylor, 1978). Skin problems include the development of erythema (redness), lentigines (or ephelides, deeply pigmented areas with irregular borders), freckles (small round pigmented spots), pachyderma (thickening and wrinkling of the skin) and keratoses (premalignant or malignant lesions). The Negroid albinos frequently dislike the appearance of lentigines for cosmetic reasons, and because in Africa they can hardly avoid sun exposure, the solar keratoses often become malignant. Preventive treatment consists of the application of anti-actinic

barrier creams, but their long-term effectiveness has not been definitely established (Fitzpatrick, 1981).

The intelligence of Negro albinos in USA has been shown to be within the normal range (Beckham, 1946), as is the intellectual maturity in South African albinos (Manganyi et al, 1974). Nevertheless, psychological problems seem to exist (Beckham, 1946; Stewart and Keeler, 1965; Woolf and Dukepoo, 1969), and assumptions are sometimes made regarding albinos' abilities or the lack of them which may or may not be justified (Cameron, 1979). Counselling should be supplied where necessary (Kromberg and Jenkins, 1984), and community education on the nature of albinism should be more widely offered.

Marriages between albinos have been reported in which normally pigmented off-spring were produced (Witkop, 1971; Trevor-Roper, 1952). The albino couples in both these reports were composed of one partner with ty-pos albinism and the other with ty-neg albinism. Non-paternity was excluded and these findings indicated that the genes for the two types of albinism were not allelic. It has been suggested, however, that the ym and ty-neg forms are due to genes which are allelic (Warren, 1981, Hu et al, 1980).

Chemical detection for the heterozygote state for the ty-neg type was developed by King and Witkop (1977), using anagen hair-bulbs. Van Dorp et al (1982), however, found an overlap in hair-bulb tyrosinase values in heterozygotes and controls and concluded that it was therefore not possible to detect heterozygotes for ty-neg albinism using this test. A further recent study on a large sample of albinos and heterozygotes (King and Olds, 1985) nevertheless confirmed that ty-neg and ym heterozygotes can be detected by determining hair-bulb tyrosinase activity.

Prenatal diagnosis of albinism has been discussed and some parents, who would not take the one in four recurrence risk, would consider another pregnancy if this option was available. Haynes and Robertson (1981) state that diagnosis of albinism is theoretically possible prenatally. A scalp biopsy could be taken from the foetus during the second trimester of pregnancy and the hair follicles could be examined for pigment. These hair follicles have been found to be present in the scalp as early as sixteen gestational weeks so that a diagnosis could theoretically be achieved in time to allow selective termination if requested. The enzyme lacking in the ty-neg type is tyrosinase, but this enzyme is not expressed in cultured cells, and the genetic defect in ty-pos albinism is unknown. Prenatal diagnosis employing the assay

of the enzyme in cultured amniotic cells (as is sucessfully done for the enzyme hexosaminidase in the prenatal diagnosis of Tay Sachs disease) is therefore not possible.

Eady et al (1983) however sucessfully diagnosed albinism prenatally by electronmicroscopic examination of a sample of foetal scalp skin obtained in utero at twenty weeks gestation. The foetal hair-bulb melanocytes were found to have melanosomes which had progressed no further than stage II in development, although melanogenesis is normally active at twenty weeks gestation. In four control foetuses many stage IV melanosomes were observed. Termination of the pregnancy was requested and the diagnosis was confirmed in the twenty-two week foetus.

A recent study has suggested that the ty-neg albinism gene is linked to autosomal recessive cerebellar ataxia, but this linked group has not yet been assigned to a specific chromosome (Shows, Sakaguchi and Naylor, 1982).

2.1.4 Prevalence

The prevalence of albinism varies according to population group (see Table 2.2). In Europe the overall frequency is about 1:20 000 with estimates ranging from 1: 10 000 in Norway (Magnus, 1922) to 1:29 000 in Italy (Raseri, 1879, cited in Pearson et al, 1911). In contrast, the frequency among the Cuna Indians of San Blas Province, Lower Panama, is 1:200 (Keeler, 1964) and among the Hopi Indians of Arizona it is 1:227 (Woolf and Grant, 1962). In Africa prevalence estimates range from 1:5 000 in Nigeria (Barnicot, 1952) to 1:2875 in Cameroon (Vallois, 1950).

Various suggestions have been put forward to explain the high rates in groups such as the Hopi Indians. One such proposal is based on the cultural habits of the people, and it has been said that the albinos were not required to work in the fields with the other men and therefore they stayed at home and produced more children than their non-albino counterparts (Woolf and Dukepoo, 1969). Mansell (1972) proposed that, until recently, albinos have been regarded as holy beings unsuited for normal village life. He added that this was the case in some South American villages where albinos were `venerated and lead lives of cloistered luxury´.

The question of why prevalence rates in Africa are at least three times higher than those in Europe does not seem to have been tackled. Oettle (1963) however has suggested the possibility of heterozygote advantage. Random genetic drift in the form of founder effect is another possible reason (Kromberg and Jenkins 1982).

TABLE 2.2 Frequency of albinism in different populations

Population	Frequency	Source
Europe		
Russia	1:100 000	Pearson et al, 1911
Italy	1: 29 000	Pearson et al, 1911
Scotland	1: 12 000	Pearson et al, 1911
Norway	1: 9 650	Magnus, 1922
Holland	1: 20 000	Sanders, 1938
Northern Ireland	1: 10 000	Froggatt, 1960
America		
Hopi Indians	1: 227	Woolf and Grant, 1962
British Columbia	1: 20 600	McLeod and Lowry, 1976
USA	1: 16 800	Witkop, 1983
Caucasian	1: 19 000	Witkop, 1983
Afro-American	1: 10 000	Witkop, 1983
Africa		
Cameroon	1: 2 875	Vallois, 1950
Cameroon (Bamileka)	1: 3 800	Aquaron, 1980
Nigeria	1: 5 000	Barnicot, 1952
Southern Africa		
Transkei and Ciskei	1: 3 759	Oettle, 1963
Transkei	1: 3 000	Rose, 1974
Soweto, Johannesburg	1: 3 900	Kromberg and Jenkins, 1982

The prevalence rates of the ty-neg and ty-pos types of albinism have been investigated in a few populations, and generally the former is much rarer than the latter type. According to Witkop et al (1970) from all the sources tested at that stage (100 subjects), 27 per cent were ty-neg and 73 per cent ty-pos albinos. There appear however to be racial variations and in the same study Witkop found that in twenty-one North American Caucasian albinos there were 67 per cent ty-neg and 33 per cent ty-pos subjects, while in thirty-seven North American Negroes subjects 27 per cent were ty-neg and 73 per cent ty-pos. Since many of Witkop's subjects were selected from a school for the partially sighted his figures for the Caucasian group may have been biased in favour of ty-neg albinos who experience more severe eye defects. McLeod and Lowry (1976) suggest that the ty-neg type is probably less frequent than the ty-pos type in Caucasians, as well as in Negroes. In their sample of forty-six Caucasian albinos in British Columbia they found that

seventeen (37 per cent) were ty-neg and twenty-nine ty-pos (63 per cent). Similarly Van Dorp et al (1983) found that in the Netherlands among seventy-eight albinos seventeen (22 per cent) were ty-neg and sixty-one (78 per cent) were ty-pos.

Among Nigerians ty-neg albinism appears to be even rarer and none were found in a sample of seventy-nine albinos (King et al, 1980). In South Africa, however, ty-neg albinism seems to occur (Dogliotti 1973) and it has also been described in Cameroon (Aquaron et al, 1981).

The findings of many of these studies depended on the method of ascertainment of the subjects, but nevertheless ty-neg albinism appears to be quite rare in Caucasians and much rarer in Negroes, while ty-pos albinism is more common in both the USA and Africa. Detailed studies of the prevalence of other types of albinism have not been carried out. The yellow mutant type is very unusual and has not been specifically described in Africans (Witkop, personal communication, 1984). Brown albinism has only been seen in Africa and New Guinea, and rufous albinism also appears to be much more common in these two areas of the world than elsewhere (Witkop, 1983, and Barnicot, 1953).

2.1.5 Skin Cancer and albinism

The albino's high risk for developing skin cancer has been recognized for a long time. In South Africa, Watkins-Pitchford described the albinos' susceptibility to skin cancer in 1911, and it was well documented in the Transvaal in 1952 (Cohen et al). Oettle (1963) studied skin cancer in seventeen Negro albinos in the Transvaal and found that there were fourteen cases with squamous cell carcinoma and one case each with basal-cell and sweat-gland carcinoma, and melanoma. The fourteen squamous-cell carcinomas represented 9.6 per cent of all such skin cancer found in South African Negroes. The commonest site for the cancer was the head (eleven cases), followed by the upper limbs (three), neck (one), back (one) and fossae (one). King et al (1980) in a study of Nigerian ty-pos albinos noted that solar keratoses were most frequently found on the forearm and hand, closely followed by the head, and less frequently on the neck and back.

Rose (1974) reported that skin cancer appeared to be infrequent in the albinos in the Transkei and that Burrell had only noted one case, among the 309 albinos he examined, and that was a melanoma on the back of the knee of a fifty year old woman. Rose investigated twenty-seven families with albino members and questioned them regarding the cause of death. None reported skin

cancer as a cause.

In Okoro's (1975) Nigerian sample however there were no albinos over twenty years of age who were free from premalignant or malignant lesions, and 50 per cent of his subjects had keratoses and superficial ulcers. He suggested that these destructive lesions were caused by years of exposure to actinic irradiation, which resulted in slight tanning at first, followed by sunburn, blisters, solar elastosis, ephelides, facial lentiginosis, solar keratoses, chronic superficial ulcers and ultimately squamous and basal-cell epitheliomata.

Rippey and Schmaman (1972) investigated squamous and basal-cell carcinoma in black patients at Baragwanath Hospital, Johannesburg, and found 11 per cent of the former type and 41 per cent of the latter type was in albinos. In a more recent study of cancer in urban blacks of South Africa, Isaacson et al (1978) examined cancer records for the ten year period 1966 - 1975 for blacks at Baragwanath. The survey yielded 101 cases of squamous carcinoma of the skin, and seven (or 6.9 per cent of the sample) of these cases were albinos. Basal-cell carcinoma was found in only nine cases and, of these, three were albinos and one a child with xeroderma pigmentosum. Malignant melanoma was found to be very rare and the prevalence rate was 1.65 per 100 000 of the population. No cases were reported in albinos.

2.1.6 Life expectancy

As long ago as 1729 Wafer stated that albinos were but `short-liv'd' (Pearson et al, 1911) and other reports have suggested that their life expectancy is reduced (e.g. Okoro, 1975, Schrire, 1958, Aquaron, 1980). Much depends, however, on where they live and how much care they receive. King et al, (1980), stated that in Nigeria no ty-pos subjects older than forty years of age were found in a sample of seventy-nine albinos, and skin malignancies appeared to be the major factor in causing a limited life-span. It is possible that ty-pos, ym, brown and rufous albinos live longer than those with ty-neg albinism, since the former groups have some little protection from the sun in the form of the pigment which develops, but no data seem to be available. Okoro's (1975) data on 1 000 unclassified albinos, also suggested that life expectancy is decreased. In the East Central State of Nigeria Okoro found that only 10 per cent of albinos were in the age range thirty-one to sixty years, whereas 20 per cent of non-albino skin patients and 20.7 per cent of the normal population in a developing country (unnamed by Okoro) were in that age group. Okoro, however, admitted that his methods

of ascertainment might have caused the older patients to be missed.

Barnicot (1952) on the other hand found that the ages of his forty Nigerian albino subjects were not markedly divergent from those of the general population. There were six or 22.5 per cent of his sample and 20 per cent of the general population in the age range thirty-five years and older. These results are, however, subject to the limitations of his small sample. Burrell (unpublished papers) and Oettle (1963) also found that the ages of their groups of Transkei albinos showed a normal distribution.

Froggatt (1960) studied 122 albinos in Northern Ireland and compared their age distribution with that of the general population. From the figures he presented it is apparent that: 43 per cent of his subjects and 28 per cent of the general population were in the age range 0-14 years; 47 per cent of both groups were aged 15-49 years; 10 per cent of his subjects and 24 per cent of the general population were in the age group fifty plus years. It was evident, therefore, that for the albinos the younger age group was over-represented and the older age group under-represented. Froggatt suggests that there are four possible explanations for this situation. Firstly, the life expectancy of the albino might be less than that of the general population, but clinical examination did not suggest systemic weaknesses or the susceptibility to any life-threatening conditions; nevertheless albinism might be incompatible with normal life expectancy. Secondly, the frequency of albinism might have been increasing, either due to a change in mating patterns, or an increase in the mutation rate, both of which seemed unlikely. Thirdly, the criteria of acceptance into the study might have led to more young than old people being included. It was possible that older people were missed, because if they had white hair they did not attract attention, and old albinos might not have had nystagmus (since it is said to improve with age), the presence of which was one of the criteria for inclusion. Fourthly, the methods of ascertainment meant that the study was more comprehensive for the younger age group.

There is, therefore, still some debate on the life expectancy of both Causasoid and Negroid albinos.

2.1.7 Skin colour and pigmentary changes in heterozygotes

Waardenburg, Klein and Franceschetti (1961) stated that evidence seemed to show that in the dark skinned races heterozygotes for generalized albinism tend to have lighter skin pigmentation than their normal fellows. Keeler (1953) observed lighter skin colour in many sibs, parents and child-

ren of albinos among the San Blas Indians. Harris (1926) was also convinced that albino families in San Blas contained persons of fairer complexion than that of average Indians. No attempt was made by these researchers to measure skin colour however, although reflectance spectrophotemetry was used by Barnicot (1952) and Froggatt (1960) to measure hair colour in albinos and their normal relatives, and by Barnicot (1958) to investigate skin colour in in normally pigmented Nigerians.

Skin colour has been measured by a variety of methods over the years. Probably the first attempt at measurement involved the rather subjective arbitrary categorization of the whole range of colour into a number of shades, and later, sets of colour standards, such as the von Luschan tiles. These were highly unsatisfactory methods both because of the subjectivity of the visual matching and because of the necessity of imposing discrete units upon a natural continuum. Nevertheless, they provided the basis for the subsequent attempts at objective measurements of skin colour, and gave data which indicated how pigmentation was geographically distributed throughout the world (Roberts, 1977). A third method was used with some success by Davenport and Davenport (1910) in determining skin colour in `Negro X White' crosses. Their quantitative data were obtained by means of the Bradley Color top, using the standard colours of the Milton Bradley company of Springfield, USA. In this top, segments of four coloured papers, viz black, white, red and yellow, were sorted in differing amounts and whirled so that they blended into colours ranging from light to dark brown. The comparison between the skin and the colour, however, still had to be made visually and was therefore somewhat subjective, but at least there was a continuous scale of measurement.

None of these methods is now used in studies on skin colour. The portable spectrophotometer surpassed all previous methods of measurement by providing not only a continuous but also an objective and accurate scale of measurement. The original EEL (Evans Electroselenium Limited, London) reflectance spectrophotometer was designed for use in industry. Weiner (1951) decided that, because of its portability and simplicity, it would be useful for anthropological field studies. The results from his trial study showed that the spectral reflectance values obtained with the EEL instrument were comparable to those obtained with the cumbersome and elaborate Hardy photoelectric recording spectrophotometer (Hardy, 1936) used successfully by Edwards and Duntley (1939) for measurement and analysis of the properties of the human skin.

Few quantitative studies of skin colour have been completed in Southern Africa. The one of most revelance to the present study is that of Wassermann and Heyl (1968). They obtained data by means of spectrophotometry on skin pigmentation in South Africa Whites, Cape Coloureds (of mixed ancestry) and blacks. They found that females tended to be lighter than the males in all three groups and that the Cape Coloured group showed a wide overlap in skin colour with both whites and blacks. They stated that `seen through the melanised epidermis, whose maximal absorption due to melanin lies towards the blue part of the spectrum, the increase in reflectance with diminishing concentration of melanin would be relatively greater in this area'. The data they obtained on blacks were from 104 males and 100 females, the majority of whom belonged to the Xhosa ethnic group. There were nineteen females who were pregnant and probably should have been excluded, since skin colour has been shown to darken in pregnancy (Rook, 1969, p145). Their results on the females are therefore slightly biased but nevertheless are useful for the purpose of comparison in the present study.

Tobias (1961) also used the EEL instrument and published quantitative data on skin reflectance in the San (`Bushmen') and San-European hybrids. Weiner et al (1964) took measurements on the skin colour of four different groups, the Bushmen, San, Khoikhoi (`Hottentots'), hybrids and Okavango Bantu-speaking Negroes, most of whom were living in South West Africa/Namibia. Although skin reflectance can be measured at nine different wavelengths, the reflectance at 685 mu (609 filter) was the only measurement systematically recorded in these samples, probably because reflectance at this wavelength gives a good index of the melanin content of the skin (Harrison and Owen, 1956).

Barnicot (1958) carried out his studies on skin colour on two indigenous groups (both Yorubas), in the summer in Southern Nigeria. He compared the two groups with one another, with Europeans, and with a small sample of African-European hybrids living in the Niger Delta region. He took his measurements on the flexor surface of the right forearm and most other workers (eg Weiner et al 1964) have preferred the medial aspect of the left upper arm which is less exposed to the effects of tanning.

A study which investigated the effect of homozygosity and heterozygosity for the phenylketonuria gene on skin colour in one Yemeni family was carried out by Roberts (1977). He found that the affected child had appreciably lightened skin pigmentation, in comparison with his father and mother, when mea-

sured at nine different wavelengths. This work appears to be the only one in which a recessive genetic disorder and skin colour were studied in homo- and heterozygotes, although the connection between skin colour and psychiatric illness (Robins, 1970), and between skin colour and vitamin D resistant rickets (Richardson, 1974) have been investigated in South Africa.

Various studies have attempted to examine heterozygotes for the albinism gene to assess whether or not pigmentary changes occur in them. Waardenburg (1947) found that in obligatory heterozygotes the iris was translucent in some but not all cases of oculocutaneous and ocular albinism, and he suggested that this information was of practical use for genetic purposes. Froggatt (1960) also found that although abnormally translucent irides were not a constant finding in all carriers, they were significantly more frequent in them than in the controls. Witkop et al (1973), however, believe that iris translucency is not a reliable indicator of heterozygosity for ty-pos and ym albinism. In a small series of Negro ty-neg obligatory heterozygotes, they found only about half their subjects had translucent irides, so that it is not even a consistent finding in this group. Froggatt (1960) noted the eye and hair colour of the parents and children of albinos during his study, and preliminary analysis suggested that blue eyes and fair hair were more common in this group than in the general population of Northern Ireland.

Other pigmentary anomalies have also been reported in heterozygotes and relatives of albinos. These include white hair locks (Sanders, 1938), white eyebrows and eyelashes (Usher, 1906) and one or more depigmented patches (Stannus, 1913). Froggatt (1960) reported three presumed carriers who had fair tufts of hair in the occipital region and one who had white eyebrows and eyelashes.

From these studies it seems that, although research workers such as Harris (1926) and Keeler (1953) commented upon the light skin colour of the parents of albinos in the dark skinned races studied by them, and although pigmentary changes of the eye and hair have been investigated, skin colour does not seem to have been objectively measured in either heterozygotes or homozygotes.

2.1.8 Fitness and fertility in homozygotes and heterozygotes

The biological fitness of individuals is generally assessed by the number of their offspring who reach reproductive age (infant deaths and still-births are therefore excluded). Fitness is unity (or 100 per cent) if an individual and spouse have two such offspring (Emery, 1979, p225). Fertility on the

other hand, is indicated by the total number of offspring produced, whether or not they survive.

Neither the fitness nor the fertility of albinos has been thoroughly assessed. Their fertility is affected by the fact that many do not marry, an observation made in both Caucasoid (Froggatt, 1960) and Negroid (Vallois, 1950, Barnicot, 1952, Okoro, 1975, Kromberg and Jenkins, 1984) societies. Among African societies marriage is virtually universal and almost every woman marries at least once in her life-time (Rinehart, 1979), so when albinos do not marry the fact is very striking. Froggatt estimated that the chance of an albino marrying was 70 per cent in comparison with that of non-albinos in Northern Ireland. He found that in his sample only twelve of thirty affected females and twenty of forty-one affected males, in the over twenty year old group, were married. Okoro (1975) stated that in Nigeria it is very difficult for albinos to marry, and he found that a very small proportion of affected women in his sample were married. Similarly, in Barnicot's (1952) Nigerian study only five of eight females and eight of thirteen males aged twenty years old or older were married. Barnicot added that he could not consider the question of fertility because of the limited data and diverse ages of the subjects in his group.

Freire-Maia et al (1978), however, discussed fertility in a small group of albinos who came from an isolate on Lencois Island (a Northern Brazilian island). The ten albinos in the sample had on average 2.90 children per person, in comparison with thirty-seven of their normal relatives who had 2.46 children per person. There was therefore no reason to conclude, from their findings, that fertility was reduced in albinos. They did not, however, assess the fitness of their subjects in terms of survival of their children to reproductive age.*

Fitness and fertility have been investigated in at least two other genetic disorders. In a study on Huntington's chorea in South Wales, increased values of fertility and fitness were found in affected individuals in comparison with both relatives and the general population (Walker et al, 1983). In another study on recessively inherited congenital glaucoma in a Slovakia Gypsy population, decreased fertility (36 per cent that of healthy siblings) was found in individuals with the condition (Ferak et al, 1982). These studies were carried out because the investigators were looking for reasons for the high prevalence of the two disorders in these specific population groups, and the possibility of selective advantage in affected individuals.

No reports on fertility and fitness of heterozygotes for the albinism genes could be found, although such studies have been carried out for other genetic conditions such as cystic fibrosis (Knudson et al, 1967) and Tay Sachs disease(Myrianthopoulos and Aronson, 1966). In both these latter studies the grandparents of affected children were found to have a reproductive advantage over the controls, and in the case of cystic fibrosis there was a statistically significant difference in favour of the heterozygotes. In the Tay Sachs disease study, survival to the age of twenty-one years was significantly higher in affected sibships.

Since there is such a paucity of data on fitness and fertility in both homozygotes and heterozygotes for the albinism genes, further studies are required for the purpose of assessing possible selective advantage in this condition.

2.1.9 Conclusion

Selected studies on albinism, and occasionally on other genetic disorders (where information on albinism is lacking), which have relevance for the present study have been reviewed. Gaps in the published reports have been identified and it is evident that data are not available for South Africa on: the prevalence rates in the major black ethnic groups; the types of albinos found; the cancer risks associated with age; the life-expectancy in males and females; and the skin colour and fertility in homozygotes and heterozygotes. The present study attempts to provide data to fill some of these gaps and to compare the findings with those reported in the literature reviewed above.

2.2 Psychosocial studies

Although, some of the psychosocial aspects of being an albino have been mentioned in passing in several of the studies reviewed above, none of the authors appear to have touched directly upon the effect on a black family of the birth of an albino infant. There are a few studies, however, which have investigated the psychology of the albino in the Negro and other dark-skinned populations and others have mentioned maternal and societal attitudes. This section of the available literature on albinism will be briefly reviewed in chronological order here.

Because there appear to be no available reports on albino infants and their families, selected reports on the impact of the birth of babies with other genetic defects will be included, and available data on parents' reac-

tions will be discussed. The investigation of the maternal- and paternal-infant relationship is an integral part of the present study and selected reports on this subject where the infants are normal and where they are defective will also be critically presented, so that a general perspective might be obtained.

2.2.1 Psychosocial aspects of albinism

Although Pearson et al, (1911), and those who sent reports to them, did not specifically examine psychosocial aspects of albinism, community attitudes towards albinism throughout Africa were mentioned in passing. In some parts of Africa the cause of albinism was believed to be sexual intercourse between a negress and a gorilla or a water spirit. In Cameroon people believed that albinos were born as punishment for the sins of the parents or for adultery, or because of maternal impression or heredity. Albinos were treated with often extreme reactions from being considered with respect and fear as sacred and inviolate in some parts of Africa, to being thought unlucky and/or killed in Gabon, Malawi, Cameroons, and South Africa. Livingstone (1857) presented one of the few detailed reports of a black mother's dilemma in Botswana. He described how the woman cared for her albino child for many years and was rejected by the father while doing so, she eventually tired of living apart from him, killed the child and returned to the father.

Probably the first psychological study on albinos was carried out by Beckham (1946) in Chicago. He was interested in the question `Does the albino present a psychology peculiar to himself?' He interviewed and conducted psychological examinations on forty-two Negro albinos. Ten years later he carried out a follow-up study and reinvestigated his subjects. Social histories were taken from all the subjects by a social worker and, from these histories, Beckham stated that albinos have some specific problems, particularly related to family and personal adjustment. Based upon his observations he stated that: many Negro albinos, even at an early age, develop a feeling of insecurity in their homes and society generally because of discrimination against them and the feeling of `being different'; psychological and psychiatric treatment are often necessary to help them face their problems; albinos have acute visual problems, it is the duty of parents, teachers and other persons concerned to see that they obtain the best visual care possible.

Beckham also measured the IQs of his subjects with the Stanford Binet scale and found that their intelligence was within the normal range. His re-

sults indicated the need for a controlled study so that the psychosocial problems of the albino could be understood and differentiated from those of the normally pigmented individual.

Barnicot (1952) studied albinos in Nigeria, but admitted that his sociological information was superficial and incomplete. His impressions were that albinos were not markedly handicapped with regard to occupation, they were not defective in intelligence, and they did not seem to experience much discrimination. In his small sample of twenty-one albinos, however, only thirteen were married. Barnicot observed that the mothers of albino children seemed to regard the condition as a misfortune, which they attributed to a variety of causes, such as `maternal impression or incorrect worship of their gods´; nevertheless they did not appear to discriminate against their affected children. Barnicot added that attitudes towards albinos seemed to have varied considerably in different parts of West Africa, and that it was impossible to tell for how long a particular local attitude had prevailed.

In the early 1960s Woolf initiated a series of studies on albinism in the Hopi Indians of Arizona. Reasons for the high prevalence of albinism (about 1 in 227 (Woolf and Grant, 1962)) were sought. Woolf and his coworkers were informed that albinos were `good luck charms´ and had some religious significance in Hopi Indian culture (Woolf and Dukepoo, 1969). After investigating the local culture and attitudes they found that albinos had no supernatural influence and were not considered to bring good luck. Albinos were viewed as any other individual with a congenital defect and were well integrated into Hopi Society (Woolf and Dukepoo, 1962).

Woolf and Dukepoo (1969) also noted the changing attitudes towards the albino. The younger Hopi Indians who had learned that the condition was a genetic defect might show some discrimination against albinos, whereas older traditional Hopis made comments which reflected only positive attitudes towards them. Even in this generally accepting society, however, many albinos remained unmarried. Woolf and Dukepoo (1969) commented that in a dark-skinned population albinism seemed to have such a repressive effect on the affected person that suitors were rejected,and they felt that this point required further investigation. They found, however, that the real or subconscious bias towards marrying an albino did not manifest itself in the selection of a sex partner. There was a local legend that one old albino, who never married, had as many as fifteen children.

Woolf and Dukepoo (1969) also discussed the intriguing finding that albinos were regarded affectionately in Hopi villages. They commented that the admiration for their whiteness was not an identification with white Americans, rather they felt it represented `an association of whiteness with cleanliness, goodness and purity; attributes honoured by traditional Hopis´.

Stewart and Keeler (1965) investigated a group of six albinos and six normally pigmented control Cuna Indians from San Blas. Their subjects were flown from Panama to Georgia, USA, and submitted to extensive medical and psychological studies and a battery of intelligence tests. The conclusions were that the albinos revealed a passive, dependent, feminine orientation with little interest in the opposite sex. They were anxious, insecure and relied on defence mechanisms, such as intellectualization, religious preoccupation and denial, to allay anxiety. The albinos tended to deny the existence of any difference between themselves and the controls, but this denial broke down under moderate stress, and hostile, resentful and angry responses, and even withdrawal, often resulted. The subjects also appeared to show a markedly less accurate perception of reality. Their emotional immaturity was attributed to either early parental overprotection or the inheritance of a poorer basic personality, or to an interaction of both factors. The albinos and controls were, however, examined in a very stressful situation far from their native land, and further confirmation of the findings with a larger sample in a less stressful environment was recommended by the authors.

In an effort to assess the intellectual maturity and body image boundary characteristics of albinos in South Africa, Manganyi et al (1974) carried out a study on affected subjects in the local black population. The results indicated that the twenty-eight albinos tested had an intellectual status well within the normal range and a less diffuse body boundary differentiation than the controls.

This study was followed by a second study on mothers of albinos in a black South African population (Kromberg and Jenkins, in preparation). Interviews were conducted with fifty-six mothers and information was collected on their attitudes to having albino children. The preliminary findings from this study support the suggestion made by Stewart and Keeler (1965) that mothers tend to overprotect their albino offspring. The mothers also differentiated between their albino and their normal offspring, expecting more from the albinos and giving them better opportunities for education and advancement. Stewart and Keeler´s (1965) comments on disinterest in the oppo-

site sex, and Vallois' (1950) and Barnicot's (1952) observations on the difficulty of marrying were partly substantiated, and the fifty-six mothers in this study showed some reluctance to permit their albino children, particularly the females, to marry. This finding, however, requires confirmation using a matched control group.

A third study in this series on Negro albinos in South Africa examined the adjustment of thirty-five young albinos and the attitudes of young normally pigmented black subjects towards them (Kromberg, 1977, Kromberg and Jenkins, 1984). The results indicated that the albinos showed a similar level of adjustment to that of the matched control group, and that the albinos were quite well accepted in the local community, where the prevalence is relatively high (1 in 4 000, Kromberg and Jenkins, 1982) and everyone is familiar with the condition. The main problems of the albinos were those associated with psychosomatic symptoms, with the physical symptoms attributed to the condition, with employment (since they believed they would have difficulties in obtaining work), with the widely held belief that albinos do not die natural deaths (which leads to some anxiety) and with the partial acceptance in the community (which resulted in many of them being rejected as marriage partners, and the term `monkey' being applied to them in many community subgroups). Much ignorance surrounded the condition and it was attributed to maternal impression, or maternal behaviour during the pregnancy, or to divine intervention (seen either as punishment or as a gift). Although the subjects in the normal control group had a generally positive attitude towards albinos, some considered that albinos had psychosocial problems. Examples of their comments in this connection are worth reporting here: `being black by birth but not by colour may be one of their major problems, this deprives them of the companionship of other black children'; `they cannot easily make friends because of skin and eye problems'; `they keep to themselves'; and `they are never relaxed in company' (Kromberg and Jenkins, 1984).

In summary then, the psychological and social studies of albinos, although not always methodologically sound, and sometimes based only on observation, have nevertheless provided some material which is of relevance as a background for the present study. The intelligence of the albino has been found to be within the normal range. Personality problems, however, have been described, particularly those relating to feelings of insecurity, to society's limited acceptance, to extreme parental reaction, and to emotional

immaturity, anxiety and denial. In South Africa, studies indicate that young albinos are intellectually average, adjusted to the same extent as other young people are, generally accepted (although this acceptance stops short of marriage in many cases), probably overprotected by their mothers, and disturbed by the widely held belief that they do not die naturally. Attitudes towards albinos in Africa appear to be changing and infanticide has generally been replaced by a measure of acceptance and sometimes overprotection.

2.2.2 Studies on the impact on the family of the birth of a defective child

Apart from the odd description of individual cases (such as in Livingstone, 1857) there appear to be no reports on parent-infant relationships where the infant is an albino, or on the impact of the birth of an albino on a black family. Neither do there appear to be reports on the impact of other genetic defects, which are obvious at birth, on black families. Reports concerning a variety of defects, generally on white families, have therefore been reviewed as a background for this part of the present study.

The birth of a baby is a dramatic event in the life of any family. Planning is usually involved, and the birth is the culmination of months of preparation and represents the future hopes of the parents. The birth of a defective baby therefore presents an unexpected crisis which engulfs not only the parents, but all those significant others who have taken part in the event.

In every 100 births world-wide, there are at least two babies born with major or minor malformations which will handicap the baby to a greater or lesser degree (Lancet, Special Article, 1971). These handicapped children and their parents' reaction at the birth have been investigated in several studies. Some workers have grouped the different malformations together and examined parental responses without reference to the specific disorder (e.g. Johns, 1971; Kennedy, 1970). Other workers have taken common disorders such as Down Syndrome and examined the family's adaptation to the birth of an affected child (Emde and Brown, 1978, Gath, 1978, and Cunningham, 1982). It appears, however, that no reported research on the effect of the birth of an albino child on the family has been included in a general group.

There are several obvious reasons for the neglect of this particular defect: albinos are very rare in the Caucasoid community, among which most of the specialised research projects have been conducted; albino births in a white community are not so startling, since the skin colour is at least simi-

lar to that of the parents (even though it might appear strange and slightly different, this difference is insignificant at first glance); since the albino baby is almost always perfectly formed and is not likely to require any traumatic medical treatment, the nature and severity of the defect and its possible future psychological sequelae tend to be ignored initially. Albinism occurring in the black community, however, cannot be overlooked and none of the above reasons for neglect, except perhaps the last (albino babies are sometimes not reported in local black hospitals since they are healthy, Kromberg and Jenkins, 1982b) apply. Albinism is common, occurring at least at the rate of 1 in 4 000 births (Kromberg and Jenkins, 1982a); the birth is extremely startling, and the skin, hair and eye colour are dramatically different from that of the parents; although the baby is perfectly formed no-one can deny that the black mother, as well as the medical staff, suffer a shock at the sight of a white baby at the time of delivery, and the psychological effect on the family is traumatic (according to the unpublished anecdotal evidence of medical staff). Nevertheless, some might still debate whether or not albinism could be termed a `defect´ or a `handicap´, since physically, problems are minimal and can relatively easily be treated. This point required clarification before the literature on the impact of defective children on the family could be considered relevant and reviewed.

The Younghusband report (1970) on children with special needs defines handicap as `a disability which for a substantial period or permanently, retards, distorts or otherwise adversely affects normal growth, development or adjustment to life´. In the light of this definition it appears that the albino at least in a black community, has a disability, that of being different from his fellow-men, which for the whole of his life probably distorts and affects his adjustment to life. The difference is apparent at first glance (particularly when the other members of the population are darkly pigmented), and as such influences the attitudes of others and disturbs the initial formation of normal relationships and the natural identification of one person with another. Albinism therefore may, in the writer´s opinion, be classified among the defects, and a baby born with albinism may be termed a defective baby. The findings of the research work on reactions of parents to the birth of a defective baby may therefore be applied to the sitution in which an albino baby is born to a black mother. Medical personnel working with that mother can probably expect her to experience the same crisis response, behavioural changes and psychological problems, as those of other

mothers with defective babies.

The reaction of mothers with defective babies has been examined much more thoroughly and at much more depth than that of fathers, probably partly because of the accessibility of mothers and their initial ready availability in hospital in the perinatal period. Psychological processes in mothers during pregnancy have also been studied, and Bibring (1959) considers that the entire period of gestation is one of strain and enhanced narcissism for the mother. Significant changes both somatic and psychological, occur, and fears, fantasies and ambivalent belief in old wives' tales abound during this time. In a study carried out by the Royal College of Midwives (Preparation for Parenthood, 1966), in London, 85 per cent of mothers admitted to fears that their baby would have a defect and a similar study in Israel (Heymans and Winter, 1975) found that 81 per cent of the mothers who were interviewed had expressed fears to their husbands that the child would either be malformed or mentally defective, or would die before the delivery. Few of these fears had been discussed with the doctor, and more fears had been expressed by primiparous than by multiparous women. Brazelton (1973) has stated that such fears are necessary to shake up the mother and to make her reorganise herself, so that she can create her new role and relate to the child. Although the mother experiences fears for her baby she nevertheless forms an image of the baby as normal, healthy, complete and attractive. The wish for a perfect child is universal and, as Solnit and Stark (1961) state, `it is likely that there is always some discrepancy between the mother's wishes and the actual child'. When the discrepancy is too great, however, as in the birth of a defective child, the situation becomes traumatic, the parents have feelings of tremendous disappointment, helplessness and failure. Suddenly the baby which they were expecting is lost, their worst fears are realised, their fantasied normal baby is replaced by one with a defect, which they find threatening, alarming, emotionally confusing and a heavy blow to their self-esteem (Solnit and Stark, 1961).

The parents' reactions seem to be tempered by their cultural background, experiences and personalities, as well as by the type and degree of defect in the baby. Their initial response also appears to be affected by their knowledge of and belief in community myths and old wives' tales regarding the particular defect.

Throughout recorded history references have been made to community reactions (Klaus and Kennell, 1976, p168) and deformed infants have evoked a

wide range of emotional responses from awe and admiration to terror. The concomitant behaviour of the community has ranged from deifying the affected individuals to murdering them. Warkany (1971) notes that extreme measures were sometimes taken in ancient communities and man either `exterminated or adored the deformed of the species - and sometimes he did both'. After killing an infant with a disfiguring abnormality he `often made an image in its likeness and set it up as an idol-god or demigod'. Sculptures, carvings and drawings of abnormal births by ancient peoples antedate the arts of reading and writing. As Klaus and Kennell (1976, p168) comment `despite new scientific knowledge about the origins of congenital defects, ancient superstitions still plague modern parents. Because of the strong emotional reactions accompanying the birth, it is not surprising that the lore of the past surfaces to haunt parents of children with congenital malformations.'

In the case of the black mother, in South Africa, widely believed myths probably affect her perception of the event of the birth of an albino baby. She is influenced by three common beliefs: that the albino is different, special and never dies, but disappears when the time comes; that albinism is caused by God's intervention either to punish the mother for her bad deeds such as, for example, laughing at albinos during pregancy, or to bestow on her a gift or a lucky symbol; and that albinism is infectious and can be `caught' from others (Kromberg, 1977). The way in which she reacts will depend on how she views the cause of the albinism and how she sees her own contribution to the causation. In some cases albinos are also believed to be representatives of the spirit of the Gods and to have special spirit-powers (Kromberg and Jenkins, 1984). The colour white has a special significance in many of the black population groups (Hoernle, 1937), such groups will only sacrifice white goats in religious ceremonies and will give special care to white cows. It is possible that these practices also have bearing, even if subconsciously, on the mother's reaction to producing a white child.

Society is still caught between the desire to do away with defective babies and the need to treat the affected baby as actively as possible. In underdeveloped communities such as the Kalahari bushmen, malformed infants may still be buried seconds after birth before they have even breathed (Konner, 1972). In developed communities such as Germany, under the third Reich, extermination of defective people was practised. In South Africa today, black pregnant women are expected to observe certain taboos. Pedi women, for example, should not have contact with other pregnant or ritually impure wo-

men, or look at people with physical disabilities (Hammond-Tooke, 1974). In Britain, severely affected babies with spina bifida, if they meet certain criteria, are `selected out´ of the treatment programme, because of the poor quality of life they may be expected to live and the burden they will put upon their families and society (Lorber, 1971), while in the USA such babies are often treated actively regardless of the consequences.

Despite the many influences on the mother, the birth of an affected baby appears to evoke responses which are experienced in one form or another by all mothers who are faced with the situation. Freud (1924, p153) describes mourning as the response to `the loss of a loved person, or to the loss of some abstraction which has taken the place of one, such as fatherland, liberty, an ideal and so on´. Such mourning occurs when an expected and wished-for normal baby does not materialise at delivery. Beddie and Osmond (1955) have emphasised that there is a need for parents to mourn and if the mourning process is not carried through, an unaccountable depression can occur later. The mother of the defective baby is put in an often overwhelming situation, since she has not only to mourn her idealised baby and achieve decathexis of the lost child, but she has to respond to the infant that is present and demands her attention. Chronic sorrow as described by Olshansky (1962) often results.

Bowlby (1960) was one of the first writers to discuss the grief process which is associated with mourning and to differentiate the phases of the process. He noted three phases: protest, despair and detachment. Grieving is, fortunately, time-limited, and Kennedy (1970) has defined and grouped the behaviours and affect found in each of the three grief phases of a mother with a defective child: 1). Protest: includes shock, numbness, disbelief, `not hearing´, evasiveness, `shopping´, searching for magic cures and anger; 2). Despair: includes disappointment, loss, hopelessness, helplessness, futility, sorrow, guilt, loss of warmth in relationships, and physical symptoms such as insomnia and loss of appetite; 3). Detachment or withdrawal: includes recall of the prebirth longings for the idealised infant and evidence of cathexis of the live infant.

During the last phase, withdrawal occurs from the loved fantasied infant, and this is followed by a gradual recovery, acceptance and learning to cope with the demands of the live baby. This phase is often characterised by the mother referring to her baby by its name. The time sequence, time limits and specific grief phases were not so distinct, according to Kennedy in his

study, as Bowlby (1960)claimed from his findings on the behaviour of children separated from their parents.

Gath (1972 and 1978) studied the effects of having children with cleft lip and palate, or with Down Syndrome, on the family, particularly on the siblings of the affected child. Her study on Down Syndrome was a prospective one in which she followed thirty families with an affected baby, starting as soon as possible after the birth, for two years. The data were collected through six structured interviews in the home. Various tests were used at different stages, to measure parental expectations and interaction, as well as the mental and physical health and personality of the parents. Both parents and teachers rated sibling behaviour and health. Mothers were seen at every interview and fathers were present at some interviews only. All fathers were seen at least once. The main aim of the study was to `see if the birth of a mongol baby produces changes that are different in kind or degree from those changes encountered in families with normal babies'. A matched control group was therefore selected. Matching by date by birth, sex of baby and ordinal position in the family was obtained. Also it was possible to match father's occupation as being in the same socio-economic group as those of the experimental subjects, and the mother's age in most cases was similar. The home neighbourhood was comparable in the two groups.

In her conclusions Gath stated (1978, p114) that all the parents in her study expressed feelings of grief and shock when they first realised that their baby had a serious abnormality, and the time taken to reach a positive acceptance varied. One of the most striking findings of her study was that the marriages of couples who had an affected baby were significantly more likely than those with a normal baby to be unhappy one year after the birth. The birth of an abnormal baby tended to magnify pre-existing weakness in the marriages. Morbidity in these affected parents, however, was not found to be significantly higher than in parents of normal babies, and the former did not show an excess of psychiatric or physical ill-health. The siblings who were studied did not have significant health problems in comparison with controls either, but siblings' well-being was influenced by the parents' marital relationship amongst other factors. The study was confined to the first two years of life of the affected child, so the long term effects on the siblings and family were not investigated. Nevertheless, Gath emphasised that although there was the initial emotional reaction to the fact of the baby's abnormality, most families adjusted well and two years after the birth were

providing a stable and enriching environment for both their normal and handicapped children.

Another relevant study is that of D'Arcy (1968) who interviewed mothers of babies with anencephaly, spina bifida, Down Syndrome, cleft lip and palate, and congenital heart disease, amongst other rarer conditions. She was interested in how, when and from whom the mothers heard about the defect in their child. She used semi-structured interviews and encouraged mothers to voice their opinions, but she did not specifically include the mother's reactions to the baby with the defect. She found from her observations that mothers whose anencephalic babies had died perinatally seemed to suffer more acutely than parents whose malformed infant survived, but the former group also recovered from the tragedy sooner than the mothers of surviving babies. In general, the mothers wished for a sympathetic approach from the staff, explanations using simple language, a willingness to answer questions, and truthful answers avoiding 'unjustifiable pessimism and unrealistic optimism'. It is probable that the birth of an albino baby should also be handled bearing these points in mind.

A small study of twelve families with babies with a variety of defects was carried out by Johns (1971). She attempted to observe directly the earliest reactions of these families to the birth of a baby with a congenital abnormality. She interviewed the mothers with a check-list of subjects used in a flexible manner, within the first day after delivery, and again three months later at the hospital. A third interview was conducted at home when the child was six months. She found that the height of the crisis occurred in the hours after the parents had been told of the child's abnormality. The parents had feelings of shock, dismay and failure, instead of happiness and a sense of achievement usually observed after a normal delivery. They needed time to absorb the unexpected information,and an opportunity for discussion and questions. The study seemed to show that the parents of the children with the most visible or more severe defects were most anxious, and facial disfigurements were especially embarrassing. Since albinism is also a visible defect this study, although of limited value because of the small sample and the variety of defects included, is of relevance.

Some of the studies on the effect of the birth of a defective baby on the family have been reviewed above and the findings summarised. Albinism in a darkly pigmented community, should in the writer's opinion, be considered a defect. The prenatal feelings and fears of having a defective child have

been mentioned and the reactions of parents discussed. The response of the community and the cultural beliefs which influence the parents' responses have also been reported briefly. All mothers appeared to experience grief and mourning as if they had lost the baby they longed for, this stage is accompanied by shock and denial, followed by despair, depression and guilt, and then a gradual recovery and acceptance. There is some oscillation between these stages and the time taken to accept the child varies. Fathers have not been investigated so thoroughly but seem to pass through a similar crisis to that of the mother. Since no examples of families with albino children appear to have been recorded, material where the babies had Down Syndrome or other congenital abnormalities has been presented. These studies were generally exploratory in nature, but their findings on the impact of the birth of a defective baby on the family are useful as comparative material for the present study.

2.2.3 Studies on the maternal-infant relationship

To assess the impact of the birth of an albino baby on the family one needs not only to know what understandings psychosocial studies on albinism, as well as those on families with defective infants, so far contribute, but also how mothers relate to and interact with their normal babies.

Various workers have studied the maternal-infant relationship which is sometimes called the 'first relationship', since it is the first to be experienced by the infant. One of these studies, which was particularly influential on the present one, because of the methodology, was that of Klaus and Kennell (1976). They studied maternal-infant bonding with reference to the impact of early separation or loss on family development, and on factors that may enhance or inhibit the formation of the earliest parent-child relationship. They were interested in the short and long-term effects on this relationship of the modern practice of removing premature or ill babies completely from their mothers, and placing them out of reach in incubators or in isolation wards. The authors pioneered the reopening of nurseries for the sick infant to parents and their families. Their studies lead them to believe in 'the existence of a sensitive period in the first minutes and hours of life during which it is necessary for optimal later development that the mother and father have close contact with their neonate' (Klaus and Kennell, 1976, px). They also suggested that the infant must signal back and cause an initial interaction which may develop into a profound and long-lasting attachment.

Klaus and Kennell (1976) did not study mothers and defective babies specifically (although their book has an excellent chapter on caring for parents of an infant with a congental malformation). They did, however, interview mothers with new-born, normal babies and observe the dyads in interaction. From their findings they formed a practical theoretical framework for the study of the first relationship. They also described in detail the behaviour of the mother that stimulates interaction, namely: touch, eye-to-eye contact and the en face position, the use of a high-pitched voice, and entrainment (including the sequence of synchronous movements accompanying communication between mother and child). Maternal attachment appeared to develop rapidly during the early postpartum period, because of these behaviours, together with the several biological contributions to the situation by both mother and baby.

Another relevant study was conducted by De Chateau and Wiberg (1977) who observed mothers and infants in the Swedish population. They studied the effect of early skin-to-skin contact in the first thirty minutes of life on infant and maternal behaviour three months later. They found that mothers in the early contact group spent significantly more time in the en face position and kissing their infants, whereas control mothers more often cleaned their infants. They observed the dyads at home during a free play period and noted such maternal behaviour as: looks en face, smiles, laughs, kisses, cleans, gives toy and rocks infant.

The maternal-infant relationship has been studied in African families by Ainsworth (1971, 1972) in Uganda. Ainsworth emphasised the need to observe the dyads rather than to use sophisticated measuring instruments that might not be valid in different and unstudied cultures (Ainsworth 1979, personal communication). She and her co-workers studied interaction between mothers and their normal babies and did not comment on mothers and defective babies. Their project was focused on the development of maternal-infant attachment and they were able to identify a number of attachment behaviours. The study on Ganda mothers and their infants was a longitudinal one, with interviews and observations of the dyads being conducted at regular two weekly intervals in the homes of the subjects. The project methods and data analysis were necessarily cruder than those of similar projects conducted in the USA (according to Ainsworth), and simple behaviour counts were done on babies who did or did not display specified behaviours. Ainsworth was impressed with the wide range of individual differences in her sample, and with the

similarities between the Ganda and the USA Samples, even though the cultural differences were great (Ainsworth, 1977).

Bowlby (1969) and Ainsworth (1972) have defined an attachment behaviour as `one that promotes proximity or contact'. It is through such behaviour, as for example, rooting, sucking, reaching, grasping, that babies become attached. To assess this attachment, Ainsworth found that observations produced more reliable information than interviews. Ainsworth visited her subjects at their homes and observed the dyad's interaction as well as the baby's behaviour when the mother left the room. The disappearance of the mother was thought to produce an alarming situation and alarm is known to activate both attachment and fear behaviour. The interpretation of the results, however, was confused, since the separation protest appeared to be influenced by the infants' confidence in their mothers' accessibility. Also the presence of a stranger, the observer, probably compounded the fear already activated by the absence of the mother and the separation protest may have been intensified as a result (Bowlby, 1973).

Because there had been so little pertinent research prior to her Ganda study, Ainsworth (1972) did not wish to limit her observations by setting up a check-list of behaviours and observing them on a time-sampling basis. She therefore used a `naturalistic' approach with narrative accounts which were extremely cumbersome and time consuming to analyse. She maintained, however, that the check-list method confines one to preconceived variables and severely reduces the possibility of discovering new and important behavioural variables. After collecting and analysing her data Ainsworth concluded that: feeding practices play an important part in the development of the formation of infant-mother attachment; the security-insecurity dimension of the attachment is linked to maternal availability and sensitivity; physical contact of of good quality in early infancy facilitates rather than hinders later development; separation protest and intensity of fear response to strangers appears to be linked to the degree of anxiety in the attachment relationship; the contingent response by the mother to the signals of the infant, his freedom to explore, and frequent opportunities for interpersonal interaction may facilitate cognitive development; locomotor development may be more advanced in Ganda babies because of their frequent upright holding position.

From this brief review of the relevant literature on the maternal-infant relationship it appears that some work has been carried out on African dyads, but probably none at all on the black mother with a defective infant in

Africa. The above studies were selected for inclusion because the information on the methods used in observation, and on the behaviours which were observed, was useful in determining methods for the present study. In some cases the results from the reported studies will also be useful for comparative purposes when the findings from the present study are discussed.

2.2.4 Studies on the Paternal-infant relationship

Fathers of albinos and their attitudes and relationships were briefly mentioned only a few times in passing in the literature on albinism. The reaction of the father of Noah, who was said to have been an albino (Sorsby, 1958), was described and he questioned his wife's faithfulness. Livingstone (1857) mentioned a father in Botswana who ordered his wife to kill their albino child and refused to live with her while she kept the child. McCrackin (1937) reported finding a father in the French Cameroon who, in 1924, threatened to kill his children if they were albinos, but could not bring himself to do so when his albino child was born. Maternal impression was considered to be the cause of albinism in many African communities (Barnicot 1950, McCrackin 1937, Kromberg 1977) and women were accused of adultery and sinning, or consorting with water spirits (McCrackin, 1937) or gorillas (Pearson et al, 1911). Albinism was also occasionally attributed to the sins of `the parents' or to heredity (McCrackin, 1937). Communities therefore generally appeared to accuse the women of causing the child's disorder and only rarely was the father implicated.

The Dutch scientist Regnier de Graaf has been given the credit for first recognizing in the second half of the seventeenth century that the union of egg and sperm was required for conception (Emery, 1979) and he suggested that both female and male parents transmitted characteristics to their offspring. This concept was not generally accepted for many years. de Maupertius however also concluded that both parents contributed equally to the make-up of the offspring (Emery 1979), from his work on animal breeding in the seventeenth century. The concept of genes is generally unfamiliar to the local black population and in a recent study only 4 per cent of black teenagers stated that albinism could be caused by hereditary factors (Kromberg, 1977). Blame for a defect in a baby is still generally allocated to the women and fathers seldom take responsibility, unless there is a family history on their side.

The attitudes of black fathers appeared to have changed over the last century and with the influx of missionaries, the increase in education and

the acceptance, even if only partially, of Christian morality, the killing of albinos appears to have decreased and such reports do not seem to occur in modern anthropological literature. Fathers therefore at least no longer appear to insist on infanticide but whether or not they accept and care for defective children does not seem to have been investigated. Research on other groups of parents who produced defective children indicates that both parents probably feel shock, denial, sadness and anger, followed by a restoration of equilibrium or adaptation and reorganization (Drotar et al, 1975) after the birth of their affected baby. The fathers' reaction is not differentiated from that of the mother in many reports and the parents have frequently been interviewed and grouped together (Eg Gath, 1978), or mothers have been interviewed alone. Cunningham (1982, p21) however states that the father's feelings may not synchronize with the mother's and their reactions may differ, producing conflict and stress. Fathers may not feel like discussing their feelings about a defective infant and Cunningham (1982, p55) observed that men were more likely than women to ask questions, where the child had Down Syndrome, about the effect of the child on the family and on the mother, in an attempt to protect the family.

Klaus and Kennell (1982, p255) state that parents who ask many questions generally appear to be making a reasonably good adaptation to their situation. These authors also suggest that fathers should be present when the wife is informed of the diagnosis, included in all discussions, and allowed to spend prolonged periods with their wives in the maternity hospital, so that they can share their feelings about the defective infant and work through their reactions in sequence as synchronously as possible. The hospital needs to be concerned with both parents. Klaus and Kennell (1982, p256) also suggest that if the parents are unable to discuss their reactions and feelings about the baby with each other, a marital rift might develop. Therefore several meetings with both parents together are advocated, at which the mother describes her feelings and how she sees the father's feelings and he does the same. They then start to think about their own and each other's adaptation, and communication between them often improves. Gath(1977) also believed that marriages were at risk and that management of the abnormal infant must include an assessment of the parents' relationship, so that intensive support can be given during the initial crisis to less stable couples.

Both Cunningham (1982, p57) and Klaus and Kennell (1982, p280) have observed that many fathers of defective infants tend to direct their energies

into their work, taking on extra employment and responsibilities in the community, so that they are constantly occupied. This behaviour helps them to avoid facing their feelings and often interferes with their communication with their wives. Mandell et al (1980) observed that fathers who had lost a child with SIDS (sudden infant death syndrome) had only a limited ability to ask for help and that paternal behaviours, such as keeping busy with increased work, obstructed the full expression of their grief. Sometimes the father also tries to keep the mother busy and this practice is anticipated by Klaus and Kennell (1982, p281) who suggest to parents that they should reduce commitments and responsibilities at this time.

Since there appears to be so little available concerning fathers and their defective infants in the literature, some of the reports on fathers and their normal infants were selected for comment. Yogman (1982a) suggests that fathers require psychological readjustment prior to the birth of a baby and they have to integrate their role of child and spouse with that of expectant parent. The new-born baby, however, has a powerful impact on the father and the father's intense engrossment has been described by Greenberg and Morris (1977). They found that the fathers experienced visual and tactile awareness of the infant, as well as noticing the distinct characteristics of the infant, perceiving it as perfect, and feeling extreme elation (the new-born's face was a particularly powerful stimulus), and an increased sense of self-esteem. The normal reflex actions and behaviour of the newborn enhanced the fathers' engrossment (Greenberg and Morris, 1974). After delivery father-infant interaction followed an orderly progression of behaviour, with touching of the infant's extremities, then touching with fingertips and palms, and increasing eye-to-eye contact (Rödholm and Larsson, 1979). Fathers appear to be just as responsive to infant vocalizations as mothers, both increase their rate of vocalizations in response to the infant, although fathers are more likely to talk rapidly than mothers (Parke, 1979). Parke (1979) noted that the father tended to hold the infant more, vocalize and touch the infant more, but smile less at the infant than the mother, in her presence. Generally however fathers gave less time in caring for their infants but spent more of their time in play with them, and this play was more physical, arousing and unpredictable than that of the mothers who made use of speech and toys during play (Klaus and Kennell, 1982, p62). The infant's appearance and abilities evoked responses from both mothers and fathers and stimulated communication, attachment and reciprocal interactions

(Klaus and Kennell, 1982, p63). The normal infant also appears capable of eliciting and engaging in differing patterns of interaction with father and mother and is not as passive as previously thought (Yogman, 1982b).

Fathers however show great variability in the involvement with their normal infants. Such variability appears to be caused by: the lack of a paternal archetype; the absence of social preparation; the paucity of institutional supports for the paternal role; the absence of predictable biological interaction patterns for fathers; the paucity of father-child interactions that are obligatory; and parents' widely differing ideas about the paternal role, child rearing, and sex roles (Palkovitz, 1984). Attitudes concerning fathering and traditional family roles are in a state of flux and a large proportion of families depend upon the income of both parents. The roles of each therefore have to be redefined. The father's role has been secondary and supportive, but men generally see their activities in the family as central and therefore some confusion and anxiety results when they first assume the paternal role (Palkovitz, 1984). Palkovitz (1984) found from his study and observation of forty father-infant dyads that significant relationships exist between parental attitudes and the fathers' involvement with their infants. The way in which the parents view the role, and the way in which the father sees his sex role, are related to levels of father-infant interaction in various social situations. The mother's concept of the father's role, in Palkovitz' study, emerged as the single best predictor of the father's involvement with the infant.

Lamb (1975) reviewed the role of fathers under the title `Fathers: forgotten contributors to child development'. He concluded that there was sufficient evidence to state that many infants interact extensively with their fathers and form attachments which differ from those with the mother. Fathers clearly do contribute to the psychological and social development of the infant. Clarke-Stewart (1982) however emphasized that concentration on the father must not be exclusive, but the family must be considered as a triadic system, with each member affecting the others.

This brief review of selected reports on fathers and albino infants, and fathers of defective and of normal infants, indicates that there is still a paucity of data especially on the first two of these topics. As Gunsberg (1982) states, there also appears to be no longitudinal study of fathers and infants from pregnancy through the first three years of life. However, what is apparent from these studies is that fathers are involved with their in-

fants from birth onwards (and probably even before birth), they can become engrossed in the baby and upset about a defect, they use different methods of coping with problems and in handling the infant, and there is much variability in their involvement with their infants. They should not, therefore, be forgotten and should be included in studies on mothers and infants, so that their roles and participation can be more fully understood, both individually and as part of the dyads and the triad which occur in families.

2.2.5 Summary

A review of the literature on the psychosocial studies of albinism indicates that although no studies of the impact of an albino baby have been reported, observations on this experience have been mentioned in passing by various writers. Attitudes of affected mothers in Africa have varied from one extreme to the other. In past centuries albinos were killed as were other congenitally defective infants, yet recent reports are available on mothers who overprotect their albino babies in Africa. Societal attitudes have also varied from ostracising and rejecting the affected individual (particularly as a marriage partner) to attributing special spiritual powers to him and believing that he is not entirely mortal. Albinos nevertheless generally appear to be reasonably well adjusted and accepted in South Africa, although feelings of insecurity, denial and anxiety have been found in San Blas Indian albinos.

Selected studies on the impact of defective infants on the family have been presented and all mothers appear to pass through stages of grief and protest, depression, and then acceptance. These reactions are affected by cultural beliefs about congenital defects and their causes and by ancient superstitions, as well as by society's attitude towards affected persons. In time the baby is generally accepted and families learn to cope. Those marriages which break up under the strain tend to be those which were stressed prior to the birth of the affected child. Parents appreciate sympathetic understanding and willingness to discuss and answer truthfully their questions and once they have accepted their situation, their normal children appear to do so as well.

Some reports on the maternal-infant relationship have been examined and the interactions between mother and baby as described in the literature have been presented. It appears that the mother's touch, eye-to-eye contact and *en face* position are important in establishing first contact with the infant and that the infant must signal back, thus causing the mutual interac-

tion. Methods of observation of mother-infant pairs have been suggested as preferable to interviews (or at least should be supplementary to them) as a means of collecting data in under-developed countries where the research work is limited and still exploratory. Nevertheless, research shows that there are many similarities between the findings on black subjects and those from in depth studies carried out in white communities.

Studies on the paternal-infant relationship suggest that fathers vary in their involvement with their infants, but they can become engrossed, and can be affected by a baby's defect in the same way as the mother.

2.3 Conclusion

A study of the literature reveals that there is some relevant material on various genetic and psychosocial aspects of albinism. The genetic section includes: a review of some of the first studies reported, with specific reference to those carried out in Africa; a definition and classification of the ten different types of albinism, and a description of the condition; the prevalence rates in various groups; a discussion of cancer risks and life expectancy of albinos; and, in both homozygotes and heterozygotes, a review of reports on skin colour, fitness and fertility. The selected reports reviewed indicate:

(i) that albinism has been observed in Africa for nearly 2 000 years, and that attitudes have occasionally been extremist;

(ii) that at least ten types with differing characteristics occur and some of these types have been described in South Africa, but have not been well characterised;

(iii) that prevalence rates vary from 1:200 (in the Hopi Indians) to 1 in 100 000 (in Russia), but such rates are not available for the major ethnic groups in the population of South Africa;

(iv) that cancer risks are high and life expectancy possibly reduced but detailed data are not available for South African albinos;

(v) that skin colour may be lighter than normal in black heterozygotes but it has not been scientifically measured;

(vi) and that fitness and fertility do not appear to have been thoroughly investigated, in heterozygotes or homozygotes in any black population.

The section on psychosocial studies incorporates a critical review of some reports on these aspects of albinism in darkly pigmented populations, on the impact of a defective baby on the family, and on the maternal-infant

and paternal-infant relationship. These studies indicate that:

(i) community attitudes to albinism have varied widely encompassing infanticide at one extreme and overprotection at the other, but attitudes at present appear to be positive, although they may stop short of marriage.

(ii) albinos do not differ in intelligence from the general population, but might have personality problems, including insecurity, anxiety, passivity, dependence, and little interest in the opposite sex.

(iii) parental attitudes to albinos have only been described briefly and superficially and no data are available on the maternal- or paternal-infant relationship where the baby is an albino.

(iv) parental reactions to the birth of babies with abnormalities other than albinism have been described and parents experience feelings of protest, despair and then acceptance.

(v) parent-infant relationships have been studied in families with normal babies, and interaction is promoted by the activities of the parents and baby and by the baby's appearance; these relationships can be investigated by observing the dyads in various situations as well as by discussion with the parents.

The present study is aimed at collecting data to provide information on some of the areas in which published reports appeared to be lacking.

CHAPTER 3

MATERIALS AND METHODS

Neither the genetic nor the psychosocial aspects of the study could be fully understood without an investigation of both the historico-theoretical background of the subject of albinism and the social structure of the environment in which the albinos were found. The psychosocial development and coping capacities of the albinos and of their parents depends to a certain extent on the attitudes of their community, as well as on such social factors as levels of education, employment opportunities, health services, overcrowding, family life, customs and habits in that community. Even the prevalence of the condition will be affected by such cultural factors as mating patterns and traditions of acceptance or rejection of affected persons and their families. Similarly, the occurrence of skin cancer and the life expectancy of albinos will be associated with life-style, and, for example, income levels and health facilities. For these reasons it was necessary to investigate and describe these social and cultural factors in Soweto, and, in a less detailed manner, in the four selected rural areas. The description of the methods used in the study is therefore preceded by a discussion of the social settings from which the samples were drawn.

3.1 Setting of the study

The focus of the study was the black population so the setting was Soweto (from $\pm$ 1979-1983), four rural areas, which were districts of Botswana, Kwazulu, Transkei and Swaziland, and the black urban areas of the greater Johannesburg metropolitan region. Soweto was chosen as the urban setting since it is the largest and closest black urban area to Johannesburg. The four rural areas were inhabited by two ethnic groups (the Tswana and Swazi) with high urban rates of albinism (based on the Soweto figures) and two with the low rates (the Zulu and Xhosa). The greater Johannesburg metropolitan region had to be included so that a sufficiently large source population would be available from which to draw the sample of new-born babies for the study. The characteristics of these settings are described below and their geographical situation may be seen in Fig 3.1.

3.1.1 Soweto

Soweto is essentially a dormitory town situated about fifteen kilometres

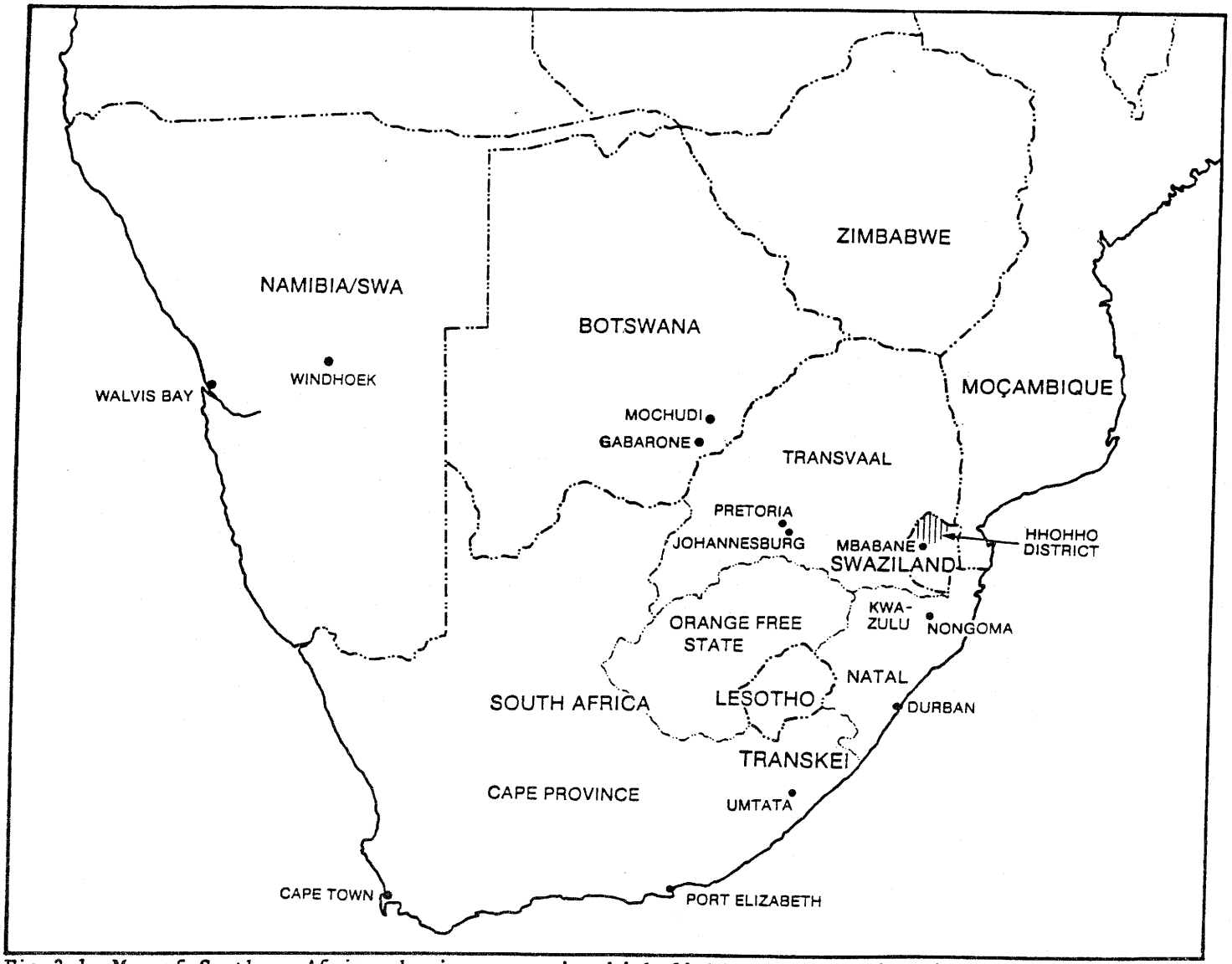

Fig 3.1 Map of Southern Africa showing areas in which field-work was undertaken

South-West of Johannesburg. It is inhabited exclusively by black people and is the largest and most advanced black urban community in South Africa. The name Soweto, introduced in 1963, was derived from the first two letters of each of the words `South Western Townships', and the area covers about ninety square kilometres (see Fig 3.2 and 3.3).

3.1.1.1 Historical background

Gold was discovered in Johannesburg in 1886 and following this discovery people streamed to the area. The first census in 1896 revealed that the population numbered 102 000, about half of whom were black (Morris, 1980), and most of these were contract workers on the mines. However, as the new town developed, other black workers moved to the area to work in homes and industry and multiracial areas developed in suburbs such as Vrededorp. In 1904 bubonic plague broke out in one of these haphazard settlements, and the slums had to be burnt to the ground. The inhabitants were moved to a municipal area fifteen kilometres to the south-west of Johannesburg and housed in temporary corrugated iron shelters and this was the beginning of the development of the area, which was initially called Klipspruit.

The black workers at this time were considered to be temporary residents in the white areas. The Transvaal local Government commission reported in 1922 that `the Native should be allowed to enter into the urban areas, which are essentially the white man's creation, when he is willing to enter and to administer to the needs of the white man and should depart therefrom when he ceases so to minister' (Morris, 1980). This principle of considering the black man as a temporary sojourner considerably hampered the development of adequate permanent urban housing and facilities for blacks for many years.

In 1927 the Johannesburg City Council set up a Native Affairs Department and in 1930 the Department purchased land on the farm Klipspruit on which part of the present-day Soweto is situated. The first township, named Orlando East, was laid out to cater for 80 000 people and houses were built. Residence rights and influx regulations promulgated at this time made it increasingly difficult for blacks to enter and settle in the area with their families.

During the years spanned by the Second World War the black population of Johannesburg increased very rapidly. The Native Affairs Department could not cope with the housing requirements because of a manpower shortage, and eleven shanty-towns mushroomed. Shacks were built of tin, cardboard, corrugated iron, hession, mud and bits of wood, and slum conditions prevailed.

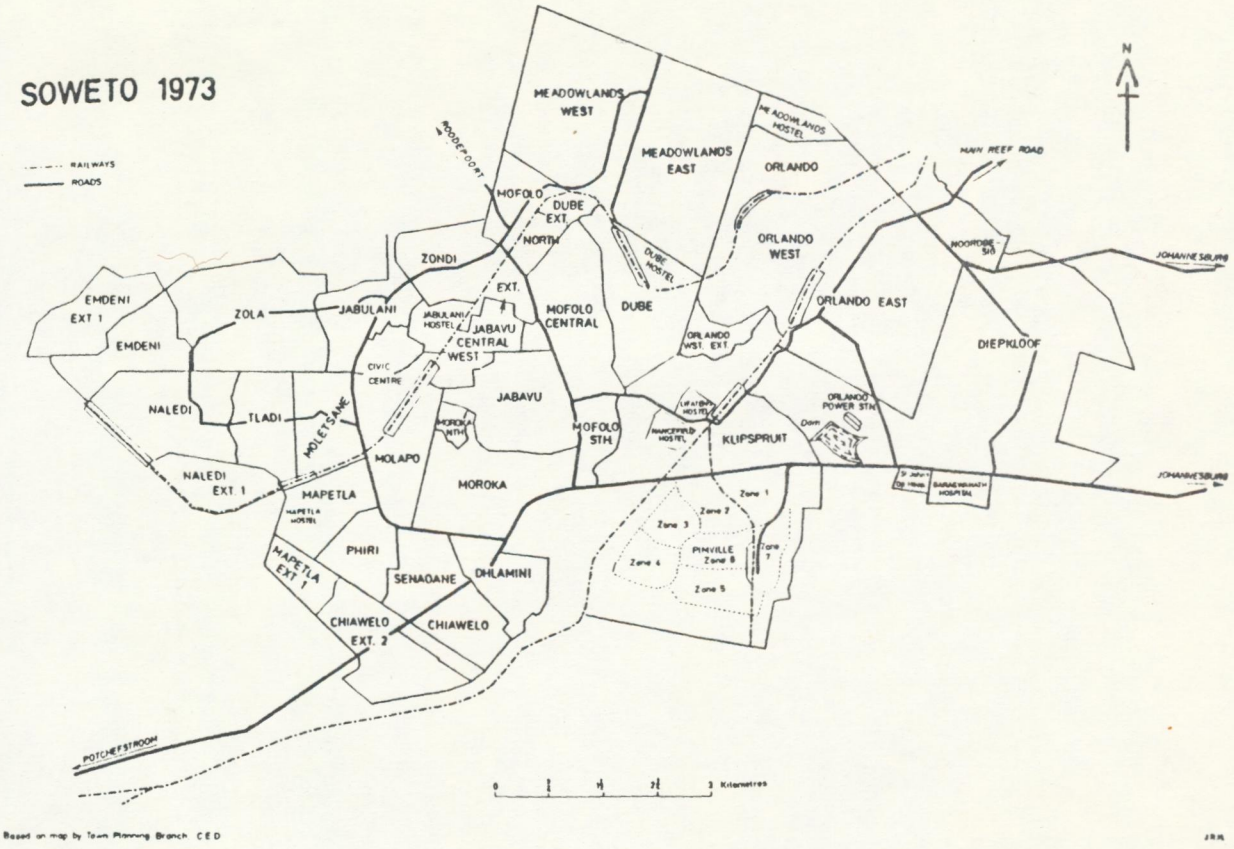

Fig 3.2 Map of Soweto showing suburbs, main roads, railways and hospitals



Fig 3.3 Houses in Soweto

Shortly after the war ended, however, work started on the improvement of these areas and on the provision of essential services such as running water and sanitation. The major part of this-work has taken place since 1956 and there are now no slum areas, such as existed before, in Soweto.

In 1976 riots broke out in Soweto. These riots were set off ostensibly by complaints about the education system, but beneath these verbalized complaints lay the miscellaneous, unspecified and officially ignored socioeconomic problems of life in Soweto. About 292 blacks and two whites died during the riots (Morris, 1980), many people were injured and schools, clinics, offices, beer halls and other buildings were burned and looted. It was only after these riots that the Government took a closer look at the rights of the urban blacks and, amongst other developments, legislation relating to leasehold was eased, restrictions on black businessmen were lifted, and the Government began to recognise the permanent status of urban blacks.

The administration of the twenty-six City Council-controlled townships that made up Soweto had been transferred in 1973 from the Council to the West Rand Administration Board (WRAB), a government controlled organization. At present WRAB is in the process of delegating powers to an elected black community council. This council has taken over certain functions, such as the allocation of housing. However, influx into the area is still very tightly controlled and it seems that in future, because of the apartheid (or separate development) policy and the development of the `Homelands´, fewer and fewer blacks will be eligible for permanent urban residential rights, and the resulting frustration and insecurity will continue.

3.1.1.2 Population statistics

Official estimates for the population vary considerably. The figure given in the last national census was 827 550 (National Census, 1980), but it is generally accepted that approximately 30 per cent of the population consists of illegal inhabitants who do not come forward for enumeration. The population at present is therefore probably nearer 1,2 million (Soweto. A Survey. Financial Mail Supplement, March 25, 1983). The population is young and in 1980, about 48.8 per cent were aged nineteen years or younger (Nel, 1982), compared with 40 per cent of the local white population (Hart and Lourens, 1977). The population is however aging since the proportion under the age of nineteen years in 1970 was 54.6 per cent (Nel, 1982). The estimated total population of Soweto is about the same as that found in Johannesburg itself, and both are balanced in terms of males and females.

The black population in South Africa has been grouped according to ethnological and linguistic characteristics and the two major groups are the Nguni and the Sotho (Seligman, 1966, p120). These two major groups are both represented in Soweto. The Nguni group includes the Zulu, who constitute 30.9 per cent of the total Soweto population, the Xhosa (9.7 per cent), the Swazi (6.6 per cent), and the Ndebele (1.6 per cent of the total) (Census, 1980). The Sotho group includes two major sub-groups, namely the Tswana, and the Northern and Southern Sotho together, these groups form 17.5 per cent and 23.1 per cent of the population respectively. Smaller groups of Shangaan/Tsonga (6.3 per cent of the population), Venda (3.8 per cent), and other peoples (unspecified, 0.5 per cent) are also living there (Census 1980). The figures for the different groups from the 1980 Census statistics appear in Table 3.1.

TABLE 3.1 Soweto Population by Ethnic Group

Ethnic group	No.
Zulu	255 540
Xhosa	79 860
Swazi	54 900
N. Ndebele	6 880
S. Ndebele	6 280
Tswana	145 140
S. Sotho	118 500
N. Sotho	73 140
Shangaan/Tsonga	52 100
Venda	31 200
Other	3 980
TOTAL	827 550

The birth rate in the South African black population is declining, as it is in all the South African groups (Randall, 1983). Estimates indicate that between 1960 and 1978 the black birth rate fell from 44 to 36 per 1000 of the population (South African Department of Health, Welfare and Pensions, Annual Report 1980). This decline is probably even more striking in Soweto (although no statistics are available) where family planning clinics are advertised and accessible and women are often the major breadwinners.

3.1.1.3 Household and family size

According to a recent Soweto survey (Soweto. A Survey. Financial Mail, Sup-

plement, March 25, 1983) there were about 105 000 houses in Soweto in 1982. Of these family houses 87 300 were rented and 18 648 owned. The shortfall of houses was about 35 000 units. The average house was originally built for a five member family and consisted of four rooms, viz. two bedrooms, a living room, and a kitchen. The average rent paid for such houses is R40 per month. Estimates suggest that these houses at present contain an average of about nine or ten people and there is therefore much overcrowding and a consequent lack of privacy (Morris, 1980). Single quarters, mostly for migrant male workers, who leave their families in the rural areas, are supplied in the form of eleven large hostels scattered throughout Soweto. Approximately 43 263 people live in these hostels (WRAB quarterly report, June 1976).

Basic services such as running water, sanitation, refuse removal and postal services are provided to all houses and work on electrification is proceeding rapidly. About 20 per cent of the houses were supplied with electricity by 1980 (Morris, 1980) and this figure is slightly higher now. In several areas high mast street floodlighting is installed.

The older suburbs of Soweto house people of different ethnic groups but, since 1955, zones have been set aside for each group. Houses that are vacated in the older areas are now filled with people from the ethnic group for which the area has been zoned. There are at present about ten predominantly Nguni areas, six Sotho, one Sotho/Nguni, two Venda/Shangaan/Tsonga, and eight mixed townships.

At least 15 800 families live in Soweto as lodgers or sub-tenants (WRAB Annual Report, 31/3/76), and many of these families are eligible for housing, but cannot obtain a house because of the critical shortage. About 40 per cent of the families living as lodgers have female heads (WRAB Annual Report, 31/3/76), whilst about 25.6 per cent of all families in Soweto have female heads (Nel,1982). The average family size is 5.3 whereas the average Soweto household is 5.51 (Nel, 1982).

The urban black family is not necessarily the extended family of the rural society, but generally consists of members of the nuclear family of the household head. According to Pauw (1963), the urban black family has a strong tendency to lose the father at an early stage and to develop a multi-generational span. Behaviour in the family is generally still based on the patriarchal tradition of the tribal society and on the principal of male dominance and female subservience. Where the female becomes the major wage-earner and assumes increasing responsibility for the budgeting, education ex-

penses and household management, the stability of the family is threatened. This alteration in the traditional roles in the family leads to marital discord, infidelity and a rising incidence of divorce (Hellman, 1971), as well as desertion, illegitimacy and associated problems in the sphere of parent-child relationships (Manganyi, 1973).

3.1.1.4 Income and unemployment

The income of Soweto inhabitants varies, there are those who are very poor and those who are very rich. According to a Bureau of Market Research report (Nel, 1982) about 20.7 per cent of black multiple households in Johannesburg earned less than R1 999 per annum in 1980 and 32.8 per cent earned more than R4 500 (the highest income category). The estimated Household Subsistence Level (HSL) for a family of six persons living in the Johannesburg area was given as R271 for the month of September 1982 (Randall, 1983), this figure showed a 12 per cent increase over that for 1981. Income and Minimum Living Level (MLL) were compared for Johannesburg for 1980 and, at that time, at least 28.6 per cent of households (the majority of whom had either uneducated, and/or female heads, and/or large families) were living below the MLL (Nel, 1982). The number of black wage-earners per household was found to be 1.51 (Nel, 1982).

Unemployment is a problem in South Africa and in Soweto. In the whole country about 24 per cent of the labour force was unemployed in 1982, of these about half were under thirty years of age, more than half had little or no education and about 25 per cent had never worked (Randall, 1983). A Market Research, Africa, survey showed that the unemployment rate in Johannesburg was 19.5 per cent and that those who were unemployed were also more likely to be in the younger age groups and have a lower standard of education (Morris, 1980).

3.1.1.5 Education

Soweto has 365 schools accommodating about 216 000 pupils (Soweto. A Survey. Financial Mail Supplement, March 25, 1983). The number of primary school children increases every year, but there are still probably only 84.5 per cent of the children in the primary school age group who are attending school in Soweto. The drop-out rate is a problem and in 1981 27.6 per cent of the pupils dropped out between the first and second years of school and only 51 per cent of scholars entered the fifth year of schooling. This fact has serious implications since many of these drop-outs with only four years

schooling are functionally illiterate. In terms of the UNESCO definition at least four years of schooling is required for literacy (Hartshorne, 1976).

The education level achieved by a sample of Sowetans studied in a Bureau of Market Research Survey (1978) is indicated in Table 3.2.

TABLE 3.2 Education levels for Sowetans*

Education level	No. of years schooling	% of sample
Nil	0	3,9
Grades	1 - 2	1,5
Standards 1 - 4	3 - 6	15,5
Standards 5 - 6	7 - 8	29,6
Standards 7 - 8	9 - 10	35,2
Standards 9 - 10	11 - 12	11,3
Post Matric		1,5
Unknown/refused		1,5
		100,0

* over 15 years old

Compulsory education is being introduced in Soweto and the State Department of Education and Training has agreed to provide the facilities, stationery, textbooks and teachers for a school, if the parents undertake to ensure that their children attend up to the age of fifteen years. Only fourteen schools in Soweto had entered into this agreement by 1982 (Soweto. A Survey. Financial Mail Supplement, March 25, 1983).

3.1.1.6 Health and Welfare Services

Health services are provided by three hospitals and nineteen clinics in Soweto. The three hospitals include one general (Baragwanath Hospital), one Tuberculosis, and one eye hospital. They are all sited on the east border of Soweto (see fig.3.2) and the medical staff are mainly white doctors who travel to Soweto daily from Johannesburg. Baragwanath has 2 700 beds (Medical instrumentation donated to Baragwanath, SA Hospital Supplies, July 1983) and 7 000 staff including 532 doctors (Morris, 1980). It is the largest hospital in South Africa and in the 1981/2 fiscal year treated 1 538 926 out-patients and admitted more than 112 000 patients (Medical instrumentation donated to Baragwanath, SA Hospital Supplies, July 1983). The hospital is heavily subsidized and a fee of approximately R10-00 (depending on income) is charged per out-patient visit.

The nineteen clinics in Soweto provide a number of services to the community such as family and child health, tuberculosis, first aid, curative, midwifery, district nursing, dental and primary health care services.

In addition to these services there were, in 1980, sixteen private practitioners in Soweto, but less than 5 per cent of Sowetans obtained medical treatment from these practitioners, and these patients paid the required fee of R4-50 per consultation (Morris, 1980).

Apart from these Western type health services there are also several forms of traditional and indigenous medicine practiced in Soweto. These indigenous healers include `nyangas' (or herbalists) and `sangomas' (who diagnose the cause of disease through the use of bones or supernatural influences, and communication with the spirits of the ancestors). The majority of people in Soweto appear to use a combination of both Western and traditional medicine (Manganyi, 1974). In one community in the Transkei collaboration between Western Medical practitioners and traditional medicine men has been proposed, particularly in the field of public health where the recognition of conditions (such as tuberculosis) which require referral to sophisticated health services is required (Ingle, 1973). Such collaboration does not however appear to occur in Soweto.

Welfare services are also available in Soweto, although the demand is far in excess of the supply (Morris, 1980). The services include those generally offered for: the child and family, the aged, the alcoholic and drug dependant, crime prevention and the rehabilitation of offenders, those with mental and other health problems, and the physically handicapped.

3.1.1.7 Amenities

Transport is available in Soweto in terms of trains, buses, and taxis. There are about 0.23 privately owned cars per family (Bureau of Market Research, 1977) and about 9.8 per cent of the commuters travel to work by car. The majority of workers use the trains (60.8 per cent) which are grossly overcrowded at peak hours when they carry an estimated 225 000 passengers. About 21.6 per cent of commuters travel by bus and 3.4 per cent use taxis (Morris, 1980).

The telephone system in Soweto is very poor and only about 16 per cent of the houses have telephones. Telecommunication is however improving, although the waiting list for telephones stands at 27 000 (Soweto. A Survey. Financial Mail Supplement, 25/3/83).

There are about 303 churches in Soweto (Horrell, 1982), and all the ma-

jor denominations are represented, including the Methodist, Anglican, Roman Catholic, Lutheran, Presbyterian and Congregational. These groups serve about 62 per cent of the church-goers. A further 22 per cent attend the African Independant churches, which were originally breakaway movements from the white run mission churches and now consist of about 900 different splinter groups and sects. These groups combine, in different degrees, elements drawn from traditional Christianity, ancestral worship and from magical systems of thought.

Other amenities in Soweto include libraries, post offices, banks, cinemas, theatres, community halls, beer halls, club houses, many illicit shebeens or bars, and some of all the usual sports facilities such as soccer fields and tennis courts (Horrell, 1982). There is one relatively big new shopping complex and there are 700 trading sites. Most people however travel to Johannesburg to do their shopping. Development of large chain stores and supermarkets in the area is at the planning stage. All these amenities fall far short of the needs of the Soweto people.

3.1.1.8 Crime

Soweto's violent crime rate is the highest in the country. The murder toll in 1982 was 1 275, and this figure exceeded, by 28 per cent, the numbers for 1981 (Soweto. A Survey. Financial Mail Supplement, March 25, 1983). The abuse of alcohol plays a large part in these murders and, for example, 86 per cent of victims were said to have had alcohol in their bloodstreams. In the average week in 1982 there were ten cases of rape, thirty robberies, 100 thefts, twenty-five cases of house-breaking, twenty car thefts and a five armed robberies.

3.1.1.9 Conclusion

Soweto has been called a `city within a city' (Hellman, 1971), since it has the population of a city but is not independent, self-supporting, or self-contained,as other cities are. It has a symbiotic relationship with the predominantly white city of Johannesburg, which is dependent on Soweto for its work-force. Soweto is an insecure city where rapid urbanization, social change and unmet needs are causing disorientation and frustration, disruption of the family, breakdown of traditions and cultural mores, and profound human maladjustment. But Soweto is also a developing city presenting opportunities, a middle class life-style and even wealth to those with ingenuity, initiative, stamina and education.

This brief description of the people and conditions in Soweto supplies the background information for the present study. Soweto's problems are multiple and this discussion deals with them only very superficially, but some knowledge of them is required to understand the nature of the subjects who took part in this study and to contribute to the interpretation of the findings.

3.1.2 Rural areas

Soweto provided a setting which was associated with all the problems as well as the facilities of urbanization. The sample of albinos obtained there might therefore have been biased either because albinos might be more likely than others to move from the rural to urban areas, in search of indoor employment, or because they might be more likely to remain in or return to the rural areas for protection. The urban sample could therefore produce inaccurate prevalence figures. Similarly, the urban ethnic groups of origin of the albinos might not be representative of these groups as a whole and for example those who migrate to the towns might be the fittest in the rural community,with no defects such as albinism in their history. The writer therefore decided to confirm the findings from the urban study by investigating the condition in four rural ethnic groups, two with high urban prevalence rates and two with low urban rates.

The rural areas selected were districts in Botswana, Kwazulu, Transkei and Swaziland (see Fig 3.1). Botswana was selected because it is peopled by the Tswana who showed a high urban rate of albinism (in Soweto), and there was an anthropologist working in one of the largest towns who knew the people and the district very well, which facilitated the preparation for and the progress of the field-work. The district of Nongoma was chosen in Kwazulu because: the Zulu people in Soweto showed a low rate of albinism and a rural Zulu sample was required; there is a central hospital at Nongoma around which the field-work could be based; the district is well defined and statistics were available. The reasons for including the Transkei were that it provided a second sample of Xhosa people for which low urban rates were found, and the writer had the opportunity of accompanying a touring team providing services for the blind in various rural districts, which were otherwise difficult to reach. The last rural area selected was in Swaziland, because the Swazis showed a high urban rate of albinism which required confirmation in a rural sample, and they are a cohesive group with a good communication network. For each of these areas the geographical position, popu-

lation statistics, cultural, mating and life-style patterns, which are important in understanding the subjects in this study, are described briefly in this section.

3.1.2.1 Botswana

Botswana is an independent country situated on the North West border of South Africa. It is surrounded by Namibia on the West and North, Zimbabwe on the North-East, and South Africa to the South-East and South. Much of the country consists of the Kalahari basin which is a region of semi-desert, inhabited by hunter-gatherers known as San (or "Bushmen") and the pastoralist Kgalagadi. In the East, however, the area changes to dry savanna woodland and it is on this Eastern North-South strip, which is the most developed and best endowed region in the country,that 80 per cent of the population of 936 000 (1981, Census) live (Africa, South of the Sahara, 1983).

Unlike many other black rural groups who live in scattered villages built around the extended family, many of the rural Botswana live in a few large towns, of mostly mud and wattle thatched dwellings. One such town provided the setting for the rural study of the Tswana people. The town, Mochudi, with a population of 18 300 (1981 Census) is situated on the Eastern strip, close to the South African border (see fig 3.1). The people living there are Bakgatla, a sub-division of the Tswana cluster of the Sotho group of Bantu-speaking people (Schapera, 1966, p20). Ox-drawn wagons form the chief means of transport. The water supply comes from bore-holes and is carried between the pump and houses. Lanterns and candles are used for light since there is no electricity. Contact with whites has extended over about 140 years, so many of the traditional cultural patterns have undergone changes. The town now has churches, schools, trading stores, a small hospital, a post-office, smithy, police station, and the dwellings of a few whites who serve as missionaries, medical practitioners, veterinary officers, traders and blacksmiths. The Bakgatla chief however still has a certain amount of power over the people, although this has to some extent been eroded by government officials and administrators.

The extended family group forms the basic social unit in Mochudi. Many years ago marriages used to be arranged preferably between cousins according to the old local proverbs: `side by side with his cousin a man is always happy´ and `child of my paternal uncle marry me so that the cattle should return to our kraal´ (Schapera, 1966, p41 and 42). Cross-cousin marriages

i.e. with the daughter of a maternal uncle or paternal aunt were preferred. Cousin marriages, it was claimed, were most likely to succeed because the girl would be well known to the boy's people and the couple, as near relatives, would be more tolerant of each other; the marriage would bind the families together, ensuring harmony and co-operation, and the cattle given at the marriage by the boy's parents to the girl's would remain in the family. In a study covering four generations, Schapera (1957) found that 32 per cent of marriages were between men and women who were near kin (second cousins or closer relatives). Such marriages appeared to be becoming less common, but the majority of marriages were nevertheless between people of the same ward or village. Most of the families living in a ward are related to their head through descent in the male line from a common ancestor, so although marriages might not have been between first or second cousins they were probably frequently between relatives, which is an important fact for the purposes of the present study.

The people of Mochudi subsist mainly on their crops, cattle, and a little hunting. Each family has its own fields outside the town and many stay near their fields during the rainy season, returning to the town during the dry season from about July to November. Much further away from the town are the cattle-posts where the cattle and other livestock, such as goats, sheep, pigs and fowls, are kept. These animals are cared for all year round by herdboys who do not have much contact with the villages at all. At present many families have members who have left temporarily to work in the urban areas of South Africa, the majority on the mines and others in industry, chiefly in Johannesburg. So there is much movement between the town and the big cities. Even in the 1930's about 40 per cent of the men were away at work every year (Schapera, 1966, p143), leaving their wives to look after the lands, homes and children and their sons to herd the cattle at the outposts.

3.1.2.2 Kwazulu

The Republic of South Africa contains several `homelands' which are areas which have been set aside or reserved entirely for black habitation, and where no whites may buy land or live. Kwazulu is one such homeland, and it is composed of several rural, fragmented and separate districts scattered mostly in the province of Natal between the escarpment of the Drakensberg mountains and the sea. The vast majority of the black people living in these districts belong to the Natal Nguni group and they are known as the Zulu

nation and speak the Zulu language. The term Zulu however is a generic term, adopted for ethnographic convenience (Hammond-Tooke, 1974, p63) since Zulu is the name of only one of the clans making up the nation. The Zulu-speaking people are not one clan nor are they one political entity, but they consist of those groups who were subjected by the Zulu king, Shaka, in the early nineteenth century, after his massive raids and massacres of his neighbours and others further afield. The Zulu-speakers are divided into about 300 groups, which the Zulu kings have ruled over for a century and a half and among which they promoted a uniformity of language and custom.

The original clan of the Zulu king lives in the district of Nongoma, which is one of the most northerly districts of Kwazulu (see Fig 3.1). The area covers about 400 square kilometres and the small town of Nongoma, with its district magistrate, officers of health and education, agricultural and other government officials, shops, churches and small hospital, is centrally situated. This district was selected for the field-work on the Zulu group because of these facilities, and the circumscribed nature of the area. The king's palace is sited a few kilometres North of Nongoma and one of his wives lives there. The king himself moves between this and his other mansions and families elsewhere in Kwazulu, but visits regularly and calls together his local Nongoma Zulu chiefs for discussions from time to time.

The population of Kwazulu was approximately 3 442 140 at the time of the 1980 Census. Nongoma district had a population of about 90 000 people, according to the figures of the district magistrate's office, at the time the field-work for the present study commenced. These people live, as is the custom in all the Nguni groups, in small, scattered, family villages. Each village has a circular cattle-fold, surrounded by a stout fence of sticks and branches, where the sheep, cattle and goats are kept at night (Seligman, 1966, p125). Around this fold the huts of various members of the family are built. These huts are no longer of the classical beehive shape constructed by the people when they were semi-nomadic pastoralists, but they are now more permanent dwellings round in shape, with mud and wattle walls, pressed dung floors and thatched roofs, and in some cases the main house is brick with a corrugated iron roof. Each of these family settlements, known as kraals, has a common open space and an enclosure reserved for the men, and the whole settlement is surrounded by a horse-shoe shaped fence. Between every settlement are the fields and grazing grounds belonging to each.

Marriages with kin, especially those of the same clan name or `isibongo',

are forbidden among the Nguni (Hammond-Tooke, 1974, p192). However, although marriage with a woman from the clans of all four grandparents is generally proscribed, it may be permitted in some groups with a woman from the maternal grandmother's clan. Polygyny exists and a man may have a few wives, but a wife may only have one husband. The society is both patrilineal and patrilocal, and the wife moves to her husband's home after marriage. The marriage is finalised by the transfer of bride-wealth or `lobola' and such lobola, although it may be paid, usually in cattle (but sometimes in cash), over many years, by the groom or his father to the father of the bride, determines the filiation of the children. These children will only belong to the father once he has paid the full lobola and they will then be given his surname.

3.1.2.3 Transkei

Apart from the `homelands' such as Kwazulu, South Africa has within her borders some independent `states'. These `states' were homelands whose leaders asked for independence, and, although still economically dependent on South Africa and not internationally recognised, these states are mainly self-governing. Transkei is one such state. It is situated on the South East coast of South Africa (see Fig 3.1) and this area has been inhabited by groups of the Nguni people, called the Cape Nguni, for several centuries (Schapera, 1937, p46). The groups living in the area speak the same language (Xhosa), with small variations, but a common line of descent cannot be traced for all of them. The Xhosa language contains many click sounds indicating that contact between these Negro people and the Khoikhoi (or Hottentot), a local small and light-skinned pastoral people, or with the hunter-gatherer San, who lived in the area many centuries ago, was extensive. The result of this contact affected not only the language but the skin colour, and the Xhosa-speaking people are lighter than those Nguni groups without this admixture in their history. Monogenic characters, like the immunoglobulin allotypes (GM) have been used by Jenkins et al (1971) to quantitate the amount of Khoisan admixture in the various Bantu-speaking populations of Southern Africa.

The Transkei extends over 41 002 square kilometres and its population was 1 751 142 at the time of the 1970 Census. Apart from the black inhabitants other population groups, including 9 556 whites, 7 645 coloureds and ten Asians, also live in the area. About 1,5 million Transkei nationals are living and working in South Africa (Africa, South of the Sahara, 1983).

The people in the Transkei have, in general, the same cultural customs as

the Nguni people described in the previous section on Kwazulu. They live in scattered homesteads and family life and marriage patterns are very similar. About 78 per cent of the population are illiterate and, since they are widely spread out over a large area, some of which is mountainous with few roads, the communication net-work is poor. Basic health services are provided at clinics, some of which are situated in the remoter areas. These clinics are often cut off during the rainy season when rivers flood and roads are washed away. Field-work for the present study was organised at seven of the accessible clinics and at one school for the blind in Umtata, the capital of the Transkei. As a result information was gathered on eight of the twenty-eight regional districts.

3.1.2.4 Swaziland

Swaziland provided the setting for the last part of the rural study. Swaziland, like Botswana, is an independent country on the border of South Africa. It is situated to the east of the Transvaal and is surrounded by South Africa on the North, West and South, and separated by the coastal plain of Mozambique from the Indian Ocean (see map, Fig 3.1). Swaziland is a kingdom and one of the smallest political entities in Africa (Africa, South of the Sahara, 1983). The area of the country is 17 363 square kilometres and the higher parts of the country on the west support temperate grassland while the eastern lower strip is characterised by dry woodland savanna. The economy is well developed: sugar, cotton, maize, tobacco, rice and fruit are produced; minerals such as asbestos, coal and anthracite are mined; and the forests are extensive.

The population of the country was 634 678 in August 1982 (Africa, South of the Sahara, 1983). According to the 1976 Census the population consisted of Swazi(97.4 per cent), Zulu (1.3 per cent), Shangaan (0.7 per cent) and other peoples (0.6 per cent). About 20 000 Swazi nationals were in South Africa as migrant workers. In 1968 at the time of independence about 40 per cent of the country was under white control, at present however, about 80 per cent is under Swazi control.

Swaziland is divided into four districts. Each district has a district commissioner who is in constant contact with community leaders and an effective net-work of communications has been established. The Hhohho district is in the North-West of the country, and since it is the smallest and most compact of the districts it was selected for the field-work for the present study. The district had a population of about 160 000 people (Central Statis-

tical Office, 1982) living in two small towns and in scattered villages. There was a network of clinics organised by the local department of health and the departments of education and agriculture had extension officials working in urban and rural areas among the people. The district commissioner was in touch with local Swazi leaders and headmen. The roads, although mostly gravel, were in reasonable condition.

The Swazi people belong to the Nguni group of Bantu-speaking peoples and have a similar culture to that described under the Kwazulu section above. The sub-group is of relatively recent origin. At the time the Zulu king, Shaka, was on the rampage, a chief, Sobhuza, was increasing his power, and the `Swazi´ people began to form a distinct group. Sobhuza´s descendent Mswazi (c 1840-1875) conquered many neighbouring groups and subjected the people living in Swaziland or expelled them; his followers took their name from him (Schapera, 1937, p51).

The choice of marriage partners in the Swazi is not subject to as many prohibitions as that found in other Nguni groups. The Swazi man may be permitted to marry into the clan of either of his maternal grandparents or into that of his paternal grandmother. The Swazi appear to particularly approve of the marriage of a man with a woman who has the clan name of his father´s, or mother´s, mother (Kuper, 1963). This marriage preference has important connotations for the present study.

3.1.3 The greater Johannesburg metropolitan region and Pretoria

In order to assess the impact of an albino baby on a black family a sample of newborn affected babies and their parents was required. Since Soweto did not have a population sufficiently large to provide a reasonable size sample of such babies in the limited time available, other smaller local areas inhabited by the black population had to be included. Most of these areas were within the boundaries of what is sometimes called the greater Johannesburg metropolitan region (see Fig 3.4) or the Witwatersrand complex.

The towns in this region lie east-west along the gold-bearing reef, the area to the East of Johannesburg is known as the East Rand and that to the West as the West Rand. Each town has its commercial and industrial centre and its white and black residential areas. Each of the black areas is similar in physical and social characteristics to Soweto and is served by a local hospital and clinics. Some of the people living in these areas travel to Johannesburg to work, but most work in commerce, or industry, or on the mines near their place of residence.

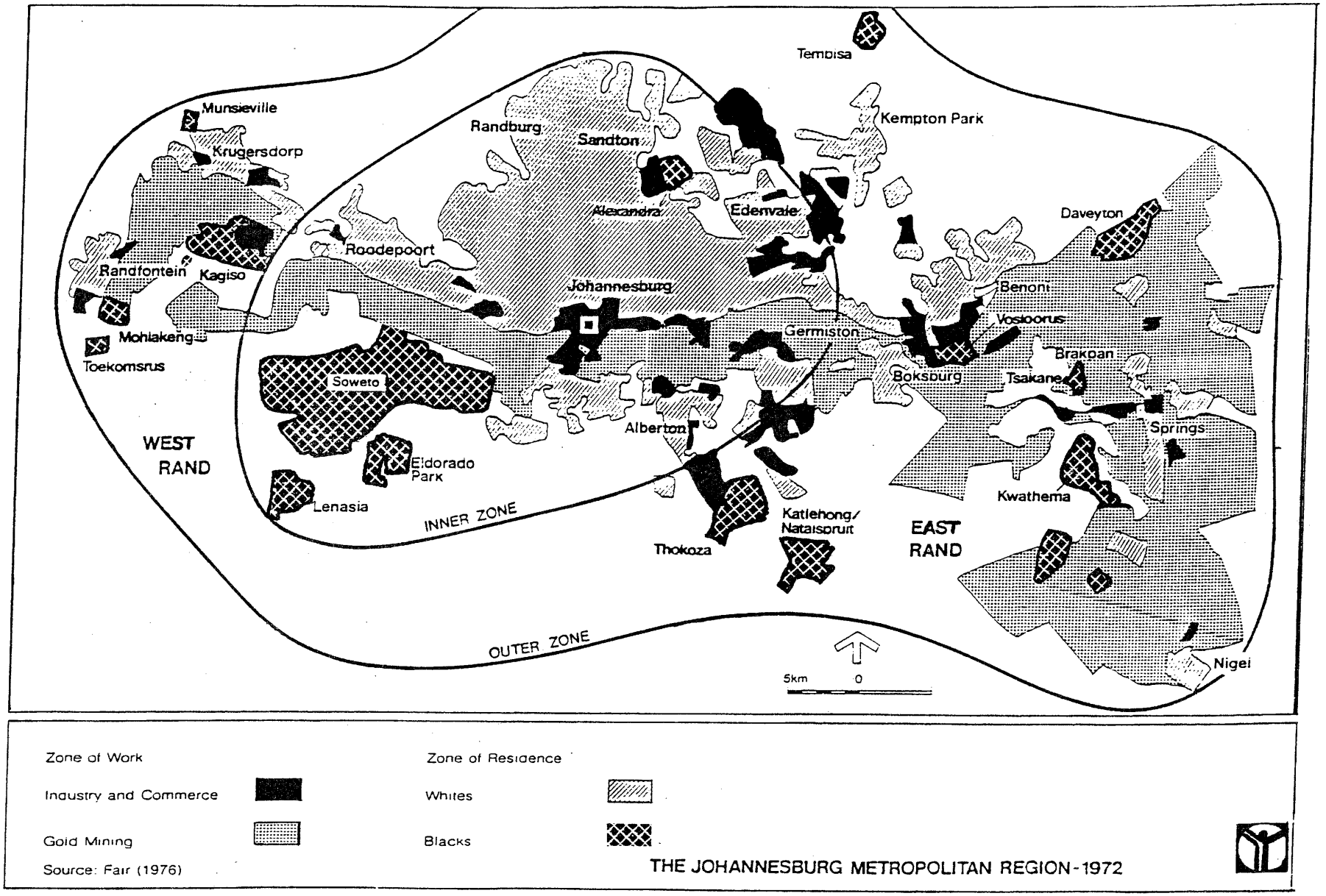

Fig 3.4 Map of the greater Johannesburg metropolitan region showing areas in which field-work was undertaken

Apart from these greater Johannesburg regional areas the black suburbs of Pretoria were also included in this study. Pretoria is situated about fifty kilometres north of Johannesburg and has a large black population living in townships, similar to, though much smaller than, Soweto, on the outskirts of the city. These suburbs have their hospitals and clinics where most babies are delivered. The newborn albino babies ascertained in these areas were admitted with their families to the study.

3.2 Materials and Methods: Genetics studies

The materials and methods chosen were based not only on their congruence with the aims of the study (see Chapter 1, 1.2) but also on an understanding of the social milieu in which the study was carried out and which is described above. The methods used in the identification and selection of experimental subjects, in the selection of controls, in data collection, and in the statistical analysis are presented below.

3.2.1 Identification and selection of subjects

The objective was not so much to find a sample population of albinos but to identify all the people reported to be albinos in the selected urban and rural areas, to see each one in order to confirm the diagnosis of oculocutaneous albinism, and to enlist their co-operation in the study.

3.2.1.1 Urban Study

The identification of albino subjects for the urban study was carried out in Soweto. The subjects were ascertained through seven different sources:

(1) the schools in Soweto (120 schools at the time of the commencement of the study)
(2) the six health clinics
(3) Baragwanath Hospital
(4) St John's Eye Hospital
(5) welfare organisations
(6) community social workers
(7) families with albino members

At each source the names and addresses of albinos were obtained, these subjects were visited and where they were found to have oculocutaneous albinism they were admitted to the study. Some subjects had partial albinism or vitiligo (these two conditions were often confused), or, in the case of babies, they were sometimes very light-skinned at birth and became darker

later. These subjects were excluded from the study, but they will be discussed briefly in the results chapter.

A secondary small group of subjects was ascertained through the specialised cancer clinic attached to the Hillbrow Hospital in Johannesburg. This hospital is used as a referral centre, and people with cancer are referred there from the Witwatersrand complex. These subjects participated in the section of the study on skin cancer and types (hairs were collected from them in order to identify whether they were tyrosinase-positive or negative albinos). Further supplementary information was obtained on the problem of skin cancer from the records of the histopathology department, although the albino subjects themselves were not seen.

The third urban group of subjects who were identified and interviewed were those at the workshop for the blind. This workshop had been removed from the Johannesburg area and resited near Pretoria, and many albinos were employed there. This group also took part in the hair-bulb tests for types.

In Soweto 250 subjects were ascertained, and those who were accessible and co-operative participated in the hair-bulb tests, physical examination, cancer, life expectancy and fertility studies. Every subject was supplied with anti-actinic cream (gratis) and bus-fare was paid wherever necessary.

3.2.1.2 Rural studies

(i) Botswana

The subjects for the rural Tswana sample were ascertained through an anthropologist, his Tswana assistant, and through the local missionary doctor in Mochudi. Names and addresses were obtained, the subjects were contacted by the anthropologist and his assistant before the visit of the writer, and arrangements were made for as many as possible of the affected people living in Mochudi to see the writer during the three days of the field-work. Altogether fourteen subjects were reported, ten were found to be oculocutaneous albinos and were interviewed and the remaining four were not available (two being at the cattle-posts far from the town) at the time of the field-work.

(ii) Kwazulu

In the Nongoma district of Kwazulu the albinos were ascertained through:

(1) the school board officials, inspectors, principals, teachers and school children
(2) the agricultural officers
(3) the magistrate and his messengers
(4) the district nurses and hospital workers

The accessible schools in the district were visited during the first week of the field-work and the pupils reported to be albinos were seen and those affected were admitted to the study. The school children were asked by their teachers to inform the local people that the study was in progress. In this way a `bush-telegraph´ system was set up and, during the second week of the field-work, several subjects presented because of this method of ascertainment. Altogether fifteen subjects were reported, thirteen were seen and two were not available for interviewing.

(iii) Transkei

The subjects were ascertained through the district health workers, particularly those who were visiting outlying clinics and offering a service to the blind. These latter workers had sent out notices to hospitals and clinics in selected Transkei districts, informing them of the dates they would be available at each local venue and asking them to call in all the blind people who required treatment. At the same time, on the writer´s behalf, they requested all the albinos to attend the clinics on the specified days. The writer then travelled with the team and interviewed the albinos who presented themselves. This method of ascertainment however was not very successful, as will be seen from the results. The area was so large and so underdeveloped, and the people so widely scattered, that it was not possible to use the methods of ascertainment which were found to be productive in the Botswana and Kwazulu studies.

A sub-group of thirteen albino subjects was however found at the local school for the blind and these pupils participated in the hair-bulb study.

The subjects who were excluded because they were not affected with oculocutaneous albinism, although reported to be albinos by the local informants, will be described briefly in the following chapter. In all forty-three subjects were seen and interviewed (including those from the school for the blind).

(iv) Swaziland

A preliminary visit was made to Swaziland, which is more easily accessible to Johannesburg than the previously mentioned rural areas, in order to initiate communication with the relevant authorities and facilitate the finding of subjects. Contacts were established with staff of the ministries of health, education, and agriculture, and with the district commissioner. Information concerning the study was discussed with these officials who were requested to inform the leaders of the community with whom they worked. The district hospitals and health clinics were visited, the staff were told

about the study, and their co-operation in informing the people in their areas was sought. Suitable places and dates for meeting with the albino subjects were assessed and selected, and a list of the finalized arrangements and the programme was widely circulated. These methods of ascertainment were very successful, forty-nine subjects were seen and admitted to the study, a further twenty-eight affected relatives were not available for interview. A small group of those reported as albinos did not have oculocutaneous albinism and they were excluded. They will however be discussed briefly.

The subjects and their families participated in the prevalence study, those albinos and their mothers (excluding those who were pregnant) who were co-operative took part in the skin reflectance study.

3.2.2 Selection of controls

Control subjects were included in two of the sections of the study, namely those dealing with skin reflectance and fertility.

For the skin reflectance study carefully matched controls were required. These controls were matched with the mothers of albinos (obligatory heterozygotes) according to ethnic group, age, sex and rural life-style. The majority of the controls were selected from a group of Swazi women, in Swaziland, who were attending the local health centre (which was used as a venue for the albinism study) for either the `well-baby´, or the family planning clinics. Pregnant women were excluded from the control group as such women are known to darken in skin colour. Women with a history of albinism were also unacceptable as controls. In six cases however, difficulty was experienced in obtaining matched controls in Swaziland. The reasons for this problem were that many of the experimental subjects were in the older age group, while the source of the controls was the younger age group attending the health centre, and time was limited. These six controls were therefore obtained from a Soweto clinic attended by Swazi women. This small group were tested within two months of the initial group testing. Although they were urban dwellers and might have been said to have had less exposure to the sun than rural dwellers and therefore be lighter in skin colour, this factor, if it affected the control group at all, would have probably biased it towards the lighter side of the scale of measurement.

Matched controls were also required for the fertility studies. These controls were selected from the Diepkloof suburb of Soweto to participate in both the psychosocial and fertility studies. A description of the methods

used in their selection appears in the section 3.3 below.

3.2.3 Methods of data collection

Once the subjects and controls were identified and their co-operation obtained for the study they participated in the methods of data collection and submitted to various tests. In brief the following techniques were used: structured interviews; hair-bulb incubation tests; physical examinations; and the measurement of skin reflectance. Additional data (to supplement that obtained at the physical examination) on skin cancer was collected from records at the local Histopathology department and the cancer clinic. These techniques and reasons for their choice are detailed below.

3.2.3.1 The interviews

Personal interviews were used as a method of data collection since this appeared to be the best way of obtaining information on the subjects, their families and their social background. The posting of questionnaires to the subjects was not considered since the population under study was generally quite unsophisticated and had not been exposed much to this type of data collection. The interviews were structured and a brief schedule (see Appendix A) was used to maintain uniformity. The advantages of using such interviews were that personal knowledge of the subjects and their circumstances could be gained. The disadvantages of this method were that the interviewees might modify facts, suffer from faulty memory, lack of insight or inability to articulate (Young, 1956, p208).

The interview was generally conducted by the writer in the home of the subject in the presence of the mother and often other relatives as well. Since most of the subjects and their families did not speak English, interpreters were used in most cases. These interpreters were usually black nursing sisters from the local clinics, or black research assistants, who helped in translating as well as in the tracing of subjects and drawing up of pedigrees.

The schedule used at the interview contained items on: name, address, method of ascertainment, sex, ethnic group, date and place of birth, alternative residences (for purposes of maintaining contact with the subject all possible addresses were taken), employment (past and present), income, education, periods of and reasons for hospitalization, year of marriage (if applicable), the spouse (normally pigmented and/or a relative), offspring and/or siblings (where applicable), and the family history of albinism. After this

schedule was completed a family pedigree (see Fig 3.5) was constructed for as far back as the subject and relatives could remember. For the purposes of the fertility study particular emphasis was laid on the collection of information on the children of the albinos, their siblings and on the siblings of the obligatory heterozygotes in the family, and details of those alive and deceased were noted. The age at death of any affected members of the family was recorded, so that information on life expectancy could be obtained. The respondents were also asked about consanguineous marriages and the nature of the relationship between those who were related. Several interviews were necessary in some cases for the collection of all this information.

3.2.3.2 The hair-bulb incubation test

The hair-bulb incubation test as described by Witkop et al (1970) was selected as the method by which tyrosinase-negative and tyrosinase-positive albinos could be identified. This test provides an objective and scientific assessment of the tyrosinase status of individual albinos which a physical examination cannot give. The test requires laboratory resources, so it was only carried out when these were easily accessible or when the hairs could be collected and transported to the laboratory within twenty-four hours.

A local superstition prevalent in the black community, holds that anyone who obtains, for example, hair or nails from a person will have power over that person or may cast a spell on him or her. For this reason some subjects refused to allow hair to be taken and others insisted that if it was taken it must be disposed of carefully after testing. Notwithstanding this belief, hairs were collected from seventy-nine subjects in Soweto as well as twenty-five subjects at the two schools for the blind (in the Transkei and Pretoria) and eight at the local cancer clinics.

When a subject agreed to co-operate at least twenty hairs were plucked from the crown of the head using forceps with curved tips. The hairs were then separated into two groups, one was placed in a tyrosine solution and the other was used as a control and immersed in a 0.I M potassium phosphate buffer with a pH of 6.8. The tyrosine solution (which was generally prepared by a laboratory technologist) consisted of L-tyrosine (80 mg per 100 ml) dissolved in 0.1 M potassium phosphate buffer at a pH of 6.8. Both the experimental and control hairs were incubated at 37°C for twelve to fifteen hours and they were then examined for the presence of pigment using a low-power microscope. If pigment was present the subject was tentatively assigned to the tyrosinase-positive group of albinos, if no pigment was observed the ten-

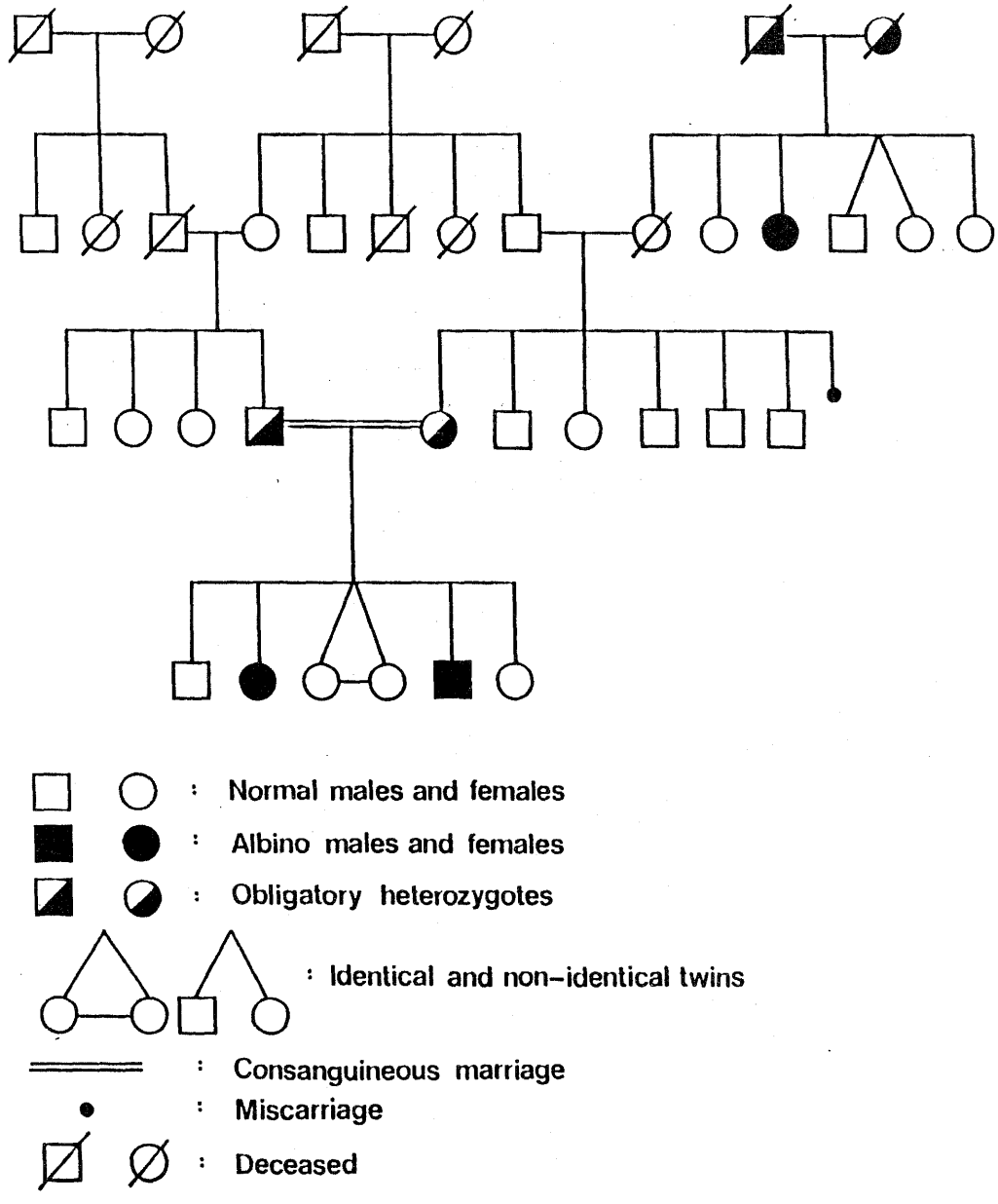

Fig 3.5 Sample pedigree

tative category was tyrosinase-negative.

Only anagen hairs (Kukita, 1957) with well-formed intact hair-bulbs were suitable for this test. Because it was not always apparent, when the result was negative and no pigment was observed, whether this finding was due to the hairs being from a true tyrosinase-negative albino, or whether the hairs plucked had been damaged in collection (Taylor (1983) has reported that hair can be very fragile in albinos) or were by chance catagen hairs, the test was repeated, whenever possible, on those with negative results. Also when siblings were tested and, for example, one was found to have a negative result and two had positive results, it was assumed that the negative result was probably due to the hairs being inadequate for testing and the three siblings were all classified tyrosinase-positive.

When the result from the hair-bulb incubation test was available the findings were correlated, in cases where a physical examination had been carried out, with the phenotype, before any subject was assigned to a particularly category.

3.2.3.3 The Physical examination and criteria for classification

A physical examination was organized for the subjects wherever possible since a clinical description of the albinos was required, together with the hair-bulb test results, for the purposes of classification. This examination was carried out by three medically qualified staff of the department of Human Genetics, to which the writer is attached.

In conjunction with the medical practitioners, a list of features to be noted at the examination was drawn up, by the writer, in the form of a schedule, which was duly completed for each subject. This schedule (see Appendix B) included items on the name, date, accompanying persons, ethnic group, general build and health status, skin (description of exposed and unexposed areas, presence of malignant or premalignant lesions and ephelides) and hair colour, eyes (colour, translucency, nystagmus and photophobia), teeth, trunk, heart and lungs. A note was also made on whether hairs were collected for the incubation test and whether a photograph of the subject was taken. The examiner then remarked on anything unusual found during the course of the examination and, if the subject was co-operative, a specimen of blood was collected for a parallel study on blood-grouping and enzyme studies, which was being conducted by the medical practitioners (the results of these tests will not be reported in the present study).

Altogether 140 albinos were examined, sixty-one did not have hair-bulb

incubation tests, and these subjects could not be classified. The five groups used for the purpose of classification were tyrosinase-negative (ty-neg) albinism, tyrosinase-positive (ty-pos) albinism (with its two sub-types, ty-pos A and ty-pos B), yellow mutant (ym), brown albinism (BA) and rufous albinism (RA). These five groups were selected from the ten defined by Witkop et al (1983), their remaining five groups consisted of syndromes associated with albinism in four cases and autosomal dominant albinism in the other (see Chapter II Table 2.1b). It is possible, but very unlikely, that the subjects in this study had any of the latter five conditions, their characteristics as reported by Witkop et al (1983) are quite distinct and would probably have been detected had they occurred.

The criteria (modified from Witkop et al, 1983 and Witkop, personal communication, 1984) used to characterise each of the five selected types were as follows:

(1) Tyrosinase-negative albinism: total absence of any pigment in the skin, light yellow or white hair, blue irides, nystagmus, photophobia, and a negative hair-bulb test.

(2a) Tyrosinase-positive albinism (A): lack of generalized pigment in the skin, but localized pigmented ephelides, naevi, or spots could be present, yellow hair, blue to light brown irides, nystagmus, and a positive hair-bulb incubation test.

(2b) Tyrosinase-positive albinism (B): lack of generalized pigment in the skin and of any pigmented spots, light yellow hair, blue irides with some visible pigment radiating outwards from the pupil, nystagmus, and a negative hair-bulb test.

(3) Yellow mutant albinism: lack of generalized skin pigment, localized pigmented spots in some cases, dark yellow-orange or gold hair, dark blue or light brown irides, nystagmus, and a hair-bulb test showing no (or a doubtful) increase in pigmentation.

(4) Brown albinism: lack of generalized skin pigment, but the skin colour is light brown or tan and pigmented freckles may be present, light brown to brown hair, blue, hazel to light brown irides, nystagmus not always present.

(5) Rufous Albinism: lack of generalized pigment but the skin is red to copper-coloured, light gold to light reddish-brown hair colour, light to dark brown irides, and nystagmus may or may not be present.

Fig 3.6 shows an example of each of these types from the local series of



(a) Male with?tyrosinase-negative albinism

Fig 3.6 Types of albinism: examples from the present study



(b) Female with tyrosinase-positive A albinism



(c) Female with tyrosinase-positive B albinism, showing spokes of pigment in the irides



(d) Male with?yellow mutant albinism



(e) Female with brown albinism



(f) Female with rufous albinism with normally pigmented nurse



(g) Young male with rufous albinism, showing skin colour

albinos.

The frequency of each physical characteristic such as hair colour, skin pigment, and ocular findings, described by the medical practitioners was calculated for each group.

3.2.3.4 Collection of data on skin cancer

In order to obtain information on the type and site of skin cancer found in albinos and the risks for developing cancer three different methods were used. Firstly, during the physical examination malignant and premalignant lesions in the subjects were noted by the medical practitioners. Secondly, the records on skin biopsy specimens, obtained from surgical operations at Baragwanath Hospital, on all patients between January 1976 and March 1980, and sent for investigation (for ? carcinoma) to the Histopathology Department at the S A Institute for Medical Research, were examined. The numbers and sites of squamous and basal cell carcinoma in albino and normally pigmented black patients were noted from these Histopathology records. Thirdly, the patient records for albinos attending the local cancer clinic for dermatological assessment and treatment were obtained and details on the types and sites of skin cancer in this group were noted and analysed.

By means of these three methods data were collected on skin cancer in albinos, so that risks according to age could be assessed and the common types and most susceptible sites could be determined.

3.2.3.5 The Skin reflectance test

Skin reflectance measurement by means of a reflectance spectrophotometer was the method used to assess skin colour. This method appears to give the most reliable and objective data on skin colour at present available and therefore it was the method of choice for the purposes of the present study.

A sample of eighteen Swazi mothers of albinos, eighteen normally pigmented matched controls (twelve in Swaziland and six in Soweto) and forty-three Swazi albino subjects, participated in the skin reflectance study.

The portable EEL reflectance spectrophotometer was used to measure the reflectance values in each subject at six different wavelengths. The instrument consists of a galvanometer and a detachable reflectometer head (see Fig 3.7). The machine was run on the mains where electricity was available, and on a 12-volt battery, where mains electricity was unreliable or unavailable (as at two rural centres).The reflectometer head directs light from a

lamp through a 3/8 inch diameter aperture on to the surface of the skin at an angle of 45°. A wheel of nine filters, which typically sample the visual spectrum and have known dominant wavelengths, is mounted inside the head. The wheel can be turned and the required filter selected. The light is then passed through the filter onto the skin, some of the light is absorbed, while the reflected light is picked up by a photocell and measured on the galvanometer unit. Before the reflectometer head is connected to the galvanometer the zero setting is adjusted, and for each filter the sensitivity is set to give a reading of 100 per cent against a standard white tile every time the filter is used. The reflectance from the skin is then measured at the selected wavelength by reading off the percentage reflectance on the linear 0 - 100 scale. If the nine filters are used in turn the instrument gives an abbreviated spectrophotometric curve of light reflected from the skin.

The movable head attached to the galvanometer unit can be applied to the skin at any site. Some workers have used the flexor surface of the right forearm (Barnicot, 1958) but most agree that the medial aspect of the left upper arm is the preferred site (Weiner et al, 1964). This position is easily accessible and relatively protected from the effects of tanning and exposure, and it was therefore selected for use in the present study (see Fig 3.8).

Measurements were taken at six different wavelengths namely 426, 465, 515, 545, 575 and 685 nm represented by filters 601, 602, 604, 605, 606 and 609, respectively. About half the measurements were made by Professor D F Roberts, who was visiting South Africa from Newcastle, England, and who trained the writer in the technique of using the EEL instrument, and the remaining half were then made by the writer.

3.2.4 Methods of Statistical Analysis

Gene frequencies and carrier rates were calculated by means of the Hardy-Weinberg formula (as discussed by Clarke, 1977). Segregation analysis was carried out using Hogben's method as outlined by Emery (1976). 'Student's' t tests and chi-square tests were used where applicable to determine whether two groups showed differences that were significant.

Due to the nature of the measurements in the skin reflectance study non-parametric statistical tests were applied, since these tests do not require that the underlying variable be continuous and normally distributed, as is required for the t test. The Wilcoxon signed ranks test (Lehmann, 1975),



Fig 3.7 EEL reflectance spectrophotometer

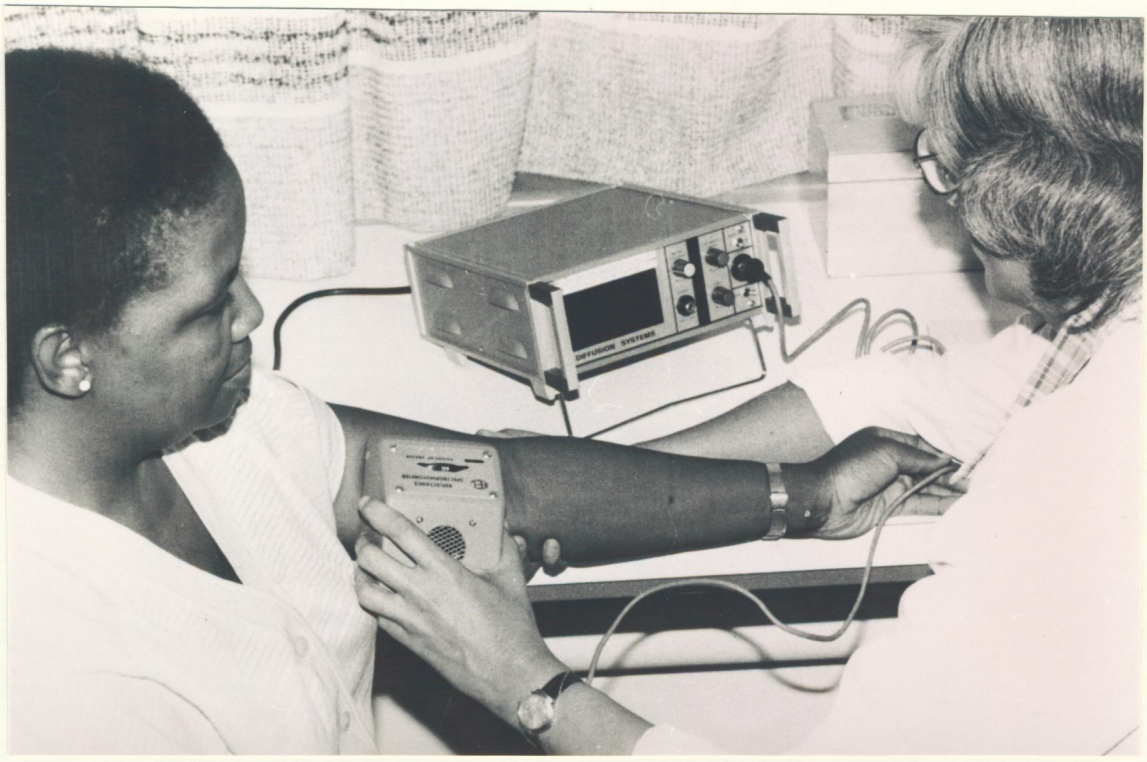

Fig 3.8 Taking skin reflectance measurements

which is based on the differences between experimental and control scores for every matched pair, was selected for analysing the data. The p values were determined by means of a one-tailed test and where a p value of 0.05 or less was found the differences between the groups were considered significant.

Since the chi-square test does not take into account the order of age categories, the Kolmogorov-Smirnov test (Siegel 1956, p47) was also applied to the data on ages. This test is concerned with the agreement between two cumulative distributions. For comparing life-time infertility rates the exact Binomial test (Snedecor, 1980) was used since the population consisted of only two classes. Values for p of 0.05 or less were considered significant throughout these tests.

3.2.5 Summary

Southern African Negro populations provided the albino and normally pigmented control subjects for this study. These subjects were drawn from the large urban community of Soweto, from four rural populations in Botswana, Kwazulu, Transkei and Swaziland, and from the black suburbs of the greater Johannesburg metropolitan region and Pretoria. A number of methods of ascertainment were used and prevalence rates were estimated by standard methods.

Interviews were conducted with the subjects using a specially designed schedule. Detailed pedigrees were constructed for each family for the assessment of fertility and life expectancy. The hair-bulb incubation test was used to assign the albinos into ty-pos and ty-neg categories. Subjects were examined physically both to facilitate and determine their tentative classification and so that their characteristics and problems could be described.

During the physical examination the presence of malignant and premalignant lesions was noted by the assisting medical practitioners. Further information on skin cancer was collected from the records on skin biopsies in the local Histopathology department and from the files of the cancer clinic.

Skin colour and fertility of both homozygotes and obligatory heterozygotes were investigated and compared with normal matched controls, and/or with the general population.

The data collected by the methods detailed above were analysed by means of statistical tests, whenever applicable, and the findings are presented in the following chapter.

3.3 Materials and Methods - Psychosocial study

This part of the study was considered to be exploratory, since neither

the subject of the albino baby and the black family, nor that of maternal-infant interaction had been investigated at any depth in the local black community, prior to the present study, and standardised reliable and validated methods of data collection were not available. The most comprehensive way of examining the infant's impact on the family would have been to look at the albino baby in the context of the family, including parents and siblings. This however was not feasible since some of the affected babies had no siblings and in other families the siblings were living in the rural areas with relatives. It was also difficult to involve fathers, since many mothers were single, and in other cases working fathers were unco-operative. The focus of the study was therefore placed on the mother and her relationship with and attachment to her infant, and fathers were included whenever possible. The purpose was: to obtain information by means of interviews and observations from mothers and fathers of albino, and as controls, of normally pigmented babies, on their feelings about, behaviour towards and attachment to their babies; and to compare the two groups to determine the differences between them, in an attempt at elucidating the impact of the birth of an albino baby on black parents. In addition, to get some idea of the effect of the infant on the siblings and the grandparents, the parents were asked for their perception of the responses of these members of the family. Parents' attitudes towards their new babies are probably altered by time and in order to examine any possible changes the writer decided upon a longitudinal study to include at least the first year of the infant's life.

A programme of field-work was designed to obtain the required information. Methods of collecting the data, of finding the subjects, and of training an interviewer were assessed and selected. Plans were made for interviewing and observing black families into which an albino baby had been born and, for comparison, black families with a new-born normally pigmented infant. Statistical methods were sought for the analysis of the data. These methods are discussed and oulined in the following sections.

3.3.1 Design of the study

Information concerning the feasibility of the study was obtained prior to the initiation of the field-work. It was found that if approximately twelve hospitals with maternity wards in Johannesburg and the satellite towns were willing to co-operate by reporting affected babies, sufficient black babies would be delivered in those wards to make it likely that one albino baby would be born every month in one of the participating hospitals. There would

then be eighteen babies available for study over eighteen months. Since a longitudinal study of babies and their families over fifteen months was planned, it was envisaged that the study would be a long-term one and that the data would be collected over approximately three years.

Once the feasibility of the study had been established the study was designed and applicable methods of obtaining information relevant to the aims (See Chapter 1, 1.2) were sought. Two methods were selected: firstly, semi-structured interviews using a specially constructed schedule would be conducted for the collection of reported information; secondly, observations of specific maternal and paternal behaviours and maternal- and paternal-infant interactions would be made during a predetermined activity for the collection of more systematic and measurable data. Once the subjects had been identified both methods would be used at every visit to them. Such contacts would be made during the first week after the birth of the baby and subsequently at three monthly intervals, until the babies reached the age of fifteen months and six interviews had been completed.

The main group of subjects were the dyads of mother and infant. A secondary group of subjects, the fathers of the infants, were included and interviewed wherever possible, at least once during the study.

To compare the impact of the birth of an albino baby on a black family with the impact of the birth of a normal baby on a family the design of the study included the selection, interviewing and observation of a matched control group.

The interviewer and observers to be used in the study were carefully selected and trained in the required methods prior to their involvement with the subjects. A pilot study was carried out both in order to train the observers and interviewer in the methods of data collection and to determine and correct any problems arising with the schedule and observations.

The results from the data collected from the subjects and their matched controls were analysed by means of statistical tests wherever possible. Thus information was obtained both on the nature of the initial impact of the birth of an albino baby on a black mother and father, and on any changes which might have occurred in the parents over time, in comparison with parents of normally pigmented babies.

3.3.2 Identification and selection of subjects

Subjects were sought over a period of two years, from May 1979 to April 1981. All the albino babies who survived the neonatal period were admitted to the

study. The minimum sample size was set at twelve, but the writer hoped to have at least fifteen subjects participating throughout the study.

Approaches were made to twelve hospitals and clinics, since a monthly birth rate of at least 4 000 babies was necessary to ensure the delivery of, on the average, one albino baby every month (the prevalence of albinism in this area being approximately 1:4 000, Kromberg and Jenkins, 1982). The co-operating hospitals were all within an eighty kilometer radius of Johannesburg (see Fig 3.4) and their monthly birth rates are shown on Table 3.3.

TABLE 3.3 Participating Hospitals and their approximate monthly Birth Rates (1979)

Hospital	Approximate Birth Rate per month
West Rand	
1. Baragwanath Hospital	1 200
2. Soweto clinics (4)	1 000
3. Leratong Hospital	300
East Rand	
4. Alexander clinic	120
5. Natalspruit Hospital	600
6. Boksburg-Benoni Hospital	400
7. Far East Rand Hospital	300
8. Tembisa Hospital	400
Pretoria & districts	
9. Kalafong Hospital	230
10. Mamelodi Maternity Home	200
11. Holy Cross Maternity Home	250
12. Garankuwa Hospital	350
TOTAL	5 350

Each of these hospitals (with the exception of the Far East Rand, where the Superintendent was contacted telephonically) were visited by the writer and the research assistant and the project was discussed with the superintendent, the matron, and usually also the senior sisters in the maternity wards. They were asked to inform the writer immediately by telephone when an albino baby was born. To reinforce this request luminous green stickers reminding staff to report albino births to the writer were printed and displayed in each ward where deliveries were performed. Since it was expected that some albino births would not be reported each hospital was routinely telephoned

once a fortnight.

When the hospital staff telephoned concerning an albino baby, the name and address of the mother, and the date of delivery were noted, and the nurse was requested to inform the mother that, unless she objected, she would be visited at home soon after discharge from the hospital.

Although albinos were familiar to the nurses and doctors at the hospital it was not always easy to make the diagnosis in the immediate postnatal period, since many black babies have a very light skin colour at birth. Thus, after one experience where the family home was situated eighty kilometres from Johannesburg and a visit and examination of the baby indicated that it was not an albino, the new-born babies were seen in hospital by either the writer or the assistant or both to check on the diagnosis, soon after the birth. At the same time the writer and assistant introduced themselves to the mother, briefly informed her about the project, obtained her co-operation and requested permission to visit her at home.

3.3.3 Selection of Controls

In order to discover whether the parents of albino babies were behaving differently to other parents with new-born babies a group of controls was selected. This group was matched with the experimental group for the sex and date of birth of the baby.

Since occasionally long distances had to be travelled in order to interview the experimental group the control group was selected from the nearest available and accessible black area, which was the suburb of Diepkloof in Soweto.

Once the albino baby was born and had been seen at home for the first time then the Diepkloof Clinic (where approximately 200 babies are born each month) was visited. The records of the clinic were examined and the names of six babies of the same sex as the albino baby and born either the same day or the day before or after were noted. The names of these babies were written on pieces of paper which were folded and one was randomly selected. If this family was not at the given address then another name was drawn, until a family was found. The nature of the study was explained, and the families were informed that the writer was interested in the relationship between mothers and fathers and their new-born babies and how these relationships developed. If the family agreed to co-operate then they were told that they would be visited every three months until six visits had been made. In this way the matched randomly selected control group was constituted. The writer

and assistant together made the selection of controls for the first two cases and thereafter the assistant herself used the predetermined procedure for selecting controls.

3.3.4 Selection of the research assistant/interviewer/observer

The decision regarding the use of either a black or a white interviewer, or both, for the in-depth interviews required for the psychosocial study, was discussed at length by the writer with anthropologists and psychologists working in the field. The writer then decided that a black research assistant should complete the interviews and observations during home visits, but that the writer would attend at least two visits, one near the beginning and one at the end of the study on each family. The black research assistant would be carefully trained by the writer in the methods to be used for the observations as well as in administering the schedule, and would be in consultation with the writer throughout the project. This decision was made because certain items in the schedule were involved with personal details and feelings, and it was assumed that the families would feel less inhibited with a black worker, and consequently she would probably obtain more reliable information. Also, a white interviewer would often have to use an interpreter, which causes communication problems, disjointed interviews and sometimes creates misunderstandings.

The research assistant selected had had three years' experience in interviewing research subjects and using schedules for projects conducted by the department of African Studies at the University of the Witwatersrand. She was proficient in the commonly used Bantu languages and could translate the items on the schedule into the home language of the subject in most cases. She was also a nurse and had good contacts with many of the hospitals, as well as some experience of new-born babies and their problems. These aspects assisted in making the research project proceed smoothly.

For the observations of the mothers and fathers and their babies the writer decided that it would be preferable to have two black observers, one of whom would be the chosen research assistant. A second black observer (also an experienced research worker) was selected and trained in the methods of observation, until the scores of the two observers for a subject were very similar. The scores of both observers for each subject could then be assessed, for the final study, by means of correlation coefficients, and the subjects could be given a mean score. This method would have prevented to a certain extent, possible observer bias. However soon after the training ses-

sions were completed the second observer was offered a better job, transferred out of Johannesburg, and no other suitable replacement could be found.

3.3.5 The interview and schedule construction

`In social research the interview is used for securing information in face-to-face associations which aid in gaining `a portrait of human personalities', broad enough to encompass the social background which influences behaviour, and deep enough to reveal inner strivings, and tensions, attitudes and wishes' (Young, 1956, p206). A specialized form of the interview is useful if personal details, responses to predetermined specific questions and quantification of the data is required. Interviews provide information, on the behaviour of the informant, which is not only dependent on the accuracy and validity of the responses of that individual, but also on the skills of the interviewer in eliciting the information. Nevertheless, interviews enrich the study of people and permit the good interviewer to gain knowledge on `motivation, emotional responses, and social processes as they are reflected in human experience and social situations' (Young 1956, p207). A semi-structured standardized interview was therefore the method of choice for the psychosocial section of the present study.

Schedules (see Appendix C, D and E) were constructed by the writer for use in the interview, so that comparable date would be collected from all the subjects. These schedules included both dichotomous and open-ended items. Various sources were consulted such as the study by Gath (1978) on Down's Syndrome and the family and Klaus and Kennell's (1976) work on maternal-infant bonding. Items were compiled using material from these sources as well as from discussions with psychologists, medical practitioners and from the writer's own experience.

The schedule for the first interview was designed to obtain information in the following areas:

(1) Identifying details: names, dates of birth, address, marital status, ethnic group, occupation, education, consanguineous marriages.

(2) Medical history: operations, fractures, hospitalisation, headaches, sleeplessness, tiredness, stomach pain, breathlessness, depression, `grumbles' (general complaints).

(3) Childhood history of mother: brought up by father/mother/both/relatives, rural or urban childhood, number of siblings, history of albinism.

(4) Details about the baby: length and problems of pregnancy and deli-

very, special care in the neonatal period, feeding methods, habits and milestones; future plans for the baby, expected problems and medical treatment required.

(5) Relationships and feelings: feelings of parents, siblings and grandparents about the baby; involvement of the siblings with the baby; the marriage relationship; fathers' activities and attention before and after the birth; plans for future babies; and any changes in family organization.

(6) Beliefs on the causes of albinism.

(7) Any comments or complaints about members of the family or any concurrent problems.

A slightly different schedule was designed for the second interview, since on some items (e.g. See 1 and 3 above) information was only required once, whereas on others it was repeatedly required. The third and all subsequent interviews were very similar to the second, except that an item on medical problems of other members of the family was inserted. The items on relatives' reactions were omitted as the responses were becoming repetitious. Five other items were added regarding the changing patterns of behaviour and attitude of the father and siblings, and the marital relationship. For example, in addition to the item `are you planning to have another baby now', a new item to assess action in this regard was inserted `are you using contraceptives now'.

At the last interview the Taylor Manifest Anxiety scale (Taylor, 1953) was given to each mother (see Appendix F). This scale was selected, since it had been adapted by a local research worker, validated and found to be reliable for South African black miners (White, personal communication, 1979) and was available in four of the major languages (Zulu, Xhosa, Tswana and Southern Sotho).

A separate schedule was drawn up for the father of the baby (see Appendix G). The items in this schedule were very similar to those in the mother's schedule. It was hoped that the schedule could be completed with the fathers around the same time as the mother's second interview i.e. when the baby was about three months of age.

The schedules were scored by the writer by comparing the responses of the matched pairs item by item and assessing the differences. The results were based on those differences which were found to be statistically significant. For the Anxiety Scale (which requires true/false responses) however, a

total score was compiled for each subject by using a simple 1 - 0 scale, where 1 represented a negative response indicating anxiety and 0 a positive response indicating no anxiety for each item. The scores for each item were then totalled, and a high score indicated a high anxiety level.

3.3.6 Methods of Observation

`Observation is a systematic and deliberate study through the eye of spontaneous occurrences at the time they occur' (Young, 1956, p154). The purpose of such observation is to `perceive the nature and extent of significant interrelated elements within complex social phenomena, culture patterns, or human conduct' (Young, 1956, p154). The method of observation was therefore selecto supplement the material gathered at the interview. The objective for the observation was to see the mother, and, separately, father, relating to the baby in a specific situation, and to specify and record the behaviours exhibited in this situation. The assumption was that such behaviour would reflect on the parents' attachment to and feelings for their child (see Klaus and Kennell, 1976, p65 and p84 - 85). The addition of this method was considered important, since the mothers and fathers in the study might have been unable or reluctant to verbalize their feelings about their babies, but their attachment to these infants might become apparent during observation sessions.

Various activities were considered during which the parents would have to interact with the baby, and their relationship and behaviour with the infant could be observed. Observations have been made for this purpose using activities such as a physical examination of the baby by a doctor, breastfeeding, and free play (Klaus and Kennell, 1976). The activity chosen for the observation on mothers was napkin-changing, which was suggested as a suitable method for stimulating maternal-infant interaction by members of the local paediatrics department. These staff had casually observed that mothers appeared to interact a great deal with their babies in this situation and show a range of behaviours which could be recorded. For the purposes of the present study the mother was asked to pick up the baby, put him on the bed, change his napkin and replace him in his crib or sleeping place. It was explained to her that there was no right or wrong way of doing this and that she should act as she normally did with her child.

The interaction was scored for the presence or absence of the following maternal attachment behaviours: eye-to-eye contact, en face position, smiling at, talking to, kissing and stroking the baby, holding the baby close,

naming the baby, supporting the head, placing him/her securely on the bed, careful handling, appearing relaxed and caring, keeping the baby clean and settling him well, and asking questions about the baby (the full list appears on the Observation form in the Appendix H). The first seven of these behaviours were based upon Klaus and Kennell's work (1976) and the remaining behaviours were added to provide further information.

It was noted after a few trial observations, during the pilot study, that the black baby's head is very strong and does not need support to the same extent that a white baby's head needs to be held (confirming Geber's (1958) findings of precocious development in African infants). The relevant item on the observation form was therefore omitted and the subjects were scored on fifteen items. Where the behaviour occurred during the observation the observer circled the `yes' response on the observation sheet, where it was not observed the observer circled `no', and where the observer was not sure `don't know' was marked. A simple 2-1-0 scale was used where `yes' was scored 2, `no' scored 0 and `don't know' scored 1. The scores of all items were then summed to give each subject a total score. The matched pairs in the two groups were then compared on individual items and on total scores using the McNemar test for matched samples (Siegel, 1956).

The fathers were observed in interaction with their babies at the three month interview. They were requested to pick up and hold the baby while the interviewer made some notes on the baby's appearance, and then put the child down again. The behaviour of the father was scored in the same way, on the fifteen items, as that of the mother (see Appendix I).

Since few black babies are still wearing napkins at twelve months the observations on napkin-changing were only carried out at one week, three, six and nine months. At the fifteen month interview a new activity was introduced and the mother was asked to sit with her infant while he played. Carefully selected toys, including a white doll, two black dolls, two cars, four blocks, two boxes containing paper clips, and one rattle, were given to the dyad to encourage play and interaction. The mother was informed that we were interested in observing the child at play. She was asked to sit on a squared mat and after being given the bag of toys she was told that she could do as she pleased. The time limit set for the observation session was ten minutes. The mat was marked out in twenty-five centimetre squares so that the maximum distance between mother and child could be estimated. The behaviours that ensued during the play and concomitant interaction were counted by making

strokes on the form on which selected behaviours for both mother and baby were listed. The number of times the behaviour occurred was then summed so that each subject was given a total score for each behaviour, and the experimental and control groups were compared on these scores.

The behaviours noted were: touching, holding, smiling, gazing, distress, vocalization, fuss cry, mediating the environment and behaviour not contingent to the infant. The maximum physical distance between the mother and child was also entered onto the form (see Appendix J).

The research assistant, after training during the pilot study carried out the observations, with the writer making spot checks and participating in a few observations at random, and the writer scored and analysed the results.

3.3.7 Pilot studies

In order to find out whether the schedules were relevant and suitable, whether the questions were easily understood, whether or not the time taken to respond was too long, and whether the behaviours selected for observation actually occurred and could be counted, a pilot study was organized. For this investigation twenty mothers were selected at random from the maternity wards at Baragwanath Hospital. They were interviewed and then observed while changing the napkins of their new-born babies. As a result of this pilot study some items in the schedule were altered and others were omitted.

The pilot study provided a means of training the research assistant. All the pilot interviews and observations were carried out with the writer present and participating. Discussion after each session provided the assistant with an opportunity of learning, and the meaning of items in the schedule and behaviours for observation were clarified. By the end of the pilot study the writer and the assistant were in agreement on the scoring and the interpretation of the behaviours in the observation situation.

Similarly, before the play situation was used for observation purposes with the experimental and control groups, a pilot study was conducted with eight normal mothers and babies who were selected from the Diepkloof and Moroka clinics in Soweto. The writer and the assistant observed the behaviour of these dyads during the selected activity and, after discussion, agreement was reached on what was required from these observations.

3.3.8 Procedure for data collection

The procedure for collecting the data may be summarized as follows:

(1) The feasibility of the study was determined.

(2) Schedules for interviews with mothers and fathers were constructed and lists of behaviours for observation on both parents were compiled.

(3) A pilot study using the constructed schedule and list of behaviours for observation was conducted with twenty black mothers with new-born normally pigmented babies.

(4) Liaison with participating hospital staff was maintained, so that albino babies would be reported soon after their birth and the experimental subjects (the parents of the affected babies) obtained.

(5) A control group consisting of parents of normally pigmented babies, matched with the experimental babies for age and sex, was selected.

(6) Mothers in both the experimental and control groups were interviewed at one week, three, six, nine, twelve and fifteen months after the birth of the baby.

(7) Observations of the mother's behaviour in relation to her infant were made during a napkin-changing activity at one week, three, six and nine months.

(8) Fathers were interviewed three months after the baby's birth.

(9) Observations on the father's behaviour in relation to his infant were made by means of an activity at three months.

(10) A pilot study on making observations of maternal-infant interaction in a play situation was conducted with eight black dyads.

(11) Maternal-infant behaviour and interaction was observed in experimental and control dyads in a play situation at fifteen months.

(12) Participating mothers completed the anxiety scale.

By these means it was hoped that sufficient data would become available from this exploratory study to score, analyse and reach conclusions on the maternal and paternal feelings and attachment behaviour in relation to the infant, as well as to provide some information (collected from the parents) on other members of the black family, so that the impact of the new-born albino baby on the family could be assessed.

3.3.9 Characteristics of the sample

The sample initially consisted of thirty-seven mothers and their albino babies born between February 1979 and April 1981. At least one interview was conducted with the mothers of these thirty-seven babies and matched controls were selected according to the predetermined procedure.

Two sets of twins were reported, one set where both babies were albinos,

but they were small, weak and premature and both died neonatally, and the other where one baby was albino and the other normal, they were healthy and the albino twin was admitted to the study. Another mother who had an albino baby had come into the city for the delivery, and she intended to leave for the rural areas immediately after discharge from hospital, so she was not included in the study.

As expected the ratio of boys to girls in the albino babies was 1:1 and a normally pigmented baby of the same sex was selected as a control in each case. The date of birth of the controls was between 0-13 days after the birth of the albino baby, and in the majority of cases the control baby was one born within 0-5 days of the albino's birth. The control group was therefore very closely matched with the experimental group in terms of the selected criteria.

Although many of both the experimental group and the controls did not complete the required six interviews the data on all thirty-seven subjects in both groups will be presented here as they were initially admitted to the study. The reasons for subjects being dropped and for those who dropped out will be discussed in a subsequent section.

3.3.9.1 Age

The age in years of the mothers and fathers in the experimental and control groups is presented in Table 3.4.

TABLE 3.4 Age at first interview

	Experimental Group				Control Group			
	Mothers		Fathers		Mothers		Fathers	
Age in years	No	%	No	%	No	%	No	%
15 - 19	8	21.6	1	2.7	7	18.9	1	2.7
20 - 24	15	40.6	6	16.3	18	48.7	12	32.5
25 - 29	10	27.0	11	29.7	7	18.9	11	29.7
30 - 34	3	8.1	11	29.7	4	10.8	9	24.3
35 - 39			4	10.8	1	2.7	1	2.7
40 - 44			1	2.7			3	8.1
45 - 49			1	2.7				
50 +			1	2.7				
No information	1	2.7	1	2.7				
TOTAL	37	100	37	100	37	100	37	100

The mothers in both groups were much younger than the fathers, 21.6 per

cent and 18.9 per cent of the mothers in the experimental and control groups respectively being aged less than nineteen years, whereas 2.7 per cent of fathers in both groups were nineteen years or younger. The mother's age range was also shorter than the fathers.

The groups showed a very similar pattern, the majority of mothers being in the twenty to twenty-nine year age group (67.6 per cent of both experimental and control subjects). Also in both groups the number of younger parents under twenty years of age was similar (21.6 per cent of mothers in the experimental group and 18.9 per cent in the controls, and only 2.7 per cent of fathers in both groups). The control group mothers were however slightly younger (mean age 23.4 years) than the experimental group (mean age 25.8 years) but the difference was not statistically significant ($p > .20$).

3.3.9.2 Place of Residence

The address where the subjects were found at the time of the first interview is shown on Table 3.5. There was much movement during the study, since it

TABLE 3.5 Place of residence at first interview

Place of residence	Experimental Group		Control Group	
	No	%	No	%
Johannesburg				
Soweto	16	43.3	37	100
Alexandra	4	10.8		
City	2	5.4		
East Rand				
Natalspruit	3	8.1		
Tembisa	2	5.4		
Vosloorus	2	5.4		
Brakpan	1	2.7		
Thokoza	1	2.7		
West Rand				
Krugersdorp	1	2.7		
Randfontein	1	2.7		
Westonaria	1	2.7		
North				
Pretoria	2	5.4		
Quaggasfontein	1	2.7		
TOTAL	37	100	37	100

was a longitudinal one covering almost three years, and moving occurred both within the urban areas and between the urban and rural areas.

The majority of subjects were from Soweto (43.3 per cent), Alexandra Township (10.8 per cent) and Natalspruit (Germiston, 8.1 per cent). The remaining experimental subjects were from outlying areas ranging from Vosloorus on the East Rand to Westonaria on the West Rand.

The control group was selected entirely from Diepkloof, an accessible and convenient area in Soweto and there was not as much movement in this group as in the experimental group.

3.3.9.3 Marital status

The details regarding the marital status of the sample and whether the parents of the selected babies were living together or apart are shown in Tables 3.6 and 3.7.

TABLE 3.6 Marital status at first interview

	Experimental group		Control group	
Marital status	No	%	No	%
Married	18	48.6	10	27.0
Single	16	43.3	27	73.0
Divorced	1	2.7		
No information	2	5.4		
TOTAL	37	100	37	100

TABLE 3.7 Father and Mother Living together/apart

	Experimental group		Control group	
	No	%	No	%
Living together	20	54.0	16	43.3
Living apart	14	37.9	20	54.0
No information	3	8.1	1	2.7
TOTAL	37	100	37	100

In the experimental group there were more mothers who said they were married (48.6 per cent) than there were in the control group (27.0 per cent). Consequently single mothers represented 43.3 per cent of experimental group but the majority (73.0 per cent) of the control group. This difference was statistically significant (chi-square 6.56, $p<.05$). Divorce was admitted in only one case in the total sample. Only 54 per cent of the experimental and 43.3 per cent of the control women were living with the fathers of their

babies at the time of the first interview. The difference between the groups was not significant (chi-square 2.50, p>.20), so that the groups were similar regarding living arrangements.

Among the mothers in the experimental group 23.7 per cent had had children by two fathers and in the control group 26.3 per cent of mothers were in the same situation, and one control mother had had children by three different men.

This information indicated that there was a great deal of family disorganization in both groups represented in the study, which reflects the general trend in Soweto as described in Section 3.1.1.3 of this chapter.

3.3.9.4 Other children

The findings concerning the children in the sample families are reported in Table 3.8.

TABLE 3.8 Number of children

	Experimental group		Control group	
Number of children	No	%	No	%
1	17	46.0	15	40.6
2 - 4	17	46.0	20	54.0
5+	2	5.4	2	5.4
No information	1	2.6		
TOTAL	37	100	37	100

There was a large proportion of mothers (46 per cent of experimental and 40.6 per cent of controls) where the baby under consideration was their first child. There were 46.0 per cent of the experimental subjects and 54.0 per cent of the controls who had 2 - 4 children, but the majority of these children were not living with their biological mothers. The two groups were similar regarding numbers of children and in both groups 5.4 per cent had had five or more children.

3.3.9.5 Ethnic affiliation

The mother was requested to give information on her own and the baby's father's ethnic affiliation during the first interview. The data obtained are presented in Table 3.9.

Approximately half of the experimental and control subjects were from the Sotho-Tswana speaking groups, i.e. groups 1 - 3 in Table 3.9, (56.7 per cent of mothers and 51.3 per cent of fathers in the experimental group, and

48.8 per cent of mothers and 46.1 per cent of fathers in the control group). A smaller proportion of each group were from the Nguni population group, i.e. groups 6 - 8 (37.9 per cent of mothers, 40.6 per cent of fathers in the experimental group, and 40.4 and 32.3 per cent respectively in the controls). The

TABLE 3.9 Ethnic Affiliation

Ethnic group	Experimental group				Control group			
	Mothers		Fathers		Mothers		Fathers	
	No	%	No	%	No	%	No	%
1. Tswana	10	27.0	7	18.9	5	13.6	3	8.1
2. Sotho	8	21.6	7	18.9	5	13.6	3	8.1
3. Pedi	3	8.1	5	13.5	8	21.6	11	29.9
4. Venda							2	5.4
5. Shangaan			2	5.4	3	8.1	3	8.1
6. Zulu	7	18.9	7	18.9	9	24.2	9	24.2
7. Xhosa	5	13.6	3	8.1	4	10.8	2	5.4
8. Swazi	2	5.4	5	13.6	2	5.4	1	2.7
9. Ndebele					1	2.7	3	8.1
10. Coloured *	1	2.7						
No information	1	2.7	1	2.7				
TOTAL	37	100	37	100	37	100	37	100

* of mixed ancestry

main difference between the groups was that there were more members of the Pedi group among the controls (21.6 per cent mothers and 29.9 per cent of the fathers) than among the experimental subjects (only 8.1 per cent mothers and 13.5 per cent fathers). This finding is probably not of much practical significance to the study since the Pedi were originally (about a century ago) an off-shoot of the Kgatla group of the Tswana people (Seligman, 1966, p123) and therefore they have cultural customs and a life-style similar to other Tswana groups.

Many marriages were between members of different ethnic groups except in the case of the Zulus, in both the experimental and control groups, who all had Zulu partners. There was one marriage where a coloured woman (with both black and white ancestors) was married to a Pedi man. There were only 42 per cent of the experimental group couples and 28 per cent of the control group where both members of the couple were from the same ethnic group. This finding might be expected for the controls since they were all selected from

Soweto where there is less ethnic cohesion and more intermarriage between ethnic groups than occurs in outlying areas. In none of the total of seventy-four couples was a consanguineous mating reported.

3.3.9.6 Education

All subjects were asked to give information on their own and their baby's father's level of eduction. Some mothers did not know these details about the baby's father but the responses from those that did are reported in Table 3.10.

TABLE 3.10 Level of education

	Experimental group				Control group			
	Mothers		Fathers		Mothers		Fathers	
	No	%	No	%	No	%	No	%
No schooling	2	5.4	2	5.4	1	2.7		
Sub A - Std 2	1	2.7	2	5.4	1	2.7		
Std 3 - Std 5	8	21.6	3	8.1	11	29.8	3	8.1
Std 6 - Std 7	15	40.6	9	24.3	14	37.8	13	35.2
Std 8 - Std 9	8	21.6	7	18.9	7	18.9	8	21.6
Std 10	1	2.7	2	5.4	2	5.4	4	10.8
Teachers Diploma	1	2.7			1	2.7		
Degree			1	2.7			1	2.7
No information	1	2.7	11	29.8			8	21.6
TOTAL	37	100	37	100	37	100	37	100

The two groups showed a similar picture and the majority of subjects had more than four years of education. Although there appeared to be a slightly higher level of education in the control group there was no statistically significant difference for the mothers (chi-square 2.12, $p > .90$) or for the fathers (chi-square 5.76, $p > .30$). More fathers and mothers (10.8 per cent and 8.1 per cent respectively) in the experimental than in the control group (0 and 5.4 per cent respectively) had four or less years of education and could thus be classified as functionally illiterate. These findings would be expected since the control group was drawn exclusively from Soweto, where there is a higher educational level generally than is found in other areas, whereas only 43.3 per cent of the experimental group lived in Soweto. The level of education however was similar to that found generally in Soweto (see Section 3.1.1.5 of this Chapter).

3.3.10 Composition of the sample

The sample was composed of different numbers of subjects at different stages of the longitudinal study. The population for the study was mothers and new-born babies which is, at best, in the black urban population, a very unstable group. Some mothers come into the urban area for hospital delivery, they may remain with relatives living close to the hospital for a few weeks or months, and then they return to the rural areas. Other mothers, particularly in families where the mother is the bread-winner, find that they cannot support themselves and their new-born baby. These mothers have several alternatives: either they may obtain the services of a baby-minder and return to work quite soon after the birth, or they take the baby to the grandmother in the rural area and return to work in the urban area, or they may return to the rural area and stay at the ancestral home there, until the child is big enough to be left with relatives. Many subjects therefore dropped out of the study at different stages of the research for one or other of these reasons. The number of subjects at each stage of the research is shown on Table 3.11.

TABLE 3.11 Number of subjects at each interview

Interview (Baby's age)		Experimental	Control	Matched pairs
1	(1 week)	37	37	37
2	($\pm$3 mo)	34	34	33
3	($\pm$6 mo)	26	23	22
4	($\pm$9 mo)	25	18	18
5	($\pm$12 mo)	23	19	17
6	($\pm$15 mo)	19	15	15

In three cases mothers went to the rural areas for a few months and then returned and they were retained in the sample, but skipped an interview and they therefore account for the information being incomplete at that particular stage. One mother missed two interviews in this way and the other two mothers missed one each. In Table 3.11 this is reflected most obviously in the fifth interview of the control group when there were nineteen subjects although there were only eighteen at the fourth interview. Because it was planned to use statistical tests for matched pairs only the data collected on these pairs could be used in the analysis and the remaining interviews were discarded.

Table 3.12 reflects the reasons why subjects dropped out of the study and could not complete all the interviews.

TABLE 3.12 Reasons for subject drop-out

Interview	2		3		4		5		6		Total	
Group	Ex*	Co*	Ex	Co	Ex	Co	Ex	Co	Ex	Co	Ex	Co
Reason												
Return to rural area	1		4	4	1				2	2	8	6
Return to work		1		2		2					0	5
Match dropped		1		2		2					0	5
Lived far away	1										1	0
Unco-operative			1			1					1	1
Skipped interview			1	1	1	1					2	2
Arrested								1			0	1
Untraceable							1				1	0
Baby too old	1	1	2	2							3	3
Baby died							2				2	0
TOTAL	3	3	8	11	2	6	3	1	2	2	18	23

* Ex = Experimental, Co = Control

A large group of subjects (eight experimentals and six controls) who dropped out did so because of unsuitable accommodation and the necessity to go to live with someone willing to support them, usually in the rural areas. There were however two subjects, one in the experimental group and one in the control group, who refused to co-operate. The experimental subject at the second interview, denied that her baby was an albino and her mother phoned, prior to the date arranged for the third interview, to say that her daughter was `being hurt' by the interviews and therefore they should cease. This request was respected and the family was invited to approach the writer if they required assistance, but for the purposes of this study was not contacted again. The control subject who was unco-operative, registered her unwillingness to participate by not keeping appointments and disappearing on the days arranged for the interviews. The experimenter or interviewer effect may have been a factor in these cases, however levels of co-operation and rapport were generally high and problems of this nature were rare.

There were seven subjects (four experimentals and three controls) who were excluded by the researcher during the course of the study. Originally

it was envisaged that it would be difficult to obtain new-born babies in the neonatal period for the study. Therefore every albino baby under nine months of age, who was reported to the writer from whatever source was incorporated into the programme. After a few months however, when the hospital staff began to co-operate actively and the new-born babies were reported soon after birth, the writer decided to drop those mothers and babies who had been reported only after the baby was one month of age. The exclusion of these subjects left a more homogeneous group who had all had their initial interview within a month of the birth. Three experimental and three control subjects were excluded for this reason. One further albino subject was initially accepted into the study, although she lived about 100 kilometres away, in Quaggasfontein. She was subsequently dropped when it was found that there would be sufficient subjects and it was not feasible to travel this distance to interview her.

A second group of subjects who were dropped by the researcher consisted of four control subjects. These subjects were dropped in the initial stages of the research when their matched experimental subject left for the rural areas. At a later stage in the study, when it was observed that the numbers in the control group were falling below those in the experimental group, control subjects were retained when their matched pair subject dropped out. These control subjects were rematched with experimental subjects who had babies of the same sex and whose controls had dropped out in six cases. In this way the maximum number of pairs were retained to the end of the project.

Many mothers returned to work during the fifteen months of the study. The majority were, nevertheless, amenable to continuing with the study and left information at their homes concerning their employer. In these cases the employer was contacted and permission obtained for the subject to stay at home for the morning of the interview, without forfeiting pay or leave. There were five cases, however, in the control group where the mothers refused to leave information about their employers, or where a day off was arranged but the mother failed to keep the appointment and could not be traced, these subjects also had to be dropped.

Towards the end of the study two albino babies died, one because of gastroenteritis caused partly by neglect (which had been apparent during the home visits) and the other from diarrhoea, vomiting and massive septicaemia, caused by an enema given to the child by a baby-minder. Also at this time one experimental subject, who had changed her address many times, could not

be traced, and a control subject was arrested and given a prison sentence for shop-lifting.

The total drop-out rate was nineteen, or 51.3 per cent of the experimental group, and twenty-three, or 59.4 per cent of the control group, which, although high, was not unexpected over fifteen months in a community as unstable as that found in Soweto and particularly in the group of mothers (many of whom were single) with new-born babies represented in the study.

3.3.11 Selection of statistical tests

Since the researcher preferred not to make the assumptions required for the use of parametric statistical tests and since in most cases measurements were represented not by scores, but by signs (i.e. it could be determined which member of any pair of subjects was `greater than' or `less than' but not how much greater or less, or which member had or did not have the attribute under investigation) non-parametric statistics were selected for use in the analysis of the data.

The pairing method was used for the selection of controls, since the researcher wanted to have controls who were similar to the subjects with respect to some extraneous variables, such as sex and age of the baby, which might influence the outcome of the research. The two groups therefore were matched and were considered as related rather than independent samples.

The non-parametric statistical tests for use with related samples were investigated, and two tests were selected. The McNemar test for the significance of changes (Siegel, 1956, p63) was chosen for comparison of the differences between the groups at the different stages of the study. The McNemar test was found to be very suitable for the data, which were in frequencies (collected from responses to generally dichotomous questions), which could only be classified in terms of separate categories having no relation to each other, so that measurement was merely nominal (or classificatory). The Binomial test (Siegel, 1956, p36) was used where the number of expected frequencies was very small, particularly at the last interview when the number of subject pairs was only fifteen, and where, within the group, each mother's performance was compared with her performance at a previous stage (i.e. the `before and after' design was used and each person acted as her own control). The Binomial test was found to be suitable since it is also used with data measured in a nominal or ordinal scale.

For assessing the changes within and between the groups over time various more sophisticated methods of analysis, including Friedman's two way

analysis of variance (Siegel, 1956, p166) and Castellan's (1965) partitioning of contingency tables, were considered. The data from the study were found to be unsuitable for analysis using Friedman's test since scoring and ranking is required. Castellan's test was found to be specifically for independent samples, whereas the groups in the present study were matched. No suitable and convenient test could be found for related samples, so the groups were compared with each other at each stage using McNemar's and the Binomial test (when the expected frequencies were less than five) and, separately, the subjects within each group were compared at the beginning and end of the study, using the same two tests.

The direction of the predicted difference between the experimental and control groups was not specified so two-tailed tests were used throughout the study. Prior to the analysis of the data the levels of significance were determined. According to Siegel (1956, p9) the level at which the researcher chooses to set the level of significance `should be determined by his estimate of the importance or possible practical significance of his findings'. Since no vital decisions regarding, for example, therapeutic treatment, were to be made on the basis of the results of the research, the differences between the groups were accepted as significant if the p value was found to be less than 0.05. In consideration of the results the reservation (discussed by Wilkinson, 1951), that since many tests were being carried out there would be a chance that 5 per cent would show differences anyway by definition of the significance level, was borne in mind. In reporting the results the actual probability level associated with each finding is indicated, where the p value is less than 0.05.

3.3.12 Summary

In order to study the impact of the birth of an albino baby on a black family a programme of research was designed. A feasibility study was undertaken and the results indicated that sufficient albino babies should be born to black mothers in accessible areas over a limited time span to make the project worthwhile, and to give a large enough sample so that statistical methods of analysis could be applied.

Possible methods in initiating contact with mothers of new-born albino babies were explored. Hospitals and clinics, where deliveries were performed within the area of greater Johannesburg, were contacted and lines of communication established so that when an albino baby was born it was reported al-

most immediately. A control mother with a normally pigmented new-born baby was selected to match each albino baby for sex and date of birth, so that the subjects were paired.

A black research assistant, proficient in the languages of the subjects and familiar with their culture, was selected. By means of a pilot study, the assistant was trained in the methods of interviewing and observation to be used for data collection.

Three schedules were constructed to include items relevant to the aims of the study. The items selected were based primarily on items used by other workers in similar studies on different genetic disorders. The first schedule was used at the first interview, conducted when the baby was approximately two weeks of age; the second schedule (which was very similar to the first but which omitted the family details) was used at the second interview with the mother when the baby was three months of age; the third schedule (which again was basically similar but omitted items found to give repetitive responses and included a few new items found to be of relevance) was used for the third to the sixth interview, at six, nine, twelve and fifteen months respectively. The first schedule was tested in a pilot study for length and complex or irrelevant questions, and was revised according to the findings. A very similar schedule was completed with the available fathers when the baby was about three months of age.

A list of maternal attachment behaviours frequently found in maternal-infant interaction was compiled. This list was used for recording the selected behaviours during observation sessions on the mother and infant, while the baby's napkin was being changed. There were four of these observation sessions which took place at the same time as interviews 1 to 4 i.e. when the baby was two weeks, three, six and nine months of age. One observation session was also held with the fathers, using a similar list of behaviours to that used with the mothers, while the father held his baby for assessment of its characteristics.

A second list of behaviours was drawn up for use with the mother and baby in a set play situation when the baby was fifteen months of age. The aim of this exercise was to assess maternal-infant interaction in mothers with albino infants at the end of the project by comparing the findings in the two groups. The lists of behaviours for counting during the napkin changing and play activity were assessed during the pilot studies and revised where necessary.

Lastly, an anxiety scale, which had been used successfully with a South African black group previously, was given to the subjects after the play observation.

The interviews and observations were conducted over a three and a half year period. The sample number was thirty-seven pairs of subjects at the first interview and, because of the nearly 60 per cent drop-out rate, only fifteen pairs at the last interview. The mothers in the two groups were very similar, many being young (under twenty-four years of age) and unmarried, with a primary school education and a first baby. The control mothers were, however, slightly more urbanized, more educated, younger and less likely to be living with the fathers of their babies than the experimental mothers (but these differences did not reach levels of statistical significance).

The data collected at the interviews, observation sessions, and from the anxiety scale, were analysed and statistical tests for related samples were applied. Results were based on the statistically significant differences found between the experimental and control group at the six different stages of research. From these findings an attempt was made to assess the impact of the birth of an albino baby on the black mother and, more superficially, on the father and siblings.

3.4 Summary and Conclusion

For the purposes of obtaining reliable and relevant information in the areas under investigation for the present study, materials and methods were assessed and selected and a programme of field-work was planned.

The major setting of the study was Soweto with its large black population representative of most of the ethnic groups living in South Africa. Secondary settings selected were from rural areas in Botswana, Kwazulu, Transkei and Swaziland, the black urban suburbs in the greater Johannesburg metropolitan region and Pretoria. Subjects for the genetic and the psychosocial studies were obtained from these areas. The subjects for the latter studies lived in Soweto, the Johannesburg metropolitan region and Pretoria, since they had to be accessible for the longitudinal study, while subjects for the genetic studies were ascertained in all the areas mentioned.

Normally pigmented matched control subjects were required for the skin reflectance, fertility and the maternal-infant study. These subjects were selected in Swaziland (since Swazi mothers formed the major subject group for the skin colour studies and ethnic group matching was important), and in Soweto for the fertility and maternal-infant studies.

The data was collected by means of interviews using constructed schedules for the genetic and psychosocial studies and by means of hair-bulb incubation tests, physical examination, investigation of patient records and skin refectance measurements, where relevant. Further information for the maternal-infant and paternal-infant study was obtained from observations on the dyads once in the case of fathers and five times over fifteen months in the case of the mothers. The interviews and observations were carried out, sometimes in the presence of the writer, by a black research assistant who was specially trained during the pilot study and who worked closely with the writer. The hair-bulb and skin reflectance tests were performed by the writer in most cases and subjects were examined by two medical practitioners.

The data were analysed by the writer and suitable statistical tests were applied, where the determination of the nature of the differences between groups was required, and the results are presented in the following chapter.

CHAPTER 4

RESULTS

4.1 Introduction

The results of the analyses of the data collected by means of the methods specified in the previous chapter will be presented in consecutive major sections on the genetic and psychosocial parts of the study below. A summary of findings for each part of the study will appear at the end of each major section and the results for the whole study will be summarized briefly at the end of the chapter.

4.2 Genetic studies

This section is divided into six sub-sections, reporting findings from the study on the prevalence rate of albinism in the selected urban and rural samples, the identification and description of types, by means of hair-bulb incubation tests and physical examination, the cancer risks, the life expectancy, and the skin colour and fertility of both homozygotes and heterozygotes.

4.2.1 Prevalence rates

The results from the urban prevalence study will be presented first, followed by those from the rural prevalence studies.

4.2.1.1 Urban prevalence rates

Altogether 213 albino subjects were reported in 126 families. Of this group 206 were personally seen, their families were interviewed by the writer and pedigrees constructed. The remaining seven were unavailable being in the rural areas or absent from home at the time of the interview. Five different aspects from the data on these families are selected for attention here: the the prevalence rates for the total and by ethnic group, the consanguinity rates, the mode of inheritance, and the sex ratio.

(i) General prevalence rate

Using the total figure of 206 affected individuals found in Soweto and the population census figure for Johannesburg blacks (most of whom lived in Soweto) of 803 511 (National Census, 1970) a prevalence rate was calculated and the gene frequency and carrier rate were derived using the Hardy-Weinberg formula. The results of these calculations appear in Table 4.1.

TABLE 4.1 Prevalence of albinism in Soweto

Number of albinos	206
Total population	803 511
Prevalence	1 in 3 900
Gene frequency	0.0160
Carrier rate	1 in 32

The albino subjects were collected during the decade 1970 - 1980 so the census figure for 1970 was used for comparison. This figure is probably too low, both because the population was increasing throughout the decade and because, during a local census, some of the black inhabitants of Soweto avoid being counted since they are illegal immigrants (a work permit and housing permit are required for one to become a legal inhabitant of Soweto). Underenumeration is probably in the region of 17 per cent (Hellman, 1973). However the ascertainment of albinos is no doubt incomplete, and the number of affected individuals is therefore a minimum number, so that if this minimum is compared to the minimum total population the estimated prevalence rate may approach the actual rate.

(ii) Prevalence rate by ethnic group

In order to determine if some ethnic groups showed higher rates than others the total number for each black ethnic group living in Johannesburg was obtained from the census figures. The subjects were then classified according to their ethnic group affiliation and the number in each group was compared to the total for that group in the general Johannesburg black population. The results are shown in Table 4.2.

As may be seen from the Table the highest rate found was that of 1 in 2 254 for the Southern Sotho and the lowest that of 1 in 28 614 for the Shangaan people. The Swazi rate of 1 in 2 716 was also high, but the group of Swazis in Johannesburg only consisted of 29 872 people which might be considered too small to give reliable results. The largest Nguni groups, the Zulu and Xhosa, who historically stem from the same original clan, had similar low rates, and the largest Sotho groups, the Tswana and Southern Sotho, who also had common ancestors, had similar high rates. The low rates in the Shangaan and Pedi groups might reflect the true situation, but might be caused by poor ascertainment, or reluctance on the part of affected individuals in these groups to come forward to participate in research, or they may not migrate to the cities. These two groups together with the Venda, probably represent the least sophisticated of all the South African peoples.

TABLE 4.2 Prevalence of albinism in Soweto by ethnic group

Ethnic group	No of albinos	Johannesburg population	Prevalence	Gene frequency
Zulu	55	245 258	1/4 459	0.0149
Xhosa	17	81 511	1/4 794	0.0144
Swazi	11	29 872	1/2 716	0.0192
Ndebele	3	13 593	1/4 531	0.0148
Tswana	42	146 184	1/3 481	0.0169
S Sotho	53	108 190	1/2 041	0.0211
Pedi	8	77 604	1/9 700	0.0101
Venda	6	31 936	1/5 323	0.0137
Shangaan	2	57 228	1/28 614	0.0059
Other	0	12 135		
No information	9			
TOTAL	206	803 511	1/3 900	0.0160

* after Kromberg and Jenkins, 1982

Using the chi-square test significant differences were found between the rates for the Zulu and Southern Sotho ($p<.01$), and between the Xhosa and Southern Sotho groups ($p<.01$).

(iii) Consanguinity rates

Reasons for the finding of the high prevalence rates were sought and since a higher rate of consanguinity than that occurring in the general population might provide one possible alternative, the information on consanguineous marriages, provided in the interview by the subjects, was analysed. The rates of such marriages for the different ethnic groups appear in Table 4.3.

TABLE 4.3 Rates of consanguineous marriages among parents of albino children, according to ethnic group

		Cousins				
Ethnic group	No of families	1st	2nd	Distantly related	Total	Consanguinity rate (%)
Zulu	23	1			1	4.3
Xhosa	10			2	2	20.0
Swazi	6	1		1	2	33.3
Tswana	24	6		4	10	41.7
S Sotho	22	1	1	4	6	27.3
Pedi	2				0	-
Venda	2		1		1	-
Shangaan	1				0	-
TOTAL	90	9	2	11	22	24.4

Information was available for only ninety of the total of 126 families in the Soweto sample, since some albinos did not know one or both of their parents well, nor whether there was a blood relationship between them.

The consanguinity rate for the total sample of families with an albino child was 24.4 per cent. The consanguinity rate for each ethnic group was compared with the albinism prevalence rate for that group. The group with the lowest consanguinity rate (the Zulus with the low rate of 4.3 per cent), was found to have a low prevalence rate, and those groups with high consanguinity rates (e.g. Tswana, Sotho, and Swazi), also had high albinism rates.

(iv) Mode of inheritance

To test whether the data collected from the sample of affected families fitted with the recessive pattern of inheritance for albinism, an analysis was carried out using Hogben's method as described by Emery (1976). The total number of sibships was 126 containing 213 affected individuals (including those seen personally and those reported) and 467 normally pigmented siblings. The data for these sibships together with the expected numbers of affected individuals and the variances are shown in Table 4.4

TABLE 4.4 Observed and expected numbers of albinos according to sibship size in 126 Soweto families*

Size of sibships	No of sibships	Total No individuals	No of affected individuals	
			Obs	Exp. (variance)**
1	11	11	11	11.0 (0)
2	12	24	13	13.7 (1.5)
3	17	51	22	22.1 (4.5)
4	8	32	10	11.7 (3.4)
5	18	90	19	29.5 (9.0)
6	18	108	33	32.9 (13.0)
7	16	112	34	32.3 (15.5)
8	8	64	20	17.8 (9.4)
9	7	63	20	17.0 (9.7)
10	3	30	7	7.9 (4.8)
11	3	33	8	8.6 (5.4)
12	3	36	8	9.3 (6.1)
13	2	26	8	6.7 (4.5)
	126	680	213	220.5 (87.6)

* after Kromberg and Jenkins, 1982

** Expected numbers calculated assuming complete ascertainment.

Obs = observed; Exp = expected.

Close agreement was found between the observed and expected numbers of affected individuals and the data are therefore, compatible with the autosomal recessive hypothesis.

Using the `singles' method of Li and Mantel (1976) of testing for recessive inheritance when there is complete ascertainment, the data were shown to be equally compatible with the recessive hypothesis. The number of singles was seventy-seven and the proportion of affected individuals in the 126 sibships was 0.23 with a standard error of 0.02. This method is, in the opinion of its originators, just as reliable as more involved methods of assessing mode of inheritance.

(v) Sex ratio

Lastly, in order to determine whether albinism was more common in males than in females the sex ratio was calculated. Although this ratio was 1.21 and more males (113) than females (ninety-three) were observed in the sample, the difference was not significant ($p>.10$).

In summary, then, the findings on urban prevalence showed a rate of albinism of 1 in 3 900 per head of the population, with high rates generally in the Sotho ethnic groups and low rates in the Nguni groups. The consanguinity rates were also found to be generally higher in the Sotho groups and lower in the Nguni groups. The exception was the Swazi group who are of Nguni descent, but have a high albinism and a high consanguineous marriage rate. An analysis of the expected and observed numbers of affected individuals, as opposed to the numbers of their normal siblings, showed that the data were compatible with the hypothesis that albinism is recessively inherited. As was also expected there was no significant difference between the numbers of affected males and females in the sample.

4.2.1.2 Prevalence in rural populations

The data collected during the four rural studies on selected areas of Botswana, Kwazulu, Transkei and Swaziland are presented below. The prevalence rates were calculated and compared with the findings from the urban study. The rare types of albinism encountered in each area are briefly described.

(i) Botswana study

The study of the population of 12 000 Tswana people living in the village of Mochudi produced fourteen albino subjects. Of this group ten albinos were seen personally and interviewed at their homes by the writer and a further four were not seen but were included, since they were reported by seemingly

reliable witnesses (an anthropologist, his trained assistant and a medical practitioner), who had had personal experience of albinism. The results of the Hardy-Weinberg calculations for the gene frequency and heterozygote frequency using these figures appear in Table 4.5.

TABLE 4.5 Prevalence of albinism in Mochudi, Botswana

Number of albinos	:	14
Number of families	:	9
Total population	:	18 300
Prevalence	:	1 in 1 307
Gene frequency	:	0.0537
Carrier rate	:	1 in 19

The prevalence rate of 1 in 1 307 per head of the population was approximately three times greater than the urban rate found for the Tswana group living in Soweto (1:3481, see Table 4.2). The consanguinity rate was 11 per cent, i.e. one of the nine couples with an albino child had a consanguineous marriage, which because of the small sample size perhaps does not reflect the true rate. In comparison the urban Tswana group had a consanguineous marriage rate of 41 per cent.

A family history of albinism was reported in five out of the six famifamilies for which information was available, and one family had multiple cases of albinos in the extended family.

In the course of this study one interesting child was observed in the street and subsequently his family was visited at home. This four year old child had light reddish yellow healthy looking skin, very pale yellow hair and light brown eyes (see Fig 4.1). His mother and three of his eight siblings were seen and were normally pigmented. A further three siblings were not seen but were said to be dark skinned. However two of his eight siblings, both older boys, were said to have the same skin, hair and eye colour as the proband. Whether this child represents a brown albino (as described by King et al, 1980, in Nigeria), whether he is a tanned yellow mutant, or whether he could be called a `red skin´ similar to those of Papua New Guinea (Walsh, 1971 and Hornabrook et al, 1980), is debatable. From the colour photographs which appear in Walsh´s (1971) paper, the writer would favour classifying this subject in the latter category. Whatever the type it appears to be recessive in this family, since three of the nine children of normal parents were said to be affected with this unusual pigmentation.



Fig 4.1 `Red-skinned' child and his friends in Mochudi, Botswana

(ii) Kwazulu study

The study of albinism in the Nongoma district of Kwazulu resulted in the identification of fifteen albinos from eleven families in the population of approximately 90 000 people (unpublished data from the Nongoma Magistrates' Court files assembled by Klopper, 1979, personal communication). Of this group of albinos thirteen were seen and interviewed either alone or together with one or more relatives, and two were unavailable or living too far away to come into the town of Nongoma. The interviews were all carried out at the hospital in Nongoma, which was a well-known landmark in the area, or at the schools visited, since the homes and villages were frequently inaccessible by car. The results of the prevalence rate calculations for the district as a whole and for the school population are presented in Table 4.6.

TABLE 4.6 Prevalence of albinism in Nongoma district and in Nongoma school children, Kwazulu

	Nongoma district	Schools
Number of albinos	15	7
Number of families	11	7
Total population	90 000	18 036*
Prevalence	1 in 6 000	1 in 2 576
Gene frequency	0.0255	0.0386
Carrier rate	1 in 39	1 in 26

* in 48 schools

Since the people had to walk long distances or catch infrequent buses in order to participate in the survey, and since the 'bush-telegraph' seemed to be slow in working because of the mountain ous nature and remoteness of some areas, the ascertainment in the general population was probably incomplete. A better estimate of the prevalence was provided by the figures for the school children, and their rate was found to be more than twice as high as that for the general Nongoma population.

The consanguinity rate in this sample of eleven affected families was zero. The family details however were obtained from the affected school children themselves, in several cases, and were sometimes very limited. There is a local taboo against marriage within the fathers', mothers', or either grandmothers' clans (according to a local social anthropologist, Clegg 1979, personal communication)so that marriages are generally exogamous. Consanguineous marriages if they occurred might, therefore, not be reported or even admitted.

During the course of this study two unusual subjects were reported. The first was a school-child of about eleven years who was seen by the writer. He had normal black skin and hair and dark eyes, except for a large white patch of skin almost surrounding his right eye and his eyelashes were also white. He reported that the patch had been developing over the years, he therefore probably had vitiligo rather than partial albinism. The second subject was the paternal uncle of an albino. He was said to have partial albinism and half his face was apparently white although his hair was black. He was reported by the informant to have been that way since birth, but was not available for examination.

(iii) The Transkei study

The field-work on albinism in selected districts of the Transkei resulted in the identification and interviewing of forty-three people reported as albinos (i.e. the Xhosa word for albino was used to refer to them). A further fifteen albinos were described by the subjects. On examination it was found that thirty-nine of the forty-three subjects were typical albinos (thirteen were seen at the local school for the blind), one was a partial albino with distinct white patches since birth, and three had light-reddish brown skin and pale yellow hair.

Although clinics drawing patients from eleven of the twenty-eight different magisterial districts in the Transkei were visited the ascertainment of subjects was very poor. Some clinic staff stated that they had not been no-

tified of the survey at all, although they knew of several albinos in their district. As a result the prevalence rates, when calculated for each of the eleven districts were found to be far lower than expected (e.g. the highest rate was 1 in 10 000 in the district with the best ascertainment), and the writer judged them to be unreliable and not worth presenting.

The consanguinity rate for the sample was calculated and, although no subjects admitted that their parents were consanguineous, 6.3 per cent were said to come `from the same family' and the same village, while 15.6 per cent were from the same village. It should be noted here that the Xhosa people have similar taboos against marriage between relatives to those of the Zulus, so information on this matter is difficult to collect reliably.

A partial albino boy, aged about six years, was brought to the writer during the study. He had two distinct ragged white patches, about three centimeters in diameter on his right knee, another measuring about four by two centimeters on his left knee, at least four smaller patches on his face and five small patches of white hair scattered on the crown and back of his head. He had had these patches since birth and had been left, apparently by his mother, at about five days of age outside the local shop and deserted, so that no family history was available. The shop-keeper had taken him in and had recently officially adopted him.

Of the two families reported with members with reddish coloured skin and fair hair, one proband who was examined stated that she had two siblings similarly affected, and the other stated that he had four siblings, but none of them were affected. The parents of the former siblings were reported by the clinic nurse to have dark skin and black hair, so it seems that this unusual type of pigmentation is recessively inherited, as was found to be the case in the one family reported in the Botswana study.

Also during the course of the study two families were described which were said to contain both albinos and red-skinned individuals. However it was not possible to substantiate these reports as the albino members were seen but the red-skinned relatives were not available. A marriage between two albinos was also reported and the affected father was seen together with two of five affected children. The albino mother had died of tuberculosis some years previously and the husband reported that she had had one normal child who had died neonatally. It seems that if both he and his wife had the same type of albinism as evidenced by the affected children, that the normal child must have been conceived extramaritally.

(iv) The Swaziland study

During the study of albinism in the Hhohho district of Swaziland forty-nine albinos from thirty-six families were personally seen and interviewed by the writer. In these families a further twenty-eight affected relatives who were living in the district were reported. Apart from these seventy-seven albinos, one teenage girl and one man of about thirty years were observed in a town street, and three further albinos (an old woman, and two young children) were reported by reliable witnesses (a nursing sister, a local chief and a local historian). These five albinos could be identified as not having been included in the initial group. So the final total for the albinos observed and reported during the study was eighty-two. The prevalence rate, calculated from this figure together with the total Hhohho district population figure, is presented in Table 4.7.

TABLE 4.7 Prevalence of albinism in Hhohho district, Swaziland

Number of albinos	:	82
Number of families	:	36
Total population	:	160 000
Prevalence	:	1 in 1 951
Gene frequency	:	0.0442
Carrier rate	:	1 in 23

The prevalence rate found in this rural study of the Swazis was high and verified the rate of 1 in 2 716 found in the small sample of Swazis (only 29 872) studied in urban Soweto during the present study.

The consanguinity rate for the thirty-six rural families was calculated from the information given at the interview and was found to be 17 per cent. In the course of the study skin reflectance measurements were carried out on parents of albinos and on a control group. Altogether 100 normally pigmented controls were involved, a pedigree was drawn up for each one and information was requested on consanguineous marriages. In this group of controls 6 per cent reported such marriages in their parents, so that the rate for the albino families (of 17 per cent) was nearly three times higher than that for these controls. A certain amount of underreporting of consanguineous marriages may have occurred, since in the Swazis, who are of Nguni descent (as are the Zulus and Xhosas), such marriages are not strictly permitted. These

motives for underreporting however, would have presumably affected the controls and albino family informants in the same way.

In addition to the thirty-six affected families three other families were brought to the writer's attention during the survey of the district. Of these families two had members affected with what seemed to be vitiligo, and the other had a very fair child, who, on examination, appeared to be a healthy normal 'coloured' child (of mixed ancestry) and on history apparently had a white grandfather.

Albinism is so common in Swaziland that 58 per cent of the affected families in the sample reported that they had a family history. A further 23 per cent of the group denied any knowledge of family members with albinism, and in 19 per cent of the families the informant could not give the required information. A history of albinism was also reported in 4 per cent of the controls for the skin reflectance study (the subjects with this history were considered unsuitable and were later dropped from the control group).

The 'red-skinned' children examined during this study had the characteristic light copper-coloured skin with fair gold hair seen in Botswana and the Transkei. Of the five children examined in two families only one child had mild nystagmus, none had obvious photophobia, two had brown eyes, one light brown eyes and the other two (including a six year old male child) had grey-green eyes. They were difficult to classify, since nystagmus appeared to be a rare feature and the hair was fair, so that they did not seem to fit into the category of 'brown albinos' described by King et al (1980), 96 per cent of whom had nystagmus, or that of xanthous individuals who had 'red' skin and 'brown' hair (Pearson et al, 1911), or that of rufous albinos with red hair (Witkop et al, 1983). They appeared, however, to be similar to the 'reds' described in Papua New Guinea (Walsh, 1971).

4.2.2 Identification and description of types

In order to classify albinos and identify their types at least the hair-bulb incubation test and a physical examination of the subject must be performed. To clarify the status of border-line subjects further studies such as tyrosinase assays, incubation of hair-bulbs in various substrates and electron-microscopy examination of melanosomes should be undertaken. Only the hair-bulb incubation test and phenotype examination were carried out in the present study.

4.2.2.1 Hair-bulb incubation tests

Hair-bulb tests using the methods described in the previous chapter were performed on all subjects in Soweto who agreed to have hairs collected and on whom it was feasible to do the test. In addition two schools for the blind (one near Pretoria and one in the Transkei) and one cancer clinic were visited and the co-operative albinos were tested. Where pigment was found in the hair-bulb after incubation (see Fig 4.2) a ty-pos result was recorded.

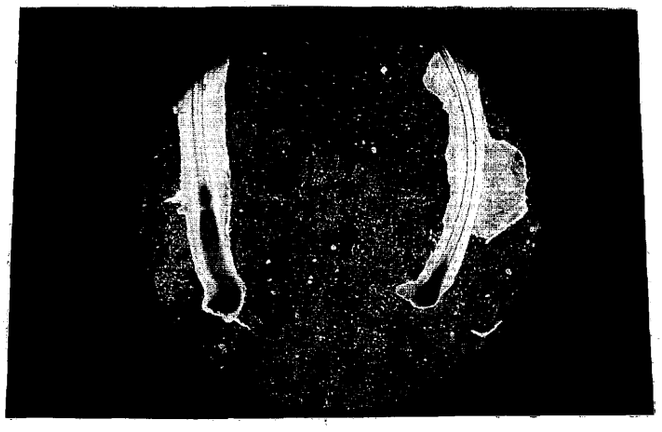

Fig 4.2 Hair-bulbs from a tyrosinase-positive subject after incubation in tyrosine (left) and buffer (right) solutions, showing dark pigment in hair-bulb on left.

The findings from the hair-bulb tests are presented in Table 4.8.

TABLE 4.8 Results of hair-bulb tests in three groups of albino subjects

Source	Total Number*	Tyrosinase-neg No*	Tyrosinase-neg %	Tyrosinase-pos No*	Tyrosinase-pos %
1. Soweto	79(52)	25(17)	31.6	54(35)	68.4
2. Blind schools	25(24)	20(19)	80.0	5(5)	20.0
3. Cancer clinics	8(8)	6(6)	75.0	2(2)	25.0
TOTAL	112(84)	51(42)		61(42)	

* Numbers represent individuals (families)

Those individuals found to have a tyrosinase-negative test result will be discussed below, since examination of the phenotype of some of the Soweto subjects showed that they did not have the typical characteristics of ty-neg albinism.

The figures in Table 4.8 suggest that in the Soweto sample, which consisted of albinos living in the community as normal individuals, the general trend showed an excess of ty-pos over ty-neg test results. The proportion of those with ty-neg compared to ty-pos hair-bulb test results was approximate-

ly 1:2. The ratio of ty-neg to ty-pos tests in subjects at the schools for the blind however, was reversed i.e. 4:1, and in cancer clinic subjects the ratio was 3:1. It is possible therefore that subjects found in a general community survey are more likely to have ty-pos albinism. Those with ty-neg albinism, who have the severe side-effects of very poor eyesight and skin highly susceptible to skin cancer, are more likely to be found in institutions, such as schools or workshops for the blind, or in hospitals, for the treatment of their skin cancers.

The type of albinism in any one subject cannot, however, be determined by the hair-bulb test alone, but the physical characteristics have to be taken into consideration and these will be discussed in the following section.

4.2.2.2 Classification by hair-bulb tests and phenotypes

An attempt was made to classify those Soweto subjects who had had both hair-bulb incubation tests and physical examination, and this group numbered seventy-nine individuals altogether. In addition, those individuals who, on physical examination only, appeared to be brown or rufous albinos, were included in the classification and there were seventeen in this group.

With data available from the hair-bulb test and the physical examination only, there was difficulty in classifying some albinos into the five classes described in Chapter 3 Section 3.2.3.3 of the present study. Several individuals appeared to be border-line between two categories showing some features of both. Nevertheless, the results of a preliminary attempt at classification are presented in Table 4.9. This should be accepted as a tentative

TABLE 4.9 Types of albinism found in Soweto

Type	Families	Individuals		
	Number	Number		%
Tyrosinase-negative	7	8		8.3
Tyrosinase-positive A	40	61)	66	68.8
Tyrosinase-positive B	3	5)		
Yellow mutant	3	5		5.2
Brown albinism	8	11		11.5
Rufous albinism	5	6		6.2
TOTAL	66	96		100

classification only, since additional studies such as those on visual acuity,

on transillumination of the iris, on the stages and types of the melanosomes in the hair-bulbs, and actual measurement of the tyrosinase in the incubated hair-bulb, would give further information which might clarify the status of some subjects and indicate that they should be reclassified.

Wherever possible hair-bulb tests were repeated to confirm the findings of the first test. There were however seven subjects who had tyrosinase negative hair-bulb results on a single test, but on examination they were found to have pigmented patches of skin in the form of ephelides. Since it was not feasible to repeat the tests on these subjects they were classified in the tyrosinase-positive group, because of their ability to make pigment.

One family who was difficult to classify had three albino children, two males and a female aged ten, fourteen and twelve years, respectively, at the time of the first examination in 1978. Hair-bulb tests were carried out three times on two of the children and twice on the oldest child. The first two tests on all three children were performed in 1978 and the third test on the two younger children in 1984. The first test showed a negative result on the younger boy and the girl and an equivocal result on the older boy. The second test showed an equivocal result on the older and younger boy and a negative result on the girl. The third test showed four positive hair-bulbs in the girl (then aged eighteen years) and two in those of the younger boy (aged sixteen years). All three children had hazel eyes and the two boys had dark yellow hair (which was observed as distinct in the younger boy when he was first seen aged two years), while the girl had lighter coloured hair. The younger boy had been seen with very badly sunburnt skin in the summer. From their skin colour the children did not appear to be brown or rufous albinos. They were, therefore, classified as yellow mutant, since the distinct yellow colour of the hair had been observed in the youngest child at two years of age. It is, however, possible that they had ty-pos albinism, but that there were either insufficient complete hair-bulbs in the first two tests to show the expected dramatic increase in pigmentation seen in the hair-bulbs of other subjects with ty-pos albinism, or that they were too young (two of them being twelve years or younger). Many of their hair-bulbs appeared to be broken and in two of the eight tests completed only four adequate hair-bulbs were present. Only one of these children, the girl, had a mild strabismus, she also had only mild nystagmus, while the boys had moderate nystagmus, and none of them had ephelides. The older boy had a marked scoliosis and the father had developed Vogt-Koyanagi-Harada Syndrome (See

Fig 4.3) resulting in bilateral hypopigmentation around the eyes, after an eye operation (for cataracts). The parents were both of the Xhosa population group and said they were distantly related, but the exact nature of this relationship could not be established. There was one relative who was an albino and the paternal grandfather's sister was said to have `ginger' hair and eyes, hypopigmented patches and normal skin colour. There were eleven children altogether and the last three, born when the father was in his sixties, had albinism. A hair-bulb test with tyrosine-cysteine incubation might clarify whether or not these subjects should have been classified in the YM group, since hair-bulbs from YM subjects show increased red or yellow pigment with this test (Witkop, 1983). If they are not YM albinos the other alternatives are: that these children have ty-pos albinism, but the hair-bulbs were not adequate in the first two tests; that they have a form of BA with a much lighter coloured skin which is much more susceptible to sunburn than the other BA subjects in this study; or that the successful collection of complete hair-bulbs and/or the expression of pigmentation in the hair-bulb after incubation in tyrosine, is dependent on age in some cases, or on nutrition (since the pattern and amount of hair growth is evidently affected by malnutrition (Ebling, 1981), and this family was very poor and probably malnourished).

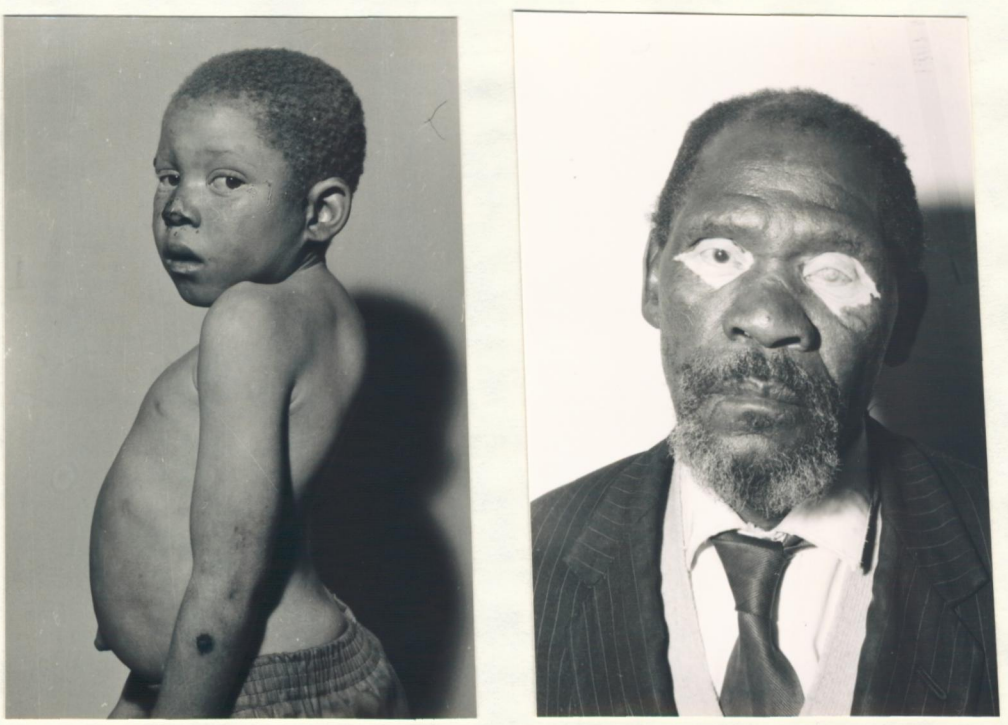

Fig 4.3 Albino boy with scoliosis and his father with Vogt-Koyanayi-Harada Syndrome

A second problem sibship consisted of two brothers who were assigned to

the ty-neg group. They both had negative hair-bulb tests (two tests were done on each), and the older brother, aged thirty-one years at the time of examination, had yellowish-white hair, pale blue grey eyes, and no ephelides. The younger brother, aged fifteen years at the time, however had blue eyes with a few light brown rays around the pupil and no ephelides. These sibs were classified as ty-neg albinos but might, on further investigation, prove to have ty-pos B.

The classification as it appears in Table 4.9 was therefore tentative and preliminary, giving only a general estimate of the types of albinos, according to hair-bulb tests and physical description, found in Soweto. The majority of albinos appeared to have ty-pos albinism, a few had brown albinism and the number of albinos with ty-neg, yellow mutant or rufous albinism was comparatively small. These groups were comprised of those subjects who agreed to participate in all the tests, so that the sample does not represent a random sample of the albinos in Soweto.

4.2.2.3 Physical characteristics

The physical characteristics of the ninety-six albinos who were classified (see Table 4.9) are presented in Table 4.10 and 4.11. Those classified as ty-neg, ty-pos (A and B) and ym albinos all had hair-bulb tests, whereas those in the brown and rufous group were generally classified according to their physical characteristics only. One brown albino subject had a hair-bulb test and the result was a dramatic increase in pigmentation in the hair-bulbs after incubation.

The findings regarding the pigment noted in the hair and skin of the subjects are presented in Table 4.10. In general the ty-neg albinos were found to have very pale yellow coloured hair, although none of them could be said to have white hair. In one sibling pair in this group, one brother (the older of the two, aged thirty-eight years) had very pale yellowish white hair while his younger brother (aged twenty-two) had hair with a distinct yellow tinge. The accumulation of pigment also varied within sibships in the ty-pos A group. Hair colour appeared to darken with age in two of nine sibships, in a further five sibships the sibs of different ages had more or less the same colour hair, and in the remaining two the younger sibs had darker hair than their older sibs.

In the group of brown albinos the majority of subjects (73 per cent) had fair or light yellow-brown hair colour. A similar hair colouring was found in the rufous group, where in five subjects examined four had fair hair and

only one had light reddish-brown hair. A few albinos had dyed their hair black and it was difficult to assess the original hair colour in these subjects.

TABLE 4.10 Hair colour and skin pigment in 96 albinos classified according to type of albinism

	Tyrosinase-neg	Tyrosinase-pos A	Tyrosinase-pos B	Yellow Mutants	Brown Albinos	Rufous Albinos
	No(%)	No(%)	No(%)	No(%)	No(%)	No(%)
Hair colour						
White	0					
Light yellow	6(75)	17(28)	4(80)	1(20)		
Yellow	2(25)	32(52)				
Dark yellow gold		9(15)	1(20)	4(80)		
Light yellow brown		2(3)			8(73)	4(66)
Red brown						1(17)
Brown					3(27)	
No information		1(2)				1(17)
TOTAL	8(100)	61(100)	5(100)	5(100)	11(100)	6(100)
Skin pigment						
Ephelides*						
Present		32(52)			7(64)	
Absent	8(100)	25(41)	3(60)	5(100)	2(18)	3(50)
No information		4(7)	2(40)		2(18)	3(50)
TOTAL	8(100)	61(100)	5(100)	5(100)	11(100)	6(100)

* includes freckles and lentigenes

Skin pigment in the form of ephelides was not apparent in any ty-neg albinos (since this characteristic was a criterion for inclusion in the group) and was only found in half (52 per cent) of the ty-pos A group, and 64 per cent of the BA group. The presence of ephelides varied within sibships but was generally age dependent. In nine sibships in the ty-pos A group there were three where all sibs had ephelides, one where none had ephelides, four where ephelides were present in older sibs and absent in younger sibs, and one sibship in which the younger sib had ephelides, but the older sib had none. There was one ty-pos subject in whom the ephelides took the unusual

form of small freckles ($\pm$ 3 mm in diameter) which were almost confluent over the face and other exposed parts of the body. The few RA albinos (three) examined for this characteristic did not appear to have any ephelides, although the three subjects were aged three, eight and fifteen years at the time they were investigated. In most cases where ephelides occurred they were about a half to one centimetre in diameter, dark brown in colour and with a ragged edge. Several subjects disliked the appearance of these pigmented patches and requested methods of preventing their development.

The colour of the irides of the albinos and the presence of nystagmus and strabismus were also noted wherever possible during the medical examination. The findings are reported in Table 4.11.

TABLE 4.11 Ocular findings in albinos

	Albino type					
	Tyrosi-nase-neg	Tyrosinase-pos A	Tyrosinase-pos B	Yellow Mutant	Brown Albinos	Rufous Albinos
Characteristic						
	No (%)	No (%)	No (%)	No (%)	No (%)	No (%)
Iris colour						
Blue	1(12)	9(15)		2(40)	2(18)	
Blue with yellow rays	7(88)	38(62)	4(80)			
Hazel		9(15)		3(60)	3(27)	1(17)
Brown					4(37)	3(50)
No information		5(8)	1(20)		2(18)	2(33)
TOTAL	8(100)	61(100)	5(100)	5(100)	11(100)	6(100)
	N/NE* (%)	N/NE (%)	N/NE (%)	N/NE (%)	N/NE (%)	N/NE (%)
Nystagmus	8/8 (100)	56/56 (100)	4/4 (100)	5/5 (100)	5/9 (55)	0/3
Strabismus	4/8 (50)	24/44 (54)	1/2 (50)	1/5 (20)	2/9 (22)	0/3

* Number with characteristic/total number examined (% with characteristic)

There were very few subjects who could be said to have pure blue eyes. The majority of those with blue eyes, had some form of yellow or light brown colour radiating out from around the pupil. Blue eyes were however found in a few subjects in all categories except for the ty-pos B, in which a criterion for inclusion was blue eyes with radiating pigment, and the RA group, in which the numbers of subjects examined was very small. Hazel eyes were

observed in only 15 per cent of the ty-pos group, but were more frequent in the ym, BA and RA groups. In the latter two groups brown eyes, somewhat lighter in colour than those of the general population, were found quite commonly.

Nystagmus was observed in all subjects with the ty-neg, ty-pos and ym types of albinism. In the BA group however, only 55 per cent had nystagmus and none of the rufous albinos examined showed any signs of nystagmus. The severity of the nystagmus varied from a mild type, which only occurred in response to light, to a severe continuous roving nystagmus.

Strabismus was a common finding among the subjects, and it was observed in about half of the ty-neg and ty-pos albinos examined and in about 20 per cent of the ym and BA subjects. The finding of strabismus was not consistent in a family and in several cases one sib had severe strabismus, while the other one or two sibs were not affected at all (See Fig 4.4). In the nine sibships examined in the ty-pos group there was only one where both sibs had strabismus. The presence of this characteristic did not appear to be related to age.



Fig 4.4 Siblings: two without strabismus (on left) and one with strabismus (on right)

In conclusion, from this small and limited attempt at classifying a cooperative sample of the albinos certain trends have been identified: the five types of albinism selected for attention appear to occur in the South African black population; the number of albinos with ty-pos albinism exceeded that for other types of albinism; subjects in schools for the blind and cancer clinics seemed to show a higher proportion of ty-neg hair-bulb results than albinos in the general population; the albinos very seldom had a total absence of pigment, and some form of pigment was generally present in the eyes and hair.

4.2.3 Skin cancer in albinos

The data collected at the physical examination on malignant and premalignant lesions in Soweto albinos were analysed according to the age of the subject. The results of this analysis appear in Table 4.12.

TABLE 4.12 Rate of malignant (or premalignant) lesions in albinos by age

Age group (in years)	Total number	Cancer present	
		Number	%
10 - 19	48	2	4.2
20 - 29	32	7	21.9
30 - 49	13	8	61.5
>50 years	6	6	100.0
TOTAL	99	23	23.2

No lesions were found in the subjects aged nine years or less and the rate of lesions increased in albinos with age; in the over fifty year group all were affected. Lesions were found in albinos of all types (Fig 4.5 shows an affected brown albino) except those with rufous albinism.

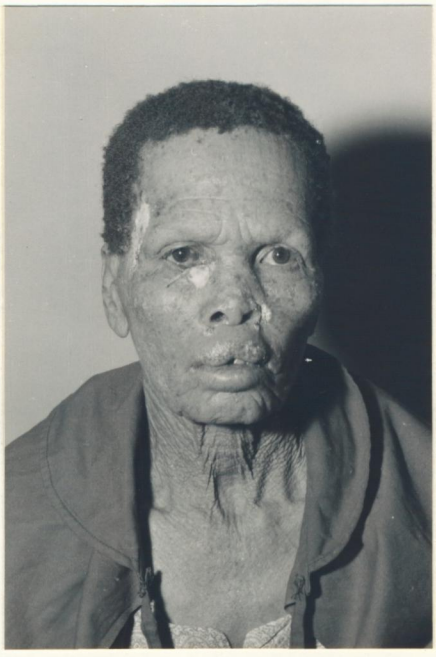

Fig 4.5 Squamous carcinoma in a brown albino

In order to assess whether lesions occurred more commonly in individuals of any particular ethnic group the data which were available for 109 albinos living in Soweto were analysed by ethnic group and age (see Table 4.13).

TABLE 4.13 Albinos in Soweto with malignant or premalignant lesions by ethnic group and age

Ethnic group	With Lesions		Without Lesions		Total	
	No (%)	Mean Age (years)	No (%)	Mean Age (years)	No	Mean Age (years)
Sotho						
Tswana	16(64)	48.1	9(36)	23.9	25	39.4
Pedi	2(50)	33	2(50)	32.5	4	32.7
S Sotho	12(33.3)	34.1	24(66.6)	20.5	36	25.0
Sub-total	30(46.2)	38.4	35(53.8)	25.6	65(100)	32.4
Nguni						
Zulu	4(15)	56.2	22(85)	24.4	26	29.3
Xhosa	1(9.1)	68	10(90.9)	24	11	28
Swazi	4(57.1)	27.2	3(42.9)	22	7	25.1
Sub-total	9(20.5)	50.4	35(79.5)	23.5	44(100)	27.5
TOTAL	39(35.8)	44.4	70(64.2)	24.5	109(100)	29.9

Significantly more of the albinos in the Sotho group were found to have malignant lesions than those in the Nguni group (chi-square 6,46, $p<.01$). The two groups however showed a similar age distribution pattern, with those having skin cancer being older than those without skin cancer (and this age difference was significant for the total group, chi-square 5.80, $p <.01$, and for the Nguni group, chi-square 9.77, $p <.01$). The average age of the subjects in the Sotho group was slightly higher than that for the Nguni group, but the difference was not significant (chi-square 0.40, $p >.50$). Age therefore could not explain the excess of albinos with malignant lesions found in the Sotho compared with the Nguni group in Soweto.

The second method of assessing the nature of the skin cancer in albinos was to examine the histopathology records on the results from all specimens of skin biopsies from black patients showing squamous and basal cell carcinoma. The information obtained from this investigation is shown in Table 4.14. Specimens from albinos were found to represent 12.7 per cent of all specimens. The majority of the specimens from albinos (twelve) had squamous carcinoma and very few (two) had basal cell carcinomas.

The site of the carcinoma biopsied was not specified in all cases. In the two albino patients with basal cell carcinoma the nose was affected in one and the eye-lid and cheek in the other. In the group of patients with

TABLE 4.14 Squamous and basal cell carcinoma in skin biopsies from albinos and the general population (1976 - 1980)

	Albinos		General population		
Type of Carcinoma	Number	%	Number	%	Total
Squamous	12	(12.4)	85	(87.6)	97
Basal cell	2	(15.4)	11	(84.6)	13
TOTAL	14	(12.7)	96	(87.3)	110

squamous cell carcinoma the site was stated in all but one subject, and three subjects had lesions in more than one site, so that a total of seventeen sites were reported. The site found in albinos was compared with the site specified in the other black patients (this information was only available for eighty-five biopsies) and the findings appear in Table 4.15.

TABLE 4.15 Site of biopsied carcinomas reported in albinos and the general black population (1976 to 1980)

	Albinos			General population		
	Total	Number	%	Total	Number	%
Head	14		(82.3)	46		(54.1)
Face (unspecified)		3			4	
Forehead		0			3	
Eye (+ eye-lid)		4			8	
Cheek		4			2	
Nose		1			1	
Mouth (+ lip)		1			6	
Jaw		1			2	
Scalp		0			10	
Neck		0			10	
Torso	0		(0)	21		(24.7)
Breast		0			1	
Glands (lymph, parotid)		0			2	
Genitalia		0			9	
Chest		0			6	
Back		0			3	
Limbs	3		(17.7)	17		(20.0)
Leg		3			13	
Arm		0			4	
Burns	0			1	1	(1.2)
TOTAL	17	17	(100%)	85	85	(100%)

The site of the cancer in albinos differed significantly from that found in normally pigmented subjects (chi-square 6.07, $p < .05$). In the majority of albinos the cancer occurred on the head (82 per cent of subjects), whereas only about half (54 per cent) of the cancer found in the black subjects was on the head. The eye-lids and cheek appeared to be most susceptible in this sample of albinos, whereas the scalp, neck and eye-lid were the commonest sites on the head in non-albinos. There were no albino subjects who had had biopsies for cancer on the torso, whereas almost a quarter of the non-albino subjects had cancer on this part of the body, and in almost half of these the genitalia were involved. Sites on the limbs were specified in 17 and 20 per cent of the albino and black group, respectively, and in the majority of these subjects in both these groups it was the legs which were affected. Specifically, susceptible areas of the legs in the black subjects were the foot, ankle and heel.

The fourteen specimens obtained from albinos were from ten patients, and in one of these patients squamous cell carcinoma was diagnosed in September 1976, at which stage she was described in the records as having `hundreds of hyperkeratoses´, and basal cell carcinoma was diagnosed in May 1977.

The majority (90 per cent) of albinos with diagnosed carcinoma were aged twenty years or older, half were over thirty years of age, and the only two with basal cell carcinoma were in their forties. The results on this small sample agreed with those found on the larger sample of Soweto albinos with malignant and premalignant lesions, the occurrence of which also appeared to increase with age.

The third method of assessing skin cancer in albinos was by examining the records of the albino patients, who attended the cancer clinic for treatment, and obtaining the dermatologists´ diagnosis of the type and its site. Nineteen subjects were investigated in this way and the findings are presented in Tables 4.16 and 4.17. In thirteen subjects only one cancerous site was reported and in six two or more sites were specified.

The findings from the cancer clinic subjects show similar trends to those obtained from the Histopathology reports on skin biopsies in albinos. In the majority of clinic patients the cancer was found on the head and diagnosed as squamous cell carcinoma. Two patients had both squamous and basal cell carcinomas, two other patients, a male aged forty years with squamous carcinoma of the neck, and a female, aged sixty-six years, with squamous carcinoma of the right leg, died during the course of the study.

TABLE 4.16 Types and sites of skin cancer (ca) in nineteen albino patients attending a cancer clinic

	Squamous ca			Basal cell ca		
Site	Total	Number	%	Total	Number	%
Head	14		(70)	6		(75)
Face (unspecified)		4			3	
Cheek		4				
Ear		1			2	
Neck		5			1	
Torso	3		(15)			
Cervix		1				
Chest		1				
Back		1				
Limbs	3		(15)	2		(25)
Legs		3			1	
Hands					1	
TOTAL	20*	20	(100%)	8*	8	(100%)

* in six subjects two or more sites were reported.

The older age groups were well represented at the clinic, 68 per cent of the sample being forty years of age or over and more than one third were fifty years and older.

TABLE 4.17 Ages of nineteen albinos attending a cancer clinic for treatment

Age (years)	No	%
20 - 29	2	(10.5%)
30 - 39	4	(21.1%)
40 - 49	6	(31.6%)
50 - 59	5	(26.3%)
60 - 69	2	(10.5%)
TOTAL	19	

Squamous carcinoma was found on the torso and on the hands in this sample of albinos, whereas these sites were not reported in the skin biopsy study. Similarly, the ear was reported as the site in two cases at the cancer clinic, but in none of the skin biopsy series, whereas the eye was reported as the site in four of the latter series, but in none of the cancer clinic patients.

In summary, the data collected on cancer in albinos indicated that they were at risk even in the teenage stage and that this risk increased with advancing age. The Sotho albinos were at higher risk for having skin cancer than the Nguni albinos. Sites differed from those found in the general population and the head and neck were the commonest sites, but limbs and torso were also at risk in some cases. The type of cancer found was usually squamous carcinoma although basal cell carcinoma also occurred. No cases of melanoma were reported in the series of patients investigated here.

4.2.4 Life expectancy

Information was obtained on the ages of 254 Soweto subjects. The majority had been seen a number of times over the twelve years of the study and only three deaths were reported during this time. The three who died included two females, aged approximately one year and sixty-eight years, respectively, and a male aged forty-four years. Since the number of deaths was so few it was impossible to construct a life-table for the albinos in the sample. Therefore, in order to obtain a general idea of how long the albinos were living the ages of those in the sample were classified, according to the age groups used in the Census (1980), and the figures for these groups were compared with those from the Census data for the local black population. The results are presented in Table 4.18.

TABLE 4.18 Ages of 254 albinos compared with the ages of the general population in Soweto

	Albinos			General population	
	Expected*	Observed			
Age (years)	Number	Number	%	Number	%
<4	26	34	(13.5)	103 480	(10.0)
5 - 14	40	37	(14.6)	162 260	(14.6)
15 - 24	54	82	(32.3)	218 340	(21.2)
25 - 34	53	49	(19.3)	211 600	(20.5)
35 - 44	37	28	(11.0)	147 180	(14.3)
45 - 54	26	8	(3.1)	106 120	(10.3)
55 - 64	14	7	(2.7)	57 140	(5.6)
65 - 74	5	8	(3.1)	19 000	(1.8)
75+	2	1	(0.4)	6 580	(0.6)
TOTAL	257	254	(100)	1 031 700	(100)

* Calculated using prevalence rate of 1:4000

The Kolmogorov-Smirnov test was used to establish if the albino group had the same age distribution as the general population. The age distribution of the albinos was found to be significantly different from that of the general population ($p<.001$). The albino group tended to be younger than the general population, the mean age for the two groups being 24.2 and 28.2 years, respectively.

The albinos were over-represented when compared with the figures for the general population in three age groups: those aged four years and less, fifteen to twenty-four years, and sixty-five to seventy-four years. It is possible that the ascertainment methods led to a more complete ascertainment of albinos in the first of these two age groups, because the psychosocial component of the study required newborn subjects and hence the ascertainment was good in this group. The initial prevalence study was conducted mainly through visits to schools in the decade 1971 to 1981, so that these subjects were in the fifteen to twenty-four age group at the time the ages were analysed. It is, however, difficult to explain the excess of subjects in the sixty-five to seventy-four year old group.

The numbers of observed subjects compared with those expected were found to be fewer in the thirty-five to fifty-four year age group. Only 14.1 per cent of the albino subjects belonged to this group, whereas 24.5 per cent of subjects were expected. The subjects in this age group either did not come forward to participate in the study or they had a higher mortality rate than other groups. In the forty-five years and over age group forty-seven (18.3 per cent) albinos were expected but only twenty-four (9.3 per cent) were observed. These findings suggest that albinos may have a shorter life-span.

In order to determine whether the data collected in Swaziland also suggested that life expectancy might be reduced for albinos, the age groups of the Swazi sample were analysed. There were forty-nine albinos in this sample and their ages were compared with those available for the general population from the latest census (1976). The figures are presented in Table 4.19.

Again there were trends (possibly caused partly by the methods of ascertainment) in the Swazi data showing that albinos were over-represented in the younger age group, and, under-represented in the age group forty-five years and older (12.9 per cent of the general population compared with only 2.0 per cent of the albinos were in this group). Nevertheless, in this small sample from a predominantly rural area, there was one albino subject in the sixty to seventy-four year age group.

TABLE 4.19 Age distribution of albinos and the general population in Hhohho district, Swaziland

	Albinos		General population*
Age (years)	Number	%	%
0 - 14	29	59.2	47.6
15 - 29	14	28.6	25.4
30 - 44	5	10.2	14.2
45 - 59	0	0	7.6
60 - 74	1	2.0	3.6
75+	0	0	1.4
No information	0	0	0.2
TOTAL	49	100	100

* From the Report on the Swaziland Population Census (1976) only percentages not actual numbers, were given.

In order to assess whether there were differences in the age structure of the albino male and female population, when compared with that of the general population, the Soweto data were analysed by sex. The information for males is presented in Table 4.20.

TABLE 4.20 Ages of 132 male albinos compared with ages of males in the general population in Soweto

	Albinos			General population	
	Expected	Observed			
Age (years)	No*	No	%	No	%
<4	13	20	(14.8)	52 440	(9.7)
5 - 14	20	22	(16.2)	80 680	(14.9)
15 - 24	28	41	(30.4)	111 580	(20.6)
25 - 34	29	24	(17.8)	117 100	(21.7)
35 - 44	20	18	(13.4)	78 280	(14.5)
45 - 54	14	7	(5.2)	56 580	(10.5)
55 - 64	8	3	(2.2)	31 340	(5.8)
65 - 74	2	0		9 860	(1.8)
75+	0.7	0		2 720	(0.5)
TOTAL	134.7	135	(100)	540 580	(100)

* Calculated using the prevalence rate of 1:4000 (Kromberg and Jenkins, 1982)

The Kolmogorov-Smirnov test (Siegel, 1956) was applied to the data and the results indicated that the distribution of ages in male albinos and the general population was highly significantly different ($p<.001$).

Male albinos were under-represented in the over forty-five year age group, in which about twenty-five were expected but only ten were found. These findings suggest that albino males might not live as long as their normal counterparts.

The information concerning the ages of albino females is reported in Table 4.21. There were fewer albino females (fourteen) in the age group forty-five years and over than expected (twenty-one). However, in the sixty-five years and over age group the number of female albinos (nine) exceeded the expected number (three). The Kolmogorov-Smirnov test (Siegel, 1956) results showed only marginal evidence, that was not significant, of a difference in the distribution of ages in albino females and the general population ($p>.10$).

TABLE 4.21 Ages of 117 female albinos compared with ages of females in the general population

	Albinos			General population	
	Expected	Observed			
Age (years)	No*	No	%	No	%
<4	12	14	(11.8)	51 040	(10.4)
5 - 14	20	15	(12.6)	81 580	(16.6)
15 - 24	27	41	(34.5)	106 760	(21.7)
25 - 34	24	25	(21.0)	94 500	(19.2)
35 - 44	17	10	(8.4)	68 900	(14.0)
45 - 54	12	1	(0.8)	49 540	(10.1)
55 - 64	6	4	(3.4)	25 800	(5.3)
65 - 74	2	8	(6.7)	9 140	(1.9)
75+	1	1	(0.8)	3 860	(0.8)
TOTAL	122	119	(100)	491 120	(100)

* Calculated using the prevalence rate of 1:4000 (Kromberg and Jenkins, 1982)

In summary then, the investigation on life expectancy suggested that it is reduced in albinos compared with the general population. Male albinos were found to have a highly significantly different age distribution, being over-represented in the younger age groups, whereas females only had a marginally different age distribution, compared with the general population. Affected females therefore probably do not have a reduced life-expectancy, whereas for affected males longevity is probably decreased.

4.2.5 Skin colour in heterozygotes and homozygotes

Data on skin colour were collected during the Swaziland study. Reflectance spectrophotometric measurements were taken on eighteen obligatory heterozygotes (mothers of albinos) and eighteen normal matched controls. For purposes of comparison, measurements were also collected on a series of forty-three Swazi albino subjects.

The mean percentage reflectance values for the heterozygotes and controls at six wavelengths, together with the p values obtained by means of the Wilcoxon signed ranks test (Lehmann 1975) appear in Table 4.22.

TABLE 4.22 Mean reflectance values for female heterozygotes and controls (N = 18)

Filter No	Wavelength (nm)	Mean value Heterozygotes	Controls	V$	P
601	425	11.50	10.50	-1.624	0.05*
602	465	13.28	12.05	-1.930	0.02*
604	515	16.00	14.50	-1.803	0.03*
605	545	18.00	16.16	-1.572	0.05*
606	575	19.05	17.50	-1.289	0.09
609	685	41.11	38.88	-1.309	0.09

$ the sum of the ranks of the positive differences (Wilcoxon test)
* significant differences

Significant differences between the two groups were found at four of the six wavelengths, i.e. at 425, 465, 515 and 545 nm, which fall at the blue end of the visual spectrum. At each of these wavelengths the heterozygotes were found to be significantly lighter in skin colour than the controls (See Fig 4.6). Skin colour in mothers of albinos was also lighter at the remain-



Fig 4.6 Teacher (mother of an albino), on left, with light skin colour and school principal with normally pigmented skin.

ing two wavelengths, but the differences did not attain signficance.

The results of the measurements of skin colour in albinos are shown in Table 4.23.

TABLE 4.23 Mean reflectance values for albinos subjects

Filter No	Wavelength (nm)	Mean reflectance value	SD	No of subjects
601	425	35.95	5.02	43
602	465	45.56	5.81	43
604	515	48.88	6.27	43
605	545	45.60	5.20	43
606	575	48.26	5.22	42*
609	685	66.92	6.56	39*

* the missing subjects were babies who became irritable and the measurements could not be completed.

The homozygotes had much higher reflectance values indicating that they were very much lighter in skin colour than the heterozygotes and controls. The reflectance values for South African Caucasoid females from Wasserman and Heyl's (1968) study were extracted for the wavelengths used in the present study for purposes of comparison. The data for the albinos and Caucasoids, as well as those for the heterozygote and control females, are plotted in Fig 4.7. As expected, the albinos show the highest percentage reflectance at all wavelengths followed by the Caucasoids, whose skin colour is slightly darker. Heterozygotes and controls have much lower values indicating much darker skin colour. The haemoglobin absorption band is clearly seen at 545nm in the albinos, less clearly in the Caucasoids whose curve shows a slight depression at that wavelength, and not at all in the heterozygotes and controls, since its effect is masked by the presence of increased amounts of melanin.

The results, therefore, indicate that, as a group, female carriers for the albinism genes have skin colour which is measurably lighter at four of six wavelengths used, than normal matched female controls, but much darker than Caucasoid females and albinos.

4.2.6 Fertility

The data on family members collected in the course of the study and during the construction of the pedigrees were analysed in two ways. Firstly, the information concerning the Soweto albinos, their children, sibs and parents'

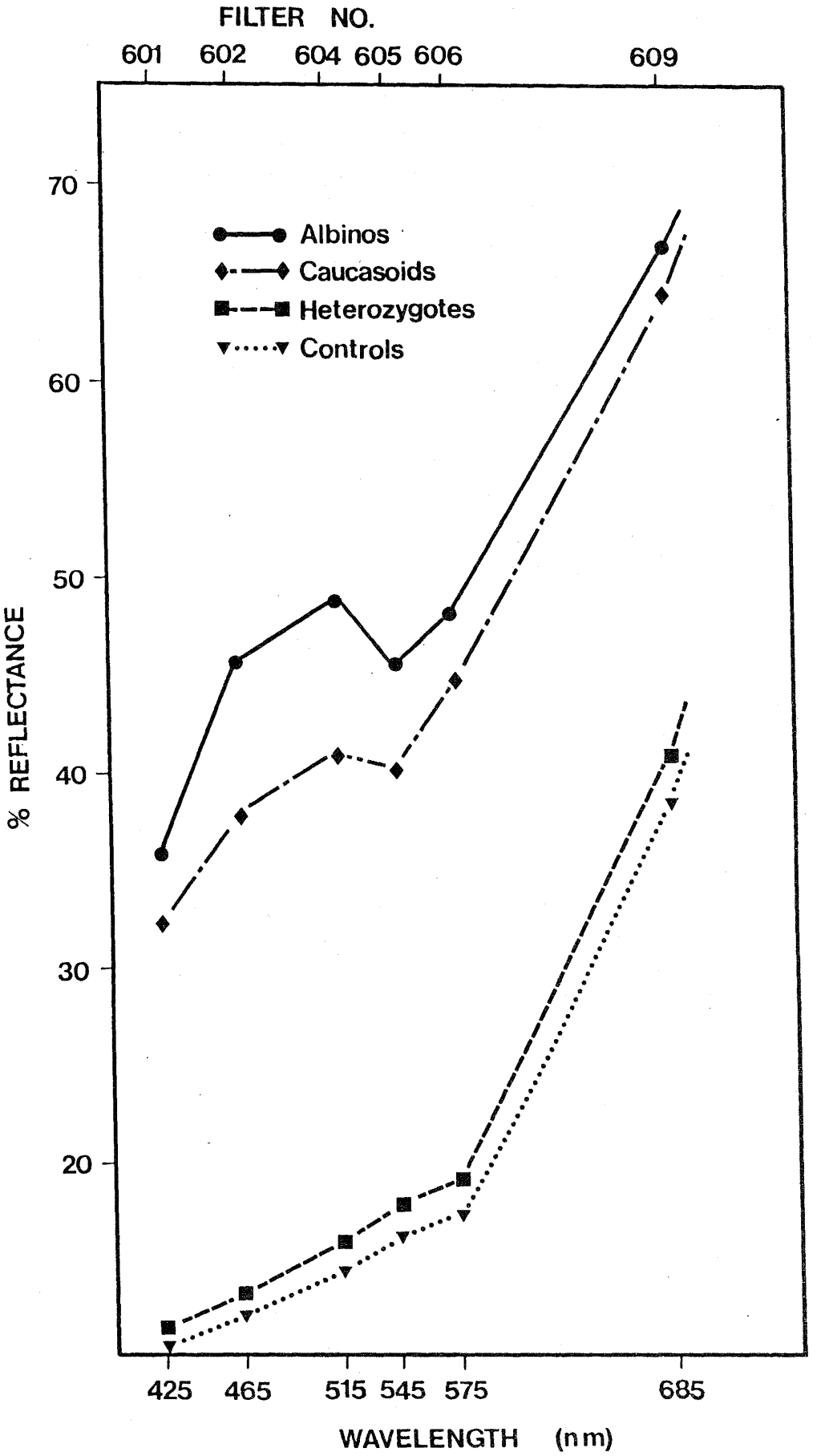

Fig 4.7 Skin reflectance measurements in albino, caucasoid, heterozygote, and control females

sibs was studied and categorised and the findings were compared wherever possible with relevant statistics in the general population, in order to assess the fertility of homozygotes and heterozygotes. Details, although not always complete, were obtained on 166 affected families. Secondly, the material gathered for the pedigrees of the matched pairs of mothers of albinos and mothers of normal babies, who participated in the maternal-infant study was analysed, and the two groups were compared. Information, which was again incomplete in a few cases, was collected on thirty-six matched pairs.

4.2.6.1 Fertility in homozygotes

The fertility of the albinos was investigated by examining the pedigrees of 128 subjects aged fifteen or more years. The results are presented in Table 4.24. Altogether 35.9 per cent of the sample had children and in this group of subjects with children there were fewer male albinos (20 per cent) than females (48 per cent). The mean age of the subjects was 29.2 years, but for the females who had children it was 33.7 and for males with children 40.9 years.

TABLE 4.24 Number of female and male albinos with children

	Females		Males		Total	
Age group (years)	No	No with children (%)	No	No with children (%)	No	No with children (%)
15 - 19	17	6(35.3)	16	0(0)	33	6(18.2)
20 - 29	30	11(36.7)	22	3(13.6)	52	14(26.9)
30 - 39	8	6(75.0)	10	2(20.0)	18	8(44.4)
40 - 49	4	3(75.0)	12	7(58.3)	16	10(62.5)
50 - 59	3	3(100)	1	1(100)	4	4(100)
60 - 69	2	1(50.0)	1	1(100)	3	2(66.7)
70+	2	2(100)	0	0	2	2(100)
TOTAL	66	32(48.5)	62	14(22.6)	128	46(35.9)
Mean age (years)	28.9	33.7	29.2	40.9	29.1	35.9

The number of children per albino for males and females over all age groups (i.e. 15 - 70+), and also excluding fifteen to nineteen year olds, since no males in this group had offspring, and the number of children per albino who had children, are reflected in Table 4.25. Male albinos had fewer children per person compared with female albinos, even when the fifteen to nineteen year group was excluded.

In the group with children the males reported that they had more child-

ren (2.78 per albino) than the females (who only reported 1.84 children per person), which might be expected since the mean age of these males was 40.9 years while that of the females was only 33.7 years.

TABLE 4.25 Number of children per albino (for all subjects and for those with children) by sex of the subjects

	Females			Males		
Age group (years)	No	No of children	Children per female	No	No of children	Children per male
All subjects						
15 - 70+	66	59	0.89	62	39	0.63
20 - 70+	49	53	1.08	46	39	0.85
Subjects with children						
15 - 70+	32	59	1.84	14	39	2.78

The life-time infertility rate has been defined as 'not ever having live-born children and being thirty years or older' (Irwig 1984, personal communication). This rate was estimated for males, females and the total group, and the findings are presented in Table 4.26.

TABLE 4.26 Life-time infertility rate for female and male albinos (aged thirty to fifty-nine years)

	Subjects		
Sex	No	No without offspring	Life-time infertility per 1 000 people
Females	19	4	210
Males	24	13	542
TOTAL	43	17	395

The only comparable available rate was that found in a study on Xhosa women in the Transkei by Irwig and Ingle (unpublished data, 1984). They found a life-time infertility rate of 57 per 1 000 women. This rate was compared with that of 210 per 1 000 albino women and the albino infertility rate was found to be significantly higher ($p = 0.02$, Binomial test, Siegel, 1956).

The number of albino and Xhosa (Transkei) females who have never had live-born children, by age group, is reported in Table 4.27. The figures suggest that albinos are less likely to have children than Xhosa women in every age group, except fifteen to nineteen years. In this youngest age group the albinos proportionately had more children than Xhosa women, al-

though the numbers in the albino group were very small. These results also suggest that the fertility of albino females is reduced.

TABLE 4.27 Albino and Xhosa* women who have never had live-born children, by age

Age (years)	No of women		No without live-born children		Without live-born children/1000 women	
	Albino	Xhosa	Albino	Xhosa	Albino	Xhosa
15 - 19	17	984	11	845	647	859
20 - 24	15	823	10	233	666	283
25 - 29	15	724	9	82	600	113
30 - 39	8	1 037	2	55	250	51
40 - 49	4	898	1	50	250	56
50 - 59	7	555	1	38	143	68

* derived from a survey by Irwig and Ingle (1984, unpublished data).

In the present sample therefore the statistics indicated that: only about half the albino females (mean age 33.7 years), and about a quarter of the males (mean age 40.9 years) claimed to have children; the number of children per head was 0.89 per female and 0.63 per male; the life-time infertility rate was greater in female albinos than in Xhosa women. Although strictly comparable data were not available these figures suggested that fertility was reduced in both male and female albinos.

4.2.6.2 Fertility in heterozygotes

The heterozygote group was divided into two sub-groups; obligatory heterozygote matings (i.e. parents of an albino child) and probable heterozygote probable normal matings (i.e. the grandparent of an albino child and his or her spouse).

(i) Fertility in heterozygote x heterozygote matings

Information was collected on 143 heterozygote x heterozygote matings in Soweto, and on thirty-six such matings in the Johannesburg area, together with thirty-six matched controls with no history of albinism. The ascertainment of these subjects followed the detection of an albino in the family or the birth of an affected baby, so that childless couples were not included in this analysis.

The 143 Soweto couples who had one or more albino children were found to have had 563 children between them, or 3.93 children per couple. Some of these couples had not yet completed their families, so that this figure was not a total fertility rate, and could not be compared with such figures for

the general Soweto population (if they had been available which was not the case).

The data on the thirty-six matched pairs are presented in Table 4.28.

TABLE 4.28 Offspring in heterozygote x heteroxygote and in normal control matings

Group	No	Mean age (years)	No of Offspring	Offspring per couple
Heterozygotes	36	25.8	75	2.08
Controls	36	23.4	75	2.08

The obligatory heterozygotes in this small sample had had the same number of children as the controls, although they were, on average, about two years older than the controls. This finding suggested that heterozygote x heterozygote matings did not have increased fertility compared with a normal matched control group.

(ii) Fertility in heterozygote x normal matings

In order to obtain an assessment of whether heterozygote x normal matings produced more or fewer children than the general population, data were collected either from parents of albinos on their siblings, or from the grandparents (if they were available) on their children, or from the albinos (where the other informants were lacking) on their parents' siblings. Unless there is a high mutation rate for the gene for albinism, one of each couple, among the grandparents of albinos, will be heterozygous for the gene. Since these grandparents were not influenced to increase or reduce their number of offspring by the birth of an albino child, they might provide a more reliable indication of the fertility of heterozygotes than do the figures for heterozygote x heterozygote matings.

Pedigrees were drawn up for 138 mothers and they were found to have 605 siblings, or 4.38 per subject. Details were available on only 112 fathers of albinos and they were reported to have 472 sibs or 4.21 siblings per subject. These figures gave an estimate of the total fertility in heterozygote x normal matings, since in most cases the grandparents of albinos had completed their child-bearing years.

The pedigrees of thirty-five of the matched pairs, for whom information was available, were also analysed, and the results on the siblings of the heterozygote and control females and their 'husbands' appear in Table 4.29.

A reliable total fertility figure does not appear to be available for the urban Soweto population. It is probably lower than the rate (5.2) given

for the South African black population (Report of the Science Committee of the President's Council, 1983), since this rate is based on rural and urban estimates combined, and urban figures are generally believed to be lower than those from the rural areas. The heterozygote group in this study can therefore only be compared with the matched control group, although the sample size in both was rather small.

TABLE 4.29 Siblings of heterozygotes and control females and males

	Total No		No of Siblings		Siblings per subject	
Group	Het*	Con*	Het	Con	Het	Con
Females	35	35	146	145	4.17	4.14
Males	30	24	128	75	4.27	3.12
TOTAL	65	59	274	202	4.22	3.63

* Het = heterozygotes, Con = Controls

The information collected on fathers was incomplete, since in a few cases the pregnancy was the result of a short-term liaison, and in others the mother did not have a good knowledge of the father's family. It was only possible to verify the mothers' report with the father in a few cases. The heterozygote fathers, however, were reported to have more siblings per subject than the controls. If the two groups of mothers' siblings and fathers' siblings are combined the experimental (heterozygote) group has a fertility rate of 4.22, while the control group has a fertility rate of 3.63. The Wilcoxon rank-sum test (Snedecor and Cochran, 1980)was used to compare the number of siblings in the experimental and control groups. The results showed no significant difference within the female, male or the combined male and female groups. These findings therefore suggested that the heterozygote did not have an advantage as regards fertility.

In summary, then, the life-time infertility rate appeared to be significantly increased in albinos compared with the Xhosa group of the black population, and the homozygote fertility rate was therefore probably reduced. The heterozygote fertility rate however did not appear to differ from a matched control group in either heterozygote x heterozygote matings (parents of albinos) or in probable heterozygote x probable normal matings (grandparents of albinos).

4.2.7 Summary

The results on the genetic studies indicated that:

(i) The prevalence rate of albinism among the mixed ethnic groups of the black urban population of Soweto was 1 in 3 900. Different ethnic groups had different prevalence rates, the Nguni (e.g. Zulu and Xhosa) generally had low rates and the Sotho (e.g. Tswana and Southern Sotho) high rates. Consanguineous marriages were found to be much commoner in the Sotho than in the Nguni affected families, confirming local cultural traditions in these ethnic groups. Males and females were found to be equally affected and a study of sibships indicated that the data were compatible with recessive inheritance.

The prevalence rate in the rural Tswana group in Botswana was found to be 1 in 1 307 in a village of 18 300 people. In the rural Zulu however, the rate was 1 in 2 576 among school children in Kwazulu. While in the rural Swazi the rate was 1 in 1 951. All these rates were higher than those found for these groups in urban Soweto.

(ii) By means of hair-bulb incubation tests and physical examination most albinos were tentatively classified into one of five groups: tyrosinase-negative tyrosinase-positive (A and B), yellow mutant, brown albinism, or rufous albinism. In the small sample from the schools for the blind and cancer clinics the tyrosinase-negative hair-bulb test results occurred quite frequently, but in the general Soweto community tyrosinase-negative type albinos were rare and tyrosinase-positive followed by brown albinos were most commonly identified.

A study of the physical characteristics indicated that very few albinos had no pigment at all, the majority had yellow or darker coloured hair and about half had skin pigment in the form of ephelides. More than half the sample had blue eyes, but the irides frequently had streaks of yellow or light brown pigment radiating from the centre. Nystagmus occurred in the majority of subjects, but was unusual in the brown albinos and not found at all in the rufous albinos. Strabismus was found in about half the tyrosinase-negative and positive types, but it occurred in only a few brown albinos and in none of the rufous albinos.

(iii) Skin cancer was a common problem for the albinos and malignant or premalignant lesions (solar keratoses) were noted in one fifth of the albinos aged twenty to twenty-nine years, more than half those aged thirty to forty-nine years and all those older than fifty. Such lesions were found significantly more frequently in Sotho than in Zulu albinos. Squamous and basal cell carcinomas were far more common in albinos than in the general

black population and biopsies from albinos represented nearly 13 per cent of a sample of all skin biopsies (from black patients) examined for suspected carcinomas.

The commonest site for the carcinoma in albinos was on the head, and the cheek was particularly susceptible, but the lower limbs and torso were also occasionally affected. The risk for skin cancer increased with age starting in some as early as the teenage years.

(iv) Life expectancy appeared to be reduced in albinos and their age distribution was significantly different from that of the general population in Soweto. Affected males particularly were significantly underrepresented in the older age groups, whereas the age distribution in females was only marginally different from that in the general population.

(v) Skin colour in heterozygotes was found to be measurably lighter (and the difference was statistically significant) at four different wavelengths compared with controls. Albinos had skin colour which was lighter than caucasoid females and both these groups were much lighter than the heterozygotes, and normally pigmented controls.

(vi) The fertility in albinos appeared to be reduced and their life-time infertility rate was significantly greater than that of one local black ethnic group (the Xhosa).

Fertility in both the parents and grandparents of albinos, however, appeared to be similar to that in the general population.

4.3 Psychosocial Studies

4.3.1 Introduction

The information collected during the interview and observation sessions with mothers and fathers was entered onto the schedules by the trained interviewer. The schedules were then returned to the writer, the responses were analysed and the findings are presented below.

The results are divided into two main sections, the first dealing with the information received from mothers, and the second with that received from fathers, and finally all the significant findings are summarized.

In the first section the data obtained at the first interview with the mothers is presented in greater detail than those collected at later interviews. The results from the first interview are presented in subsections on the mothers' medical and childhood history, the pregnancy, delivery and neonatal problems, infant-care, the maternal and paternal response to the in-

fant (according to the mother), the maternal-paternal relationship, siblings' response to the new baby, and the mother's behaviour in the observation situation.

The second to sixth interviews with the mothers are not reported in such detail, but rather the changes that occurred (particularly in the items showing significant differences at the first interview), in each interview and observation as compared with the previous interview and observation are outlined. The differences found between the groups at each stage are described. The findings from the sixth and final interview were also compared with those from the first interview, so that results could be obtained concerning the changes which occurred over time, within the experimental and control groups, in the couples who remained in the study throughout the specified time period of fifteen months.

After the last interview an observation of the mothers and their babies in a play situation was made and an anxiety scale was administered. The results of this observation and scale are reported separately.

4.3.2 Results of data obtained from interviews and observation sessions with the mothers

4.3.2.1 Interview 1

There were thirty-seven experimental and thirty-seven matched controls at this initial stage of the study. The results obtained from the responses to the schedule items (see Appendix C) are presented here in terms of proportions and percentages in most instances, and the raw data on the matched pairs, where significant differences were found, appear in Appendix K.

(i) Medical History of Subjects

The subjects were asked ten dichotomous questions on their medical problems and their responses are shown in Table 4.30.

As can be seen from the table significantly more experimental than control subjects claimed to have headaches and to feel depressed. On the other items the subjects in the two groups showed similar responses.

There were twelve experimental subjects and five controls who had been hospitalised and this difference was not statistically significant. The varied reasons for their hospitalization are, however, worth recording as an indication of the medical problems experienced by heterozygotes (see Table 4.31). Tonsillitis and tuberculosis were the commonest problems in the group of subjects as a whole.

TABLE 4.30 Comparison of the medical history in two groups of mothers at the first interview

History	Experimental group (N = 37) No	%	Control group (N = 37) No	%	chi-square	p
Operations	2	(5.3)	0			
Fractures	2	(5.3)	1	(2.7)		
Hospitalization	12	(31.8)	5	(13.5)		
Headaches	17	(44.7)	8	(21.6)	4.27	<.05*
Sleeplessness	4	(10.5)	2	(5.4)		
Tiredness	1	(2.6)	2	(5.4)		
Stomach pain	1	(2.6)	2	(5.4)		
Breathlessness	3	(7.9)	1	(2.7)		
Depression	7	(18.4)	0		5.14	<.05*
`Grumble' factor	2	(5.3)	0			

* significant differences found using McNemar's test for comparing frequencies in matched samples (for raw data see Appendix K Table I).

TABLE 4.31 Reasons for Hospitalization

Reason	Experimental group	Control group
Tonsillitis	2	1
Injuries	2	
`Pelvic' infection	1	
Tuberculosis	1	2
Diphtheria		1
Asthma	1	
`Swollen legs'	1	
Abscess	1	
Cardiac problems	1	
Abortion		1
Dilatation and curettage	1	
`Don't know'	1	
TOTAL	12	5

(ii) Childhood history

After the medical history was obtained from the subjects they were asked three questions about their childhood.

The first item concerned their upbringing and whether they were raised by their mother, father, both parents or relatives. There were four experi-

mental subjects and eight controls who were brought up by their mothers alone, and only two controls who were raised by their fathers alone. A further twenty-three experimentals and nineteen controls grew up with both parents. Altogether eleven experimentals and eight controls were raised by relatives and, in about half of these cases, the relative was the grandmother. There were no significant differences between the groups on this item.

Secondly, the subjects were asked whether they had had a rural or an urban childhood. In the experimental group 47 per cent of subjects had had a rural childhood and 53 per cent an urban one, while for the control group the figures were 24 per cent and 76 per cent respectively. This finding was to be expected since all the controls were selected from an urban area, whereas some of the experimental group were rural dwellers.

Information on the sibship size of the mothers themselves was then requested. The means for the experimental and control group were 5.0 and 5.2 siblings per subject, respectively, so that family size was similar in the two groups of mothers.

(iii) The pregnancy

The subjects were questioned on any unusual event which occurred during the pregnancy which gave rise to the index baby. There were seven experimental subjects and five controls who mentioned strange dreams as being an important part of their pregnancies. Dreams of snakes were reported by two experimental and one control subject and two experimental subjects mentioned dreams of white objects (a dog in one and a child in the other). Two further experimental subjects reported contact with albinos during their pregnancies. The majority in both groups however had experienced nothing unusual.

In the experimental group 45 per cent of subjects claimed that they had planned the pregnancy, whereas only 32 per cent of the controls did so. This finding was probably associated with the fact that more of the experimental subjects were married than the controls. Full-term pregnancies occurred in the majority of both groups (87 per cent of experimental and 97 per cent of controls) and premature births were experienced by five experimental subjects and one control.

(iv) Delivery and neonatal problems

There were five (13.1 per cent) of the mothers in the albino group who had difficult deliveries. In this group three mothers had to have delivery by

Caesarian section, one required a vacuum extraction, and a further one had a difficult delivery because the baby was `too big´. In the control group no mothers had difficult deliveries, but one had a post-partum haemorrhage and a second one had an adverse reaction to a blood transfusion, and both these mothers required special care. In comparison with these two mothers (5.4 per cent of the control group) seven of the albino mothers (19 per cent) required special post-delivery care. The difference between the two groups was however not statistically significant.

Special care, mostly due to prematurity, was required by four albino babies (11 per cent of the group). No normal babies had any problems neonatally according to their mothers.

The mothers were all asked whether or not they wanted to hold and breast-feed their babies immediately after birth. In the experimental group only eighteen (49 per cent) wanted to hold their babies immediately and six of these women qualified their statements with remarks such as `Yes, I wanted to hold the baby, but I did not know he was an albino´, or `Yes, I wanted to hold him, but with a sore heart´. In comparison thirty-four (92 per cent) of the control mothers wanted to hold their babies immediately, and the three who did not said they were tired, dizzy or had pain. The difference between the two groups on this item was significant ($p<.001$). There were eighteen (49 per cent) of the albino mothers who did not want to hold their babies. The reasons they gave for this behaviour was that they: were shocked because the baby was an albino (five subjects); were too tired (five subjects); were surprised (three subjects); were not feeling well, or fainting, or crying (two subjects); were convinced a mistake had been made (one subject); were not brought the baby (one subject); and could not give reasons (one subject). Only one subject said she did not know whether or not she wanted to hold her baby.

The behaviour, as reported by the mothers, in connection with breast-feeding, was also found to be significantly different ($p <.05$) in the two groups. Only fifteen (40.5 per cent) of the mothers of albinos wanted to breast-feed their babies immediately after delivery, while twenty-six (70 per cent) of the mothers with normal babies wanted to do so. Among the twenty-two (59.5 per cent) of the mothers of albinos who did not want to breast-feed their babies (sixteen subjects) gave the following reasons: they were too shocked (five subjects), too tired (three subjects), scared (one subject), sad (one subject), in pain (one subject), not well (two subjects), or they said that they had no milk (two subjects) or that the baby refused

the breast (one subject). There were eleven subjects in the control group who did not want to breast-feed their babies and of those who gave reasons, the majority said that they were too tired (six subjects), were dizzy (one subject) or had no milk (one subject).

The group of mothers of the albino babies therefore showed a reaction to the birth of their unexpectedly different babies, which was evidenced in their reluctance, in comparison with the normal control group, to hold and breast-feed their babies. Some experimental mothers also expressed feelings of shock and surprise, as might be expected in mothers in this situation.

(v) Infant care

Information was obtained on several different aspects of baby-care and the first item in this series concerned the method of feeding. The results can be seen in Table 4.32.

TABLE 4.32 Breast/bottle feeding at $\pm$ 2 weeks

	Experimental group		Control group	
	No	%	No	%
Breast feeding	27	(73)	33	(89)
Bottle	4	(11)	0	(0)
Both	6	(16)	4	(11)
TOTAL	37	(100)	37	(100)

The majority of both groups were breast-feeding their babies at the first interview. There were however four (11 per cent) of the mothers of albinos who had already put their babies entirely onto the bottle and, although no controls had behaved in a similar fashion, the difference between the groups on this item was not significant.

The mothers were then asked if their babies were feeding well and the large majority (97 per cent) of both groups responded positively. They also stated that their babies were sleeping well (97 per cent of the experimental and 94 per cent of the control mothers). Among the mothers of albinos eight (21 per cent) complained that their babies cried a lot, and one gave the culturally believed reason that, when the weather was cloudy, the wound on the umbilicus caused pain and made the baby cry. In the controls six mothers (16 per cent of the group) said their babies cried a lot and the reasons given were that the umbilicus hurt, the baby had no name, or had a cold, or liked milk too much. There were no statistically significant differences in this series of items.

The nicknaming of the infants was also discussed with the mothers and 29 per cent of the mothers of albinos and 16 per cent of the controls had given their babies nicknames at the time of the first interview, which was not a statistically significant difference.

The responses to the item on plans for the future of the baby showed that the two groups planned generally to behave in a similar fashion. Only about half of the mothers in the experimental and control group were planning to stay at home with their babies. Of the control mothers thirteen (or 35 per cent) were planning to return to work, and ten of the thirteen intended to seek work either when the baby reached six months' of age (five subjects) or at one year (five subjects), the remaining three would wait until the baby reached two years of age. There were seven (18.5 per cent) mothers of albinos who planned to return to work, three when the baby was six months old and four at one year. One mother's response was that she could not return to work since child-minders do not like albinos. Details of the results on this item are presented in Table 4.33.

TABLE 4.33 Plans for the baby

	Experimental group		Control group	
	No	%	No	%
Stay at home with baby	20	(54)	22	(59)
Return to work	7	(19)	13	(35)
Baby to go to child-minder	0		1	(3)
Baby to go to grandmother	2	(5.5)	1	(3)
Baby to be given good education	2	(5.5)	0	
Mother and baby leaving town	6*	(16)	0*	
	37	(100)	37	(100)

* a significant difference (chi-square 4.17, $p<.05$, found using McNemar's test for comparing frequencies in matched samples, for raw data see Appendix K)

Among the mothers of albinos six (16 per cent of the group) were planning to leave town, and to go `home' to the rural areas with their albino babies. None of the controls admitted to having similar plans at this stage, and the responses of the groups were significantly different on this item ($p<.05$).

As a final open-ended item in this section the mothers were asked if they had any complaints about their babies. There were six mothers of albino babies (16 per cent of the group) and no mothers of normal babies who had

complaints and this difference was significant at the $p < .05$ level. The mothers of albinos stated that they did not like the hair and skin colour of their babies (two subjects), wished the hair would turn black (one subject), asked `why must she be an albino' (one subject), and complained of sensitive skin and rashes in their babies (two subjects).

(vi) Maternal response

In order to collect data on the maternal response the mothers were questioned on their attitudes and feelings about their new babies.

The first item in this group was for introductory purposes and the mothers were asked who their babies looked like. The groups responded similarly and there were twenty-four and twenty-three of the mothers in the experimental and control groups respectively who said the baby looked like the father, and only six and seven respectively who said it looked like its mother. The remaining mothers did not know who the baby looked like, or said it looked like other relatives.

When asked if they liked the way the baby looked thirty-six (97 per cent) controls and eleven (29 per cent) mothers of albinos said `Yes', this difference was significant ($p < .01$, for raw data see Appendix K). A further seventeen (45 per cent) of the latter group answered with a qualified yes, adding, for example, `she's God's gift', `nothing can be done', `he's a human being', `its my child'. There were nine (24 per cent) mothers in the albino group, and one in the control group, who admitted that they did not like the way the baby looked, one mother of an albino baby added that she was intending to give it away for adoption, and another that the father would deny paternity.

The mothers' emotional responses to their new babies were investigated by two general items on what they were feeling and thinking. The results on the feelings appear in Table 4.34. Significantly fewer of the albinos' mothers said that they were happy about the birth. Altogether seventeen (46 per cent) said they felt `alright' and were neither happy nor sad, perhaps indicating some indifference or emotional numbness. A third group of mothers admitted quite frankly to being heartbroken, sad, worried and surprised. The control mothers however were happy to have the baby in twenty-nine (78 per cent) cases and felt alright in eight (22 per cent) of cases.

When asked what they thought of their situation some mothers of albinos made positive responses and stated that the child must be educated, be kept healthy, survive, be brought up well, work for and help its mother, that the

child would sleep well, was alright and was a person. On the negative side the mothers reported that: nothing could be done, they must avoid thinking, they wanted to cry, they felt confused and surprised, the child must go to the grandmother or the `farms', and that hopefully it might be like other children. In contrast the mothers of normal children thought that they must educate their children, keep them healthy and bring them up well, and two mothers in this group hoped that their children would not become `tsotsis' (delinquents) or truants.

TABLE 4.34 Mother's feelings about the birth of their babies

	Experimental group		Control group	
	No	%	No	%
Happy	14*	(38)	29*	(78)
Sad	2	(5)	0	
Heartbroken	2	(5)	0	
Alright	17	(46)	8	(22)
Worried	1	(3)	0	
Surprised	1	(3)	0	
	37	(100)	37	(100)

* significantly different (chi-square 8.45, $p<.01$, found using McNemar's test for comparing frequencies in matched samples; for raw data see Appendix K)

The mother's view of her baby was explored and two questions were put to each subject concerning how the baby looked and what caused it to look that way. The majority of mothers said their babies looked beautiful (88 per cent of experimentals and 82 per cent of controls) and one mother of an albino and three mothers of normal babies thought their babies looked ugly. The remainder said their babies looked alright. There were ten (26 per cent) experimental and seven (19 per cent) control mothers who claimed that God was responsible for the way the baby looked. A further seven (18 per cent) and thirteen (33 per cent) subjects in the respective groups admitted that inheritance was responsible. The remaining mothers in the albino group said that the children looked the way they did because of: the mothers contact with whites, the coloured blood in the family, witchcraft, dreams of sleeping with a white man or of the dead, laughing at and disliking albinos, having bad blood, the parents being light-skinned, drinking Fanta orange cold drink, eating certain foods, worrying and fighting with the father of the baby during the pregnancy, and `lack of sperms' in the boyfriend. Whereas

the other mothers of normal babies said that the baby looked the way it did because it was well-looked after and well fed, loved, thriving on breast-milk, and because the mother had been happy, well cared for and well fed in pregnancy.

The majority of the mothers in both groups however stated that their babies were good babies and only five (13 per cent) of the mothers of albinos and two (five per cent) of the mothers of the controls said that their babies were not so good, for example, because they cried too much. When asked if they showed their new babies to their friends sixteen (42 per cent) mothers of albino babies and nineteen (52 per cent) controls stated that, because of cultural beliefs and rules concerning the behaviour of mothers after the birth of a baby, they were not allowed to take the baby out at all or show it to friends or visitors to the house. There were however fifteen (40 per cent) experimental mothers and eighteen (49 per cent) controls who nevertheless said they showed the baby to friends. There were significantly more mothers of albinos than controls who said they had no friends (seven and none respectively, $p < .05$).

The subjects' ideas on the care of their babies were then probed in an attempt to identify whether they proposed to give any special treatment to these babies. The mothers were asked three questions about whether they would give their new babies more clothing, food and/or schooling than they would give to their other children. The groups showed no statistically significant differences in their responses to the first two items, but the response to the item on schooling showed that ten subjects (26 per cent) in the experimental group planned to give this baby more schooling than its siblings, while only two (5 per cent) of mothers in the control group planned to act in a similar fashion (this difference was significant at the 5 per cent level).

Family planning was also discussed with the subjects and the mothers in both groups responded similarly. The majority (56 per cent in the experimental and 62 per cent in the control group respectively) stated that they would have more children, but thirteen (34 per cent) in the albino group and twelve (33 per cent) in the control group said they did not plan to have any more children. When asked their reasons for not having more children several said `because of the cost of living', but four subjects (10.5 per cent) in the albino group said that they were afraid of having another albino child. In addition, twenty-two (58 per cent) mothers of albinos said if they did

have another child they would prefer not to have one like the present baby, four (10.5 per cent) said they would like another albino baby and ten (26 per cent) said they did not mind what the next one was like. Whereas in the control group thirty-one (84 per cent) of the subjects, said they would be quite happy if the next baby was like this one. There was a highly significant difference between the groups on this item ($p < .001$, for raw data see Appendix K).

The last two items in this section were aimed at assessing the mother's behaviour in connection with her visits to friends with or without the baby. Again the majority in both groups emphasized that due to cultural restrictions they were not allowed out with the baby while it was still very small, and only four (10.5 per cent) mothers of albinos and six (16 per cent) control mothers admitted to visiting friends with their babies.

(vii) Paternal response according to the mother

There were five items on the schedule which dealt with the mothers' view of the paternal response to the birth of the baby under attention.

The mother was first asked what the father felt about the birth of the baby. The responses to this item are shown in Table 4.35.

TABLE 4.35 Mothers' impression of the paternal response to the birth of the baby

	Experimental group		Control group	
Paternal response	No	%	No	%
Happy	8*	(21.6)	28*	(75.7)
Loves/likes child	6	(16.2)	4	(10.8)
Alright	4	(10.8)		
Comforts me	1			
Shocked/surprised	2	(5.4)		
Confused	1			
Feels nothing	4	(10.8)		
Denied paternity	1		1	
Deserted	1			
Denies albinism	1			
Do not know	2	(5.4)	1	
Not seen baby	6	(16.2)	3	(8.1)
TOTAL	37		37	

* significantly different (chi-square 12.04, $p < .001$, found using McNemar's test for comparing frequencies in matched samples, for raw data see Appendix K)

It is apparent from this table that, if the mothers' opinion can be accepted, the fathers of the albinos responded to the birth of their babies in a similar way to the mothers, and in a significantly different way to the control fathers. Only eight fathers (21.6 per cent) were happy, whereas in the control group twenty-eight (75.7 per cent) were happy with their new baby. At the time of the first interview one father of an albino baby and one father of a normal baby had denied paternity, and six fathers of albinos and three fathers of normal babies had not yet seen their babies.

The majority of the mothers (twenty in each group) reported that the father's behaviour, regarding time spent in the home, had not changed since the birth. There were, however nine subjects in each group who said the father was at home more since the birth, and eight experimentals and six controls stated that he was at home less often. Two controls explained that the father was not at home so much, because the aunt wanted `lobolo' (the payment of bride-price) in one case, and custom dictated his absence for ten days after the birth of the baby in the other case.

The drinking habits of the fathers were discussed with the mothers. Again the two groups showed a similar picture and there were no significant differences concerning whether the fathers were drinking more since the birth than they did prior to it. Only a short time had elapsed between the birth of the baby and the first interview, so the mothers might not have had time to formulate a realistic opinion on this problem.

The majority of both groups of mothers thought that the fathers gave attention to the siblings both before and after the birth of the present baby. One mother in the albino group added that her husband gave more attention to the albino sibling than to the normal siblings.

Lastly the mothers were asked whether they had any complaints about their husbands. There was a significant difference ($p<.05$) between the groups and ten (26 per cent) mothers of albinos and three (8 per cent) control mothers made complaints about the fathers of their babies. The complaints concerning the fathers of albinos were that they: had not seen the baby (two subjects), did not support the baby (two subjects), did not love the mother any more, were out of work, in jail, short-tempered, and swore at the mother (one subject each). The main complaint against the fathers in the control group were that they drank too much.

(viii) The maternal-paternal relationship

One item was inserted into the schedule to obtain data on the mother's view

of the maternal-paternal relationship and whether or not there had been a change before or after the birth of the baby. The results are shown in Table 4.36.

TABLE 4.36 Mothers' view of the relationship between mother and father

	Experimental group				Control group			
	Now*		Before birth		Now*		Before birth	
	No	%	No	%	No	%	No	%
Good	27	(73.0)	26	(70.2)	34	(91.9)	29	(78.4)
Bad	5	(13.5)	11	(29.8)	3	(8.1)	8	(21.6)
Do not know	3	(8.1)						
Father elsewhere	2	(5.4)						
TOTAL	37	(100)	37	(100)	37	(100)	37	(100)

* at first interview

The mothers in both groups perceived the relationship with the fathers in a similar way, more subjects seeing it as good after than before the birth of the baby. No significant differences between the groups were detected.

(ix) Mothers' view of the siblings' response

There were eighteen (49 per cent) mothers in the experimental group and sixteen (43 per cent) in the control group who had no other children and so the items in this section were not applicable to them. The totals for each group on the items on siblings were therefore reduced to nineteen and twenty-one for experimental and control groups respectively. Since the number of matched pairs where siblings were present in both members of the pair was too small statistical tests were not performed on this data. Observations only are therefore presented here.

The mothers reported on the siblings' responses and remarks when they saw the baby. In the albino group there were four children who said the baby was a white child, and one asked the mother why she had bought a white child. There was however one child in the control group who asked the mother to throw the baby away. Eleven (58 per cent) mothers of albinos and seventeen controls (81 per cent) said their children were happy to have a new sibling.

The mothers said that sibs were permitted to handle the infant in 58 per cent and 62 per cent of the experimental and control groups respectively. Among the mothers of albinos however three added that the sibs refused to handle the child and one would not even enter the room where the baby slept.

There were four (21 per cent) experimental and six (29 per cent) control mothers who would not allow their other children to handle the new baby, frequently because it was `too small'.

With regard to school attendance the behaviour of the sibs had not changed according to the mothers, in the two groups, since the birth of the new baby. All those of school-going age were said to be attending school regularly.

Most mothers stated that the sibs had brought friends in to play at home. In only two and three cases in the experimental and control groups respectively were the sibs not allowed to show the baby to their friends. However there were three sibs of albino babies who did not show the baby to their friends as, according to their mothers, they did not want their friends to know the baby was an albino.

(x) Information from observations on the mothers' behaviour towards their infants

After the interview was concluded the mother was asked by the interviewer to change her baby's napkin. Her actions in dealing with her baby in this situation were observed and scored according to the predetermined list of sixteen specific behaviours (see Appendix H). For eleven of the sixteen behaviours it was easy for the observer to note whether or not the behaviour had occurred. In four items, however, the observer had to judge whether or not the mothers had behaved in a certain way. When the results were examined it appeared that the majority of the `don't know' responses occurred in connection with these four behaviours, i.e. `handled the baby carefully', `appeared relaxed', `settled baby well', `appeared to care'. The data collected on these items were therefore considered to be unreliable and discarded. A fifth item was treated separately, it was: `Asked questions about the baby'. The results on this item showed that the mothers of albino babies asked questions related to, and because of, their baby's albinism, and therefore this item could not be included in the total. There were eleven items remaining on the scale after these items had been discarded. For the analysis of differences between the groups on separate items,the pairs in which `don't know' was recorded for one member were dropped. The results appear in Table 4.37.

Altogether there were four significant differences between the groups in the total of twelve observed behaviours. The two-tailed probability that

this number of significant statistics could occur at the 5 per cent level by chance alone is. 0002 (Wilkinson, 1951), therefore the number is sufficiently great to indicate a non-chance occurrence.

TABLE 4.37 Observations of behaviour in experimental and control mothers with their infants (aged ± two weeks) in a set situation at first interview (N = 36)[x]

	Tied pairs		Differing pairs		chi-square *	p*
Behaviour	++	--	+-	-+ **		
Eye-to-eye contact	33	0	0	2		
en face position	35	0	0	1		
Smiles at baby	9	11	3	13	5.06	<.05
Talks to baby	8	14	7	7		
Sings to baby	0	32	3	1		.36
Kisses baby	1	27	2	6		.28
Holds baby close	21	2	1	9	4.90	<.05
Strokes baby	13	9	2	10	4.08	<.05
Places baby securely on bed	23	1	2	6		.28
Names baby	1	28	2	5		.45
Keeps baby clean	35	0	0	1		
TOTAL SCORE	4	0	7	25	9.03	<.01
Asks questions about baby	2	27	7	0	5.14	<.05

[x] Pairs in which one member responded `Don´t know´ were excluded
* McNemar´s test for comparison of frequencies in matched samples (Binomial tests were used where expected frequencies were = or <5)
**++ behaviour present in both experimental and control subject
-- behaviour absent in both experimental and control subject
+- behaviour present in experimental subject absent in control
-+ behaviour absent in experimental subject present in control

The total score for each experimental subject was compared to the matched control subject and the results showed that there was a significant difference between the groups. The group of mothers of albinos had significantly lower scores compared with the control mothers, indicating that the mothers of albinos showed fewer observed behaviours in the set situation than the mothers of normal babies.

There were three specific behaviours among the total of eleven, that indicated significant differences between the two groups. These behaviours were `smiles at the baby´, `holds the baby close´, and `strokes the baby´,

and they were observed significantly less often in mothers of albinos babies, than in the mothers of normal babies.

The last item which was analysed separately and concerned whether or not the mother asked questions about her baby, also showed a significant difference between the groups ($p < .05$). More mothers of albino babies asked questions than did mothers of normal babies, the numbers in each group being fourteen and four respectively. The experimental group asked questions relating to their babies' colour, future and problems.

(xi) Summary of findings at the first interview

In the total of fifty-one items investigated at this interview there were thirteen significant differences (at the 5 per cent level) in the responses made by the two groups. The probability of obtaining thirteen significant statistics in a set of fifty-one on the basis of chance alone is .0088 (Wilkinson, 1951), so that these results are unlikely to have have been due to chance.

The significant findings may be summarized as follows:

(1) Medical History. There were significant differences on two items: more mothers of albino babies than mothers of control babies claimed to have headaches and to feel depressed. There were no significant differences between the groups, in either childhood history, family patterns, rural or urban origins, age, education or marital status, which might have explained these findings.

(2) Neonatal period. There were two items which produced data indicating significant differences between the groups: there were more mothers in the experimental group than in the control group, who did not want to hold their babies shortly after the birth, nor to breast-feed their babies at this time.

(3) Plans for the baby. There was a significantly greater proportion of mothers of albinos than of controls who planned to leave town and return to the rural areas, and who expressed complaints about their babies, stating, inter alia, that they did not like the infants' hair and skin colour.

(4) Maternal response. There was a significantly greater proportion of mothers in the experimental than in the control group who stated that they did not like the way the baby looked, that they felt unhappy about the birth, that they had no friends, that they planned to give their affected babies more education than their normal children, and that they would not like their next baby to be like the present one.

(5) Paternal response. Only a minority of mothers of albinos said that the fathers of their babies were happy about the birth, and there were significantly more subjects in the control group expressing this feeling. More experimental than control mothers stated that they had complaints about their husbands and this item also showed a significant difference between the groups.

(6) Observations. In comparison with the controls fewer specific maternal behaviours were observed in mothers of albino babies, while the mothers were changing their babies' napkins. There was also a smaller proportion of mothers of albinos, than mothers of controls, who smiled at their babies, held them close and stroked them.

The findings based upon the mothers' reports on the siblings' reactions can be expressed as impressions only, as the number of siblings in the two groups were not large enough for statistical tests to be applied to the data. Some of the mothers of albinos stated that their children were afraid to handle the affected child, were not happy with their new sibling, had said that the child was a white child, and did not show the baby to their friends, because it was an albino. Whereas the majority of the mothers in the control group said that their children were happy to have a new sibling.

From these findings one can conclude that the mothers of albino babies in this sample reacted differently to their babies in several ways, in comparison with the the mothers of normal babies. They felt depressed and unhappy about their babies, initially did not want the close contact connected with holding and breast-feeding them, and disliked their appearance. On the other hand they did not desert or totally reject the baby and some planned to provide extra education possibly to compensate for the defect. Their behaviour in interaction with their new-born babies could be observed to differ from that of the controls and they exhibited fewer behaviours in the set situation selected for the purposes of this study.

4.3.2.2 Interview 2

The interval between the first and the second interview was about three months and during this time four mothers in each group dropped out of the study themselves or were dropped by the writer. In the experimental group one mother had sent her albino baby to relatives in the rural areas, one had left with her baby for the rural areas, one lived too far away for regular visits and interviews to be feasible, and one mother's baby was too old and

she was excluded so that the group would be more homogeneous. In the control group three mothers, who were matches for the experimental mothers who dropped out, had to be excluded. One control mother was retained, although her matched experimental mother had dropped out, because she also provided a good match for another experimental mother, whose paired mother had returned to work and could not be traced. In this way the maximum number of matched pairs possible continued in the study. Altogether there were thirty-three pairs who were interviewed at the second stage of the study. There were however only thirty-two pairs who took part in the observation study, as it was not feasible to do the observation on one subject at the time of the interview.

The results of the data collected at the second interview, particularly on those items found to show significant differences between the groups at the first interview, as well as on the other items which showed significant differences, appear in Table 4.38. Several results in this interview were different from those found at the first interview and changes had occurred. There was no longer a significant difference between the groups regarding headaches and depression nor about having no friends. In connection with the plans for the baby, the experimental and control mothers gave similar responses concerning education at the second interview. Complaints about the fathers of their babies were also similar in both groups.

Significantly more experimental than control mothers, however, still claimed to feel very disturbed about their babies and reported that the fathers were also still unhappy. A greater proportion of experimental mothers than control mothers also stated that their babies would have more problems than other babies, planned to leave town with their babies, added that they would prefer the next baby not to be like this one, and that this infant cried too much.

The results obtained from the maternal-infant observations showed that there were no longer significant differences between the two groups on the total behaviour score or on any specific behaviour observed. The two groups showed similar scores for the behaviours relating to smiling at the babies, holding them close and stroking them, all of which had shown significant differences between the groups at the first observation session.

4.3.2.3 Interview 3

The third interview took place at a time as close as possible to the date on

TABLE 4.38 Comparisons between experimental and control mothers' (matched pairs) responses to dichotomous items at 6 interviews (significant chi-square values) $

	Interview	1	2	3	4	5	6
	Age of baby	2 wks	3 mo.	6 mo.	9 mo.	12 mo.	15.mo.
	No of pairs	37	33	22	18	17	15
	Experimental group response						
MEDICAL HISTORY							
Headaches	+	4.27*	NS	NS	NS	NS	NS
Depression	+	5.14*	NS	NS	NS	NS	NS
INFANT CARE							
Want to hold baby	-	12.19***	N/A	N/A	N/A	N/A	N/A
Want to breast-feed	-	4.76*	NS	NS	NS	NS	NS
Extra solids	+	N/AP	N/AP	N/AP	6.12*	NS	NS
Leaving town with baby	+	4.17*	4.0*	NS	NS	NS	NS
Problems with baby	+	4.17*	5.14*	7.11**	5.14*	6.12*	5.14*
More education	+	4.9*	NS	NS	NS	NS	NS
Baby cries too much	+	NS	4.0*	NS	NS	NS	NS
MATERNAL RESPONSE							
Unhappy/Disturbed	+	8.45**	11.08***	4.16*	NS	NS	NS
No friends	+	5.14*	NS	NS	NS	NS	NS
Next baby different	+	23.04***	5.78*	7.69**	9.09**	6.12*	9.09**
Grandmother's support	+	N/A	N/A	4.0*	4.16*	NS	NS
Complaints about father	+	5.14*	NS	N/A	N/A	N/A	N/A
PATERNAL RESPONSE							
Father unhappy	+	12.04***	6.75*	4.16*	NS	NS	NS
Supports baby		NS	NS	NS	NS	NS	NS
BABIES' MILESTONES							
Smiles		N/AP	NS	NS	NS	NS	NS
Rolls over		N/AP	NS	NS	NS	NS	NS
Sits	-	N/AP	N/AP	NS	NS	4.92*	NS
Crawls	-	N/AP	N/AP	NS	NS	5.78*	NS
Walks		N/AP	N/AP	N/AP	N/AP	NS	NS
OBSERVATIONS ON MOTHERS (NAPKIN CHANGING ACTIVITY)							
Smiles at baby	-	5.06*	NS	NS	NS		
Holds baby	-	4.9*	NS	NS	NS		
Strokes baby	-	4.08*	NS	NS	NS	No observations	
Total behaviours	-	9.03*	NS	NS	NS		
Asks questions	+	5.14*	NS	NS	NS		

$ obtained by McNemar's test for comparing frequencies in matched samples (for raw data see appendix K, Tables I, II, III, IV)

+/- + = increased, - = decreased (compared to controls);

* = significant difference, * p <0.05, ** p <0.01, *** p <0.001;

NS = not significant; N/A = not asked; N/AP = not applicable

which the baby reached six months of age. It was between the third and sixth month of the child's life that there was a large reduction in the numbers of subjects. In one third of the matched pairs one or other subject either dropped out, or had to be excluded. There were four subjects who left for the rural areas, and two subjects had babies who were too old and they were dropped to make the group more homogeneous. One mother of an albino adamantly denied that her baby was an albino and her mother telephoned to say that she refused to co-operate any further. Another mother could not be located and had to miss this interview, a further three subjects had to be dropped because their controls had withdrawn. The result was that the number of matched pairs was reduced from thirty-three to twenty-two in total.

The results on selected items which showed differences between the groups at the first interview are shown in Table 4.38. Possibly because so many of the subjects who had intended to leave town had left, it was noted at this stage that there was no significant difference between the groups regarding plans for moving out of town. Results also indicated that there were no longer more mothers of albinos than controls complaining that their babies cried too much. A significant difference was found in the responses to the new item which concerned whether the mothers' mothers gave them moral support. More mothers of albinos stated that they received such support from their mothers than did controls.

The significant differences that remained at this stage were those related to the responses concerning the disturbed feelings of the experimental mothers and fathers, compared with the controls towards their babies. In the experimental group a large proportion of both parents (according to the mothers) professed these disturbed feelings. A significantly larger group of mothers of albinos than mothers of controls also continued to complain that their children would have problems and stated that they would prefer the next child not to be the same as the present infant.

The scores on the observations of the two groups of mothers and babies were similar and there were no significant differences for any of the behaviours observed, which confirmed the findings of the previous interview.

4.3.2.4 Interview 4

When the babies reached nine months of age the fourth interview was carried out with the same schedule as that used at the third interview.

The numbers of pairs had been further reduced from twenty-two to eigh-

teen in total. There was one mother of an albino who returned to the rural areas and one missed the fourth interview. There were also two subjects who had to be dropped as in one case the matched control had returned to work and could not be contacted, and in the other the control was unco-operative and refused to continue with the interviews.

The results of selected items from Interview 4 appear on Table 4.38. At this stage some of the mothers of albinos stated that they did not feel as disturbed about their babies as they had previously, and that they had accepted their infants. There was therefore no longer a significant difference between the two groups on this item. It is possible that those who were most disturbed had left for the rural areas to obtain support from their families or to leave their child there out of the public eye, but nevertheless there were many who admitted that they had been unhappy, but were now resigned to their lot. A larger proportion of mothers of albinos also indicated at this stage that the fathers were happy about the birth and there was no longer a significant difference between the groups on this item.

Significantly more mothers of albinos than controls continued to report that their babies would probably have more problems than other children, that they would prefer the next child not to be the same, and that they obtained moral support from their mothers. In addition, the results showed that the mothers of albinos claimed to give their babies a wider variety of solids than did the mothers of the normal babies. Bottled baby foods, as well as fruit, were added to the diet of many of the albinos, whereas most of the controls were still on the staple diet of porridge alone.

The last observation in the napkin-changing activity was made at this stage, since it was found that mothers proposed to take their babies out of napkins when they started walking. Again, confirming the findings in the second and third interviews, there were no significant differences between the groups on scores for the behaviours selected for observation.

4.3.2.5 Interview 5

This interview was conducted when the babies were one year old. There were two albino babies who died between their ninth and twelfth month of life. One baby died as a result of septicaemia after an enema which was given to her by a baby-minder, and the other died as a result of neglect leading to gastroenteritis. There was also one control mother who was arrested and imprisoned. It was possible by changing around some controls who were reason-

ably good matches for more than one subject to retain seventeen pairs in the sample at this stage.

The results on selected items from Interview 5 are presented in Table 4.38 and compared with those for the other interviews in the same table. There was no difference between the groups on the variety of solids given to the infants at this stage. Regarding grandmothers' support, there was no longer a difference between the groups on this item either. It is possible that since the mothers had apparently started accepting their albino babies they did not need, or did not comment upon, the support given them by their mothers.

There were, however, differences in the achievement of milestones of the babies in the two groups at this stage. The results indicated that, according to the information on milestones obtained from the mothers, a significantly larger proportion of albinos babies sat and crawled later than did the control babies.

Some mothers of albino babies continued to report at this interview that they thought their babies would have more problems than other normal children, and that they would prefer it if the next baby were not the same as the present one. On both these items the differences between the groups remained significant.

4.3.2.6 Interview 6

The last interview was carried out with the mothers when the babies reached the age of fifteen months. The number of subjects was further reduced from seventeen to fifteen pairs by the departure of two mothers of albinos and two controls to the rural areas. Again it was possible by transferring controls who matched several experimental subjects sufficiently well, to retain as many pairs as possible.

As may be seen on Table 4.38 there were very few significant differences between the groups at this stage and it might be said that the study could have been terminated after interview 5. On the other hand the fact that the conclusions from the two successive interviews were similar, could indicate that stable and reliable results were obtained. A significant proportion of mothers continued to state that their albino babies would have more problems than normal children, and this was a realistic approach since they frequently mentioned skin and eye problems. Although there were several subjects who mentioned that they would like to have another albino child to keep the in-

dex case company, or that they would accept whatever God gave them, the majority of the mothers of albinos said they would like their next child to be normally pigmented.

4.3.2.7 Comparison at the first and sixth interviews of results from two sub-samples of subjects, one in the experimental and one in the control group

Since one of the major purposes of the study was to determine the nature of changes over time in the mothers of albinos and in the mothers of normal babies, the responses of a sub-sample of experimental subjects (those who participated throughout the study) to certain selected items at the beginning and end of the study were compared. An identical `before and after' study was carried out on the controls. The results from the first and sixth interview, of each of these sub-samples of fifteen subjects (in both experimental and control groups) are presented in Table 4.39 for the experimental group and in Table 4.40 for the control group. The scores on the observations were compared within the two separate groups at stage 1 and stage 4, (when there were sixteen pairs in the study) since no further observations were made thereafter.

Although there was no difference between the numbers of experimental or control subjects complaining of headaches or depression at the first as opposed to the sixth interview the reasons given at the two stages were diffeent. At the first interview some experimental subjects stated that they were depressed because of their albino baby and at the sixth interview those who were depressed said it was because they could not find employment, or the father was not paying maintenance, or they had housing problems.

There was a significant difference, between the first and sixth interview, within both groups, on the infant-feeding items since many had ceased breast-feeding by the time the infant was fifteen months of age. A comparison was also made between responses of the mothers of albinos concerning the perinatal period (a large proportion stated that they did not want to breast-feed their babies immediately), and their responses concerning how they were feeding at the first interview (when most of them were found to be breast-feeding, as were the control mothers). There was a significant difference between the two stages on this item within the albino group (chi-square 7.11, $p<.02$), but not within the control group. This finding indicated that although many of the mothers of albinos felt that they did not want to breast-feed their babies soon after the birth, they either changed their attitude

towards the baby or were forced to do so by circumstance, and within two weeks they were feeding their infants in this way.

TABLE 4.39 Comparisons for experimental subjects on selected item responses (N = 15) and observed behaviours (N = 16) at first and last interview

	Tied Responses		Differing responses		p value$
	++	--	+-	-+*	
Medical History					
Headaches	2	6	5	2	
Depression	2	8	2	3	
Infant Care					
Breast feeding	8	1	6	0	0.03**
Problems with baby	2	6	1	6	0.12
Maternal Response					
Unhappy	2	6	7	0	0.02**
Shows baby to friends	6	0	0	9	0.004**
Next baby different	5	6	4	0	
Paternal Response					
Unhappy	3	6	5	1	0.21
Observed Behaviour					
Smiles	3	7	0	6	0.03**
Holds baby close	8	2	3	3	
Strokes baby	6	1	2	7	0.18
Total behaviours	1		3	12	0.03**

$ obtained by Binomial two-tailed test for small matched samples (Siegel, 1956)

* ++ = condition present at both first and last interview
-- = condition absent at both first and last interview
+- = condition present at first, absent at last interview
-+ = condition absent at first, present at last interview

** significant differences

One further item should be mentioned and this referred to the mothers' claims to show their babies to their friends. From the figures presented in Table 4.39 and 4.40 it can be seen that there was a significant change over time in both groups and that neither displayed their babies at the beginning, but the majority of both groups did so at the end of the study.

The only items where a response, which differed significantly between the groups, was retained from the first to the last interview, were those concerned with the extra problems of the albino babies and the desire by mothers of albinos to have a different baby next time.

TABLE 4.40 Comparisons for control subjects on selected item responses (N = 15) and observed behaviours (N = 16) at first and last interview

	Tied Responses ++	Tied Responses --	Differing Responses +-	Differing Responses -+*	p value$
Medical History					
Headaches	0	9	1	5	0.22
Depression	0	11	0	4	
Infant Care					
Breast feeding	4	0	11	0	0.006**
Problems with baby	0	15	0	0	
Maternal Response					
Unhappy	0	11	4	0	
Shows baby to friends	7	0	0	8	0.008**
Next baby different	0	10	3	2	
Paternal Response					
Unhappy	3	9	2	1	
Observed Behaviour					
Smiles	6	2	2	6	0.29
Holds baby close	13	0	3	0	
Strokes baby	8	2	4	2	0.68
Total behaviours	6		4	6	0.75

$ obtained by means of Binomial two-tailed test for matched samples (Siegel, 1956)

* for explanation of these symbols see Table 4.39 footnote

** significant differences

The results of the observation study support those which indicated the decreasing unhappiness in mothers of albinos over time. Significantly more maternal behaviours were observed during the interaction with the infant at the final session (at nine months) than at the first observation. Specifically, a significantly greater proportion of mothers were observed to smile at their albino babies at the last observation than at the first. The control mothers however showed no significant change in numbers of counted behaviours from the first to the last observation.

The most dramatic change from the first to the sixth interview was that related to the mothers' feelings towards their albino babies. The data concerning these feelings of unhappiness are presented in Table 4.41. A significantly smaller proportion of mothers of albinos felt unhappy and disturbed at the fourth and sixth interview, in comparison with the proportion at the

first interview. Although there were two mothers in this group of fifteen who maintained throughout the study that they were upset about their babies,

TABLE 4.41 Feelings of unhappiness in experimental and control mothers (N = 15) at the first compared to subsequent interviews

Interviews	Tied pairs		Pairs showing differences			
	++	--	+-	-+*	chi-square $	p
Experimentals						
1 vs 2	7	4	2	2	0.25	>0.05
1 vs 3	4	5	5	1	1.50	>0.05
1 vs 4	2	6	7	0	5.14	<0.05**
1 vs 5	2	5	7	1	3.12	>0.05
1 vs 6	2	6	7	0	5.14	<0.05**
Controls						
1 vs 2	0	11	4	0	2.25	>0.05
1 vs 3	0	15	0	0		
1 vs 4	0	15	0	0		
1 vs 5	0	15	0	0		
1 vs 6	0	15	0	0		

$ = obtained by means of Binomial test for matched samples (Siegel, 1956)
*++ = unhappiness present at both interviews
-- = unhappiness absent at both interviews
+- = unhappiness present at first absent at subsequent interview
-+ = unhappiness absent at first present at subsequent interview
** = significant difference

the majority of mothers appeared to move from a state of disturbance and unhappiness to one of acceptance, some taking longer than others, but most reaching the latter stage at about nine months after the birth. This change has been indicated graphically in Fig 4.8 in which the proportion of mothers and fathers in both groups showing feelings of unhappiness at each stage of the study is presented.

As can be seen in Fig 4.8, the fathers' feelings, as perceived by the mothers, also showed some change over time. The proportion of fathers reportedly showing change was not, however, significantly different at subsequent interviews in comparison with the first interview within the experimental or the control group over the fifteen month period. The number of fathers present in the home was very small and in fact may have been too small to show changes to any significant degree. Two fathers in both groups

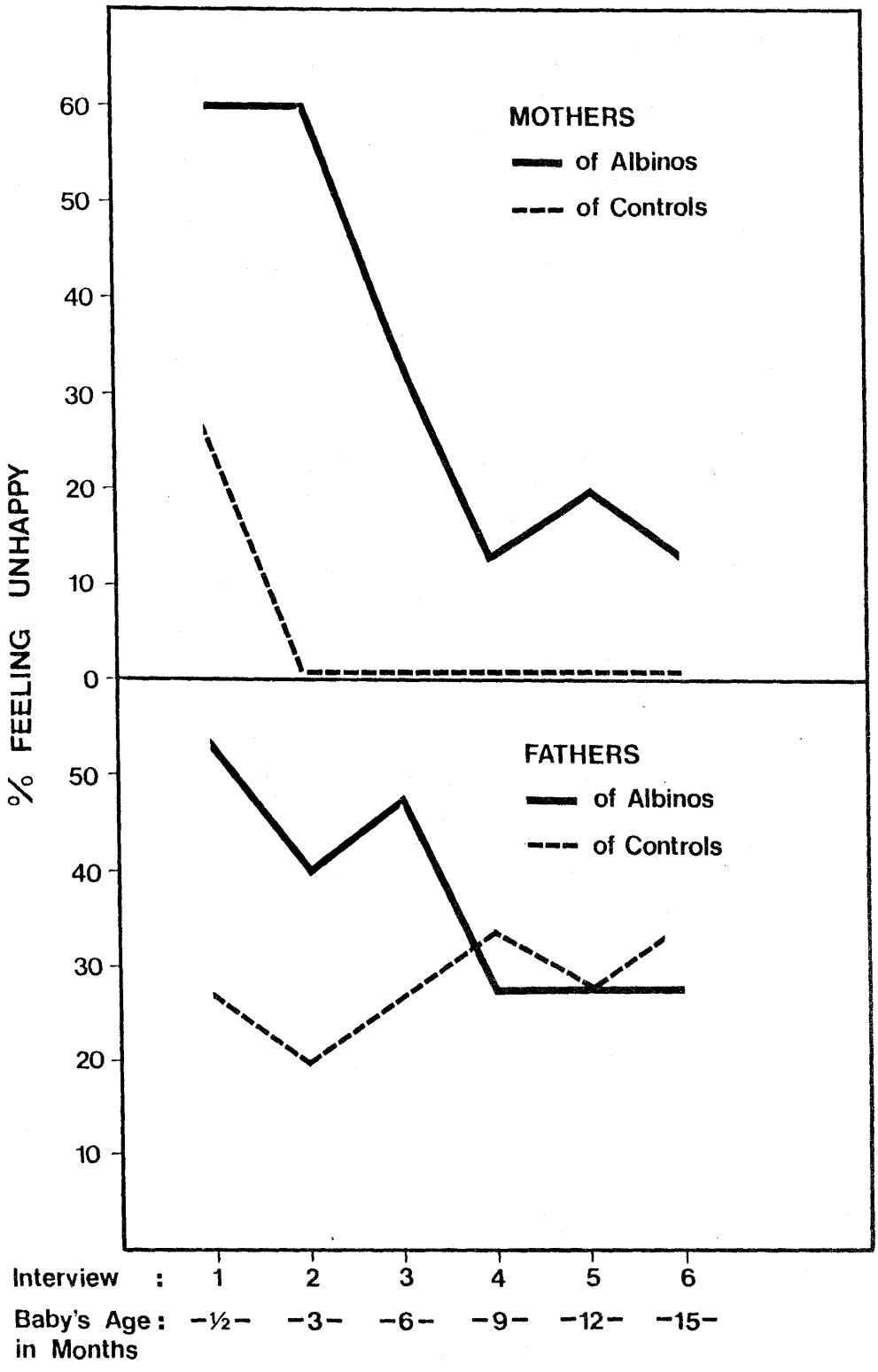

Fig 4.8 Feelings of unhappiness (according to mothers) in mothers and fathers of albino and normally pigmented babies

denied paternity, so the defect did not seem to cause more outright paternal rejection than would normally occur in this society. There were four control mothers and one experimental mother who stated that they were separated from their husbands.

In summary then, the mothers of albinos appeared to have reverted to 'normal' and to show similar responses and behaviours to the mothers of controls, by the time their babies reached fifteen months. Many of the changes that took place in the experimental mothers occurred by the time the babies were nine to twelve months of age. The control group as a whole consisted of mothers who were slightly younger, more urbanized, and more often single than the experimental group. These factors could have caused the controls to show less attachment to their babies and more disruption in their maternal-infant relationship. The fact that there were differences between the experimental and control groups therefore takes on added significance and suggests that the birth of an albino to a black mother causes a disturbance in the normal maternal pattern of functioning, which can be detected even in comparison with a relatively unstable control group.

It was necessary to compare the fifteen experimental and control dyads who participated throughout the study since the changes reflected over time in the comparisons between the larger total of matched pairs (as presented in Table 4.38) might have been said to have been due to the dropping out of the study of those subjects who were most disturbed by the birth of an albino baby. The results of the investigation of these fifteen pairs indicated that there was change over time even within this small sub-sample. Significantly fewer mothers of albinos expressed feelings of unhappiness, by the end of the study, about having an affected baby, and significantly more maternal behaviours were observed at the last observation session than at the first, whereas in the control group no such changes occurred. These findings therefore confirm those resulting from the analysis of all the subjects' responses in the main study.

4.3.2.8 Observation of maternal-infant interaction in a play situation

The observation of the mothers and babies at play was made after the last interview when the babies were about fifteen months old. There were seven specific behaviours of the mothers and of the infants which were noted (see Appendix J) and the number of times they occurred in the ten minute observa-

tion session was counted. Although initially a white (the writer) and a black observer both made the observations and counted the behaviours, it was found that since the white observer did not fully understand the language it was difficult for her both to separate and count the vocalizations, and, because of the cultural differences, to interpret the meaning of the behaviour in certain circumstances. Also one infant, whose mother had used threats that white people would punish him, refused to leave his mother's arms or to play at all in the presence of the white observer and the session had to be repeated later in the presence of the black observer alone. For these reasons it was decided that only the scores of the black observer should be used in the analysis of the results of these observations.

The findings from these results are shown in Table 4.42. Statistical tests were not carried out on this material since the numbers in each group (seven) were so small. From this purely descriptive data the mothers of albinos tended to hold their babies and talk to them slightly more, but to gaze at them and mediate their environment less than the control mothers did. The experimental mothers also behaved in a way which was more contingent to the needs of the infant than the controls did, and the albino infants showed fewer `fuss' cries and less distress in the play situation than the control babies. The differences between the groups however was very small and only general observations on the results can be made.

Although there was much movement of mothers and babies during the observation, the mean maximum distance at which mothers of albinos placed themselves from their babies (50.4 cm) on the play mat was nearly double the mean maximum distance between mothers and their black babies (27.6 cm). Other behaviours which might also suggest some distancing between the mother and her albino child, included less maternal mediating behaviour, less gazing, and also fewer emotional fuss or distress reactions, suggesting fewer demands for attention, on the part of the albino babies, in comparison with the controls.

Although it was difficult to observe both mother and baby, and the observer tended to look first at the mother, the information obtained on the babies is worth a comment. From Table 4.42 it can be seen that the babies' behaviour in both groups followed very closely the pattern of the mothers' behaviour. The babies in both groups touched their mothers more often than the mothers touched their babies, but otherwise the more the mothers smiled, gazed at the baby and vocalized, the more the babies behaved in the same man-

ner. For example, the experimental group vocalized more often with their babies, and the babies responded with a higher number of vocalizations than the babies in the control group. Similarly, the experimental mothers gazed less at their babies and the babies in response gazed less than the control babies, whose mothers gazed more frequently at them.

TABLE 4.42 Maternal-infant interaction in experimental and control dyads in a play observation situation (seven matched pairs)

	Mean scores*			
	Experimental group		Control group	
	Mothers	Babies	Mothers	Babies
Maternal-infant interaction				
Touch	2.86	3.14	2.86	3.57
Hold	0.71	0	0.28	0
Smile	6.14	4.43	6.00	3.57
Gaze	2.14	0.57	2.57	0.71
Vocalization	22.28	11.71	18.85	8.00
Maternal behaviour				
Mediating environment	9.28		11.57	
Not contingent to infant	0.57		2.00	
Infant behaviour				
Distress		0.14		1.00
Fuss cry		0.43		1.28
Maximum distance				
Between mother and infant	50.4cm		27.6cm	

* based on number of times each behaviour was observed

No conclusions can be drawn from this information because of the small sample size, but nevertheless the trends described above are worth reporting as a base on which future research may be developed.

4.3.2.9 Anxiety scale

In order to gather additional data on the anxiety state of the mothers, a standardised anxiety scale was given to them after the final interview and the play observation. Again, since the matched groups were so small (seven subjects in each), no statistical tests were carried out on the data collected. The mean score (the higher the score the higher was the level of anxiety) for each group was calculated and the score for the mothers of albinos was slightly higher (9.28) than the score for the control group (8.71). If

however, the additional four mothers of albinos who completed the scale (but who were initally excluded because their matched controls did not complete the scale) were included, then the mean for the experimental group was even higher (11.27). It is therefore at least possible that the level of anxiety in the mothers of albinos was generally higher than that of the mothers of controls and that, with a larger sample, this finding might be confirmed.

4.3.3 Results of data obtained from interviews and observations sessions with fathers

Initially it was intended that the fathers of albino babies should be interviewed three times, when their babies were three, nine and fifteen months old, i.e. at the same time as the mothers' second, fourth and sixth interviews. However at the mothers' second interview it became apparent that almost half the sample were unmarried, that many were not living with the fathers of the baby, that in some cases the fathers had never seen the baby, in others they had denied paternity, and in still others they were inaccessible or living far away. It was obvious therefore that it would be very difficult both to obtain a good sample of fathers and to involve them in a longitudinal study. It was therefore decided that the interviews with fathers should be limited to one, to be conducted as close to the mothers' second interview as possible, when the baby was three months of age.

Even this limited goal was difficult to attain and only ten of the thirty-seven fathers of albinos and three of the thirty-seven controls were interviewed. The schedule used for the interview was very similar to that used for the mother's second interview except for the minor adaptations required for its use with the fathers (see Appendix G).

Of the ten available fathers, nine were observed in interaction with their babies and in the tenth case the mother and baby were in hospital, so only the interview schedule could be completed with the father. The list of behaviours used in the observation (see Appendix I) was again adapted from that used with the mothers, but the activity differed. The fathers were requested to pick up and hold their babies, while the interviewer made some notes on the features of the baby. The interviewer observed the fathers' behaviour and checked off on the list whether or not the specific behaviours occurred.

The sample of fathers who were interviewed and observed consisted of those who were co-operative, self-selected and accessible and as such the sample was probably biased towards those fathers who were accepting and par-

ticipating in their situation. Due to the small size of the sample the data collected were not amenable to analysis by means of statistical tests, the results therefore are presented as observations and should be considered as descriptive material only. The information is also useful to supplement that given by the mothers on the paternal reaction to the birth of the affected baby.

Due to the difficulty in obtaining the fathers' co-operation the babies had reached different ages by the time the interview and observation session were completed, as indicated on Table 4.43. This fact certainly influenced the results since some fathers had had more time to adapt to their babies than others.

TABLE 4.43 Age of babies at father's interview

Age of baby	Experimental group	Control group
1 week	1	0
3 months	4	1
6 months	2	1
9 months	1	0
12 months	2	1
TOTAL	10	3

4.3.3.1 Results from the interview

The fathers' responses on selected items are shown in Table 4.44. In the section on medical history there were five of the ten experimental fathers and two of the three control fathers who reported feeling depressed. In none of the experimental fathers was the reason for depression said to be the albino baby. Three of these fathers stated they did not know why they felt depressed, one said he had financial difficulties and another that his wife was in hospital. The reasons the two control fathers gave for their depression was `permit problems' in one and `thinking of someone' (not specified) in the other.

Regarding special problems that their babies might have, two fathers of albinos responsed realistically that they would have eye and skin problems, another father said the albino child would not have problems because she would be well cared for by her parents. There were two fathers who thought their albino child should have more education than other children and one father stated specifically that the child required such education `because she is an albino'. These findings complement the results found at the ini-

tial interview with the mothers of albinos, among whom there was a significantly greater proportion who said their child should receive more education, than there was among the control mothers.

TABLE 4.44 Experimental and control fathers' responses to schedule items

	Experimental group (N = 10)			Control group (N = 3)		
	Yes	No	D.K.$	Yes	No	D.K.$
Medical history						
Headaches	2	8		1	1	
Fractures	0	10		2	1	
Depression	5	5		2	1	
Hospitalization	3*	7		1**	2	
Infant care						
Problems	2	8		0	3	
More education	2	6	2	0	3	
Paternal response						
Disturbed	2	8		1	2	
Next baby different	2	2	6	1	2	
Baby is like myself	6	4		1	2	
Plans for another baby						
Father	7	2	1	1	2	
Mother (according to father)	6	1	3	1	1	1
Asked questions about baby	6	4		0	3	
Maternal response						
Disturbed	0	10		1	2	
Marital relationship						
Good	10	0		3	0	

$ D.K. = don't know
* for injury from a car (one), assault (one), and a hand injury (one)
** for an ankle injury

When asked if they felt disturbed about the baby, two out of the ten fathers of albinos said they did, one because of the family's reaction, and the other particularly when the child was sick. The others denied any feelings of disturbance and unhappiness, although the experimental mothers' view of the paternal response was that many were disturbed, and there was a significant difference on this item between the experimental and control mothers' responses at the first, second and third interview, when the baby reached six months. The mothers' and fathers' responses to this item however are not

strictly comparable since in half the cases of fathers the babies were between six and twelve months of age at the time of the interview, and changes in feelings could have occurred by this time. One control father also commented that he was disturbed because his baby's one eye appeared to be smaller than the other.

The fathers' view of the maternal response to the item on feeling disturbed was also anomalous, since none of the experimental fathers admitted that the mothers of their babies felt disturbed about the baby, although a large proportion of the mothers stated that they themselves felt disturbed, for at least six months after the birth. Again this apparently inconsistent finding may have been due to the time at which the fathers were interviewed, and/or to the possibility that their perception was inaccurate, or that this particular sample of men had 'wives' who had accepted their babies well.

The fathers gave various responses to the item on whether or not they preferred the next baby to be different. There were two fathers who wanted normally pigmented babies (one because 'albinos need a lot of care') and two fathers who wanted further albino babies (one because he already had two, and the other because he said he had always wanted an albino and would now like another). Then there were six fathers of albinos who said they did not know whether or not they wanted the next baby to be different (one was indifferent, and five said they would accept 'God's gift' of an affected or unaffected child). By comparison many of the mothers wanted the next baby to be normally pigmented and there was a significant difference between experimental and control mothers, until the end of the study when the babies reached fifteen months. There were six of the ten fathers of albinos who stated that the albinos looked like themselves, suggesting a measure of acceptance of and identification with the child and perhaps a certain amount of attachment.

The majority of the fathers were planning further children. Only two fathers did not want more children and they gave as their reasons 'the cost of living' and 'the wife's poor health', and made no mention of albinism. According to the fathers, the majority of the mothers of their babies also wanted more children. These data confirms the findings on the mothers; there was no significant difference between the mothers of albinos and the controls on plans for future children, and the majority wanted more children.

Another indication that the group of fathers interviewed may have been a biased group was that six of them asked questions about their babies at the

end of the interview and appeared concerned and involved. There were two who requested information on the causes of albinism, and the four others asked about nystagmus, hair-bulb tests, the right food to give the baby, and the nature of the research project. By comparison none of the control fathers asked questions.

4.3.3.2 Results from the observation session

The results concerning three of the observed paternal behaviours which showed differences, together with the total behaviour score, are reflected in Table 4.45.

TABLE 4.45 Observations on fathers' behaviour

	Experimental group (N = 9)		Control group (N = 3)	
Behaviour	Yes	No	Yes	No
Names baby	4	5	2	1
Strokes baby	7	2	3	0
Kisses baby	4	5	2	1
Total behaviour score*: Mean	13.55		14.00	

* Maximum score possible = 18

Again, since the numbers are so small especially in the control group, no statistical tests could be performed and general comments only can be made. There were two of the fathers who did not stroke their infants, and significantly fewer mothers of albinos stroked their infants than did the mothers in the control group. Only four of the nine fathers named and kissed their babies during the observation, whereas in the control group the numbers were two out of three in each case. The experimental father, who showed the fewest behaviours and lowest score (twelve out of eighteen points), stated that, although the child was already nine months old, he was still afraid to hold the baby and never did so. This father also admitted to feeling depressed and occasionally feeling disturbed about the baby.

The mean total behaviour score for the two groups showed that there were slightly fewer behaviours in the fathers of albinos group, but the difference was very small and no conclusion could be drawn.

4.3.3.3 Summary of findings on fathers

The study of the ten experimental and three control fathers showed certain trends some of which supported the findings on the study of the mothers. As

a group the experimental fathers (interviewed on average 5.7 months after the birth of the albino baby), in general, did not admit to feelings of disturbance in themselves or the mothers of their infants. The majority of the fathers added that they and most of the mothers were planning more children and this confirmed the findings from the mothers' study. Although a large proportion of fathers did not admit to wanting the next baby to be different (which many mothers stated openly) there was a large group who were indecisive. Many of the fathers asked questions about their affected baby. In the observation situation there was very little difference between the experimental and control groups for mean number of behaviours, except that only about half the group of fathers of albinos named and kissed their babies. These results suggest that these fathers were generally quite accepting of their albino babies, but they must be treated with much reserve and merely as descriptive data which give some insight and are useful for amplifying the more extensive information collected from the mothers of albinos.

4.3.4 Summary and concluding comments

Results have been obtained from the analysis of the data on some aspects of the impact of the birth of an albino baby, primarily, on the mother, secondarily on the father, and, from the mothers' perspective, on the siblings and grandmothers.

Those findings that showed significant differences between the groups indicated that immediately after the birth of the baby a greater proportion of experimental than control mothers were reluctant to hold and breast-feed their babies. When observed handling their infants about two weeks after the birth, the experimental mothers as a group showed fewer maternal behaviours than the controls. Specifically, a smaller proportion of experimental mothers smiled at, stroked, and held their babies close than did the control mothers. At this early stage the mothers of albinos were significantly more likely than controls to complain of depression and headaches and to state that they were disturbed and unhappy. Several experimental subjects planned to leave town and to join their families in the rural areas. Regarding their babies, a significant proportion of the mothers of albinos stated realistically that the affected babies would have more problems than other children, that they would give them more education, and that they would prefer the next baby to be different. Regarding the fathers of their babies, significantly more experimental than control mothers reported that these men were

also disturbed and unhappy. The experimental mothers as a group had more general complaints about these men than the control group had about the fathers of their babies.

By the time their albino babies had reached three months some aspects of the mothers' behaviour and some of their attitudes had apparently changed (as evidenced by the items which had shown statistical differences at the first interview , but which no longer showed such differences between the experimental and control groups at the second interview). The majority of the experimental mothers were breast-feeding their babies and, at observation, were seen to smile at, stroke and hold their babies closely, to the same extent as the control mothers did. Although most of the group no longer complained of headaches and depression a significant number of experimental mothers continued to state that they felt disturbed and unhappy and some still planned to leave town. More experimental than control mothers added that the babies would have many problems, that they cried too much, and that they would prefer a `normal' baby next time. Significantly more mothers of albinos than controls felt that the fathers were still unhappy and disturbed about the baby.

There were further changes six months after the birth of the albino baby. Most experimental mothers continued to breast-feed their babies and on observation, to behave as the controls did. A significantly greater proportion of the mothers of albinos than controls stated that their babies would have more problems than other children. However, those experimental mothers who planned to leave town had presumably done so, and those who remained in the study did not express such plans. Nevertheless significantly more mothers of albinos than controls maintained that they and the fathers of their babies were still unhappy and disturbed about the child, but that they were receiving support and comfort from their mothers.

At the stage at which the babies reached nine months there were no longer more experimental mothers than controls reporting that they and the fathers of the babies felt unhappy and disturbed, and some affected mothers said they had accepted the baby `as he is'. A significant proportion of these mothers still, however, stated that they would prefer it if the next baby was different (i.e. normally pigmented) and mentioned that the albino baby would have more problems. As a group, mothers of albinos had treated their babies significantly differently to the mothers of normal babies, by introducing them to a greater variety of solids. More experimental mothers

than controls also claimed to be receiving continuing support from their mothers.

A year after the birth of the babies there were very few differences between the experimental and control mothers. The only two items that continued to show significant differences were those concerning the albino babies' problems (more mothers of albinos thought their babies would have many problems than did the control mothers) and the desire to have a `different' baby next time (a larger proportion of experimentals than controls wanted the next baby to be different). However, in connection with the babies' progress, at this stage it was noted that albino babies had been significantly slower to sit and crawl than the controls according to the mothers' reports.

The information gathered when the baby reached fifteen months reinforced that obtained at earlier interviews and there were few changes or differences between the groups. The support of the grandmother was no longer mentioned, but the problems of the baby still bothered more of the experimental than the control mothers, and more of the former group retained the desire to have a subsequently different (implying normal) baby.

In the play observation session held at this stage, it was noted (although not statistically evaluated because of the small sample size) that mothers of albinos as a group did not sit as close to their babies as the control group did, they also appeared to vocalise more with their babies, who consequently responded with more vocalisations than the control babies, but to mediate the babies' environment less.

It is possible that the changes observed over time might have been caused by the drop-out of those experimental mothers who were feeling extremely disturbed or negative towards their babies, and therefore removed themselves to the security of their rural family, so that the impression that remained was that the group as a whole felt disturbed about the babies at first but later appeared to accept them. For this reason the findings from the responses at the sixth interview with the fifteen mothers of albinos who continued to participate throughout the study were compared with the findings on their responses at the first interview. Significant differences were found, and the majority of experimental mothers, at the sixth interview were no longer expressing the feelings of unhappiness with the baby which they had verbalized at the first interview. The mothers of albinos also showed significantly more behaviours at the last observation session (at nine months) and smiled significantly more at their babies than they had done at the first session

(at $\pm$ two months after the birth). These findings suggest that, within this sub-group, the mothers accepted their affected infants after a time (about six to nine months) and, by fifteen months, had established patterns of behaviour towards the infant which appeared similar to those of the control group, for the aspects investigated.

The data collected on the other members of the family were superficial, limited and unsuitable for statistical analysis. Nevertheless, as indications of general trends the findings are summarized here. The interview with, and observations on, the small partially self-selected and co-operative group of fathers of albinos showed that: the majority did not admit to excess feelings of disturbance regarding their child, nor that the mothers felt disturbed; most were planning future children and stated that the mothers were too; many were indecisive about whether they would like the next child to be different; and, on observation with their babies, they were less likely to name and kiss their babies than were the few unmatched controls.

The mothers reported that in general the albino babies' grandparents, where available, accepted the child, but the grandmother tended to give her daughter more comfort and support than was reported by the control mothers. The mothers also gave information on the siblings' reponse, and in some cases this was quite extreme, indicating fear and rejection in some and misunderstanding of the condition and curiosity in others. Several children were also reported to experience difficulty in touching and looking at the affected baby. The group was, however, small, since in several families the albino baby was a first child and in others the siblings were living elsewhere, and since it was not possible to use many of the matched controls no statistical tests could be applied nor reliable conclusions drawn.

In conclusion, the results of this study indicated that the birth of an albino baby affected the mother's behaviour, feelings and attitudes in several specific ways which seemed to change over time. The descriptive data on the father, grandmother and siblings suggest that they might have been sufficiently disturbed by the birth to alter their behaviour towards the baby and, in the case of the grandmother, towards the mother. The normal functioning and dynamics of the black family therefore appeared to be disrupted on several planes by the unexpected and unanticipated event of the birth of an albino baby.

CHAPTER V

DISCUSSION AND CONCLUSION

5.1 Introduction

From the analysis of the data presented in the previous chapter several significant findings became apparent. These findings will be discussed below and compared with similar findings, where these are available, from the literature. The conclusions arising from this discussion will then be reported in relation to the aims of the study. Several limitations became apparent during the collection, and analysis of the data, and these limitations and the areas requiring further research will be presented. In the writer's opinion all research should be action oriented and discussion therefore includes the various applications and recommendations arising from the study.

5.2 Discussion of the findings

5.2.1 Genetic studies

5.2.1.1 Prevalence

The urban prevalence rate for albinism found during the study in Soweto was 1:3 900. This rate is remarkably high, being approximately four times higher than that reported for many Caucasoid populations (eg. Witkop, 1983, Sanders, 1938, Pearson *et al*, 1911). It is likely to be a minimum rate, since ascertainment in Soweto was probably incomplete, and only an estimate, because the Soweto population fluctuates and is difficult to ennumerate precisely.

It is not clear why the prevalence of albinism should be raised in a sub-tropical country with high daily sunshine rates and an environment generally detrimental to the homozygote's well-being. It is possible that founder effect has contributed to the carrier rate of 1:32 in this population. It is also possible that those ethnic groups who carried the albinism gene were more likely to migrate from tropical Africa, to escape the deleterious effects of the weather and the heat of the sun experienced there, especially if their health was threatened by these factors. Alternatively, there could be some form of heterozygote advantage, such as immunity to one of the major killers in the tropics, as has been found for sickle cell anaemia. Resistance towards malaria for example, would have given carriers an advantage in the past in tropical Africa. The investigation of endemic diseases in carr-

iers is required. Another form of heterozygote advantage, was mentioned in passing by Oettle (1963) who suggested that if the light skin colour he observed in carriers was considered a sign of beauty they would have a selective advantage.

Selection in favour of homozygotes might have occurred if albinos were considered special or honoured people. This attitude does not seem to have been prevalent locally, however, since the killing of albinos in Southern Africa was reported by travellers, such as Livingstone (1857), and by Stannus (1913), and according to Turner (in Pearson, Nettleship and Usher, 1911, p144) albinos were considered as undesirable wives and husbands. Considering the information from the present study regarding possibly reduced fertility in albinos, even if they have been well cared for for many years, it seems very unlikely that selective homozygote advantage could have accounted for the high rate in the past or present. The solution to this problem seems much more likely to originate in some heterozygote advantage. Local high consanguineous marriage rates would also partly explain the prevalence rate.

Although there are many reports of albinos in Africa not many systematic prevalence studies have been performed. The available rates for the condition vary from 1 in 2 858 to 1 in 5 000 (see Vallois, 1950, Barnicot, 1952, Oettle, 1963, and Aquaron, 1980) and approximate those found in the present study, suggesting that albinism may occur at a similar rate throughout Africa. The African rates, however, are not as high as those for certain isolates, such as occur in San Blas (Woolf and Grant, 1962).

The urban prevalence rate found here might have been affected by various social factors. The urban Soweto population figure used to determine the prevalence consists of people who have been in the city for at most two generations. It is possible therefore that the actual prevalence is higher, since albinos and their families might not migrate to the cities where competition for employment and facilities is strong. They might prefer to remain in the rural areas where they can maintain a sheltered existence within the extended family. Alternatively, albinos may be taken or sent out of the urban areas (as was suggested by the findings of the present study) to be brought up by relatives in the rural villages and this pattern of behaviour would also reduce the urban prevalence. On the other hand the rate may be too high if albinos tend to seek indoor industrial employment in the urban areas, rather than having to submit to rural occupations frequently associated with excess sun exposure. These factors however could counterbalance each other

and the estimates of prevalence would therefore seem fairly reasonable.

A break-down by ethnic group of the urban statistics indicated that the rates for albinism varied, the Zulu and Xhosa (two of the Nguni groups) having significantly lower rates than the Southern Sotho group. The Southern Sotho subjects however had a consanguineous marriage rate which was more than twice as high as the combined Zulu/Xhosa rate. An investigation of local customs confirmed this finding and Sotho culture condones and even promotes consanguineous marriages, while in Zulu/Xhosa culture such marriages are taboo (Hammond-Tooke, 1974). Specific details on the types of consanguineous marriages in affected families from the present study and in the general population were not available, so calculations to take into account the effect of the consanguinity rate on the gene frequency could not be completed. The data however suggest that the heterozygote rate might be similar in all ethnic groups, but the homozygote rate might be exaggerated in certain groups due to their encouragement of consanguineous marriages.

The prevalence in males and females was found to be similar during the present study, although some authors have found higher rates in males than in females (Barnicot, 1952, Froggatt, 1960, Aquaron, 1980). Barnicot (1953) suggested that the excess of males in his sample was due to his ascertainment methods and to local social pressure, which might encourage males to participate in research projects, whereas females would be less likely to volunteer.

The four studies on the prevalence in rural populations indicated a wide range of local rates, which were higher than those found in urban Soweto. The highest rate (1:1 307) was found in Mochudi (Botswana) where the acknowledged consanguinity rate was only 11 per cent. It is possible that the urban Tswana send their albinos to live with their rural relatives, and one definite case where this had happened was described in Mochudi. It is also possible that this large village might be considered an isolate where inbreeding though not admitted, nor perhaps even realised, may frequently occur. The rural far exceeded the urban prevalence rate of 1:3 481 (with a consanguinity rate of 41 per cent).

In Kwazulu the prevalence rate was found to be 1:6 000 for the general population and 1:2 576 in school children (compared with 1:4 459 for Zulus in Soweto). Barnicot (1952) also reported a higher rate in school children in in Nigeria and it may be that in a community where not everyone can have education albinos are more likely to be favoured above their normal siblings,

and sent to school, alternatively the ascertainment in schools might be more complete. Also, if albinos have shorter life-spans they will be over-represented in the younger age groups.

Although thirty-nine albino subjects were investigated during the Transkei study, the ascertainment from the eleven remote rural districts visited was very poor. The district prevalence rates were obviously too low (the highest being 1:10 000) and taking into account local reports of families who had been missed, as well as Oettle's (1960) and Burrell's (unpublished) figures, they were considered unreliable. The only estimate to arise from this study for the Xhosa group was therefore the urban rate of 1:4 794.

In Swaziland the lines of communication were much more effective and well established and ascertainment of subjects was consequently easier than in the Transkei. Both the prevalence (1:1 951) and the consanguinity rate (17 per cent) were found to be higher than might be expected in an Nguni group (and this finding is supported by Hitzeroth and Hofmeyer's, 1964, data). The Swazis appear to be an exception and, whereas the Nguni eschew consanguineous marriages, the Swazi have tended to promote such marriages, especially among the ruling families to maintain a homogeneous power group. The urban Swazi rate of 1:2 716 was also high compared to those in the other Nguni groups. From discussions in Swaziland it appears that albinos are generally well accepted there, even as marriage partners, and the superstitions which surround albinism elsewhere (see Kromberg and Jenkins, 1984) do not seem to be prevalent. Such social factors together with the increased consanguineous marriage rate could have contributed to the high rate in this ethnic group.

The findings of ethnic differences in prevalence rates in the present study substantiates the results from Hitzeroth and Hofmeyers' (1964) study. They too found the highest rates were in the Tswana (a Sotho group) and significantly lower rates in the Zulu and Xhosa (Nguni groups).

In summary, the urban prevalence of albinism in South African blacks was approximately 1 in 3 900 which is similar to that in other Africac countries. Males and females were equally affected. The Nguni groups (e.g. Zulu and Xhosa) generally showed both a low prevalence for albinism and a low consanguinity rate. The exception to this rule was the Swazi, who are of Nguni extraction, but had a high albinism and high consanguinity rate. The Sotho groups (e.g. Tswana and Southern Sotho), who favour consanguineous marriages also had high albinism rates. It is therefore possible that the gene fre-

quency is similar in all ethnic groups, but that the homozygote frequency is affected by the varying consanguineous marriage rates, and perhaps by the possible selective advantage of heterozygotes which, if such advantage is based upon skin colour, could differ from group to group.

5.2.1.2 Identification and description of types of albinism found in the study

Only a tentative classification of a sub-sample of the subjects could be carried out during the study. The subjects were classified on the basis of hair-bulb incubation tests and physical description.

The majority (68 per cent) of the oculocutaneous albinos assessed during the study were of the tyrosinase-positive type (which included a few ty-pos type B subjects). Very few subjects belonged to tyrosinase-negative group (8.3 per cent), 11.5 per cent were classified brown and 6.2 per cent as rufous albinos. Only 5.2 per cent were considered to fall in the yellow mutant group and these were rather border-line young subjects, who might upon further investigation prove to be tyrosinase-positives or brown albinos. This sample of co-operative subjects was not a random one and therefore it does not necessarily reflect the proportion of these types in the general population.

The tyrosinase-negative group might not have been fully ascertained, since findings from the present study (as well as those of Dogliotti, 1973) suggest that there might be a greater proportion of this type in schools for the blind and cancer clinics than in the general community. On the other hand, the tyrosinase-negative albinos might present with such severe eye and skin problems, that they might not venture out, and might be more likely to be recluses remaining in the shadow of their huts, as suggested by Burrell (unpublished papers). Alternatively, tyrosinase-negative albinos might actually be very uncommon in African populations as proposed by King et al, (1980). This type was also rare in Dutch albinos (Van Dorp et al, 1982) whereas almost half the British and USA albinos (Jay et al, 1982 and Witkop et al, 1978, respectively) and about one third of the albinos in British Columbia were tyrosinase-negative (McLeod and Lowry, 1976).

The classification of subjects into the rufous group presented some problems. Those investigated during the present study appeared to be very similar to those described in Papua New Guinea (Walsh,1971) having red-skin and fair hair, and to those described as having xanthism in Malawi (Stannus, 1913), but they did not fit with the description of Witkop et al (1983)

who stated that the hair colour is generally red in the rufous albinos. Those classified rufous here might therefore have been called brown albinos by another research worker, although their skin has a very distinctive reddish tinge. Since the New Guinea red-skins seem to be classified in the rufous group (Witkop et al,1983), however, those with similar features were also classified in this group during the present study. Ascertainment in this group is likely to have been poor (since they were not specifically sought) and they are probably more common than is suggested here. Barnicots' 1953 study indicates that they occur quite frequently in Nigeria and they have been described in other countries throughout Africa (Eg. Pearson et al, 1911, Stannus, 1913, Loewenthal, 1944, Rose, 1973, Aquaron, Ronge and Aubert, 1981). This rufous group of people have minimal problems and are therefore unlikely to volunteer for research projects and, since they are not always considered to be typical albinos by local generally lay informants, they might not be reported. In order to obtain an estimate of their frequency a study aimed at ascertaining them specifically is required.

Subjects classified as having oculocutaneous albinism are a very heterogeneous group which is still being investigated in attempts to define and clarify the types, particularly in terms of the specific block in the metabolic pathway in each (see King and Olds, 1985). The results from the attempt at classification of the subjects in the present study suggest that all the common types mentioned so far in the literature occur locally. No attempt was made to identify the rarer types, such as the Hermansky-Pudlak Syndrome. The findings of the study suggested that the tyrosinase-negative albinism and yellow mutant types were very rare, while the tyrosinase-positive type was very common, and the brown and rufous types also occurred. This distribution of the types is probably similar to that observed in other parts of Africa, and Stannus' (1913) classification suggests that he saw examples of these five types in Malawi.

From the description of physical characteristics in the sample of albinos in the present study it appears that very few of these subjects have the white hair, white skin and pale blue eyes of the classical albino. None of the subjects in the study could be said to have white hair and all had some form of pigment, in the form of light yellow or yellow hair, even those in the tyrosinase-negative group. It is probable that the phenotypic expression of the albinism gene is age and race dependent. As King and Olds (1985) state the affected individuals are generally very pale at birth but pigment

appears, as they develop, in the hair, eye and skin, and these changes are accelerated in the darker ethnic groups. This accumulation of pigment in the subjects in the present study also seemed to depend on life-style and sun exposure, and patterns of pigmentation could vary even in siblings living together in the same household. These factors had to be taken into account when classifying the subjects, and in siblings the phenotype of both had to be assessed before they could be assigned to a particular class.

Eyes which were basically blue in colour occurred in all the tyrosinase-negative and the majority of the tyrosinase-positive albinos. There was however, in the majority of cases, a more or less clearly defined ring of yellow-brown pigment radiating out from around the pupil. This pigment ring has been described by King and Olds (1982 and 1985) and, in the absence of any other signs of pigmentation and of the development of tyrosinase in the hair-bulb incubation test, they suggested that this presents a different type of albinism called OCA Type II-B (by King and Olds in 1982) and OCA Type III (or minimal pigment, by King and Olds, 1985). The classification of albinos with a negative hair-bulb incubation test result into the tyrosinase-negative or minimal pigment group (Type II-B) was rather arbitrary in the present study. Those with a distinctive clearly visible ring in both sibs were placed in the minimal pigment group, whereas those with an indistinct paler ring or without a ring in one or both sibs were placed in the tyrosinase-negative group. Since seven out of eight subjects in the tyrosinase-negative group had such indistinct rings, it is possible that all these should have been classified in the minimal pigment group. The one subject with blue eyes with no apparent ring however had a younger brother with blue eyes with brownish rays of pigment and therefore even this sibling pair might fit better in the Type II B or minimal pigment group, and the tyrosinase-negative type might not have been detected at all in the series investigated in Soweto.

Blue eyes and strabismus occurred more frequently in the Nigerian albinos (King et al, 1980) than in the local sample of ty-pos and brown albinos. Ephelides however were not a consistent finding in the tyrosinase-positive group in either study, and only about half the subjects in both were affected. This finding might indicate the need for further sub-divisions of the tyrosinase-positive group into the pigment-makers (albinos who make pigment, in the form of ephelides, upon sun exposure) and the non-pigment-makers (albinos who cannot make pigment upon sun exposure).

From this discussion one can conclude that even when a detailed description,as well as a hair-bulb incubation test result, is available it can be difficult to classify albinos. In the present study all the subjects were assigned to a class, but it may be that those on the border-line of two groups should have been left unclassified as in the study of Jay et al (1982), who could not make a definitive diagnosis in 18 per cent of their 201 albino subjects. Further tests are required to clarify and confirm the diagnosis in many of the local subjects. Nevertheless certain trends were apparent from the findings: the tyrosinase-negative type was rare; the tyrosinase-positive A type was common, although about half the subjects in this group could make pigment in the form of ephelides and half could not; the minimal pigment or tyrosinase-positive B group is probably commoner than might be thought; the yellow mutant group was rare; the brown albino and rufous albinos (which might be sub-divided into those with red-skin and fair hair and those with fairer skin and red hair) might be quite common, but were not specifically ascertained during the study. All these albinos appeared to have some form of pigment even if only in the form of pale yellow hair and the classical totally hypopigmented albino was not seen.

5.2.1.3 Cancer risks

The findings from the present study indicated that skin cancer is a common problem for albinos, and that their susceptibility for developing this condition depends upon age, ethnic group, probably environment, and also on life-style. In Central Africa premalignant and malignant lesions appear to develop in the second decade and advanced lesions in the third decade of life (Okoro, 1975, and King et al,1980). In Johannesburg , however, 90 per cent of the skin cancer clinic albino patients were in their fourth decade or older. Albinos living here therefore appear to be at lower risk earlier in life than those living in Central Africa.

The Sotho group of albinos were significantly more susceptible to skin cancer than the Nguni group in the urban part of the present study. Several previous workers (Burrell, unpublished findings quoted by Rose, 1973, Rose, 1974, and Keen, personal communication, 1977) have observed that skin cancer in albinos is very infrequent in the Transkei where the large Xhosa group of the Nguni people live.The Transkei is however further from the Equator and at a lower altitude than the areas inhabited by the Sotho groups. Latitude has been found to be inversely correlated with prevalence rates for skin can-

cer (Scotto et al, 1982a). An ecogenetic study of human malignancy suggested that the chief environmental agent affecting the genetic condition of albinism and causing skin cancer was ultraviolet (uv) radiation (Arrighi et al, 1981 p507) and Scotto et al (1982a) stated that there was good evidence that skin cancer arose from uv - induced changes in DNA, the uv-B portion of the spectrum being especially implicated. The amount of uv-B reaching the earth's surface was found to vary with latitude, altitude, and sky cover (Scotto et al 1980a). Botswana, Lesotho and the Transvaal Province of South Africa where the majority of the Sotho people live, are areas at a lower latitude and at a higher altitude, and have more sunny days and less sky cover than the coastal areas of the Transkei and Kwazulu (where the Nguni live), it is likely therefore that the amount of uv-B is greater in the former than in the latter areas. These findings could explain why the Sotho albinos appear to develop skin cancer more frequently than the Nguni. There may however be other factors, such as varying diet, cultural habits, skin colour (which is generally darker in the Nguni than in the Sotho), infections, viruses and/or the genetic constitution of these two groups of people, contributing to the situation. If the trend suggested by the findings for the Transvaal urban sample (in the present study) in which the Sotho albinos had a significantly higher rate of premalignant and malignant lesions than the Nguni albinos (although they all lived in Soweto) continues over the next few decades, when these groups will be completely urbanized, then genetic factors might be found to be more important than the environmental factors. The susceptibility of albinos to skin cancer may then depend in the first instance upon their ethnic group, rather than on the environmental factors which will be similar for all those living in the same area.

The investigation of the histopathologists' reports on skin biopsy specimens in the present study indicated that at one hospital (Baragwanath) nearly 13 per cent of the specimens investigated and found to have squamous or basal cell carcinoma were from albinos. Figures from other studies (Eg. Cohen et al, 1952, Oettle, 1963, and Isaacson, 1978) also show that the risk for albinos developing skin cancer is dramatically increased over that of the general black population. Oettle (1963) found that in comparison with US whites South African Negroid albinos had approximately ten times the incidence of skin cancer. Higginson and Oettle (1960) estimated that the albino in Johannesburg had a mean annual risk of 1:80 of developing skin cancer and that this risk was 1 000 times as high as that of the local pigment-

ed black population.

The commonest site for the skin carcinomas reported in albinos in the histopathology records was the head, particularly the face (unspecified) eye and eye-lid, and cheek. The limbs were involved in only a minority of cases and in each of these it was the lower limb. Oettle (1963), King et al (1980) and Luande et al (1985) found a similar distribution, although upper limbs were also affected in their series. In the present study the normally pigmented black population had a different distribution of sites and the head was only involved in half the cases, the limbs (particularly the lower limbs) in a quarter and the torso in a further quarter of the cases. Oettle (1963) on the other hand, found the lower limbs associated mainly with previous trauma, burn scars and chronic ulcers, to be most susceptible, followed by the head in his series of black patients. His subjects however, included many from the rural areas where shoes were not often worn, and the feet and ankles were thus very susceptible to trauma resulting in ulcers and eventually skin cancer. The back and chest were found occasionally as affected sites in albinos in both Oettle's (1963) and the present study, and this suggests that either these areas were still sometimes exposed to the sun in this population, or that factors other than sun exposure contribute to the development of cancer, in these sites, or that clothing is not always adequate as a protection for the body from ultraviolet radiation. Cervanka et al (1979) suggest that events, such as unusual diets, chronic bacterial or parasitic infections and/or oncogenic viruses, which are not directly involved with malignant transformation might play an important role by non-specifically interfering with immunological defenses. If such factors are important this theory could perhaps partially explain the occurrence of cancers in unexposed as well as exposed sites in albinos.

The proportion of basal cell to squamous cell carcinoma appears to be similar in the present study and in the studies of Oettle (1963), Isaacson et al (1978) and Luande et al (1985), the former type occurring much less frequently than the latter in albinos and in the general population. Malignant melanoma was not reported in albinos in the present study, but Oettle (1963) however found one affected albino in his series and Rose (1973) reported one in the Transkei. This type of carcinoma therefore appears to be very rare in albinos.

In summary, then, skin cancer (particulary on the head) was found to be a high risk for the albinos compared with the pigmented population in the

present study, as has been found in other studies. The risk increased with age, but was also partially determined by ethnic origin, since the albinos in the Sotho group appeared to have a higher risk than those belonging to the Nguni groups (when both were residing in Soweto), and by environmental and geographical factors (such as latitude, altitude and sunlight).

5.2.1.4 Life expectancy

The results on the analysis of the age distribution in the albino group in the present study compared with that for the general population of Soweto indicated that there was a statistically significant difference between these two groups. The albinos were younger than the general population and the average ages of the two groups were 24.2 and 28.2 years respectively. Although some ascertainment bias is possible, this finding suggested that since albinos were over-represented in the younger age groups and under-represented in the older age groups their life expectancy is reduced. The age distribution of the albinos was skewed at two points particularly, there were more albinos than expected in the fifteen to twenty-four year age group and less than expected in the age group forty-five and over. This pattern suggests that the albinos might be sent from the rural to the urban area for educational and employment reasons, while other pigmented people finding limited opportunities might leave as adolescents for the rural areas; then occasionally after the age of thirty-five, but generally after forty-five many either die of skin cancer, or leave the city for their rural village. Those who do not die at this stage, it seems, can live to a ripe old age and in the sixty-five and over age group, although the sample was very small (eight subjects), albinos were again over-represented. This latter group presumably represented those who were well cared for, nourished and sheltered throughout their lives and who did not develop malignancies at a younger age.

For purposes of comparison the Swazi data on ages of albinos and the general population were analysed and a very similar pattern to that found in Soweto emerged. Again there were more albinos than expected in the younger age groups and less than expected in the older age groups, suggesting a reduced life expectancy. Okoro (1975) stated that the age distribution of his sample of Nigerian albinos showed a rapid decline in numbers after the third decade, and although he admitted that this might also have been due to his methods of ascertainment, he nevertheless concluded that albinos probably had a shortened life-span.

Oettle (1963), however, presented age statistics for the Transkei, which

indicated that there was no discrepancy between those for albinos and the general population, and he concluded that the albino's life expectancy did not seem to be reduced. This finding fits well with that observed during the present study on skin cancer in Soweto, since Xhosa albinos (who generally live in the Transkei and Ciskei) were noted to have a markedly reduced rate of skin cancer compared with albinos from some of the other ethnic groups. It appears therefore from Oettle's study that albinos in the Transkei, at least, can probably expect a normal life-span, since they do not appear to succumb to skin cancer.

Aquaron (1980), claimed that in the Cameroon, albinos had a shortened life-span although there were, in his sample, a few albinos in the fifty to sixty year age groups. In Nigeria no tyrosinase-positive albinos older than forty years of age were seen by King et al (1980). The other different types of albinism have not however been investigated separately for longevity and it might be assumed that the milder the type the less effect the condition will have on life-span. Barnicot's (1952), claim that longevity was not compromised in Nigerian albinos is at variance with the findings of King et al (1980), but it is possible that regional differences could explain these two findings, since Barnicot's work was conducted in a coastal region and King et al's in the East central region of Nigeria. In addition the life-styles which existed in the early 1950's, when Barnicot carried out his study, may have undergone radical changes in the intervening thirty years. From the findings of the present study in comparison with Oettle's study regional variations also appear to occur in South Africa.

Albinos were more common in the younger age groups in Ireland, but Froggatt (1960) could not find any clinical problems that could account for concluding that their life-span was shortened. He therefore suggested that his findings were probably due to ascertainment bias, and/or that possible changes in mutation rate or mating patterns, might cause a rise in the prevalence of the condition over different time periods.

The analysis of age distribution in male and female albinos in the present study indicated that both groups were younger than their counterparts in the general population. For males the difference was highly significant, whereas for females it was border-line not significant. Expectation of life at birth for local black males and females was estimated as fifty-five years and sixty years, respectively, in 1980 (Mostert and van Tonder, 1982). The present study has shown that both male and female albinos could reach the

expectation of life in the normal population. The difference in life expectancy for albino men and women might be explained by their differing lifestyles. It is probable, in the local situation,that albino men are obliged to receive more sun exposure because of their limited education which results in restricted employment opportunities and, often, in out-door manual labour. Whereas the women, although in the rural areas they may tend the fields, may be able to avoid exposure to sunlight more successfully. Furthermore women, especially if they do not marry, may remain sheltered by their biological family for life, while the men may leave the security of the home in search of independence and employment.

5.2.1.5 Skin colour studies

The data collected on skin colour showed that Swazi female heterozygotes for the albinism gene were significantly lighter in colour than matched pigmented females in the general population. The refectance values for the matched control group (Swazis, one of the Nguni groups) were compared with those of Wasserman and Heyl (1968), who measured skin reflectance in a group of Xhosa (another Nguni group) women. The two studies showed very similar results suggesting that the small sample (eighteen subjects) in the present study was probably quite representative of the general Nguni population.

The suggestion of lighter skin colour in heterozygotes adds to the continuing discussion in the literature on pigmentary changes in carriers. It has been proposed that iris translucency is significantly more frequent in carriers than in the general population (Waardenburg, 1947 and Froggatt, 1960). Witkop et al (1973) however believed that translucency was not a reliable indicator of the tyrosinase-positive and yellow mutant carriers, and found that even in the tyrosinase-negative group only half the carriers showed translucency. Although the type of the albinos found among the Swazi was not identified in the present study, very few ty-neg albinos were detected in the Soweto sample of albinos. It can therefore be assumed that most of the albinos in the Swazi sample were ty-pos and that nevertheless, their mothers appeared to have lighter skin colour than controls. Fair hair and blue eyes have been described as more common in unclassified Irish carriers than in the general population (Froggatt, 1960). Pigmentary changes therefore seem to occur in groups of unclassified carriers, among whom there are generally a minority of tyrosinase-negative types.

Since the general observations of the writer and others (e.g. Oettle, 1963, Waardenberg et al, 1961, and Keeler, 1953) that parents of albinos

were lighter in skin colour than the general population stimulated the present investigation, it might be assumed that the lightness of the skin colour is such that it can be detected quite easily by any member of the general population. Such light skin colour has been considered to be beautiful in many African groups over the years. Sir Harry Johnston (reported by Pearson et al, 1911) stated that in British Central Africa local people admired light brown skin colour especially in women, and the wives of chiefs were often of this colouring. Similarly in Somaliland people admired the lighter shades of skin colour and tended to marry into families of this description (Paulitshke, reported by Pearson et al, 1911). The generally admired type of woman who was preferred as a marriage partner in a Tswana community, was `light-skinned' (Schapera, 1940) and those with a light skin colour have been admired by the Swazi at least since the late nineteenth century (Kuper, 1947, p33). In the Transkei witches worked by means of `familiars' (sometimes called tikoloshe) who took the form of a beautiful girl or handsome man, very light in colour (Wilson, 1951). Among the Southern black peoples of South Africa the members of the chiefs' families were usually lighter in skin colour than the ordinary members of the group, and chiefs were known to select especially light-skinned wives (Bothma, 1969). Dark negroes generally tend to prefer light wives and fair skinned negroes prefer to have fair skinned partners (Robins, 1970). Consequently it is very possible that, since carriers of the albinism gene seem to have lighter skin colour they have been preferentially selected for as marriage partners in the past. This selective advantage could have contributed to the high local prevalence of the albinism gene. Skin lightening creams are presently very popular among black women in South Africa, suggesting that woman wish to have lighter skins, since they seem to assume that such a feature is beautiful. The light skinned person may therefore still be at an advantage in present day society.

For the purposes of comparison with the reflectance values for albinos the figures for South African Caucasoid females were extracted from Wasserman and Heyl's (1968) work. The albinos were found to have reflectance values which were higher at all wavelengths than the Caucasoid females. This result might have been expected since albinos have skin which can be observed with the naked eye to be unusually light and less tanned than that of most South African Caucasoid women.

In summary then, the skin reflectance measurements made during the present study indicate that the albinos had the highest values followed by the

heterozygotes, who were found to be lighter than the normally pigmented matched controls. Light skin colour appears to give an advantage in the local community and there is probably some selective mating in favour of the lighter skinned members of society. This cultural practice may represent one of the factors which has contributed and may still contribute to the high prevalence of albinism found in indigenous Southern African populations.

5.2.1.6 Fertility studies

The fertility in the homozygotes in the present study appeared to be reduced. Life-time infertility rates for males were more than twice as high as those for females. Rates for males however are generally considered unreliable, and the only comparable figure that could be found for the female rate was that for rural Xhosa women (Irwig and Ingle, unpublished data, 1984). The urban albino women (from Soweto) showed a significantly higher infertility rate than the rural Xhosa. This however might be an unfair comparison since urban women generally have lower fertility rates than rural women. The number of children per albino woman was therefore calculated and compared with that reported for urban Xhosa women living in the Cape (Roberts and Rip, 1984). The albinos, had 0.89 children per person, while the urban Xhosas, had 2.9 living children and had had 3.9 pregnancies, although the albinos had a mean age of only two years less than the Xhosa women. There were 52 per cent of albino women and 5 per cent of Xhosa women without children, confirming the finding of increased life-time infertility rates in albino females.

Increased levels of plasma melatonin which were constant during the day and night have been found in two albino males (Oosthuizen et al, 1983). Melatonin has been reported to inhibit reproductive function in the rat (Reppert and Klein, 1980) and to inhibit ovulation and modify the secretion of other hormones in man (Wurtman, 1975). If melatonin is generally over produced in albinos this may be associated with their reduced fertility. On the other hand social stigmatization may also exacerbate the problem for albinos. Locally, however, the albinos seem to have difficulty in marrying, but not in obtaining sex partners, and although few are married, several have children (Kromberg and Jenkins, 1984). The apparent reduced fertility in local males and females is therefore probably not entirely due to the social stigma associated with albinism and other possibly physiological factors might be involved. In Brazil 10 albinos were found to have normal fertility (Freire-Maia et al, 1978), but in a Zuni group 11 male albinos had no

offspring and in a Brandywine isolate reproductivity was not increased (Witkop et al, 1972), so the matter requires further clarification.

Heterozygotes in Soweto (some of whom might not have completed their families) were found to have 3.93 children per couple. In comparison with the total fertility rate of 5.2 children per woman for the South African black population (Report of the Science Committee of the President's Council, 1983) this figure for parents of albinos was not raised. The latter report included rural and urban families, while the albino families were all from urban Soweto. No comparable figure could be found for pigmented families in Soweto, so the figure for parents of albinos was again compared with that for a sample of urban Xhosa women (mean age thirty-one years); they had 2.9 children and had had 3.5 pregnancies per person (Roberts and Rip, 1984). In this group of Xhosa women only 10 per cent of the pregnancies had occurred over the age of twenty-five years, so that most of them had completed their child-bearing, and therefore their fertility rate could possibly be comparable with the Soweto heterozygotes figure. Although the latter is higher than the former no conclusions could be drawn, since both figures were not sufficiently specific or closely comparable, and an undetermined number in each group had probably not completed their families.

The heterozygotes and matched controls participating in the psychosocial part of the present study were therefore investigated. The mothers of albinos (mean age 25.8 years) had 2.08 children and the mothers of controls (mean age 23.4 years) also had 2.08 children. Although the mothers of albinos on average were two years older than the controls, the differnce between the groups appeared negligible.

Lastly, fertility in heterozygote x normal matings (i.e. grandparents of albinos, among which one was assumed to be a carrier and the other normal) was assessed. The group of Soweto mothers of albinos (143 subjects) had 4.38 siblings, per person which was lower than the rate, of 5.3 which was the figure given for family size in Soweto (Bureau of Market Reseach, 1977) and 5.2 which was given in the President's Council Report (1983), but again both these latter figures are probably not strictly comparable. The information from the matched pairs was therefore also analysed in terms of maternal and paternal siblings per person. The combined rates reported for the parents of albinos were 4.22 per person, while the controls reported 3.63 siblings per person. These findings, although they did not quite reach statistical significance, suggest that there may have been an excess of children in the het-

erozygote x normal matings, possibly giving the heterozygotes a slight fertility advantage over the normal controls. If this is so then such increased fertility might contribute to maintaining the high local frequency of the gene for albinism. Increased values for fertility have been found in grandparents in families with recessives, such as cystic fibrosis (Knudson et al, 1967) and Tay Sachs Disease (Myrianthopoulos, and Aronson 1966), which also have high prevalance rates in well defined communities.

5.2.2 Psychosocial studies

The results on the longitudinal study of the impact of the birth of an albino baby on the mother and, secondarily, on the father, indicated that there were several statistically significant differences between the experimental group and the control group, and within the experimental group, when the findings at the beginning and end of the study were compared. These differences will be discussed here.

5.2.2.1 The impact of the birth on the mother

In the neonatal period the mothers of albino babies were more reluctant than the controls to hold and breast-feed their babies. Such behaviours are essential, together with, amongst others, eye-to-eye contact with the baby, an en face position, the use of high pitched voice, and entrainment,for the development of maternal attachment to the baby, and for the stimulation of interaction between the mother and her child (Klaus and Kennell, 1976). Feeding practices also play an important part in the development of maternal-infant attachment, and physical contact of good quality in early infancy has been found to facilitate later development (Ainsworth, 1972). The newborn albino babies in the present study were therefore partially denied the necessary interaction with their mothers which appears to be conducive to normal development.

As we have seen (see chapter 2 of the present study) other studies have not examined the black African mother's behaviour in interaction with her handicapped infant during the neonatal period. In general terms, however, the mother's initial reaction has been described as one of protest, shock and numbness (Kennedy, 1970; Drotar et al, 1975), followed by feelings of helplessness (Solnit and Stark, 1961). Such a reaction may naturally lead to an inability to interact with the baby, a reluctance to be close to it and to hold and breast-feed it, as was observed in the present study. From the experimental mothers behaviour at this time it appears that the impact of an

albino baby is similar to that described in the literature in connection with other defective babies.

Parents of children with visible defects tend to be more anxious than those with babies with other defects (Johns, 1971). Since albinism is visible not only to the mother, but also cannot be hidden from those around her, it might be associated with greater anxiety and shock than some other disorders. Skin colour seems to be important, particularly as a means of establishing identity and affinity to a particular population. The sight of her white baby might therefore upset a black mother in many ways; she may not feel pacified by the fact that the baby is physically normal, apart from the absence of pigment, and her behaviour might thus be affected, as evidenced in this study. As Slutsky (1969) has suggested, from the point of view of rehabilitation, the psychosocial problems for a mother in this situation may transcend in importance the actual physical disability. This is not surprising since society generally has a negative response to atypical physical appearance and functioning, and places highly positive values on normal appearance and adequate functioning. The mother has to adapt to the wider milieu and the requirements of the interpersonal and social relationships, which may initially involve rejection, within her environment and culture.

Two weeks after the birth the mothers of albino babies showed significantly fewer interactive behaviours in relation to their babies, and they were less likely to smile at, stroke, and hold their babies close, than the controls. Recovery from the effects of having a defective baby is gradual and takes time (Kennedy, 1970). This lack of interactive behaviour probably indicated that development of attachment to the infant was somewhat delayed.

Also at the two week interview, more mothers of albinos than controls complained of headaches, feelings of depression and unhappiness, and admitted to being disturbed by the baby's defect. Apart from the claim to have headaches, which might be said to be of psychosomatic origin in some cases, most of the experimental mothers did not admit to other health problems to a greater degree than contols. Similarly parents of Down Syndrome children showed no excess of symptoms of physical ill-health, in comparison with a control group (Gath, 1978, p55). The feelings of depression and unhappiness which were reported were probably related to the second stage after the birth of a defective child as described by Kennedy (1970), during this stage the parents tend to feel despair, disappointment, hopelessness and futility. More mothers of albino babies than controls planned to leave town and join

their extended families in the rural areas. A loss of warmth in previously close relationships has been observed in families with a defective child (Kennedy, 1970). This factor might have contributed to the mothers need to go to her wider, probably more supportive, family.

The mothers of albinos also stated, quite realistically, that their babies would have more problems than other normal babies and that they would give them more education, but that they would prefer the next baby to be different. Whether these findings indicate that the mothers felt the need to overprotect their child in the future is a moot point. As Shakespeare (1975, p53) states, in practice it is difficult to define the difference between the protection which is necessary, because of the defect, and overprotection. The albino's physical problems are related in many instances to sun exposure and, therefore, more education would probably result in less likelihood that the albino would have to work outdoors as a manual labourer. The mothers might therefore have been indicating realistic coping with the problem by these responses, rather than a tendency to overprotect.

The findings showed that the mother's behaviour and some of her attitudes had changed by the time the baby had reached three months' of age. As many mothers of albinos as controls were breast-feeding their babies. They were also observed to smile at, stroke and hold their babies close, and there was no significant differences between the groups for these observations. As Gath (1978, p121) states, although there was the initial reaction to the fact of the baby's abnormality (Down Syndrome) most families adjusted well, in her study.

Although the experimental mothers were observed to behave in the same way as the controls towards their infants, at three months, some nevertheless still claimed to feel disturbed and unhappy and planned to join their families in the rural areas. More experimentals than controls added that their babies cried too much and Elster et al (1983) suggest that mothers who are stressed may be less tolerant of adverse stimuli such as infant cries. These findings of intolerance and unhappiness can be accepted in the light of those of other research workers (e.g. Kennedy, 1970; Drotar, 1975; Cunningham, 1979) who state that stages overlap, feelings fluctuate, and sorrow can be chronic (Solnit and Stark, 1961). Learning to cope in this situation is a gradual process which might take longer to achieve in thought than in action. The affected mothers, it appeared, coped with interacting with their babies at this stage, but not with their own thoughts and feel-

ings. Another finding in partial support of this statement is that the mothers added that they would prefer a normal baby if they had another child. It might be argued, however, that such a statement might be made by any mother of a defective child, even where the child is totally accepted.

The observations conducted when the babies were six months' of age again showed no significant differences in behaviour between the groups. As many experimental as control mothers breast-fed their babies and the experimental mothers continued to express their concern over the excess of problems the affected child might have. Those who planned to leave town had left, and those who remained did not express the desire to leave. According to the custom of many of the South African black ethnic groups, the child is not allowed out of the hut where it was born until it is two or three months' old, or even older in some cases. The child is believed to be weak and in danger of being injured by harmful forces in the outside world (Krige, 1937, p95). This belief is still widely held even in the urban areas and for this reason many of those mothers who planned to leave for the rural areas only did so after their babies were three months' of age or older. Although the mothers were more settled at the six month interview, nevertheless some still maintained that they felt unhappy and disturbed about their albino child. They no longer complained of the babies' excessive crying, however, possibly suggesting that they were beginning to accept the baby and were not feeling so stressed. They admitted that their mothers were supportive and comforting, and there was a significant difference between the mothers in the two groups for this item. Again, it is possible that those who stayed in town remained there because of their mother's presence and support, whereas those who left might have done so to join their mothers in the rural villages. If this was the case then this finding could be caused by selective dropping out and the remaining subjects being a biased sample for this particular aspect.

At the nine month interview the sample consisted of eighteen pairs. The mothers professed to some feelings of acceptance and stated that they were no longer unhappy or disturbed by their babies' defect, although they would have excess problems. They, nevertheless, were found to treat their albino babies differently and had introduced them to a wider range of solids than the control infants were receiving. This finding again might indicate a tendency to overprotect the albinos and to compensate for the fact of their defect. It lends weight to the finding mentioned above that mothers wanted

their affected children to have more education, and to the results from a previous unreported study (Kromberg, unpublished data), that mothers gave their albino children more food than their normal children. The mothers of albinos also admitted to receiving continuing support from their mothers.

There were very few differences between the groups from the data collected when the infant was one year old. The only two items which showed differences were those relating to the problems of the albino baby and the probably normal desire of the mothers to have a `different´, implying normal, baby next time. However, a significant result was found when the milestones of the babies in the two groups were assessed. The albino babies were reported by their mothers to have been slower to sit and crawl than the normal babies. The African child is precocious in psychomotor development, and the majority of infants can sit alone at four months, stand alone at eight months and walk at ten months (Geber, 1958). The albino babies took significantly longer than the controls to sit and crawl.

There are at least three possible explanations for this finding of delayed development in albinos. Firstly, the mothers might have reported a distorted perception of their baby´s achievements. Secondly, as mentioned above, the mothers of albinos showed less neonatal interaction behaviour with their infants than did the controls, and such maternal deprivation can lead to slower development (Ainsworth, 1962, p108). It is possible that the deprivation has to be grossly insufficient (and such deprivation was not observed in this sample) to cause delayed development. Thirdly, the albino children might have been slower to explore, and therefore to sit and crawl, because of their poor eye-sight. Blind children have been found to be slower to crawl and walk than normal children, and the median age at walking alone in one study was 15.25 months for blind subjects and 11.7 months for sighted children (Fraiberg, 1977, p204). Vision is very important in the infant´s learning of control and body movement (Scott et al, 1977, p30). Babies with normal vision will lift and move their heads and be rewarded by seeing different objects and colours, but babies with limited vision are not fully aware of these sights, they lack external stimuli and therefore are not motivated to move their bodies purposefully and self-initiated mobility takes longer to achieve (Fraiberg, 1977, p62). An intervention programme that provided early auditory-tactile experiences, encouraged physical activity, and permitted `sound to serve as a lure for forward progression´ (Fraiberg, 1977, p209) resulted in the earlier achievement of mobility. The albino child is,

however, not blind but partially sighted, and therefore it is possible that the slow development observed during this study was the result of a combination of factors, including partial sight and partial maternal deprivation.

The information obtained at fifteen months indicated that the development of the albino babies was not delayed for long and there was no significant difference between the two groups at this stage. The results confirmed those found previously, that the two groups were essentially functioning similarly, but that the mothers of albinos wanted a normal baby next, and that they were still concerned that the affected child would have more problems than the normal child. Since no changes were found at this fifteen month stage, in comparison with the twelve month stage the study was then concluded. Families with Down Syndrome children were found to be providing a stable and enriching environment for both the affected and normal children two years after the birth of the affected child (Gath, 1978). Mothers of cleft lip and palate children (a visible defect), reported anxieties about the reactions of others in the first year of the child's life but these anxieties lessened during the second year (Slutsky, 1969). From the findings of the present study it appears that the mothers, of albino children had more or less accepted them by nine months' of age and that there were no observable differences on the aspects selected for study between the experimental and control groups at fifteen months' of age.

An observation session was held at the last interview to assess the mother-infant interaction in a play situation. The numbers were too small for statistical analysis, but the findings suggest that the mothers of albinos did not sit as close to their babies as the control mothers did. The experimental mothers also tended to vocalize more and their babies vocalized more in response, but they gazed less at them and mediated the babies' environment less by, for example, passing him toys less often than did the controls. These trends are worth recording as preliminary observations which require further study. Withdrawal, indicated here by the increased distance between the mother and albino child, and indifference, suggested by the lesser mediating of the environment, have been observed in connection with people who are different and unfamiliar (Wright, 1960, p263). Those with an atypical physique may experience a certain emotional distance and sometimes even antipathy from others. Whether this emotional distance can be measured by physical distance, is debatable. Increased distance might also be a sign of continuing partial rejection of the child by the mother, and a

reluctance to be close to a child with whom she cannot identify, because of his strange and different skin colour. This finding could be associated with the post-natal behaviour of the mothers when they did not wish to hold their albino babies close and showed reduced interaction with them. The strange and unfamiliar involves a negative reaction in most people resulting in some distancing from the object that causes it. This theory might explain the mothers' reluctance both to hold their unexpected albino babies close soon after the birth and, later, to sit near them while playing. Although the mothers claimed and appeared to be accepting their babies by fifteen months, this might not have been a complete acceptance, which probably takes longer or might, in fact, never be achieved.

The results associated with the anxiety scale can also only be reported as observations since the sample numbers were too small. Raised levels of anxiety have been found in mothers with babies with a visible defect (cleft lip and palate), in the first year of the baby's life, in comparison with controls who had babies with an invisible defect (cleft palate) (Slutsky, 1969). Similarly in a small sample of mothers of babies with a variety of defects, those with the visible and more severe defects were found to be most anxious (Johns, 1971). The tentative observations, from the small number of mothers in the present study, support these findings and suggest that the mothers of albinos were more anxious than mothers of controls, even fifteen months after the birth of their babies when they were tested.

Two further factors should be considered in this discussion of the impact of the birth on the mother. The first is a reservation which should be mentioned in connection with these results. The interviews were conducted with the mothers alone and many of the items referred to feelings and attitudes, while others concerned the babies' progress. In response, the mothers reported their own retrospective view of their feelings and attitudes and of the development of their babies over the time period between each interview. Such parental unconfirmed reporting has been looked at askance by some research workers. In connection with the child's development, parental reporting has been found to be unreliable in some normal families, and it is suggested that unreliability might be even greater when the child has a behaviour disorder (Thomas et al, 1968, p7). If, however, the behaviour to be described is not too remote in time and if descriptive factual information is requested then accurate parental reporting is possible (Thomas et al, 1968, p17). In the present study information was requested retrospec-

tively on the previous three months, and the mothers were only asked to report facts about the age of their children when they reached various milestones. Hopefully, therefore the information given was reasonably accurate. The mother's emotional response to her defective child, however, could have affected her memory of the baby's achievement of milestones and caused her to report them as later than they really were. Such reports should ideally be substantiated if possible in further research such as this.

A second factor which should be considered when taking a general view of the findings on the mothers in this study, is the effect of the infant's characteristics on the mother. Maternal-infant interaction is a two-way process, the infant must respond to the mother's approaches and vice versa. If, however, the infant's sight is not perfect, as is the case in many albinos, the infant may not be capable of becoming fully involved in the visual communication aspect of the process, and may be slow to recognise the mother's approaches and signals and to appreciate her smiles and looks. If this is the case the mother may not receive the necessary rewards that she requires to maintain good interaction and might withdraw , or assume that the baby does not like being played with and leave him alone (Scott et al, 1977). This situation might therefore be similar to that suggested by Thomas et al, (1968, p183) in families with autistic children, where good mothering may be provided, but the infant may be so constituted that he cannot take part in the usual interaction patterns. Another characteristic of the albino baby is the whiteness of his skin and hair. The mother cannot even look at the infant without being reminded of his condition and his imperfections. This might initially cause her to look less at her child, resulting in reduced maternal-infant interaction. Some writers (such as Stewart and Keeler, 1965) have described the albinos they investigated as insecure and emotionally immature and attributed these characteristics to overprotection. The findings of this study, however, suggest that the initial reduced maternal-infant interaction might also be a factor.

5.2.2.2 The impact of the birth on the father

Only superficial information was collected in a single interview with a few of the fathers, but since there is so little in the literature on black paternal-infant relationships the observations made on this group merit some discussion. The fathers generally did not admit to feelings of being disturbed and unhappy about their babies when interviewed on average six months after the birth. This finding differed from that expressed by the mothers

who said (six months after the birth) that many of the fathers of their babies were unhappy. Mothers however are not always reliable informants or interpreters of fathers' behaviour (Eron, et al, 1961). On the other hand it is possible that since those fathers who were interviewed were those who were co-operative and, to a certain extent, self-selected, they were the ones with a positive view-point. Alternatively, they might have been reflecting societal values that men are stoical, less emotional and strong, and fathers in general might have only a limited ability to ask for help, in this situation (Mandell et al, 1980). It might also be that the mothers were projecting their feelings on to the fathers and that fathers did not feel as disturbed as the mothers implied. Cunningham (1982, p217) suggested that mothers and fathers can have quite different reactions to the birth of a Down Syndrome baby and that such dissynchrony might cause conflict and increase emotional stress. Kennedy (1970) stated that when parents are at the stage of dispair and depression after the birth of an affected baby there is a loss in the warmth of their relationship, and they cannot converse freely with each other, partly because of the need to apportion guilt and blame. If this was the case in the families in this study, the mother possibly should not have been asked about the father's feelings, since communication may have decreased and she may not have been fully aware of them. There are therefore at least three possible interpretations of these results: either the fathers were not unhappy because they had accepted their babies; or they were not admitting their unhappiness, for stoical reasons; or their feelings were dissynchronous with the mothers, whose perception of the fathers was inaccurate. Further research on fathers and mothers is required to clarify this matter.

During the observation session on the fathers only half the fathers of albinos (compared with two of the three control fathers) called their babies by name or kissed them. This behaviour should be viewed against the background of the local culture. The father in local traditional society has comparatively little to do with his children during infancy (Schapera, 1937, p244). His interaction with his new-born child, is minimal and, in some tribal groups, subject to a strict ritual in the first few weeks of life. In the Tswana group for example, the father must not even sleep in the same hut as his wife and child after the birth (Schapera, 1966 p234). He is forbidden to enter the hut at all while the `cross-poles' lie outside the door, lest he trample on, injure or fatally retard the progress of mother and child.

Once the umbilical cord has fallen away and, in some cases, after the baby has been presented to the first new moon following its birth, the father may cross the poles, enter the hut and be shown the baby for the first time. The mother and baby may, however, remain in the hut for one or two months and no other men may cross the poles. During this time the child has no name, but just before it is due to `come out´ the father recognizes the child as his own by naming it, after discussion with his relatives. It is only at this stage that the father returns to sleep in the same hut as his wife and baby. As the infant grows, it spends most of its time on the mother´s back, or with the mother, another female relative, or a sister. The father plays a very small role and has only limited opportunities to interact with the child, and, although he may play with the baby, he seldom feeds it or cleans it, or puts it to sleep (Schapera, 1966, p239). With this background it is not surprising to find that only half the fathers in this study referred to their babies by name or kissed them and one father of an albino even admitted to having hardly ever held his baby (before being requested to do so during the observation), although the baby was nine months old.

The fathers in both the experimental and control groups expressed the desire to have more children, but the experimental fathers did not commit themselves to stating that they would like the next child to be different (and not an albino). Throughout Africa the black population has been characterised by a high birth rate (40 to 50 per 1 000 inhabitants, as compared with 18.5 in France and 25 in the USSR (Collomb and Valantin 1970, p363) and children have been looked upon as part of a man´s wealth. So again this finding is to be expected in these fathers. However, whether they planned further children sooner than fathers who had had a normal child was not established, nor was the question of whether an extra child was planned to compensate for the defective child put to them. Nevertheless the presence of the abnormal child did not seem to be acting as a deterrent in the planning of future children, as has been described elsewhere (Shakespeare, 1975, p62).

The paternal-infant relationship does not develop in isolation and the contribution of the mother cannot be ignored. Every family therefore has to be seen as a triadic system (Clarke-Stewart, 1982) and the interaction between all three members must be analysed in context. In the present study however, fathers and mothers were interviewed and observed separately and the extent of the influence of the other parent was not assessed. Nevertheless it is possible that some fathers might have had reduced access to their

child, if the mothers were overprotecting the child and did not allow much paternal-infant interaction. The information on the fathers is too limited and the sample too small to draw any firm conclusions.

5.2.2.3 The impact of the birth on grandmother and siblings

It was not feasible to interview grandmothers during the study and where there were siblings they were unavailable in most cases. The information on these family members is therefore obtained from the mothers' reports and is limited and unsubstantiated. Nevertheless a brief discussion of the findings and a comparison with the data from the literature is worthwhile, since grandmothers and siblings of albinos in black African families have not been investigated by social scientists as far as the writer is aware.

The grandmothers in the experimental group were found to be more supportive than those in the control group according to the mothers. The role of the maternal grandmother in local black society is a significant one. She provides her daughter with utensils and seed-corn (if necessary) after her marriage, to take to the new home, she visits her daughter frequently, helps her with household tasks and attends to her when she is sick (Schapera, 1966, p244). After confinement she assists her daughter in bringing up the children and advises her on any problems she experiences. It appears that those grandmothers who were present during the study were playing their traditional role. Their daughters saw them as comforting, supportive and reassuring, especially when the baby was an albino. Grandparents have been reported as being difficult and interfering on occasions where the child has Down Syndrome (Cunningham 1982, p49-52). They may also become as bewildered and upset as the parents, although the mothers in the present study did not report observing these feelings in their own mothers. There was only one grandmother who directly interfered with the study and asked that her daughter be allowed to withdraw, since she did not accept that her child was an albino. This grandmother might also have been denying the condition of her grandchild, and such feelings have been reported in grandparents in regard to other conditions (Cunningham, 1982, p49). Cunningham (1982, p51) also found that grandparents can feel pessimistic, and can accuse the other side of the family of being to blame for the baby's defect. This attitude can result in an increase in marital tension. Grandparents on the other hand have to accept that they have a grandchild with a defect and they have their own difficulties in dealing with this situation. Presumably, however, those grandmothers who had negative attitudes towards their daughters and their

albino babies did not visit so frequently, or at all (as one subject reported), and the daughters may not have been aware of these attitudes or may not have reported them.

The siblings were another group that were investigated only superficially through information given by the mother. Siblings of defective children have been found to reflect the parental attitudes to the defective child, or to show reactions which are related to their parents' reactions. Only half the siblings in the present study were reported by the mothers to be happy to have a new sibling. According to the mothers, some of the siblings of the albino babies showed extreme reactions, going so far as being afraid to enter the room where the child slept, or refusing either to touch the child, or to show the new baby to their friends. Similarly, another study has reported that some siblings of physically and mentally disabled children were deterred from entertaining friends at home, because of the presence of the affected child (Shakespeare, 1975, p62). Depending on age and experience some children will be upset about the different and unexpected child, some will deny that the baby is different, some will feel embarrassed, others will require an explanation (as one child asked of his mother in the present study `why did you buy a white baby?'), and several will not even admit that they have the affected sibling, either because they wish to deny his existence, or because they find it difficult to explain the situation (Cunningham, 1982, p45). The majority of siblings however, learn to cope, as their parents do, and are not detectably disturbed by the presence of the affected child in the home (Shakespeare, 1975, p62). The number of siblings, according to Shakespeare (1975, p63) who do show problems differs in various studies, and in the present study there was only a minority of cases, in which the mothers described extreme reactions in the siblings. As a group siblings' feelings were mixed and they were said to react differently to the birth of the albino, but the mothers did not report physical side-effects, such as excessive visits to the doctor, nor truancy, nor absence from school, in the small sample available.

Sometimes strong bonds of affection occur between siblings and the retarded child, and brothers may be found to be less disturbed than their sisters by the experience of having a retarded sibling (Gath, 1978, p69). This could not be investigated in the present study, but in general sisters have greater responsibilities in child-rearing than brothers and often carry younger siblings on their backs at quite a young age. It may be however,

that, if albinos are over-protected by their mothers, they are not left in the care of siblings as many other young children are. These aspects have yet to be examined in the local context.

5.3 The conclusions relating to the aims

5.3.1 Genetics

(i) Prevalence: the urban prevalence (1:3 900) was similar to that found in most other parts of Africa, but at least two or three times higher than is reported in many European countries. The rates for different ethnic groups varied, but were generally higher in the Sotho-speaking groups ($\pm$1: 2 500) and lower in the Nguni groups ($\pm$1:5 000), with the exception of the Swazi who were found to have high rates. Those groups with high prevalence rates were also found to have high consanguinity rates. The sex ratio was not found to diverge from the normal. The rural prevalence studies suggested that there are some semi-isolates in South Africa with high rates of albinism, such as the large village in Botswana and the Hhohho district of Swaziland, both of which had rates in excess of 1:2 000. These were higher than the urban rates for the same ethnic groups.

(ii) Identification, classification and description: The results on a co-operative sample of the subjects who had hair-bulb tests and physical examinations showed that although in most cases the types could be identified on the basis of these tests, there was difficulty in classifying some individuals. Nevertheless, the majority of albinos in this sample appeared to be of the tyrosinase-positive type, and very few were tyrosinase-negative. Brown and rufous albinos were also found and two families were described where the possible diagnosis was yellow mutant albinism. All the albinos appeared to have some pigment in the form of pale yellow hair, or a ring or spokes of pigment around the irides, although only about half the subjects in the large tyrosinase-positive group presented with pigmented ephelides.

(iii) Skin cancer: The risks for skin cancer were found to be high, to increase with age, and to vary according to ethnic group and geographical area. The Sotho group of albinos residing in Soweto had a statistically significantly higher rate of malignant and premalignant lesions than the Nguni albinos in Soweto, suggesting that other genetic factors which increase the susceptibility of the Sothos, might be involved. Both squamous and basal cell carcinomas were described, but the former was much more frequent than the latter. Malignant melanoma was not found in this series. The site most

often affected was the face, but other areas of the head and neck, and rarely the limbs and torso, could be affected.

(iv) Life expectancy: The results suggested that life expectancy is reduced. Specifically, male albinos were found to have a significantly different and younger age distribution than the general male population. Life expectancy appeared to vary according to geographical location and skin cancer rates, as well as sex of the individual.

(v) Skin colour: Skin colour was found to be lighter in Swazi heterozygous females than in matched controls. Anthropological studies suggest that a light-skinned person has often been considered the preferred mate. It is possible, therefore, that selective mating has occurred in this population favouring the individual heterozygous for the albinism gene.

(vi) Fertility: The albinos themselves were found to have a markedly decreased fertility. This could be due to their partial social stigmatization or partial rejection as marriage partners or there may be a physiological factor responsible. The matings between heterozygotes showed a similar fertility to that of the matched control group, and the grandparents of albinos had a slightly increased fertility (in terms of numbers of children per person) although the difference did not quite reach statistical significance.

5.3.2 Psychosocial studies

The aim of the psychosocial component of the study was to investigate the impact of the birth of an albino child on a black mother and father and, where possible, on other members of the family.

(i) The mothers: The mothers of albinos verbalized their distress and unhappiness and described their reluctance to hold and breast-feed their babies soon after the birth. They exhibited altered behaviour patterns and interacted less with their babies. This lack of the normal response to the baby probably indicated some depression, and an initial delay in the development of full maternal attachment. Some attachment seemed to have occurred by three months after the birth, and maternal behaviour and interaction with the baby was no longer different from the controls. Many mothers, however, still described feelings of sadness and unhappiness and their coping appeared to have been achieved more quickly in action than in thought. Unhappiness, was expressed until the infant was nine months by which time most mothers had accepted their babies, although they recognized that they would have excess problems and they had given them a wider variety of solids than the

controls babies had received. By one year the albino babies were reported to be slower in development; this could have been due to either the initial partial maternal deprivation, to the poor sight in the albino babies, or to the mothers' inaccurate reporting, or to a combination of these factors. The mothers continued through to the last interview at fifteen months, stating that they would prefer a `different' baby next time, and that their albino babies would have many problems. The impact on the mothers therefore, appears to have been long-lasting and, although the baby was generally well accepted and maternal-infant interaction established by nine months, nevertheless these reservations were still held at the end of the study, six months later. The findings therefore, suggest that a mild form of `chronic sorrow' might be a feature in mothers with albino children.

According to the mothers the grandmothers of the albino infants appeared to accept the baby and to assist and support their daughters to a greater extent than grandmothers of normal babies. The siblings showed a variety of responses, some indicative of quite extreme forms of rejection.

(ii) The fathers: The few available fathers, who were only interviewed some months after the birth, did not verbalize any negative feelings, but seemed to have accepted their affected babies, although according to the mothers they were unhappy. The birth of the albino did not seem to affect the fathers' reproductive plans. Nevertheless, the fathers showed reduced interaction with their children suggesting that there might have been some distancing and a lack of closeness between the father and his albino child, compared to fathers with normal babies. No reliable conclusions could be drawn however, due to the small sample size.

Although the study was exploratory the findings strongly suggest that the birth of an albino baby has an impact on the close family members causing them much unhappiness and resulting in altered behaviour patterns.

5.4 Suggestions for further research

5.4.1 Genetic studies

The question concerning why the prevalence of albinism should be so high in Africa, especially Southern Africa, where because of the high risk of sun exposure it is a most debilitating condition, is worth investigating further. The answer does not seem to be associated with the homozygote who appears not only to be susceptible to skin cancer, but also to have a shortened life expectancy and reduced fertility. Further investigations should therefore

probably be directed towards the heterozygote. One possible selective advantage for the heterozygote has been detected during the present study, and that is associated with the carrier's lighter skin colour which is reportedly considered beautiful by the local population. Further studies might be directed towards assessing present day trends regarding choice of marriage partners. If light-skinned mates are still being chosen in preference to those with darker skin colour, and if light-skinned people choose others with the same skin colour, the high prevalence of albinism could be maintained as long as these practices continue. Also, if, for example, light-skinned children are considered beautiful by their parents and are nurtured and given extra care and therefore have better survival rates than darker siblings, this could also contribute to a selective advantage for heterozygotes. Other possible heterozygote advantages, such as immunity to common endemic conditions might also be investigated.

The calculation of the gene frequency in the different ethnic groups could also be refined if data were collected on the consanguinity rates in the general population. The gene frequencies in the different ethnic groups might in fact be similar, but the prevalence rates might be higher in the Sotho-Tswana than in the Zulu, because the former favour consanguineous marriages. This theory cannot be tested at present because of the lack of data both for the general population, and on the specific relationship between some consanguineous couples in the sample of families with albino offspring.

Cultural factors surrounding the albinos in different ethnic groups and the societal attitudes towards them also require further investigation. During the present study, for example, two fathers were proud of having produced an albino and said they would like to have another, and the majority of fathers were not intending to restrict their plans for further children because of the presence of the albino. Therefore, if albinos are well accepted in some communities, but not in others, this might partially explain the differing ethnic group rates. Such detailed cultural data are not yet available.

Regarding the classification of albinos the present study was hampered by the fact that only data on hair-bulb incubation tests and on clinical descriptions on the subjects were collected. On this basis it appears to be difficult to classify some individuals. Further tests such as tyrosinase assays, the addition of cystein to the solution in which the hair-bulbs are

incubated, the examination of skin biopsies by electronmicroscopy, and visual acuity studies, are required if the tentative classification is to be confirmed. Visual evoked potentials have also been found to be helpful in making the diagnosis of albinism in Holland (Apkarian et al, 1983) and may be useful in confirming some of the border-line cases in the present series.

Another question that arose during the present study was why, in sibling pairs, skin pigmentation, particularly in the form of ephelides, could vary markedly, and the younger could have more than their older siblings. Also why pigmented ephelides occurred in some tyrosinase-positive individuals and not at all in others. Whether the development of pigmented patches is dependent upon life-style or genetic factors, merits further investigation, perhaps exploiting the twin pairs ascertained in this study.

Skin cancer in albinism also requires further research. Probably the most important area, from the practical point of view, concerns the apparent success of barrier creams in preventing skin cancer in albinos. This matter requires urgent attention if albinos are to be protected from the ill-effects of sun-exposure, including the high risk of developing solar keratoses some of which result in malignancies. Why the Sotho-Tswana albinos appear to be at greater risk of developing carcinomas than the Nguni albinos should also be investigated.

The reasons for the apparently shortened life expectancy, especially in males, as well as the causes of death in albinos should be assessed. It is generally assumed that skin cancer plays a major role in the morbidity and mortality of albinos, but there may be other contributory factors, such as susceptibility to infections, that may cause their early demise. It is noteworthy that two of the sample of thirty-eight new born albino babies in the present study died, whereas none of the control babies died in the first year of life. These babies died of gastroenteritis and mulnutrition, but whether these conditions were due to neglect, or infections and inefficient immune responses in the albino babies, is not apparent. Changing mating patterns might also contribute towards the disproportionate age distribution of albinos in the present study, and it would be useful to have more data on these practices and their fluctuations.

Another study which requires attention has been suggested by the apparently reduced fertility rates in albinos and the finding of increased melatonin levels in two albinos by Oosthuizen et al (1983). The study on mela-

tonin levels should be expanded and the association with fertility investigated.

In order to properly assess fertility in heterozygotes further data on fertility in the general population is required before accurate conclusions can be drawn. Information on trends and fluctuations in fertility, life-time infertility rates, and urban and rural statistics, are required for comparison with the data from the present study.

Suggestions for future research therefore include not only questions concerning albinism which have arisen from the present study, but also projects which involve basic research in the general population for which comparative data are frequently lacking.

5.4.2 Psychosocial studies

The psychosocial study also brought to light several unexplored areas which require elucidation. The data collected on fathers was limited and since the crisis situation occurs soon after the birth of the affected child and the mother showed the most dramatic changes in her behaviour at that stage, it would be worthwhile investigating the fathers' reaction in the neonatal period. Most of the fathers in the present study were only interviewed much later, when the initial effects of the birth had been tempered by time. For the same reasons and for the collection of further insights on the siblings' reactions, this group should also be interviewed within the first few weeks after the birth and perhaps observed in an interaction situation with the baby. The information on the grandmothers was secondhand and, in order to obtain a more complete picture, it would be worth extending future research work to include this important member of the family.

Most of the mothers in the present study stated that they had accepted their babies by the time they were nine months of age, whether this time span is typical of any mother with a defective baby and how long mothers generally take to adjust to such a birth would be an interesting topic to pursue. Many mothers claimed initially to be planning more education for the albino than for their other children and gave their albino children a greater variety of food at one year than in the controls. Whether or not such factors are indicative of the mothers' attempts to compensate for the defect by extra caring are debatable and worth investigating further.

The observations, although making use of crude methods of counting behaviours, provided some valuable material and this sort of study could be extended by using more precise methods of measurement, such as time-sampling.

The use of video film might also be reconsidered so that more specific details of the maternal-infant interaction might be detected, particularly regarding the albino baby's eye problems, the nature of the eye contact and visual communication. It is possible that the albino babies did not look or gaze at their mothers so frequently because of their nystagmus and visual defects; this could have caused the mothers initially to limit their behaviour and to reduce their interaction with the babies and this matter requires clarification.

The play situation at fifteen months and the results on the anxiety scale also indicated trends that should be pursued. The mothers appeared to sit further from their albino babies, when compared with control dyads. This suggests that there was a gap between the mother and her `different' baby, which might not close for a long time, and, in fact, an abnormal distance might persist. A long-term study extending perhaps into adolescence may be necessary to investigate this suggestion. Similarly, maternal anxiety in the mothers might continue or fluctuate, or may recede in time and this aspect requires confirmation and clarification. Anxiety should probably also be investigated in fathers, especially since the anxiety scale which was used with the mothers in the present study, has been validated for use in South Africa.

5.5 Applications and Recommendations

5.5.1 Genetic Studies

The genetic component of the study resulted in a number of findings which can be applied in different ways to the handling and treatment of albinos, and recommendations can be made which will be to their own, their family's and society's advantage.

Firstly, because of the high prevalence of the condition in the local population, education of the community is essential. Such education should probably be initiated in school biology courses and should include discussions on the cause of albinism and the genetics; on the types and the associated health problems, with particular emphasis on the high risks of skin cancer, on methods of preventing it, and on obtaining early treatment. This information may be communicated to the target group (the black scholars) via the teachers, and albinism should be one of the disorders stressed in any biology teachers' refresher course, which includes discussions on genetics.

Secondly, classification of individual albinos is possible and counsell-

ing of albinos in future should follow testing and determination of the type, since different types seem to have different health risks (Witkop, 1983). Very few tyrosinase-negative individuals were identified in the present study, but if these individuals are diagnosed their prognosis appears to be worse, particularly as regards eye problems and skin cancer, than the other types, and therefore more attention must be paid to emphasizing protective measures and to dealing with their problems. Similarly, turosinase-positive albinos would require more time and attention than brown and rufous albinos, who appear to have fewer problems.

Thirdly, the findings from the study can also be applied to the field of cancer treatment since the specific sites and the ethnic groups most at risk in the local situation have been identified. When anti-actinic creams are offered to the patients it should be emphasized that it is essential to apply these to the face including the ear, neck and eye-lids, and less importantly to the hands, arms and legs. Male albinos should probably get preference since they seem to be more susceptible (in Tanzania the male to female ratio in albinos with skin cancer was 9:1, Alexander and Henschke, 1981). At any clinic where albinos are treated a preventive health counselling service is strongly recommended. In particularly the albinos should be advised to take such protective measures as: using barrier skin creams; using sunshades or hats with large brims; wearing long sleeved dresses or shirts and long trousers (all clothing should be cotton rather than nylon, which does not seem as effective a barrier to the damaging ultraviolet rays, (Margolius, personal communication)); working indoors if possible; at least avoiding sunlight between 11.00 to 13.00 hours, thus reducing ultraviolet exposure by 50 per cent (Scotto and Fraumeni, 1982), and perhaps even from 10.00 to 15.00 hours as is recommended in Tanzania (Alexander and Henschke, 1981); and keeping the hair long, rather than shaving the head, which is sometimes practised locally. Among white populations the highest rates for skin cancer are in areas of greatest insolation, and one of the highest rates in the world has been found in South Africa (Scotto and Fraumeni, 1982). Albinos in South Africa therefore might have similar raised risks which are likely to be increased over those for albinos living elsewhere (excluding perhaps those in Equatorial regions). If the albinos follow the recommended advice their skin cancer risks should be reduced. If, however, sores develop they should be treated immediately as cure rates are generally high (very few skin cancers metastacize and only 1 per cent are fatal in the

USA, Scotto and Fraumeni, 1982). It has however been suggested that basal cell carcinomas may behave more aggressively in Africans, especially albinos, than in Caucasiods (Itayemi et al, 1979).

These recommendations can best be implemented by establishing a specialist clinic, specifically for albinos, and probably sited in a dermatology clinic. This approach has been taken in Dar-es-Salaam and found to be very effective (Alexander and Henschke, 1982). The setting-up of such a clinic has been discussed with local dermatologists at Baragwanath Hospital, Johannesburg, and moves have been made towards implementing the proposal.

Fourthly, questions are sometimes asked about life expectancy in the discussion of the prognosis for albinos. The information found during this study can be used to partially supply an answer based on the local situation. Parents can be advised that it seems that females have an almost normal, but males have a slightly reduced, life expectancy, but that it is possible, if they follow the recommendations concerning the avoidance of sun exposure, that life expectancy may be nearer to the norm for males, as well as females.

Fifthly, the skin colour studies have le d to the detection of lighter skin colour in heterozygotes, but at present this finding cannot be reliably applied to the detection of carriers in the general population, because of the wide range of skin colour which occurs. However, if general advice is required in a genetic counselling situation, and if a light sibling of a tyrosinase-positive albino requests information on how to avoid having an albino child (such a question is unknown in the writer's experience, but is a possibility for the future), marriage with a dark skinned person would probably reduce the risks, but more specific advice cannot yet be given.

The results of the present study therefore have many applications, and recommendations can be made which upon implementation will hopefully offer an improvement in the services for albinos, as well as in their quality of life, and in the understanding of their condition and its associated problems.

5.5.2 Psychosocial studies

The birth of an albino baby appears to have an effect on the family which is somewhat similar to the effect of any 'different' or handicapped child on the family. Strong feelings are aroused which cause behavioural disturbance and the families should be offered a counselling and support service to enable them to cope with these emotions, so that the affected babies do not become deprived of the love and attention they require. Such counselling

should also provide answers to the many questions asked by both parents about the condition and how to cope with it.

If a local clinic specifically for albinos and their families is established, it should provide a team service where the families have the opportunity to discuss their problems with a social worker or psychological counsellor, as well as obtaining medical treatment and preventive health counselling. The psychological counsellor and social worker should be aware of the chronic sorrow that can surround the birth of a child who is `different', and of the sadness and depression which can follow. Such counsellors should also be informed on the reactions of mothers and significant others, on their feelings of inadequacy, ambivalence and vulnerability, and on the initial crisis which appears to result in delayed maternal-infant interaction, which all require sensitive, supportive and accepting handling. Crisis counselling, with `a little help rationally directed and purposefully focussed at a stategic time is more effective than more extensive help given at a period of less emotional accessibility' (Rapoport, 1970, p287), and should be included in the role of the counsellor. The introduction of peer group counselling by selected, well-adjusted and trained mothers of albinos should also be considered, since such a service is much appreciated by mothers with babies with other disorders in the writer's experience. The families involved in the present study showed a need for such a service and a genuine concern for their albino child, so that they would be well motivated to use the specialist clinic if it was organized.

Another proposal that was discussed during the research work concerned therapy to assist the mother in coping with her partially sighted child. A local service has been initiated specifically for blind infants, and their mothers, to improve maternal-infant interaction, infant mobility, performance, the achievement of skills, and the mothers' awareness of the problems and ability to cope. This service has been offered to the mothers of partially sighted albino children, and the mothers should be encouraged to have their affected children visually assessed, and to make use of this service if necessary.

Families with many different inherited disorders have been found to benefit from having an Association of their own. This has been found to be the case in the Albino Fellowship in Great Britain (Taylor, 1980). This fellowship aims to provide mutual support and information for affected families and their community. Such an Association would meet many of the social

and emotional needs that have been high-lighted in the present study, could also assist in the education of both the albinos and the community regarding albinism and its problems, and could encourage participation in a specialist clinic. The formation of a local Albino Fellowship or group is therefore strongly recommended.

Genetic counselling is available in greater Johannesburg but the clinic is situated in the city centre far from where the majority of blacks live, and they do not often use the service. A second service provided in conjunction with the specialist dermatology clinic for albinos is recommended and a genetic counsellor could be included in the team. Most parents in the present study planned further children in apparent ignorance of the nature of the recurrence risks. The genetic counselling that is provided should be the broadly defined type, as proposed by Fraser (1974), which includes assisting the family and individual in five areas: the comprehension of the medical facts, diagnosis, prognosis and management; the appreciation of the genetics and risk of recurrence; the discussion of options for dealing with the risk; the choice and instigation of an appropriate course of action; and the making of the best possible adjustment to the disorder. The families in the present study seemed to indicate needs in every one of these areas and would benefit from such a broadly based service.

The psychosocial studies highlighted certain needs and the establishment of services to meet these needs should be the practical outcome of the study.

5.6 Summary and Conclusion

An attempt has been made in the present study to gain new insights into seven different aspects of albinism on which detailed information was lacking locally. In brief the findings indicate that: the prevalence of albinism in Southern Africa is similar to that in other African countries, but varies between local ethnic groups; most of the albinos fall into the tyrosinase-positive group, and almost all have some form of pigment; skin cancer risks are high especially for those of the Sotho-Tswana group; life expectancy is reduced, for males in particular; skin colour is lighter in heterozygotes and this appears to give them a selective advantage; fertility is reduced in albinos, but is normal in their parents and grandparents; the birth of an albino baby has a profound impact on the mother causing her to experience depression and sadness for on average, nine months, and to interact less with the baby in the neonatal period, and according to the mother the birth also affected the father, siblings and grandmother in different ways.

During the study it became apparent that various topics require further research. Why the prevalence is high is curious and whether the heterozygotes have other selective advantages, apart from a lighter skin colour, that could account for the high prevalance is not as yet known. Methods of preventing skin cancer must be tested and the best preventive treatment offered to albinos. Why life expectancy is shorter especially in males must be determined and life-styles particularly require investigation. In addition, insufficient data were collected during the present study to elucidate fully the effect of the birth of an albino baby on the father, and whether mothers experience some form of sorrow and some distancing from their affected child in the long-term is still to be investigated.

Upon consideration of the study's conclusions recommendations can be made. A specialist clinic service should be established with medical treatment, health, genetic and psychological counselling being provided for albinos and their families. An Albino Fellowship group should also be initiated so that affected persons and their families can meet, share their problems, promote education of themselves and the community, and encourage the formation of and participation in a specialist clinic.

Although this study was focussed on the albino in a black African society the findings may have implications relating to broader issues. The birth of a defective baby probably constitutes a stressful life-event for any mother and may cause a delay in the achievement of maternal attachment regardless of the nature of the defect. The disruptive psychosocial effects of such a birth might result in the mother experiencing several stages in coming to terms with the event. These stages may be similar to those associated with grieving after the death of a loved one or with the acceptance of ones own approaching death in a terminal situation. The progress through these stages may vary and fluctuate and acceptance may be achieved at different times at different levels of functioning. From the present study it appears that of the cognitive, affective and behavioural components of the mother's attitude towards her affected baby it is functioning at the behavioural level which changes first. If three stages could be identified and derived from the data they might be: first a stage (within the first month after the birth) when the mother could not accept the child at any level and showed unhappiness and reduced interaction with the baby; second, a stage (at about three months after the birth) when the mother was still unhappy and not accepting on the cognitive and affective levels but able to behave

normally with her baby; third, a last stage (at about nine months) where the mother showed cognitive and behavioural acceptance and implied that affective acceptance was achieved as well.

The present study, however, was an exploratory one and was not chiefly concerned with theories; and theoretical inferences, therefore, cannot easily be made. Further theorising will have to deal with such issues as: crisis theory and the experiences of parents at the birth of a defective infant; how long a mother takes to adjust to the infant; through what stages she passes to achieve such adjustment and acceptance; what attributes those mothers who adjust quickly have; whether mothers compensate for their child's defect by extra caring; and whether some societies are more caring in relevant situations than others (either directly by encouraging mothers to care, or indirectly by providing social support services).

The nature of the factors which determine interaction between individuals and, in particular, between mother and child remains to be clarified. It seems that maternal-infant attachment is not purely instinctive, but to a certain extent, may be determined by the infants' physical and/or temperamental characteristics, as well as the mothers' personality, parental attitudes and practices, together with various environmental factors. As Emde and Brown (1975) state `it is difficult to appreciate the impact on a mother of a smile which disappoints or of a look which is fleeting instead of engaging' (which is likely to be the case in the albino baby). Virgil (about 30 BC) too recognized the importance of the infant's contribution and wrote `incipe, parve puer, risu cognoscere matrem' (begin, baby boy, to know your mother with a smile). The writer would therefore propose an interactionist approach, and the findings of the present study appear to point to the necessity of taking all the factors relating to the child and the mother into consideration, when investigating maternal attachment, maternal-infant interaction, or the impact of the birth of a defective baby on the parents.

The visual defect of the albino infant is one of the problems which occurs at the interface between the genetic and psychosocial aspects associated with albinism. This defect is caused by the presence of the albinism genes, but it might have an effect on maternal-infant gaze and the concomitant development of maternal-infant interaction. Another such problem for the albinos is the development of skin cancer, which is partly caused by various psychosocial factors, which influence life-style interacting with both environmental factors, such as high sunshine rates, and their susceptibility due

to their genetic constitution. The lesions that develop, if they become unsightly, may further lead to some societal rejection, specifically where choice of a marriage partner is concerned. Albinism is therefore a good example of a genetic condition which, in the black population at least, has wide-ranging psychosocial effects, which are only beginning to be elucidated.

In conclusion the present study is a contribution towards the solution of the multi-faceted problems posed by birth defects. The prevalence of the problem has been seen to be associated partly with the ways in which local living is organized and the health hazards are the result of behaviour which is not harmonized with the environment. These aspects of genetic disorders have not been given priority but attention to them is essential if the understanding of the effects of disorders on individuals, families and communities is to keep pace with the developments on the biochemical and molecular levels.

APPENDIX A

Form for Survey of Albinos

Name: File No:

Address:

Ascertained through: Date:

Date of Birth: Sex: Tribe:

Place of Birth:

Residences:

Employment: Present:

Past:

Income:

Education and school career:

Periods of hospitalization: (Dates and diagnoses)

Year of marriage:

Spouse normally pigmented? (heresay/seen)

Spouse blood relative?

Offspring: (including miscarriages)

	Name	Date of birth	Sex	Albino	
1					If Yes: see No:
2					
3					
4					
5					
6					
7					
8					

Any other albino relatives?

Draw pedigree on separate sheet.

APPENDIX B

Medical Examination

Name: File No:

Date:

Accompanied by:

Ethnic affiliations:

Build and Health Status:

Skin: (describe and note malignant and premalignant lesions)

Exposed areas:

Unexposed areas:

Ephelides:

Naevi:

Hair:

Colour:

Tyrosinase test:

Eyes:

Pigmentation:

Transluscency:

Nystagmus:

Photophobia:

Teeth:

Trunk:

Heart:

Lungs:

Blood collected:

Photographs taken:

Remarks:

Signature:

APPENDIX C

Schedule I (for Mothers of New-Born Babies)

(for Interview 1)

Researcher: Mrs J G R Kromberg

Department of Human Genetics, School of Pathology,
The South African Institute for Medical Research
and University of the Witwatersrand, Johannesburg.

Introduction

The researcher is a social worker who is interested in the relationship between mothers and fathers and their new-born babies. She would like to find out how this relationship develops and therefore she would be glad of your help. If you agree to help there will be one visit to you every three months, with six visits altogether in fifteen months. At every visit you will be asked questions and if you answer these questions honestly and thoughtfully you will understand yourself and your baby better and you will help the research worker to understand you both too.

There are no right or wrong answers to these questions, just tell the interviewer what you think in each case.

File No:

Name of mother: Name of baby:

Address: Date of birth:

Interviewer:

Interview No: Date:

I. Identifying Details

1. Name Mother: Date of Birth/Age:
2. Maiden Name:
3. Name Father: Date of Birth/Age:
4. Address:
5. Married/Single/Divorced/Widowed:
6. Tribe: Mother: Father:
7. Consanguinity:
8. Occupation: Mother: Father:
9. Education: Mother: Father:
10. Living together:
11. Children

Name	Sex	Age	Alive/Dead	Albino	Father's Name

12. Abortions: Yes/No No:
    Miscarriages: Yes/No No:
    Stillborns: Yes/No No:

Medical History

13. Operations: Yes/No/Don't know
    Why:
14. Fractures: Yes/No/Don't know
15. Hospitalization: Yes/No/Don't know
    Why/where:
16. Headaches: Yes/No/Don't know
    How often:
17. Sleeplessness: Yes/No/Don't know
    How often:
18. Tiredness: Yes/No/Don't know
    How often:
19. Stomach pain Yes/No/Don't know
    How often:
20. Breathlessness: Yes/No/Don't know
    How often:
21. Depression: Yes/No/Don't know
    How often:
22. Feel well/sick (grumble factor):

Childhood history

23. Brought up by: Mother/Father/Both/Relatives:
24. Rural/urban childhood:
25. Number of siblings:
26. History of albinism: Yes/No:
    Affected relative/s:
    (draw pedigree on separate sheet)

II. Items on Baby

Weight: Sex:

1. Did anything unusual happen to you during the pregnancy: Yes/No
   Describe:
2. Planned pregnancy: Yes/No
3. Length of pregnancy - full-term/premature:
4. Special care in neonatal period:

   Baby: Yes/No
   Why:

   Mother: Yes/No
   Why:

   Separation from Mother: Yes/No
   How long:

   Why:
5. Breast/Bottle feeding:

6. Wanted: Boy/Girl: Mother: Father:
7. Who does baby look like:
8. Do you like the way the baby looks: Yes/No
   Why:
9. How do you feel about the baby:
   What do you think about the baby:
10. What does the father feel about the baby:
11. What do grandparents say about the baby. Maternal:
    Paternal:
12. What did siblings say about the baby:
    Were they happy/sad:
13. Did you want to hold the baby straight away: Yes/No
14. Did you want to breast-feed the baby: Yes/No
15. Does the baby look beautiful/ugly:
16. What do you think caused the baby to look the way it does:
17. Is the baby good: Yes/No
18. Do you show the baby to your friends: Yes/No/Don't know
19. Does baby feed well: Yes/No/Don't know
20. Does baby sleep well: Yes/No/Don't know
21. Does baby cry a lot: Yes/No/Don't know
22. Is there a nickname: Yes/No
    What is it:
23. What are your education plans for the baby:
24. What is relationship like between yourself and the father of this child:
    Now:
    Before birth:
25. Is baby different from previous baby: How: Yes/No
26. Will you give this baby more clothes: Yes/No
27. Will you give this baby more food: Yes/No
28. Will you give this baby more schooling: Yes/No
29. Are sibs allowed to handle baby: Yes/No
30. Are sibs attending school regularly:
    Now: Yes/No
    Before birth: Yes/No
31. Do sibs have friends to play: Yes/No
32. Do sibs show baby to friends: Yes/No
33. Is father at home more/less often than before birth:
34. Does father drink: Yes/No
    Now: Yes/No
    Before birth: Yes/No
    More/Less:

35. Is father giving attention to sibs: Yes/No
    Now: Yes/No
    Before birth: Yes/No

36. Are you planning to have another baby:
    Father: Yes/No
    Mother: Yes/No
    If you have another baby, would you like it to look like this one: Yes/No

37. Do you visit friends with the baby: Yes/No
    Now: Yes/No
    Before birth: Yes/No

38. Do you visit friends without baby: Yes/No

39. Do you have any complaints about the father: Yes/No
    What:

40. Do you have any complaints about the siblings: Yes/No
    What:

41. Do you have any complaints about the baby: Yes/No
    What:

42. Any questions:

43. Any comments:

APPENDIX D

Schedule II (for Mothers of New-Born Babies)

(for Interview 2)

File No:
Albino/Control

Name of mother: Name of baby:

Address: Date of birth:

Interviewer:

Interview No: Date:

I. Medical History (since I last saw you)

1. Have you had an operation: Yes/Don't know/No
   Why:
2. Have you had a fracture: Yes/Don't know/No
   Why:
3. Have you been in hospital: Yes/Don't know/No
   Why:
4. Do you have frequent headaches: Yes/Don't know/No
   Why:
5. Do you have difficulty getting to sleep at night: Yes/Don't know/No
   Why:
6. Do you wake earlier than you need to in the morning: Yes/Don't know/No
   Why:
7. Do you feel tired all the time: Yes/Don't know/No
   Why:
8. Have you been suffering from stomach pain: Yes/Don't know/No
   Why:
9. Have you been getting breathless recently: Yes/Don't know/No
   Why:
10. Have you been feeling depressed/sad: Yes/Don't know/No
    Why:
11. Do you often have bad dreams: Yes/Don't know/No
    Why:
12. How do you feel generally (grumble factor): Good/Don't know/Bad
    Why:
13. Has there been a change in your health: Yes/Don't know/No
    Why:

II. Items on Maternal/Infant relationship

14. How are you feeding your baby: Breast/Bottle/Both
    Why:
15. Who does baby look like:
16. Do you like the way the baby looks: Yes/Don't know/No
    Why:

17. Do you feel disturbed that the baby is like he is: Yes/Don't know/No
    Why:
18. How do you feel about the baby now: Happy/Don't know/Sad
    Why:
19. How does the father feel about the baby now: Happy/Don't know/Sad
    Why:
20. What do your parents feel about the baby: Happy/Don't know/Sad
    Why:
21. What do they say:
22. How do the father's parents feel: Happy/Don't know/Sad
23. What do they say:
24. What do your other children feel about the baby: Happy/Don't know/Sad
25. What do they say:
26. Does the baby look beautiful/don't know/ugly to you:
27. What do you think caused the baby to look the way he/she does:
28. Is the baby good: Yes/Don't know/No
    Why:
29. Do you show the baby to your friends: Yes/Don't know/No
    Why:
30. Does baby feed well: Yes/Don't know/No
31. Does baby sleep well: Yes/Don't know/No
32. Does baby cry too much: Yes/Don't know/No
33. Is baby rolling over: Yes/Don't know/No
34. Is baby sitting up alone: Yes/Don't know/No
35. Is baby crawling: Yes/Don't know/No
36. Is baby walking: Yes/Don't know/No
37. Is baby saying any words: Yes/Don't know/No
38. Is there a nickname for the baby: Yes/Don't know/No
    What:
39. What are your plans for the baby in the next two years:
40. What is the relationship like between yourself and the father of this child: Good/Don't know/Bad
    Why:
41. Is he supporting this baby: Yes/Don't know/No
42. Are you happy with the sexual relationship: Yes/Don't know/No
43. Are you happy with your relationship with the father: Yes/Don't know/No
    Why:
44. Is this baby different from the previous baby: Yes/Don't know/No
    Why:
45. Do you expect the baby to remain here: Yes/Don't know/No
    Why:

46. What are your long-term plans for the baby:

47. Will you give this baby more clothes: Yes/Don't know/No
Why:

48. Will you give this baby more food: Yes/Don't know/No
Why:

49. Will you give this baby more schooling: Yes/Don't know/No
Why:

50. Do you enjoy your children: Yes/Don't know/No
Why:

51. Are your other children allowed to play with this child: Yes/Don't know/No
Why:

52. Do your children play with this child: Yes/Don't know/No
Why:

53. Do the siblings attend school regularly: Yes/Don't know/No
Why:

54. Do the siblings bring friends home here to play: Yes/Don't know/No

55. Do the siblings show this baby to their friends: Yes/Don't know/No
Why:

56. Do the siblings like this baby: Yes/Don't know/No
why:

57. Is the father at home less often than before the birth: Yes/Don't know/No

58. Does the father drink: Yes/Don't know/No

59. Is the father drinking now: Yes/Don't know/No
Why:

60. Is the father giving attention to the siblings: Yes/Don't know/No

61. Is the father interested in this baby: Yes/Don't know/No
Why:

62. Are you planning to have another baby: Yes/Don't know/No
Why:

63. Does the father want another baby: Yes/Don't know/No
Why:

64. If you have another baby, would you like it to look like this one: Yes/Don't know/No
Why:

65. Do you often visit friends: Yes/Don't know/No
Why:

66. Do you take the baby with you when you visit friends: Yes/Don't know/No

67. Do you think the baby will have any particular problems: Yes/Don't know/No
Why:

68. Do you think this child will have more problems than other children: Yes/Don't know/No
Why:

69. Do you think other children will accept this child as a friend: Yes/Don't know/No
    Why:
70. Do you live in the same house as your mother: Yes/Don't know/No
71. If she lives in another house how often do you see her: Once a day/twice a week/once a week/other (specify):
    Why:
72. Do you get much support and help from her: Yes/Don't know/No
    Why:
73. Has there been any change in the attitude of your relatives since the birth of this baby: Yes/Don't know/No
    Why:
74. Do you have anything you wish to add:
75. Do you have any questions to ask:

Interviewer's comments:

APPENDIX E

Schedule III (for Mothers of New-born babies)

(for interviews 3 - 6)

File No:
Albino/Control

Name of mother:

Address:

Name of baby: Weight: Length:

Interviewer:

Interview No: Date:

I. Medical History (Since I last saw you)

1. Have you had an operation: Yes/Don't know/No
   Why:
2. Have you had a fracture: Yes/Don't know/No
   Why:
3. Have you been in hospital: Yes/Don't know/No
   Why:
4. Have you had a headache in the last 2 weeks: Yes/Don't know/No
   Why:
   How many:
5. Did you find it difficult getting to sleep at night in the last 2 weeks: Yes/Don't know/No
   Why:
6. Did you wake earlier than you needed to in the morning in the last 2 weeks: Yes/Don't know/No
   Why:
7. Do you feel tired all the time: Yes/Don't know/No
   Why:
8. Have you had a stomach pain in the last 2 weeks: (excluding menstrual pain): Yes/Don't know/No
   Why:
9. Have you been feeling breathless in the last 2 weeks: Yes/Don't know/No
10. Have you felt very depressed/sad in the last 2 weeks: Yes/Don't know/No
    Why:
11. Do you often have bad dreams: Yes/Don't know/No
    What is the message of the dreams:
    Why:
12. How do you feel generally (grumble factor) Good/Don't know/Bad
    Why:

    (a) Are you eating well: Yes/No

    (b) What are your bowel habits:

13. Has there been a change in your health: Yes/Don't know/No

    (a) Have you been to anyone for medical treatment: Yes/Don't know/No
    Why:
    To whom:

    (b) Has the baby's father been for treatment: Yes/Don't know/No
    Why:
    To whom:

    (c) Have the children been for treatment: Yes/Don't know/No
    Why:
    To whom:

    (d) Who took the children to the Doctor:

    (e) Did the father give the money for the treatment: Yes/No

    (f) Did the father ask questions about the child after the visit to the doctor: Yes/No

II. Items on Maternal/Infant relationship (Since I last saw you)

14. How are you feeding your baby: Breast/Bottle/Both
    Why:

    (a) Is the baby eating solids now: Yes/No
    What:

15. Has the baby had diarrhoea and vomiting: Yes/No

16. When did you start taking the baby to the clinic: Date:
    (a) Do you take the baby regularly: Yes/No

    (b) Has the baby been immunized: Yes/No

17. Do you feel disturbed that the baby is like he is: Yes/Don't know/No
    Why:

18. How do you feel about the baby now:

19. How does the father feel about the baby now:

(20-26) Omitted for interview III

27. What do you think caused the baby to look the way he does:

28. Is the baby healthy: Yes/Don't know/No
    Why:

29. Do you show the baby to your friends: Yes/Don't know/No
    Why:

30. Does the baby feed well: Yes/Don't know/No

31. Does the baby sleep through the night: Yes/Don't know/No

32. Does the baby cry more than other babies: Yes/Don't know/No

    (a) At what age did the baby smile:

33. Is the baby rolling over: Yes/Don't know/No

34. Is the baby sitting up alone unsupported: Yes/Don't know/No

35. Is the baby crawling on hands and knees: Yes/Don't know/No

36. Is the baby walking without support: Yes/Don't know/No

37. Is the baby saying words: Yes/Don't know/No
    What words:

38. Is there a nickname for the baby: Yes/Don't know/No
    What:
39. What are your plans for the baby in the next two years:
    (a) Where is the baby going to live:
40. What is the relationship like between yourself and the father of this child:
41. Is he supporting the baby: Yes/Don't know/No
    (a) Did he give you money for the baby in the last month: Yes/No
42. Are you happy with the sexual relationship between yourself and the father of this child: Yes/Don't know/No
43. Did father take baby out with him to see other people in the last 2 weeks: Yes/No
44. Did the father feed the baby in the last 2 weeks: Yes/No
45. Is the baby different from the previous baby now: Yes/No
    How:
46. What do you want baby to be when he is big:
47. Will you give this baby more clothes than your other children: Yes/Don't know/No
    Why:
48. Will you give this baby more food: Yes/Don't know/No
    Why:
49. Will you give this baby more schooling: Yes/Don't know/No
    Why:
50. Do you enjoy your children Yes/Don't know/No
    Why:
51. Are your other children allowed to play with this child: Yes/Don't know/No
    Why:
52. Do your children play with this child: Yes/Don't know/No
    Why:
    (a) Do your children take this child out of the the house to play: Yes/Don't know/No
53. Have siblings missed school in the last 2 weeks: Yes/Don't know/No
    Why:
54. Have the siblings brought friends home here to play in the last 2 weeks: Yes/Don't know/No
55. Have the siblings shown this baby to their friends in the last 2 weeks: Yes/Don't know/No
    Why:
56. Are the siblings afraid of this baby: Yes/Don't know/No
    Why:
57. Is the father at home less often than before the birth: Yes/Don't know/No
    Why:

58. Does the father drink: Yes/Don't know/No

    (a) Has the father been drinking heavily in the past 2 weeks: Yes/Don't know/No
    Why:

59. Has the father been drunk (with staggering walk) in the last 2 weeks: Yes/Don't know/No

60. Has the father played with the other children in the last 2 weeks: Yes/Don't know/No

61. Did the father participate in the care of the baby in the last 2 weeks: Yes/Don't know/No
    Why:

62. Are you planning to have another baby: Yes/Don't know/No
    Why:

    (a) Are you using contraceptives now: Yes/Don't know/No
    Why:

63. Does the father want another baby: Yes/Don't know/No
    Why:

64. If you have another baby, would you like it to look like this one: Yes/Don't know/No
    Why:

65. Have you visited friends in the last 2 weeks: Yes/Don't know/No
    Why:

66. Did you take the baby with you when you visited friends: Yes/Don't know/No
    Why:

67. What problems (different from the problems of other children) do you think the baby will have:
    Why:

68. Do you think this child will get sick more often than other children: Yes/Don't know/No
    Why:

    (a) Do you think he will be cleverer at school: Yes/Don't know/No

69. Do you think other children will be friends with this child: Yes/Don't know/No
    Why:

    (a) Do you think he will be bad tempered: Yes/Don't know/No

70. Do you live in the same house as your mother: Yes/Don't know/No

71. If she lives in another house how often do you see her:
    Once a day/twice a week/once a week/ other (specify)
    Why:

72. Has your mother looked after the baby for you in the last 2 weeks: Yes/Don't know/No
    Why:

    (a) Has your mother given you money for the baby in the last 2 weeks: Yes/Don't know/No

(b) Has your mother talked with you and comforted you in the last 2 weeks: Yes/Don't know/No

73. Did your relatives visit you in the last 2 weeks: Yes/Don't know/No
Why:

74. Do you have anything you wish to add:

75. Do you have any questions to ask:

Interviewer's comments:

APPENDIX F

ANXIETY SCALE

BIOGRAPHICAL INVENTORY

Please indicate whether you experience the feelings described below by making a cross over the appropriate word:

1. I believe I am no more nervous than most others.	False/True
2. I work under a great deal of strain.	True/False
3. I cannot keep my mind on one thing.	True/False
4. My feelings are more easily hurt than those of most other people.	True/False
5. I often find myself worrying about something.	True/False
6. I am usually calm and not easily upset.	False/True
7. I feel anxious about something or someone almost all of the time.	True/False
8. I am happy most of the time.	False/True
9. At times I am so restless that I cannot sit in a chair for very long.	True/False
10. I have often felt that I faced so many difficulties that I could not overcome them.	True/False
11. I certainly feel useless at times.	True/False
12. I find it hard to keep my mind on a task or job.	True/False
13. I am more self-conscious than most people.	True/False
14. I am the kind of person who takes things hard.	True/False
15. I am a very nervous person.	True/False
16. Life is often a strain for me.	True/False
17. At times I think I am no good at all.	True/False
18. I am not at all confident of myself.	True/False
19. At times I feel that I am going to crack up.	True/False
20. I don't like to face a difficulty or make an important decision.	True/False

(This scale was also available in Tswana, Southern Sotho and Xhosa).

APPENDIX G

Schedule IV (for Fathers of New-Born Babies)

(for Interview 1)

Researcher: Mrs J G R Kromberg

Department of Human Genetics, School of Pathology,
The South African Institute for Medical Research
and University of the Witwatersrand, Johannesburg.

Introduction

The researcher is a social worker who is interested in the relationship between mothers and fathers and their new-born babies. She would like to find out how this relationship develops and therefore she would be glad of your help. If you agree to help there will be one visit to you every six months, with three visits altogether in fifteen months. At every visit you will be asked questions and if you answer these questions honestly and thoughfully you will understand yourself and your baby better and you will help the research worker to understand you both too.

There are no right or wrong answers to these questions, just tell the interviewer what you think in each case.

File No:

Name of father: Name of baby:

Address: Date of birth:

Interviewer:

Interview No: Date:

I. Medical History

1. Have you ever had an operation: Yes/Don't know/No
   Why:
2. Have you ever had a fracture: Yes/Don't know/No
   Why:
3. Have you ever been in hospital: Yes/Don't know/No
   Why:
4. Do you have frequent headaches: Yes/Don't know/No
   Why:
5. Do you have difficulty getting to sleep at night: Yes/Don't know/No
   Why:
6. Do you wake earlier than you need to in the morning: Yes/Don't know/No
   Why:
7. Do you feel tired all the time: Yes/Don't know/No
   Why:
8. Have you been suffering from stomach pain: Yes/Don't know/No
   Why:
9. Have you been getting breathless recently: Yes/Don't know/No
   Why:

10. Have you been feeling depressed/sad: Yes/Don't know/No
    Why:
11. Do you often have bad dreams: Yes/Don't know/No
    Why:
12. How do you feel generally (grumble factor): Good/Don't know/Bad
    Why:
13. Has there been a change in your health: Yes/Don't know/No
    Why:

II. Items on Paternal/Infant relationship

14. Who does baby look like:
15. Do you like the way the baby looks: Yes/Don't know/No
    Why:
16. Do you feel disturbed that the baby is like he is: Yes/Don't know/No
    Why:
17. How do you feel about the baby now: Happy/Don't know/Sad
    Why:
18. How does the mother feel about the baby: Happy/Don't know/Sad
    Why:
19. What do your parents feel about the baby: Happy/Don't know/Sad
    Why:
20. What do they say:
21. How do the mother's parents feel: Happy/Don't know/Sad
22. What do they say:
23. What do your other children feel about the baby: Happy/Don't know/Sad
24. What do they say:
25. Does the baby look beautiful/don't know/ugly to you:
26. What do you think caused the baby to look the way he does:
27. Is the baby good: Yes/Don't know/No
    Why:
28. Do you show the baby to your friends: Yes/Don't know/No
    Why:
29. Does baby feed well: Yes/Don't know/No
30. Does baby sleep well: Yes/Don't know/No
31. Does baby cry too much: Yes/Don't know/No
32. Is baby rolling over: Yes/Don't know/No
33. Is baby sitting up alone: Yes/Don't know/No
34. Is baby crawling: Yes/Don't know/No
35. Is baby walking: Yes/Don't know/No
36. Is baby saying any words: Yes/Don't know/No
37. Is there a nickname for the baby: Yes/Don't know/No
    What:

38. What are your plans for the baby in the next two years:

39. What is the relationship like between yourself and the mother of this child: Good/Don't know/Bad
Why:

40. Is she caring for this baby: Yes/Don't know/No

41. Are you happy with the sexual relationship: Yes/Don't know/No

42. Are you happy with your relationship with the mother: Yes/Don't know/No
Why:

43. Is this baby different from the previous baby: Yes/Don't know/No
Why:

44. Do you expect the baby to remain here: Yes/Don't know/No
Why:

45. What are your long-term plans for the baby:

46. Will you give this baby more clothes: Yes/Don't know/No
Why:

47. Will you give this baby more food: Yes/Don't know/No
Why:

48. Will you give this baby more schooling: Yes/Don't know/No
Why:

49. Do you enjoy your children: Yes/Don't know/No
Why:

50. Are your other children allowed to play with this child: Yes/Don't know/No
Why:

51. Do your children play with this child: Yes/Don't know/No
Why:

52. Do the siblings attend school regularly: Yes/Don't know/No
Why:

53. Do the siblings bring friends home here to play: Yes/Don't know/No

54. Do the siblings show this baby to their friends: Yes/Don't know/No
Why:

55. Do the siblings like this baby: Yes/Don't know/No
why:

56. Is the mother at home less often than before the birth: Yes/Don't know/No

57. Does the mother drink: Yes/Don't know/No

58. Is the mother drinking now: Yes/Don't know/No
Why:

59. Is the mother giving attention to the siblings: Yes/Don't know/No

60. Is the mother interested in this baby: Yes/Don't know/No
Why:

61. Are you planning to have another baby: Yes/Don't know/No
Why:

62. Does the mother want another baby: Yes/Don't know/No
    Why:
63. If you have another baby, would you like it to look like this one: Yes/Don't know/No
    Why:
64. Do you often visit friends: Yes/Don't know/No
    Why:
65. Do you take the baby with you when you visit friends: Yes/Don't know/No
66. Do you think the baby will have any particular problems: Yes/Don't know/No
    Why:
67. Do you think this child will have more problems than other children: Yes/Don't know/No
    Why:
68. Do you think other children will accept this child as a friend: Yes/Don't know/No
    Why:
69. Do you live in the same house as your mother: Yes/Don't know/No
70. If she lives in another house how often do you see her: Once a day/twice a week/once a week/other (specify):
    Why:
71. Do you get much support and help from her: Yes/Don't know/No
    Why:
72. Has there been any change in the attitude of your relatives since the birth of this baby: Yes/Don't know/No
    Why:
73. Do you have anything you wish to add:
74. Do you have any questions to ask:

Interviewer's comments :

APPENDIX H

Observation Form (for mothers of New-Born babies)

(for Pilot Study)*

Instructions to mothers: We are trying to learn how mothers and babies respond to each other and we would like your help. Would you please pick up your baby, put him/her on the bed, change his/her napkin and replace him/her in the crib. There is no right or wrong way of doing this and there is no hurry, just be yourself and act as you would normally with your baby.

Name: Observer: Date:

1. Eye-to-eye contact	Yes/No/Don't know
2. *En face* position	Yes/No/Don't know
3. Smiles at baby	Yes/No/Don't know
4. Talks to baby	Yes/No/Don't know
5. Sings to baby	Yes/No/Don't know
6. Kisses baby	Yes/No/Don't know
7. Holds baby close	Yes/No/Don't know
8. Supports head	Yes/No/Don't know
9. Strokes baby	Yes/No/Don't know
10. Places baby securely on bed	Yes/No/Don't know
11. Handles baby carefully	Yes/No/Don't know
12. Names baby	Yes/No/Don't know
13. Appears relaxed	Yes/No/Don't know
14. Keeps baby clean	Yes/No/Don't know
15. Settles baby well	Yes/No/Don't know
16. Appears to care	Yes/No/Don't know
17. Asks questions about baby	Yes/No/Don't know

Observer's comments:

* The same form, excluding item 8, was used in the final study.

APPENDIX I

Observation Form (for fathers of New-Born babies)

(Instructions to fathers: We are trying to learn how fathers and babies respond to each other and we would like your help. Would you please pick up your baby and hold him/her for me while I make some notes on his appearance, and then put him/her down again).

Name: Observer: Date:

1. Eye-to-eye contact	Yes/No/Don't know
2. En face position	Yes/No/Don't know
3. Smiles at baby	Yes/No/Don't know
4. Talks to baby	Yes/No/Don't know
5. Sings to baby	Yes/No/Don't know
6. Kisses baby	Yes/No/Don't know
7. Holds baby close	Yes/No/Don't know
8. Strokes baby	Yes/No/Don't know
9. Handles baby carefully	Yes/No/Don't know
10. Names baby	Yes/No/Don't know
11. Appears relaxed	Yes/No/Don't know
12. Keeps baby clean	Yes/No/Don't know
13. Settles baby well	Yes/No/Don't know
14. Appears to care	Yes/No/Don't know
15. Asks questions about baby	Yes/No/Don't know

Observer's comments:

APPENDIX J

Observation form (for play situation at interview 6)

Instructions

We are interested in observing your baby at play. Here are some toys with which he/she can play. Please sit on this mat with him/her. You may do as you please.

Behaviours for counting

	(Count each response with 1 stroke)			
	MOTHER	TOTAL	BABY	TOTAL
TOUCHING				
HOLDING				
SMILING				
GAZING				
DISTRESS				
VOCALIZATION				
FUSS CRY				
MEDIATING ENVIRONMENT				
BEHAVIOUR NOT CONTINGENT TO INFANT				
PHYSICAL DISTANCE (MAX) BETWEEN MOTHER AND CHILD				

APPENDIX K

TABLE I Comparison for headaches and depression in experimental and control mothers at first interview (N = 37)

	Response					
	Tied pairs		Differing pairs			
Item	++	--	+-	-+ **	chi-square	p*
Headaches	5	17	12	3	4.27	<.05
Depression	0	30	7	0	5.14	<.05

* McNemar's test for comparison of frequencies in matched samples

** ++ = condition present in both experimental and control subject
-- = condition absent in both experimental and control subject
+- = condition present in experimental subject, absent in control
-+ = condition absent in experimental subject, present in control

TABLE II Delivery, neonatal problems and feelings in matched pairs of experimental and control mothers at first interview (N = 37)

Item	Response: Tied pairs ++	Tied pairs --	Differing pairs +-	Differing pairs -+ **	chi-square	p*
Medical History						
Complications at or after delivery	0	28	7	2	1.77	>.10
Infant care						
Desire to hold baby	15	1	2	19	12.19	<.001
Desire to breast-feed baby	10	6	5	16	4.76	<.05
Leaving town	0	31	6	0	4.17	<.05
Complaints about baby	0	31	6	0	4.17	<.05
More education for baby	1	26	9	1	4.90	<.05
Maternal response						
Mother unhappy about birth	5	12	17	3	8.45	<.01
No friends	0	30	7	0	5.14	<.05
Next baby different	3	4	25	0	23.04	<.001
Paternal response (according to mother)						
Father unhappy about birth	3	10	21	3	12.04	<.001
Complaints about father	3	27	7	0	5.14	<.05

* obtained using McNemar's test for comparison of frequencies in matched samples

** for explanation of these symbols see Appendix K Table I footnote

TABLE III Observations of behaviour in experimental and control mothers with their infants (aged ± three months) at second interview (N = 32)[x]

Behaviour [z]	Tied pairs ++	Tied pairs --	Differing pairs +-	Differing pairs -+ **	chi-square *	p*
Eye-to-eye contact	31	0	1	0	0	
en face position	32	0	0	0	0	
Smiles at baby	10	5	6	11	0.94	
Talks to baby	7	12	4	9	1.23	
Sings to baby	0	32	0	0	0	
Kisses baby	1	26	2	3		1.00
Holds baby close	24	3	1	3		0.36
Strokes baby	16	3	7	5	0.08	
Places baby securely on bed	23	2	5	2		0.45
Names baby	2	23	5	2		0.45
Keeps baby clean	27	1	2	2	0	
TOTAL SCORE	7	0	10	15	0.64	
Asks questions about baby	0	29	4	0		0.36

[x] Pairs in which one member responded `Don´t know´ were excluded

* McNemar´s test for comparison of frequencies in matched samples (Binomial tests were used where expected frequencies were = or <5)

[z] Symbol + was awarded if behaviour was displayed and - if behavour was not displayed

** For explanation of these symbols see Appendix K Table I footnote

TABLE IV Observations of behaviour in experimental and control mothers with their infants (aged ± six months) at third interview (N = 22)[x]

Behaviour[z]	Tied pairs ++	Tied pairs --	Differing pairs +-	Differing pairs -+ **	chi-square *	p*
Eye-to-eye contact	22	0	0	0	0	
en face position	22	0	0	0	0	
Smiles at baby	9	4	2	7		0.18
Talks to baby	3	10	4	5		1.00
Sings to baby	0	22	0	0	0	
Kisses baby	1	10	8	3	1.45	>.20
Holds baby close	17	0	2	3		1.00
Strokes baby	13	1	5	3		0.72
Places baby securely on bed	21	0	1	0	0	
Names baby	2	15	3	2	0	1.00
Keeps baby clean	17	0	5	0	0	0.62
TOTAL SCORE	7	0	11	4	2.40	>.10
Asks questions about baby	0	19	2	1	0.00	

[x] Pairs in which one member responded `Don't know' were excluded

* McNemar's test for comparison of frequencies in matched samples (Binomial tests were used where expected frequencies were = or <5)

[z] Symbol + was awarded if behaviour was displayed and - if behaviour was not displayed

** For explanation of these symbols see Appendix K Table I footnote

TABLE V Observations of behaviour in experimental and control mothers with their infants (aged ± nine months) at fourth interview (N = 18)[x]

Behaviour[z]	Tied pairs ++	--	Differing pairs +-	-+ **	chi-square *	p*
Eye-to-eye contact	18	0	0	0	0	
en face position	18	0	0	0	0	
Smiles at baby	6	0	6	6	0	
Talks to baby	9	4	2	3		1.00
Sings to baby	0	18	0	0	0	
Kisses baby	1	14	1	2	0	
Holds baby close	13	1	1	3		0.36
Strokes baby	7	1	4	6		0.74
Places baby securely on bed	17	0	1	0	0	
Names baby	4	12	1	1	0	
Keeps baby clean	12	1	4	1		0.36
TOTAL SCORE	3	0	7	8	0.90	
Asks questions about baby	0	18	0	0	0	

[x] Pairs in which one member responded `Don´t know´ were excluded

* McNemar´s test for comparison of frequencies matched in samples (Binomial tests were used where expected frequencies were = or <5)

[z] Symbol + was awarded if behaviour was displayed and - if behaviour was not displayed

** For explanation of these symbols see Appendix K Table I footnote

TABLE VI Comparison of matched pairs of experimental and control responses at interviews 2 to 6 on dichotomous items (significant differences only)[x]

Item	Interview No	Tied pairs ++	Tied pairs --	Differing pairs +-	Differing pairs -+	chi-square	p*
Infant Care							
Extra solids	4	0	10	8	0	6.12	<.02
Leaving town	2	1	27	8	1	4.0	<.05
Complaints about baby	2	1	25	7	0	5.14	<.05
	3	0	13	9	0	7.11	<.01
	4	0	10	7	0	5.14	<.05
	5	1	6	8	0	6.12	<.02
	6	0	7	7	0	5.14	<.05
Maternal Response							
Mother unhappy	2	0	16	13	0	11.08	<.001
	3	0	16	6	0	4.17	<.05
Next baby different	2	2	4	12	2	5.78	<.02
	3	1	7	12	1	7.69	<.01
	4	2	5	11	0	9.09	<.01
	5	1	3	8	0	6.12	<.02
	6	2	2	11	0	9.09	<.01
Grandmother comforts	3	2	11	8	1	4.00	<.05
Paternal Response							
Father unhappy	2	2	18	11	1	6.75	<.05
	3	0	16	6	0	4.17	<.05
Babies' behaviour							
Baby cries too much	2	0	24	8	1	4.00	<.05
sits late	5	0	2	11	2	4.92	<.05
crawls late	5	0	1	12	2	5.78	<.02

[x] Pairs where one member responded `Don't know' were excluded.
* McNemar's test for comparison of frequencies in matched samples
** For explanation of these symbols see Appendix K Table I footnote

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