It’s my genetic make-up, part of me, who I am.

A psychotherapeutic exploration into the experiences of female fragile X premutation carriers: an interpretative phenomenological analysis.

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Supervisor of Research: Dr. Gráinne Donohue.
But, as we are bone, so are we dream,
as we are ash, so are we seed.

I know a place
where poets and painters trace the faces of God,
and within a leaf, the other side of the moon.

It is that space between skin and bone,
I call “Life”.

~ Neil Frederick Sharpe
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Table 1 – Participant Sample

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For Dad, Bish, Janina and Margo

To Michael, my constant companion, thank you for holding the light when I lost my way.

To my mother, without whose genetic material I would not have had the stamina to complete this project!

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Abstract

The completion of the Human Genome Project (HGP) and the on-going advances in genetic science mean that more people will become aware of their genetic risk. While the ‘lived experience’ is increasingly acknowledged as being significant to wellbeing, it is often overlooked in genetic medicine. This study presents a deeper understanding into the nature of genetic illness by illuminating the world of experience and by exploring the question of meaning in relation to carriers of a genetic mutation on the fragile X gene (FMR1), which is responsible for a family of disorders (FXD) including fragile X syndrome (FXS). A psychological approach to genetic counselling has evolved to meet the needs of those confronting genetic risk yet significant challenges in genetic counselling have been identified which suggests a deficit in the care available to genetic carriers. Three individuals, all female fragile X carriers diagnosed at least three years before the time of the study, were invited to participate in semi-structured interviews. An Interpretative Phenomenological Analysis (IPA) was used to elicit a nuanced analysis of individual narratives and, using an Object Relations framework the study examines the implicit meaning of a fragile X diagnosis thus illustrating how a diagnosis of a genetic condition might manifest in the psyche. The study found that an FXS diagnosis in their children was experienced as deeply traumatic for the participants and the feeling of unconnectedness was experienced in their journey within an unresponsive medical system, thereby exacerbating the trauma. The study suggests that anxiety arising out of a genetic diagnosis, which is experienced as a trauma, cannot be fully understood without the acknowledgement of the unconscious processing of loss. The study addresses the paucity of psychotherapy research into the experiences of genetic carriers and offers new insight into genetic disease. The study recommends additional psychotherapeutic support in the clinical setting for those confronting genetic illness as well as further research using other psychotherapy frameworks to explore the experiences of genetic carriers.
Chapter 1 – Introduction

1.1 Context of the Study

Genetic material containing mutations in the gene structure puts individuals and their children at risk of disease (Lewis, Skirton and Jones, 2011). Individuals who have inherited a mutated gene with future disease potential are known as genetic carriers (Capelli, et al., 2009). This study uses an Object Relations framework to examine the subjective experiences of three women who were diagnosed as fragile X premutation carriers (NB: will be referred to as *fragile X carriers* throughout this study). By conducting an Interpretative Phenomenological Analysis (IPA) of the narratives gathered in three semi-structured interviews, the study illuminates the ways in which a genetic diagnosis of fragile X manifests in the psyche.

The National Human Genome Research Institute states that advances in genomic science “allow us to read nature’s complete genetic blueprint for building a human being” (NHGRI, 2013). However, Todres, Galvin and Holloway (2009) argue that in order to fully understand what it means to be human one must uphold the unique essence of the human being. Scientific advances, while radically improving the health of the population, have neglected the place of subjectivity and the sense of self, both of which are essential to a deeper understanding of health and well-being (Todres, Galvin and Holloway, 2009). In addition, Brocki and Wearden (2006) present mounting evidence of the relevance of IPA in health psychology and describe the beginnings of a new paradigm for understanding illness:

“…with a move away from a simple biomedical model of disease and illness, where an observable bodily process is held to map onto a predictable illness experience in a fairly simple way, there has come an increasing recognition of the constructed nature of illness” (p. 88).
Targum (1981) describes genetic disease as intrinsically human and he emphasises the human suffering experienced when faced with our own vulnerability. Therefore, confronting genetic illness raises fundamental questions of what it means to be human. McDaniel (2005) asserts that our genetic inheritance governs our genetic future and adapting to new information on our current or future health has profound implications for our internal experience as efforts are made to integrate the meaning of a genetic diagnosis.

Esplen, Hunter and Kash (2012) add that an increased awareness of genetic risk triggers a host of emotions that may significantly impact a person’s quality of life. They identify issues around unresolved grief and loss as complicating both self-image and perceptions of genetic risk. They further note that complex emotions triggered by perceived risk impacts on decisions around testing, understanding test-results, accurate communication of risk information to family members and decisions regarding treatment.

A psychological approach to genetic counselling has evolved to meet the growing recognition for the need for psychological support for those confronting genetic risk (Biesecker, 1998; Esplen et al., 2012; Rantanen et al., 2008). However, the research identifies significant challenges for both genetic counsellors and clients within this discipline (Hallowell, 1999; McAllister, 2001; Petersen 1999; Kessler, 1992) which raises the fundamental question as to whether genetic carriers are being met as they try to manage the emotional impact of a genetic diagnosis.

Despite these considerations, there is a paucity of research from the field of psychotherapy into the experiences of genetic carriers as a whole, much less the experiences of fragile X carriers. Consequently, this study addresses this disparity by capturing rich descriptions of the
experiences of three fragile X carriers and by exploring the themes emerging from their internalised experiences through the lens of psychotherapy. The study also builds on the existing research from health psychology which highlights the psychosocial impact of a genetic diagnosis. Finally, the study upholds the significance of the lived experience as relevant to generating new insights into genetic disease.

1.2 Fragile X – a genetic mutation

In 1991, researchers identified a genetic mutation in a gene, known as the Fragile X Mental Retardation 1 (FMR1) located on the X chromosome (Verkerk et al. as cited in Abrams et al. 2012). They discovered the FMR1 gene had the propensity to fully mutate causing the neurodevelopmental disorder fragile X syndrome (FXS). The inheritance pattern of the faulty gene follows the norm of any X-linked condition (Miller, 2006). In short, a mother will pass it to both her sons and her daughters, whereas a father will only pass it to his daughters and not to his sons. However, this single gene gains complexity as it is transmitted through the generations (Bourgeois et al, 2009). Mutations are observed as repeats of a DNA pattern on the gene, known as CGG repeat expansions. The length of the CGG repeat, as it is transmitted, is significant in that it determines whether the inherited altered gene has a normal/intermediate expansion (5-55), a premutation expansion (55-200) or a full mutation expansion (>200). Girls who inherit the altered gene from their father will remain in the premutation range but girls and boys who inherit the altered gene from their mother may remain premutation or they may inherit the full mutation. Individuals who have inherited a full mutation will be affected by FXS (Abrams et al, 2012).

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1 A sequence of trinucleotides: a cytosine followed by two guanines (Miller, 2006).
1.3 Fragile X Syndrome

Fragile X Syndrome (FXS) is considered to be the most common single-gene cause of inherited intellectual and developmental disability and autism (Hagerman, Rivera and Hagerman, 2008). FXS is observed in both males and females and is characterised by difficulties in physical, behavioural and cognitive development. According to Abrams et al. (2012), physical features may include a long face, low muscle tone and prominent ears; behavioural challenges often include social anxiety, impulsiveness, hand-flapping and poor eye contact; and cognitive features manifest along the spectrum of mild to severe learning difficulties. Abrams et al. (2012) add that motor and speech delays are commonplace and often prompt an initial medical consultation. Miller (2006) explains that, because the FMR1 gene is found on the X chromosome, a female who inherits the mutated gene may be somewhat protected by a copy of the gene on her other X chromosome. A male however with the full mutation will always present with FXS as they do not have another X chromosome to compensate. Boys in this respect are often more severely affected by FXS whereas it is possible that girls show little or no signs of FXS.

1.4 Fragile X Premutation Carriers

For a decade after its discovery, clinicians were mostly concerned with the fully mutated FMR1 gene which causes FXS. Individuals with the premutation expansion, known as fragile X premutation carriers, were thought to follow the traditional definition of a genetic carrier as one who inherits an altered gene with the propensity to pass on a genetic condition to their children, while they themselves remain asymptomatic (www.fragilex.org). However, recent research implicates the FMR1 gene as the single-gene cause of a family of fragile X clinical disorders (Tassone, Hagerman and Hagerman, 2014). Accordingly, clinical involvement in fragile X premutation carriers has gained greater acknowledgment and, while not at risk from
FXS, premutation carriers are at increased risk of developing a range of fragile X disorders (FXD).

1.5 Fragile X Disorders

Fragile X Disorders (FXD) include the late-onset neurodegenerative disorder fragile X tremor ataxia syndrome (FXTAS) and primary ovarian insufficiency (FXPOI) as well as risk of weakened immune systems and other neurological problems (Hagerman and Hagerman, 2008; Tassone, Hagerman and Hagerman, 2014). Abrams et al. (2012) define FXTAS, which affects approximately 46% of male carriers and 8% of female carriers after the age of 50, as characterized by severe tremor, gait ataxia and cognitive decline. Furthermore, Abrams et al. (2012) report that approximately 20% of female carriers will experience FXPOI which leads to infertility and menopause before the age of 40.

Within the family of FXD it is then possible that a woman may give birth to a child affected by FXS and her father or brother may be affected by FXTAS while she herself may experience future fertility issues and uncertainty around developing FXTAS (Visootsak, Charen, Rohr, Allen and Sherman, 2011). For these reasons, this study has chosen to explore, from a psychotherapeutic perspective, the lived experiences of female fragile X premutation carriers.

1.6 Aims and Objectives

The overall aim of this study is to offer a deeper understanding of the nature of genetic risk and genetic illness by illuminating the world of experience and by addressing the question of meaning in relation to being diagnosed as a fragile X premutation carrier. Using Object Relations theory to identify how a fragile X carrier diagnosis affects the inner lives of the
participants, the study also addresses the remarkable paucity of psychotherapy research into genetic carriers and in particular into fragile X carriers.

The objectives of the study are as follows:

- To explore participants’ perceptions and interpretations of their status as a fragile X premutation carrier.
- To investigate how participants have managed and contained the risks of their genetic inheritance.
- To explore the participants’ journey within the medical framework thus illustrating how the nature of this interaction creates psychological states which hold the influence of the internal working models of the mind.

The researcher hopes to generate new knowledge and understanding of what it means to be a fragile X carrier. By illuminating the inner worlds of the participants, the study illustrates how a genetic diagnosis should not be reductive as to overshadow the experience of being human. Lastly, the study will broaden psychotherapy research, thus contributing to comprehensive care for genetic carriers and adding to current discourse around the ethics of medical genetics.
Chapter 2 – Literature Review

2.1 Introduction

Due to the absence of qualitative studies on the implicit meaning women have made of a fragile X carrier diagnosis, the literature review begins by presenting a small number of studies from the discipline of genetic medicine which highlight the emotional impact of undergoing fragile X genetic testing. In addition, the researcher reviews medical genetic research which explores the comorbidity of anxiety and depression in female fragile X carriers.

Following on, articles by authors who hold an existential view of the impact of a genetic diagnosis or who have a special interest in psychotherapeutic interventions in the field of genetic medicine is explored. Due to the dearth of literature on fragile X carriers from the field of psychotherapy, further articles included for review are written by psychiatrists and genetic counsellors specialising in psychosocial research in genetic medicine. This literature identifies areas of importance around genetic testing and genetic counselling as significant to a greater understanding of individuals confronting genetic disease.

Finally, the researcher addresses the lack of psychotherapy research into the subjective experience of fragile X carriers by exploring Object Relations literature, in particular the theories of Melanie Klein and Ronald Fairbairn who present the psyche as a dynamic entity which constructs itself, using defences, to create a sense of coherency when disruption is experienced.
2.2 Previous Studies

Studies involving fragile X carriers have, broadly speaking, focussed on women’s reactions to undergoing carrier testing (Anido, Carlson, Taft and Sherman, 2005; McConkie-Rosell, Spiridigliozzi, Sullivan, Dawson and Lachiewicz, 2000). These studies, which sought to determine the psychosocial impact of fragile X genetic testing, identified anxiety, depression, guilt, responsibility and feelings of loss among the participants.

A qualitative study conducted by Visootsak et al. (2011) identified significant delays of FXS diagnoses for children of ten African American mothers. The delays in ordering the tests and the results, along with the insensitive nature of the disclosure of results, caused distress in this group of women and their families.

A study carried out by Lewis Skirton and Jones (2011), including female fragile X carriers, identified the opportunity to engage in appropriate family planning as a benefit of receiving genetic information. Although Anido, Carlson and Sherman (2007) reported feelings of ambivalence in women who had newly learned of their positive fragile X carrier status, they note that the women in this study were not aware of their risk when they agreed to participate in the study, thus suggesting that they had not yet made sense of their new genetic information.

Carmichael (2007) and Heath (2008) state that while a positive diagnosis for FXD in families often provides the affected individual with the support they need around their illness, it is important to recognise the feelings of loss, hopelessness and unresolved grief that often accompany a genetic diagnosis of FXS. Carmichael (2007) adds that anger and denial are commonly witnessed in family members affected by a FX diagnosis.
Medical research into fragile X carriers reports that females, in particular, are at an increased risk of mood and psychiatric disorders such as anxiety, depression and difficulties in social functioning (Hagerman and Hagerman, 2008; Lachiewicz et al 2010; Tassone, Hagerman and Hagerman, 2014). Much of the medical research focuses on the correlation between CGG repeats and psychiatric manifestations in premutation carriers (Bourgeois et al., 2011). Although initial studies carried out on female premutation carriers found no evidence of increased anxiety and depression, later studies reported evidence of anxiety and low level depression (Bourgeois et al., 2011). Franke et al. (1998) report that lifetime prevalence of major depression has been found among female premutation carriers of children with FXS. Bourgeois et al. (2008) questioned whether the difficulties associated with caring for a child with FXS could contribute to feelings of anxiety and depression; however they confirmed that the same group of women had also experienced these symptoms before their children were born. A study conducted by Rodriguez-Revenga et al. (2008) further suggests that the aetiology of the depression experienced by female premutation carriers may be entirely genetic, regardless of the challenges in raising a child with FXS.

2.3 Genetic Testing

With the increasing availability of genetic testing many people will learn that they, or their children, face significant health risk (Biesecker, 1998). McDaniel (2005) believes the experience of being at risk begins when a genetic condition is known to run in the family, after which, a decision is made to take a genetic test. Abrams et al. (2012) emphasise the importance of early detection and management of those at risk of FXD and have stressed that the “diagnostic odyssey” (p.4) of ongoing delays in the identification and diagnosis of FXS in infants is deeply distressing for families. Sharpe and Carter (2006) state it is the uncertainty of genetic risk that allows for inaccurate and elevated risk perception which, in turn,
heightens emotional distress. Studies have shown that it is perceived risk rather than scientific risk that contributes to difficult psychosocial and emotional responses to testing (Lerman, Croyle, Tercyak and Hamann, 2002). Todres, Galvin and Holloway (2009) add that efforts to make sense of a medical diagnosis do not align the human experience with statistical truth. McDaniel (2005) writes “It is the meaning we make of these data, not necessarily the data themselves that affects how we respond” (p. 27).

Sharpe and Carter (2006) assert that an understanding of the relationship between risk perception and emotional response is fundamental to understanding the psychodynamics of genetic testing. However, Todres, Galvin and Holloway (2009, p. 73) note that the medical system relates to ‘how’ the individual is in the here-and-now, rather than ‘who’ they are in the context of their personal journey.

Targum (1981) considers parents’ experience of their child’s genetic diagnosis as a trauma which activates the immediate use of defences. Oftentimes, denial is the first attempt made to manage feelings of loss and hopelessness. Similarly, shame or guilt may lead to anger which is then projected onto spouses or medical staff in a further attempt to bear the unbearable. In some cases, a parent may experience narcissistic injury wherein exposure to their own faults may arise. According to Targum (1981), loss is commonplace in parents’ experiences of genetic illness in their children. He stresses the importance of parents being able to successfully mourn the loss of the healthy or idealized child and describes this as “…a process of emancipation from bondage to a lost object (in this case the idealized child), adjustment to the environment in which the lost object is missing, and the formation of new object relations (or goals)” (Lindemann (1944) as cited in Targum, 1981).
2.4 Genetic Counselling

In relation to a genetic diagnosis, it is important to consider the support currently available in the clinical setting. Rantanen et al. (2008) describes genetic counselling as the practice whereby an appropriately trained person presents medical and genetic facts to individuals and their families in a way that facilitates an understanding of the meaning of the genetic information and the course of action most appropriate to the family. Evans (2006) agrees that the principle aim of the genetic counsellor is to impart complex medical information in a way that increases personal knowledge about genetic risk for the individual and their family. However, Sharpe and Carter (2006) argue that the emotional responses triggered from the process of genetic testing “impairs the communication, understanding and retention of information” (p. 53), all of which is essential to well-being. Targum (1981) adds that the anxiety provoked by a genetic diagnosis inhibits the individual’s capacity to be psychological, thus the goals of genetic counselling cannot be reached.

The professional accreditation standards of the American Board of Medical Genetics (2004) require that genetic counsellors are able to recognise the psychosocial implications of a genetic problem in the family. Furthermore, these standards require that counsellors recognise the need for a psychiatric referral where appropriate (as cited in Sharpe and Carter, 2006). Accordingly, a study conducted by Capelli et al. (2009) among genetic healthcare workers, assessing their ability to deal with individuals experiencing high levels of anxiety due to genetic test results, highlighted a significant lack of confidence in dealing with acute distress, particularly suicide ideation, due to a diagnosis of a progressive neurodegenerative condition. The study suggests that some genetic healthcare workers feel they are not adequately trained regarding the psychological and emotional aspects in their role as a genetic counsellor.
Evans (2006) however, maintains that genetic counsellors need not be psychotherapists and contends that it is possible for genetic counsellors, using the concept of non-directiveness borrowed from Carl Rogers’ client-centred therapy, to recognise clients’ psychological complexities and to facilitate their autonomous decision-making when confronting genetic risk. However, Petersen (1999) argues that the concept of non-directiveness is widely debated among genetic healthcare professionals who argue that non-directiveness is an ideal which is impossible to uphold given the value-laden medical information being presented. Furthermore, McAllister (2001) notes that the lack of a theoretical framework in genetic counselling brings uncertainty for counsellors and is therefore detrimental to the work with clients. In response, Evans (2006) recommends the use of attachment theory by way of an integrated framework so that genetic counsellors understand how the client’s early attachment style influences the counselling relationship, as well as their processing of grief and loss. Similarly, Eunpu (1997) argues that the short-term solution-focussed nature of genetic counselling would benefit more from a psychodynamic or client-centred therapeutic orientation.

In addition, Kessler (1992) raises concerns about counter-transference issues wherein genetic counsellors may associate their clients’ struggles with their own issues, potentially re-opening old wounds and reminding them of previous losses and past traumas. Sharpe and Carter (2006) support this argument by noting the client’s perception of genetic risk and suggest that results may be influenced by the counsellor’s own experience. Although Evans (2006) notes that personal therapy and supervision, which supports psychotherapy trainees with issues of counter-transference, have been introduced to some genetic counselling training.
2.5 An Introduction to Object Relations

This study seeks to explore the subjective experiences of three fragile X carriers through the lens of psychotherapy. In order to understand how a diagnosis of fragile X might manifest in the psyche, the researcher has chosen to review literature from the Object Relations School as it centres on the interplay of the inner and outer worlds.

The Object Relations perspective, as advanced by Klein and Fairbairn, moved psychoanalytic theory from a pleasure-seeking biologically-driven concept of relating to an object-seeking social concept, whereby connection with the Other is recognised as the primary need of the organism (as cited in Gomez, 1997). Central to this premise, Object Relations views the psyche as a self-organising dynamic system, wherein the Self relates to internal representations of early external relationships (Jacobs, 2012). These internal relationships, known as internal objects, are not formed entirely from, nor are they an exact representation of, early relationships. Rather, they represent aspects of external relationships in the form of feelings towards, or fantasies of, the Other. While all psychodynamic theory holds the child’s early caregiving relationships as central to the healthy development of the psyche, Jacobs (2012) emphasises the powerful role of fantasy in the creation of internal objects.

According to Gomez (1997), impingements in the oral stage of development are of fundamental importance to Object Relations theorists. The oral stage, which is based on the infant’s utter dependency and helplessness in the first year of life gives rise to primitive anxieties when needs are not met. Howe (2005) asserts that children whose caregivers are deeply unresponsive to them instil a level of fear in the child that is intolerable for the developing psyche to bear. Furthermore, when the child is left in an interminable state of unmanageable emotion due to lack of connection with the parent, segregated mental
representations of the self and others arise. If the environment is limited in terms of holding, the organism patterns itself in such way as to make sense of its world and the psyche relies on the use of defences to achieve a sense of coherency (Gomez, 1997). As defences are employed by the developing psyche, the internal world becomes increasingly livelier in the face of what is viewed as a hostile external world.

Psychodynamic activity therefore constitutes an active inner life, wherein the individual relates, both consciously and unconsciously, to internal objects as different parts of the self. Gomez (1997) adds that the internal objects are influenced by each other just as they continue to be influenced by external experience. Jacobs (2012) expands on this idea and states that although altered by ongoing reality and maturity, the early patterns of relating, laid down in infancy, continue to influence how we relate to ourselves and others, particularly in times of trauma. In psychodynamic terms, reverting back to these early modes of relating is known as regressing to a more primitive state (Jacobs, 2012).

2.6 Kleinian Theory

Klein (1959) held the development of the Self in early infancy as crucial to an understanding of the adult in maturity. For Klein (1959), the development of the Self centres on the interplay of inner and outer worlds, which are mutually dependent, and where projection and introjection, as psychic defences, operate in tandem. Klein (1959) claims that all aspects of mental life are concerned with Object Relations which have become internalised to construct the child’s inner world. Gomez (1997) describes Kleinian theory as “…a constant interplay, a recycling almost, of perception and feeling between the outer and inner worlds, so that both are experienced partially in the light of the other” (p. 35).
Klein portrays a deeply conflicted infant psyche and attributed this partly to the death instinct, wherein the infant feels a sense of annihilation from within. In an effort to rid themselves of the dread of annihilation the baby projects unbearable feelings outwards into the world, however, when met by a now hostile world the baby must project out goodness from the life instinct to counteract the bad (Gomez, 1997).

Referring to the first few months of life Klein presents two stages of development; the paranoid-schizoid position, characterized by persecutory anxiety and splitting processes, and the depressive position, characterised by the infant’s ability to integrate good and bad in the same object (Gomez, 1997).

Klein (1946) describes the baby in the paranoid-schizoid position from birth to six months, during which the infant relates to parts of the mother, one such part-object being the breast. During this time the infant manages disruptions and deprivations, such as hunger and separation, by splitting these experiences into total goodness as separate from total badness. Howard (2011) writes “when the baby is hungry he fantasizes the breast as something bad through the process of projecting his discomfort of hunger into it; in turn he feels he is being persecuted by the bad breast” (p.44). The infant longs for the ever-present breast where no destructive impulses are present and the idealizing of this breast deepens the phantasy of good and bad (Klein, 1946). Efforts to assuage the internal conflict and to make sense of the disruptions of hunger and separation, by splitting everything bad from everything good, allow the baby to begin to feel safe from attack. Splitting the experience into total goodness and total badness allows the baby to introject the goodness which is needed to begin to develop a sense of self. In this way, the developing psyche is further protected from the intolerable feelings of loss (Klein, 1946).
In time, the transition from splitting to integration of both good and bad signifies the infant having arrived at the depressive position (Gomez, 1997). Klein (1946) describes the depressive position wherein the baby learns to integrate internal and external experiences, good and bad, and recognises sadness and loss where grief overtakes anger. For Klein (1946) the depressive position was central to the healthy development of the psyche.

Jacobs (2012) adds that the psychic defences, employed in Klein’s paranoid-schizoid position, allows the infant to continue relating to the Other for whom they have ambivalent feelings and for whom they depend on for survival. Gomez (1997) adds that where projection and introjection is employed, denial is also in operation. She describes this as “the superficial plastering over...of the persecutory experience of frustration with the imagined experience of satisfaction” (Gomez, 1997, p. 39).

2.7 Fairbairn’s Theory

Like Klein, Fairbairn (1946) asserts that the primary aim of the organism is to seek connection with the Other. If, however, the libidinal connection with the object is broken, in the form of neglect or abandonment by the caregiver, the infant experiences what Fairbairn (1946) calls a primary trauma. In an effort to continue to manage the relationship, on which it is wholly dependent for survival, the infant relocates the traumatic experience inside. The ego splits the experience into a tolerable ideal-object and an intolerable bad-object. The ideal-object reflects how we like others to be all of the time, while the intolerable object is further split into an exciting-object and a rejecting-object, representing the aspect of the Other that we painfully yearn for and need, and the aspect of the Other that has painfully rejected and abandoned us. Attached to the exciting-object is the aspect of the Self that represents intense
neediness, and attached to the rejecting-object is the aspect of the Self that becomes contemptuous of this neediness (Gomez, 1997).

Fairbairn (1943) describes the introjection and repression of bad-objects as “taking upon himself the burden of badness… and in doing so he is rewarded by that sense of security which an environment of good objects so characteristically confers” (p. 65). While the child continues to relate to the external object and creates for itself a sense of security, the internal object-relating intensifies and guilt and shame operate within the child that has now become ‘bad’ (Fairbairn, 1943). Gomez (1997) states that the intense and powerful dynamics of the internal objects and repressed feelings give rise to persecutory guilt and rage resulting in the despising of oneself or others. According to Fairbairn (1946), the process of internal split, known as the schizoid position, structures the development of all personalities.

2.8 The Lost-Object

Gomez (1997) writes that “our most basic anxiety is separation anxiety; the dread of the loss of the other, on whom our physical and psychological survival depends” (p.58). Bowlby (1988) asserts that the objective of the attachment system is to propel the infant towards its caregiver to seek protection and ensure survival. The infant’s attachment system is triggered when discomfort due to separation is experienced and the baby activates the caregiver to respond in a way which meets its needs. Depending on the reaction of the caregiver, infants will organise their experience as mental representations of memories, feelings and behaviour thus developing their knowledge of how relationships work, particularly with attachment figures, in times of need. Van der Kolk (2005) writes that “the security of the attachment bond mitigates against trauma-induced terror” (p.4), as in the loss of the Other. If the experience of the early attachment relationship has been good enough, the infant internalises
the good-enough object, integrates somatic expression of emotion and develops the ability to regulate feelings, allowing for mentalisation and reflective capacity (Gerhardt, 2004).

Children who experience early loss, however, do not experience this sense of protection resulting in insecure avoidant and ambivalent patterns of attachment. This creates distorted mental representations of themselves and others wherein unconscious adaptive strategies are used to manage their anxieties (Bowlby, 1988). He stresses the importance of helping the child to mourn the loss of a parent so that the inner representation of their relational world does not become distorted.

Freud (1917) described the states of mourning and melancholia that arise in response to the experience of loss. In his essay Mourning and Melancholia, he refers states that:

“Mourning is commonly the reaction to the loss of a beloved person or an abstraction taking the place of the person, such as fatherland, freedom, an ideal and so on. In some people, whom we for this reason suspect of having a pathological disposition, melancholia appears in place of mourning” (p. 203).

Due to the painful dejection of their loss, the mourner experiences a cessation of activity in the world and a loss of the capacity to love another as this would mean neglecting the memory of the deceased. In the state of mourning, the loss is identifiable to the mourner in that they know who and what is being mourned. According to Freud (1917) mourning is time-limited and its function is to detach the survivor’s hope and memories from the lost-object; in this way “…reality-testing has revealed that the beloved object no longer exists and demands that the libido as a whole sever its bonds with that object” (p. 204).
When the work of healthy mourning is complete, the original lost-object has been abandoned in death and the libido has been displaced onto another object. This allows energy to be reinvested in life and the ego to remain intact (Freud, 1917).

The melancholic, in their grief, remains unclear about what has been lost even when they know who has been lost. By comparison to the mourner, the melancholic experiences the same lack of interest in life; however there is an added lowering of self-regard wherein self-reproaches are commonplace. The interior script of the melancholic states that they are truly worthless and their deeply-felt sense of inadequacy reflects an ego that is wholly deprived (Freud, 1917). The tendency for the melancholic to admonish themselves before others is a “consequence of internal work…that is devouring his ego” (p. 206). Unlike the mourner, whose libido is displaced onto another object, the melancholic’s libido, through the disappointment of the lost-object, is transferred onto their own ego. The insults and criticisms directed at the self are therefore meant for the lost-object who has been internalised. In this way, the melancholic identifies entirely with the lost object. Freud (1917) describes the internalisation and identification with the lost-object as when “the shadow of the object fell upon the ego” (p. 209). Freud (1917) highlights the ambivalence that arises out of feelings of the love for and the fear of the lost-object. Affection and hatred for the one who has abandoned us serve to further complicate the grieving as anger and guilt arises to meet the loss.

Finally, Leader (2008) describes reactions to the death of a loved one where individuals, in their grief, displace their anger onto doctors, partners and family members without realising that the feelings are meant for the deceased. Leader (2008) maintains that human emotions are all too often pathologised wherein anxiety and depression are understood as clinical
symptoms with a definite organic aetiology. He suggests that these symptoms cannot be fully understood without the acknowledgement of unconscious processing of the experience of loss. To use the term ‘depression’ is to attempt to refine and sanitise the complex and multifaceted response to loss (Leader, 2008). Likewise, ‘anxiety’ which is nowadays treated as a separate psychiatric illness is in fact an appropriate human response, encompassing a deep sense of fear, to the experience of loss (Leader, 2008). According to Gomez (1997) depression and anxiety may arise out of prolonged mourning where new attachments have not been made. Therefore it is necessary that feelings of sadness, guilt and anger which stem from loss are accepted so that the mourning process can be completed.

2.9 Conclusion

Genetic illness is often experienced as a trauma which affects the Self deeply and profoundly (McDaniel, 2005; Targum, 1981). Previous studies have highlighted anxiety, depression, loss and guilt among the challenging emotions experienced by fragile X carriers. Despite this, there is a remarkable paucity of research in relation to the implicit meaning women have made of their fragile X carrier status. In exploring the psychological support available to genetic carriers through genetic counselling, the research suggests that the profession struggles to meet individuals and families in their struggle to understand and integrate their new genetic information.

In order to examine the subjective experiences of fragile X carriers the researcher chose to review literature from the Object Relations School which understands emotional difficulties experienced in adult life in light of the early development of the Self. Object Relations pays particular attention to the degree to which the psyche uses defences to manage the anxieties brought about by a traumatic experience and in particular when feelings of loss are evoked.
By focusing on the inner life as it is changed by, and as it changes, the outer life, the implicit meaning of life’s experiences remains the focal point of Object Relations theory. An understanding of the Object Relations framework is therefore necessary to offer a psychotherapeutic exploration of the experiences of fragile X premutation carriers.
Chapter 3 - Methodology

3.1 Research Design

Qualitative research is an inquiry process whereby the researcher seeks to develop a deep understanding of peoples’ lived experiences and how they make sense of these experiences (McLeod, 2011). Qualitative research is neither prescriptive in its approach nor does it determine what is true “…since to limit the criteria for truth would mean restricting the possibilities for knowledge…” (Yardley, 2000, p. 217).

Unlike quantitative research methods, which abstract from and measure static properties of larger populations, qualitative research is concerned with the qualities of the individual experience (Yardley, 2000). However, Yardley (2000) rejects the dichotomous framing of quantitative versus qualitative research, particularly in health psychology, and insists that qualitative research alongside quantitative research offers greater insight for health and caring professions. Likewise, Todres, Galvin and Holloway (2009) argue that the valuable information elicited by qualitative studies be used to inform and expand the healthcare service which so often reduces the body to biological processes resulting in a loss of meaning for the patient.

Thus, the researcher believes that a qualitative approach, using Interpretative Phenomenological Analysis (IPA) (discussed in the Section 3.5) which upholds the individual narrative within a homogenous participant sample, is most suited to a psychotherapeutic exploration of the experiences of female fragile X carriers.
Furthermore, qualitative research which seeks to illuminate details of peoples’ lived experience, and seeks to understand their meaning-making capacities within these experiences, must be conducted in such a way as to ensure validity and quality of the research. To establish validity, Yardley (2000) suggests the research meets the following criteria: sensitivity to context, commitment and rigour, transparency and coherence and impact and importance. The researcher’s adherence to these criteria is demonstrated in the sections below.

3.2 Participant Sample

McLeod (2011) suggests that a small number of participants are best suited to a qualitative study which seeks to identify the essence of the lived experience. Smith and Osborn (2003) suggest using approximately three participants for a qualitative study where IPA is used to elicit a nuanced analysis of individual narratives. Further, the IPA researcher who employs purposive sampling selects participants according to criteria relevant to the study. In this way, the group is considered a homogenous sample (Smith and Osborn, 2003). A small homogenous sample allows for greater depth and complexity and offers the qualitative researcher the opportunity to explore in detail a particular area of interest.

Keeping in mind Smith and Osborn’s (2003) recommendations, three participants were selected for this study. The inclusion criteria stipulated that the participants be women who had been diagnosed as a fragile X premutation carrier no less than three years before the time of the study. In this timeframe, the researcher believes that the participants would have made sense of their new genetic information. Although it was not specified in the inclusion criteria, each participant is a mother of a child diagnosed with FXS. The sample is described in Table 1.
Participants were recruited via the Irish Fragile X Society (IFXS). When an email to the IFXS via their website, detailing the nature of the study, went unanswered the researcher contacted the IFXS via their Facebook page. The Chair of the IFXS expressed interest in the study and offered to recruit three potential participants, thus snowballing was used in the recruitment process.

Three individuals identified via the IFXS were invited to read the Participant Information Sheet (Appendix A) and the Consent Form (Appendix B) which were sent to them by email. All three individuals replied expressing interest in the study and the interviews were arranged by organising a suitable date, time and location.

Table 1 – Participant Sample

<table>
<thead>
<tr>
<th>Participant</th>
<th>Years since FX carrier diagnosis</th>
<th>From whom did participant inherit the gene</th>
<th>Children</th>
<th>Age when FXS diagnosed</th>
</tr>
</thead>
<tbody>
<tr>
<td>Laura</td>
<td>3 ½</td>
<td>Father</td>
<td>1. Son (Jack) – FXS&lt;br&gt;2. Daughter – asymptomatic</td>
<td>3 ½</td>
</tr>
<tr>
<td>Catherine</td>
<td>11</td>
<td>Mother</td>
<td>1. Daughter (Rachel) – FXS&lt;br&gt;2. Daughter – asymptomatic</td>
<td>2 ½</td>
</tr>
<tr>
<td>Sarah</td>
<td>7</td>
<td>Father</td>
<td>1. Son (Frankie) – FXS&lt;br&gt;2. Son – asymptomatic&lt;br&gt;3. Daughter – asymptomatic</td>
<td>3</td>
</tr>
</tbody>
</table>
3.3 Pilot Study

A pilot study was conducted with an interviewee who is a female carrier of another x-linked genetic condition and who is known to the researcher. Reservations regarding the familiarity of the relationship were initially noted by the researcher who became aware of feeling protective of the interviewee when deep feelings of loss were evoked for her. However, the interviewee later spoke of her regret that her father, from whom she inherited the gene and whom she lost due to a condition related to the gene, was reluctant to discuss her genetic risk in his effort to protect her. The pilot study proved useful as the feelings evoked in the researcher were reflected upon and encouraged the researcher to consider more fully the dynamic process of engagement of a semi-structured IPA interview. Furthermore, the researcher was reminded of, and kept in mind, the sensitive nature of the fragile X carrier study. The pilot interview thus helped to define the duality of roles: of researcher, managing the clarity of the dialogue, and of therapist, providing safety and containment.

3.4 Data Collection

Smith and Osborn (2003) note that semi-structured interviews are the ideal method for collecting data that will be subject to IPA. Researchers conducting semi-structured interviews are guided, rather than dictated, by the use of open ended and non-determined questions. According to Smith (2004), researchers are free to probe participants’ responses and change the sequence of questioning thus encouraging a unique and unpredictable story to emerge. The researcher does not hold the expert position. Rather, they establish a rapport with the participant which facilitates a dialogue that elicits rich and descriptive personal accounts (Smith and Osborn, 2003). So that a sense of safety is created that allows the participant’s story to emerge, Kvale (1996) states that the researcher must strike “…a delicate balance
This study used semi-structured interviews comprising of thirteen open-ended questions (Appendix C) which were informed by the literature review and reviewed by the academic supervisor after which some proposals were adopted by the researcher. Due to the lack of psychotherapy research on the subject, the researcher felt it was important that the questions be framed broadly and openly so that participants’ perceptions and views, that may hitherto not have been expressed, were allowed a voice.

The questions covered the following themes:

- How they (the participant) have made sense of their new genetic information in the time since diagnosis;
- How they managed their feelings at the time of diagnosis;
- Impact of their diagnosis on their sense of self;
- Impact of their diagnosis on their relationship with others;
- How they relate to the FX gene;
- How they manage genetic risk.

The duration of the interviews varied greatly. Catherine’s interview lasted 1 hour 20 minutes, Sarah’s lasted 36 minutes and Laura’s lasted 16 minutes. Interestingly, the researcher notes that the interview times are proportionate to the years since diagnosis, perhaps an indication of how the participant may have been able to process the genetic information in the time since diagnosis.
3.5 Data Analysis

Keeping in mind the dearth of psychotherapy research on the experiences of genetic carriers and given that most of the research on this topic has come from the disciplines of genetic medicine and health psychology, the researcher finds the Interpretative Phenomenological Analysis (IPA) model advocated by Smith and Osborn (2003) most appropriate for this psychotherapeutic exploration into the experiences of female fragile X premutation carriers. The aim of IPA is the in-depth exploration and detailed analysis of personal experiences and perceptions, thus capturing the true meaning of the personal and social world of the individual (Smith and Osborn, 2003).

By adopting a phenomenological approach to data interpretation the IPA researcher is wholly concerned with the experience of being human, which, according to Charon (2006) is absent from medical research (as cited in Todres, Galvin and Holloway, 2009). Smith (2011) confirms that illness experience has increasingly lent itself to IPA. This study uses IPA to present three participants whose rich and varied experience highlights the complexity of genetic illness and offers a vivid portrayal of their journey through the medical model.

Smith and Osborn (2003) describe the interpretative relationship with the transcribed text as a “two-stage interpretation process or double hermeneutic” (p. 53). This means that IPA is a dynamic process involving a researcher who is actively interpreting personal meaning and sense-making in order to fully understand the experience of the participant. Both empathic and questioning hermeneutics form part of the interpretative process allowing for “a richer analysis and…greater justice to the totality of the person, ‘warts and all’” (Smith and Osborn, 2003, p. 54). Within the exploration of meaning and sense-making, this in-depth qualitative approach recognises a sensory, cognitive and affective individual who sometimes experiences
difficulty in disclosing what they are thinking or feeling. As a result, the IPA researcher remains sensitive and curious and is able to sustain engagement with the text and the process of interpretation (Smith and Osborn, 2003).

The researcher immersed herself in the transcription of the interviews through repeated listening to the audio transcripts, manually transcribing them, and by reading the transcripts a number of times thereafter. In this way, the researcher hoped to capture and understand the meaning of the lived experience of the participant (Smith and Osborn, 2003). According to the idiographic, inductive and interrogative features of IPA laid out by Smith (2004), the researcher employed in-depth and complete analysis of one transcript before moving to each of the others. The researcher remained open to the possibility of unexpected themes emerging which did not fit any particular hypothesis. As themes began to emerge (Appendix D) in each of the transcripts a cross examination was carried out which saw a number of themes emerge across the transcripts. The process of checking for similarities and differences led to the emergence of three major themes which were further lent to interpretation through an Object Relations perspective. Verbatim extracts were used to illustrate the themes therefore grounding them in the actual words of the participant.

3.6 Ethical Considerations

The research proposal was submitted for approval to the Department of Psychotherapy Ethics Committee at Dublin Business School in April 2014. Approval was granted without conditions albeit the sensitive nature of the study was noted and it was recommended that the researcher remain mindful of this.
In keeping with Kvale’s (2007) guidelines, the researcher provided each participant with a Consent Form as well as a Participant Information Sheet on the study. The information was sent by email in advance of the study and it specified the voluntary nature of the study, the right to withdraw at any time, the potential risks of participating in the study and the participant’s right to disclose or withhold information at their own discretion. Participants were assured of their anonymity by the researcher’s use of pseudonyms on all documents for all those mentioned in the narrative. Confidentiality was ensured by protecting all documents with passwords known only to the researcher and these documents will only be stored on the researcher’s computer.

At the beginning of each interview the researcher read out the Participant Information Sheet and the Consent Form with the participant. When participants indicated that they were satisfied with the content both the participant and the researcher signed and dated two copies of the Consent Form.

Due to the sensitive and personal information imparted, the researcher checked with the participants at the end of the interview to ensure that they were sufficiently grounded. The researcher reminded them that should they wish to see a psychotherapist due to feelings that may arise after the interview, they could contact a number of psychotherapy practices, details of which were provided on the Participant Information Sheet.
Chapter 4 – Findings

4.1 Introduction

This chapter examines the research data gathered from individual semi-structured interviews that took place with three participants: Laura, Catherine and Sarah. In the interviews, participants were asked to describe their experiences as a fragile X premutation carrier. Although it was not specified in the inclusion criteria, each participant is a mother of a child diagnosed with FXS. Consequently, this diagnosis prompted each participant to undergo a genetic test which confirmed their status as a fragile X carrier. The traumatic nature of learning of their childrens’ diagnoses, rather than the confirmation of their own diagnosis, features predominantly across the interviews.

Upon analysing the interview transcripts, three main themes emerged which will contribute to a psychotherapeutic understanding of the experiences of female fragile X premutation carriers. The themes presented are ‘Suspended in The Unknown, ‘Fragile X: A Threatening Presence’ and ‘Primitive Defenses’. Themes are illustrated below using excerpts from the verbatim transcripts which are subjected to interpretation by the researcher using IPA.

4.2 Suspended in The Unknown

FXS was not known to the participants during the initial investigations into their childrens’ developmental delays. Therefore, in each case shock and denial permeates the experience of what was the sudden presentation of the genetic diagnosis of FXS. A further sense of uncertainty manifests, in different ways, throughout the participants’ interaction within the medical framework, as they recall their struggles to make sense of their new genetic information.
When asked how she became aware of her status as a fragile X premutation carrier, Sarah refers to her son’s diagnosis which marked her initial discovery with FXS. Her recollection below illustrates the abrupt and unexpected arrival of FXS which saw her move from the secure and familiar surroundings of her sunny garden to darker more unfamiliar territory:

Well eh…y’see I still you know have to keep going back to when my son was diagnosed really because the consultant just rang us up and my husband’s German so he…and I remember it was a sunny July afternoon and eh so he just sort of said it said it on the phone to him and my husband didn’t y’ know, kind of…kind of y’know didn’t really know what he was talking about but then he put him on to me and ehm [silence]…so it was just y’know it was just I didn’t know, I didn’t know what it meant y’know.

Sarah’s initial sense is that her German husband does not understand what is being said to him in english, however, as she takes the phone, the information being imparted to her remains incomprehensible as she struggles to make sense of it herself.

Here, she likens the disruption of the diagnosis, received in a phone call from the geneticist, to the experience of death. Sarah had known death due to the loss of her father when she was seven and learning of her son Frankie’s diagnosis she describes a similar feeling, yet the loss she describes this time seems to have a different complexion not entirely known to her:

It was devastating! It was just so…I mean that, that I can still see so clearly like where I was on the day and it was a beautiful July afternoon and the children were outside and it was sunny and you know just sort of awful news like you know somebody was dead...almost. So
that was bad. Like a death. You know it... kind of everything changed but nothing changed, you know cos he’s still the same you know and yet everything changed so ehm yeah.

Sarah’s shock of learning that her child had been diagnosed with FXS was deepened by the fact that she was unaware that the test had taken place. Although she refers to FXS later in the interview, Sarah does not name it when remembering the initial diagnosis, revealing a sense of the unfamiliar:

…it was completely out of the blue there was nobody else in the family and I...I mean I wasn’t told he was being tested for it or anything no genetic...and I remember signing something but being told it was just sort of just to rule out stuff so I certainly didn’t know or wasn’t aware that he was being tested for it so when he was diagnosed that was the main shock because I didn’t know what it was and it was on the phone...

I: So, if I understand correctly, you were not aware that they were testing Frankie for FXS?
Sarah] No, now I must...I did sign something so presumably it would have said it but he wasn’t saying ‘now read it’, you know it wasn’t like you ‘now read this with me’

[referring to the Participant Information Sheet and Consent Form that the researcher and Sarah had reviewed together before the interview].

While Sarah’s hazy recollection of the possibility of having given consent may indicate her reluctance to accept her child’s genetic condition, her description of the interaction with the medical consultant illuminates their failure to remain in contact, thus adding to her later distress. She offers further confirmation of this failure when she compares the
communication between herself and the researcher in a more favourable light, and one which
seems to have facilitated her understanding of what this study entailed.

Although Laura and Catherine were aware that their children had undergone testing for FXS,
the positive test results were also unexpected. Laura confirms that fragile X was not known to
her family and, like Sarah, she is unable to name it:

Absolutely nothing at all nobody in the family had...for generations that we knew of had
anything at all.

Despite having researched FXS in the interim between testing and results, Catherine
describes her denial and shock upon receiving her daughter’s diagnosis:

It was with my daughter’s that the geneticist came back and said ‘Well you’re daughter’s
came back with something’ and before she even opened her mouth again I said ‘don’t say
fragile X, don’t say fragile X, don’t say fragile X, don’t say fragile X’ like panicking...cos
she’d left it so long that I just done so much research we just parked fragile X saying it
couldn’t be it-could-not-be fragile X because it has never appeared in our family, it’s not, it’s
just not in our family, like how could this gene just suddenly pop up?

Descriptions of the participants’ interaction within the medical framework suggest that the
trauma of the diagnosis was compounded by their experience of not being met or even
responded to in their fearful state of unknowing. Like Sarah, Laura’s description of the
emotional impact of the lack of communication and the lengthy delays within the medical
framework communicates a sense of helplessness that left her in a state of fearful uncertainty:
when we got the diagnosis she did say about Jack that he would never live independently and she said he might never be able to take public transport or have a bank account and when you’re told that when your child is four, it’s kinda devastating, we didn’t know what we were supposed to do... we were literally on wait lists all the time...for the first two years. There was no central point at all, if, looking back the consultant had have said to us ‘he has fragile X and this is what he needs, he should be getting this this and this’...we were referred to the early intervention team and that’s so fragmented there’s a high turnover of staff that you’re ringing these people and they’re saying call back in a few months’ time and it’s all wait lists.

In a similar vein, Catherine shares the belief that lengthy waiting lists served to prolong her anxiety:

...here they just keep you waiting and waiting and waiting which causes a lot of anxiety and stress... that waiting for people who are naturally anxious [laughs] is horrible...

She describes the impact of the numerous appointments and the interaction with the medical system as if on a journey without any sense of direction. Her vivid portrayal of the clinical setting illustrates her deeply felt sense of isolation, as if frozen in trauma and suspended in time and space:

It wasn’t nice at all the journey...it didn’t feel like 2005. I felt like I was in the 18th Century. I don’t know where I felt. It did not feel like 2005, or 2007/2008 yeah she was born in 2005. It just didn’t feel like modern or with it, I felt like I was surrounded by Old-fashioned Ireland, dinosaurs if you like...at my diagnosis it was equally as dark and as cold. So I had done my
research so by the time I met with the genetics counsellor I remember going in there and it was February and again like I don’t know why but I suppose, maybe with traumatic events in life like they’re always darker, I felt…like it was probably just a regular waiting room but it felt dark and even like visually I can still see like it was cold and dark. And again like, again I just felt like I was in some sort of flippin’ time-warp.

4.3 Fragile X: A Threatening Presence

The meaningless dynamic of the unknown, without any contextual elements, created a sense of fear which facilitated a mental representation of fragile X as a threatening presence. Fragile X was construed as looming in the background and possessed the capability of launching an unexpected attack. Fragile X, with its capacity to devastate, threatened survival and therefore evoked feelings of loss.

Here Sarah describes her own and Frankie’s diagnosis similar to an ambush against which she tried and failed to defend:

...y’see, I don’t think you can isolate it because it would have started with him...getting the the...the results...ehm because it was completely out of the blue there was nobody else in the family...

Laura also portrays fragile X as having the potential to strike:

...we were literally in the room with the genetic counsellor, she was lovely, she went through everything, the family tree and everything and how it would work but it still doesn’t hit you and have time to settle in, sink in. [Jack’s diagnosis]
Where I would have worried about it implicating was if I wanted more children and how would it affect them so that was...that’s where it would have hit me, that aspect of it...

Laura’s diagnosis

As the interviews proceeded it was apparent that the place occupied by fragile X in the participants’ inner world became more threatening than the external threat posed by the unstable gene. Here, Catherine appears to confirm, at least unconsciously, that the medical risks of fragile X are more manageable than the threat from within:

There’s still not enough awareness about fragile X and what it is what it, what carriers...I mean OK we know that auto-immune disorders are common in carriers, we still don’t have any definite answers, there’s health concerns that I have but when I go to GPs still it’s not addressed and that’s...you don’t know like it’s not a degenerative condition, well apart from FXTAS, which is like it’s neurological so that the guys well and women, the ladies who get FXTAS which is not unpleasant but for younger carriers that just have thyroid issues or...well maybe anxiety is the big predominant feature that’s causing the whole load of problems, who knows?

Catherine portrays fragile X as a more sinister presence lurking in the background with the capability of multiplying as if to engulf the family:

...so there’s me going...focused on, focused on ironically genetics on his side whereas I had something lurking in my genes...

...I remember saying to him ‘this has just opened a can of worms for the whole family’.
Similarly, Sarah describes her struggle to make sense of the diagnosis and can be seen to separate off what she calls ‘the genetics of it’ and ‘the actual fragile X’ from fragile X with a specific nature that can work its way through the family threatening survival:

...I remember he didn’t really say much about Frankie it was more just the genetics of it and then about then sort of suggested that if we ever wanted to have more children that ehm you know you can tested and you know some people you know if they have it they can go abroad for a termination and that was all he really said and you know that was really it.

It was...it was devastating, it really was because first of all I didn’t really understand it, what it meant, I mean I still have...people ask me how it works and...I googled it first the actual fragile X and it was so negative...

...this cousin had just moved in with her partner and ehm...so she was over and I said ‘just so you know this is the way it works’...

Further, a medically confirmed reduced risk of fragile X does nothing to ease the anxiety which comes from the fragile X-enemy who is lying in wait, ready to strike Catherine’s family:

...so it’s just lying there in the genes, my sister is, let’s say, an intermediate carrier...you know the CGG repeats are around the 40s. She was anxious about you know when she was having her baby and her husband kept saying, ‘you’re intermediate it’s not gonna happen, it’s not going to expand, it’s not going to expand!’ She was like ‘what if I’m the first, what if I’m the first?’
Although recent medical research suggests to Catherine that she is less likely to have a child affected by FXS, than first indicated, her internal world reflects the threat of fragile X where the odds are less certain. Here she explains her decision as if to cut her losses against a crafty opponent in a game of chance:

*If I’d known now what the research is telling about the AGG interruptions so y’know it’s not just enough to have your CGG repeats and even though the tables say that I’m not a 50/50 chance cos my repeats are in the 70s so I would have about a 30% chance of having a child affected…still wasn’t…even a 30% chance still wasn’t a gamble I was willing to take, a role of the dice as we say… so that was the big thing it stopped the family planning, I wasn’t going to have any more children.*

Further, Catherine’s description of fragile X being ‘rare but not rare’ suggests to the researcher that the statistical occurrence does not match her terrifying feelings of isolation that felt so rare as if she were entirely alone in her feelings. In an effort to establish a connection she found herself reaching out to work for the charity.

*...it’s rare but it’s not rare but the sense of isolation was really horrible, actually it was terrifying the sense of isolation…those feelings are what prompted me to go on and work for the charity.*

Catherine states explicitly how fragile X robbed her of her son and, with that, her ideal family:
...when I found out my second child was a girl I said ‘oh I’m gonna be back’ I said it in the hospital before fragile X before I ever knew that I had fragile X I said that I was gonna be back for my boy [soft tone] and I was convinced like that I wanted...I had this idealistic notion that I was going to have two girls and two boys well fragile X just [harsh tone]...so that was the big thing it stopped the family planning, I wasn’t going to have any more children.

Likewise, painful feelings of loss are evoked for Sarah wherein no amount of medical reassurance helps to soothe her anxiety. Her rich description of receiving Frankie’s diagnosis illustrates feelings of loss which are implicitly known to her, feelings which can be attributed to her seven year old self who had lost her father:

...getting a phone call in the middle of the afternoon to say that you know that your child has fragile X and you know not knowing anything about it and I had seen actually a programme the night before or I had seen it advertised and it was about some chromosome thing where the child is perfect and you know then at seven they just get really sick and die so I didn’t know what it was and that’s all I could think of so I was going ‘is he going to die?’ And he was going ‘no, no, no he won’t, it’s just you know after Down syndrome it’s the second most common inherited intellectual disability and it’s a big range...’ and you know he could have been telling me he got the flu you know...like you know, like you know when people die and you know that’s all you can think about and you just cry, it was like that. It really was. I suppose that’s part of the process.

For Laura, loss is acknowledged but quickly defended against by focussing on her son’s achievements. In addition, humour is used to defend against the experience of future loss:
...when we got the diagnosis she did say about Jack that he would never live independently and she said he might never be able to take public transport or have a bank account and when you’re told that when your child is four, it is kinda devastating so I suppose all that but now he’s exceeded things that we’d never thought so there is...y’ know every day brings us something new.

I: So would you say your diagnosis impacted on your plan to have more children?

[Laura]...Well I was still recovering from the other two! [laughs]. Ehm, it would definitely be a factor for consideration, yeah.

4.4 Primitive Defenses

Defense against the anxiety arising from the turbulent fragile x diagnosis is seen to varying degrees across the interviews. The strength of the regression sees Catherine split her experience of others, and of herself, into good and bad. This is particularly evident as she and projects her intolerable feelings onto a medical system that she feels is entirely bad. Laura, on the other hand, continues to relate to a medical system she finds satisfactory while introjecting the frustrations of that same system. Sarah’s use of denial is evident throughout her interview. Significantly, two weeks after the interview in a phone call to the researcher, Sarah disclosed the impact of her own diagnosis, denied at the time of interview, which illuminated feelings that were disavowed after the early loss of her father.

Catherine defines herself as a split subject by making the distinction between the good dominant Spanish genes inherited from her father and the weaker Irish genes she inherited
from her mother. She accepts the faulty gene as part of herself while at the same time wishing to reject it.

…when she married my Dad they were saying ‘it was good you went outside the gene pool’ because he’s like Spanish, you know and it was really good and whoever said that to her was right because I feel that sometimes the Spanish side of me is more dominant than so I’m a Spanish x and an Irish x and I feel sometimes that the Spanish x would be more dominant than the Irish x which is the fragile x. So I suppose yeah, I call the fragile x the Irish x and Mom always had this sense that I kind of rejected the Irish side of the family and then I suppose then fragile x made me probably cement it further like.

I mean she’s be a bit more…very anxious, very OCD, very religious and like her focus is my daughter…is a gift and a blessing and you know this kind of stuff, the old-fashioned, you know the Irish thing...

…it’s just who I am [sounding tired]…you know, what can I do, it’s who I am. It’s my genetic make-up, part of me, who I am.

Moreover, Catherine is intolerant of the vulnerability that she associates with the ‘fragile’ gene. Evidently, Catherine demonstrated further disdain for vulnerability throughout the interview; firstly, when reading the Participant Information Sheet, Catherine seemed amused at the idea that the researcher would ensure participants were aware that the interview may evoke difficult emotions and she questioned if the Ethics Board had enforced this. Secondly, towards the end of the interview she split carriers into those who help other fragile X carriers and those who have a victim-mentality.
Here, Catherine describes intense feelings of isolation despite having her husband by her side. As she describes her guilt at having passed the fragile x gene to her daughter she begins to communicate her frustration with the medical system she comes to view as inherently bad.

_I had my husband by my side and I felt like I was alone, like I was…in this horrible vacuum alone, it was just me in a bubble, my own and it was all my fault. I started to blame myself for giving her this. And I was annoyed like that…what were they doing testing my husband I mean he had no reason to be tested…I was the cause of her fragile x if you like…so so yeah it was cold, I wouldn’t recommend seeing a genetic counsellor in Ireland, it was just old, not warm, horrible cold experience._

Laura describes the impact of her son’s diagnosis as not having affected her relationship with her husband:

_…when his meltdowns were really bad so [inaudible] stress between us but otherwise no we try and make the most of it [laughs]. So it’s the day-to-day managing._

Likewise with her father, from whom she inherited the gene, the status quo has remained:

_Yeah, no we don’t really even talk about it and we focus more on Jack than anything we just kinda get on with it…we talk about it if we need to but again we focus on Jack and getting him the day-to-day what he needs._

_I: So it hasn’t impacted on your relationship with your Dad?_
Although she had spoken of a fragmented medical system with its long delays and lack of information, she aims her frustration away from the medical system towards herself so that she can manage day-to-day and get what she needs. Here, her self-reproaches are evident:

...we just didn’t know I suppose, looking back now we wasted two years on wait lists...we didn’t know what we were supposed to do, I suppose we should have... I suppose it’s about getting on with it, rather than...I do regret hugely the first two or three years that we didn’t do, we didn’t know what we should be doing. I already had this discussion with my husband two or three nights ago that it’s lack of education on our half or not knowing but...eh...there’s an awful lot more available in the last few years, knowing where to go as well and parents are a huge support too so.

On the other hand, Catherine’s feelings of fear, anger and guilt encourage a reorganisation as if to take up combat:

*When I got the diagnosis I said ‘OK now we need to act, act, act!’* [sounds angry] ...I was annoyed as well because you know we were in the system and the system had already failed us...so I suppose I was in fight mode there because I was going ‘aaaargh have to get her help’...there’s guilt again, Mommy’s guilt, Irish guilt whatever it is, that I hadn’t done enough. Because she needed help and they kept telling me...they kept dismissing me with ‘it’s just behaviours, it’s just behaviours, it’s just behaviours’, uuugh that it was just her personality...I was assured that the system was good once you were in it but the system isn’t good once you’re in it, it still isn’t good once you’re in it.
Her vivid account of a genetic consultation illustrates her projections which suspend her in stasis and highlights a parallel process wherein the medical system splits off that which is unacceptable. The potent mix of medical and historical factors are observed wherein the institution holds the shame projected out of a society that fears being less abled. Here, Catherine describes a traumatic experience:

So going into this dark, dark institution was horrible because I was scared, fear of the unknown and having to bring my little, my perfect little girl into these places...scared the crap outta me.

The paediatrician stupidly said to me ‘you don’t have to tell anybody’ and that’s like big old Ireland kinda thing...I was going ‘what does she mean I don’t have to tell anybody? [sounding angry] I’m just after finding out this genetic information of course I have to tell people!’ She was like ‘no you don’t have to tell anybody about this it’s your own information’. But that’s not my nature and plus I felt it was nearly like shameful Old Ireland like you know these kids could be put into institutions you know and you forget about them.

They’re still, they’re still institutions...they’re trying to make it modern and move away from them but it’s still...you feel the history...she was only a toddler at the time and she banged her head on the door by accident... the doctor at the time said ‘Oh you’re in the right place’ [whispering]. I was like somebody get me out of this place, like some kind of bad, bad story and it was almost surreal like ‘is this really happening’? Is this really happening?

Oh my God so much shame! And I was annoyed at myself because say when correspondence would come in from the institution where we had our appointments and it was on the envelope and I was ‘uuargh this is stamped like uuargh the postman knows, the neighbours know’, I was like ‘Oh my God I’m getting completely sucked into this’. 
Laura maintains her satisfaction with the genetic counsellor, while taking the hit herself:

...she was lovely, she went through everything the family tree and how it would work but it still doesn’t hit you and have time to settle in, sink in.

... we’re on top of it now I think we’re getting there but...coming back from the diagnosis yeah it was like a big black hole [speaks very quickly and laughs].

Finally, in making the distinction between the UC Davis research centre in the US and the Irish medical system, Laura implicates herself in the failure of the Irish system whereas in Davis she allows herself to be separate. She confirms, albeit unconsciously, the introjection of the bad Irish experience in her efforts to achieve coherency:

In Davis they did in 2 ½ days what we had tried to get done in 3 years here, 3 years and only 2 days over there. It was definitely worth the trip.

Sarah’s defense had been to deny her son’s diagnosis in the hope that it would go away, thus replacing the intolerable feelings with more manageable ones. Furthermore, to accept support at that time would mean to invite the unbearable feelings back.

I was kinda hoping that still there would be a mistake but ehm...it was pretty devastating. There was no support. Now that said I have to say that you know I would have been given the...the...the leaflets for the fragile x society...to be honest with you I didn’t really want to go there so you know cos...if you do that then you’re accepting it and it can’t go away whereas if you don’t really give in to it, it might go away.
She struggles to understand her feelings of grief around her son’s diagnosis:

*Yeah and like nothing has changed so it isn’t like a death in that somebody’s there and they’re gone. It was very hard, very, very hard [voice fragile] like you know, like you know when people die and you know that’s all you can think about and you just cry, it was like that. It really was. I suppose that’s part of the process [silence].*

Throughout the interview Sarah denies the impact of her own diagnosis as something separate from her son’s and offers the following description of what it was like to receive her own results:

*Well it had started with him…kind of the nail in the coffin then was when my results came back.*

Two weeks after the interview had taken place, Sarah called the researcher to say that her father (from whom she inherited the fragile x gene) and to whom she had prayed every day since the age of seven, had disappeared very suddenly on the day of her diagnosis. Until that day she felt that he had always been there for her and had firmly believed that he would not have let this happen. When it did, she no longer felt his presence.
Chapter 5 – Discussion

5.1 Introduction

The aim of this study is to build upon the existing research from genetic medicine, using an Object Relations framework, to offer a deeper understanding of the experiences of female fragile X carriers and to address the lack of on psychotherapy research in this area. The findings illustrate the participants’ perceptions and interpretations of their genetic status, thus illustrating the implicit meaning of confronting a genetic diagnosis of fragile X.

The themes of ‘Suspended in The Unknown’, ‘Fragile X: A Threatening Presence’ and ‘Primitive Defenses’ presented in Chapter 4 illustrate the participants’ experiences as a fragile X carrier and the ways in which they have attempted to make sense of these experiences. The findings are discussed here in relation to the literature reviewed in Chapters 1 and 2.

5.2 Suspended in The Unknown

The key finding common to all participants was the unexpected and unknown nature of the diagnoses of fragile X syndrome which was experienced as deeply traumatic. The trauma of the diagnoses was exacerbated by a lack of engagement by a medical system which ultimately failed to contain the participants in their distress. The findings illuminate the participants’ journey within the medical framework as an experience of unconnectedness, which along with the traumatic impact of the diagnosis, created a deep sense of fear.
According to Hagerman, Rivera and Hagerman (2008), FXS is the most common inherited cause of intellectual disabilities and the most common known cause of autism. Despite this statistic, the findings show that none of the participants were aware of the existence of the FXS until their child’s diagnosis and the shock expressed correlates with Targum’s (1981) belief that a parent’s experience of their child’s genetic diagnosis is experienced as a trauma. McDaniel (2005) states that an individual may decide to undergo genetic testing when they become aware of their risk of a genetic condition that runs in the family. This corresponds with the findings, whereby the participants became aware of their risk through their children’s FXS diagnosis. However, the abrupt and unexpected nature of FXS presenting for the first time in the family, in their children, was a further source of anxiety for the participants. Research suggests that heightened emotional states interfere with the integration of genetic results and decisions around testing or treatment (Sharpe and Carter, 2006). Equally, this study suggests that the trauma of their child’s diagnosis may have prevented the integration of their own genetic information as a fragile X carrier.

A lack of engagement within the medical framework was experienced across all interviews. From an Object Relations perspective, the unresponsiveness of a caregiver (in this case the medical system) is experienced as a trauma and instils a level of fear in the child which has a profound effect on the developing psyche. What is more, when the infant is left in an interminable state of unmanageable emotion due to lack of connection with the parent, segregated mental representations of the self and others arise (Bowlby, 1988; Fairbairn, 1946; Gerhardt, 2004). It is important to keep in mind Bowlby (1998) and Klein’s (1959) assertions that an understanding of how the adult psyche relates is brought about by an understanding of the development of the infant psyche. It is therefore understood that the experiences of the
original trauma and the failure to hold shocked the adult psyche resulting in a defensive organisation which is discussed in Section 5.3.

Catherine and Laura’s accounts of lengthy waiting lists illustrate this sense of fear and anxiety. Likewise, Sarah’s description of the abrupt phone call she received with the results of a genetic test, which she was unaware had been conducted, demonstrates the importance of what Todres, Galvin and Holloway (2009) advocate in terms of future healthcare “…individuals can be dislocated and shocked by the unfamiliarity of events that excessively wrench them away from the familiar…this engagement with temporality thus needs to be understood when considering a more humanised form of care” (p.73).

Additionally, the vivid accounts of the challenges faced within an unresponsive medical system support the calls for earlier intervention in genetics which would prevent the “diagnostic odyssey” Abrams et al. (2012, p.4) discuss, wherein ongoing delays in the identification and diagnosis of FXS creates challenging experiences for parents. Catherine’s description of her journey through diagnosis illustrates feelings of isolation, wherein she is utterly disconnected from time and space, giving weight to the importance of the narrative in the illness experience. This is reflected in Todres, Galvin and Holloway (2009) who write:

“To be human is to be on a journey…we move through time meaningfully and do not exist in a vacuum; to be human is to be connected to a sense of continuity…the meaningfulness of a person’s journey can either be supported or lost” (p.72-73).

The findings are therefore consistent with previous studies which identify anxiety among fragile X carriers (Hagerman and Hagerman, 2008; Lachiewicz et al 2010; Tassone, Hagerman and Hagerman, 2014). While medical research seeks to understand the aetiology
of anxiety in biological terms, this study offers another paradigm by which the anxiety experienced by fragile X carriers may be understood. Kleinian theory (1946) asserts that anxiety is central to our being and therefore the anxiety stirred from internal conflicts may be more frightening than the fear encountered from external perils. Likewise, Fairbairn (1946) and Bowlby (1998) theorize that the organism is essentially object-seeking and disruption to the psyche in the form of separation or loss evokes an intense anxiety that is almost impossible to bear.

5.3 Fragile X: A Threatening Presence

Jacobs (2012) emphasises the importance of unconscious mental life in understanding how individuals relate to themselves and others. He draws on Object Relations theory which defines the creation of mental representations of external relationships as part reality and part unconscious fantasy. The study found that for each participant an inner engagement with fragile X was framed as a Threatening Presence which evoked feelings of loss and anxiety in which the participants were not supported within the medical framework.

The study found that participants constructed meaningful narratives in which the sinister fragile X-object played a part in their life decisions, particularly in relation to family planning. The sense of threat evoked unbearable anxiety in the participants and fits with Klein’s (1946) paranoid-schizoid position, wherein persecutory anxiety and the dread of annihilation are felt by the intense interplay of “…externally-derived bad experience…[and] the rebounding-back of the death instinct which we project into the other” (as cited in Gomez, 1997).
Targum (1981) describes feelings of loss as common to parents’ experiences of a diagnosis of a genetic condition in their children and this is consistent with the findings. Catherine’s vivid recollection of leaving hospital with the thoughts of coming back for her boy and the tones in her voice express sadness and anger at having lost her much-longed-for son and her ideal family. Targum (1981) emphasises the need for parents to grieve for the loss of their healthy child or the loss of future children. In this way, separation from the lost-object by way of mourning allows the individual to move on and to reinvest their energies in other activities (Freud, 1917). To this end, Catherine describes her intense feelings as motivating her to begin working for a fragile X charity, thus indicating an integration of her loss.

Likewise, Sarah’s rich description of receiving Frankie’s diagnosis illustrates feelings of loss which are implicitly known to her. Her portrayal illustrates her struggle to make sense of the experience which she describes like a death but where no one had died. Her experience correlates to Freud’s (1914) state of melancholia wherein the person who grieves is unsure who or what is being mourned. Significantly, at a later stage Sarah’s own diagnosis evoked feelings of the earlier loss of her father. Upon receiving her own test results, her father (the lost-object) with whom she remained identified with in prayer (Freud, 1917), suddenly disappeared. The researcher proposes that as a child she may not have been able to mourn her father and had instead internalised the trauma of the loss which allowed her to continue relating to a spiritual idealised father who continued to look out for her (Fairbairn, 1946). When he failed her by not protecting her against fragile X her repressed feelings relating to the earlier loss were reawakened and his death was realised.

The study builds upon and deepens the understanding of previous studies of genetic carriers which identify loss (Anido, Carlson, Taft and Sherman, 2005; McConkie-Rosell,

Furthermore, the study found the participants’ inner engagement with fragile X became more threatening than the external threat and efforts to assuage their fears through genetic facts were unsuccessful. This supports Todres, Galvin and Holloway’s (2009) idea that life as it is lived from the inside does not always reflect the statistical truth, which remains the primary focus in medicine while the subjective experience is overlooked. Participants’ accounts unanimously demonstrated frustrations experienced within the medical system regarding their unmet expectations of genetic counselling which led to difficulties in integrating the trauma of the diagnosis. The findings support Sharpe and Carter’s (2006) claims that complex psychological responses often arise from genetic diagnoses and should therefore be adequately assessed and supported.

The literature raises concerns about the appropriateness of genetic counselling in managing the emotional distress experienced by genetic carriers. While psychotherapeutic interventions have been noted in genetic counselling, major discrepancies have been found throughout the practice which is designed to be both educative and supportive in autonomous decision-making. Evans (2006) advises the competent use of non-directiveness, borrowed from psychotherapy, while Petersen (1999) finds it unsuitable for genetic counselling which sits within the medical model. The genetic counsellor remains the medical expert in the counsellor-client relationship and is responsible for imparting value-laden medical facts so it raises the question if they should ever hold the not-knowing position taken up by a psychotherapist.
The research highlights frustrations within the genetic counselling profession regarding training (Capelli et al., 2009) wherein counsellors feel they have inadequate skills to deal with high levels of anxiety experienced in their clients. Furthermore, the extent of the emotional responses experienced by individuals and families undergoing genetic testing raises issues of counter-transference (Kessler, 1992) and begs the question: can the true nature of their anxieties be fully explored in a clinical genetic setting where the sessions are found to be short-term solution-focussed? McAllister’s (2001) concern around the lack of a theoretical framework in genetic counselling may also be detrimental to the work with clients as it brings uncertainty for counsellors. The frustrations expressed by the participants reflect a lack of psychotherapeutic services whether provided by genetic counsellors or otherwise.

5.4 Primitive Defenses

Object Relations theory holds that the self needs to feel connected, coherent and understood in a world that makes sense. The developing psyche is a self-organising, dynamic entity which relates to the Other, in reality and fantasy, via splits and conflicts and by introjecting and projecting (Fairbairn, 1946; Klein, 1946). When its needs are met by an attuned caregiver the infant internalises the good-object, which facilitates integration and brings about a sense of continuity and meaning (Bowlby, 1988). The extent to which the infant organises its inner world using defenses depends on the degree of care and the ability of the Other to contain the infant’s anxieties (Fairbairn, 1946). In this way, the psyche achieves a sense of coherency which enables the infant to engage with and live in its environment. Although the defenses may become modified over time, the adult psyche may regress to these primitive ways of relating in times of trauma (Jacobs, 2012). The findings identified across the interviews are consistent with Object Relations theory in that the trauma of a FX diagnosis gave rise to the use of primitive defense mechanisms of splitting, projection, introjection and denial.
Targum (1981) maintains that a diagnosis of a child’s genetic condition is experienced by the parents as a trauma which activates the use of unconscious defenses. He notes that denial is most often used to defend against the intense anxieties which arise out of the feelings of loss that accompany the diagnosis. The use of denial, which defends against that which is unacceptable to the psyche (Jacobs, 2012), is most evident in Sarah’s experience where she continued to deny her son’s condition in the hope that it would go away. Throughout the interview, Sarah also denied any impact of her own diagnosis as a carrier and insisted that the researcher come back to questions relating to her son’s diagnosis. However, the phone call two weeks after the interview, wherein Sarah disclosed her experience after her own diagnosis, suggests that through the mechanism of repression she denied the impact of her own diagnosis as it was connected to the original loss of her father which had also been disavowed.

Gomez (1997) notes that both projection and introjection “strengthen the division between good and bad” (p. 38). For Catherine and Laura, these mechanisms are evident in their management of the anxiety experienced within the intersubjective framework of the medical system. According to Fairbairn (1943), “it is better to be a sinner in a world ruled by God than to live in a world ruled by the devil” (p. 67). This is consistent with Laura’s introjection of the trauma of her unmet needs which saw her blame herself for not doing enough to protect her child and allowed her to go on relating to a satisfactory medical system. The same may be said of her familial relationships in which nothing much seems to have changed despite the disruption of the distressing diagnosis.

The findings identify Catherine as projecting intense anxieties associated with the FX gene onto a system that is already failing her. The FX gene has been internalised both as a
threatening bad-object and also as a fragile, vulnerable and Irish bad-object that is accepted as genetically part of her. In this way, Catherine’s inner life holds many powerful emotions and the study identifies deep feelings of guilt and persecutory anxiety as she constantly questions if she is doing enough for her daughter. Catherine’s disdain of her own vulnerability and at times of others’ vulnerability is consistent with Fairbairn’s (1946) process of the internal split in the schizoid position where part of the ego attached to the rejecting-object fears vulnerability (as cited in Gomez, 1997).

The study also highlighted a parallel process of projection within society. Catherine’s vivid portrayal of the appointment in a clinic housed in a former ‘mental institution’ suggests that her intense feelings of shame not only arose from her own bad-objects but from shame that had been disavowed in society (Sinason, 1992 as cited in Gomez, 1997). Catherine is incredulous at having carried the feeling of shame home with her, thus suggesting that she may have felt the shame projected out by the genetic counsellor through ‘projective identification’ (Klein, 1943).
Chapter 6 - Conclusion

6.1 Overview of Study

This study examines, through the lens of psychotherapy and from an Object Relations informed perspective, the implicit meaning three women have made of their experience as fragile X premutation carriers. Due to the dearth of literature from the field of psychotherapy, the researcher initially drew on literature from the disciplines of genetic medicine and health psychology. The literature describes the confrontation with genetic illness as a profoundly human encounter and that the experience of a genetic diagnosis in a child is often experienced as traumatic for parents (Targum, 1981; McDaniel, 2005). However, the research identifies challenges in genetic counselling which raises the fundamental question as to whether genetic carriers are contained in their efforts to manage the anxieties arising out of this new genetic information (Capelli et al., 2009).

To develop a deeper understanding of the difficult emotions experienced by those confronting genetic illness identified in the introductory literature, the researcher further explored literature from Object Relations which presents the psyche as a self-organising dynamic entity which relies on the use of defences to achieve a sense of coherency when separation and loss is experienced. Object Relations theories as outlined by Klein (1946), Fairbairn (1946) and Freud (1917) are discussed in respect of the findings of the study, thus illustrating how a genetic diagnosis of fragile X manifests in the adult psyche.

For all participants, the diagnosis of the previously unknown fragile X syndrome in their child was experienced as traumatic. By exploring how the participants have managed and contained the impact of the trauma, the study highlighted a difficult journey within an
unresponsive medical framework which added to the fear and anxiety already experienced by the participants. The study found that the inner engagement with fragile X as a mental representation became more threatening than the external threat, and unresolved grief and feelings of loss are identified across the interviews. The study suggests that the anxiety arising out of a genetic diagnosis cannot be fully understood without the acknowledgement of unconscious processing of the experience of loss. In other words, feelings of loss need to be mourned. Lastly, in an effort to manage the anxieties arising out of their experiences, a regression to primitive defences is identified in different degrees across the interviews.

6.2 Strengths and Limitations

There is a paucity of research in the field of psychotherapy on the experiences of genetic carriers and specifically that of fragile X carriers which this study seeks to address. By upholding the significance of the lived experience, the study generates new knowledge and understanding of what it means to be a fragile X carrier. This is of particular importance in the light of genetic advances which radically improve health yet often overlook the significance of the subjectivity of genetic illness in wellbeing. Furthermore, the study highlights the need for psychotherapeutic support for genetic carriers in the clinical setting.

In outlining the therapeutic efficacy of psychoanalysis, Freud (1904) stated that the difficult emotions attached to painful events from consciousness which have been banished to an unconscious level of mental life, by an active repressing mechanism, are offered the opportunity of being expressed and integrated through talking. The study supports this view through Sarah’s post-interview process which indicates her separation from the internalised lost-object.
A number of limitations were identified at the beginning and during the course of the study. Based on the lack of psychotherapy research into the experiences of genetic carriers, the researcher was limited in choosing a specific psychotherapy framework that would expand upon the psychosocial studies from other disciplines. The researcher felt, however, Object Relations theory allows a meaningful framework from which the experiences of fragile X carriers can be explored. However, other psychotherapy frameworks may be useful in further research.

The phenomenological analysis of the data collected in three semi-structured interviews is limited by its size and may not be applied to the general population. The researcher chose a small sample group, as recommended for IPA, in order to elicit a richer understanding of how the participants attempted to make sense of their experiences. Furthermore, because the participants were all fragile X carriers the findings may not be applicable to genetic carriers of other conditions. However, the researcher understood the importance of having a homogenous sample group and further felt that female fragile X carriers will have direct experience of the family of fragile X disorders.

The impact of the unresponsive medical system on the participants in the study was experienced in the context of the Irish medical system and therefore cannot be applied to the wider international field. However, the researcher understands the psychological impact of an unresponsive healthcare system as universal.

Finally, the researcher is a fragile X premutation carrier. The researcher attempted to manage this bias through regular contact with the research supervisor, by regular attendance at the research seminars and with weekly engagement in personal therapy.
6.3 Further Recommendations

Due to the dearth of literature from the field of psychotherapy in the area of genetic illness, it is hoped that this study will broaden psychotherapy research. The word count limited the depth to which the rich data, elicited in the interviews, could be explored and the themes discussed in this study should be seen as an initial premise for further research. As noted above, other theoretical frameworks in psychotherapy may be useful in the further exploration of the experiences of genetic carriers.

Overall, additional studies from psychotherapy will contribute to a fuller understanding and comprehensive care for genetic carriers while adding to current discourse around the ethics of medical genetics.
Bibliography


Appendices

Tania Kacperski
MA in Psychotherapy
School of Arts
Dublin Business School (DBS)  
February/March 2015

Appendix A

Qualitative Research Study:
A psychotherapeutic exploration into the experiences of female fragile X premutation carriers: an interpretative phenomenological analysis.

PARTICIPANT INFORMATION SHEET

Introduction
My name is Tania Kacperski and I am a student in the final year of a Master of Arts in Psychotherapy at Dublin Business School (DBS). I am currently undertaking a qualitative research study which seeks to understand the experiences of women who carry a premutation of the fragile X gene.

The study has received approval from the DBS Ethics Committee.

What is the purpose of the study?
You are invited to participate in this study if you are a woman who has been diagnosed as a fragile X premutation carrier three or more years ago. The purpose of the study is to explore what your status as a fragile X premutation carrier has meant for you since your diagnosis and how this new genetic information has impacted you.

What are the criteria for participation in the study?
Participants of this study must be females who have been diagnosed as a fragile X premutation carrier three or more years ago.

What is involved in participation in the study?
Should you choose to participate in the study, you will be invited to take part in an hour long individual interview, with myself the researcher, at a private location in Dublin city centre. The interview will consist of a series of questions relating to the research question. The interview will be recorded and later transcribed by the researcher.

Will my identity and information be protected?
Your identity will be protected by a code known only to the researcher. This code will be used on all forms relating to your participation and on any additional notes taken in relation to the information obtained from. All identifying information will be removed during transcription of your interview to protect your anonymity, for example, pseudonyms will be given to those mentioned in your narrative. All data, including transcripts, will be coded and...
stored on the researcher’s computer in a folder which is protected by a password known only to the researcher. All paper records will be kept in a file which will be locked. Audio recordings will be destroyed once transcripts have been made.

**Can I withdraw from the study?**
Participation in the study is entirely voluntary and you can withdraw consent at any time without prejudice.

**Are there any risks of participating in the study?**
If you feel distressed in any way during the interview, due to the sensitive and personal information being explored, please make this known to the researcher and the interview can be stopped at any time. Likewise, if painful or distressing issues are evoked at a later stage, the opportunity of a therapy session, by way of de-briefing with a psychotherapist, can be availed of - see information below:

<table>
<thead>
<tr>
<th>Aris Psychotherapy &amp; Counselling Centre Dublin 2 or Balbriggan, North Co. Dublin</th>
<th>01-8020437</th>
<th><a href="mailto:info@ariscentre.ie">info@ariscentre.ie</a></th>
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<tr>
<td>Loreto Centre Crumlin Counselling &amp; Psychotherapy Service</td>
<td>01-454 1078</td>
<td><a href="mailto:loretocentrecrumlin@eircom.net">loretocentrecrumlin@eircom.net</a></td>
</tr>
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</table>

Alternatively, you can search for a registered psychotherapist, anywhere in Ireland, on the Irish Council for Psychotherapy’s website [http://www.psychotherapy-ireland.com/find/](http://www.psychotherapy-ireland.com/find/)

**How can I get further information?**
Should you have further questions regarding this study, please do not hesitate to contact either person below:

**Researcher:** Tania Kacperski tankacperski@gmail.com  
**Research Supervisor:** Dr. Gráinne Donohue grainne.donohue@dbs.ie
Appendix B

Qualitative Research Study:
A psychotherapeutic exploration into the experiences of female fragile X premutation carriers: an interpretative phenomenological analysis.

CONSENT FORM

Please tick the appropriate answer

<table>
<thead>
<tr>
<th>Question</th>
<th>Yes</th>
<th>No</th>
</tr>
</thead>
<tbody>
<tr>
<td>I confirm that I have read and understood the Participant Information Sheet attached and that I have been given ample opportunity to ask questions all of which have been satisfactorily answered by the researcher.</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>I understand that my participation in this study is entirely voluntary and that I may withdraw at any time.</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>I understand that my identity will remain confidential at all times.</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>I am aware of the potential risks of this research study.</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>I am aware that audio recordings will be made of interviews.</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>I have been given a copy of the Participant Information Sheet and this Consent Form for my records.</td>
<td>Yes</td>
<td>No</td>
</tr>
</tbody>
</table>

Participant ____________________________ ____________________________ __________
Signature                                                  Print Name                                                  Date

To be completed by the researcher.
I, the undersigned have taken the time to fully explain to the above participant the nature and purpose of this study in a manner that she understands. We have discussed risks involved and I have invited her to ask questions on any aspect of the study that concerns her.

Researcher ____________________________ ____________________________ __________
Signature                                                  Print Name                                                  Date
Appendix C

Qualitative Research Study:
A psychotherapeutic exploration into the experiences of female fragile X premutation carriers: an interpretative phenomenological analysis.

1. When were you diagnosed as a fragile X premutation carrier?
2. Can you tell me about the time you first became aware that you might be at risk of being a carrier of fragile X?
3. What prompted you to undergo genetic testing for fragile X?
4. Can you describe what that was like for you to receive your genetic test results?
5. Did you receive genetic counselling? If so, can you describe this experience?
6. How has your new genetic information impacted you?
7. What does it mean for you to be a FX carrier?
8. When you think of yourself before you received your diagnosis, would you describe yourself as any different than today?
9. From whom did you inherit the fragile x gene?
10. Describe the impact, if any, being a FX carrier has had on your relationships with others?
11. Can you describe a time when you when you have felt that being an FX carrier impacts upon your health?
12. I’m interested to know what it’s like for you to participate in this study, can you tell me?
13. Is there anything else you would like to add or questions you wish I had asked you?
Appendix D

Themes – Phase 1

Participant 1
THEME:
Loss: a threat to survival?
The Unknown – what is not known?

A sense of loss of certainty/what is known, rising anxiety
Loss of future children - ‘family planning’ – a threat to generational survival
Failure of holding/managing loss:
Helplessness
Trauma
Anxiety
Difficult to manage the unknown
Failure of Genetic Counselling/Medical to provide holding – ‘unresponsive’
Resulting trauma.
Cold – frozen? Trauma
Sense of suspension in the unknown; Other-wordly, depersonalisation, time-warp, difficulty remembering, vacuum, dark, horrible, bubble.

“Trauma, it was trauma!”
Fight or flight –“Act! Act! Act!” - Helplessness to powerful when suspended in the unknown have to defend against the anxiety, Act! Act! Act!

“There but not there. I just knew”

Participant 2
THEME:
Loss: a threat to survival?
The Unknown – what is not known?

Sunny day before hand, difficulty remembering.
It – what is it?
I didn’t know what it was? If I’d known?
Is he going to die?
Like a death but not like a death
Nail in the coffin
It taking away from, destroying. “IT” – Mental Representation Theme?