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Psycho-social Aspects of Turner Syndrome

A Qualitative Study

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**Dissertation submitted as part requirement for the
degree MMed (O&G) at the Faculty of Health Science
University of Cape Town**

DECLARATION

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ABSTRACT

INTRODUCTION:

The psychosocial aspects of Turner Syndrome have been mainly studied in developed countries but the impact of this syndrome in women in developing countries is unknown. The aims of the study were to explore the experiences of women affected by Turner Syndrome and living in an urban community in South Africa.

METHODS

A qualitative study, using semi structured in-depth interviews, of women with Turner Syndrome was conducted in order to evaluate their psychosocial experiences.

RESULTS

Thirteen women with Turner Syndrome were recruited for this study. The age of the women was between 19 and 45 years and the average height was 1,39meters. (range 1,22m — 1,52m)

Most of the participants had poor knowledge about their condition. Infertility was considered the most traumatic consequence of the syndrome by most of these women and a cause for low self image. Short stature was not associated with low self image for most of the women in this study. Interpersonal relationships were not negatively affected by having Turner Syndrome.

These women, however, displayed some evidence of delayed partner relationships as eight of them were still staying with their parents. Two were married and most of the respondents were not in a relationship at the time of the study. There was evidence of some cognitive deficits, as three of the participants had required special schooling, six repeated a grade and most had difficulties with mathematics. Eight of the thirteen women were employed, which suggested no negative impact of Turner Syndrome on employment and job opportunities. Most

of the participants felt that support was lacking from their doctors regarding the psychosocial impact of their medical condition.

CONCLUSION

The results of this study provide insights about the psychosocial impact of Turner Syndrome on women in our clinical setting. The study has highlighted some opportunities for interventions to improve the healthcare of these women. Medical care givers need to focus, not only on medical aspects of the condition, but also on the psychosocial aspects. Patients must be given adequate information about Turner Syndrome and its effects and their families must be included in the counselling. This may improve the quality of support we can offer these women. Feedback about the results of this study needs to be given to our paediatric unit. Psychosocial support for women with Turner Syndrome should be initiated during their childhood. Studies on the psychosocial impact of Turner Syndrome on women elsewhere in South Africa are needed. The results of this study may not reflect the experiences of women in other provinces or those living elsewhere in Africa.

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CHAPTER 1: INTRODUCTION AND LITERATURE REVIEW

1.1 Etiology and Incidence of Turner Syndrome.

Turner Syndrome is a genetic disorder first described by Henry Turner at the annual meeting of the Association for the Study of Internal Secretions in San Francisco, California, 1938 (Turner, 1938). Turner Syndrome occurs secondary, either to an abnormality, or the absence of one of the X chromosomes in all cell lines. Approximately 60% of affected patients have only a single X chromosome (45 XO) and the remainder have a structural abnormality of the X chromosome or a mosaicism with two or more cell lines such as (45 XO/46 XX) (Ross et al., 2000).

Table 1

| STRUCTURAL ABNORMALITIES OF THE X CHROMOSOME | |
|--|--|
| 46, X del [Xp] | Deletion of the short arm |
| 46, X del [Xq] | Deletion of the long arm |
| 46, Xr [X] | Ring Chromosome |
| 46, Xi [Xq] | Isochromosome with 2 long arms and missing the short arm |
| 46, Xi [Xp] | Isochromosome with 2 short arms and missing the long arm |

The structural abnormalities of the X chromosome include a deletion of the short arm (46, X del [Xp]) or long arm (46, X del [Xq]), a ring chromosome (46,Xr [X]), an isochromosome in which the chromosome consists of two long arms and is missing the short arm (46, Xi [Xq]) or two short arms and missing the long arm (46, Xi [Xp]) (Ross et al., 2000).

The incidence of Turner Syndrome is estimated to be 1 in 2500 female infants at birth. In contrast the prenatal incidence is thought to be about 2% of all human conceptions, but less than 1% of embryos survive to term (Kaneko et al., 1990; Nilsson et al., 1996; Pasquino et al., 1997; Belin et al., 1998; Rosenfeld et al., 1998; Ross et al., 2000).

1.2 Physical Characteristics of Turner Syndrome.

The physical characteristics of Turner Syndrome includes disorders of skeletal growth and features related to lymphatic obstruction as illustrated in a series of 165 patients with Turner Syndrome presented by Lippe in 1991. Some women may have only one of the many anomalies described, whereas others may have multiple features of the syndrome (Kaneko et al., 1990; Nilsson et al., 1996; Pasquino et al., 1997; Rosenfeld et al., 1998; Belin et al., 1998; Ross et al., 2000).

Skeletal abnormalities include short stature, short neck, cubitus valgus, short metacarpals and characteristic facies with micrognathia and high arched palate (Turner. 1938; 1998; Belin et al., 1998; Ross et al., 2000). Abnormal growth of the cranial base of the skull occurs in some women with Turner Syndrome and it results in the abnormal position of the external auditory meatus with an increased incidence of otitis media (Mc Cauley., 1990).

Physical features related to lymphatic obstruction include webbed neck, low posterior hairline, rotated ears, oedema of hands and feet and severe nail dysplasia (Turner. 1938; Belin et al., 1998; Ross et al., 2000). The endocrinological abnormalities in Turner Syndrome are the consequence of gonadal dysgenesis and gonadal failure, which result in failure of pubertal development and subsequent infertility (Turner. 1938; Belin et al., 1998; Ross et al., 2000).

Table 2

PHYSICAL CHARACTERISTICS OF TURNER SYNDROME

| SKELETAL ABNORMALITIES | ABNORMALITIES RELATED TO LYMPHATIC OBSTRUCTION |
|-------------------------------|---|
| Short stature | Webbed neck |
| Short neck | Low posterior hairline |
| Short metacarpals | Rotated ears |
| Cubitus vulgus | Oedema of hands |
| Micrognathia | Oedema of feet |
| High arched palate | Severe nail dysplasia |

Cardiac abnormalities, renal abnormalities and autoimmune disorders are also associated with Turner Syndrome. Reported cardiac abnormalities include coarctation of the aorta, bicuspid aortic valves, mitral valve prolapse and aortic aneurysm. Renal abnormalities include unilateral pelvic kidney, horseshoe kidney and partial or complete duplication of the collecting system. Autoimmune disorders associated with Turner Syndrome are Hashimoto's thyroiditis, Addison's disease, alopecia and vitiligo. (Turner. 1938; Belin et al., 1998; Ross et al., 2000).

Table 3

STRUCTURAL ANOMALIES

Cardiac Anomalies

Coarctation of the aorta
Bicuspid aortic valves
Mitral valve prolapse
Aortic Aneurysm

Renal Anomalies

Unilateral pelvic kidney
Horseshoe kidney
Partial or complete duplication of the collecting system

Autoimmune Disorders

Hashimoto's thyroiditis
Addison disease
Alopecia

In patients with 46 X, Xi [Xq]) karyotype, who have an isochromosome, in which the chromosome consists of two long arms and is missing the short arm, the complete clinical picture of Turner Syndrome is seen. In contrast patients with 46 X, Xi (Xp]) karyotype in whom the chromosome has two short arms and is missing a long arm, the stature is normal and the gonadal dysgenesis seems to be less severe. (Ostberg and Conway. 2003). Patients with a ring chromosome karyotype have a more severe phenotype than those with the other structural abnormalities of the X chromosome. (Ostberg and Conway. 2003).

Auto-immune disorders and hearing loss are more frequently found in patients with isochromosomes. (Kaneko et al.1990). Mosaicism generally results in a milder phenotype with up to 40% of the women entering puberty,

although premature ovarian failure always occurs. (Ostberg and Conway. 2003).

1.3 Neuro-developmental and cognitive aspects of Turner Syndrome.

Specific neuro-developmental and cognitive behavioural aspects of patients with Turner Syndrome have been identified. A review by Nijhuis-van der Sanden et al (2003) identified 385 publications on neuro-developmental and cognitive behavioural aspects of patients with Turner Syndrome published between the years 1962 — 2003.

The literature has been consistent about the deficiencies in visual-spatial skills, visual memory, attention and facial affect recognition in patients with Turner Syndrome (Turner., 1938; Reiss et al., 1995; Sarah et al., 1998; Ross et al., 2000; 2003; Nijhuis-van der Sanden et al., 2003; Ostberg and Conway. 2003). Lawrence et al (2003) demonstrated that women with Turner Syndrome showed impairment at recognising faces and reading facial expression of emotion, especially with regard to the expression of fear. Difficulties with directional sense, driving and multitasking have also been described in these women (Sybert and Mc Cauley., 2004).

Van Borsel et al (1999), in their survey of 128 girls with Turner Syndrome, in Belgium reported that almost a third had repeated a class at school with most of these young women having difficulties with mathematics. Difficulties with

mathematics were also evident in 22 girls with Turner Syndrome followed up for 3 years by Siegel et al (1998) in America.

Women with 45 XO karyotype appear to perform worse in cognitive and motor skills than women with mosaics. (Nilsson et al., 1996). Overt mental retardation is seen in Turner Syndrome women with a ring chromosome and this is thought to be due to failure of X inactivation, which occurs more commonly with a smaller X ring chromosome (Van Dyke et al., 1991; Borman et al., 1998; Ostberg and Conway., 2003).

Importantly, the parental origin of the X chromosome influences cognitive functioning. Women with Turner Syndrome who have a paternally derived X chromosome tend to have superior verbal and executive skills compared with those with a maternally derived X chromosome (Scourfield et al., 1997; Skuse et al., 1999; Skuse and Lawrence., 2005; Cutter et al., 2006). There appear to be one or more imprinted loci on the paternally derived X chromosome, which influence social functioning. Skuse et al (1999) studied 80 girls with Turner Syndrome at the Institute of Child Health (U.K), 55 with maternally derived X chromosome and 25 girls with paternally derived X chromosome. Girls with a paternally derived X chromosome had higher verbal intelligence and fewer social difficulties when compared with girls with a maternally derived X chromosome (Skuse et al. 1997).

Many studies have attempted to identify the actual brain area responsible for the cognitive deficits seen in Turner Syndrome. There seems to be evidence for abnormalities localised to the posterior part of the right hemisphere of the

brain (Reiss et al., 1995; Brown et al., 2002; Nijhuis van-den Sanden et al. 2003). In a study by Reiss et al (1995) at the Johns Hopkins University School of Medicine in Baltimore, which evaluated volumetric brain measures derived from Magnetic Resonance Imaging, significant differences in the grey and white matter ratios were seen exclusively in the right parietal regions of 30 girls with Turner Syndrome compared to 30 controls.

1.4 Psycho-social Function:

A wide range of psycho-social difficulties relating to Turner Syndrome are described in the literature with problems noted from early childhood to adulthood. Mathisen et al (1992) studied oral motor skills in ten babies with Turner Syndrome recruited from specialist clinics for children with growth problems in London. Findings revealed oral motor dysfunction with resultant feeding difficulties, stuttering and articulation and language problems in early childhood.

A sample survey by Van Borsel et al (1999) of 128 girls with Turner Syndrome done at the University of Gent in Belgium, also concluded that due to the presence of cranio-facial and oral abnormalities, difficulties with speech and language were apparent. Of the 128 girls studied, a quarter had received treatment for stuttering, articulation problems or delayed language.

Many studies, mainly done in Europe, have concluded that children with Turner Syndrome tend to have immature behaviour compared to their peers, are more withdrawn, have lower self esteem, have increased anxiety

problems and have a higher incidence of attention deficit hyperactivity disorder (Reiss et al., 1995; Siegel et al., 1998; Borman et al., 1998; Ross et al., 2000; Mc Cauley et al., 2001; Mc Cauley and Sybert., 2006).

Twenty two girls with Turner Syndrome were compared to 25 controls in a study by Siegel et al (1998) through the use of psychological tests. The study also included a Child Behaviour Checklist completed by the parent of each girl. The findings indicated that the girls with Turner Syndrome had fewer friends and were involved in fewer activities than the controls. Similar findings were reported by McCauley et al (2001). They compared Child Behaviour Checklist completed by mothers of 122 girls with Turner Syndrome versus those of 108 controls. The findings indicated that girls with Turner Syndrome had fewer friends and had difficulty socializing with other children compared to the controls. They also reported a higher incidence of attention deficit hyperactivity disorder in the Turner Syndrome group compared to the control group.

McCauley and Sybert (2006) in their review on social and behavioural development of girls and women with Turner Syndrome suggest that the social and behavioural problems tend to affect girls with Turner Syndrome but that adult women with Turner Syndrome are emotionally stable and live self- sufficient lives.

A study of 22 adult women with Turner Syndrome which evaluated, through the use of semi- structured interviews, how the condition affected their lives and coping style, was conducted in Sweden. This study showed that while

social isolation was a feature in adolescent girls with Turner Syndrome, none of the adult women felt they were socially isolated. (Sylven et al., 1993)

The neuro-behavioural and cognitive deficiencies in girls with Turner Syndrome have been noted to cause psychosocial problems in adult women. The first studies of the personality of girls and women with Turner Syndrome were performed in the 1970's. Money et al (1970) conducted a study of seventy three patients with Turner Syndrome. They evaluated the impact that physical appearance and parental support had on psychosocial functioning of these women. They showed that these women had a high stress tolerance, meaning that they had an ability to adapt to life much better than might be expected. They reported that psychopathology was not a significant feature in patients with Turner Syndrome, despite the short stature and physical deformities associated with the condition. They also concluded, however, that lack of parental support was associated with poor psychosocial functioning in these women.

In several studies, mainly in Europe, women with Turner Syndrome have been described as being less likely to leave the parental home, to have less frequent or delayed partner relationships, to have a lower occupational status than would be expected for their level of education and a slightly increased rate of unemployment (Borman et al., 1998; Ross et al., 2000; Borman et al., 2001; Saenger et al., 2001; Lawrence et al., 2003; Ostberg and Conway., 2003., Mc Cauley and Sybert. 2006).

In contrast, some studies from Europe suggest that women with Turner Syndrome have excellent educational and occupational achievement. They do, however, document delays with adult development such as living independent of parents or marriage (McCauley and Sybert. 2006).

The social isolation among adults with Turner Syndrome is thought to be also linked to the high prevalence of deafness in these women (Conway., 2002; Serra et al. 2003).

1.5 Aetiology of the Impaired Psychosocial Function

Premature ovarian failure and infertility have been shown to be the main causes of inhibited social behaviour in these women. Cardoso et al (2004) evaluated the prevalence of mood and psychiatric syndromes in women with Turner Syndrome at the National Institution of Mental Health, Bethesda ,Maryland, USA . They concluded that the rates of depression were higher in these women, relative to community samples, but women with Turner Syndrome had similar rates of depression to women in gynaecology clinics. They also showed similar levels of shyness in women with Turner Syndrome and those with premature ovarian failure.

The physical characteristics of patients with Turner Syndrome, in particular short stature, have an impact on psychosocial functioning. In their longitudinal study of 47 American girls with short stature, 22 of whom had Turner Syndrome, Siegel et al (1998) found that girls with short stature had

fewer friends and engaged in fewer activities compared to 25 girls with normal stature for age.

A study by Van Pareren et al (2005) of 50 young women with Turner Syndrome who had achieved final heights within the normal range after treatment with growth hormone showed that girls with Turner syndrome had low self esteem compared to their peers despite having achieved heights within the normal range.

Havenkamp et al (2000) assessed the influence of short stature on self image of 50 women with Turner Syndrome at the University of Bonn, Germany. They concluded that height and physical appearance was associated with low self esteem but general satisfaction with life was mainly dependant on the support the women had received from their parents. In the Swedish study of 22 women, Sylven et al (1993) also found that ovarian failure and infertility, not short stature were mostly associated with depression in women with Turner Syndrome. (Sylven et al., 1993)

The role of the endocrine profile in psycho-social functioning in patients with Turner Syndrome is still unclear. There is a suggestion in the literature that adolescent girls with Turner Syndrome showed improvement in baseline self-esteem after being started on oestrogen treatment.

(Ross et al., 2000; Borman et al., 2001; Nijhuis-van der Sanden., 2003)

Ross et al suggest that deficits such as motor function, executive ability and memory appear to improve in Turner Syndrome patients treated with

oestrogen. (Ross et al., 2000) The mechanism of these oestrogen effects is not known and a cause and effect relationship has not been proven .

Environmental factors have also been shown to have an impact on psychosocial function of women with Turner Syndrome as many of these women experience overprotection by family or ridicule by peers. (Rickert et al., 1996)

In the developing world, due to lack of adequate health resources, the psycho-social aspects relating to reproductive health care are often neglected. The psychosocial impact of Turner Syndrome on women in South Africa or, indeed, elsewhere in Africa have not been previously explored. The aim of this study was to evaluate the psychosocial experiences related to Turner Syndrome in women living in Cape Town and attending the Gynaecological Endocrinology Clinic at Groote Schuur Hospital. It is anticipated that the results of this study will help inform the clinical management of these women.

CHAPTER 2: PARTICIPANT RECRUITMENT AND METHODS

2.1 Participants

Women with Turner Syndrome were identified from the records of the Gynaecological Endocrinology Clinic at Groote Schuur Hospital. Of a total of 26 women who were identified with Turner Syndrome, 9 could not be contacted as their telephone numbers were invalid and one of the women had died from Acquired Immune Deficiency Syndrome.

The remaining 16 women were contacted telephonically and informed about the study. Three of the 16 women declined to participate in of the study due to time constraints and work commitments. Dates and times were arranged for interviews with those women who consented to participate.

On arrival at the clinic the women were issued with an information leaflet containing the details of the study [See Appendix A]. All women were informed that declining to participate would not impact on their current or future medical care.

2.2 Setting

All interviews were conducted at the Gynaecological Endocrinology Clinic at Groote Schuur Hospital in a quiet and private room. Groote Schuur Hospital is a tertiary hospital and an academic institution attached to the University of Cape Town. Women are referred to this clinic from the general gynaecology clinic at Groote Schuur Hospital and from secondary and primary level hospitals. The team of doctors working at this clinic includes three reproductive medicine consultants and two registrars.

2.3 Data collection

Qualitative research methods were used. Semi-structured in-depth interviews were conducted. An interview guide was used to assist the researcher in covering the key areas of enquiry [See Appendix B]. The interview took the form of a conversation to allow the informants to discuss issues freely. These interviews lasted an average of forty five minutes to an hour and were tape recorded, with the consent of the informants. Interviews were conducted by the primary investigator in the language of choice of each informant - English, Afrikaans or Xhosa.

The key areas of enquiry included the following concepts:

- 1. The basic knowledge of the informants about Turner Syndrome and the impact of this knowledge on their self image.**
- 2. Experiences relating to interpersonal relationships with parents, siblings, extended family, friends, parents, and community members.**
- 3. Marital and sexual relationships**
- 4. Educational experiences**
- 5. Job experiences**
- 6. Support and coping mechanisms.**

2.4 Data analysis and interpretation

The tape recorded interviews were transcribed verbatim with the inclusion of nonverbal elements into written transcripts by the primary investigator. This gave the primary investigator the opportunity to listen to each interview and to identify recurring themes. The written transcripts were then analysed according to the principles of descriptive analysis.

Qualitative descriptive studies have the main aim of providing a comprehensive summary of events in the everyday terms of those events. Qualitative description is the method of choice when straight description of phenomena are desired. Basic qualitative description entails an interpretation of data that is very close to the data without much interference or overinterpretation of the data. This interpretation is more likely to result in easier consensus among researchers. The description in qualitative description involves the presentation of data in everyday language. The researchers stay closer to their data and to the surface of words and events (Sandelowski, 2000).

Qualitative content analysis is the analysis strategy of choice in qualitative descriptive studies. Qualitative content analysis is a dynamic form of analysis of verbal and visual data with the aim to summarize the informational contents of that data. Codes that are generated from the data during the course of the study are systematically applied to the data (Sandelowski, 2000).

Numerically defined verbal counting was used to express the results. This is the use of words such as most, many, some and few to express the results. In this study 'most' refers to nine or more participants, 'many' refers to more than seven but less than nine, 'some' refers to more than four but less than seven participants and 'few' refers to less than four participants.

2.5 Qualitative Research

Qualitative research is a method of research aimed at understanding human behaviour (Bergsjö, 1999). It is a useful method when quantitative measures cannot adequately describe or interpret a situation. In contrast to quantitative research, there is no hypothesis to be tested (Sandelowski, 1995). The data in qualitative research are collected by means of interviews, questionnaires or participant observation (Pope and Mays, 1995). Semi-structured interviews provide a general guide that is uniform enough to generate comparable responses but flexible enough to accommodate the variety of people encountered in the interview (Firestone, 1993).

The final sample size is determined during the data collection. New areas of enquiry may evolve during data collection. This can then guide further data collection and necessitate an increase in sample size (Bergsjö, 1999). Adequacy in sample size is achieved when additional interviews fail to provide fresh insights.

Data in qualitative research are collected until repetition of the information or until recurrent themes are identified (Sandelowski, 1995). This can result in a smaller sample size to what was originally planned. In quantitative research, analysis of data can only begin once all the data have been collected. In contrast, there is no clear distinction between data collection and data analysis in qualitative research (Sandelowsk, 1995). Qualitative content analysis is similarly reflexive and interactive as researchers continuously modify their treatment of data to accommodate new insights about those data (Sandelowski, 2000).

CHAPTER 3: RESULTS

3.1 Socio-Demographic Characteristics of the Participants

Sixteen women were invited to participate in the study. Three declined to be interviewed because of time constraints. Thirteen women were interviewed. Nine of the women interviewed were of mixed ancestry. Four were black African women. Twelve of the informants were South African citizens. One participant was from the Democratic Republic of Congo and she was in South Africa to seek medical assistance for her condition of Turner Syndrome. Eleven women belonged to the Christian faith and two of them to the Islamic faith.

The age of the informants was between 19 and 45 years. Eight of the women were below the age of 30 whilst five women were above the age of 30. The average height was 1.39 meters. (range 1,22m - 1,52m)

Table 1

| socio - DEMOGRAPHIC CHARACTERISTICS | |
|--|---------------|
| AGE | |
| <30 | 8 |
| >30 | 5 |
| RACE | |
| Black | 4 |
| Coloured | 9 |
| RELIGION | |
| Christian | 11 |
| Muslim | 2 |
| HEIGHT | |
| Average height | 1,39m |
| Range | 1,22m – 1,52m |

Three informants had attended "special school" which they all described as a school for people who are "slow". Two participants were still at high school (grade 11 and 12), four had left school before matriculation and seven had obtained a matriculation certificate. Two participants had entered into tertiary education but did not complete their tertiary education because of lack of finance. One woman was still at University at the time of the interview. All women who had matriculated indicated that they would have wanted to study further but did not do so due to lack of financial support. Eight of the women were still living with their parents.

3.2 Women's knowledge about Turner Syndrome

Two of the 13 women appeared well informed about Turner Syndrome. In contrast, the other eleven participants said that their knowledge about their medical condition was deficient. This lack of knowledge was reflected by statements such as "*I don't know much*", or "*I don't understand much except for what the doctor told to me*".

The majority of these women appeared to paraphrase what the doctor had told them. Only very few seemed to provide their own concept about their medical condition. Many women were reluctant to talk about their medical condition and needed encouragement. This reluctance appeared to be related to their lack of knowledge rather than an unwillingness to talk about their particular problems.

When asked about the causes of Turner Syndrome, six informants mentioned that there was a genetic or chromosomal aetiology. One of them said that they had an "X" instead of "XX", another one spoke about deficiency in either the X or Y chromosome. A third woman said that her eggs had "45" instead of "46". One woman believed that the chromosomal disorder was caused by one of her parents.

Many of the participants said that Turner Syndrome was associated with difficulty or the inability to have children. This was the main theme in terms of women's knowledge about Turner Syndrome. Even though many of the women associated Turner Syndrome with infertility, very few had a clear understanding of the reasons for the infertility or the fertility options available to them. Three women alluded to "having no eggs "and one said: "my one egg is collapsed". Most of the informants had no knowledge of the options available as a resolution to their infertility. Only three women knew about oocyte donation.

Only four informants referred to the absence of their menstrual periods as being part of their medical condition. Two of these women tried to provide an explanation for their amenorrhoea, with one woman mentioning "a problem with hormones" and another saying; "there is a problem with the development of my reproductive organs".

Seven informants alluded to short stature. Three women mentioned other effects of Turner Syndrome on the skeletal system, with one woman indicating that she had a short finger. Another woman stated that her spine was very prominent and a third women mentioned deformities in her hands which she attributed, correctly, to her condition.

3.3 Self — Image and well-being

For eleven women Turner Syndrome appeared to have a negative impact on their self-image and well-being. These women spoke about being *"incomplete"*, not being *"full women"* and *"being different"*. This sense of low self-image appeared to be especially linked to their loss of fertility.

Most women became tearful whilst discussing fertility issues. This is evident in the following interview extracts:

"Having children is part of being a complete person". "I think I am not going to be a full woman".

"I would like to have my own child, like any other normal person".

Five informants said that not having periods had caused them to have a low self esteem. They said they did not feel that they were like other *"normal girls"*. One woman experienced this and said *"I had low self esteem, because I was not like other girls, I did not even have periods; I felt that I was different"*.

Short stature was not a cause of low self image for most of the informants. Three women said that they were occasionally teased about their height and called "shorty". These three women however said that this was said "jokingly" and did not affect them negatively.

In contrast, two informants related being short to not being normal, one of them said, "I am not normal, I was told I would not grow like a normal person does".

Most of the women did not have any negative perceptions about their intellectual abilities. The six informants who repeated a grade at school attributed this to lack of effort on their part rather than to their medical condition. Only three women said they had attended "Special School" because they were "slow".

One woman suffered from very low self image and had many visible skeletal abnormalities. When being questioned about her knowledge of Turner Syndrome, her initial response was: "It worries me, I have low self esteem. I feel useless in life. I feel I have no future. I feel like people look down upon me, I mostly do not want to be with other people".

3.4 Relationships with parents and siblings

Nine women had a good relationship with their parents and eight informants were still staying with their parents. Despite these good relationships, most said that their parents also had very little knowledge about Turner Syndrome and therefore could not offer much support.

Three participants found their parents to be over protective. They attributed this to their short stature. One woman explained that her mother treated her as though she was the youngest child despite there being younger siblings. Another informant said: "*I think the fact that I'm short makes my parents more protective of me. Although they give me freedom to do what I want to do, they ..., wanting to protect me all the time.*"

One woman had a poor relationships with her parents. Both of her parents were alcoholics. She described their relationship as a "*hands off relationship*", and she related this to her mother's feelings of guilt. She said: "*I know my mom feels guilty about it, she thinks it was brought on by her smoking and drinking during her pregnancy*".

Most participants also had close and good relationships with their siblings. Two informants however said that they needed to enforce their authority over their younger siblings. One woman said, "I*need* to speak very loud ... before they listen to me". The other women said, "My mouth always makes up for my height".

Most women felt that their siblings were not aware of their condition and had very little knowledge about it. Two participants, however, had a bad relationship with their siblings. One of them said that she was frequently being called "*stupid*" by her siblings. The second women always felt like an outsider in her family, saying this is about her siblings: "I do not chat to them about my medical condition because they don't know me. They are just not interested."

3.5 Relationship with extended family, friends and community members

Good sentiments were expressed by most of the participants regarding relationships with extended family members. One woman described being looked down upon by some of her extended family members. She attributed that to the fact that her cousin would not assist her to get employment because she was ashamed of her physical appearance. This woman had visible deformities of her hands. Many informants had "lots" of friends and did not perceive themselves as having difficulty in making friends.

Only three women, however, had ever discussed their condition with their friends. Most of the other informants said that their friends had no knowledge about their condition.

Six women reported that they had few friends and mainly attributed this to being very selective or being reserved. One woman had only a single friend. She explained this by saying: "*I want to protect myself from negative comments that people might have about me*".

Seven informants felt that they were treated well by people in their community. Three women however alluded to having had some bad experiences in their communities. One woman said that people often stared at her and called her "small aunty". Another woman said: "*they talk about me as if I'm not there ... they ... say: 'Is she a child or an adult?'*" The third women said that people stared at her when she was with her male partner as they think she is a child having a relationship with an older man.

3.6 Experiences relating to sexual or marital relationship

Eleven of the women interviewed were not married. The remaining two were married and had good relationships with their spouses. Their spouses were aware that they had Turner Syndrome and understood the fertility implications.

They were also informed about the option of oocyte donation but because of socio-economic circumstances had opted to adopt children. Both of these women said they lacked the finances and therefore could not attempt oocyte donation. These women had good relationships with their adopted children and seemed to be living happy and fulfilled lives.

Many of the unmarried women were also not in a relationship at the time of the interview. The reasons were variable, with a few women saying they were focussing their energy on their studies or work. Others expressed the view that relationships required too much commitment or that they were not interested in relationships at present in their lives.

Most of these women did not attribute being single to having Turner Syndrome. One woman, however, experienced this differently and said: *"Well, it's because I feel different from other women, especially because I don't get my periods. The fact that I can't have children worries me. I think a man might not want me because of that"*. Some of the single women expressed fear that a future relationship might break down because they were unable to have children. One of them said: *"I know that one needs a child to strengthen a relationship"*. Two women evidently spoke from experience as they had been in relationships which ended after informing their partners about Turner Syndrome and its association with infertility.

All the informants said they were of heterosexual orientation. Six of them had been sexually active. These six women said they had normal sexual relationships. Of the seven participants who had never been sexually active, none attributed this to having Turner Syndrome. Some of them said this was because of religious beliefs, and others said they did not trust their partners.

3.7 Educational Experiences

As previously mentioned, six of the participants had repeated a grade at school. In addition, most of the women had had difficulty with mathematics with two saying that they "hated maths". A few others reported having needed extra lessons.

Most informants had good relationships with other learners. Three women however said that they were often teased about their height. One participant who did not enjoy school attributed this to constant teasing about her short stature. She said: *"Oh yes, when I was in school, I had it at high school. The very first day I came there, the matric guys used to come to me and pick me up"*.

Twelve informants had experienced good relationships with their teachers and had not felt discriminated against in any way. One woman even felt that because of her short stature the teachers treated her particularly well. She said, "*I was always the teacher's pet*".

A very different experience was related by one informant. She entered into a relationship with her school teacher at the age of 12 years which, by definition was abusive. The relationship ended traumatically six years later, after she had informed him of being diagnosed with Turner Syndrome.

3.8 Experience Relating To Employment and Job Opportunities

Two of the participants were still studying and had never attempted to seek employment. Eight of the informants were employed at the time of being interviewed. Six of these women, however, previously struggled to get employment. The three who were unemployed were having difficulty getting employment and thought that this might be related to their short stature. The words of this woman appear to reflect the experiences of others: "*I always have a feeling that people might think I am a child and therefore not wanting to employ me*"

Five informants worked with children, mostly at a crèche. One was an au pair. The few that were unemployed expressed the sentiment that they would love to work with children. The women employed at the crèche said that being with children "filled the void" created by not having children of their own.

Most participants who were employed had a good relationship with their employers. Three informants however felt that they were treated unfairly by their employers. One woman said that she was made to do the jobs that no one else wanted, such as changing children's nappies. She also felt that she was being underpaid. Another informant experienced this differently and said that her employer treated her badly because she was seen to be "slow". Many of the women had good relationships with their fellow employees. Only a few felt that they were taken for granted by some of their fellow employees, but none of them attributed this to their condition of Turner Syndrome.

3.9 Coping mechanisms and Support systems

Most informants said that they were coping well with their medical condition and felt that they had learnt to live with it. Some emphasized that although they were coping this was hard, but since nothing could be done about their condition, they felt they just had to manage.

When asked about their sources of support, many spoke about their family members and friends. Most of the informants relied heavily on their parents for emotional support. This is highlighted in this interview extract:

"My parents love me a lot. Sometimes I wish I were dead, but when I think about how much they care for me ...they don't reject me, that gives me the strength to go on".

A few participants felt that having friends did not necessarily translate into support, as the friends had no knowledge about Turner Syndrome. The negative impact of this lack of knowledge is highlighted in this interview extract:

"I talk to nobody really about it, because they would not know how to support me either. Anyway, I would not know where to start and what support to expect".

Three participants felt that depression was a major part of their lives. One of them said: *"I'm not coping, I feel useless, I can't even help my younger siblings, especially financially. I'm still dependant on my parents financially. I wish I could also help them sometimes".*

Other reasons given for the depression included being treated badly by people in the community, not having children and the need to take daily medication. One woman said: *"Just as I get older, I see people with children. I think I could have had a daughter, a son, or I could be a grandmother, but I don't have that, and it makes me sad".*

The majority of the women appeared to lack support from their medical care givers. These women felt that the hospital was always busy and the doctors had very little time to explain to them and discuss their medical condition. Several of the women said that they needed more information about Turner Syndrome. Only one participant knew about support groups for women with Turner Syndrome. When others were asked about their attitude towards a support group, many said that it would be useful to belong to a support group expressing the need to converse with women who had similar problems to them. Two women however said that they do not think it was necessary to belong to a support group. These women felt well supported and did not perceive themselves to be different from other people. In addition one of them thought that belonging to a support group might make her more distressed by constantly reminding her about her medical condition.

CHAPTER 4: DISCUSSION

The psychosocial impact of Turner Syndrome on women in South Africa is not known. Most studies have been done in developed countries. South Africa is a developing country with women from diverse cultural and socio-economic backgrounds. The results of studies on the psychosocial impact of Turner Syndrome done in developed countries may not reflect the experiences of women in South Africa. It is hoped that the results of this study will help inform the management of women living with Turner Syndrome in South Africa.

The key findings of the study were as follows: Most of the participants had poor knowledge about their condition. Infertility was considered to be the most traumatic consequence of the syndrome. Short stature was not a cause of a low self image for most of these women. Interpersonal relationships were not negatively affected by having Turner Syndrome. There was evidence of delayed partner relationships and of cognitive deficits. Employment and job opportunities were usually not negatively influenced by having Turner Syndrome. Support was lacking for these women regarding their medical condition.

4.1 Women's Knowledge about Turner Syndrome

The lack of knowledge about Turner Syndrome had a negative impact on the psychological well being of the women in this study and their families. This

lack of knowledge was attributed to the inadequate information given to these patients by their doctors.

The parents of these informants had insufficient knowledge about Turner Syndrome and as a consequence could not discuss the condition with their daughters. One woman had a bad relationship with her mother. She related this to her mother who was an alcoholic having feelings of guilt about her medical condition. She thought that her consumption of alcohol during her pregnancy had caused her daughter to have Turner Syndrome. In their study of 73 women with Turner Syndrome, evaluating personality pathology, social maturity and psychosexual development Money et al (1970) concluded that parental rejection and psychopathology were directly associated with psychopathology in their daughters. Of particular interest is that drunkenness was one of the criteria used to define parental psychopathology in that study.

One of the participants in our study blamed her parents for her medical condition. She thought that she inherited Turner Syndrome from one of her parents.

Sutton et al (2006) conducted a qualitative study in Bethesda to evaluate the importance of diagnostic disclosure amongst 97 women with Turner Syndrome. Thirty percent of these women mentioned that their health care providers or parents had withheld all or part of their diagnosis. The reasons given by the parents for non disclosure to their daughters about the diagnosis of Turner Syndrome were that the parents themselves had inadequate information about the condition as the medical care givers had not provided

them with enough information and some of the parents reported wanting to protect their child from the heartbreak of knowing they were infertile.

A Swedish study on psychosocial aspects of twenty two women living with Turner Syndrome, evaluated, through semi-structured interviews, their social functioning, emotional development, sexuality and coping style. **Sixty** percent of these women stated that the information given to them about their medical condition had been insufficient. Six of these women said they knew too little about their condition to even ask questions from their doctors (Sylven et al; 1993).

The lack of knowledge of the women in our study about their condition affected their compliance with medication. Most of these women had no understanding of why they had to be on long term hormonal treatment. This added to their distress as many expressed unhappiness about taking daily medication. There is a need for clinicians to spend more time on education and counselling about Turner Syndrome. The clinicians can benefit from training sessions on disclosure of the diagnosis to patients and how to counsel these patients. Information in the form of a pamphlet should be given to these women. This, however, may not always be helpful due to the varying degrees of literacy of women seen in our clinical service.

4.2 The Psychosocial Impact of Infertility

Most of the informants considered infertility and premature ovarian failure to be the most traumatic consequence of having Turner Syndrome. Most of

these women reported having a low self image and related this to being infertile. The inability to have children impacted negatively on their sexual and marital relationships. Most of the participants felt that the success of their relationships depended on their ability to have children.

Similar findings about this negative impact of infertility on women with Turner Syndrome are reported in studies from the developed countries (Sylven et al; 1993; Tang. 1989; Cardoso et al; 2004; MacCauley and Sybert 2006; Schmidt et al; 2006, Downey et al; 1989). In their psychosocial study of twenty two women with Turner Syndrome, Sylven et al reported that fifty percent of these women had been deeply affected by infertility and had depression as a result of this.

Downey et al (1989), in their report on interview and questionnaire data evaluating psychopathology and social functioning in twenty three women with Turner Syndrome compared with twenty three closely matched women with constitutional short stature, reported that many of those women were concerned about finding a partner who would accept them despite their infertility. These women had major fears of being rejected by men, to the extent of being reluctant to put themselves in situations where they could meet men.

The two women in our study who were married had not been able to access in vitro fertilization and oocyte donation due to financial constraints. In South Africa in vitro fertilisation is expensive and therefore inaccessible to many women. Infertility in South Africa has been shown to have a major

psychosocial impact due to various cultural beliefs (Dyer et al 2002; Dyer et al 2006). Children are often seen as an extension of wealth and infertile women are often ridiculed by their in-laws and may be abandoned by their husbands (Dyer et al; 2002). Only two women had adopted children, which perhaps reflects a lack of acceptance of the option of adoption among our respondents.

4.3 The Psychosocial Impact of Short Stature

Short stature was not a cause of low self image for women in our study. In a North American study evaluating social relationships and behavioural characteristics of 17 girls with Turner Syndrome, in comparison with 16 girls with short stature, the girls with Turner Syndrome were reported to have poor peer relationships and to have more behavioural problems such as needing more structure to socialise and to complete tasks than the controls. The authors concluded that short stature is not the reason for social and emotional difficulties of patients with Turner Syndrome (McCauley et al; 1986)

Ricket et al; (1996) conducted a prospective; cross sectional survey of 59 women with Turner Syndrome in Texas. They studied the effects of body image, height dissatisfaction; and peer ridicule on depression and self image on these women and found that short stature did not cause depression and low self image for these women. They concluded however, that peer teasing about general body appearance was the most significant factor related to

depression and a low self image in their cohort of women with Turner Syndrome.

In another American study on gender role development, Downey et al (1987) compared gender development between 23 women with Turner Syndrome versus 23 closely matched women with constitutional short stature. Their results showed more feminine behaviour in the women with Turner Syndrome than the controls. They concluded that the clinical features of Turner Syndrome including short stature do not impede normal female gender development.

Van Pareren et al (2000), in their study of 50 young women with Turner Syndrome who had achieved final height within the normal range after growth hormone treatment showed that girls with Turner Syndrome had low self esteem compared to their peers despite having achieved heights within the normal range. Their results showed that short stature was not the cause of psychosocial difficulties experienced by these girls.

4.4 Interpersonal Relationships

Most of the women in this study had good relationships with their parents and siblings. Two participants found their parents to be overprotective. They did not, however, find this to impact negatively on their relationship with their parents.

The one woman who had a bad relationship with her parents had parents who were abusing alcohol. Her bad relationship with her parents and siblings is a reflection of her dysfunctional family and not of having Turner Syndrome.

MacCauley and Sybert et al (2006) in their review article on psychosocial aspects of Turner Syndrome concluded that adult women with Turner Syndrome had socially fulfilling lives. Our results are similar in that most of the women reported no difficulties with making friends and felt they had fulfilling social lives.

The two women who were married reported living happy lives with their husbands and adopted children.

One woman, however, had an abnormal relationship. She had a love relationship with her school teacher at the age of 12 years. This was obviously inappropriate and an abusive relationship and perhaps illustrates the vulnerability to abuse of these girls with Turner Syndrome.

4.5 Delays with Partner Relationships

Downey et al (1989) in their study of 23 women with Turner Syndrome compared with 23 closely matched women with constitutional short stature, evaluated various milestones associated with adulthood such as leaving the parent's home, finding a partner and becoming sexually active. They found that fewer women with Turner Syndrome lived independently of their parents and fewer have partners than the general population. Several other studies

have shown some delay with adult development in women with Turner Syndrome (Borman et al; 2001, Ross et al; 2000, Saenger et al; 2001, Lawrence et al; 2003, McCauley et al; 2006)

Borman et al (2001), in Sweden, conducted a cross-sectional study using semi-structured interviews and two standardised self rating scales to evaluate psychological well-being, self rated health and social life amongst 63 women with Turner Syndrome. One of their significant findings was that only 18 of those women were living with a partner and only ten of them were married.

The women in our study demonstrated delays with adult development. Eight of the thirteen women were still living with their parents, however, it is not uncommon in Cape Town especially for Muslim women to live with their parents until such time that they get married. Most of these women were not in a relationship at the time of the interview. Only two of the women in this study were married.

4.6 Cognitive Deficits

Some of the women in our study demonstrated some cognitive deficits. Deficiencies in cognition among women with Turner Syndrome have been described (Siegel et al; 1998. Van Borsel et al, 1999; Nilsson et al; 1996; Murphy et al; 2006; Mazzocco; 2006). Three of the women in this study had required "special schooling" which suggested severe educational problems, six had repeated a grade and most had struggled with mathematics.

Swillen et al (1993) evaluated cognitive function in a cross-sectional study of fifty women with Turner Syndrome. Only five of these women were intellectually impaired. Poor results in mathematics and scientific drawing were present in this study and more so in the primary school group. This study highlights the importance of early recognition and remedial teaching for children with Turner Syndrome. Similarly, a study by Bruandet et al in France (2004), in which 12 women with Turner Syndrome and thirteen control subjects were subjected to cognitive tests including tests for arithmetic, demonstrated impairments with arithmetic amongst the women with Turner Syndrome.

The fact that only three women had tertiary education, however, is possibly a reflection of the financial difficulties which women in South Africa encounter. Tertiary education in South Africa is expensive and not easily accessible to many of the population unless they have exceptional academic results or are judged to have considerable potential.

4.7 Employment and Job Opportunities

Studies done in developed countries show that women with Turner Syndrome tend to have a lower occupational status than would be expected for their level of education and a slightly increased rate of unemployment (Borman et al; 1998; Ross et al; 2000; Borman et al; 2001; Saenger et al; 2001, Lawrence et al; 2003; Ostberg and Conway; 2003; McCauley and

Sybert 2006). The women in our study had employment that was compatible with their level of education.

None of the women in our study had high powered jobs because none of them had a university qualification. In South Africa, the rate of unemployment is high. The fact that five of the informants in this study were unemployed is probably a reflection of this rather than of the effects of Turner Syndrome. Turner Syndrome has not been shown to impact negatively on job opportunities in our study. Despite having no other formal training after matric, eight of the thirteen informants were employed.

Another relevant finding in this study is that five of the eight women who were employed were working with children. There is a suggestion in the literature that childcare or health care professions appear to be a frequent choice of career for women with Turner Syndrome (De Looz et al: 1993, Downey. 1989). De Looz et al (1993) in Belgium, evaluated psychosocial functioning of 20 adult women with Turner Syndrome. Their results showed that 16 of those women were employed and, of significance, is that more than half of these women had the ambition to work in nursing care, however, only three had succeeded and the rest were working as a nurse or a crèche teacher.

4.8 Support

The results of this study show that support for these women regarding their condition is lacking. Ideally, referral to a psychologist for counselling

especially after receiving the diagnosis should be mandatory. The lack of resources has made this practice unavailable in our clinic.

A support group needs to be established for these women. This should enable them to discuss issues and concerns about Turner Syndrome with other women who have similar experiences. The difficulty, however, would be access to the support group as most of these women are in full time employment. Information needs to be given to these women about other support groups in South Africa for women with Turner Syndrome.

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CHAPTER 5: CONCLUSION

The results of this study have assisted us in gaining insight about the psychosocial impact of Turner Syndrome on women in our clinical setting.

The study has highlighted some opportunities for interventions to improve the healthcare of these women. Clinicians need to focus, not only on medical aspects of the condition, but also on the psychosocial aspects. The women must be given adequate information about Turner Syndrome and its effects. Ideally the families of these women need to be included in the counselling sessions. This should improve the quality of support they can offer these women.

Feedback about the results of this study will be provided to our paediatric unit. Psychosocial support for women Turner Syndrome and their families should start early in their lives. Support for these women has to be improved. Referral to an informed social worker for counselling should be offered especially after informing them about the diagnosis.

More studies are needed to assess the psychosocial impact of Turner Syndrome on women in South Africa. Due to the diversity of cultures and socio-economic backgrounds of women in this country, the results of this study may not reflect the experiences of women in other parts of South Africa.

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INFORMATION LEAFLET

A qualitative study to evaluate the psychosocial experiences related to the condition of women with Turner Syndrome in Cape Town.

Turner Syndrome is a genetic disorder characterized by physical and endocrinological abnormalities. The psychosocial aspects relating to Turner Syndrome have been mainly studied in developed countries and the impact of this syndrome in women in Southern Africa is unknown.

We are conducting a qualitative study using in-depth interviews to evaluate the psychosocial experiences of women in Cape Town who have Turner Syndrome. The interview will focus mainly on the basic knowledge of the women about their condition, their experiences relating to interpersonal relationships, experiences relating to education, employment and their experiences relating to fertility issues.

Participation in the study is voluntary and we would appreciate if you would assist us by willing to be interviewed. The interview will take an average of about 30 minutes. The interviews will be tape recorded, but participants will be given the option to refuse having the interview taped. The tape recorded interviews will be transcribed into anonymous written transcripts and the tape recorded interviews will then be destroyed. There are no direct benefits or disadvantages in participating, but we hope to use the information obtained to improve the service we provide.

Confidentiality and anonymity will be strictly adhered to regarding the interview information. This qualitative study is being conducted at the University of Cape Town.

Should you have any further questions on this study, please contact:

- 1. Dr. N.T. Matebese Telephone: 021-404-6027/8
Cell: 083 282 8875**

- 2. Prof. M. Blockman
Chairman: Research Ethics Committee
(For questions related to your rights and welfare as a research participant)**

INTERVIEW GUIDE

Study number:

- 1. Age**
 - 2. Marital status**
 - 3. Height**
 - 4. Highest level of education**
 - 5. Occupation**
 - 6. Ethnic group**
 - 7. Religion**
- 1. Basic knowledge about the condition**
 - a. What does Turner Syndrome mean to you?**
 - 2. Experiences relating to interpersonal relationships**
 - a. Can you describe your relationship with your**
 - Parents**
 - Siblings**
 - Other family: cousins, aunts, uncles etc.**
 - Friends**
 - Community members**
 - b. What are your experiences relating to:**
 - Sexual/marital relationships**
 - Fertility issues**
 - 3. Educational experiences**
 - a. Can you describe your experiences relating to education and educational opportunities?**
 - b. How were the relationships with your fellow students and teachers?**

- 4. Experiences relating to employment and job opportunities**
 - a. Can you describe your experiences relating to employment and job opportunities?**
 - b. Relationships with colleagues and employees**
- 5. Support**
 - a. How are you coping with your condition?**
 - b. Are you aware of any support groups for your condition?**
- 6. Is there any additional information that you would like to contribute regarding how your medical condition affects your life?**

University of Cape Town