An investigation into the level of genetic knowledge and family communication regarding genetic risk in parents of children with cystic fibrosis

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DECLARATION

I, Mardelle Schoeman, hereby declare that the work on which this dissertation is based is my original work (except where acknowledgements indicate otherwise) and that neither the whole work nor any part of it has been, is being, or is to be submitted for another degree in this or any other university.

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ABSTRACT

Cystic fibrosis is the most common severe, autosomal recessive disorder in populations of caucasian descent with an incidence of 1 in 2500 live births worldwide. The Red Cross War Memorial Children's Hospital has a dedicated weekly cystic fibrosis clinic, attended by children with cystic fibrosis and their parents/caregivers from all over the Western Cape. The aims of the present study were to determine the level of genetic knowledge of parents with a child with cystic fibrosis; to determine the impact of the birth of a child with cystic fibrosis upon subsequent reproductive choices and to investigate family communication about genetic risk.

A qualitative approach was selected as it aims to understand, attempts to make sense of and provides descriptions that portray the richness and complexity of ordinary events from the perspective of the participants. Ten semi-structured qualitative interviews were conducted with parents who had a child with cystic fibrosis. Interviews were conducted in the language of the participants' choice and signed informed consent was obtained prior to the interview.

The participants in this study generally had a flawed understanding of the genetics of cystic fibrosis. The level of understanding was identified as being related to socioeconomic status. The birth of a child with cystic fibrosis had a major impact on subsequent reproductive decisions for participants. Most of them chose to reduce the number of children they had originally planned to have, following the diagnosis of their affected child. Knowledge about the genetics of the condition was not readily disseminated in the majority of these families. A lack of genetic knowledge was found to be the main barrier to risk communication.

Although cystic fibrosis knowledge has been previously assessed in this population, this was the first research of its kind to look at how the genetic knowledge impacts on reproductive decisions and how risk information is disseminated in cystic fibrosis families in South Africa. The findings of this study will help healthcare professionals involved in the cystic fibrosis clinic to address gaps and misconceptions in parents' knowledge and to understand the barriers to risk communication in these families. The service that is delivered at The Red Cross War Memorial Children's Hospital cystic fibrosis clinic may be improved by having a genetic counsellor as a regular member of the interdisciplinary team involved with all families with cystic fibrosis. A genetic counsellor could play an important role in facilitating information giving; knowledge gain and the dissemination of risk information in the family; assist in the reproductive decision-making process of carrier couples; and in contributing to providing the necessary ongoing psychosocial support.

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LIST OF TERMS AND ABBREVIATIONS

CF	Cystic Fibrosis
CFTR	Cystic Fibrosis Transmembrane Conductance Regulator
Creon	Pancreatic enzyme supplement
DNA	Deoxyribonucleic acid
Gr	Grade
HBOC	Hereditary Breast and Ovarian Cancer
No	Number
P	Participant number
R	Rand
RCCH	Red Cross War Memorial Children's Hospital
SA	South Africa
Steatorrhoea	Non-digestion of fat leading to bulky, smelly stool.
TOP	Termination of Pregnancy
USA	United States of America
WC	Western Cape
Yrs	Years

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Chapter 1

INTRODUCTION

1.1 INTRODUCTION

Cystic fibrosis (CF) is the most common severe, autosomal recessive disorder in populations of Caucasian descent, with a carrier rate of 1 in 25 and an incidence of 1 in 2500 live births worldwide (Massie *et al.* 2005). It occurs in all population groups in South Africa (SA) at different frequencies. CF is a multi-system disease with highly variable clinical characteristics. As a result of early diagnosis and advances in treatment and care, the life expectancy of individuals with CF has increased dramatically over the last two decades. The median life expectancy is now 33.4 years (Szyndler *et al.* 2005). Despite this, it remains a distressing and debilitating disease for the affected individuals and their families, for which there is no cure.

The Red Cross War Memorial Children's Hospital (RCCH) has a dedicated weekly CF clinic, attended by children with CF and their parents/caregivers. The clinic has an interdisciplinary team approach, offering expert care to any child with CF in the Western Cape. Once the diagnosis of CF is confirmed clinically and, if possible, molecularly, the parents/caregivers of the affected child are counselled about the genetics of the disease. This includes the prognosis, implications for further pregnancies, as well as possible risks for other family members. The understanding of the genetics of the condition directly impacts on decisions regarding further pregnancies and is also likely to have implications for risk communication with relatives (Wilson *et al.* 2004).

Parents of a child with CF have a 1in 4 risk of having another affected child in subsequent pregnancies. Various reproductive options are available to these couples, including prenatal diagnosis and selective termination of an affected foetus. Investigating the reproductive choices of parents of children with CF is important in gaining insight into the possible needs for counselling of carrier couples in the reproductive decision-making process (Henneman *et al.* 2001).

Family communication is important when dealing with genetic conditions. However, very few studies have specifically explored the dissemination of genetic knowledge to family members. Although the birth of a child with CF implies an increased risk for other relatives of being carriers, the hereditary aspect of the disease is not frequently discussed with these relatives. Genetic counsellors and health care professionals need to develop effective ways to help patients and clients to deal with communication issues in families. In order to do this, it is essential to explore how communication occurs in families. This includes with whom

parents communicate; how and what information is shared; and what leads to information being disseminated or not (Forrest *et al.* 2003).

1.2 AIMS

The aims of the study were to:

- Investigate the level of genetic knowledge of parents with a child with CF
- Investigate the impact the birth of a child with CF had on further reproductive choices and
- Investigate family communication about genetic risk.

1.3 OBJECTIVES

- To investigate the level of knowledge of the inheritance of CF and knowledge of the carrier status of parents, other offspring and family members
- To investigate the sources of genetic information
- To investigate how this knowledge influences or has influenced reproductive choices
- To investigate the transmission of information about genetic risk within a family
    - Whether the information shared with relatives
    - With whom is the information shared with
- To identify which factors facilitate or hinder information dissemination of genetic risk to relatives
- To investigate the sociodemographics of the group of participants
- To facilitate improved knowledge and communication about CF in families affected by the condition to enable them to make informed reproductive decisions.

1.4 ORGANIZATION OF THE STUDY

In Chapter Two a literature review is presented on various aspects of CF. The methodology of the study is described in Chapter Three. This includes the process by which participants were recruited, as well as a description of the measurement instruments in terms of appropriateness and validity/trustworthiness. A brief explanation of the data gathering and analysis is provided, as well as a description of the procedure of the research study. The presentation of the analysis, findings and discussion are intermixed in Chapter Four as is customary in qualitative research (McMillan and Schumacher 2001). In Chapter Five the main findings of the research are summarized in the conclusion. Recommendations as a result of the outcome of the study are discussed in Chapter Six.

Chapter 2
LITERATURE REVIEW

2.1 INTRODUCTION

This chapter includes literature reviews of the clinical aspects of cystic fibrosis (CF), including diagnosis and management thereof, the level of understanding of genetics following genetic counselling; communication about risk in CF families; and the impact the birth of a CF child has on future reproductive choices. Literature searches were performed using PubMed, Ebscohost, ScienceDirect and Google Scholar research databases.

2.2 CLINICAL ASPECTS OF CYSTIC FIBROSIS

Cystic fibrosis is caused by mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene on chromosome 7, which codes for a complex chloride channel, responsible for regulating the transport of chloride ions across epithelial cell membranes in certain organs (Brice *et al.* 2007). When CFTR is not produced or is altered in structure or function as a result of mutations in the CF gene, defective chloride transport leads to thick, viscous secretions in the lungs, pancreas, liver, intestine and reproductive tract, as well as an increased salt content in sweat gland secretions (Moskowitz *et al.* 2005). CF is therefore a multi-system disease with highly variable clinical symptoms.

CF is usually characterized by the triad of recurrent respiratory infections, pancreatic insufficiency causing malabsorption of nutrients and abnormally high sweat chloride levels. However, the spectrum of the clinical phenotype of CF is highly variable, some patients present with a full range of clinical features, while others may only show a single feature, such as male infertility. Many patients also demonstrate mild or atypical symptoms (Orenstein *et al.* 2002; Goldman *et al.* 2001).

a) Gastrointestinal and nutritional abnormalities:

Intestine: Meconium ileus presents in 15-20% of newborn infants with CF. This may be detected antenatally by the presence of echogenic (bright) bowel (Smyth 2005). Signs of intestinal obstruction can occur within 48 hours after birth. There is a delay or failure in passing meconium because of sticky secretions blocking the bowel (Ratjen and Döring

2003). This can also manifest as distal intestinal obstruction syndrome (DIOS) in older patients (Jackson 1989).

Pancreas: Infants and children may present with a voracious appetite, failure to thrive and/or steatorrhoea, due to pancreatic malabsorption. Pancreatic insufficiency occurs in 85% of cases (Turcios 2005). In infants, steatorrhea may be accompanied by rectal prolapse.

Liver: A spectrum of liver abnormalities may occur. Bile flow is compromised by the salt and water imbalance, leading to stasis and bile duct obstruction. The consequence of this is focal biliary cirrhosis which leads to liver cirrhosis and portal hypertension in 10-15% of CF patients (Ratjen and Döring 2003).

The resulting poor nutritional status from these gastrointestinal and nutritional abnormalities can affect growth and lowered resistance to infection. These patients are known to have higher total energy expenditure than other individuals and they therefore have increased nutritional requirements.

b) Respiratory tract

Both the upper and lower respiratory tracts are affected. In the upper respiratory tract recurrent infections lead to thickening and hypertrophy of the mucous membranes of the nose and nasal sinuses, resulting in upper airway obstruction and, in many cases, polyp formation (Smyth 2005).

Lung disease is the major cause of morbidity and mortality in CF. Affected individuals have lower airway inflammation and chronic endobronchial infection (Turcios 2005). Failure of lung defenses leave cystic fibrosis sufferers prone to recurrent chest infections caused by bacteria, notably *Staphylococcus aureus, Pseudomonas aeruginosa and Hemophilus influenzae* (Harris and Super 1987), resulting in airway obstruction and intense neutrophilic inflammation. Early manifestations are a chronic cough, intermittent sputum production and exertional dyspnea. This can progress to end-stage lung disease

characterized by extensive airway damage (bronchiectasis, cysts, abscesses) and fibrosis of lung parenchyma (Davis 2005). Finally, death usually results from a combination of respiratory and cardiac failure.

c) Sweat glands: The sweat glands produce sweat that is abnormally high in sodium and chloride, both at rest and during exercise (Orenstein *et al.* 2002). This can lead to the clinical consequences of salt deficiency, namely: failure to thrive, hypochloraemic metabolic alkalosis and heat prostration. This defect is a hallmark of CF and also provides the basis for the best diagnostic test, the "sweat test" (Westwood *et al.* 2006).

d) Male genitalia tract: The great majority of males (99%) are infertile because of developmental defects of the vas deferens which is either absent or blocked, leading to obstructive azoospermia (Ratjen and Döring 2003).

Superimposed on these direct effects, secondary effects on skin, bones and joints, blood and the endocrine system are not uncommon. Cystic fibrosis-related diabetes can occur in patients with pancreatic insufficiency. This increases with age, reaching a prevalence of over 30% in patients over the age of 30 (Turcios 2005).

2.2.1 Diagnosis of cystic fibrosis

For CF to be diagnosed the patient is required to have suggestive clinical features, as described above, as well as two positive sweat tests and/or two identified disease-causing mutations in the CFTR gene (Westwood *et al.* 2006).

a) *Sweat testing:* The gold standard for diagnosis of the classical form of CF remains the sweat test. The diagnosis rests on the presence of an excessive quantity of sodium and chloride in the sweat of an individual with clinical features suggestive of CF (Westwood *et al.* 2006). The test must be carried out in an experienced laboratory using standardized methods (Goldman *et al.* 2003. A sweat chloride concentration of more than 60mmol/L is consistent with the diagnosis of CF, if it was obtained on two separate occasions in a patient with

one or more clinical features consistent with the phenotype or a history of the condition in a sibling (Rosenstein and Cutting 1998).

In those cases in which sweat tests repeatedly give ambiguous results, measurements of nasal potential difference may aid the diagnosis (Murray *et al.* 1999). This method of testing is not yet available in SA. Alternatively, genetic testing can be used. Analysis of rectal mucosal biopsy or pancreatic stimulation testing may also be indicated to aid diagnosis (Ratjen and Döring 2003).

b) *Genetic testing*: The diagnosis of CF can be confirmed by DNA analysis in many cases by finding two known disease-causing mutations in a patient with the appropriate clinical presentation (Smyth 2005). To date, over 1400 disease-causing mutations of the CFTR gene have been identified (Cystic Fibrosis Mutation Data Base 2006). Very few of these are common and some are population- or family-specific. The ethnic origin of a patient or family must be considered when doing genetic testing. The most common mutation in the white population is ΔF508, occurring at a slightly lower frequency in the mixed ancestry population of SA. The 3120+1G>A mutation has been identified in the majority of CF cases in the black population (Denter *et al.* 1992). Because of the large number of mutations that have been identified as well as the variable distribution of these among the different population groups, diagnosis by DNA analysis is not always possible. Therefore, molecular diagnosis can be used to confirm a diagnosis, but cannot exclude it if fewer than two mutations are found (Goldman *et al.* 2001; Rosenstein and Cutting 1998). Goldman *et al.* (2003) described a panel of population-specific mutations that should be used when testing for CF in SA to give the best results. Using this panel, the mutation detection rate is 91% in white, 74% in coloureds and 46% in black African CF patients. Testing only for the ΔF508 mutation in white South African CF patients would give a diagnostic result in 58% of cases (Goldman *et al.* 2003).

Alternatively, a non-symptomatic diagnosis of CF can be made by means of as a result of screening for the disease either as part of a neonatal screening programme or because of a family history of CF ('cascade screening'). Another means of making a non-symptomatic diagnosis is by prenatal diagnosis through amniocentesis or chorionic villus sampling, followed by genotyping for CFTR mutations (Smyth 2005).

c) *Newborn screening*: Newborn screening facilitates the early diagnosis of CF and genetic counselling for affected families. The rationale is that early treatment with physiotherapy and antibiotics will improve the long-term prognosis (Comeau *et al.* 2007). Newborn screening for CF is possible in most cases in the first days of life by measuring the blood immunoreactive trypsin (IRT), followed by genetic testing for CFTR mutations in infants with raised IRT (Smyth 2005). This is not yet generally undertaken in SA.

d) *Carrier testing*: The aim of carrier testing is to identify carrier couples at risk of having offspring with a serious genetic disorder. It is considered good medical practice to offer carrier testing and genetic counselling to the relatives of those affected by recessive disorders such as CF (Roberts *et al.* 2003). Close relatives are tested first and depending on the results, more distant relatives can be offered testing. This is known as "cascade screening". Cascade screening essentially means extended family testing and the identification of carriers within a family radiating outward from the affected individual (Super *et al.* 1994). Carrier testing is generally aimed at individuals of reproductive age, as well as members of the family approaching reproductive age. Once a carrier has been identified in the family, the partner can be offered testing. Testing of children at risk of being carriers is generally delayed until at least the age of 18, when they are old enough to make their own informed judgement (Roberts *et al.* 2003). As mentioned above, there are limitations to genetic testing, because not all mutations in each ethnic group can be detected.

e) *Prenatal diagnosis*: In pregnancy, if a couple are both known CF carriers, they can be offered antenatal foetal genetic testing via amniocentesis or chorionic villus sampling to determine whether the foetus is affected. These procedures are associated with a miscarriage risk of around 1%. Both mutations must be known and although the genetic test is almost 100% accurate, the severity of the disease cannot be predicted (Brice *et al.* 2007).

2.2.2 Inheritance and incidence of cystic fibrosis in South Africa

Cystic fibrosis is one of the most common life-limiting genetic disorders in South Africans of Caucasian descent, with a prevalence of 1 in 2000 (Goldman *et al.* 2003). Cystic fibrosis is inherited in an autosomal recessive manner. To be affected a child must inherit a CF mutation from both of his/her parents. The chances of two carrier parents conceiving an affected child are 1 in 4 for each pregnancy. Cystic fibrosis is present in all population groups in SA, however at very different frequencies. A recent study of CF patients in the Western Cape by Westwood (2005) found an incidence of 1:3000 and 1:10 300 live births in the white population and coloured (mixed ancestry) population respectively. Carrier rates in these populations were found to be 1:32 and 1:61 respectively. Estimated carrier frequencies of between 1:14 in 1:59 have been proposed in the black South African population (Goldman *et al.* 2001). Padoa *et al.* (1999) showed a carrier rate of 1:34 and a calculated incidence of 1:4624 live births. The majority of clinically identified cases of CF in SA are, however, found in the white and coloured populations. To date, only 4 cases of CF have been identified in the Xhosa-speaking black population in the Western Cape.

2.2.3 Management of cystic fibrosis

Treatment is directed towards preventing disease progression. Although advances in research and treatment over the last 30 years have dramatically improved the longevity as well as quality of life for individuals with CF, the diagnosis is still associated with a reduced life expectancy (Orenstein *et al.* 2002). Cystic fibrosis is a complex disease requiring a holistic, multi-disciplinary approach to treatment. Center care by a team of experienced health professionals is essential for optimal patient management and

outcome (Mahadeva *et al.* 1998). There is still no cure. Gene replacement therapy is far from being used in patients with CF, mostly because of difficulties in targeting the appropriate cells (Turcios 2005).

Treatment of respiratory problems aims to minimise the effects of chest infections, to clear the lungs of obstructive and damaging secretions and to delay the onset of bronchiectasis. Management strategies include chest physiotherapy and physical exercise; mucolytic agents to reduce the viscosity of sputum; bronchodilator therapy; and antibiotics (Smith and Ball 1998). Minimising exposure to colds and viral respiratory infections, ensuring adequate immunisation, avoidance of both passive and active smoking and good nutrition are also essential (Ratjen and Döring 2003).

Due to the reduced secretion of digestive juice from the pancreas in most CF patients, pancreatic enzyme supplements are needed to help with digestion and absorption of food. Children with CF should have a normal balanced diet high in calories and receive Vitamin A, B, C, D, E and K supplementation (Turcios 2005). Treatment is tailored to the needs of each patient.

2.2.4 The cystic fibrosis clinic at RCCH

CF centers have played a key role in the improved prognosis in these patients. The CF service at RCCH was established in 1973. As part of this service a weekly clinic takes place, attended by children with CF and their parents/caregivers. The clinic has an interdisciplinary team approach, comprising a paediatrician, gastroenterologist, pulmonologist, physiotherapist, dietician, social worker and the liaison secretary, whom together offer expert care in a tertiary health care environment to any child with CF in the Western Cape.

2.3 THE PSYCHOSOCIAL IMPACT OF CYSTIC FIBROSIS

Chronic illness or disability has a dramatic and far-reaching impact on the affected individuals and their families. All aspects of the lives of the affected individuals and the

people around them are affected, including the physical, psychological, social and economic functioning (Falvo 2005). The profound effect of chronic illness is compounded by the prolonged, intensive course of treatment, the often uncertain prognosis, the constant psychosocial stress and the gradual interference with daily activities and family roles (Livneh and Antonak 1997).

2.3.1 Reactions to the diagnosis

All couples expect and wish for a normal, healthy baby. Parents start fantasizing and forming images of their expected child long before the actual birth. Learning that their child has a chronic illness or disabling condition, such as CF, is an all-encompassing, life shattering experience to parents (Carpenter and Narsavage 2004). All the hopes and dreams of the perfect child are lost. In most people, this produces emotions of loss that are associated with grief. Parents grieve the loss of the 'perfect child' they wished for and have to accept the realities and emotional impact of the child's condition (Weil 2000). Kubler-Ross (1969) described the main stages of the grieving process, including denial and isolation, anger, bargaining, guilt, depression and acceptance. Parents can move through these stages in a cyclical fashion throughout the different phases of the child's life and whenever the child has a period of hospitalisation for an acute episode of symptoms.

Götz and Götz (2000) described parents' reactions to the diagnosis of CF and found that they experienced all the emotions related to the grieving process. Emotions experienced include: shock, devastation, denial, anger, guilt, despair, and depression, intense feelings of fear, disappointment and sadness.

The diagnosis of a chronic illness or disabling condition in a child may have a traumatic effect on the entire family and may predispose both the child and family to concomitant problems with adjustment (Ziolko 1991). Stages of adjustment are individual and varied and the ultimate goal of adjustment is acceptance of the condition.

2.3.2 Information and acceptance

Parents can only constructively begin to cope after they have acknowledged the irreversibility of the condition and understood and accepted its implications (Ziolko 1991). The information parents receive affects their coping strategies and adjustment. A realistic image of the child's condition and its implications for their lives and their other children's lives, together with adequate information and practical advice on how to cope with their affected child in everyday life has been shown to have a protective effect on the parents' relationship and family functioning. Parents need information regarding the nature of CF and its daily management as well as the growth and developmental needs of their children (Hymovich and Baker 1985). This facilitates the process of "assigning meaning to the illness", which is an important coping strategy (Coyne 1997). Better parental counselling and information leads to fewer conflicts caused by insecurity and helplessness engendered by the child's condition (Taanila *et al.* 2002).

2.3.3 Impact on family functioning

Caring for a child with CF is stressful and has implications for the main carer, the parental relationship, family functioning, unaffected siblings as well as the affected child (Hodgkinson and Lester 2002). The family is a social system in which the individual's behaviour and emotions influence and are themselves influenced by other family members in an ongoing manner (Weil 2000). Families must make adjustments, adaptations and role changes both as a unit and as individual family members (Falvo 2005). The family affects the child's adjustment and in turn, the child's illness affects the family functioning (Götz and Götz 2000).

Children with CF are now living well into adulthood and parents are faced with many of the same additional childrearing issues encountered by all parents (Hymovich and Baker 1985). Although the presence of CF does not lead to psychopathology in families, it increases the vulnerability of family members to the stresses of life. The nature of the intensive treatment, including daily therapy, the potential for hospitalization on a

repeated basis and the uncertain course of the illness can lead to considerable stress in families (Götz and Götz 2000; Hymovich and Baker 1985).

Conflict over responsibilities for caring for the affected child and decision-making are the most commonly reported stresses experienced by parents caring for a child with a chronic illness (Quittner *et al.* 1998). Specific issues related to CF include diet and mealtime behaviours, social development and adjustment, and non-cooperation with treatments (Blair *et al.* 1994). Higher levels of distress, an avoidant coping strategy and low levels of family support have been associated with poor psychological adjustment in CF families (Ievers and Drotar 1996). The family needs to accommodate the complex treatment into a schedule which permits family activities to continue as normal (Coyne 1997).

There is evidence that the major responsibility of caring for a child with CF, still tends to be borne by the mother and that this burden of caring can cause increased illness, depression and loneliness (Hodgkinson and Lester 2002; Coyne 1997).

Siblings of children with genetic conditions are affected in many ways. They may be affected by their parents' sadness, grief, fear or sense of stigmatization. The needs of the affected child and the natural over-involvement and overprotection may leave other children feeling that they are not getting enough attention from their parents (Weil 2000). Emotional strain may be created amongst family members, with resulting feelings of resentment, antagonism and frustration (Falvo 2005). Unaffected siblings may have fears relating to their own vulnerability of being affected or of being carriers of the condition. There may be confusion arising from a lack of understanding, or as a result of a deeper psychological response to family or personal vulnerability or identification with the affected individual (Weil 2000; Patterson *et al.* 1990).

2.3.4 The importance of social support

Social support has been shown to be an important influence on how an individual copes with a number of stresses (Ziolko 1991). A support system within the family is an important coping strategy which helps to share the burden of the illness and, therefore, decrease the family's vulnerability to stress. Family cohesion and feelings of togetherness and co-operation are important in allowing the family to cope (Taanila *et al.* 2002). Along with family support, it seems that families derive a great deal of benefit from sharing experiences with families in similar situations. Families and patients affected by the same specific condition are an extremely practical and rich source of information (Smith 1998).

2.3.5 Psychosocial problems in children with cystic fibrosis

As a result of the increase in life expectancy of children born with CF, issues such as quality of life, adherence to treatment and psychological well-being, although previously believed to be of lesser importance than physical well-being, are now recognised as significant factors (Szyndler *et al.* 2005).

Family relationships including levels of cohesion, conflict and expressiveness have a significant impact on the affected young person's psychological well-being, functioning and adjustment (Szyndler *et al.* 2005). Communication difficulties within the family may lead to an increased risk of developing psychosocial problems (Chesson *et al.* 2004). There is evidence that adequate family functioning is more important in terms of psychological impact on the child, than the illness itself (Cowen *et al.* 1986; Lewis and Khaw 1982). Lower family stress, parental availability and integrative family coping have been associated with better health outcomes in affected children (Patterson *et al.* 1990). Emotional well-being, quality of life and adherence to treatment has also been linked to hopefulness and optimism, despite the severity of the physical disease (Szyndler *et al.* 2005; Abbott and Gee 1998).

Patients with CF have been found to generally cope well with their disease, to be well-adjusted and to successfully handle life tasks, despite their illness. The medical outcome of the condition is not only determined by the objective pathophysiological characteristics of the disease, but also, importantly, by the subjective psychological reactions to the illness and the coping behaviour of the patients (De Jong *et al.* 1997).

Identifying stresses and supporting healthy coping strategies in CF families is important, not only for maintaining good family functioning, but also in improving the physical health of the affected child (Hodgkinson and Lester 2002).

2.4 GENETIC COUNSELLING

"Genetic Counselling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates the following:

- Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.
- Education about inheritance, testing, management, prevention, resources and research.
- Counselling to promote informed choices and adaptation to the risk or condition" (The National Society of Genetic Counsellors Definition Task Force 2006:79).

Genetic counselling for CF involves discussing the prognosis and management of CF; the cause and inheritance of the condition, including risks for future pregnancies; options for future pregnancies; and implications for other family members. It also involves addressing how the condition affects the psychosocial, medical and financial aspects of the families' lives and providing support to the family in the process of adjusting to and coping with the diagnosis (Bennett *et al.* 2003). Genetic counselling is an important intervention for all newly-diagnosed CF families (Kerem *et al.* 2005).

Considering the complex, consequential and personal nature of genetic risk information, genetic counselling is a suitable way of informing individuals about the nature and

outcomes of genetic conditions (Dillard *et al.* 2006). Risk information is however, one of the most complicated types of data that can be presented in genetic counselling. Not only is the term "risk" defined differently by individuals, people also vary in the understanding and relevance they attach to risk information, for themselves and for others (Veach *et al.* 2003).

2.5 LEVEL OF GENETIC KNOWLEDGE

For the counselling component of a genetic service to be effective, a good level of understanding of the genetics of the condition is important. This raises the question of how much parents understand about the genetics of CF. A major obstacle in the provision of genetic counselling is the lack of public knowledge of basic human biology and genetics (Chapple *et al.* 1997; Leonard *et al.* 1972). The importance of genetic aspects in the management of a CF family is often underestimated. Counselling needs to include greater detail than warning parents of the 1 in 4 recurrence risk. Knowing the risk, but not fully understanding the pattern of inheritance, may lead parents to continue to worry over the years and may lead to them being unable to ask questions to dispel their fears (Hodson *et al.* 1983). Several CF studies have reported low levels of genetic knowledge amongst parents of children with CF.

Leonard *et al.* (1972) conducted a study on a sample of 76 families with various genetic conditions. Of these families, 39 had at least one child affected with CF. They found a flawed understanding of genetics in 25% of these CF families and no understanding in a further 25%. In a sample of Scottish and Irish CF families, McCrae *et al.* (1973) reported the parents' understanding of the genetic basis of the disease to be unsatisfactory and that 75% of parents had a poor understanding of the inheritance of CF. They found psychological barriers and poorly-timed counselling to be major contributing factors to the lack of knowledge. Nolan *et al.* (1986) found that CF patients and their parents were well informed about disease pathophysiology and treatment, but had only a fair understanding of genetics and a poor understanding of reproductive risks and male sterility. He noted that parents found questions involving risk estimation particularly difficult.

Henley (1988) conducted a large study in the Western Cape assessing CF knowledge in patients and their families. She found parental knowledge of the genetics of CF and associated reproductive risks to be mediocre and that of patients and siblings to be poor. Social class was shown to be a significant predictor of knowledge in parents. The ongoing need for assessing parents' and patients' knowledge of CF was highlighted in the study (Henley and Hill 1990). This is the only study of its kind that has been published in SA.

Studies have shown that although parents sometimes have the genetic knowledge and can answer questions, they are unable to apply this knowledge, indicating poor understanding (Henley and Hill 1990; Nolan *et al.* 1986). Rona *et al.* (1994) found that although couples at risk of having a child with a severe genetic disorder value the genetic counselling they received, many of them did not remember important facts in relation to their risk status.

Denayer *et al.* (1990) found that although most parents (94%) of CF patients knew they were obligate carriers, only 18% knew that the risk of their siblings being carriers is 1 in 2. Similarly, only 1% knew that their healthy children have a 2 in 3 risk of being carriers of the CF gene. Similarly, in a study comparing client's genetic knowledge before and after genetic counselling, Seidenfeld and Antley (1981) interviewed mothers of children with Down syndrome. They found that, although counselling was very effective in enhancing knowledge about general genetics, clients had difficulty comprehending recurrence risks for relatives, siblings and the general population.

Only one study (De Braekeleer *et al.* 2001) conducted in Canada, reported that CF patients and their parents had a particularly good knowledge of the genetics of the disorder. In contrast to other studies on this topic, they found that individuals in their study had no difficulty with risk calculations in various scenarios.

A recent study by James *et al.* (2006) investigated the influence of mode of inheritance in families affected by autosomal recessive and X-linked conditions. The participants in

their study had a moderate understanding of inheritance, with X-linked families significantly more likely to understand inheritance and their own reproductive risks. They also found that autosomal recessive family members overestimated the reproductive risks of siblings and affected individuals, findings which have been reported elsewhere (Fanos and Gatti 1999). This misunderstanding of risk has potential harmful implications. Overestimating their reproductive risks has led to long term decisions to forgo marriage and childbearing in siblings of individuals with CF (Fanos and Johnson 1995).

Another important factor to consider is that the majority of parents lack prediagnostic knowledge of CF and therefore have to acquire a great deal of new, and often very unfamiliar, information. Parents are faced with unfamiliar medical information, such as progressive lung disease and pancreatic insufficiency, as well as genetic information including terms like 'carrier status', 'inheritance pattern' and 'recurrence risk'. The provision of written and illustrated materials is of vital importance in facilitating this knowledge gain (Jedlicka-Köhler *et al.* 1996).

Facilitating an accurate perception of risk is a constant concern in genetic counselling and it is likely to have implications for risk communication with relatives (Wilson *et al.* 2004). The understanding of the genetics of the condition also directly impacts on decisions about further pregnancies. Information giving at the time of diagnosis should be limited as this is a time parents are known to be suffering from stress and shock and could therefore lead to lack of retention of information. Explanations should be provided in a clear, simple non-technical language and should be repeated, clarified and provided in written form (Henley and Hill 1990).

2.6 RISK COMMUNICATION

Patients attending genetic clinics are often the main gatekeepers of information for other family members (Clarke *et al.* 2005). Genetics is a family matter. Knowledge of an individual's genetic risk inevitably provides information about his or her biological

relatives as well, thus conferring responsibility to decide whether and how to share this information with others in the family (Peterson 2005).

Communication among family members is integral to the process of genetic counselling and to the coping and adaptation of the family and its members (Weil 2000). Communication is critical in identifying and notifying extended family members who may be at increased risk or whose present or future children may be at risk. Not conveying risk information may deny relatives the opportunity to make informed reproductive choices (Clarke *et al.* 2005). Beyond the counselling session, open communication is, in general, associated with more effective coping, including greater emotional support, sharing of relevant information and effective, informed planning (Weil 2000).

Although family communication is important in clinical genetics, very few studies have specifically explored the dissemination of genetic knowledge to family members. Wilson *et al.* (2004) conducted a meta-analysis of literature related to the disclosure of genetic information. They described disclosure as more than a single event in which there is transfer of information, but rather that it has to be seen in the context of complex individual, familial and socio-cultural beliefs, behaviours and often tensions. Telling relatives is more usefully viewed as a process, rather than an act (Forrest *et al.* 2003). This process could be affected by several factors including: the nature of the disease, whether or not preventative measures were available and, the overall pattern of family communication; including family rules, roles and beliefs, geographical and emotional distancing and the individual coping styles of family members (Wilson *et al.* 2004; Weil 2000).

Although the birth of a child with CF implies an increased risk for other relatives of being carriers, the hereditary aspect of the disease is not often discussed with these relatives. Studies have shown that there is often poor communication about genetic risk between relatives in families (Forrest *et al.* 2003; Denayer *et al.* 1992a). As early as

1964, Turk described CF family members as being caught in a "web of silence" (Turk 1964).

2.6.1 Telling siblings

Aunts and uncles of a CF patient have a 1 in 2 chance of being a carrier of the CF gene. The little work that has been done on the topic suggests there are difficulties in disclosing carrier information to family members. Only partial information may be passed on by parents of an affected child to their siblings or risk information may be shared in a way that many are not made fully aware of the risk of being a carrier themselves (Wilson *et al.* 2004; Denayer *et al.* 1992a). In a sample of 105 CF-parents in Belgium, Denayer *et al.* (1990) found that only 19% discussed the genetic aspect of CF regularly with their siblings and 36% did now and then. Twenty percent of the CF-parents had never mentioned the hereditary aspect of CF to their family.

A further study by the same authors looked at the level of knowledge of CF and its genetic transmission among aunts and uncles of CF children and found that although they were relatively well informed about the main symptoms of CF, they only had a very superficial knowledge about the genetic transmission (Denayer *et al.* 1992b; Denayer *et al.* 1992a). Only a small proportion of the respondents (20%) were aware of their approximate carrier risk and/or risk of having a child with CF. They found that parents of the child with CF were the major source of genetic information in these individuals. Importantly, the risk of having a CF child played a part in the decision-making about future pregnancies for at least 39% of the participants. One individual completely refrained from having children because her sister told her the risk of having an affected child was 1 in 4. Similarly, Lafayette *et al.* (1999) found a lack of understanding of the inheritance of CF amongst the majority of relatives of individuals with CF, suggesting that relatives were not receiving adequate information. In general, they underestimated their carrier risk, but overestimated their risk of having a child with CF.

2.6.2 Telling the children

For parents, in particular, the issue of disclosure can be very difficult when considering what, if anything, to tell their children about their genetic risk status (Forrest *et al.* 2003). Informing children is often viewed as particularly difficult because of the sense of guilt it evokes, knowing that they may have passed the mutation on to their children. When and how to tell presents a further dilemma (D'Agincourt-Canning 2001). It has been shown that parents in Huntington Disease (HD) families often find themselves torn between wanting to protect their children for as long as they can, but being aware that they need to discuss the genetic information in time for their children to make key life decisions (Forrest *et al.* 2003). Ormond *et al.* (2003) found that the primary reason parents gave for not informing their children of their CF carrier risk was age; parents felt the children were too young to understand the significance of the information. Delaying disclosure may provide parents with a temporary sense of control (Wilson *et al.* 2004).

Siblings of children with CF have a 2 in 3 risk of being carriers themselves and depend on parental transmission of information. Because of the emotional impact of CF on parents, coping responses are often initiated by the diagnosis. This disrupts the normal environment in which children develop. Communication patterns in CF families, which evolve over years of dealing with the condition, are often not conducive to an environment in which genetic possibilities can be discussed openly. Parents rarely sit down and talk with their unaffected children about CF and siblings do not generally discuss the condition with each other or their affected brother/sister. Fanos and Johnson (1995) found that 63% of their sample of adult CF siblings had problems with obtaining information about their risk status and the possibility of carrier testing from their parents. In contrast, individuals who knew about testing came from families where there was open communication about the illness. Siblings from these families indicated that their parents had managed to face the genetic transmission of the condition, without placing undue blame on themselves or redirecting it to "the other side of the family". Because of this, an atmosphere was created in which genetic concerns could be confronted openly.

Evidence that individuals do not always share information about genetic risk with other family members has also been found in other genetic conditions (Claes *et al.* 2003; Green *et al.* 1997; Varekamp *et al.*; 1992; Suslak *et al.* 1985)

2.6.3 Barriers to risk communication

Barriers to information dissemination include lack of information about CF, the perception that relatives would not be interested in carrier screening, as well as gender; studies have shown that men are less likely than women to inform family members of their genetic risks (Sorenson *et al.* 1996; Fanos and Johnson 1995). McConkie-Rosell and DeVellis (2000) found misunderstanding of genetic risk to be a major reason for carriers not sharing their status with family members. For risk information to be distributed in a family, knowledge and understanding of genetic risk is essential. Lack of understanding may cause inaccurate or selective information to be passed on (Claes *et al.* 2003). Individuals need to make sense of their own risk and have time to do so, before dealing with what and whether to tell their relatives (Forrest *et al.* 2003). Information provided in genetic counselling may be misunderstood, leaving the genetic counsellor and family members with different views because of their different understandings of the risks and benefits of disclosure (Clarke *et al.* 2005).

Geographical distance, lack of social closeness, divorce, separation, adoption or internal family rifts mean that individuals are often no longer in contact with some members of their family (Featherstone *et al.* 2006; Ormond *et al.* 2003; Weil 2000). CF is often viewed as located within the nuclear family, not the extended family (Fanos and Johnson 1995). Members of families may simply choose not to share information about genetic risk with one another (Featherstone *et al.* 2006).

Limited research also suggests that, when the knowledge of carrier risk comes after the birth of an affected child, the emotional difficulty in coming to terms with the child's condition may present an important barrier to communication. The diagnosis of a chronic illness, such as CF, initiates a process of coping responses on the part of the parents. Guilt, hostility, grief and mourning and a sense of defectiveness are all feelings

of individuals coping with the burden of genetic disease (Fanos and Johnson 1995). This may be further compounded by the incomplete comprehension of complex genetic information, feelings of denial, rationalization, the desire to protect others or perceptions of stigma related to genetic disease (Wilson *et al.* 2004; Holt 2006). Parental guilt and blame around having a child with CF can prevent any discussion about genetics in a family. If one parent uses blaming 'the other side of the family' as a coping mechanism to deal with guilt, that parent would choose to avoid evidence that may point to their own genetic contribution to the condition (Fanos and Johnson 1995). Although parents generally regard the dissemination of risk information as their responsibility, they may feel burdened by having to deliver what they consider to be bad news (Clarke *et al.* 2005). There may be an internal struggle between the perceived obligation to provide information and a concern not to alarm (D'Agincourt-Canning 2001).

Another suggestion has been that there may be too few cases of affected members in a family for them to recognise a clear pattern of inheritance. If the affected person is a more distant relative there may be less discussion in the family about the hereditary implications of the disease and an underestimation of potential carrier risk. Close presence of an affected family member may increase the likelihood of carrier risk information being disseminated (Henneman *et al.* 2002).

Research suggests that disclosure of genetic information is a gendered activity, with the benefits and burdens of the task falling mainly on women (D'Agincourt-Canning 2001). Richards (1996) observed that female family members take the primary role in almost all aspects of genetic activity. Women are most often the ones to collect information about their own family and that of their partner. They take the lead in exchanging information about a genetic condition, even in cases where it originates on the paternal side of the family.

2.6.4 Facilitators of risk communication

The occurrence of effective communication should not be underestimated. Family members are an important source of information about the nature and inheritance of genetic disorders (Weil 2000). Common reasons for disclosing information about carrier status have been a close social relationship with the relatives and the need for social support in a time of crisis (Ormond *et al.* 2003). Studies suggest that some individuals regard their relatives as having a right to the information they receive and perceive themselves, rather than genetic counsellors or clinicians as having a moral obligation to disseminate this information to their at-risk family members (Hallowell *et al.* 2005). Genetic counsellors' work is not with and about individuals, but is directed at families. Communication within the family should receive special attention during counselling. The genetic counsellor can explain the determination of genetic risk to the proband or close kin, but cannot directly control how such information is transmitted or not transmitted among the social network of the extended family (Featherstone *et al.* 2006). Written material describing the implications of a diagnosis, may aid parents in informing their families and thus assist them in fulfilling their moral responsibilities to others (D'Agincourt-Canning 2001).

Genetic counsellors and health care professionals need to develop effective ways to help patients and clients to deal with communication issues in families, because this is by no means a straightforward issue. In order to do this, it is essential to explore how communication occurs in families. This includes who parents communicate with; how and what information is shared; to whom they feel responsible and what leads to information being passed on or not (Forrest *et al.* 2003).

2.7 IMPACT ON REPRODUCTIVE CHOICES

The genetic cause of CF has an impact on reproductive choices of the affected child, parents and extended family. The autosomal recessive nature of this condition means that parents of children with CF have the following risks with each pregnancy: 1 in 4 chance of having another affected child, 50% risk of having a child who is a carrier of a

CFTR mutation and 25% chance of having a child who is neither a carrier, nor has CF (Nussbaum *et al.* 2004). Investigating the reproductive choices of parents of children with CF is important in gaining insight into the possible needs for counselling of carrier couples in the reproductive decision-making process.

Studies have shown that the birth of a CF child has a major impact on subsequent family planning (Henneman *et al.* 2001; De Braekeleer *et al.* 2000; Evers-Kiebooms *et al.* 1990). This is attributable to the recurrence risk of CF and also to problems parents have experienced with their CF-child in the past, as well as to future problems they fear (Evers-Kiebooms *et al.* 1990). In their survey amongst parents of children with CF, Henneman *et al.* (2001) found that the major reason the diagnosis influenced their reproductive decisions was that they did not want another child with CF. The couple's experiences with CF had a major influence on future choices. Knowledge of recurrence risk has resulted in parents postponing pregnancies or deciding against further progeny (Evers-Kiebooms *et al.* 1990; De Braekeleer *et al.* 2000). The decision of having another pregnancy after the birth of a CF child is partly determined by the presence or absence of another healthy child in the family (Evers-Kiebooms *et al.* 1990).

A few studies have focused on the effect of the birth rank of the affected child and have reported that couples were much more likely to have more children when the affected child was firstborn (De Braekeleer *et al.* 2004; Evers-Kiebooms *et al.* 1990; Steele *et al.* 1986; Kaback *et al.* 1984). These studies reported that that the probability of initiating a pregnancy was between 47 and 69% when the CF child was first-born and between 12 and 22% when he/she was second-born. Possible explanations for these findings could be the desire for a healthy child or to reach an ideal number of children. Other reasons include misunderstanding of the recurrence risk or the perception that it is not enough to prevent the parents from having another child (De Braekeleer *et al.* 2000). Parents have also been shown to use more reliable contraceptive methods after the birth of their CF child (Evers-Kiebooms *et al.* 1990). There is a lack of recently published data on these topics.

Faced with the knowledge that future children have a 25% chance of suffering from CF, parents are presented with a dilemma. A genetic counsellor should point out the various options that they can consider (Hodson *et al.* 1983). Several reproductive options are available to parents of a child with CF. They can choose not to have more children; accept the risk of giving birth to a child with CF; opt for prenatal diagnosis, possibly followed by selective termination of an affected pregnancy; or consider options such as artificial insemination, prenatal genetic diagnosis (PGD) or adoption (Henneman *et al.* 2001).

Although most parents believe that prenatal diagnosis is an important reproductive option for families at risk (Watson *et al.* 1992; Conway *et al.* 1994), the actual utilisation of prenatal diagnosis is, however, generally lower than was expected from earlier studies when this option was not yet available (Jedlicka-Köhler *et al.* 1994; Mischler *et al.* 1998; Sawyer *et al.* 2006). The use or intention to use prenatal diagnosis is influenced by factors such as religion, family income, education, family size, the presence or absence of healthy children born before the affected child and the willingness to terminate a pregnancy for CF (Evers-Kiebooms *et al.* 1990; Borgo *et al.* 1992; De Braekeleer *et al.* 2004).

Attitudes toward abortion among parents of children with CF showed a wide spectrum. The majority of studies report that surveyed parents support termination of an affected pregnancy (Al-Jader *et al.* 1990; Evers-Kiebooms *et al.* 1990). Correlations have been shown between abortion attitudes and religion, education and interpretation of risk. Parents would often indicate their intentions to make use of prenatal diagnosis, but not opt for termination should the foetus be affected (Henneman *et al.* 2001). Despite the fact that pregnancy termination is a logical implication of prenatal diagnosis on the cognitive level, it is not experienced as such on the emotional level (Evers-Kiebooms *et al.* 1990). Furthermore, parents who consider terminating an affected foetus may feel guilty because of the feeling that it is an implicit rejection of their child with CF (Polnay *et al.* 2002). Close attention should be given to the counselling and support of carrier couples in the decision-making process (Henneman *et al.* 2001).

Chapter 3

METHODOLOGY

3.1 INTRODUCTION

In this chapter the methodological process used to conduct this research study is discussed. The reasons and motivations for having chosen a particular methodology are provided, potential sources of bias are identified and attempts at minimising these are described in the relevant sections.

3.2 RESEARCH DESIGN

This qualitative research project was designed as an interview-based cross-sectional descriptive study, using a 'multi-method' approach of both qualitative and quantitative methods.

3.2.1 Qualitative Research

A qualitative approach was selected as it aims to understand and describe ordinary events in natural settings, attempts to make sense of, or interpret phenomena in terms of the meanings the people concerned bring to them (McMillan and Schumacher 2001). In this way it provides one with a strong handle on reality. Qualitative research, with the emphasis on people's lived experience is well-suited for identifying the meanings people place on the events, processes and structures of their lives and for connecting these meanings to the social world around them (Miles and Huberman 1994). Qualitative research is based on the premise that individuals are best placed to describe situations and feelings in their own words, adding richness to the data (Holloway and Wheeler 1996). The researcher attempted to examine the perspectives and experiences of parents of children with cystic fibrosis attending the CF clinic at RCCH (Holloway 1996).

A cross-sectional study investigates a sample of the population at a single point in time, rather than the same subjects at different points in time, as in longitudinal studies (McMillan and Schumacher 2001). In this study data were collected in a single session by means of interviews conducted with the participants. To provide a broad representation of the CF population attending RCCH in this study, individuals in the sample were of different ages, ethnic groups, educational and income levels (Bailey

1994). Cross-sectional studies are essentially descriptive. Although this method is less costly and simpler than longitudinal studies, the disadvantage of this kind of study is that the circumstances which influence the lives of the participants over time are not captured (Neuman 2006).

Descriptive studies aim to describe phenomenon in detail. They are concerned with the current or past status of phenomenon and describe characteristics of a group of subjects, without manipulation of independent variables (McMillan and Schumacher 2001). The researcher searches for accurate information about the characteristics of a sample or about the frequency of the occurrence of a phenomenon. After identifying and defining variables of interest, such as opinions, attitudes, needs or facts, they are described to provide a full, clear picture of the phenomenon as it exists in reality (Brink 2006).

3.3 SAMPLE

3.3.1 Population

There are over 60 children with CF ranging from the age of 1 month to 18 years old attending the CF clinic at the RCCH.

For the purpose of this study, the parents of 12 children from different families with CF who met the inclusion criteria and were from different socioeconomic groups were recruited from the CF clinic.

Time and cost constraints prevented the inclusion of a larger sample from a larger geographical area.

3.3.2 Eligibility criteria

i) Inclusion criteria

- Parents of children with CF between the age of 2 and 18. Since the majority of CF cases are diagnosed during the first year of life, the lower age limit of 2 years

was chosen to ensure that parents had adequate time following the diagnosis of their child to disclose CF carrier information to relatives. Individuals older than 18 do not attend the CF clinic at RCCH.

- Parents who regularly attend the CF clinic at RCCH with their child.
- Parents of children who have been diagnosed with CF for at least 1 year prior to being approached for this study.
- Parents who live within a 60 km radius of RCCH.
- Parents of children with CF who consented to audio-taped interviews.
- Parents with either Afrikaans or English as home language.

ii) Exclusion criteria

- Parents of children who have other medical conditions in addition to CF.
- Parents who have lost a child to CF.

3.3.3 Sampling method

Based on the underlying principle of qualitative research to gain rich, in-depth information, a small sample was selected for a specified purpose. Convenience sampling was used, where participants are selected on the basis of being accessible to the researcher and being representative of the general population of children with CF attending the CF clinic at RCCH (McMillan and Shumacher 2001). The first 12 parents of children from different families who met the eligibility criteria and attended the CF clinic during the period of November 2006 – January 2007 were identified by the paediatrician. Appropriate individuals from a variety of socio-economic and ethnic backgrounds attending the CF clinic during this time were identified and informed about the objectives and method of the study. Individuals who agreed to participate were then contacted by the researcher to arrange a suitable date, time and venue for the interviews, which took place during the period of February - May 2007.

3.4 STUDY METHODS AND MEASURING INSTRUMENTS

3.4.1 Interviews

A semi-structured interview schedule designed by the researcher was used to gather the data (Appendix II). Individual interviews are frequently used in descriptive studies as they provide the most direct method of obtaining facts from the participant and are particularly useful in ascertaining values, beliefs, attitudes and experiences (Brink 2006). Semi-structured interviews allow the researcher to address questions of interest with flexibility to explore in depth other emergent issues (Patton 1987). A variety of questioning techniques were used as each provides different outcomes. Open-ended questions were framed to invite broad responses from participants and therefore allowed them to respond as extensively or specifically as they chose. It also provided the opportunity for in-depth questioning of the topic of concern. Closed-ended questions were those that generally have "yes" or "no" answers or ask for a specific aspect of information, such as socio-demographic details. Prompt questions were used to guide the discussion to maximise the amount of relevant information gathered during the interview (Smith *et al.* 1995). The researcher started the interview by taking a family history and drawing a genetic pedigree to identify the relatives relevant to this study and the participants' relationship to each relative. A pedigree or family tree provides a permanent record in a standardized format of the most important family information and is a valuable adjunct to clear thinking about family relationships (Schuette and Bennett 1998).

The section of the interview schedule on level of understanding of the genetics of CF, was adapted from interviews used by Hames *et al.* (1991) and Somer *et al.* (1988). Content validity of the interview schedule was established by being critically reviewed by two research supervisors to ensure that all the relevant questions had been included and that they had been sequenced appropriately (Neuman 2006).

All interview schedules, information sheets and consent forms were available in English and Afrikaans. As the researcher is fluent in both English and Afrikaans, the interview was conducted and the data captured in the language of the participants' choice.

3.4.2 Research setting

The interviews were conducted in a private venue of the participants' choice. Ten interviews took place at the homes of the participants. Two participants chose to have the interviews at their place of work, in a private room. As individuals are likely to feel more comfortable in their home environment when discussing issues they might find sensitive, this was the preferred site for interviews. The researcher chose not to have any interviews in the clinic environment, because a period of observation in the home environment may have given more information than is obtained in a formal clinic meeting (Smith *et al.* 1995). It allowed the researcher the opportunity to observe reactions to questions; both verbal and non-verbal; interaction of the individuals with other family members, the environment in which they live, the home circumstances which are likely to affect their daily lives; as well as the level of support they receive from their extended families. These aspects could easily be observed by the researcher without the need for direct questioning.

3.5 PROCEDURE

The paediatrician at the CF clinic at RCCH informed the parents of the purpose and method of the study at a routine clinic visit. The consenting individuals were then contacted by the researcher to arrange interview times and venues.

3.5.1 Piloting

A pilot study was undertaken with two participants to test the interview schedule to determine if questions were easily understandable. It ensured that no ambiguous questions were asked and established how much time was needed for the interview. The procedures in the pilot interviews were identical to those implemented during the study. A pilot study is a good way of improving the reliability of the interview schedule by checking for bias in the questions, interviewer and procedures (Neuman 2006).

Following the two pilot interviews, adjustments were made to the interview schedule to ensure the logical order and clarity of questions. In this study interviews were found to

take approximately 45–60 minutes. The data obtained in the pilot study was not included in the results.

3.5.2 Recruitment

The researcher attended the weekly CF clinic during the recruitment period of November 2006 to January 2007, to establish contact with recruited individuals and to repeat the aims of the research. If both parents were available they were interviewed together. If only one parent was available or, in the case of single parents, only one parent was interviewed. When contacting the individuals, the researcher informed them that there should be no extended family members present at the time of the interview.

3.5.3 Data collection

At the beginning of the interview, the participants were reassured of the following:

- The information they provided during the interview (which would be audio-taped) would be kept confidential, apart from a possible publication in a scientific journal where no names would be used;
- Information would not be discussed with extended family members; and
- Participation was completely voluntary and that they were free to choose not to participate or withdraw from the study at any time without jeopardizing their access to the medical services to which they were entitled (Appendix I).

All semi-structured interviews were conducted personally by the researcher. Each interview was audio-taped and transcribed verbatim by a professional transcriber who was fluent in both English and Afrikaans, for data analysis. Audio-taping allows the researcher to capture the exact words of the interview, preventing the possibility of forgetting important areas and allowing the researcher to maintain eye contact with the participant and concentrate on the interview procedure (Holloway and Wheeler 1996). Two transcripts were randomly selected and then checked for accuracy by the researcher and research supervisor.

Due to the fact that some of the questions asked during the interview may have raised sensitive issues, a follow-up visit was scheduled if the participants felt the need. The researcher contacted the participants a month after the interview to arrange a follow-up visit. The follow-up visit provided the opportunity for counselling the parents regarding sensitive issues evoked during the interview and to answer any questions they may have had.

3.6 DATA ANALYSIS

The transcripts of the interviews provide the raw data for and a descriptive record of the research, but cannot be meaningful as it is. The researcher has to make sense of it by systematically sifting through the data and interpreting it (Pope *et al.* 2000). In this qualitative study, textual data was explored by content analysis and, contrary to quantitative analysis, the aim was not to quantify data, but rather to use analytical categories to describe and explain social phenomena (Neuman 2006; Pope *et al.* 2000).

Content analysis is most often used for descriptive purposes (Neuman 2006). It involves identifying coherent, important themes and patterns in data. The researcher identified observations or quotations that described the same underlying issue, idea or concept (Patton 1987). Qualitative studies rely on inductive reasoning processes to interpret and structure meanings that can be derived from the data (Thorne 2000). Data was read and reread to identify themes and categories. The interpretation of the data into themes and categories was verified by the research supervisors.

Simple counts are sometimes used in qualitative data analysis to provide a useful summary of certain aspects of the analysis (Pope *et al.* 2000). Responses to certain sections of the interview schedule were categorized by frequency and descriptive statistics such as percentages, means and averages as calculated to describe the occurrence of the data.

3.7 TRUSTWORTHINESS/VALIDITY

Trustworthiness is used to measure the validity or truthfulness of qualitative research (Holloway and Wheeler 1996). A qualitative study is trustworthy when it represents reality. Qualitative researchers are interested in authenticity. Authenticity implies giving a fair, honest and balanced account of social life from the viewpoint of someone who lives it every day (Neuman 2006). Guba and Lincoln (1989) identified the following elements of trustworthiness: credibility, transferability, dependability and confirmability.

Credibility can be compared to internal validity, which is used in quantitative research. The researcher needs to demonstrate that he/she has presented the reality of the participants through excerpts from their interviews. The researcher's findings have to be compatible with the perceptions of the individuals involved in the study and the participants must be identified and described accurately (Holloway 1997). Peer debriefing is an important means of improving the credibility of a study. The researcher met with her supervisors on a regular basis to ensure that the data were interpreted accurately.

Transferability is about how the findings of a study can be generalised or transferred from a representative sample of a population to the whole group (Holloway and Wheeler 1996). It is therefore important for researchers to describe the data in their context as accurately and in as much detail as possible in order to provide peers and readers with a full, clear picture of how the study was conducted. This in turn allows them to decide whether the findings of the study are applicable in another context or setting.

For a study to be dependable it must be consistent and accurate. This was demonstrated by providing detailed descriptions of the methodology (Holloway 1997).

Confirmability assures that the findings, conclusions and recommendations of the study are supported by the data and that there is internal agreement between the researcher's interpretation and the actual evidence. This was achieved by confirming data with a telephone call to the participants (Brink 2006).

3.8 ETHICAL CONSIDERATIONS

3.8.1 Ethical approval

This study was granted approval without reservations by the Medical Research Ethics Committee of the University of Cape Town (Reference number 380/2006) (Appendix III).

3.8.2 Consent

The individuals were identified by the paediatrician involved in the CF clinic at the RCWCH who explained the purpose of the research to be conducted. No form of persuasion was used to encourage the individuals to participate. The paediatrician informed the individuals that a home visit would be undertaken by the researcher where an interview would take place, unless otherwise requested by the participant. Written consent was obtained by the researcher from each participant before commencement of the interviews (Appendix I). Where both parents were present for the interview, signed consent was obtained from each. Consent included permission to audio-tape each interview. Participants were assured that if they did not wish to participate it would not have any influence on their future medical management at the CF clinic.

3.8.3 Confidentiality

As confidentiality was of central concern, audio-taped recordings were transcribed as soon as possible following each interview. The audio-tapes and transcriptions were stored in a safe in the Division of Human Genetics and were destroyed once the study had been written up. The participants received numerical codes and their names did not appear on the interview schedules, the tapes, the transcripts or spreadsheets.

3.8.4 Risk Benefit

The risk to the participants in this study was the discussion of sensitive information and the possibility of evoking distressing emotions. The researcher ensured that confidentiality and anonymity were maintained. The researcher remained sensitive to the

emotional state of the participant throughout the interview. They had the opportunity of a second session where any emotional issues evoked during the initial interview could be dealt with or any questions that the participant may have had could be answered.

The long term benefit of this study will be to use the information to, if necessary, improve the genetic counselling processes to support the individuals and their families that attend the CF clinic at RCCH.

3.9 ASSUMPTIONS

The researcher assumed that the responses of the participants were honest and a true reflection of their lives.

3.10 LIMITATIONS AND STRENGTHS OF THE STUDY

3.10.1 Limitations of the study

- The major limitation of this study was the small sample size, due to budget and time constraints. Thus, the findings will not be generalizable to a different population;

- Another limitation is the paucity of information, literature and research concerning CF in developing countries. Where relevant, information gathered in other countries was used, while bearing in mind that caution needs to be exercised in generalising from communities of one country to another, from one geographical area to another and from one socio-economic area to another; and

- The researcher had limited interviewing and counselling experience and may therefore not have been able to elicit information to its full potential during the interview.

3.10.2 Strengths of the study

- The researcher conducted all interviews in person;
- Open-ended questions in the interview schedule allowed participants to express themselves without being limited by set categories;
- Audio-taping of all interviews allowed for a more complete record than handwritten notes by the researcher;
- The trustworthiness of the study was increased by including questions in the interview schedule that were used in similar international research; and
- As most of the interviews were conducted in participants' homes, they were more likely to feel comfortable to discuss emotional issues than in a clinical environment.
- The researcher was an outsider to the CF clinic team and therefore, had no vested interest or private agenda for personal gains. This allowed the participants to talk more freely about personal and emotional aspects than with people they were accustomed to seeing at the RCCH CF clinic

Chapter 4

ANALYSIS, FINDINGS AND DISCUSSION

4.1 INTRODUCTION

The analysis and findings of the research are presented in this chapter. Data are presented in tables, followed by a discussion. When possible, reference is made to literature in order to demonstrate similarities and differences to other studies on CF. In total, 10 interviews were conducted with the parents of children with CF.

4.2 INTERVIEW PROCEDURE

Six of the interviews were conducted with the mother of the affected child. Both parents were present at the other four interviews. With the exception of two, all interviews were conducted in the participants' homes. The other two interviews took place in private rooms at the participants' place of work.

All of the interviews took less than an hour and a half to complete. None of the participants expressed the need to have a second interview.

To ensure confidentiality, the participants will be referred to by number throughout this chapter. The participant number refers to the adult/adults who were interviewed. Direct quotes of the participants' responses are included to provide the reader with greater insight into their thinking.

4.3 SOCIODEMOGRAPHIC INFORMATION OF PARTICIPANTS

A summary of the sociodemographic information of the participants is presented in Table 4.1.

Table 4.1: Summary of the sociodemographic information of the participants

P	Age of affected child	Marital status	Level of education		Current occupation		Household income per month (indicated by an income bracket)
			Mother	Father	Mother	Father	
1	16	Married	Gr 12	N/A	Housewife	Building contractor	R12801-25 600
2	13	Married	Tertiary	Gr 12	Teacher	Technician	R12 801-25 600
3	10	Divorced	Gr 12	N/A	Police sergeant	N/A	R6401-12 800
4	4	Married	Gr 12	Gr 12	Food and beverage distributor	Administration	R25 601-51 200
5	7	Married	Tertiary	Gr 9	Housewife	Nurse	R6401-12 800
6	15	Divorced	Gr 8	N/A	Self-employed	N/A	R3201-6400
7	13	Divorced	Tertiary	N/A	Self-employed	N/A	R25 601-51 200
8	10	Remarried	Gr 12	N/A	Human resources	N/A	R3201-6400
9	16	Single	Gr 10	N/A	Part-time carer	N/A	R1200
10	3	Married	Tertiary	Gr 12	Part-time Admin assistant	Teacher	R12 801-25 600

The participants included ten females and four males ranging in ages between 30-47 years old. Six individuals were of Caucasian origin and the remaining eight belonged to the Mixed Ancestry Population group. The majority of participants had completed high school and four individuals had gone on to complete tertiary education. The majority of participants were of a higher socioeconomic group. The average household income for each participant is presented in Figure 4.1.

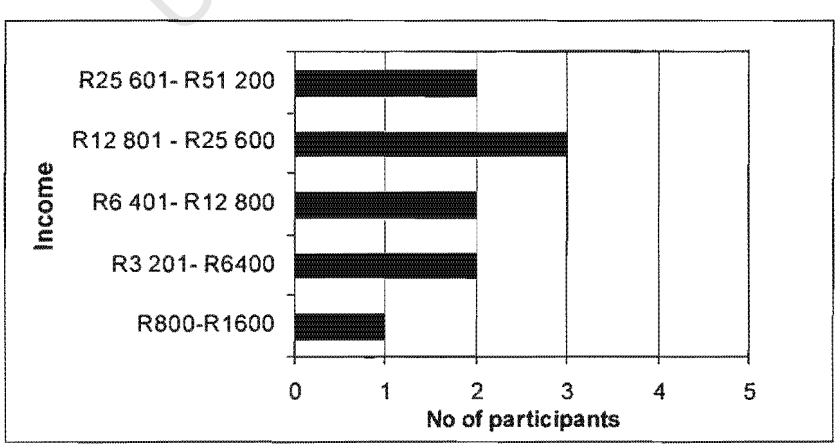

Figure 4.1: Average household income per month (n=10)

According to the latest information available from Statistics SA (2001), the average annual household income in the Western Cape (WC) in 2000 was R45 000 (R3750/ month). With the exception of P9, all individuals had a monthly income above the average income in WC. P9 was a single mother living with an elderly lady who raised her in a household with 16 other people. She is employed on a part-time basis on weekends to take care of a pensioner, for which she receives R300/week. R200 of this goes toward board and lodging every week, leaving her with R100/week for herself. Her affected son has been removed from her care, because she is unable to provide for him. He only comes to visit her during school holidays and she accompanies him to clinic visits at RCCH once a month.

Four of the participants were divorced mothers, one of whom had remarried. Only one of these women (P6) reported that CF played a big role in her reason for separating. After their child was diagnosed with CF, her husband blamed her and her family for the condition:

"*It put strain on everything, they (his family) were always the ones that did nothing wrong and it always came from us*". Blame, particularly directed at the mother, is facilitated by the male tendency to externalize their emotional responses to devastating news in the form of blame, while women are likely to internalize their responses and accept the blame. These gender differences in placing and accepting blame, allows the father the opportunity to blame the mother and in this way he is able to separate himself from the shame of the ill child. This can lead to poor family functioning (James *et al.* 2006).

There was one case of consanguinity. P7 was from an arranged marriage with a first-cousin; their parents were siblings. She ascribed the divorce to the fact that it was an arranged marriage with a man she never loved, and not in any way related to CF.

Studies have shown that the incidence of divorce and separation is no higher than average amongst parents of children with CF (Gayer and Ganong 2006; Cowen *et al.* 1986; De Wet and Cywes 1984).

4.4 RCCH CYSTIC FIBROSIS CLINIC

Participants were asked various questions regarding the RCCH CF clinic, to determine the accessibility of the clinic and the frequency of visits. Table 4.2 illustrates some of the important aspects of the clinic.

Table 4.2: Accessibility of and frequency of visits to the CF clinic

P	Referred by	How often	Public Transport	Cost	Travelling time
1	CF clinic, Johannesburg	2 months	No	Unknown	25 min
2	Pulmonologist	2 months	No	Unknown	30 min
3	Paediatrician	3 months	No	Unknown	20 min
4	Paediatrician	2-3 months	No	Unknown	20 min
5	RCCH	3 months	Yes	R32	1 hr
6	GP	1 month	Yes	R40	45 min
7	Great Ormond Street Hospital, London	3 months	No	Unknown	20 min
8	Paediatrician	3 months	No	R50	30 min
9	RCCH	1 month	Yes	R24	30 min
10	Paediatrician	3 months	No	Unknown	50 min

The majority of participants in this study had access to their own transport. Clinic visits ranged between monthly to three-monthly visits, depending on the affected child's health. No participants mentioned any particular difficulties in getting to and from the CF clinic. Travelling time ranged between 20 to 60 minutes.

Participants were asked whether they had medical insurance and to what extent this covered their child's medical needs, to gauge the financial impact of the condition on the family.

Table 4.3 Medical insurance for each participant

P No	Do you have medical aid?	What does it cover?			Comments
		Consultation	Medication	Hospital stays	
1	Yes	✓	✓	✓	Until depleted
2	Yes	✓	✓	✓	Until depleted
3	Yes	✓	✓	✓	Pay up to R30 000/year
4	Yes	✓	✓	✓	None
5	Yes. "Was forced to join because the bills were too sky high"	✓	X	Don't know yet	Doesn't cover medication
6	Yes, only affected son. Through Grandfather.	✓	✓	✓	Worried about depletion
7	Yes	✓	"Only up to a point"	✓	"They choose when they pay and what they pay"
8	Yes	✓	X	✓	Only R30 000/year
9	No	n/a	n/a	n/a	None
10	Yes	✓	X	✓	Only R6500 available for medication/ year

4.4.1 Discussion on medical insurance

All the participants, except P9 had medical insurance for their affected children to varying extents. The father of P6 worked for the City council before he died in September 2006. He had arranged that his grandson be put on his medical insurance, which covered all his medical costs. P6 was extremely worried about what was going to happen with her son's medical costs after her father passed away, but when she enquired they told her that her father had made arrangements for his grandson. She commented: "*All I have to do is pay R40 a month to the medical aid. I was over the moon.*" She was however very concerned that with the current monthly bills from RCCH of R10 000-12 000, the medical aid would soon be depleted and this was of great concern to her, since she explained that there was no way she could personally pay for any of his medical expenses.

Several participants reported having problems with depletion of medical insurance, because CF is not recognised as a chronic illness on most medical aid schemes.
P3: "*Halfway through the year the R30 000 is completely depleted and then we have to start buying medication for cash and that's like, yes, well, you can imagine…*"

P8 indicated that her medical insurance provided her affected son with R30 000/year to cover all his medical expenses.
P8: "*It's nothing. It's only 3 months worth of medication. RCCH phones me all the time, my bills add up to R50 000, what must I do? His father refuses to contribute.*"(Translated)

P10: *In the beginning the funds lasted half a year, because Creon is so expensive. The next year it was only 4 months, this year only 3 months....I've written letters, the doctors at the clinic have written letters.*" (Translated)

By the time all the deductions have been made, including medical insurance, P6 is left with R3000/month. Because the medical insurance doesn't cover the affected child's medication, once this has been deducted from their monthly income, they have R1000 to support a family of 7. "*I've received lawyers' letters...It's a bit heavy. Every month is a struggle in this household*" (Translated)

4.4.2 Diagnosis of cystic fibrosis

As part of the section on the CF clinic, participants were asked questions to determine how they experienced the period of diagnosis. Their responses to certain questions are summarized in Table 4.4.

Table 4.4: Experiences of the diagnosis of CF

P	Age at diagnosis	Presenting Symptoms	Reactions to diagnosis
1	Few days	"Born with a blockage, operated when 2 days old, tested for CF"	"Devastated"
2	9 yrs	Chronic cough, sick baby, sick child. Constant lung problems	"Cried for nights on end…slept very little. It was like a death sentence
3	3 mnths	Losing weight after birth…in and out of hospital…different tests done. Couldn't figure out what was wrong	"It was a stressful period…but in a sense it was better knowing than not knowing"
4	4 wks	"Wasn't putting on weight, had a lot of pooh nappies." After trying everything referred to paediatrician who had diagnosed CF before.	"Devastated. At the time it was horrific…It gets a lot easier with time"
5	3 mnths	Started with a cold, became bronchitis and didn't get better.	"Shock…you think it could have been prevented somehow. We had to work through the trauma ourselves…
6	2 ½ yrs	Chronic bronchial bronchitis and diarrhoea	"Completely devastated. It was really bad"
7	18 mnths	Wasn't putting on weight, but happy toddler. Got a cold on her 1st birthday and couldn't get rid of it. Then the extreme coughing…	"Devastated"
8	5 mnths	Went from doctor to doctor with blocked nose, phlegm, cough. One says its sinus the next says it can't be. Until found doctor who had worked in CF clinic in London	"Broken, terrible. I cried for days. The doctor tells you your child is dying and it's the utmost worst thing anyone can tell you"
9	2 wks	"Everything he ate and drank just came out again"	"Heartsore"
10	6 wks	"Didn't put on weight, lots of dirty nappies after each other"	Shock

Shock and devastation were the most common reactions to the diagnosis of CF. Shock is a common reaction, especially when the diagnosis was not expected (Ross and Deverell 2004). Jedlicka-Köhler (1996) described initial shock-like reactions in 54% of parents following the diagnosis of CF in their child.

Only one participant described feeling a sense of relief at finally understanding what was wrong with her child after three months of being in hospital and having every test imaginable:

P3: "*You can deal with something if you know what you're dealing with, but when you don't know what is happening, then you are stressed out all the time.*"

At the time of diagnosis P4 was the only participant in which there was another individual in the family with CF. Although they reported that it meant it was not the first time they'd heard about the condition, it still came as a big shock:

P4: "*But you still never believe that this person you've met and decided to marry would be a carrier...or that I was a carrier, you know it's in your family and what not, but you still don't think about it.*"

The affected child of P2 was only diagnosed at the age of nine years and mother and father admitted to reacting differently to the diagnosis:

Mom: "*I cried for nights on end...slept very little. It was like a death sentence.*" (Translated)

Dad: "*I felt completely different. To me it was like they still had to convince me. I'm a believer and I want to see hard facts.*" (Translated)

Differences between men and women in their response to stress and their processing of loss and grief have been well described. Women are generally more emotionally expressive and consciously affected by grief over a longer period of time. Men on the other hand, are less open to discussing their emotions and return more quickly to social involvement and work activities (Weil 2000). This father was also experiencing a certain amount of denial. Denial, the inability to acknowledge to oneself certain information or news, is a common response when information elicits shock or fear, as in the case of the

diagnosis of a genetic condition in one's child (Djurdjinovic 1998). Denial is a defense mechanism used to ward off anxiety and other distressing emotions. It is used to avoid the reality of the situation, until an individual is ready to come to terms with it (Weil 2000). Denial can manifest in various ways; it may be the rejection of the diagnosis, its permanence or its impact (Falvo 2005).

P4 voiced her feelings and initial concerns about what the future might entail when faced with the diagnosis of genetic condition and chronic illness, such as CF.
P4: "*Just devastated...at the time it was horrific...with time it obviously gets a lot easier...as time goes by you see you can live quite a normal life...at that time you have no idea of what the future entails, so then it is terrible. And being your first child, you don't know if it's going to be your only child, how bad life's going to be. But yes, it gets a lot easier as time goes by.*"

P10 also described that after the initial shock it does get better with time:
"*We were very shocked. It felt like the whole world was on our shoulders. It just came down on us too quickly. We just never expected to have a child with a chronic illness. But with time, we've learned to cope.*" (Translated)
With time, together with being confronted by the reality of the situation, the initial shock subsides and intellectual acceptance of parents increases (Weil 2000). Venters (1981) reported that by the end of the first year after the diagnosis had been made, family life became less stressful, strained family relationships eased and new routines lessened the initial disruption caused by the diagnosis and its implications. Furthermore, a certain amount of agreement had been reached among family members about future goals and aspirations.

Two participants mentioned the way in which the paediatrician who gave them the diagnosis handled the situation and how welcoming the CF clinic was the first time they attended:

"The private paediatrician gave us a crash course on CF. He wasn't very passionate...compassionate. He did not come down lightly. He was very bombastic and very direct. He could have put it a little better. He just said: you can't wish it away, you can't medicine it away. When we got to Red Cross it was like walking into a place of angels. It was like a haven, because of the way things were presented to us." (Translated)

P10 gave a similar description of their first experience at the CF clinic after the diagnosis was given to them the month before by their private paediatrician:
"We got a big fright when the doctor told us. He was talking about how long our child would live and things like that. When we walked out of there it felt like the whole world was on our shoulders. It was the first time we'd heard of it and it all sounded so weird....When we got to Red Cross we were expecting the worst because of what we'd been told. But when we left it felt like a mountain had been lifted from our shoulders." (Translated)

4.5 LEVEL OF GENETIC KNOWLEDGE

Several studies have been conducted to determine the level of genetic knowledge of families with CF (De Braekeleer *et al.* 2001; Hames *et al.* 1991; Denayer *et al.* 1990; Henley 1988; Nolan *et al.*1986; McCrae *et al.* 1973, Leonard *et al.* 1972). Participants in this study were asked a range of questions to assess their knowledge about the genetics of CF. The responses to selected questions are presented in Table 4.5 (p51).

Table 4.5: Participants' understanding of the genetics of CF

P	Cause of CF	Inheritance	Recurrence risk	Unaffected children carriers	Siblings carriers	Siblings affected child
1	"Double dose of faulty gene"	"Both me and my husband are carriers of the Δf508 gene"	1 in 4	"1 in 4. She is a carrier, she was tested"	"If I had to guess…50%"	50/50
2	"Inherited condition"	"Carrier marrying a carrier- ¼ children will have CF"	Chance is high 1 in 4 is just a statistic	"1 out of 3 remaining kids"	We were told if it was somewhere it would be in the cousins	"Don't know, but you would have seen it by now"
3	"Thick secretions prevent pancreatic enzymes from breaking down food"	Through the generations	"Probably the 3rd child would be affected"	Good possibility	"Really don't know"	"Don't know, but they're not having anymore"
4	"Both of us having a faulty gene"	"Both parents having gene, then 1 in 4 chance of having child with it"	1 in 4	50%	50%	"Same as me if their partner also has the gene, if not, no chance"
5	"Too much salt in the blood"	We are both carriers and passed it on to our child	100%	Nil	"Don't know"	"Actually negative"
6	Genetic disorder. Bad genes that don't develop properly	Didn't know it was an inherited disorder until another child diagnosed	"60 out of a 100"…small chance	Very big 100%	"Very slim"	"Very slim. There are no signs in their children of being CF of carriers".
7	"2 parents have faulty gene and they come together and cause CF"	"Genetically"	1 in 4	1 in 4	1 in 4	"They don't have the genes, they've been tested"
8	"Genetic disorder"	Two carriers	2 out of 3	It can happen, good chance	"There is a chance"	There is a chance
9	Problem lay with the father	Don't know	Don't know	Don't know	Don't know	"He (brother) doesn't have it and his child doesn't have it"
10	"2 negative cells from mom and dad melt together"	"They talked about carriers"	"Something like 1 in 4"	"He can be a carrier"	"They can"	"The possibility is there"

4.5.1 Misconceptions regarding the genetics of cystic fibrosis

There were many misconceptions and inaccurate responses with regards to the genetics of CF.

P9 had no idea about the genetic nature or cause of CF. She believed that CF came from the father. He was murdered while she was pregnant with her affected child. When asked about what she understood the cause of CF to be, she said:
"*The way the doctor explained to me, they did a sputum sample on me and the problem didn't lay with me, so it must have lay with the father, but they couldn't test him.*" (Translated).

P6 knew CF was a genetic disorder, but was completely unaware of the inheritance of the condition, she thought it was just:
"two genes that didn't develop properly", from both her and her husband. Until a few months ago when another child (son of her husband's sister) was also diagnosed with CF. This came as a big shock to them, because her impression was:
*"It's a genetic disorder. It is not inherited. It's not passed on in the family."(*Translated)
She further believed that neither of her other two children were carriers because they had a sweat test after their brother was diagnosed. "*They went for a sweat test and they aren't even carriers of CF.*"(Translated). Further indicating that she wasn't aware that carriers could not be detected with a sweat test or indicating a lack of understanding of the carrier concept. Difficulty with fully understanding the carrier concept was also indicated by the responses of other participants.

When P3 was asked about the chance of her unaffected daughter being a carrier, she explained that "*there was a good possibility*" that she could be, but she wasn't sure. The unaffected child had a sweat test after she was born and the mother was told:
"*No she is not a carrier. But I don't know if that means she is not an active carrier like (the affected child), or if she perhaps is just a carrier of CF until such time as when she has her own children.*"

Furthermore, despite not knowing whether she inherited the CF mutation from her mother or her father, P3 was not worried about her brother's potential carrier risk, because they didn't share the same mother.

P2 mentioned that the inheritance of CF involved a carrier marrying a carrier, but when asked a question relating to whether the faulty gene was transmitted by father, mother, both or neither, they responded:
Mom: "*For our own peace of mind we say both…*"
Dad: "*But I don't think you can determine that, or can you? If I haven't got it wrong it's a weakness from the father's side. Bad genes...these boys can't perform so well.*" (Translated)

There were several misconceptions about the recurrence risk and difficulty understanding the risk-concept. Several participants didn't understand that the question about what their chances were of having another affected child and what their chances were of having another child with CF after having three affected children, related to the same concept (i.e. the same risk in all subsequent pregnancies) and gave very different responses.

Although P2 knew the correct answer (1 in 4) to the question about recurrence risk, she understood this to be "just a statistic" and that in fact the chance would be higher: "*Statistics are just averages, you can't really say what your chances are. With one couple it could come out strongly, with another it won't*" (translated). To the question about the chances of having a fourth affected child, the father responded:
"I thought you only get that bad luck once." (Translated)

P5 believed that because they were already carriers, the recurrence risk was 100%, but they thought their chance of having a fourth affected child was zero.

P3 understood that CF was passed on through the generations and that every second child would be affected:

P3: "If your first child has CF then normally it would skip one and the third child would have CF again." She didn't know what the chances were of having a fourth affected child, but felt that:
"No, I think then it is safe to jump off a building!"

P8 thought the chance of having another affected child to be 2 out of 3, but when asked about the possibility of having a fourth affected child she answered:
"*I wouldn't even try, but I guess 50/50*". She did however understand that because she was remarried and not with the father of her CF child, unless he was a carrier, there was no chance of her having another affected child.

4.5.2 Discussion of the general level of understanding of genetics

The results in this study confirm the findings of the majority of studies that have been conducted to determine the level of genetic knowledge of parents of children with CF, which have reported a generally low or flawed understanding (James *et al.* 2006; Hames *et al.*1991; Denayer *et al.* 1990; Henley 1988; Nolan *et al.* 1986; McCrae *et al.* 1973, Leonard *et al.* 1972).

All of the participants knew that CF affects both males and females and the majority (eight out of ten) knew that both parents had to be carriers to give birth to an affected child. None of the participants used the term "autosomal recessive inheritance". Half of the participants correctly answered that the risk of having another child with CF was 1 in 4. Kaback *et al.* (1984) obtained correct answers about recurrence risk in 86% of participants, while Somer *et al.* (1988) reported that 74% of their sample in Finland knew the correct risk. In the study of Leonard *et al.* (1972), 64% of parents of children with CF remembered the recurrence risk. There were several misconceptions about inheritance risks. Other studies have also shown that parents often have difficulties with questions involving risk estimates (Nolan *et al.* 1986), applying risk concepts (Denayer *et al.* 1990) or mathematical probabilities (Hames *et al.* 1991). Consistent with findings in this study, previous research has shown that parents of children with CF often fail to grasp the critical fact that with autosomal recessive inheritance, all pregnancies carry a 1

in 4 risk, irrespective of how many affected children may have already been born (Denayer *et al.* 1990).

In the only study looking at this topic to be conducted in South Africa to date, Henley (1988) reported a mediocre knowledge of the genetics of CF among parents of CF children. Similar to this study, she found that 80% of parents knew both parents had to be carriers, but in her study as many as 75% of the parents knew that this implied a 1 in 4 recurrence risk, compared to only half of the participants in the current study. Henley (1988) found that only 40% of mothers and 30% of fathers correctly estimated their chances of having a third affected child. This is similar to 40% correct answers to having a fourth affected child by participants in this study.

The probability that specific relatives could be CF carriers was very poorly known. The majority of parents were aware that there was a chance their unaffected children could be carriers, but none of the participants knew the exact chance (2 in 3). Incorrect responses ranged from zero to 100% (Table 4.4, p51). Genetic risks are often underestimated or overestimated (Fransen *et al.* 2006). In a Belgian study (Denayer *et al.* 1990), only 2% of parents knew that their unaffected children had a 2 in 3 chance of being carriers.

Only two participants correctly estimated the risk of their siblings being carriers to be 1 in 2 (50%) and one of them, P1, admitted that she was guessing. Similarly, Denayer *et al.* (1990) found that although most parents (94%) of CF patients in their sample in Belgium knew they were obligate carriers, only 18% were aware of the 1 in 2 risk of their siblings being carriers. In a British study (Hames *et al.* 1991), only 5% of parents gave the correct answer. Parents often have difficulty comprehending recurrence risks for siblings and relatives (Seidenfeld and Antley 1981). Studies by James *et al.* (2006) and Fanos and Gatti (1999) reported that parents of CF children often overestimate the reproductive risks of siblings. This was true for only 1 participant in this study who thought the chance of her sibling having an affected child was 50/50 (P1). The other participants either didn't know or thought the chance was very slim.

Consistent with the findings of Henley (1988), several participants in this study could be described as having knowledge about genetics, but without understanding. Although the majority knew that the mutation in the CF gene had to be carried by both parents to give birth to an affected child, a significantly smaller group were able to translate this knowledge into actual recurrence risks. A poor grasp of concepts such as probability has previously been described as a main cause for parents' poor recall and misunderstanding of genetic information (Leonard *et al.* 1972).

Similar to previous studies, the level of genetic knowledge among participants was found to be related to socioeconomic status (De Pina-Neto and Petean 1999, Denayer *et al.* 1990; Hames *et al.* 1991; Henley 1988). Individuals from the higher socio-economic groups showed a better understanding.

Participants were asked when the genetics of CF was discussed with them by a health professional. The results are presented in Table 4.6.

Table 4.6: When the genetics of CF was discussed

Time	No of Participants
At the time of diagnosis	6 (60%)
At a later stage	4 (40%)

The majority of participants reported that the genetics of CF was discussed with them at the time of diagnosis. This has been well described as a time when parents are suffering from extreme distress learning of their child's condition and the ultimate limited life expectancy, leading to a lack of understanding of information received (Jedlicka-Köhler *et al.* 1996; McCrae *et al.* 1973). This is illustrated well in the response of P8 to the question of when the genetics of CF was discussed:

"*At the time of diagnosis, I believe, but at that stage the doctors were telling your child is dying and that was the last thing you heard, nothing after that.*" (Translated)

Care should to be taken as to how much information parents that have just received a diagnosis can absorb and cope with. During this initial phase information provided should be limited to and mainly designed to set the scene for later discussion. Information should be presented in a step-wise manner which includes regularly assessing their understanding and emotional responses (Weil 2000). Because of the complex nature of genetic information, poorly timed counselling can lead to lack of retention of information (Henley and Hill 1990).

Another possible explanation could be that a member of the health care team felt it unnecessary to cover certain aspects about CF, such as the genetics, because of being under the impression that it had been covered by others.

With the exception of P4 in whose family there was another individual affected with CF, none of the other participants had ever heard of the disease before the discovery of their affected child. Genetic terminology is generally less intelligible to lay people and this may lead to limited knowledge, a lack of understanding and misconceptions (Chapple *et al* 1997; Leonard *et al.* 1972), as illustrated by P4's comment: "*To really understand genetics, you have to be a doctor.*" (Translated)

Based on similar findings, Hames *et al.* (1991) suggested that information on the probability of children and siblings inheriting CF should be explained more clearly and on more than one occasion.

Participants were asked to indicate what their main sources of information about the genetic aspects of CF were. The first five items in Table 4.7 were presented to the participants as options and they were asked to identify additional or other sources, which are provided in the last column of Table 4. 7.

Table 4.7: Sources of information about genetics of CF

Source of genetic information	No of Participants
Doctors at the CF clinic	8
CF association	3
Internet	4
Media	2
Literature	2

The doctors at the CF clinic were the main source of genetic information for the majority of participants in this study. Four participants indicated that they had used the internet for research on the topic. Two of these mentioned being warned by the doctors to be careful of the information available on the internet. Vast amounts of genetic and medical information are now available through the internet directly to the public, including patients and their families (Christian *et al.* 2001). While this can be very beneficial it also has its drawbacks. Individuals may often be misled or confused by detailed information that would have been much more understandable and useful if presented in a structured way in a professional consultation. Some of the information encountered on the net may also be distressing to people (Harper 2004).

Findings in this study confirm reports by Henley (1988) and others showing that parents of CF children have "errors, gaps and misconceptions" in their knowledge of the genetics of CF that need to be addressed.

4.6 IMPACT ON REPRODUCTIVE CHOICES

Participants were asked whether the birth of their child with CF had any impact on further reproductive choices and if it had, how or why it had influenced their decisions. Their responses are presented in Table 4.8.

Table 4.8: Reproductive choices of participants

P	Impact	How or why?	Changed number of children
1	Yes	"I was afraid another child of mine would suffer as she has."	Yes "I wanted more but I was too afraid"
2	No	"We wouldn't have had more children"	No
3	Yes†	"Second child was not planned, we were actually trying to steer clear of having another one"	Yes Didn't plan to have second child, because 1st one had CF
4	Yes†	"It has affected us to the point that this is where we'll stop"	"Yes and no, maybe we would have only had 2, but now we'll definitely only have 2".
5	No	"I'm done with having babies. But I must say I think it had a lasting effect, because you always think you could have another child with CF"	No "We wanted 4 now we have 4.
6	Yes†	Already pregnant when child diagnosed. "If I'd known I wouldn't have had more kids".	Yes
7	Yes†	"I was 7 months pregnant when I found out my 1st child has CF. It was too late to do anything then."	Yes "I wanted 4".
8	Yes	"I've always said there is no way I'd take the chance again."	Yes "Wanted two"
9	No	No comment	Yes "Two is enough"
10	Yes	"We are afraid it would happen again, but it's not 100% because of the CF. I've also had difficult pregnancies"	Yes definitely "We wanted 3, but have now decided on only 2"

† Participants who indicated that their method of contraception had changed after the diagnosis of CF in their child.

4.6.1 Discussion of the impact on reproductive choices

Seven out of ten participants indicated that the birth of a child with CF had an impact on their further reproductive choices. The biggest reason for this was the fear of having another child with CF. Amongst those in whom it had no impact; two of the three participants (P2 and P5) indicated that the reason was because they had completed their

families. They had reached the ideal number of children and were not intending to have any more children.

A possible explanation for P9 feeling that the birth of her affected child did not have an impact on her deciding to have further pregnancies, was a lack of knowledge of the genetics of CF. As discussed in an earlier section (p52), P9 thought the problem lay with the deceased father and not with her, because they had done a sputum test on her and found nothing. She also had no idea what the recurrence risk was for future pregnancies. It is possible that she didn't see future pregnancies as a risk. Although the reality is that future pregnancies are at a lower risk because the carrier father is deceased, the point is that she was completely unaware of the fact that she was a carrier. Lack of understanding or misunderstanding of recurrence risk has been shown to be a reason for parents embarking on pregnancies after the diagnosis of a child with CF (De Braekeleer *et al.* 2004; Evers-Kiebooms *et al.* 1990). Knowledge of recurrence risk is important when evaluating the impact on family planning. Fifty percent of the participants in this study were aware of the 1 in 4 risk of having another affected child. Three participants overestimated their risks of having another affected child. Because genetic risks are often underestimated or overestimated, reproductive decisions may be based on incorrect assumptions (Fransen *et al.* 2006).

Despite answering 'no', P5 admitted that it still had an influence to some extent:
P5: "*I'm done with having babies. But I must say I think it had a lasting effect, because you always think you could have another child with CF*" (Translated)

Reproductive decisions in couples with a CF child, have been shown to predominantly be influenced by their experiences with CF (Henneman *et al.* 2001), as confirmed in this quote by P6:
"To take care of a sick child is not nice. It takes everything away from you, everything. You don't have any time for yourself, you don't have time for your house, and you don't have time for anything. You only concentrate on the child, you're too scared to go to sleep. Is he not breathing anymore, you only focus on the child, because you don't know if you'll have the child for tomorrow, what happens if the child dies? All of this goes through your mind and to go through all of that again? No thank you."(Translated)

Seven of the ten participants reported that the diagnosis of CF in their child changed the number of children they had originally wanted to have, resulting in them deciding to have fewer children than previously intended. P4 was undecided and said:
"*Yes and no. Maybe we would have only had two, but now we'll definitely only have two.*" Both participants (P2 and P5) who reported 'no change' felt that they had reached the ideal number of children.

With the exception of P8 and P9, all participants indicated that they would seek counselling if they were considering another pregnancy. P8 felt that because she was no longer with the father of her child with CF, she didn't need counselling. She did express her concerns by saying:
"I would maybe want my new husband to be tested to see if he is a carrier." (Translated)

Table 4.9: Birth rank of the child affected with CF

P	No of children	Birth rank of affected child
1	2	1st
2	2	Last
3	2	1st
4	2	1st
5	4	Last
6	3	2nd
7	2	1st
8	1	1st
9	2	1st
10	2	Last

Results in this study are supported by other studies which have shown that couples are more likely to have more children when the affected child was firstborn, than when there was already a healthy child before the birth of the child with CF (De Braekeleer *et al.* 2004; Evers-Kiebooms *et al.* 1990; Steele *et al.* 1986; Kaback *et al.* 1984). The desire

for a healthy child or to reach an ideal number of children has been described as possible explanations for these findings (De Brakeleer *et al.* 2000).

In six of the ten families the affected child was firstborn and with the exception of P8, who had separated from her husband, all of these couples went on to have a second child. P7 was however already seven months pregnant with her second child when her firstborn was diagnosed and P3 acknowledged that her second child was not planned:
P3: *They said it would be the first and the third child and we were scared that with our luck, it would be the second one. So that is why were trying to steer clear of having another one. We didn't plan to have the second one, because the first one had CF*".
She explained that she fell pregnant because she was spending so much time in hospital with her affected child and she forgot to take her contraceptive pill:
"because I was sitting at the hospital all the time, it just slipped my mind."

P1 and P4 wanted to have another child and were willing to take the risk, as described by P4:
"*We definitely wanted to have a 2nd child, so we were willing to take the chance...but I don't' think we would take the chance to have a 3rd child."*

With the exception of P6, who was already pregnant with her third child when the second born was diagnosed, in all the other families where the affected child was second or last born, there were no further pregnancies and the couples weren't planning any further pregnancies. P6 also acknowledged that she wouldn't have had another child if she had known about the diagnosis before her pregnancy.

Parents have been shown to use more reliable contraceptive methods following the birth of their CF child (Boué *et al.* 1991; De Braekeleer *et al.* 2000). In this study, four participants reported that their contraceptive methods had changed to more reliable methods following the birth of their child with CF. Only one participant opted for sterilization. In a Belgian study, 23% of participants opted for this method (Evers-Kiebooms *et al.* 1990), while 51% of the couples in New England chose sterilization

(Wertz *et al.* 1992), and as many as 84, 4% of parents in a Canadian study (De Braekeleer *et al.* 2000).

Attitudes to prenatal diagnosis and termination of pregnancy

Participants were asked whether they would/ have considered prenatal diagnosis and/or termination of an affected pregnancy, because this has become an important reproductive option available to parents with a child with CF (Henneman *et al.* 2001). Their responses are presented in Table 4.10.

Table 4.10: Attitudes to prenatal diagnosis and termination of pregnancy

P	Would/have consider/ed prenatal diagnosis	Would consider TOP?
1	Yes, been through it	"I don't know what I would have done"
2	No	No
3	No	No For religious reasons
4	Yes, been through it	No
5	Yes	No "We are Christian and it's a sin".
6	Yes	Yes
7	No…there are no guarantees	"I have no idea"
8	No, I don't believe in it	No May not end a life
9	Yes	No Would raise the child
10	No	No, never. "It's against our beliefs"

Half of the participants indicated their intention of using or that they had used prenatal diagnosis in further pregnancies. The other half would not consider it (Table 4.10). P6 did enquire about the possibility of prenatal diagnosis in her second pregnancy, but was told it couldn't be done, because in her affected child's case, they couldn't detect CF by means of a "blood test" (genetic test), only a sweat test.

P6: "*They told me there was no point. We had to wait until 6 weeks for a sweat test. They said I must make up my mind whether I wanted her or didn't want her and we couldn't, where our religion was concerned, we didn't just want to have an abortion, we wouldn't have been able to live with ourselves if we did it, even if she had CF again, because you can't just kill human life in the name of convenience.*"

The affected child's medical records at RCCH showed that genetic testing did not detect the common mutations; therefore the disease-causing mutations in this family were unknown.

The use or intention to make use of prenatal diagnosis is influenced by several factors including: religion, family income, the presence or absence of healthy children and the willingness to terminate an affected foetus (De Braekeleer *et al.* 2000; Borgo *et al.* 1992; Wertz *et al.* 1992; Jedlicka-Köhler *et al.* 1994)

Two participants (P1 and P4) had been through prenatal diagnosis in their second pregnancies and described it as being very traumatic. Both of these couples wanted a second child. P4 originally didn't want prenatal diagnosis, but halfway through her pregnancy she changed her mind:

"*I felt like I didn't want to know and then all of a sudden I just felt like I couldn't not know...so it was like things just totally changed.*"

P4 was then told by one of the other mothers at the CF clinic that she could go for a scan with which they could pick up certain things that indicate the baby is at a high risk and you could then have further tests.

P4: "*So we went for the scan and they were pretty sure it was CF, because they look for something called blocked bowel and it was apparently very evident....they checked and they called in someone else, there were two women and they pretty much both started crying. I then had an amnio and he wasn't even a carrier*"

Although this couple felt they wouldn't have terminated even if it was an affected foetus, they just had to know before the child was born.

P1 described why she wanted prenatal diagnosis and how anxious she was about the outcome:

P1: "*I wanted another child, so that (affected child) could have a playmate. That's why I fell pregnant again. I was very worried about the answer when I had the test done....I was afraid that another child of mine would suffer as she had.*" Her second child was found to be a carrier and she was extremely relieved. She insisted she wouldn't go through it again as it was too traumatic. When asked whether she would have considered termination of pregnancy for an affected foetus, she replied:

"*I don't know what I would have done*".

Both these participants described the waiting time to get the results of the prenatal testing as very stressful. Evers-Kiebooms *et al.* (1990) described a similar finding in their sample. The author explained that the waiting time was particularly stressful for these parents, because parents who plan a pregnancy with prenatal diagnosis mostly have a strong desire to have a child, but not to have an affected child.

Only one participant (P6) gave a clear "yes" response to whether she would consider terminating an affected pregnancy following prenatal diagnosis. The majority of participants (7 out of 10) said "no" for various reasons. Correlations have been found between abortion attitudes and religion, education and interpretation of risk (De Braekeleer *et al.* 2004; Al-Jader *et al.* 1990).

Three participants mentioned religious or moral beliefs. Two participants indicated that it was not in their power to decide about a life:

P2: "*To me its murder.*" (Translated)

P8: "*I may not end a life.*" (Translated)

P4 felt that they may have considered termination for another condition, such as Down syndrome, but not for CF:

P4: "I think because we have a daughter who is really very well and lives a very normal life, I wouldn't have considered it. Obviously, you never know, the other child could be

way worse. But you'd never know and you can't terminate on that basis." Termination of pregnancy is not necessarily regarded as an option by women, even when the foetus is known to be affected. This may in part be due to the fact that CF is not associated with mental retardation or dysmorphic features, as well as because of the steadily increasing life expectancy of individuals with CF, due to improved therapies and medications (Wei *et al.* 2007).

P9 felt she would raise the child and the remaining two participants (P1 and P7) were undecided:

P7: "I *have no idea...it would depend on my state of mind at the time*"

The literature shows that there is a wide spectrum in the available studies in attitudes toward abortion among parents of children with CF. Although the majority of studies have shown that parents are in favour of termination (De Braekeleer *et al.* 2004; Al-Jader *et al.* 1990; Evers-Kiebooms *et al.* 1990), some studies have found low levels of interest (Wertz *et al.* 1992). In contrast to the majority of available research, this sample predominantly indicated that they were against this option.

4.6.2 Summary of results on the impact on reproductive choices

The findings in this study are supported by studies which have shown that the birth of a CF child has a major impact on subsequent family planning for parents (De Braekeleer *et al*, 2004; Henneman *et al.* 2001; De Braekeleer *et al.* 2000; Evers-Kiebooms *et al.* 1990).

The biggest way in which the diagnosis of the CF child impacted on participants' reproductive choices in this study was by reducing the number of children they originally planned to have, as indicated by seven out of ten participants. The most important reason given for the impact on reproductive decisions was the fear of the recurrence risk, the fear that another child would have CF. This is in alignment with other studies (De Braekeleer *et al.* 2000; Evers-Kiebooms *et al.* 1990). Henneman *et al.* (2001) found this to be the biggest reason in 70% of cases in their study of a Dutch

population. Similar to this study, the major reason for reporting no influence on reproductive planning was that the initially-planned number of children had been reached at the time of diagnosis.

In total, there were five further pregnancies after the diagnosis of a child with CF. However, one of these pregnancies (P3) was unplanned and two participants were already pregnant with their next child when the diagnosis was made (P6, P7). Of the remaining three pregnancies, two participants were willing to take the risk, because they wanted another child. P9 was unaware that there was a risk in future pregnancies. None of the participants were planning any further pregnancies. Similar to previous research, the birth rank of the affected child was found to have an influence on reproductive planning. Where the affected child was first-born, couples were more likely to embark on further pregnancies.

Fifty percent of the participants indicated their intention to use or that they have used prenatal diagnosis in further pregnancies, but there was far less consensus about termination of pregnancy should prenatal diagnosis reveal an affected foetus, with only one participant convinced that she would consider the option. These results are similar to other studies in which prenatal diagnosis has been shown to be an important reproductive option for parents of children with CF and several parents have indicated their intention to use it/ or have used it. However, termination of an affected pregnancy has been shown to be far less acceptable (Henneman *et al.* 2001; Evers-Kiebooms *et al.* 1990).

4.7 RISK COMMUNICATION

As an introduction to the section on communication about the genetics of CF, participants were asked various questions relating to information sharing about CF with their children. Responses to selected questions are presented in Table 4.11.

Table 4.11: Results on communication about CF of parents with children

P	Age of affected child (yrs)	What has ill child been told?	Questions he/she has asked?	Affected child(ren) understanding of CF	No of Sibs	Age(s)	Unaffected child(ren) understanding
1	16	"It can shorten her life if she doesn't look after herself"	"Not much…but she does get angry"	"She doesn't want to talk about it really. She doesn't like me to tell people about it…"	1	13yrs	She understands it all. "She knows she's a carrier"
2	13	"He knows he's got it mildly, we made him aware of how potentially dangerous it can be"	"Is it contagious? Will I still be able to fish and live a normal life? Am I also dying?"	"It helped that when he was diagnosed, he could already understand a bit about how dangerous this thing is"	1	15yrs	"Talked openly about it. Explained the same way we did to (affected child)"
3	10	"I told him that I'm not going to hide anything from him…I explained that if he is not taking care of his lungs, not exercising, not taking meds…he could get an infection and die"	"He just wants to know: Why him?".	He understands his condition.	1	8 yrs	"She knows. She listens every time I speak to him and she also tells him: you know you're going to die, you better take your tablets."
4	4	"Just to explain the Creon she has when she eats that her body doesn't make the soldiers she needs to digest her food, so these are her soldiers"	"Would I stop taking Creon when I'm big?"	"She knows she's got CF. She knows she's different, but she obviously doesn't understand really"	1	7 mnths	N/A
5	7	"Not much. I tell him he must take it so that his food can be digested so that he can put on weight and grow"	Only about pills	"He'll tell you he has a bad chest or tummy"	3	20 yrs 17 yrs 13 yrs	"We explained to them how the pills work, but we haven't really sat down and discussed the illness with them"
6	15	"Every month, he sees the doctor alone. He knows, the doctor told him. He tells the doctor where it hurts and how he's taking his pills"	"He always asks difficult questions that you can't answer. Why did it have to happen to me?"	"He understands what is expected of him"	2	22 yrs 15 yrs	"They understand everything"
7	13	"Whatever she needs to know according to her age…relevant to her age and appropriateness. Nothing unnecessary."	"Obviously she's said: Why do I have to have this?"	"She understands it's genetic, that the mucous is thick in her body, that she has to exercise and eat a lot"	2	11yrs	"Everything that goes on in the household goes on in front of her, so she knows most things as well. She knows about

							treatment, enzymes, that she needs to be away from people with colds"
8	10	"The doctors at RCCH explained it to him"	None	"At this stage, I don't think he realises the magnitude of it"	0	n/a	n/a
9	16	"God has tested you and its not something to ask too many questions about"	"He once asked how long he has to live? He doesn't discuss these things with me"	"He told the other women he's satisfied with this illness he has. He doesn't blame anyone"	1	14 yrs	"He knows he can't hit his brother on the back because he gets tired"
10	3	"Only that if he doesn't take his pills, his food will fall right through. Nothing else, he's too young"	None	"He knows about the pill story, we haven't told him why he has to drink it"	1	7 yrs	"He knows its CF, but he doesn't know what that means. He knows his brother is not well and when you're sick, you have to stay away from him. He's not ready yet to know all the rest"

4.7.1 Discussion of communication with children about cystic fibrosis

Research has described the sharing of information about genetic conditions from parents to children as an unfolding process which continues throughout childhood. Parents base their decisions about what and when to share on the child's developmental readiness and interest (Gallo *et al.* 2005).

Similar to other studies, parents in this study based the information they shared with their children about CF on their child's age and maturity, with the main reason for not sharing information being that they felt their children were too young to understand. Information given was based on age-appropriateness:

P5:"*We haven't really explained things to him, but we will from now on, he's turning eight now, so we can start explaining why he has to go to clinic, why he has to take his pills, what it is.*" (Translated)

P7:"*Whatever she needs to know according to her age, relevant to her age and appropriateness. Nothing unnecessary.*"

With younger children, explanations of the condition were kept simple and mainly focused on the daily management tasks. A good example is the way in which P4 described the pancreatic enzyme supplement (Creon) to her daughter: "*Just to say that her body does not make the soldiers that she needs to digest her food, so these are her soldiers.*"
This is in line with guidelines that have been published for the disclosure of a chronic illness to a child (Thorne *et al.* 2000; AAP 1999; Bibace and Walsh 1980).

Some participants waited for their children to ask questions and used it to explain certain aspects of the illness. Two participants relied on the doctors at the CF clinic to give their child information about the disease (P6 and P8). The majority of parents of older children expressed that they tried to be open and honest with their children about the condition. They had addressed the shortened life expectancy, by explaining that their

lives could be shortened if they didn't take care of themselves. P3 for example, told her affected son from the beginning that she was going to be very honest with him and not hide anything from him. She felt he understood in his own way, because when she got angry with him he would say:

"*Yes mommy I know what I must do: I must take my tablets, I must use my pump, I must do my lung exercises and I must do my physio, otherwise I am going to die. He tells me the whole little speech, so he listens, but it doesn't sink in, in a way.*" (Translated)

P8 had not discussed the shortened life expectancy aspect with her 10 year old son, because she didn't want to burden him, she felt it keeps improving and believes there is still hope: "*There are people with CF that are in their fifties and sixties and are still alive. On the other hand there are people of 20 that die in motor vehicle accidents. I don't want to confuse him or burden him with it. Perhaps by the time he's 20 he can go for some or other therapy*" (Translated).

Parents of children with CF or other life-shortening conditions have been shown to be more likely to selectively share information related to issues such as death and dying (Gallo *et al.* 2005).

P5 gave a basic description to their seven year old affected child of the reason for having to take pills: "*so that his food can be digested so that he can put on weight and grow*". However, this is also the extent of the information on CF that they have given to their three older unaffected children.

P5: "*We explained to them how the pills work, but we haven't really sat down and discussed the illness with them.*" (Translated)

Furthermore, the genetics of the condition had not been discussed with them and the parents were under the impression that the risk of their unaffected children being carriers was "nil" (p51). Thus, leaving the older children unaware of their potential carrier risk. The couple's seventeen year old daughter had already had her first baby. Studies have shown that parents rarely sit down and talk with their unaffected children about CF, as indicated in P2's response:

P2: "*It's been a while since we've explained it to her, maybe not a bad idea to talk to her again.*" (Translated). Importantly however, siblings of affected children are dependant on parental transmission of information about CF (Fanos and Johnson 1995) and they may have fears relating to their own vulnerability of being affected or of being carriers of the condition. There may be confusion arising from a lack of understanding (Weil 2000; Patterson *et al.* 1990). The majority of parents reported giving the same information to their affected child and his/her unaffected siblings.

Although P6 reported that her two unaffected children "understand everything" about CF, it is worrying that she was under the impression that neither of them were carriers, based on a sweat test that was done after the affected sibling was diagnosed. She also mainly relied on the doctors at the clinic to inform her child about his illness, so it is unclear how much they really understand about CF. Similarly, P9 has not really discussed the condition (or genetics thereof) with either her affected or unaffected child. Lack of knowledge was the major limiting factor in these families. For risk information to be distributed, knowledge and understanding of genetic risk is essential. Misunderstanding of genetic risk has been identified as a major barrier to risk communication in families (McConkie-Rosell and DeVellis 2000).

On the other hand, P1, who had a very open approach to information sharing with her children, had already disclosed to her thirteen year old unaffected daughter that she was a carrier. Following the diagnosis of her affected child, P1 had prenatal diagnosis in her second pregnancy. The carrier status of the foetus was revealed to the parents prenatally. Over the last few years, concerns about carrier testing in minors have been raised and debated. Concerns relate mainly to the psychological or social harm of genetic information, the lack of autonomy of minors, as well as the potential inability of children to fully comprehend the risks and benefits of genetic information (McConkie-Rosell & Spiridigliozzi 2004).

Several participants reported that their affected child had asked the question: 'Why me?' All of them reported having difficulty answering this question. Below are some of the responses:

P3: "*I have to explain to him, nobody could have planned this and it's not as if we can plan anything in our lives. Our lives are planned for us. I explain more of the spiritual side than what I can explain 'why him'.*"

P6: "*I always wait a while before I answer and then I say: the reason is that you were one of the unfortunate ones. But God has blessed you so that I can look after you because you are my special child. And then he'd ask: but what about the other two? And I'd say: yes, they are also special, but not as special as you.*"

P7: "*You've got to have a lot of faith in God and it is always: Allah knows best, God knows best. And not to sit and dwell on these things, but to deal with it day to day. We live a very day to day life. Positivity is the most important thing.*"

Parents should be advised on how to transfer information about CF to their children from very early on, in a way that is neither threatening to child nor to the parent. In this way independence of both the family and the patient is promoted. To some extent this could further prevent displacing of patient dependence from the family to the clinic (Nolan *et al.* 1986).

4.7.2 Discussion of risk communication in families

Despite the fact that the birth of a child with CF implies an increased risk for other relatives of being carriers, the hereditary aspect of the disease is not often discussed with these relatives. Although there is not a vast amount of literature available on the topic, studies that have been done have shown that there is often poor communication about genetic risk between relatives in CF families (Forrest *et al.* 2003; Denayer *et al.* 1992a; Turk 1964).

Participants were asked to indicate with which family members they had discussed the genetics of CF and therefore their potential carrier risk. The results are summarized in Table 4.12

Table 4.12: Summary of informed family members as indicated by participants

Discussed Genetics of CF with:	Participants	Total no of participants
Close and extended family	1,3,4,7	4
Close family (siblings, parents, children)	2	1
Siblings	10	1
1 parent	8	1
Nobody	5,6,9	3

Four participants (P1, P3, P4, P7) reported that they had notified all their family members- close and extended, of their potential risk of being carriers of CF. Ormond *et al.* (2003) found that parents of an affected child told 100% of their living parents, siblings and half-siblings, compared to 50% of participants in this study. More selective communication to extended family members generally occur because of a lack of closeness, familiarity and uncertainty about sharing genetic risk information (Wilson *et al.* 2004).

P4 agreed that it had been easier for them, because there was already another affected individual in the family (maternal cousin). At least on one side of the family, people were already aware of it. Research has shown that where there is more than one affected family member, it is easier to recognise a pattern of inheritance and therefore a higher chance that carrier risk information will be disseminated (Henneman *et al.* 2002).

They explained that it had been more difficult explaining the genetics of CF to the father's side of the family, because: "*nobody knew anything about it, it was totally new*". But he felt that everyone went out and did their homework on the condition, although he still felt that: "*I'm sure that parts of my family still don't get what we're talking about.*"

P7 went through a lot of trouble to contact the whole family to inform them of the condition and explain the potential carrier risk:

"*I told everybody. I'm a big talker, as you can see. I just made everybody know about it. I insisted that when they got married or chose their partners that they got tested.*"

She did feel however that they didn't listen to her or really take note of what she was saying:

"*They don't want to know about it. It's too close to home, like, they don't want to know. And they could be carriers and their children could be carriers, but if they're willing to take that risk, at least I've done my job and informed them.*"

In a study on the disclosure of genetic risk information in HBOC families, D'Agincourt-Canning (2001) found that several individuals who had made the effort to inform family members were troubled that not all relatives took this information seriously.

It was interesting to note that two of the four participants who had reported informing all of their family members and gave explanations that were accurate, had been the only two participants in this study who had been for a genetic counselling session with a genetic counsellor following the diagnosis of their affected child. P1 was seen by a genetic counsellor at the Johannesburg Hospital in Johannesburg, Gauteng and P7 was residing in London at the time of diagnosis of her affected child. She was seen by a genetic counsellor at Great Ormond Street Hospital for Children in London. They were also two of the participants with the most accurate knowledge on all aspects of the genetics of CF.

Studies have shown that only partial information may be passed on by parents or it may be done in such a way that family members are not made fully aware their carrier risk (Wilson *et al.* 2004; Denayer *et al.* 1992a). Participants were asked how they had explained the information about the genetics of CF and their possible risk status to family members. Some of their explanations are good illustrations of how partial or perhaps misleading information was disseminated, leaving the researcher to wonder what family members actually understood.

P2: *I didn't explain in detail, for me it's just about the fact that it was inherited from both sides. There's enough in the media about it. They can just look at those programs."* (Translated).

P2 only partly discussed the genetics of the condition with siblings, telling them that if CF was to "come out somewhere", it would be in their children. She also indicated:

"*It's never been an issue for me to really go into the genetics. It's inherited and we're coping with it.*" (Translated)

Because all of their siblings had completed their families and none of the children were affected, P2 felt that they wouldn't be interested in further information, not considering the possibility that these children were also at risk of being carriers.

P3:"*I explained to them that it was in our family, it was just misdiagnosed. Everyone then got involved in trying to track down the guilty party…*"

P5 felt that they had planted the seed that: "*just because it happened to us, doesn't mean it couldn't happen to you.*" (Translated)

P10 had only discussed the potential carrier risk with their siblings. The reason they didn't want to tell their parents was because they felt that they had to deal with this thing by themselves without burdening other people: The desire to protect others has been shown to be a barrier to communication about risk in families (Holt 2006; Wilson *et al.* 2004). They also indicated that they didn't think any of their siblings took the information seriously:

"*I don't think they've really thought about it or realised the full implications of the story yet.*" (Translated)

Although P8 had the knowledge, she selected only to discuss the genetics of the condition with her father and step-mother. She didn't have any contact with the rest of the family and felt that they weren't interested and therefore chose not to tell anyone else: "*If they'd asked, I would have told them* (translated)". She also had no contact with her mother or anyone on that side of the family, no-one was even made aware of the

affected child's condition. She expected her step-mother to inform her two half-sisters, but was unaware whether this had in fact happened. P8 also said she didn't think her ex-husband would have informed any of his family members either and she wanted nothing to do with his family. As families become more diverse through separation, divorce and remarriage, it is likely that the disclosure process will become more complex (Wilson *et al.* 2004).

P6 didn't think it was necessary to talk to any family members, because as discussed earlier (p52), she wasn't aware that any other family members could be at risk of being carriers or of having a child with CF until another affected child was born on her ex-husband's side of the family (sister's son). CF is often viewed as located within the nuclear family, not the extended family (Fanos and Johnson 1995).

Furthermore, her ex-husband blamed her and her side of the family for the condition in their child. Parental guilt and blame around having a child with CF can prevent any discussion about genetics in a family. If one parent uses blaming 'the other side of the family' as a coping mechanism to deal with guilt, that parent would choose to avoid evidence that may point to their own genetic contribution to the condition (Fanos and Johnson 1995). Therefore, he didn't provide any information to his side of the family and his sister had a child with CF. In this small sample, the birth of at least one affected child could have possibly been prevented if risk communication had taken place effectively in this family. Even if the parents of the recently diagnosed affected child had chosen not to have prenatal diagnosis and/or termination of the affected foetus, they would have had the opportunity to make an autonomous decision. Not conveying risk information denies relatives the opportunity to make informed reproductive choices (Clarke *et al.* 2005).

The genetics of the condition had been discussed with both affected and unaffected children in four families (P1, P2, P3, P7). Both P2 and P7 had explained to their unaffected children that there was a risk that they could be carriers, but that they could only be tested if they so wished when they were older. Testing of children at risk of

being carriers is generally delayed until at least the age of 18, when they are old enough to make their own informed judgement (Roberts *et al.* 2003). As mentioned, P1 had disclosed her carrier status to her unaffected daughter after careful explanation of the genetics of the condition when she was about eight years old. Although P3 had discussed the genetics with her unaffected daughter, she was under the impression (like P6), that her daughter was not a carrier based on a sweat test that was done after she was born. In a study in Chicago, USA, Ormond *et al.* (2003) found that one third of the children at risk of being carriers had been informed of their potential carrier status by their parents. For those who were not informed, the major reason provided was that parents thought their children were too young to understand, results reported by other studies (Gallo *et al.* 2005; Canam 1986). In this study, this was the main reason given by two participants (P4 and P10). Other reasons for not informing children in the present study were a lack of knowledge of the genetics of CF in parents and misconceptions about children not being at risk.

Awareness of and discussion about carrier testing:

Three participants had never heard of carrier testing (P5, P9, P10). Five out of ten participants could more or less correctly explain what it was and how it worked. The other two participants had misconceptions:

P3: "*I would think it would be another sweat test. I know Prof....said they normally do these tests at Tygerberg Hospital. That's all I know, I don't know exactly what happens.*"

P6 explained that they had taken blood from her last year to do carrier testing but that she never got the results of that test.
"*I don't understand how it works, but I must ask them (doctors at CF clinic) what happened about that, because I might be a carrier...I must know.* " (Translated)
This participant had difficulty understanding the carrier concept.

Only three participants (P1, P4, P7) indicated that they had mentioned the possibility of carrier testing to some family members. P4 told only her sister: "*that she could have a*

test done to determine whether she is a carrier or not". P7 advised all her family members to do it and P1 discussed it with her siblings. It was only in these three families that participants reported being aware of any family members who had acted on the knowledge that they could be carriers. Some of the siblings in these families had already been for carrier testing and others indicated their intent to go for testing when they started planning their families. In the Belgian study (Denayer *et al.* 1990) reported that 58% of his sample of parents had informed their siblings about the possibility of carrier testing. One of the main barriers to carrier testing is lack of awareness of the availability thereof and what it can and cannot do (Ormond *et al.* 2003). Studies have shown that barriers to the dissemination of information about carrier testing from parents of a child with CF to their relatives include: lack of information and the perception that relatives would not be interested in carrier screening (Sorenson *et al.* 1996; Fanos and Johnson 1995).

Participants were asked to give their reasons for either choosing to inform family members about their possible genetic risk or choosing not to inform them. The responses of the participants are summarized in Table 4.13.

Table 4.13: Reasons for and against informing family members

Reasons for telling	Reasons for not telling
P1: "They could have a child with CF…to prevent them from having a child with CF"	P5: "We didn't really understand or know ourselves (translated)"
P2: It made it easier for them to understand, because CF was as much of a mystery to them as it was to us in the beginning (translated)"	P6: "Didn't know it was in the family…and its too much explaining to do (translated)"
P3: "They need to know where the disease comes from, because in the end none of my brothers or sisters or their children were tested…we all know there might be a risk there"	P8: Lack of interest Didn't have contact with the rest of the family. Told step-mom and expected her to inform her own children.
P4: "It's the beginning point of explaining the disease as a whole, you need to explain where things come from"	P9: "They misuse alcohol (translated)" Didn't have contact with her family
P7: "To make them aware…so that their child doesn't suffer"	P10: "Felt like we didn't want to burden other people (like parents) with it. Felt we had to carry this thing by ourselves (translated)"

The main reasons for sharing information about the genetics of CF was to help family members understand where the disease comes from, and to make them aware that there was a risk that it could happen to one of them too, that another child in the family could be born with CF.

The majority of participants who had discussed risk information with family members reported that they did so almost immediately after they had received the diagnosis. Ormond *et al.* (2003) found that disclosure of carrier status information usually takes place within six months of becoming aware thereof. It most often takes place in the context of explaining the affected child's disease as a whole, as was the case for several participants in this study. In contrast, the major reasons for disclosure given by Ormond *et al.* (2003) were a close social relationship and the need for support in a time of crisis. Neither of these reasons were mentioned by participants.

The main reasons for not sharing risk information was a lack of contact with certain family members, as well as a lack of knowledge about risk genetic information in participants. For risk information to be distributed in a family, knowledge and understanding of genetic risk is essential (Forrest *et al.* 2003). There are several possible reasons why individuals may not be in contact with their family, including: geographical distance, lack of social closeness, internal family rifts, divorce and separation (Featherstone *et al.* 2006; Ormond *et al.* 2003). No individuals reported negative feelings about their CF carrier status (e.g. guilt or shame) or perceptions of stigma as reasons for nondisclosure, factors that have influenced disclosure in other studies (Wilson *et al.* 2004; Holt 2006; Fanos and Johnson 1995).

Across a range of disorders with different modes of inheritance it has been suggested that genetic risk information is passed on to family members to which individuals feel emotionally close, whilst the information is shared much more selectively with more distant relatives (Claes *et al.* 2003; Peterson *et al.* 2003; Suslak *et al.* 1985). Although a close social relationship was not explicitly mentioned as a reason for disclosure, as found by other studies (Ormond *et al.* 2003; Fanos and Johnson), most participants did

not feel an obligation to inform family members they did not know or did not have contact with.

Participants were asked which parent took the biggest responsibility for discussing risk information with family members.

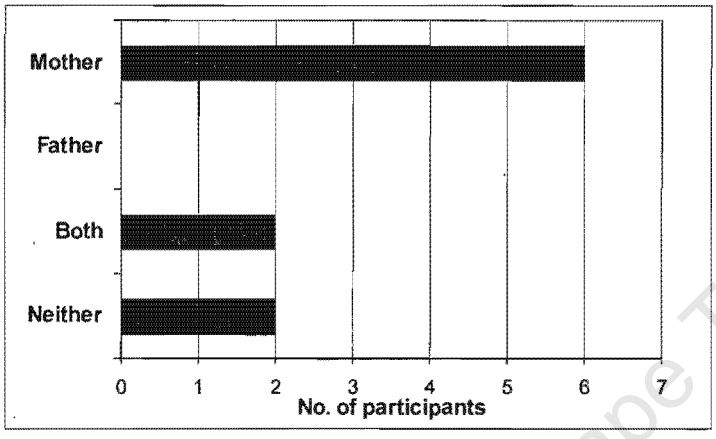

Figure 4.2: Results of responsibility for risk communication (n=10)

In the majority of couples (60%), the mother was responsible for the dissemination of risk information in the family and beyond. These findings are consistent with other research which suggests that disclosure of genetic information is a gendered activity, with the benefits and burdens of the task falling mainly on women (Hallowell *et al.* 2005; D'Agincourt-Canning 2001; Richards 1996).

Participants were asked whether they thought it was their responsibility as parents of the affected child or that of the health professional (such as doctor or genetic counsellor) to inform family members of their possible genetic risk. With the exception of P1 and P9, all participants (80%), felt it was their responsibility as parents, because doctors didn't have the time to go from family member to family member talking to them about it.

P1 felt that: "*They (doctors) are more convincing. They (the family) won't listen to you as they will to a professional.*" P7 agreed that although it was her job, "*they (the family) might take it more seriously if it came from a health professional.*"

This corresponds well with findings in other studies which have shown that parents generally regard dissemination of risk information as their responsibility (Green *et al.* 1997; Hallowell *et al.* 2005), although they may feel burdened by being the bearers of bad news (Clarke *et al.* 2005). The participants in this study did however feel that the doctors or genetic counsellors had to make sure they had sufficient information to feed back to the family.

Although eight out of ten participants felt they had sufficient information about the genetics of CF to discuss it with family members, seven out of ten indicated that they would like more information about this topic. There was a general opinion that they had the basics and this was sufficient, for example:

P3: "*I'm just trying to keep it basic and not over explain in too much detail*"

Written material, such as pamphlets and booklets were found to be useful in explaining information to family members by some participants. Studies have emphasized the important role written material on the implications of the diagnosis of a child with CF in the family can play in assisting parents to inform family members (Claes *et al.* 2003; D'Agincourt-Canning 2001).

Participants were asked how regularly they discussed the genetics of CF with relatives. Their responses are presented in Figure 4.3.

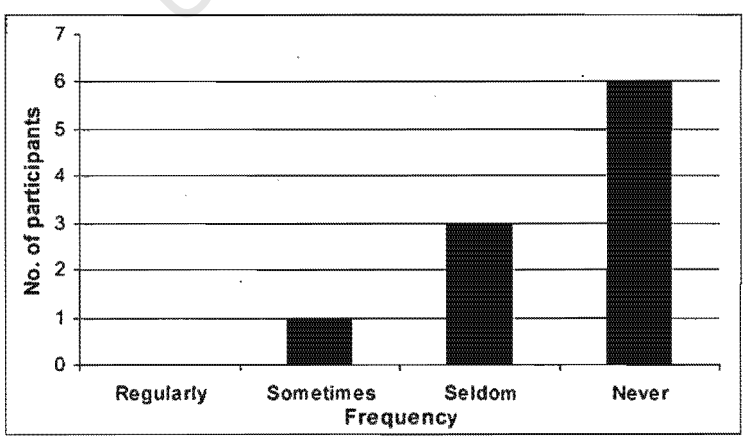

Figure 4.3: Frequency at which genetics is discussed with relatives (n=10)

In a sample of 105 CF-parents in Belgium, Denayer *et al.* (1990) found although 80% of the parents of a child with CF informed their bothers and sisters about the genetic aspect of CF, only one fifth discussed the genetic aspect of CF regularly with their siblings and 36% do now and then. The majority of participants in the current study (60%) reported never discussing the genetics of CF with relatives. Some of these participants felt that they had discussed this issue with family members after the diagnosis and therefore did not see the need to talk about it again:

P1: "*I feel that they all know about it, so there's no use talking about it anymore.*"

Denayer *et al.* (1992a) found that the major source of information for aunts and uncles of a child with CF was the parents of the affected child. However, their knowledge on the genetics of CF was found to be poor, with only 7% aware of their 1 in 2 carrier risk. Nevertheless, the risk of having an affected child played a major part in decision making about further pregnancies for 39% of individuals (Denayer *et al.* 1992b), indicating the importance of accurate transfer of information to these relatives.

4.7.3 Summary of discussion on risk communication

The extent of disclosure of genetic risk information varied greatly among the participants in this study. While some participants kept information sharing to close relatives (parents, siblings, children), a few participants went to great lengths to inform the extended family (cousins, aunts, uncles). In one family (P4) this process was facilitated by the fact that there was another affected individual on one side of the family and therefore several family members had already been aware of the condition. Three participants reported never having discussed the genetic aspect of CF with any family members. With the exception of these three participants, the remainder told at least one biological relative.

Siblings of parents with a CF child have a 1 in 2 chance of being carriers of the CF gene themselves. In this study risk information was reported to have been discussed with siblings by six out of ten participants. The remaining four had never mentioned it to their siblings. It is important to note that of these participants; only two had correctly

estimated their siblings' carrier risk (p55). In some of these cases information was disseminated only partly or in such a way that relatives may not have been made fully aware of their potential carrier risk. Only three participants had mentioned carrier testing to some family members.

The majority of participants had not yet discussed the genetic aspect of the condition with their children (affected or unaffected). The main reasons being that parents felt they were too young to understand or a lack of understanding of the genetics of the condition. P1 on the other hand, had already informed her 13-year old daughter about her carrier status. Genetic counsellors and doctors working with CF families need to be aware that the information sharing process continues throughout a child's lifetime. This allows health professionals the opportunity to help parents develop strategies on how to talk to their children and to discuss with parents their views and attitudes related to sharing information about CF, including the genetic aspects. Parents need to be aware of the important role they play in the developmental process of sharing information with their children (Gallo *et al.* 2005).

Participants who had disseminated risk information to family members did so mostly in the context of explaining the affected child's disease as a whole, as well as to make family members aware that they were at risk of having a child with CF. The main barriers to sharing risk information identified in this study was a lack of knowledge about risk information in participants, as well as a lack of contact with certain family members.

The task of disseminating risk information to family members fell mostly upon the women in this study. The doctors at the CF clinic were identified as the main source of information about the genetics of CF for the majority of participants. Sixty percent of parents reported never discussing the hereditary aspect of their child's condition with any family members.

Preparing parents of children with CF to inform their relatives about the genetics of CF and therefore their potential carrier risk should be an integrated component of genetic counselling for CF. It is an aspect that needs special attention, particularly because of the availability of carrier detection in most families with a living child with CF. Counselling should include discussion on those relatives who might be told, how to go about telling them and their possible reactions to the information. Ways of coping with anticipated difficulties could also be suggested (Green *et al.* 1997). Indeed, in an earlier study of women attending a HBOC clinic, one of the main dissatisfactions reported by several women was the lack of support or help in informing relatives who might be at risk (Richards 1999).

Risk information is often poorly retained and misunderstood and written material in a clear, precise, non-technical language can be a helpful aid. Professionals need to develop tools and strategies to facilitate the communication process in these families, it is very important that assisting in this process is given sufficient attention during counselling sessions (Claes *et al.* 2003).

Chapter 5
CONCLUSION

5.1 CONCLUSION

The aims of the present study were to determine the level of genetic knowledge of parents with a child with CF; to determine the impact of the birth of a child with CF upon subsequent reproductive choices and to investigate family communication about genetic risk

Participants in this study generally had a flawed understanding of the genetics of CF. This study has been able to highlight gaps and misconceptions in parental knowledge of the genetics of CF that need to be addressed. There were several misconceptions about the recurrence risk in future pregnancies and the probability that relatives (siblings of the parents and their unaffected children) could be CF carriers was poorly understood. A strong correlation was found between level of genetic knowledge and the socioeconomic status of participants. Lower socio-economic status resulted in a lower level of knowledge about the genetics of CF. A good understanding of the genetics of CF in parents of affected children is important because it impacts directly on further reproductive choices and is also required for effective risk communication in families.

Parents of a child with CF need to understand and to weigh the risks and options involved in undertaking further pregnancies. Genetic counselling is a valuable way of assisting couples in this process. The birth of a child with CF had a major impact on subsequent reproductive decisions for participants in this study. Most participants chose to reduce the number of children they originally planned to have following the diagnosis of their affected child. The most important reason given for the impact on reproductive decisions was the fear of the recurrence risk, the fear that another child would have CF.

Participants were generally aware of and in favour of prenatal diagnosis, but the majority indicated that they would not consider termination of pregnancy. The intention to use prenatal diagnosis was not necessarily linked to the intention to terminate in the case of an affected pregnancy.

Results from this study show that although the birth of a child with CF implies a risk for other relatives of being carriers, the genetics of CF is not often discussed with these family members. The extent of disclosure of genetic risk information to family members varied greatly among the participants in this study. Participants who had disseminated risk information to family members did so mostly in the context of explaining the disease as a whole, as well as to make family members aware that they were at risk of having a child with CF. The task of disseminating risk information fell mostly upon the women in this study.

The main barriers to sharing risk information was found to be a lack of knowledge about risk information in participants, as well as a lack of contact with certain family members, for reasons including divorce, separation, geographical distance and internal family rifts. There was a general lack of awareness of carrier testing among participants, including the application and limitations thereof; and poor dissemination of information about this topic in families.

Parents who had not yet informed their children about the genetics of the condition thought their children were too young to understand the significance of this information. A lack of knowledge about the genetics of CF in parents and misconceptions about children not being at risk were other reasons for non-disclosure to children. Genetic counsellors and doctors working with CF families need to guide parents on how, when and what information about CF, including the genetic aspects, to share with their affected and unaffected children.

In this small sample, the birth of at least one affected child could have possibly been prevented if risk communication had taken place effectively in this family. A lack of genetic knowledge and misconceptions prevented the dissemination of genetic risk information, resulting in family members being unaware of their potential carrier risk and ending in the birth of another child with CF in this family. Not conveying risk information denies relatives the opportunity to make informed reproductive choices.

Only two participants in this study had been for a genetic counselling session with a genetic counsellor following the diagnosis of their affected child. Genetic counselling was effective in enhancing knowledge about the genetics of CF in these individuals as indicated by their good performance on the majority of questions related to the genetics of CF and the availability of testing. Furthermore, genetic counselling facilitated risk communication as these participants also belonged to two of the families in which the dissemination of risk information was reported to have been done extensively and accurately.

The service that is delivered at the RCCH may be improved by having a genetic counsellor as a member of the interdisciplinary team involved with all families in which a diagnosis of CF is made. A genetic counsellor could play an important role in facilitating information giving, knowledge gain and the dissemination of risk information in the family. A genetic counsellor could help at risk couples understand the risks involved in undertaking further pregnancies and explain the various options available to them to allow them to make informed reproductive decisions. Finally, he/she could also assist in providing the necessary ongoing psychosocial support to CF families.

Chapter 6

RECOMMENDATIONS

5.1 RECOMMENDATIONS

Based on the outcomes of the study, the following recommendations are made:

- Information giving should not end at the initial session when the diagnosis is given. It should be followed up on a regular basis, at follow-up or routine clinic appointments by involving a genetic counsellor with all families in which a diagnosis of CF is made.
- More attention needs to be given to discussing the dissemination of risk information and possibility of carrier testing to relatives during counselling sessions with parents of affected children.
- Simple, written information should be provided in the home language of the families. It should include the implications of the diagnosis of a child with CF in the family and the importance of sharing this information with other relatives.
- Unaffected siblings of children with CF face the impact of a genetic condition without the support of genetic counselling (Fanos and Johnson 1995). Ways of ensuring they are accurately informed about the condition, their possible risk status and options available to them, including carrier testing, need to be considered.
- Considering the limited knowledge about carrier testing amongst the participants, a simple fact sheet explaining carrier testing, its applications and limitations would aid parents' understanding and facilitate sharing this information with relatives. It should be provided in the three official languages of the Western Cape. One of the main barriers to carrier testing is lack of awareness of the availability thereof and what it can and cannot do (Ormond *et al.* 2003).

6.2 FUTURE RESEARCH

It will be of great value to:

- Explore the knowledge of patients on the genetics of CF.
- Evaluate the knowledge of unaffected siblings and other relatives of children with CF and to explore effective ways of meeting their information needs.
- Continue to assess and develop effective interventions related to disclosure and communication of risk information in families with genetic conditions.
- Explore the attitudes to and knowledge of relatives of an affected child about carrier testing and reproductive options, such as prenatal diagnosis and termination of pregnancy.
- Evaluate the impact of genetic counselling by using pre and post genetic counselling genetic data collection projects.
- Include participants from all over SA to explore the experiences of a larger population, from a larger geographical area, including urban and rural areas.
- Compare the experiences of families living in rural areas to that of families living in urban areas in different areas of South Africa. This study only investigated families living in an urban area in the Western Cape.

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M.Sc in Genetic Counselling Research Project

An investigation into the level of genetic knowledge and family communication about genetic risk in parents of children with Cystic Fibrosis

PARTICIPANT INFORMATION SHEET AND CONSENT FORM

STATEMENT BY PARTICIPANT

I, ..., living at (address)

.....

confirm that:

1. I have been invited to participate in the above research project which has been initiated through the Division of Human Genetics, University of Cape Town because I have a child who is affected by Cystic Fibrosis (CF) and attends the CF clinic at Red Cross War Memorial Children's Hospital (RCCH).

I understand that the objective of this study is to investigate:

the level of knowledge of the inheritance of CF and knowledge of the carrier status of family members;

how this knowledge influences or has influenced reproductive choices; and

family communication about genetic risk information

I understand that the interview will take place in my home or another venue of my choice and that it may take one or two visits of two hours each.

2.3 I am aware that this is a once-off procedure that will be implemented in 2007 at a time convenient to me and my family.

2.4 I understand that some of the questions may make me angry or sad, but the risks to me from the study are minimal. The researcher will refer me to a genetic counsellor if necessary. She will show me respect, acceptance and empathy during the interview.

3.1 I have been assured that all information will be handled confidentially. Information may be used for a thesis, publications in scientific journals and presentations at professional congresses, but names will not be included.

3.2 I understand that the interview will be tape recorded so that the researcher does not have to write too much during the interview. The tape will be secured in a safe until the research has been written up and will then be destroyed immediately. The data stored on the computer will have a numerical code only and my name does not appear anywhere.

4. I have been assured that the recorded and transcribed information discussed at the meeting will only be made available to the researcher's supervisors with my study code number and that they do not know that it refers to my name.

5. I willingly agree to consent to taking part in the study and I have been informed that I may refuse to participate in this project and that I may stop participating at any stage, and that such refusal or stoppage will not in any way negatively affect my future access to medical and genetic services to which I am entitled.

6. .....has explained the information of the study to me in English/Afrikaans/Xhosa. I am proficient in that language and my questions have been answered satisfactorily.

7. I understand that there will be no medical benefits to me from this study.

8. I have been assured that participation in this project will not lead to additional costs for me or my family and I will not benefit from it financially.

I HEREBY DECLARE THAT I HAVE VOLUNTARILY AGREED TO PARTICIPATE IN THE ABOVE RESEARCH STUDY

Signed at:
(address) ..... on..... 2007

....
Participant's signature Witness

I HEREBY DECLARE THAT I AGREE TO HAVE MY INTERVIEW AUDIOTAPE RECORDED

Signed at:
(address) ..... on 2007

....
Participant's signature Witness

IMPORTANT INFORMATION

Dear Participant

Thank you for your participation in this study. Should you have any questions during the duration of this study regarding:

problems as a result of the research, or
questions regarding information about the project

please contact me at the following telephone number:

Mardelle Schoeman: (021) 406-6373
Email: schoeman@cormack.uct.ac.za

Prof Jacquie Greenberg: (021) 406-6299

If you have any questions about your rights as a research participant please contact Dr M Blockman, Chair of the Research Ethics Committee, Faculty of Health Sciences, University of Cape Town Ethics Review Committee on (021) 406-6496.

AFRIKAANS VERSION OF THE INFORMATION AND CONSENT FORM

M.Sc in Genetiese Berading Navorsingsprojek

'n Ondersoek na die kennis van genetika van ouers van kinders met Sistiese Fibrose en kommunikasie oor genetiese risiko in die familie

INLIGING EN TOESTEMMIMG VORM

VERKLARING DEUR DEELNEMER

Ek, ...,

(adres)

.....

bevestig dat:

Ek is uitgenooi om aan die bogenoemde navorsings projek wat deur die Divisie van Mensgenetika, Universiteit van Kaapstad geinisieer is, deel te neem aangesien ek 'n kind/kinders het met Sistiese Fibrose en die Sistiese Fibrose kliniek by Rooi Kruis Kinderhospitaal bywoon.

2.1. Ek verstaan dat die doel van hierdie projek is om die volgende te ondersoek:
- Kennis van die oorerflikheid van Sistiese Fibrose en kennis van die draer status van familielede;
- Hoe die kennis u besluite aangaande verdere voortplanting beïnvloed het
- Kommunikasie in die familie oor die genetiese aspek van Sistiese Fibrose

2.2. Ek verstaan dat die onderhoud of by my huis of by 'n ander plek van my keuse sal plaasvind en dat dit een of twee besoeke van twee ure elk behels.

2.3. Ek is bewus dat dit 'n eenmalige ondersoek is wat in 2007 sal plaasvind op 'n tyd wat vir my en my gesin gerieflik is.

2.4. Ek verstaan dat van die vra my hartseer of ongelukkig mag maak, maar dat die risiko's van die studie minimaal is. Die navorser sal my na 'n genetiese raadgewer verwys indien nodig. Sy sal my met respek, aanvaarding en empatie behandel geduurende die onderhoud.

3.1. Ek is verseker dat alle inligting vertroulik behandel sal word. Inligting mag vir 'n tesis, publikasies in wetenskaplike tydskrifte en aanbiedings by professionele kongresse gebruik word, maar name sal nie ingesluit word nie.

3.2. Ek verstaan dat die onderhoud op band opgeneem sal word sodat die navorser nie te veel hoef te skryf geduurende die onderhoud nie. Die band sal in 'n kluis gestoor word tot dat die navorsing opgeskryf is en sal daarna dadelik vernietig word. Die band en die data op die rekenaar sal slegs 'n numeriese kode op hê en my naam sal nie daarop verskyn nie.

4. Ek is verseker dat die inligting wat opgeneem en getranskribeer is slegs aan die navorser se mentor bekend gemaak sal word, maar dit sal slegs my numeriese studie kode bevat en my naam sal nie daarop verskyn nie.

5. Ek is nie oorreed om aan die die projek deel te neem nie en ek is bewus dat ek mag weier om deel te neem, en ek kan op enige staduim besluit om te onttrek. My onttrekking sal op geen manier my huidige of toekomstige toegang tot die mediese of genetiese dienste, waarop ek geregtig, is beïnvloed nie.

6. ..... het die inligting van die projek in Engels/Afrikaans aan my verduidelik. Ek is vlot is hierdie taal en my vra is ten volle beantwoord.

7. Ek verstaan dat daar geen mediese voordele vir my sal wees as gevolg van hierdie projek nie.

8. Ek is verseker dat my deelname aan hierdie projek nie tot enige additionele koste vir my familie sal lei nie en dat ek nie finansieel gaan baat daarby nie.

EK VERKLAAR HIERMEE DAT EK VRYWILLIG AAN DIE BOGENOEMDE NAVORSINGS PROJEK DEELNEEM

Geteken te:

(Adres) .....

op 2007

....

Deelnemer se handtekening Getuie

EK VERKLAAR HIERMEE DAT EK MY ONDERHOUD OP BAND OPGENEEM MAG WORD

Geteken te:

(Adres) .....

op 2007

....

Deelnemer se handtekening Getuie

BELANRIKE INLIGTING

Geagte deelnemer,

Baie dankie vir u deelname aan hierdie studie. As U gedurende die verloop van die navorsing enige vra het aangande:

1. probleme as gevolg van die navorsing, of
2. vrae aangaande inligting oor die projek

kontak my of Prof. Greenberg gerus op die volgende telefoon nommers:

Mardelle Schoeman (021) 406-6373
Email: schoeman@cormack.uct.ac.za

Prof Jacquie Greenberg: (021) 406-6299

As u enige vrae het in verband met u reg as 'n deelnemer, kontak D M Blockman, die Voorsitter van die Etiese Hersiening Komitee van die Universiteit van Kaapstad by (021) 406-6496.

INTERVIEW SCHEDULE

Participant No:.....

A. SOCIODEMOGRAPHIC INFORMATION

1. Family history

   - Date of birth
   - Marital status
     Single/Married/Divorced/Widowed
   - Consanguinity
   - How many children?
   - Ages of children
   - Age of affected child?
   - Rank of affected child?
   - Do you have siblings?
   - If yes, How many brothers and how many sisters?
   - Do they have children? Are they all well?
   - Are your parents still alive?

2. Diagnosis of CF
   - At what age was your child diagnosed with CF?
   - Is he/she the first individual in the family to have CF?
   - Tell me about the time leading up to your child being diagnosed with CF

3. Which grade/standard did you complete at school?
   (To be answered by father and mother)
   - Grade 12 (matric, Std 10)
   - Grade 11
   - Grade 10
   - Grade 9
   - Grade 8
   - Grade 7
   - Other

4. Have you started any further courses/training since leaving school?

   - Yes
   - No

5. If Yes to question 5, what?

   - No post-school

- Certificate from college
- Diploma (beyond Grade 12)
- Bachelors degree
- Postgraduate diploma/degree
- Other

6. Have you completed it?

7. If not, why did you not continue at school/ tertiary education?

8. Are you employed?

   - Yes
   - No

9. If Yes to question 8, what kind of work?

   - Self-employed
   - Full-time employed
   - Part-time employed
   - Casual

10. If No to question 9, are you:

    - Unemployed
    - Housewife
    - Full-time student
    - Part-time student
    - Permanently unable to work
    - Retired or pensioner
    - Other

11. Is the household income pooled together to pay for living?

    - Yes
    - No

12. What is your current household income per month?

    - No income
    - Disability grant
    - R1 – R400
    - R401 – R800
    - R801 – R1 600
    - R1 601 – R3 200

- R3 201 – R6 400
- R6 401 – R12 800
- R12 801 – R25 600
- R25 601 – R51 200
- R52 201 – R102 400
- R102 401 – R204 800
- More than R204 800

13. How many people does the income support?

B. RED CROSS WAR MEMORIAL CHILDREN'S HOSPITAL (RCWMCH) CYSTIC FIBROSIS CLINIC.

14. How often do you attend the CF clinic at RCWMCH?

15. How far is it from your home?

16. How do you get to the clinic? (Own transport, access to vehicle, bus, train, taxi?

17. If you use public transport, how far is this from you home and then how long is the journey to Red Cross?

18. How long does it take to get to the clinic?

19. How much does it cost to get there?

20. Do you have medical aid?
    - Yes
    - No

21. Does your medical aid cover your child's medical needs
    - Consultations
    - Medication
    - Hospital stays

C. LEVEL OF UNDERSTANDING OF GENETICS (ADAPTED FROM HAMES *ET AL.* 1991 and SOMER ET AL.1988)

22. What is the cause of CF?

23. Is CF caused by genes
    - entirely,
    - partly, or
    - not at all?

24. How is CF passed on?

25. What is the chance of having another child with CF?

26. Does CF affect both males and females?

27. In your family has (name of patient) inherited the abnormal gene(s) from:
    - father,
    - mother,
    - both, or
    - neither?

28. What is the chance of your unaffected children being carriers?

29. What is the chance of your siblings being carriers?

30. What is the chance of one of your siblings having a child with CF?

31. After having three affected children, what is the chance of having another affected child?

32. Is it possible to detect CF before a baby is born?

33. Is it possible to detect CF by amniocentesis?

34. After you received the diagnosis of CF, when was the genetics of the condition discussed with you?
    - At the time of diagnosis,
    - at a later stage
    - frequently
    - not at all

35. What are the source/sources of your information about the genetics of CF?
    - Doctors at CF clinic
    - CF association
    - Media
    - Internet
    - Other (please specify)

36. After you received the diagnosis of CF and understood the cause and the genetics, how did you feel?

D: IMPACT ON REPRODUCTIVE CHOICES

37. Have you had carrier testing?

38. If yes, what did you understand about it the test, what it means, and what it can or cannot tell you?

39. If no, what do you understand about carrier testing and how it works?

40. Has the diagnosis of cystic fibrosis in your child had an impact on further reproductive choices?

41. If yes, how and why?

42. Has it influenced the number of children you wanted to have?

43. Has your use of contraceptive methods changed since the diagnosis of CF was made in your child?

44. Did you/would you seek advice prior to the next pregnancy?

45. Would you/have you considered prenatal testing in future pregnancies?

46. If yes, would you consider terminating an affected fetus?

E. RISK COMMUNICATION WITHIN FAMILY.

47. Have you shared any information about (child name)'s CF with other immediate or extended family members? (Unaffected children, siblings, parents. Aunts, uncles, cousins)

48. How did you explain it to them?

49. If no to question 44, why not?

50. What have you told your child with CF about the disease?

51. What is your ill child's understanding about the disease?

52. What do your other children understand about the disease?

53. What questions have your unaffected children asked about the disease? How have you answered them?

54. What questions has you child with CF asked about the disease? How have you answered them?

55. Do your children discuss the condition with each other?

56. Have you shared any information specifically about the genetics of CF with other immediate or extended family members?

57. If yes, with whom?(Affected child., unaffected children, siblings, parents)

58. How did you explain it to them?

59. How long after being informed of the diagnosis of CF did you discuss the genetics of CF with immediate or extended family members?

60. How did they react?

61. Have any of your brothers and sisters been worried about their chance of having a child with CF?

62. How of ten do you discuss the genetic aspect of CF with family members
    - Regularly
    - Sometimes
    - Seldom
    - Never

63. Did you mention carrier testing to any immediate or extended relatives?

64. If yes, what did you tell them about it?

65. What are your reasons for giving them information about the genetics of CF?

66. Have any of them acted on this knowledge? (Seeked counselling, considered carrier testing, provided you with support?)

67. If you haven't told immediate or extended family members about the genetics of CF, what were your reasons for not communicating this information to them?

68. Who takes primary responsibility for disclosure and communication of information about CF in your family?

69. Do you feel confident that you had enough information about the genetic aspect of CF to explain it to family members?

70. Would you like more information about the genetics of CF?

71. In your opinion, is it your responsibility or that of the health care professionals to inform your relatives of the potential genetic risk?

ONDERHOUD SKEDULE

Deelnemer No:.....

B. SOSIODEMOGRAFIESE INLIGTING

1. Familie geskiedenis

   - Geboortedatum
   - Huwelikstatus
   -
   - Hoeveel kinders?
   - Ouderdomme van kinders?
   - Ouderdom van kind met CF?
   - U kind met CF, is hy/sy eersgeborene/tweede/derde/vierde/vyfde kind?
   - Het u broers en susters?
   - As ja, Hoeveel van elk?
   - Het hulle kinders? Is hulle almal gesond?
   - Leef u ouers nog?

2. Diagnose van CF
   - Hoe oud was u kind toe hy/sy gediagnoseer is met CF?
   - Is hy/sy die eerste persoon in die familie met CF?
   - Vertel vir my van die tyd wat gelei het daartoe dat u kind met CF gediagnoseer word

3. Watter graad/standerd het u voltooi op skool?

   - Graad 12 (matriek, Std 10)
   - Graad 11
   - Graad 10
   - Graad 9
   - Graad 8
   - Graad 7
   - Ander

13. Het jy enige verdere kursusse/opleiding gedoen na u skool verlaat het?
    - Ja
    - Nee

14. Indien JA op vraag 4, wat?

    - Ambag
    - Sertifikaat van kollege

- Diploma (na Graad 12)
- Bacculureurs graad
- Nagraadse opleiding of diploma
- Ander

15. Het u dit voltooi?

16. Indien NEE, hoekom het u nie skool/tersiëre opleiding voltooi nie?

17. Werk u?

- Ja
- Nee

18. Indien JA op vraag 8, watter tipe werk doen u?

- U eie besigheid
- Voltydse werk
- Deeltydse werk
- Casual/kontrak werk

19. Indien NEE op vraag 9, is u:

- Werkloos
- Huisvrou
- Voltydse student
- Deeltydse student
- Ongeskik vir werk
- Afgetree of pensionaris
- Ander

20. Hoeveel mense dra by tot die gesamentelike inkomste?

21. Wat is u huidige gesamentelike huishoudelike inkomste per maand?

- Geen inkomste
- Ongeskikheids toelaag R780

Salaris inkomste:

- R1 – R400
- R401 – R800
- R801 – R1 600
- R1 601 – R3 200
- R3 201 – R6 400

- R6 401 – R12 800

- R12 801 – R25 600
- R25 601 – R51 200
- R52 201 – R102 400
- R102 401 – R204 800
- Meer as R204 800

22. Hoeveel mense word deur hierdie inkomste onderhou?

B. ROOI KRUIS OORLOG GEDENK KINDER HOSPITAAL SISTIESE FIBROSE KLINIEK

14.Hoe gereeld woon un en u kind die CF kliniek by RCWMCH by?

15. Hoe ver is dit van u huis af?

16. Hoe kom u by d ie kliniek? (Eie vervoer, toegang tot motor, bus, trein, taxi)

17.Indien u van publieke vervoer bebruik maak, hoe ver moet u stap om daarby uit te kom en hoe lank is die trip Rooi Kruis toe?

18. How lank vat dit u om by die kliniek te kom?

19. Hoeveel kos dit om kliniek toe te gaan?

20. Het u mediese fonds?
- Ja
- Nee

21. Dek u mediese fonds u kind se mediese behoeftes?
- Konsultatises
- Medikasie
- Hospitaal besoeke

C. VLAK VAN VERSTAAN VAN GENETIKA (AANGEPAS VAN HAMES *ET AL.* 1991 EN SOMER ET AL.1988)

22.Wat verstaan u is die oorsaak van CF?

23. Word CF versoorsaak deur gene:
- heeltemaal,
- gedeeltelik, or
- geensins?

24. Wat verstaan u van die manier wat CF oorgeërf werd?
25. Wat is die kans om nog 'n kind met CF te hê?

26. Affekteer CF beide mans en vrouens?

27. Wie in 'n familie dra gewoonlik die abnormale geen oor:
    - vader,
    - moeder,
    - beide, of
    - nie een nie?

28. Wat is die kans dat u kinders wat nie met CF geaffekteer is nie draers van die CF geen is?

29. Wat is die kans dat u broers en susters draers van die CF geen is?

30. Wat is die kans date en van u broers of susters 'n kind met CF kan kry?

31. Nadat ouers drie kinders met CF gehad het, wat is die kans om 'n vierde kind met CF te kry?

32. Is dit moontlik om CF voorgeboorte te diagnoseer?

33. Kan CF deur middel van amniosintese gediagnoseer word?

34. Nadat u die diagnose van CF in u kind gekry het, wanneer is die genetika van die kondisie met u bespreek:
    - Terselftertyd wat die diagnose gemaak is
    - Op 'n latere stadium
    - Gereeld
    - Glad nie

35. Wat is die grootste bron/bronne van u inligting oor die genetika van CF?
    - Dokters by die CF kliniek
    - CF vereniging
    - Media
    - Internet
    - Ander (spesifiseer asb)

36. Nadat u die diagnose ontvang het en die oorsaak en genetika van CF vertaan het, hoe het u gevoel?

D: IMPAK OP VOORTPLANTINGS BESLUITE

37. Het u al draer toetsing gehad?

38. Indien ja, wat het u verstaan omtrent hierdie toets, wat dit beteken, en wat dit vir mens kan en nie kan sê nie
39. Indien nee, wat verstaan u van draer toetsing en hoe dit werk?

40. Het die feit dat jou kind met sistiese fibrose gediagnoseer is, jou besluit om weer swanger te raak beinvloed?

41. Indien ja, hoe?

42. Het dit die hoeveelheid kinders wat u wou gehad het, verander?

43. Het u gebruik van voorbehoedmiddeld verander na dit u kind met CF gebore is?

44. Het u/sal u raad soek oor die onderwerp voor u volgende swangerskap??

45. Het u/sal u voorgeboorte diagnostiese toetse oorweeg in volgende swangerskappe?

46. Indien ja, sal u dit oorweeg om 'n fetus geaffekteer met CF te termineer?

E. KOMMUNIKASIE OOR GENETIESE RISIKO IN DIE FAMILIE.

47. Het u enige inligting omtrent (kind se naam) se CF met enige van u nabye en ander familielede gedeel?(Ongeaffekteerde kinders, broers en susters, ouers, ooms, tannies, niggies, nefies?)

48. Hoe het u dit aan hulle verduidelik?

49. Indien nee op vraag 44, hoekom nie?

50. Wat het u vir u kind met CF van die siekte verduidelik?

51. Wat verstaan u kind met CF van sy/haar siekte?

52. Wat verstaan u ander kinders van die siekte?

53. Watter tipe vrae het u ongeaffekteerde kinders al gevra oor die siekte? Hoe het u dit geantwoord?

54. Watter tipe vrae het u kind met CF al gevra oor die kondisie? Hoe het u dit geantwoord?

55. Bespreek u kinders die kondisie met mekaar?
56. Het u enige inligting, spesifiek aangaande die genetiese aspek van CF, bv., u status as draer en daarom hulle risiko om draers te wees met enige van u nabye en ander familielede gedeel??

57. Indien ja op vraag op 53, met wie? (Ongeaffekteerde kinders, broers en susters, ouers, ooms, tannies, niggies, nefies?)

58. Hoe het u dit aan hulle verduidelik?

59. Hoe lank na u uitgevind het van die CF diagnose het u met ander familielede oor CF en hulle en hul genetiese risiko gesels?

60. Hoe het hulle daarop reageer?

61. Was enige van u broers of susters al bekommerd oor hul risiko om 'n kind met CF te hê?

62. Hoe gereeld bespreek u die genetiese aspek van CF met ander familielede?
    - Gereeld
    - Somtyds
    - Selde
    - Glad nie

63. Het u die moontliheid van draer toetsing met enige ander familielede bespreek?

64. Indien ja, wat het u hul daarvan vertel?

65. Wat is u redes waarom u vir hulle inligting omtrent die genetiese aspek van CF gegee het?

66. Het enige van u familielede al die inligting verder gevat of gebruik? (Gegaan vir berading, dokter gaan sien, toetsing oorweeg om te bepaal of hulle draers is, vir u ondersteuning gebied?)

67. Indien u nie vir familielede inligting omtrent die genetiese aspek van CF gegee het nie, wat was u redes waarom u nie die inligting aan hulle oorgedra het nie?

68. Wie in u familie het die grootste verantwoordelikheid geneem om inligting oor CF aan die familie oor te dra?

69. Voel u dat u genoeg inligting oor die genetiese aspek van CF het/gehad het om dit met ander familielede te bespreek?

70. Sou u graag meer inligting oor die genetiese aspek van CF wou hê?

71. In u opinie, is dit u verantwoordelikheid of die van die dokters en ander gesondheidswerkers om u familielede in te lig oor hul genetiese risiko?

UNIVERSITY OF CAPE TOWN

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16 October 2006

REC REF: 385/2006

Prof LJ Greenberg
Clinical Laboratory Sciences
Human Genetics
IIDMM

Dear Prof Greenberg

PROJECT TITLE: AN INVESTIGATION INTO THE LEVEL OF GENETIC KNOWLEDGE AND FAMILY COMMUNICATION ABOUT GENETIC RISK IN PARENTS OF CHILDREN WITH CYSTIC FIBROSIS.

Thank you for submitting your study to the Research Ethics Committee for review.

It is a pleasure to inform you that the Ethics Committee has **formally approved** the above-mentioned study.

Are you proficient in Xhosa (page of information sheet)? On page 11 you indicate that interviews will be conducted in English and Afrikaans, do you intend including Xhosa families which might require using an interpreter.

This serves to confirm that the University of Cape Town Research Ethics Committee complies to the Ethics Standards for Clinical Research with a new drug in patients, based on the Medical Research Council (MRC-SA), Food and Drug Administration (FDA-USA), International Convention on Harmonisation Good Clinical Practice (ICH GCP) and Declaration of Helsinki guidelines.

The Research Ethics Committee granting this approval is in compliance with the ICH Harmonised Tripartite Guidelines E6: Note for Guidance on Good Clinical Practice (CPMP/ICH/135/95) and FDA Code Federal Regulation Part 50, 56 and 312.

Please note that the ongoing ethical conduct of the study remains the responsibility of the principal investigator.

Please quote the REC. REF in all your correspondence.

kmjedi