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**DISORDERS OF SEX DEVELOPMENT:  
DEVELOPMENTAL CHALLENGES AND MOTHERS'  
EXPERIENCES OF SUPPORT.**

**Section A:** What is currently understood about the impact of Disorders of Sex Development on the Psychosocial and Psychological Development of young people?

Word Count: 8,091

**Section B:** Disorders of Sex Development: Mothers' Experiences of Accessing Support

Word Count: 8,511

**Section C:** Appendices of Supporting Material

Overall Word Count: 16,602

A thesis submitted in partial fulfilment of the requirements of  
Canterbury Christ Church University for the degree of  
Doctor of Clinical Psychology  
April 2014

SALOMONS  
CANTERBURY CHRIST CHURCH UNIVERSITY

## Acknowledgements

Firstly, I would like to thank all the mothers who participated so openly in this research and who enabled me to learn so much. I valued their honesty and openness and without them this research would not have been possible. I would also like to thank my two supervisors, Professor Jan Burns, and Dr Martha Deiros-Collado for offering their expertise, guidance, and encouragement throughout the project. Lastly, to Paul, my family, and my friends, who have supported me endlessly throughout this time. I cannot thank you enough.

## **Summary of the MRP portfolio**

**Section A** is a review of the literature relating the psychosocial and psychological development of young people with Disorders of Sex Development (DSD). The literature is summarised across four areas; psychosexual development, social and sexual adaptation, cognition, and psychological wellbeing. The findings of the review are discussed in relation to developmental theory. Methodological limitations are discussed and gaps in the research are outlined. Recommendations for further research are made.

**Section B** describes a qualitative study which explores the support experiences of mothers of children with DSD. Semi-structured interviews were conducted with eight mothers and the results were analysed using Interpretative Phenomenological Analysis. Four master themes were identified and these are discussed in light of existing research. Limitations of the study are outlined along with areas for future research. The clinical implications are discussed.

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**Section A: Literature Review**

What is currently understood about the impact of Disorders of Sex Development on the Psychosocial and Psychological Development of Young People?

Word Count: 7662 (429)

A thesis submitted in partial fulfilment of the requirements of  
Canterbury Christ Church University for the degree of  
Doctor of Clinical Psychology  
April 2014

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CANTERBURY CHRIST CHURCH UNIVERSITY



### Abstract

Disorders of Sex Development (DSD) are a range of congenital conditions in which the development of chromosomal, gonadal, or anatomical sex is atypical. DSD are often identified at birth when a baby is born with ambiguous genitalia, and occur most frequently because there is a problem with the genetic make-up or the body does not respond to sex hormones in the correct way. DSD present families and clinicians with many distinct demands and the child faces a number of unique developmental challenges. This review provides a summary of the literature relevant to the psychosocial and psychological development of young people with DSD. Relevant papers were identified following a systematic search of the electronic databases Science Direct, Medline, BioMed, and PsycInfo, using a variety of key terms relevant to DSD, and psychosocial and psychological development. Thirty papers relevant to psychosexual development, social and sexual adaptation, cognition, and psychological wellbeing were identified for the review. While overall many inconsistent findings were uncovered, there is a significant body of evidence indicating that some developmental tasks of childhood and adolescence may be impacted by DSD. These include the development of gender role, aspects of social functioning including peer, and intimate relationships, and psychological adjustment. Methodological concerns are discussed along with recommendations for future research.

*Keywords:* Disorders of sex development, psychosocial development, psychosexual development, mental health



## What is currently understood about the impact of Disorders of Sex Development on the Psychosocial and Psychological Development of Young People?

The congenital conditions classified as Disorders of Sex Development (DSD) represent a potential disruption to the typical psychosocial development of children and adolescents. This review, of empirical research, provides a summary of what is currently understood about the impact of DSD on psychosocial and psychosexual development, as well as psychological wellbeing. Important gaps in this understanding are identified, as well as recommendations for future research.

### **Developmental Theory**

Theories of child development have attempted to provide a framework for thinking about the transitions involved in human growth, development, and learning. Developmental theories provide guiding principles which describe and explain typical human development and are considered essential to allow an appreciation of the growth that children go through from birth into early adulthood including cognitive, emotional, physical, and social growth.

A variety of frameworks exist that describe human development. While some focus on a specific developmental domain, for example moral development (Kohlberg, 1984), others describe processes of growth that occur across the lifespan, such as Erikson's theory of psychosocial development (Erikson, 1956). Erikson's theory of psychosocial development is well supported by empirical research and can be applied across different genders and cultures (Newman & Newman, 2011). A key component of Erikson's stage theory is the development of identity: the conscious sense of self which is developed through social interaction with others and which is constantly changing due to new experiences. Erikson proposed eight stages of development, each of which is concerned with becoming competent in an area of life through the resolution of a specific conflict. Each stage is associated with developmental tasks which reflect accomplishment in physical, cognitive, social, and emotional development

(Newman & Newman, 2011). The development of identity, which begins with the infant's discovery of self, continues throughout childhood and becomes the focus of adolescence, is considered to include physical and sexual identity, occupational goals, religious beliefs, and ethnic background (Marcia, 1991). Adolescents are thought to explore these dimensions, making commitments to aspects of their identity as they move into early adulthood. While adolescence is an important phase in the development of identity, gender identity theory (Kohlberg, 1984) suggests that an individual's understanding of gender moves forward in stages, starting in childhood. By the age of two, the child is thought to be able to recognize its own gender, and by age four, to recognize that gender is something which remains the same over time. The third stage, reached by around age seven, is gender constancy in which the child begins to realize that gender is independent of external features. The physical changes associated with puberty initiate adolescents' exploration of their physical and sexual identity before a sexual identity is committed to (Marcia, 1991). Another important aspect of development is the development of peer relationships and the later development of intimate relationships. While there are several perspectives on this, Sullivan (1953) proposed that there are five basic social needs across the period from infancy to adolescence which include tenderness, co-participation in playful activity, acceptance by others, interpersonal intimacy, and sexual contact. Sullivan (1953) proposes that these needs are fulfilled by specific individuals- parents, peers, same sex best friends, and sexual partners, and this is considered a sequential and cumulative process. Sullivan (1953) proposes that the period of late childhood (aged six to nine years) is marked by the increased need for approval from peers and the need for relationships which are equal. During the later pre-adolescent years (nine to twelve years) the needs of relationships shift from this more general need for group approval to a need for a close and intimate relationship with an individual same sex peer. Sullivan (1953) suggests

that the onset of puberty brings with it the need for sexual contact and intimacy in the opposite sex friendship.

While many individuals will move through the developmental stages without difficulty, resulting in a cohesive sense of self, for others the completion of developmental tasks may be delayed or interrupted resulting in a sense of inadequacy, or poor sense of self. Such deviations from the typical path of development have been used to offer perspectives on difficult childhood behaviours, emotional difficulties, and poor psychological wellbeing (Kroger, 2007). Therefore, understanding possible factors which may lead to interruptions during development is important in preventing and reducing emotional distress.

Life-long childhood health conditions are one important factor which can result in disruptions to typical childhood psychosocial development (Mattson, 1972). The development of independence, a sense of self, and peer and intimate relationships, have all been reported as disrupted or delayed in individuals with chronic health conditions (Suris, Michaud, & Viner, 2004). With this in mind, childhood health conditions are likely to have important implications for psychological wellbeing, and among other things, health difficulties have been identified as a risk factor for emotional difficulties (DeHart, Sroufe, & Cooper, 2000).

### **Impact of Childhood Health conditions on Wellbeing**

Research evaluating the impact of on-going illness in childhood has demonstrated that in addition to the physical effects, there are a number of psychological consequences which may have a significant impact on children's overall quality of life (QOL) (Shaw & Páez, 2002). Difficulties have been identified such as low mood, poor educational attainment, social withdrawal, peer conflicts, and problems with adaptive behaviour (Shaw & Páez, 2002; Silver, Westbrook, & Stein, 1998). Additionally children with on-going health conditions are at greater risk of developing a diagnosable mental health problem (Pless & Nolan, 1991).



Accordingly, research has sought to develop a more thorough understanding of the factors which promote good adaptation for the child. Interpersonal factors within the family, family cohesion, parenting satisfaction, socio-economic resources, and social support, have all been positively associated with good psychological adjustment in children with health problems (Hamlett, Pellegrini, & Katz, 1992). Additionally parental adaptation to their child's health condition is considered key for the subsequent wellbeing of the child (Carmichael & Alderson, 2004). Such findings highlight the potential risks to child emotional wellbeing in the presence of an ongoing health condition. Many health conditions are well researched in this regard, including diabetes, epilepsy, heart disease, and cancer, however other health conditions have received relatively little attention concerning psychological impact and factors associated with positive adjustment. One such condition is Disorders of Sex Development.

### **Disorders of Sex Development**

The congenital conditions classified under the term Disorders of sex development (DSD) occur when the development of the chromosomal, gonadal, or anatomical sex is atypical (Lee, Houk, Ahmed, & Hughes, 2006). Such disorders occur because there is a problem with the genetic make-up and/or the body does not respond to sex hormones in the correct way. There are many variants of DSD and the current classification system (Lee et al., 2006) distinguishes between those who are genetically female (46,XX DSD), those who are genetically male (46,XY DSD), and those whose sex chromosomes are neither typically male or female, (Sex Chromosome DSD). These will be considered further below. For a more detailed description of different DSD please refer to Wisniewski, Chernausek, and Kropp (2012).

**46,XX DSD.** Here, the individual possesses the usual number of chromosomes (46), and the sex chromosome of the individual is female- XX. However, the external genitalia

may appear male or enlarged due to a process called virilisation<sup>1</sup> caused by excess androgen exposure. The most frequently diagnosed 46,XX DSD is Congenital Adrenal Hyperplasia (CAH) in which individuals presents with varying degrees of virilisation, and in its most serious form, individuals are unable to store salt in the body.

**46,XY DSD.** Here, the individual again possesses the usual number of chromosomes, and the sex chromosome of the individual is male-XY, however the external genitalia may appear female or ambiguous. Additionally, testes may be absent or not properly formed. While 46,XY DSD can occur for a variety of reasons, it is generally attributed to the inability either to produce, or respond to testicular hormones during foetal life. More frequently occurring diagnoses in 46,XY DSD are Partial Androgen Insensitivity Syndrome (PAIS) or Complete Androgen Insensitivity Syndrome (CAIS), and CAH can also be diagnosed in this category.

**Sex Chromosome DSD.** This refers to a number of DSD in which the individual does not have either normal male (XY) or female (XX) sex chromosomes. Examples are Klinefelter's in which a male has an additional X chromosome, known as XXY, or 47,XXY, and Turner Syndrome affecting females, where one, or part of one, of the X chromosomes is absent or has abnormalities.

While some children affected by DSD are identified at birth others will not present until adolescence when it is noticed that their pubertal development is unusual in some way (Ahmed et al., 2011). Although the presentation and course of treatment for every individual with DSD varies, treatment can be challenging for both the family and the individual. At birth, decisions may be required about the gender in which the child should be raised. There

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<sup>1</sup> Virilisation refers to the development of adult male physical characteristics in a genetically female individual or a young boy. Specifically, a genetically female baby develops male (complete virilisation) or ambiguous genitalia (partial virilisation).

may be a need for surgery to protect the child from serious health risks if organ development is incomplete, and some DSD will require daily medication over the life course. Genital reconstructive surgery may take place within the first one to two years of life, and/or around the time of puberty. Decisions around the use of sex steroid replacement and delaying puberty are necessary during early adolescence, and consideration also needs to be given to psychosocial factors for the child (Lee et al., 2006). While figures on the incidence of DSD vary, a recent estimate reports that DSD occur in every 1-2:10,000 live births (Woodward & Patwardhan, 2010).

### **Impact of DSD on Psychological Wellbeing**

DSD present a range of unique challenges to parents, young people, and professionals (Brain et al. 2010). Due to the nature of the medical interventions which may be required, research on outcomes has centred on medical issues such as sex of rearing and surgical outcomes, including functionality and appearance of genitalia. Longitudinal psychological research has tended to focus on gender identity and sexuality, with less attention to satisfaction of relationships and overall quality of life (QOL) and wellbeing (Sandberg, 2012).

The Consensus Statement for DSD (Lee et al., 2006) has set out a number of guiding principles with regard to research and identifies that outcomes in DSD should consider a range of psychological outcomes including sexual function, social and psychosexual adjustment, mental health, QOL and social participation. Psychological research which has considered the impact of DSD on wellbeing has reported mixed findings. Johannsen, Ripa, Mortensen, and Main, (2006) reported that those with DSD had an overall poorer QOL, and Scützmann, Brinkmann, Schacht, and Richter-Appelt (2009) found that individuals with DSD may experience higher levels of distress, deliberate self harm, and suicidal tendencies in adulthood. Conversely, Berenbaum, Bryk, Duck, & Resnick (2004), reported that

psychological adjustment in adults with CAH was no different to that of unaffected adults. Qualitative research has begun to reveal particular aspects of DSD and its care which have resulted in feelings of shame and isolation as a result of repeated medical examinations and surgery. The negative impact that secrecy about the diagnosis has on relationships has also been reported (MacKenzie, Huntington, & Gilmour, 2009; Sutton et al., 2006).

### **Development and DSD**

It has been acknowledged that children with DSD encounter developmental hurdles that are both unique and emotionally challenging (Schober et al., 2012). This is reflected in the need for long term multi-disciplinary care (Lee, et al., 2006). Psychosocial developmental tasks such as peer play, friendship development, membership in peer groups, accepting physical maturation, the development of romantic and sexual relationships, and the development of gender identity, are likely to be particularly challenging for individuals with DSD. These developmental tasks, which span from early school age through to later adolescence, have been recognised by the Consortium on the Management of DSD (2006), as potential areas of difficulty. The impact of completing such developmental tasks successfully on emotional wellbeing has been highlighted (Wenar & Kerig 2000), and with this in mind, understanding more about the impact of DSD on particular developmental tasks is important in furthering our understanding about its long-term impact on individual wellbeing and QOL.

Although several recent reviews have focused on the QOL and psychological outcomes for adults with DSD (Malouf, Inman, Carr, Franco, & Brooks, 2010; Zainuddin, Grover, Shamsuddin, & Mahdy, 2013), there are no current reviews which focus on child, adolescent or young adult populations. Understanding what is currently known about the impact of DSD on child and young adult development is important in providing clarity about specific developmental tasks which pose additional challenges for young people affected by DSD. It will also ensure a better understanding of long-term outcomes. Understanding if and

when these children may be more vulnerable will enable better targeted support, improving outcomes for individuals with DSD. This systematic search and review (Grant & Booth, 2009) therefore aims to establish what is currently understood about the impact of DSD on aspects of psychosocial and psychological development by drawing together literature specific to child, adolescent and young adult populations, and across different DSD.

The main aims of the review are:

- 1) Drawing on empirical research, what is known about the impact of DSD on psychosocial developmental tasks?
- 2) How have DSD been shown to impact upon child, adolescent, and young adult, psychological wellbeing?

### **Method**

A systematic search (Appendix A) of the databases Science Direct, Medline, BioMed, and PsycInfo was performed. The key search terms of disorders of sex development, intersex, hermaphrodite, ambiguous genitalia, congenital adrenal hyperplasia, and androgen insensitivity syndrome were used. These terms were combined with a range of psychological and psychosocial developmental terms to identify relevant papers. Publications from 1955 to March 2014 were included and search results were screened, initially by looking at the abstracts to determine relevance to the topic area and, where necessary, viewing the participants section to determine the age range of participants. Only those papers published in peer reviewed journals in English, were considered for inclusion. Papers which were based on samples of children, adolescents and young adults, up to the age of 25 years, were included in the review. Individual case studies were excluded, as were those which focused solely on medical outcomes and the physicality or satisfaction with surgical outcomes.

## Literature Review

A total of 30 papers were included in the review (Appendix B). Relevant papers were identified as covering four main topics relevant to psychosocial development. These topics are psychosexual development, social and sexual adaptation, cognition, and psychological wellbeing. Many of the included papers provided information relevant to more than one of these domains. The literature relevant to each domain will be considered in turn below.

### Psychosexual Development

It has been suggested that a person's psychosexual development consists of three parts: gender identity, gender role, and sexual orientation. Gender identity refers to whether the individual views and presents them self as male or female. Gender role relates to aspects of behaviour and personality which are typically attributed within society to males or females, i.e. play choices and aggressive behaviour. Sexual orientation concerns a person's focus of sexual interest, attraction and fantasy (Hughes, Houk, Ahmed, & Lee, 2006). Gender role is the most extensively researched area and thus this will be considered first.

**Gender role.** The research in this area with young people predominantly focuses on childhood play preferences, with fewer studies considering gender typical behaviours, activities and interests, and childhood drawings. The majority of studies in this area have focused on individuals, both male and female, with CAH, drawing comparisons between those with CAH and unaffected controls, typically consisting of siblings.

Studies have consistently demonstrated that girls with CAH have a greater interest in masculine toys and play activities, and less interest in feminine toys and play activities than unaffected females (Berenbaum & Hines, 1992; Berenbaum & Snyder, 1995; Meyer-Bahlburg et al., 2004; Oner et al., 2009; Pasterski et al., 2005; Pasterski et al., 2011; Servin, Nordenström, Larsson, & Bohlin, 2003). Girls with CAH have also been found as more likely than unaffected girls to report having boy playmates (Pasterski et al., 2011; Servin et

al., 2003), and to choose playmates that are engaged in male-typical activities (Pasterski et al., 2011). In addition it has been shown that adolescent girls with CAH show increased interest in male-typical activities and careers, and less interest in female typical activities and careers (Berenbaum, 1999; Servin et al., 2003). Studies which have looked at behaviour in children and adolescents with CAH have also consistently reported that females with CAH are more aggressive (Berenbaum & Resnick, 1997; Pasterski et al., 2007) more active (Pasterski et al., 2007), behave in a more 'boylike' manner (Servin et al., 2003), and display less interest in infants (Leveroni & Berenbaum, 1998) than unaffected girls. Sex differences in the free drawings of girls with CAH have also been reported (Iijima, Arisaka, Minamoto, & Arai, 2001) where CAH girls' drawings display more masculine characteristics than unaffected girls. Such findings in females with CAH are attributed to prenatal androgen excess and are thought to support the hypothesis that early androgen exposure has a major effect on gender related play and interests (Berenbaum, 1999). While few studies have accounted for the effects of socialisation, a recent study by Pasterski et al. (2005), reported that more frequent positive feedback for play with girls' toys was received by girls with CAH, in comparison to unaffected girls. They concluded that male typical toy play by girls with CAH cannot be attributed to parental encouragement of male typical play, further enhancing support for the role of androgen exposure in gender related play and interests.

Overall boys with CAH have not been shown to differ from unaffected boys in their preferences for male or female typical play (Berenbaum & Hines, 1992; Berenbaum & Snyder, 1995; Pasterski et al., 2005), male and female typical activities, or gender typical careers (Berenbaum, 1999; Servin et al., 2003). This lack of a difference between boys with and without CAH is explained by the very similar level of foetal androgen exposure of these two groups; i.e., the androgen levels of boys with CAH are not outside the typical range observed in unaffected boys.

Only one study was identified which looked at gender role differentiation in DSD, other than CAH. Jürgensen, Hiort, Holterhus, and Thyen, (2007) reported that those with DSD and reared as female were more likely to express an interest in male typical activities than the control group females on a measure of gender typical behaviour and attitudes. Jürgensen et al. (2007) concluded that androgen exposure in the pre- and postnatal period plays a decisive role in the development of gender-typical behaviours in children.

**Gender identity.** Seven studies were identified which reported on gender identity in samples of young people (Crawford, Warne, Grover, Southwell, & Hutson, 2009; Gupta, Bhardwaj, Sharma, Ammini, & Gupta, 2010; Jürgensen et al., 2007; Liang et al., 2008; Mayer-Bahlburg et al., 2004; Oner et al., 2009; Slijper, Drop, Molenaar, & de Muinck Keizer-Schrama 1998) and have used a range of measures to assess indicators of gender dysphoria. Two studies, with small samples, reported that a minority of children in their sample expressed or displayed behaviours which may indicate gender dysphoria (Crawford, Warne, Grover, Southwell, & Hutson, 2009; Liang et al., 2008). Liang et al. (2008) also reported a correlation between atypical sex-type behaviour in childhood and subsequent gender dysphoria. In a larger sample of 59 individuals with DSD, Slijper et al. (1998) reported that 13% of girls, but no boys, were identified as having gender identity disorder based on Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition (DSM-IV) criteria. However, indicators of gender confusion or dysphoria, and associations between gender atypical play and gender confusion, have not always been reported in samples of young people with DSD. Mayer-Bahlburg et al. (2004) compared girls with CAH to unaffected siblings and detected increased levels of male typical play in girls with CAH using the Child Game Participation Questionnaire (CGPQ; Meyer-Bahlburg, Sandberg, Dolezal, & Yager, 1994), but did not identify any signs of gender confusion in the girls with CAH using the Child Behaviour and Attitudes Questionnaire (CBAQ; Meyer-Bahlburg, Sandberg, Yager,



Dolezal, & Ehrhardt, 1994). This finding was replicated by Jürgensen et al. (2007) using similar measures. Oner et al. (2009) reported that in a sample of females with CAH, although elevated masculinisation scores were reported on the Gender Identity Questionnaire (GIQ; Johnson et al., 2004), no participants met the criteria for a diagnosis of gender identity disorder. The authors concluded that while prenatal androgen effects lead to the masculinisation of subsequent gender related behaviour, increased gender-identity confusion/dysphoria was not reported, indicating that gender identity is not directly determined by prenatal androgens. A final study reporting on gender identity assessed satisfaction with gender assignment. Gupta et al. (2010) reported that 85% of males with DSD felt satisfied with their gender assignment, and 100% of their parents were satisfied with the gender assignment of their child.

**Sexual orientation.** The only study identified to comment on sexual orientation in DSD (Gupta et al., 2010) took place in India, and reported that 85% (51/60) of the all male sample identified with a heterosexual orientation, 7% were undecided, and 8% did not wish to answer the question.

In summary, studies reporting on psychosexual development in DSD have consistently reported an increased occurrence of gender atypical play, interests, and activities in girls with DSD as a result of increased androgen exposure in the prenatal period. Mixed findings are reported with regard to gender dysphoria, however even among studies which did report possible gender dysphoria, this seemed to be for a small minority of individuals. The majority of research did not support associations between increased gender atypical activity and increased gender dysphoria.

### **Social and Sexual Adaptation**

Six studies were identified as reporting on either social functioning, or the development of intimate relationships. Four reported on the social function of individuals

with DSD (Gupta et al., 2010; Zhu et al., 2012) or CAH (Gordon, Lee, Dulcan, & Finegold, 1986; Sanches, Wiegers, Otten, and Claahsen-van der Grinten, 2012), and three on intimate relationships and sexual behaviour among those with DSD (Gupta et al., 2010; Kleinenmeier et al., 2010) or CAH (Liang et al., 2008).

**Social adaptation.** In the first of the studies (Gupta et al., 2010) which looked exclusively at those with DSD and male gender assignment in India, 60 young people, aged between 15 and 25 years, were interviewed about aspects of psychosocial adjustment. Peer relationships were considered 'good' by 72% participants, and 'poor' by 12%. However 70% of the sample reported apprehensions about their future, including employment, marriage, and acceptance in society, and only 15% felt that they fitted into society. Similarly, in a Chinese study, Zhu et al. (2012) identified that boys and girls with DSD, aged between 6 and 17 years old, reported social limitations as measured by the socio-emotional sub-scale of the Child Behaviour Checklist (CBCL; Achenbach, 1991). Specifically, it was reported that four of eight boys with DSD exhibited social limitations, including having few friends, poor communication with family and peers, and academic struggles, in comparison to a control group of boys who did not report these difficulties. Of the 42 girls with DSD in this study, 28.6% showed social limitations, had poor communication, and poor academic achievement. However the scores in this group were not significantly different from girls in the unaffected control group, indicating that although girls with DSD may experience social limitations, they are no more frequent than those reported by unaffected adolescent girls.

Studies focusing on individuals with CAH reported different results, with social functioning largely unaffected (Gordon et al., 1986; Sanches et al., 2012). In an American study which assessed the social competency of girls, aged 6 to 16, using a subscale of the CBCL, no difference was reported between those with CAH and a control group (Gordon et al., 1986). Likewise, results from a Dutch study of boys and girls under the age of 18 with

CAH demonstrated that social participation, as measured by a specifically designed questionnaire, was similar to that of unaffected peers, and only 8% of parents felt that CAH constrained their child's daily life (Sanches et al., 2012).

**Sexual adaptation.** Three studies were identified which provided information about intimate relationships and sexual behaviour (Gupta et al., 2010; Kleinemeier, et al., 2010; Liang et al., 2008).

In a Taiwanese sample of 11 females with CAH, aged between 8 and 25, none had experienced sexual intercourse or were currently involved in an intimate relationship (Liang et al., 2008). Given the age and size of the sample this does not seem unreasonable. Gupta et al. (2010) reported that in a sample of 60 males, 15 to 25 years, with DSD, two were married. A further 26 reported they would consider marriage in the future, however the majority of participants reported they would not consider marriage due to a fear of being rejected because of infertility or a small phallus. In a more detailed assessment of sexual activities in adolescents aged 13 to 16 with DSD from Germany, Switzerland, and Austria (Kleinemeier et al., 2010) no overall differences in romantic relationships were reported between adolescents with and without DSD. However, female adolescents with DSD were less sexually active overall than unaffected females. These differences were only significant for falling in love, frequency of dating, kissing, and cuddling. There were no reported differences in sexual activities between boys with and without DSD. Importantly, no difference was found between different DSD diagnoses, however those adolescents with induced puberty had fallen in love less frequently, experienced less cuddling and had less experience with sexual intercourse.

In summary, those with DSD have been reported as demonstrating reduced social functioning, however among those with CAH only, no such finding was reported. Although this may be as a result of the differences in the diagnostic presentations, it may also be as a

result of the different cultures in which the CAH and DSD studies were completed. With regard to intimate relationships, the only study to use a control group reported that there was no difference in romantic relationships overall, between those with and without DSD, however females with DSD may have a tendency to be less sexually active.

### **Cognition**

Research in this area has focussed on the assessment of cognitive function in individuals with CAH and has occurred primarily as it provides a natural model of endocrine dysfunction for which the impact of abnormal hormonal levels on cognitive function can be observed. Five studies were identified which assessed aspects of cognitive function in individuals with CAH (Hampson, Rovet, & Altmann, 1998; Mueller et al., 2013; Nass & Baker, 1991; Plante, Boliek, Binkiewicz, & Erly, 1996; Sinforiani et al., 1994 ), and one additional paper reported on cognition in individuals with Klinefelter's syndrome (Temple & Sanfilippo, 2003).

One study assessed the spatial reasoning of girls and boys with CAH (Hampson et al., 1998), and reported that girls with CAH demonstrated higher spatial reasoning abilities than unaffected females, while boys with CAH demonstrated poorer spatial abilities than unaffected boys. It was concluded that levels of pre- and perinatal androgen exposure were linked to the development of spatial ability. Another study investigated the impact of prenatal androgen exposure on cognitive control and motivation during adolescence (Mueller et al., 2013). In this Belgium based study, 27 adolescents with CAH, and 36 unaffected adolescents, completed a mixed-saccade<sup>2</sup> task with incentive and non-incentive trials. Inhibitory control was improved during incentive trials, relative to no-incentive trials in unaffected adolescents, but not in those with CAH. These results were taken to suggest that

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<sup>2</sup> A saccade task requires a participant to spot or look in the direction of an object displayed to them on a screen. In an anti-saccade task the participant is required to inhibit this response and to look away from the object of interest

early androgen exposure in CAH may disrupt the processes by which motivation can improve voluntary action (Muller et al., 2013).

Four studies reported on aspects of intelligence in individuals with CAH using a variety of measures. In the first of these studies, eleven participants with CAH were compared to five of their unaffected siblings and 16 matched control subjects using the Leiter International Performance Scale (Leiter, 1969), a non verbal test of intelligence, and either the Clinical Evaluation of Language Fundamentals- Revised (CELF-R; Semel, Wiig, & Secord, 1987) or the Structured Photographic Elicited Language Test-II (SPELT-II; Werner & Kresheck, 1983) (Plante et al., 1996). Language/learning disability was reported to be more prevalent in both the CAH participants and their families than in the control subjects. These findings were taken to indicate an elevated familial rate for language-based learning disabilities in families with the gene for CAH. A second study which tested the hypothesis that early androgen exposure may be the cause of the higher incidence of Learning Disability (LD) among males, used individuals with CAH as a natural model to investigate this (Nass & Baker, 1991). The Wechsler Intelligence Scale for Children (WISC; Wechsler, 1974) was used to determine the extent of Verbal-Performance IQ discrepancy. The study reported a significantly larger verbal-performance IQ discrepancy in girls with CAH than unaffected female relatives. Additionally the Verbal-IQ discrepancy of females with CAH was reported to be in the range shown by both CAH and unaffected males (Nass & Baker, 1991). The authors concluded that early androgen exposure, equivalent to that in unaffected males, increases the incidence of LD in females with CAH (Nass & Baker, 1991). In contrast, both Hampson et al. (1998) and Sinforiani et al. (1994) reported that there were no significant differences in IQ, as measured by the age appropriate WISC, between those with CAH and unaffected controls. Those with CAH were reported to have slightly higher IQs with respect to the expected distribution and no significant LD was detected. No differences were

reported between types of CAH, or between males and females with CAH (Sinfioriani et al., 1994).

One study assessed the executive function of individuals with Klinefelter's syndrome (KS), and compared three boys with KS to unaffected boys, matched for age, sex, and intelligence (Temple & Sanfilippo, 2003). Those with KS were reported to display impairments in executive skills, but only for inhibitory control. These results were considered to demonstrate that an additional x-chromosome has very specific effects on the cognitive profile seen in KS development.

In summary, one study has identified that those with CAH demonstrate increased spatial ability, and there is very limited research indicating that they also have reduced inhibitory control. Inconsistent findings are reported with regard to IQ and there is very limited research to suggest that language/learning disability may be associated with CAH.

### **Psychological Wellbeing**

Studies which have addressed psychological factors in DSD have tended to focus on QOL, behavioural adjustment, mental health diagnosis, and self-image. In total, 11 studies were identified which provided information in at least one of these areas.

**Quality of life.** Three studies assessed QOL in young people with DSD (Crawford et al., 2009; Kleinemeier et al., 2010; Willihnganz-Lawson et al., 2013). In an Australian study (Crawford et al., 2009), both children with a range of DSD, and their parents, rated child QOL using the Pediatric Quality of Life Inventory (PedsQL; Varni, Seid, Knight, Uzark, & Szer, 2002), which consist of two subscales, physical and psychosocial. The sample consisted of boys and girls aged between five and ten years old. Results were considered in relation to published values for the PedsQL in healthy children. Physical QOL was reported to be in the same range as that of healthy children, with the exception of male children, who self-reported their physical QOL as lower, but not significantly, than unaffected males.

Psychosocial QOL was reported by both parents and children as lower than unaffected children, and differences were greater between DSD and unaffected males than DSD and unaffected females. The authors concluded that these results were an indicator to continue with early intervention in DSD in order to minimise the impact on QOL. Similarly, in a study of 60 adolescent girls and boys, with a range of DSD it was reported that Health Related Quality of Life (HrQOL), as measured by the KINDL-R<sup>3</sup> Questionnaire for Measuring Health-Related Quality of Life in Children and Adolescents (Ravens-Sieberer & Bullinger, 2001) was not impaired in comparison to a reference group of school children (Kleinemeier et al., 2010). Girls with DSD showed higher HrQOL regarding physical wellbeing than girls in the reference group. No differences were observed between boys with DSD and boys in the reference group. Possible explanations provided for better physical wellbeing among girls with DSD were that the exposure of prenatal androgens has a positive effect on girls' physical wellbeing in adolescence, or that those with DSD do not experience such intense hormonal fluctuations as unaffected girls (Kleinemeier et al., 2010). Interestingly no differences were reported between the different DSD diagnoses, or between those individuals who had 'natural' or hormonally induced puberties.

The final study which assessed QOL specifically reported on QOL following secondary vaginoplasty in five girls with CAH, between the ages of 7 and 24 (Willihnganz-Lawson et al., 2013). QOL was assessed using the Glasgow Children's Benefit Inventory (GCBI; Schwentner et al., 2007) and responses varied. Positive scores were reported by younger participants, and negative scores by older participants. The authors made little comment about the implication of these findings, but noted that the more positive response of

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<sup>3</sup> KINDL –R is not an abbreviation, it refers to 'Kind' meaning child in German, and Kindl is a dialect for 'Kind', therefore this indicates that this is a questionnaire for children

the younger participants were completed by parents, rather than the individual themselves. It is also possible that older girls were more aware of the impact of their diagnosis.

**Behavioural adjustment.** Four studies have assessed behavioural adjustment in young people using the CBCL with mixed findings (Berenbaum et al., 2004; Gordon et al., 1986; Oner et al., 2009; Zhu et al., 2012).

The CBCL is a standardised parent report questionnaire which provides measures of Externalising and Internalising behaviours, as well as a Total Problems score. Two studies of individuals with CAH reported that, overall, there were no differences for CBCL scores between individuals with CAH and a control group of unaffected individuals (Berenbaum et al., 2004; Gordon et al., 1986). It was also reported that psychological adjustment was not related to disease severity or genital characteristics (Berenbaum et al., 2004). However, Gordon et al. (1986) who focused on girls with CAH reported that differences were observed for younger girls, 6 to 11 years, on subscales of somatic complaints, and schizoid/obsessive and this was attributed to the frequency and nature of repeated hospital visits necessary in these younger years.

Conversely, two additional studies reported that there were increased behavioural problems among those with CAH (Oner et al., 2009) and DSD (Zhu et al., 2012). A Turkish study of 28 females with CAH, aged between 8 and 20 years, reported that externalising behaviours, aggressive behaviours, and total problems, as measured on the CBCL, were higher among those with CAH than those with Diabetes Mellitus (DM) or those unaffected by either (Oner et al., 2009). Factors found to be associated with behavioural and emotional problems were higher mean testosterone levels, satisfaction with the appearance of genitalia, quality of surgical procedures, and hydrocortisone dose. Type of CAH was not associated with behavioural and emotional difficulties when the effects of other variables were taken out (Oner et al., 2009). The authors concluded that the success of surgical treatments, patients'



favourable perception of their genitalia, and good endocrine control were associated with better behavioural outcomes for individuals with CAH (Oner et al., 2009). Finally, Zhu et al, (2012) compared 50 boys and girls with DSD aged between 6 and 17 years old with matched controls of unaffected individuals. All eight boys with DSD were reported to have psychological difficulties, as measured by the psychological component of the CBCL. Depression, problems with communication, hyperactivity, and discipline problems were reported as significantly higher among boys with DSD than in unaffected males. Psychological and behavioural problems were also reported as significantly higher among girls with DSD than for unaffected girls. Problems reported included depression, somatic complaints, hyperactivity, discipline problems, violent behaviour, and immaturity.

A further study looked at behavioural adjustment among those with DSD using the Strengths and Difficulties Questionnaire. This study reported no significant overall differences between those with DSD and the reference group (Kleinemeier et al., 2010).

**Mental Health Diagnosis.** Four studies were identified which reported on the prevalence of diagnosable mental health problems in samples of individuals with DSD. Three studies looked at psychiatric diagnosis in CAH (Liang et al., 2008; Mueller et al., 2010; Oner et al., 2009) and one across a range of DSD conditions (Slijper et al., 1998).

In the first of these studies, 11 women between the ages of 8 and 25 years with CAH, living in Taiwan, were either assessed using the Chinese version of the Kiddie Schedule for Affective Disorders and Schizophrenia for School Aged Children (K-SADS-E) or interviewed and their presentation considered in relation to the DSM-IV criteria, depending on age (Liang et al., 2008). It was reported that 36.4% of those with CAH met the criteria for psychiatric diagnosis at some point in the year previous to the study. Participants were more likely than the general population to report anxiety, depressed mood, and sleep disturbances,

and it was noted that increased atypical sex-role behaviour in childhood was associated with greater severity of the psychiatric illness (Liang et al., 2008).

More recently, in a study which assessed psychiatric diagnoses in children, 8 to 18 years, with CAH, 44.4% of participants with CAH were found to meet the criteria for at least one psychiatric diagnosis in their lifetime, as assessed using the Kiddie Schedule for Affective Disorders and Schizophrenia-Present and Lifetime Version (K-SADS-PL; Kaufman et al., 1997) (Mueller et al., 2010). In comparison to the estimated population prevalence, males with CAH exhibited significantly increased rates of anxiety disorders and Attention Deficit Hyperactivity Disorder (ADHD), while females showed significantly increased rates of anxiety disorders only.

A third study which looked at the presence of psychiatric diagnosis in 28 females with CAH, aged between 8 and 20 years, reported that although diagnosis was higher among those with CAH (57.1%) than in those with DM and unaffected individuals, the difference between these groups was not significant (Oner et al., 2009). However it was reported that those with CAH were significantly more likely to have an anxiety disorder.

In the only study to look at psychiatric diagnosis in a sample of those with a range of DSD diagnoses it was reported that 39% of the sample had a diagnosable psychiatric problem, and a further 19% had mild psychiatric problems, when assessed using a semi structured psychiatric interview with both parents and children (Slijper et al., 1998). In this study, which took place in The Netherlands, the psychological care of children with intersex conditions over a 10 year period was evaluated. The study reported that psychiatric problems occurred despite early sex assignment, corrective genital surgery, psychological support of parents, and intensive psychotherapy of the children.

**Self-image.** Three studies were identified which assessed self-image (Berenbaum et al., 2004), self-perception (Gordon et al., 1986), and body-image (Kleinemeier et al., 2010).

Using the Self Image Questionnaire for Young Adolescents (SIQYA; Petersen, Schulenberg, Abramowitz, Offer, & Jarcho, 1984), it was reported that there was no difference in self-image between boys and girls with CAH and unaffected boys and girls (Berenbaum et al., 2004). Likewise, no differences were identified in scores on the Self Perception Profile for Children, between a group of 16 girls, aged 6 to 16, with CAH and a group of unaffected girls (Gordon et al., 1986). With regard to body image, it was reported that boys with DSD had a more negative body image than unaffected males, but that this difference did not exist for girls (Kleinemeier et al., 2010).

In summary, regarding QOL, based on those studies which provided a comparison group, it seems that overall, QOL is not reduced among those with DSD and results for behavioural adaptation currently present an unclear picture. Rates of diagnosable mental health problems ranged from 36.4% to 57.1% and although a mixed picture emerged with regard to whether those with DSD were at increased risk compared to unaffected individuals, it seems that there may be a trend for increased presence of anxiety disorders among those with DSD. Over all, self-image was comparable to unaffected individuals, with the exception of boys' body image.

### **Discussion**

This review has summarised literature relating to psychosocial and psychological outcomes in child, adolescent, and young adult populations with DSD. The literature identified was of relevance to several important areas of psychosocial development, including psychosexual development, social and sexual adaptation, cognitive development, and psychological wellbeing. While the literature in each of these areas was somewhat sparse, with several inconsistent findings, there is evidence to suggest that particular developmental tasks may be impacted by DSD.

The literature surrounding gender role reported consistently that girls with DSD engage in a range of sex-atypical play and activity preferences, and have a preference for male playmates. Literature did not report on whether individuals are particularly aware of or troubled by this difference, or whether their gender role has a direct impact on their ability to form friendships and participate in peer play from a young age. How these sex-atypical play preferences may continue into adolescence, and whether there may be implications for the development of later friendships and peer group membership in the teenage years is unclear.

Some research summarised in this review indicated that individuals with DSD may experience difficulties with peer relationships, show poor communication skills, and have worries about acceptance in society. Although limited to date, this research suggests that individuals with DSD may find it harder to develop friendships and feel accepted into peer groups, two important tasks of development during later childhood and adolescence.

Other important developmental tasks in the adolescent period are the development of romantic and sexual relationships, and the forming of a gender identity and sexual orientation. Research relating to the development of romantic and sexual relationships has produced inconsistent findings; however one study identified that girls with DSD may engage in aspects of sexual relationships less than their unaffected peers, and that some men experience anxieties about entering into marriage. Very few studies considered gender identity and among these, few reported the presence of gender dysphoria in a minority of participants. Further research is required to understand whether these important developmental tasks may be affected by having a DSD. As outlined earlier, Sullivan (1953) proposed stages of the development of friendships which included co-participation in play, acceptance by others, interpersonal intimacy, and the progression to intimate relationships. The research findings summarised here suggest that these stages in the development of friendships may be interrupted for those with a DSD.

Research assessing the psychological impact of DSD identified mixed findings with some suggestion of increased behavioural difficulties and anxiety among those with DSD. This may suggest that for some, their development is impacted in a way which results in poor adaptation to the diagnosis.

Although a number of important areas of psychosocial development have been considered in the DSD literature, few areas have been researched in sufficient depth across this age group to enable a satisfactory understanding of the impact of DSD on development. Additionally, there are a number of important and potentially relevant areas of development which have not been given consideration across this age group. These include attachment, and developing autonomy from parents.

While there are several possible reasons for the inconsistent findings reported in this review, there are a number of methodological concerns which may contribute to this and also add to the difficulty of generalising these findings. With consideration to research quality assurance criteria set out in the Critical Appraisal Skills Programme (CASP, 2013), the following methodological issues have been identified. First, different measures, some non-standardised, are used to assess the same outcome. For example, QOL encompasses a range of factors, both physical and psychological. As such, two different QOL measures are unlikely to assess the same specific factors, making comparison between studies problematic. Second, research in this area often recruits participants from a wide age range, possibly in order to increase sample sizes. While this review focused on children, adolescents and young adults, the papers still tended to have age ranges around 10 years, and up to 16 years. Whether results from such an age range, particularly over childhood and adolescence, can usefully be summarised together is questionable, particularly as some younger individuals may not yet have had a full disclosure about their DSD, and others may not yet have realised its potential implications for their future. Furthermore, research has indicated that acceptance

of the condition may change with age (Garrett & Kirkman, 2009). Third, several studies lacked a control group and as a result it is difficult to determine whether the reported findings were different to those of unaffected populations. Fourth, research often groups together a range of different DSD diagnoses. While this is beneficial in order to increase overall sample size and power, there are rarely enough participants in each diagnostic group to make comparisons between the different diagnoses. While studies which have made such comparisons have generally reported no difference in their outcome of interest between different DSD, where individual studies consist of different and mixed diagnostic groups the ease and usefulness of comparing outcomes is reduced. Fifth, although not provided for all of the papers in this review, response rates as low as 30% were noted. It is acknowledged that this is a hard to reach participant group, however representativeness of the sample needs to be considered. Finally, the research reviewed here is from a range of cultures and societies. Research has indicated that societal responses to DSD vary between cultures, as do treatment options and preferences (Lee et al., 2006). With this in mind, comparing or generalising results across differing cultures may be problematic and studies reporting outcomes such as QOL in different societies may be expected to produce different results.

Some of the methodological difficulties outlined above provide some general principles for future research including using standardised and consistent measures, focusing research on those facing particular developmental tasks, ensuring the use of control groups, and aiming to ensure, when looking at a range of DSD, that this is justifiable considering the aims of the research. Further research is required which focuses on child, adolescent and young adult populations with DSD, and there is a need for further culturally specific research. Research should also seek to identify the possible impact of DSD on other important developmental tasks such as attachment and developing independence from parents. In view of the mixed findings which have been reported to date, research should also begin to explore

factors which may contribute to good or poor adaptation to a DSD. While it is important to consider a number of individual factors such as vulnerability and resilience, literature from child health also demonstrates the importance of the family response, and family support, in child adaptation (Hamlett et al., 1992). An emerging body of research is beginning to consider the impact of DSD on parents, however little is currently understood about specific factors which are important for families coping with DSD. Understanding more about the role of the family in coping with DSD, and how families need to be supported throughout their children's development is important in facilitating positive outcomes for the child.

### **Summary**

This review has summarised literature evaluating a variety of psychosocial and psychological outcomes relating to DSD in child, adolescent, and young adult populations. The research has focused on four areas important in development; psychosexual development, social and sexual adaptation, cognition, and psychological wellbeing. Research in this area was found to be insufficient particularly in relation to aspects of psychosexual development, and sexual and social adaptation, and a number of methodological limitations, and gaps in the research contribute to limiting what can currently be understood about the impact of DSD on psychosocial development. Despite contradictory findings across most areas, there is research to suggest that, for some individuals with DSD, important areas of development may be impacted upon, such as gender role development, aspects of social functioning including peer, and intimate relationships, and psychological adjustment. Future research should seek to identify what contributes to good adaptation for individuals with DSD, and this should include consideration of individual and family factors.

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**Section B: Empirical Paper**

Disorders of Sex Development: Mothers' Experiences  
of Accessing Support

Word Count: 7926 (585)

A thesis submitted in partial fulfilment of the requirements of  
Canterbury Christ Church University for the degree of  
Doctor of Clinical Psychology  
April 2014

SALOMONS  
CANTERBURY CHRIST CHURCH UNIVERSITY



### **Abstract**

**Background:** An increasing body of research has sought to determine the impact of Disorders of Sex Development (DSD) on the family of the affected child. Little is currently understood about the support needs of the family and how well these needs are met.

**Methods:** With a focus on mothers as primary caregivers, Interpretive Phenomenological Analysis was used to analyse semi-structured interviews with eight mothers of children with DSD about their experiences of support.

**Results:** Four master themes emerged which encapsulated the stages in their child's development when mothers most needed support, the importance of developing an understanding of the child's condition, the lack of an acknowledgement of the emotional needs of the parent, and the importance of having close and trusted networks for support. Continuity and availability of support were considered important and while all participants prioritised maintaining privacy about the condition, a minority felt that this impacted on the level of support they received.

**Conclusions:** Key periods of time for support were identified and while some felt that they were well supported others felt that their support did not meet their emotional needs. The results were discussed in light of previous research, and the clinical implications considered.

*Keywords: Disorders of sex development, support needs, family impact, mothers' experiences, social support*



### Disorders of Sex Development: Mothers' Experiences of Accessing Support

Disorders of sex development (DSD) are a class of congenital conditions in which the development of chromosomal, gonadal, or anatomical sex is atypical (Lee, Houk, Ahmed, & Hughes, 2006). The condition is most often identified in newborns or adolescents, however an increasing number of individuals present either antenatally or during childhood (Brain et al., 2010). Affected newborns typically present with atypical genitalia, and those who are identified in adolescence present with atypical sexual development during the pubertal years, such as delayed puberty (Ahmed et al., 2011).

The diagnosis of a DSD presents a range of unique challenges to the individual, as well as parents and professionals (Brain et al., 2010). The need for assessment and management by a multidisciplinary team (MDT) has been emphasised (Ahmed et al., 2011; Brain et al., 2010; Lee et al., 2006) regardless of the type of DSD and when it has been identified. While the course and treatment for individuals with DSD vary considerably, treatment may include gender assignment at birth, genital reconstructive surgery, sex steroid replacement, and psychosocial management (Lee et al., 2006).

### **DSD and Wellbeing**

Since DSD are often first treated by paediatric endocrinologists, surgeons, and urologists, research on outcomes and interventions has centred on issues most relevant to these fields, such as gender assignment, psychosexual outcomes including gender role and gender identity, and ethical considerations related to treatment. Although less is known about the impact of DSD on quality of life (QOL) and mental wellbeing, an increasing field of literature demonstrates inconsistent findings across a number of DSD (Wisniewski & Mazur, 2009). The psychological adjustment of adolescents with DSD has been reported as unaffected (Berenbaum, Bryk, Duck, & Resnick, 2004), and adult women with DSD have also shown comparable levels of self esteem and psychological wellbeing in relation to

unaffected individuals (Hines, Ahmed & Hughes, 2003). Elsewhere, it has been reported that adults with DSD demonstrate an impaired QOL (Johannsen, Ripa, Mortensen, & Main, 2006) and higher levels of affective distress and deliberate self-harm, (Brinkmann, Scützmann, Schacht, & Richter-Appelt, 2009). Adults with DSD have spoken of the experience of managing silence and secrecy around their condition, coping with difference, and developing acceptance as particular challenges (MacKenzie, Huntington, & Gilmour, 2009).

### **The Impact of DSD on Parents**

Emerging research has sought to understand the impact of DSD on the parents of affected children. It has been demonstrated that carers of children with a DSD are at risk for overprotection of the child, increased perceived child vulnerability and parenting stress, and reduced self-esteem and psychological stability (Duguid et al., 2007; Kirk et al., 2011). In addition, high levels of post-traumatic stress symptoms (PTSS) have been identified in some parents of children with DSD, with 31% of mothers, and 18% of fathers meeting the threshold for caseness of post-traumatic stress disorder (PTSD) (Pasterski et al., 2014).

Genitoplasty (a form of corrective genital surgery), the degree of masculinisation of the genitalia, and the developmental stage of the child have all been considered in relation to parental stress, overprotection, and caregiver depression and anxiety. Among female caregivers whose children did not receive corrective surgery, there was a tendency for increased stress, while among male caregivers, where corrective surgery had taken place, increased stress and overprotection was reported (Fedele et al., 2010). Also, though the caregivers of male children with under masculinisation of the genitalia reported higher levels of caregiver depression, over masculinisation of the genitals of female children was unrelated to caregiver depression or anxiety (Wolfe-Christensen et al., 2012). Over protection of children with DSD was greatest for infants and toddlers (Hullman, Fedele, Wolfe-Christensen, Mullins, & Wisniewski, 2011).

A number of qualitative studies have explored parental emotional response to the diagnosis of a DSD and have identified shock, grief, anger, guilt, and shame in response to a diagnosis of Androgen Insensitivity Syndrome<sup>1</sup> (AIS) (Slijper, Frets, Boehmer, Drop, & Niermeijer, 2000). In addition, the parental struggle to negotiate a coherent sex identity for their child, and parental difficulties in sharing their story with others has been highlighted (Gough, 2008; Sanders, Carter, & Goodacre, 2007). Similarly, shock, disbelief, and strong protective instincts, have been reported in relation to children's reconstructive surgeries for ambiguous genitalia, (Sanders et al., 2007; Sanders, Carter, & Goodacre, 2012). Crissman et al. (2011) explored the early parental experience of having a child with DSD and identified the gender assignment process, decisions about genital surgery, disclosing information about the child's condition, and interacting with healthcare professionals, as salient aspects of the process for parents.

### **Family Coping and Support**

Drawing upon literature from childhood chronic health, some parenting characteristics have been demonstrated to be predictive of children's emotional, behavioural, and social adjustment (e.g. Colletti et al., 2008). Specifically, parental overprotection has been associated with poorer quality of life (QOL) and higher rates of behavioural problems in the child (Hullmann, Wolfe-Christensen, Meyer, McNall-Knapp, & Mullins, 2010). Additionally, parental adaptation to a child's health condition has been identified as key for the subsequent wellbeing of the child (Carmichael & Alderson, 2004) and higher levels of parenting stress are related to poorer emotional, behavioural, and social adjustment in children with chronic illness (Colletti et al., 2008). Therefore, reducing stress, and increasing

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<sup>1</sup> Androgen Insensitivity Syndrome is a condition in which the sex chromosome of the individual is male-XY, however the external genitalia may be female or ambiguous. Additionally, testes may be absent or not properly formed.



adaptation and coping skills for parents, can have important implications for the child and whole family coping.

The resiliency model of family stress, adjustment, and adaptation (McCubbin & McCubbin, 1993) identifies protective factors which sustain families through stressful periods such as those when a child is born with a health condition. Alongside factors such as family cohesion, family communication, and financial management, social support is identified as important in the relationship between family stress and family coping (McCubbin & McCubbin, 1993).

Empirical research from the childhood chronic illness literature has demonstrated the importance of sharing worries about the illness and social support in parental and family coping (Gannoni, & Shute, 2010; Tak, & McCubbin, 2002; Ware, & Raval, 2007). It may therefore also be important for parents of those with DSD to communicate with others and access support in order to achieve improved family coping, and in turn, positive child adaptation and wellbeing.

### **Parent Support and DSD**

While no studies to date have specifically looked at the process or impact of support for the parents of those with a DSD, an intricate picture is emerging through studies assessing the effect of DSD on the family. Such studies have pointed towards reduced coping skills, including a reduction in social support, and a reduction in the utilisation of communication with medical professionals among parents of children with DSD (Duguid et al., 2007). Additionally, tensions may exist between maintaining privacy for the child, which has been identified as increasing parental stress (Crissman et al., 2011), and disclosing the condition in order to access social support. The need for support from professionals has been highlighted as an important factor in coping (Liao & Boyle, 2004; Sanders, Carter, & Goodacre, 2011),

however parents' perceptions about how helpful it is to seek support from relatives is more varied (Duguid et al. 2007).

### **Rationale and Aims**

Despite the acknowledgement that families of children with DSD experience unique challenges requiring support, very little is currently understood about the specific needs and wishes for support amongst parents. Developing further understanding regarding parents' experiences of support is therefore important, as well as ascertaining the purpose of the support, and what may contribute to parents accessing support or not. To date, research assessing the impact on the family has focused on the early parental period and little is currently known about the changing needs of parents as their child continues to develop. Therefore it would be valuable to understand when, in the child's developmental pathway, parents have felt in need of support. While it is acknowledged that the experiences of both mothers and fathers are important, research has indicated that mothers may exhibit more stress than fathers in relation to their child's DSD, particularly when genitoplasty has not taken place (Fedele et al., 2010). Additionally, within Western culture, mothers tend to be the primary caregiver for the child and may be more likely to access support in the context of child illness (Ware & Ravel, 2007). For these reasons, this study will focus on mothers' experiences of accessing support. Understanding parent experiences of support will enable services to better target their support and also to provide support which is more closely attuned to the needs of the parents. Additionally, it is hoped that such information may benefit parents in similar circumstances. Therefore, this study aims to address the following research questions:

1. What are the support needs of mothers of children with DSD and how do these change over time?
2. How well have their support needs been met and was anything missing?

## **Method**

### **Research design**

As this research aimed to understand the experience of participants in an exploratory and in-depth manner, a qualitative methodology was chosen using semi-structured interviews. Interpretive Phenomenological Analysis (IPA) (Smith, Flowers, & Larkin, 2009) was selected as a method of analysis as it enables the exploration of the meaning of a particular experience, and an understanding of how the participants make sense of this experience within their personal and social worlds (Smith & Osborn, 2007). It also acknowledges the influence of the researcher's own view of the world, and the quality of the interaction between the researcher and participant in interpreting what is said by the participant (Willig, 2013).

### **Participants**

An opportunistic and homogenous sample of 8 women participated. The sampling method and size was consistent with those recommended for IPA (Smith et al., 2009). Inclusion criteria specified that all participants were the biological mother, and primary caregiver to a child with a DSD diagnosis, diagnosed a minimum of six months ago, and that the child was aged between 6 months and 18 years, inclusive. Participants were recruited through one NHS hospital and three charitable organisations.

### **Measures**

Semi-structured interviews are considered an appropriate means of collecting data for IPA analysis (Smith et al., 2009). The interview schedule (Appendix C) was developed with the exploratory nature of the research question in mind, and also following guidance on IPA interview schedules (Smith et al., 2009). Questions were developed based on previous literature and discussions with a clinical psychologist working in the area, and the schedule was piloted and revised prior to interviewing the first participant. Questions sought to

explore the experiences of mothers of children with a DSD diagnosis in accessing support. A number of prompts were identified, and follow up questions were utilised in all interviews.

### **Procedure**

Participants recruited through the NHS were identified by the clinical psychologist in an endocrinology paediatric clinic. Women were approached about the study and, if interested, permission was sought to provide the researcher with their contact details, or they were asked to contact the researcher directly. Written information about the study was provided at this time (Appendix D). Participants recruited through the three charitable organisations made direct contact with the researcher in response to brief information displayed in closed internet forums, or emails sent out by the charity (Appendix E).

Interviews were scheduled with the researcher and took place at the hospital site, the participant's home, or over the telephone. Participants were reimbursed up to £10 in travel expenses. Prior to interview, all participants read the information sheet (Appendix D) and were given the opportunity to ask questions. Participants gave their full written consent to participate (Appendix F). Interviews lasted between one and two hours and participants were informed of their right to refuse to answer any question, or to terminate the interview at any stage. All interviews were audio recorded. At the end of the interview participants were given the opportunity to reflect on the interview process and were debriefed by the researcher (Appendix G) including being given the contact details of support groups (Appendix H).

### **Analysis**

Interviews were transcribed and analysed using Interpretative Phenomenological Analysis (IPA; Smith et al., 2009). The procedure for IPA, as described by Smith et al. (2009), was followed. Interviews were transcribed, and read and re-read. On the initial reading of each transcript exploratory comments were made by the researcher, including descriptive, linguistic, and conceptual comments. Using these exploratory comments,

emerging themes were then noted alongside the transcript, (Appendix I) and these emerging themes then listed, chronologically, in a separate document. Emerging themes were moved around to form clusters, developing initial super-ordinate themes. In keeping with IPA's idiographic commitment, this process was then repeated for each interview, attempting to bracket off ideas emerging from previous cases while working on the present case (Smith et al., 2009). In looking for patterns across cases, super-ordinate and emerging themes from all cases were viewed together and consideration was then given to possible connections across the cases. This involved manually moving themes around, and themes were both renamed, and reconfigured throughout this process. When it was felt that this represented a good-fit of the researcher's interpretation of the data, this process was stopped. Transcripts were re-read to ensure that the themes were captured within the text, and suitable quotes for themes were identified.

### **Quality Assurance Checks**

Criteria for validity of qualitative research (Yardley, 2008) were considered throughout the research process and in the writing of the report. Specifically, Sensitivity to Context was considered in having an awareness of the existing literature in this area, and in discussing with a clinical psychologist the potential difficulties faced by the sample. Commitment and Rigour was considered in the selection of the sample, and in the care taken with the analysis process, which was thorough, and conducted as per IPA guidance (Smith et al., 2009). Transparency and Coherence is evidenced by the inclusion of a coded interview transcript (Appendix I), and extracts from both a research diary (Appendix J), and a bracketing interview (Appendix K). Bracketing interviews are a strategy in which the researcher is interviewed themselves about the proposed topic of the study in order to investigate their presuppositions concerning the research project (Roulston, 2010). While it is acknowledged that the researcher cannot fully 'bracket' off their knowledge or beliefs, it is

hoped that generating an awareness of these prior to commencing interviewing provides increased subjectivity and transparency. Completion of the bracketing interview brought to my attention my existing beliefs about the importance of support when individuals are experiencing difficulties. I also became aware of my preconceptions about how it might be to have a child with physical health complications, particularly with a DSD. The use of a research diary enabled me to monitor my emotional responses to the mothers I interviewed, and what was spoken about. Prior to completing the analysis, revisiting the bracketing interview and the research diary enabled me to improve subjectivity, and to consider carefully what may be influencing the way in which I interpreted the data. Finally, an independent audit trail was completed of the analysis process to further determine validity (Appendix L) (Smith et al., 2009).

### **Ethical considerations**

Ethical approval for this study was obtained from the National Research Ethics Committee and the Research and Development Department at the NHS trust through which participants were recruited (Appendix M).

### **Results**

All mothers who participated in the research seemed keen to have their experiences heard. They engaged fully, and many talked at greater length than they had expected to. Many expressed feeling glad that research was taking place in this area and hoped that their participation would ensure positive experiences of support for mothers in the future. All participants were white British, and ages ranged between 35 and 45 years. All of the mothers were in a long term relationship with the father of the affected child. Six were married, while two referred to their 'partner'. All of the mothers had at least one other child, with one family having four children. The time since diagnosis ranged from 6 months to 18 years.

Relevant information about the participants' children is displayed in Table 1. Two of the mothers interviewed had two children with a diagnosis of DSD.

The majority of the children had salt losing CAH, a form of CAH in which the individual does not store salt within the body. This form of CAH is often considered the most severe form of CAH. Non-salt losing CAH can also result in virilisation but the individual is able to store salt. It is considered to be a less severe form of CAH.

Table 1

*Participants' children's characteristics*

Child characteristics	
<b>Gender (n)</b>	
Boy	4
Girl	6
<b>Age</b>	
Mean <i>years</i> (SD)	9 (5.12)
Range <i>months/years</i>	9 months-18 years
<b>Time since diagnosis</b>	
Mean <i>years</i> (SD)	9 (5.21)
Range <i>months/years</i>	6 months- 18 years
<b>Diagnosis</b>	
CAH- Salt losing	6
CAH- Non salt-losing	2
49 Syndrome	1
46 XX/46XY mosaic DSD	1

The analysis resulted in four master themes, and 14 sub themes which are described below and illustrated with quotes. The theme development process is outlined in Appendix N and additional quotes relevant to each theme can be seen in Appendix O.

Table 2

*Themes and sub-themes*

Master themes	Sub-themes	Number of participants contributing to sub-theme (N=8)
1. Evolving support needs	• Developing support needs	8
	• Emotional support needs shifting	7
	• Learning Curve	6
2. Seeking understanding	• Uncertainty brought by the diagnosis	8
	• Professionals facilitating understanding	8
	• Independently seeking information	8
3. Parental emotional needs in a medical setting	• Emotional impact of the diagnosis	8
	• Parental emotional support needs unacknowledged	7
	• Not feeling heard	6
	• Intermittent support	8
	• Valuable experiences addressing emotional needs	7
4. Close support networks	• Privacy over support	8
	• Support network	8
	• Impact on 'normal' parental supports	5

**Evolving Support Needs**

This master theme encapsulates the evolving challenges and areas of need which are faced by the mothers as their child develops.



**Developing support needs.** Participants identified periods of time in the child's development when they had felt particularly in need of support, and also aspects of their child's future development about which they currently had worries. Five participants spoke about the years following birth as a particular time of need:

*“in the first few years of her life I think really is when I, when we both, me and my husband really felt that we did need to talk to people and professionals”* (P03, 18, 450)<sup>2</sup>.

Adolescence was the period of the child's development which was most frequently talked about by participants:

*“she's sort of started to hit puberty now and there are lots and lots of issues around that (...)”<sup>3</sup> so I am going to need to access something I think at that point”* (P06, 38, 734).

Seven participants also identified the need for support in communicating with their child about the diagnosis:

*“as they get older I want to know the right things I should be saying to help them to come to terms with it (...) I'll need support to help talk about it”* (P02, 26, 655).

The prospect of surgery also created worries *“I am worried about the next few years, you know the surgery”* (P08, 23, 464), and the need for support from professionals *“we did want to talk to people and you know we talked to the doctors at the hospital”* (P03, 19, 473).

Although separate from the development of the child, two participants identified the prospect of having another child as a time at which they needed additional support:

*“I really wanted to have another one and it was sort of like, erm I've got to get over it somehow”*, (P 05, 55, 1136).

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<sup>2</sup> (Participant number, page number, line number)

<sup>3</sup> (...) Material omitted or added for clarity or confidentiality reasons

**Emotional support needs shifting.** Three participants identified a sense of the diagnosis being on the mother, or the family, rather than the child:

*“I think especially when they’re so young, because they have absolutely no understanding, so really it is you, even though it’s not your diagnosis, it is you that’s dealing with it and coming to terms with it, not them at all (...) they don’t have clue what’s going on”* (P02, 17, 430).

With this was the notion that the need for emotional support was initially within the family. However, there was an impression from the participants that this changed over time, and this was acknowledged in their dialogue about the child’s emerging need for emotional support:

*“beyond that really to her teenage years and beyond to adult hood, it’s really about helping her and (...) allowing her to see that there can be a useful relationship with (professionals) if she chooses to have that”* (P07, 32, 654).

Similarly, *“we made it quite clear to her that these people are here for you just like the doctors were there for us”* (P03, 19, 484). It was also identified that the child may need their own private space:

*“everybody has things that they don’t want to tell anybody and they don’t want to tell their mum or dad, you know and that is the very personal things (...) so I think she needs someone”* (P06, 46, 903),

**Learning curve.** Six participants referred to a learning curve which occurred in the period initially after birth and across the first few years of the child’s life:

*“Cause at that time it’s a learning curve as well, cause when they’re little or babies you don’t know what you’re dealing with (...) you’ve got to learn”* (P06, 32, 619).

For participants whose children had CAH, this seemed particularly salient around the giving of injections:

*“because you have to administer the injection, (...) and I’m a bit of a wimp when it comes to needles, but even I’ve overcome that” (P01, 12, 241).*

### **Seeking understanding**

This master theme depicts the process of seeking knowledge and understanding about the child’s diagnosis and seeing this as a means of both coping and ensuring the best for the child.

**Uncertainty brought by the diagnosis.** All participants spoke about the uncertainty that the diagnosis created. This included a lack of awareness about the existence of DSD conditions, *“before she was born we had absolutely no idea, no idea, never heard of it before”* (P03, 4, 75), as well as the sense of uncertainty that was created around the child’s future:

*“knowing what to expect (...)looking to the future, that kind of gets taken, but not replaced with anything”* (P05, 65, 1338).

There was a sense of relieving this uncertainty by gaining knowledge about the condition and its treatment:

*“I want the truth and I want to know how it’s going to be, you know, even when she was born, I wanted to know (...)I want to know what she’s got (...), and how we’re going to deal with it”* (P03, 17, 423).

Knowledge was associated with power *“Knowledge is power”* (P08, 21, 423), and also seemed to be viewed as a means of ensuring the best for the child.

**Professionals facilitating understanding.** Participants viewed professionals as people who could support their understanding of their child’s condition, its causes, and the treatment. For four participants, there was a sense that their understanding was not well facilitated:

*“what is it that you’re measuring, what it is that you’re looking for, I just don’t really understand and still these are questions that are not really being answered”* (P02, 20, 497).

Attempts to understand the cause of the condition were hindered by the use of jargon, *“all this medical jargon(...), it does blind you with science”* (P01, 34, 740), and the timing of such explanations *“He did explain, but you’re just not in the right frame of mind to take anything in”* (P06, 13, 238).

Six participants identified the need to have contact with specialist professionals, as non-specialist professionals were felt to lack specific knowledge:

*“cause she wasn’t an expert in it, cause it’s quite an in-depth area isn’t it, and she wasn’t obviously the right person to speak to”* (P05, 47, 968).

Two participants spoke of communications with professionals which facilitated their understanding well and two had mixed experiences:

*“this was the first time we’d actually been explained the process of what actually happened when the baby is formed and why the chromosomes do that, and what it means”* (P05, 27, 355).

Two participants also spoke of feeling well guided by professionals at important times in the child’s development:

*“You’re in the hands of the professionals while you’re there, and they were fantastic really”* (P03, 6, 133).

**Independently seeking information.** Participants spoke about their search for information aside from contact with professionals. This most frequently took the form of the internet. For two participants this was in response to a lack of information from professionals:

*“I’d go back (from an appointment) and read another medical article, or you know, Google the effects of low cortisone production and try and [SIGHS] make myself feel better” (P07, 21, 421).*

Two participants found the information available on the internet beneficial:

*“It was this really good site actually (...) of their (children) so that was brilliant because that was real, and that was exactly, you’re seeing the truth of it all” (P 04, 10, 208).*

However, five participants felt that the information available on the internet was not helpful and impacted upon them in a negative way.

*“I started reading quite a lot of posts that people had put up and I didn’t find that at all useful cause I found it all very negative and quite depressing to be honest” (P02, 18, 443).*

One participant expressed that she would *“rather hear a professional answer” (P03, 6, 151)*, than look on the internet.

### **Parental emotional needs in a Medical Setting**

This master theme encapsulates the participants’ need for emotional, and ongoing support, within a medical system which prioritised the physical health of the child.

**Emotional impact of the diagnosis.** All participants reported a strong emotional response to the diagnosis reporting that it was *“painful” (P08, 26, 522)*, and *“it was just a frightening place” (P01, 5, 86)*. For three participants, the longer term impact on their wellbeing was identified, *“that triggered the depression (...) really” (P07, 21, 417)* as well as the sense of isolation that caring for a child with DSD created, *“I just kind of feel quite alone” (P02, 14, 349)*.

**Parental emotional support needs unacknowledged.** Six participants identified that the emotional impact on the parents and family was not well acknowledged among professionals:

*“we’re so blessed to have been able to get the medical support for her that we did but I think there is some aspects that are really quite overlooked with supporting the parents really and trying to keep the parents sane through the whole thing”* (P 08, 6, 109),

*“they are more interested in it from a medical point of view, whereas they don’t think, ‘how is this life going to turn out for these parents’”* (P05, 52, 1073).

Attempts to establish how parents were coping were also limited:

*“she’d also finish up by saying ‘and how are you?’ and every time she said it I’d nearly start crying cause (...) she was the only person who actually asked how I was”* (P02, 16, 385).

There was also a sense that medical professionals did not notice the signs of participants who were in need of emotional support, and it was felt that opportunities to offer support were missed:

*“I feel a bit let down the person didn’t really see, (how I was) maybe she just really didn’t understand what the implications of the condition (were)”* (P05, 49, 1012).

**Not feeling heard.** In the context of their needs not being met, six participants referred to a sense of not feeling heard by professionals, *“a lot of things get, can get brushed off, you know”* (P04, 41, 906), and that this often continued over time:

*“and I’ll say things like (...), and have done since I can remember, ‘but she’s still really rough and tough and aggressive’ and err it will just be fobbed off with, ‘oh well you know’”* (P08, 9, 174).

Five participants talked about the difficult power dynamics which can exist between medical professionals and 'patients':

*"it was intimidating and daunting(...) you're talking to doctors and nurses, (...) I think, the first time I went to the hospital there (...) was about four of them in the room, I can't tell you how daunting and scary it was"* (P06, 55, 1092).

Three participants also referred to the idea that they were viewed as 'neurotic mothers' for questioning procedures or treatments:

*"when the nurse came in I said 'can you just check cause I don't think that's what he normally has(...) when he's unwell', and the hands went on the hips and do you not trust us and everything"* (P07, 11, 214).

**Intermittent support.** Participants expressed a need for ongoing and continuous support. A sense that support was 'there' and 'available' seemed important in helping participants to feel well supported:

*"I know that if I needed to go to (professional) for anything I know, I know that I could email her and she would speak to me or she'd make an appointment and we could go and just discuss, and I feel like I've got that now if I needed it"* (P05, 43, 884).

Four participants seemed to feel that support was only there when you demonstrated that you were not coping, *"I suppose because I just hold it together and support comes in when you crumble a bit doesn't it"* (P04, 19, 409), or when there was a crisis rather than the continuous support which felt needed:

*"that's just crisis control that's just you know, getting you through the hardest days, I don't think anybody really grasps that the other days are pretty bloody hard as well"* (P08, 29, 595).

For three participants, there was also a sense that there was good support at the time of diagnosis but that this dissipated:

*“to start with I sort of thought that (the support) was fine but then I think as the times gone on (...) I’ve actually felt more alone, because I guess to start with there’s that initial kind of buzz (...), and then it all just sort of peters off and you’re just kind of left to get on with it” (P02, 12, 286).*

Additionally, four participants identified that better support would include an allocated professional, who could be the main point of contact, and take an overall view of what is going on:

*“access to, for the parents to speak to, well almost like be allocated (...) someone that you know that they could just sort of ring up and maybe have a chat to” (P06, 54, 1068).*

**Valuable experiences addressing emotional needs.** Although not all participants felt that they had been well supported overall, six participants identified some interactions or experiences, outside of those with family or friends, which had clearly been of value to them emotionally. These had occurred to some extent with medical professionals, however clearer examples of this occurred in the process of meeting or hearing about the experiences of other mothers of children with DSD, either on the internet, via support groups, or facilitated groups. What seemed to be important in all these interactions was that it reduced feelings of isolation, or being the ‘only one’:

*“just the fact that you know that someone else is going through that. I know it’s silly but, I don’t know, it just relieves you a bit, you just think oh yes someone else knows exactly how I feel and that (...) helps, really helps (...) talking to the other parents, just knowing that actually he’s not the only child (...) like that” (P04, 28, 629)*



Participants also spoke of the acceptance which seemed to be present when talking to other mothers of DSD children, *“it was like the first time ever that you felt comfortable talking about it with somebody cause they knew what it was like, what the feelings were like”* (P05, 28, 554), and the importance of having their own feelings and responses to the diagnosis normalised:

*“sharing it with everyone and everyone was doing the same and it was sort of like everyone felt the same and had the same feelings at diagnosis and the same experiences of (...) telling other people and for the first time ever (you could be open)”* (P05, 28, 560).

For participants who had not experienced a support group or talking with other mothers in the same situation there was a sense that this could be helpful, and such a process was likened to their experiences of other supportive groups which had created a sense of acceptance:

*“I went to a breast feeding support group when I had my first (child) and it was the best possible thing cause we just all used to go talk about what was crap, have a whinge, have a cup of tea and feel better (umm) and there’s no way to do that with this, cause nobody understands”* (P08, 32, 643).

### **Close support networks**

This master theme encapsulates the nature of the support networks that participants identified and the conflicts which sometimes existed for participants in maintaining privacy around the condition, and within the support network.

**Privacy over support.** All participants expressed strong protective feelings towards their children, *“it (...) must of been like a deep seated protection of your child that I didn’t really know that I had at the time”* (P05 15, 295). Such protective feelings seemed to be in relation to the child’s future and the social implications of the condition, and this resulted in participants talking minimally about their child’s diagnosis:

*“you kind of keep it minimal, cause you know she’s going to have to grow up and deal with this and you know, you don’t want people knowing her innermost secrets” (P08, 6, 121).*

Maintaining privacy for the child seemed a priority for all participants:

*“irrelevant of what I want to share with such and such or this is a burning issue that I feel like I want, you know, my friends to know or whatever, erm that’s not my priority” (P08, 19, 379).*

Three participants felt that this privacy was directly limiting in how much they were able to talk about their child’s condition *“so I guess in that way I’ve had to keep things inside, bottled up a bit more” (P02, 24, 594)*, and in the level of support that they were able to utilise:

*“that kind of stops you from perhaps having as full a support as perhaps you might be able to have” (P07, 19, 383).*

It was however acknowledged that ‘telling people’ did not equate with being supported, *“with some people, why would I be telling them (...), they’re not going to be any support to me” (P02, 25, 638).*

**Support network.** Six of the participants identified a small network of individuals who were recognised as their main supports. Who was in this network seemed largely based on who was trusted, and close enough to the participant to know about the condition and offer support:

*“cause we’re such a close family (...) you know, we trust everybody 100 % that we told” (P06, 11, 200).*

Networks were predominantly made up of family and close friends:

*“I suppose yeah sometimes (I talk) to my mum, (...) I’ve got a couple of my closest friends er, so I talk to them I suppose, and my husband of course” (P04, 18, 386).*

However three participants also identified a professional who was deemed to be a key source of support:

*“the endocrine nurse (...), I would say she’s been the biggest support, she is amazing, she is a wonderful lady” (P02, 12, 284,).*

Variations and limitations in the kinds of support that individuals in the network could provide were identified:

*“our family didn’t know anything anymore than we knew about it (...) they hadn’t heard of it either so in the medical sort of side they couldn’t help us, but you know emotionally if we were a bit down then they could help us” (P03, 5, 121),*

*“I can talk to my friend (...) about things that I can’t talk to (my husband) about it (...) I don’t want to tell him that and either make him upset or he’ll be thinking why are you thinking that” (P06, 27, 524).*

Of those who had a ‘network’ two participants expressed that it provided ‘enough’ support, *“I think that I have got what I need from who I need” (P03, 16, 393).*

Those participants who seemed to lack a sense of a support network around them identified isolating themselves from those who could offer support, and the strain that the diagnosis had placed on family relationships:

*“it pushed us to the edge and quite a few times we very nearly did split up, this being a major factor in it” (P08, 4, 72).*

**Impact on ‘normal’ parental supports.** Five participants seemed to vocalise that the child’s diagnosis had a negative impact on how they accessed the normal parental supports, such as the baby clinic, mother and baby groups, and utilising family or friends to look after the child. Attendance at such groups seemed to act as a reminder, or emphasise the child’s diagnosis:

*“to go to something that I thought was going to be nice and getting him weighed, turned into something horrific(...)I just felt so different (...) I found it quite hard”*  
(P04, 13, 291).

Difficulty in relinquishing control over administering medication also seemed to influence how readily mothers left their child in the care of others:

*“I wouldn’t ever leave him I was, I didn’t want anyone to have him I didn’t want anyone to have that role of caring for him because I didn’t think they could do it as well as I could and I thought it was too complicated”* (P01, 23, 486).

In summary, there was a clear sense of the emotional impact and the uncertainty that the diagnosis had on participants, and their desire to learn and understand the condition was generally poorly facilitated by professionals. Many participants felt that their emotional needs went unacknowledged within the medical system and while for some this was met adequately elsewhere, others felt their emotional needs were unmet. The need for continual support to be ‘there’ was emphasised.

### **Discussion**

The purpose of this study was to develop an understanding of the support needs of mothers of children with DSD and to appreciate how these may change over time. It also aimed to understand how well mothers’ experiences of support had met their needs and to identify aspects of support which may have been lacking. The results suggested that mothers of children with DSD have varying support needs which evolve over time and include the need to understand the condition, as well as the need for emotional support for the family and the child. While some mothers have had valuable experiences of support from small networks of individuals, others have found their emotional needs largely unmet. The results will be considered below in relation to the research questions and existing literature.

Limitations and areas for future research will be discussed, followed by consideration of the clinical implications.

### **Support Needs**

The themes 'evolving support needs', 'seeking understanding', and 'parental emotional needs in a medical setting' offer relevant information in understanding the support needs of participants, and how these needs may change over time in relation to the child's development. Overall, participants identified two key ways in which they needed to be supported; firstly to be supported in developing an understanding of their child's condition, their future and the treatment implications, and secondly, to be supported emotionally.

Parents identified a number of times when they felt that their need for support was increased. The need for support at the time of birth and in the year or so following birth was significant and this was emphasised by the idea of the 'learning curve'. Other times at which increased support had been sought or was anticipated included surgery, adolescence, communicating with the child about the diagnosis, and having other children. The need for support around the time of birth and surgery has been identified in previous literature (Crissman et al., 2011; Sanders et al., 2007) and the importance of developing understanding, and learning to manage medication in CAH is important in improving the self-efficacy of parents of children with CAH (Fleming, Rapp, & Sloane, 2011; Mitchelhill et al., 2013). Parental anxiety about puberty and adolescence in children with DSD has also been reported elsewhere (Crissmann et al., 2011). Psychosocial theories of development (Erikson 1956), as well as theories of gender identity, point towards the importance of puberty and adolescence in the development of the self. As such mothers' expectations that this will be a time of the child's development in which they need additional support is not surprising.

A key task at this time which was identified by the participants in the present study concerned communicating with the child about the diagnosis. The importance of children

understanding and receiving a full disclosure of their diagnosis has previously been reported (Slijper et al., 2000; Sutton et al., 2006) and research has identified that children with a genetic condition, including CAH, wish to know about their condition and its causes by the age of 12 (Szybowska, Hewson, Antle, & Babul-Hirji, 2007). While this research has highlighted that children wish for their parents to be involved in this process, less has been written about how best to guide parents in talking helpfully with their child about their condition.

### **Experiences of Support**

In considering participants' experiences of support and how well their support needs were met the themes of 'close support network', 'emotional needs in a medical setting', and 'seeking an understanding' were relevant.

Consistent with previous research, participants strongly associated with feelings of protection towards their child (Kirk et al., 2011) and maintaining privacy about their child's diagnosis in order to protect them from possible negative social and emotional outcomes (Crissman et al., 2011; Sanders et al., 2012). While maintaining privacy about DSD has been identified as stressful for parents (Crissman et al., 2011), only a minority of participants felt that maintaining privacy impacted negatively on the support they received, acknowledging that others being aware of the condition does not equate to being supported.

Though many participants identified a close support network, predominantly consisting of family, few felt that this adequately met their needs. Previous research has suggested that parents have not found it helpful to talk to relatives (Duguid et al., 2007) and the limitations of the support network identified by participants in the present study may further explain the limitations to the support that family can offer.

Those participants who seemed to lack a support network identified isolating themselves, and the strain placed on family relationships by the diagnosis. Such factors may

indicate poor family cohesion resulting in a lack of positive family coping (McCubbin & McCubbin, 1993) for these individuals.

The majority of participants described difficulties in having their emotional needs acknowledged within the medical setting. For some participants, not only did they feel that medical professionals did not understand the emotional impact of the diagnosis, some also felt that professionals missed opportunities to identify their distress and offer support. Similar findings have been reported in areas of childhood chronic illness, where professionals lack the time, or training, to address non-medical factors in care (Farmer, Marien, Clark, Sherman, & Selva, 2004).

The importance of support which generated feelings of acceptance and reduced feelings of isolation was identified and occurred occasionally with medical professionals, but predominantly through contact with other mothers in a similar situation. For mothers to find a space where these supportive qualities exist is clearly important and parent support groups may be one such space. The value of parent support groups in child chronic health is widely accepted and the potential benefits for parents of children with DSD have been highlighted here and previously by Lee et al. (2006). Despite this, complexities which may exist in accessing support or advocacy groups around DSD have been identified (Lee & Houk, 2010).

The majority of participants spoke about the need for ongoing and continuous support, rather than intermittent, or crisis based support. The importance of knowing that support was there should it be needed seemed central to participants feeling well supported, and several participants felt that it would be helpful to have an allocated worker, who could be the main point of contact, taking an overall view of the child's care.

Participants' understanding of the condition and its cause was often poorly facilitated by professionals. Previous research has highlighted the importance of gaining information from the medical team in reducing the stress of parents of children with DSD in the early

stages of diagnosis (Crissman et al., 2011). Factors contributing to the poor facilitation of understanding identified in this study are similar to those that have previously been reported including too much information at an inappropriate time (Sanders et al., 2011), the use of technical language (Crissman et al., 2011) and difficulties in communicating with professionals (Duguid et al., 2007). Participants also spoke of their own search for information, which although positive for some, proved overly negative, and difficult to contextualise for others. While no previous literature appears to address the potential benefits, or otherwise, of seeking information regarding DSD over the internet, Lee and Houk, (2010) recently identified the need for a cautious approach to information available from support and advocacy groups, particularly in light of the changing treatment of DSD conditions.

### **Limitations and areas for further research**

While this study benefitted from participants whose children were at a range of developmental stages, the variation in time since diagnosis may have impacted on the mother's adjustment to their child's diagnosis, and also the treatments which may have been received. While it is felt that participants in this research reported a wide range of experiences regarding support, it is acknowledged that the opportunistic nature of this sample may have resulted in a bias for mothers who were, at the time of the interview, generally coping with their child's diagnosis. It is also acknowledged that this sample was limited to white British mothers and therefore the findings of this study may not generalise well to mothers of other ethnicities.

Further research should seek to expand on the impact of DSD on the family. This could usefully look at the experiences and needs of fathers, as research in other areas of child health has indicated that mothers and fathers may differ in their responses to child illness (Knafl & Zoeller, 2000), and fathers are an under-represented group in child health research



(Ware & Raval, 2007). Research could also consider the impact on siblings given that for some DSD, siblings may be carriers of the condition. Understanding when and how families address this with siblings, and how they are supported in this process is important.

### **Clinical Implications**

The consensus statement on management of intersex disorders (Hughes et al., 2006) highlights psychology as a core member of the multidisciplinary team in providing treatment to these individuals. The findings of the current research reveal clear and important clinical implications for those working with individuals with DSD and their families which may be a useful addition to the consensus statement in guiding best practice guidelines in this area.

There is a need to develop awareness among all professionals working with these families about the potential emotional impact on the family and the need to make emotional support available. It needs to be understood that the medical system does not readily facilitate the emotional support which families may require and therefore this needs to be given particular consideration in addition to the medical care which is provided.

Training needs to be provided to professionals in contact with these families to ensure that they have the necessary skills to identify when the family or parents may need emotional support and to provide basic emotional support. These professionals need to be aware of how and where to refer families or parents who may need more extensive or specialist support, and families should routinely, and regularly be offered the option of meeting with a clinical psychologist throughout the child's development as the emotional support needs of the parents change over time.

Parents need to be provided with detailed information about the aetiology and course of the diagnosis and to be provided with information regarding not just the physical implications but also psychosocial implications. While this may be usefully done through the use of resource packs or books which are available, this needs to be carefully facilitated by

professionals. While mothers want accurate and clear information about the child's future, this needs to be facilitated in a manner which does not provoke anxieties about the future and allows questions to be raised. In addition, to aid this process of understanding parents or carers should be offered a follow up appointment shortly after the diagnosis has been made in which more detailed discussion about the diagnosis and its implications can take place. Reducing isolation among mothers of children with DSD is important and in the correct environment, contact with other mothers appears to facilitate acceptance and understanding. Hospitals which offer specialist DSD clinics should set up means by which parents can be in contact with other parents of children with DSD if they choose to be. This may be facilitated on a 'pairing' basis or through the facilitation of parent support groups. As such parents should be asked whether they would like to have contact with other parents.

As continuity of support was identified as key, a means of providing support outside of medical appointments is required. Allocating a clinician who is available outside of these appointments to be a main point of contact who can deal with concerns or signpost to other professionals or services is important in mothers' sense of feeling supported.

### **Conclusions**

This is the first study to specifically explore the support needs and experiences of mothers of children with DSD. Key support needs of facilitating knowledge and emotional support were identified by mothers and periods of time in the child's development when these needs were most prevalent were identified. Support at the time of diagnosis and approaching the child's puberty were key. Participants identified the difficulties in accessing emotional support in a medical setting and discussed how these needs were, for some, met elsewhere, either in small support networks, or through mothers with shared experience. While privacy about the condition was a priority, few felt that this directly impacted on support. These findings add to the existing literature on parental experiences of having a child with DSD and

expand our understanding of the role of support. The findings have important clinical implications for professionals working with families of children with DSD.

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**Section C: Appendices of Supporting Material**

A thesis submitted in partial fulfilment of the requirements of  
Canterbury Christ Church University for the degree of  
Doctor of Clinical Psychology  
April 2014

SALOMONS  
CANTERBURY CHRIST CHURCH UNIVERSITY

*Please note: Any identifying features have been removed ensure anonymity and maintain confidentiality.*



## Appendix A

### Literature search strategy for review in Part A.

#### **Inclusion criteria**

Articles were required to assess an aspect of development, or psychological wellbeing among individuals diagnosed with a DSD. The sample of individuals in the study had to consist of children, adolescents or young adults. In effect this meant an age range somewhere between 0 and 25 years. All DSD diagnoses that could be classified under the Consensus statement on the Management of Intersex Disorders (Lee, Houk, Ahmed, & Hughes, 2006) were included in the review. Articles had to be published in a peer reviewed journal, in English.

#### **Exclusion criteria**

Articles which focused on only surgical (including satisfaction with genital surgery and satisfaction with appearance of genitalia), or medical outcomes were excluded. Articles where the age range of participants could not be determined were excluded. Articles employing a sample group which included both DSD diagnoses and other related diagnoses were excluded, unless the presentation of the results for DSD were partitioned off and could be interpreted individually. Individual case studies were excluded.

#### **Search strategy**

The following databases were searched from 1955 up until the end of March 2014: *Science Direct, Medline, BioMed* and *PsycInfo*.

This range of databases was used to ensure articles published in both psychological and medical journals were captured in the search. The year 1955 was chosen as a start date as this has been cited as the time at which there began to be extensive publication of articles in

peer-reviewed scientific journals on DSD conditions, then termed Intersex. (Wisniewski & Mazur, 2009).

The key search terms of DISORDER OF SEX DEVELOPMENT or INTERSEX, HERMAPHRODITE or AMBIGUOUS GENITAL\* or CONGENITAL ADRENAL HYPERPLASIA or ANDROGEN INSENSITIVITY SYNDROME were used.

These terms were combined with PSYCHOSOCIAL or PSYCHOSEXUAL or WELLBEING or QUALITY OF LIFE or MENTAL HEALTH or GENDER IDENTITY or SEXUALITY or ATTACHMENT or FRIEND\*

### **Study selection process**

Abstracts of search results were screened to determine relevance to the topic area, and sample of interest. Where clarification of the sample age range was required, the full article was accessed and the participants section reviewed to determine the age range of participants. The reference sections of identified articles were reviewed to check for any additional articles not identified in the search. Thirty papers were identified for inclusion in the review.

### **Study Categorisation**

The identified articles covered four main topics relevant to development. These topics are psychosexual development, cognitive function, relationship and social adaptation, and psychological wellbeing. Many articles provided information relevant to more than one of these domains.

## Appendix B

Summary table of papers identified and included in the review

General Study information		Participants					Results			
Authors and country	Area of relevance to the review	Diagnostic group	n (Study group only)	Sex	Age range	Response rate	Measures	Respondent	Control group	Overall findings
<b>Berenbaum, (1999).</b>  USA	Psychosexual development- Gender role behaviour	CAH	42	Girls 34 Boys 18	9-19	90%	(a) Activity preferences, (b) Occupational interests (c) Composite measure- Male-typical interests, Female typical interests,	Parent and adolescents Adolescents	Unaffected male and female relatives	<i>CAH- Control participant comparisons</i> Girls with CAH differed from the control girls across all scales, all differences were significant except for adolescents' self report of female typical activities Boys with CAH did not significantly differ from control boys on any measure  <i>Comparison of all groups</i> On average, girls with CAH had no significant sex-typed activity and interest preference, whereas control girls and all boys showed sex-typical preferences
<b>Berenbaum,</b>	Psychological	CAH	67	Girls 54	3-19	90%	(a) Child	Parent	Unaffected	<i>CAH-Control</i>

<b>Bryk, Duck, &amp; Resnick, (2004).</b> USA	wellbeing- Behavioural Adaptation and Self Image			Boys 33	Sample 1 only		Behaviour Checklist (CBCL) (b) Self Image Questionnaire for Young Adolescents (SIQYA) (participants 9 years and above)	Children/ Adolescents	male and female relatives	<i>participant comparisons</i> Girls with CAH did not differ significantly from unaffected female controls on any measure. Boys with CAH did not differ significantly from unaffected boy controls on any measure.  Psychological adjustment was unrelated to indicators of disease or genital characteristics.
<b>Berenbaum &amp; Hines, (1992).</b> USA	Psychosexual development- Gender role behaviour	CAH	37	26 girls 11 boys	3-8 yrs	Unknown	Toy preference play task	Researcher rated	Unaffected male and female siblings	Girls with CAH showed significantly increased play with boys' toys and reduced play with girls' toys unaffected female relatives.  Boys with CAH did not differ from their male relatives in play with boys' or girls' toys
<b>Berenbaum &amp; Resnik, (1997).</b> USA	Psychosexual development- Gender role behaviour	CAH	35	Girls 20 Boys 15	2.7-12.6  Sample 3 only.	Unknown	Aggression Modified version of Reinisch Aggression Inventory (RAI).	Parent report	Unaffected male and female relatives	<i>CAH Participant- Control Comparisons</i> CAH girls had higher scores on RAI but this was non-

										significant CAH boys obtained similar scores to control boys on RAI
<b>Berenbaum &amp; Snyder, (1995).</b>  USA	Psychosexual development- Gender role behaviour	CAH	43	24 girls 19 boys	2.5-12 yrs	Unknown	(a) observation of sex-typed toy play (b) child's selection of a toy to keep (c) Child Game Participation Questionnaire (CGPQ) (d) playmate preference	Researcher  Child/ parent report	Unaffected male and females siblings and cousins	<p><i>CAH participant-Control comparisons</i> CAH girls spent significantly more time playing with boys' toys and less time playing with girls' toys compared with their controls. CAH girls were more likely to choose a boys toy.</p> <p><i>CGPQ</i> Parent and child CAH girls scored higher on Masculine non-athletic, and Athletic play and lower on Feminine Play than control girls.</p> <p>CAH boys were very similar to control boys on all measures. No significant differences on any measure.</p> <p><i>Relationship between sex typed activities and playmate</i> No significant correlations found</p>



										between playmate preference and masculine activities (composite score) for any group
<b>Crawford, Warne, Grover, Southwell, &amp; Hutson, (2009).</b>  Australia	Psychological wellbeing-QOL  Psychosexual development-Gender Identity	DSD	n= 54	Girls 19 Boys 22	5-10	Unknown	(a) PedsQL Pediatric Quality of Life Inventory (b) Gender Identity Questionnaire for Children (GIQC)	Children and Parent Parent	Published values on healthy children only for comparison	<i>PedsQOL</i> Parent and child rated physical quality of life as close to published values, with the exception of male children who rated their physical quality of life as lower. Both parents and children reported psychosocial quality of life as lower than healthy children.  <i>GIQC</i> 1/19 males and 3/19 females had a score of 3 or less, suggesting a risk of gender identity disorder if gender dysphoria.
<b>Gordon, Lee, Dulcan, &amp; Finegold, (1986).</b>  USA	Social and sexual adaptation-social adaptation  Psychological wellbeing-Behavioural adaptation and	CAH	16	Girls	6-16	80%	(a) CBCL- social competency and problem behaviours scale (b) Self-Perception Profile for Children (Under 14 years only)	Parent  Child	Age appropriate girls with minor thyroid problems and healthy community controls	<i>CBCL</i> Overall, no significant differences on CBCL for CAH and control group. For girls aged between 6-11 years, significant differences were reported for somatic

	self-perception									<p>complaints and schizoid/obsessive subscales between CAH and community controls.</p> <p><i>CBCL- Psychosocial</i> No difference between CAH and control groups for social competency scores.</p> <p><i>Self-Perception profile for Children</i> Those with CAH did not differ significantly from control groups on any subscale. Score within normal range</p>
<p><b>Gupta, Bhardwaj, Sharma, Ammini, &amp; Gupta, (2010).</b></p> <p>India</p>	<p>Psychosexual development- Gender Identity</p> <p>Social and sexual adaptation- Romantic relationships Social adjustment</p>	DSD	n= 60	Males	15-25	Unknown	<p>Interview assessing: (a) Gender acceptance, (b) Family adjustments, (c) Marriage expectations (d) Social adjustments</p>	Researcher interview	None	<p><i>Gender acceptance</i> 85% felt satisfied with their gender assignment, and 100% of parents felt satisfied with the gender assignment of their child. 92% accepted a masculine gender role.</p> <p><i>Sexual orientation</i> 85% (51/60) reported a heterosexual orientation, 7% were undecided, and 8% did not wish to answer the question</p>

										<p><i>Family adjustments</i> 85% reported “good” family relationships. 15% reported fitting in with society.</p> <p><i>Marriage expectations</i> Two participants were married. 44.8% reported considering marriage in the future, with the remainder reporting that they would not consider marriage.</p> <p><i>Social adjustments</i> 70% had apprehensions about the future including jobs, marriage, and acceptance in society.</p>
<p><b>Hampson, Rovet, &amp; Altman, (1998).</b>  Canada</p>	Cognition	CAH	n=12	Girls= 7 Boys= 5	8-12		<p>Spatial reasoning- The Spatial Relations Test Perceptual Speed Test</p> <p>IQ</p>	Researcher	Unaffected sibling controls	<p><i>Spatial reasoning</i> Girls with CAH achieved significantly higher spatial scores than control girls. Boys with CAH showed significantly lower spatial scores than control boys.</p> <p><i>IQ</i> No differences were reported between individuals with</p>



										<p>activities. <i>GBAQ</i> Boys with and without DSD were not observed to show significant differences on this scale. Those with less severe DSD raised as girls demonstrated lower scores on the femininity scale than control females but this was not significant and signs of gender identity confusion or instability were not observed.</p> <p>Androgen effects were significantly related to gender related behaviours.</p>
<p><b>Kleinemeier, Jürgensen, Lux, Widenka, &amp; Thyen, (2010).</b>  Germany</p>	<p>Psychological factors- QOL, Body Image,  Social and Sexual adaption- Sexual adaption</p>	DSD	n= 60	Girls = 54 Boys = 6	13-16	Unknown	<p>(a)HRQOL by KINDLr (b) mental health by Strengths and Difficulties questionnaire (SDQ) (c) Body Image by Body Image Scale (BIS) (d) unstandardised biographical questionnaire about sexual</p>	Adolescent	School survey respondents	<p><i>HRQOL</i> Overall, no difference between DSD and reference group</p> <p><i>Mental Health-SDQ</i> No significant difference between DSD and reference group either overall or at subscale level. No differences between different</p>

							activities and coping with DSD			<p>DSD diagnoses.</p> <p><i>Body Image</i> Boys with DSD have more negative body image concerning primary sex characteristics compared to a reference group boys (<math>t = 2.463, df = 239, p = .014</math>). Girls with DSD did not differ from girls in reference group</p> <p><i>Sexual activities</i> No difference concerning romantic relationships. Boys with DSD did not differ in their sexual activities from reference group boys. Girls with DSD had less experience in some sexual activities than reference group females. No significant difference in sexual activities between the different DSD diagnoses.</p> <p>Adolescents with induced puberty had fallen in love less</p>
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										frequently experienced less cuddling and had less experience with sexual intercourse.  <i>Coping with DSD</i> No significant differences reported between different diagnoses. Stepwise regression: significant influence of Shame/ stigmatization and openness. The lower the feelings of shame/stigmatization , and the higher the openness, the better the HrQoL
<b>Leveroni, &amp; Berenbaum, (1998)</b>  USA	Psychosexual development- Gender role	CAH	n= 39	Girls 23, Boys 16	3-12	Unknown	Melson's Questionnaire- assesses play which could be considered as 'caring' for siblings, pets, toys etc	Parent	Sibling Control	Girls with CAH had less interest in infants than their unaffected siblings  Boys with and without CAH did not differ in their caring interests.
<b>Liang, Chang, Chen, Chang, Lo, &amp; Lee (2008)</b>  Taiwan	Psychosexual development- gender identity  Sexual adaptation- Intimate relationships	CAH	n= 11	Women	8-25	22%	(a) Psychiatric diagnosis-Kiddie Schedule for Affective Disorders and Schizophrenia for School Aged Children (K-SADS-E) or	Researcher	None	<i>Psychiatric Diagnosis</i> 7/11 were assessed as meeting criteria for a psychiatric diagnosis and four were considered to have more minor psychiatric

	Mental wellbeing- Psychiatric diagnosis						Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition (DSM-IV) (b) Minor psychiatric morbidity- Chinese Health Questionnaire (CHQ) (c) Gender dysphoria- Gender Identity interview or Recalled Sex-Typed Behaviour Questionnaire	Participant  Researcher  Participant		disturbances  <i>Intimate relationships</i> None of the participants had current partners or intimate relationships, or experience of sexual intercourse  <i>Gender Identity</i> Three participants had higher scores for gender confusion. Increased atypical sex-typed behaviour in childhood was related to gender confusion and severity of psychiatric diagnosis.
<b>Meyer-Bahlburg, Dolezal, Baker, Carlson, Obeid, &amp; New (2004)</b>  USA	Psychosexual development- Gender Role	CAH	n=15	Girls	Middle childhood	Unknown	(a) The Child Game Participation Questionnaire (CGPQ) (b) Child Behaviour and Attitude Questionnaire (CBAQ) (c) Gender Observation (d) Gender Identity Interview	Parent  Parent  Researcher  Participant	Control group of unaffected boys and girls	<i>Gender related behaviour</i> CAH girls scored significantly higher for masculine behaviour  <i>Gender Identity Interview</i> No significant differences between CAH and unaffected girls.
<b>Mueller,</b>	Cognition	CAH	n=27	Female 12,	Adolescent	Unknown	Saccade accuracy,		Unaffected	Healthy participants



<b>Daniele, MacIntyre, Korelitz, Carlisi, Hardin, Van Ryzin, Merke, &amp; Ernst (2013)</b>  USA				Male 15	s		in trials with and without incentives		individuals	successfully improved inhibitory control (anti-saccade accuracy) during incentive trials relative to non-incentive trials.  CAH adolescents did not improve inhibitory control during incentive trials and this was not moderated by CAH severity or individual hormone levels
<b>Mueller, Ng, Sinaii, Lescjek, Green-Golan, VanRyzin, Ernst, &amp; Merke (2010)</b>  USA	Psychological wellbeing- Mental health diagnosis	CAH	n=54	Female 21, Male 33	8-18	100%	Psychiatric diagnosis Kiddie Schedule for Affective Disorders and Schizophrenia-Present and Lifetime Version (K-SADS-PL)	Mother respondent	Comparison with estimated population prevalence	44.4% of those with CAH met the criteria for at least one psychiatric diagnosis in their lifetime  Boys with CAH showed increased rates of anxiety disorder and ADHD, females showed increased rate of anxiety disorder (in relation to estimated population prevalence).
Nass & Baker, (1991)  USA	Cognition	CAH	n= 38	Female 18, Male 20	Children	Unknown	Verbal- Performance discrepancy as measured by WISC	Researcher	Unaffected Siblings	<i>Verbal-Performance discrepancy</i> 24% of those with CAH and 19% of unaffected siblings had a LD as defined

										by a Verbal-Performance IQ discrepancy $\geq 15$  Verbal-Performance IQ discrepancy of females with CAH is in the male range, similar to males with and without CAH
<b>Oner, Aycan, Tiryaki, Soy, Cetinkaya, &amp; Kibar (2009).</b>  Turkey	Psychological factors and Psychosexual development	CAH	n= 28	Females	8-20	(a) Kiddie Schedule for Affective Disorders and Schizophrenia-Present and Lifetime Version (K-SADS-PL) (b) Gender Identity Interview (GII) (c) Child Behaviour Bhecklist (CBCL) (d) Teacher Report Form (TRF) (e) Strenghts and Difficulties Questionnaire (SDQ) (f) Child Depression Inventory (CDI) (g) Chidlrens Play Questionnaire (CPQ) (h) Gender Identity Questionnaire	Researcher  Researcher  Parents  Teacher  Parents  Participants  Parents  Researcher	matched females with Diabetes Mellitus and healthy controls	<i>Behavioural problems</i> CBCL Externalisation Problems, CBCL Total Problems,TRF Aggressive behaviours cores were significantly higher in the CAH group.  Patients with higher mean testosterone levels had higher internalisation and externalisation and total problem scores- parent and teacher rated.  Higher total behavioural problems were significantly associated with poor satisfaction with genital appearance,	

							(GIQ) (J) Body Parts Satisfaction Scale (BPSS)	Researcher	<p>poor surgical procedures, and higher testosterone treatment.</p> <p>No significant differences in CDI and SDQ scores between groups</p> <p><i>Play</i> CAH patients significantly more likely to engage in male typed play on CPQ, and had higher masculinity and lower femininity scores in GIQ.</p> <p><i>Gender Identity</i> No participants met criteria for gender identity disorder</p> <p><i>Diagnosis</i> Diagnosis of psychiatric disorders highest in CAH group (57.1%) but not significantly different across groups. Patients with CAH had significantly higher risk of having an anxiety disorder only.</p>
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<p><b>Pasterski, Geffner, Brain, Hindmarsh, Brook, &amp; Hines (2005)</b></p> <p>USA and UK</p>	<p>Psychosexual development- Gender role</p>	<p>CAH</p>	<p>n= 65</p>	<p>Females 34 , Males 31</p>	<p>3-10</p>	<p>Unknown</p>	<p>Videotaped play sessions with male and female typical toys- toy choice and parental response</p>	<p>Researcher</p>	<p>Unaffected siblings</p>	<p><i>Toy choices</i> CAH girls played with girls toys less and boys toys more than unaffected girls.</p> <p><i>Parental responses to girls with and without CAH</i> Mothers and fathers gave more positive responses to their daughter with CAH than to unaffected daughters when playing with girls toys</p> <p><i>Parental responses to boys with and without CAH</i> Mothers gave fewer negative responses to their sons with CAH than to their unaffected sons for play with boys toys</p>
<p><b>Pasterski, Geffner, Brain, Hindmarsh, Brook, &amp; Hines (2011)</b></p> <p>UK and USA</p>	<p>Psychosexual development- Gender role</p>	<p>CAH</p>	<p>n= 57</p>	<p>Females 26, Males 31</p>	<p>3-10</p>	<p>Unknown</p>	<p>Playmate and Play style Preferences Structured Interview (PPSI)</p>	<p>Researcher administered</p>	<p>Unaffected siblings</p>	<p><i>Playmate Preference</i> Unaffected girls chose more female targets as playmates than unaffected boys, boys with CAH, or girls with CAH.</p> <p>Girls with CAH chose more female</p>

										<p>targets than unaffected boys or boys with CAH.</p> <p>No difference between boys with and without CAH.</p> <p><i>Toy choice</i>          Unaffected girls chose more girls toys than unaffected boys, boys with CAH, or girls with CAH. Girls with CAH chose girls toys more than unaffected boys or boys with CAH. No difference between boys with and without CAH</p> <p><i>Play style</i>          Girls with CAH preferred less rough and tumble play styles than unaffected boys but not boys with CAH. Boys with CAH preferred less rough and tumble play to unaffected boys</p> <p><i>Conflict situation</i>          Girls with CAH tended to choose playmates engaged in a male activity.</p>
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<p><b>Pasterski, Hindmarsh, Geffner, Brook, Brain, &amp; Hines (2007)</b></p> <p>USA and UK</p>	<p>Psychosexual development-gender role</p>	<p>CAH</p>	<p>n=67</p>	<p>Girls= 38, boys= 29</p>	<p>3-11</p>	<p>Unknown</p>	<p>Aggression and activity level assessed using the Activity Level/ Extraversion Questionnaire</p>	<p>Parent rated</p>	<p>Unaffected siblings</p>	<p><i>Aggression</i> Unaffected boys were more aggressive than unaffected girls. Girl with CAH were more aggressive than unaffected girls. Girls with CAH, boys with CAH, and unaffected boys did not differ.</p> <p><i>Activity level</i> Unaffected girls were more active than unaffected girls. Girls with CAH were more active than unaffected girls. Girls with CAH, boys with CAH and unaffected boys did not differ from one another.</p>
<p><b>Plante, Boliek, Binkiewicz, &amp; Erly (1996)</b></p> <p>USA</p>	<p>Cognition</p>	<p>CAH</p>	<p>n=11</p>	<p>Girls 9, Boys 2</p>	<p>4-15</p>		<p>(a) Edinburgh Handedness Inventory, (b) Leiter International Performance Scale, (c) Clinical Evaluation of Language Fundamentals-Revised (CELF-R)</p>	<p>Researcher administered and rated</p>	<p>Non CAH Siblings and unrelated controls</p>	<p><i>CELF-R</i> Four individuals with CAH obtained scores indicating impaired language skills.</p> <p>Six of eight CAH families were reported by parents to have a history of speech or language LD. Three of 16 non CAH families</p>

										<p>reported a history of speech or language LD.</p> <p><i>Leiter Scores</i> Leiter scores for subjects with CAH were typically lower than for control subjects.</p>
<p><b>Sanches, Wieggers, Otten, &amp; Claahsen-van der Gritten (2012)</b></p> <p>The Netherlands</p>	<p>Sexual and social adaptation- Social adaptation</p>	CAH	n= 106	Female= 50, Male= 56	0-18	69.7%	<p>(a) Burden of CAH (b) participation in school and leisure time</p>	Parent or self report	No comparison group	<p><i>Burden of the condition</i> 8% of parents reported child to have constraints on daily life due to CAH. 30% had been absent from school in the past year due to CAH. 96% of parents satisfied with child's overall health. All adolescents satisfied with their own health</p> <p><i>Participation</i> 82% of children engage in sport and 84% reported children over 4 engage in hobbies 28% of children over 12 have a part time job. 85% or parents indicated that their children sometimes</p>

										spend a night away from home.
<b>Servin, Nordenström, Larsson, &amp; Bohlin (2003).</b>  Sweden	Psychosexual development-gender role	CAH-separated into severe and mild	n= 26	Girls	2-10	84%	(a) toy play  (b) Other gender typed interests-best friend, career options  (c) child behaviour and characteristics	Researcher observation Child report  Parent report	Control group	<p><i>Toy Play</i> All groups spent more time with the masculine toys but the preference was only significant for the severe CAH group, the mild CAH group and the control group.</p> <p><i>Best Friend and Choice of Toy</i> Two thirds of the girls with a severe form of CAH reported a boy as their best friend. The majority of girls with the mild, and all unaffected girls reported being best friends with a girl. Significant differences were reported between CAH and control with CAH girls more likely to select a masculine toy, and a male best friend.</p> <p><i>Career questions</i> Girls with CAH answered 'yes' to a greater extent with regard to masculine</p>



										career questions than did control girls.
<b>Sinforiana, Livieri, Mauri, Bilio, Sibilla, Chiesa, &amp; Martelli (1994)</b>  Italy	Cognitive	CAH	n= 19	Females 7, Males 12	16-24	Unknown	Neuro-psychological evaluation including the WAIS	Researcher	Matched control group	No significant differences were found between CAH subjects' and control subjects' WAIS scores. No differences were found with regard to type of CAH. No patient had a learning disability defined as Verbal-Performance- IQ difference > 15.
<b>Slijper, Drop, Molenaar, &amp; Muinck Keizer-Schrama (1998).</b>  The Netherlands	Psychological wellbeing- Mental health diagnosis  Psychosexual development- Gender identity	DSD	n= 59	Females 54, Males 5	Children	Unknown	Semi-structured psychiatric interview		No control group	<i>Psychiatric Diagnosis</i> 39% met criteria for a psychiatric diagnosis Mild psychological problems in a further 19%.  <i>Gender Identity</i> 13% of girls were identified as having gender identity disorder based on DSM-IV criteria.
<b>Temple, &amp; Sanfilippo (2003)</b>  UK	Cognitive	Klinefelter's syndrome	n= 3	Boys	10 years		Battery of executive tasks	Researcher	Matched controls	Impairments in executive function in comparison to controls- inhibitory skills only Concept formation, problem solving, task switching, and

										speeded responses are normal
<b>Willihngaz-Lawson, Isharwal, Lewis, Sarafoglou, Boisclair-Fahey, &amp; Shukla (2012)</b>  USA	Psychological wellbeing-QOL	DSD	n= 10	Girls	5-24 years		Quality of life (QOL)- Glasgow Children's benefit inventory (GCBI)	Self report and parent report	No comparison group	3/5 indicated that there was 'no change' or 'a little better' improvement in the QOL after their surgical experiences  Total GCBI score for 3/5 was neutral or positive. The two patients with positive scores were the youngest two in the group with questionnaires completed by parents. The two patients with the negative scores were the older two of the subset.
<b>Zhu, Hu, Wan, Li, You, Gao, &amp; Feng (2012)</b>  China	Social and sexual adaptation-social adaptation  Psychological wellbeing-behavioural adjustment	DSD	n= 50	Female 42, Male 8	6-17 years		Socio- emotional evaluation and psychological evaluation as measured by Child Behaviour Checklist (CBCL)	Parent report	Matched controls	<i>Socio-emotional evaluation</i> 4/8 boys exhibited social limitations including having few friends, poor communication with others, and academic struggles. No problems were reported in males in the control group. 12/42 girls with DSD showed social limitations, with poor

									<p>communication, and poor academic performance. This rate did not differ significantly from unaffected girls.</p> <p><i>Psychological evaluation</i> All 8 boys with DSD had psychological abnormalities, whereas such psychological difficulties were rare in the control males, resulting in a significant difference between these groups. Psychological and behavioural problems were reported as significantly more frequent among girls with DSD and control girls.</p>
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## Appendix C Interview Schedule

### Introduction questions:

1. Can you tell me a little bit about your family?  
*(Prompts: Who lives at home with you? How old are they? What do they do? Are they around at home much? What kinds of things do you like doing? Either as a family or as individuals?)*
2. I know that your child (insert child's name) has a diagnosis of (insert diagnosis), could you tell me a little bit about when and how your child was diagnosed with this?  
*(Prompts: What was that like for you? Who told you and what words did they use? Did you feel you understood? Who did you tell? How did you tell others?)*
3. Earlier you were telling me about when your child was diagnosed with DSD, can you tell me about how you were supported at this time?  
*(Prompts: Who supported you? Who did you talk to? Did you seek your own support away from the professionals? Where? Who? Friends/Family/Partner? What did they do to help you? Was it the right kind of support for you at the time of the diagnosis?)*
4. Since then, in what other ways have you accessed support?  
*(Prompts: From who? What kinds of support? From friends, partner, support groups, professionals, peer groups, work colleagues, social, emotional, practical? How has it felt to receive this support?)*
5. Could you tell me a bit about your most supportive experience?  
*(Who/where did it come from? What did they do and how? What made it better than other experiences of support? Specifically what was useful about this experience? How did this experience make you feel?)*
6. Could you tell me about a time when you sought support and you feel that you did not get it?  
*(Who/where were you seeking support from? What was it about that experience that was unsupportive? Why do you think they were unable to support you in the way you wanted? Has it impacted on how you currently access support? How did this make you feel?)*
7. Thinking about from when your child was diagnosed to now, when have been the times in your child's development that you have felt most that you needed support?  
*(Prompts: How were things when (child's name) was a toddler/first went to school/ was a young child/teenager/puberty? If any, what were the particular challenges at this time? And how were you supported with these?)*
8. What are some of the things that have got in the way of you feeling able to access support?  
*(Prompts: Have you ever had doubts about asking others for support? Why? What were you concerned about? Have you had worries about talking with others about child's diagnosis in order to access support?)*
9. What might it be useful for other parents of children with DSD to know about accessing support?

10. Is there anything else about your experience of accessing support that you feel important to discuss today?

## Appendix D

### NHS Participant Information Sheet

#### **Participant information sheet- Information about the research**

##### **Mothers of children with Disorders of Sex Development: Experiences of accessing support**

You are invited to take part in a research study. This research is being carried out in part fulfilment of a Doctoral Degree in Clinical Psychology at [REDACTED]. Before you decide whether you would like to take part it is important that you understand why the research is being done and what it would involve for you.

##### **What is the purpose of the study?**

The purpose of this study is to understand more about parents' experiences of seeking support when they have a child with a diagnosis of a Disorder of Sex Development (DSD). I hope to understand more about the times when families most need support, the kinds of support that parents seek and from whom, and how they may have experienced this process. It is hoped that this will help to improve the support and advice that services are currently offering to families in similar positions.

##### **Why have I been invited?**

You have been invited to participate because you are the mother of a child with a DSD and your child has been involved with the Paediatric Endocrinology service at [REDACTED] Hospital.

##### **Do I have to take part?**

Participation in this research is completely voluntary and it is up to you to decide whether you wish to participate. If you agree to take part, you may withdraw at any stage. Whether you decide to participate or not, the standard of care that you receive from [REDACTED] Hospital will not change in any way.

##### **What will happen to me if I take part?**

Once you have read this information, if you think you might be interested in participating please contact me on [REDACTED]. This will give us the opportunity to discuss the project and what it involves in more detail, and I can answer any question you may have. In calling, you are not agreeing to participate, however you will be giving permission to have your name and contact details recorded. Please note that I am not based at this office every day, but please leave a message with your name and contact number and I will call you back as soon as possible. If when you received this information sheet you agreed to be contacted and I do not hear back from you in two weeks time, I will call you to discuss the research and find out whether you are interested in participating.

If you choose to take part, I will arrange a suitable time for you to attend an interview at [REDACTED] Hospital. Your travel expenses, up to a value of £10 can be reimbursed. Participation will involve meeting for one interview session with myself. This interview will last between 40-90 minutes but please allow two hours in total so that there is the opportunity for you to ask questions.

All interviews will be audio recorded, these recordings will be transcribed and anonymised following the interview, your name and the name of your child will be removed from these

transcriptions. During the interview you will be asked a series of questions which will focus on your experiences of accessing support since your child was born. If at any stage you wish to take a break or to end the interview you may do so.

### **What are the possible disadvantages and risks of taking part?**

Participating in the interview will involve discussing topics which are personal to you and your family. It is therefore possible that taking part may cause you some upset or distress. However, every step will be taken to ensure your wellbeing throughout the interview. The research is supervised by [REDACTED], clinical psychologist in the paediatric endocrine team. If after your interview you would like to discuss anything that the interview has brought up for you with the clinical psychologist, this can be arranged at a date that suits you.

### **What are the possible benefits of taking part?**

Although participation in the research may not directly benefit you and your child, it is hoped that the information obtained from the interviews will help families in similar circumstances in the future.

### **What if there is a problem?**

Any concern or complaint about the study will be taken very seriously. You may speak with myself or the clinical psychologist, [REDACTED], working in the paediatric endocrine team if you wish. If you would prefer to speak with someone else please [REDACTED] on the contact details provided below.

### **Who is organising and funding the research?**

This research is a collaboration between Canterbury Christ Church University and [REDACTED] NHS Foundation Trust. It is being funded by Canterbury Christ Church University. The research has been reviewed and approved by Canterbury Christ Church University, the NHS Research Ethics Committee, and the [REDACTED] Research and Development department.

### **Will my taking part in this study be kept confidential?**

Ethical and legal practices will be followed and all information about you will be handled in confidence. Any information which is collected about you during the course of the research will be kept strictly confidential, and any information about you which leaves the hospital and university grounds will have any identifiable information about you, such as your name, removed. Any identifiable information from your interview will be disguised so that your anonymity is maintained throughout the research process.

The only circumstances under which this confidentiality may be broken is if you report something in the interview which makes me feel concerned for your safety or the safety of others. In this situation I may have to discuss what you have told me with other parties, however I would always try to discuss this with you first.

### **What will happen to the results of the research study?**

The results of the study will be used in the research report submitted to Canterbury Christ Church University, and it is intended that this report will also be published. If you wish to see a copy of the report before publication please contact me. I will also arrange to meet with participants if they wish to know about the outcome of the study. Participant names, and any other information that may identify you will not be included in the report. Quotations from the interviews may be included in the report but these will be completely anonymised to maintain your confidentiality.

The data which is collected from your interview will be stored for 10 years. This will be stored anonymously and on an encrypted memory stick in a locked location at the university.

**What will happen if I don't want to carry on with the study?**

If you decide at any stage that you want to withdraw from the study you are free to do so. If analysis of the interviews has not yet taken place your data will be removed from the study entirely. If however the data analysis has already taken place and the report has been written then we will not be able to do this.

**What if there is a problem or you have a complaint?**

Any concern or complaint about this study will be taken seriously. I would encourage you to talk to myself in the first instance in the hope that we can resolve your concerns. If however you would prefer not to speak with myself, or if having spoken with me you remain unhappy and wish to make a formal complaint please contact [REDACTED] on the telephone number below.

**Contact Details and Complaints****Researcher- Clare Chivers**

[REDACTED] (Please note that this is the voicemail for [REDACTED], please leave a message if I am unavailable and I will call you back as soon as possible).

[REDACTED], **Canterbury Christ Church University**

[REDACTED], **Clinical Psychologist in Paediatric Endocrine Services**

**Further information**

For further information, or if you have any questions please contact me on the above telephone number. If I am unable to answer your call please leave a message stating who you are and that you are calling about the research project. Please leave a contact number and I will get back to you as soon as possible.



## Appendix E

### Charity Recruitment Information

Charitable organisation 1:

#### **Mothers' experiences of accessing support- Call for research participants**

I am a trainee clinical psychologist and I am completing some research about how families with children who have a diagnosis of Congenital Adrenal Hyperplasia (CAH), or a related condition, access support. I would like to hear from you if you are the mother of someone with CAH. I am interested to hear about your experiences of accessing support, for example, when you feel that you have most needed support, and also who you have approached for support. If you have not accessed support at all, or even just a little, I would still like to hear from you as I am also interested in finding out why it may be difficult to access support. I would like to meet with people on an individual basis to hear about their experiences and I hope that the information obtained from the interviews will help families in similar circumstances in the future, and also will guide services to better support families. Your confidentiality is very important and therefore your details will not be shared with anyone outside of the research, additionally any identifiable information about you will be anonymised to maintain your confidentiality.

If you might be interested in sharing some of your experiences I would like to hear from you. Please contact me by email [REDACTED], or by telephone (number below) in the first instance and we can discuss the research in some more detail. This does not mean you are committed to participating; initially it is just an opportunity to discuss the research further.

Thank you for your interest in this research.

Kind Regards,

[REDACTED]

(Researcher)

Email: [REDACTED]

Telephone: [REDACTED] (This is an answer phone service, **please leave my name, your name and a contact number**).

Charitable organisation 2:

### **Mothers' experiences of accessing support- Call for research participants**

I am a trainee clinical psychologist and I am completing some research about how families with children who have a diagnosis of a Disorder of Sex Development (DSD) access support. I would like to hear from you if you are the mother of someone with a DSD. I am interested to hear about your experiences of accessing support, for example, when you feel that you have most needed support, and also who you have approached for support. If you have not accessed support at all, or even just a little, I would still like to hear from you as I am also interested in finding out why it may be difficult to access support. I would like to meet with people on an individual basis to hear about their experiences and I hope that the information obtained from the interviews will help families in similar circumstances in the future, and also will guide services to better support families. Your confidentiality is very important and therefore your details will not be shared with anyone outside of the research, additionally any identifiable information about you will be anonymised to maintain your confidentiality.

If you might be interested in sharing some of your experiences I would like to hear from you. Please contact me by email [REDACTED], or by telephone (number below) in the first instance and we can discuss the research in some more detail. This does not mean you are committed to participating; initially it is just an opportunity to discuss the research further.

Thank you for your interest in this research.

Kind Regards,

[REDACTED]

(Researcher)

Email: [REDACTED]

Telephone: [REDACTED] (This is an answer phone service, **please leave my name, your name and a contact number**).

Charitable organisation 3:

### **Mothers' experiences of accessing support- Call for research participants**

I am a trainee clinical psychologist and I am completing some research about how families with children who have a diagnosis of Androgen Insensitivity Syndrome (AIS), or a related condition, access support. I would like to hear from you if you are the mother of someone with AIS. I am interested to hear about your experiences of accessing support, for example, when you feel that you have most needed support, and also who- if anyone, you have approached for support at different times. I am also interested what has been helpful, and what has been less helpful. I would like to meet with people on an individual basis to hear about their experiences and I hope that the information obtained from the interviews will help families in similar circumstances in the future, and also will guide services to better support families. Your confidentiality is very important and therefore your details will not be shared with anyone outside of the research, additionally any identifiable information about you will be anonymised to maintain your confidentiality.

If you might be interested in sharing some of your experiences I would like to hear from you. Please contact me by email [REDACTED], or by telephone (number below) in the first instance and we can discuss the research in some more detail. This does not mean you are committed to participating; initially it is just an opportunity to discuss the research further.

Thank you for your interest in this research.

Kind Regards,

[REDACTED]

(Researcher)

Email: [REDACTED]

Telephone: [REDACTED] (This is an answer phone service, **please leave my name, your name and a contact number**).

Appendix F  
Participant Consent Form

Centre Number: 01  
Study Number: 13/L0/0151  
Participant Identification Number for this study:

**CONSENT FORM**

**Title of Project:** Mothers of Children with Disorders of Sex Development: Experiences of Accessing Support

**Name of Researcher:** [REDACTED]

**Please initial box**

1. I confirm that I have read and understand the information sheet dated..... (version.....) for the above study. I have had the opportunity to consider the information, ask questions and have had these answered satisfactorily.

2. I understand that my participation is voluntary and that I am free to withdraw at any time without giving any reason, without my medical care or legal rights being affected.

3. I understand that the data collected during the study may be looked at by the lead supervisor, [REDACTED], and the second supervisor, [REDACTED]. I give permission for these individuals to have access to my data.

4. I agree for an audio recording of my interview to be taken.

5. I agree that anonymous quotes from my interview may be used in published reports of the study finding

6. I agree to take part in the above study.

Name of Participant \_\_\_\_\_ Date \_\_\_\_\_

Signature \_\_\_\_\_

Name of Person taking consent \_\_\_\_\_ Date \_\_\_\_\_

Signature \_\_\_\_\_

## Appendix G

### Debrief Protocol

- Allow participant time to think about and share their thoughts on the experience of the interview
- Ask participant whether they found it OK to talk about the topic and whether they are feeling OK about what they discussed
- Ask participant whether there was anything that they would like a follow-up telephone call about
- Provide participants with information about support groups if they want to take this information away
- Ask participant whether they have any questions or concerns about the research process or what will happen with the information they provided

## Appendix H

### Debrief and support group information

I would like to thank you for taking the time to participate in this research. Your opinions and thoughts about this subject area are very valuable.

If at any stage in the future you have any further questions about the research, or you would like to know the results then please contact me at [REDACTED]

Below are the details of some organisations which may be of interest to you, particularly if you would like to access some support.

Should you have any complaints about the research process then please either contact me or use the contact details given below to contact [REDACTED] or [REDACTED]

Thank-you again for your participation.

[REDACTED]  
(Researcher)

#### Additional Contacts

[REDACTED] **Canterbury Christ Church University**

[REDACTED] **Clinical Psychologist in Paediatric Endocrine Services**

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

## Appendix I

### Coded interview transcript

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Appendix J  
Extracts from research diary

*This has been removed from the electronic copy*



Appendix K  
Bracketing interview questions and key themes

Questions for bracketing interview were taken from Roulston, (2010).

1. Tell me about your research topic.
2. Tell me about some of your experiences [concerning the research topic].
3. What have you learned about the topic from your reading in the literature?
4. What do you expect to find from your study?
5. Why do you think this topic is important?
6. What audiences do you hope to inform with your research?

Key concepts and themes arising from the interview:

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## Appendix L

### Audit trail process

The following documents were given to the external supervisor:

- 1) Two complete, annotated transcripts
- 2) Two documents of the emerging themes listed chronologically for each of these participants
- 3) Two documents of the super-ordinate themes for these two participants, where emerging themes had been moved around to form clusters
- 4) Theme process development table which documents the final grouping and selection of emerging themes from across participants and how these were finally grouped into super-ordinate themes and then subsequently master themes (Appendix N)

Appendix M  
Approval Letters

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## Appendix N

## Theme development process

Master theme	Sub theme	Emergent theme related to sub theme	Participants with emergent theme	Total number contributing to theme
Evolving support needs	Developing support needs	Birth	1,3,4,5,8	5
		Adolescence	1,2,3,4,6,7,8	7
		Communicating to the child	2, 3,4,5,6,7,8	7
		Surgery	3,5,6,7,8	5
		Having more children	1,5	2
	Emotional support needs shifting	Diagnosis on the family/ mother	2,3,8	3
		Child's emerging emotional needs	2,3,4,6,7,8	6
		Child's need for own private space	1,3,4, 6,7	5
	Learning curve	Injections	1,2,3,6,7	5
		Medication management	1,3,6,7,8	5
Seeking understanding	Uncertainty brought by the diagnosis	Unheard of diagnosis	1,2,3,4,5,6,7,8	8
		Uncertain future for the child	1,4,5,8	4
		Needing to understand	1,2,3,5,6,7,8	7
	Professionals facilitating understanding	Understanding the diagnosis poorly/ well facilitated	1, 2, 4, 8 (poor) 5, 3(good),	6
		Understanding the cause poorly/ well facilitated	1, 5 (mixed), 8(poor)	3
		Feeling guided by professionals	3,7	2
		Barriers to understanding	1,2,5,6,7	5
		Specialist professionals	1,2, 4,5, 6,7	6
	Independently seeking information	The internet	1,2,3,4,5,6,7,8	8
	Parental emotional needs in a medical setting	Emotional impact of the diagnosis	Strong emotional impact	1,2,3,4,5,6,7,8

	Emotional support needs unacknowledged	Emotional needs and impact overlooked	1, 2,5,6,7,8	6
		Missed opportunities for support	1, 5, 8	3
		Feeling let down by professionals	2,5,7,8	4
	Not feeling heard	Own perspectives brushed aside	4,7,8	3
		Power dynamics	2, 4,5,6,7	5
		Viewed as neurotic	6,7,8	3
	Intermittent support	Support is 'there'	1,2, 3,5,6	5
		Availability of professionals (good/bad/ideal)	2 (unavailable) 3,5,6 (good) 8(ideal)	5
		Crisis control/ intermittent support	2, 4,7,8	4
	Valuable experiences addressing emotional needs	Reducing isolation, normalising and acceptance	1,3, 4,5, 7	5
		Absence of such experiences	2,8	2
	Close support networks	Privacy over support	Protection from social implication	1,2,3,4,5,6,7,8
Limited communication-Mantra			1,2,3,4,5,6,7,8	8
Privacy impacting support (Negative)			2,5,7	3
Support network		Closeness and trust	2, 3, 5, 6,7,	5
		Family and professionals and friends	2,3,4,6,7	5
		Limitations of the network	2, 3, 6,8	4
		Support type	2,6	2
		Absence of a network/ Strained relationships	1,5,7	3
Impact on 'normal' parental supports		Impact on normal supports- negative	1, 4,5,6,7	5
		Emphasising difference	1, 4, 5	3
		Relinquishing control	1,6	2

Appendix O  
Additional Quotes

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## Appendix P

### Proposed research summary for Ethics Panel and Research and Development

#### **Background:**

Disorders of sex development (DSD) are a class of congenital conditions in which the development of chromosomal, gonadal, or anatomical sex is atypical (Hughes, Houk, & Ahmed, 2006). Affected newborns typically present with atypical genitalia, whereas adolescents present with atypical sexual development during the pubertal years, such as delayed puberty (Ahmed et al., 2011). Research regarding the impact of DSD on quality of life (QOL) and mental wellbeing, presents a somewhat mixed picture, however there is a body of evidence to suggest a range of negative outcomes for those with DSD (Johannsen, Ripa, Mortensen, & Main, 2006; Brinkmann, Scützmann, Schacht, & Richter-Appelt, 2009).

Emerging research has sought to understand the impact of DSD on the parents of affected children and has demonstrated that carers of children with a DSD are at risk for increased overprotection, perceived child vulnerability, and parenting stress (Kirk et al., 2011) and parents have been reported to demonstrate reduced self-esteem and psychological stability (Duguid et al., 2007). Parental emotional responses to the diagnosis of a child's DSD include shock, grief, anger, guilt, and shame (Slijper, Frets, Boehmer, Drop, & Niermeijer, 2000). Parental adaption to a child's health condition has been identified as key for the subsequent wellbeing of the child (Carmichael & Alderson, 2004) and higher levels of parenting stress are related to poorer emotional, behavioural and social adjustment in children with chronic illness (Colletti et al., 2008). Therefore, reducing stress, and increasing adaption and coping skills for parents, can have important implications for the child, and whole family coping.

While no studies to date have specifically looked at the process or impact of support for the parents of children with a DSD, an intricate picture is emerging through studies assessing the effect of DSD on the family indicating that support from relatives may not be helpful (Duguid et al. 2007) and the wish to maintain privacy about the condition may impact on the support individuals are able to receive.

#### **Aims:**

Despite the acknowledgement that families of children with DSD experience unique challenges requiring support, very little is currently understood about the specific needs and wishes for support amongst parents. Understanding parent experiences of support will enable services to better target their support and also to provide support which is more closely attuned to the needs of the parents. Additionally, it is hoped that such information may benefit parents in similar circumstances. This study has chosen to focus on mothers as in Western culture they are traditionally the main caregiver and may be more likely to access support. Therefore, this study aims to address the following research questions

1. What are the support needs of mothers of children with DSD and how do these change over time?
2. How well have their support needs been met and was anything missing?

**Method:**

Eight mothers who were the primary care giver to a child with DSD and who met other study inclusion criteria were recruited through the hospital site and through charities. Semi-structured interviews were completed with participants which lasted between one and two hours. Interviews were analysed using Interpretative Phenomenological Analysis (IPA) (Smith, Flowers, & Larkin, 2009).

**Findings:**

Four master themes emerged of ‘evolving support needs’, ‘seeking understanding’, ‘parental emotional needs in a medical setting’, and ‘close support networks’. These themes encapsulated the kinds of support that mothers needed and when they most needed it. They also identified aspects of support that were missing, and the qualities of good support. Stages in their child’s development when mothers most needed support, included the birth, puberty, and communicating with the child about their diagnosis. Mothers emphasised the importance of developing an understanding of the child’s condition, including the cause, what the future would look like, and the treatment needs of the child. This was often poorly facilitated by professionals. Mothers also spoke about the lack of an acknowledgement of the emotional needs of the parent within the medical system, and that emotional support needed to be ongoing rather than just focusing on times of medical need or crisis. Six mothers spoke about close networks, of predominantly family, who supported them well, however for others, these networks either did not exist, or did not meet their needs. While the need to maintain privacy about the child’s diagnosis was important to all mothers, few felt that this impacted on the support that they received. Mothers spoke about positive experiences of support as those which had normalised their experience, and this most often occurred when talking to other mothers who had shared similar experiences. These findings have important clinical implications for those working in the area of DSD, particularly in considering how best to facilitate parent’s understanding of the condition, both physically and psychologically, and also in acknowledging and addressing with parents the psychological impact on the family.

**Feedback to participants:**

A fully anonymous, summary report will be developed to feedback the result to those who participated. All those who participated expressed an interest in receiving a copy of the completed report.



**Dissemination of findings:**

The findings will be submitted for publication to The International Journal of Pediatric Endocrinology and will also be presented at the British Psychological Society Children and Young Peoples Conference 2014 or another suitable conference.

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Appendix Q  
Publication guidelines for chosen journal of publication

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