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**Relationships, sexuality and parenting: The experience of five young women
with 22q11.2 deletion syndrome (22q11DS). An interpretative
phenomenological study.**

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Structured Abstract

Introduction Many people with intellectual disability (ID) experience problems with maintaining social relationships, have smaller social networks and experience feelings of social disconnectedness. A large majority do not enjoy intimate relationships, marry or have children. When adults with ID fulfil parenting roles, many experience significant barriers including stigma. It is clear that there is value in relationships for adults with ID and that there are some unique challenges associated.

22q11.2 deletion syndrome (22q11DS) is a genetic condition associated with developmental disabilities and has an incidence of approximately 1 in 4000 newborns. More than 90% of people with 22q11DS have developmental difficulties, with approximately half having an ID. There is a paucity of research available from the experiences of individuals with 22q11DS. This study aims to explore how young women with 22q11DS experience relationships, sexuality and parenting. The perception of parental and service provider's attitudes on relationships, sexuality and parenting among the women will also be explored, to understand the quality of support experienced.

Method Using Interpretative Phenomenological Analysis, this study explored the experiences and perceptions of five women with 22q11DS, regarding relationships, sexuality and parenting. Semi-structured interviews were conducted to elicit the themes.

Results Four Superordinate themes were evident within the women's dialogues, including several subthemes: (i) Limitations of 22q11DS (Engagement in social comparison/responsibility); (ii) Acceptance/ normality (Social competence / to be a good parent); (iii) Support (From Mum); and (iv) Individuation (Readiness for adulthood/ with parental agreement).

The women interviewed offered insights into how they experienced their limitations associated with 22q11DS, commonly reporting difficulties with learning and social skills problems. Participants engaged with their disability/ limitations through processes of social comparison; noting their individual strengths through comparisons with those with more serious disabilities; and identifying their own limitations through comparisons with typically developing peers. With regard to childbearing, the women tended to overestimate the risks of heritability; and reflected themes of responsibility, expressed as a commitment to meeting the individual needs of any children who may be affected by the deletion.

Participants ascribed value to normality, with social competence identified as a mechanism for achieving this. The women offered their experience of relationships with typically developing peers as evidence for their own perceived social competence. Being a good parent was also an important aspect for the women who expressed intentions for parenting; and parenting values were often derived from the behaviours and values of parents and extended family. Being a good parent was also viewed as a possible mechanism for ensuring acceptance from others.

Support from family, especially mothers, was perceived positively by the women interviewed. Supports provided by mothers ranged from practical assistance with daily living, through to provision of emotional support and friendship.

Themes of individuation from family and aspirations for independence were also contained in the women's dialogues. The women communicated their understanding of relationships, sexuality and parenting; and in doing so believed this inferred a readiness and competence for adulthood.

However, the expressed knowledge of adulthood had often not been derived through direct personal experiences. There was a perception that parents were supportive of the longer-term plans for adult roles held by the women with 22q11DS, inclusive of marriage and parenthood; however, the perceptions had developed often without having these explicit conversations with family.

Conclusions This study has begun to explore how women with 22q11DS experience their relationships, sexuality and parenting. The women with 22q11DS in this study had positive perceptions of support and faced their futures with optimism and confidence regarding their abilities. They expressed a desire for normalisation and a strong sense of self-determination, in a context of perceived family support. Consideration and further understanding of the unique experiences of women with 22q11DS, including the acknowledgement of expressed desires and concerns regarding relationships, sexuality and parenting, will invariably assist young women with the deletion to achieve fuller and meaningful life roles for themselves and their future families.

Table of Contents

| | |
|----------------------------|---------|
| Critical Literature Review | page 7 |
| Manuscript | page 23 |
| References | page 57 |
| Appendices | page 71 |

Relationships, sexuality and parenting: The experience of five young women with 22q11.2 deletion syndrome (22q11DS). An interpretative phenomenological study.

Introduction

For most young adults, establishing social and intimate relationships, exploring sexuality, and considering parenthood is a normal and expected part of life. However, for young people with disabilities, these areas of life are more problematic.

People with intellectual disability (ID) experience the same interest in relationships with peers and partners as their typically developing counterparts do (May & Simpson, 2003). That said, the majority of people with ID do not marry, have children, or enjoy intimate relationships (May & Simpson, 2003). The rights of people with ID to make decisions about their own lives and take on valued roles in the community, including the right to marry and form a family, is enshrined in many pieces of legislation (United Nations, 2006). Historically though, relationships and marriage between people with ID were not supported by policy makers or those who held a direct care role. Whilst many gains have been made in exercising the rights of people with ID to marry, express their sexuality and have children, as a group they still continue to experience stigma and prejudice, are socially isolated, and encounter major barriers that restrict their quality of life (Chou, Pu, Lee, Lin & Kröger, 2009). Indeed, many still experience poorer sexual education, sexual knowledge and negative experiences of sexuality (McCabe, 1999; Isler, Tas, Beytut & COUNK, 2009).

Historically, concerns associated with the sexuality and marriage of people with ID first arose with the eugenics movement (May & Simpson, 2003). Deinstitutionalisation in the late 1960's created a climate for people with ID to explore marriage and parenting without the surveillance and control of institutional care (May & Simpson, 2003). Whilst this was occurring, significant policy changes were also being made through the introduction of normalisation principles, which promoted access to what was perceived as a normal pattern of life (Bank-Mikkelsen, 1976). This included opportunities for people with ID to engage in employment, education, social and recreational activities, and independent housing. However, it was through the introduction of the contraceptive pill and the inherent decoupling of sex from procreation, that the eugenics concerns for the "genetic health" of the population and the moral fabric of society (May & Simpson, 2003, p28) then evolved to the opposition of parenting by adults with ID in the 1980's. These concerns were based on parental competence and moral rights. So despite the presence of strong legislation which supports the self-determination and individual rights of adults with ID, in reality, full access to these life domains remains restricted.

Background

22q11.2 deletion syndrome (22q11DS)

22q11.2 deletion syndrome (22q11DS), also known as Velo Cardio Facial Syndrome (VCFS) is one of the most common multiple anomaly syndromes with an incidence of approximately 1:4000 newborns (Hallberg, Óskarsdóttir & Klingberg, 2010). It is inherited in an autosomal dominant manner, which means that if a parent with the deletion has a child, there is a 50% risk of passing it on to the child. In approximately 93% of individuals the deletion occurs de novo, with 7% inheriting the 22q11.2 deletion from a parent (McDonald-McGinn, Emanuel & Zackai, 2013). The phenotype includes (but is not limited to) cardiac defects, velopharyngeal insufficiency, feeding difficulties,

learning disabilities, social problems, and psychiatric disorders (Hallberg, Óskarsdóttir & Klingberg, 2010); however, there is significant variability in the presentation of the deletion within affected individuals (Swillen, Vogels, Devriendt & Fryns, 2000). Developmental difficulties are found in more than 90% of individuals with 22q11DS (McDonald-McGinn, Tonnesen, Laufer-Cahana, Finucane, Driscoll, Emanuel, & Zackai, 2001), with approximately half having an intellectual disability (Semple, Smyth, Burns, Darjee, & McIntosh, 2005). In cases where the deletion is inherited from a parent, lower full-scale IQ scores are reported, when compared to individuals where the deletion occurs de novo (Swillen, Devriendt, Legius, Eyskens, Dumoulin, Gewillig & Fryns, 1997).

How adults experience living with an ID that has an identified genetic cause is relatively unexplored territory. With specific regard to 22q11DS, research to date has mostly focused on neurological studies; brain imaging; co morbidity with mental health disorders including schizophrenia; genetic counselling; and the study of behavioural phenotypes. For adults with ID, there is considerable research in the areas of sexuality (McCabe, 1999), relationships (Knox & Hickson, 2001), and parenting (IASSID, 2008); however, the individual perspectives of people with ID on these subject areas has been given less attention.

The relationships of adults with intellectual disability

It is recognised that relationships are an essential aspect of any person's life, including adults with ID. Having relationships is closely linked to one's quality of life (Brown, 1994; Brown, 1997; Schalock, 1996) and critical also to the well-being of adults with ID (Knox & Hickson, 2001). A recent paper, co-authored by adults with ID, reinforces this position affirming that close relationships with peers contribute to positive attributions of happiness and satisfaction within the lives of adults with ID

(Haigh, Lee, Shaw, Hawthorne, Chamberlain, Newman, Clarke & Beail, 2013). However, many researchers have reported a lack of social networks for people with ID and feelings of not being connected socially (Knox & Hickson, 2001).

A common indicator of social competence or success in relationships is time spent with peers and friends, outside of formal activities such as work and school. Australian adults reportedly spend approximately 12 hours per week socialising with friends (ABS, 2006a). This is often not the case for people with ID who report most of their social contact to be with family members or family friends (Emerson & McVilly, 2004). Further, whilst adults with ID may identify as having peer friendships within their employment and educational programs, they often do not spend time with their work and school friends outside of these facilities (Petrovski & Gleeson, 1997). These findings may be reflective of the differing nature of relationships that exist between adults with ID, or simply issues of access and availability and a necessary reliance on family and support persons to facilitate opportunities for social contact outside of these formal structures.

It is clear from the research literature that people with ID experience fewer friendships (Amado, 1993) and have difficulty maintaining such relationships where they do exist (Abery & Fahnestock, 1994); however, how people with ID understand and experience their relationships is an area of research that has received less attention. Knox and Hickson (2001) used in-depth interviews, guided by focus questions on close friendships, to further understand the “close relationships” of four adults with ID. Those interviewed differentiated between two types of close relationships: “the good mate” and “the boyfriend/ girlfriend”. Inherent in the good mate relationship was the importance of the friendship, doing lots of things together, a shared sense of history, common interests and reciprocal support. Within the girlfriend/ boyfriend relationship, meanings ascribed included:

difference from other relationships, feelings of intimacy, physical attractiveness, and an expectation of relationship change (i.e., marriage and/ or children). The study highlights the ability of adults with ID to form valued, complex and dynamic relationships.

When reporting on peer relationships and loneliness, McVilly and colleagues (2006) noted the expressed importance of a shared experience or history with other persons linked to or associated with their ID (McVilly, Stancliffe, Parmenter & Burton-Smith, 2006). Further, the adults with ID ascribed importance to also having relationships with adults who do not have disabilities. Those who perceived themselves to be part of a support network inclusive of people with and without ID, reported lower levels of loneliness. However, despite the reported preference of McVilly and colleagues (2006) for having friends both with and without an ID in their social networks, the adults interviewed by Knox and Hickson (2001) did not identify people without ID's as their good friend or mate relationship. Rather, adults without ID were noted as more distal relationships in the social networks of people with ID.

Lutfiyya (1991) commented on the friendships between four pairs of adults, inclusive of an adult with learning difficulties and a typically developing peer, finding that genuine friendships do exist. The meanings ascribed to the relationships were derived through interviews with the adult without ID and observation of the informant pairs. Common relationship characteristics were identified by all the pairs including: it being mutual, exclusive, and voluntary nature; recognition of the rights, obligations, and responsibilities of friends to each other; and also positive regard or affection between the friends. The researcher did commence interviews with the adults with ID; however, they produced brief and limited responses. Lutfiyya (1991) noted her own lack of experience interviewing adults with ID as a limitation and a possible reason for the lack of data.

People with ID have been described as one of the most stigmatised groups when compared to other disability groups, with the stigma experienced either directly, such as, insults (Jahoda, Markova & Cattermole, 1988) or through more subtle forms which limits full access to meaningful adult activities such as employment and relationships (Jahoda & Markova, 2004). It has also been reported that people with ID face stigmatisation and prejudice merely through association with each other (Chappell, 1994). Whilst, relationships with other adults who do not have ID could be viewed as avenues for enhancing one's self-concept and self-esteem, the *close mate* relationship described by Knox and Hickson (2001) may provide benefits that attenuate the stress associated with the stigma or perceived stigma of ID. That said, peer relationships vary depending on stages of development and are influenced by the definition of a friend, the behaviours that occur within those friendships, the friendship quality, and number of friends (Hartup, 1992). It seems apparent that people with ID value close relationships and are capable of achieving intimacy and differentiating between close, romantic and acquaintance relationships, thereby appreciating the role each relationship type can play in their life. However, further research from the perspective of people with ID is warranted and justified based on the richness of relationships that exist for people with ID.

Whilst the emerging literature is demonstrating interpersonal relationships for people with ID that are both enriching and critical to their ongoing development and quality of life, it would be remiss to not address the literature which indicates greater rates of abuse and interpersonal violence for people with ID when compared to the general population. Ward, Bosek and Trimble (2010) interviewed men and women with mild to moderate ID who were living in supported accommodation about their romantic relationships and experience of interpersonal violence. Their results indicate that partnered or romantic relationships were very important to the adults interviewed, even if the couples spent little time together. Further, the relationships sounded similar to those of other boy-girlfriend relationships, in what they did together; however, the amount of

time spent with each other was reduced. Regrettably violence characterised many of the relationships, with 60% of participants reporting that they had experienced some type of interpersonal violence in their past or current relationships, with both men and women reporting physical assault. This is consistent with previous research indicating that women with disabilities are at greater risk of interpersonal violence and abuse (Gill, 1996; Smith, 2008). In comparison, national statistics indicate that 15% of Australian women have experienced physical or sexual violence from a previous partner and 2.1% from current partners (ABS, 2006b). Unfortunately Ward and colleagues (2010) did not specify demographic details for the described partners (I.e., level of disability), leaving questions about the personal characteristics of the alleged perpetrators.

Sexuality and adults with ID

Sexuality is also an essential part of one's personality and sense of self, and adults with ID are sexual beings (Kijak, 2011). Young adults with ID have an understanding of their sexual rights and are able to identify the social and cultural barriers that they feel prevent them from achieving sexual autonomy (Healy, McGuire, Evans & Carley, 2009). Many misconceptions about the sexuality of people with disability exist including that they are asexual, childlike, and vulnerable and in need of protection from society (Murphy & Young, 2005). Many adults with ID also report having an awareness of these sexually limiting perceptions (Murphy & Young, 2005).

The sexual knowledge and experience of adults with ID is generally reported to be poorer and problematic. McCabe (1999) interviewed 60 people with mild ID and 60 people with a physical disability in an attempt to examine their sexual knowledge, experience and needs. McCabe found that the adults with ID had lower levels of sexual knowledge and experience, held more negative

attitudes towards sexual activity, and had stronger sexual needs, compared to the adults with physical disabilities. McCabe (1999) cited difficulty discussing sexual matters with family and friends as contributing to their sexuality not being normalised. The participants also reported lower levels of sexual experience, which was contrasted with higher levels of sexual needs; indicative of a strong need for dating and intimate relationships. Restricted access to dating experiences which manifested through limited sexual education in addition to negative feelings about sexuality was also reported.

Lower levels of sexual knowledge and poorer access to sexual education, is a consistent finding in the research literature (Isler, Tas, Beytut & Conk, 2009; Siebelink, de Jong, Taal, & Roelvink, 2006). Despite the recognition that people with ID are sexual beings and access to sexual relationships is supported by legislative frameworks and disability models of care, the issue is still contentious with many families and caregivers who feel compelled to balance the needs and sexual rights of adults with disability, with the knowledge that they as a group are a vulnerable population with traditionally poorer sexual knowledge. Support staff working with adults with intellectual disability hold generally positive views of sexuality, when compared to their families (Brown, 1994; Cuskelly & Bryde, 2004). Similarly, the research considering the views of sexuality of adults with ID held by families reports more conservative views, when compared to the more progressive attitudes of direct care staff (Brown, 1994; Cuskelly & Bryde, 2004).

Parents with intellectual disability

The right of people with ID to bear children is enshrined in many pieces of legislation. Despite this, many people with ID experience direct barriers to achieving sexual autonomy and parenthood. A large majority (60-90%) of adults with mild ID wish to marry and become parents (David, Smith &

Friedman, 1976; Bratlinger, 1985). However, the attitudes held by the care staff that support adults with ID are often limiting and generally negative with regard to parenting aspirations in particular. Further, when pregnancy or parenthood is achieved, many people with ID report experiencing their announcements of pending parenthood as being met with either disbelief or dismay (Llewellyn, 1994, as cited in Mayes & Llewellyn, 2012), the assumption that it is a mistake which should not be repeated, and in some cases encouragement to terminate the pregnancy (Booth & Booth, 1995). Once over these initial hurdles, parents with ID continue to experience barriers to successful parenting in the forms of poverty, prejudice, limited access to resources, respect, and lack of moral support and practical assistance (IASSID, 2008). The restrictions and barriers to parenting with ID are no more apparent than in the child protection literature, where a disproportionate number of cases involved parents with ID (Mayes & Llewellyn, 2012). Despite the negative perceptions and restrictions, the research literature suggests that many adults with ID do experience success in their parenting roles; however, many others indeed struggle. Support from both informal and formal structures is noted as a key factor in success; however, how the support is perceived (i.e., positively or negatively) is also significant (Aunos, Goupil & Feldman, 2004).

Gilmore and Chambers (2010) interviewed 169 disability support workers and found that they held more cautious or conservative views on the topic of parenting for people with ID, compared to the generally positive attitudes they held regarding sexuality. When questioned about their reservations, support workers cited heritability of disability, parenting capacity, and the financial and health status of the prospective parents as the reasons for their concern. As such, there seems to be a sense of permissibility by direct care staff when it comes to sexual expression; however, restrictions and limitations are applied to parenthood. Further, it appears that these negative attitudes to parenting are more prevalent for people with ID than any other special population or minority group. Giami (1987) compared attitudes of families and special education providers toward the sexuality of both

persons with intellectual disability. Giami found that the parenting rights were not afforded to people with intellectual disability; however, when pregnancy did arise accidentally, it was a source of great conflict between parents and educators with regard to who was responsible for the occurrence. The negative attitudes to parenting by people with ID were then compared with the attitudes held regarding people with physical disability, with Giami (1987) reporting a stronger taboo on parenting and sexuality for adults with ID. If these negative biases exist for persons with intellectual disability where the cause of such is often unknown, it seems logical that further biases will exist for those with learning difficulties and a genetic disorder which is heritable.

The heritability of specific disorders and syndromes is an area of research that has gained significant attention recently, especially with regard to pre-implantation genetic diagnosis (PGD) testing and the processes for reproductive decision-making for both affected and unaffected and/ or carrier couples. The cause of intellectual disability is a combination of both genetic and environmental factors and only identifiable in approximately two thirds of cases (Harris, 2005). The reviewed literature for relationships, sexuality and parenting with ID, does not routinely include demographic information such as heritability.

Ward, Howarth, and Rodgers (2002) described people with ID and their families as 'conspicuously absent' from the debates about genetic technologies. There is also additional thought within the disability sector that genetic diagnoses or reasons for intellectual disability, offer little or no value in the daily support of people with disability (Finucane, Haas-Givler, & Simon, 2003). However, advances in genetic testing and diagnosis may provide families with reasons or causes for the disability which may then impact on the quality of care provided, through the recognition of associated phenotypes or the future reproductive decision-making of the person themselves and

their extended family. Statham, Ponder, Richards, Hallowell, and Raymond (2010) interviewed families where there were a number of males with significant intellectual disability and no identifiable cause, about their reasons for participating in the Genetics of Learning Disability (GOLD) study. In their results, the theme “to give the next generation choices” was present in 38 of the 80 (47.5%) interviews, suggesting that heritability of the disorder and preventing transmissibility was an important consideration and an area of concern for families.

Kay and Kingston (2002) interviewed women who were carriers of X-linked disorders such as Duchenne muscular dystrophy and Lesch-Nyhan syndrome about how they felt about passing identified genetic disorders on to offspring. They found that personal experience of the disorder, such as being a carrier or having an affected sibling, influenced decision making regarding parenthood, genetic testing and pregnancy in women. All but one of the 14 women interviewed indicated that they would avoid having an affected child either through prenatal diagnosis or through termination of affected pregnancies. Feelings of guilt were associated with the perception of responsibility for the birth and upbringing of an affected child as well as for the babies that they had lost through termination.

Genetic guilt is also hypothesised to be present in familial cases of 22q11DS. Prinzie and colleagues (2004) investigated the association between heritability, personality characteristics, and the parenting and family context, in 48 families affected by 22q11DS. This study found higher levels of marital conflict and lower warmth in the parent-child interactions in families where there was a familial deletion (N=5) compared families where the deletion occurred de novo (N=43). In explaining these results, the authors hypothesised that “genetic guilt” from passing on the disorder, leads to increased marital conflict and self-blame, which in turn results in less parental warmth. This has

implications for prospective parents with 22q11DS, based on the reported increased incidence of marital conflict and also the greater rates of abuse and interpersonal violence typically found for adults with ID. Whilst the study attempted to describe relationship characteristics associated with 22q11DS, reliance on self-reports, a small sample size and an absence of longitudinal follow-up were identified as limitations of this study (Prinz et al. 2004). Further it is unclear in the study outcomes whether the parents with 22q11DS knew of their own diagnosis before deciding to have a family. This may have been a significant factor in their reproductive decision making.

With more couples becoming increasingly aware of their genetic risk for transmitting conditions to their offspring and the increasing availability of technology which detects genetic disorders during pregnancy and before implantation, Hershberger and colleagues (2012) sought to understand the decision-making process of genetically at-risk couples. Their qualitative study interviewed 22 couples who were actively considering whether to use PGD to prevent known genetic conditions (E.g., muscular dystrophies and cystic fibrosis). Their results suggest that couples go through a number of phases or stages in the decision-making process including: Identify, Contemplate, Resolve, and Engage. In presenting this model, the authors were cognisant that this process is a complex one with many variables to consider. The degenerative nature of the disorders in the Hershberger and colleagues study (2012) is noted to be quite different to the presentation of 22q11DS in affected individuals which is varied, non-progressive and often with primary developmental concerns.

The experience of stigma

Despite the positive changes in policy and attitudes towards people with ID, as a group many adults with ID still continue to experience stigma. How individuals with ID experience stigma is an area of

research that is relatively unexplored in Western literature (Chou *et al.* 2009). The few qualitative studies that have looked at the perceived experience of stigma by individuals with ID have shown that people with ID are aware of the stigma of their disability. Edgerton (1967) interviewed adults with ID leaving long-stay hospital, and reported an awareness of stigma which resulted in attempts to overcome the spoiled identity and to pass as normal. Jahoda and Markova (2004) found that individuals with ID face stigma from being associated with specialised services such as day programs for people with disabilities, and further that they were aware of discriminatory treatment and attitudes. This awareness resulted in attempts to distance themselves from the stigma or to engage in attempts to emphasise their achievements to counteract the stigma of being associated with specialised services.

With regard to genetic diseases or syndromes, Chapple, May, and Campion (1995) observed there to be considerable stigma attached to these disorders and further that lay people lacked knowledge of genetics and inheritance. Stigma has been reported in many disorders that have a genetic component including hemophilia (Barlow, Stapley & Ellard, 2007), albinism (Wan, 2003), Down syndrome (Sebastiano, 2003), and Huntington's disease (Wexler, 2010). Despite the prevalence of stigma associated with genetic disorders, minimal research exploring the stigma from the perspective of people living with genetic risk has been undertaken (Etchegary, 2007). Research also suggests that there is significant stigma attached to mental illness (Overton & Medina, 2008). As many as 40% of adults with 22q11DS experience significant mental health conditions such as Depression, Anxiety Disorders, and Bipolar Disorder, and approximately 30% experience psychotic disorders, such as Schizophrenia (Green, Gothelf, Glaser, Debbane, Frisch, Kotler, Weizman, & Eliez, 2000). It is likely then that persons who experience the genetic deletion with comorbid mental illness are at an even greater risk of experiencing stigma associated with their condition.

The potential stigma experienced by people with 22q11DS and ID may therefore result in further barriers to achieving many of the opportunities afforded young people without disability when entering adulthood. When taken together the literature on ID, genetic disorders and mental health suggests that this double-stigma from the environment and also intra-personal attitudes may make it difficult for adults with 22q11DS and ID to form relationships, explore their sexuality and to have children.

Purpose of the current study

There is a paucity of research from the perspective of adults with ID on their experience of and meanings made from their relationships and sexuality. Current research concerning parents with ID focusses on the determinants of positive parental support and the inherent situational and contextual influences (Llewellyn, Mayes & McConnell, 2008). As the cause of ID is only identifiable in approximately two thirds of cases, demographic information on cause of ID is rarely reported in the research literature, especially on the topics of relationships, sexuality and parenting. In the case of 22q11DS, the autosomal dominant nature of the syndrome provides predictability regarding heritability, as well as opportunities for early identification through PGD testing. The 22q11DS phenotype also includes other comorbid mental health and developmental problems, adding further predictability to the developmental and psychiatric trajectory for offspring. Despite growing acknowledgement of the rights of people with ID to have relationships and bear children, the attitudes of both the community and direct care staff remain largely negative. Of interest to the current study is whether people with 22q11DS experience similar negative attitudes, or if their unique situation presents more complications or bias.

Therefore the purpose of the study is to identify, describe and understand how young adult women with 22q11DS or VCFS experience their relationships, sexuality and parenthood. As this is relatively unexplored territory, the views of young women with 22q11DS will be sought through qualitative methods, i.e., Interpretative Phenomenological Analysis (IPA). IPA acknowledges individual differences in ways of thinking, as well as the impact human interaction and wider contextual factors (E.g., culture, environment) have on the individuals' views of the world and the meanings that they ascribe to their experiences (Smith, 1995). A number of studies have been published using IPA to explore issues in the new genetics, including issues around genetic counselling and prenatal screening (Chapman & Smith, 2002) and a handful of studies (E.g., Clarkson, Murphy, Coldwell, & Dawson, 2009; Cookson & Dickson, 2010; Isherwood, Burns, Naylor & Read, 2007) are known to have utilised IPA with people with ID. IPA is judged to be the best approach for analysing the present data as we are interested not only in individual constructs and perceptions; but also on the shared meanings of relationships, sexuality and parenting for women with 22q11DS.

To conclude, research consistently reports that people with ID experience problems with maintaining social relationships (Abery & Fahnestock, 1994), have smaller social networks and experience feelings of social disconnectedness (Amado, 1993; Knox & Hickson, 2001). In addition, many do not enjoy intimate relationships, marry or, have children (May & Simpson, 2003). When adults with ID fulfil parenting roles, many experience significant barriers and stigma (IASSID, 2008). Given the value and challenges of relationships, sexuality and parenting for adults with ID, and the paucity of research from the individual's perspective, the current study seeks to use a qualitative methodology to identify, describe and understand how young adult women with 22q11DS or VCFS experience their relationships, sexuality and future parenthood, in particular, whether the possibility of passing on the deletion to future children is met with similar cognitions and feelings of guilt. Further, the perception of parental and service provider attitudes to relationships, sexuality and

parenting among the women with 22q11DS is important, to understand the quality of support experienced.

Relationships, sexuality and parenting: The experience of five young women with 22q11.2 deletion syndrome (22q11DS). An interpretative phenomenological study.

Abstract

Introduction Many people with intellectual disability (ID) experience problems with maintaining social relationships, have smaller social networks and experience feelings of social disconnectedness. A large majority do not enjoy intimate relationships, marry or have children. There is a paucity of research on relationships, sexuality and parenting from an individual's perspective. This study aims to explore relationships for women with ID and 22q11.2 deletion syndrome (22q11DS), a genetic condition associated with developmental disabilities.

Method Using Interpretative Phenomenological Analysis, this study explored the experiences and perceptions of five women with 22q11DS, regarding relationships, sexuality and parenting.

Results Four Superordinate themes were evident within the women's dialogues, including several subthemes: (i) limitations of 22q11DS (Engagement in social comparison/responsibility); (ii) acceptance/ normality (Social competence / to be a good parent); (iii) support (From Mum); and (iv) individuation (Readiness for adulthood/ with parental agreement).

Conclusions Young women with 22q11DS approach their adulthood with a sense of optimism and personal competence; with recognition also given to their own unique limitations and the benefits of parental support. The findings provide further insights into the lived experience of women with 22q11DS. Clinical implications include giving consideration to the potential risk factors and genetic counselling to address misconceptions regarding heritability.

Introduction

For most young adults, establishing social and intimate relationships, exploring sexuality, and considering parenthood can be a difficult part of an otherwise normal and expected, part of life. However, for young people with intellectual disabilities (ID), these areas can be problematic. Research consistently reports that people with ID experience problems with maintaining social relationships (E.g., Abery & Fahnestock, 1994). They often have smaller social networks and experience feelings of social disconnectedness (Amado, 1993; Knox & Hickson, 2001) and many do not enjoy intimate relationships, marry or have children (May & Simpson, 2003). When adults with ID fulfil parenting roles, many experience significant barriers including stigma (IASSID, 2008).

Individuals with ID do not form a homogenous group and much of the reported research is categorised by severity of ID, the presence of co-morbid conditions (E.g., mental health), or by specific syndromes associated with ID (E.g., Autism Spectrum Disorders). 22q11.2 deletion syndrome (22q11DS), a genetic condition associated with ID, represents one of these subgroups.

22q11DS, also known as velo-cardio-facial syndrome (VCFS), is one of the most common multiple anomaly syndromes in the general population with an incidence of approximately 1 in 4000 newborns (Hallberg *et al.* 2010). 22q11DS is inherited in an autosomal dominant manner, which means that if a parent with the deletion has a child, there is a 50% risk of passing it on to the child. In approximately 93% of individuals the deletion occurs de novo, with 7% then inheriting the 22q11.2 deletion from a parent (McDonald-McGinn, Emanuel & Zackai, 2013). The phenotype includes (but is not limited to) cardiac defects, velopharyngeal insufficiency, feeding difficulties, learning disabilities, social problems, and psychiatric disorders (Hallberg *et al.* 2010). Whilst there is significant variability

in the presentation of the deletion between affected individuals (Swillen *et al.* 2000), more than 90% of people with 22q11DS have developmental difficulties (McDonald-McGinn *et al.* 2001) with approximately half reported as having an ID (Semple *et al.* 2005). In cases where the deletion is inherited from a parent, lower full-scale IQ scores are reported, when compared to individuals where the deletion occurs de novo (Swillen *et al.* 1997). The syndrome is recognised as one of the most common known genetic causes of developmental delay (McDonald-McGinn & Zackai, 2008).

For most adults, fulfilling close relationships are linked to quality of life (Schalock, 1996) and the evidence suggests this is also true for adults with ID (Knox & Hickson, 2001). A recent paper, co-authored by adults with ID, reinforces this position affirming that close relationships with peers contribute to positive attributions of happiness and satisfaction within the lives of adults with ID (Haigh *et al.* 2013).

Adults with ID actively participate in close and intimate relationships, although some unique challenges may exist. Knox and Hickson (2001) interviewed adults with ID on the relationships they viewed as close and valued, categorising two distinct relationship types: the “good mate” and “the boyfriend/girlfriend” or partner. The “good mate” relationship was characterised by friendship, doing lots of things together, a shared sense of history, common interests and reciprocal support. The “boyfriend/girlfriend” relationship included feelings of intimacy, physical attractiveness, and with an expectation of relationship change, (i.e., marriage and children). This study highlights the ability of adults with ID to form valued, complex and dynamic relationships.

Ward and colleagues (2010) interviewed men and women with a mild to moderate ID about their romantic relationships. Those interviewed ascribed importance to these relationships, even if they spent little time with their partner. Their relationships were noted to be similar to those of their typically developing peers in terms of how they described their partner, how they spent time together, and what they did (i.e., went to the movies). Many of the adults interviewed indicated that they would like to spend more time with their partner. An experience of personal violence characterised many of these romantic relationships, with 60% of participants reporting that they had experienced some type of interpersonal violence in their past or current relationships. In the general population, 15% of Australian women have experienced physical or sexual violence from a previous partner and 2.1% from current partners (ABS, 2006b).

Sexuality is also an essential part of one's development, personality, and sense of self. Adults with ID are sexual beings (Kijak, 2011). Many misconceptions about the sexuality of people with disability exist, for example, that adults with ID are asexual, childlike, and vulnerable (Murphy & Young, 2005). However, whilst lower levels of sexual knowledge and poorer access to sexual education are often reported (Isler *et al.* 2009; Siebelink *et al.* 2006), it is recognised that individuals with ID have the same sexual needs as people without ID (Leutar & Mihokovic, 2007). The expression of sexuality can be limited by support systems which adults with ID rely on; and many adults with ID report difficulties in exercising their sexual rights (Healy *et al.* 2009). Whilst attitudes of direct care staff towards the sexuality of adults with ID are reported to be generally positive, families tend to have more conservative views (Brown, 1994; Cuskelly & Bryde, 2004)

A large majority (60-90%) of adults with mild ID express aspirations for marriage and parenthood (Bratlinger, 1985; David *et al.* 1976). Direct care staff tend to be conservative when it comes to

issues of parenting, citing concerns such as heritability of disability, parenting capacity, and the financial and health status of the prospective parents (Gilmore & Chambers, 2010). In addition, parents often report fairly negative attitudes towards their adult children with ID becoming parents (Aunos & Feldman, 2002; Cuskelly & Bryde, 2004). Despite the reservations of parents and caregivers, ID itself is not a reliable predictor of parenting performance.

Conder and colleagues (2010) identified that individuals with ID's knowledge about fertility and contraception (to either facilitate or prevent pregnancy) was low. They found that when couples were actively attempting pregnancy; only one out of six of the couples interviewed reported that they stopped using contraceptives, indicative of not fully understanding the processes of conception and/or contraception. Where parenthood is realised, some parents with ID demonstrate adequate parenting skills, whilst others require extra support (Feldman, 1994; IASSID, 2008). Barriers to successful parenting for persons with ID include poverty, prejudice, limited access to resources, respect, a lack of moral support and practical assistance (IASSID, 2008). These barriers are thought to contribute to a disproportionate number of child protection cases involving parents with ID (Mayes & Llewellyn, 2012). When adults with ID do experience success in their parenting roles, support from informal and formal structures is a key factor, along with having a positive perception of this support (Aunos *et al.* 2004).

It is clear that there are some negative and conservative attitudes towards parenthood for adults with an ID. It is possible that further stigmatisation could exist for those with an ID that is attributable to an identified genetic disorder. The notion of 'genetic responsibility' suggests that parents should seek to actively prevent the transmission of disorders to their children (Downing, 2005; Hallowell, 1999; Kelly 2009). Indeed, the heritability of specific disorders and syndromes is an

area of research that has gained significant attention recently, especially with the availability of pre-implantation genetic diagnosis (PGD) testing.

Kay and Kingston (2002) interviewed carriers of X-linked disorders such as Duchenne muscular dystrophy and Lesch-Nyhan syndrome about their feelings of passing genetic disorders on to offspring. The findings suggest that personal experience of the disorder (i.e., close relationship with an affected sibling), influenced decision making regarding parenthood, genetic testing and pregnancy. The majority (13 of 14) of women interviewed indicated that they would avoid having an affected child either through prenatal diagnosis or through termination of affected pregnancies. Feelings of “genetic guilt” were associated with responsibility for the birth and upbringing of an affected child, as well as loss for the children they had lost through termination.

Genetic guilt is also thought to be present common in familial cases of 22q11DS. Prinzie and colleagues (2004) investigated the association between heritability, personality characteristics and the parenting and family context in 48 families affected by 22q11DS. This study found higher levels of marital conflict and lower warmth in the parent-child interactions in families where there was a familial deletion (n=5) compared with families where the deletion occurred de novo (n=43). The authors hypothesised that genetic guilt from passing on the disorder led to increased marital conflict and self-blame, resulting in less parental warmth towards children. It is possible that parents with 22q11DS experience the added stress of genetic guilt more so than adults with ID.

It is clear that there is value in relationships for adults with ID and there are some unique challenges associated with this. There is a paucity of research available from the experiences of individuals with

22q11DS. The current study seeks to use qualitative methodology to identify, describe and understand how young adult women with 22q11DS experience their relationships, sexuality and future parenthood. The perception of parental and service provider's attitudes on relationships, sexuality and parenting among the women with 22q11DS will also be explored, to understand the quality of support experienced.

As research into these areas for women with 22q11DS is relatively 'unexplored territory' Interpretative Phenomenological Analysis (IPA) was judged to be the best approach for analysing the present data as we are not only interested in the individuals perceptions and experiences; but also on the shared meanings of their experiences (Reid *et al.* 2005).

IPA acknowledges individual differences in ways of thinking, as well as the impact human interaction and wider contextual factors (E.g., culture, environment) have on the individuals' views of the world and the meanings that they ascribe to their experiences (Smith , 1995). A number of studies have been published using IPA to explore issues in the new genetics, including issues around genetic counselling and prenatal screening (Chapman & Smith, 2002) and a handful of studies (E.g., Clarkson *et al.* 2009; Cookson & Dickson, 2010; Isherwood *et al.* 2007) are known to have utilised IPA to interpret data provided by people with ID.

Subjects and methods

Participants

Seven young women with 22q11DS (*de novo*) and mild ID participated in the study. The women were aged between 20 and 28 years ($M = 24$) and had not yet started a family. Two of the women self-identified as being in current heterosexual relationships and the others were single. The participants self-identified as having either an intellectual disability, or learning problems. Intellectual ability was not formally assessed through cognitive assessment; however, all participants indicated that they either received additional supports when at school or attended disability transition programs on leaving school. Six of the seven women were living with parents, whilst one young woman was living independently with regular family contact.

Selection and recruitment

Ethics approval for the study was obtained through the University of Newcastle's Human Research Ethics Committee (Approval No. H-2011-0202). Women aged 18-35 years with 22q11DS and ID, who had not yet started a family, were invited to participate in the study. Participants were sourced through services known to provide support to people with 22q11DS including the VCFS & 22q11 Foundation, Australia. Study information inviting potential participants to participate was posted on the VCFS & 22q11 foundation's website and the study outline was also presented at the annual AGM in August 2011. Recruitment was also facilitated through a VCFS & 22q11 Foundation linked Facebook site. The initial recruitment strategy attracted one participant to the study and as a result ethics approval was sought to amend the recruitment process, allowing for the research project supervisor to send Information packs directly to potential participants who had previously been involved in other research undertaken at the University. This change in the recruitment procedure

resulted in a further six participants being recruited to the study, all of whom met the inclusion criteria of the study.

Participants also needed to be able to provide verbal accounts of their experience and, not experience a severe intellectual disability or a significant mental illness (E.g., schizophrenia).

Participants were required to be able to give informed consent, by satisfying each of the three elements of informed consent as it relates to psychological research: information, competence and voluntariness (Arscott, Dagnan, & Kroese, 1998). Once the above criteria were satisfied, participants were asked to sign a Consent Form, agreeing to participate in the study. As true informed consent is difficult to establish at the outset of a study and owing to the sensitive nature of the interview questions; verbal consent was sought again throughout the interview. Participants under the care of the Office of the Public Guardian or Protective Commissioner were excluded.

Participants were also required to be able to complete the interview, without direct support from family or other support persons. One transcript was excluded from the study results as family members were involved in the interview and their presence was deemed to be a variable that was not able to be controlled for in data analysis. A second transcript was also excluded after the interview was completed, as on review all three elements of informed consent were unmet.

The interview

A semi-structured interview was utilised consistent with guidelines for IPA prescribed by Smith (1995) and Smith and Osborn (2003). Participants were given the choice as to where they were

interviewed, with all choosing the phone whilst in their own homes. Privacy and access to support people if needed was a consideration, with the interviewer discussing this with each participant at the outset. Telephone and recording facilities in a private room at the University of Newcastle, were used by the interviewer for all interviews.

Interview questions provided a guide for the interviewer to explore the topics of interest and also allowed flexibility in following and exploring the participants' ideas and information. Demographic information was collected at the beginning of each interview. The main body of the interview included questions covering experiences and perceptions of 22q11DS; history of intimate relationships and intentions for future relationships; sexual knowledge, education and behaviour; sexuality; and attitudes toward pregnancy and parenting. Examples included "Have you had a boyfriend before?"; "What is contraception for?"; and "Would you like to have a baby?" The interviewer used probing questions, such as "can you tell me more about that?" to further investigate the responses offered by participants.

Each interview lasted between approximately 45 and 90 minutes and was digitally audio-recorded with the participants' consent. Interviews were transcribed verbatim to allow for a detailed case-by-case analysis of the transcripts. To protect anonymity transcripts use pseudonyms.

Data analysis

The analytic process followed the sequence suggested by Smith (1995). Each interview transcript was repeatedly re-read and the statements which reflected participants' perceptions, meanings and

understandings on the topic noted. These statements were then organised into groups of themes. The themes derived from each subsequent interview transcript were then continuously compared and contrasted in turn against themes derived from previous transcripts until saturation was reached. Finally, a list of superordinate and subordinate themes was developed to reflect the groups' shared views and psychological processes.

Internal validity was verified through checking individual and verbatim extracts against the corresponding sub-themes and higher order themes derived (Smith, 1995). The extent to which derived themes were clearly and adequately supported by corresponding extracts was then verified by the research team. Interviews were analysed independently by two co-authors, LP and MPJ using IPA methodology as described by Smith and colleagues (2003) themes coding was then cross checked with LEC as a strategy for achieving trustworthiness and credibility, ascertaining that themes and extracts reflected the shared experiences of respondents. To begin, the transcript of the first interview was read several times by LP and annotated with initial comments, summaries and preliminary interpretations. Next these initial notes were converted into brief phrases or themes which aimed to succinctly capture the essential meaning of what the respondent had said in the interview. These emerging themes were then listed and clustered into groups of connected themes forming a list of superordinate themes for the first respondent. This process was then repeated for each subsequent transcript looking for similar themes, whilst remaining open to allowing new themes to emerge. When all interviews had been conducted and analysed, similarities and differences between them were examined, identifying superordinate and subordinate themes which the co-authors agreed reflected the shared experiences of respondents.

Results

The findings presented are based on the derived superordinate and subordinate themes, with transcript extracts also included to evidence the themes. The nature of IPA is such that only shared experiences i.e. themes found in all interview transcripts are reported. Verbatim extracts presented are merely selective representations of that shared experience. The superordinate and subordinate themes are detailed in Table 1:

Table 1 Summary of superordinate and subordinate themes.

Limitations of 22q11DS

Engagement in social comparison

Responsibility

Acceptance (normality)

Social competence

To be a good parent

Support

With support from Mum

Individuation

Readiness for adulthood

With parental agreement

Limitations of 22q11DS

Participants had an awareness and understanding of their own limitations associated with 22q11DS, notably difficulties with learning and social relationships. Positive statements accompanied the limitations indicating their acceptance of these difficulties.

Engagement in social comparison: When discussing their limitations associated with 22q11DS, the participants reflected processes of social comparison. The comparisons were made with two reference groups: their typically developing peers and those with more 'serious disabilities'. Social comparison with their typically developing peers revealed individual limitations. Participants described being "held back" which brought about feelings of sadness, especially regarding their identified difficulties.

"I don't really worry about it really, except for there's things like going out socially and trying to get a job, I think that it does hold me back a little bit..., it's not a huge part, something we have learned to get along with...it's just a thing that is holding me back".

Carol expresses acceptance of her disability through ascribing it limited significance in her life.

However, she feels 22q11DS is providing limitations, namely: having meaningful social relationships

and satisfying employment. Carol's juxtaposition of both acceptance and awareness of limitations, suggests that she is indeed concerned about the limitations and has aspirations for a fuller participation in adult activities, such as work and social relationships.

Where specific difficulties or disabilities were disclosed, participants also highlighted their acceptance of such and their adaptive coping abilities:

"...in terms of learning, problem-solving is also hard, so if someone gave me a task to do and I had never done it before,and I had to figure out a way to do it the most efficient way, ... that would be difficult and take a lot longer than most people".

Despite Michelle's resilient attitude to her limitations, her reference to "most people" self-identifies her as part of a minority group. Recognition of her own skills allows Michelle to adopt a positive attitude to her learning difficulties, as does her belief that the problem solving task is achievable.

To counteract any difficult emotions and cognitions, participants engaged in downward social comparison with other people with 22q11DS, or with people with more severe physical and intellectual disabilities. Participants communicated an awareness of different disabilities and support needs, as well as their judgement about where they fell on this spectrum. Engaging in downward social comparisons assists participants to recognise their strengths and also engenders optimism and acceptance of the deletion:

"you wouldn't want any serious problems like diseases and things...no I was probably thinking about some other things, like the diseases that make it hard for parents out there like the disabilities, that need full-time care and are dependent."

Rochelle's use of the word "serious" minimises her limitations by differentiating and contrasting 22q11DS with other disabilities. The statements reflect the participants' positive thoughts regarding their own ability through comparisons with other individuals with more significant disabilities.

Through engaging in processes of social comparison with two reference groups, the participants' position on their limitations seemed to alternate. Understanding and acceptance of their limitations had developed over time. However, with the commencement of adulthood and opportunities for parenting, sexuality, relationships and employment, there was recognition of the limitations to their full participation in all aspects of adulthood.

Responsibility: Participants expressed a responsibility to consider the transmission of 22q11DS to future generations; however, passing on the deletion did not deter plans for parenthood. The women felt the need to protect their children from similar experiences or difficulties which they perceived to be resultant from 22q11DS. In all cases, the women assumed that their children would have more significant disabilities than themselves. Value was ascribed to people with disability along with a belief of personal capacity to parent.

"actually the only other thing that would be worrying, would be me having VCFS, and all the different symptoms I have and that sort of thing. Mum has always said to me you have to be careful, like when you get pregnant you have to do all these tests. That won't stop me from having a baby, but it will just mean that it will be a longer process. Instead of the normal check-ups that other people have, I think I will be doing a lot more tests and that sort of thing".

Michelle anticipates that her experience of pregnancy and parenthood will be more difficult compared to her typically developing peers. Michelle is not deterred, though she is cautious.

Participants also considered the chance of passing on the deletion to their children. The women had varying levels of knowledge about heritability.

“like I said, it wouldn't stop me from having a baby, you know Mum had me so it can't be that hard..... I would probably go ahead with it, because I would hopefully be in a financial situation where I could have a baby with problems”.

Michelle uses her mother (who does not have 22q11DS) as a reference point, comparing between the demonstrated parenting abilities of her mother and her own perceived parenting capacity.

Michelle identifies herself as a competent element in a parent child relationship, being a protective factor and capable of caring for a child with a disability.

Responsibility was also expressed through empathic understanding of the challenges of living with a disability, either through reflection on their own experiences of the deletion or through their risk perception of heritability:

“I think particularly the autism, I wouldn't want them to have it, even though I have a really mild case of it, I wouldn't want my kids to have the full on autism stuff...I think I would feel like, that it was happening all over again, what if they have to have the surgeries that I had to, that's what I'll worry about. But the technology now you can detect it before it is even born”.

Carol raises the issue of PGD testing for the deletion, but does not go on to comment on how she would approach the outcomes of testing or whether she would have these tests. Carol expresses concern regarding transmissibility and is anxious for the children she may have; communicating a responsibility to protect them from these symptoms and expressing empathy for others who have a similar experience.

Acceptance (normalcy)

For the women interviewed there was a sense that normalcy was desirable, as perception of normality directly translated to acceptance from others and also self. Normalcy is characterised by reducing the potential for differentiation from typically developing peers and achieving acceptance through various strategies including social competence.

Social competence, as indicated by the ability to maintain friendships and participate in socially normed activities, was valued by the women and linked to their experience of acceptance. Value was given to relationships that originated during school. Longevity in these friendships was offered as evidence for competence, especially where it included typically developing peers.

“Yes I had (when at school) a good group of friends and we still catch up every weekend now, outside school now, yes we do everything, anything really...There was a fair few, half a dozen....Like we will go to a movie or go to the theatre, go out and go to concerts, on Saturday we went to a ball, we had a meal and they had a band and we just danced”.

Carol's understands that relationships differ in intensity, nature and frequency is consistent with many of the socially constructed determinants of peer friendships. Active participation in relationships is perceived as socially competent.

"I had a married couple I kept in contact with, one of my friends, she went to school with me and then got married after school, anyway they came back and I didn't realise that I hadn't told them (that I had VCFS), I thought that I would have told them at school and we were talking about it with my other friend... and they were like "what you talking about", so they had no idea that I had any of those symptoms".

Michelle's description of the couple as "married" is significant, and provides further weight to her assumption of social competence. Michelle identifies marriage as a status relationship and her ability to maintain this relationship is offered as further evidence for her perceived social competence.

Friendships that develop from current informal activities were valued:

"...there are some friends from the gym, I do a couple of classes and I have become friends, a couple from uni also".

Carol ascribes meaning to relationships that develop independently of formal structures. This reinforces her sense of social competence, and also her experience of being accepted.

To be a good parent: Of those who expressed a desire to parent, being a good parent was an important aspect. Many of the women interviewed drew upon values developed within their own immediate families, aspiring to emulate these within their own lives.

“I would like to be a nice parent, but... if they are misbehaving and all that you would have to put your foot down and really look after them, but not be too protective because you have to let them sort some things out for themselves”.

Donna recognises the complex role of parent. Donna also considers the difficulties of parenting a child with a disability, communicating a shared understanding and empathy for the experience of not feeling trusted as the child in the parent-child relationship.

Carol also designated the role of parent to include instilling morals in children:

“Teaching them what's right and having them growing up the way that you want them to grow up... probably, having discipline, but not too firm, still flexible and teaching them what my parents taught me, respect, morals and stuff like that, I think I would be like mum and dad”.

Being a good person was viewed as a potential avenue for acceptance or normality. The fragility in her social standing is also recognised in these statements. “Teaching them” and being good to other people is a perceived parenting role and thought to advantage her future children in their own lives.

Support

Support was an important theme, with the women identifying their families, particularly their mothers as primary sources of support. Support is viewed as a necessary component of having 22q11DS or any disability and is actively embraced.

With support from Mum: The support offered by mothers fulfilled a range of functions including providing support with social situations, independent living, and with their overall experience of 22q11DS.

Mothers were identified as instrumental at the time of diagnosis for 22q11DS, either being present during initial medical appointments, or through later discussing the diagnosis with the women when they were old enough to understand it:

“she made it all pretty easy, we went to a conference about it”.

For Rochelle, there is a reliance on the support provided by her mother to assist with navigating the diagnosis. The support is perceived positively and accepted as an integral component of her experience.

Participants experienced a shifting in their support needs, from the practical support of childhood, to complex levels of support inherent in adolescence and adulthood. Emotional support was now valued by the women along with practical assistance for independent living:

“I'm actually living on my own at the moment in a one-bedroom apartment. I come to their house quite often, it's kind of like I never really left home. It's good”.

Michelle is appreciative of the support offered and views the relationship with her parents as a safety net. This allows Michelle to maintain her social status of living independently with the knowledge that she will have reliable supports when needed.

Families provided social support for many of the women in lieu of peers, or through assisting the navigation of interpersonal relationships. Inherent in the role of parents as social supports is the underlying expectation that parents will provide social relationships in the absence of established or satisfying peer friendships:

“I just socialise with family friends mainly...family events, like a birthday or something”.

Recognising her own limitations with regard to peer relationships Rochelle draws on the experiences she shares with family, communicating the extent of the support she requires. Her role within the parent- child relationships is somewhat passive, assigning great responsibility to her family and recognising that she would need to take a more active role if she were to maintain relationships with her peers outside of her family networks.

When asked about who she could talk to about sexuality, Carol replied:

“mainly mum... I talk to her about pretty much everything” .

Carol experiences the support from her mother as unconditional and without limits.

To be individuated

The women expressed a desire to be individuated from their parents. Communicating to others that they had the required knowledge to achieve this individuation was also important. Making the transition to a more independent lifestyle was not only considered to be possible but planned for in many cases.

Readiness for adulthood: the women's statements reflected the various ways in which they considered themselves to be individuated from family and ready for adulthood. The women perceived that they had a more sophisticated level of understanding of adult interpersonal relationships, including sexual relationships and parenthood, which had developed at times without direct experience of such.

The knowledge base regarding relationships, sexuality and parenting varied greatly amongst the women; however, it was the desire to express their understanding and competence that was common to all the women's transcripts:

"I think it would be really rewarding in the end as you get to bring another life, another child into this world and it would be really good to raise it, because we want to try and get a house before all that stuff kind of happens".

Donna acknowledges the perceived personal benefits and positive emotions she anticipates with parenthood. Donna is identifying how parenting can contribute to her adult experience rather than just reflecting an understanding of the demands associated.

Anna's understanding of the rudimentary tasks associated with parenting is less complex; however, it still offers evidence for her readiness for adult roles.

"Looking after it, feeding it...sometimes they're fussy. You can't get them to eat what you want them to...to get them to sleep".

Participants also communicated their understanding of intimate and sexual relationships. When asked about why adults have sexual intercourse, Michelle explained:

“the first reason is if they want to have a baby, other reasons, because they care about that person and that's the only person that they would want to do it with, pleasures as well, personal sexual pleasures that people need to have filled, and if they don't they might not feel right or something, they are the only ones I can think of really”.

Michelle expresses her appreciation of the variety of individual needs that exist for each of the partners in a relationship, which may not be mutual. Intimacy is contrasted with the recognition that sexual activity may be an enjoyable physical experience for only one of the partners. Further, the physical experience is conceptualised as a “need” to be exercised, as opposed to the themes of affection and desire for parenthood, which is viewed as a choice undertaken jointly.

Despite the considered accounts and expressed knowledge, the participants also experienced some apprehension about marriage and parenting, with many discussing their personal considerations necessary before embarking on these journeys:

“I am sure when I'm older I'll probably want them (children) and after I've been in a steady relationship for a period of time... if I was to have a family I would like to be in a steady career environment with a steady income and like I said have a partner”.

Through emphasising her own individual needs and personal aspirations, Michelle communicates her ability to make independent and competent decisions; that are both consistent with her personal values and reflective of her understanding of how adequate preparation maximises parental success.

With parental agreement: The women's dialogue is also suggestive of the need to obtain parental agreement before exploring marriage and parenting. The women had prioritised the roles they planned to explore, seemingly by whether the topic had been previously discussed with parents. There appeared to be incongruence in the statements regarding the perception of their parents' thoughts regarding their choices to undertake adult roles. Participants perceived that parents would be supportive of their plans for relationships, sexuality and parenting; however, in many cases, these conversations were not held explicitly. Participants often knew their families thoughts on relationships; however, marriage and, in particular, parenting was not discussed.

The women perceived that their parents would have positive attitudes towards parenthood and marriage; however, they were not actively engaged in discussions or precursory relationships to allow for testing of these assumptions:

"I've talked a little bit about marriage and engagement and that but not too much about having a baby...yes, they know (I want to have a baby) , I already told mum...she said I can do what I like...she will probably be happy because then she'll have someone to look after as well".

Donna's statements reflect her awareness that her parents' permissibility changes in response to the increasing levels of responsibility. Further, the statements suggest underlying cognitions that Donna's decision to have a baby would be mutually beneficial for herself and her parents as grandparents and that unless there was this mutual benefit they may not be as supportive of the decision.

The absence of demonstrated parental opposition to their personal plans for parenting and marriage was perceived to equate to positive support and acceptance of achieving these roles for those who were not currently engaged in romantic relationships. Despite this, uncertainty remains about whether parenting is a role truly available to them as women who have an ID and a genetic disorder.

Discussion

The aim of the current study was to understand the lived experience of young women with 22q11DS, specifically how they experience their relationships, sexuality and parenting. As such, a purely qualitative approach was utilised to capture and reflect the narratives of the women. Analysis of the women's discourses revealed four superordinate themes relating to: limitations of 22q11DS; acceptance; support; and individuation; along with a number of corresponding subordinate themes.

The women interviewed offered insights into how they experienced their limitations associated with 22q11DS. Although the 22q11DS phenotype can vary significantly (Swillen *et al.* 2000) the most commonly reported difficulties were learning and social skills problems. One of the ways participants appeared to engage with their disability/ limitations was through processes of social comparison; that is, the process of evaluation of oneself by comparison to others (Festinger, 1954). Similarly to studies of adults with ID (Finlay & Lyons, 2000), the women in the current study engaged in downward comparison by comparing their individual strengths and abilities to those of others with more serious disabilities. Conversely, comparisons with typically developing peers then revealed limitations or difficulties associated with 22q11DS. Upward or negative social comparisons

are not a consistent finding in the literature for adults with ID, despite assumptions of their occurrence (Cooney *et al.* 2006). For the women with 22q11DS, although their difficulties were accompanied by some sadness or despondency, there was a general sense of optimism. In particular, the current comparisons were not noted to be negative, possibly owing to an absence of perceived stigma and the presence of positive self-concepts expressed by the women (Dagnan & Sandhu, 1999).

Paterson and colleagues (2012) found that when individuals with ID perceived acceptance from their own peer group and also viewed themselves as more able in comparison, they reported higher levels of self-esteem. It was thought that the women would identify other women with 22q11DS as their peer reference group; however, only one of the participants had first-hand knowledge of other adults with the deletion. Whether the reference group was 22q11DS or other women with ID, the women expressed a connectedness with their identified group and also a sense that they were more capable. Further, the statements indicated resilience, positive self-regard and self-esteem. As the themes around social comparison were revealed through qualitative analysis of transcripts, it is not possible to generalise the results to the literature, as social comparison is typically investigated via structured interview. Nevertheless, the women's engagement in multiple forms of social comparisons was compatible with the wider literature and may be the topic of further investigation.

Themes of responsibility were also evident in the transcripts regarding future offspring and 22q11DS. Many of the women were under the impression that their potential children would most certainly be affected by the deletion and that the presentation would be more severe than their own. Owing to the autosomal dominant nature of the disorder, there is a 50% chance of heritability. With the variability of expression for the deletion, it is not possible to predict whether offspring

would have a milder or more severe presentation of the phenotype. Despite raising concerns about heritability, this was not considered by the women to be a deterrent to having a family. Rather, responsibility was reflected in their commitment to meeting the individual needs of any children who may be affected by the deletion.

Responsibility, as expressed by the women in the current study, differs to the research for other genetic disorders; where genetic responsibility is expressed through a commitment to prevent the transmission of genetic disorders to children either through PGD or by choosing not to have children (Downing 2005, Hallowell 1999, Kelly 2009). Genetic responsibility was enacted by Fragile X carrier parents through avoiding conception or by choosing not to have a biological child (Raspberry & Skinner, 2011).

Feelings of guilt were not often expressed in the current study. Guilt is often a common feeling expressed by family members known to be carriers of genetic disorders (Kay & Kingston, 2002). An individual's intention for parenting, (i.e., whether it is an expressed short or long-term goal and also the perceived likelihood of such), may potentially impact on experience of guilt and expression of genetic responsibility. Imminent plans for future parenthood were not expressed by the women with 22q11DS and as such, their responses to the interview questions were largely hypothetical in nature. Another contributing factor may have been lack of knowledge about heritability and reproductive technologies. The need for better understanding of perception of heritability and intentions for parenting, along with longitudinal studies that consider imminent and future plans for parenthood, are therefore indicated in the results.

The value ascribed to normality was evident in the women's dialogues. Social competence (SC) was one such mechanism for achieving normality, referring to the ability to form and maintain personally satisfying friendships (Nezlek, 2001). Relationships with typically developing peers were offered as evidence for perceived SC in the current study. The value ascribed to relationships with typically developing peers is consistent with the research literature (McVilly *et al.* 2006). Löfgren-Mårtenson (2004) also commented on relationships with adults perceived as more able, suggesting that adults with moderate ID prefer sexual partnerships with adults with mild ID and adults with mild ID prefer to be involved with a person without an ID. In attempting to understand these preferences they suggest that engagement in such relationships assists the person with ID to avoid stigmatisation.

Young adults with ID also desire social inclusion (Emerson *et al.* 2005); however, often experience a devaluing of their social identity by peers (Szivos-Bach, 1993). Lower perceived SC is associated with higher rates of depressive symptoms (Gable & Shean, 2000); and higher rates of depression are found for both children (Antshel *et al.* 2006) and young adults with 22q11DS (Green *et al.* 2009). The value assigned to SC by adults with ID and an experience of being devalued by peers, combined with vulnerabilities for depressive symptoms, presents a potential risk factor for young adults with 22q11DS.

Being a good parent was an important aspect for the women who expressed intentions for parenting. The parenting values were often derived through the perceived exemplary behaviour of their parents and extended family. Being a good parent was also viewed as a possible mechanism for ensuring acceptance from others. For many parents, positive models of parenting are learned through first-hand experiences of being parented themselves; however, many people with ID lack these support systems (IASSID, 2008). The women with 22q11DS all had a positive perception of

their family support and viewed their own parents as competent caregivers. Llewellyn (1997) found that learning from family parenting traditions, was instrumental in developing parenting behaviours for parents with ID. Parents with ID who viewed their own childhoods positively, often carried out parenting roles in a similar manner to how they were parented. Results of the current study highlight the women's aspirations to emulate the parenting behaviours of their own parents and also the positive regard for the values instilled by their parents. The women expressed similar intentions for parenting to the parents in the Llewellyn study; however, the themes were generated from prospective thoughts on parenting rather than actual experience.

The women's dialogues reflect the value ascribed to family support; with mothers identified as the primary source of this support. In addition to providing the practical support, especially at time of diagnosis, mothers were also conceptualised as friends by the women in the current study. The results are consistent with the wider literature which finds mothers are more actively involved in the direct care needs of their adult children with ID, as compared to fathers (Rowbotham *et al.* 2011).

A positive perception of support is consistent with the research literature for adults with ID (Haigh *et al.* 2013; Keogh *et al.* 2004; Ward *et al.* 2003). However, support from family is not universally considered favourably by adults with ID. Walmsley (1996) also described parental support for adults with ID. Support was experienced either positively, that is, very supportive or mutually supportive; was characterised by role reversals where parents were dependent on children; or in some cases the support was perceived negatively and inclusive of significant levels of conflict regarding choices for independent living (Walmsley, 1996). There are some notable differences between the participants in the current study and the adults who participated in the Walmsley study. The mean age of the women with 22q11DS was 24 years, they were mostly without children or partners, and mostly living

at home with parents. This is contrasted with older participants (mean age 43), more life experience (including parenthood and independent living) and both genders. The discrepancy in findings, notably the unanimous positive regard for support from mothers expressed in the current study, indicates the need for longitudinal studies, which are considerate of age and life experience, and inclusive of males with 22q11DS.

Themes of individuation from family and aspirations for independence were also contained in the women's dialogues. Individuation from family and establishing one's independence has traditionally been recognised as a key developmental task in the transition to adulthood (Erikson, 1968); however, for adults with ID the transition to adulthood has been described as 'extended, partial and with a stable and dominant identity (of ID) continuous across the years' (Baron *et al.* 1999, p496). The women in the current study communicated their understanding of the more complex aspects of adulthood (relationships, sexuality and parenting), and in doing so believed this inferred a readiness and competence for adulthood. In many cases, knowledge of adulthood was not expressed through direct experiences, rather through knowledge and experience gleaned from the collective experiences of peers and family, formal and informal education, and their understanding of social norms.

The transition to adulthood after formal schooling can be an especially stressful experience for young adults with developmental disorders (Heal *et al.* 1998) and may trigger depressive experiences for adults with 22q11DS (Green *et al.* 2009). Whilst mood and psychological well-being was not directly assessed as part of the current study, the women expressed positivity and optimism, perceiving opportunities for change and goal attainment. Consistent with the wider research, the transition to adulthood was ongoing and contained opportunities for support.

There was a perception that parents were supportive of the longer-term plans for adult roles expressed by the women with 22q11DS, inclusive of marriage and parenthood. The women recalled having conversations with their parents about plans for relationships and marriage; however, discussions regarding parenthood were conspicuously absent. The literature reports fairly conservative parental attitudes towards sexuality and parenthood by adults with ID (Cuskelly & Bryde, 2004). With regard to relationships and sexuality, family members typically indicate their preference for less intimate relationships (Evans *et al.* 2009; Löfgren-Mårtenson, 2004); express concerns regarding sexual vulnerability; and introduce fewer topics of sexual education at later stages as compared to typically developing peers (Pownall *et al.* 2012). Further, heritability of ID and preventing transmissibility are important considerations for the families affected by learning disabilities (Statham *et al.* 2010). The research literature for sexuality and parenting by adults with ID is inconsistent with the perceptions held by the women in the current study. As such, a future area of interest concerns parental attitudes to relationships, sexuality and parenthood for adults with 22q11DS and the accuracy of these attitudes as perceived by their adult children.

Considerations

The study aimed to explore the lived experiences of young women with 22q11DS who also presented with ID, with regard to their personal relationships, sexuality and parenthood. The accounts of five participants with 22q11DS form the basis of the reported results. Whilst the themes are thought to be representative of the women interviewed, they do not account for all women with 22q11DS. Future studies might also include the views of men with 22q11DS and also additional quantitative measures to further explore the current data. The current study was prospective in nature with regard to relationship and parenting intentions. Further understanding of the experiences of women with 22q11DS could be gained from considering these results with additional

longitudinal or retrospective studies involving participants who had actively undertaken these adulthood roles or who had made conscious decisions not to explore them. Despite attempts to homogenise the group, the women presented with a diverse range of abilities and experience. As such, cognitive level and personal experience with relationships, sexuality and parenting, may have influenced the development of themes and a more heterogeneous group may have resulted in more confident findings. Formal assessment of cognitive ability may also have further homogenised the group.

Optimal research conditions for adults with ID include opportunities for face-to-face meetings and multiple interview sessions where necessary (Booth & Booth, 1996). Some of the participant responses were at times brief and limited contact with the interviewer prior to the interview may have engendered social desirability. Whilst acknowledgement is given to these recommendations, some participants suggested that their responses were less censored and more honest due to the anonymity provided by phone interviews. Lastly, IPA as a qualitative research method with adults with ID is still emerging. The participant responses were at times brief and limited and this may have affected the process of analysis.

Clinical Implications

Mental health concerns are part of the 22q11DS phenotype and the present findings highlight some risk factors for women with 22q11DS. Transition to adulthood and difficulties in peer relationships trigger depressive symptoms in young adults with 22q11DS. Whilst the women had an awareness of personal difficulties associated with 22q11DS, limited consideration was given to the potential

mental health challenges they may face. The need for proactive monitoring of the mental health by service providers and families is indicated for adults with 22q11DS as they transition to adulthood.

In recognition of the rights of adults with disabilities to have children and the expressed intentions of the participants', the importance of providing information on family planning is indicated. Many of the women did not have an accurate understanding of heritability; however they all held concerns about transmissibility. Psycho-education or genetic counselling regarding risk and management of transmissibility is recommended to allow for informed decision making regarding parenthood.

Parenting capacity and factors which maximise parental outcomes for parents with ID is also an important consideration for service providers and family members. Direct care staff have reported concerns regarding parenting by adults with ID, including heritability of disability, parenting capacity, and the financial and health status of the prospective parents (Gilmore & Chambers, 2010). These considerations are important for all prospective parents and require consideration by parents with ID and their support systems to ensure parenting outcomes are maximised for both parents with ID and their children. The women's positive perception of family support and values for parenting are noted as protective factors and important considerations in future family planning.

Conclusion

This study has begun to explore how women with 22q11DS experience their relationships, sexuality and parenting. The women with 22q11DS in this study had positive perceptions of support and faced their futures with optimism. They expressed a desire for normalisation and a strong sense of self-determination, in a context of perceived family support. Consideration and further understanding of the unique experiences of women with 22q11DS, including the acknowledgement of expressed

desires and concerns regarding relationships, sexuality and parenting will invariably assist young women with the deletion to achieve fuller and meaningful life roles for themselves and their future families.

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Appendices

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|-------------------|---|
| Appendix 1 | Results: Additional Participant Background/ Demographic data |
| Appendix 2 | Semi-structured Interview Schedule |
| Appendix 3 | Ethics NEAF Application |
| Appendix 4 | Participant Information Statement |
| Appendix 5 | Project Flyer |
| Appendix 6 | VCFS Website Information |
| Appendix 7 | Participant Letter (Recruitment) |
| Appendix 8 | Consent Form |
| Appendix 9 | Aims and Scope of Journal for Submission (including relevant submission guidelines) |

Results: Additional demographic data for 5 participants.

| | |
|---|---|
| No. of participants with boyfriends: | 2 |
| Participants who have not had a romantic relationship (past or current): | 2 |
| No. with current peer group/ relationships: | 3 |
| No. live with family: | 4 |
| No. Live independently (no support service): | 1 |
| No. using contraception (i.e., the pill or Depo Provera) | 5 |
| No using for contraceptive reasons: | 2 |
| No. wanting to have children: | 2 |
| Knowledge of heritability: Transmissibility is: <ul style="list-style-type: none"> • a possibility: (111 • almost certain:1 • unknown: (1 | <ul style="list-style-type: none"> • 3 • 1 • 1 |
| Highest level of education: University: TAFE/ short courses: High school: | 1 (current enrolment) 3 1 |
| Vocational experience/s: Post School Options: Supported employment: Previously employed or current: self-employed: Working for family: | 1 1 2 1 1 |

