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The effect of Fragile X syndrome on family relations

by

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DECLARATION

I declare that the dissertation, which I hereby submit for the degree Magister Educationis at the University of Pretoria, is my own work and has not previously been submitted by me for a degree at this or any other tertiary institution.

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MELISSA PIENAAR

30 November 2017

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DEDICATION

I dedicate this research to my husband, Helmut. Without you, I would have never taken this road. You are my true north.

~My True North~
Pamela Pante

I close my eyes and listen
To the soft sound of your voice as
You weave a new tale for me to hear
I see the pictures evolve before my eyes
As characters begin to appear
You take me by the heart
To places we dream to be
Painting our lives with your words
Endearing you more to me
Other may have touched my skin

You have touched my soul

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- Last, but not the least, my family, co-workers and those crossed my path in the last two years – it had to be fate.

"There is no greater disability in society, than the inability to see a person as more"

- Robert M Hensel -

ABSTRACT

Fragile X syndrome (FXS) is one of the most common forms of intellectual and developmental disabilities, as well as the most widely inherited intellectual disability, which has a tremendous impact on the family. FXS is, apart from Down syndrome, the most common form of an intellectual and developmental disability. The literature indicates that one in 2 500 to 4 000 females and one in 4 000 to 6000 females are affected by FXS. These numbers indicate that one in 260 females has the full-mutation gene, while one in 300 to 800 males has the premutation gene. Studies show that little research and resources with regard to FXS are available internationally, especially in South Africa.

Parents are often plagued by feelings of guilt as they are the carriers of the FMR1 gene that causes FXS. The mother is particularly affected, as she is often the main caregiver, as well the one who carried the FMR1 gene to her child. However, both parents suffer because they experience profound emotions such as the loss of their dreams for their child. These families are also socially isolated, as the extended family and friends often neither understand, nor accept the behaviour of a child with FXS. These children present with behavioural, emotional and physical difficulties. Some of the behaviour difficulties are hand-flapping, hand-biting, anxiety, tantrums and aggression. Furthermore, these children experience emotional difficulties, such as extreme shyness and feelings of rejection. They very often do not know how to deal with their emotions. These children also present with physical manifestations, such as prominent ears, long faces and double-jointedness. The marital relationship of the parents is affected, as they have to take care of a child with an intellectual disability and this often puts a lot of strain on the marriage. The siblings tend to feel rejected and neglected. As the extended family is also supposed to go for genetic counselling, denial and subsequent avoidance of social contact with the affected family are often experienced.

Situated within an interpretivist paradigm, this study followed a qualitative approach and employed a multiple case study design. Three experts on FXS were purposively selected to participate in the study based on their knowledge and

experience with families affected by FXS. The data were collected through interviews and a narrative. The findings indicate that in order for families to deal with the various challenges that FXS presents, support is needed from society, family and friends. The recommendations include that more awareness needs to be created about FXS. The empirical analysis in this study revealed that FXS has a severe impact on family relations.

Key terminology: Fragile X syndrome, premutation, full-mutation.

CERTIFICATE OF LANGUAGE EDITING

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DECLARATION

To whom it may concern

I hereby certify that the English language of the following dissertation meets the requirements of academic publishing. This dissertation was linguistically edited and proofread by me, Dr. L. Hoffman.

Title of dissertation

The effect of Fragile X syndrome on family relations

Candidate

Melissa Pienaar

Lariza Hoffman

Kroonstad

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CHAPTER ONE

INTRODUCTION AND ORIENTATION

1.1 INTRODUCTION

Fragile X syndrome (FXS), previously known as “Martin-Bell syndrome”, is one of the most common causes of intellectual and developmental disabilities, as well as the most widely inherited intellectual disability (Cornish, Turk & Hagerman, 2008:469). Landsberg and Swart (2011:422) add that FXS is the most common form of intellectual impairment, apart from Down syndrome.

Abrams et al. (2012:1126) define FXS as an intellectual disability, which can be either mild or severe and which affects a child’s intellectual and physical development. These authors further maintain that physical manifestations are often elusive in infants and young children but tend to be visible from the age of nine to twelve months in developmental and behavioural features, such as anxiety, attention problems and social avoidance (Abrams et al., 2012:1128). According to Howlin, Charman and Ghaziuddin (2011), developmental manifistations tend to be in factors such as speech, crawling and walking. Furtmemore, aspects of irritability and anxiety are also seen in the early developmental years of a child with FXS (Howlin et al., 2011).

Hayes and Matalon (2009:790) state that FXS was first diagnosed showing the fragility at the distal end of the X chromosome. Only later, when the FXS gene was discovered, did the diagnosis of FXS become more accurate. Raspa, Bailey, Bann and Bishop (2014:33) emphasise that FXS is passed to children through their parents, who unknowingly carry the Fragile X gene. Approximately 25% of families have more than one affected child because it is often only discovered at the age of 36 months, which results in some families already having conceived another child.

More recent research indicates that FXS is often first diagnosed between 36 and 40 months (Hayes & Matalon, 2009:790). Lisik (2017:28) argues that FXS is sometimes misdiagnosed and only later diagnosed due to little knowledge of this syndrome. Lisik also states that due to late diagnosis, families are affected not only in terms of the child being diagnosed but the families' entire structure that is turned upside down.

An intellectual disability, such as FXS, has an impact on families on various levels, as it is a complex condition, which, according to Cornish et al. (2008:469), has far-reaching implications for the developmental progress of the affected person. Finucane, McConkie-Rosell and Cronister (2002:18) point out that the child's development when he or she is diagnosed with FXS, depends largely on the way the family adapts after the identification of the disability. In this regard, Finucane et al. (2002:18) note that it is very difficult for most parents to accept the fact that their child has an intellectual disability and Abbeduto (2007:294) observes that most parents who have a child with FXS report high levels of stress.

This study explored the relations of families living with a young child with FXS and, furthermore, suggests guidelines for parents with children with FXS.

1.2 RATIONALE

Reinsberg (2015, n.p.) succinctly states, "As parents, we develop hopes and dreams about who our baby will be in the world and how we will be as parents." Therefore, Lee in Wall (2011) suggests that very often the diagnosis of an intellectual disability can be perceived as traumatic due to feelings of loss and grief. Parents are also often very confused as medical personnel do not always diagnose FXS correctly, which results in parents visiting various specialists before their child is correctly diagnosed. This author concludes that "parents of young children with special needs often tread a path that is steep, rocky and fraught with pitfalls" (Lee in Wall, 2011:38).

As a learning support teacher for the past three years, I developed a passion for learners with unique barriers, as I have always been interested in children, especially those with intellectual disabilities. I have often encountered parents who

have one normal and healthy child, and one child with a disability. This aspect enthused a keen interest in me as to how families cope with the news of knowing their newly born or recently diagnosed toddler has an intellectual disability. Hallberg (2014:1-2) asserts that having a child with a disability creates a lot of stress for parents, as the rearing of such a child is emotionally and financially taxing and has a tremendous impact on the family.

Growing up, I had a cousin who seemingly had an intellectual disability because of a lack of oxygen at birth. During this time, I often had the privilege of spending time with him and often saw the frustrations my aunt endured as my cousin developed and could not always reach the milestones typical of his age. It left scars on her marriage, finances and very often was the reason for him changing schools. Today, he is in a life-long care facility, because after my uncle's death, my aunt could not cope with the demands of having a child with an intellectual disability. This was my very first encounter with a child who had an intellectual disability and I saw the definite effects it had on my family. So many dreams and aspirations for my cousin either changed or were simply in vain. I often wondered if my cousin's life would have been any different if he were correctly and timeously diagnosed and given the appropriate therapy as a toddler. It is most likely that the family situation would have been different if they had family counselling and guidelines to help my cousin grow into an intellectually differently abled young man.

The past three years I also had the privilege to work with a girl who was diagnosed with autism. Having read all the available reports of her doctors and therapists, a clear picture formed of her early developmental years. She started to walk only at the age of three and talked at the age of five. She could not communicate properly during those early years and very often became frustrated. Clear patterns of autism, such as poor communication skills, sometimes inappropriate social behaviour, as well as struggles in her early school years, became part of the family's daily encounters. Although her parents did not cease to visit doctors and therapists on a regular basis and provided her with the best educational support, it also caused some difficulties in their family structure. The family had more expenses, had to endure long sessions of counselling and had to support their

elder daughter, who, at an earlier stage, could not understand her younger sister's behaviour at times. The mother admitted having an anxious personality, as she had to deal with many predicaments, having a child who was demanding and different.

Bröberg (2009:93) mentions that becoming a parent is, in most cases, overwhelming and accompanied by some lifestyle changes, but having a child with an intellectual disability can be even more overwhelming due to the child's temperament, personality traits, characteristics and other aspects pertaining to the specific intellectual disability. Chang and McConkey (2008:27) note that parents vary in the way they cope if they have a child with an intellectual disability and this experience may cause the parents additional stress and challenges. Roper (2014:241) elaborates by saying that parental stress is often exacerbated by maintaining sibling relationships.

Numerous studies focus on children with disabilities, but according to Raspa et al. (2014:44), more research should be conducted on the parents of children with disabilities.

Although new research is being conducted on the anxiety of the mothers of children with FXS or intellectual developmental disabilities, authors such as Raspa et al. (2014:44) encourage research with a focus on family adaptation when one of their members has been diagnosed with FXS. This study fills this gap by focusing on the experiences of parents coming to terms with having a child with FXS and the impact of such a syndrome on family relations. Mulroy (2008:2) posits that research in the previous decade revolved around parents' experiences concerning caring for children with an intellectual disability; however, current research focuses on the potential effects on the siblings.

I decided to focus on the impact of a child with FXS on family relations in its entirety. The insights derived from this study may serve as guidelines to parents who have children with intellectual disabilities. The insights derived from this study might also serve as guidelines for health professionals.

1.3 PROBLEM STATEMENT

Carvajal (2011:11) refers to the difficult journey of parents who have children with FXS and mentions that these parents usually visit various specialists and therapists before the final diagnosis is given. This author further maintains that apart from having to deal with having a child with an intellectual disability, the parents need to process the fact that FXS is a genetically transferrable condition (Carvajal, 2011:11). Cornish et al. (2008:469) refers to the emotional impact of the diagnosis, as the parents usually feel guilty because they believe that they are responsible for the disability. In addition, this syndrome drains the parent emotionally, as most carriers of FXS have a low self-esteem, which leads to the parents giving these children more attention. Finucane et al. (2002:19) and Abbeduto (2007:240) also identify challenges in raising children with FXS, as these children often have the characteristics of autism spectrum disorder and, therefore, have problems in communicating. The financial implications of having a child with FXS are high, and as such, the children require numerous therapies. Neely-Barnes (2008:93) concludes that parents who have children with FXS, therefore, require more support educationally, socially and emotionally.

The following research questions guided this study:

1.3.1 Primary research question

- What is the effect of Fragile X syndrome on family relations?

1.3.2 Secondary questions

- What is the profile of a child who presents with FXS?
- What challenges do families with children with FXS experience?
- What coping strategies do parents use in raising a child with FXS?

The aim of this study was to explore the impact of FXS on family relations with a child or children with FXS. The research specifically focused on the challenges and coping experiences of families who have a child with FXS.

1.4 LITERATURE REVIEW

A literature review reflects on current as well as historical research (Maree, 2013:26). In order to contextualise the study, the broader concept of intellectual disabilities is discussed first, followed by a discussion on the historical aspects of FXS, as well as the genetic implications of FXS.

1.4.1 Intellectual disabilities

A definition of intellectual disability, which had a profound impact on me, was that of the American Association on Mental Retardation (AAMR):

> [Intellectual disability] is not something you have, like blue eyes or a bad heart. Nor is it something you are, like short or thin. It is not a medical disorder or a mental disorder. A particular state of functioning begins in childhood and is characterized by limitations in both intelligence and adaptive skills. Intellectual impairment reflects the 'fit' between the capabilities of the individual and the structure and expectations of their environment. (Bornman & Rose, 2014:113)

An intellectual disability can be referred to as a "disorder" of incomplete or detained mental development, principally characterised by the deterioration of concrete functions at each stage of development and contributing to the overall level of intelligence, such as cognitive, language, motor and socialisation functions, where, in this anomaly, adaptation to the environment is always affected (Katz & Lazcano-Ponce, 2007:133). According to the Intellectual Disability Rights Service in Australia (2009:1), an intellectual disability is a disability that occurs during or after birth in the developmental period of life before the age of 18 years and is typically characterised by below-average intellectual functioning. Bornman and Rose (2014:113) allude to the ambiguity in the terminology used when people refer to people who are intellectually impaired. Concepts such as "mentally retarded", "cognitive impairment" and "intellectual disability" are also used. Notwithstanding an intellectual impairment, depending on how the family of origin handles such a family member, a child with a disability such as FXS has a place in society and has a distinct identity.

1.4.2 Aetiology of FXS

First discovered by James Purdon Martin and Julia Bell in 1943, FXS was first known as “Martin-Bell syndrome” and is in modern times referred to by either this name or “Fragile X syndrome” (Phadke, 2005:1). According to the National Fragile X Foundation (2009:1), the name of this syndrome was derived from the Fragile X gene, FMR1, which looks like the letter “X”. Under the microscope, a portion of the X chromosome appears to be “broken” or “fragile”. FXS is a genetic condition caused by gene-related changes in the FMR1 gene (Bornman & Rose, 2014:116). The cause of this condition can be traced back to mutations in the FMR1 gene on the X chromosome, which are the reason for the changes in the gene as well (Landsberg & Swart, 2011:422).

FXS affects twice as many males as females, and individual needs concerning someone diagnosed with FXS, vary according to the seriousness of the diagnosis (Landsberg & Swart, 2011:422). Kidd (2014:1001) further reports that FXS is associated with other syndromes such as cardiac disorders, otolaryngology disorders, gastrointestinal disorders, ocular disorders, sleep problems, growth problems and neurological disorders, such as autism and movement disorder.

Several manifestations and physical features may be present in a child who has FXS. Bornman and Rose (2014:117) identify five categories that present the most prominent features of a child with FXS. The first category refers to intelligence and learning, and involves those children with FXS who are severely intellectually impaired. These children usually struggle to process information, especially problem-solving skills. Landsberg and Swart (2011:451) add that FXS is a profound cognitive impairment with usually severe learning difficulties. The next category involves the physical features of FXS. A prominent feature of individuals with FXS is their extraordinary physical appearance. Bornman and Rose (2014), therefore, list a second group of characteristics, labelled “physical appearance”, when they describe the facial and bodily appearance of individuals with FXS. According Bornman and Rose (2014:117), the Department of Communities (2014:4) and Fernandez (2011:41-44), children with FXS may have the following physical features:

- A larger head than that of the average child
- A chest that appears to be resonating
- A long face and prominent chin
- Protuberant ears
- Particularly soft and smooth skin
- Puffy eyelids
- A single palmar crease
- A high-arched palate

Another group of manifestations involves behavioural aspects. Even prior to a diagnosis, other physical and behavioural aspects, such as anxiety, poor concentration, fixation, biting hands, tantrums, late toilet-training, sleep disturbances, avoidance of touch and difficulty in changing routines, may become discernible (Department of Communities, 2014:4). Many of these aspects will start to show when the child is a toddler and, therefore, diagnosis is an important part of the therapeutic intervention a child with FXS may need (Department of Communities, 2014:6).

Those with FXS often have few social and emotional skills. They often present challenging behaviour and stereotypical movements, such as hand-flapping (Bornman & Rose, 2014:117). These seemingly uncontrolled movements may, according to Kaufmann et al. (2004:225), contribute to the fact that many boys with FXS also display characteristics of autism, such as hand-flapping, little to no eye contact and shyness. An interesting fact is that at least 10% of boys with autism also have FXS (Bornman & Rose, 2014:117). Cuzzocrea (2014:14) also mentions that children with FXS may have difficulties interacting on a social and emotional level with their families and peers.

Speech and language skills is another challenging aspect, identified by Bornman and Rose (2014:117). These authors explain that a child with FXS may have a speech and language impairment depending on the degree of intellectual impairment. Their speech may sound cluttered and dysfluent, and they tend to repeat certain words and sounds (Bornman & Rose, 2014:117).

The last category involves the sensory skills of children with FXS (Bornman & Rose, 2014:117). Scerif, Cornish, Wilding, Driver and Karmiloff-Smith (2004:116) state that children with FXS are prone to have middle-ear infections and visual difficulties. According to Baranek, Chin, Hess, Yankee, Hatton and Hooper (2002:539), children with FXS tend to struggle with sensory processing and responses. Furthemore, they also state that spesific difficulties, such as sensory stimuli, were one of the aspects which children with FXS were found to have difficulties with (Baranek et al., 2002:539).

1.5 THEORETICAL FRAMEWORK

According to Anfara and Mertz (2015:15), a theoretical framework does not have a clear definition, but can be viewed as any empirical or quasi-empirical theory of social and/or psychological processes at a variety of levels, which can be applied to the understanding of a phenomenon. In the case of this study, I aimed to understand the impact of FXS on family relations. As the family is part of a broader community, the reciprocal impact of the various social systems must be explored in order to obtain an in-depth understanding of how a syndrome, such as FXS, can influence a family. I, therefore, opted to use Murray Bowen's family systems theory as the theoretical framework of this study. Dr Murray Bowen designed the family systems theory in 1974, which was one of the first complete theories of family systems functioning (Brown, 1999:94).

According to Hall (2013:15-16), Bowen's theory describes the emotional processes believed to have an influence on human behaviour. Additionally, she argues that Bowen's theory theorises human behaviour in a broad context and then assumes the existence of behaviour, as well as suggests that the intense emotional interdependency in families may predict family interaction and behaviour in other groups or settings. Furthermore, Hall also gives a perspective that families have a strong influence on one another's behaviour (Hall, 2013:15-16).

Bowen's family systems theory was, therefore, applicable to this study, as it explores the family as a unit and gives an in-depth view of a family experience of an emotional and societal matter. Titelman (1999:64) states that Bowen's model enables the researcher to gain a perspective on the phenomenon under study.

1.6 RESEARCH METHODOLOGY

Maree (2013:31) states that research is not only about understanding the world, but also about understanding the world through one's own views. Research methodology is seen as a manner to solve a research problem systematically (Sivasubramaniyan, 2012:8) and, therefore, stipulates the manner in which the research will be conducted. Thomas (2010:8) suggests that the research methodology consists of a research design and research methods.

1.6.1 Research design

Creswell (2003:3) explains that a research design is a manner of showing how the research will be conducted, whereas Maree (2013:34) suggests that a research design in a qualitative study can be interactive in nature, especially when using a case study as a research type. In relation to the research design, the research paradigm, research approach and type of research that were used, are discussed next.

1.6.1.1 Research paradigm

A research paradigm incorporates systems of consistent practice and thinking that defines the nature of the research (Terre Blanche, 2006:6). Nieuwenhuis (2007:47) defines a paradigm as the broad framework of assumptions or beliefs about fundamental aspects of reality in which yield themselves to a particular world-view. According to Nieuwenhuis (2007), three main paradigms can be distinguished, namely positivism, interpretivism and critical theory. This study is situated within the interpretive paradigm.

Interpretivism involves the practice of interpretation and providing a particular belief system about something (Nieuwenhuis, 2013:58). Considering FXS, the interpretive paradigm allowed me to understand the manner in which parents deal with their child who has FXS, as well as the belief system of a family with a member affected by FXS and the way they deal with the challenges posed by the disability. Nieuwenhuis (2007:60) makes the following statement:

> The aim of interpretivist research is to make available a perspective of a situation and to analyse the situation under study so as to provide an

> understanding into the way in which a particular group of people make sense of their situations or phenomena they encounter.

I attempted to understand how a family with a child with FXS makes meaning of their situation.

1.6.1.2 Research approach

This study followed a qualitative research approach. Creswell (2009:173) states that a qualitative study involves different philosophical assumptions, strategies of inquiry and methods of data collection, analysis and interpretation. Nieuwenhuis (2013:51) argues that qualitative research is a research methodology concerned with understanding the processes and the social and cultural contexts of a phenomenon.

Nieuwenhuis (2013:59-60) gives the four characteristics of qualitative research, which were followed in this research:

- Human life can be understood from within.
- Social life is a distinctively human product.
- The human mind is the purposive source or origin of meaning.
- Human behaviour is affected by knowledge of the social world.

In Chapter 3, all four these aspects, as well as their application to this study are discussed.

1.6.1.3 Research type: case study

According to Creswell (2003:15), a case study is a programme in which the researcher explores a process and certain individuals through a variety of data collection methods over a sustained period. A case study involves an in-depth investigation of “a program, an event, an activity, a process or one or more individuals” (Creswell, 2013:15). This study involved a comprehensive investigation into three professionals in the field of FXS, who have worked with families affected by FXS. One of these experts is also the mother of a child with FXS. Nieuwenhuis (2013:75) explains that the term “case study” has multiple meanings, as it can be used to describe a unit of analysis or to describe a research method as such. As this study was situated within an interpretivist paradigm, I strove to give an overall understanding of how the participants behave and feel in a family context, given the fact that one of their family members has

FXS. Hence, the aim of using the case study as a research type was to provide a comprehensive understanding of the possible effect of FXS on family relations.

Rule and John (2011:7) assert that case studies are generally used for many purposes in research, such as to generate understanding, explore a problem, generate theoretical insights, provide a level of generalisation on concepts and, lastly, for teaching purposes in order to illuminate broader concepts. These aspects are the reason I used a case study design, as it allowed me to provide insight on FXS and to understand the possible effect of FXS on a family.

1.6.2 Research methods

According to Creswell (2009:15), research methods elaborate on how the research is conducted in the form of data-gathering techniques, analysis, interpretation and the researcher's purpose for the research. As I followed a qualitative research approach, I used qualitative research methods, which, according to Creswell (2009) were established in the social sciences to allow researchers to study social issues as well as cultural phenomena. As part of research methods, I will discuss the role of the researcher, the research site, the participants, the data collection methods and the data analysis and interpretation.

1.6.3 Participants and research site

Three participants were selected to participate in the study, based on their expertise and experience with FXS. The first participant is a paediatrician in children's health, who specialises in autism, among other health concerns. She was also part of a team who opened a clinic for children. Participant one was specifically chosen for her 30 plus years' experience in children's health. She has encountered a few families that have been affected by FXS. The second participant is a keen researcher in the field of FXS. She was chosen as a participant in this study as she has worked and lived with families affected by FXS. The third participant is the founder of the first organisation for FXS in South Africa. She has experience in working with families affected by FXS. Moreover, she helped these families through her own personal experience of having a son with FXS.

For the purpose of this study, the participants consisted of professionals in FXS who have worked closely with families affected by FXS. I chose to work only with experts in FXS, as I wanted to gain insight into their experiences of having worked with FXS families.

In an endeavour to identify suitable participants, I approached the Genetics Centre at the University of Johannesburg, as well as several doctors in the Randburg area to assist in identifying participants. I also visited a school that indicated its willingness to enlist the help of a volunteer family. However, as I struggled to gain participants, I chose to work with only professionals in FXS, as this would also allow for a more objective view of FXS and its effect on families.

I interviewed all the participants through email by asking about their experiences and knowledge of FXS.

1.6.4 Data collection

According to Nieuwenhuis (2013:81), most qualitative studies treat data collection and data analysis as an ongoing, iterative and cyclical process. This was done to gain deeper insight into the study. I gathered information through structured interviews and a narrative from the expert who is also the mother of a child with FXS.

Interviews in case studies have long been conversations between the researcher and the research participants about the phenomena of interest in the study (Rule & John, 2011:64). In this research, I followed a structured interview approach. Structured interviews are seen as thorough questions that are set in advance and very often used in case study research (Niewenhuis, 2013:87). In this research, I gave the participants a set of six questions that was developed in advance, which they had to answer and return to me. I also asked one of the participants to write a short narrative about her experience working with families affected by FXS as well as her journey of her own personal experience, if she would be comfortable to do so. Creswell (2009:13) states that narratives are used when participants are asked to write their own “life story”.

1.6.5 Data analysis and interpretation

Creswell (2009:14) states that the process of data analysis and interpreting involve many components of making sense of text and image data. The data analysis strategy that was followed in this study was an inductive approach. Inductive analysis is used when a study is conducted in a natural setting in order to gain more understanding of the context (Maree, 2013:37). Maree and Van der Westhuizen (2013:37) claim that inductive data analysis is more likely to identify multiple realities potentially presented in the data, which is in line with an interpretivist paradigm. According to an interpretivist approach, there is more than one reality to a phenomenon and, therefore, inductive data analysis will provide the opportunity to present each reality in its essence to the study.

The data were, therefore, considerate of the multiple meanings of the situation in which the families found themselves. These findings were organised into meaningful themes and categories to simplify the task of analysis.

1.7 ETHICAL CONSIDERATIONS

Before starting a research project, it is important to consider the ethical implications of the study (Creswell, 2009:73), since ethical factors in social research are important, as the research deals with an important social issue. As I was exploring FXS in a social context and working with professionals, it was important that proper ethical caution was taken, not only to make sure the study was dependable, but also to ensure the participants' anonymity.

1.8 TRUSTWORTHINESS

According to Nieuwenhuis (2013:80), it is generally accepted that the use of multiple data-gathering techniques, such as observations and interviews, leads to trustworthiness. In this study, I used both interviews and a narrative, which contributed to the trustworthiness, conformability, credibility and transferability of the study.

1.8.1 Credibility

Shenton (2004:64) states that credibility has to do with how the findings of a study correspond with reality. In this study, I aimed to ensure credibility by persistent observation in the field and engagements with my participants.

1.8.2 Dependability

Dependability is seen as a substitute for reliability (Rule & John, 2011:107). Morrow (2005:2252) states that dependability refers to the way in which the study is conducted through consistency across time. Maree (2013:299) argues that dependability also denotes the degree to which the reader can be assured that the findings occurred as the researcher say they did. This research was conducted by a single researcher and all the activities in this study was consistent and chronological.

1.8.3 Transferability

Transferability is referred to as the alternative to generalisability (Rule & John, 2011:105). Morrow (2005:252) adds that transferability also refers to the extent to which the readers can generalise the findings of the study to their own context, in other words, how the findings of a study can be applied in similar situations. I believe that my research findings may be applied to other contexts where children with disabilities are part of a family.

1.8.4 Conformability

Conformability refers to a qualitative investigator's concern to objectivity in a study (Shenton, 2004:72). In order to ensure an objective approach, I conducted various readings and studied the findings of previous researchers' ideas in the field of FXS.

1.8.5 Final remarks on ethical considerations

Ethical considerations are important in research, as it promotes the norms and aims of the research (Resnik, 2015:15). Maree (2013:300) reports that students must obtain permission from the relevant board of ethics in order to work with

people in their research. I applied for ethical clearance from the Ethics Committee of the Faculty of Education, University of Pretoria, to conduct my empirical study.

Hill (2005:61) states that involving children in research requires care and thoughtfulness. I did not use children in this study, but still considered the professionals who work with children and families with FXS. The participants were assured of their voluntary participation and informed that they could withdraw at any time during the course of the study. Their identities were protected, and pseudonyms were used.

1.9 OVERVIEW OF CHAPTERS

Below is a short overview of the chapters in this study. The purpose of this overview is to interest the reader in the research that was conducted.

Chapter 1 – Introduction and orientation

Chapter 1 introduces the reader to this study, exploring the effect of FXS on families. This chapter discusses the rationale, problem statement, research methodology, and so forth. This chapter is concerned with giving the reader a brief, but concise overview of what will be investigated and how it will be done.

Chapter 2 – Theoretical framework and perspectives from literature

Chapter 2 discusses the literature relating to FXS as it applies to this study. The literature that is discussed, involves the key literature details of FXS, as well as the impact of intellectual disability on families. Bowen's family systems theory as the theoretical framework of the study is deliberated on.

Chapter 3 – Research methodology

This chapter is of the utmost importance for this study, as it explains how the study was conducted. Moreover, it specifically deals with the design, participants, data collection methods, data analysis, conclusions and ethical considerations of the study, of which each of these elements is discussed in depth in Chapter 3.

Chapter 4 – Data analysis and research findings

This chapter discusses the research analysis and findings. This chapter is divided into two main parts. The first part elaborates on the interviews with the professionals, as well as the observations and insights derived from the narratives. The last part of this chapter discusses the findings derived from the data that were collected.

Chapter 5 – Interpretation of research findings

This chapter deliberates on the findings and observations I made throughout my study. These findings are interconnected with those of Bowen's family systems framework, which I used as the theoretical framework on which the study is based.

Chapter 6 – Summary, conclusions and recommendations

This chapter presents an overall summary of the study, as well as the insights derived, and the conclusions made. Furthermore, it presents recommendations to the families affected by FXS, which will contribute to the research in the field of FXS.

1.10 CONCLUDING REMARKS

Chapter 1 introduced the study to the reader. The focus of this chapter was to provide the rationale, problem statement, research methodology and ethical considerations of this study.

Chapter 2 discusses the literature in the field of FXS, with the emphasis on intellectual disabilities, FXS as an intellectual disability and the impact of FXS on families. Correspondingly, Chapter 2 discusses Bowen's family systems theory as the theoretical framework of this study.

CHAPTER 2

LITERATURE FINDINGS

2.1 INTRODUCTION

The focus of this study is to explore the impact of a child with Fragile X syndrome (FXS) on the family. In the previous chapter, an outline of the study was provided by discussing what the study entailed and how it was conducted.

In this chapter, specific attention is given to the incidence of intellectual disabilities, with the emphasis on FXS and the impact of this syndrome on family relations. Accepting a child with disabilities in a family can be very challenging. In this regard, Taanila, Syrjälä, Kokkonen and Järvelin (2002:73) note that the "birth of a disabled child is an event that affects all the family members as well as their internal and external relationships". Having a child with an intellectual disability is often seen as a situation to which families struggle to adapt (Koller et al., 1992:315). In some cases, intellectual disabilities are passed on from one generation to another, as is the case with FXS (Hall et al., 2007:29). Passing on a genetic syndrome, such as FXS, very often causes parents to feel guilty and responsible for their child's disability (Caravajal & Aldridge, 2011:16). It is documented that mothers experience the most stressors when they have a child with FXS (Abbeduto et al., 2004:238). The reason for this is that mothers are often the main caregivers of children with FXS and blame themselves for passing the gene to their children (Caravajal & Aldridge, 2011:18). Therefore, a closer look is given to the mother's experience of having a child with FXS. Furthermore, specific attention is given to the manifestations of FXS as an intellectual disability and also how FXS affects family relations.

The chapter concludes with a discussion on Bowen's family systems theory.

2.2 THE IMPACT OF INTELLECTUAL DISABILITIES ON THE FAMILY

Simpson, Mizen and Cooper (2016:679) suggest that intellectual disabilities are seen as a group of “causes” or diverse conditions that occur during the developmental phases of an individual. It is further characterised by a considerably below-average intellectual functioning and adaptive behaviour. These authors further state that intellectual disabilities refer to functioning and adaptive behaviour, which are at least two, but can be more standard deviations below the mean (Simpson et al., 2016:679). This means that intellectual disabilities are characterised by the limitations of intellectual functioning, as well as adaptive behaviour, in social, practical and adaptive skills. In most cases, intellectual disabilities occur before the age of 18 years (Schalock et al., 2007:118). In the case of FXS, individuals with FXS often have severe cognitive and adaptive failure (Dean et al., 2016:304), often first presented within the first few years of the individual’s life (Caravajal & Aldridge, 2011:33).

Fragile X Syndrome (FXS) is the most common inherited form of intellectual disability (Hersh & Saul, 2015:995), which is, according to Bagni, Tassone, Neri and Hagerman (2012:4214), concerned with mutations in the FMR1 gene in the X chromosome, causing various difficulties such as academic, communication and emotional problems. Therefore, Herefordshire (2016:1) coins FXS as a genetic intellectual disability for which affected families often seek genetic counselling (Bagni et al., 2012:4315). FXS is both an intellectual and a developmental disability, often resulting in affected families being forced to accept that there is no cure for this syndrome and that they will be forced to make some lifestyle changes and resort to special education and vocational therapy for their children (Dean et al., 2016:306).

An intellectual disability affects the functioning of an individual’s communication skills, sensory and gross motor development and behavioural aspects, which usually appear to be normal. It also affects an individual’s cognitive functioning and the overall limitations of an individual in terms of adaptive behaviour (Schalock et al., 2007:118). Many parents suspect that something might be wrong with their child due to poor development, which is usually apparent in the child’s communication (Caravajal & Aldridge, 2011:13).

The child's reduced intellectual capacity has a profound impact on his or her family, especially if the individual needs intense support. Very often the individual's family members are the prime caregivers (Neely-Barnes & Dia, 2008:93). Lackwitz (2012:656) furthermore asserts that although there has been an improvement in the quality of life for families with intellectually disabled individuals, as well as for the disabled individuals themselves, there are still families that suffer due to the unavailability of resources. This author also refers to the number of stressors on mothers, as they usually are the main caregivers (Lachwitz, 2012:655).

Parents often feel guilty, lonely and totally isolated when they have a child with an intellectual disability (Caravajal & Aldridge, 2011:19). Research on families who have to deal with an intellectual disability within their family context is a recent topic in the field of research and, according to Blacher et al. (2013:166), most research focuses on the negative impact of intellectual disabilities on families. However, recent studies have been reporting on the positive aspects of a family member with an intellectual disability, which is due to the current age in which we live and the resources these families have at their disposal (Neely-Barnes & Dia, 2008:95). In many cases, an intellectual disability, such as FXS, is manageable, but families need to accept a few facts about the syndrome (Caravajal & Aldridge, 2011:82).

Families in their greater context are often seen as the most influential relationship in the early years of a child's development, as the most crucial relationships are formed within the family context (Wall, 2006:34). Bronfenbrenner (1994), in his ecological systems theory, refers to the interactions and relationships between family members, which exert a determining influence on a child's development. However, Bronfenbrenner (1994:4) also points to the reciprocal relationship between family members. In this regard, Wall (2006: 35) argues that family has a direct influence on a person's development, whereas Kearney and Griffin (2001:582) maintain that in many instances where a child has an intellectual disability, it holds far-reaching implications for the family relationships. In this regard, Beresford (2007:2) postulates that parenting a child with an intellectual disability has long been seen as going further and beyond "ordinary" parenting (Beresford, 2007:2). Ungar (2016:23) refers to "challenging contexts", implying that

parents often struggle to raise children with intellectual disabilities, as the disability causes much stress.

There are many reasons why an intellectual disability may have an impact on a family. Researchers, such as Bailey et al. (2003), Abbeduto et al. (2006), Raspa, Sacco, Candrilli, Bishop and Petrillo (2016) and Weber (2016), focused in their studies on caring for an individual with an intellectual disability. However, there is a scarcity of research on how a child with an intellectual disability affects the identity of the family, as well as the extent of the emotional impact of such a child on the family. Parents often feel that they lose their personal identities, as they not only need to play the role of parent and dominant caregiver, but also need to deal with how the society sees them as a family (Beresford, Rabiee & Sloper 2007:2). These authors add that the parents of children with intellectual disabilities see themselves as different from parents with "normal" children (Beresford et al. 2007:2). In this regard, Kearney and Griffin (2001:583) mention that many parents are seen as "almost dysfunctional" because they "shop around" for opinions to confirm their unrealistic wishes that there is nothing wrong with their children. This aspect truly emphasises that not only is raising a child with an intellectual disability a difficult fact indeed to make peace with, but also accepting the fact that one's child's journey in life will be different from that of others. In a way, this aspect could also contribute to the parents' emotional state when they raise a child with an intellectual disability. Beresford et al. (2007) maintain that these parents often express a need for emotional support, as society does not always understand their circumstances and is quick to judge. According to Kearny and Griffin (2001:582), even though parents often find joy in their child with an intellectual disability, it is most often feelings of sorrow that prevail when they take care of their child. The parents are faced with not only the initial diagnosis, but also the financial burden and difficulties in schooling, limited support in many instances and internal family relations, such as sibling relationships (Schladant, 2011:39).

Another factor that needs to be considered, is cultural stigmatisation when the individuals and their families are faced with an intellectual disability. In order to illustrate this, Begum (2007:12) reports that in many cultures, receiving therapy might be considered a foreign concept, as there are certain stigmas around people

who are intellectually disabled. She uses the example of being blind, which in the Bangladeshi culture, is seen as the result of being bewitched or having dangerous powers (Begum, 2007:14). Pitten (2008:1) mentions that cultural values form people's roles and perceptions, especially when they are confronted with a life-changing event. In her study, she pays specific attention to the role of the families' culture when a child has been diagnosed with autism (Pitten, 2008:3).

It is, therefore, clear that intellectual disabilities have an impact on families. However, to truly understand the impact of intellectual disabilities on a family, FXS, as one of the most common intellectual disabilities, is discussed in the following section.

2.3 FRAGILE X SYNDROME (FXS)

Beresford (2007:3) believes that in order to understand FXS and its origin, this disability needs to be studied in detail. Consequently, the Genetics Home Reference (GHR, 2012:2) – a guide to understand genetic conditions – explains that genes are transferred on structures called "chromosomes". A foetus inherits a set of 23 chromosomes from its mother and a set of 23 chromosomes from its father. The chromosomes are numbered according to size, starting with the largest, which is chromosome 1 up to 22, which is the smallest of them all. The last pair of chromosomes (23), determines the gender of the child (GHR, 2012:2). There are two kinds of chromosomes, X and Y chromosomes, which control whether a person is a male or female (Anonymous, 2012:2). Females inherit two X chromosomes, one from her mother and one from her father, while a male inherits an X chromosome from his mother and a Y chromosome from his father (Anonymous, 2012:2). Bornman and Rose (2014:117) add that as males contribute a Y chromosome to their sons, and not an X chromosome, FXS cannot be transmitted by the father. However, if the male, in this case the father, carries the full-mutation gene, it will be passed to his daughter. If a female carries the Fragile X gene, it can be transmitted to both her sons and daughters (Bornman & Rose, 2014:117). Thus, each gender produced by the female has a 50% chance of inheriting the Fragile X gene. Raspa et al. (2016:844-845) explain that FXS is caused by the silence of one single gene, called the "gene 1" in the X chromosome (Hall et al., 2008:489). Within the FMR1 gene, there are many

mutations, causing a “trinucleotide repeat”. In an FXS case, this repeat is between 50 and 200 repeats, causing the FXS “premutation”. Typically, men are more affected than women, since men only have one X chromosome and women are born with two X chromosomes.

Bornman and Rose (2014:117) furthermore state that not everyone with FXS has the same signs and symptoms. The Department of Communities, Child Safety and Disability in Australia (2014:4) agrees by stating that FXS can affect individuals in different ways, as some individuals with FXS may have mild learning difficulties, while others may have severe intellectual impairments and behavioural difficulties.

FXS is one of the most common inherited genetic forms of mental incapacity among families (Hersh & Saul, 2015:994). Mental impairments are associated with more than 500 conditions and FXS is one of the most overlooked mental impairments (Duane, 2003:2), which Hall et al. (2008:469) maintain can be found in all population groups. Researchers such as Garber, Vistootsak, Abbeduto and Warren (2008:1), as well as Raspa et al. (2016:844), regard FXS as the most common inherited mental disability. Hagerman (2008:498) adds there are a great variety of factors that characterise FXS, which can be found on a continuum, implying that the intelligence quotients (IQ) of the individuals can range from low to high levels. Thus, many individuals who have this intellectual impairment may present normal emotional behaviour or IQ levels (Hagerman, 2008:499). Individuals with FXS often fail to develop in terms of learning, maturation and social adjustment (Dean et al., 2016:303), therefore indicating that FXS, as an intellectual disability, affects the development of an individual on various levels.

It is stated that all the sons of a father with a premutation will inherit his Y chromosome and that the father will pass on his X chromosome to all his daughters, which, in turn, will produce a premutated X chromosome. Moreover, a mother carrying the fully mutated X chromosome will have a 50% chance of having a son or daughter without FXS. If she passes on her fully mutated X chromosome, there is a 50% risk that her sons and daughters may have FXS. Females have two X chromosomes, which may lower the risk of showing too many signs of FXS. Males, on the other hand, inherit an X chromosome from their

mother and a Y chromosome from their father. In turn, if the X chromosome is affected, the male will have FXS (Caravajal & Aldridge, 2011:25). Diagnosing FXS is often viewed as an uneven road due to the lack of information on this syndrome, as well as a resistance to accept the diagnosis thereof (Caravajal & Aldridge, 2011:56).

2.3.1 Diagnosing Fragile X Syndrome

In a study concerning FXS as a common, but underdiagnosed disease, Lisik (2017:27) remarks that the diagnosis of FXS is difficult due to nonspecific symptoms. Ciaccio et al. (2017:2) state that prenatal and neonatal diagnosis is not always possible when there is no history of the FXS gene in families due to the lack of ultrasounds and clinical features. They further state that children with FXS often have normal height and weight at birth. Genetic counselling is thus of the utmost importance when any family, especially a family with no family history of FXS, receives a diagnosis.

The National Fragile X Foundation in San Francisco (NFXF, 2002:18) states that parents often feel devastated when their child is diagnosed with FXS. As children are usually diagnosed around 36 months to around four years, parents may often already have another child, which may cause uncertainty about the second child's wellbeing (Raspa et al., 2014:43).

According to the FRAXA website (2017:1), although FXS is one of the most common intellectual disabilities, most people who have FXS have not been diagnosed yet. More awareness should be created with regard to FXS as, although the syndrome is common, many people in the medical field, as well as the public, are not aware of it (Krause, 2015:544). Lachiewicz, Dawson and Spiridigliozzi (2000:800) mention that FXS is the leading cause of mental retardation and yet is underdiagnosed, especially among boys. According to Kaufmann et al. (2004:224), FXS is almost synonymous with autism and around 15 to 33% of boys with FXS will be autistic. Unless professionals have the knowledge of the different FXS phenotypes often present, a misdiagnosis can be made (Krause, 2015:544).

Diagnosing FXS is an important matter, as families need the necessary guidance, counselling and support to deal with it (Neely-Barnes & Dis, 2008:93). Not knowing what is wrong with one's child is yet another important reason why parents should consider an FXS screening or diagnosis. After such a diagnosis, the parents should become involved in a community of support. Genetic counselling is always recommended after the completion of tests and the diagnosis of FXS, as parents have much to come to terms with (Caravajal & Aldridge, 2011:82). Ungar (2016:9) also mentions that a diagnosis enables the parents to help the child develop within their capabilities.

2.3.2 Implications of a Fragile X Syndrome diagnosis

Parents are usually continually aware of their children's development, and when their child does not develop as he or she should, they immediately start seeking advice and help from their family members and professionals (Caravajal & Aldridge, 2011:33). Diagnosing FXS is a complicated process, as the early signs and symptoms of FXS are not visible at birth and are actually "discovered" (Bailey et al., 2009:527). The first signs and symptoms of FXS only present themselves at the first developmental phases of a child's development (Caravajal & Aldridge, 2011:53). The early signs of FXS are often seen at month 32, when parents may see certain physical features, such as a long face with prominent ears (Hersh & Saul, 2015:995). Parents often only receive a diagnosis after 36 months, which leaves them with a delay in the therapy and genetic counselling families must receive (Pembrey et al., 2001:4). Bagni et al. (2012:4314) suggest that the diagnosis can be either early, but more often quite late. In this regard, Carvajal and Aldridge (2011:11-12) refer to a case study indicating that parents often tread a steep path of going from one professional to another before being confronted with an FXS diagnosis. After such a diagnosis, the parents often feel confused because, in many cases, they have never heard of FXS and now they have to face not only the diagnosis, but also the reality that if they have more than one child, the siblings may also have FXS due to the genetic components of FXS (Caravajal & Aldridge, 2011:58).

Diagnosis before three years of age is usually seen as early. By this time, the parents may already have noticed some speech difficulties, motor difficulties,

hand-flapping (boys), poor eye contact and irritability (Bagni et al., 2012:4315). There is a difference in the symptoms of FXS between boys and girls, which will be discussed later in this chapter. A late diagnosis usually occurs after three years of age, with patterns of symptoms presenting themselves to be fixed (Bagni et al., 2012:4315).

The diagnosis of FXS has varied over the past years. In 1969, Herbert Lubs introduced a “chromosome/cytogenetic” test, which indicated whether an individual had FXS. This was replaced in 1991 by a molecular test, which tested the FMR1 gene (Caravajal & Aldridge, 2011:53). The FMR1 gene is the gene where the “trinucleotide” repeat shows, as discussed earlier in this study. A more complex description of FXS is provided by Garber et al. (2008:666):

> FXS, an X-linked dominant disorder with reduced penetrance is associated with intellectual and emotional disabilities ranging from learning problems to mental retardation, and mood instability to autism. It is most often caused by the transcriptional silencing of the FMR1 gene, due to an expansion of a CGG repeat found in the 50 - untranslated region. The FMR1 gene product, FMRP, is a selective RNA-binding protein that negatively regulates local protein synthesis in neuronal dendrites. In its absence, the transcripts normally regulated by FMRP are over translated. The resulting overabundance of certain proteins results in reduced synaptic strength due to AMPA receptor trafficking abnormalities that lead, at least in part, to the fragile X phenotype.

A much more simplified description of the genetic component of FXS is that the mutations in the FMR1 gene occur on the X chromosome, which causes FXS (Jooste & Jooste, 2011:422). Bornmann and Rose (2014:116) state that the tip of the X chromosome seems to be “fragile” or damaged. Before blood tests are taken, one can also identify certain features of FXS, according to a checklist developed by Meas (Meas et al., 2000:212), which was used to identify men and boys who might have FXS. Another questionnaire for families was developed in order to identify FXS in the families that are supposedly carrying the FXS gene (Garber et al., 2008:669). Both these methods are merely a screening for FXS in families. The most accurate method of knowing whether one has a child with FXS or if one’s family is affected by FXS, is doing a genetic test at an institution, such as a genetic clinic. A DNA test is done, which is seen as the most accurate way to

test for the FMR-1 gene. Another manner would be to combine the DNA test with a PCR test. A PCR test is a test where the possible Fragile X mutations in an individual are identified. PCR products are used on silver-stained polyacrylamide gels, which then clearly shows the chain reaction of mutations, identifying FXS with a 98% accuracy (Haddad et al., 1996:808). A combination of the DNA test and the PCR test will give a 99% accurate result whether an individual has FXS (Caravajal & Aldridge, 2011:55). Prenatal diagnosis is available, but only if the mother is a premutation or a full mutation, due to the fact that she can pass the gene to her son (Macpherson & Murray, 2016:8). A premutation or a full mutation is the mother who carries the FMR-1 gene and run the risk of having children with FXS and FXTAS. Mothers have a 50% chance of giving it to their sons. Fathers with the FMR1-related gene have a 50% chance of giving it to their daughters (Saul & Tarleton, 2008:2).

Before any diagnosis, the parents usually notice symptoms that are quite alarming to them, usually consisting of cognitive, emotional and behavioural symptoms (Caravajal & Aldridge, 2011:31-32). According to Tranfaglia (2012:282), persons with FXS suffer enormous functional impairment, which is beyond their intellectual impairment. This is usually as a result of maladaptive behaviour and emotional disturbance, called the “behavioural manifestation” of FXS. Further attention will be given to the specific symptoms later in this chapter.

Diagnosing FXS is not just as simple as a blood test. One needs to consider the fact that it is a genetic disease that needs genetic counselling and therapy, not only for the individual, but also for the family. In an article about new-born FXS screening, Christie et al. (2013:302) assert that no condition is incurable, due to the availability of programmes designed for the prevention of and support for conditions such as FXS. Although this statement does not say that FXS is curable or preventable, it postulates that it is possible to live with FXS, as support is available in the form of therapy for the individual and for his or her family members

Carvajal and Aldridge (2011:63) advise that when one considers a screening test for FXS, it is important to acknowledge the fact that a positive diagnosis has consequences, not only for the individual, but also for the family. Therefore,

genetic counselling is recommended when a family encounters an FXS diagnosis, as parents often experience feelings of loss and grief, as such a diagnosis is neither easy to accept, nor to adapt to. Yet again, one must consider the impact of the diagnosis, as well as the aspect of support in some cultural systems. According to Begum (2007:12), there must be culturally appropriate methods to provide counselling not only for those who have been diagnosed with FXS, but also for their family members.

In order to understand the family's experiences and emotions, one has to understand the long-term effects and feelings, as well as the adaptations with which the family must make peace. These factors are discussed in the following section.

2.3.3 Impact of the Fragile X Syndrome diagnosis on the family

Caravel and Aldridge (2011:82), in their book, *Understanding FXS, a guide for families and professionals*, aptly remark as follows:

> It is understandable that when a family are confronted with such a diagnosis and receive a confirmation of the condition, that they may feel emotionally overwhelmed by the complexity of the disorder and the implications this has for the family.

To me, this quotation confirms the tremendous emotional impact of such a diagnosis and the subsequent changes the diagnosis implies. The early identification of children with FXS is of great importance, but the ramifications of such a diagnosis may have a great impact on the family (Bailey et al., 2000:315). The family has to adapt to the member who has FXS, especially after the diagnosis. This is often a complex process (Raspa et al., 2014:33).

Families learn much about FXS when they are confronted with the diagnosis of a child and/or sibling with the syndrome. Learning about FXS after diagnosis is often perceived as overwhelming and complex (Caravajal & Aldridge, 2011:82). It is also important to note that by the time the parents receive the diagnosis of FXS, they often have had numerous visits to various doctors and specialists, and they may already feel overwhelmed (Bailey et al., 2003:407). Although the parents learn

about the learning and developmental aspects of their child from the specialists after an FXS diagnosis, each individual with FXS develop in his or her own unique way (Caravajal & Aldridge, 2011:116).

As there is no cure for FXS (Picker & Sudhalter, 2012:1), the parents of a child with FXS must adhere to all the available treatments and support from their doctors and the community in order to maintain a healthy family environment and a place where their child with FXS can have a family life. Although they often experience feelings of loss and mourning due to the loss of their initial dreams they had for their child (Caravajal & Aldridge, 2011:63), Roper (2014:241) warns that such a diagnosis may affect family and sibling relationships negatively.

Although few studies have been done on the effect of FXS on siblings in a family context (Hall et al., 2007:29), one report (Anonymous, 2016) mentions that siblings are often tormented by feelings of embarrassment, isolation, resentment, responsibility and pressure to succeed; on the other hand, siblings may also experience maturity, insight, tolerance, patience and loyalty.

Socially, FXS has a huge impact on families and the social wellbeing of families, especially the mothers, who experience stress (Weber, 2016:2). Socially, many factors influence the parents of a child with FXS, such as social support, social life and knowledge with regard to FXS (Raspa et al., 2014:33). Weber (2016:6) also states that the mothers of children with FXS report that their children struggle to interact socially with their peers (Weber, 2016:6). The mother of a child with FXS described her child's social wellbeing as follows in Weber's study:

> They (my children) live a bit of an isolated life, because I always kept them for myself. I did what I needed to do to help them by to the best I can. But as far as going out and being with people and socialising, I didn't do enough of that. And that, it thinks, that, it was like not good. She did not learn how to socialise. (Weber, 2016:6)

From the above description, for most families, socialising with what is perceived as a "normal" family, may be overwhelming and difficult. Various consequences arise when parents have a child with FXS. Hal, Burns and Reiss (2007:29) state that in most cases, there have been reports of stress and marital problems. Mothers pass

the FXS gene along to their sons, while fathers pass the gene along to their daughters (Caravajal & Aldridge, 2011:25). Girls have only a 50% chance of presenting symptoms of FXS due to the fact that they have another X chromosome, while boys only have one X chromosome inherited from their mother. This means boys present with the most severe symptoms of FXS, implying that FXS is presented more in boys than in girls (Saldarriaga et al., 2014:190). McCarthy et al. (2006:688) comment that mothers often feel guilty as they are responsible for the presentation of FXS in their sons, which occurs more often than in girls. This explains why the mothers generally feel much guiltier than the fathers of children with FXS.

Mothers tend to have the primary responsibility of child care and often, in cases where the child has a disability, are the ones who take care of and face the challenges associated with their child's disability. The mothers of children with FXS often tend to be anxious and depressed (Abbeduto et al., 2004:237). In an article written by Ruth, this mother of two middle-aged sons with FXS narrates about the challenges of having not only one, but two sons with FXS (Ruth & Rachel, 2017). Rachel, another mother, mentions that due to few resources and little support from specialists, dealing with a child with FXS, who shows behavioural difficulties, are often the burden of their mothers, which adds to the pressure they feel. Rachel also mentions that to many people, FXS is a foreign concept, which aggravates the diagnosis, resulting in a lonely and unsure process, especially when looking at the future of one's child (Ruth & Rachel, 2017). Since FXS in the family is an aspect that often makes parents feel lonely and isolated (Caravajal & Aldridge, 2011:82), it is important that these families have the support and guidance they require (Abbeduto et al., 2004:238).

Hall et al. (2008:46) report that children with FXS often present with various emotional, behavioural and learning difficulties associated with developmental difficulties. This can be difficult for parents to adapt to when their child is diagnosed with FXS. The parents need the necessary support and, therefore, support groups and genetic counselling are recommended (Care, 1997:166). The parents also ought to deal with how FXS manifests, which Garber et al. (2008:667) lists as behavioural difficulties, learning difficulties and physical features, such as

flat feet. Autism is also part of the package among 30% of boys with FXS, as well as emotional difficulties, social difficulties and hyperactivity.

For the parents, not only the stress of the initial diagnosis, but also the realisation that their child is “different”, are difficult to accept (Minnes et al., 2007:120). However, Reilly (2016:93) mentions that there are parents who report positive aspects with regard to raising a child with FXS. In his study, 95% of the parents mentioned the sense of humour of these children, as well as other aspects, such as joy, a changed outlook in life, more tolerance for others, being more patient, having compassion, friendship, the opportunity to learn and develop and even positive sibling relations, although Reilly’s research indicates that 30% of the participants indicated a negative impact on sibling relationships

Closer to home, Weber (2016:1) argues that there is little understanding of FXS in South Africa, which leads to families perhaps not knowing from what condition a loved one is suffering (Weber, 2016:1). This aspect may also lead to families struggling to understand the sibling with FXS, which may lead to families being isolated from one another and experiencing stress in the family (Caravajal & Aldridge, 2011:82). Greenberg, Kromberg, Loggenberg and Wessels (2012:253) state that although much progress has been made with regard to genetic disorders, such as FXS, specifically in South Africa, cultural and religious beliefs about this syndrome may still cause problems. Some cultures do not believe in mental disorders and, therefore, also do not pursue any therapy for these individuals. Although families with an individual with FXS may face many challenges with regard to the diagnosis, relationships, as well as religious and cultural beliefs, they further face the fact that their preconceived ideas and dreams for the family member with FXS may have to change.

Receiving an FXS diagnosis and accepting the difficulties and even the challenges that come with FXS can be overwhelming. Even more than this, the initial aspect of not knowing what is wrong can also be problematic. Therefore, it is important to make sure of the initial symptoms of FXS in order to obtain clarity. FXS can be both rewarding and overwhelming in a family context. The symptoms of FXS are discussed in the following section.

2.4 MANIFESTATIONS OF FRAGILE X SYNDROME

FXS presents with symptoms that affect the individual's behaviour, emotions and physical as well as intellectual capabilities (Saldarriaga et al., 2014:190). These aspects with regard to FXS are discussed as follows.

2.4.1 Behavioural symptoms

The following behavioural symptoms often occur in individuals with FXS (Hagerman et al., 2009:3):

2.4.1.1 Poor eye contact

Caravajal and Aldridge (2011:39) explain that eye contact regulates someone's needs, apart from the fact that it is perceived as proper social behaviour. Individuals with FXS often show little to no eye contact (Bornman & Rose, 2014). According to Ke et al. (2005:255), people with FXS are completely at a loss when it comes to eye contact, facial and bodily expression. This factor often prevents them from giving a clear perception of their needs and desires. Poor eye contact can also be problematic when an individual with FXS encounters someone from a different culture. In Western cultures, poor eye contact may be perceived as disrespect or dishonesty.

2.4.1.2 Excessive shyness

Excessive shyness is another manifestation of FXS. According to Bagni, Tassone, Neri and Hagerman (2012: 4315), one of the most common characteristics among people with FXS is shyness, which also affects their social behaviour. People with FXS may struggle to communicate their needs due to the fact that they are shy (Powell, 2004:37-38).

2.4.1.3 Anxiety

According to Bailey, Skinner and Sparkman (2003:408), both male and female persons with FXS often present with high anxiety levels. Hersh and Saul (2015:998) suggest that very often, anxiety-related disorders, such as obsessive-compulsive disorder and perseverative behaviour, as well as aggressive and self-injurious behaviour, are seen in both male and female carriers, often resulting in

emotional problems and outbursts. Furthermore, anxiety is often detected in the way people with FXS behave in social contexts, as perceived in behaviour, such as hand-flapping and -biting (Hersh & Saul, 2015:996), which are discussed further in the next section.

2.4.1.4 Hand-flapping, hand-biting and sensory difficulties

Hand-flapping is one of the earliest signs of FXS (Bagni et al., 2012:4314). According to Kaufmann et al. (2004:232), hand-flapping is often associated with autism. This aspect also contributes to the fact that most boys with FXS often present autism. Hand-biting, according to Picker and Sudhalter (2012:3), is a self-abuse behavioural characteristic in many individuals with FXS, which results in substantial oral tendencies where these individuals try to put things, such as too much food, toys or fabric, into their mouths.

Another manifestation among people with FXS is tactile defensiveness (Garber, Visootsak & Warren, 2008:2). In a study done by Baranek et al. (2002:538), it was noted that children with FXS struggle with sensory processing and, therefore, need therapy as infants or children to develop coping strategies.

2.4.1.5 Aggression

In a study by Hall, Barnet and Hustyi (2016:1191), around 80% of individuals with FXS display aggressive behaviour. Antipsychotic drugs are usually prescribed to individuals with FXS in order to control signs of irritability, aggression and mood instability among these individuals (Caravajal & Aldridge, 2011:66-67). Hagerman (1999:1) explains that persons with FXS often feel aggression due to boredom and frustration, especially during the adolescent years when hormonal changes are presented (Hagerman, 1999:1).

2.4.1.6 Attention difficulties

Both boys and girls with FXS present with attention difficulties (Kapalu & Gartstein, 2016:891). Tranfaglia (2012:16) notes that in most cases, people with FXS have attention deficit hyperactivity disorder (ADHD), which makes educating them challenging. O'Connor (2008:1) agrees when she mentions that around 80% of

children with an FXS diagnosis will also have an ADHD diagnosis, often accompanied by attention difficulties, hyperactivity and impulsivity.

2.4.1.7 Hyperactivity and impulsivity

Chromik et al. (2012:4315) argue that other than the cognitive and social difficulties of a person with FXS, they also often present with symptoms of ADHD, such as hyperactivity and impulsivity. Often these manifestations of FXS lead to the individual being anxious and then behaving even more hyperactively or impulsively. Hyperactivity, impulsivity and attention difficulties in FXS are seen as underdeveloped milestones; these difficulties are typically seen in the early years of individuals with FXS (Cornish & Scerif, 2004:128).

2.4.1.8 Hyperarousal to sensory stimuli

Hyperarousal and sensory difficulties are further alarming characteristics of FXS in the early years. O'Connor (2008:1) states that sensory and tactile difficulties are important characteristics of those with FXS. Often sensitivity to sensory stimuli in the environment leads to people with FXS being anxious and difficult (Bagdasarian, 2014:12). Individuals with FXS are often hyperaroused due to extensive sensory stimuli. This aspect often leads to heightened and prolonged arousal, where there are excessive auditory, visual or tactile stimuli, which may lead to the individual acting out in terms of tantrums, hyperactivity, anxiety and restricted verbal output (Hersh & Saul, 2015:996).

Furthermore, the aspect of hyperarousal to sensory stimuli may contribute to financial strain and the emotional draining of the parents, as they have to deal with medical bills and nurturing their children with FXS when they are sick or struggle with, for example, communication difficulties. Fernandez (2011:76) states that occupational therapists can work with children with FXS, focusing on sensory integration, but yet again, this contributes to financial pressure on the parents. Children and especially toddlers with FXS display hypersensitivity to sounds, smells and tastes (Fernandez, 2011:77). Baranek et al. (2002:544) postulate that sensory difficulties are more present in boys than in girls. It is evident from the aforementioned aspects that there are clear physical and behavioural signs that

are visible in children with FXS, but diagnosis is a key element of providing clarity to the families who are the carriers of the Fragile X gene.

2.4.1.9 Autism

Fragile X syndrome and autism are associated disorders due to the shared characteristics that are presented in these syndromes (Demark, 2003:29). According to Devitt, Gallagher and Reilly (2015:92), both FXS and autism are neurobiological disorders, which often exhibit overlapping behavioural symptoms, although the disorders have different neurobiological underpinnings. Bagdasarian (2014:22), the president of the Fragile X Alliance of Ohio and the mother of two children diagnosed with FXS, emphasises that there are three areas of impairment in autism, namely social impairment, communication impairment and restricted interest or repetitive movement (Bagdasarian, 2014:22). These areas identified in autism are often also presented as behavioural problems in FXS (Kaufmann et al., 2004:231).

Around 30% of people with FXS present with symptoms of autism, such as hand-flapping, hand-biting, perseverative speech, poor eye contact and social problems (Hersh & Saul, 2015:996). Autism is more common in boys than in girls with FXS and is accompanied by severe socialisation difficulties in some cases (Kaufmann et al., 2004:232). It is important to note that according to Bagdasarian (2014:22), the manifestations of autism in FXS are often more apparent before the age of five years and as the individual grows older, they decrease.

2.1.4.10 Mood lability

Aspects of mood lability are often associated with FXS. These are common behavioural symptoms displayed by both boys and girls who have FXS. However, there are clear differences between the symptoms displayed by boys and girls (Hagerman et al., 2009:3). Girls with FXS display relatively milder cognitive and behavioural problems, including shyness, anxiety, social anxiety and some features of ADHD (Hagerman et al., 2009:3). Boys with FXS often display autistic behaviour (Kaufmann et al., 2004:225). According to Hagerman et al. (2009:3), 30% of boys with FXS meet the formal diagnostic and statistical manual disorders

of autism. These symptoms can be seen and tested through the screening tool presented in Practical Genetics (Garber et al., 2008:669).

2.1.4.11 “Splinter skills”

Splinter skills are the indices of isolated abilities that do not replicate accurate functional abilities (Bagdasarian, 2014:22).

2.4.2 Emotional difficulties

Emotional difficulties are one of the symptoms displayed by individuals with FXS. According to Practical Genetics (Garber et al., 2008:667), emotional difficulties include social interaction difficulties and controlling emotions, as well as other emotional problems associated with FXS. Emotional problems include behavioural difficulties, such as outbursts, loss of self-motivation, difficulty with social integration and self-abusive behaviour (Picker & Sudhalter, 2012:1). Other emotional difficulties that are often quite visible are anxiety, obsessive thoughts, schizophrenia and depression (Hersh & Saul, 2015:997). At this stage, it seems quite clear that there may be a link between the emotional and behavioural patterns of a child with FXS.

2.4.3 Physical features of FXS

Bornman and Rose (2014:117) present the following image of the facial features of a child with FXS:

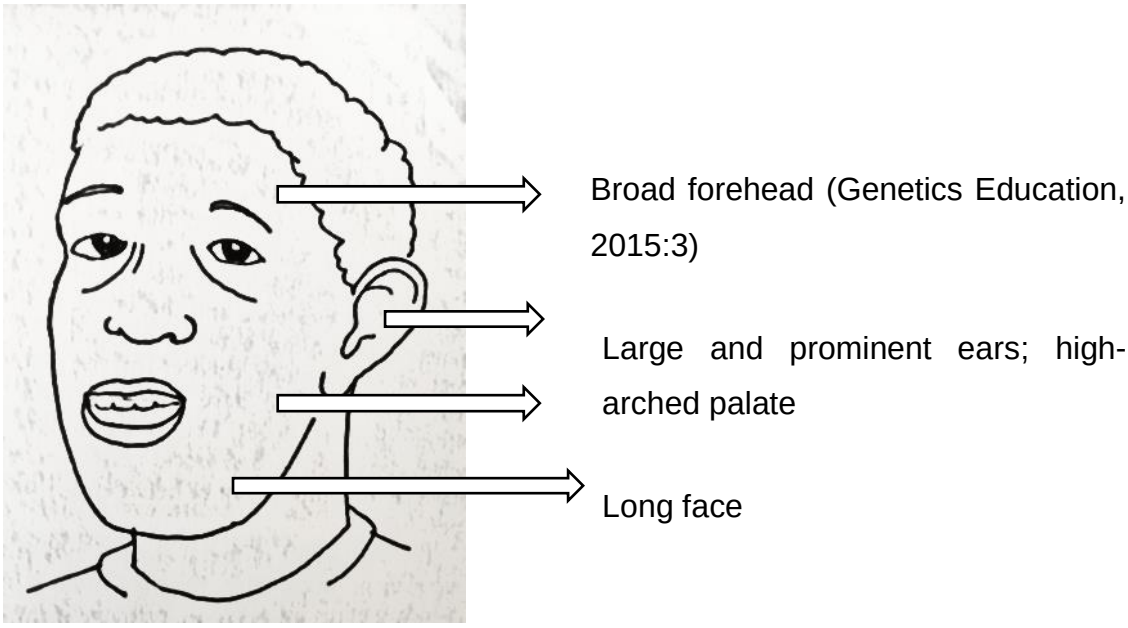

Figure 2.1: Facial characteristics of a child with FXS (Bornman & Rose, 2014:117)

Figure 2.1 depicts the physical features of an individual with FXS, in this case, a boy. Note the broad forehead, large ears and long face. However, there are many more physical characteristics with regard to FXS, as stated by the Centre for Genetics Education in Australia (Genetics Education, 2015:3). According to this organisation, boys with FXS also have enlarged testicles. Furthermore, those with FXS also have connective tissue problems (e.g. flat feet and loose joints). Hagerman (2004:11) adds that children with FXS also often have hyperextendable finger joints, double-jointed thumbs, single palmar crease, hand callouses, in some cases a heart murmur or click, vision problems, soft and stretchable skin, sinusitis, ear infections and, often, seizures. This author also notes that in most cases, children with FXS do not show these physical symptoms at infancy (Hagerman, 2004:11). Numerous therapies for these physical symptoms are usually required, which places more stress on the parents, as well as pressure on the relationships in the family (Raspa et al., 2014:34).

2.4.4 Intellectual disabilities in FXS

Most of the young children with FXS present with intellectual difficulties (Saldarriaga et al., 2014:190). Toddlers with FXS often show speech and language delays as well as other intellectual impairments (Kapalu & Gartstein, 2016:891). Since girls have two X chromosomes and boys only have one X chromosome, boys with FXS are often more affected by intellectual impairments than girls with FXS (Garber et al., 2008). In this regard Fernandez and Aldridge (2011:68) mention that more than 80% of males with FXS are cognitively delayed and, in general, cognitive challenges manifest in problems with logical thinking, problem-solving and overall intelligence challenges.

Children with FXS also present with ADHD, with the emphasis on concentration difficulties in academic situations, as well as impulsivity (Hagerman et al., 2009:3). Developmental delays are almost synonymous with FXS and the parents of children with FXS have reported that their children's academic situation is delayed (Hersh & Saul, 2015:999). Bagni et al. (2012:4317) elaborate by explaining that children with FXS have trouble with academics due to their difficulties with memory, perception, language and processing information, as a result of their anxiety levels. Boys with FXS often present with autism and, therefore, have difficulty expressing themselves through language (Ke et al., 2005:1).

It is important to remember that although children with FXS have academic difficulties, family involvement in academic situations is a way of making academics not only more bearable, but also more enjoyable for the learner with FXS (Hagerman et al., 2009:477). According to Hall et al. (2008:477), numerous studies indicate that people with FXS do struggle academically, but it is important to ensure realistic academic expectations for the child with FXS. This study used Bowen's theoretical framework of the family as a system. Bowen's family systems theory, as it is incorporated in this study, is discussed in the following section.

2.4.5 Speech and language difficulties

Ungrund (1998:13) elaborates by placing specific emphasis on echolalia, which is specific communication difficulty, resulting in even more frustration and challenges

for the parents and siblings when the affected person cannot communicate effectively. Brady, Skinner, Roberts and Hennon (2006:345) point to the fact that children with FXS tend to have distinct patterns of language and speech delays in their development. These authors assert that moderate to severe delays in both female and male persons have been reported, but more impairments presented in males. Therefore, children with FXS may have difficulty communicating their needs and frustrations to their families and peers, which may contribute to the families becoming frustrated when they are not able to communicate properly with their child with FXS.

2.5 THEORETICAL FRAMEWORK

Bowen's family systems theory was developed in 1974 with the aim of giving a comprehensive understanding of how family systems function (Brown, 1999:94). Titelman (1998:51) explains that Bowen's family systems theory starts by gathering facts and patterns that comprise the family system. This material becomes a map in which the researcher obtains facts concerning the planning of the intervention needed. These facts become the basis of gaining an understanding of what each family member's role is (Titelman, 1998:52). In this study, data were collected with regard to the impact of a child with FXS on family relations.

Rabstejnek (2012:4) states that Bowen's theory consists of eight key concepts, which are discussed and applied to this study as explained in the following section.

2.5.1 Concepts in Bowen's theory and the relevance to this study

Table 2.1: Concepts in Bowen's theory and their relevance to this study

Bowen's concept	Relevance to this study
1. Triangles Bowen identifies triangles as the first concept in his study concerning adequacy and inadequacy in families (Rabstejnek, 2012:4).	In this study, I focused on the triangles, referring to the emotional units presented in families.
Triangles in Bowen's theory also refer to the smallest relationship unit in a family. It is a central theme in this system, as it refers to the smallest, but core relationships in a family (Brown, 1999:3).	The smallest unit refers to the mother's intrapersonal relationship – in other words, her relationship with herself. In a family affected by FXS, it has often been noted that mothers experience feelings of guilt, depression and loss (Caravajal & Aldridge, 2011:16), as they are the carriers of the gene that causes FXS in their children (Bailey et al., 2003:411).

Bowen's concept	Relevance to this study
In a family, the relationship between the mother, the father and the child are a triangle. When there is tension between the mother and the father, the presence of a third person, such as the child, may minimise this tension. Although the tension may now be altered, it still does not relieve the core of the tension between the two parties (Haefner, 2014:3).	It has been reported that a child with FXS can have both positive or negative effects on family relations (see Section 2.5). According to Weber (2016:2), FXS often presents with behavioural problems, which are the main cause of a negative impact on parents' marriages.
Hall states that the triangle concept in Bowen's theory refers furthermore to the interdependence and interaction in families' emotional relationships with one another (Hall, 2013:17).	Julia Beyer (2009:444) states that sibling relationships are typically the longest relationships most people have and also provide experiences that enable emotional understanding, self-regulation and a sense of belonging among individuals. Smith and Hart (2002) add that sibling relationships play a substantial role in the development of children's consideration of other individuals' emotions and thoughts. With regard to this study, Lesniak-Karpiak, Mazzocco and Ross (2003:55) highlight that families affected by FXS have to deal with the emotional and behavioural imbalance of a family member with FXS, which leads to intense relationships within the nuclear family. Bowen's concept on triangles enabled

Bowen's concept	Relevance to this study
	me to gain insight into family relationships – especially the relationship between an individual with FXS and his or her siblings, and the effect FXS may have on the relationship.
2. Differentiation of self Differentiation of the self, another concept identified in Bowen's family systems theory, is linked with emotional fusion, which Brown (1999:95) explains as the "lack of differentiation" in order to create harmony in a system.	Mothers of children with FXS often present with emotional problems and guilt when taking care of their children with FXS (Raspa et al., 2016:845). This aspect allowed me to gain insight into the emotional journey the mother and the whole family go through when a child has FXS.
3. Nuclear family emotional process Titelman (1998:51) states that the nuclear family refers to the individual's immediate family. The term "emotional" is used to denote the family as a unit that deals with changes together and individually. The nuclear family is also interlocked with the extended families, which sprout from both the mother's and the father's side (Titelman, 1998:52).	In this study, I gained insight from experts in the field of FXS and how they, through their expertise, have experienced families affected by FXS. Because FXS is a genetic syndrome, it will have been encountered before in the family, either in a full mutation or through the associated syndromes (Caravajal & Aldridge, 2011:45). Associated syndromes are fragile X-associated tremor/ataxia syndrome (FXTAS) and primary ovarian insufficiency (FXPO1) (Garber et al., 2008:2).

Bowen's concept	Relevance to this study
For Bowen, family is seen as a system, and the doings and relationships between the immediate and extended family have an influence on an individual, as well as the family as a whole (Brown, 1999:3).	As mentioned before, FXS has an impact on marriages, finances and the quality of family life (Weber, 2016:2). This, as well as the fact that FXS is genetic and mothers often experience guilt and loss (Caravajal & Aldridge, 2011:13), may influence not only the nuclear family, but also the extended family.
4. Family projection process Haefner (2014) argues that the family projection process refers to the process in which parents pass on their emotional problems to a child. This involves three steps, namely 1) the parents focus on the child, fearing that something is wrong, 2) the parents interpret the child's behaviour and confirm their fears, and 3) the parents treat the child as if there is something truly wrong with the child. Haefner (2014:836) further argues that these aspects may be associated with maternal instincts and could initiate anxiety and further behavioural problems in the family.	FXS is a syndrome associated with an intense emotional journey, which not only influences the whole family, but also the family members individually (Raspa et al., 2016:847). The challenges faced by families, individuals and especially mothers with children with FXS stretch far beyond those arising from problems within a family. The challenges are often a reflection of the mother's problems, such as social anxiety (Abbeduto et al., 2004:109-110). Mankowski (2007:4) states that the mothers of children with FXS very often have emotional problems, which may be projected to the siblings with or without FXS.
5. Multigenerational transmission processes Although in his theory, Bowen	FXS is a genetic intellectual disability.

Bowen's concept	Relevance to this study
(Rabstejnek, 2012:8) focuses on schizophrenia and neurosis, he mentions that symptoms of emotional disorders may be intensified over generations until someone is diagnosed with one of the syndromes.	Caravajal and Aldridge (2011:18) report that this syndrome can be passed on through generations and show no effect at all and then later be passed on in a full mutation state, which will then be full-mutation FXS. Just as Bowen predicts with schizophrenia and neurosis, FXS can also be passed on without any specific elements of concern, and then finally have someone being diagnosed with FXS full mutation.
However, the focus of multigenerational transmission as a process lies far in-between the emotional demands of the triangle, where the family may use mental impairment, trauma or anxiety to "target" the child emotionally (Rabstejnek, 2012:8).	Apart from feelings of guilt, as has been alluded to before, Abbeduto et al. (2004:239) furthermore note that mothers often suffer from depression, guilt and anxiety before and after the diagnosis of FXS and, in many cases, their children experience the same emotional experiences (Caravajal & Aldridge, 2011:82). Through the experience of people who have worked with the mothers of children with FXS, this research also aimed to understand whether the emotional "baggage" the mother carries, has an actual effect on the child with FXS.
6. Sibling position Sibling position refers literary to the overall position of the child and/or sibling in the family (Haefner,	Hall, Reiss and Burns (2007:28) argue that a child with FXS will have an impact on a sibling without FXS.

Bowen's concept	Relevance to this study
2014:836). It furthermore gives information towards the influence a sibling may have on aspects such as finances, marriage and relationships in the family (Haefner, 2014:836).	Siblings of children with FXS might present with behavioural problems and be affected by maternal stress (Hall et al., 2007:29).
Emotional symptoms, as identified in Bowen's theory, are sometimes a contributor to family dysfunction (Maloni, 1998:147). Furthermore, Maloni (1998) states that sibling position, gender and the anxiety felt at the time of the child's birth contribute to the everyday life of the family. This is seen in whether this child receives special attention and what the effect of the child's position in the family may be on the siblings (Maloni, 1998:147).	According to Okashah, Schoch, Hooper, Shashi and Callanan (2015:2), most siblings affected by a brother or sister with a disability of some sort will be affected emotionally. Families diagnosed with FXS feel much uncertainty after such a diagnosis and this very often takes its toll on the siblings in the family structure (Abbeduto et al., 2004:240). The mothers of children with FXS often struggle with anxiety, depression and stress-related factors, which have an impact on their families, especially those members without the syndrome (Hall et al., 2007:30).
Reed (1998) states that when a child is diagnosed with a medical condition, the family tend to live in a great range of uncertainty and this aspect contributes to major stress in the family. Furthermore, the main caregiver in the family very often struggles with not only emotional problems, but also with physical	Mothers cope differently with their child who has FXS (Abbeduto et al., 2004:241); however, it is argued that the mothers of children with FXS often cope with high levels of stress.

Bowen's concept	Relevance to this study
problems, such as stress and pain. Adaptation in Bowen's theory is the process to which individuals in a family is seen to fit into their environment (Reed, 1998:179). This is how the family survives through difficult encounters. Sibling position is determined in the experiences the family faces and this is done through a contribution of the family's history (Haefner, 2014:8).	Adaptation in a family affected with FXS can be both positive and negative and this differs from each family as each child with FXS are unique (Raspa et al., 2014:42). Beyer (2009:445) reports relationship barriers in between siblings and the child with FXS. FXS is a genetic mental impairment, often combined with other medical conditions related to FXS (Caravajal & Aldridge, 2011:18). A family history, including associated FXS syndromes, as well as sibling relationships with a child with FXS was a focus in this study. Using Bowen's factor of sibling position, I gathered information from the experts with regard to sibling relations in a family affected by FXS.

Bowen's concept	Relevance to this study
7. Emotional cut-off Sholevar and Schwoerie (2003:71) refer to emotional cut-off as the termination of a relationship.	At times, families affected by FXS live in denial and cannot except the fact that there is a genetic component to FXS (Caravajal & Aldridge, 2011:79), especially when the family's hopes and aspirations for their child might not be realised (Caravajal & Aldridge, 2011:80).

Bowen's concept	Relevance to this study
8. Emotional processes in society Society parallels, anxiety and stress in the family regress to a lower level of functioning in the family (Haefner, 2014:836). Emotional processes in the society are furthermore seen as triangles of relationships outside the family domain and the emotional forces that tend to diminish functioning and anxiety (Rabstejnek, 2012:8). From this factor in Bowen's theory, societal effects and opinions may affect families (Hall, 2013:19).	Families coping with FXS often have to deal with all the implications of FXS. At times families choose not to inform other members of the family or members of the community, which make their adaptation process within a community difficult (Caravajal & Aldridge, 2011:83). Of course, a lack of support and knowledge of FXS in a system also contributes to societal stressors. Children with FXS often have emotional outbursts in public due to anxiety or overstimulation, which may put the family in a difficult situation (Caravajal & Aldridge, 2011:37).

Bowen's theory of family systems assessment was ideal for this study because it provided a lens to understand the phenomenon of family life when FXS is part thereof. Not only did it provide insight, but also theory, which may help newly diagnosed families gain an understanding of FXS and find coping mechanisms in order to live a full family life with FXS.

2.6 CONCLUDING REMARKS

This chapter paid attention to the impact of intellectual disabilities on families, with specific reference to FXS. It became clear that families face many challenges associated with a child presenting with intellectual disabilities. FXS was discussed in detail, with the focus on diagnosing the syndrome and the manifestation thereof. It became evident that FXS is a serious mental impairment, which needs more awareness. Various emotional, physical and cognitive aspects related to FXS were discussed, with specific attention awarded to the emotional difficulties the mothers and siblings experience when they have a family member with FXS. FXS in the

family context was also discussed and it became apparent that FXS has a huge impact on a family's everyday life. Additionally, Bowen's family systems theory was discussed and applied to this study. The theory by Bowen is ideal for this study, as it provides the researcher with an understanding of how a family with FXS operates as a system. The eight components of Bowen's theory were used as a guide when experts in the field of FXS were interviewed, as discussed in Chapter 4. The following chapter discusses the research methodology that was followed in this study.

CHAPTER 3

RESEARCH METHODOLOGY

3.1 INTRODUCTION

In the previous chapter, the concept of intellectual disabilities with the emphasis on Fragile X syndrome (FXS) was discussed. The nature and manifestations of FXS were explored and it became apparent that FXS has an impact on family relations. It was also mentioned that diagnosing FXS is challenging due to ignorance about the syndrome. Another aspect that was highlighted, relates to the impact intellectual disabilities, such as FXS, have on how families see themselves, especially in a social and cultural context. As the aim of this study was to explore how FXS affects family relations, I needed to collect data from people who are familiar with FXS. This chapter, therefore, describes the research methodology I followed to gather the data in order to answer the research questions (see Section 1.3.1).

3.2 RESEARCH METHODOLOGY

The research methodology refers to the approaches used to conduct the research. In other words, the research methodology is the process followed to produce research (Cohen, Lawrence & Morrison, 2007:47). The research methodology consists of a research design, research methods, research strategy, data collection methods, data analysis and quality assurance (Maree & Van der Westhuizen, 2013:34-37). The research design is discussed first.

3.2.1 Research design

The research design is a set of plans to present the way the researcher will conduct the research. This is done by identifying what type of data is required and how the questions will be asked (Van Wyk, 2009:2). The research design is a

“blueprint” for the research, dealing with the following four important concepts: what questions should be studied, what data are relevant to the study, what data should be collected and how should the gathered data be interpreted (Yin, 2013:89). In the case of this study, the impact of FXS on family relations was investigated, in other words, how the family copes with such a diagnosis, their lives after the diagnosis and how their initial hopes and aspirations for their child have to be adapted throughout their experiences. For this study, the research design included the research paradigm, research approach and the type of study that was conducted. The research paradigm is the perspective of the study (Johnson & Christensen, 2010:31), as discussed below.

3.2.1.1 Research paradigm

The research paradigm is the perspective about the research held by a community of researchers, based on a set of shared ideas, assumptions, concepts, values and practices (Johnson & Christensen, 2010:31). Nieuwenhuis (2013:47-48) defines the research paradigm as a set of beliefs about fundamental aspects of reality, which yield themselves to a world-view. Furthermore, it is a manner of “as we think, we act” and can also be defined as the lenses or principles through which we interpret our reality (Nieuwenhuis, 2013:48).

This study was situated within an interpretive paradigm. Creswell (2009:176) defines an interpretive paradigm as a form of interpretive inquiry; thus, researchers make their own interpretation of what they see, hear and understand. De Vaus (2001:10), on the other hand, regards interpretivism as a manner of seeing “things” within their context and considering the context and meanings people bring to a situation. This study aimed to understand how family relations are affected when a child with FXS is part of the family. One participant who is not only part of a family with FXS, but also an expert in the field of FXS, was involved in the empirical study, as well as two professionals in the field of FXS. Their experiences working with families affected by FXS provided insight into the daily lives of families affected by FXS. This information provided me with insight into families’ experiences, in terms of their values, beliefs and knowledge, which they shared with the professionals.

I also regarded the interpretive paradigm as suitable for this study, as I made use of interviews in collecting data. The interviews were conducted in the participants' own environment or where they preferred. This is in line with what Chowdhury (2014:435) recommends, namely that one should observe the participants in their social and personal contexts in order to gain an understanding of their situation. In this study, I interviewed experts to gain insight into their experiences working with families affected with FXS. Through this I gained knowledge of not only how they perceived families with FXS, but also what their thoughts were on how the parents behaved before and after the diagnosis of FXS. Furthermore, I gained an understanding of how the parents' preconceived ideas had changed after the FXS diagnosis – not only via the experts, but also via one expert who is also the mother of a child with FXS. This was done by exploring her lived experiences with a child with FXS.

Nieuwenhuis (2007:57) states that the interpretivist perspectives are based on the certain assumptions, as set out in Table 3.1.

Table 3.1: Interpretivist perspectives and their application to this study (adapted from Nieuwenhuis, 2007:57)

Core Belief	Application to study
Human life can be understood from within (Nieuwenhuis, 2013:59)	The literature review allowed me to gain insight into the research that has been conducted on FXS, but I had to hear from people who have personal experience with FXS. I therefore interviewed an expert in the field of FXS, who is also the mother of a child with FXS, as well as other professionals in the field of FXS. This allowed me to understand how they experience life with a child who has FXS.

Core Belief	Application to study
Social life is a distinctively human product (Nieuwenhuis, 2013:59)	This study focused on the family in their social context to obtain a true perspective of their coping mechanisms and understanding of family dynamics. The focus was on their experience as a family with FXS, their social stance and how having a family member with FXS has affected their social and cultural life. Lin (1998:163) states that interpretivists not only look for the presence or absence of relationships, but the ways in which the relationships manifest and the context in which they occur. I gained insight on family relationships through the experience of experts, as they were able to share their experiences of what they have observed from families affected by FXS.
The human mind is the purposive source or origin of meaning (Nieuwenhuis, 2013:60)	By understanding the parents' experiences and opinions of having a child with FXS, I could improve my understanding of the impact thereof on the family. By understanding their perceptions, dreams and aspirations for their child, before and after the diagnosis, I was able to gain knowledge on how these aspects might have changed over time. By interviewing the experts, I gained further insight, as they were able to give information on their experiences with families before and after the FXS diagnosis.

Core Belief	Application to study
Human behaviour is affected by knowledge of the social	FXS is managed daily by different people and families, who, in turn, are affected by each of

Core Belief	Application to study
world (Nieuwenhuis, 2013:60)	the individuals' understanding and experiences of FXS. Thus, one might assume that the manner in which society views a family with FXS may affect the family members' opinion of themselves, as well as their social position. Experts in the field of FXS were able to give more insight on what they have encountered in families, as well as how the parents and siblings behave around the FXS carrier. Jansen (2013:21) postulates that the social context, norms and standards of a person or community are crucial elements to understand human behaviour and that these parallels give understanding (Jansen, 2013:21). My interviews with experts in the field of FXS helped me to gain true insight into and an understanding of families affected by FXS.

Gadamer (1970:87) succinctly remarks, "To understand is always to understand differently." Gaining insight into a family affected by FXS allowed me to develop guidelines, as obtained through the experts in the field of FXS, which would enable newly diagnosed families to gain support and guidance.

3.2.1.2 Qualitative approach

The purpose of this study was to understand the effect an intellectual disability, such as FXS, has on family relations. Furthermore, this study gained knowledge and insight from the views of experts in the field of FXS on their experiences with families who have children with FXS. Qualitative research involves the way the researcher develops an understanding of what is observed and studied. It focuses on individuals' views and understanding of a phenomenon and how meaning is constructed from their experiences (Nieuwenhuis, 2013:50). According to Black (1994:425), qualitative research is necessary because it helps us understand the nature of a perception of events. It asks the "what" questions that enrich our

understanding of a concept (Black, 1994:425). Furthermore, Maykut and Morehouse (1994: viii) regard qualitative research as especially well suited for sociology and the educational field. As I wanted to understand the life worlds of a family who has a child with FXS, a qualitative approach was suitable, as I was focusing on the social aspect of the family. Following a qualitative method, I focused on the characteristics of the qualitative approach.

The following table describes the characteristics of qualitative research and how it is applicable to this study.

Table 3.2: The characteristics of qualitative research in this research study

Characteristics of qualitative research	Application to this study
To explore and understand a phenomenon (Ivankova, Creswell & Clark, 2013:259-260).	The phenomenon in this study is FXS and the aim of the study was to explore the effect this syndrome has on families. This was done by focusing on aspects such as family dynamics, culture and social aspects, as gathered through the views of the experts. It is important to note that one of the experts in this study is also the mother of a child with FXS. Through her view, as a professional in FXS, I gained a mother's personal experience.

Characteristics of qualitative research	Application to this study
Qualitative methods are used to seek a deeper truth (Greenhalgh & Taylor, 1997:2).	By making use of interviews and interpreting the data, I was able to make sense of the FXS

Characteristics of qualitative research	Application to this study
	phenomenon by gaining a deeper perspective of family relations in terms of FXS.
The researcher as key instrument: The qualitative researcher collects data through documents, literature and interviews (Creswell, 2007:38).	I realised the important role I played in the research, as I was the person who collected the data. During the research process, I had to be aware of any biases. For example, in the case of the one expert who is the mother of a child with FXS, I had to ensure that I did not ask questions about her child, but rather focused on questions that could give me an understanding of what it means to be a parent of a child with FXS.
Qualitative studies often use more than one data collection method, such as observations, documents and interviews (Creswell & Poth, 2017).	In this study, I made use of interviews and a literature review to collect the data. The views of experts in the field of FXS also provided better insight into the perspective of FXS.

Characteristics of qualitative research	Application to this study
Purposive sampling: Maree and Pietersen (2013:178) state that purposive sampling is used in	I wanted to investigate a specific phenomenon, namely FXS;

Characteristics of qualitative research	Application to this study
special occasions where the researcher wants to focus on something specific (Maree & Pietersen, 2013:178).	therefore, purposive sampling was the appropriate means of selecting the participants, whom I chose based on their expertise in the field of FXS.
Inductive data analysis: Maree and Van der Westhuizen (2013:37) state that inductive analysis gives more realities presented in the data.	I followed an inductive approach in analysing the data, as accounted for later in this chapter.

A qualitative approach was thus the most appropriate approach to use, as it enabled me to gather rich, in-depth information on the experiences of a family with FXS. I also made use of case study research, which, according to Rule and John (2011:3), focuses on a specific problem or circumstance. Making use of a case study allowed me to gain insight into the lives of families affected by FXS

3.2.1.3 Research type: case study

Nieuwenhuis (2013:75) states that the term “case study” has multiple meanings. Zucker (2009:1) agrees and asserts that it can be a systematic investigation into either an event, or into a set of related events, aiming to describe a phenomenon of interest. Furthermore, Nieuwenhuis (2013:75) argues that a case study allows the researcher to discover multiple meanings about a phenomenon. A case study allows the researcher to gain insightful information to support the conclusion of the study (Rule & John, 2011:3). Moreover, case study research usually involves a sensitive matter (Struwig & Stead, 2001:8). A case study was, therefore, ideal for this study, with my research topic being the effects of FXS on a family with the emphasis on family relations, sibling relationships, preconceived ideas, finances, social and cultural aspects through the experiences of professionals in FXS.

By means of the knowledge, experience and expertise of experts in the field of FXS, I gained further insight on FXS. As this study is situated within an

interpretivist paradigm, I strove to make meaning of how families see themselves when they are affected by FXS, as perceived through the expertise of the professionals. I also gained a personal view from one of the experts being the mother of a child with FXS. Hence, the aim of using the case study as a research type was to provide a comprehensive understanding of the effect of FXS on a family, with the further help of experts in the field of FXS.

3.3 RESEARCH METHODS

Research methods involve the forms of data collection, analysis and interpretation the researcher proposes for his or her studies (Creswell, 2009:15 & 87). Additionally, research methods involve the role of the researcher as well as the specifics of the research site and the participants. These aspects are discussed in the following section as they applied to this study.

3.3.1 The role of the researcher

Maree (2013:41) states that the role of the researcher should empower the researcher to gather the data from the participants. From an interpretive perspective, the researcher plays a prominent role in the data-gathering process (Maree, 2013:298). As the researcher, my role was mainly to interview the experts who participated in this study. As a qualitative researcher, I realised the importance of having a good relationship with the professionals. Rule and John (2011:35-36) state that knowing one's position as a researcher is of the utmost importance in a qualitative study; therefore, I was objective towards the participants at all times and did not allow my own assumptions and opinions to guide them.

FXS is an intellectual disability and families that are affected by these abilities are often very sensitive about it. Furthermore, the professionals who work with these families and share their insights had to be considered in an ethical manner (Griffin & Balandin, 2004:65). Thus, in my position as a qualitative researcher, I was aware of being sensitive and objective and I conducted the interviews in an honourable manner, as advised by Blancher and Mink (2004:65). In doing so, I posed my questions in an ethical and considerable manner, so that no

professional was guided into giving private information about any family (Griffin & Balandin, 2004:137).

3.3.2 Participants and research site

As mentioned before, I used purposive sampling in this study. In such cases, Rule and John (2004:64) state that the researcher must include people who can shed more light on a situation (Rule & John, 2011:64). Hence, the people selected for this study were chosen deliberately, since they had worked with families affected by FXS. Furthermore, the experts who were chosen for this study are well-known specialists in the field of FXS, who have worked with families affected by FXS.

I made use of three experts in the field of FXS. One of these experts was also the mother of a child with FXS. In this regard, Sergeant (2012:1) mentions that the sample size is usually small when qualitative research is conducted. The reasoning behind using these participants was twofold. Firstly, it was a struggle to obtain participants, as FXS is not well known or well supported in South Africa. Secondly, I had to consider easy access to the participants. Lastly, using experts allowed me to obtain precise and to-the-point data.

Creswell (2009:178) refers to the participants as actors. My participants also had a specific role to play in the study. Finding families that are affected by FXS was problematic, as few families were available due to little knowledge about the syndrome (Carvajal & Aldridge, 2011:18). I made use of three experts in the field of FXS. These experts I selected, had to meet specific criteria, such as that they should have had considerable experience in working with families where a child had FXS and, therefore, have extensive knowledge of the effects of FXS on families. The criteria are discussed in detail in Section 3.3.1.3. Creswell (2009:178) suggests four aspects to consider when qualitative research is conducted, namely the setting, events, actors and process.

3.3.2.1 The participants/actors

Those persons who are interviewed, are the actors in the research study (Creswell, 2009:178). The participants in this study chose to stay anonymous, although they are specialists in the field of FXS. One of them is a paediatrician, the

other a highly educated educational specialist in the field of FXS and the third a founder of the first organisation for FXS in South Africa and mother of a child with FXS. All the specialists who participated in this study had experience and valuable knowledge of FXS.

3.3.2.2 Criteria for research participants

The selection of the participants in a qualitative study is purposeful (Sargeant, 2012:1). Hence, the participants were chosen to give an understanding of the phenomenon under study. The participants in this study were thus carefully selected as the outcome of the study was dependent on their experiences with families affected by FXS.

3.3.2.3 Language

The experts who participated in this study had to be fully bilingual in both Afrikaans and English to make the communication between the researcher and the participants clear and understandable. Creswell (2013:8) states that the researcher gains valuable insight into the study from the participants.

3.3.2.4 Willingness to share information

The participants in this study shared not only their experiences, but also valuable knowledge of FXS. However, no participants were required to give personal information of their clients or patients. The information that was valuable for this study was only the information with regard to the experiences and unique circumstances families affected by FXS face daily. I asked one of the participants to write a narrative to explain her experiences, as well as advice to families affected by FXS. This narrative was written in general about the circumstances families with FXS face and not only about her own circumstances, having a child with FXS.

3.3.2.5 Research site

Creswell (2009:178) explains that the setting of the study is the actual place where the research takes place. The interviews took place through email, as it was requested by all three participants due to time constraints. The events are the

participants' doings and the process refer to the nature of events undertaken by the actor within the setting (Creswell, 2009:178). The participants were asked a set of questions about FXS (see Appendix 1).

3.3 DATA COLLECTION

Maxwell (2013:24) states that data collection is the manner in which the researcher aims to collect the data that will be used in the study. I used the following data collection instruments: interviews, narratives and general notes I made throughout the study. The data collection methods, as they apply to this study, are discussed next.

3.3.1 Interviews

Interviews in case studies are regarded as conversations between the researcher and the research participants about the phenomena of interest in the study (Rule & John, 2011:64). I conducted the interviews in my study, as it allowed me easy access to the participants on their own terms. Interviewing the participants allowed me to gain a deeper understanding and insight into the responses given.

I followed a structured interview approach. Structured interviews are seen as pre-set questions, which are direct (Ashenqeeti, 2014:40). Furthermore, Rule and John (2011:64) state that structured interviews are a set of questions that guides the interview as an interview schedule and the interviewer does not deviate from the set of questions provided to the participants. As the interviews were conducted via email, I had easy access to them, as the data/interview conversations were saved in my emails. According to Meho (2006:1285), email interviews are rapidly increasing in qualitative research as it provides access to more participants. Therefore, due to all three participants' busy schedules, as well as geographical aspects, I chose to conduct this study via email. Each interview was recorded, as this allowed me to go back to information during the data interpretation process.

3.3.2 Narrative

Nieuwenhuis (2013:102) states that narratives are often used to tell the "story" of one's personal experience. In this study, the one participant and founder of the

FXS Foundation in South Africa was asked to write a narrative about her experiences as the mother of a child with FXS. This was not done directly from her own experiences, but rather to gain a general insight on what it is like to have a child with FXS. The following guidelines were considered in the writing of the narrative (Nieuwenhuis, 2013:102):

- The impact of FXS on the families. The aspirations the parents had for their children before and after the diagnosis.
- The journey most parents follow from the diagnosis onward and their coping with the diagnosis.
- The impact of social, emotional and cultural beliefs on the diagnosis.
- The support available for families that are affected by FXS. Another aspect the participant wrote about was how little knowledge about and support for FXS in South Africa are available and what, in her opinion, was needed.
- A general view about FXS through the eyes of a mother.

Narratives must be informal and nonthreatening (Rule & John, 2011:65). In this study, no names of particular families were mentioned. This allowed me to gain an insight on the day-by-day existence of a mother, through her description of her journey with her child who has FXS. Through the use of her narrative, I was able to gather a personal but general view of what might be easier to write than to say out loud about the intellectual disability, FXS.

3.4 DATA ANALYSIS

Data analysis is the process in which the data gathered are interpreted and examined (Creswell, 2009:14). In this study, the data analysis strategy was inductive data analysis, which Schwandt (2011:1) defines as the analysis of specific data in a specific case. Inductive analysis is used when a study is conducted in the participants' natural context (Maree & Pietersen, 2013:37), such as was the case with this study.

This study was situated within the interpretivist paradigm. Therefore, an inductive data analysis approach was best suited for this study, as Maree and Pietersen (2013:37) maintain that the researcher is more likely to gain more than one

perspective of the phenomenon through the use of inductive data analysis. Nieuwenhuis (2013:107) defines inductive data analysis as the coding of data into categories to allow the researcher to examine the data directly.

In order to allow for the breakdown of the data, I followed the advice of Creswell (2009:183), who states that analysing data involves making sense out of the text and the recorded data by moving deeper and deeper until the researcher is able to interpret the data. The inductive data analysis allowed me to invent codes from the data in order to make the process of data analysis, transcribing and saving the data easier. The following table represents the way in which the data were analysed according to nine steps.

Table 3.3: The steps to be followed in order to inductively analyse and interpret the data (Nieuwenhuis, 2007:103)

Steps	Application in study
1. Describing the data and participants When using a quantitative research methodology, the focus of the data will remove any influences of external variables. However, as this was a qualitative study, the focus was on participants who could best deliver valuable and accurate data to contribute to the study.	Finding families who were affected by FXS proved to be difficult. I, therefore, focused on experts in the field of FXS. In this study, I used three participants.

Steps	Application in study
2. Organising interview data	The data gathered from the interviews were carefully organised according to the following aspects: • The roles of the participants as they were interviewed • Exact quotes given by the

Steps	Application in study
	participants These were possible breakdowns and organising it this way allowed me, as the researcher, to follow my thoughts and explain them to the reader in a simpler, clearer way. Furthermore, it would be easier accessible at a later stage.
3. Transcribing the data after I have carefully worked through it	This was the first step in analysing the captured data. The researcher transcribed only the data that were important to the study. Great care was given to details within the data.
4. Familiarising myself with the data I gathered	The researcher read through all the interview data and the narrative.
5. Saving the data	All the data were typed and saved. Thereafter, I emailed a copy of the data to myself and to my supervisor as backup. In this day and age with the current technology, it is easy to only have a copy of one's research stored on one's notebook or laptop.

Steps	Application in study
6. Coding the data according to meaningful units By doing this, I made the data easier to access when discussing the outcomes.	By doing this, I made the data easier to access when discussing the outcomes. In order to make the data easier to understand and to allow for interpretation, the data were coded into units that made sense and would allow any reader to follow.

Steps	Application in study
7. Identifying categories and themes	The data were analysed inductively by allocating codes and themes to the data, which made the data accessible to both the researcher and the reader.
8. Structuring and analysing the data	The data were structured in a way that flows logically and makes sense to the reader. This structure was used as a foundation for the analysis of the data.
9. Interpreting the data	The data were interpreted and analysed to answer the initial research question.

Once all the data had been structured and analysed, the interpretation of the data could be conducted. However, for me to provide clear and concise conclusions, I had to pay attention to whether the data I had gathered were trustworthy. This aspect is discussed in the following section.

3.5 TRUSTWORTHINESS

Creswell (2013:256) states that trustworthiness refers to the believability and truthfulness of a study. The elements related to trustworthiness are discussed as they apply to this study in terms of credibility, dependability, transferability and conformability.

3.5.1 Credibility

Shenton (2004:3) states that the term “credibility” asks the question, “How congruent are the findings with reality?” Moreover, it deals with the fact whether a situation is more likely true, given the existing state of knowledge of that situation (Smith, 2011:3). Westhof (2014) suggests that credibility implies the retrieved data are not biased.

In terms of this study, the aim was to give a true account of whether the knowledge gained from the experts on FXS was a true account of the experience of having a child with FXS and the effects thereof on the family. Having kept this in

mind, I gave credibility to this study by the following aspects, as identified by Morrow (2005:252):

- Making use of various data collection methods, namely interviews and a narrative.
- Being open to the participants with regard to my findings. I supplied them with my findings to verify and to ensure that these are a true reflection of the information they had shared.
- I also made sure I engaged with my participants on a regular basis and I was persistent in seeking the knowledge I needed for this study.

3.5.2 Dependability

Morrow (2005:252) advises that the way in which a study is conducted, must be consistent. Shenton (2004:71) also states that the process followed within the study should be reported in detail, for future research to be conducted from the study. I strove to describe the measures and strategies that can be undertaken to conduct the same study under similar circumstances and in similar contexts. I endeavoured to discuss how the research design and research questions were formulated within clear and explicit theoretical and philosophical traditions.

3.5.3 Transferability

Morrow (2005:252) states that transferability refers to the extent to which the findings of a study can be generalised. It also refers to how the data can be used to make comparisons about the phenomenon (Shenton, 2004:73). I believe that through the literature I read and presented in both Chapter 1 and 2, along with presenting the data I gathered from experts in the field of FXS, the findings of this study are proven to be valid and transferable to other families affected by an intellectual disability.

3.5.4 Conformability

Conformability refers to the validity of one's research. This concerns the interpretation of what the researcher has observed throughout the study and whether it was done objectively, and the application thereof is sensible and reliable

(Perakyla, 2016:412). In order to maintain conformability in the study, I conducted an in-depth investigation into the literature on FXS and the effect this study might have in future studies. These steps are confirmed by Westhof (2016), who refers to Ary et al. (2010) when they stated that conformability suggests that the findings are supported by similar studies. Chapter 2 attests to the relevant literature that has been consulted.

3.6 ETHICAL CONSIDERATIONS

With any study, ethical considerations are of the utmost importance to make sure that no harm is done to any individual, institution or the researcher (Morrow, 2008:4,12). Ethical considerations are concerned with the moral stance of the research being conducted (Payne & Payne, 2011). These authors, furthermore, explain that the research is conducted not only to achieve high professional standards, but also to show respect and protection towards those who gave their consent to be involved in the study (Payne & Payne, 2011:1). In social research, where people are involved, ethical applications are even more important (Neale & Bishop, 2012:2). Hence, it is the duty of the researcher to take care of the welfare of the participants by being ethical, concise, clear and to treat the information and participants with confidentiality (Neale & Bishop, 2012:2).

For this study, I submitted an ethical application to the Ethics Committee of the Faculty of Education at the University of Pretoria (EC 17/03/02). I also considered the Code of Ethics (Rt 429/99) of the University of Pretoria, which places emphasis on social justice and the protection of the families participating in a research study.

The ethical principles adhered to in this research are discussed below.

3.6.1 Informed consent

Through letters of informed consent, I requested the experts in the field of FXS to participate in the study. They were assured of the protection of their identities by the use of pseudonyms. It was emphasised that they could withdraw from the study at any time. They gave me permission to ask them questions about their patients and clients. However, no questions were asked concerning names or

specific cases. None of the findings, if too personal, was shared unless the participants were comfortable with it.

Greenstein (2011:3) mentions that two factors are important when conducting family and social research, namely that the knowledge gained should be valuable to the research field and taking into account that the participants are human beings with their own lives and homes. I made sure that the data I collected were relevant to the research field, as very little South African research exists on families with children who have FXS. The data collection took place in a place where the participants felt comfortable and, in the way,, they chose to conduct the interviews.

3.6.2 No harm

Creswell (2009:91) makes it clear that researchers need to take into consideration that some information might be harmful to participators. Therefore, due to the sensitivity of this topic, I was very aware of the fact that I had to be sensitive while conducting the interviews, but I realised that through my indirect involvement with FXS, there was no possibility of any heinous discovery. The third participant, who is the mother of a child with FXS, is a resilient lady, as is evident in her also being the head of Fragile X South Africa. Furthermore, as I have mentioned, the interview questions were made available to the participants beforehand, so that they could decide whether they wanted to answer the questions or not.

3.6.3 Confidentiality

Orb et al. (2001:93) state that researchers doing qualitative research should not only promise confidentiality but must also adhere to their promise. One of the participants, who is the main caregiver of a child with FXS and who has worked with several families affected by FXS, was asked to write a narrative about her personal encounters of families with children with FXS. The experts gave their opinions, knowledge and experiences of having worked closely with families affected by FXS. Therefore, each of the participants was assured of anonymity and pseudonyms were used when they were referred to. The experts were not

asked to share any patient information, as it was not required for this study and not ethical to do so.

Further ethical considerations involve the safekeeping of the data. I emailed a copy of my research to my supervisor and kept a copy on a Google Drive account. This was to ensure multiple backups. The Google Drive solution, as well as the email solution, is fully ISO 27001 certified and SOC 2 and SOC 3 type II audited to ensure the stored data were safe and would not be compromised. Furthermore, the Faculty of Education of the University of Pretoria will also keep all the data for the next 15 years.

3.7 CONCLUDING REMARKS

In this chapter, the methodology I followed in conducting the empirical research on the impact of a child with Fragile X syndrome on family relations was described. I discussed the approach and paradigm of the study and gave detailed descriptions of the methods I used in this study. This research was done in the form of a case study with experts who have knowledge and experience in the field of FXS. The importance of trustworthiness and ethical considerations was also referred to. The next chapter discusses the data analysis and interpretation.

CHAPTER 4

DATA ANALYSIS AND INTERPRETATION

4.1 INTRODUCTION

Chapter 3 discussed the research methodology followed in the study. The study is situated within an interpretive paradigm and followed a qualitative approach. The case study consists of three experts in the field of Fragile X syndrome (FXS). Each participant was purposefully selected by means of their expertise with regard to FXS. Structured interviews were used to gather data, as it allowed me to gain insight into the phenomenon of FXS and the impact of this syndrome on family relations. This data collection method also allowed for further questioning on the initial responses. The interviews were conducted through a set of eight questions put to all three the experts in the field of FXS. One of the participants, who is also the mother of a child with FXS, was asked to write a narrative, documenting her experience as a mother in a family with a child with FXS.

The purpose of this chapter is to describe the case first, followed by an analysis of the data gathered through the interviews and the narrative. The chapter concludes by interpreting the data by means of the themes and categories that emerged through the data analysis process.

4.2 DESCRIPTION OF THE CASE

Three professionals who had been working with individuals and families affected by FXS were chosen for this study. Initially, I wanted to use families as cases, but because of the underdiagnosed status of FXS, it was difficult to find families affected by FXS and moreover, families that would have been willing to participate in this study. After much consideration, I decided that a case study based on the experience of experts who have worked with families affected by FXS would also

provide insight of how this syndrome affects families. A more objective opinion of just how FXS affects families would also be a possibility, which would have arisen if I only worked with people who have expert opinions on FXS. All three the participants chose to stay anonymous in the study and pseudonyms were used to respect their privacy.

4.2.1 Biographical data

The biographical data below give a perspective of the professional participants selected for this study.

Table 4.1: Biographical data of professionals whom participated in this study

Participant	Gender	Pseu-do-nyms	Field of expertise	Years experience	Language used for communica-tion
Participant A	Female	Ann	Paediatrics & Child Health (Johannes-burg Hospital)	Approximately 35 years	English
Participant B	Female	Mia	Researcher (PhD) *Resilience in FXS*	10 years	English
Participant C	Female	Kelly	Founder of FXS Organisa-tion South Africa and mother of a son with FXS	17 years	English

Table 4.1 indicates the participants in the study, through whom the information was gathered with regard to FXS and the effect thereof in a family context. Only three participants were used, along with the literature from previous research into FXS.

Purposive sampling was used in this research. The participants were selected based on their extensive knowledge and experience, working with families affected by FXS. Face-to-face interviews were originally chosen as the data-gathering technique. However, due to the participants' busy schedules and the fact that Participant C lives in Cape Town, they chose to conduct the interviews through emails. The interviews were conducted in English, as it was the language they were comfortable with to use.

4.2.2 Participant 1: Ann

Ann is an associate professor at a renowned university in South Africa. In 1986, she opened a clinic for disadvantaged children with visual impairments and autism. Her interest in helping children with visual impairments started when she helped a child who was blind and realised there was no facility to deal with these children at that time – *"I helped treat a blind child over 26 years ago and it struck me that there was no facility to deal with these children as they grow*." At that time there were no longitudinal assistance available to children with special needs – "*We also realised that we needed to work with the child from diagnosis onwards and to assist their parents and caregivers from when they are very young until their school years*." She also mentioned that she saw more and more children who were diagnosed with autism and then started to include these children in the programme at the centre. In 1986 she opened the so-called "Teddy Bear Clinic" for children with special needs, which *"offers a range of specialists and caregivers who help parents of children with special needs".*

As a specialist in children's health, she gained a lot of experience in autism and autistic-associated syndromes, such as FXS. Professor Ann was chosen due to her 30 plus years of experience in children's health and autism. I contacted her and she was willing to conduct an interview.

4.2.3 Participant 2: Mia

Mia is an educational psychologist and specialist in the research field of FXS. She had worked with families in the United States of America and South Africa. She received her doctoral degree in 2016, specialising in resilience in families affected by FXS. Moreover, she had spent time with families affected by FXS in South Africa. She helped spread awareness of FXS through her study and by working with families affected by FXS. In 2017 she was asked to give her views on FXS in a local magazine, as part of an awareness article on FXS. Her experience and values towards FXS and those affected thereby were a key reason for her being selected to partake in this study. When asked why her interest was specifically in FXS, she mentioned that while working for a family affected by FXS, she came to realise how little information was available on this syndrome. It also bothered her that so many people may be affected by FXS without knowing so. Her experience in working with families affected by FXS, as well as her excellent knowledge in the research field of FXS served as the reasons for her being selected as a participant.

4.2.4 Participant 3: Kelly

Kelly is a remarkable woman, who not only dealt with her personal experience of FXS, but also made the time and effort to help others affected by FXS. Her son was born with FXS and diagnosed at the age of four and a half years. He is currently 17 years old and, as she stated, *"still learn[s] new things every day*". After the diagnosis of FXS, Kelly soon realised that there is little knowledge and information about the syndrome and, therefore, a lack of support in South Africa. As most families, this family also received genetic counselling and started their long journey of family adaptation and gaining knowledge of the syndrome. They also had to inform their extended family about the diagnosis, which was rather a battle, as some family members failed either to accept her son, or to accept the fact that they might be affected by FXS as well. Kelly mentioned, for instance, that her father did not accept the diagnosis, claiming that it cannot come from his side of the family. He rejected her son and had very little contact with him. Ironically, after testing the FMR1 gene, it was traced back into her father's side of the family. Other family members also chose to ignore the facts, while others accepted that

they too might be affected and started their process of testing and, of course, supporting Kelly and one another. Kelly later created a Facebook page and website to find other families affected by FXS to share their stories and support to one another. Her valuable insights as the mother of a boy with FXS, as well as her narrative of her emotions and her family's journey, were of the utmost importance to this study.

The following part of this chapter discusses the data gathered from the participants.

4.3 DATA ANALYSIS

Qualitative data analysis is usually done to gain a broad perspective of the participants' attitudes, perceptions, knowledge and understanding of a phenomenon (Nieuwenhuis, 2013:99). Moreover, to construct an understanding of the phenomenon, the data are usually analysed through inductive analysis (Nieuwenhuis, 2013:99).

A structured interview schedule was followed, which allowed for data emerging from other data (Nieuwenhuis, 2013:87). All three the participants chose to conduct the interviews through email due to their busy schedules. According to Meho (2006:1284), interviews through email help with cost efficiency, as well as considering the interviewees in terms of their schedules and privacy. The interviewees were emailed a questionnaire, consisting of a set of eight questions, which was to be completed in the same format. Through the structured interviews, I gained a descriptive view of each participant's perspective of FXS in families. I asked Participant 3 to include a narrative of no longer than 100 words, which allowed me to gain insight into her family's own experience, as she had a child with FXS.

The data collected through the structured interviews were analysed first, followed by the narrative data.

4.3.1 Interview data

The interview data are presented by the various questions posed to participants.

QUESTION 1:
What are the challenges that families with children with FXS usually experience?

With this introductory question, I wanted to obtain a bird's eye view on the challenges the family of a child with FXS experiences. With this question, I wanted to explore the various challenges that accompany FXS. The participants referred to the following challenges: finances, feelings of guilt, social life, sibling relationships, as well as developmental and physical challenges.

As an introductory part in this section, Mia stated, "*I have met and worked with families in the USA as well as South Africa, and a big challenge is the financial aspect.*" Ann agreed when she stated that if there is a lack of finances, this may also affect the family. She elaborated, saying that the mothers often implement therapeutic interventions, which can be very costly. Children with FXS also present with medical problems, which may contribute to high costs. Kelly did not specifically mention anything about finances in her experience, but argued that there are extra medical costs for therapy, schooling and caretakers. She also mentioned that government grants and medical support from the government are available to help if a family does not have any medical aid.

Mia, on the other hand, describes the families affected by FXS as "*fantastic people*", as, in her experience working with these families, they are resilient. However, some days may be experienced as tough. She pays specific attention to tantrums, focusing on the behavioural problems these children present with. Yet, Mia also mentions that in many cases, they have very good days as well. Just as other parents, the parents of children with FXS also want the best for their children. These families tend support and teach others about FXS and try to create social groups, as Kelly did.

Families affected by FXS are also affected by socialising with other or "normal" families. This aspect is addressed in some of the other questions. Another challenge mentioned, relates to social challenges – in a social environment, families affected by FXS are often seen as being "different". Kelly referred to the peculiar physical manifestation of FXS, such as poor eye contact, hand-flapping and rocking, as well as obsessions, anger outbursts and separation anxiety. Society finds these manifestations strange and tends to avoid families with children with FXS. Mia referred to the adaptations these families need to make –

> *These families also can't really do what other families with unaffected children do. All activities have to be altered to meet the needs of the individual diagnosed with FXS. A child with FXS experiences great anxiety when it comes to social activities, and also for the unknown. So, when these families plan something, they have to prepare the child way in advance and it should preferably be an activity that the child is comfortable with, one he or she has experienced before. So, they know what to expect.*

Kelly spoke from personal experience when she mentioned that separation anxiety was yet another aspect that makes a family's social life challenging. Many children with FXS battle with separation anxiety due to the fear of not being with his or her mother or the family he or she knows. This often leads to tantrums at social gatherings, in shops and even if the mother just needs to go to the restroom – "*For about three years, I could not step out of Jadin's eye sight. He was figuratively 'glued to my hip'. I felt trapped and needed freedom of my own.*"

Certain aspects, such as socialisation, are very difficult for families. Mia emphasised this aspect when she mentioned that "*all activities have to be altered to meet the needs of the individual diagnosed child with FXS*". This is often due to anxiety experiences by the child with FXS. If and when the families plan a social activity, they have to prepare the child in advance. The activity should also be something which the child with FXS is comfortable with. Ann mentioned that a child with FXS can sometimes disrupt the family and this may also take place in a social context. Another aspect Ann mentioned, was that the mothers wanted acceptance. However, to contribute to this, I am of the opinion that the mothers crave acceptance, not only for themselves and their families in general, but also for their children.

Kelly, as the mother of a child with FXS, had much to say about the challenges that accompany having a child with FXS. She referred to the developmental challenges as follows:

> *…unlike a child with a physical disability there is no need to alter your home or purchase wheelchairs, etc. With a healthy FXS child, your only medical expenses are seeing specialists to get the initial diagnosis and cost of necessary medication and therapies e.g.: speech, occupation and maybe physical therapies. For some there is an additional cost of nappies for many years and possibly the cost of adult nappies.*
>
> *Speech is the main challenge of Fragile X and this creates many challenges for the family. A Fragile X child gets frustrated because they cannot communicate with others and thus reacts with tantrums and meltdowns. There is frustration for the family members as they cannot adequately communicate with the Fragile X child. Speech therapy and alternative forms of communication should be introduced as early as possible. Echolalia / palilalia is a speech behaviour that is common in Fragile X people.*

Kelly also mentioned how anxious her son would become –

> *To help cope with the anxieties that they face; the child will constantly repeat a question or a phrase. For Jadin, he would ask repeatedly where we were going even though he knew and could verbalise the answer. In a two-kilometre journey to Pick 'n Pay, he could ask up to a hundred times: 'Where we go?'*

She mentioned that potty-training can be very challenging –

> *This is dependent on the level of mental development delays and physical development in each child. For some it takes years to toilet-train and for some it follows the normal chronological developments. For us it took 7 years and for someone whom I have met, up to 12 years before nappies could be eliminated. I have only met one child who was toilet-independent by the age of 5 years.*

The biggest stressor for parents seems to be their concern about their child's future, including his or her education and care. Kelly referred to her personal experience, saying that parents are often concerned with two things – education or schooling and who will be taking care of their child if the parents pass away. She emphasised the lack of support: "*[C]are centres provided by the government are*

almost non-existent. Private centres are very costly." She verbalised a parent's greatest concern: "*Knowing that your Fragile X child, when an adult, will not be able to work and live completely independently.*"

This concern was echoed by the other two participants as well, when they stated that through their experience, most parents are concerned of what might happen if they are no longer there to take care of their child with FXS. Mia strongly emphasised this point: *"Definitely what will happen to their child or children when they are no longer there to care.*" She added the following:

> *Parents often feel guilt with regards to the fact that often siblings would have to care for their brother or sister diagnosed with FXS, when the parents are no longer there to do so. They might feel that it would be a burden for their siblings to care of them for the rest of their lives, as some of the children diagnosed with FXS would need assisted living throughout their lives.*

Based on her experience, Mia postulated that most parents are also concerned with the financial aspect of taking care of their child with FXS throughout his or her life. Ann, however, stated that it also depends on the time of the parents' life, adding the concern, in retrospect, of who will in the end take care of their child.

QUESTION 2:
In your opinion, what is the impact on family relations when one of the children have been diagnosed with FXS?

As this study focused on family relations, my second question probed the various relations of the families of children with FXS. Ann emphasised that a child with a disability of any kind imposes stress on a family. Mia mentioned the first relationship that is affected by FXS – the intrapersonal relationship of the mother – "*A genetic problem such as FXS comes with its own issues because there is the burden of guilt and blame.*" Mia agreed and explained that mothers who are full-mutation carriers blame themselves, which can also lead to depression. Kelly also mentioned that the mother experiences a lot of guilt, as she is the source of her son's condition, as with the father when he passes on his premutation X chromosome to all his daughters

According to Ann, FXS also presents with quite a few behavioural challenges and because of the fact that these children can be difficult, more stress is put onto families. In my opinion, Ann gave a more medical perspective on what parents might experience if they have a child with FXS.

Mia, on the other hand, not only gave a more emotional perspective of the phenomenon, having lived with families affected by FXS. Mia mentioned that FXS may impose stress on marital matters and referred to an increase in the divorce rate. She explained that being the cause of FXS may have an effect on the marriage when the mother blames herself for bringing the syndrome into the family, sometimes even feeling that her husband blames her, even though this might not be the case at all. Kelly illustrated the challenges in the marital relationship:

> *My husband, Matthew, could not cope with the problems we were facing and distanced himself from the family by working long hours, partying with colleagues and friends and finding many excuses to not be at home on weekends…went on for over seven years… he now regrets the time he lost to bond with our children. In many cases, the father will leave and divorce his wife. I was grateful that Matthew 'came back to us' in time.*

Having a child with FXS may also create behavioural problems in siblings. Kelly mentioned that her relationship with her daughter was rather difficult at some stages. The mothers deal with guilt and blame, as well as depression in many cases, as stated by Mia. Kelly mentioned, "*As a mother, you expect your child to be normal and when they are not you face many mixed emotions.*" As a mother, Kelly affirmed that the mothers deal with many emotions they need to address –

> *The emotions I faced were different to most mothers (as I've learnt through the years), I was upset that I couldn't adequately care for him, I was nervous that I would fail him, and I feared for his future as a special needs child in South Africa*

Throughout her answers, it became apparent that Kelly made her peace with the situation and accepted what she cannot change, enjoying her son and loving him for who he is –

> *Jadin is a child, my child, despite his differences and challenges. I was his mother and I loved him no matter what. I come from a family where special needs are common and not an embarrassment. I have a lot of compassion for people with challenges, so the idea did not scare or embarrass me. I was told to place Jadin in a facility to allow for our family unit to function "normally" but I would not even consider this option.*

Mia, who has spent time with numerous families affected by FXS, mentioned that the mothers often interact differently with their children with FXS, which may result in feelings of guilt towards the siblings who are affected by FXS. Mia gave a very good description of how FXS in a family may affect the siblings. She argued as follows:

> *When there are siblings that are not diagnosed with FXS, the parents also feel guilt with regards to the fact that these siblings would have to care for their brother or sister diagnosed with FXS, when the parents are no longer there to do so. They might feel that it would be a burden for their siblings to care for them for the rest of their lives, as some of the children diagnosed with FXS would need assisted living throughout their lives. The siblings might feel frustrated as a lot of the attention goes to the affected sibling. But it could also be the other way around. And then the siblings might get frustrated with not being able to do what other families with unaffected children do. So, this again puts a lot of strain on the parents, trying to juggle between the affected child and his or her siblings.*

Ann stated that there are factors that might have an impact on the siblings of a child with FXS. These factors include neglect due to the care the disabled child requires, rejection by peers, disruptions for the family, the acceptance of an abnormal sibling and a lack of finances for the child with FXS.

In her own experience as the mother of a child with FXS, Kelly gave a description of a real-life event that occurred. She said that her son with FXS influenced her relationship with her daughter. Her daughter did not have FXS and she was tested to see whether she is also a carrier like her mother. Kelly then had to explain the concept of FXS to her five-and-a-half-year-old daughter, who then replied, "*So is that why you don't spend any time with me!*" For Kelly, the founder of FXS South Africa and, more importantly, a mother, this was a "*hard blow*". As mentioned at Question 1, Mia states that in most cases, relationships, such as marital and sibling relationships, are affected due to FXS. Kelly states that another aspect that

affects the family is that a change in routine for a child with FXS could lead to a meltdown.

QUESTION 3:
In your opinion, what are the needs of parents who have a child with FXS?

Mia responded from her experiences working with these families. The first thing she referred to was the need of these families to belong. They need support groups and the mothers especially need to connect with other mothers who also have children with FXS – "*They also need social groups where their children can connect with other children that are on the same level as them, or higher*." Mia also mentioned that children with FXS learn from their peers. Thus, connecting on a social level with others with the same circumstances is important. Ann stated that families affected by FXS need emotional support, counselling, affordable medical care, affordable therapy and appropriate education. Referring to Ann's statement with regard to the need of affordable medical care and therapy, Mia indicated that for parents, it is severely frustrating when they consult specialists, doctors and therapists, and these professionals have not heard of FXS yet. This means that the parents must educate the professionals, yet the professionals do not have the skills to support these families anyway.

Kelly, as a mother and through her interaction with other parents, mentioned the following about what parents of children with FXS need:

> *These parents need emotional support through support groups and educational centres such as Autism SA. Furthermore, medical support in terms of affordable medical aids and government grands are needed. Social interaction and family support will also help these parents to not feel isolated. Special needs schooling in terms of preschool, school and afterschool is essential. Respite care is also needed when the main caregiver has to go and do tasks such as grocery shopping et cetera.*

Finally, a routine is important. Under the previous question, I mentioned that a change in routine has an impact on families affected by FXS. Both Mia and Kelly stated that not having a routine places emotional challenges on the family and their social network. Both Mia and Kelly argued that another challenge is that few people and specialists know about FXS, which is often frustrating for the parents when they visit specialists as a last resort. Thus, having specialists who know about FXS and the support of networks and social aspects may help these parents cope.

QUESTION 4:
What coping mechanisms do family members usually employ to deal with a diagnosis such as FXS?

As mentioned before, each family affected by FXS is unique and, therefore, each family deals with FXS in its own way. This is also the case with coping mechanisms. Ann specifically mentioned that coping mechanisms vary from one family to another. In general, in order to cope, families need support from their community, church (depending on their religion), support groups and other families in similar situations. Ann also mentioned that when a family member is diagnosed with FXS, the family needs to make the whole extended family aware of this matter in order for them to be counselled and tested. Mia, speaking from her own experience working with families affected by FXS, mentioned that there are several things families need to cope with this syndrome, which can be internal (within themselves) or external (within their families and communities). She further emphasised the necessity of sound marital relations – "…*have a solid marriage… The families I have worked with have made a point of making time for themselves as a couple."* She further stated that a healthy marriage plays a big role in a healthy family.

Resources in the community is yet another aspect Mia emphasised. She argued that countries such as the United States of America have much more support and resources than South Africa due to the fact that FXS is not commonly known in

South Africa. Mia also placed much emphasis on religious and spiritual aspects, playing an important role in the affected families – "*This goes hand in hand with a supportive community. But it also encourages the family's individual strengths, like having a positive outlook on life, believing that everything has a purpose*."

From these experts' responses, it became quite clear that support is needed for families affected by FXS, especially in a country where little is known about FXS. Kelly referred to care centres that offer support to parents in the sense that they take the burden off the parents' shoulders by looking after the child during the day or during the week:

> *The most common coping mechanism I have seen over the years is to place the child in a care facility, placing the day-to-day responsibilities on that of the care centre. Parents will bring their child home for special occasions or weekend visit.*

Furthermore, there is also the option of hiring a nanny who has experience and training in dealing with autism or autistic behaviour in children. Kelly referred once again to the fathers when she asserted,

> *Fathers tend to cope by running away from the problem and pretending it does not exist. They absolve themselves from any form of responsibility. For the family unit to stay together; routine, taking turns in the child care and finding alternative communication styles seem to bring balance. Having a hobby or external interest also helps to take your mind of the day-to-day issues.*

Kelly also gave specific information with regard to behavioural aspects and discipline, as discussed in Section 4.3.2.

QUESTION 5:

How would you describe the social network of a family that has a child affected by FXS?

Ann and Mia both mentioned that a family's social network varies from one family to another. It would, for instance, depend on their culture, their religion and their country. She mentions as follows:

> *A Muslim family in South Africa reported that they did not have a lot of friends, but their extended family were their anchor… [whereas] the American families I have worked with are often wealthy families with a lot of friends, and they have made a lot of these friends though the FXS support groups and community. Also, their extended families were also very involved and supportive.*

From these quotes, it became apparent that the social existence of families varies according to their support system, whether it is based in their religion, their extended family, their knowledge of FXS or their community.

With regard to cultures, it is important to note, as stated by Ann that in her experience, all families with a child affected by an intellectual disability that have a religious background seem to cope better –

> *I worked with a father who never once mentioned their special needs daughter and when I met their daughter at our son's special needs school, the father would not let me talk about it at work. It was as if she never existed. Another family whose special needs child was placed in my day-care, treated her like an object they owned, not as a person. They brought her to day-care when it suited their routine, they would not spend money on education or therapies and at home she spent her days propped up on a couch watching TV.*

Talking from personal experience, Kelly lamented as follows:

> *The social life and network of the family diminishes when there is a Fragile X family member. Routine is imperative for any Fragile X family, this means that social nights out and doing things on the spur of the moment become less and less.*

In her response, she also mentioned that "*friends sometimes pull away due to not knowing how to deal with your new lifestyle and your child's behaviour"* and that *"finding baby-sitters and respite care is near to impossible when it comes to Fragile X and autism*".

QUESTION 6:
From an educational perspective, do you think more awareness is needed with regard to FXS? What would you suggest?

Ann specifically stated that more awareness should be created with regard to FXS. She stated, "*This is an X-linked recessive condition and is the second most common genetic cause of intellectual disability.*" As an educational psychologist and passionate FXS ambassador, this participant could not make it any clearer that people do not know about FXS and that more education should be provided, and awareness of the syndrome should be created – "*Too many individuals are undiagnosed or misdiagnosed."* Mia also argued that health practitioners, paediatricians, gynaecologists, teachers and therapists should spread the word on FXS and should educate themselves and the community. Academic journals, schools, magazines and seminars should also spread more awareness. Kelly gave specific advice:

> *A Fragile X SA web-site is needed – maintained regularly with updates and interesting articles. An NGO/NPO support group is needed – like Autism SA. Fragile X needs to be communicated and taught to doctors and specialists during their university studies. All special needs schools – government and private – need to be educated on identifying and working with a child with FXS. Fragile X must be listed as a common special need in all manuals used for training preschool teachers and Primary school teachers. The common ones to date are Down's syndrome and Autism, along with Cerebral Palsy. We need local media to do more articles in general magazine such a Huisgenoot and You – not only in specific medical or educational magazine where the market is limited. Forming a NGO for Fragile X and managing a South African based FXS website.*

4.3.2 Narrative data

Kelly has a 17-year-old son with FXS and has, over the years, been able to establish a few aspects of routine, which have proven to help her family in terms of having a family member with FXS. She gave advice on behavioural aspects and discipline, as well as specific recommendations with regard to the following aspects, which are given individually.

4.3.2.1 Behaviour

For behavioural challenges, Kelly mentioned that routine is the best way to prevent outbursts of anxiety. She mentioned, "*Picture routine works very well in the form of photos of the child's daily activities. For example, brushing teeth, getting dressed, washing face, putting on pyjamas for bed et cetera."* Kelly gave this heart-wrecking statement:

> *When I fetched Jadin from "school" in the evenings, he had memorised the route home. One evening to avoid traffic, I took a different route. Jadin had a massive tantrum – kicking the dashboard, screaming uncontrollably and hitting the window. It took me a long time to realise what had caused this sudden outburst. Thereafter I had to teach him that we had the possible routes to go home, the route to Claire's friends' home and the route to Dad's office. When I fetched him, I had to let him know where we were going to avoid these dashboard attacks.*

In order to avoid tantrums and to make Jadin feel safe, she recommended the following, based on their personal experience:

> *When a change in routine is needed, the child needs to be notified and prepared for these changes. We learnt some "best practices" along the way which I am happy to share with other parents. For each family this will be different. I had to prepare Jadin for short trips to the shops or visiting friends. If he knew where we were going, he was content.*

4.3.2.2 Discipline

Kelly argued that discipline goes hand in hand with routine. She argued as follows:

> *Many parents feel that it is wrong or cruel to discipline their special needs child. However, a child is a child and needs discipline for the*

> *family unit to function effectively. The discipline needs to be standardised for both the FXS child and other siblings.*

She said that many parents cannot cope with anger outbursts and tantrums and then give in to avoid tantrums and outbursts. Kelly argued as follows:

> *Wrong! As early as possible a child needs to learn that these outbursts are unacceptable and there are consequences for negative behaviour. I'm not saying, "smack the child" but find ways to let them know they are being punished for a negative behaviour. For Jadin, we would put him in safe place and let his tantrum pass with no recognition/reward for the tantrum. If we were in a public place, one of us will remove him and take him to the car.*

She advised that good behaviour should be rewarded –

> *The child also needs to understand the concept of good behaviour… [and] many people argue this concept but in doing so you are preparing the child for life. In life we all get rewarded for good behaviour – an adult goes to work and gets paid. If they don't perform they get fired and get no pay!*

Finally, Kelly argued that a child with FXS also needs to be entertained. However, "*because of the developmental delays, the FXS child has a very short concentration span (as with a toddler)".* She gave her guideline – *"…five minutes concentration time for each developmental year. E.g.: If the child's mental developmental age is two years but they are 7 years of age, you cannot expect more than ten minutes of concentration time.*"

She stated that this short concentration time causes pressure on the mother and the family –

> *In a "normal" home, a child will sit and playing with toys or watch a movie whilst the mother does housework or cooking but with a child whose concentration is limited this means the mother must constantly find something new to keep the child entertained. To do so, a variety of toys is needed. Help in the form of a spouse, grandparent or older sibling is needed to keep the child entertained whilst the mother continues with her normal household chores.*

4.5 THEMES AND CATEGORIES

Through the responses of the participants in this study, I collected enough data to use inductive analysis to identify themes and categories. The following themes and categories emerged from the data collected through the interviews as well as the narrative.

Table 4.2: Themes and categories that emerged from the data

THEMES	CATEGORIES
1. Mother	Guilt Loss Fear Isolation
2. Family relations	Siblings Marriage Extended family
3. Challenges	Socialisation Finances Concerns regarding caretaking Lack of awareness
4. Coping mechanisms	Support

Table 4.2 portrays the four main themes identified in the study. The first three themes – the mother, family relations and challenges – are connected, as it illustrates the extent of the factors having an impact on families with a member with FXS. The last theme deals with the mechanisms of coping with FXS in a family. Each theme also presents specific related categories.

4.6 DATA INTERPRETATION

Nieuwenhuis (2013:111) states that data interpretation refers to the themes and categories that emerge from the data analysis. Here the researcher should look for patterns, concepts and explanations in the data. The themes and categories that emerged from the data (see Table 4.2) were used to interpret the data by referring to the literature, theoretical framework and research questions.

4.6.1 Theme 1: Mother

The first theme that emerged, was the mother. Sousa (2017:220) states that the image of a good mother is “socially constructed”, which is influenced by historical and cultural determinants. In many cases this is seen as thorough mothering. The mothers of children with intellectual disabilities are under severe emotional pressure (Ellingsen, Baker, Blacher & Crnic, 2014:666; Section 2.3.2). This is the person who is the carrier of the gene that causes FXS and, therefore, this theme produced categories related to guilt, loss, fear and isolation. The mother also represents the first triangle in Bowen’s theory, where he refers to the concept of triangles. Triangles refer to the smallest relationship unit in the family, which for the purpose of my study, is the intrarelationship of the mother, in other words, her relationship with herself and the various emotions she experiences as the mother of a child with FXS.

FXS is seen as one of the most common inherited forms of intellectual disability (see Section 2.2). The mothers, who are the full-mutation carriers and who, therefore, are responsible for passing the FMR1 full-mutation gene to their sons, often present with feelings of guilt and loss – guilt because they blame themselves for passing the syndrome to their children and loss because the dreams they had for their children as fully functioning members of society, free of any barriers, are now shattered (see Section 2.3.3). In order to illustrate this, Kelly, the mother of a son with FXS, stated, “*As a mother, you expect your child to be normal and when they are not, you face many mixed emotions*.” Mia referred to a mother with four daughters with FXS, who experiences immense sadness, since she would never be able to arrange their weddings and have grandchildren.

Mia, furthermore, mentioned that the mothers deal with a lot of guilt, knowing that they have transferred the gene to their children. This is confirmed by Fernandez and Aldridge (2011:7), when they state that the mothers blame themselves or feel as if they are being punished when they have children with FXS. The guilt also extends to how such a mother reacts to her child with FXS, as well as her other children. Mia mentioned that one of the mothers she had worked with, stated that if she had known that she had FXS and would pass it to her children, she would have never have had any children.

The protective instinct of the mother is also often activated. Kelly illustrated the feelings of a mother when she said the following:

> *I was told to place Jadin in a facility to allow for our family unit to function "normally" but I would not even consider this option. Jadin is a child, my child, in spite of his differences and challenges. I was his mother and I loved him no matter what. The emotions I faced were different to most mothers (as I've learnt through the years), I was upset that I couldn't adequately care for him, I was nervous that I would fail him, and I feared for his future as a special needs child in South Africa.*

Furthermore, the mothers often experience isolation, since their children's behavioural problems cause their friends and even family members to avoid them. The social isolation also has an impact on the mother's identity, as she is not regarded as an individual person anymore, but as the mother of a child with FXS (see Section 2.2).

4.6.2 Theme 2: Relations

The second theme is linked to my primary research question, which asked, "What is the effect of Fragile X syndrome on family relations?" The categories that emerged, therefore, refer to the mother's relationship with her other children, the marital relationship as well as the family's relationship with the extended family. Bowen's concept of triangles is also applicable here. Titelman (1998:51) explains the relationship between the mother, father and children as a triangle, which is characterised by interdependence and interaction as stated in Bowens theory. Should a child be diagnosed with FXS, the result thereof can be tension in the family relations (see Table 2.1).

When they are confronted by the FXS diagnosis, families experience turbulent emotions. Kelly mentioned that parents often consult numerous specialists and when they receive the diagnosis, they may experience not only feelings of shock, but also relief, as they can finally put a name to what is wrong with their child. Kelly, who is herself a mother of a child with FXS, mentioned that the diagnosis of FXS had an enormous impact on her family. Her husband pulled away and she, as the mother, had to care for the family and deal with her emotions as well. It is also important to note that parents, especially the mother, is not at all the only one who has to deal with the emotional impact of FXS. The siblings of the child with FXS are also affected by the syndrome. All three the participants mentioned that the mothers of children with FXS often end up feeling guilty, which affects the way they take care of their children with FXS, as well as how they take care of the rest of their families.

The parents are often under a lot of pressure because not only must they try to give an equal amount of attention to the child with FXS and his or her siblings, but they must also at times accept and deal with the child's siblings being unhappy due to the amount of attention given to the child with FXS. Kelly specifically mentioned that her son's FXS diagnosis affected her relationship with her daughter. The siblings experience many stressors, including encountering a difficult relationship with their affected brother or sister, as stated by Mandleco, Freeborn and Dyches (2014:24). Within a family, the siblings share a unique bond with one another and Beyer (2009:444) mentions that it is typically the longest relationship in their lifetime. Kelly stated that there was a unique bond between Jadin and his sister, who was described as "amazing" and someone who took it on herself to be Jadin's other "mother" in order to help Kelly. This corresponds with Bowen's sixth concept of sibling position (see Table 2.1). Seeing that the sister is older than the child with FXS, she adopted a maternal role and was very protective of Jadin.

De Caroli and Sagone (2013:1222), on the other hand, argue that the siblings of children with autism-related disorders, such as FXS, have been found to be more negative towards their sibling with FXS. They also appear to have social issues, which indicates that having a child affected by FXS may not just have an impact

on the family, but more specifically on sibling relationships. In relation to these statements, I want to emphasise that the effect of FXS on siblings appears to depend on the family context. Moreover, it is also important to note that great emphasis is placed on genetic counselling, not only for the parents, but for the entire family in order to gain help from therapies and strategies to deal with a sibling with FXS (see Section 2.3.2). Sibling relationships may also be affected when the parents feel guilty with regard to the fact that the siblings often have to take care of their brother or sister with FXS when their parents are no longer there. Mia suggested that the siblings might feel burdened, as people with FXS may need lifelong assistance. The effect of FXS on the broader network of the family is also important.

Mia, who has stayed with families affected by FXS, is of the opinion that a solid and maintained marriage helps parents cope better with the diagnosis of FXS. Moreover, a solid marriage will allow the parents not to lose contact with each other. Kelly did not specifically say that her marriage was affected by FXS, but she mentioned that after the diagnosis, her husband distanced himself from her, Jadin and their daughter. He simply could not cope with the problems they, as a family, were facing and, therefore, started working long hours, partying and making excuses not be home over the weekends. This type of behaviour corresponds with the seventh concept of Bowen's theory, namely "emotional cut-off" (see Table 2.1), and refers to the termination of an emotional relationship, which fortunately was only a temporary stage in Kelly's marriage. For Kelly and her family, this behaviour went on for seven years, after which her husband seemed to realise the importance of his family – "*I was grateful that Matthew came back to us.*" Research also points to the pressure in a marriage when a diagnosis, such as FXS, is made (see Section 2.3.2). The mothers may feel that they have burdened their families and that they have failed them. Beresford et al. (2007:3) advise that parents need to put certain measures in place to preserve their relationship.

Another category that emerged was the fact that the diagnosis of FXS not only implies serious implications for the immediate family, but for the extended family as well. Kelly mentioned that in her personal encounters with families, some family members tend to be supportive, while others distance themselves and are in

denial. As mentioned before with regard to Kelly's case, her father stated that the syndrome could not come from his side of the family. Ironically, after doing tests, the FXS gene was traced back to his line of the family. According to Vistootsak, Charen, Rohr, Allen and Sharman (2012:7), families are often in denial after the FXS diagnosis, while others do not want to be tested at all due to the possible implications of such a diagnosis. Similarly, research conducted by Fernandez Carvajal and Aldridge (2011:79) indicates that some families live in denial when they are faced with an FXS diagnosis. Visootsak, Charen, Rohr, Allen and Sherman (2012:7) report that reactions ranging from defensiveness, denial and anger to disinterest result when some extended family members hear that they might be the inheritors of a genetic condition. Thus, a shift should be made from the concept of the individual patient to that of helping the family (Fernandez Carvajal & Aldridge, 2011:83). Both Ann and Mia argued that families affected by FXS need support, as well as supporting one another.

4.6.3 Theme 3: Challenges

Humans are social beings (Herrmann, Call, Hernández-Llorenda, Hare & Tomasello, 2007:1360) and, therefore, need interaction with other human beings. However, socialising with other families that are not affected by FXS may be difficult (see Section 2.3.3). Bowen's eighth concept, namely "emotional processes in society" (see Table 2.1), especially refers to this aspect where reference is made to societal effects and opinions that may affect families. As children with FXS very often have strange behavioural features, such as hand-flapping due to excessive anxiety (see Section 2.4.1.3), this may have an impact on how other people react to children and their loved ones. Kelly gave the example that her family's entire social life diminished after they received the diagnosis, as some friends and family members did not know how to behave towards or treat the child with FXS. Mia added that each family is different and that in her opinion, socialisation goes hand in hand with aspects of culture, religion and the country. For instance, a Muslim family she had worked with in South Africa reported not to have many friends, but that their extended families were their anchor. In contrast, she mentioned that in America, the families she had worked with had many friends and family members with whom they could socialise. Kaminsky and Dewey (2001:399) state that some children with FXS have autistic temperaments and that

socialising are very difficult for them. In many cases, the siblings of the child with FXS may be more interactive with their brother or sister with FXS. This may very well be due to their understanding of their sibling's behaviour (Kaminsky & Dewey, 2001:399). Ann mentioned that the social networks of those families affected by FXS are variable, since no two families are the same. She specifically mentioned that the child with FXS may also experience side-effects of medication. Because of the lack of friends and understanding, Dew-Hudges (2004:46) mentions that these children often experience rejection and, subsequently, have a low self-esteem.

One of the major challenges parents experience when their child is diagnosed with FXS, relates to financial concerns, as extra money is needed for schooling, therapy and support (see Section 2.3.3). Beresford et al. (2007:3) mention that in many cases there might be extraordinary demands associated with having a child with FXS, such as full-time caring, equipment and suitable housing. Kelly mentioned that there are numerous financial concerns associated with a child with FXS. Financial provision has to be made for:

- family support, such as support groups and educational centres;
- medical support, such as medical aid and support in terms of therapy and medication;
- family support by allowing social interaction and alone time for the parents;
- special needs schooling; and
- respite care.

From a clinical view, interventions when a family member is a full- or premutation carrier, necessitate genetic counselling. It is important to gain an understanding of any effects on insurance, as well as to the potential impact of genetic counselling. Moreover, prenatal screening may be done in order to find out whether one is a carrier.

Another challenge parents are confronted with when they have a child with FXS, was mentioned by Mia when she said that parents very often are concerned of what might happen to their child with FXS when they are no longer there to support and take care of him or her. This boils down to whether a sibling or

siblings would take care of this child. Parents often have to deal with the guilt of having to burden their other children with having to take care of the sibling with FXS.

Another challenge parents experience is the lack of awareness of FXS even though FXS is among the most common inherited genetic intellectual disabilities and yet either misdiagnosed or not diagnosed at all (see Section 2.3.1). This aspect was mentioned by all three experts and they related to the lack of awareness of FXS internationally and nationally. During the interviews, the participants often mentioned that more awareness should be spread of FXS, especially because FXS is the second most common form of an intellectual disability in the world. Parents are often compelled to explain to other people, at times even doctors and specialists who have never heard of FXS, what this syndrome entails (Fernandez Carvajal & Aldridge, 2011:84). Moreover, both Ann and Mia mentioned that families affected by FXS often feel that they experience a lack of support due to the absence of awareness of the syndrome. Mia mentioned that in America and other parts of the world, there is more awareness about this intellectual disability, yet still not enough. Closer to home, the lack of awareness and subsequent support for families affected by FXS is a matter of concern.

4.6.4 Theme 4: Coping strategies

Throughout the responses by the participants it became apparent that the lack of awareness of FXS, especially in South Africa affects the support of families affected by FXS. Ann mentioned that families need emotional, financial and clinical support, whereas Mia argued that not only is support from the family and extended family important, but also support from their community and church in order to maintain marital and family relations. From her experience, Ann mentioned that families affected by FXS need emotional support and counselling. Genetic counselling is seen as a manner in which families receive professional help upon receiving an FXS diagnosis (Fernandez Carvajal & Aldridge, 2011:63). According to Kelly, there are few records of families being diagnosed with FXS in South Africa, as FXS is not commonly known. Thus, feeling isolated and having few social resources and little support might contribute to families feeling overwhelmed and rejected (see Section 2.3.3). Genetic counselling is a way in which families

are helped to deal with the diagnosis, not only for the immediate, but also the extended family. Both premutation and full-mutation carriers should receive genetic counselling.

4.7 CONCLUDING REMARKS

Even though FXS is found in all different races and on all economic and social levels (Fernandez Carvajal & Aldridge, 2011:13), it does not mean all the affected families deal with it the same way. As stated earlier, 1 in 2 500 to 4 000 males and 1 in 4 000 to 6 000 females are affected by FXS and according to these numbers, around 1 in 260 females will have the full mutation and 1 in 300 to 800 males will be carriers of the premutation gene. Again, no race and no culture can escape this intellectual disability. Furthermore, awareness would be created because by creating more awareness of the syndrome, more people will be able to help and develop ways to support these individuals and families. Families, especially the mothers of children with FXS experience FXS as emotionally and intellectually overwhelming. In many of the responses, FXS is seen as a loss, as a result of the loss of the perfect child the parents had anticipated. Some people may even feel they are being punished in some way. The diagnosis of FXS has many implications for a family, yet if approached and educated in the right way by the right people, it may just be the blessing Kelly and Mia refers to.

CHAPTER 5

SUMMARY, CONCLUSIONS AND RECOMMENDATIONS

5.1 INTRODUCTION

Chapter 5 concludes the study by summarising the key findings from the literature review and the empirical study. The research conclusions are drawn by answering the research questions (see Appendix 1). Based on these conclusions, recommendations are suggested for all role players involved in FXS.

5.2 SUMMARY OF LITERATURE AND EMPIRICAL RESEARCH FINDINGS

This section contains a summary of the main literature and the empirical findings made in this study.

5.2.1 Summary of key literature findings

Through an in-depth study of the literature pertaining to FXS, the following important facts emerged: Fragile X syndrome (FXS) is one of the most common forms of intellectual disabilities, which can be found across all boundaries, be that cultural, racial, demographic or financial. In spite of the high incidence of this syndrome, very little awareness of FXS exists and, therefore, many people may go through their lives not knowing that they may be a pre- or full-mutation carrier. As a result of their unfamiliarity with or lack of knowledge of FXS, this gene will be passed to future generations (see Section 2.3.1).

The literature points to the profound impact of FXS on family relations, which goes beyond the direct familial unit, such as the mother and father, to include the siblings, as well as the extended family (see Sections 2.3.2 and 2.3.3).

As the mother is the primary carrier of the gene, she is hit hardest, with feelings of guilt, anxiety and, in some cases, social isolation and depression (see Section 2.3.3). The parents experience loss, as their dreams for the child are shattered and often the marital relationship is also affected negatively. The siblings often experience feelings of neglect and are burdened by the responsibility that accompanies having a sibling with FXS. Bowen's theory on family systems theory (see Section 2.5) uses the concept of "triangles" to illustrate the interdependence and interaction of a family's emotional relationships and the impact it has on family relations, should one family member be affected in some way (see Table 2.1).

Due to the fact that the child with FXS presents with behavioural, social, emotional and intellectual challenges, their families are often socially isolated, not only by their friends, but also their extended family (see Section 2.3.3). Children with FXS are often aggressive and impulsive, displaying behaviour which society may perceive as undisciplined and unacceptable. A child affected by FXS may also be seen as rude in some cultures due to the fact that they struggle to make eye contact with people. Emotional difficulties are also seen in children who are affected by FXS. They may have emotional meltdowns in front of other people, which can be misunderstood and cause people to withdraw from not only these children, but also their families. Bowen's seventh concept, namely "emotional cut-off" is applicable here, as it refers to the termination of relationships.

5.2.2 A short overview of the empirical research findings of this study

In this study, three participants were selected to partake in this study based on their experience within the field of FXS and working with families affected by FXS. The data collection methods involved structured interviews and one narrative. Four themes from the data analysis emerged, namely the mother, family relations, challenges and coping mechanisms.

As the primary caregiver and main carrier of the FXS gene, the mothers experience feelings of guilt because of transmitting this syndrome to her children and the resultant impact thereof on her child's life. The participants also mentioned feelings of loss, when they think about all their unfulfilled dreams being shattered when they picture the future they had dreamed of for their children. All their hopes

for what their children would achieve, seem in vain in light of such a diagnosis. Some mothers were fearful, as they were not sure what to expect in the future, which can be brought back to the lack of information currently available to anyone struggling through a diagnosis such as FXS (see Section 4.4.1). Isolation from other families and children, and sometimes even their own extended family, was also experienced (Section 4.4.1).

The family relations are also affected, and the siblings may feel rejected or cast aside (see Section 4.4.2). The parents often spend a lot of time on the child with FXS, leaving the “normal” child or children feeling rejected and experiencing a lack of care and appreciation. Additionally, there are financial implications, as the siblings often cannot have the things they want (see Section 4.4.2). The marital relationship is often affected, and Kelly specifically referred to her husband who had withdrawn from her and the children, as he could not face their circumstances. Mention was also made that the marriage tends to head into one of two directions, either a stronger unit emerges when the parents are confronted with such a diagnosis, or the unit splits up due to the stress caused by the care needed (see Section 4.4.2). Having a child with FXS also affects the extended family, where denial is often experienced (see Section 4.4.2).

The challenges the families affected by FXS experience, involves socialisation, as society and the extended family members tend to avoid social contact with these families. Socialising one’s child with other children becomes a task that requires foresight and planning when a child has FXS versus a child that is developing normally (see Section 4.4.3). This, furthermore, affects the caregivers’ social life, as often, their children inability to socialise normally excludes them from spending time with the families and friends they had made before the diagnosis The financial impact on the family cannot be ignored, as it is not only the financial implication of the testing needed to conclude the diagnosis, but the costs of additional care need to be taken into account as well, which, as mentioned earlier, may lead to parents having to limit expenditure, sometimes at the siblings’ expense.

There is also the concern of ongoing care for the person that has been diagnosed, which on top of being expensive, often means that the siblings are expected to carry that burden, should the parents no longer be there to take care of the child with FXS. The parents are also concerned about their child's future, as they often do not know who will take care of their child once they are not there anymore. They also experience feelings of guilt because the siblings are expected to be responsible for possible future care. Finally, the core finding of the study relates to the lack of information available on FXS. The ignorance of society and, often, medical personnel, results in social avoidance, misunderstanding and parents having to bustle around in search of support and general information.

Support was identified as the only way in which parents can cope with a diagnosis of FXS. Ann specifically mentioned the need for emotional support counselling in order for these parents to cope. From the empirical research it became clear that FXS has a tremendous impact on the family, involving the relationships and various challenges. Awareness of this syndrome is, therefore, crucial.

5.3 RESEARCH CONCLUSIONS

The research questions (see Section 4.3.1) were answered to enable me to draw the final conclusions of the study. The secondary questions are attended to first, followed by the main research question.

5.3.1 Secondary research question 1: What is the profile of a child who presents with FXS?

FXS manifests with behavioural, emotional, physical and social characteristics that make it difficult for families to cope and have a social life, especially if the child cannot cope in social situations. The child with FXS often presents with anxiety, which can cause them to do inappropriate things in public, such as hand-flapping, tantrums and seeming unmannered when they do not make eye contact or socialise at all. The child's physical appearance may also contribute to people staring and making the family affected with FXS even more uncomfortable in social situations. As Kelly mentioned, some family and friends desert one if one's child has FXS, due to the lack of knowledge of FXS.

Children with FXS often have emotional difficulties because they may feel rejected by their peers and are excessively shy. Thus, they often withdraw from other people. Children with FXS also present with hyperactivity, which may lead to their being bored and then getting irritated and aggressive. Furthermore, children with FXS are severely tactile defensive and will often avoid being touched or avoid certain clothing. If then forced in certain situations, such as being hugged or to wear certain clothing, they may start presenting with behavioural manifestations, such as hand-flapping or hand-biting.

Communication is yet another aspect that is difficult for a child with FXS. Communicating their needs is difficult and very often the reason they become frustrated and then lash out or present with behavioural manifestations, such as hand-flapping and so forth.

The profile of a child with FXS is broad and even though there are certain manifestations that are seen as indications of children with FXS, each child with FXS is still unique and should be treated uniquely.

5.3.2 Secondary question 2: What challenges do families with children with FXS experience?

This study found that the main challenges families face relates to their socialisation, as society often do not understand the manifestations of FXS. These families often experience isolation, as people do not want to associate with them because they do not understand the syndrome. Financial challenges represent a large burden for the family, as provision needs to be made for additional support, such as therapy, doctors' fees and so forth. The parents are also often concerned about their child's future and have questions relating to caretaking and guilt, as the siblings will often be responsible to take over the caretaking role in the future. The need to have someone to take care of a child with the syndrome can be overwhelming, even when it to comes to simple tasks, such as the parents going out for an evening. This becomes an even larger concern once the parents themselves are no longer able to provide that care. Furthermore, due to the lack of awareness, these parents do not always know how to deal with their child. Most

doctors and therapists do not know about FXS and, therefore, cannot always support families affected by FXS.

5.3.3 Secondary question 3: What coping strategies do parents use in raising a child with FXS?

As each family is unique, it was found that in most cases, FXS is dealt with differently. Support is necessary for the parents to cope and certain strategies, such as keeping to a routine and preparing the child if one needs to deviate from the normal routine, were mentioned (see Section 4.3.2). A strong marriage was also mentioned as a supporting mechanism (see Section 4.3.1) and from the literature and empirical findings it could be deducted that a close family bond is fundamental in dealing with the diagnosis of FXS.

5.3.4 Main research question: What is the effect of Fragile X syndrome on family relations?

This research has found that FXS has a determining impact on family relations, which was illustrated by the concept of “triangles” in Bowen’s family systems theory (see Section 4.4.2). The mother or primary caregiver experiences multiple emotions and often needs to negotiate her feelings of guilt, loss, fear and isolation when being confronted with the diagnosis and subsequent life with a child with FXS. The core familial unit of the child diagnosed with FXS is also affected, as the marriage is often under a lot of strain. The siblings may experience feelings of neglect and rejection and may have to make a lot of sacrifices. The social network of the family is also affected, as the parents often lose friends because of their child’s behaviour. Their relationship with the extended family is affected, as the latter have to come to terms with the possibility of their having passed this gene on, which can lead to fractures within a family.

5.4 RECOMMENDATIONS

With reference to the findings of the research, the following recommendations are made to the Department of Health, the extended family and parents of persons with FXS and for further research.

5.4.1 Recommendation directed at tthe Department of Health

Recommendation 1: Creating awareness of FXS

The Department of Health needs to arrange awareness campaigns to inform the public about FXS as one of the main forms of intellectual disabilities. The following advice emerged from the empirical study, which can inform the guidelines emanating from the Department of Health:

- Informing health professionals and paraprofessionals about the aetiology, diagnosis and incidence of FXS.
- Creating awareness about FXS by informing schools.
- Arranging conferences to disseminate relevant research about FXS.
- Designing a website on FXS, which can be regularly updated and maintained by a professional to support the public with the necessary information regarding FXS.
- Educating the public in general about FXS.
- Including FXS as a special need in all education policy documents.
- Using the media as a platform to create awareness through talk-shows, magazines articles and radio talk shows to create general awareness.

By focusing on these awareness campaigns, resources will emerge, which will, in turn, help the parents and families affected by FXS gain knowledge and insight.

5.4.2 Recommendations directed to the extended family

The following recommendations are directed towards newly diagnosed parents, as well as parents who seek advice in coping with their child and his or her siblings.

Recommendation 2: Testing for Fragile X syndrome

Extended family members from the same bloodline should go for screening tests to determine whether they may be carriers of a premutation or full-mutation gene. They should receive genetic counselling to explain the impact of the diagnosis.

Recommendation 3: Taking on a supportive role

Extended family members need to be aware of the impact a diagnosis of FXS may have on a family. Through obtaining the necessary information on FXS, they need to support the family through visits, showing interest and being available for emotional support.

5.4.3 Recommendations directed to the families of children with FXS

Recommendation 4: Attitudes of parents

The parents need to see their child with FXS as an individual in his or her own right and not as a burden. They should contemplate what would be best for themselves, their child with FXS and their other children, and focus on familial relations as a strong support system.

Recommendation 5: Joining a support group

The parents should join support groups at genetic clinics to interact with other parents who also have children with FXS or autism. Knowing that somebody is in the same boat, or even seeing that one is a bit more privileged may also give a new perspective. Thus, gaining support not only from parents in a similar situation, but also gaining support from the general community, family and church may also help the parents and siblings to cope better with FXS.

Recommendation 6: Counselling for mothers of children with FXS

The mothers are often the main caregiver and mostly the ones who transmit the gene that causes FXS to their children. Because of feelings of guilt, depression and isolation, the mothers need to talk to someone, such as a close friend or a counsellor, on a regular basis.

Recommendation 7: Gaining financial support

Parents who do not belong to a medical scheme can approach the Department of Health for financial assistance. Such support may alleviate financial concerns and ensure that specialist help in terms of therapy and special education can be gained.

Recommendation 7: Strengthening of the marital relationship

The parents need to spend some alone time together to help strengthen their marriage and to maintain a good relationship. If possible, it might be a good idea for these parents to ask a family member with whom the child is comfortable to take care of the child for an evening.

Recommendation 8: Strategies to deal with communication challenges and routine

The following strategies can be recommended to support parents with discipline, setting routines and communication:

- It is important for the parents to explain any deviation to a routine to the child with FXS. Explaining that there are alternatives and that sometimes things may change, will help prepare the child that change is inevitable.
- A system in order to encourage good behaviour, such as rewards for good behaviour and intolerance for outbursts, is recommended.
- Entertaining a child with FXS is recommended. Due to the child's short attention span and developmental delays, entertainment should be highly attended to. A variety of toys, a movie while the mother is doing household work and creating a "normal" home is highly recommended. The siblings are also a source of entertainment. If the siblings and the child with FXS play together regularly, it may also strengthen their relationship.
- Communication plays a significant role in any relationship. As children with FXS often struggle to communicate their needs, it is recommended that creative measures should be taken to help the child communicate, such as a cue card system, which involves making cards representing needs such

as food or water. This may contribute to their social issues and anger outbursts.

Recommendation 9: Planning for the future

Planning for the future of the child with FXS is very important. The parents should decide who would be taking care of the child once they are not able to fulfil this role anymore. This decision needs to be communicated to the child as well as the person(s) who will be taking over this role.

5.4.4 Recommendations for further research

- The data for this study were gathered from experts in the field of FXS. It is recommended that families who are affected by FXS should be involved to gain broader insight into the challenges and support they experience. Taking into account that FXS is one of the most prevalent disabilities, there is still very little research available about this phenomenon. One of the key findings of this study is that more education and learning on this subject would assist families to cope better with the diagnosis.

- This study focused on the impact of FXS on a family. It was a rather general concept. More in-depth research should be done into what can be done to create awareness and to provide techniques and help for parents of children with FXS all over the world. Thus, a manual with methods or conferences should be developed to stimulate more research topics on FXS.

5.5 CONCLUDING REMARKS

This study aimed to investigate how family relations are affected by FXS. Although I could not access families of children with FXS, the three participants provided sufficient information to ensure that in-depth insight into the functioning of such families could be garnered. I was very lucky that one of the participants is the mother of a boy with FXS. I am, therefore, satisfied that my study and subsequent recommendations may assist various role players in dealing with FXS. It is my

wish that further research may be conducted in this field in order to help parents, therapists and researchers to gain more insight into FXS.

Personally, I regard this study as a learning curve, as I gained so much understanding of this syndrome. I also wish to gain further knowledge by completing my doctoral degree in FXS in order to help families by means of a manual including strategies to cope with FXS. FXS is both a steep and a smooth road because, in the words of my participants, children with FXS are “awesome” and “fantastic”. FXS is now part of me and a passion I would like to pursue through further research.

Throughout this study I gained the insight that even if a child is not born into the world as perfect as we had hoped for, there is always something that we can love and see as being perfect in that small individual. Children with FXS are precious gifts and their parents were chosen specifically to nurture these gifts.

I hope that wherever my studies go and wherever it is distributed, I may touch the life of someone, perhaps provide some comfort and help those affected by this syndrome. It is my biggest wish that no-one needs to go through something like this on their own and that my study is a stepping stone to greater understanding.

Life is not about waiting for the storm to pass,

it’s about learning to dance in the rain.

- Viviane Green -

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ANNEXURE 1: INTERVIEW QUESTIONS

UNIVERSITEIT VAN PRETORIA
UNIVERSITY OF PRETORIA
YUNIBESITHI YA PRETORIA

September 2017

Interview questions to professionals in FXS

The effect of Fragile X Syndrome on family relations

Dear Mrs _____

I would like to thank you for taking the time to answer the questions about my study. This study, *The effect of Fragile X Syndrome on family relations*, involves understanding the phenomenon of Fragile X syndrome and the effects of a child with this syndrome on a family. I would, therefore, appreciate your extensive knowledge and understanding through your years of experience on this matter.

Important factors to consider before answering the questions:

1. Please use this document and reply to each question underneath the question.
2. Answers may not be handwritten.
3. Please do not provide any personal details of patients. If you choose to make your name available, please type your name at the end of the questions.
4. Please note that a copy of the data analysis will be sent to you before the study is sent for publishing.
5. Please write a narrative of no longer than 100 words at the end of these questions providing insight into your experience of having worked with and encountered families affected by FXS.

Research questions to experts in the field of Fragile X syndrome:

1. What are the challenges that families with children with FXS usually experience?
2. In your opinion, what is the impact on family relations when one of the children has been diagnosed with FXS?
3. In your opinion, what are the needs of parents who have a child with FXS?
4. What coping mechanisms do family members usually employ to deal with a diagnosis such as FXS?
5. How would you describe the social network of a family that has a child affected by FXS?
6. From an educational perspective, do you think more awareness is needed with regard to FXS? What would you suggest?

Should you prefer, the study will be made available to you, once it is finalised. If you have any questions about the consent letter you may contact me or my supervisor. Should you be willing to participate in the study, please sign the permission slip supplied below. This document can be emailed to me.

Yours sincerely,

Melissa Pienaar
Signature ……
MEd student (Department of Early Childhood Education)
University of Pretoria
Pretoria 0002
Republic of South Africa
Mobile: 072900 5886
Email: u14329655@tuks.co.za

ANNEXURE 2: CONSENT FORM

UNIVERSITEIT VAN PRETORIA
UNIVERSITY OF PRETORIA
YUNIBESITHI YA PRETORIA

DEPARTMENT OF DEPARTMENT EARLY CHILDHOOD DEVELOPMENT

FACULTY OF EDUCATION

Groenkloof Campus
Pretoria 0002
Republic of South Africa
Tel: (012) 420 5569
Fax: (012) 420 5621
http://www.up.ac.za

- I have read the consent letter and understand the terms of participation as explained in the letter of consent.
- I do agree/do not agree (delete what applies to you) to participate in the research entitled: The effect of Fragile X Syndrome on family relations
- I do agree/do not agree (delete what applies to you) to be interviewed via a personal visit or via skype or email on a suitable time for me and the researcher for a period of at least an hour. I understand that the interview will be audio-taped and transcribed by the researcher.
- I do understand that my participation is voluntary and that I have the right to withdraw during any stage of the research project.

Print name in full: _____

Signature: _____

Date: _____

ANNEXURE 3: EXAMPLE OF ANSWERS FROM A RESPONDENT

UNIVERSITEIT VAN PRETORIA
UNIVERSITY OF PRETORIA
YUNIBESITHI YA PRETORIA

RESPONDENT 2: Mia

1. **In your opinion, what is the impact on family relations when one of the children has been diagnosed with FXS?**

"There are many ways in which FXS affects family relations. Due to the fact that the mothers are the carriers where children have full mutation, they blame themselves a lot. And because of this blame and guilt that they experience, they tend to have depression. In a way I think that this affects the way in which they interact with their children. I am generalising now, but this is what I experienced."

"As a mother myself, I can understand that this can cause great sadness and blame for a mother. Mothers are quick to blame themselves for something that goes wrong, how much more must these mothers feel when they are the cause of their child's intellectual impairments and other challenges."

"When there are siblings that are not diagnosed with FXS, the parents also feel guilt with regards to the fact that these siblings would have to care for their brother or sister diagnosed with FXS, when the parents are no longer there to do so. They might feel that it would be a burden for their siblings to care for them for the rest of their lives, as some of the children diagnosed with FXS would need assisted living throughout their lives."

"Another impact could be on the marriage of the parents. Because of the blame that the mother experiences, this could negatively affect their marriage. They might feel that their husbands blame them for bringing FXS into the family, although this might not be the case from the husband's side."

2. **What do you think are the challenges that such families experience? Please refer, among others, to the mother as well as the siblings in your answer.**

"I have met and worked with families in the USA as well as South Africa, and a big challenge is the financial aspect. This is more challenging for families in South Africa, as FXS is not nearly as well-known as in the USA, and furthermore, there are just not as much resources available as in a first world country such as the USA. Mothers often become stay at home moms, or they have to get a caregiver for the child, which is extra expenditure. There is a lot of therapies and remedial work that these children need. And they also need to be in special schools very often."

"Due to the lack of awareness of FXS around the world and even more so in South Africa, this is challenging for the parents as they always have to explain to people what it entails. Be it at therapy, on a more informal situation."

"These families also can't really do what other families with unaffected children do. All activities have to be altered to meet the needs of the individual diagnosed with FXS. A child with FXS experience great anxiety when it comes to social activities, and also for the unknown. So, when these families plan something, they have to prepare the child way in advance and it should preferably be an activity that the child is comfortable with, one he or she has experienced before. So, they know what to expect."

"When it comes to the mother, it would be like I said before, the guilt and depression. Now again, I am generalising. The siblings might feel frustrated as a lot of the attention goes to the affected sibling. But it could also be the other way around. And then the siblings might get frustrated with not being able to do what other families with unaffected children do. So, this again puts a lot of strain on the parents, trying to juggle between the affected child and his or her siblings."

"And then of course you have a family with three or four children who have all been diagnosed with FXS. This again is a whole new ballgame. One mother told me that if she knew that she had FXS and would pass it on to her children she would rather not have had children. Another mother said (she had four girls with

full mutation FXS) that it saddened her that she won't be able to have weddings for her children and that she won't have grandchildren."

3. **How would you describe the typical family who has a member with FXS?**

"Fantastic people!" "That is one thing that I have come to known working and getting to know families with children diagnosed with FXS, they are resilient. Yes, they have days that are tough, but they get through it and carry on with their lives. They strive to give their children the best they can. The mothers give up everything to keep the family going. At the end of the day they are just like any other family. They have children that have tantrums, they have good days, they have bad days. And they want the best for their children, be it with a syndrome or without. But they are so resilient. When people don't know about FXS they go out and teach them, when there is no social group for their children, they create a social group. The family I mentioned before with the four girls, their one daughter actually got married recently."

4. **In your opinion, what are the needs of parents who have a child with FXS?**

"The one thing that came across quite clearly with all the families I have worked with is that they all want to belong. They need support groups. The mothers have a need to connect with other mothers who are in the same situation. And they also need social groups where their children can connect with other children that are on the same level as them, or higher. These children learn a lot from their peers. They enjoy being social. Also, they need others to know. It is very frustrating for these parents when professionals, doctors, teachers, therapist's etcetera *have never heard of FXS. That means they don't have the necessary skills and knowledge to work with their children."*

5. **What would you say are parents' biggest concern about their child with FXS?**

"Definitely what will happen to their child or children when they are no longer there to care. They worry about the future. Not all have the financial ability to ensure that their children are cared for after they pass away. So, this worries them a lot. Who

will look after these individuals when they are no longer here, if they can't do it for themselves."

6. **What coping mechanisms do family members usually employ to deal with a diagnosis such as FXS?**

"I have found that there are a number of things. It can be things within themselves as well as things within the family and community. Like I said before, a major thing that helps them deal with their situation is that of a support group. Now it doesn't only have to be a support group of parents with similar situations, this helps too. But also, a supportive extended family, or a supportive community. Maybe their church community. Also, have a solid marriage can help them cope. The families I have worked with have made a point there of to make time for themselves as a couple. I believe that a healthy marriage plays a big role in a health family. Having resources within the community also helps. This is much more common in countries like the USA. FXS families do not have as many resources in South Africa as in the USA.
Their religion and spirituality also play a part. This can again go hand in hand with a supportive community. But is also encourages the family's individual strengths, like having a positive outlook on life, believing that all have a purpose, etcetera."

7. **How would you describe the social network of a family that has a child affected by FXS?**

"That is difficult to say. Each family would differ. It would depend on their culture, their religion, the country. A Muslim family from South Africa reported that they did not have a lot of friends, but that their extended family were their anchor. Whereas the American families I worked with were often wealthy families with a lot of friends, and also, they have made a lot of these friends through the FXS support groups and community. However, their extended families were also very involved and supportive."

8. **Would you say different cultures approach a diagnosis such as FXS differently?**

"No, I don't think so. I think that we are all human. And we all experience negative and positive things the same. Parents want the best for their children, doesn't matter what religion they are or what they believe or from which part of the world they are. I think the one thing that one can say would influence the way they handle their specific situation is religion."

9. **From an educational perspective, do you think more awareness is needed with regard to FXS? What would you suggest?**

"YES, YES YES!!! People don't know about FXS. Gosh, and it is the most common genetic cause of intellectual impairment. We need to do whatever we can to tell the world about it. There are so many individuals that are undiagnosed or misdiagnosed.

There is so much that can be done. But getting health practitioners such as doctors, pediatricians, gynecologists on board. And then of course teaches and therapists. It will take some time, but we need to spread the word. Write up on it in academic journals, in magazines, have seminars, have presentations at schools, especially special needs schools. So much can be done, where to start!!!"