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THE REPRODUCTIVE CHOICES MADE BY SOUTH AFRICAN MOTHERS WHO HAVE CHILDREN WITH DOWN SYNDROME

Julie Clare Lampret

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
Master of Science in Medicine

Johannesburg, 2006

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DECLARATION

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

_____

(Julie Lampret)

On this_____ day of_____, 2006

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DEDICATION

In loving memory of my aunt
Irene Broekmann
1954-2005

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PRESENTATIONS ARISING FROM STUDY

Oral Presentations:

- Lampret JC, Christianson AL. The Reproductive Choices made by South African Mothers who have Children with Down Syndrome. Eleventh biennial congress of the Southern Africa Society of Human Genetics held at Glenburn Lodge, Muldersdrift, 14-16 March 2005.

- Lampret JC, Christianson AL. The Reproductive Choices made by South African Mothers who have Children with Down Syndrome. Human Genetics Departmental Seminar held at the National Health Laboratory Service, Johannesburg, 8 September 2005.

- Lampret JC, Christianson AL. The Reproductive Choices made by South African Mothers who have Children with Down Syndrome. 2nd Southern African Conference on Down syndrome and Intellectual Disabilities held at Birchwood Conference Centre, Boksburg, 29 September-1 October 2005.

Poster Presentations:

- Lampret JC, Christianson AL. The Reproductive Choices made by South African Mothers who have Children with Down Syndrome. Eleventh biennial congress of the Southern Africa Society of Human Genetics held at Glenburn Lodge, Muldersdrift, 14-16 March 2005.

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ABSTRACT

Down syndrome is the commonest cause of congenital developmental disability in industrialized countries, where it occurs in approximately 1.4 per 1000 live births. In South Africa, the birth prevalence of Down syndrome was documented as 1.8 and 2.09 per 1000 live births in urban and rural populations, respectively. The physical, psychosocial and emotional burden of Down syndrome on affected families is significant.

The aim of this study was to determine the reproductive choices of women with a child with Down syndrome, aged 1 year or older. The survey was conducted using a structured questionnaire. The sample consisted of fifty women; 36 African, 4 Asian and 10 Caucasian. The questionnaire assessed the mothers' knowledge of Down syndrome prior to diagnosis, what counselling was received and how this knowledge was utilised. Information was also obtained on the mothers' use of family planning, the knowledge and use of prenatal medical genetic screening and diagnosis, and what decisions would be made in future pregnancies.

None of the sample group of mothers had prenatal diagnosis in their pregnancy with their Down syndrome child, but 76% (38) said that they would want prenatal diagnosis in any future pregnancies. Of the 50 mothers, 21 (42%) said they would terminate a pregnancy if Down syndrome had been detected, 26 (52%) said they would not, and 3 (6%) said they were unsure what they would have done if faced with this decision. Of the Caucasian women, 40% (4) said they would opt for termination of pregnancy, 40% (4) said they would not and 20% (2) were unsure. Of the African and Asian women, 52.8% (19) and 75% (3) respectively said they would not terminate an affected fetus.

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The information from this study can be used to improve the understanding of how women and their families cope with their children with Down syndrome and give insight for the provision of more effective and comprehensive genetic counselling.

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- My parents, Joe and Beth who have blessed me with every opportunity in life, always support me with every decision I make, and give endless love and encouragement. You mean the world to me, I love you and thank you.
- The Lord God, Whom without I am nothing and can do nothing

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NOMENCLATURE

- AMA = Advanced maternal age: Pregnant women over the age of 35 years
- Amniocentesis = Procedure to obtain a sample of amniotic fluid from the uterus
- Anaphase = The stage of cell division when the chromosomes leave the equatorial plate and move to opposite poles of the spindle
- Anaphase Lag = The loss of a chromosome as it migrates to the pole of the cell during anaphase leading to monosomy in the resulting cell
- Chromosomes = the structures within each cell nucleus containing the genetic information (DNA). They are transmitted from one generation to the next via gametes
- Cordocentesis = Procedure of obtaining fetal blood samples for prenatal diagnosis
- CVS = Chorionic villus sampling: Procedure to obtain chorionic villi from the chorionic membrane of the fetus
- Homolog = one of the chromosomes in a pair of chromosomes that contain identical loci in meiosis
- Meiosis = Cell division which occurs in the gamete formation with halving of the somatic number of chromosomes resulting in haploid (one set of chromosomes i.e. 23) gametes
- NT = Nuchal Translucency: Prenatal test involving measuring of the thickness of a fluid filled space behind the neck of the fetus
- PD = Prenatal diagnosis: Diagnostic testing during pregnancy (e.g. amniocentesis, CVS or cordocentesis) to confirm whether or not a fetus is affected with a particular disorder
- PCR = Polymerase chain reaction: Amplification of a particular piece of DNA

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- TOP = Selective termination of pregnancy
- Translocation = The transfer of genetic material from one chromosome to another
- Triple Test = A screening test done during the second trimester of pregnancy which gives a risk for having a fetus with Down's syndrome by measuring and computational analyzing of the levels of serum α-fetoprotein, oestriol and human chorionic gonadotrophin
- Trisomy = The presence of three copies of a chromosome

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CHAPTER 1

INTRODUCTION

During a pregnancy, parents prepare psychologically for the birth of a healthy normal child. With the birth of a child with a congenital disorder such as Down syndrome, their loss of the anticipated healthy child is immediately experienced. This creates a crisis for parents, the psychosocial effects of which are immense (Hobdell & Deatrick 1996). Amongst other issues that they will have to confront in the future will be whether or not to have more children.

Down syndrome was first described in 1866 by Langdon Down. In 1959, Lejeune, Gautier, and Turpin (1959) discovered that the cause of Down syndrome was the presence of an extra chromosome 21. Subsequently Down syndrome has become the most highly researched and well documented genetic condition. Over 6000 papers were published on Down syndrome before 1980 (Peuschel & Steinberg 1980) with more than 6000 publications since (Christianson 1996). Although there are over 12000 publications in the world literature on Down syndrome, there are less than 25 papers on Down syndrome in African populations (Christianson 1996).

Review of the literature revealed that few papers have dealt with the reproductive behaviour of mothers after the birth of a child with Down syndrome. Tips, Smith, Perkins, Bergman and Meyer (1963) reported from a study on 24 American women with previously high reproductive rates, that there was a sharp decline in their reproductive activity after the

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birth of a child with Down syndrome. An average of 49% of mothers had had pregnancies prior to the birth of their child with Down syndrome and this rate dropped to 25% subsequently. No correlation was seen in the control group. The reason given by the mothers was that their experiences of having a child with Down syndrome were very traumatic. In addition, amongst the aunts of the children with Down syndrome, 47% had had pregnancies prior to the birth of this child and this rate dropped subsequently to 9%. Ando (1978) found that reproductive activity decreased significantly in Japanese mothers who had children with Down syndrome, with the decline of reproduction observed more clearly amongst younger mothers. Elkins, Stovall, Wilroy and Dacus (1986) documented that 61 (60.4%) from a sample of 101 American women, who had a child with Down syndrome, had no further children. Currently there is only one study on the reproductive behaviour of African mothers who have had children with Down syndrome (Mgone 1982), and it showed that reproductive behaviour in those mothers remained unchanged after the birth of a child with Down syndrome.

1.1 DOWN SYNDROME

Chromosome abnormalities account for about 50% of all spontaneous miscarriages and 0.5-1% of live born babies (Mueller & Young 2005). Down syndrome is the most common autosomal chromosomal syndrome in live born infants and is the commonest cause of congenital developmental disability in industrialized countries (Baird & Sadovnick 1989).

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1.1.1 Causes of Down syndrome

Down syndrome is due to the presence of extra chromosome 21 material, which can be acquired by non-disjunction (trisomy), translocation or mosaicism.

1.1.1.1 Non-disjunction Down syndrome

Most Down syndrome, found in 94% of affected individuals, results from a free trisomy of chromosome 21 due to a non-disjunction event during meiosis (Jones 1997). Because of the increasing risk of meiotic error as a woman gets older, the occurrence of non-disjunction Down syndrome increases as the maternal age increases (Harper 2004). The birth prevalence of Down syndrome in infants of women under 25 years of age is considered to be low, less than 1 in 1000, although it rises slightly in very young mothers. The risk rises above the overall population risk at a maternal age of 30 years. At a maternal age of 40 years, the risk is approximately 1% and rises significantly thereafter (Harper 2004). Figure 1.1 shows the risk of Down syndrome increases in a gradual, linear fashion until about age 30 and increases exponentially thereafter.

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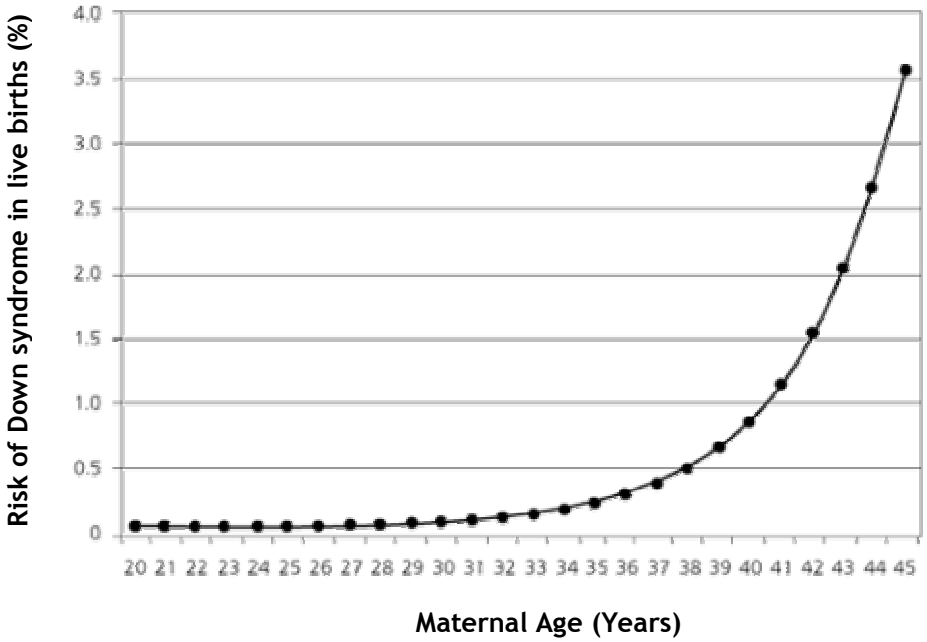

FIGURE 1.1: Estimated risk of Down syndrome according to maternal age (Newberger 2000)

Recurrence risks for non-disjunction Down syndrome are increased over the normal risk, but the increase does not show a simple relationship to maternal age (Harper 2004). Recurrence for Down syndrome is about 1% for women under 35 years of age. The recurrence risk for a mother over the age of 37 is calculated as the age related risk + 1% (Harper 2004).

1.1.1.2 Translocation Down syndrome

Another cause of Down syndrome is chromosomal translocation, which occurs in about 3.3% of individuals with Down syndrome (Jones 1997). The rearrangement of

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chromosomes in translocation Down syndrome usually involves the acrocentric chromosome 14, less commonly chromosomes 22, 13, or 15 and rarely the other chromosome 21 (Jones 1997) with chromosome 21, therefore representing an unbalanced translocation.

The genetic risks of recurrence depend on the type of translocation and whether one of the parents carries a balanced translocation i.e. an accurate amount of genetic material. If there is a balanced translocation in the parental chromosomes, the recurrence risk can be up to 10%, unless the rare 21/21 translocation occurs where recurrence risks are 100% (Harper 2004).

1.1.1.3 Mosaic Down syndrome

Mosaicism occurs in about 2.4% of individuals with Down syndrome (Jones 1997). In mosaicism, two cell lines are present, some of which have 46 chromosomes and the other having 47 chromosomes due to the presence of an additional chromosome 21. Mosaicism usually results from a mal-segregation of homologs, or an anaphase lag of one homolog, which occurs postzygotically; the cell mixture arises after fertilization, usually when some of the cells in the developing fetus lose one of the chromosome number 21's at anaphase lag. Alternatively, a normal gamete arises and due to non-disjunction as cell division occurs, a cell line arises with the presence of an extra chromosome 21 (Mckinlay Gardner & Sutherland 2004). The recurrence risk for mosaic Down syndrome is approximately 1% (Jones 1997).

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1.1.2 Birth Prevalence of Down syndrome

Down syndrome is the most common genetic pattern of malformation in man, occurring in 1 in 660 newborns in industrialised countries (Jones 1997). The rate of trisomies is however subject to marked variation due to maternal age and the birth prevalence will thus vary according to the maternal age structure of the population (Brock, Rodeck & Ferguson-Smith 1992) as reflected in developing countries such as South Africa. The birth prevalence also varies according to the availability of medical genetic antenatal screening, prenatal diagnosis and the option of termination of pregnancy. In industrialised countries, about 50% of fetuses with Down syndrome are detected prenatally. With the impact of antenatal screening and prenatal diagnosis, the birth prevalence of Down syndrome is adjusted to approximately 1.43 to 1.54 per 1000 live births (Mueller & Young 2001). Birth prevalence has been documented as 2.3 per 1000 in middle and low-income countries but as low as 1.2 per 1000 in high-income countries (Christianson, Howson & Modell 2006).

Modell, Kuliev & Wagner (1992) documented that in Western Europe in the 1950's, the birth prevalence of Down syndrome was approximately 2.5 per 1000 live births, of which the majority were born to mothers over the age of 35 years. By 1980, the proportion of older mothers decreased to between 5% and 9% due to the use of family planning, with the birth prevalence of Down syndrome decreasing to as low as 1.2 per 1000 live births, of which the majority were born to younger mothers. Since the mid-1980's the proportion of older mothers in Europe has been on the increase because of women delaying reproduction until later in life. However, the minority of infants born with Down syndrome are born to older mothers (Christianson & Modell 2004; Christianson *et al* 2006), because of the

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availability and use of services for antenatal screening and for prenatal diagnosis, and the option of termination of pregnancy (Christianson *et al* 2006).

The situation is remarkably different in developing countries such as South Africa, where the birth prevalence of Down syndrome is higher due to a high percentage of women being of advanced maternal age (United Nations Fund for Population Activities 1998) and the availability of antenatal screening and prenatal diagnosis services and the option of termination of pregnancy being either not available to a large proportion of the population or not accessed by those who have it available (Schön 2004).

The percentages of births to women over 35 years of age range from 11-18% in developing countries and 5-9% in industrialized countries (Christianson *et al* 2006; United Nations Fund for Population Activities 1998). In industrialized countries, the minority of infants with Down syndrome were born to mothers over the age of 35 years (advanced maternal age), whereas in South Africa, studies of black neonates with Down syndrome have shown that 55% of infants with Down syndrome were born to mothers of advanced maternal age (Kromberg *et al* 1992).

It was not until 1955 that the first black African children with Down syndrome were described (Luder & Musoke 1955). The previously held belief that Down syndrome was rare amongst black Africans was shown to be incorrect when Adeyokunno (1982) reported in a retrospective study a birth prevalence of 1.16 per 1000 live births at an academic hospital in Nigeria. In South Africa, Kromberg, Christianson, Duthie-Nurse, Zwane and &

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Jenkins (1992) reported a birth prevalence of 1.68 per 1000 live births at the Chris Hani Baragwanath Hospital, refined to 1.8 per 1000 live births for infants born in urban Johannesburg, Gauteng (Christianson 1996). Delport *et al* (1995) documented a birth prevalence of 1.33 per 1000 live births in a Pretoria academic hospital. The birth prevalence of Down syndrome in rural South Africa was documented as 2.01 per 1000 live births in Limpopo (Venter *et al* 1995)

1.1.3 Clinical Features of Down syndrome

An individual with Down syndrome is recognised by a collection of common identifiable clinical features which include: hypotonia, a protruding tongue, hyperflexibility of joints with short stature, an awkward gait and increased weight as features of the older child and adolescence (Jones 1997). Other features may include microcephaly, brachycephaly, small ears, flat nasal bridge, epicanthic folds, short thumbs and fingers, sandal gap, and duodenal atresia (Winship 2003; Jones 1997). About 40% of infants with Down syndrome have cardiac anomalies, including endocardial cushion defects, ventricular septal defects, patent ductus arteriosus, atrial septal defects, and an aberrant subclavian artery. Hypothyroidism may occur in as many as 28% of individuals affected with Down syndrome. Thyroid function should be checked early in life and treated if indicated. Annual thyroid functions tests should be performed thereafter. In childhood, individuals with Down syndrome may have recurrent respiratory infections, intestinal obstruction, hearing loss, eye abnormalities and irregular dentition (Jones 1997).

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The defining clinical features of Down syndrome, which are mostly similar in different ethnic groups, and their frequency, are shown in Table 1.1 (Jones 1997).

TABLE 1.1: Principle features in neonates with Down syndrome

Organ System	Clinical Feature	Frequency (%)
Craniofacial features	Flat facial profile	90
	Excess skin on back of neck	80
	Slanted palpebral fissures	80
Central Nervous System	Hypotonia	80
	Poor Moro reflex	85
	Hyperflexibility of large joints	80
Musculoskeletal System	Dysplasia of pelvis on radiographs	70
	Anomalous auricles	60
	Dysplasia of midphalanx of fifth finger	60
	Clinodactyly	50
	Single palmar crease	45

The diagnosis of Down syndrome can be made soon after birth with the most common presenting feature being hypotonia. The newborn period is also characterized by lethargy and excess nuchal skin (Mueller & Young 2001). Christianson, Kromberg and Viljoen (1995) reported that some of the craniofacial features in black neonates with Down syndrome overlap significantly with the features found in normal black neonates. This

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made the diagnosis more difficult, especially for health care workers with limited experience of Down syndrome. They also stated that there was a lack of awareness by mothers of infants with Down syndrome who did not recognize the differences between their infant with Down syndrome and normal newborns. Christianson and Kromberg (1996) found that 83% of mothers with African infants with Down syndrome did not recognise any facial differences in their affected child compared to other normal infants. They observed that awareness of these mothers was often delayed until delayed milestones in their infant with Down syndrome were noticed and subsequently this led them to bring the problem to the attention of medical personnel.

Individuals with Down syndrome all have intellectual disability and delayed milestones. The rate of developmental progress slows with age (Jones 1997). Intellectual disability (ID) is defined as limitations in intellectual functioning, which includes difficulties in learning and performing daily life skills. Intellectual disability can be classified as mild, moderate, severe or profound, based upon IQ (intelligence quotient) ranges. A person with a "severe" or "profound" disability may be unable to learn basic social skills such as speech, basic motor skills including walking and personal care, and is likely to require supported accommodation (WHO 1992).

Intellectual disability places a great burden on individuals, their families, society and the health care system, as it is serious and lifelong (Christianson, Zwane, Manga, Rosen, Venter, Downs & Kromberg 2002). Limited information is available on the prevalence of intellectual disability and on associated epidemiological factors in developing countries.

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Christianson *et al* (2002) performed a study on intellectual disability in rural South Africa and defined severe intellectual disability (ID) when general intelligence quotient (GIQ) was less than 55 and mild ID when GIQ was 56-80. Jones (1997) reports that the IQ range for children with Down syndrome is generally 25 to 50. The IQ in individuals with Down syndrome decreases as the child gets older (Jones 1997; Gath 1994). The mean IQ for older individuals is 24 although there have been reports on individuals with that over 50 (Jones 1997).

Behavioural problems are common in Down syndrome and are a common and perturbing complication of intellectual disability (Gath 1994). Epidemiological studies show that behavioural disorders become more common with decreasing levels of intellectual functioning (Gath & Gumley 1986). Gath (1994) reported that half the children with Down syndrome at the age of 8 years had significantly more behaviour problems than unaffected children of the same age. She noted that 25% of the children with Down syndrome had attention deficit disorder. Individuals may be mischievous and obstinate and 13% have serious emotional problems (Jones 1997).

Down syndrome adults are at high risk of developing Alzheimer's disease, becoming apparent usually in the fifth decade but earlier in some cases. This is much earlier than the general population (Dalton & Wisniewski 1990). Distress, confusion and other signs of premature ageing accompany the loss of skills (Gath 1994). Approximately 30 -60% of adults acquire cataracts (Jones 1997).

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1.1.4 Prognosis of Individuals with Down Syndrome

The prognosis of infants born with a severe congenital disorder depends on the level of development and availability of health services. In high-resource settings, up to 70% of all birth defects can be avoided, infant lives saved or disability considerably improved (Christianson & Modell 2004) by comparison to low-income settings where a majority of affected infants die undiagnosed or are disabled (WHO 1985; Christianson, Howson & Modell 2006). More than 30% of infants with a serious congenital disorder in middle-income countries, and at least 50 % in low-resource countries die within infancy or childhood (Christianson & Modell 2004). The population prevalence of Down syndrome in high resource countries is close to 70% of its birth prevalence (Christianson & Modell 2004). In industrialized settings, the life expectancy of individuals with Down syndrome has improved in recent years compared to what it was decades ago (see figure 1.2) as complications are treated and educational and social support is available (Christianson & Modell 2004).

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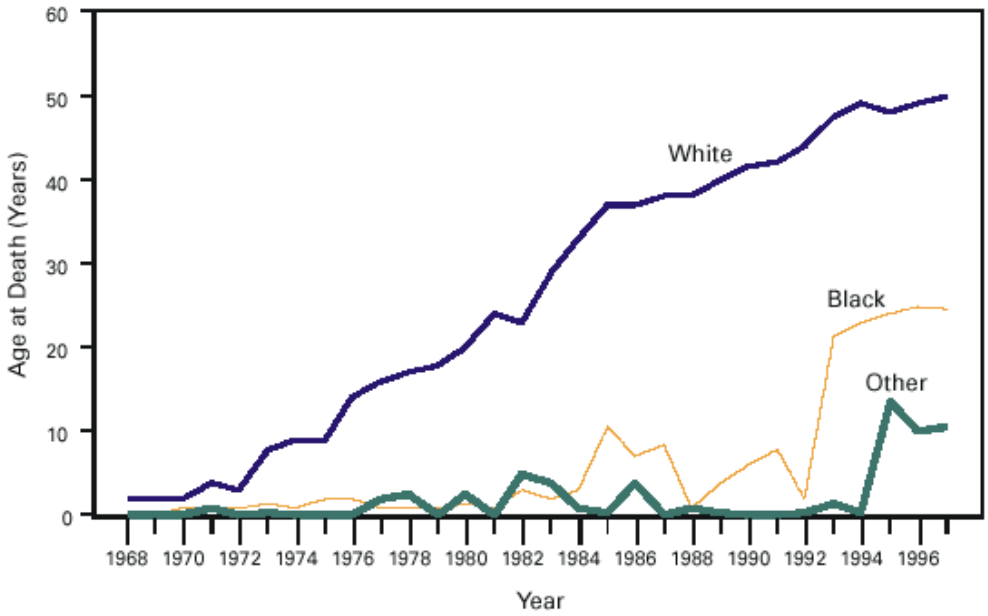

FIGURE 1.2: Median age at death of people with Down syndrome by race – United States 1968-1997 (CDC data 1968-1997)

A cohort study in France (1984-1990) of 280 Down syndrome fetuses, recorded that 43% (120) had died by the end of the first year of life. Termination of pregnancy had been carried out in 76 (27%) of the pregnancies. The remaining mortality was due to late spontaneous abortion or stillbirth (11 or 4%), or death during the first year of life (33 or 12%). Of the 192 live born infants with Down syndrome, 17% (33) died during the first year of life, with half of these in the neonatal period (Julian-Reynier *et al* 1995). Baird & Sadovnick in 1989 showed that life expectancy in children with Down syndrome is significantly poorer if congenital heart disease is present. They had also documented (1987) that for patients with congenital heart disease (CHD), 76.3% survive to 1 year of age; 61.8% to age 5; 57.1% to age 10; 53.1% to age 20 and 49.9% to age 30. For patients

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without CHD, survival to the same ages is 90.7%; 87.2%; 84.9%; 81.9% and 70.2%, respectively. Mortality from respiratory disease, predominantly pneumonia, and also other infectious diseases is much higher than in the general population (Jones 1997).

In lower resource nations, the mortality in infancy and early childhood of children with Down syndrome is far greater (Christianson & Modell 2004). In South America 26.4% of infants with Down syndrome die before their first birthday (Castilla, Rittler, Dutra, Lopez-Camelo, Campana 1998). Christianson *et al* (2002) documented that two out of three children with Down syndrome in South Africa die prior to 2 years of age.

For those children who survive, there is lifelong chronic disability with needs that exceed those of most other children (Gath 1990). These needs include early developmental enrichment programs, including physiotherapy and speech therapy which are recommended (Jones 1997; Cohen, Nadel, Madnick 2002), special education needs, and medical needs due to recurrent respiratory infections, and other complications such as those listed above (Jones 1997).

It was estimated that the cost of medical care in the public service in South Africa for one individual with Down syndrome was on average R20 000 per year in 2001, excluding the care of cardiac abnormalities, which would cost in the region of R50 000 if surgery were required (Policy Guidelines for the management and prevention of Genetic Disorders Birth Defects and Disabilities 2001). Given that 2300 infants with Down syndrome will enter the

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health care system in South Africa per year, the estimated cost of caring for each annual cohort of these children is approximately R100 million (Christianson *et al* 2006).

1.1.5 Care of People with Down syndrome

The burden of congenital disorders worldwide has been greatly underestimated. It has not been recognised, especially in middle and low-income nations, that there is a great need, and that possibilities exist for care and prevention of congenital disorders. However, prognosis improves as care improves; an increasing proportion of infants with congenital disabilities survive as services become available. As the number of infants that survive increases, the number requiring care increases incrementally (Christianson & Modell 2004).

Care for individuals with congenital disorders includes diagnosis, therapeutic intervention and genetic counselling with psychosocial support (WHO 1999; Christianson & Modell 2004; Christianson *et al* 2006).

Care for individuals with Down syndrome therefore involves:

1) Early postnatal diagnosis
2) Treatment
    i) Medical treatment for congenital heart defects, recurrent infections, hypothyroidism and other medical complications
    ii) Surgical treatment for congenital heart defects, duodenal atresia and other malformations
    iii) Neurodevelopmental therapy

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3) Genetic Counselling

1.1.5.1 Diagnosis

A clinical and family history, a physical examination and laboratory testing are required to make a medical genetic diagnosis. A realistic care plan, taking the family, community circumstances and available health care facilities into account, can then be considered once an accurate and hopefully early diagnosis has been confirmed (Christianson & Modell 2004). This is as important for Down syndrome as any other birth defect, especially in the situation in South Africa where the clinical diagnosis of Down syndrome is poor (Christianson & Kromberg 1996).

1.1.5.2 Treatment

Medical care requires an organised approach of assessment, monitoring, and vigilance. Once an early and correct diagnosis is made in the individual, the associated problems can be recognised. Treatment for an individual who has been diagnosed with Down syndrome includes cardiac evaluation with treatment if necessary, prompt treatment of infection, regular dental care, monitoring of thyroid function and treatment of hypothyroidism when indicated and prevention of obesity (Roizen & Patterson 2003). The best option for neurodevelopmental therapy to help an affected individual in middle and low-income settings is community based rehabilitation run by primary health care practitioners with local people trained to assist (Christianson & Modell 2004).

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Many simple and therapeutic measures for people with Down syndrome exist that can be given in the primary health care setting. The more complex interventions such as cardiac assessment and surgery for children with Down syndrome may be available in secondary and tertiary hospitals. However complex procedures such as cardiac surgery may not be offered to such a child, because of lack of facilities and competing priorities (Christianson & Modell 2004).

Improvements in the quality of life of individuals with Down syndrome have resulted from advances in medical care, early educational intervention with support in educational settings, and the identification and treatment of psychiatric disorders found in Down syndrome such as depression, disruptive behaviour disorders, and autism (Roizen & Patterson 2003).

1.1.5.3 Genetic Counselling

Genetic counselling is an educational process by which individuals or relatives, at risk of a congenital disorder, are informed of the nature of the genetic disorder, the probability of developing or transmitting it and the options available to them in which it can "be prevented, avoided or ameliorated" (Harper 2004 p.3). Genetic counselling plays an important role in the co-ordination of the multi-disciplinary and long-term management of patients as the necessary referrals are made and genetic counsellors are aware of what services are available to patients. Genetic counselling also aims to provide psychosocial support. Starke & Moller (2002) reported that by seeking information and gaining knowledge about the diagnosis, parents can handle their reactions of having a child with a

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chronic illness. It is thought that this is a way for parents to restore order in a "chaotic existence". Genetic counsellors are trained to meet these needs in parents of children with a congenital disorder.

Genetic counselling for Down syndrome occurs in two different settings: prenatal and postnatal. In prenatal counselling for Down syndrome, the risk of having a baby with Down syndrome would be discussed, depending on the family history, the mother's age and results of medical genetic screening (see section 1.1.6.2). Prenatal diagnostic situations can be complex and always carry some degree of prognostic uncertainty, making decisions and preparation for the future difficult (Fonda Allen & Mulhauser 1995). The options available after prenatal diagnosis of a congenital disorder are often limited, and the resulting psychosocial and emotional consequences are far-reaching, making the experience for parents a significant burden. Previous dreams and hopes for a healthy baby are threatened, and their ability to cope and their views of life may be challenged. An abnormal fetus may greatly affect their bonding with the infant, their belief in a good world, interactions with each other, as well as their perception of future risks (Fonda Allen & Mulhauser 1995). The genetic counselling process aims to empower individuals and their families experiencing such turmoil through informing them and providing psychosocial support, thus enabling them to make suitable decisions.

In post-natal counselling where a baby is suspected of having Down syndrome, an examination and chromosome analysis is performed, or parents are counselled on a confirmed result for Down syndrome. The clarification of the diagnosis, the cause, clinical

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features, prognosis, treatment options and risks for future pregnancies are discussed with parents and the necessary referrals are made. A congenital disability has lifelong implications for the individual and family and genetic counselling provides a support service.

Genetic counselling is non-directive and focuses on the accurate and clear presentation of facts, risks and options, and one of the most important goals of genetic counselling is to help patients understand the reproductive options available to them (D'Alton & DeCherney 1993).

1.1.6 Reproductive Options of Women at Risk

The prevention of congenital disorders based on reproductive options includes family planning, public education about prenatal risks, and prenatal screening and diagnosis for fetal anomalies (Penchaszadeh 2002; Christianson *et al* 2006). Using these options, women at risk thus have the reproductive choice to prevent occurrence or recurrence of a child with Down syndrome (Christianson & Modell 2004).

1.1.6.1 Family Planning

Family planning allows couples to have the option of when to start and complete their family, to determine their family size, and to plan spacing between children. It is a woman's basic right, and plays a positive role in reproductive health and in the improvement of pregnancy outcomes (United Nations Fund for Population Activities

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1998). When women in a population choose to complete their families at a younger age, it is associated with the reduction of chromosomal trisomies, particularly Down syndrome (Christianson & Modell 2004).

Penchaszadeh (2002) summarised that in developing countries, rates of Down syndrome that are maternal age-specific are similar to those in developed countries but that there is data suggesting that the overall birth prevalence of chromosome disorders is higher in developing countries. This could either be as a result of the lack of family planning services or lack of access to them resulting in a higher proportion of births to women of advanced maternal age. As discussed previously (in section 1.1.2), births to women over 35 years of age are more common in developing nations than in industrialized countries (United Nations Fund for Population Activities 1998). The WHO (2000) estimated that in developing countries with high fertility, the reduction to 2 to 3 children per family together with encouragement to complete reproduction before the age of 35, family planning can reduce the birth prevalence of Down syndrome by 50%.

1.1.6.2 Medical Genetic Screening for Down syndrome

Medical screening is a public health activity that aims at the prevention of disease on a population basis, where screening is offered to all people in a defined population. Down syndrome screening programmes aim to identify mothers at risk of having a child with Down syndrome so that they may receive genetic counselling regarding their specific circumstance and the options available (Christianson & Modell 2004).

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Medical genetic screening for Down syndrome during pregnancy is undertaken by determination of advanced maternal age, biochemical screening in the first and second trimesters, and ultrasound scanning. Women identified at being at an increased risk for having a fetus with Down syndrome should then have the choice of having the diagnosis confirmed by prenatal testing. Once confirmed, women through genetic counselling can be offered the choice of TOP or continuing with the pregnancy.

1.1.6.2.1 Advanced Maternal Age

As discussed in section 1.1.2, the rate of trisomy 21 increases with maternal age. A woman should be referred for genetic counselling when she is of advanced maternal age (AMA), which is from the age of 35 years and older. At the age of 35, a woman has a risk of 1 in 386 of having a baby with Down syndrome, and this risk increases with advancing maternal age (Hook *et al* 1983).

1.1.6.2.2 First Trimester Screening

First trimester screening differs substantially in the public and private (fee for service) health sectors in South Africa. In the private sector, first trimester biochemical screening for Down syndrome has been established using pregnancy-associated plasma protein-A (PAPP-A) and the free beta subunit of human chorionic gonadotropin (Fß-hCG) levels present in maternal blood. Reduced levels of PAPP-A and increased levels of ß-hCG are found in the first trimester of pregnancies with fetuses affected by Down syndrome (Wheeler & Sinosich 1998). One fetal ultrasound marker, nuchal translucency (NT), has

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been shown to be effective for first trimester screening for Down syndrome. NT screening can be offered to women to a limited extent in the private and public sector, if they book for antenatal care in their first trimester of pregnancy. However, it is a procedure that requires appropriately trained operators, of which there are few in South Africa. NT screening is performed between 11 and 14 weeks gestation and involves measuring the thickness of a fluid filled space behind the neck of the fetus. The NT thickness changes with gestational age and measurement of the NT provides a modified risk, relative to the age-related risk, for having a fetus with a chromosome abnormality/Down syndrome. Increased nuchal translucency is also associated with congenital heart disease, severe skeletal dysplasias and other fetal abnormalities. Approximately 5% of normal fetuses would also have an increased NT measurement (Nicolaides, Sebire & Snijders 1999). Prospective studies have shown that screening by a combination of NT and Fß-hCG and PAPP-A can identify 90% of fetuses with trisomy 21 and other major chromosomal abnormalities, with a false-positive rate of 5% (Nicolaides 2005).

1.1.6.2.3 Second Trimester Screening

The maternal serum triple test (MSTS) which measures AFP, human chorionic gonadotrophin (hCG), and oestriol is used to screen for Down syndrome in pregnant women in the second trimester of pregnancy (Chard & Macintosh 1995). It identifies approximately 60% of cases of Down's syndrome, with a false positive rate of 6.6%. If ultrasonography is used to verify gestational age, the false positive rate is reduced to 3.8%. It is essential that this test be done between 16 and 18 weeks gestation (Haddow, Palomaki, Knight *et al* 1992). Other factors, such as maternal weight, maternal diabetes, ethnicity,

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twin pregnancy and the previous maternal history can influence the results of the MSTS (Wald, Cuckle, Densem *et al* 1988), and women should be made aware of the limitations.

Fetal anomaly scanning is used in the second trimester, between 18 and 20 weeks gestation. It is important that it is performed by skilled operators using high quality equipment. It is also important that referring doctors understand the possible anomalies that may be seen and how to explain these findings to the patient. Ultrasound in the second trimester diagnoses 50-70% of cases of Down syndrome by detecting soft markers (Shipp & Benacerraf 2002).

Screening for Down syndrome in pregnant women ultimately increases or decreases their risks for having a baby with Down syndrome and thus influences a woman's decision as to whether or not she should have prenatal invasive testing.

1.1.6.3 Prenatal Diagnostic Testing

Prenatal diagnosis is the identification of disease or condition in a fetus or embryo prior to delivery. Ultrasound and amniocentesis were first used for prenatal diagnosis of fetal abnormalities due to neural tube defects and chromosomal abnormalities in the 1970's (Fonda Allen & Mulhauser 1995). Amniocentesis is the most common invasive procedure used to obtain fetal cells for prenatal diagnosis of chromosomal abnormalities (Brock, Rodeck & Ferguson-Smith 1992). The procedure is done by direct needling of the pregnant uterus with the aid of an ultrasound scan, to obtain a small sample of amniotic fluid ($\leq$ 20ml), usually done between 16 and 20 weeks gestation (Harper 2004).

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Amniocentesis has an associated risk of fetal loss estimated as 0.5 to 1.0% (D'Alton & DeCherney 1993). Cells can also be obtained by chorionic villi sampling (CVS), where samples of developing placenta are obtained transcervically or transabdominally at 11-14 weeks of pregnancy, and by cordocentesis, the extraction of fetal blood from the umbilical cord, after 20 weeks gestation (Harper 2004). The rate of miscarriage proceeding CVS exceeds that in amniocentesis by 0.8% (Rhoads, Jackson, Schlesselman *et al* 1989). Other studies suggest a 2-3% risk of fetal loss and give reports of an excess of limb abnormalities possibly related to CVS, especially when done at a gestation earlier than 9 weeks (Harper 2004). Cordocentesis was reported to have an associated fetal loss rate of 2.5-5% (Brock, Rodeck & Ferguson-Smith 1992).

After amniocentesis or CVS when the pregnancy is still relatively early, cytogenetic analysis (karyotyping) on fetal cells is performed, from which major structural and numerical chromosomal abnormalities can be detected (Valenti, Schutta & Kehaty 1968). Culture failure may occur after amniocentesis in less than 1% of cases and maternal contamination is approximately 2% in CVS samples (D'Alton & DeCherney 1993).

Amniocentesis can be performed up to 24 weeks gestation to obtain fetal cells for Polymerase Chain Reaction (PCR) and direct DNA analysis without the need to culture the cells (Mueller & Young 2001). This analysis, PCR aneuploidy, can detect the common numerical chromosome abnormalities (which include trisomy 21) and has been available at the National Health Laboratory Service (Central) in Johannesburg since 2001 (Lane 2005).

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Harper (2004) reports that most couples that have had a child with Down syndrome do elect for prenatal diagnosis in a subsequent pregnancy and that the strength and indication for prenatal diagnosis will depend on their perception of the magnitude of the recurrence risk and their attitude and own experience. Before prenatal procedures are contemplated, the acceptability of termination of pregnancy to a couple should be considered. Sometimes it is unacceptable to couples based on religious grounds, or because of the attitude of the community, or due to a personal ethical view. Unacceptability of termination however, should not rule out the option of prenatal diagnosis as some parents feel it is a way they can be better prepared for an abnormal child (Harper 2004).

A person's previous experience, as well as ethnic and cultural background and religious beliefs will affect the acceptability of prenatal diagnosis and the choices to be made if a fetal abnormality is diagnosed. Genetic counselling is essential as parents need to understand that the central issue is balancing the risk of an investigative invasive procedure against the risk of the birth of an affected child (Harper 2004).

1.1.7 Prenatal diagnosis for Down syndrome in South Africa

In South Africa, prenatal diagnostic services exist in the private and public sector. They are however only available to the public sector at selected Academic and State hospitals. The resources are limited as many of the techniques require high quality, expensive equipment and trained technicians.

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Some developing countries have followed the example of industrialized nations in implementing prenatal screening programs, prenatal diagnosis, the option of amniocentesis, and the option of termination of pregnancy if the fetus has Down syndrome. South Africa is a country where improvement of health conditions exposed birth defects, including Down syndrome, as a major cause of infant mortality (Penchaszadeh 2002). Genetic services for genetic counselling and prenatal diagnosis have been available in Johannesburg since 1979, but have been underutilised by the African population (Kromberg *et* al 1989). However, a large proportion of the population remain unaware of this genetic service (Schön 2004). The mean gestational age that many women present at a tertiary care centre such as Baragwanath Hospital is 28 weeks (Kromberg *et al* 1992), which is too far advanced to offer invasive prenatal testing.

Although it is thought that women who receive prenatal care have fewer complications in pregnancy and during delivery, it has been found that there is a difference in the utilisation rate of services that offer prenatal diagnosis by people of different ethnic origin (Sokal *et al* 1980; Brett *et al* 1994; Kupperman *et al* 1996). Reasons for the difference in utilisation have been suggested; Epstein *et al* (1972) and Shino & Kellogg (1977) reported that it was due to lack of education; Kupperman *et al* (1996) proposed that lack of education and socio-economic factors were the major factors. Pelser (1998) proposed that the lack of awareness and under-utilization of services in South Africa could be due to limited knowledge regarding prenatal diagnostic services, as well as the lack of availability of prenatal tests. Pelser (1998) and Schön (2004) reported that medical personnel fail to inform women regarding the tests. Schön (2004) found that women of advanced maternal

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age present to clinics in time for genetic counselling and prenatal diagnosis but are not referred on time by medical and nursing staff. The researcher feels that this is one of the critical issues that needs to be addressed in South Africa.

The prenatal diagnostic techniques described above (section 1.1.6.3) are currently not offered routinely in South Africa. There are few centres with suitably trained doctors to offer NT screening, and the accuracy is reduced in MSTS screening due to the lack of appropriate equipment and adequately trained professionals to assess correct gestational dating.

South Africa has however made tremendous progress over the last decade in the development of principles, practice and policy for the initiation and development of a national community-based medical genetics programme. The programme exists for the care of people with genetic disorders, birth defects, disability as well as prevention (Christianson 2000).

Infant and childhood mortality and morbidity has until recently predominantly been caused by infectious diseases and malnutrition in developing nations. These problems have been addressed and mortality is thus declining. An epidemiological shift is beginning to occur in South Africa (Christianson *et al* 2002). Genetic disorders and birth defects which previously received limited attention from health care planners, have begun to "emerge and demand due consideration" (Christianson *et al* 2002 p.180), as these contribute increasingly to infant and childhood mortality. However, a major threat to this health transition exists in

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South Africa through the current Human Immunodeficiency Virus (HIV)/ Acquired Immune Deficiency syndrome (AIDS) pandemic. It affects infants, children and adults, causing death and disability and is using up an increasing amount of the National healthcare budget (Christianson 2000).

HIV/AIDS indirectly creates a quandary in diagnosing fetal chromosomal abnormalities, as the invasive procedures used to obtain cells create an increased risk of transmission of the virus to the fetus (Tess *et al* 1998). Thus after counselling, women can either opt not to have prenatal diagnosis for chromosome abnormalities or if they opt for prenatal diagnosis be made aware that the fetus is at an increased risk of contracting HIV/AIDS. Antiretroviral treatment is not currently available at all the hospitals where the invasive procedures are performed and many women decline prenatal testing due to their HIV status alone. An increasing number of women of advanced maternal age who are HIV positive are presenting at Genetic counselling clinics (Bee 2005). Bee (2005) showed that 39% of HIV negative women opted to have amniocentesis, whereas only 14% of HIV positive women choose this option. The study concluded that there is no clear understanding of mother to child transmission during second trimester amniocentesis, and that there are no clear policies in South Africa regarding HIV prophylaxis prior to and after amniocentesis.

Although the local situation has improved over the years, and an essential service is now offered to patients, there are many issues that need to be addressed and resolved.

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1.2 AIMS OF THE STUDY

The aim of this study was to determine what reproductive decisions are made by South African women who live in Johannesburg who have previously had a child with Down syndrome. Attitudes of women who have had children with Down syndrome to having further children were explored, as well as to family planning, prenatal screening, prenatal diagnosis, their considerations about termination of pregnancy and their emotional experience when initially given the diagnosis of their child with Down syndrome. Information gained will aim to improve understanding of maternal psychological and social factors and responses surrounding the diagnosis of a child with Down syndrome.

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CHAPTER 2

STUDY PARTICIPANTS & METHODOLOGY

Relevant aspects of Down syndrome including the reproductive options available to women were discussed in the previous chapter. In this chapter the study participants and methodology will be discussed.

The research design was a retrospective study, the aim met through the administration of face-to-face interviews using a structured questionnaire. Ethical clearance was obtained from the Medical Human Research Ethics Committee (clearance number: M040226).

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2.1 RESEARCH TOOL

The survey was conducted through the use of a structured questionnaire in an interview format (Appendix A). An explanation of the study and informed consent was obtained from all the women that participated in the study (Appendix B and C). To minimize inter-personal differences and interviewer bias, the questionnaire was completed by the researcher on a face-to-face basis. The researcher ensured that all the questions were explained uniformly to all participants and that all questions were fully understood. A translator was used to interpret the questions to mothers, and also the answers from those participants who could not speak or understand English.

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The research questionnaire was administered in a structured and standard manner. The following information was gained:

i) General Information
   a. Social information
   b. Socio-economic status

ii) Biomedical Information
   a. Previous pregnancy history
   b. Prenatal diagnosis in pregnancy involving child with Down syndrome
   c. Reproductive decisions

iii) Information on child with Down syndrome
   a. Age of child
   b. Maternal knowledge of Down syndrome prior to birth of child

iv) Genetic Counselling
   a. Whether mother received counselling
   b. From whom counselling was received

v) Reproductive decisions made, after birth of child with Down syndrome

2.2 RESEARCH METHOD

2.2.1 The Population

The women participants for the research study were South Africans who had a child with Down syndrome who had survived to at least 1 year of age. The sample consisted of 50 women.

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2.2.2 The Sampling Method

Women with children with Down syndrome were accessed from seven sources, listed below. The women were given an explanation of the study and an information sheet (Appendix B) and then asked to sign consent (Appendix C) before taking part in the study.

2.2.3 The Sample

The women were accessed through:

1) Division of Human Genetics

The names of mothers of children with Down syndrome were obtained from files of the Division of Human Genetics, National Health Laboratory Service (Central) for the years 1980-2003. These mothers had attended a Genetic Counselling clinic.

2) Genetic Counselling Clinics

Mothers of children with Down syndrome who were seen at the Genetic Counselling Clinics run weekly at Chris Hani Baragwanath, Johannesburg General and Coronation Hospitals were included in the study. These mothers were either coming for genetic counselling for the first time or were attending a follow-up appointment.

3) The Memorial Institute (TMI)

Clinics for children with Down syndrome are run on a monthly basis at the TMI, where they attend for ongoing developmental assessment. These clinics were attended by the researcher and mothers were asked if they would participate in the study.

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4) Down syndrome South Africa (DSSA)

The mothers of children with Down syndrome who attended weekly DSSA support group meetings at TMI were approached to take part in the study. The Down syndrome support group in Lenasia was attended by the researcher and mothers of children with Down syndrome were approached. The researcher also interviewed the mothers on the DSSA committee who have children with Down syndrome.

5) Word of Mouth

Families known to colleagues were introduced to the interviewer and home visits arranged to conduct the interviews.

6) Cardiac Clinic

Contact was made with a paediatric cardiologist working at the Johannesburg Hospital. Permission was obtained to approach mothers of children with Down syndrome attending paediatric cardiology clinic.

7) Speech and Hearing Department

Interviews were conducted with mothers who attend a Down syndrome support group run by the social work department at Chris Hani Baragwanath Hospital.

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2.3 DATA COLLECTION AND ANALYSIS

2.3.1 Pilot Study

A pilot study using the questionnaire (Appendix A) was done on 10 women, prior to commencement of the study. No changes were considered necessary after the pilot study and the remaining interviews were conducted with the original questionnaire.

2.3.2 Data Analysis

The results from the questionnaire were coded and entered onto a spread sheet using Microsoft Excel. Chi square testing was performed on the data where comparison was to be made between populations. The following website was used for this purpose: http://www.graphpad.com/quickcalcs/contingency1.cfm.

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2.4 ETHICAL CONSIDERATIONS

Ethical clearance was obtained from the Human Research Ethics Committee (Medical) on 09/02/2004 to undertake this study (Appendix D). The mothers that were interviewed were ensured that confidentiality would be maintained and they were informed that they could withdraw from the study at any stage without any detrimental effect in the management or treatment of child.

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CHAPTER 3

RESULTS AND DISCUSSION

The questionnaire (Appendix A) was administered to South African mothers living in Johannesburg who have children with Down syndrome in order to determine their knowledge regarding Down syndrome, their reproductive choices after having a child with Down syndrome and what may have influenced their decisions. In this chapter, the results will be presented in the order elicited from the questionnaire and will be depicted in tables, histograms and picture charts. A discussion is included to further elucidate the particular category addressed in the questionnaire.

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3.1 DEMOGRAPHICS

The mothers of children with Down syndrome interviewed in this study were from all social classes and various education levels. They all have a living child with Down syndrome who was at least 1 year of age at the time of the study. Fifty women, 36 African, 4 Indian and 10 Caucasian (figure 3.1.1), were interviewed. It was required for this study that the child was older than one year of age as these women were considered to have experienced some of the emotional and social issues associated with having a child with Down syndrome, and also to have had sufficient time to have made choices regarding reproduction.

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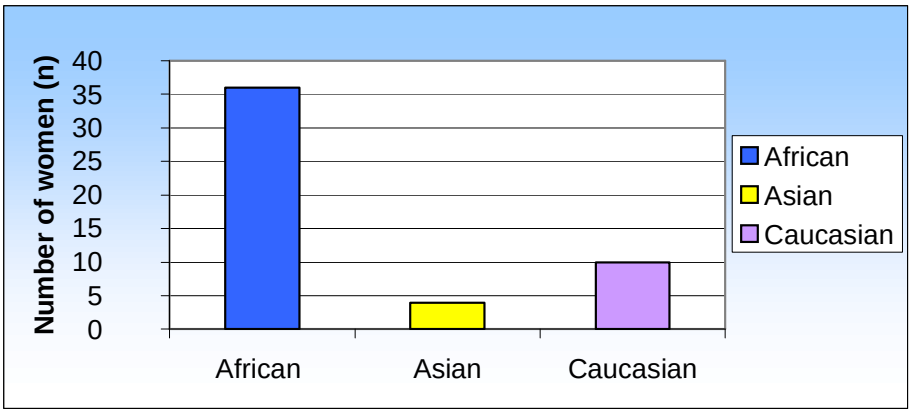

FIGURE 3.1.1: Total number of women included in the study

Most of the mothers interviewed, 72% (36), were African women, who used public sector medical services. This was expected as the majority of children with Down syndrome born in South Africa are African (Christianson 1996). The ages of the mothers interviewed ranged from 21 years to 56 years. The mean age was 37.8 years.

The social class of each woman was determined by assessing the woman's occupation. In the cases where women were currently unemployed, their previous occupation, or the occupation of their spouse/partner was taken into account. Based on international models, Schlemmer and Stopforth's (1979) guide to coding of occupations, was adapted for the South African situation. All occupations were divided into 5 groups, with group 1 being the highest social class and group 5 being the lowest:

Group 1: Professional and Managerial

Group 2: Middle White-collar

Group 3: Manual Foreman, Skilled Artisans, Farmers, and Status Equivalent

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Group 4: Routine Non-Manual and Semi-Skilled Manual

Group 5: Unskilled Manual and Menial

For this study, the five groups were narrowed down to two classes. The first, indicating higher social status, consists of groups 1-3, and the second, lower social status, consists of groups 4 and 5. The total household income of the woman was determined to ensure accuracy of the grouping. The income was found to be congruent with the occupation for each woman with respect to categorization into the social class.

Of the 50 women interviewed, 35 (70%) were from a lower social background and 15 (30%) were from a higher social class. Of the African women, 2 (4%) were from a high socio-economic background and 34 (68%) were from a low social background, whereas 9 (18%) of the Caucasian women were from the former and 1 (2%) from the latter (figure 3.1.2).

For comparison, in South Africa the unemployment rate of women was found to be approximately 35% (and 26.2% if men are included) according to the General Household survey July 2004 (Statistics South Africa 2005). Of the category of employed people, 22.4% were in the three most senior occupation categories, which included managers, professionals, semi-professionals, and technicians (termed “more skilled”). Therefore, based on occupation, 83.5% of people in South Africa are either unemployed or part of the less skilled occupation categories (lower social status) and 16.5% are of higher social class. Therefore when comparing the social status of the sample of this study to the statistics of the population of South Africa, the Pearson Chi-square value was 6.614 and subsequent p-

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value of 0.01 indicates a significant difference between the two groups and that the sample in this study is not an accurate representation of the population of South Africa as a whole.

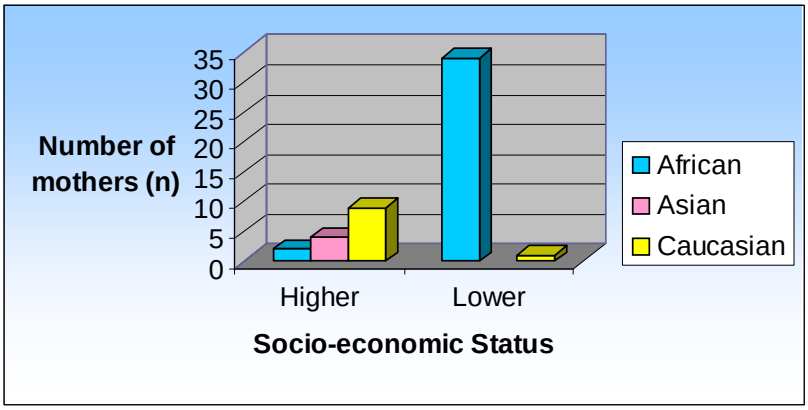

FIGURE 3.1.2: Distribution of socio-economic status among ethnic groups

The mothers were divided into four groups according to their level of education:

a) Group 1 included women who had no schooling at all

b) Group 2 included women who had 1 to 7 years schooling

c) Group 3 included women who had 7 to 12 years schooling

d) Group 4 included women who had tertiary education

Of the 50 women in the sample, 5 (10%) had no schooling, 11 (22%) had 1 to 7 years schooling, 21 (42%) had 7 to 12 years schooling and 13 (26%) had obtained tertiary education (figure 3.1.3).

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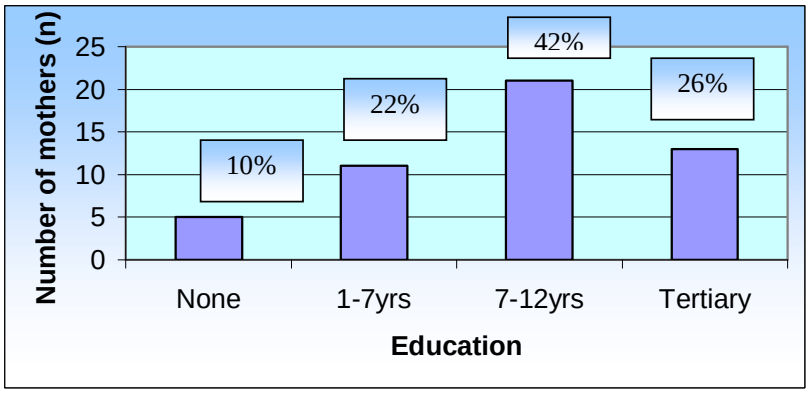

FIGURE 3.1.3: Distribution of education level of mothers

For comparison with the South African population, Statistics South Africa (2003) reported the level of education of people 20 years and older in the census 2001, and documented that 17.9% of the population had no education, 22.4% had 1 to 7 years of schooling, 51.2% had 7 to 12 years of schooling and 8.4% had tertiary education. The number of women in the education groups in this study with reference to their ethnicity, is shown in figure 3.1.4.

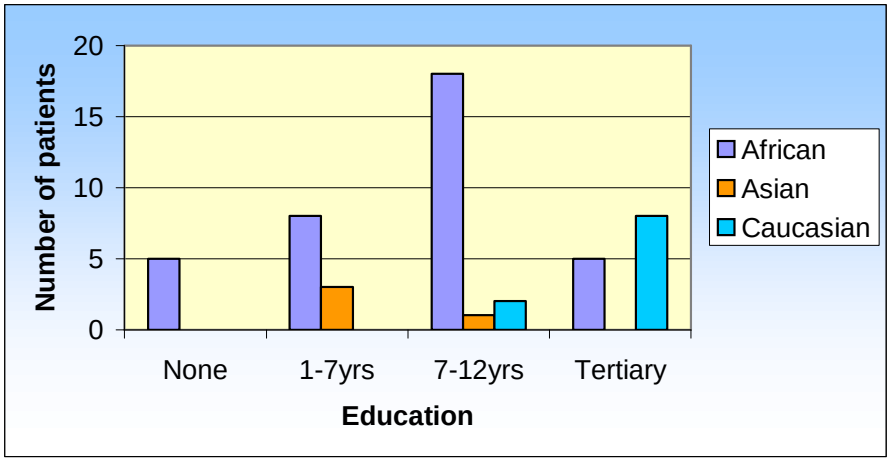

FIGURE 3.1.4: Distribution of education level among ethnic groups

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3.2 INFORMATION ON PREVIOUS PREGNANCIES

The average number of pregnancies that the mothers of children with Down syndrome had was 3.4, with a range of 1 to 10 pregnancies and the average number of live born infants was 3.0, with a range of 1 to 10. At the time of the interview, the number of children per mother ranged from 1 to 10, with a mean value of 2.9. The average number of children per mother was 2.1 in the Caucasian population, 3.1 in the African population and 3.0 in the Asian population.

For comparison, the total fertility rate in South Africa is 2.8. The total fertility rate amongst different ethnic groups in South Africa was recorded to be 3.0 in the African population, 1.7 in the Caucasian population and 1.9 in the Asian population (Health Systems Trust 2005).

Africans have more children per family on average compared to the Caucasian population, thus the birth prevalence of African babies with Down syndrome would be expected to be higher. In addition to having more children (Health Systems Trust 2005), many African mothers have their children when they are over the age of 35, increasing their risk for having a baby with Down syndrome (Kromberg *et al* 1992).

3.3 MATERNAL KNOWLEDGE OF DOWN SYNDROME PRIOR TO BIRTH OF CHILD WITH DOWN SYNDROME

The mothers were questioned on their knowledge of Down syndrome prior to their child with Down syndrome being born. Of the mothers, 17 (34%) knew about Down syndrome

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compared to 33 (66%) who did not. Of the Caucasian women, 90% (9) knew what Down syndrome was prior to the birth of their child while only 19% (7) of African women could answer in the affirmative (figure 3.3.1). To confirm this knowledge regarding Down syndrome, they were asked to give a short description about what they knew about Down syndrome. A mother was regarded as having knowledge of Down syndrome when her description included that it was a chromosome abnormality or that affected individuals were "slow" or intellectually delayed.

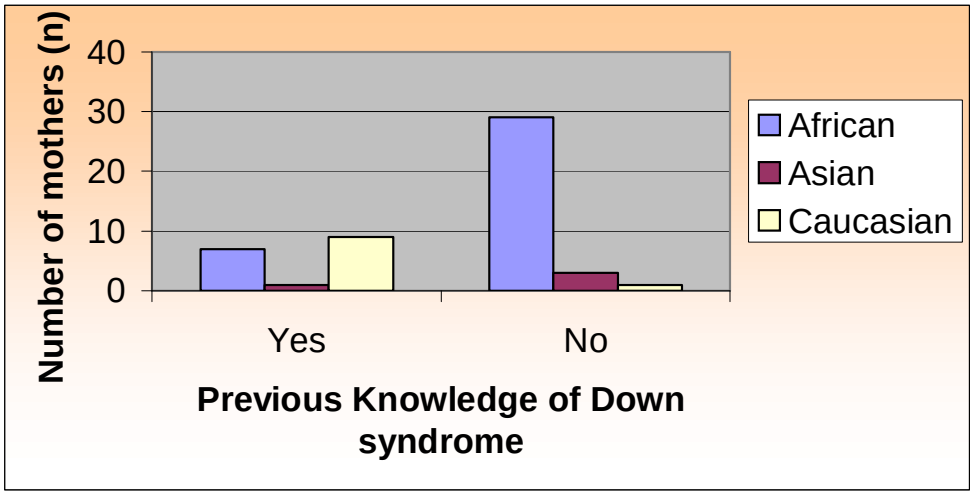

FIGURE 3.3.1: Mother's knowledge of Down syndrome prior to birth of child

Using the Fisher's exact test to compare the prior knowledge of Down syndrome between the African and Caucasian groups, the two-tailed P value of less than 0.0001 was statistically significant. Of the mothers who did know about Down syndrome (17 or 34%) however, most (15 of the 17) had only a very basic knowledge and did not fully understand the severity or prognosis of the disorder.

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Pelser (1998) documented in a study done in Pretoria, Gauteng, on women of advanced maternal age presenting for genetic counselling that 54.4% of African women and 96.7% of the Caucasian women knew about Down syndrome. However, this study was done at the time when there was an extensive education drive in Pretoria to educate the public on Down syndrome. Schön (2004) in a study assessing missed opportunities for AMA genetic counselling found that 31% of African women of advanced maternal age presenting at an academic clinic knew about Down syndrome. This figure is comparable to the knowledge of Down syndrome in mothers in this study.

Christianson *et al* (1995) proposed possible reasons for the difference in knowledge between Caucasian and African South Africans. Down syndrome was previously considered by the medical profession to be rare in the African population in sub-Saharan Africa, including in South Africa. This has since been proven erroneous. Christianson *et al* (1995) documented that it was more difficult to recognize Down syndrome in African neonates, and Christianson & Kromberg (1996) stated that even after counselling African mothers did not recognize the differences in their newborns that had Down syndrome. In the African population, there is no word for Down syndrome and it was not recognized as an entity (Christianson 1996). Due to the significant mortality of Down syndrome infants between birth and 2 years, individuals with Down syndrome do not form a significant or recognisable group in the population due to this early mortality (Christianson 1996). Affected individuals in the African population are therefore grouped together with other intellectual disabilities and are not distinguished specifically or individually as Down syndrome.

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A contributory reason may also be that as Down syndrome was thought to be rare in Africans, doctors did not and still do not refer African women who are of AMA for genetic counselling. These women are therefore not being educated about Down syndrome and the community remains unaware (Kromberg *et al* 1992). The Caucasian population has been made more aware of Down syndrome by public education, information from literature and because of their access to more comprehensive health care in their medical insurance based health care system.

The researcher determined whether the knowledge of women who have children with Down syndrome was influenced by education level (figure 3.3.2). Of the women who had no education, none of them knew what Down syndrome was prior to the diagnosis of their child; 1 of the 11 women who had less than 7 years of schooling knew what Down syndrome was, whereas 6 of the 21 women with 7-12 years schooling, and 10 of the 13 women with tertiary education knew what Down syndrome was.

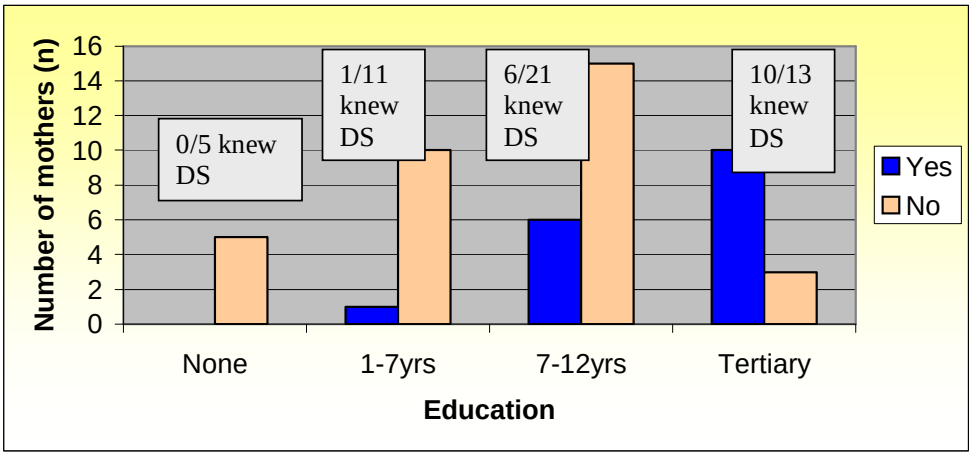

FIGURE 3.3.2: Did education level influence knowledge of Down syndrome prior to birth of child

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The Pearson Chi-square for the data in figure 1.6 of 16.57 and subsequent p-value of 0.00087 was significant evidence that education level influenced knowledge regarding Down syndrome.

Of the 13 women who had tertiary education, 8 were Caucasian (61.5%). Three of those with tertiary education (two African and one Caucasian) did not know what Down syndrome was prior to the birth of their child with Down syndrome.

Pelser (1998) also documented that knowledge was dependant on the level of education in women of advanced maternal age when questioned about their knowledge of Down syndrome. Of the women with less than 10 years education, 42.42% knew what Down syndrome was, whereas 88% and 100% of women with 10-12 years education and over 12 years education respectively knew Down syndrome.

3.4 INFORMATION ON THE PREGNANCY WITH THE CHILD WITH DOWN SYNDROME

At the time of the pregnancy with the child with Down syndrome, 21 (42%) of the women were of advanced maternal age (AMA). Of these women, 3 were Caucasian (30% of Caucasian group) and 18 were African (50% of African group). The maternal ages at the time of the birth of their child with Down syndrome ranged from 18 years to 45 years (Table 3.4.1), with a mean age at the time of pregnancy of 33 years.

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TABLE 3.4.1: Maternal Age Distribution at Time of Birth of child with Down syndrome

AGE (years)	African	Caucasian	Indian	TOTAL No.	Percent (%) of total
Under 20			1	1	2
20-24	2	2		4	8
25-29	6	1	2	9	18
30-34	9	4	1	14	28
35-39	12	2		14	28
40-44	6	1		7	14
45-49	1			1	2

Statistics South Africa (2004) reported the recorded live births according to the age of the mother. This is shown in comparison to the maternal ages of the mothers of this study in Table 3.4.2, as well as in Figure 3.4.1. These show that there is a greater proportion of older mothers who have children with Down syndrome in comparison to the general population.

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TABLE 3.4.2: Maternal age at time of birth of child with DS compared to maternal age distribution of general population

AGE (years)	Percent (%) of mothers with children with DS	Percent (%) of mothers of general population
Under 20	2	16
20-24	8	26
25-29	18	22
30-34	28	18
35-39	28	11
40-44	14	5
45-54	2	1
Other & Unspecified	0	1

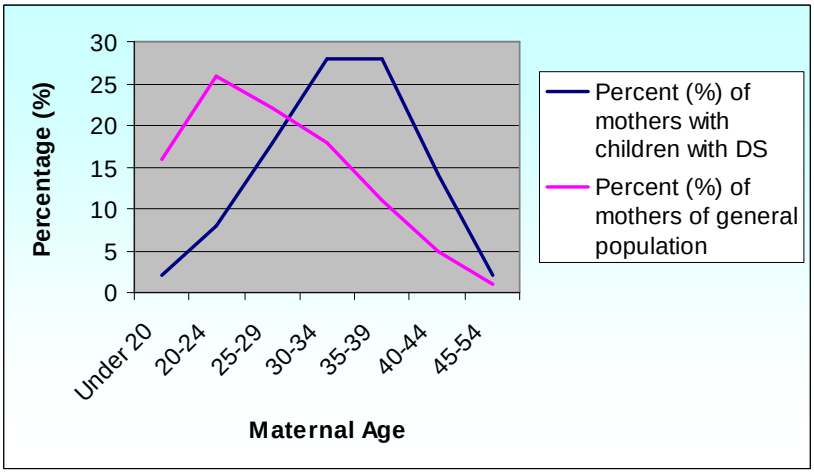

FIGURE 3.4.1: The distribution of maternal ages in the mothers who have children with Down syndrome compared to the maternal ages in the general population

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This is in keeping with the three prospective studies in South Africa in which the Down syndrome birth prevalence in African newborns was shown to be as high as or higher than that occurring in populations in industrialized nations (Christianson 1996). These studies showed that 52-56% of infants with Down syndrome are born to mothers of advanced maternal age (Kromberg *et al* 1992).

The prenatal diagnostic technique performed to diagnose chromosome abnormalities specifically Down syndrome in pregnancies of women who are of advanced maternal age is usually amniocentesis (Harper 2004). This has been available to a limited extent in the public health sector and more accessibly in the private health sector in South Africa since the late 1970's (Kromberg *et al* 1989).

It has been suggested in studies from developed countries, that one of the reasons women did not have amniocentesis performed was due to lack of education (Epstein *et al* 1972, Shino & Kellogg 1977). Kromberg *et al* (1992) documented that the majority of African women were unaware of the existence of prenatal amniotic services and that only 5% of amniocenteses performed in Johannesburg in 1990 were on African women, who comprise 90% of the pregnant population. Schön (2004) found that only 12% (9 out of 70) of African women were aware of prenatal diagnosis. She also documented that a majority of AMA women in her study (70%) were not referred for genetic counselling when their pregnancy was initially confirmed while a minority presented too late in their pregnancy to be offered genetic counselling and invasive testing.

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In the present study, none of the women had had prenatal diagnosis during the pregnancy that resulted in the birth of a child with Down syndrome. In question number 21 (Appendix A) asking about whether or not they would have opted for prenatal diagnosis had it been offered to them, 70% of the mothers answered in the affirmative. Of the African women 29 (81% of the African population) answered yes, 6 (17% of African population) answered no and 1 (2%) answered maybe to this question. Of the Caucasian women, 5 (50%) answered yes, 4 (40%) answered no and 1 (10%) answered maybe to the question (figure 3.4.2).

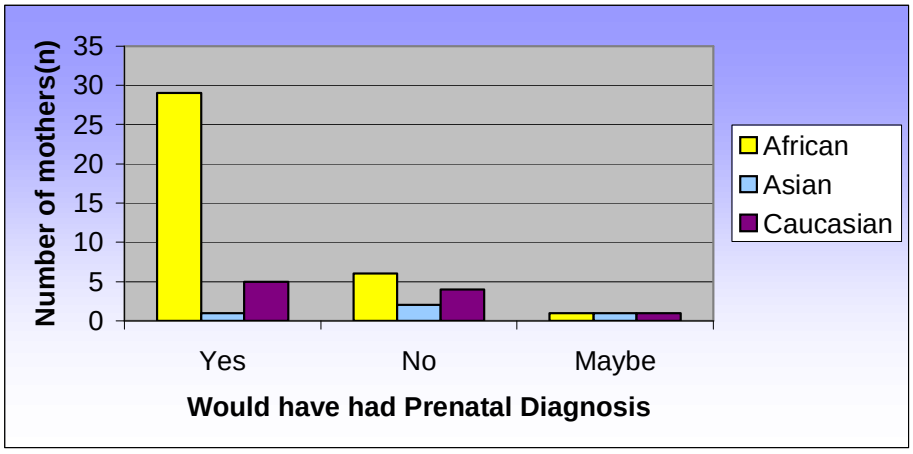

FIGURE 3.4.2: Would mothers have opted to have prenatal diagnosis had they been offered

The Pearson Chi-square statistic for this data was 3.89 (df=2 p=0.14), which indicated that the decision making process of all women with respect to their decision to accept or reject prenatal diagnosis was independent of ethnicity (p-value > 0.05).

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For comparison, in a previous study, from a sample of African women in South Africa with Down syndrome infants, none of whom had been offered prenatal diagnosis during pregnancy, 73% would have asked for amniocentesis if it had been offered (Kromberg *et al* 1992). In another South African study at the Groote Schuur Hospital, Cape Town, 316 women, from mixed ancestry, and black and white population groups, were offered amniocentesis, and 75.9% accepted to have the procedure (Viljoen, Oosthuizen & van der Westhuizen 1996). Pelser (1998) also documented that 89.13% (41) of African women and 93.3% (28) of Caucasian women in the study opted to have amniocentesis performed based on their risks related to advanced maternal age.

3.4.1 Information on child with Down syndrome

All the children with Down syndrome were still alive at the time of the interview, with their ages ranging from 1 year to 32 years of age, with a mean age of 4.6 years. Most of the mothers that were interviewed were attending or had attended clinics because of their child with Down syndrome. Death amongst children with Down syndrome is common (discussed in section 1.1.4) but only mothers with live children with Down syndrome were interviewed. It is a limitation of the study and possibly a more comprehensive conclusion could be made on the topic if the opinion of mothers who had children with Down syndrome that had died was obtained. However, getting access to these mothers would be difficult and it would be traumatic for the mothers to be interviewed on the subject.

The ages of the African children with Down syndrome ranged from 1 to 10 years with a mean age of 3.2 years. The Caucasian children with Down syndrome were between 2 and

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32 years of age and the mean age was 10.5 years. In the Caucasian population, when excluding the two older individuals of 22 and 32 years, the mean age drops to 5.8 years. This indicates that in both populations, the children were of an age that ensured most of the mothers had to have confronted reproductive issues and made decisions.

3.5 REPRODUCTIVE CHOICES OF WOMEN AFTER HAVING A CHILD WITH DOWN SYNDROME

After the birth of their child with Down syndrome, the majority of the women did not have further children. However, there was a significant difference between the African and Caucasian groups. Almost all (35 out of 36 or 97%) of the African women had not had children after their child with Down syndrome was born, whereas 5 (50%) of the Caucasian women did have more children (figure 3.5.1). The Pearson Chi-square statistic for this data was 15.88 (df=2 p=0.00036), which indicates that there is a significant difference between the two groups.

This may be explained by the fact that more of the African women were of advanced maternal age and were possibly not going to have more children regardless of whether the child had Down syndrome or not. It may also be explained by the fact that more of the Caucasian women were aware of the risks for their next pregnancies (in most cases, the recurrence risk was considered to be low, i.e. 1%) as well as the options available for them with regards to prenatal testing (see figure 1.10). Mgone (1982) noted in a study of African mothers with children with Down syndrome that their reproductive attitudes and behaviour were unchanged after the birth of their child with Down syndrome. The mothers who did not want to have further children gave the reason that it was due to their advanced maternal

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age rather than their Down syndrome infant. Christianson (1996) noted that the mothers may have given this reason due to the lack of awareness of the aetiology, features and prognosis of Down syndrome. That African women in the study did not have further children may indicate that with counselling and information, they decided against having future children.

The birth order of when the child with Down syndrome was born in comparison to siblings was determined. In the African population, 26 of the 36 children (72%) were third or higher in the birth order, but only 1 (10%) in the Caucasian population was third in the birth order. In the African population, 11 (30.6%) were fourth or higher in the birth order. The average was 1.6 in the Caucasian population whereas in the African population, the average was 3.14, and 3 in the Asian population. Christianson *et al* (1996) reported that 60% of children with Down syndrome, born to African mothers in South Africa, were fourth or higher in the birth order and Adeyokunnu (1982) noted this in 76% of his cases.

Only 7 (14%) of the women (5 Caucasian women, 1 African woman and 1 Asian woman) had subsequent children after their child with Down syndrome was born. Subsequent to their child with Down syndrome, five of these women had one child and two had two children each

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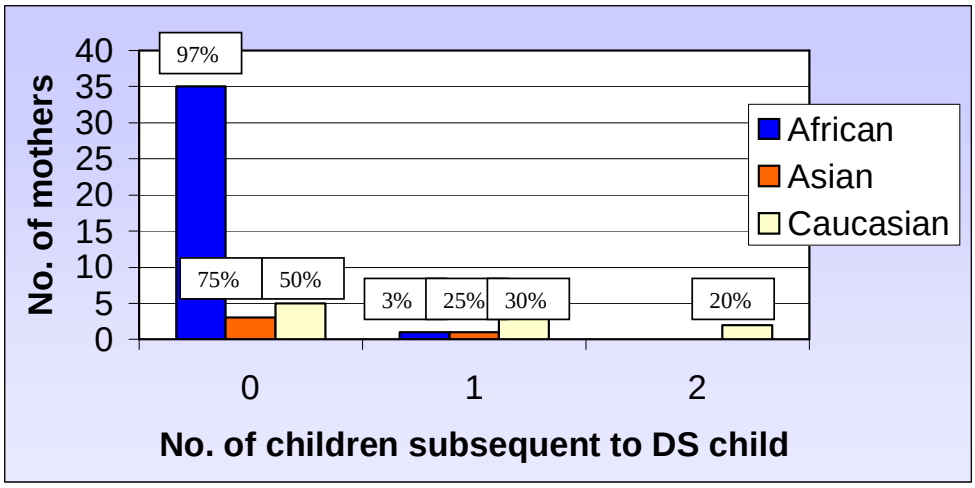

FIGURE 3.5.1: Number of children that mothers had subsequent to their child with Down syndrome

3.6 GENETIC COUNSELLING

Once the child had been diagnosed with Down syndrome, 38 (76%) of the mothers received some form of counselling from a nurse, doctor or genetic counsellor, and 12 (24%) did not (figure 3.6.1). It was not possible however, to ascertain as to how comprehensive it was, or to establish the specific details of the counselling.

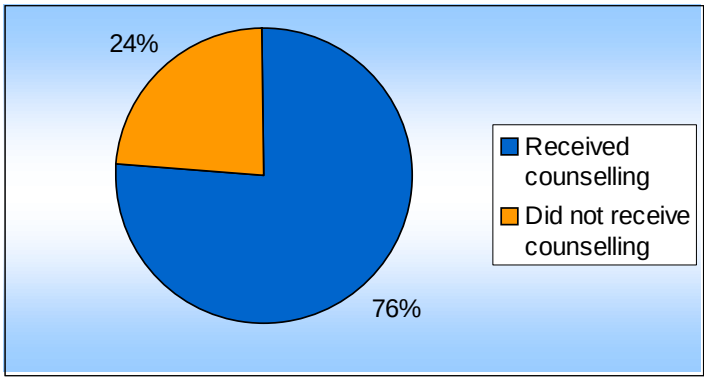

FIGURE 3.6.1: Did mothers receive genetic counselling

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Comparing the ethnic groups, the majority, 90% (9) of the Caucasian women received counselling, compared to 72% (26) of the African women and 75% (3) of the Asian women (figure 3.6.2).

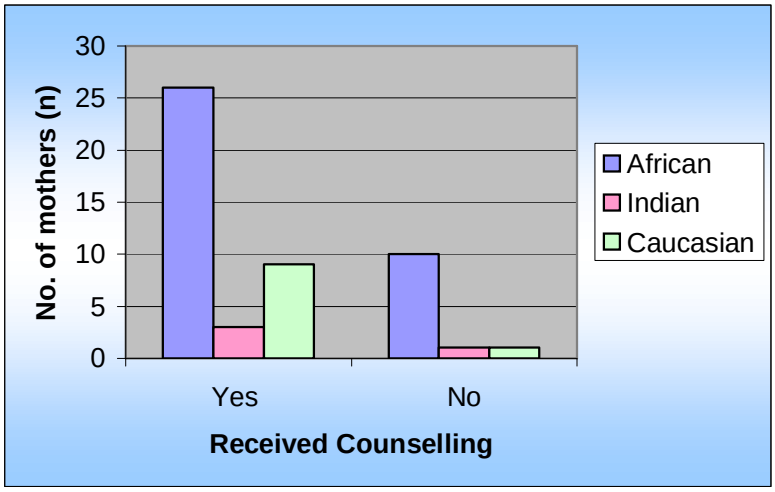

FIGURE 3.6.2: Distribution of mothers who received counselling among ethnic groups

These results differ from the cohort of African mothers seen by Mgone (1982). He found in the mothers in his study whose reproductive behaviour had remained unchanged after the birth of their child with Down syndrome that none of them had received any counselling for Down syndrome, whereas 76% of the mothers interviewed in this study received counselling and few (16%) had further children.

If the sample group of mothers had received counselling, they were asked by whom they had been counselled. Most of the mothers, 22 (44%), were counselled by a doctor. Five of these mothers were unsure which type of doctor specifically, whereas 12 said they were

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counselled by a paediatrician, 3 by a gynaecologist and 1 by a geneticist. Ten mothers (20%) received counselling from a Genetic counsellor, 2 (4%) from a nurse, 3 (6%) from the Down Syndrome Association, and 1 (2%) from a social worker.

Of these women (n=38) who were counselled, 28 (73%) were told their risks in future pregnancies and 10 (26%) reported that they were not told risks. Of the African women with a child with Down syndrome, 21 (58%) said that they were told their risks in future pregnancies and 7 (70%) Caucasian women reported that they were told their risks. Counselling seemed to be variable. Only 12 (31.6% of the women counselled) women remembered their specific risk. One African woman said she was told her risk to have another baby with Down syndrome was 100% despite having two other children who do not have Down syndrome. Three Caucasian women said they were told that their risk was 50%, 4% and 25% respectively. The rest of the women said that they were told their risks were 1% or less. The researcher feels that these women may not have been correctly counselled, as some of the risks seem very high, or considers that some of the women did not recall the information accurately.

With regards to future pregnancies, 29 (58%) of the mothers were advised about the option of prenatal diagnosis and 17 (34%) of the mothers were not. The option was not applicable to 4 (8%) of the women as they were not going to have more children, as 3 had undergone tubal ligation and 1 woman stated that she was not sexually active.

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3.7 FAMILY PLANNING

When the mothers were asked whether they were using any contraception after the birth of their child with Down syndrome, 12 (24%) said they had not used or were not using contraception, although the majority, 38 (76%) of the mothers had used and were still using contraception. These figures include the women who had tubal ligation and the one who said she was not sexually active.

The contraceptive prevalence in South Africa was found to be 56% (UNICEF 2004). This is substantially less than the 76% of women who have children with Down syndrome using contraceptives, which possibly indicates that the majority of women did not want to have further children after a child with Down syndrome, and implies that their reproductive behaviour changed subsequently. This change occurred subsequently to the genetic counselling received by these mothers.

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3.8 INFORMATION ON PREGNANCIES SUBSEQUENT TO THE BIRTH OF THE CHILD WITH DOWN SYNDROME

Amongst the 7 (14%) mothers who did have children subsequently to their Down syndrome child, there were 8 pregnancies altogether. Of these pregnancies, 6 (75%) were planned and two (25%) were unplanned. Five of these women said that the pregnancies were planned; one said that it was unplanned and one woman said that one of her pregnancies was planned and one was unplanned. Of these women, 5 were Caucasian, one was Indian and one was African. Table 3.8.1 summarises the prenatal testing during the subsequent pregnancies.

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TABLE 3.8.1: Prenatal testing in pregnancies subsequent to DS child

			SCREENING TESTS			**DIAGNOSTIC TESTS**	
MOTHER	RECEIVED COUNSELLING	PLANNED PREGNANCY	ULTRASOUND	NT	TRIPLE TEST	AMNIOCENTESIS	CVS
1	Y	One: Y Two: N	Y Y	N N	Y Y	Y Y	N N
2	Y	Y	Y	Y	Y	N	N
3	Y	N	Y	Y	Y	Y	N
4	N	Y	Y	Y	N	N	N
5	Y	Y	Y	Y	Y	N	N
6	Y	Y	N	N	N	Y	N
7	Y	Y	Y	N	N	Y	N

KEY: Y= Yes; N= No

Six of the 7 mothers had counselling after their child with Down syndrome was born. With regard to the 8 subsequent pregnancies, 7 ultrasound examinations were performed and one woman, although she did not have ultrasound, had an amniocentesis in the 1970's. All mothers thus had some form of prenatal screening or diagnosis. In the 7 pregnancies where women had ultrasound, at least one other prenatal test was performed: 4 nuchal translucencies (see glossary), 5 triple tests (see glossary), and 5 amniocenteses were performed. The results of prenatal testing were all normal and all the women continued with their pregnancies.

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All 7 women wanted to have some form of prenatal testing in their pregnancies after their child with Down syndrome was born and in 5 of the 8 pregnancies (62.5%) they opted for invasive testing. Of the 3 women who did not have amniocentesis, all had other prenatal testing and 2 received genetic counselling, suggesting that they had made an informed decision not to have amniocentesis performed.

This is a further indication that the counselling that the women interviewed had received after the birth of their child with Down syndrome had impacted on their future reproductive behaviour.

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3.9 WOULD WOMEN WANT PRENATAL DIAGNOSIS IN FUTURE PREGNANCIES

The women were asked whether they would want to have prenatal diagnosis (PD) in future pregnancies. Of all the women interviewed, 38 (76%) said that they would have prenatal diagnosis in any future pregnancies, 7 (14%) said they would not, and 5 (10%) said they were unsure whether they would have prenatal diagnosis (figure 3.9.1).

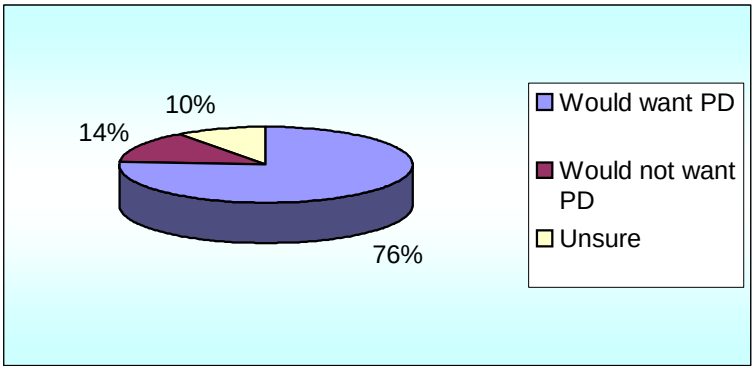

FIGURE 3.9.1: Would mothers want prenatal diagnosis in future pregnancies

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These data confirm that the majority of women would opt to have prenatal diagnosis. Of the Caucasian women, 80% (8) would want prenatal diagnosis in future pregnancies and 20% (2) would not. Similarly, 80.6% (29) of the African women would want prenatal diagnosis, 8.3% (3) would not and 12.1% (4) were unsure. Of the Asian women, 25% (1) answered yes, 50% (2) answered no and 25% (1) were unsure when questioned whether they would want prenatal diagnosis in future pregnancies.

Prenatal diagnostic techniques, such as amniocentesis and ultrasonography, are available to women in the private health care sector and at selected academic and state hospitals for the women who cannot afford private fees. Pelser (1998) interviewed women who were of AMA and found that 90% of Caucasian women, but only 52% of African women, were aware that prenatal diagnostic tests were available to them. She proposed that the lack of awareness and underutilization of services in South Africa could be due to:

- Lack of availability of prenatal tests
- Limited knowledge regarding prenatal diagnostic services
- Medical personnel failing to inform women regarding the tests
- DS and the risk of AMA previously being unrecognized by African women (Christianson 1996)

This highlights the need to increase awareness of prenatal diagnostic services, as well as referrals from practitioners, as many women would opt for testing. Pelser (1998) reported that in Pretoria a large number of women who obtained prenatal care at peripheral clinics were not counselled about advanced maternal age and their risks and thus missed the

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opportunity for genetic counselling and prenatal diagnosis. Many women also presented too late in pregnancy to be offered amniocentesis. Public education regarding advanced maternal age, the associated risks and the availability of genetic counselling is therefore essential. Many of the women who do book in early are not referred. Schön (2004), in a study, "Missed opportunities for prenatal diagnosis in women of AMA", found that 70% of these women presented for pregnancy confirmation early enough to be offered genetic counselling and prenatal testing. The average gestational age, at which the pregnancy of these women was confirmed, was 12.8 weeks gestation, yet none were appropriately referred for genetic counselling or given the option of prenatal diagnosis. Medical and nursing staff in primary, secondary and tertiary hospitals should be aware of the availability of amniocentesis so as to increase the number of women referred early in pregnancy for genetic counselling and to be given the option of amniocentesis. The public and medical personnel therefore should be informed about the risks of AMA and the availability of prenatal diagnosis, made aware of women at risk and as to which women should be referred for these tests.

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3.10 WOULD WOMAN CONSIDER TOP IF FETUS FOUND TO HAVE DOWN SYNDROME

Of the 50 women interviewed, 21 (42%) said they would terminate a pregnancy (TOP) if Down syndrome were to be detected in a future pregnancy, 26 (52%) said they would not, and 3 (6%) said they were unsure what they would do if faced with this problem (figure 3.10.1).

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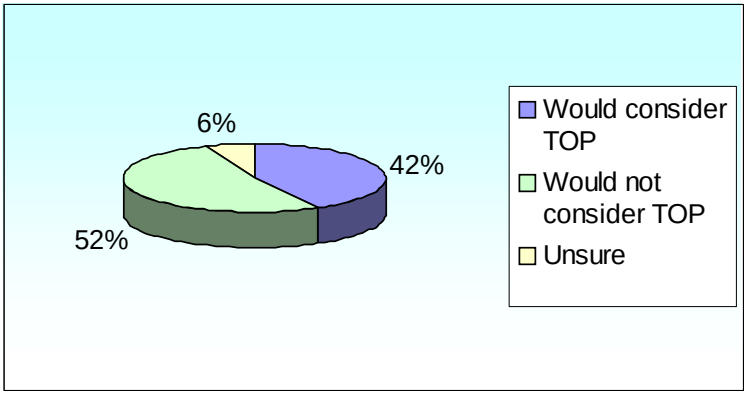

FIGURE 3.10.1: Mothers who would consider TOP in a subsequent pregnancy if fetus found to have Down syndrome

Of the Caucasian women, 40% (4) said they would opt for TOP, 40% (4) said they would not and 20% (2) were unsure. Of the African women, 52.7% (19) said they would not terminate an affected fetus, 44.4% (16) said that they would and one (2.8%) woman was unsure. Of the Asian women, 75% (3) said that they would not terminate an affected fetus and one (25%) said she would.

In a previous study, from a sample of African women in South Africa with Down syndrome infants, 52% would have terminated the pregnancy if prenatal diagnosis had been offered to them and showed an abnormal result (Kromberg *et al* 1992).

In a survey by Elkins *et al* (1986), mothers of children with Down syndrome in an American population were questioned concerning their attitudes with respect to prenatal diagnosis and TOP. The sample consisted of 101 women of whom 40 had children subsequently to their child with Down syndrome. Of these women, 50% had amniocentesis

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in subsequent pregnancies and only half of these women (25%) said they would terminate the pregnancy if Down syndrome were confirmed.

Figure 3.10.2 represents the comparison between the Caucasian and African women.

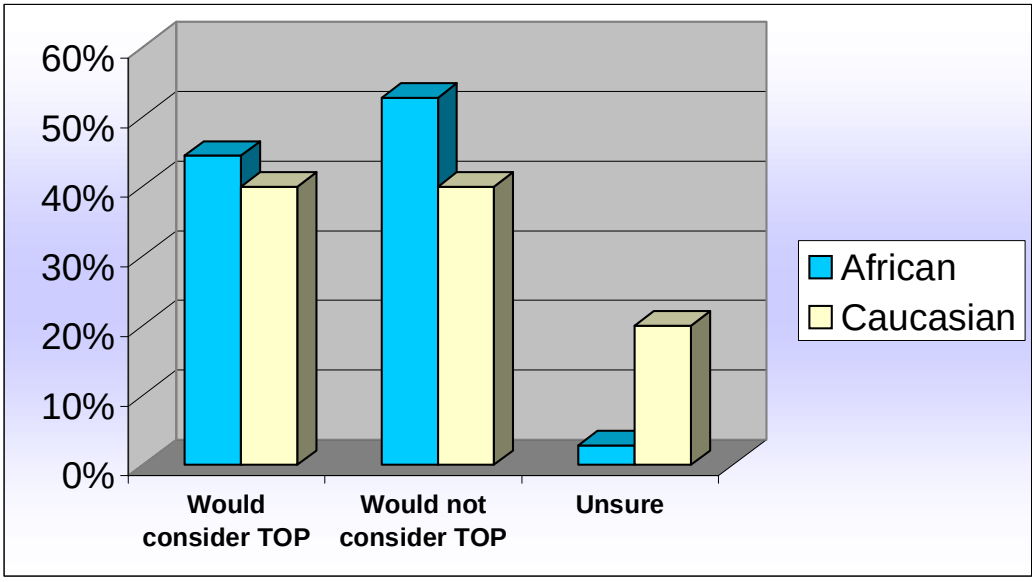

FIGURE 3.10.2: Comparison of Caucasian and African mothers' choice with regards to TOP of a fetus with Down syndrome

The Pearson Chi-square statistic for these data was 3.85 (df=2 p=0.15), which indicated that the decision making process of all women was independent of ethnicity (p-value > 0.05). These results compare favourably with other studies; Pelser (1998) concluded that acceptance of TOP, after non-directive counselling for a birth defect, was not influenced by ethnicity and Viljoen *et al* (1996) documented that ethnicity, education and socio-economic

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class did not influence woman's acceptance of prenatal diagnosis, but that religion influenced decisions regarding amniocentesis and TOP.

It is interesting to note that a recent study on South African women revealed that 80% of Caucasian women would consider TOP for a severe anomaly compared to 63% of African women. The overall acceptance rate of TOP for a birth defect anomaly was 69.7% (Pelser 1998). Similarly, Viljoen *et al* (1996) documented that of 466 women, from African, Caucasian and mixed ancestry, the overall TOP acceptance rate was 76.3%. Although these figures are much higher than the figures for women who have had a child with Down syndrome, all severe congenital abnormalities are included, and the study did not necessarily include women who previously had a child with Down syndrome.

In a study on women with Down syndrome children of primary school age, results indicated that negative feelings that they had felt initially at the birth of the child with Down syndrome had almost invariably changed in a positive direction and the experience of depression or anxiety among these mothers was uncommon. The families were described as enmeshed and controlled, and the experience of the family situation was generally positive (Ryde-Brandt 1988). This may explain why only a limited number of mothers in the present study would consider TOP of a fetus with Down syndrome in a future pregnancy.

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3.11 LIMITATIONS OF THE STUDY

The researcher recognized the following limitations of the study:

- The sample in this study does not necessarily represent the attitudes of all women with children with Down syndrome in the South African population as only mothers who currently live in Johannesburg, Gauteng were interviewed.
- The ages of the children with Down syndrome in this study ranged from 1 to 32 years and may present a limitation in that mothers who have very young children may not have experienced the same long-term complications as mothers of older children with Down syndrome.
- Responses from mothers whose first language differed from that of the interviewer may not have been accurately interpreted either by the interpreter or by the interviewer.
- Women may not have been completely comfortable to discuss sensitive issues such as their attitudes towards prenatal testing and TOP. This may have biased the results.

3.12 RECOMMENDATIONS

The following recommendations can be made from the findings of this study:

- Further research in other areas of Gauteng and in other provinces of South Africa could provide more in-depth insight into the reproductive choices of women with children with Down syndrome.

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- Doctors, nursing staff and the lay public should be made aware with respect to risks associated with advanced maternal age, genetic counselling services and the reproductive options available to women.

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CHAPTER 4

CONCLUSION

This study documented the reproductive choices made by South African mothers who have children with Down syndrome (DS). The reproductive decisions with respect to future family planning, prenatal diagnosis and termination of pregnancy, were assessed by using a questionnaire administered by the researcher to the women. The analysis of the questionnaire was performed with respect to ethnicity, education level and socio-economic status.

The information gained from the interviews showed that only one third of mothers knew about Down syndrome before their child with DS was born. Of these mothers, most had only a very basic knowledge and did not understand the severity and prognosis of the disorder. However most of the women received early counselling upon the diagnosis of Down syndrome in their child and those who did not, received counselling subsequently as they were attending DS clinics. After counselling, almost all of the African women stopped having further children while half of the Caucasian women had further children. Of the women who did have further children, all of them had some form of prenatal testing.

These results correlate significantly to those of other studies confirming that:

1) African women do not know about Down syndrome or are aware of the risks to women of advanced maternal age of having a baby with Down syndrome (Pelser 1998; Schön 2004).

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2) Most African women of advanced maternal age are not being offered genetic counselling and prenatal diagnosis (Kromberg *et al* 1992; Pelser 1998; Schön 2004).

However, after the birth of a child with Down syndrome, many South African women in this study received some counselling that seemed to impact on their subsequent reproductive behaviour. After the birth of their child with DS the majority of women in this study used contraception, and only few have had subsequent children. This is in contrast to Mgone's (1982) findings that the reproductive behaviour of Tanzanian women remained unchanged after the birth of a child with Down syndrome. These women had however not received counselling. The women who did have children subsequent to the birth of their child with Down syndrome all had some form of prenatal screening or diagnosis. The behaviour of women in this study is more similar to that of women in industrialised countries, such as Japan and America (Ando 1978; Elkins *et al* 1986).

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This study showed that in Johannesburg postnatal counselling to women who had a child with Down syndrome appears to have changed their subsequent reproductive behaviour. Genetic counselling for DS aims to inform individuals and families of the nature of Down syndrome, the probability of having children with Down syndrome in future pregnancies and the options available in which this can be detected prenatally and prevented (Harper 2004). The results documented in this study indicate that the majority of South African mothers of children with DS living in Gauteng are receiving counselling that impacts their

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future reproductive behaviour. This confirms the need for parents of infants and children with Down syndrome to be referred and given genetic counselling.

The researcher considered it important to document this information to assist in the understanding of mothers and families that receive genetic counselling for Down syndrome and thus enhance and give increased insight to the Genetic Counselling services offered by the Department of Human Genetics, University of the Witwatersrand & the National Health Laboratory Service in South Africa.

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CHAPTER 5

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APPENDIX A

QUESTIONNAIRE

Research No:_____

General Information:

1. Age:_____
2. Home Language:_____
3. How many years of school did you complete?
   - None
   - 1-7
   - 7-12
   - Tertiary
4. Which province do you live in?_____
5. What area do you live in?
   - Urban
   - Peri-urban
   - Rural
6. Occupation:_____
7. Husband's occupation:_____
8. What is your household salary?
   - 0 - R1000
   - R1000 - R5000
   - R5000 - R10 000
   - R10 000 - R20 000
   - Over R20 000

Previous Pregnancy History:

9. Para (how many deliveries you have had):_____
10. Gravida (how many pregnancies you have had):_____

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11. How many live children do you have?_____

12. Age of child with Down Syndrome:_____

13. Is this child still alive?

- ❑ Yes
- ❑ No

14. Did you know about Down syndrome before the birth of the affected child?

- ❑ Yes
- ❑ No

15. If yes, what did you know?_____

16. How many children have you had subsequent to your child with Down Syndrome?_____

17. Did you have prenatal diagnosis during the pregnancy with your child with Down syndrome?

- ❑ Yes
- ❑ No

18. If yes, why?

- ❑ Because of an increased risk due to AMA
- ❑ Because of an increased risk from prenatal screening
- ❑ Because of an increased risk due to previous child with Down syndrome

19. Were there any abnormal findings after prenatal diagnosis?

- ❑ Yes
- ❑ No

20. If yes, what were the findings?_____

21. If you did not have prenatal diagnosis, would you have chosen to have prenatal diagnosis if you knew you had an increased risk for Down syndrome?

- ❑ Yes
- ❑ No
- ❑ Maybe

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22. After the diagnosis of your child with Down syndrome, did you receive counselling?

- ❑ Yes
- ❑ No

23. If yes, who counselled you about your child with Down syndrome?

- ❑ Nurse
- ❑ Doctor
  a) Paediatrician
  b) Gynaecologist
  c) General Practitioner
  d) Geneticist
- ❑ Genetic Counsellor

24. During counselling for another child with Down Syndrome, were you told about your risks in future pregnancies?

- ❑ Yes
- ❑ No

25. If yes, what were the risks you were given?

- ❑ Increased
- ❑ Decreased
- ❑ Specific figure:_____

26. Were you advised about the option of prenatal diagnosis in future pregnancies?

- ❑ Yes
- ❑ No

27. Did you use contraception after the birth of your child with Down syndrome?

- ❑ Yes
- ❑ No

28. Were your pregnancies after your child with Down syndrome planned?

- ❑ Yes
- ❑ No

29. If yes, what were you advised?_____

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30. Did you have prenatal diagnosis in any of the pregnancies after the birth of your child with Down syndrome?

- ❑ Yes
- ❑ No

31. If yes, what prenatal diagnosis?

- ❑ Ultrasound
- ❑ Nuchal Translucency
- ❑ Triple Test
- ❑ Amniocentesis
- ❑ CVS

32. What were the results of the prenatal diagnosis?_____

33. If an abnormality was found, what was the diagnosis?_____

34. Did you:

- ❑ Continue with the pregnancy
- ❑ Have a TOP
- ❑ I miscarried

35. In any future pregnancies, would you want to have prenatal diagnosis?

- ❑ Yes
- ❑ No
- ❑ Not sure

36. In subsequent pregnancies, would you consider (or would you have considered) termination of pregnancy if it was discovered that the fetus had Down syndrome?

- ❑ Yes
- ❑ No
- ❑ Not sure

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APPENDIX B

PATIENT INFORMATION SHEET

Research Title: **THE REPRODUCTIVE CHOICES MADE BY SOUTH AFRICAN MOTHERS WHO HAVE CHILDREN WITH DOWN SYNDROME**

Researcher: Julie Lampret
MSc(Med) Genetic Counselling, 2nd year
Department of Human Genetics, University of the Witwatersrand

Supervisor: Professor AL Christianson, FRCP Ed
Department of Human Genetics, NHLS
University of the Witwatersrand (011) 489-9223

I am an MSc(Med) student at the Department of Human Genetics of the National Health Laboratory Service, School of Pathology, University of the Witwatersrand. I am conducting a study to determine what reproductive decisions are made by South African women, who have previously had a child with Down syndrome.

As a mother of a child with Down syndrome, we would appreciate your help and participation in this study. This would assist us in determining what reproductive decisions (choices made during pregnancy) are made by women like yourself with regard to family planning (e.g. contraception) and prenatal diagnosis (diagnosis of the baby during pregnancy). You will therefore be asked questions relating to the choices you made in any pregnancies before or after your child with Down syndrome was born. This will help us improve our understanding of the psychological (emotional) and social factors surrounding Down syndrome.

If you agree to help with the study, you will be asked to answer a number of questions in an interview by myself, which should not take more than an hour of your time. The information you give will remain strictly confidential and your name will not appear on the questionnaire.

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The results of this study will be analysed anonymously and will be used to help us better understand how women and their families cope with children with Down syndrome. This may give us further insight into managing our services effectively.

If you choose not to participate, this will not affect the management or treatment of you or your child in any way. Your participation in this study is entirely voluntary and you are free to withdraw at any time.

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APPENDIX C

INFORMED CONSENT

I,..... consent to participate in a study which involves the completion of a questionnaire regarding my experiences as a mother of a child with Down Syndrome. I understand that the information will remain confidential, and is for research purposes. I understand that I may withdraw from this study without any negative impact on myself or my child.

Signature:.....

Date:.....

Phone Number:.....

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APPENDIX D

ETHICAL CLEARANCE CERTIFICATE

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