

Sociocultural Aspects of Disorders of Sex Development

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Disorders of sex development (DSD) is a congenital condition in which the development of chromosomes, gonads, hormones, and reproductive structures are atypical. DSD brings with it a psychological impact on the affected individual and their families. The consensus statement on management of DSD strongly advised an integrated and multidisciplinary approach in providing care to the affected individuals. Studies have been conducted focusing on medical intervention, and more recently, there is increasing attention paid to psychological aspects of DSD. However, studies reporting cultural aspects of DSD are lacking. This review provides an overview on how culture impacts the affected individuals in coping with DSD and making decisions with regard to gender assignment or reassignment,

help-seeking behavior for medical treatments, attitudes toward medical treatment, religious beliefs, and values concerning marriage and fertility. The involvement of social scientists is needed to study sociocultural aspects of DSD from more diverse cultures, to help affected individuals and their families in gaining better social acceptance.

Birth Defects Research (Part C) 108:380–383, 2016.
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Key words: culture; socio-cultural; genital anomalies; disorders of sex development; stigma

Introduction

The management practice of patients with a disorder of sex development (DSD) in Western countries differs greatly from those approaches applied in Asian countries (Warne and Bhatia, 2006). Late presentation, delayed identification, lack of diagnostic facilities and treatment options, and lack of expertise have been reported as major challenges in the management of DSD in poor-resource countries (Armstrong et al., 2006; Warne and Bhatia, 2006; Warne and Raza, 2008; Özbey and Etker, 2013; Zainuddin et al., 2013). It was argued that poverty and cultural aspects influence the management practices between Western and non-Western countries differently (Warne and Bhatia, 2006). Poverty makes health-care facilities not only less available, but if available also less accessible for poor patients. Cultural background and social context both influence the patients', as well as health-care professionals' cognition about illness and their decisions in dealing with it. In poor-resource countries of Asia, it is common that children born with ambiguous genitals grow up with their original anatomy, due to unaffordable medical treatment or struggles for cultural acceptance (Armstrong et al., 2006; Warne and Bhatia, 2006).

This article was published online on 29 December 2016. An error was subsequently identified. This notice is included in the online and print versions to indicate that both have been corrected 6 January 2017.

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Published online in Wiley Online Library (wileyonlinelibrary.com). Doi: 10.1002/bdrc.21144

DSD at a Glance

DSD is a group of congenital conditions which include atypical conditions of chromosomal, gonadal, and phenotypical sex (Hughes et al., 2006). The incidence of DSD has been estimated to be approximately 1 in 4500–5500 without distinguishing the specific type of DSDs (Lee et al., 2016). In 2006, the LWPES/ESPE (Lawson Wilkins Pediatric Endocrine Society/European Society for Paediatric Endocrinology) consensus group proposed the classification of DSD and divided it into three groups: (1) Sex Chromosome DSD; (2) 46,XY DSD; and (3) 46,XX DSD (Hughes et al., 2006).

Sex chromosome DSD consists of 45,X Turner and variants, 47,XXY Klinefelter and variants, 45X/46XY MGD, and chromosomal ovotesticular DSD (46XX/46XY chimeric type or mosaic type). In this type of DSD, there is abnormal gonadal development, due to numerical sex chromosome abnormalities (Hughes et al., 2006; Houk and Lee, 2008).

The most common form of 46 XX DSD is congenital adrenal hyperplasia (CAH), particularly 21-hydroxylase deficiency. CAH is the autosomal recessive inheritance that impairs normal steroidogenesis that leads to cortisol deficiencies and an excess of adrenal androgens (Speiser et al., 2010; Nimkarn et al., 2011). The incidence of CAH has been estimated to be 1:10,000–1:15,000 live births with approximately 75% being salt wasters (Webb and Krone, 2015). Besides the problem of ambiguous genitalia in female CAH, this condition was potentially lethal, and was related to increased mortality due to adrenal crises (Falhammar et al., 2014).

The 46 XY DSD include disorders of gonadal development and disorders in androgen synthesis or action with the commonest form being Androgen Insensitivity Syndrome (Hughes et al., 2006; Ahmed et al., 2013).

Although many individuals with DSD are recognized in the newborn, the clinical manifestations sometimes occur later, presenting in children, adolescents, or adults as progressive clitoromegaly, inguinal, or labial mass in a phenotypic female, delayed puberty, progressive virilization in

girls, and clinical hematuria in phenotypic boys (Lee et al., 2016). Individuals with DSD will encounter different problems across developmental stages. A multidisciplinary team of DSD specialists would be ideal to provide the different approaches required in helping children, adolescents, and adults with DSD.

Gender Assignment or Reassignment

The birth of a baby with ambiguous genitalia can be stressful for parents because often gender assignment is delayed and parents have to cope with an awkward moment when they cannot answer the most common question “Is your baby a boy or a girl?” In a village setting, newborns with ambiguous genitalia often cannot avoid being identified with DSD (Armstrong et al., 2006; Warne and Raza, 2008). As ambiguous genitalia are very rare and DSD is not known widely, the birth of a newborn with ambiguous genitalia can shock neighbors and family members, and the news can be spread throughout the village. Soon both the baby and the affected family become objects of usually unwanted social attention (Armstrong et al., 2006; Warne and Raza, 2008).

Being a member of society is part of an individual’s personal identity, and so is sex and gender. Being neither male nor female is uncommon in some societies, like in India, Vietnam, and Indonesia. There are several local terms assigned to people with DSD. Like “*waria*, *banci*, *wadam*” (Indonesian terms for male transvestite) or “*bencong* or *wandu*” (Indonesian terms for male transvestite), *hijra* (in Indian), and *ai nam ai nu* (in Vietnam) (Kuhnle and Krahl, 2002; Warne and Raza, 2008; Ediati, 2014). None of these terms are respectful. Being labeled with these terms comes with a strong message of social exclusion, which is painful, as reported by affected individuals who experienced different forms of social stigmatization due to their DSD condition. It is obvious that there is a lack of proper knowledge about DSD in many societies. As a consequence, patients with visible DSD features (i.e., a girl with a virilized body, ambiguous genitalia in the newborn) experience stressful situations due to social stigmatization.

Gender assignment or reassignment could be complicated when religious beliefs and preference for a son in the family is taken into account. These complicated problems were described thoroughly in Egyptian and Malaysian cultures (Dessouky, 2001; Kuhnle and Krahl, 2002; Zainuddin et al., 2013). Parents of children with DSD preferred a male gender assignment for their children (if that was possible) because of the social advantages of growing up male in a patriarchal society (Kulkarni et al., 2009; Gupta et al., 2010; Chowdhury et al., 2015; Ediati et al., 2015b; Joseph et al., 2016).

Social Cognition About DSD

The ethical principles proposed by Wiesemann et al. (2010) highlight the importance of endorsing good relationships

between the affected children/adolescents and their family members, as well as building supportive peer relationships for the affected children/adolescents. Thus, care for the affected youths with DSD should not be limited to providing medical care for the affected individual, but also helping them to adjust to their social world. A good relationship among family members will support affected youths and their mothers, particularly because genetic inheritance of DSD (i.e., CAH) is often being misinterpreted as the responsibility of mothers who are blamed for the DSD outcome (Joseph et al., 2016). Moreover, lack of knowledge about DSD in a society leads to affected individuals experiencing considerable social discrimination, not only from relatives and friends, but also from medical and paramedical staff in hospitals (Joseph et al., 2016).

Help-Seeking Behavior and Attitude Toward Medical Treatment

Culture does influence management of DSD (Kuhnle and Krahl, 2002; Warne and Raza, 2008). In societies where discussing sexuality is considered taboo, especially when DSD is not known, it is difficult for individuals with DSD to express their feeling about anomalies in their bodies or discussing problems related to dissatisfaction with sexual activities among married couples with DSD. Even for health practitioners who have no experience or knowledge related to ambiguous genitalia, discussing this topic might feel uncomfortable.

In the case of genetically females with CAH who live without treatment, a virilized body triggers negative social reactions which lead the affected individuals to withdraw from social relationships (Ediati, 2014) or decide not to seek medical treatment. Lack of knowledge about DSD and an unsupportive society play important roles in influencing individuals with DSD to seek medical treatment early in life. In a case where parents recognized ambiguous genitalia as a social emergency, referral to a specialized medical center does not happen, because the medical practitioners or the patients and their families are unaware of such centers. Consequently, individuals with DSD live in an ambiguous body and gender without sufficient or proper information and treatment for their conditions (Ediati et al., 2015a). In contrast, in Western countries, most patients with DSD are identified and treated early in life, and medical care for DSD is available and accessible for patients and the families.

Sexuality, Marriage, and Infertility

Marriage and infertility are major concerns for parents of youngsters with DSD, as well as adults with DSD. In procreation societies, marriage is a precondition for becoming a fully respected member of society and parents have a great responsibility to initiate their children’s marriage. The newlywed couples are expected to have children soon after being married. It is common that friends, relatives, or neighbors ask

them for signs of pregnancy to show their caring, whereas in Western society this practice would be considered rude or disrespectful to one's privacy. Social consequences are far more severe for infertile women than for infertile men (Ediati et al., 2013). Perhaps it is easier for men than for women to hide their infertility from society. Being unmarried, particularly for a woman, is considered bringing shame to the family or bringing more disadvantages for women, as reported in the Indian, Egyptian, Sudan, and Indonesian societies (Gupta et al., 2010; Abdullah et al., 2012; Ediati et al., 2013; Özbey and Etker, 2013). In India, 55.2% patients with DSD do not consider marriage due to fear of rejection. For example, a male with DSD would have a small phallus and could be infertile. In contrast, 85% of parents expect their children to get married. In dealing with society, although 85% of affected individuals reported they have good relationships with their family members, they also reported that they did not feel that they fit into society because of their intense distrust of the society. Study from Indonesia reported that parents and affected children keep DSD a secret (Gupta et al., 2010).

Social Stigma and Social System

In a collective-driven society, people have been taught to follow social norms. Individuals behave in accordance to what others think about it. The tendency to seek social acceptance is stronger in people who live in a collective-driven society. Individuals with DSD, particularly those who had visible features of DSD (for example, a virilized face or body and low voices in females with CAH) are prone to experience social stigmatization. Fear of being teased, rejected by peers or partners, and in most females, fear of being divorced because of their infertility, are reported among parents and affected individuals with DSD (Ediati, 2014). Avoiding social relationships, delays in entering romantic relationships, and leaving their DSD condition undisclosed are common ways of coping with having a DSD condition in the family (Ediati et al., 2013).

As society only accepts binary gender categories, the option of having a third gender is not popular, particularly in a society which applies religious values strictly. For example, in Moslem society, it is important to distinguish and separate male and female in clothing, prayer, and other religious practices, and in education as well as social setting. Individuals with DSD, and often their parents as well, encounter stigmatizing situations in all social contexts of everyday life: family, peers, colleagues, strangers, and the media (Ediati, 2014; Wisniewski and Sandberg, 2015; Meyer-Bahlburg et al., 2016). To anticipate social stigmatization, parents of newborns with genital anomalies often develop protective instincts, which lead them to make decisions for early genital surgeries for their babies (Sanders et al., 2008, 2012). Living with a DSD is hard and even harder for patients who live in a society which neither understands nor supports patients with a DSD. In response to this situation, the involvement of

social scientists and social workers are needed to develop a social system or network that endorses social acceptance and serves as the source of social support for the affected individual and their families. Educating people about DSD is a way to raise awareness and social support for individuals with DSD and their families.

Summary and Recommendation

As patients with DSD interact with their society, there is considerable impact of the society on the patients' problems and experiences. In a collective-driven society, the society exerts more power than individuals, in the sense that they contribute to people's cognitions, attitudes, and behaviors (Hofstede, 2011). In a culture that emphasizes social bonds, particularly in a village (Armstrong et al., 2006), people know each other very well and share their concern about everything. What other people will say or do in response to DSD is of major concern for the affected individuals and their families. Therefore, it is not surprising that problems with social and emotional functioning are evident among individuals with DSD (Ediati et al., 2015a).

Living in a society where DSD is poorly understood and unaccepted is problematic. For the affected families, it is like living in a crisis across the life span (Santos and Araujo, 2008). At this point, it is crucial to develop a social intervention to endorse social acceptance for individuals with DSD and their families. For this reason, it is important to provide educational materials in local languages, tailored to the local cultures. Sex, gender, and sexuality are social constructions that influence the ways people think and interact with others (Bhavsar and Bhugra, 2013). In this digital era, where information can be accessed from anywhere at any time, the borders between cultures are getting blurred. With ease of mobility around the world, marriage across cultures or nationalities are common now. With the help of information technology, e-learning platforms for DSD specialists are now available to help health practitioners around the world to consult with other DSD specialists so that they can give better advice and care for patients with DSD and their families (Drop et al., 2012; Muscarella et al., 2014). These learning platforms, which currently focus on the medical aspects of DSD, would be greatly enhanced by including educational materials related to coping with sociocultural aspects of DSD.

Acknowledgment

We received no funding. We declare that we have no conflicts of interest.

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