



# The Psychosocial Impact of Disorders of Sexual Development

Kavita Ravendran<sup>1,2\*</sup> and Rebecca Deans<sup>1,2</sup>

<sup>1</sup>Gynaecology Research and Clinical Excellence (GRACE), Royal Hospital for Women, Australia

<sup>2</sup>University of New South Wales, Australia

## Abstract

Disorders of Sexual Development (DSD) consist of 'congenital conditions with atypical development of chromosomal, gonadal, or anatomic sex'. They cause significant psychological impact upon patients and their families. A search of MEDLINE, EMBASE and Clinical trials USA and Australia was conducted using MESH terms 'disorders of sexual development,' 'psychology' and 'quality of life.' Four themes emerged from the literature search; psychological support and treatment for parents and patients, the need for holistic management, quality of life assessment and identification of sociocultural factors that impact psychological health in DSD patients. Educating medical and paramedical staff is crucial to ensure adequate early assessment, diagnosis, counselling, and where appropriate management. Caution should be exercised in early genital surgery to ease parental distress; rather this should be a team-based decision at an appropriate age where the individual can be involved in the consent process. Psychological input should be specialised to the individual and more readily accessible.

## Introduction

Disorders of sexual development (DSD) consist of 'congenital conditions with atypical development of chromosomal, gonadal, or anatomic sex' as defined in the 2006 consensus statement(1). These disorders are rare and data on incidence is limited however they are estimated to affect 1 in 5500 new born's. The majority of DSDs are secondary to congenital adrenal hyperplasia (CAH), androgen insensitivity syndrome (AIS) and mixed gonadal dysgenesis [1]. However, it is important to note that not all patients are diagnosed at birth. These disorders are now classified into; 1) Sex chromosome DSD such as 45X Turners syndrome, XX Male and XY Female, 2) 46 XY DSD including androgen insensitivity syndrome (AIS) and testicular regression syndrome (TRS); and 3) 46 XX DSD including pure gonadal dysgenesis (PGD), and congenital adrenal hyperplasia (CAH) [2-4].

These disorders confer significant psychological impact upon patients and their families and remain challenging to holistically manage in the medical setting. Psychosocial care in these patients and families has previously been described as inadequate with minimal research in the area [5]. This review of current literature aims to shed some insight into current methods to evaluate psychological outcomes and co morbidities of DSD patients and their families, associated psychosocial and cultural factors as well as current management approaches.

## Method

A search of MEDLINE, EMBASE and Clinical trials USA and Australia was conducted using MESH terms 'disorders of sexual development,' 'psychology' and 'quality of life.' Article titles and abstracts were screened prior to evaluating individual articles. Results predominantly consisted of review articles, qualitative prospective studies and narrative articles. Articles had to include a measure of psychological impact or quality of life, or include description of contributing psychosocial factors affecting patient outcomes.

Articles that focused on psychosexual development, sexuality, gender identity or only surgical management were excluded. Non-English articles were also excluded. The time frame consisted of articles from 1990 to present acknowledging that prior research was predominantly focused on early surgical management or these disorders rather than psychosocial impact, and a paradigm shift in the management of these patients has since occurred.

## Results

Four themes merged from the literature search, namely psychological support and treatment for parents and patients, the need for holistic management, quality of life assessment and identification of sociocultural factors that impact psychological health in DSD patients. Psychosexual development, sexuality and gender identification are substantive issues that will not be discussed in depth.

## Discussion

### Parental distress

A diagnosis of DSD can be traumatic for parents, as shown in multiple qualitative studies [6-8] and appropriate parental counselling, psychological and educational support is important [9-11]. A recent study surveyed 51 parents of infants under the age of 2, diagnosed with moderate to severe DSD with no decisions made regarding gender assignment at that stage. Over 25% of parents reported reduced quality of life, 18% reported clinically

Submitted: 20 June 2019 | Accepted: 29 June 2019 | Published: 30 June 2019

\*Corresponding author: Kavita Ravendran, University of New South Wales, Australia, Tel: 614-2319-5215; Email: kavita.ravendran@gmail.com

Copyright: © 2019 Ravendran K and Deans R. This is an open-access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.

Citation: Ravendran K, Deans R (2019) The Psychosocial Impact of Disorders of Sexual Development J Sexual Med Reprod Health 2: 4.



significant symptoms of depression and 25.4% had moderate to severe anxiety. A subset (15%) reported post-traumatic stress symptoms (PTSS). Uncertainty regarding the health of the child is also expressed by parents, with higher uncertainty reported for boys, or those unsure of their child's gender [6].

Genital surgery in infancy is controversial. The traditional binary gender classification system was at the core of decision-making with the belief that children require 'unambiguous genitalia' to develop healthy gender identity [12]. This binary classification has been subject to bioethical criticism and challenged with the suggestion of incorporating 'intersex' into the classification system to allow for gender fluidity [13-15]. There is minimal outcome data to support genital surgery, with concerns that capacity for orgasm may be severely affected by early surgery as well as risks of scar tissue, stenosis and multiple procedures [12,16-18]. An older study of 59 patients found higher rates of psychopathology despite early surgical intervention [19]. Since the early 1990s, critics and lobby groups have expressed the need to end social prejudice, stigma and forced medical interventions. They have instead emphasised the need for family and patient-centres care, advocacy for self-determination and bodily integrity for these individuals [20].

Previously, early surgical intervention and gender reassignment was encouraged with a view to improving parental psychological distress [16]. On the contrary, a recent prospective study of 49 parents of children with DSD measured parental distress before and 6 months after genitoplasty, and found no significant change in parental anxiety, depression or quality life. PTSS reduced, which the authors postulated could represent parents' having come to terms with the acute stress of initial diagnosis [8]. Another similar study interviewed 45 parents prior to, and 12 months after their children underwent genitoplasty. They found significant improvement in psychological distress symptoms; however 21% of parents reported ongoing clinically relevant distress. It was unclear whether resolution in symptoms is specific to parents whose children underwent genitoplasty or if this represented adaptation consistent with passing the acute stress phase [21].

### **Individuals quality of life**

There have been multiple largely observational studies examining quality of life for individuals with DSD, mental health diagnoses and barriers to seeking psychological help [22-30]. A recent study of 110 adults with DSD found lower mental health related quality of life, and significantly higher distress including; obsessive-compulsive, depression, anxiety, paranoid ideation, hostility and psychoticism [22]. Multiple studies of school aged children have shown similar results with reduced paediatric quality of life inventory scores, and lower school functioning, particularly for subjects of assigned female sex [23,31,32]. A recent review of the literature found that dissatisfaction resulted from inadequate management rather than assigned sex or condition. Another cross-sectional study also found that individual's health status, rather than diagnosis is important [33]. This indicates the need for specialised, multidisciplinary, tailored care for these individuals.

Additionally, repeated genital examinations have been identified as a factor in reduced quality of life and dissatisfaction. A recent literature review was conducted pertaining to childhood sexual assault (CSA) in order to better quantify potential psychological harm from repeated genital examinations in individuals with DSD. Multiple recommendations were made including: giving information to parents and subjects prior to medical appointments, encouraging parents to discuss the examination with their children prior to the appointment, training physicians to provide reassurance about normal aspects of the examination while practising full disclosure, making efforts to reduce stigma, empowering youth to have some control of aspects of the examination and limiting the number of medical professionals present. The authors identified management of DSD as an area requiring further research, particularly in regards to anxiety and distress related to the genital examination [34].

### **Holistic management**

Ideally a multidisciplinary team should be involved in caring for DSD individuals and their families consisting of; an endocrinologist, urologist, gynaecologist, psychologist, biochemist, geneticists and social workers with access to ethical advice and specialist investigations [9,35]. However, given the rarity of these disorders and subsequent lack in specialists with adequate knowledge and skills, access to multidisciplinary care can be difficult. It has been suggested that official 'Centres of Excellence' be established to care for DSD individuals and families to mitigate this, however internationally limited public health care and finances limits this strategy [3,36]. Finally, given the heterogeneity of the disorders, care must be tailored to the individual [37].

### **Sociocultural factors**

Gender assignment or 'sex of rearing' choices are strongly affected by individuals' cultural contexts, with religious and traditional beliefs impacting parents' decisions. In cultures where males achieve privileged financial and social position, there may be increased pressure to raise children as male [38]. Additionally, concerns about infertility and potential social stigma may also affect decision making. There have been limited studies examining these sociocultural factors in DSD individuals internationally, the majority of current research data comes from Caucasia as patients from Europe and America [39].

Are view of subjects with CAH from developing countries noted additional challenges regarding quality of life including poverty, ignorance, illiteracy, lack of medical access, lack of medical specialists and psychological services [40]. A recent study conducted psychologist led interviews of 205 Indian children with DSD and their parents and uncovered a myriad of issues including: high maternal distress as many mothers felt the condition had been transmitted through them, late or misdiagnosis, significant discrimination from medical and paramedical staff, inadequate information provided to parent pre-operatively and parental preference to raise children as male due to social advantages in a patriarchal society [41]. Higher rates of anxiety and depression were also reported in Indonesian



adult patients who received late diagnoses [42]. A prospective study of 40 Nigerian subjects predominantly residing in rural communities uncovered similar social issues and barriers to care, with 4 individuals attempting suicide due to social stigma and lack of acceptance [43].

### Future practise

This review highlights the need for clear communication, accurate diagnosis and multidisciplinary care to mitigate psychological harm. Development of hospital or health-network based guidelines for assessment of newborns with ambiguous genitalia would be effective in educating staff and ensuring parents receive accurate information early on. Streamlining referral pathways to tertiary centres which can act as 'centres of excellence' where possible, would be beneficial. Finally, utilising validated quality of life measures and systematic, regular screening of psychosocial outcomes will help identify targeted areas to improve psychological status [44]. Further research and education will be necessary to improve standards of care in the developed, and particularly the developing world.

### Conclusion

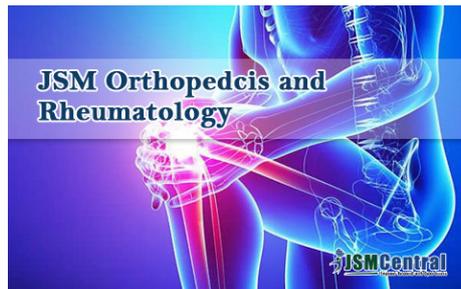
While DSD are rare, they certainly have significant impact upon individuals and their families' due to their sensitive nature and stigma surrounding gender identity and gender roles. Parental and patient mental health and quality of life are often adversely affected by these diagnoses. Sociocultural factors further affect those individuals who reside in developing countries with limited access to healthcare. Educating medical and paramedical staff is crucial to ensure adequate early assessment, diagnosis, counselling, and where appropriate management. Caution should be exercised in early genital surgery to ease parental distress; rather this should be a team-based decision at an appropriate age where the individual can be involved in the consent process. Psychological input should be specialised and accessible. Further research is necessary to improve standards of care.

### References

1. Houk CP, Hughes IA, Ahmed SF, Lee PA. Summary of consensus statement on intersex disorders and their management. *Pediatrics*. 2006; 118: 753-757.
2. Kim KS, Kim J. Disorders of sex development. *Korean J Urol*. 2012; 53: 1-8.
3. Gomez-Lobo V. Multidisciplinary care for individuals with disorders of sex development. *Curr Opin Obstet Gynecol*. 2014; 26: 366-371.
4. Wang C, Tian Q. The Investigation of Quality of Life in 87 Chinese Patients with Disorders of Sex Development. *BioMed Research International*. 2015.
5. Ernst MM, Liao L-M, Baratz AB, Sandberg DE. Disorders of sex development/intersex: gaps in psychosocial care for children. *Pediatrics*. 2018; 142: e20174045.
6. Suorsa KI, Mullins AJ, Tackett AP, Reyes KJS, Austin P, Baskin L, et al. Characterizing Early Psychosocial Functioning of Parents of Children with Moderate to Severe Genital Ambiguity due to Disorders of Sex Development. *J Urol*. 2015; 194: 1737.
7. Pasterski V, Mastroyannopoulou K, Wright D, Zucker KJ, Hughes IA. Predictors of Posttraumatic Stress in Parents of Children Diagnosed with a Disorder of Sex Development. *Arch Sex Behav*. 2014; 43: 369.
8. Wolfe-Christensen C, Wisniewski AB, Mullins AJ, Reyes KJ, Austin P, Baskin L, et al. Changes in levels of parental distress after their child with atypical genitalia undergoes genitoplasty. *J Pediatr Urol*. 2017; 13: 32.
9. Mendonca BB. Gender assignment in patients with disorder of sex development. *Curr Opin Endocrinol Diabetes Obes*. 2014; 21: 511-514.
10. Magritte E. Working together in placing the long term interests of the child at the heart of the DSD evaluation. *J Pediatr Urol*. 2012; 8: 571-575.
11. Leidolf EM, Curran M, Scout, Bradford J. Intersex mental health and social support options in pediatric endocrinology training programs. *J Homosex*. 2008; 54: 233-242.
12. Creighton S, Minto C. Managing intersex. *BMJ: British Medical Journal*. 2001; 323: 1264.
13. Viau-Colindres J, Axelrad M, Karaviti LP. Bringing Back the Term "Intersex". *Pediatrics*. 2017; 140: e20170505.
14. Cresti M, Nave E, Lala R. Intersexual births: The epistemology of sex and ethics of sex assignment. *J Bioeth Inq*. 2018; 15: 557-568.
15. Dickens BM. Management of intersex newborns: Legal and ethical developments. *Int J Gynaecol Obstet*. 2018; 143: 255-259.
16. Karkazis K. Disorders of sex development: Improving care for affected persons and their families. *Am J Psychiatry*. 2008; 165: 265-266.
17. Schober JM, Jamieson MA. Early feminizing genitoplasty or watchful waiting. *Journal of pediatric and adolescent gynecology*. 1998; 11: 154-156.
18. Liao LM, Wood D, Creighton SM. Parental choice on normalising cosmetic genital surgery. *BMJ*. 2015; 351.
19. Slijper FM, Drop SL, Molenaar JC, de Muinck Keizer-Schrama SM. Long-term psychological evaluation of intersex children. *Arch Sex Behav*. 1998. 27: 125-144.
20. Carpenter M. The "normalization" of intersex bodies and "othering" of intersex identities in Australia. Springer; 2018.
21. Ellens RE, Bakula DM, Mullins AJ, Reyes KJS, Austin P, Baskin L, et al. Psychological adjustment of parents of children born with atypical genitalia 1 year after genitoplasty. *J Urol*. 2017; 198: 914-920.
22. Bennecke E, Thyen U, Grüters A, Lux A, Köhler B. Health-related quality of life and psychological well-being in adults with differences/disorders of sex development. *Clin Endocrinol (Oxf)*. 2017; 86: 634-643.
23. Selveindran NM, Zakaria SZS, Jalaludin MY, Rasat R. Quality of life in children with disorders of sex development. *Hormone Research in Paediatrics*. 2017; 88: 324-330.
24. Alpern AN, Gardner M, Kogan B, Sandberg DE, Quittner AL. Development of Health-Related Quality of Life Instruments for Young Children With Disorders of Sex Development (DSD) and Their Parents. *J Pediatr Psychol*. 2017; 42: 544.
25. Amaral RC, Inacio M, Brito VN, Bachega TA, Domenice S, Arnhold IJ, et al. Quality of life of patients with 46, XX and 46, XY disorders of sex development. *Clin Endocrinol (Oxf)*. 2015; 82: 159-164.
26. Sani AM, Soh KL, Ismail IA, Arshad MM, Mungadi IA, Yau SL, et al. Experiences of people living with disorders of sex development and



- sex reassignment: Meta-ethnography of qualitative studies. *J Adv Nurs*. 2019; 75: 277-290.
27. Fagerholm R, Mattila AK, Roine RP, Sintonen H, Taskinen S. Mental health and quality of life after feminizing genitoplasty. *J Pediatr Surg*. 2012; 47: 747-751.
28. Schober JM. Quality-of-life studies in patients with ambiguous genitalia. *World Journal of Urology*. 1999; 17: 249-252.
29. Özbaran B, Özen S, Gökşen D, Korkmaz Ö, Onay H, Özkınay F, et al. Psychiatric Approaches for Disorders of Sex Development: Experience of a Multidisciplinary Team. *J Clin Res Pediatr Endocrinol*. 2013; 5: 229.
30. Schützmann K, Brinkmann L, Schacht M, Richter-Appelt H. Psychological Distress, Self-Harming Behavior, and Suicidal Tendencies in Adults with Disorders of Sex Development. *Arch Sex Behav*. 2009; 38: 16-33.
31. Jürgensen M, Lux A, Wien SB, Kleinemeier E, Hiort O, Thyen U. Health-related quality of life in children with disorders of sex development (DSD). *Eur J Pediatr*. 2014; 173: 893-903.
32. Schweizer K, Brunner F, Gedrose B, Handford C, Richter-Appelt H. Coping with diverse sex development: treatment experiences and psychosocial support during childhood and adolescence and adult well-being. *J Pediatr Psychol*. 2016; 42: 504-519.
33. Rapp M, Mueller-Godeffroy E, Lee P, Roehle R, Kreukels BP, Köhler B, et al. Multicentre cross-sectional clinical evaluation study about quality of life in adults with disorders/differences of sex development (DSD) compared to country specific reference populations (dsd-LIFE). *Health and Quality of Life Outcomes*. 2018; 16: 54.
34. Tishelman AC. Disorders of Sex Development: Pediatric Psychology and the Genital Exam. *J Pediatr Psychol*. 2016; 1: 14.
35. Brain CE, Creighton SM, Mushtaq I, Carmichael PA, Barnicoat A, Honour JW, et al. Holistic management of DSD. *Best Pract Res Clin Endocrinol Metab*. 2010; 24: 335-354.
36. Thyen U, Ittermann T, Flessa S, Muehlan H, Birnbaum W, Rapp M, et al. Quality of health care in adolescents and adults with disorders/differences of sex development (DSD) in six European countries (dsd-LIFE). *BMC Health Serv Res*. 2018; 18.
37. de Neve-Enthoven NG, Callens N, van Kuyk M, van Kuppenveld JH, Drop SL, Cohen-Kettenis PT. Psychosocial well-being in Dutch adults with disorders of sex development. *J Psychosom Res*. 2016; 83.
38. Abdullah M, Katugampola M, Al-Habib S, Al-Jurayyan N. Ambiguous genitalia: medical, socio-cultural and religious factors affecting management in Saudi Arabia. *Ann Trop Paediatr*. 1991; 11: 343-348.
39. Bakula DM, Mullins AJ, Sharkey CM, Wolfe-Christensen C, Mullins LL, Wisniewski AB. Gender identity outcomes in children with disorders/differences of sex development: Predictive factors. *Semin Perinatol*. 2017; 41: 214-217.
40. Zainuddin AA, Grover SR, Shamsuddin K, Mahdy ZA. Research on Quality of Life in Female Patients with Congenital Adrenal Hyperplasia and Issues in Developing Nations. *J Pediatr Adolesc Gynecol*. 2013; 26: 296-304.
41. Joseph AA, Kulshreshtha B, Shabir I, Marumudi E, George TS, Sagar R, et al. Gender issues and related social stigma affecting patients with a disorder of sex development in India. *Arch Sex Behav*. 2017; 46: 361-367.
42. Ediati A, Faradz SM, Juniarto AZ, van der Ende J, Drop SL, Dessens AB. Emotional and behavioral problems in late-identified Indonesian patients with disorders of sex development. *J Psychosom Res*. 2015; 79: 76-84.
43. Osifo OD, Amusan TI. Female children with ambiguous genitalia in awareness-poor subregion. *Afr J Reprod Health*. 2009; 13: 129-136.
44. Sandberg DE, Gardner M, Callens N, Mazur T, DSD-TRN Psychosocial Workgroup tDTAAN, Alliance A, editors. Interdisciplinary care in disorders/differences of sex development (DSD): The psychosocial component of the DSD - Translational research network. *American Journal of Medical Genetics Part C: Seminars in Medical Genetics*. 2017. Wiley Online Library.



Our motto is to advance scientific excellence by promoting open access. We are committed in the widest possible dissemination of research and uplift future innovation



[Submit Manuscript](#)