# An Assessment of the Genetic Knowledge of Final Year Diploma Nursing Students

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A Research Report submitted to the Faculty of Health Sciences, University of the Witwatersrand, Johannesburg in partial fulfilment of the requirements for the degree of

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# Declaration

I, Merlyn Glass declare that this research report is my own work. It is being submitted for the Degree of Master of Science (Nursing) in the University of the Witwatersrand, Johannesburg. It has not been submitted for any degree or examination at this or any other university.

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(Merlyn Glass)

On this_____day of _____ 2004

# Dedication

To my mother, Vanessa Glass and my late father, Reuben Barney Glass both of whom always encouraged, supported and guided me through all my endeavours

# Abstract

The purpose of this research was to ascertain and examine the current genetic knowledge of the nurses in their final year of the Diploma in Comprehensive Nursing (General Nursing, Community Nursing, Psychiatric Nursing) and Midwifery.

The research design was an exploratory, descriptive, quantitative survey, using a sample of final year student nurses at two of the three nursing colleges in Gauteng. Data were collected through the use of a self-administered paper-based questionnaire, which was adapted from a questionnaire used for a study to assess the genetic knowledge of general practitioners (Trenton, 2003). The questionnaire was adapted using the broad outline for subject content given in the curriculum for the four-year Diploma in Comprehensive Nursing (General Nursing, Community Health and Psychiatric Nursing) and Midwifery, the South African Nursing Council regulations, and the Department of Health Policy Guidelines. Random sampling was utilised to select two of the three nursing colleges in Gauteng and the total population of final year students was used as the sample. Data were analysed by means of descriptive statistics.

It was concluded through this study that, although nurses showed fairly adequate knowledge in the areas of teratogens and multifactorial inheritance, they showed a lack of knowledge with regard to basic genetic inheritance patterns, genetic conditions, epidemiology and prenatal diagnostic techniques. It can thus be recommended that an expanded component of genetics be included in the basic curriculum of the four year Diploma in Comprehensive Nursing (General, Community, Psychiatric) and Midwifery programme in accordance with programmes initiated in other countries.

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**"Sometimes our light goes out, but is blown into flame by another human being. Each of us owes deepest thanks to those who have rekindled this light." Albert Schweitzer**

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# Chapter One

## 1. Overview of the study

## 1.1 Introduction

In this chapter the background, rationale and motivation for the study will be discussed. The background to the study covers the history of genetic disease, from when first recognition of genetic disorders took place. It also covers the researcher's personal experience when dealing with genetic disorders in the maternity wards in the public hospitals. The prevalence of various conditions is mentioned, and the importance of integrating genetics into the nursing curricula is addressed.

Emphasis is placed on the impact of the Human Genome Project and the exponential increase in knowledge of genetic conditions, on the prevalence and incidence of genetic conditions in South Africa, and the relevant changes that must be initiated and included into the education and practice for nurses in all fields in order to equip them with relevant and updated knowledge. The aims and objectives will be addressed and a brief description of the methodology will be given. A brief discussion regarding the validity and reliability of the study is included into this chapter and a description of the ethical considerations is given. The limitations and the potential usefulness of the study will be discussed.

With the inception of the Human Genome Project (HGP) in 1989 and the identification of genes responsible for so many common diseases, the identification, diagnosis, treatment and care of "at risk" and affected people has transformed dramatically on all levels. The HGP was founded on the premise that the understanding of the entire sequence of human DNA and the ways in which it differs between individuals will provide information that will be beneficial to health. The genome contains the genetic information present in each cell of the body, and the sequences that code for proteins, essential for life, are the most important. These proteins correspond to specific genes. Sequences of genes vary from individual to individual, leading to different characteristics, physical and physiological, and may influence susceptibility to specific disease (Burton & Stewart, 2003). The impact that the Human Genome Project is

making on the management of patients emphasises the need for nurses in all spheres of nursing to understand the implications of new genetic knowledge. (Anderson, Monsen, Prows, Tinley & Jenkins, 2000). It is therefore essential that nurses in all fields of care, education and management be kept informed of the rapid developments in this area enabling them to provide comprehensive care and service to their communities.

## 1.2 Motivation for the study

The motivation for this study was guided by the researcher's interest in the field of genetics. The researcher has been a genetic nurse counsellor since 1987 and has motivated for genetics to be integrated into primary health care, both in a clinical, counselling and educational capacity. Working at the various antenatal clinics in the Gauteng Provincial Hospitals, the researcher noted that the nurses appeared to be unaware of the importance of obtaining a detailed family history for all patients, a thorough obstetrical history or the significance for early referral of women of advanced maternal age. It was also recognised that the medical and nursing staff are unaware of the importance of genetic investigations of infants with multiple congenital abnormalities and similarly detailed investigation into the cause of stillbirths. The foetus needs to be fully examined taking note of any dysmorphic or abnormal features, X-rays need to be done, especially where there is a skeletal abnormality, bloods from the foetus must be taken for genetic investigations, as well as photographs to try and establish a diagnosis so that a couple can be fully informed in order to make reproductive decisions for future pregnancies. If nurses, during their midwifery training were informed about the importance of diagnosis and risks to such a family, there would possibly be more concern as to the management of such a case. The researcher conducted informal inservice training for the labour ward staff at one of the regional hospitals regarding these issues. Subsequently the midwives have been referring cases to the genetic counselling team.

The researcher was concerned as to whether any studies investigating the genetic knowledge of nurses had been initiated in South Africa. To date, no other research, other than the study by Lemkus, Van der Merwe, and Op't Hof (1978) has been conducted. The study conducted by Lemkus et al (1978) examined knowledge and attitudes to genetic and congenital disorders on

White women (n=2500), White nurses (n=826, inclusive of students, sample not clearly demonstrated in the study) and doctors (n=61). These were three separate surveys, and the conclusive findings in all three studies showed that there was an overall need for further genetic knowledge, a recommendation to execute a genetic education programme as well as a plan to increase the genetic content in the nursing curriculum. However, it was not clear as to whether any of these recommendations were implemented. In the 21st century in South Africa, the demographics of such a study would be not be representative of the general population, nor of the nursing fraternity and therefore, the researcher felt that it would be of value to examine and assess genetic knowledge of final year student nurses to establish a current baseline knowledge.

Extensive input, in the form of inclusion of a genetic component into nursing curricula and summer schools for the nursing education faculty, appears to have been made in the United Kingdom and the United States of America (Jenkins, Prows, Dimond, Monsen & Williams, 2001). South Africa is far behind the United States of America and the United Kingdom in this area of development of genetic knowledge for nurses.

With the rapid developments in the field of genetics, including diagnostic techniques and treatment available, nurses and midwives should be aware of the implications for individuals and families with a history of birth defects and genetic disorders or for those potentially at risk for having a baby with a congenital defect. Nurses need to keep up to date with available resources and genetic testing, to be able to provide a comprehensive service in the form of education and counselling, to the families, the patients and those individuals with potential for genetic disease.

## 1.3 Prevalence and significance of genetic disorders

Genetic disorders have been recognised since biblical times. The earliest recognition of patterns of inheritance was recorded in the Talmud, where it was noted that if two boys in a family died from bleeding following circumcision, the parents were exempted from having further sons circumcised (Sutton, 1980). Three percent of all births have a congenital disorder

(Department of Health, Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities, 2001), and the commonest chromosomal disorder, Down syndrome, has a prevalence of 1 in 650 births (Harper, 1998).

In South Africa, we face a major problem with Foetal Alcohol syndrome (FAS). In a study conducted by May, Brooke, Gossage, Croxford, Adnams, Jones, Robinson and Viljoen in 2000, it was established that the rate of FAS in the Western Cape is approximately 8% which is higher than any other country in the world. Viljoen (2003) showed that the prevalence of foetal alcohol syndrome in three provinces had risen: in the Western Cape from 46, 4/1000 in 1997 to 110, 2 /1000 in 2002; in Gauteng, a prevalence rate of 37/1000 was determined for FAS in 2000 and in the Northern Cape the rate was established as 52/1000. (Viljoen, Proceedings of the 22$^{nd}$ Conference on Priorities in Perinatal Care in Southern Africa, 2003). Education about the risks associated with alcohol use and abuse in pregnancy needs to be emphasised. There is also a high rate of skin cancer in people with albinism (DoH [a] 2001); therefore, nurses need to be educated about the high prevalence of this condition and the appropriate management of affected individuals and their families.

The curriculum as it stands at present, includes a component of genetics in the midwifery curriculum of the four year diploma in Comprehensive Nursing (General, Community, Psychiatric) and Midwifery (South African Nursing Council, Reg. 425 of 1992), through the introduction of the more common birth disorders, including Down syndrome, Turner syndrome, trisomy 13 and trisomy 18. The textbooks include very little on the importance of prenatal counselling and the availability of prenatal testing with the option to terminate an affected foetus. In the Textbook for Midwives (Myles, 1999) currently used in the nursing colleges in Gauteng, there is a section dedicated to prenatal diagnosis but there is no reference to the Human Genome project or newer prenatal and diagnostic techniques available.

When discussing genetic problems in the labour wards and antenatal clinics at the provincial hospitals in Gauteng with the researcher, the nurses participating in the discussions expressed that they feel that they do not receive sufficient training in counselling of families with genetic

disorders and therefore neither feel confident nor have the clinical expertise to manage these cases.

According to the Primary Health Care Package for South Africa in the section on norms and standards, the intent is to transform the health services in South Africa. "An integrated package of essential primary health care services available to the entire population will provide the solid foundations of a single, unified health system" (Department of Health, Primary Health Care Package, 2000, p 3 of 82). Primary Health Care (PHC) clinics render comprehensive integrated PHC using a one-stop method 8 hours a day, five days a week. These clinics are primarily manned by nursing staff and doctors. Other specialised professionals visit periodically and are accessible for consultation, support and referral. Management and prevention of genetic disorders and birth defects forms part of the integrated maternal, child and women's health care. The norms and standards set in the PHC package state that within each clinic, there should be at least one staff member who has been trained to recognise, counsel, treat, manage and refer most common genetic conditions (DoH[b] 2000 p 16). It stipulates that clinic staff should receive regular genetic training and updating from the regional genetic co-ordinator as well as receiving support from visiting specialists, clinical geneticists and other academic experts. The package emphasises patient education by the provision of posters, pamphlets and other educational material on genetic conditions, and that all patients receive health education with regard to genetic disorders, birth defects and disabilities. Encouragement of women to have their children at the ideal reproductive age, thus reducing the risk of chromosomal abnormalities, is recommended as well as the education of mothers concerning teratogenic effects of alcohol, recreational drugs and infections on the unborn foetus. The nursing staff, specifically midwives, are responsible for the safe and comprehensive care of all women. The competence of the staff is dependent on their ongoing training in the perinatal education programmes (PEP), and subsequently their ability to provide effective contraception methods to the clients (also utilising this as a primary prevention method for genetic disorders) and provision of post-abortion care management. The staff is responsible for the health of the mothers during pregnancy including education and counselling to each pregnant woman and partner on the monitoring of problems that may

arise, nutrition, sexually transmitted diseases (STD's), human immuno-deficiency virus (HIV), delivery, newborn and child care and advanced maternal age (DoH [b], 2000 p 15).

## 1.4 Problem statement

With a lack of available literature in South Africa regarding the knowledge of nurses in the field of genetics, it is questionable whether nurses are able to provide a comprehensive service to their clients, patients and their families with regard to genetic disease. From personal experience, a large number of patients who attend the genetic counselling clinics who have had previous stillbirths or children with abnormalities do not know why the child died, or what condition the child had. It presents a difficult situation for the genetic counsellor who is asked what the risks would be for having further affected children. If a diagnosis in an abnormal baby or child were established, and the importance and relevance of this recognised by professionals working in labour wards, postnatal and neonatal units as well as in the community environments, comprehensive management for those at risk individuals, especially for planning further pregnancies, would be enhanced.

## 1.5 Rationale for the study

In South Africa, nurses provide the bulk of Primary Health Care and Maternity Services, either in their capacity as primary practitioner primary health care nurse or midwife, or in collaboration with a medical practitioner.

With the advent of the Human Genome Project and the potential to detect and treat common conditions that have a basic genetic component, for example, breast cancer, colon cancer, diabetes, cardiac disease and hypercholesterolaemia, it would be of great advantage to the communities of South Africa if health professionals, inclusive of nurses, doctors and members of allied medical professions (physiotherapists, speech therapists and occupational therapists) had a more comprehensive knowledge of genetic disease, diagnostic techniques and treatment. The families and communities would be able to benefit from the possibility of early diagnosis,

prevention or treatment of disease and would be in a position to make informed choices regarding their genetic health. They would also be able to make decisions regarding their reproduction with reference to their risks of passing on inherited disorders. Nurses are in a position where they have the opportunity and the responsibility of giving this important information to their patients and the families at risk, as they are frequently the first or the only contact that the patient may have.

## 1.6 The purpose of the research

The purpose of this research is to ascertain the current genetic knowledge of the nurses in their final year of the comprehensive programme leading to registration as a general, psychiatric and community nurse and midwife. By assessing the genetic knowledge and recognising if there are deficits, recommendations can be made to the South African Nursing Council for inclusion of a more intensive genetic component into the nursing curriculum as it currently stands.

## 1.7 Aims and Objectives

**Aim:**

To examine the genetic knowledge of students in their final year of the Diploma in Comprehensive Nursing (General Nursing, Community Health and Psychiatric Nursing) and Midwifery.

**Objectives:**

1. To examine the final year student nurses' current genetic knowledge through the administration of a questionnaire
2. To elicit the student nurses' opinions regarding the adequacy of the current genetic component in their curriculum and whether, in their opinion, there is a need for inclusion of a more extensive genetic component in their syllabus.
3. To make recommendations according to the identified needs of nurses as to any changes that may be required in the content of the current curriculum

## 1.8 Definitions

The following definitions were delineated for the specific purpose of this study.

**Student nurse**

Refers to a nurse currently in his/her final year of study, in the Diploma course leading to the registration as a general, psychiatric and community nurse and midwife (SANC Regulation No R 425, 1992).

**Genetics**

Refers to the study of heredity, including a knowledge of genes, chromosomes, DNA, and the transmission of inherited disorders through the modes of inheritance (Mueller & Young, 2001, Harper, 1998)

**Genome**

The full set of genes in an individual, either haploid (the set derived from one parent) or diploid (the double set, derived from both parents). In a human being the haploid set contains about 3 billion base pairs of DNA and 30,000-100, 00 genes. (Dorland's, 2003.)

**Genomics**

The study of the structure and function of the genome, including information about sequence, mapping and expression and how genes and their products work in organisms (Dorland's 2003).

**Genetic education**

Refers to the education and delivery of information about genetics and genomics and the responsibility that is implied by having that knowledge in terms of advice, counselling, knowledge and referral resources.

**Genetic knowledge**

Refers to basic knowledge in the science of genetics, including meiosis, mitosis, chromosomal abnormalities, single gene disorders, multifactorial disorders and the effects of teratogens. Included in this definition would be knowledge of prenatal testing, counselling and epidemiology and referral resources.

## 1.9 Methodology

### 1.9.1 Research design

The research design was an exploratory, descriptive, quantitative survey, using a sample comprised of the total population of final year student nurses at two of the three randomly selected nursing colleges in Gauteng. A self-administered paper-based questionnaire, adapted from a questionnaire used for a study to assess the genetic knowledge of general practitioners (Trenton, 2003) was distributed. The questionnaire was adapted using the broad outline for subject content given in the curriculum for the four-year Diploma in Comprehensive Nursing (General Nursing, Community Health and Psychiatric Nursing) and Midwifery, the South African Nursing Council regulations, and the Department of Health Policy Guidelines, and included questions covering basic genetics, epidemiology, prenatal diagnosis, genetic counselling and new advances in the genetics field.

### 1.9.2 Population, Sample and selection

**The population** studied were all students in their final year of the four-year Diploma in Comprehensive Nursing in the three Nursing colleges in Gauteng. Two of the three colleges were randomly selected and the third college was used as the site for the pilot study.

**Total population** consisted of final year diploma students at the three nursing colleges in Gauteng (N= 242)

**Sample**: Inclusion criteria for the sample were that the nurses had to be in their final year of study of the four-year Diploma of Comprehensive Nursing, and willing to participate

voluntarily. Nurses were invited to participate, those willing to be included in the study (n=131) comprised the sample.

### 1.9.3 Instrument

A self-administered, paper-based questionnaire including questions on basic genetics, epidemiology, prenatal testing and counselling was developed. This questionnaire was adapted from a questionnaire used for a study to assess the genetic knowledge of general practitioners (Trenton, 2003). The questionnaire was adapted by assessing the validity and importance of which aspects of genetics were considered appropriate for nurses' knowledge, after a literature review, consultation with nurse educators, clinical geneticists and genetic counsellors was conducted. It was modified for nurses, using the SANC regulations and curricula for general nursing and midwifery, and the National Policy Guidelines of the Department of Health for Management and Prevention of Genetic Disorders and Birth Defects as guidance and reference. A statistician was consulted to assess if the coding and scoring of the study was feasible.

The instrument was used in a pilot study at the third nursing college in Gauteng to determine clarity of the questionnaire and to exclude any ambiguity of the questions posed. Fourteen out of fifteen volunteers participated in the pilot study, and the questionnaire was found to be neither ambiguous nor to have confusing questions, thus no adaptation was required

## 1.10 Data collection

An information sheet was given to each student, which they read prior to voluntary participation in the study. The researcher was present to supervise the administration of the questionnaire at a set time arranged through the principals at the two nursing colleges in Gauteng. The researcher collected the completed questionnaires, which were individually placed in sealed envelopes by the participants to ensure anonymity.

## 1.11 Data Analysis

A statistician was consulted and data were entered into the Excell programme and analysed using the Statistical Package for the Social Sciences (SPSS) Version 11 for Windows. Descriptive statistics were applied and the results are depicted in a variety of visual presentation methods including tables, pie graphs and histograms.

### 1.11.1 Validity and Reliability

**Reliability** is concerned with the consistency, stability and repeatability of the informant's accounts as well as the investigator's ability to collect and record information accurately (Brink, 1999). In quantitative research, if information is gathered with a standardised questionnaire, the study will be considered to have a comprehensive level of reliability. The standardisation of the questionnaire and the researcher's analysis of the data ensured reliability.

**Internal validity** may be threatened by various factors. One of these factors is testing by the use of a questionnaire. The researcher ensured that the questionnaire used was not too lengthy, as this may have caused participant fatigue. A pilot study conducted prior to the research validated this not to be the case. Selection bias may influence the internal validity and thus the pilot study sample utilised was taken from the total population of final year diploma nursing students from one of the three nursing colleges in Gauteng. They were similar in that they met the inclusion criteria of the sample. All nurses studying at the three colleges meet the criteria in that the initial selection of students applying for the course is performed centrally and students are then allocated randomly to any of the three colleges.

**External validity** was managed through the use of a standardised questionnaire based on a questionnaire used to evaluate the genetic knowledge of General Practitioners in Gauteng (Trenton, 2003). The questionnaire for this study was adapted to suit the needs of this research in accordance with the SANC curriculum for the four year Diploma in Nursing (General

nursing, Community nursing, Psychiatric nursing) and Midwifery, and the South African Nursing Council regulations (SANC Reg. 425 of 1992) and the Department of Health Policy Guidelines, 2000. The questionnaire can be repeated and used to re-evaluate the students' knowledge after an intervention programme has been implemented thus ensuring generalisability of the instrument. It may also be repeated to assess the knowledge of students at different colleges in the country and may be used to assess final year students in subsequent years. Thus, the external validity was not threatened. Threats to the external validity may be reactive effects, for example, test anxiety (Brink, 1999). The questionnaires were administered without the prior knowledge of the students to avoid the possibility that the nurses would look up the information, which could bias the study. Confounding variables may have included that some of the students may have been absent or may have felt unwell on the day that the questionnaire was administered. Participation was voluntary therefore the students who participated did not fall into either of these categories.

**Content validity:** Content validity refers to the appropriate and adequate information dealt with in the questionnaire, which covers "the scope and range of the information that is sought" (Brink, 1999 pp 207). In this study, the content of the questionnaire included the specific genetic information that is in the current curriculum. Two experts, a clinician and a genetic counsellor, from whom permission had been granted for adaptation of the questionnaire from a previous study (Trenton, 2003) evaluated the questionnaire, as well as nurse educators at the University of the Witwatersrand. The questionnaire was amended to comply with SANC regulations by ensuring the inclusion of content mentioned in the current curriculum and recommendations from the Department of Health Policy Guidelines (2000).

**Instrument validity:** The instrument used is the questionnaire. The instrument was divided into seven sections. The first section covered aspects of basic genetics, chromosomes and chromosome anomalies, the second section dealt with single gene disorders, the third section included multifactorial disorders and teratogens, the fourth section dealt with epidemiology, the fifth section included questions about prenatal diagnosis, the sixth section was on genetic counselling and the students' perceptions about their competence in the field, and the seventh section dealt with knowledge about new changes in the genetics field. Although these subjects

are not categorised in the curriculum set out by the South African Nursing Council *per se*, individual tutors at the colleges teaching midwifery include the subject matter that they feel is relevant in the different aspects of the course; this was established through perusal of midwifery lecture content and discussion with individual tutors. Thus, the validity of the instrument was ensured.

## 1.12 Ethical Considerations

Application to the Postgraduate Committee of the Faculty of Health Sciences of the University of the Witwatersrand was undertaken prior to commencement of the study. Application to The Committee for Research on Human Subjects (Medical) of the University of the Witwatersrand was undertaken prior to commencing with the study for ethical clearance (Appendix 3). An application was made to The Gauteng Provincial Administration's Head of the Department of Nursing Schools for permission to undertake the study (Appendix 1). Permission from the Principal of each Nursing School was obtained, and formalisation of the study by means of arranging a suitable date and time for each college.

An information sheet was given to each participant to provide the individuals with adequate comprehensible information, inviting voluntary participation in a research project (Appendix 5). An informed written consent is not required for this study as completion and submission of the questionnaire implied consent. However, in accordance with the ethical principal of human rights, no person was coerced to participate without his or her full and voluntary co-operation. Their studies at the college were not jeopardised in any way whatsoever by non-participation in the study, and they could withdraw at any stage without any impact on their studies.

Confidentiality and anonymity of each participant as well as the nursing schools was ensured. No names or identifying information appeared on the questionnaires and each answer sheet was placed by the participant in a sealed envelope on completion, which was opened by the researcher when analysing the data. Emphasis was placed on the fact that participation in the research would have no detrimental influence on their examination marks or tests.

Scientific honesty is of major importance. The researcher has acknowledged any work that is done by anyone other than herself, and there was no manipulation of design or methods, or selective retaining or manipulation of data.

## 1.13 Limitations of the study

The limitations of this study were that only nurses in Gauteng were used as a sample. The results only indicate the knowledge level of final year nurses in the nursing colleges in Gauteng offering the four year diploma in comprehensive nursing, and are not representative of the educators, students following the four year Degree course at Universities, post graduate or post basic students. The results of the study cannot be generalised to nurses in the final year of study at other Colleges of Nursing Education in South Africa.

## 1.14 Potential usefulness of the study

After identification of results regarding the knowledge of student nurses of the genetic component of the curriculum, recommendations will be forwarded to the South African Nursing Council for inclusion of a comprehensive genetic component into the four- year diploma curriculum. After completion of the four-year diploma course, nurses in all fields of care should have a broad understanding of the implications that genetic disease has on an individual, the family and communities. Nurses would feel more confident with regard to their genetic knowledge and be able to identify "at risk" members of the community and thus be in a position to discuss genetic conditions and refer “at risk” individuals for genetic counselling and where appropriate, specialist medical interventions, for example, invasive prenatal testing including chorionic villus sampling and amniocentesis. In all fields of nursing, knowledge of the availability of genetic testing, gene replacement therapy, prenatal diagnosis, and pre-symptomatic diagnosis is essential to manage patients comprehensively. Genetic disease has an immense impact on individuals, families and the communities, physically, emotionally and financially. Management of genetic disease should improve the individuals' and families' ability to cope with genetic disease and make informed choices with regard to testing, prenatal

testing and pre-symptomatic testing which could impact profoundly on their lifestyle, financial and emotional resources.

## 1.15 Conclusions

In this chapter an overview was given regarding the importance of genetic knowledge for nursing with reference to the rapid changes that have occurred. The motivation for the study was guided by the researcher's interest in the field of genetics and the potential value of integration of a comprehensive genetic component into the nursing curriculum. The objectives of this study were to examine the final year nurses' current genetic knowledge, and to elicit whether in the nurses' opinions there was an adequate genetic component in their curriculum. The survey was done through the use of a self-administered questionnaire which covered a general overview of genetics, inclusive of chromosome abnormalities, single gene disorders, multifactorial and teratogenic disorders, prenatal diagnosis, epidemiology and new changes in genetics.

The data were analysed using SPSS and depicted graphically through the use of pie graphs and histograms. The limitations of the study were that only the genetic knowledge of final year diploma students in Gauteng was determined, and this does not necessarily reflect the knowledge of nurse educators, students following the four year course at universities, post-basic or postgraduate students, or necessarily the genetic knowledge of -year diploma student nurses in other regions in South Africa.

Throughout the chapter, it is emphasised that nurses need to keep up to date with available knowledge, testing, management and resources, so they can provide a comprehensive service in the form of education and counselling to the families, the patients and potential individuals with genetic disease and thus be in a position to make informed decisions about their potential for genetic disease, be aware of their choices regarding reproduction and enhance the management of their conditions.

With the advancing changes in genetic information, diagnostic techniques and treatment available, doctors, the allied medical professionals and especially nurses, as they are in the forefront of management of patients, should be aware of the implications for families with genetic conditions or who are at risk for having babies with genetic disorders.
With reference to the literature (Jenkins et al, 2001), extensive input, in the form of inclusion of a genetic component into nursing curricula and summer schools for the nursing education faculty, appears to have been made in the United Kingdom and the United States of America. In South Africa, we have not yet reached this level and this chapter emphasises the relevance and importance of the changes that need to occur.

In the next chapter a literature review will cover genomics and the Human Genome Project, Epidemiology of Genetic Disease, Diagnostic techniques, curriculum issues, and nursing responsibilities with regard to genetic conditions.

# Chapter Two

## 2. Literature review

## 2.1 Introduction

In this chapter, the importance of genomics, and the integration of this science into nursing education and practice will be explored and discussed with reference to the literature reviewed.

The epidemiological aspect, specifically the prevalence and incidence of genetic conditions in South Africa will be addressed. The importance and availability of laboratory testing, the genetic content of nursing curricula and the nurse's responsibility with reference to the changes in management, because of the implementation of the human genome project and it's implications on health care, will be discussed.

## 2.2 Genomics and the Human Genome Project

Genetics is the study of inheritance. The Human Genome project (HGP), initiated in 1989, was developed with a view to sequence the entire human genome (Monsen & Anderson, 1999). The aim of this project being to identify genes that cause disease and to be able to diagnose, treat and possibly prevent certain conditions. Monsen & Anderson(1999) state that new advances need to be included into clinical practice, research and education.

With the Human Genome Project nearing final completion, it is essential for all nurses in all spheres of nursing to be *au fait* with all updated knowledge with regard to genetic discoveries. Skirton and Patch (2000) state that genetics, being a previously specialised subject, has become essential knowledge for all nurses. Genomics can no longer be seen as a luxury which is not essential in nursing education (Lea & Monsen, 2003). A family tree drawn up accurately can reveal numerous health issues and determine possible risks for inherited disorders. The American Medical Association (AMA) stated that one of the most important facets of obtaining a thorough genetic history is the family tree. Careful evaluation of a family history

will be the first clue as to whether a family is at a risk of inheriting a specific genetic disorder (Lea & Monsen, 2003).

## 2.3 Epidemiology

Worldwide, it is reported that approximately 40-50% of all first trimester pregnancy losses are a result of a chromosomal abnormality and two to three percent of neonates have at least one major congenital abnormality, of which at least 50% are due completely or partially to genetic factors. Other statistics worldwide indicate that fifty percent of all childhood blindness, 50% of all childhood deafness, and 50% of all cases of severe mental retardation is due to a genetic disorder (DoH[a] 2001). Genetics also plays a major role in the common and complex disorders such as various cancers, infectious diseases, Alzheimer disease, heart disease and chronic obstructive pulmonary disease (Lashley, 2000).

Internationally, five to ten percent of common cancers such as breast, colon and ovary have a strong genetic component and by the age of 25 years, approximately 5% of the population will have a disorder in which genetic factors play a role (Mueller & Young, 2001). Over 50% of the older adult population will have a genetically determined medical problem, taking into account the contribution genetics has in cancers and cardiovascular disease (Mueller & Young, 2001). Updating genetic knowledge will also shed a new light on management of patients potentially at risk for genetic disease. From a management perspective, it will have a significant impact on the emotional, ethical and psychological implications of the conditions and the repercussions thereof, including the impact on other family members who may also be at risk. Whether it is in oncology or paediatrics, renal care units or the cardiac intensive care unit, it is of utmost importance that the nursing and medical staff are knowledgeable about genetics, so that they in turn can inform their patients and provide health care based on current information.

In South Africa, there are various conditions that are specific to particular communities and nurses need to be familiar with them in order to provide a comprehensive service. Albinism, for example, was recognised by Kromberg and Jenkins (1982) to have a carrier status of 1 in

32 in the Southern African Black population, but it was suggested in a later study to be approximately 1 in 40 (Stevens, Ramsay & Jenkins, 1997). The incidence of skin cancer in affected individuals is extremely high. Affected individuals present with visual impairment and need early intervention to facilitate the prevention of developmental or motor delay. If a couple both carry the gene for albinism, the risk for having an affected child is 25% with each pregnancy. The gene has been identified and prenatal testing is available for couples who may request this. Some couples who choose to have prenatal testing may opt for termination of an affected foetus. In South Africa, other priority genetic conditions include Down syndrome, neural tube defects, foetal alcohol syndrome, cleft lip and palate, talipes equinovarus, congenital infections and genetic deafness, blindness, physical handicap and mental retardation (DoH[a], 2001).

In the United States of America, it has been recommended that all couples planning pregnancies should be tested for cystic fibrosis as the carrier rate in the Caucasian community is 1 in 20 (Lea & Monsen, 2003). If both of the couple were found to be carriers, they would then be in a position to make informed decisions regarding their reproductive choices. In South Africa too, the carrier rate for cystic fibrosis is 1 in 20 in Caucasians, and the mutation is known in about 80% of carriers. In our black community, from a recent study, it has been shown to be 1 in 34 with a calculated incidence of 1 in 4624 births (Goldman, Graf & Ramsay, 2003), but only about 46% of affected people show the known common mutation, therefore limiting carrier detection in this community (Padoa, Goldman, Jenkins & Ramsay,1999).

In a study done by Viljoen, it was established that the prevalence of foetal alcohol syndrome (FAS) in the Western Cape has risen from 46, 4/1000 in 1997 to an estimated 100, 2/1000 in 2002. In Gauteng, the prevalence of FAS has been shown to be 37/1000 in 2000 (Viljoen, 2003).

When the infant mortality rate (IMR) falls below 40-50 per 1000, it is recognised that genetic disorders and birth defects are contributing significantly to this number. At this stage, infectious diseases and malnutrition are notably controlled (with the exception of tuberculosis and HIV/AIDS). In South Africa in 1999, the IMR was 54/1000 indicating that health care

plans should be initiated through strategic planning and the care, prevention and control of genetic disorders must be implemented with appropriate services (Wessels, 2002).

## 2.4 Diagnostic techniques

Confirmation of diagnosis can now be offered for many of the inherited disorders and prenatal testing for many of the conditions is a feasible and available option. Previously, DNA testing could not be offered for common conditions like haemophilia A and prenatal testing and termination of pregnancy for this condition was based on sexing of the foetus. DNA analysis can now be offered prenatally and families can make decisions about termination of a pregnancy, based on the confirmation of an affected foetus.

Testing by the National Health Laboratory Services in Gauteng can be offered for chromosomal abnormalities as well as cystic fibrosis, Duchenne muscular dystrophy, sickle cell anaemia, thalassaemia and albinism to name a few. Genetic Counselling Clinics are held weekly in Gauteng at three academic hospitals in Johannesburg (Chris Hani Baragwanath, Coronation, and Johannesburg Hospitals). Clinics for private patients on medical insurance schemes, are run by the University of the Witwatersrand's Division of Human Genetics and are held at the Donald Gordon Medical Centre, a privately run facility. Outreach clinics are offered every three months in Sebokeng, which is a large peri-urban community in Gauteng. Nurses at the various hospitals and community and primary health care clinics in Gauteng should be made aware of all these facilities. Genetic clinics are also held in Pretoria, Cape Town, Bloemfontein, and Durban.

To detect colon cancer, a gene test done on stool samples will be of utmost importance in the primary health care setting. A recent study of individuals at high risk of developing colorectal cancer, identified early colorectal tumours in 60% of these individuals thus indicating that this is an innovative and non-invasive approach for early diagnosis and management (Lea & Monsen, 2003).

An important discovery was the gene for breast cancer, which was identified in 1999. Family members who are identified as being at risk can be offered pre-symptomatic testing and if the gene is found in an individual, early intervention can be implemented.

## 2.5 Curriculum Related Issues

An overview of the literature revealed that, although the recommendation that a genetic component be included in the nursing curricula as early as 1962 by Brantl and Esslinger (quoted by Jenkins, Prows, Dimond, Monsen and Williams, 2001), the genetic knowledge of faculty members and curriculum content for nurses in the United States of America is indeed still deficient. Moreover, since Monsen had measured the genetics content of nursing curricula in the USA in 1980, there had been no significant improvement (Jenkins et al, 2001). Since then, however, three nursing bodies, including the American Nurses Association, The American Association of Critical Care nurses and the Oncology Nursing Society, initiated the International Society of Nurses in Genetics (ISONG). This society is made up of a group of professional experts who can offer direction in influencing the genetic education for the practising nurse. Their focus is on management and care for the genetic health of individuals. Their mission is to manage genetic information through the promotion of their members from a scientific, professional and personal perspective (Jenkins et al, 2001).

The International Society of Nurses in Genetics (ISONG) together with the American Nurses Association (ANA) have set out standards that should be met in the integration of genetics into clinical and education practices. These standards are available at their websites, www.ISONG.org and www.ANA.org. Another initiative is practice-based genetics self-learning modules created by Dale Halsey Lea aiming to support and educate faculty to increase genetics into curricula. (Jenkins et al, 2001). These modules are available on the March of Dimes website, www.marchofdimes.com. Other online ongoing education modules are available at www.webct.isu.edu.

Lea and Monsen (2003), suggest that to provide genomics-based health care, nurses will need to incorporate three key aspects: firstly, knowledge of current gene function and the

mechanisms involved in causing disease, diagnosis and prevention. Secondly, interpretation of the concepts of genetic and inherited risks and thirdly, the co-ordination of services of multidisciplinary teams and other service providers. The role of a Genomics Nurse Case Co-ordinator will be essential as the demand on specialised clinical geneticists and genetic counsellors increases, with the simultaneous increase of the knowledge of the members of the communities and their demand to have improved health care.

In a study done in Taiwan, it was emphasised that genetic technology, such as DNA analysis, should be included in the content of nursing education programs (Huang, Liu, Tsai, Lin, 1997). If it is established that the nurses' knowledge is insufficient, it may be necessary to establish whether the nurse tutors are being kept fully informed of accessible treatment and management for genetic conditions. In a study done by Prows and Latta (1995), it was shown that, after a two-day workshop, a three-month preceptorship, and regularly scheduled continuing education meetings, scores from pre and post-workshop tests indicated a considerable improvement in nurses' knowledge of genetic concepts and resources (Prows & Latta, 1995).

Various studies have shown that there is a deficit in the human genetics component in nursing curricula (Jenkins et al, 2001, Lea et al, 2002, Prows et al, 2003). Lashley (2000) examining genetics in nursing education, noted that the genetic content in nursing and other health profession curricula was deficient. She emphasised that it was unrealistic to expect graduating nurses to be confident to do general genetic counselling without specific training and education. The author also states that important knowledge for the basic nursing course should include the basic mechanisms of inheritance, information about the common inherited disorders, interaction of genetics and environment, care of adults and children with genetic disorders, interpretation of genetic risks, and information about genetic testing and screening and the interpretation of the results of genetic tests. She also emphasised the importance of their recognition of social legal and ethical issues, and an awareness of familial predisposition to genetic disease. Lashley (2000) also states that both didactic and clinical teaching of a genetic component should be included into nursing curricula and one of the initial phases would be to meet with teachers to establish what is already included.

Lashley (2000) identified that barriers that could be encountered when including genetics at a deeper level in the curricula are often at faculty level. The faculty barriers included a perceived lack of knowledge and insecurity in their knowledge. The teaching faculty did not perceive genetic content as important and failed to recognise the vast potential of genetics as influential to health care. Genetic conditions were viewed as rare, and not as conditions that would be observed in the actual nursing environment (Lashley, 2000). In the United States of America, major organisations have recognised and have implemented the updating of the knowledge of the nurse educators through different programmes. The aim of these programmes is to stimulate and educate faculty, so the content of genetics in entry level nursing is increased (Jenkins, Prows, Dimond, Monsen & Williams, 2001).

A Genetics Program for Nursing Faculty (GPNF) funded by the Ethical, Legal and Social Implications Research Program of the National Institutes of Health (ELSI) in the United States of America was initiated as a response to these recognised needs (Prows et al, 2003). An annual Genetics Summer Institute (GSI) with continuing support for attendees through a variety of media was developed. The aims of the GPNF are to increase nursing faculty's genetic knowledge and clinical application, and to increase genetic content into nursing education curricula. The programme includes the provision of updates on genetics topics, nursing practice and curriculum issues, and the drafting of recommendations for increasing genetic content into the nursing curricula (Prows, Hetteberg, Johnson, Latta, Lovell, Saal & Steinberg Warren, 2003).

The aims were supported through the conduction of an annual Genetics Summer Institute and the provision of ongoing support for previous participants. Many of the past participants stated that their incentive to continue efforts to increase genetics content in the nursing curricula was encouraged through the opportunity to ask questions and receive positive reinforcement (Prows et al, 2003). Continuing online education modules are posted at the website www.cincinattichildrens.org.

Prows et al (2003) indicated that the GPNF succeeded in accomplishing its aims. Participants' knowledge was determined using pre-test/post-test methods. It was shown that there was a significant improvement in post-test scores with reference to participants' knowledge in 1997 and 1998 and again in 1999 and 2000. Prior to each GSI, a curriculum survey was sent to each participant to determine change in nursing curricula genetics content. It was established that there was a statistically significant improvement in the total number of genetic topics and conditions included and taught by the 39 of the 53 participants representing different nursing schools after attendance at a GSI and 86% of the respondents reported that adding genetic content to their own lectures was the most practical method (Prows et al, 2003). Barriers to curriculum change identified in this study included that faculty felt that there was not enough time in the curriculum to add genetics content (69%) and that they did not have time for additional commitments (45%).

Recommendations for inclusion of basic genetic principles into the basic curricula were listed on the GPNF website. Participants were required, through workshop attendance, to identify clinical outcomes with specific regard to genetics. Lea, Feetham & Monsen (2002) conducted a systematic analysis of the genetics education literature and conducted surveys of nurses in genetics and nursing leaders involved in national initiatives, genetics research training programmes and genetics education models in the United States of America. They researched the genetics nursing education literature in the second half of the 20$^{th}$ century and conducted a focused survey where they used multiple networks of professionals in health care, education and research. The survey asked for content in genetics nursing education programme. The authors identified their sample from leaders of master's programmes with a speciality in genetics, those offering continuing education in genetics and those in nursing education programmes with some level of genetics in their requirements for degree completion. They concluded from their survey that successful initiatives in genetics education for nurses have come from both a top down model of national organisations and from grass roots efforts. They recognised however, that none of the health professions are prepared sufficiently for genetic based health care, and that all professionals working in the health environment need to reach efficiency in genetics. All participants in the survey were shown to be ready to accept the challenges and provide leadership for genetic based health care (Lea, 2002).

Following the Genetics Summer Institute (GSI), a Web-based Genetics Institute, which covered the same content as the GSI was established (WBGI) and initially offered in 2002. Summaries from the participants' ideas were used to establish a checklist for the integration of genetics into the nursing curricula (Hetteberg & Prows, 2004). The checklist is divided into four sections: the first section aims at establishing the existing genetics content, the second, to increase the faculty awareness about the need to include genetics, the third to increase faculty knowledge regarding genetics and lastly, the integration of genetic content into the nursing curricula (Hetteberg & Prows, 2004).

According to ISONG, a nurse graduate should be competent in obtaining a genetic history inclusive of ethnic, racial and cultural background, in the examination of an individual with recognition of characteristics suggestive of a genetic condition, and should be able to construct a family tree. This should enable the nurse to determine at risk individuals. Inclusion of a more comprehensive genetic component into basic curricula will be delayed until nursing faculty have a sound knowledge (Jenkins, Prows, Dimond et al., 2001) and commitment to the importance of human genetics being an integral part of nursing care.

The curriculum in nursing and other health care professions as it presently stands is deficient in genetic content (Lashley, 2000). According to the South African Nursing Council, minimum requirements for the education concerning the teaching of students in the programme leading to registration as a nurse (general, psychiatric and community) and midwife, which was last amended in 1994, a midwifery student "also needs subject content in respect of the health of the mother……the development and health of the foetus……..early identification of mother and child (foetus, neonate and infant) at risk" (SANC Regulation R.425 p 5). No direct references to the incorporation of issues such as advanced maternal age or high-risk pregnancies with regard to genetic disorders are included. It is stated that "the student shall apply the components fundamental to a scientific approach in carrying out the comprehensive ante-natal assessment….and recording all relevant information" (SANC Regulation R.425 p 19).

The nursing curricula in South Africa need to be examined with specific reference to the inclusion of a genetic component in all subjects. To provide safe, knowledgeable, effective and relevant care to their clients and families, nurses must keep up their knowledge. Genetics must be included into the curriculum to produce a competent and reliable professional who is equipped to practice currently and in the future of health care (Lashley, 2000). Recommendations in the Department of Health Policy Guidelines (2001) state that standardisation of content and assessment of the medical genetics component of the nursing curriculum should be assessed on both basic, post-basic undergraduate and post-graduate level. Revision of genetic education in faculty must be offered and aim to increase confidence levels for staff teaching the various programmes. This can be addressed through continuing education and workshops. Forsman (1988) states that, prior to teaching genetic content, faculty must feel secure.

"Nursing and genetics must be in the mainstream of health care together, and nursing must be ready to integrate genetic content or be left behind" (Lashley, 2000 p 804). According to literature from the United Kingdom and the United States of America, in-service training programmes, in the form of updating and training for tutors in genetics have been highly successful in integrating a "genetic thread" throughout the teaching of the nursing curriculum, and genetics is not taught solely as an exclusive subject (Prows, Personal communication, 2003).

## 2.6 Nursing Knowledge, Education and Nursing responsibility

In the United States of America, the National Coalition of Health Professional Education in Genetics (NCHPEG) is a coalition of professional organisations, the aim of which is to promote professional education in genetics.

The nursing process emphasises the importance of managing patients in a holistic manner. “We want to develop a professional who can think genetically when approaching a clinical situation or problem” (Lashley, 2000, p 797).

Jenkins (2003 p 13), states that "it's patient care that should motivate all nurses to become genetically literate". She implies in her article that nurses who are uninformed about genomics will severely limit its use in improving health and health care. With the genes known, there is the potential to provide possible gene therapy, predictive and diagnostic testing, and early detection of inherited disorders and disease. Nurses must be in the forefront of discussing and explaining genetic testing and implications to patients. Jenkins & Collins (2003) mention that these tests guarantee the provision of rational and powerful foundations for the art of healing.

Grady and Collins (2003), state that the future of healthcare will benefit from genetics, as genetic knowledge facilitates both the identification of specific disease mechanisms and diagnostic test availability. They also emphasise that nurses must also be aware of the ethical, social and legal aspects of genetic information and how this impacts on individuals and families. They must be empowered to be able to assist individuals and families make informed decisions regarding their genetic risks. Nurses should be able to use genome-based knowledge effectively for the provision of healthcare for society (Grady & Collins, 2003).

Scientific information emerging from human genome research has considerable implications for the practice of professional nursing. Both valuable and harmful effects can arise as a result of gene identification. Nurses need to be fully aware and be prepared for their changing roles that may occur due to genetic testing (Williams, 1998).

In every sphere of nursing, be it critical care, oncology, paediatrics or midwifery, as well as geriatric, medical and surgical, it is becoming more and more essential that the nurse has a basic knowledge of the genetics of the conditions. The nurse must be aware of patients who may be at risk, and be knowledgeable as to any available testing. To offer a comprehensive health service the nurse must be updated with changes that are rapidly occurring in the genetic field and must be given in-service education, including training in genetic counselling. Almost all disease, including cardiac disease, diabetes and cancers, has some basis of genetic inheritance. Those patients treated in secondary and tertiary health care environments deserve comprehensive care from the health care personnel. Feetham (2000), states that, by the year 2010, more than 25 common conditions such as diabetes and hypertension may have

predictive genetic testing. Unless nurses are well informed about the genes that have been identified, are fully aware of testing that is available, and are confident that they can manage their patients with genetic conditions from medical, psychosocial and ethical aspects, will they really be offering a comprehensive service? Nurses should become active in dealing with the concerns resulting from this new knowledge specifically for dealing with aspects of education, practice and policy (Jenkins, 2000).

An emphasis has been placed on a bi-directional approach in integrating and initiating change in genetic knowledge within nursing spheres. Lea (2002) states that primary health care providers should be able to recognise the role of genetic factors in health and to use genetic information for explanation to at risk members of the community.

Although it is expected that individuals will take cognisance of the impact genetics has on the more common conditions, nurses must form active partnerships with their clients for the management of their disease. They must therefore become genetically literate, and genetic education opportunities that integrate the genetic knowledge into clinical practice, emphasising a multidisciplinary and holistic approach have been initiated (Lea, Feetham & Monsen, 2002). A bi-directional approach (from top-down to grass roots and vice versa) has been developed by nurses in the United States of America for their colleagues in nursing and other health disciplines in the form of national conferences, research training, education models, and nursing literature.

Although for the last two decades, recommendations have been made for genetics to be included into nursing curricula in the United Kingdom and the United States of America, according to Anderson's (1996) summaries of nine surveys done during the 1980's and early 1990's, the documentation recorded a lack of genetic content in the nursing curricula. Insufficient educational material supportive of an integration of genetics into the body of knowledge was noted.

According to the Department of Health Primary Health Care Package for South Africa – a set of norms and standards (2000) in the section titled “Management and Prevention of Genetic

Disorders and Birth Defects" (Appendix 7, p 17 of 85), emphasis is placed on Genetic services forming part of the integrated maternal, child and women's health care. The components required in the primary health care setting as stated in the document are clinical diagnostic services, counselling, and laboratory support and prevention strategies. Public awareness campaigns should be initiated in collaboration with non-governmental organisations (NGO's) and other governmental sectors. The recommended norms are that at least one clinic staff member is trained to recognise, treat, manage and refer most common conditions; that clinic staff receive regular genetic training and updating from the regional co-ordinator for genetic services; and that clinic staff receive support from visiting specialists, clinical geneticists and other academic experts. It states in the standards that the clinics should have the latest copy of the Human Genetics Guidelines for Management and Prevention of Genetic Disorders, Birth Defects and Disabilities (DOH, Primary Health Care Package, 2000, p 17 of 85) and that patient education should be provided through the distribution of posters and other educational material, that patients and caretakers receive health education on genetic disorders, birth defects and disabilities; that women should be encouraged to "procreate at the ideal reproductive age to reduce the risk for chromosome abnormalities" and that women should be educated about the effect of teratogens, including alcohol, drugs and infections on the unborn foetus ( DoH[b], 2000).

Responding to requests from the various clinics in the regions and provinces, the researcher is directly involved in the teaching and organising several two part, four-day genetic workshops annually in conjunction with the Department of Health, the South African Inherited Disorders Association and the Division of Human Genetics of the University of the Witwatersrand. The targeted audience includes midwives, community health nurses and primary health care nurses. They are trained in basic genetics, single gene disorders, inheritance patterns, multifactorial conditions and teratogens, causes of mental retardation, visual and hearing impairment as well as prenatal options and basic counselling skills.

However, personal experience has shown that although the nurses receive a certificate after completing part two of the course, few of them have expressed confidence in managing genetic cases alone. It is stressed at the courses that the primary goal is that they recognise and

refer genetic cases, and acknowledge that education of our communities with regard to the common conditions, including albinism, neural tube defects, Down syndrome and Foetal Alcohol syndrome is of utmost importance.

Christianson reported that medical and nursing staff have difficulty in the recognition and diagnosis of black neonates with Down syndrome. He noted that there were similar craniofacial features in the black infant with Down syndrome and in unaffected black babies, particularly epicanthic folds and oblique palpebral fissures, compared with Caucasian Down syndrome and unaffected infants. A protruding tongue and excess neck skin were significantly less frequent in black compared to white Down syndrome infants (Christianson, 1997). Christianson noted that congenital heart disease was notably higher in infants under 12 months of age than in children 13 months of age or older, suggesting that congenital heart disease is a major cause of mortality in black Down syndrome individuals in sub-Saharan Africa (Christianson, 1996).

To improve and enhance genetic knowledge in South Africa, a proposal of a distance education programme specifically directed at the ongoing in-service training of midwives in South Africa, was initiated in 2001 following the guidelines of previous programmes, including advanced midwifery and management in the primary health care facilities. These programmes were referred to as the Perinatal Education Programme (PEP) developed by Dr David L. Woods. These can be viewed at www.pepcourse.co.za. The programme is comprises modules in maternal care, newborn care and perinatal/AIDS, but these are not endorsed by the South African Nursing Council, as there is no formal teaching through recognised and formal nursing colleges. They are, however, are accepted and acknowledged by the Health Professionals Council of South Africa and registered for earning Continuing Professional Development (CPD) points (Woods, 2004).

In collaboration with the PEP authors, a Genetic Education Manual (GEM) has been formulated with the primary goal being to educate and empower nurses working in primary health care facilities about the care and prevention of common birth defects seen in the South African population. The developer of GEM is a professor in the Division of Human Genetics,

National Health Laboratory Services, Gauteng, in conjunction with contributing authors and reviewers. The first phase of the GEM, a Birth Defects Manual (BDM) for distance education, has six chapters. The first is an introduction that covers the definitions, epidemiology and the causes of birth defects. The second chapter teaches the basics of care and prevention of birth defects and genetic counselling. The other four chapters address Down syndrome, neural tube defects, foetal alcohol syndrome and single gene disorders including oculocutaneous albinism, Waardenburg syndrome and haemophilia. While undertaking the BDM over a six month period, nurses will have four contact days during which they will have workshops on the basics of birth defect recognition and diagnosis, including history taking, drawing and interpretation of family trees, external clinical examination and basic investigations for common defects. After successfully completing an exit examination they will be eligible to undertake an intensive two-week training programme on medical genetics at an academic unit. The pilot study was initiated in November 2004 and the initial groups of nurses were randomly selected from two rural areas in the Free State. The researcher is involved as a reviewer and contributor to the programme and will assist in the facilitation and running of the programmes once they have been initiated.

Continuous updated knowledge should be included in the curricula, taking into account the numerous genes that are being identified daily by researchers involved in the Human Genome Project. Nurse educators should ideally be offered in-service training and workshops to offer updating of diagnostic testing and services available. They could subsequently include these subjects in their teaching, thus empowering the students to provide a comprehensive service to the communities.

## 2.7 Conclusion

In this chapter, the literature was reviewed specifically with reference to the importance of Genomics and the Human Genome project and the impact that it has on the potential and effective management and care of individuals and families with genetic conditions.
The worldwide epidemiology of genetic conditions was addressed and conditions specific to South Africa were discussed.

The relevance and importance of up-to-date diagnostic testing was emphasised as well as curriculum related issues. If nurses at all levels are kept fully informed and their genetic knowledge is continuously updated, they will be able to be more proficient in their management of their patients and the community. They will be more aware of the rapid and continuous changes and be knowledgeable about techniques that may be available in areas of genetic engineering, gene therapy, and prenatal or pre-implantation diagnoses. This could impact profoundly in all the spheres of health care.

Issues surrounding the nursing responsibility towards the treatment and management of individuals and families were considered, and the researcher expressed the opinion that there was a need to assess South African nurses' genetic knowledge with specific reference to the changes that have occurred in the field of human genetics and genomics. This research examined whether there is a need to make recommended changes in the curriculum as it presently stands.

A detailed description of the methodology used to achieve the objectives of this study will be discussed in Chapter Three.

# Chapter Three

## 3. Methodology

## 3.1 Introduction

In the previous chapter the literature review covered the aspects of the relevance of the human genome project and genomics to nurses, epidemiology of genetic conditions, diagnostic techniques available for genetic disease and the genetic content in the various nursing curricula in the United Kingdom, United States of America, and South Africa. The importance of nurses' responsibility towards affected individuals and those at risk for inherited conditions was discussed as well as the importance of including a wider genetic component into the education and training of nurses in all fields of care.

In this chapter the materials and methods will be discussed in the context of this study. The design for this study was an exploratory, descriptive, quantitative survey.

## 3.2 Research Aims and Objectives

**Aim** To elicit the genetic knowledge of students in their final year of the Diploma in Comprehensive Nursing (General Nursing, Community Health and Psychiatric Nursing) and Midwifery.

**Objectives**

1. To examine the final year student nurses' current genetic knowledge through the administration of a questionnaire.
2. To elicit the student nurses' opinions regarding the adequacy of the current genetic component in the curriculum and whether, in their opinion, there is a need for inclusion of a more extensive genetic component in their syllabus.
3. To make recommendations according to the identified needs of nurses as to any changes that may be required in the content of the current curriculum.

## 3.3 Research design

A research design is a “blueprint for conducting the study that maximises control over factors that could interfere with the validity of the findings” (Burns, & Grove, 2001 pp 223). The design as well as the sampling method, type of measurement tool used, the technique of collection of data and data analysis and interpretation guides the choice of the population and sample. Brink (1999) describes the design as emerging from the particular research question or hypothesis posed and from the purpose of the study.

It is essential that a design provide sufficient control in order that the results of the research are valid and produce accurate evidence of the actuality. The design of the study is decided upon after careful evaluation by the researcher as to how the study will actually take place and what decisions are to be made. A design has to be adapted to be specific for a particular study.

An exploratory, descriptive quantitative survey was conducted. It was exploratory in order to provide an accurate and more insightful knowledge of the dimensions of the problem, and indicate the level of the genetic knowledge of the final year diploma students in comprehensive nursing. It was descriptive in order to describe information gathered about the nurses' genetic knowledge, and thus explore and elucidate whether the genetic knowledge of the sample group was adequate or insufficient. In this study, a survey was done to gather information from a sample of the population in question by using a research tool in the form of a questionnaire developed specifically for this research study through adaptation and modification of a previous questionnaire used to determine the knowledge of general practitioners in Gauteng (Trenton, 2003).

An exploratory study is one where generalisation to large population is not the objective. Striving towards increased knowledge in the specific area of study is the primary goal. Population parameters may be apparent from the data analysis of these studies (Burns & Grove, 2001). An exploratory study will "explore the dimensions of a phenomenon, the manner in which it is manifested…. it provides more insight about the nature of the phenomenon" (Brink, 1999 p 11).

In this study, the genetic knowledge of the final year student nurses in the four-year-diploma in general, psychiatric nursing, community health nursing and midwifery has been explored, thus increasing the information as to the extent of their particular knowledge. This study can thus possibly be used to determine the genetic knowledge of a larger sample of final year student nurses countrywide, or practising nurses in general.

A descriptive study according to Burns and Grove (2001 p 795) "provides an accurate portrayal or account of the characteristics of a particular individual, event, or group in real-life situations for the purpose of discovering new meaning, describing what exists, determining the frequency with which something occurs, and categorising information". The acquisition of comprehensive and exact information about a particular observable fact, through examination, explanation and organisation, thus providing new information on that particular event or incident is the primary purpose of descriptive studies as described by Brink (1999).

In this particular research study, the genetic knowledge of student nurses in their final year of the four-year diploma in general, community, psychiatric nursing and midwifery is described, thus establishing the current and actual knowledge that they possess.

A quantitative study is one in which the focus is on a small number of concepts, and it is succinct and restricted. A quantitative study is initiated with established ideas about how the concepts are interconnected (Brink, 1999). Structured measures and instruments are utilised to gather information. Strict measures of control are used during the collection and analysis of information, which must be stringently objective. Statistical procedures are used to analyse the information numerically. The researcher is not directly involved in the gathering of the information; this can be done at a distance. Logical and deductive reasoning are incorporated into quantitative studies.

In this study, the concept of the genetic knowledge of the students in their final year of the four-year comprehensive programme in general, community, psychiatric nursing and midwifery was addressed. Through a formal instrument, a restricted and concise spectrum of

information was gathered on a specific subject; their knowledge of human genetics. The collection of the data was strictly controlled and the data were analysed descriptively, using the Statistical Package for Social Sciences (SPSS) Version 11 for Windows.

## 3.4 Research Method

The research method describes the process of data collection, the population and sampling, data analysis, reliability and validity.

### 3.4.1 Setting and scope

A setting is the site, which is unforced and natural, fairly or extremely controlled, where research is conducted. The scope is the range or opportunity available for the particular research. In this study two of the three colleges in Gauteng were used as the setting. The third college was used for the pilot study. The three colleges follow the identical curriculum outline and it is the individual responsibility of the lecturers to provide the appropriate prescribed genetic input.

### 3.4.2 The population (N)

The population is the entire group of persons or objects that meet the criteria for the research study that the researcher is exploring (Brink, 1999). In this study, the inclusion criterion was that subjects must be in their final year of the four-year diploma leading to the registration as a general, psychiatric and community health nurse and midwife. The population in this study thus comprised all student nurses in their final year of study of the four-year diploma in comprehensive nursing, (general, community health and psychiatric nursing) and midwifery in three Nursing Colleges in the province of Gauteng. The total population at the three colleges in Gauteng was 242.

### 3.4.3 A sample

A sample is a representative section of that population (n) that is chosen by the researcher for a particular study. Brink (1999) describes a sample as a specific group of elements from a distinct population. Sampling is the selection method best suited to gather information about a specific phenomenon, which will represent the population of interest. In this study the sample (n) selected were from the total population of final year student nurses in the comprehensive programme (general, community, psychiatry) and midwifery at these three nursing colleges in Gauteng.

### 3.4.4 Sampling method

All candidates for admission into the three colleges of Nursing in Gauteng are chosen through a central selection committee and thus are similar. Colleges in Gauteng were used as the researcher is based in Johannesburg. Simple random selection of the two of the three colleges in Gauteng was undertaken. The names of the colleges were written on cards and placed into a sealed box. The researcher requested that a colleague select two of the cards. The names of the colleges that appeared on the chosen cards were used for the selection of the sample for this study. The name of the college on the card not chosen was used for the pilot study.

The total population at these two randomly selected colleges was 192. A random sample from this group of 192 students was utilised, in that the elements were typical of the population sought for the study. The sample selection was taken from students who volunteered for the study. The students were given the information sheet to read and the sample was thus drawn from those students who agreed verbally and by action to participate in the study.

The advantage of random sampling is that the sample chosen is specific for the phenomenon examined. The disadvantage of this type of sampling method is that the sample may be biased in that it does not represent the entire population of four-year student nurses, and in this study, is representative only of student nurses in Gauteng.

### 3.4.5 Sample size

The total number of students currently in their final year of the diploma in comprehensive nursing (general, psychiatry and community) and midwifery at the three colleges during 2002 in Gauteng was 242. There are no other colleges teaching this course in this Province. The sample size from the two colleges randomly selected was 78 volunteering individuals out of a total of 87 final year students from the first college and 53 from the total of 105 students from the second college. The sample size for the study was thus 131 (n= 131). The sample size was thus larger than the critical size of 30 necessary for descriptive statistics and representative of the population. The sample size was large enough to represent the population regardless of whether there was non-response to some of the questions in the questionnaire.

## 3.5 Instrument

The instrument or tool used in a quantitative research study is developed with stringent criteria to meet instrument validity and reliability thus the research conducted is trustworthy.

In this study, a self-administered paper-based questionnaire, adapted for nurses from a similar study done by a student doing her master's degree in Genetic counselling to examine the genetic knowledge of General Practitioners (Trenton, 2003), and modified using the general guidelines from the South African Nursing Council curriculum, the South African Nursing Council regulations and the Department of Health Policy Guidelines, was developed and distributed. Questions in Trenton's study that were specific to the knowledge and practice of General Practitioners were eliminated. Questions that did not fall into a category that would be of relevance to nurses were eliminated. After a careful and thorough literature review further items of direct relevance to nursing practice were added to the revised questionnaire. A genetic specialist clinician, a genetic counsellor and nurse educators at the Department of Education, University of the Witwatersrand examined the adapted questionnaire to enhance content validity and concurred that the items accurately interrogated the SANC requirements and the recommended proposals of the Department of Health Policy Guidelines. The instrument was divided into seven sections. The first section covered aspects of basic genetics,

chromosomes and chromosome anomalies. The second section dealt with single gene disorders and the third section included multifactorial disorders and teratogens. The section dealt with epidemiology, the fifth section included questions about prenatal diagnosis, the sixth section was on genetic counselling and nurses' perceptions about their competence in the field, and the seventh section dealt with knowledge about new changes in the genetics field and educational needs. Questions 1-37, 40, 42 and 43, were asked in a multiple-choice format, whereas questions 38, 39, 41, and 44 were open-ended. The open-ended questions were included in order to gather specific information regarding the nurses' concepts of self-confidence, competence and needs regarding genetic information and management of patients with genetic conditions.

## 3.6 A pilot study

A pilot study was distributed to final year nursing students at the third nursing college in Gauteng. The total population of final year nursing students at this college was 50. All the names of the final year student nurses were placed in a sealed box. Fifteen names were drawn. One student did not want to volunteer and the remaining fourteen students volunteered to participate in the study. An information sheet was given to each student, which they read prior to participation in the study. The researcher was present to administer the questionnaire at a set time, arranged through the principal, at the nursing college, and was present to collect the completed questionnaires, which were individually placed in sealed envelopes by the participants to ensure anonymity. Fourteen questionnaires were distributed to the volunteers to check for acceptability, clarity, feasibility and possible errors. On analysis, there were responses by the fourteen participants to all questions, the questionnaire was not found to be ambiguous nor have confusing questions, and thus no adaptation was required.

## 3.7 Data collection

Data collection is "the precise, systematic gathering of information relevant to the research purpose of the specific objectives, questions or hypotheses of a study" (Burns & Grove, 2001 p 794). Data collection must be done with due consideration to the type of measurement used,

the manner in which and by whom the data is collected, the location or setting that will be used for the collection and adherence to an allotted time frame.

The instrument used for the data collection in this study was a self-administered, paper-based questionnaire. An information sheet was given to each student, which was read prior to participation in the study. The researcher was present to distribute the questionnaire at a set time, arranged through the Principals of both nursing colleges in Gauteng, and was present to collect the completed questionnaires, which were individually placed in sealed envelopes by the participants to ensure anonymity.

## 3.8 Data analysis

Data analysis is the implementation of amalgamation, synthesis and accordance of significance to the information gathered about the specific phenomenon studied (Burns & Grove, 2001). It is the process by which raw data are converted into explicable terms to enable conclusions to be drawn from the specified objectives.

It is the method used to determine and depict the accurate and factual results of the study intended. This is done through reduction, organisation and clarification of the outcomes. (Burns & Grove, 2001). In this study, the statistic results were organised according to the seven categories in the questionnaire. The data were analysed using the computer programme SPSS Version 11 for Windows, in conjunction with the assistance from a statistician using descriptive statistics and are depicted in pie graphs, histograms and tables.

The data were analysed through quantitative means by examination, assessment and elucidation thus meeting the objectives of the research study. Descriptive statistics were used to organise data and thus graphic representation of the results could be made.

## 3.9 Validity and reliability

"The reliability of an instrument denotes the consistency of measures obtained in the use of a particular instrument and is an indication of the extent of random error in the measurement method." (Burns & Grove, 2000 p 395) If measurements that are obtained by different researchers on the same subject or element are comparable then the reliability of the instrument is ensured. Reliable instruments increase the power of a study by detecting significant differences or associations that occur in the population under study accurately. The researcher attempted to ensure rigour through the use of a pilot study and the expert advice and opinions from nurse educators, genetic clinicians and genetic counselling specialists. The validity of an instrument is the extent to which the instrument reflects the conceptual construct in question (Burns & Grove, 2001). Validity, according to Burns and Grove, will serve to determine appropriate, meaningful usage from the specific deductions made from outcomes of a study and that the inferences made, and not necessarily the score, is what makes a study valid. The validity of the instrument used in a study must be re-examined in each study situation (Burns & Grove, 2001). A previously used questionnaire (Trenton, 2003) for General Practitioners was specifically adapted for assessing the nurses' genetic knowledge.

A literature survey was conducted, the South African Nursing Council regulations (SANC Reg 425, 1992) were used to clarify the current curriculum content, recommendations from the Department of Health Policy Guidelines (2000) were given consideration and a previously administered instrument (Trenton, 2003) was consulted for the compilation of the questionnaire for this study. Thereafter, the draft questionnaire was given to experts in the field of clinical genetics, genetic counselling and in the field of nursing education for their perusal and input as to the face and content validity of the instrument. The questionnaire used for this study after adaptation for nurses was used in a pilot study to clarify the validity of the instrument as well as to test the reliability of the study.

## 3.10 Ethical considerations

Research in any field must maintain ethical principles. This research maintains all ethical principles of client autonomy, anonymity and confidentiality. The research was conducted only after extensive communication and co-operation with the heads of faculties and departments that had to be contacted and permission obtained to proceed.

Application to the Postgraduate Committee of the Faculty of Health Sciences of the University of the Witwatersrand was undertaken prior to commencement with the study. Permission was granted. Application to the Human Research Ethical Committee (Medical) of the University of the Witwatersrand was undertaken prior to commencing with the study for ethical clearance. This was granted (Appendix 3). An application was made to the Head of the Department of Nursing Schools, Department of Health, for permission to undertake the study (Appendix 1). After permission was obtained from the University and Provincial authorities, formalisation of the study was arranged after letters of request to the principals of each nursing school were sent (Appendix 2) and suitable dates and times for data collection at each college were negotiated.

To ensure that participation was entirely voluntary and thus ethical, an information sheet was given to each potential participant to provide the individuals with adequate comprehensible information inviting voluntary participation in a research project. An informed written consent was not required for this study; however, in accordance with human rights, no person was coerced to participate without his or her full and voluntary co-operation. Consent was implied through their voluntary completion and submission of the questionnaire. It was clarified that their studies at the college would not jeopardised in any way whatsoever by non-participation in the study, and that they could withdraw at any stage without it having any impact on their studies. Emphasis was placed on the fact that participation in the research would have no detrimental influence on their examination marks or tests. For the research to be ethical, the confidentiality and anonymity of each participant and the nursing schools was ensured. No names appeared on the questionnaires and each answer sheet was placed by the participant in a sealed envelope on completion, which was opened by the researcher when analysing the data.

Scientific honesty is of major importance. The researcher has acknowledged any work done by anyone other than herself, and there was no manipulation of design or methods, or selective retaining or manipulation of data.

## 3.11 Conclusion

In this chapter the design of the study and methodology techniques were discussed. The design for this study was an exploratory, descriptive, quantitative survey.

The aim of the study was to assess the genetic knowledge of students in their final year of the Diploma in Comprehensive Nursing (General Nursing, Community Health and Psychiatric Nursing) and Midwifery.

The objectives of the study were to examine the current genetic knowledge of the final year student nurses through the administration of a questionnaire; to elicit whether, in the nurses' opinion, there is a need for inclusion of a more extensive genetic component in their syllabus and to make recommendations according to the needs of nurses as to any changes, which might be required in the content of the current curriculum

The research design was an exploratory, quantitative, descriptive survey in that it would provide more insight into the dimensions of the problem and descriptive in that it would provide new information on the research problem. A pilot study was conducted at one of the three nursing colleges in Gauteng, selected randomly. A quantitative survey gathered information from a random sample (n=131) from the total population of final year student nurses at the other two (of three) colleges in Gauteng (N=192) using a research tool in the form of a questionnaire.

An information sheet was given to each student, which was read prior to his or her voluntary participation in the study. The researcher was present to distribute and collect the

questionnaires, which were placed by the participants in a sealed envelope after completion, to ensure anonymity and confidentiality.

The data were analysed using SPSS Version 11 for Windows and are depicted in pie graphs and histograms. The rigour of the study was discussed in detail as were the ethical considerations.

In Chapter Four, the data analysis and findings of each section of the questionnaire will be discussed in detail.

# Chapter Four

# 4. Results and Discussion

## 4.1 Introduction

In this chapter the results will be presented in the order elicited from the questionnaire and address the study objectives. These will be illustrated by means of tables, histograms and pie graphs. A discussion is included to further explicate the particular category addressed in the questionnaire.

The first objective of the study was to examine the final year student nurses' current genetic knowledge through the administration of a questionnaire, which consisted of seven sections interrogating knowledge of:

1. Chromosomes and chromosome abnormalities
2. Single gene abnormalities
3. Multifactorial inheritance and teratogens
4. Epidemiology
5. Prenatal diagnosis

The second objective was to elicit the student nurses' opinions regarding the adequacy of the current genetic component in their curriculum and whether, in their opinion, there is a need for inclusion of a more extensive genetic component in their syllabus. This objective was ascertained through the next two sections of the questionnaire namely:

6. Genetic Counselling
7. New genetics and educational needs

## 4.2 Section One: Chromosomes and chromosome abnormalities

This section included questions relating to basic genetic knowledge included in the course in the anatomy and physiology and midwifery section of the curriculum. Questions addressed basic mitosis and meiosis, common chromosome abnormalities, signs and symptoms of Down syndrome and the frequency of liveborn babies with Down syndrome in South Africa.

**Table 4.1: Section one covering chromosomes and chromosome abnormalities**

| Question ( N=131) | Answered | Correct answer % (n) | Incorrect answer % (n) | Unsure % (n) | Non-responders % (n) |
|---|---|---|---|---|---|
| Q1 Number of chromosomes in a human cell | 130 | 83,2 (109) | 14,5 (14,5) | 1,5 (2) | 0,8(1) |
| Q2 Number of chromosomes in sperm/egg | 131 | 84 (110) | 10,7 (14) | 5,3 (7) | 0 (0) |
| Q3 Name of the cell division producing sperm/eggs | 127 | 23,7 (31) | 70,2 (92) | 3,1 (4) | 3,1(4) |
| Q4 In individuals with Down syndrome (DS) for which chromosome are there three copies? | 130 | 60,3 (79) | 20,6 (27) | 18,3 (24) | 0,8 (1) |
| Q5 Presence of cleft lip and palate as a clinical feature of DS | 24 | — | 18,3 (24) | — | 81,7 (107) |
| Q5 Presence of epicanthic folds in DS | 20 | 15,3 (20) | — | — | 84,7 (111) |
| Q5 Presence of polydactyly in DS | 3 | — | 2,3 (3) | — | 97,7 (128) |

**Question 1** asked how many chromosomes are there in a normal human cell? One hundred and thirty responders attempted this question; the majority, 83, 2% of the nurses were correct in their answer in that there were 46 chromosomes in a normal human cell. Only 19 nurses (14, 5%) were incorrect and 2 (1, 5%) nurses were unsure of the correct answer.

**Question 2** asked how many chromosomes there are in a normal sperm or egg cell. Of the 131 responders to this question, 84 % (110) of the students were correct in their answer that there

are 23 chromosomes in the egg or sperm and 10,7% ( 14) were incorrect, 5,3% (7) were unsure of the correct answer.

**Question 3** gave three options as to the name of the cell division which takes place for the production of sperm and egg cells. The correct answer, 'meiosis' was given correctly by only 23, 7% (31) of the students and 70, 2% (92) were incorrect in their answer **(See Figure 4.1).** Meiosis is covered during the course in basic anatomy and physiology and the study of cells (SANC, Reg. 425, 1992). It is important for midwives in a primary health care setting or nurses working in antenatal clinics to have a basic understanding of the concept of meiosis in order to explain this to mothers who are potentially at risk for having a baby with a chromosome abnormality; for example those mothers with a previous child with a chromosome abnormality or mothers of advanced maternal age (Burton & Stewart, 2003).

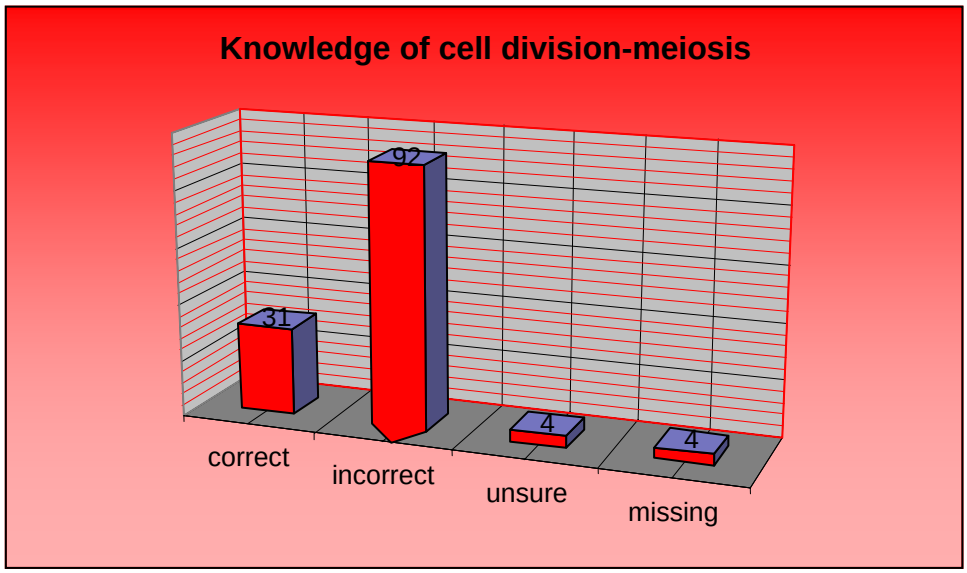


**Figure 4.1 Cell division that takes place for the production of sperm and egg cells**

**Question 4** asked as to which chromosome there would be three copies if an individual was diagnosed as having Down syndrome and gave five options including one of unsure. The correct answer, which is three copies of chromosome 21, was answered correctly by 60, 3% (79), 20, 6% (27) students were incorrect in their answer and 18, 3% (24) were unsure of the correct answer. One (0, 8%) student did not respond. The majority of individuals with Down

syndrome have the usual non-disjunction type, where there are three copies of chromosome 21. Approximately 4% of Down syndrome individuals may have the translocation type, where the additional chromosome 21 is “translocated” onto another autosome, either number 14, 15 or 21. In one third of the cases, a parent is found to be a balanced carrier of the translocation (Mueller & Young, 2001). There is also a mosaic form of Down syndrome, in which a percentage of the cells have 47 chromosomes and the rest have 46 chromosomes. It is essential that when diagnosing an individual clinically with Down syndrome, a chromosome analysis is performed to establish the type as this would alter the recurrence risks.

**Question 5** presented various options for clinical features in a child with Down syndrome; 86,3% (113) students identified correctly that individuals with Down syndrome have mental retardation (Mueller & Young, 2001), **(Figure 4.2)**, 18 (13,7%) did not respond; 15,3% (20) identified correctly that individuals with Down syndrome have associated cardiac defects **(Figure 4.3)**, 111 (84,7%) did not respond. Cardiac abnormalities occur in 40% of individuals with Down syndrome (Jones, 1997).

Only 15, 3% (20) students identified correctly that individuals with Down syndrome have epicanthic folds **(see Table 4.1)**, 111 (87, 7%) Christianson (1997) reported that this clinical feature was present in 94,5% of children in his particular study, compared to 98% of black children, 82,5% black neonates, and 70,3% white children in other studies.

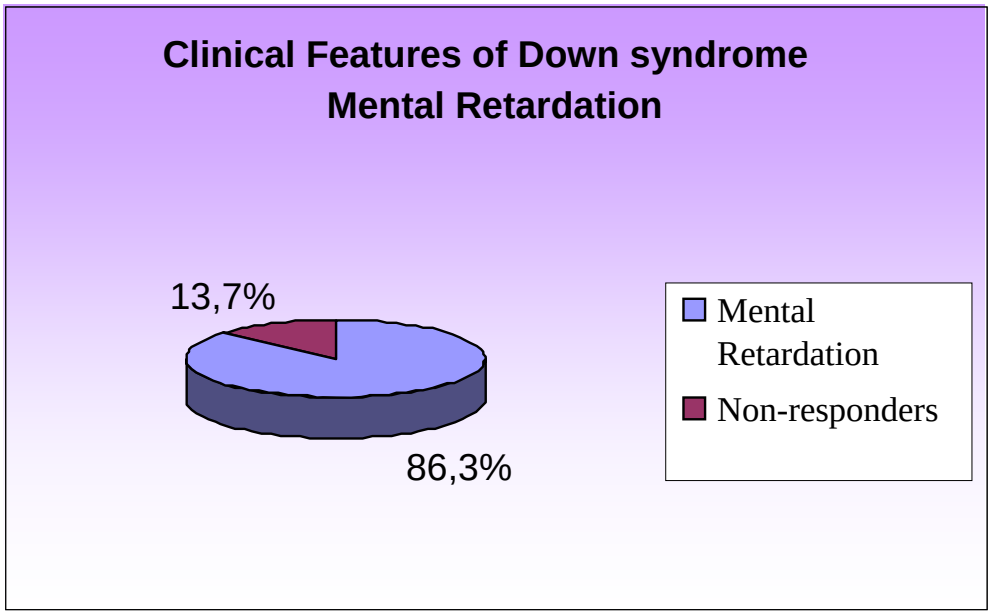


**Figure 4.2: A clinical feature of Down syndrome is mental retardation**

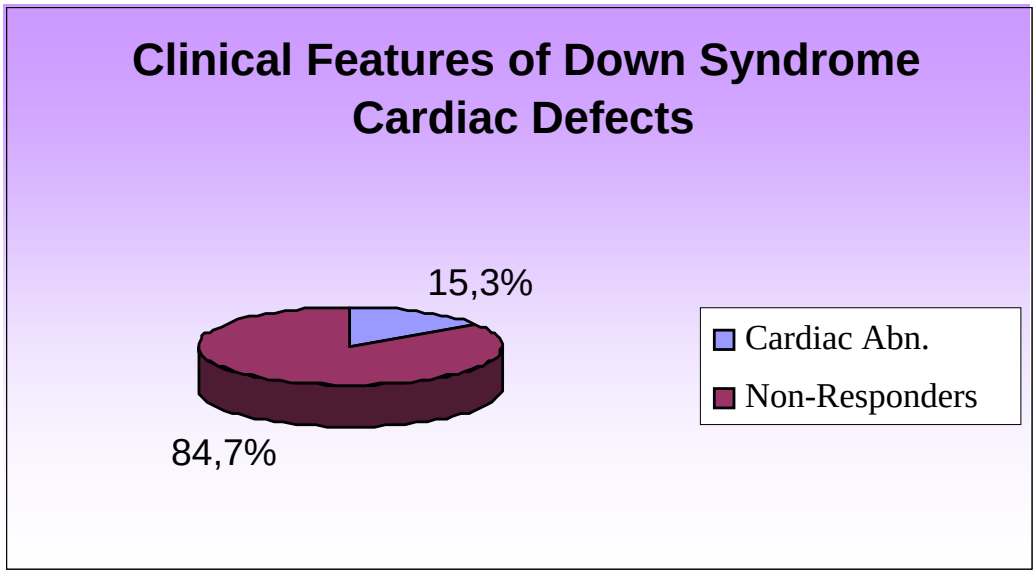


**Figure 4.3: A Clinical feature of Down syndrome is a cardiac abnormality**

Cleft lip and palate is not a feature of Down syndrome and this was incorrectly identified by 18, 3% (24) of the students; 107 (81, 7%) did not respond; 2, 3% (3) students identified incorrectly that polydactyly was a feature of Down syndrome, 128 (97, 7%) did not respond. Although the majority of the student nurses appear to be knowledgeable with regard to the

cause of Down syndrome and are aware that these individuals have mental retardation, it is of particular concern that they do not appear to be aware of the associated cardiac abnormalities and this could impact immensely on their management of affected individuals.

During the discussion of results, **a limitation in the questionnaire** was identified. Question five gave five options for clinical features in Down syndrome; it was not possible to ascertain from a non-response to a specific option, whether the student was clear that the answer was incorrect, or whether he/she was unsure.

**Question 6** asked for the frequency of liveborn Down syndrome babies in South Africa **(Figure 4.4)**. Out of the 116 students who responded to this question, 15 (11, 5%) answered correctly that the frequency is approximately 1 in 700; 53, 4% (70) answered incorrectly, 23, 7% (31) were unsure of the correct answer, and 11, 5% (15) students did not respond to the question. Down syndrome is the commonest chromosomal abnormality with a prevalence rate of approximately 1 in 650 livebirths (Harper, 1998). Of the 116 nurses who responded to the question as to the prevalence of Down syndrome, 15 (11, 5%) nurses answered correctly, 70 (53, 4%) answered incorrectly and 31 (23, 7%) were unsure of the correct answer. Fifteen students (11, 5%) did not respond to this question. With the availability of prenatal screening and diagnostic testing, women of all ages should be offered the option of knowing whether they are at risk for having a baby with Down syndrome or are carrying an affected foetus. Accessibility to this and all other health care facilities is one of the twelve patient rights stated in the Patient's Rights Charter (DoH[b], 2000).

The minority of the student nurses indicated that they were aware of the frequency of Down syndrome in South Africa. Of the 116 (88, 5%) who responded, 15 (11, 5%) were correct in their answer, 70 (53, 4%) were incorrect and 31 (23, 7%) were unsure. Fifteen nurses (11, 5%) did not respond. In a review article in the Journal of Medical Genetics in 1996, Christianson noted that several authors stated that it may be more difficult to diagnose Down syndrome in African infants than in other ethnic groups. The authors in this study had noted and confirmed that the phenotypic presentation of normal black children was similar to Down syndrome infants and children. Epicanthic folds and oblique palpebral fissures were present in more

unaffected black infants and children than in unaffected white infants and children thus indicating that the craniofacial features of African Down syndrome individuals and normal infants are similar, and suggesting that the diagnosis in African Down syndrome children may indeed be more subtle than in white babies (Christianson, 1996). If nurses were more aware of the high prevalence of this condition, they would be more alert when examining neonates and be able to recognise the various clinical features of Down syndrome more easily, and thus offer a more comprehensive service to the parents of, and those individuals affected with Down syndrome.

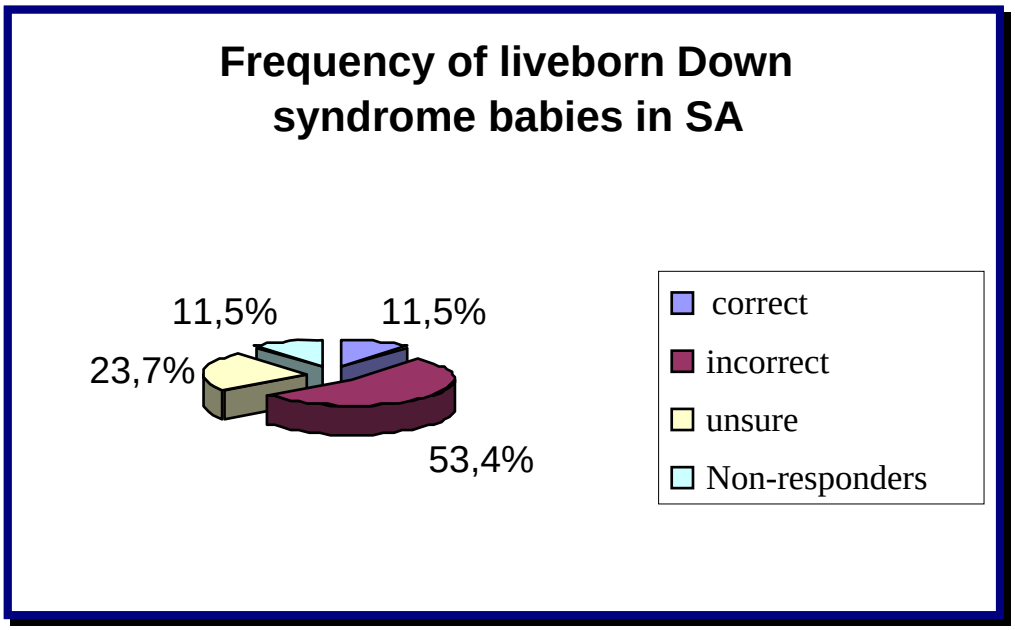


**Figure 4.4: Frequency of Liveborn babies with Down syndrome in South Africa**

## 4.3 Section Two: Single gene abnormalities

This section dealt specifically with single gene abnormalities, and included questions regarding patterns of inheritance in dominant, recessive and X-linked disorders and presentation in males and females, risks of recurrence of genetic conditions and examples of conditions that follow specific patterns of inheritance.

### 4.3.1 Patterns of inheritance: dominant and recessive

The first question in this section, Q**uestion 7**, gave 3 options for the correct number of autosomal chromosomes in a human cell; 14, 5% (19) students were correct, 34, 4% (45) were incorrect, 45% (59) were unsure of the answer and 8 (6,1%) students did not respond.

**Question 8** asked what risk a person with an autosomal dominant condition would have for passing on the gene to his child; of the 126 respondents who answered the question, sixty six of the students (50, 4%) answered correctly, 38 (29%) were incorrect and 22 (16, 8%) were unsure of the correct answer. Five (3, 8%) students did not respond. Of the 123 students who answered **Question 9** about whether males and females are equally affected by autosomal dominant conditions, 48, 9% (64) were correct, 45% (59) were incorrect, 8 (6,1%) did not respond to this question.

This lack of knowledge regarding the inheritance patterns of dominant disorders could impact on their treatment and management of individuals with dominant conditions in South African communities; lack of knowledge could result in the non-recognition of the inheritance patterns in particular families and subsequently information and education regarding reproductive choices may not be imparted to “at risk” individuals. Autosomal dominant conditions often have a variation of expression of clinical features and may be missed through the non-recognition of the condition (Harper, 1998). One of the priority conditions mentioned in the Department of Health Policy Guidelines is Waardenburg Syndrome. The clinical features included are a white forelock of hair and heterochromia (different colour eyes), but the most important feature would be the risk of deafness (DoH[a], 2001). Other dominant conditions seen at the Genetic counselling clinics in South Africa, include neurofibromatosis, Huntington’s disease, polyposis coli (cancer of the colon) and achondroplasia.

**Question 10**, asked what the risk would be for a couple, who both carry a recessive gene, to have a child with a recessive disorder. This question was answered by a total of 120 students; 5, 3% (7) were correct in their answer that there is a 25% risk for a couple to have an affected child if both of them were carriers whereas 70, 2% (92) were incorrect. Sixteen percent (21) of the students were unsure of the correct answer and 11 (8, 4%) did not respond.

**Table 4.2: Single Gene Disorders**

| Question (N=131) | Answered | Correct answer % | Incorrect answer % | Unsure % | Non-responders % (n) |
|---|---|---|---|---|---|
| Q7 The number of autosomes in a human cell? | 123 | 14,5 (19) | 34,4 (45) | 45 (59) | 6,1 (8) |
| Q8 What is the risk of passing on the gene in autosomal dominant (AD) inheritance/ | 126 | 50,4 (66) | 29 (38) | 16,8 (22) | 3,8 (5) |
| Q9 Are males and females affected equally in AD conditions? | 123 | 48,9 (64) | 45 (59) | — | 6,1 (8) |
| Q10 Risk for having a child with a autosomal recessive disorder | 120 | 5,3 (7) | 70,2 (92) | 16 (21) | 8,4 (11) |

Albinism is a common recessive condition in South Africa, the carrier status initially thought to be 1 in 32 (Kromberg & Jenkins, 1982) but recognised in a later study to be approximately 1 in 40 (Stevens et al, 1997); an individual with albinism is at risk for developing skin cancer and may present with delayed motor development due to impaired vision. Early recognition of the condition, and early treatment would enhance the management of affected individuals. Albinism has been noted as a priority condition in the National Health Policy Guidelines for the management and prevention of genetic disorders, birth defects and disabilities with the emphasis being on prevention at the primary level (DoH[a], 2001). Nurses should be aware of the risks for people with recessive conditions so they can be referred appropriately and manage the families accordingly. Prevention strategies for albinism according to the Policy Guidelines, include Genetic Counselling for couples with an affected child or family history, and the early clinical detection and management for affected individuals with appropriate referral where indicated.

### 4.3.2 Patterns of inheritance: X-linked

**Question 11**, asked whether males or females are mainly affected in X-linked inheritance. Of the 128 students who answered this question, 44 (33, 6%) of them were correct in their answer

that X-linked conditions affect mainly males. Fifty five students (42%) gave the incorrect answer, 29 (22, 1%) were unsure and 3(2, 3%) students did not respond.
The deficit of this knowledge concerning conditions that follow an X-linked pattern of inheritance could have implications for the management of conditions such as haemophilia, that with early recognition, diagnosis and management, long term complications may be avoided.

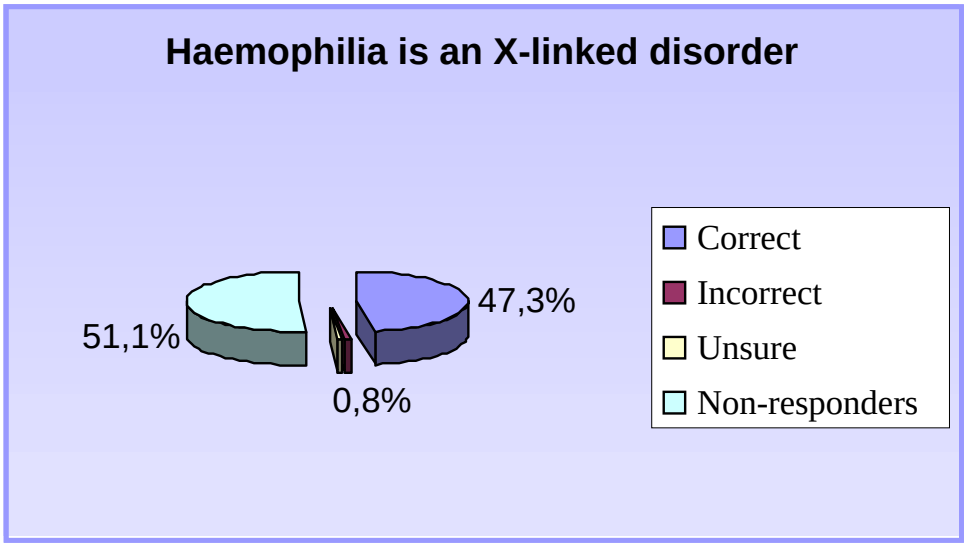


**Figure 4.5: Haemophilia is an X-linked disorder**

**Question 12** gave four genetic conditions, and requested that the students indicate which ones they thought were inherited in an X-linked manner. Sixty two (47, 3%) of the 64 students who responded to this question indicated correctly that haemophilia is an X-linked condition; 67 (51, 1%) did not respond. Ten (6, 1%) students responded to the option of Duchenne muscular dystrophy, and 8 (6, 1%) recognised correctly that this is an X-linked condition, 121(92, 4%) students did not respond. The majority of the students correctly indicated that neither cystic fibrosis nor achondroplasia was inherited as X-linked conditions (79, 4% and 90, 8% respectively) by not marking that option. Nineteen (14, 5%) of the students incorrectly indicated that none of the mentioned conditions were X-linked, 110 (84%) did not respond.

**A limitation in the questionnaire was identified in the discussion of question 12**. It was not possible to ascertain whether a non-response to the particular option was because the student did not recognise that particular option as being correct or if he/she was unsure.

These results clearly indicate that the students are not fully aware as to which genetic conditions are inherited in an X-linked manner. This could have implications on their management of the individuals and families at risk, and may influence the reproductive choices that carriers of the X-linked conditions make as they may not receive adequate information from the nurses working in antenatal and primary health care facilities; reproductive choices may be influenced if individuals at risk are fully aware of the implications and the impact that genetic disorders may have on them.

**Table 4.3: X-linked inheritance**

| Question (n=131) | Answered | Correct answer % | Incorrect answer % | Unsure % | Non-responders % |
|---|---|---|---|---|---|
| Q11 Does X-linked inheritance affect mainly males? | 128 | 33,6 ( 44) | 42 (55) | 22,1 (29) | 2.3 (3) |
| Q12 Is Achondroplasia an X-linked disorder? | 12 | _ | 8,4 (11) | __ | 90,8 (119) |
| Q12 Is Duchenne muscular dystrophy an X-linked disorder? | 10 | 6,1 ( 8) | 0,8 (1) | 0,8 (1) | 92,4 (121) |
| Q12 Is Cystic fibrosis an X-linked disorder (recessive)? | 27 | __ | 19,8 (26) | __ | 79,4 (104) |
| Q12 None of the last three mentioned conditions X-linked? | 21 | 1,5 (2) | 14,5 (19) | ___ | 84 (110) |

Lashley (2000) addressing the needed genetic knowledge for nursing graduates states that nurses should be aware of common inherited disorders, including cystic fibrosis, phenylketonuria, sickle cell anaemia and haemochromatosis. The author also mentions that disorders that are common in the geographic region of the nursing schools should be included, and therefore change the thinking about health management. In South Africa, our priority conditions would include cystic fibrosis, specifically in the Caucasian community,

thalassaemia in our Greek and Indian communities, and albinism in our black communities. Also to be included in education of the nurse should be information about more complex diseases such as breast cancer, also inherited in some families as a dominant trait, colorectal cancers, heart disease, hypercholesterolaemia and mental illness. Lashley (2000) also states that nurses should be able to interpret simple genetic risks, for example a risk of 1 in 4 for a recessive condition.

In this section, it was clear that knowledge regarding single gene abnormalities was limited and this area needs to be addressed at a more formal level.

## 4.4 Section Three: Multifactorial inheritance and teratogenesis

This section dealt with questions around definitions of multifactorial inheritance and teratogens, examples of conditions that follow multifactorial inheritance, infectious agents and drugs that pose risks to the developing foetus during pregnancy, alcohol use during pregnancy and fetal alcohol syndrome. Multifactorial inheritance refers to conditions that have arisen due to an interaction of a multitude of genes and the environment, both contributing to the disorder. Single gene disorders, for example cystic fibrosis, have no contributing factors from the influence of the environment and there are those conditions or disorders that are wholly the result of environmental sources, for example rubella. Conditions inherited in a multifactorial manner, influenced by the genetics of the individual and individual susceptible threshold, include Diabetes Mellitus Type II, hypertension, cardiac disease, neural tube defects and cleft lip and palate.

### 4.4.1 Multifactorial inheritance

The first question in this section, Q**uestion 13**, asked for a definition of multifactorial inheritance. Eighty one (61, 8%) of the students were correct in the definition of multifactorial inheritance, 6, 1% (8) of the students were incorrect, 24, 4% (32) of the students were unsure of the correct answer and 9 (6, 9%) did not respond.

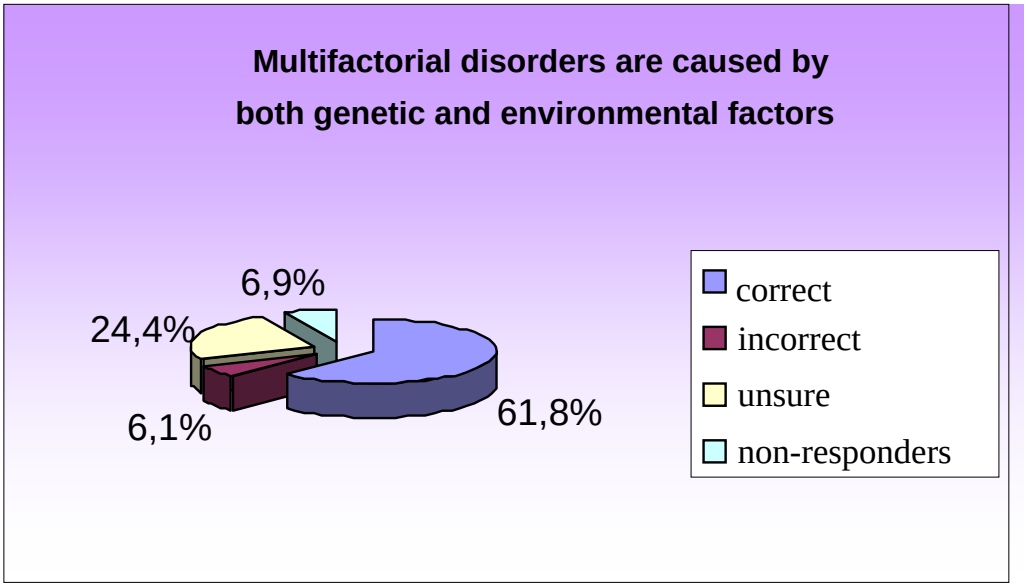


**Figure 4.6: Definition of multifactorial inheritance**

**Question 14** gave four options of possible multifactorial conditions and forty five students (34, 4%) correctly stated that neural tube defects are inherited in a multifactorial manner and 64, 9% (85) did not respond to this option. It was disconcerting for the researcher to note that although the majority of the students, 81 (61, 8%) correctly identified the definition of multifactorial inheritance, only 45 (34, 4%) of the students recognized that neural tube defects are inherited in a multifactorial manner.

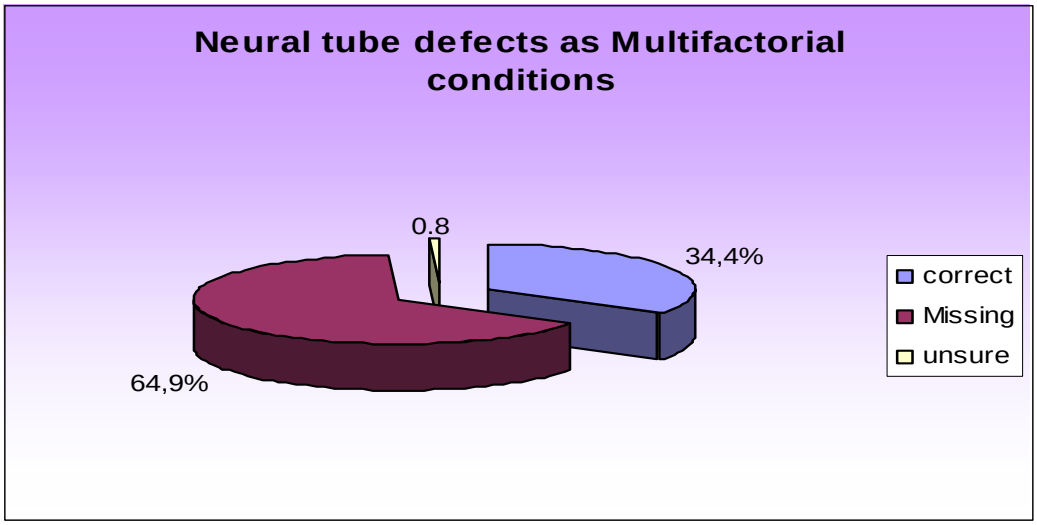


**Figure 4.7: Neural tube defects as multifactorial conditions**

Neural tube defects are common in South Africa and according to the National Health Policy Guidelines should be considered high priority in the Primary Health Care environment (DoH[a], 2001). Early recognition during pregnancy, by sonar examination or by elevated alpha-fetoprotein levels in maternal serum, would give those women carrying affected foetuses the opportunity to make an informed decision based on the prognosis determined by the severity of the lesion. Management and treatment of an affected infant, undiagnosed prenatally, should be early recognition and referral to appropriate centres, thus preventing secondary complications such as infections and irreversible hydrocephalus. An anencephalic foetus, one in which there is complete exposure of an abnormally developed brain and an absence of cranial bones, would have a minimal chance of survival. However, a foetus with a small myelomeningocoele, a defect in the neural tube, with protrusion of neural tissue and without the complication of a hydrocephalus or other congenital abnormalities prior to delivery, or those who have early intervention by insertion of a ventriculo-peritoneal shunt would have a better prognosis. Although there are associated complications with shunts, including infections and blockage, with this intervention an affected individual would have a better prognosis than one who presented with enlarged ventricles in utero early in the pregnancy (Stevenson, Hall & Goodman, 1993).

According to the Department of Health Policy Guidelines, prevention strategies for neural tube defects would include folic acid supplementation (which has been shown to reduce the recurrence risk of neural tube defects, cardiac defects and cleft lip and palate) the identification of “at-risk” individuals, mid-trimester screening for maternal serum alpha-fetoprotein, ultrasound examination and a voluntary termination of an affected pregnancy (DoH[a], 2001).

One of the options given in question 14 asking which conditions may be inherited in a multifactorial manner was Diabetes Type II. Thirty seven (28, 2%) of the students correctly indicated that this condition is inherited in a multifactorial manner and 92 (70, 2%) did not mark this option. Effective management of affected individuals may be affected through lack of awareness of the inheritance pattern of this common condition in all population groups in South Africa.

Forty two students (32, 1%), incorrectly identified albinism as inherited as a multifactorial trait; this would have implications in their management as registered nurses and midwives of the clients through the information that they impart to the affected individuals and their families, in particular with respect to reproductive decisions and risks of recurrence.

**Table 4.4 Multifactorial disorders**

| Question (N=131) | n | Correct answer % | Incorrect answer % | Unsure % | Non-responders % (n) |
|---|---|---|---|---|---|
| Q14 Diabetes type II is a multifactorial condition | 39 | 28,2 (37) | 0,8 (1) | 0,8 (1) | 70,2 (92) |
| Q14 Albinism as a multifactorial condition | 42 | — | 32,1 (42) | — | 67,9 (89) |
| Q14 Cleft lip &palate as a multifactorial condition | 27 | 19,8 (26) | 0,8 (1) | — | 79,4 (104) |
| Q14 Neural tube defects as multifactorial | 46 | 34,4 (45) | — | 0,8 (1) | 64,9 (85) |

Only twenty six (19, 8%) students recognised correctly that cleft lip and palate is inherited mainly as a multifactorial condition. Cleft lip and palate may be inherited in a variety of ways, but the majority of cases are inherited as a multifactorial condition. The risks of recurrence would depend on the number of individuals affected in a family and whether the affected individuals were male or female (Harper, 1998). The majority of the students, 105 (80, 2%) did not identify this condition to be multifactorial by not marking this option. This would have implications for their management and influence the information that they communicate to the families. Cleft lip and palate is an easily repairable condition, and is considered by the Department of Health to fall into the category of a high priority condition in the Primary

Health Care setting (DoH[a], 2001). Women on drugs such as phenotoin (and other anticonvulsants) for epilepsy are at a 2-3 times higher risk than that of the general population for having a baby with congenital abnormalities, including neural tube defects and cleft lip and palate (Briggs, Freeman & Yaffe, 2002).

Thirty nine (28, 2%) students indicated that they were unsure as to which conditions were inherited in a multifactorial manner.

**A limitation in the questionnaire was recognised in the discussion of question 14**. A non-response by a student to one of the four options of examples of multifactorial conditions, did not clarify whether a student was clear as to whether the option was correct or whether he/she was unsure of the answer.

### 4.4.2 Teratogenesis

**Question 15** in this section, asked for a definition of teratogenesis, and of the 116 students who responded to this question, 87 (66, 4%) gave the correct answer, 29 (22, 1%) gave an incorrect answer and 15 (11, 5%) did not respond.

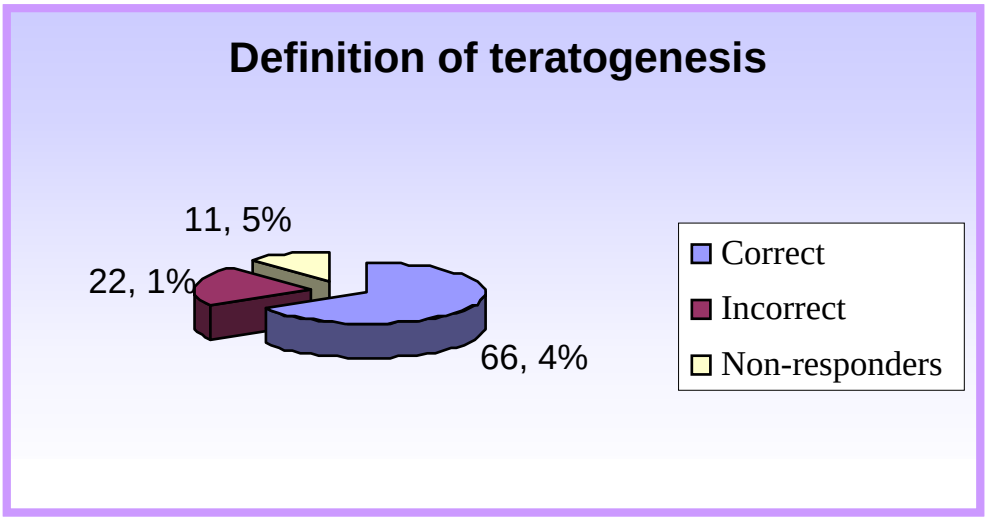


**Figure 4.8: Definition of Teratogenesis**

### 4.4.3 Infectious agents

**Question 16** gave four options of various infectious agents that pose a risk to the foetus; 116 (88, 5%) students answered correctly that syphilis poses a risk, 87 (66, 4%) correctly indicated that rubella poses a risk to the unborn foetus, 44 (33, 6%) did not indicate that they knew that rubella posed a risk. Rubella, in the first trimester of pregnancy, can cause cataracts, deafness, congenital heart defects, microcephaly and mental retardation. Contracting rubella in the first month of pregnancy poses a risk of foetal abnormality of around 60%. In the second month the risk is around 25% and then the risk during the third month falls to 8 %. In the second trimester, the risks of having a baby with a serious abnormality are negligible (Harper, 1998). Congenital deafness is one of the high priority conditions stated in the Department of Health Policy Guidelines, and all nurses working in the field of Primary Health Care and specifically in antenatal care facilities should be aware of the importance of immunizing young girls prior to reproductive age for rubella, thus reducing the risk of infections during pregnancy (Mueller & Young, 2002).

Thirty two students (24, 4%) were aware that cytomegalovirus (CMV) is a risk factor in pregnancy, 97(74%) students did not indicate that they considered this virus to be a risk factor. There is no immunisation against CMV and the risk is highest if an infection occurs during the first trimester of pregnancy. However, this virus is teratogenic in 5% of affected pregnancies. Effects in an exposed foetus include chorioretinitis, deafness and microcephaly (Mueller & Young, 2002). With regards toxoplasmosis in pregnancy, 42 (32, 1%) of the students recognised that this virus would pose a risk to the foetus. Toxoplasmosis would have a teratogenic effect in 20% of foetuses in the first trimester of pregnancy and a risk of 75% in the second and third trimesters. A foetus affected by toxoplasmosis would present with hydrocephalus, microcephaly, cataracts, chorioretinitis and deafness (Mueller & Young, 2002). If a woman knows that she has been exposed to these infectious agents during pregnancy, a foetal blood sample could be taken and IgM antibodies measured. The results indicate that the majority of student nurses are aware of syphilis as a risk factor, but only a small majority were aware that rubella and other viral infections could pose a risk to the foetus in pregnancy.

All four options of infectious diseases given in question 16 pose a risk to the foetus. **A limitation in the questionnaire was identified in this question**, as a non-response to one of the options did not clarify whether the student was unaware whether that agent posed a risk, or whether he/she was unsure.

### 4.4.4 Teratogenic drugs in pregnancy

**Question 17** asked which drugs in pregnancy were considered to be a risk for the foetus and were contraindicated for use during pregnancy. Warfarin, was recognised to be a risk by 79, 4% (104) of the students, 26 (19, 8%) did not respond to this option. Prescription drugs taken in pregnancy are known to have potentially damaging effects on the developing foetus. Foetal Warfarin Syndrome (FWS) is considered to be relatively common in South Africa due to the high incidence of rheumatic heart disease, subsequent to which, the women have surgical correction of valve abnormalities and warfarin therapy is not discontinued during pregnancy. In 50% of cases there is a poor prognosis for the foetus, including Foetal Warfarin syndrome (Gregersen, 2004). Warfarin used in the first trimester of pregnancy may result in a foetus with FWS which includes the characteristics of nasal hypoplasia, and stippled epiphyses. The baby may have a birth weight less than the $10^{th}$ percentile, eye defects, hypoplasia of the extremities, developmental retardation and seizures amongst others. The critical period appears to be between the $6^{th}$ and $9^{th}$ week of gestation. In the second and third trimesters, complications due to foetal haemorrhage have been observed, including central nervous system defects, agenesis of corpus callosum, Dandy Walker malformations, midline cerebellar atrophy and optic atrophy (Briggs et al, 2002).

**Table 4.5 Drugs contraindicated in pregnancy**

| Question N=131 | n | Correct answer (%) | Incorrect (%) | Unsure (%) | Non-responders (%) |
|---|---|---|---|---|---|
| Q 17 Warfarin is contraindicated in pregnancy | 105 | 79,4% (104) | -- | 0,8 %(1) | 19,8 (26) |
| Q17 Valproic acid is contraindicated in pregnancy | 70 | 52,7% (69) | -- | 0,8% (1) | 46,6% (61) |
| Q17 Retinoic acid is contraindicated in pregnancy | 26 | 19,1% (25) | -- | 0,8% (1) | 80,2 (105) |
| Q17 Panado is NOT contraindicated in pregnancy | 8 | ----- | 5,3%(7) | 0,8%(1) | 93,9% (123) |
| Q17 Don't know | 11 | --- | --- | 8,4%(11) | 91,6% (120) |

Valproic Acid was recognised as posing a risk to the foetus by 52, 7% (69) of the students; 62 (46, 6% and 0, 8%) of the students did not indicate this drug as being contraindicated in pregnancy by their non-response. Valproic acid is an anti-convulsant which readily crosses the placenta to the foetus. Abnormalities in the foetus and newborn include intrauterine growth retardation, and defects in neural tube closure, although the risk for these is approximately 1%-2%, but in conjunction with other anticonvulsant therapy, the risk for these and other abnormalities including cardiac defects, facial defects, skeletal and head and neck defects are increased (Briggs et al, 2002).

Retinoic acid was indicated by 19, 1% (25) of the students to pose a risk to the foetus. One hundred and five (105) of the students did not respond to the question on valproic acid. Retinoic Acid (Vitamin A) is a retinoid used for the treatment of acne and other skin disorders, and for the treatment of leukaemia. The effects in a foetus include damage to the central nervous system, craniofacial deformities, cardiovascular abnormalities and thymic anomalies. The combination of these defects is termed retinoic acid embryopathy (Jones, 1997). Twenty five (19, 1%) students indicated that they were aware that retinoic acid could be considered a risk in pregnancy, and 81% (105 and 1) did not indicate this drug as contraindicated during pregnancy. Seven students (5, 3%) incorrectly stated that Panado was a drug that was contraindicated in pregnancy, and the majority of the nurses, 123 (93, 9%), by their non-response to this option, indicated that Panado does not pose a risk to the foetus nor is contraindicated in pregnancy. **A limitation in this question was identified** in that a non-response by a student did not clearly establish whether a student did not identify that particular drug as posing a risk to the foetus, or whether he/she was unsure.

Foetal alcohol syndrome (FAS) is caused by in utero exposure to large amounts of alcohol. The diagnosis was initiated in 1973 by Kenneth L Jones and David Smith (May, Brooke, Gossage, Croxford, Adnams, Jones, Robinson & Viljoen, 2000). A characteristic pattern of abnormalities was found in children who had been exposed to large amounts of alcohol prenatally. The characteristic features include growth retardation, a long smooth philtrum, small palpebral fissures, microcephaly, and neurological deficit. A confirmed diagnosis can be made by a clinical and neurological examination and must include a history of alcohol consumption by the mother during the pregnancy (May et al, 2000). FAS, being the common preventable cause of mental retardation worldwide, has reached vast proportions in socio-economically compromised communities in the Northern Cape, Gauteng and Western Cape Provinces (Viljoen, 2003).

**Question 18** asked which alcoholic beverages could cause foetal alcohol syndrome and the majority of the students, 83 (63, 4%) indicated that beer, wine and spirits contributed to the syndrome. **Question 19** gave four options of clinical features of children with FAS. The majority of the students 107 (81, 7%) recognised that developmental delay was a feature of

foetal alcohol syndrome, 24 (18, 3%) did not respond; 22 (16, 8%) indicated that a long smooth philtrum was a feature, 108 (82,4%) did not respond; 46 (35, 1%) indicated short palpebral fissures to be a feature, 85 (64, 9%) did not respond to this option; 77 (58, 8%) indicated that these individuals have microcephaly, 53 (40,5%) did not respond to this option. Once again, **a limitation in the questionnaire was identified during the discussion of question 19**, in that a non-response by a student to one of the options did not clarify whether the student did not recognise the feature as one of FAS or was unsure. FAS is considered to be a priority condition in South Africa according to the National Health Policy Guidelines and primary prevention should include the discouragement of alcohol consumption during pregnancy (DoH[a], 2001).

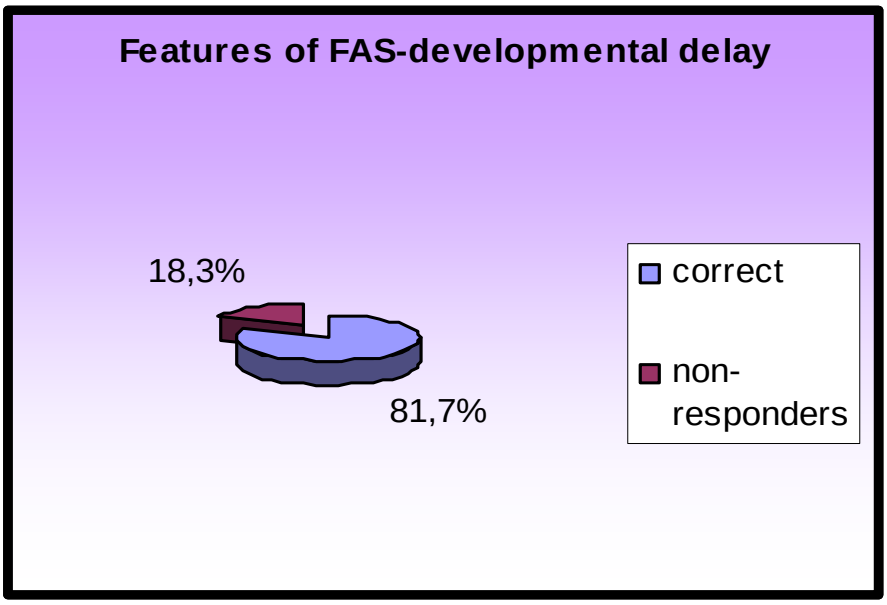


**Figure 4.9: Developmental delay as a feature of Fetal Alcohol Syndrome**

Although the majority of the students appear to be aware of the definition of multifactorial inheritance and seem to understand the concept of teratogenesis, the results indicate that clarification is needed as to which congenital abnormalities fall into these two categories, and as to which women are at risk, and what medication could potentially cause congenital birth defects.

## 4.5 Section four: Epidemiology of inherited conditions commonly found in South Africa

This section covered the epidemiology of conditions commonly found in South Africa, eliciting responses as to whether the conditions mentioned were common, very common or very uncommon. Included in this section were questions relating to the incidence of specific conditions in specific ethnic or language groups and the rate of birth defects in South Africa.

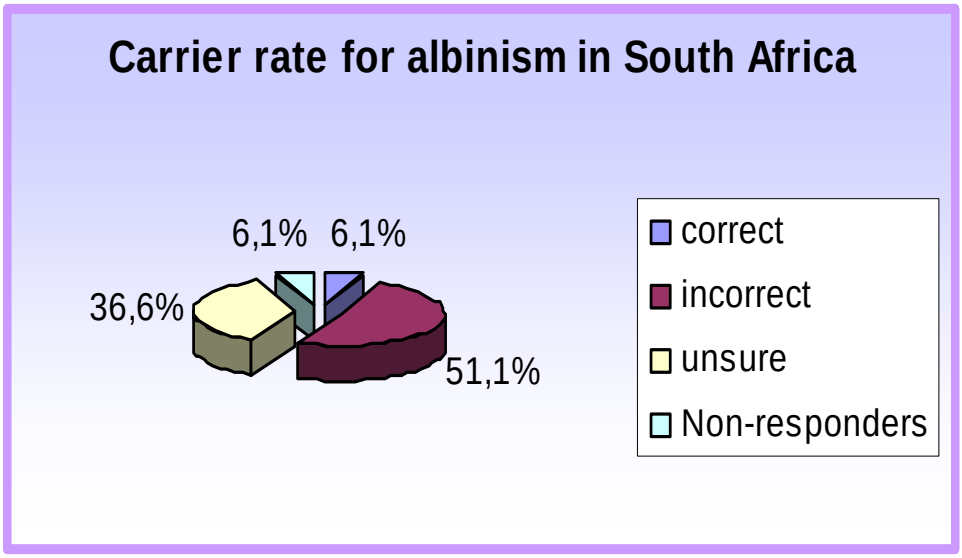


**Figure 4.10: Carrier rate for albinism in South Africa**

Of 123 students who responded to Q**uestion 20** addressing the carrier frequency of albinism in the black population in South Africa, 6, 1% (8) were correct in their answer that the carrier frequency is very common. Sixty seven of the students (51, 1%) were incorrect and 36, 6% (48) were unsure of the correct answer. Eight (6, 1%) did not respond. The carrier rate of albinism is South Africa is approximately 1 in 32, and the frequency of affected individuals is therefore approximately 1 in 3,900 (Kromberg & Jenkins, 1982) There are many myths and legends associated with the condition, including that a person with albinism does not “die like other people”, but just “disappears”. Another myth exists in that it is said that a baby with albinism is born if the mother conceived during menstruation or as a punishment for bad deeds done by the parents (Kromberg, 1990). The results clearly indicate that nurses are unaware of the high carrier frequency in the black community, and thus they would not be able to advise couples of their risks of having an affected baby, therefore impacting on the patients’ rights to

accurate information regarding their reproductive choices. The importance of understanding and rectifying the beliefs surrounding the myths and legends should be emphasised so that couples having affected babies are alleviated of inherent fears associated with these beliefs.

The carrier frequency of cystic fibrosis (CF) is very common in Caucasians and is approximately 1 in 25 (Harper, 1998). Of the 123 students who responded to **Question 21** asking the approximate carrier frequency of CF in the South African white population, 17, 6% (23) were correct, 28, 2% (37) were incorrect and 48, 1% (63) were unsure of the correct answer. There were 8 (6, 1%) non-responders to this question. In view of the fact that the majority of the students were black South Africans, it may be an explanation of the poor knowledge of this particular carrier frequency. However, their knowledge of the carrier frequency of albinism in the previous question was also poor, thus refuting this argument.

Cystic fibrosis (CF) is an important cause of chronic illness and death in childhood and early adult life. It is one of the most common autosomal recessive disorders, affecting the lungs and the pancreas, with a variety of clinical presentations, including meconium ileus in the neonate (obstruction in the small bowel caused by viscous meconium), recurrent chest infections and significant failure to thrive (Mueller and Young, 2002).

CF was previously unrecognised as a condition in the black communities of South Africa. However, children diagnosed with chronic and repeated chest infections and failure to thrive with subsequent malnutrition were possibly misdiagnosed and a diagnosis of CF not recognised (Carles, Desgeorges, Goldman, Thiart, Guittard, Kitazos, de Ravel, Westwood, Claustres & Ramsey, 1996). The carrier frequency in the black South African community has now been reported to be 1 in 34 with a projected incidence of 1 in 4624 births (Goldman, Graf & Ramsay, 2003). The gene for CF was mapped to chromosome 7q31 in 1985 and approximately 70% of mutations are accounted for by delta F508. There are now more than 27 known mutations that can be tested for within the CF gene, and thus approximately 85% of all carriers can be identified (Mueller & Young, 2001).

If individuals and communities are made more aware of the carrier risks in their particular population, they may request testing prior to making reproductive choices, specifically if a couple is Caucasian. If a couple is consanguineous, there would be a 1 in 16 chance of both of them carrying a common recessive gene (Harper, 1998), such as CF, and it would be advisable to test at least one of the couple.

**Table 4.6: Epidemiology**

| Question (N=131) | n | Correct answer % | Incorrect answer % | Unsure % | Non-responders |
|---|---|---|---|---|---|
| Q20 The carrier frequency of albinism in SA | 123 | 6,1 (8) | 51,1 (67) | 36,6 (48) | 6,1 (8) |
| Q 21 Carrier frequency of cystic fibrosis in Caucasians | 123 | 17,6 (23) | 28,2 (370 | 48,1 (63) | 6,1 (8) |
| Q22 For whom should testing for porphyria be offered? | 115 | 12,2 (16) | 14,5 (19) | 61,1 (80) | 12,2 (16) |
| Q23 Which genetic test should be offered to people of Indian origin? | 109 | 9,9 (13) | 23,7 (31) | 49,6 (65) | 16,8 (22) |
| Q24 What genetic test should be offered to French speaking Africans? | 101 | 19,1 (25) | 10,7 (14) | 47,3 (62) | 22,9 (30) |

**Question 22** asked as to which population in South Africa Porphyria testing should be recommended; 12, 2% (16) of the students correctly identified the Afrikaans speaking individuals as those that should have testing, 14, 5 % (19) answered incorrectly, 61, 1% (80) were unsure of the correct answer and 12,2% (16) did not respond.

Porphyria variegata is a common and manageable condition, the condition originally traced back to 1688 when Ariaanntje Jacobs (also known as Ariaantje Adriaannse) an orphan child brought out to marry the first free burghers in the Cape, married Gerrit Jansz van Deventer

(from the Deventer district in Holland). Their daughter, married Cornelis van Rooyen and thus it was initially thought that all people with a surname of Van Rooyen (Porphyria was also referred to as van Rooyen's skin), would be at risk. With intermarriage of families and language groups, all people with Afrikaans ancestry would be at risk for inheriting this dominant gene (Dean, 1963).

Approximately 95% of South African individuals with variegate Porphyria have a common mutation as they are all descended from the original Dutch couple (Dean, 1963). Porphyria testing should be offered to Afrikaans speaking individuals in South Africa, specifically if there is a family history or if an individual is symptomatic. Features of porphyria variegata include sensitivity to sunlight causing lesions on exposed skin, sensitivity to certain foods, for example pork and red meat, and negative reactions to anaesthetic and other drugs (Hift, Meissner, Meissner & Petersen, 1999).

**Question 23** addressed which test a couple of Indian origin should be offered prior to conception. Thalassaemia, being the correct response was answered correctly by 13 (9, 9%) students, 23, 7% (31) answered incorrectly and 49, 6% (65) were unsure of the correct answer. Twenty two (16, 8%) did not respond.

Thalassaemia is a severe form of anaemia with where the homozygote usually presents in the first year of life. It is common for affected individuals to die in their late teens or early twenties, but with iron-chelating treatment, their survival is considerably better (Mueller & Young, 2002); if a couple both carry the faulty gene, there would be a 25% chance of offspring inheriting this disorder. In South Africa, there is a large Indian community and consanguinity is encouraged in some of the castes. As the carrier rate is 1 in 30 (Rimoin, Connor, Pyeritz & Korf, 2002), the risk of a couple both having the gene would be 1 in 90, and thus the overall risk for a couple who has not been tested for their carrier status to have an affected baby would be 1 in 360. Nurses must be aware of particular conditions associated with the variety of population groups in South Africa in order to provide a comprehensive service and to advise couples regarding their reproductive risks and choices.

**Question 24** asked which test should be offered to a French-speaking Black couple from Central Africa. If a black French-speaking couple request carrier testing, exclusion of sickle cell anaemia should be offered; the sickle cell trait is carried by 1 in 12 black individuals in Central Africa (Rimoin et al, 2002); and is as high as 1 in 5 in the Democratic Republic of the Congo (Rimoin et al 2002). Of the 101 respondents to this question, thirteen (9, 9%) students answered correctly, 23, 7 % (31) answered incorrectly and 49, 6% (65) were unsure of he correct answer and thirty (22, 9%) students did not respond.

Sickle cell anaemia is a common haemoglobinopathy and the presenting signs and symptoms, associated with the abnormal "sickling", causing clumping and increased viscosity of the red blood cells. The sickle cells cause obstruction to the small arteries and result in poor oxygen supply to the tissues. Other symptoms arising from this include sudden limb or chest pain, a high temperature and dark urine. This is referred to as a sickle cell crisis. Affected individuals have a shortened lifespan but, with early recognition and treatment, this can be improved (Mueller & Young, 2002).

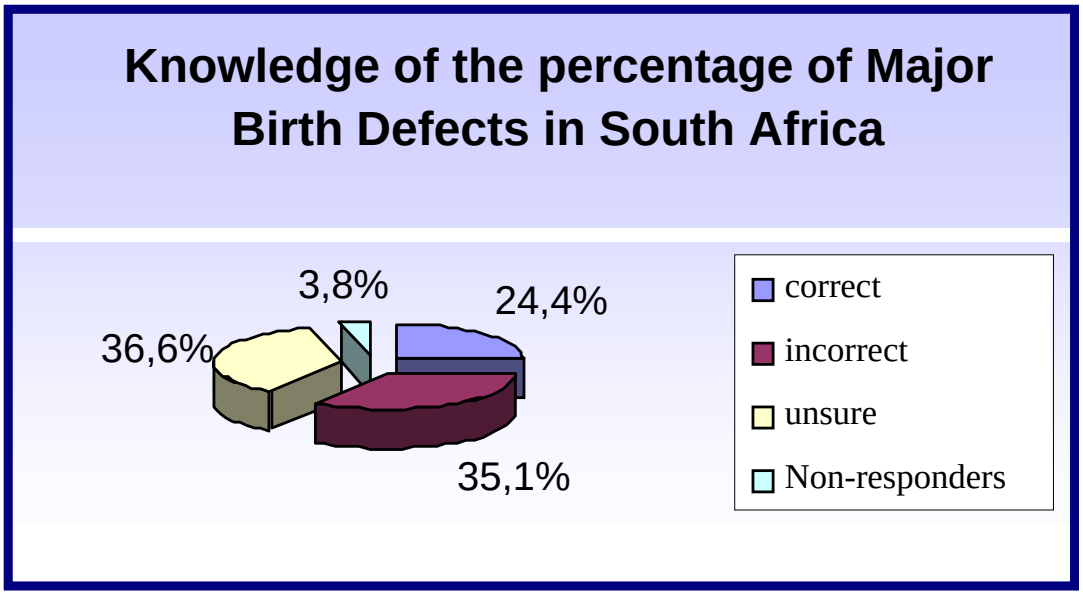


**Figure 4.11: Percentage of Major birth defects in South Africa**

Of the 126 students who responded to **question 25** enquiring as to the percentage of major birth defects in South African liveborn babies, 32 (24, 4%) were correct, 46 (35,1%) and 48 (36,6%) were incorrect and unsure respectively, and 5(3,8%) did not respond. Thus the majority of the nurses did not know the answer. The incidence of birth defects in developing

countries, according to the Department of Health Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities (2000), is not well documented, but it is stated that the “cumulative incidence of severe (significant) birth defects and genetic disorders reaches eight percent by the age of five years”. It is also estimated that if all late onset genetic conditions were included in the equation, including hypertension, diabetes, and inherited cancers, that 60-65% of the population will have a genetic condition in their lifetime.

The Department of Health National Health Policy Guidelines (2001) emphasise that at a Primary Prevention level, every effort must be made to improve health, nutrition and education, particularly of women. Women should have improved access to quality prenatal care and genetic counselling, including information about teratogens, infective agents causing birth defects, like rubella, substances such as alcohol and other recreational drugs that can cause birth defects, and should receive folic acid to reduce the risks of particular birth defects such as neural tube defects cardiac abnormalities and cleft lip and palate (DoH[a], 2000).

Individuals from high risk populations attending Primary Health Care and antenatal clinics must be informed timeously about their possible risks and options available to them. Nurses working in these facilities should be aware of the prevalence of the various inherited conditions in their cross-cultural and cross-racial communities and advise members of the community accordingly. Unless nurses working in the Primary Health Care facilities are aware of the carrier status of specific conditions in specific ethnic groups and are alert of the high frequency of birth defects, they may not recognise the importance of providing these amenities at the Primary Health Care level.

## 4.6 Section five: Indications for an increased risk for having a baby with a birth defect, prenatal testing and diagnostic techniques

This section covered questions about increased risks for having a baby with a chromosome abnormality, prenatal testing availability, risks and limitations associated with the tests, and the use of folic acid in pregnancy to decrease risks of certain abnormalities.

**Table 4.7: Risks and Tests**

| Question (N= 131) | n | Correct answer % | Incorrect answer % | Unsure % | Non responders % (n) |
|---|---|---|---|---|---|
| Q26 There is an increased risk for having a child with a chromosome abnormality if a couple has had a previous child with a chromosome abnormality | 46 | 35.1 (46) | __ | __ | 64,9 (85) |
| Q26 If the mother >35 there is an increased risk for having a baby with a chromosome abnormality | 66 | 50,4 (66) | __ | __ | 49,6 (65) |
| Q26 The risk for having a baby with a chromosome abnormality with high exposure to radiation | 52 | 39,7 (52) | __ | __ | 60,3 (79) |
| Do all of options in Q26 pose a risk | 76 | 55,7 (73) | 2,3 (3) | __ | 42 (55) |
| Do none of the options in Q26 pose a risk | 1 | __ | 0,8 (1) | __ | 99,2 (130) |

The first question in this section, Q**uestion 26**, addressed the possible increased risks for having a baby with a chromosome abnormality. Forty six (35, 1%) of the students answered correctly that there was an increased risk for having a baby with a chromosome abnormality where there is a previous child with an abnormality, 64,9% (85) did not respond. If a mother is over 35 years of age, her risk will be increased; 50, 4% (66) of the students answered correctly, 49, 6% (65) did not respond. Fifty two (39, 7%) students answered correctly about

radiation as a risk factor, and 60, 3% (79) did not respond to this question. The majority of the nurses, 55, 7% (73) recognised correctly that all three options posed an increased risk to the mother for having a baby with a chromosome abnormality.

Chorionic villus sampling is the earliest invasive test available for prenatal DNA testing. In Q**uestion 27**, two (1, 5 %) students answered this correctly, 55% (72) answered incorrectly, 19, 8% (26) were unsure of the correct answer and 31 (23, 7%) did not respond. Individuals who are at high risk for having a baby with a genetic condition, or who have had a previously affected child, often wish to have a very early invasive test for reassurance or to allow them the option to terminate an affected foetus earlier rather than later in the pregnancy. Enlightened midwives in an antenatal setting can inform "at risk" individuals of the earliest testing available and refer them appropriately to centres that can offer these tests should the patients so request.

An amniocentesis involves the extraction of amniotic fluid, which contains amniocytes, fibroblast and other cells from the foetus, for chromosome analysis, DNA analysis and biochemical testing. **Question 28** presented four options for facts regarding the procedure of amniocentesis which is ideally performed between 16-20 weeks gestation. Amniocentesis poses a risk in that there is an approximate risk of miscarriage of <1% and this figure would also be dependent on the expertise of the technician (Harper, 1998). Other risks associated with amniocentesis would include Rhesus sensitisation if the mother is Rhesus negative, and anti-D antibody treatment should be given after the procedure (Harper, 1998).

In South Africa, with the increasing numbers of HIV positive pregnant mothers, care must be taken to ensure minimal mother to child transmission of the HIV virus through invasive procedures such as amniocentesis. To date there are no data to support the increased risk of transmission of the HIV virus to the foetus via such a procedure. However, precautions need to be in place, in the form of giving the mother antiretroviral therapy, prior to any invasive procedure. Fifty three (40, 5%) student nurses answered incorrectly that an amniocentesis is best performed at 12 weeks. Of the 17 nurses who responded to the option that there is <1%

risk of miscarriage, 17 (13%) answered correctly. One hundred and fourteen (87%) students did not recognise this as a fact by their non-response. The majority of the students, 125 (95, 4%) correctly, did not indicate that amniocentesis allows for the examination of chromosomes only, and 86 (65, 6%) acknowledged accurately that amniocentesis can be used for biochemical and DNA studies; 44 (33, 6%) did not respond to this option. Only 15 (11,5%) were not sure of any of the correct answers, of which there were only two correct options, namely that amniocentesis has a low miscarriage risk of less that 1% and that amniocentesis can be used for biochemical and DNA studies. **A limitation in this question** was identified in that a non-response by a student did not clarify whether the student did not know the answer or whether he/she was unsure for that particular option.

**Table 4.8 Prenatal tests**

| Question (N=131) | n | Correct answer 5 | Incorrect answer % | Unsure % | Non-responders % |
|---|---|---|---|---|---|
| Q27 What is the earliest test available for DNA testing | 100 | 1,5 ( 2) | 55 (72) | 19,8 ( 26) | 23,7 (31) |
| Q28 An amniocentesis is best performed at 12 weeks | 53 | ` __ | 40,5 (53) | __ | 59,5 (78) |
| Q28 Amniocentesis has a low miscarriage risk of less than 1% | 17 | 13 (13) | __ | __ | 87 (114) |
| Q28 Amniocentesis allows only for chromosome analysis | 6 | __ | 4,6 (6) | __ | 95,4 (125) |
| Q28 Amniocentesis can be used for biochemistry testing and DNA testing | 87 | 65,6 (86) | __ | 0,8 (1) | 33,6 (44) |
| Unsure of the correct answers for Q28 | 16 | | __ | 12,2 (16) | 87,8 (115) |

**Question 29** gave four options for possible associations with an increased maternal serum alpha-fetoprotein. An increased maternal serum alpha-fetoprotein (AFP) can be associated with neural tube defects and multiple pregnancies; 15, 3 % (20) of the students were correct in their response to the association with neural tube defects, and 84% (110) did not recognise or indicate that they knew that this was true by their non-response. An increased AFP is also

associated with multiple pregnancies and 8, 4% (11) students indicated that they knew of this association, 91, 6% (120) students did mark this option and 25, 2% (33) were incorrect in their indication that an increase in AFP levels is associated with chromosome abnormalities, 97 (74%) did not respond, which could indicate that they are aware that this is not a fact. The majority, 51% (68) of the students were unsure of the answers to this question. **A limitation in this question** was identified as a non-response by a student did not clarify whether the student knew the answer to be incorrect or correct, or whether he/she was unsure.

In the maternal serum triple test, an increased maternal serum AFP may be associated with incorrect gestational age of the foetus, intrauterine bleed, threatened miscarriage or abdominal wall defect (Mueller & Young, 2002).The maternal serum triple screen was introduced as a screening technique specifically aimed at mothers under the age of thirty five. It was noted that at 16 weeks gestation, maternal serum AFP and unconjugated oestriol (E3) levels appeared to be decreased in pregnancies with Down syndrome foetuses as compared to normal pregnancies. Levels of human chorionic gonadatrophin appeared to be elevated. These levels in conjunction with the maternal age, the gestational age of the pregnancy, the maternal weight and history of diabetes, neural tube defects or Down syndrome are analysed with a computerised programme which will adjust the maternal age related risk for having a baby with Down syndrome (Mueller & Young, 2002). If the test shows a positive result, that is, when the patient's risk is higher than the cut off level (which varies between laboratories) she may then opt to have a diagnostic invasive test.

**Table 4.9: Knowledge about levels of AFP in maternal serum and use of folic acid in pregnancy**

| Question (N=131) | n | Correct answer % | Incorrect answer % | Unsure % | non-responders % (n) |
|---|---|---|---|---|---|
| (Q29) ↑ AFP is associated with neural tube defects (NTD's) | 21 | 15,3 (20) | __ | 0,8 (1) | 84 (110) |
| (Q29)↑AFP is associated with multiple pregnancies | 11 | 8,4 (11) | __ | __ | 91,6 (120) |
| (Q29)↑ risk of AFP is associated with chromosome abnormalities | 34 | 0,8 (1) | 25,2 (33) | __ | 74 (97) |
| (Q29)↑ AFP is not associated with an increased risk for albinism | 9 | __ | 6,9 ( 9) | __ | 93,1 (122) |
| unsure of answers in Q 29 | 68 | __ | __ | 51,9 (68) | 48,1 (63) |

In response to Q**uestion 30** regarding the significance of a positive maternal serum triple screen, only 29 (22, 1%) student nurses indicated that they were aware that this indicated a risk for having a baby with Down syndrome. Fifteen (11, 5%) were wrong, the majority of the students, 71(54, 2%) were unsure of the correct answer and 16 (12, 2%) did not respond.

Midwives should be aware of associated risks with abnormal biochemical markers. This will enable them to manage and treat their patients competently and effectively, and refer to appropriate resources if indicated. The Department of Health Policy Guidelines (2001) suggests that primary prevention includes maternal serum triple screening to detect certain chromosome disorders and neural tube defects. In order to manage patients efficiently, nurses must be in a position where they can explain the advantages and limitations of the maternal serum triple test to patients and be able to interpret the results.

**Question 31** gave four possible options for reduced risks of the occurrence of specific conditions if folic acid was taken during pregnancy. Folic acid has been shown to decrease the risk of having a baby with a cleft lip and palate; 14, 5 % (19) students identified this correctly, 84, 7% (111) did not respond. Folic acid has also been shown to reduce the incidence of neural tube defects (NTD); 66, 4% (87) were correct in their answer to this section of question 31 and 32, 8% (43) did not respond. Folic acid does not reduce the risk of increased amniotic fluid, 5,3% (7) students answered this incorrectly stating that it does, and folic acid taken in the pregnancy will not reduce the risks of infections in the baby; 14,5% (19) students indicated incorrectly that it does reduce this risk, 112 (85,5%) did not respond. Seventeen (13%) students indicated that they were unsure of any of the correct answers. **A limitation was recognised in this question** as a non-response to one of the options did not clarify whether the student did not know the answer to be correct or incorrect, or whether he/she was unsure of that particular option.

It has been well documented in literature that folic acid taken prior to conception and during the first trimester of pregnancy can reduce the risk of neural tube defects, cardiac defects and cleft lip and palate. Women who have had a previous child with a neural tube defect should take 4-5mg of folic acid daily prior to and during the first trimester of pregnancy, and all women of childbearing age who are planning to conceive should take 0,4mg of folic acid daily (Mueller & Young, 2002).

**Table 4.10 Maternal serum triple screen and folic acid**

| Question N=131 | n | Correct answer % | Incorrect answer % | Unsure % | Non-responders % |
|---|---|---|---|---|---|
| (Q30)A Positive triple screen may indicate a baby with Down syndrome | 115 | 22,1 (29) | 11,5 (15) | 54,2 (71) | 12,2 (16) |
| (Q31)folic acid ↓ risk for cleft lip and palate | 20 | 14,5 (19) | — | 0,8 (1) | 84,7 (111) |
| (Q31) folic acid ↓ risk NTD's | 88 | 66,4 (87) | 0,8 (1) | — | 32,8 (43) |
| (Q31) folic acid ↓ increased amniotic fluid | 7 | — | 5,3 (7) | — | 94,7 (124) |
| folic acid↓ risk for infections | 19 | — | 14,5 (19) | — | 85,5 (112) |
| Unsure of the answers in Q31 | 17 | — | — | 13 (17) | 87 (114) |

Prenatal testing is available in South Africa at a limited number of facilities in the public sector, namely at the academic hospitals. In Johannesburg, Gauteng, facilities for prenatal ultrasound examination and invasive prenatal testing are available at the Coronation, Johannesburg and Chris Hani Baragwanath Hospitals.

From the researcher's experience, many of the women referred to the Genetic Counselling Clinics either with a child with a birth defect or for prenatal counselling and prenatal testing, were unaware that these facilities were available to them or expressed that they did not know why they had been referred to the Genetic Counselling Clinic.

The results from this section of the questionnaire clearly indicated that the nurses lack the knowledge as to which patients should be offered prenatal testing, the tests available and the associated risks. This aspect should be addressed when the students are in the process of

studying the midwifery component of their course to enable them to be more knowledgeable and thus more confident and competent to manage their antenatal patients effectively and be in a position to offer the clients options of prenatal testing where appropriate.

## 4.7 Section six: Genetic Counselling and students' perceptions of confidence with genetic conditions

This section concentrated on aspects of genetic counselling; descriptions of genetic counselling and the students' perceptions of their confidence to discuss genetic risks and genetic test results with their patients, with justification of their answers.

Genetic counselling is described by Professor Peter Harper (1998 p 3) as " ..the process by which patients or relatives at risk of a disorder that may be hereditary are advised of the consequences of the disorder, the probability of developing or transmitting it and of the ways in which this may be prevented, avoided or ameliorated".

Communication, being a two way process is an integral part of genetic counselling as the clients express verbally or non-verbally their anxiety, fears and hopes. The Genetic Counsellor, as well as imparting valuable facts and information to the client must show empathic understanding and have excellent listening skills to be able to support and guide the client during sensitive and often very emotional decision making processes. Genetic Counselling is regarded as being non-directive in that the counsellor does not direct the client into making decisions one way or another, but rather, after offering the various options available, clearly indicates that the decision reached would be fully supported. A competent genetic counsellor should be non-judgemental and not show prejudice even if a decision reached by the patient may not comply with his/her own moral or religious beliefs.

**Question 32** addressed whether the nurses had heard of Genetic counselling; All 131 nurses responded to this question; 96, 2 % (126) nurses indicated that they had heard of it, and 5 (3, 8%) indicated that they had not heard of it. **Question 33** asked the source from where they had

heard of it and 23, 7% (31) had heard of it through colleagues, 89, 3% (117) had heard of it at college, 40, 5 % (53) through the media, 35, 9% (47) through magazines or journals.

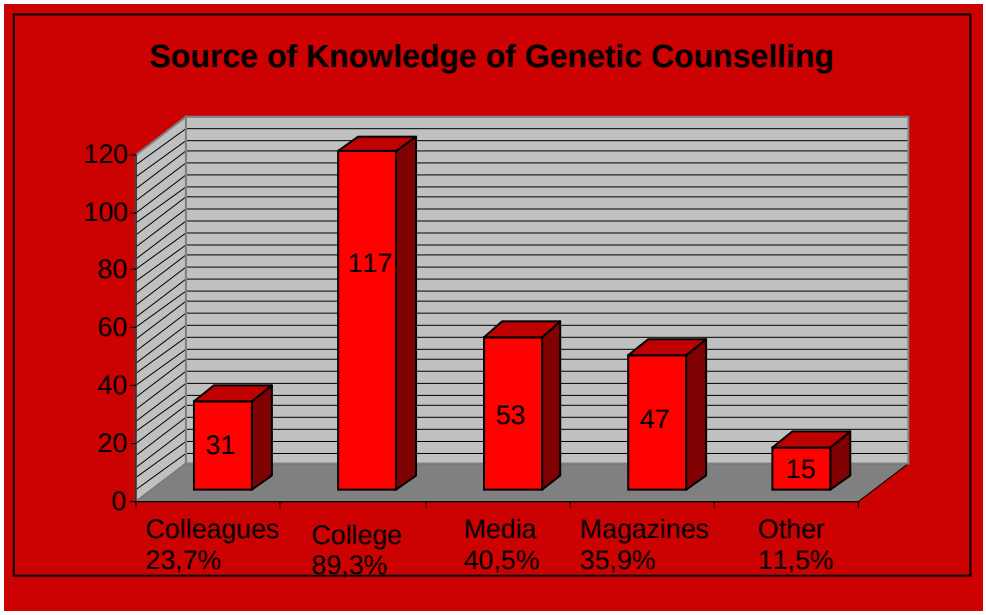


**Figure 4.12: Source of Knowledge of Genetic Counselling**

In **Question 34**, the nurses were asked to mark the factors that they felt described genetic counselling. The options that that they were given are in **table 4.11**.

**Table 4.11: Factors that describe Genetic Counselling**

| Question (N=131) | Ticked (factor described Genetic Counselling) % (n) | Not ticked % (n) |
|---|---|---|
| Explaining the inheritance pattern of a specific condition | 95,4 (125) | 3,8 (5) |
| Avoiding the truth so as not to alarm the family | 1,5 (2) | 96,9 (127) |
| Psychosocial support | 82,4 (108) | 16,8 (22) |
| Advising that they shouldn't have any further children because there is a risk | 73,3 (96) | 26 (34) |
| None of the above | 2,3 (3) | 95,4 (125) |

As a description of genetic counselling, 95, 4% (125) marked that it included an explanation of the inheritance pattern of a specific condition, 96, 9% (127) did not agree that they felt that the truth should be avoided, 82, 4% (108) agreed that it should include psychosocial support

and although the majority of the nurses had “heard” of Genetic Counselling as indicated in question 32, it was disturbing for the researcher to note that 96 (73, 3%) of the nurses indicated that one of the descriptions should be “advising that they shouldn’t have any further children because there is a risk” clearly indicating a directive approach to the students’ perception of Genetic Counselling.

In **Question 35** addressing whether the students felt that genetic counselling be part of comprehensive health care, the majority of the students, 129 (98, 5%), agreed with this statement, one student (0,8%) did not agree, and one student (0,8%) did not respond. In question 36 addressing whether the nurses felt that patients with hereditary disorders would benefit from genetic counselling, 98, 5% (129) agreed, one (0, 8%) student disagreed with this statement and one (0,8%) did not respond. These results clearly indicate that the students realised the importance of integration of Genetic Counselling into the comprehensive management of their patients, which is a priority in the current Primary Health Care Policies (DoH[b], 2000).

It is important for nurses in a primary health care environment to be aware of facilities and resources available to them. In Q**uestion 37**, the students were asked if they knew where the nearest genetic counselling clinic was in their area. It was interesting to note that the majority of the students, in question 32, indicated that they had heard of Genetic Counselling. However, there did not seem to be any correlation of how they would access that service, as only 50 (38, 2 %) of the students in response to question 37, indicated that they were aware of where the nearest genetic clinic in their area was. The majority of the students, 78 (59, 5%) indicated that they did not know where the nearest Genetic Counselling facility was to their area and 3 (2,3%) did not respond. This clearly indicates a need for marketing the Genetic Counselling services through to the Primary Health Care facilities, with leaflets and pamphlets providing addresses and contact telephone numbers for the various clinics held at the academic hospitals as indicated in the Department of Health Primary Health Care Package (2000).

**Question 38** asked if the nurse felt confident to discuss genetic *risks* with their patients. Sixty one percent (81) nurses indicated that they felt confident to discuss genetic risks with their

patients. The themes that emerged in the justification of a "yes" answer to question 38 were increasing patient knowledge, and the patients' right to informed decisions. The majority 96 (73, 3%) and 109 (83, 2%) of the student nurses did not give any justification for their answer. From this, it is not possible to elucidate whether these answers are realistic or expected.
One of the students, in response to question 38 as to whether the student felt confident to discuss genetic risks, indicated that he\she did not feel confident to discuss genetic risks with the patients, and in the justification for this response, stated " it's embarrassing (sic) and unrespecting to other generations''. It is a concern for the researcher that this particular nurse may not be fully aware of his/her responsibilities as a midwife.

Although thirty five (26,7%) of the students indicated that they were confident to discuss risks and justified their answer by indicating the importance of the patients' right to informed choices and fairness, the responses to the question however, did not clearly indicate whether these were personal answers, or anticipated or expected responses to the question. One of the students who answered "yes" to question 38 as to whether he/she felt confident to discuss risks with the patients and justified the answer by responding that "our focus is to help our patients understand the genetic risks therefore I feel confident to discuss but the problem is that I don't have enough information", thus contradicting the "yes" response. One student, indicating in question 38 that he/she did not feel confident to discuss genetic risks with patients justified the answer thus: "I was not socialised to be open about birth, pregnancy etc". This response is curious as this student is currently studying to become a midwife and is dealing directly with pregnancy and birth. It would be presumptuous of the researcher to assume that this student was male; however with regards to his answer, in some South African cultures, it is only the women who deal with issues around pregnancy and birth.

Forty four (33, 6%) of the students answered "no" to question 38. The theme that emerged from the justification of their answer was one of insufficient knowledge. Thirty four (26%) gave this justification, 10 of the nurses who answered "no" to this question, did not justify their answer.

**Question 39** enquired as to whether the nurse felt confident to discuss genetic test *results* with the patients. The response showed that 51, 1% (67) of the nurses felt confident, of which only 32, 1% (42) justified their answer that they felt that it was fair to the patient. Of the 45, 8% (60) nurses who answered that they did not feel confident to discuss genetic test results, only 27, 5% (36) justified their answered by responding that they felt that they had insufficient knowledge and lack of experience. The majority of the nurses did not justify their answers.

From the responses to the questions in this section, although through the pilot study it was confirmed that there were no ambiguities or confusion surrounding any of the questions, it clearly shows that some of the students misunderstood the focus of the questions; the questions aimed to elicit the students' individual feelings regarding confidence in discussing tests and results with patients, rather than what is expected of them. Some of the individual responses indicate that the students may not have understood the inference of the questions clearly.

## 4.8 Section Seven: Human genome project and education issues

This section dealt with the human genome project, and also addressed issues around their current genetic education, and what they felt should be included in their studies to be able to offer a comprehensive genetic service to their patients.

Of the 129 nurses who responded to **question 40** asking whether they had heard of the Human Genome Project, 5,3% (7) indicated that they had heard of it, 93,1% (122) indicated that they had not heard of it and 2 (1,5%) did not respond.

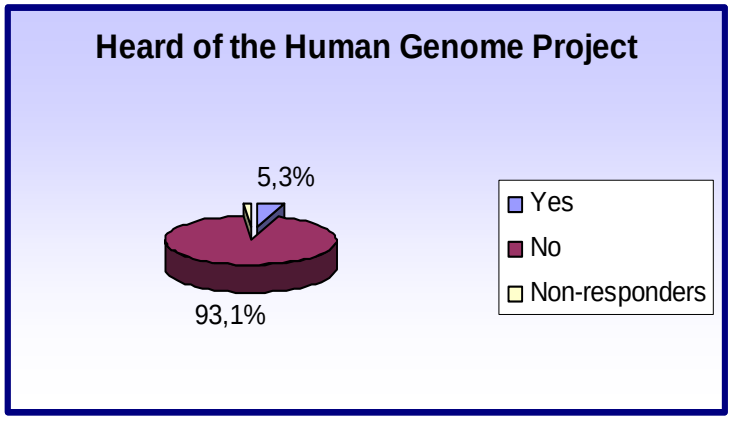


**Figure 4.13: Human Genome Project**

**Question 41** asked for clarification of question 40. If the nurses indicated that they had heard of the Human Genome Project, they were asked to explain what they understood it to be. Of the seven students (5,3%) who indicated that they had heard of the Human Genome Project, one of them explained that what he/she understood it to be was the "cloning of humans for medical purposes (research) for diseases like Parkinson's, Alzheimer etc" . Another student indicated that the HGP was "an artificial way of interacting genes such as cloning". A third student, indicating that he\she had heard of the project questioned "can humans truly be cloned?" Another student understood it to be "study of genes and how they affect individuals and is it hereditary or not". The majority of the nurses 122 (93, 1%) had not heard of the HGP and thus did not respond to question 41.

**Question 42** enquired of those students who indicated that they had heard of the Human Genome Project as to where they had obtained the information giving the options of newspapers, magazines, television, in their course or from colleagues. One of the seven students who said that they had heard of the HGP did not respond to question 42. One student said that he/she had heard of it through television, one said she/he had heard of it during the course, two students indicated that they had read about it in newspapers, and one student indicated that he/she had heard about the HGP through newspapers, magazines and television.

There is a growing concern that nurses globally, in all spheres of nursing care, become more aware of the implications of genetics on disease and their role in management of affected and

"at risk" individuals and families (Lashley, 2000, Burton & Stewart, 2003, Hetteberg & Prows, 2004).

Medical genetics, which was once considered a speciality, now provides a basis for the understanding of illness and health at a different level, as it impacts in every discipline of medicine and nursing. Nurses need to have a clear understanding of genetics, to enable them to become capable and proficient in recognising and managing individuals and families who are potentially at risk for having or developing genetic conditions (Burton & Stewart, 2003). The Human Genome Project (HGP) has changed the face of the management of genetic disease. When once, families were only given empirical risks of inheriting certain conditions including breast cancer and colon cancer, now, nurses, by doing a thorough investigation of a family history, may identify those at risk for developing genetic conditions, and refer them for early intervention by genetic counselling and the subsequent option to test for specific genes. The HGP affects not only the management of genetic disease, but also the prediction and prevention as well as diagnosis  Midwives are often the first to recognise possible risks of genetic disease, but they need to have skills to recognise this and offer advice and referral where indicated. Nurses need to be able to discuss advantages and disadvantages of available tests and be confident to explain results. Paediatric nurses work with children with genetic disease and they also need to assist families with the implication for risks for further affected children and consequences of the disease manifestation (Burton & Stewart, 2003)

**Question 43** asked if the student nurses felt that the genetics that they have studied equipped them sufficiently with relevant knowledge to help patients confidently. Fifty three (40, 5%) of the nurses felt that the genetics that they have studied equips them sufficiently with relevant knowledge to help patients confidently, and the majority, although small, 52, 7% (69) did not feel that the genetics that they have studied equips them with relevant knowledge. Seven percent of the nurses (9) did not respond. These responses do not correlate entirely with the 61% (81) who in question 38 in the previous section, indicated confidence to discuss genetic risks with their patients. However these figures do correlate closely with the 51, 1% (67) who indicated that they feel confident to discuss genetic test results with their patients. More than half of the student nurses feel that their studies do not equip them adequately with relevant

genetic knowledge, indicating that the course content is either insufficient or possibly presented in a manner without clarity as to how they should incorporate genetic knowledge into practice.

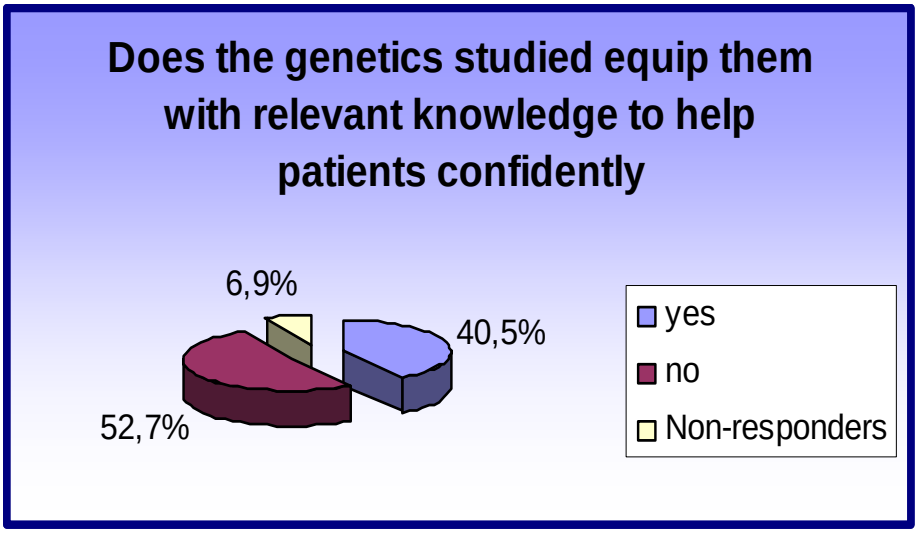


**Figure 4.14 Does the genetics studied equip them with relevant knowledge**

The final question, Q**uestion 44** in the instrument, addressed what additional information the students felt should be received during their training to be able to offer a comprehensive service to their patients. Ninety three students (79, 9%) responded to the question. Two major themes emerged from the answers given by the students. These were firstly, an inclusion of a genetic counselling component and secondly, a comprehensive genetic course. The majority of the students, 95 (72, 5%) and 83 (63, 4%) nurses did not respond to either of these themes respectively. Thirty six (27.5%) of the students felt that there should be a counselling component added to the curriculum and 48 (36.6%) indicated that they felt that there should be a more comprehensive genetic component in the curriculum. It was interesting to note that the nurses did not suggest incorporating genetics into their other subjects, and that genetics is still viewed as a separate branch of learning.

## 4.9 Conclusion

Genetics has historically been taught as a separate subject, and with the advent of the Human Genome Project and the implication that it has on all medical disease, it should ideally be

introduced as a thread running throughout all subjects and disciplines taught at nursing schools (Lea & Monsen, 2003, Prows, 2004). With the new method of teaching medical students in some universities in South Africa, using a problem based learning (PBL) method; this matter is being addressed at medical schools.

Lea and Monsen in 2003 made recommendations for the integration of genetics into the nursing curricula, by incorporating a component including prenatal genetic family history risk assessment, risks associated with advanced maternal age, prenatal testing and diagnosis, prenatal drug and medication use and associated birth defects into the course in reproductive and sexual health. The authors suggested nurses should be taught to take a family history risk assessment using a three generation pedigree. It was recommended that issues surrounding predisposition to adult onset conditions such as Alzheimer's disease, Huntington's disease, cancer, heart disease and bi-polar disease be incorporated. In community health, nurses should be taught how to identify detrimental effects of substances during pregnancy, and they should be trained to be aware of actions that can be taken to prevent birth defects, for example the ingestion of folic acid and carrier screening (Lea & Monsen, 2003).

Hetteberg and Prows (2004) suggest that through the use of their "checklist", basic genetic topics such as cell division chromosomes, DNA, and patterns of inheritance could be included in pathophysiology and reinforced during maternity, newborn, child health, adult health and mental health nursing courses. The authors recommend that history taking, dysmorphology assessment, the drawing of pedigrees, genetic testing, and genetic conditions be covered during maternity, neonatal, child health, adult health and mental health nursing courses. Ethics, psychosocial needs, legal and social implications of genetic disorders could be included and integrated into all the disciplines.

To conceptualise the relationship between genetics, health and nursing, nurses must have an understanding of genetics. To be able to integrate genetic concepts into nursing science the development of co-operative relationships among nursing researchers, as well as with those in genetics and related disciplines is vital (Williams, Tripp-Reimer, Schutte, Barnette 2003).

In this chapter each section of the questionnaire was examined, and the students' responses to the questions elucidated through the use of tables and graphs depicting the descriptive statistics obtained for the analysis. Each section was thoroughly discussed, specifically relating to the first objective of this study namely the examination of the current genetic knowledge of the final year student nurses and the second objective, to elicit whether in the nurses' opinion, there is a need for inclusion of a more extensive genetic component in their syllabus.

In the next chapter the research study is summarised, the recommendations from suggestions from the student nurses are addressed, as well as the limitations and conclusions of the study.

# Chapter Five

# 5. Conclusions, Recommendations and Limitations

## 5.1 Introduction

In this chapter, the conclusions relating to the aim and objectives of the study will be reported. The limitations of the study will be discussed and the third objective, namely, to formulate recommendations of genetic advancements and developments for inclusion into the four-year-diploma nursing curricula based on findings of the study will be addressed. The aim of this study was to examine the genetic knowledge (as prescribed by the South African Nursing Council curriculum and South African Nursing Council (SANC) regulations) of students in their final year of the Diploma in Comprehensive Nursing (General Nursing, Community Health and Psychiatric Nursing) and Midwifery through the research objectives.

### 5.1.1 Summary

**The first objective** was to elicit the final year student nurses' current genetic knowledge through the administration of a questionnaire. After the administration and collection of the instrument at two of the three nursing colleges in Gauteng, an analysis of the answers was done through the statistical package, SPSS, version 11 for Windows, and results were depicted descriptively through the use of pie graphs and histograms. The analysis of the results was discussed in Chapter Four and the overall results indicated a lack of knowledge in various categories in the subject of genetics.

Chromosomes and chromosome abnormalities were addressed in the first section. Although the students indicated that they knew the correct number of chromosomes in a human cell, they had poor understanding of the process of the formation of egg and sperm cells, meiosis. The majority of them knew that there is an extra chromosome 21 in individuals with Down syndrome and recognised the feature of mental retardation, but had poor recognition of other clinical features associated with this condition. Their knowledge of the prevalence of Down syndrome in liveborn babies in South Africa was inadequate. The majority of the students did not know the number of autosomes (44) in a human cell.

Only 50, 4% of the students recognised that there is a 50% chance of passing on a dominant condition, when one parent is affected. This lack of knowledge of the inheritance pattern in dominant disorders could have an impact on their management of families with dominantly inherited conditions, such as breast cancer, colon cancer and hypercholesterolemia, which are all common conditions across all race groups in South African communities. Less than half of the students were aware that conditions inherited in an autosomal dominant manner would affect males and females equally; this could have an implication on their identification of at risk individuals and the management of affected families.

The vast majority of students (70, 2%) were incorrect in their answer that there is a 25% risk for having a baby with a recessive condition if both of the couple carry the gene. With regards to albinism, a common recessively inherited condition in South Africa (where one in thirty black individuals carries the gene), all nurses should be enlightened as to the risks of recurrence and be aware of the associated risks for affected individuals, for example skin cancer.

Only 33, 6% of the students were aware that in X-linked conditions, mainly males are affected, once again, having a potential impact on their management of families affected by this condition. Haemophilia was recognised by less than half of the students as being inherited in an X-linked manner and the majority did not indicate this to be an X-linked condition. This could have major implications on what appear to be “minor” nursing practices, for example, intramuscular injections for immunisation. If an affected baby boy receives an intramuscular injection without prior treatment with Factor VIII, he could present with an intramuscular bleed, which if not treated may cause pain and dysfunction of the limb. Nurses need to be informed and aware of the complications that may occur even in an individual with a mild presentation of this condition.

Duchenne muscular dystrophy is a genetic disease found in all race and ethnic groups. The history of the presentation is classic although parents may not “know” the name of the condition, even though they may be aware of the clinical presentation and the poor prognosis

of the males in their families. A nurse working in a primary health care facility or antenatal or paediatric outpatients department, when taking a history from a mother, must be aware of inherited conditions like Duchenne Muscular dystrophy and should be able to advise families accordingly. Eight student nurses (6, 1%) were aware that this is an X-linked condition. Female siblings of children affected with an X-linked condition are at risk for being carriers and for having affected children. Parents with one affected son would have a 50% risk of having another affected male child. Nurses should be aware of and be attentive to the devastation that such a condition has on an individual and family and be in a position to offer counselling and support, prenatal options and referral to appropriate resources.

Eleven nurses (8, 4%), although a small percentage, indicated that achondroplasia is inherited in an X-linked manner, and 26 (19, 8%) indicated that they thought that cystic fibrosis is an X-linked condition. It concerns the researcher that these students may not be clear as to what these conditions are.

A small majority (61, 8%) of the students correctly recognised the definition of multifactorial disorders, being caused by both genetic and environmental factors, but only 37 recognised that diabetes type II is one of these conditions, forty two nurses (32,1%) incorrectly marked that albinism is inherited in a multifactorial manner, only 26 (19,8%) recognised cleft lip and palate as a multifactorial condition and only 45 (34, 4%) indicated correctly that neural tube defects are inherited as multifactorial defects. This indicates clearly that although many of them correctly identified the definition of multifactorial inheritance, that there is a lack of knowledge by the student nurses as to which conditions are inherited in this manner. This deficit of knowledge could impact on their management of affected individuals planning pregnancies, or those parents with affected children, specifically with regard to risks of recurrence.

The term teratogenesis was understood by a small majority (66.4%) of the students. The majority recognised syphilis as being a teratogenic agent and a small majority (66.4%) recognised rubella as posing a risk to the foetus, but very few students indicated that they

knew that cytomegalovirus (24,4%) and toxoplasmosis (32, 1%) could cause teratogenic effects in a foetus. This could impact on their education of their antenatal patients. Warfarin embryopathy is recognised as a complication of ingestion of this drug during pregnancy; it was interesting to note that student nurses seem to be aware of this complication. However their knowledge regarding other drugs such as valproic acid and retinoic acid as being teratogenic in a foetus was poor.

In South Africa, the rate of foetal alcohol syndrome (FAS) is higher than in any other country worldwide (Viljoen, 2003) and the author noted that the student nurses' knowledge with regard to this condition indicated a clear recognition of this "local" problem. However, the majority of the nurses, although they recognised that there is developmental delay in those affected with FAS, were unaware of the other typical facial clinical features. This could influence their recognition of affected children, and would not prompt them into questioning the mother appropriately, thus the risk to subsequent children could not be necessarily avoided or decreased.

The student nurses' overall knowledge of the epidemiology in the South Africa population is poor. This could be attributed to their lack of exposure to communities other than their own. However, even for the very common conditions, such as albinism, the students were not aware as to carrier rates in the South African communities. This lack of knowledge could lead to incorrect or lack of education to their patients and to the communities for whom they are responsible for health care and management. In the section on prenatal testing and prenatal diagnosis it was noted by the researcher that the nurses have inadequate knowledge with regards to types of testing available and associated risks; but it was reassuring that the majority of the student nurses, although a small majority, (66.4%) of those who answered knew that the ingestion of folic acid during a pregnancy could reduce the risk of neural tube defects.

**The second objective** of this study was to elicit the student nurses' opinions regarding the adequacy of the current genetic component in their curriculum and whether, in their opinion, there is a need for inclusion of a more extensive genetic component.

Although there had been a recommendation over forty years ago that genetics be included n nursing curricula, it still appears that worldwide, genetic content is still lacking (Hetteberg & Prows, 2004). The majority of nurses (96, 2%) had heard of genetic counselling but it was disconcerting for the author to note that 96 (73, 3%) of the student nurses felt that a description of genetic counselling includes "advising that they shouldn't have any further children because there is a risk". Genetic counselling, worldwide, is recognised to be non-directive (Harper, 2000).

Although the majority of the students (98, 5%) indicated that they felt that genetic counselling be a part of a comprehensive health care and patients with hereditary disorders would benefit from genetic counselling, only a small majority 61, 8% felt confident to discuss genetic risks and 51,1% felt confident to discuss genetic test results with their patients. If genetics was an integral part of the curriculum, nurses would ultimately, by the end of their four year training period, be in a position where they felt confident to deal with genetic issues.

The majority of the student nurses, 122 (93, 1%) had not heard of the human genome project. With the imminent finalisation of this project, it is of concern that so few nurses are aware of current events such as this that have an enormous impact on health care from all perspectives; with an emphasis on management of common conditions such as cancer, heart disease, hypertension and diabetes type II to name but a few. Hetteberg and Prows (2004) recommend that nurses must be aware of the challenges health care now faces with the developments from the Human Genome Project, and that should keep up to date with practices that have an effect on management and care in the primary health care setting.

The majority of the nurses (52, 7%) indicated that they did not feel that the genetics that they studied equipped them with the relevant knowledge to help their patients confidently. This clearly indicates a need to incorporate a genetic component into the syllabus. Of the 93 students (79, 7%) who responded to the final open ended question requesting their opinion as to what additional information they feel they should receive in their training, the two major

themes that emerged were that of an inclusion of a complete genetics course and one of a counselling component.

**The third objective** of this study was to make recommendations according to the needs of nurses as to any changes which might be required in the content of the current curriculum. There is clearly a need from the nurses that a more integrated genetic component should be incorporated into the basic curriculum although a number of them stated that they felt the curriculum was “packed” and there was enough to study.

In South Africa, we need to address this issue, with reference to programmes that have succeeded elsewhere, namely the United States of America. Hetteberg and Prows (2004) citing “The Report of the Expert Panel on Genetics and Nursing: Implications for Education and Practice” from Health Resources and Services Administration (2000), emphasise a venture designed to address the need for genetics knowledge and skill competencies for nurses. The Statement on the Scope and Standards of Genetics Clinical Nursing Practice from the International Society of Nurses in Genetics (ISONG), American Nurses Association (1998) urge that all registered nurses perform an important task in the delivery of genetic services and that genetic knowledge is needed so that they are able to recognize and refer individuals and families affected by, or at risk for, developing or passing on genetic conditions (Hetteberg & Prows, 2004).

Prior to introducing a genetic component into the basic curriculum, the authors suggested that workshops to increase the genetic knowledge of tutors and clinical teaching staff be initiated. This would provide them with the competence and confidence to introduce this subject at all levels and in all areas of nursing practice to their students. In the United States of America, a Web-based Genetics Institute (WBGI) was developed, and covered content as the Summer Institutes initiated by the Genetics program for Nursing Faculty (GPNF), funded by the Ethical, Legal and Social Implications Research Program of the National Human Genome Research Institutes of Health. The participants discussed ideas and created plans for the incorporation of genetics into their school’s curriculum and were used to develop a checklist format. The checklist as mentioned in the literature review is divided into four sections: the

first section was to determine existing genetics content, the second to increase faculty awareness about need to include genetics, the third to increase faculty knowledge about genetics and the fourth, to integrate genetics content in nursing curriculum.
Hetteberg and Prows state that there are “three essential steps that must be accomplished before genetics content can be added or improved upon in the nursing curriculum”. Firstly, the amount of material, the type of material and where it should be placed needs to be determined. Secondly that faculty should be encouraged to understand the need to increase the genetic content of the curriculum and thirdly that nursing faculty must become educated about genetics. Each faculty member involved in the teaching of the basic curriculum would need education dependent on her current knowledge base and how she intends to increase the genetics content into the curriculum (Hetteberg & Prows 2004). Lea & Monsen, 2003, discussed that a programme with case-based instructional modules, emphasising clinical applications of genetic principles, was effective in assisting educators to incorporate genetics into existing curricula and giving educators the knowledge and confidence to teach genomics.

At present, in South Africa, an introduction of the GEM (Genetics Education Manual) is to be initiated as a distance education programme, primarily aiming to increase the genetic knowledge of Primary Health Care Nurses, in order to empower them to manage patients with genetics conditions effectively and comrehesively. The researcher feels that if a genetic component is introduced in the four year diploma course that ultimately, distance education and in-service training programmes would suffice as “refresher” courses for registered nurses working in the field. It is also felt that with a comprehensive genetic component introduced into the basic curriculum, nurses, once qualified, would feel confident and competent to address genetic issues as part of their “holistic” management and not see genetics as being separate and unattainable knowledge, available to only a “select” few.

Hetteberg & Prows (2004) recommend that for a basic curriculum, besides the basic human and molecular topics such as cell division, chromosomes, DNA genes patterns of inheritance being addressed in pathophysiology, and being reinforced in midwifery, newborn health, and mental health nursing, other topics including history taking, assessment of the dysmorphic child, drawing of three generation pedigrees, genetic testing and genetic conditions could be

strategically placed in the various subjects in the curriculum. Psychosocial needs of the clients need to be addressed inclusive of ethnic, racial and cultural considerations, as well as ethical legal and social implications (Hetteberg & Prows, 2004). These latter topics would be of vital importance given the vast diversity of our South African communities. The midwife is often the first to recognise a genetic birth defect or the potential risk for a mother to have a child with a birth defect; and it is essential for the midwife taking the antenatal history to be aware of risk factors associated with pregnancy and be able to advise mothers accordingly. The paediatric nurse also deals with genetic conditions. They may be caring for children with life-threatening conditions, and play an important role in the psychosocial support of the parents, siblings and other family members. The primary health care nurse, who is at the forefront of health care and management in South African communities, in accordance with the Primary Health Care Norms and Standards (2000), is often the first to become aware of a family history of cancer or heart disease.

In all areas of nursing, before birth, during birth and postnatally, in childhood and adolescence, adulthood and old age, the nurse is indispensable in care and management.

Genomics is considered to be an integral part of the future of health care and the nurse has a responsibility to keep up with changes in order to provide comprehensive and committed care and management to the communities of South Africa.

## 5.2 Recommendations

### 5.2.1 Nursing Education

The researcher identified the following recommendations for nursing education:

- The researcher, in collaboration with interested stake holders would draw up an updated education programme for nursing educators, based on programmes used in the United States of America and adapted specifically for South Africa.

- A recommendation, based on the findings of the study, is that a genetic component be included in all the basic curricula of the four year diploma in nursing (General nursing, Community nursing, Psychiatric nursing) and Midwifery, the university degree courses for nurses (BA Nursing), as well as in post-basic and postgraduate courses.

- A formal course, in the vein of the Genetics Summer Institute (Prows et al, 2003) could be held for nursing educators, representative of each of the nine provinces, and each of the nursing schools.

### 5.2.2 Nursing Practice

To enhance nursing practice in South Africa, the recommendations from the researcher are as follows:

- In-service genetic training, as a "hands on" experience could be offered at the clinical training centres of the nursing schools. Experienced genetic clinicians and trained genetic nurses would be approached in the various areas in South Africa to assist with the training of clinical tutors and student nurses to examine individuals with genetic conditions, and recognise individuals and families at risk for inheriting a genetic disorder.

- Visits to the schools for the intellectually and physically disabled would be arranged, as well as visits and practical "hands on" training at the schools for the visually and hearing impaired in order to familiarise tutors and students with common genetic conditions.

## 5.3 Further Research

Arising from this study, the researcher identified the following recommendations for further research in the field of genetics and nursing education:

- An assessment of the genetic knowledge of diploma student nurses in other provinces in South Africa
- Assessment of the genetic knowledge of nurses doing the four-year Bachelor of Science degree course in South Africa
- An assessment of the genetic knowledge of nurse educators at the nursing schools in South Africa

## 5.4 Limitations

The following limitations in this study were identified:

- Demographics were not included. These would have elucidated the ethnic or language background of the students and this may have given a clearer depiction of some of the responses to the questions. A goal of the research programme ELSI (Ethical, Legal, and Social Implications) of the National Human Genome Research Institute of Health (USA) is to explore the use, understanding and interpretation of genetic information and the use of genetic services by different ethnic and socioeconomic populations. (Williams, Tipp-Reimer, Schutte & Barnette, 2003) and this aspect needs to be explored in South Africa.

- The ages of the students may have clarified questions relating to confidence about dealing with genetic risks and explanation of results to patients. The majority of students in South African Nursing Schools are black and the first language is generally not English. Although the students study in English, there may have been

misunderstanding of some of the questions asked, thus giving a rationalization to the contradictory statements and answers given by some of the students. However, the pilot study did not suggest any irregularities, any ambiguities or any confusing issues.

- The sample from one of the colleges should have been larger, but due to miscommunication from the tutor who co-ordinated the date and time for the distribution and completion of the questionnaire, a large percentage of the students did not arrive for the selection.

- Only the genetic knowledge of nurses in Gauteng was examined. This may be indicative of the overall knowledge of student nurses in South Africa, but this aspect would need to be explored through further research.

- A limitation in the design of some of the questions in the instrument was recognised during the analysis and discussion of the results. It was not possible to clarify from the particular questions, discussed in detail in Chapter Four, whether a non-response to a specific option in these questions indicated clearly whether the student felt the answer to be incorrect or whether he/she was unsure of the answer. Should this questionnaire be used for other research studies, it is recommended that those particular questions be modified accordingly, and thus attain more conclusive results.

## 5.5 Conclusion

From this study, it can be concluded that the overall genetic knowledge of the final year diploma student nurses is deficient. With reference to their knowledge of chromosomes and chromosome abnormalities, their knowledge was limited. Their understanding of modes of inheritance and specific risks related to the single gene disorders inherited in a dominant, recessive or X-linked manner is lacking. There were areas in which their knowledge was greater, namely with reference to multifactorial inheritance and the effect of drugs and infections in pregnancy, although the minority of the students knew which genetic conditions are inherited in a multifactorial manner. The students were also knowledgeable about foetal

alcohol syndrome. However their epidemiological knowledge was poor, with direct reference to their lack of knowledge as to the frequency of common conditions in South Africa, specifically albinism and Down syndrome. Their understanding of prenatal testing was insufficient, and their knowledge of specifics about prenatal tests was lacking. However, most of the students were aware that the ingestion of folic acid during a pregnancy could reduce the risks of having a baby with a neural tube defect. Although the majority of the nurses had heard of genetic counselling, the majority of them felt that a description of genetic counselling included "advising that they shouldn't have any further children because there is a risk", which is against the basic non-directive manner in which genetic counselling is advocated. Though many of the nurses indicated that they felt confident to discuss genetic test results and risks with their patients, their answers and justification did not appear to correlate appropriately.

Ninety three (79,9%) students responded to the final question in the instrument asking what additional information they felt nurses should receive during their training to be able to offer a comprehensive genetic service to their patients. The two major themes that emerged were that they felt there should be a comprehensive genetic course as well as counselling courses.

In conclusion, it was noted that there is a deficiency of genetic content in the current nursing curriculum and that the genetic knowledge gained by the students following the four year diploma in nursing (general, community, psychiatric) and midwifery, is not sufficient to provide them with the knowledge, confidence and competence to manage patients and families with genetic conditions.

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# Appendix 1

## Letter of permission from the Head of Nursing Education

GAUTENG PROVINCIAL GOVERNMENT

DEPARTMENT OF HEALTH

**ENQ.** : Ms. P. C Nel
☎ : 011 355 3522
*fax* : 011 355 3549

14 June 2002

MS. M. GLASS
P.O. BOX 3030
BROMHOF
2154

REQUEST FOR PERMISSION TO CONDUCT RESEARCH: AN ASSESSMENT OF THE GENETIC KNOWLEDGE OF FOURTH YEAR DIPLOMA NURSING STUDENTS.

Your application dated the 14 April 2002, received on the 12 June 2002, refers.

I have the pleasure in informing you that you are hereby granted permission to conduct the above research in Nursing Colleges in the Gauteng Province provided that the following conditions are met:

⇒ **You receive permission from the Principals of the Colleges.**
⇒ **The Management is kept informed regarding progress of the research.**
⇒ **The research does not interfere with the learning environment of the respondents.**
⇒ **The confidentiality and the anonymity of the respondents and the institution is maintained at all times.**
⇒ **A copy of your completed study is donated to this Department.**

Best wishes for your research. The Department looks forward in hearing about your progress in this study. Please do not hesitate to contact us if we can be of any assistance.

**MS. P.C NEL**
**ASSISTANT DIRECTOR : PROFFESSIONAL DEVELOPMENT**

# Appendix 2

## Letter of request to Heads of Departments at Nursing Schools

P.O.Box 3030
Bromhof
2154
April 14th, 2002

Attention: Ms L.Van Wyk
Gauteng Department of Health
Sub directorate: Professional Development
Private Bag X085
Marshalltown
2107

Dear Ms van Wyk

I am currently doing research for a Master's degree through the Department of Nursing Education at the University of the Witwatersrand. The topic that I have chosen is "An Assessment of the Genetic Knowledge of Fourth-Year Diploma students ".

I would like to request permission to administer a paper-based questionnaire to the fourth- year Diploma Nursing students during mid 2002 at the nursing colleges in Gauteng. The questionnaire will be answered in the college at a time that is suitable for the lecturers and should take approximately 45 minutes to complete.

I have attached a copy of my research proposal and outline for your perusal. Approval has been obtained from the Postgraduate committee and the Committee for Research on Human Subjects (Medical) of the University of the Witwatersrand.

Should your permission be granted, I will forward my request to the principals of the Nursing schools, and upon their permission and co-operation, arrange a suitable date and time to administer the questionnaire.
I hope that you will find my request acceptable and acknowledge that the research I am planning is valid and relevant. Should you have any queries, I can be contacted on 011-7920122 or 0828537113.
Thanking you in anticipation,
Yours faithfully,

Merlyn Glass(RN, RM, Dip Paeds)

P.O.Box 3030
Bromhof
2154
April 14th, 2002

The Principal
Baragwanath College of Nursing

Dear Mrs Ndlovu,

I am currently doing research for a Master's degree through the Department of Nursing Education at Wits University. The topic that I have chosen is “An Assessment of the Genetic Knowledge of Fourth-Year Diploma students”.

I would like to request permission to administer a paper-based questionnaire to the fourth- year Diploma nursing students during mid 2002. The questionnaire should be answered in the classrooms at a time that is suitable for you, and should take approximately 45 minutes to complete.

I have attached a copy of my research proposal and outline for your perusal. Permission has been granted from the Head of Nursing Education at the Bank of Lisbon, and should you agree, could we arrange a suitable date and time to administer the questionnaire.

I hope that you will find my request acceptable and acknowledge that the research I am planning is valid and relevant. On completion of the study, a copy of the findings will be available for you, should you so request.

Thanking you in anticipation,
Yours faithfully,

Merlyn Glass(RN, RM, Dip Paeds)

P.O.Box 3030
Bromhof
2154
April 14th, 2002

The Principal
S.G. Lourens School of Nursing
Private Bag X755
Pretoria 0001

Dear Mrs Rambau,

I am currently doing research for a Master's degree through the Department of Nursing Education at Wits University. The topic that I have chosen is “An Assessment of the Genetic Knowledge of Fourth-Year Diploma students”.

I would like to request permission to administer a paper-based questionnaire to the fourth- year Diploma nursing students during mid 2002 as my pilot study. I would like approximately 10-15 students to participate. The questionnaire should be answered in the classrooms at a time that is suitable for you, and should take approximately 45 minutes to complete.

I have attached a copy of my research proposal and outline for your perusal. Permission has been granted from the Head of Nursing Education at the Bank of Lisbon, and should you agree, could we arrange a suitable date and time to administer the questionnaire.

I hope that you will find my request acceptable and acknowledge that the research I am planning is valid and relevant. On completion of the study, a copy of the findings will be available for you, should you so request.

Thanking you in anticipation,
Yours faithfully,

Merlyn Glass(RN, RM, Dip Paeds)

P.O.Box 3030
Bromhof
2154
April 14th, 2002

The Principal
Ann Latsky School of Nursing
Private Bag 40
Auckland Park
2006

Dear Mrs Harms,

I am currently doing research for a Master's degree through the Department of Nursing Education at Wits University. The topic that I have chosen is “An Assessment of the Genetic Knowledge of Fourth-Year Diploma students”.

I would like to request permission to administer a paper-based questionnaire to the fourth- year Diploma nursing students during mid 2002. The questionnaire should be answered in the classrooms at a time that is suitable for you, and should take approximately 45 minutes to complete.

I have attached a copy of my research proposal and outline for your perusal. Permission has been granted from the Head of Nursing Education at the Bank of Lisbon, and should you agree, could we arrange a suitable date and time to administer the questionnaire.

I hope that you will find my request acceptable and acknowledge that the research I am planning is valid and relevant. On completion of the study, a copy of the findings will be available for you, should you so request.

Thanking you in anticipation,
Yours faithfully,

Merlyn Glass(RN, RM, Dip Paeds)

# Appendix 3

# Ethical Clearance Certificate

**UNIVERSITY OF THE WITWATERSRAND, JOHANNESBURG**

Division of the Deputy Registrar (Research)

**COMMITTEE FOR RESEARCH ON HUMAN SUBJECTS (MEDICAL)**
Ref: R14/49 Glass

**CLEARANCE CERTIFICATE** **PROTOCOL NUMBER** M02-05-10

**PROJECT** As Assessment of The Genetic Knowledge of Fourth Year Diploma Nursing Students

**INVESTIGATORS** Ms M Glass

**DEPARTMENT** School of Therapeutic Sci, Johannesburg Hospital

**DATE CONSIDERED** 02-05-31

**DECISION OF THE COMMITTEE** *

Approved unconditionally

**DATE** 02-07-18 **CHAIRMAN** .....(Professor P E Cleaton-Jones)

* Guidelines for written "informed consent" attached where applicable.

c c Supervisor: Mrs G Langley
Dept of School of Therapeutic Sci, Johannesburg Hospital

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==========

**DECLARATION OF INVESTIGATOR(S)**

To be completed in duplicate and **ONE COPY** returned to the Secretary at Room 10001, 10th Floor, Senate House, University.

I/we fully understand the conditions under which I am/we are authorized to carry out the abovementioned research and I/we guarantee to ensure compliance with these conditions. Should any departure to be contemplated from the research procedure as approved I/we undertake to resubmit the protocol to the Committee.

DATE 5/8/2002 SIGNATURE .....

**PROTOCOL NO.:** M 02-05-10

PLEASE QUOTE THE PROTOCOL NUMBER IN ALL ENQUIRIES

# Appendix 4

# SANC reg 425 of 1992 as amended 1994

Die Suid-Afrikaanse Raad op Verpleging

(ingestel ingevolge die Wet op Verpleging, 1978)

The South African Nursing Council

(Established under the Nursing Act, 1978)

# MINIMUM REQUIREMENTS FOR THE EDUCATION AND GUIDE CONCERNING THE TEACHING OF STUDENTS IN THE PROGRAMME LEADING TO REGISTRATION AS A NURSE (GENERAL, PSYCHIATRIC AND COMMUNITY) AND MIDWIFE

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# MINIMUM VEREISTES VIR DIE OPLEIDING EN GIDS BETREFFENDE DIE ONDERRIG VAN STUDENTE IN DIE PROGRAM WAT LEI TOT REGISTRASIE AS 'N VERPLEEGKUNDIGE (ALGEMENE, PSIGIATRIESE EN GEMEENSKAPS-) EN VROEDVROU

Regulations/Regulasies
R. 425

Date/Datum
22/02/1985

1992 Guide/Gids
1994 Amended/Gewysig

To achieve this, this the student also needs subject content in respect of

- the health of the mother, the course of normal pregnancy, labour, and puerperium, and the development and health of the foetus, neonate and infant, including
  * anatomical and physiological changes
  * diagnosis
  * management
  * early identification of mother and child (foetus, neonate and infant) at risk
  * psychosocial aspects
- abnormalities/complications in respect of of pregnancy, labour, puerperium, and the foetus, neonate and infant, including the
  * aetiology
  * pathology
  * clinical presentation
  * diagnosis
  * prevention
  * management

  of such abnormalities/complications.

## 1.6 BIOLOGICAL SCIENCES -

### 1.6.1 Anatomy, Physiology, Chemistry, Biophysics, Microbiology and Parasitology

An overview enabling the student to understand and to apply the following:

- the structural composition of the body
- the anatomical relations and structures
- the functions of the body as a whole
- the relevant
  . chemistry and biophysics
  . microbiology and parasitology

Practising within the legal scope of midwifery practice, the student shall, in the course of such allocation and with suitable accompaniment -

- apply the components fundamental to a scientific approach in -

  * carrying out the comprehensive ante-natal assessment and care of at least 30 pregnant women and recording all relevant information;

  * carrying out pelvic assessments cases of primigravida's or at the onset of labour where pelvi disproportion is suspected, the findings of which shall be checked by a registered midwife or medical practitioner;

  * recognizing the different stages of labour;

  * critically observing, monitoring and interpreting th findings in the course of all stages of labour, and providing appropriate nursing, including the provision of pain relief, in order to ensure the safety of mother and child throughout;

    The findings shall be checked by a registered midwife of a medical practitioner. The use of simulation to practise the skill is permissible, but should not be applied exclusively;

    Opportunities should be created for students to experience continuity of care throughout all stages of labour.

  * delivering at least 15 patients and conducting the 2nd and 3rd stages of labour of at least 5 patients either in the course of carrying out the deliveries indicated above, or conducting deliveries for other students;

  * mastering the skills needed for the the cutting of a episiotomy to prevent a severe tear of the perineum or complication relating to the child, provided the head is distending the perineum;

  * mastering the skills needed for administering local anaesthetic, excluding pudendal block and epidural anaesthesia, and the suturing of first and second degree tears and episiotomies;

# Appendix 5

# Information Sheet

## Information sheet

Dear Colleague,

My name is Merlyn Glass and I am currently doing a Master's Degree in Nursing Education at the University of the Witwatersrand. I am conducting a study, for research purposes, to determine whether nurses in their fourth year of study have obtained adequate and relevant genetic knowledge in their basic course, and are able to apply this in their patient care. I would also like to establish whether fourth-year student nurses have any knowledge of the rapid advances that are taking place in the field of genetics.

Based on the outcome of this study, an intervention in the form of information sheets, in-service training and recommendations for the curricula will be initiated if indicated.

If you participate in this study, you will be required to answer a questionnaire, which will take approximately 45 minutes to complete. You are not obliged to answer all questions should you not choose to do so. Participation in this study is entirely voluntary. Should you choose to participate in the study, you will remain anonymous, as no names will appear on the questionnaire, which will be kept strictly confidential. On completion of the questionnaire you will be required to place it in the provided, unmarked envelope which you will seal yourself. You will not be penalised for your answers, nor will the results of the study have any influence on your course or you examination or test results. You also have the right to withdraw from the study at any time with no detrimental effect whatsoever.

The appropriate committees of the University of the Witwatersrand as well as the head of the Gauteng Nursing Department and principals of the nursing schools have granted approval for this study.

Should you have any queries regarding this study or about your participation in the study, I can be contacted on the following numbers:

082 853 7113 or 011 792 0122

Thank you for your co-operation,

Merlyn Glass. RN, RM, Dip Paeds

# Appendix 6

# Questionnaire

**Genetic Questionnaire**

Please answer by ticking the correct answer/s or in written form where requested. **IN SOME OF THE QUESTIONS, MORE THAN ONE ANSWER MAY BE CORRECT**. This questionnaire is for research purposes only, and the results will not have any effect on your marks for your course. You will remain anonymous.

This section covers questions regarding genetic facts.

**Chromosomes and chromosomal anomalies**

1. How many chromosomes are there in a normal human cell?

☐ 23
☐ 48
☐ 46
☐ 47
☐ Don't know

2. How many chromosomes in a normal sperm or egg cell?

☐ 22
☐ 23
☐ 24
☐ 26
☐ Don't know

3. The cell division that takes place for the production of sperm and egg cells is:

☐ meiosis
☐ mitosis
☐ DNA duplication
☐ None of the above
☐ Don't know

4. If a baby is diagnosed as having Down syndrome, the chromosome result will show three copies of:

☐ chromosome 21
☐ chromosome 18
☐ chromosome 13
☐ chromosome 22
☐ Don't know

5. A child with Down syndrome may present with the following:

☐ cleft lip and palate
☐ epicanthic folds
☐ polydactyly
☐ mental retardation
☐ cardiac defect

6. The frequency of liveborn babies with Down syndrome in South Africa

☐ Is approximately 1 in 700
☐ Is greater in the white population
☐ Is lower in South Africa than in First world countries
☐ Is higher in women of advanced maternal age
☐ Don't know

**<u>This section deals with single gene abnormalities</u>**

7. How many autosomal chromosomes are there:

☐ 48
☐ 46
☐ 44
☐ Not sure

8. A person with an autosomal dominant condition will have a risk of passing on the gene to his child of:

☐ 25%
☐ 50%
☐ 100%
☐ Don't know

9. In autosomal dominant conditions:

Males and females are affected equally

☐ yes
☐ no

10. If a couple both carry an autosomal recessive gene, their risks of having a child with this disorder would be:

☐ 0%
☐ 25%
☐ 50%
☐ 100%
☐ Unsure of the correct answer

11. An X-linked recessive disorder affects mainly

☐ males
☐ females
☐ Neither
☐ Unsure

12. Which of the following are X-linked disorders

☐ haemophilia
☐ duchenne muscular dystrophy
☐ achondroplasia
☐ cystic fibrosis
☐ none of the above

**<u>This section deals with other types of conditions</u>**

13. Multifactorial disorders are

☐ Single gene abnormalities
☐ Caused by abnormal genes only
☐ Caused by environmental factors only
☐ Caused by both genetic and environmental factors
☐ Unsure of the correct answers

14. Multifactorial conditions include

☐ Diabetes type II
☐ Albinism
☐ Cleft lip and palate
☐ Neural tube defects
☐ Unsure of the answers

15. Teratogenesis is

☐ The study of abnormalities
☐ The effect of environmental agents on the developing foetus
☐ An abnormal growth
☐ Tumour development
☐ None of the above

16. Which of the following infectious agents poses a risk to the foetus

☐ syphilis
☐ rubella
☐ cytomegalovirus
☐ toxoplasmosis
☐ unsure

17. Which of the following drugs are considered risk factors for the foetus and are contraindicated in pregnancy?

☐ Warfarin
☐ Valproic acid
☐ Retinoic acid
☐ Panado
☐ don't know

18. Foetal alcohol syndrome is caused by the consumption of the following during a pregnancy

☐ Beer
☐ Wine
☐ Spirits
☐ None of the above
☐ All of the above

19. Children with Foetal Alcohol syndrome(FAS) may have the following features:

☐ Short palpebral fissures
☐ Long smooth philtrum
☐ Developmental delay
☐ microcephaly
☐ none of the above

**Genetic Epidemiology**

20. The carrier frequency of albinism in the black population in South Africa is approximately:

☐ 1 in 50(common)
☐ 1 in 30 (very common)
☐ 1 in 500 (uncommon)
☐ 1 in 1000 (very uncommon)
☐ Don't know

21. The carrier frequency of cystic fibrosis in the South African white population is approximately:

☐ 1 in 1000(very uncommon)
☐ 1 in 500 (uncommon)
☐ 1 in 50 (common)
☐ 1 in 20 (very common)
☐ Don't know

22. Testing for Porphyria is recommended in which population group

☐ Afrikaans-speaking individuals
☐ Asian individuals
☐ Individuals from Greece and Cyprus
☐ Ashkenazi Jewish individuals
☐ Uncertain of the correct answer

23. A couple of Indian origin asks you whether they should be tested for any genetic conditions before planning a pregnancy. Which of the following tests would you advise?

☐ thalassaemia
☐ albinism
☐ porphyria
☐ tay sachs
☐ unsure

24. A French- speaking Black couple from Central Africa request testing for genetic conditions. Which would you offer them?

- ☐ cystic fibrosis
- ☐ Haemophilia
- ☐ Duchenne muscular dystrophy
- ☐ Sickle cell anaemia
- ☐ Unsure of the correct answer

25. The percentage of Major birth defects in South African live born babies is:

- ☐ Below 1%
- ☐ Between 3% and 5%
- ☐ Between 10% and 15%
- ☐ Between 15% and 20%
- ☐ Uncertain of the correct answer

**Prenatal diagnosis**

26. The risk of having a child with a chromosomal abnormality increases:

- ☐ Where there is a previous child with a chromosomal abnormality
- ☐ If mother is over the age of 35
- ☐ If the parent has major exposure to radiation
- ☐ All of the above
- ☐ None of the above

27. A 22-year-old woman requests prenatal testing and wants the earliest test possible. She has a family history of a genetic disorder where DNA testing is possible. Which test would you offer her?

- ☐ Maternal serum triple screen
- ☐ Amniocentesis
- ☐ chorionic villus sampling
- ☐ High level ultrasound
- ☐ Uncertain of the correct answer

28. Amniocentesis:

- ☐ Is best performed at 12 weeks
- ☐ Has a low miscarriage risk of less than 1%
- ☐ Allows the examination of chromosomes only
- ☐ Can be used for biochemical and DNA studies
- ☐ Unsure of the correct answers

29. Increased maternal serum alpha-fetoprotein (AFP) is associated with:

- ☐ Neural tube defects
- ☐ Multiple pregnancies
- ☐ Chromosomal anomalies
- ☐ Albinism
- ☐ Unsure

30. A positive maternal serum triple screen (AFP, hCG, E3) may indicate a risk for:

- ☐ A baby with Down syndrome
- ☐ A baby with albinism
- ☐ A normal baby
- ☐ A baby with cystic fibrosis
- ☐ Unsure of the correct answer

31. Folic acid taken in the pregnancy will reduce the risks of:

- ☐ cleft lip and palate
- ☐ neural tube defects
- ☐ increased amniotic fluid
- ☐ infections in the baby
- ☐ unsure of the answers

**Genetic counselling**

Please tick where necessary and comment fully where indicated.

32. Have you heard of genetic counselling? ☐ Yes ☐ No

33. How did you hear of it?

☐ Colleagues
☐ At college
☐ Through the media
☐ Through magazines/journals
☐ Other

34. Please tick three factors that you feel describe genetic counselling

☐ Explaining the inheritance pattern of a specific condition
☐ Avoiding the truth so as not to alarm the family
☐ Psychosocial support
☐ Advising that they shouldn't have any further children because there is a risk
☐ None of the above

35. Do you feel that genetic counselling be a part of comprehensive health care?

☐ yes
☐ no

36. Do you think patients with hereditary disorders would benefit from genetic counselling?

☐ yes
☐ no

37. Do you know where the nearest genetic counselling clinic is in your area?

☐ Yes ☐ No

38. Do you feel confident to discuss **genetic risks** with your patients?
Please justify your answer.
☐ yes ☐ no

_____

_____

39. Do you feel confident to discuss **genetic test results** with your patients.
Please justify your answer.

☐ yes ☐ no

_____
_____
_____

**New Genetics**

40.Have you heard about the Human Genome project?

☐ Yes ☐ No

41. If yes, what do you understand it to be?

_____
_____
_____

42. If yes, where did you obtain the information?

☐ Newspapers
☐ Magazines
☐ Television
☐ During your course
☐ From colleagues

43. Do you feel that the genetics that you have studied equips you with relevant knowledge to help your patients confidently?

☐ Yes ☐ No

44. What additional information do you think nurses should receive during their training to be able to offer a comprehensive genetic service to their patients?

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This questionnaire was used with kind permission from Dr Amanda Krause and Ms Pat Craig and was adapted from a similar questionnaire used by a student doing a Master's degree in Genetic Counselling.

# Appendix 7

## Primary Health Care Package, Department of Health, Standards and Norms

**The Primary Health Care Package for South Africa – a set of norms and standards**

Part 1 Norms and standards for health clinics
Part 2 Norms and standards for community based clinic initiated services

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Department of Health
Pretoria
March 2000

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## ACKNOWLEDGEMENTS

### STAKEHOLDERS

- The production of this document is the culmination of a task that has involved many people in a great deal of work and effort. It benefited greatly from ideas, inputs and critical review from a broad range of participants from National Department of Health, Provincial Health Departments, other Governments Departments (Correctional Services, SAHMS ), Non Governmental Organisations, Universities, Private Hospitals, Professional bodies, Labour organisations and the South African Local Government Association (SALGA)

**The National Department of Health thanks all these contributors.**

### OTHER SUPPORT

- To individuals not part of either the stakeholder group nor Technical Team Task Team but who were requested to critique and advise on specific chapters related to their areas of specialty (both public and private), **our sincere thanks for their for co-operation and assistance.**

### TECHNICAL TASK TEAM

- A special tribute is paid to the Technical Task Team Members who contributed their time and experience to produce this document. Without their continuous contribution and tireless hard work for the Department of Health, equity would have remained beyond our reach. **We applaud them for their efforts.**

### TECHNICAL TASK TEAM MEMBERS

- Dr John Bennett — MSH Equity Project
- Dr Gary Morris — Child Survival Project, Bergville Kwa Zulu Natal
- Dr Abdul G. Elgoni — Centre for Health Systems Research and Development, University of Free State
- Dr Cameron Bowie — UK DFID consultant for Department of Health

### QUALITY ASSURANCE TECHNICAL AND SUPPORT STAFF

The project was co-ordinated for the National Department of Health by The Directorate: Quality Assurance (QA) located within the Cluster: Health Information, Evaluation and Research (HIER).

**Thanks to:**

- Mrs. Nthari Matsau — Chief Director HIER
- Dr Louis Claassens — Director QA
- Mrs Myra Tshabalala — Deputy Director QA
- Ms Assy Moraka — Assistant Director QA and project co-ordinator.

**QA SUPPORT STAFF**

Sincere thanks to the following staff members :

- Mrs Cora De Groot,
- Mrs Caroline Mangwane, and
- Mr Abram Shakoane for all the secretarial and administrative assistance throughout the project.

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## CONTENTS

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**The Primary Health Care Package for South Africa -- a set of norms and standards**

## INTRODUCTION

Primary health care is at the heart of the plans to transform the health services in South Africa. An integrated package of essential primary health care services available to the entire population will provide the solid foundations of a single, unified health system. It will be the driving force in promoting equity in health care. This document sets out the norms and standards that are to be made available in the essential package of primary care services. For the first time it will be possible for individuals to see what quality of primary care services they can expect to receive. It also acts as guidance for provincial and district health authorities to provide these services.

This introduction describes the background to the work, the way the package and standards have been produced, their potential uses and how they are likely to evolve with time and experience.

## THE BACKGROUND

The draft Health Bill requires the production of norms and standards to be used by provinces to provide health services at acceptable levels. Providing acceptable levels of service to all people will help the process of redistribution and reduce inequalities. The Year 2000 targets included the objective of having "*defined comprehensive services which are to be delivered at primary care level of health service delivery"*. The task to define and produce norms and standards falls to the Directorate: Quality Assurance, Department of Health.

A primary health care package was defined following detailed consultation over four years with national experts and provincial staff. It forms the basis of this document, which contains norms and standards for clinic and community services. A national task team has undertaken the production of the norms and standards. Norms and standards for community health centres and Level 1 hospitals will follow.

## THE CHOICE OF NORMS AND STANDARDS

All necessary components of a comprehensive primary care package are described and norms and standards for each component are provided. The norms and standards are largely derived from existing national policy documents or, if unavailable, other authoritative sources such as WHO and research work undertaken in the country. All the norms and standards are verifiable (some more easily than others) by staff providing the service. Some of the norms were taken from the Year 2000 Objectives and Indicators. An attempt has been made to ensure that the standards are practical, essential and comprehensive and describe the range of services that should be available to all South Africans.

## POTENTIAL USES

It is hoped that the norms and standards are comprehensive enough to be used: --

- By local staff to help assess their own performance and that of their clinic.
- By the community who are able to see the range and quality of services to which they are entitled.
- As planning guidelines by district and provincial health planners to help assess the unmet needs of their population and draw up plans to bring services up to national standards.
- By provincial governments to guide resource allocation.

This wide range of uses requires the document to be available in different formats and selecting particular sections. Once this core document is published, it will be widely distributed to all stakeholders. Components can for example be adapted for use as checklists for local staff.

## A LIVING DOCUMENT

The document has two parts – one on clinic services, the other on community services. The community health centre and level-1 hospital sections are given a separate document. The

choice of separate documents follows the precedent set by the EDL and permits each document to remain of reasonable size.

Not every primary health care component has been fully documented. National policies will change and service standards will be able to be enhanced, as more resources are made available. The document is the first of its kind. The task group believes that, with experience of its use, many things will be found that can be improved. Feedback from patients and staff is essential. Some provinces have set up norms and standards initiatives themselves. This is good as the more experience that is gained with their use the more can be shared.

## DEFINITION OF NORMS AND STANDARDS

## FOR THE PURPOSE OF THIS DOCUMENT NORMS AND STANDARDS ARE DEFINED THUS:

**A NORM** is defined as ***a statistical normative rate of provision or measurable target outcome over a specified period of time.***

**A STANDARD** is defined as ***a statement about a desired and acceptable level of health care.***

A common framework used to develop these standards addresses health service inputs, processes, outputs and outcomes. This approach has been adopted. Standards are best developed in incremental stages and according to national priorities. These represent the first stage of this process for primary health care.

Standard setting takes place within specific dimensions of quality -- acceptability, accessibility, appropriateness, continuity, effectiveness, efficiency, equity, interpersonal relations, technical competence and safety. The most important dimensions have been chosen for each service.

## INTERPRETATION

Two important issues need to be taken into account when interpreting these norms and standards in the local setting. The first relates to the role of national and provincial health authorities. The second relates to staff competency.

## WHAT SERVICES ARE REQUIRED NOT HOW SERVICES ARE PROVIDED

The national task is to define **what** services are required to best meet the health needs of the nation. It is for provinces and local government to decide, in the light of local circumstances, **how** these services are to be provided. Because of these different roles this national document is about **what** services at **what** standard are required. The standards do not specify **how** the services are to be provided and at what level the standards will be met. It is for provinces and Local Gto harden up the standards with verifiable time limited measures based on existing performance and anticipated improvements.

Different kinds of facilities will be required to provide the same services in different situations. Take for instance the use of mobile clinics in remote rural areas compared to polyclinics in high-density urban areas. For this reason national standards about facilities and staffing norms are not offered. In some instances some standards about special facilities are

included without which a service would be impossible to provide, for example a confidential room to talk to a sexually abused patient.

**STAFF COMPETENCY**

Many standards are about staff competency. It is to be expected that some staff will not be trained, or if trained, remain competent to provide all the services specified. It is the responsibility of professional staff to seek to rectify the deficit in themselves and their staff by arranging appropriate training. It goes without saying that no members of staff should undertake tasks unless they are competent to do so. The safety of the patient is paramount.

**CONTENT**

The document is arranged in a logical order. There are two parts; the first deals with health clinics and the second section with community based services. The part on health clinics starts with a chapter on patient rights, which is followed by one on core norms and standards for all clinics whatever services they are providing. For instance all clinics are expected to have and use the Essential Drug List. The standard is therefore included as a core standard. It is not repeated in later chapters although its use is essential for most if not all services. Chapters succeeding the core standards one do not duplicate core standards.

Then follows chapters on individual services in life cycle order starting with maternity care and women's health through children and adolescent services to communicable diseases and finally non-communicable diseases.

Each chapter has three paragraphs. The first describes the service to be provided and is taken from the document "The Primary Health Care Package. The second paragraph describes the norms, chosen to represent key measures of what is required. All clinics should be aspiring to measure and reach these norms. The third paragraph describes the standards for each service and it is divided into 9 sections. The first three sections describe the essential written material, equipment, supplies and medicines required. Successful performance to meet these standards requires good organisation and logistics.

**Sections 4 and 5** are perhaps the most important of all in describing the required competence of staff, without which services will be of poor quality. These sections will be of help to individual professionals as they assess their own capabilities against what is required of them. They will also be of help to managers and training departments in offering a backbone for training curricula and supervisory support.

**Sections 6 – 9** relate to other professional tasks required but which are not directly related to individual patient care. They are nevertheless important, as they are to do with improving the health of the local community.

**Part 2** is about community based clinic initiated services. The format is similar.

Documentary sources are listed at the back, which together with the documents listed in sections 1 of each chapter, reference the authoritative evidence on which the norms and standards are based.

***Your comments and feedback***

## PART 1

## NORMS AND STANDARDS FOR HEALTH CLINICS

## BATHO PELE -- PEOPLE FIRST

**INTRODUCTION**

Access to decent public services is the rightful expectation of all citizens especially those previously disadvantaged. Communities are encouraged to participate in planning services to improve and optimize service delivery for the benefit of the people who come first.

**STANDARDS**

`

All communities will know from displayed posters about the eight principles of Batho Pele, which are:

CONSULTATION
Communities will be consulted about the level and quality of public services they receive and where possible will be given a choice about the services offered.

SERVICE STANDARDS
Citizens would know the level and quality of public service they are to receive and know what to expect

ACCESS
All citizens have equal access to the services to which they are entitled

COURTESY
Citizens should be treated with courtesy and consideration.

INFORMATION
Citizens should be given full accurate information about the public service they are entitled to receive.

OPENNESS and TRANSPARENCY
Citizens should be told how national and provisional departments are run, how much they cost and who is in charge.

REDRESS
If the promised standard of service is not delivered they should be offered an apology, an explanation and an effective remedy, when complaints are made, citizens should receive a sympathetic positive response.

VALUE FOR MONEY
Public services should be provided economically and efficiently in order to give citizens and communities the best possible value for money.

**Implications for health staff**

In line with these principles the local health services for a community will provide:

- services with a high standard of professional ethics
- a missions statement for service delivery
- services which are measured with performance indicators displayed, so community can understand the level of achievement
- services which are in partnership with or complement other sectors e.g. the private sector and non-government organizations and community based organizations
- services which are customer friendly and confidential
- opportunities for community consultation
- types of outreach which can reach to all communities and to families in greatest need
- easily accessible and effective ways of dealing with complaints or suggestions for improvement
- current information on services available and hours of service, staff changes of movements and extra activities such as health days.

**PATIENTS RIGHTS CHARTER**

## SERVICE DESCRIPTION

The purpose and expected outcome of the patients rights charter and complaints procedure is to deal effectively with complaints and rectify service delivery problems and so improve the quality of care, raise awareness of rights and responsibilities, raise expectations and empowerment of users, change attitudes by strengthening the relationship between providers and users, improve the use of services and develop a mechanism for enforcing and measuring the quality of health services.

### STANDARDS

`

1. Each clinic displays the patients rights charter and patient responsibilities at the entrance in local languages.
2. The twelve patient's rights are observed and implemented. Every patient has the right to:

- a healthy and safe environment
- access to health care
- confidentiality and privacy
- informed consent

- be referred for a second opinion
- exercise choice in health care
- continuity of care
- participation in decision making that affect his/her health
- be treated by a named health care provider
- refuse treatment and
- knowledge of their health insurance/medical aid scheme policies
- complain about the health service they receive.

3. The ten patient's responsibilities are displayed alongside the patients rights charter. These include:

- Living a healthy lifestyle
- Care and protect the environment
- Respect the rights of other patients and health staff
- Utilise the health system optimally without abuse
- Know the health services available locally and what they offer
- Provide health staff with accurate information for diagnosis, treatment, counselling and rehabilitation purposes
- Advise health staff on his or her wishes with regard to death
- Comply with the prescribed treatment and rehabilitation procedures
- Ask about management costs and arrange for payment
- Take care of the patient carried health cards and records.

4. There is provision for the special needs of people such as a woman in labour, a blind person or a person in pain.
5. Services are provided with courtesy, kindness, empathy, tolerance and dignity.
6. Information about a patient is confidential and is only disclosed after informed and appropriate consent.
7. Informed consent for clinical procedures is based on a patient being fully informed of the state of the illness, the diagnostic procedures, the treatment and its side effects, the possible costs and how lifestyle might be affected. If a patient is unable to give informed consent the family is consulted.

8. When there is a problem the health care user is informed verbally of the health rights charter with emphasis on the right to complain and the complaints procedure is explained and handed over.
9. The clinic has a formal, clear, structured complaint procedure and illiterate patients and those with disabilities are assisted in laying complaints.
10. All complaints or suggestions are forwarded to the appropriate authority if they cannot be dealt with in the clinic.
11. A register of complaints and how they were addressed is maintained.
12. The name, address, telephone number of the person in charge of the clinic is displayed.

## CORE NORMS AND STANDARDS FOR HEALTH CLINICS

**CORE NORMS**

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1. The clinic renders comprehensive integrated PHC services using a one-stop approach for at least 8 hours a day, five days a week.
2. Access, as measured by the proportion of people living within 5km of a clinic, is improved.
3. The clinic receives a supportive monitoring visit at least once a month to support personnel, monitor the quality of service and identify needs and priorities.
4. The clinic has at least one member of staff who has completed a recognised PHC course.
5. Doctors and other specialised professionals are accessible for consultation, support and referral and provide periodic visits.
6. Clinic managers receive training in facilitation skills and primary health care management.
7. There is an annual evaluation of the provision of the PHC services to reduce the gap between needs and service provision using a situation analysis of the community's health needs and the regular health information data collected at the clinic.
8. There is annual plan based on this evaluation.
9. The clinic has a mechanism for monitoring services and quality assurance and at least one annual service audit.
10. Community perception of services is tested at least twice a year through patient interviews or anonymous patient questionnaires.

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**CORE STANDARDS**

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### 1. References, prints and educational materials

1.1 Standard treatment guidelines and the essential drug list (EDL) manual.
1.2 A library of useful health, medical and nursing reference books kept up to date.
1.3 All relevant national and provincial health related circulars, policy documents, acts and protocols that impact on service delivery.
1.4 Copies of the Patients Charter and Batho Pele documents available.
1.5 Supplies of appropriate health learning materials in local languages.

### 2. Equipment

2.1 A diagnostic set.
2.2 A blood pressure machines with appropriate cuffs and stethoscope.
2.3 Scales for adults and young children and measuring tapes for height and circumference.
2.4 Haemoglobinometer, glucometer, pregnancy test, and urine test strips.
2.5 Speculums of different sizes
2.6 A reliable means of communication (two-way radio or telephone).
2.7 Emergency transport available reliably when needed.
2.8 An oxygen cylinder and mask of various sizes.
2.9 Two working refrigerators one for vaccines with a thermometer and another for medicines. If one is a gas fridge a spare cylinder is always available.
2.10 Condom dispensers are placed where condoms can be obtained with ease.
2.11 A sharps disposal system and sterilisation system.
2.12 Equipment and containers for taking blood and other samples.
2.13 Adequate number of toilets for staff and users in working order and accessible to wheelchairs.
2.14 A sluice room and a suitable storeroom or cupboard for cleaning solutions, linen and gardening tools.
2.15 Suitable dressing/procedure room with washable surfaces.
2.16 A space with a table and ORT equipment and needs
2.17 Adequate number of consulting rooms with wash basins, diagnostic light (one for each professional nurse and medical officer working on the same shift).

### 3. Medicines and Supplies

3.1 Suitable medicine room and medicine cupboards that are kept locked with burglar bars.
3.2 Medicines and Supplies as per the essential drug list for Primary Health Care, with a mechanism in place for stock control and ordering of stock.
3.3 Medicines and Supplies always in stock, with a mechanism for obtaining emergency supplies when needed.
3.4 A battery and spare globes for auroscopes and other equipment.
3.5 Available electricity, cold and warm water.

### 4. Competence of Health Staff

**Organising the clinic**

4.1 Staff are able to

4.1.1 map the clinic catchment area and draw specific and achievable PHC objectives set using district, national and provincial goals and objectives as a framework.
4.1.2 Organise outreach services for the clinic catchment area.
4.1.3 Organise the clinic to reduce waiting times to a minimum and initiate an appointment system when necessary.
4.1.4 Train community health care promoters to educate caretakers and facilitate community action.
4.1.5 Plan and implement a district focused and community based activities, where health workers are familiar with their catchment area population profile, health problems and needs and use data collected at clinic level for this purpose.

**Caring for patients**

4.2 Staff are able to follow the disease management protocols and standard treatment guidelines, and provide compassionate counselling that is sensitive to culture and the social circumstances of patients.
4.3 Staff are positive in their approach to patients, evaluating their needs, correcting misinformation and giving each patient a feeling of always being welcome.
4.4 Patients are treated with courtesy in a client-oriented manner to reduce the emotional barriers to access of health facilities and prevent the breakdown in communication between patients and staff.
4.5 The rights of patients are observed.

**Running the clinic**

4.6 A clear system for referrals and feedback on referrals is in place.
4.7 All personnel wear uniforms and insignia in accordance with the South African Professional Councils' specifications.
4.8 The clinic has a strong link with the community, civic organisations, schools and workplaces in the catchment area.
4.9 The clinic is clean, organised and convenient and accommodates the needs of patients' confidentiality and easy access for older persons and people with disability.
4.10 Every clinic has a house keeping system to ensure regular removal and safe disposal of medical waste, dirt and refuse.
4.11 Every clinic provides comprehensive security services to protect property and ensure safety of all people at all times.
4.12 The clinic has a supply of electricity, running water and proper sanitation.
4.13 The clinic has a written infection control policy, which is followed and monitored, on protective clothing, handling of sharps, incineration, cleaning, hand hygiene, wound care, patient isolation and infection control data.

**5. Patient Education**

5.1 Staff are able to approach the health problems of the catchment area hand in hand with the

clinic health committee and community civic organisations to identify needs, maintain surveillance of cases, reduce common risk factors and give appropriate education to improve health awareness.
5.2 Culturally and linguistically appropriate patients' educational pamphlets are available on different health issues for free distribution.
5.3 Appropriate educational posters are posted on the wall for information and education of patients.
5.4 Educational videos in those clinics with audio-visual equipment are on show while patients are waiting for services.

### 6. Records

6.1 The clinic utilises an integrated standard health information system that enables and assists in collecting and using data.
6.2 The clinic has daily service registers, road to health charts, patient treatment cards, notification forms, and all needed laboratory request and transfer forms.
6.3 All information on cases seen and discharged or referred is correctly recorded on the registers.
6.4 All notifiable medical conditions are reported according to protocol.
6.5 All registers and monthly reports are kept up to date.
6.6 The clinic has a patient carry card or filing system that allows continuity of health care.

### 7. Community and Home Based Activity

7.1 There is a functioning community health committee in the clinic catchment area.
7.2 The clinic has links with the community health committee, civic organisations, schools, workplaces, political leaders and ward councillors in the catchment area.
7.3 The clinic has sensitised, and receives support from, the community health committee.
7.4 Staff conduct regular home visits using a home visit checklist.

### 8. Referral

8.1 All patients are referred to the next level of care when their needs fall beyond the scope of clinic staff competence.
8.2 Patients with a need for additional health or social services are referred as appropriate.
8.3 Every clinic is able to arrange transport for an emergency within one hour.
8.4 Referrals within and outside the clinic are recorded appropriately in the registers.
8.5 Merits of referrals are assessed and discussed as part of the continuing education of the referring health professional to improve outcomes of referrals.

### 9. Collaboration

9.1 Clinic staff collaborate with social welfare for social assistance and with other health related public sectors as appropriate.
9.2 Clinic staff collaborate with health orientated civic organisations and workplaces in the catchment area to enhance the promotion of health.

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**CORE MANAGEMENT STANDARDS**

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### 10. Leadership and planning

10.1 Each clinic has a vision/mission statement developed and posted in the clinic.
10.2 Core values are developed by the clinic staff and posted.
10.3 An operational plan or business plan is written each year.

### 11. Staff

11.1 New clinic staff are oriented.
11.2 District personnel policies on recruitment, grievance and disciplinary procedures are available in the clinic for staff to refer to.
11.3 The staff establishment for all categories is known and vacancies discussed with the supervisor.
11.4 Job descriptions for each staff category are in the clinic file.
11.5 There is a performance plan/agreement and training plan made and a performance appraisal carried out for each member of staff each year.
11.6 The on-call roster and the clinic task list with appropriate rotation of tasks are posted.
11.7 An attendance register is in use.
11.8 There are regular staff meetings (at least once a month).
11.9 Services and tasks not carried out due to lack of skills are identified and new training sought.
11.10 In-service training takes place on a regular basis.
11.11 Disciplinary problems are documented and copied to supervisor.

### 12. Finance

12.1 The clinic, as a cost centre, has a budget divided into main categories.
12.2 The monthly expenditure of each main category is known.
12.3 Under and over spending is identified and dealt with including requests for the transfer of funds between line items where permitted and appropriate.

### 13. Transport and communication

13.1 A weekly or monthly transport plan is submitted to the supervisor or transport co-ordinator.
13.2 The telephone or radio is working.
13.3 The ambulance can be contacted for urgent patient transport to be available within two hours.

### 14. Visits to clinic by unit supervisor

14.1 There is a schedule of monthly visits stating date and time of supervisory support visits.
14.2 There is a written record kept of results of visits.

### 15. Community

15.1 The community is involved in helping with clinic facility needs.
15.2 The community health committee is in place and meets monthly.

**16. Facilities and equipment**

16.1 There is an up-to-date inventory of clinic equipment and a list of broken equipment.
16.2 There is a list of required repairs (doors, windows, water) and these have been discussed with the supervisor and clinic committee.

**17. Drugs and supplies**

17.1 Stocks are secure with stock cards used and up-to-date.
17.2 Orders are placed regularly and on time and checked when received against the order.
17.3 Stocks are kept orderly, with FEFO (first expiry, first out) followed and no expired stock.
17.4 The drugs ordered follow EDL principles.

**18. Information and documentation**

18.1 New patient cards and medico-legal forms are available.
18.2 The laboratory specimen register is kept updated and missing results are followed up.
18.3 Births and deaths are reported on time and on the correct form.
18.4 The monthly PHC statistics report is accurate, done on time and filed/sent.
18.5 Monthly and annual data are checked, graphed, displayed and discussed with staff and the health committee.
18.6 There is a catchment area map showing the important features, location of mobile clinic stops, DOTS supporters, CHWs and other outreach activities.

## WOMEN'S REPRODUCTIVE HEALTH

**SERVICE DESCRIPTION**

Reproductive services for women are provided in an integrated comprehensive manner covering preventive, promotive, curative and rehabilitative aspects of care. The focus is on antenatal, delivery, postnatal and family planning care.

**NORMS**

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1. Increase the percentage of pregnant women receiving antenatal care (ANC) from the existing level to at least 70%.
2. Increase the deliveries in institutions by trained birth attendants from the existing level to at least 75%.
3. Reduce the proportion of pre-term deliveries and low birth weight babies by at least 20%.

4. Reduce the proportion of births in women below 16 years and 16-18 years from the existing level (13.2% in 1998).

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**STANDARDS**

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### 1. References, prints and educational materials

1.1 Midwifery protocols
1.2 Contraception protocols
1.3 Termination of pregnancy protocols
1.4 Sterilisation act
1.5 All Provincial circulars and policy guidelines regarding women's health issues
1.6 A library of suitable references and learning material on women's health issues

### 2. Equipment and special facilities

2.1 Delivery set
2.2 Neonatal resuscitation trolley
2.3 Specula
2.4 Fetalscope
2.5 Women's Health charts

### 3. Medicines and Supplies

3.1 Ferrous and folic acid tablets
3.2 Oxytocin
3.3 Vit K injections
3.4 Contraceptive barrier methods e.g. condoms
3.5 Vaginal contraceptives e.g. spermicidal jelly
3.6 Intrauterine contraceptive devices
3.7 Injectable hormonal contraceptives
3.8 Oral hormonal contraceptives
3.9 Post-coital contraceptives

### 4. Competence of Health Staff

4.1 Nurses receive training in the perinatal education programme (PEP), contraception and post-abortion care management.
4.2 Staff are able to take a history and perform a physical examination and tests according to protocols and guidelines.
4.3 Staff provide routine management, observations and service according to the ANC protocol at each step of the pregnancy including at least three visits during pregnancy.
4.4 Staff provide education and counselling to each pregnant woman and partner on monitoring signs of problems (e.g. bleeding), nutrition, child feeding and weaning, STDs / HIV, delivery, newborn and child care, advanced maternal age, family planning and child

spacing.
4.5 Staff offer appropriate counselling, advice and service to pregnant women requesting termination of pregnancy.
4.6 At least one member of staff is able to:-

4.6.1 Deliver uncomplicated pregnancies.
4.6.2 Make routine observations according to the postnatal care protocol.
4.6.3 Make usual routine observations and select and prescribe appropriate family planning methods according to national protocol.
4.6.4 Screen, advice and refer infertility cases as per national guidelines.
4.6.5 Conduct breast cancer and cervical screening for women older than 35 years as per protocols.
4.6.6 Conduct home visits to provide support and supervise care.
4.6.7 Provide appropriate adolescent/youth services on family planning, sexuality, health education and counselling.

### 5. Patient Education

5.1 Information is given to mothers on booking for delivery, child preventive care, education about child feeding and the introduction of solid food.
5.2 Further information is given to mothers on the care of breasts, vaginal bleeding and scars, signs of hypertension, diabetes, anaemia, return to usual physical efforts, labour rights, rights of the child and advice on family planning.
5.3 Patients are given group education.
5.4 Patients' relatives and the community receive continuous, appropriate high quality information on the importance of antenatal care and institutional deliveries.
5.5 Information, education and counselling are offered to adolescents and youth.

### 6. Records

6.1 All information on cases and outcome of deliveries are correctly recorded on the register.
6.2 All registers and monthly reports are kept up to date.

### 7. Community and Home Based Activity

7.1 The clinic has sensitised, and receives support from, the community health committee about the positive encouragement of attendance at clinic of all pregnant women.
7.2 Staff conduct regular home visits using a home visit checklist.

### 8. Referral

8.1 All referrals within and outside the clinic are motivated and indications for referral written clearly on the referral form.
8.2 Patients with need for additional health or social services are referred according to protocols.
8.3 Referrals from traditional birth attendants (TBA) should be encouraged and associated with the training of the TBAs and follow up of the training.

### 9. Collaboration

9.1 Clinic staff collaborate with social welfare for social assistance and other role players.
9.2 Clinic staff collaborate with clinic health committee, the civic organisations and workplaces in the catchment area to enhance health promotion.

## MANAGEMENT AND PREVENTION OF GENETIC DISORDERS AND BIRTH DEFECTS

**SERVICE DESCRIPTION**

Genetic services are forming part of the integrated maternal, child and women's health care. It aims to assist individuals with a genetic disadvantage to live and reproduce as normally and responsibly as possible .The components include clinical diagnostic services, counseling, laboratory support, prevention strategies and public awareness campaigns in collaboration with NGOs, CBOs and other government sectors.

**NORMS**

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1. At least one clinic staff member trained to recognize, counsel, treat manage and refer most common conditions.
2. Clinic staff receive regular genetic training and update from the regional genetic cordinator.
3. Clinic staff receive support from visiting specialist, clinical geneticist and other academic experts.

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**STANDARDS**

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### 1. References, prints and educational materials

1. The clinic has the latest copy of the Human Genetics Guidelines for Management and Prevention of Genetic Disorders, Birth Defects and Disabilities.

### 2. Equipment

2.1 .

### 3. Medicines and Supplies

3.1 List of drugs in accordance with the Essential Drugs List

### 4. Competence of Health Staff

4.1 At least one clinic staff is able to recognize, counsel, treat, manage and refer most common genetic conditions

### 5. Referral

5.1 Referrals for further support as per guidelines

### 6. Patient Education

6.1 Provide posters, pamphlets and other educational materials on genetics for patients.
6.2 All patients and caretakers receive health education on genetic disorders, birth defects and disabilities.
6.3 Encourage women to procreate at the ideal reproductive age (25-35 years) to reduce the risk of chromosomal abnormalities.
6.4 Educate women to avoid exposure to teratogens during pregnancy e.g. alcohol, recreational drugs and certain chemical and infecting agents.

### 7. Records

7.1 Notification forms to notify genetic disorders and birth defects in the immediate post-natal period and later in life.

### 8. Community Based Services

8.1 Clinic staff to work with South African Inherited Disorders Association and other NGOs and CBOs to support affected individuals and families at community level.

### 9. Collaboration

9.1 Clinic staff collaborate with social workers, physiotherapists, speech therapists and other support staff to provide comprehensive care.
9.2 Clinic staff to work with South African Inherited Disorders Association, school teachers, and other NGOs and CBOs to provide information and raise awareness on genetic disorders, birth defects and disabilities.