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Gender identity: A psychosocial primer for providing care to patients with a Disorder/difference of Sex Development and their families [Individualized Care for Patients with Intersex (Disorders/Differences of Sex Development): Part 2]

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Summary

Introduction/background: Many parents of infants born with a DSD describe the process of initial sex assignment at birth as highly stressful. Parents of children with a DSD also note high distress when their children engage in behaviors that are not considered typical for their gender.

Objective: The goal of this article is to provide members of the health care team a brief overview of psychosocial facets of gender and gender identity particularly relevant to DSD for the purposes of enhancing shared decision-making and optimizing support for individuals with a DSD and their families.

Discussion: Gender identity is a multidimensional construct involving related but distinct concepts such as gender typicality, gender contentedness and felt pressure for gender differentiation, and can be assessed via standardized measures. Gender dysphoria is associated with poor psychological adjustment, and is mitigated by family and peer support. Family influences on gender identity include parental modeling of gender behavior and family composition (e.g., same-sex children vs both sons and daughters in a family). Cultural factors that may influence sex assignment include societal views on gender, and gender-related differential resource allocation within a society. In addition, religious beliefs and the presence of a “third-sex” category within a culture may also influence parental gender ideology.

Clinical application: Health care providers who work with patients with a DSD must have a strong grasp on the construct of gender identity, and must be able to clearly and consistently

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communicate with patients and families about gender beliefs in order to optimize family support and gender-related decisions.

Keywords

Gender identity; Disorder of Sex Development; Cultural; Intersex

For the vast majority of humans, being labeled a boy vs girl at birth is straightforward, based on genital anatomy, and is experienced as “obvious” - without need for a “choice”. Similarly, for many diagnoses that fall within the Disorders/Differences of Sex Development (DSD) umbrella, clear recommendations for male versus female assignment can be made on the basis of outcome data, even when genital anatomy is atypical. However, for some DSD diagnoses, recommended sex assignment at birth¹ is less clear, and a “choice” must be made in terms of which gender label (boy vs. girl) a child is to be assigned [1]. Parents of children with a DSD identify uncertainty about initial gender assignment as very stressful; as children grow older, parents also experience considerable distress if their child displays behaviors they deem typical of the “other” gender [2, 3]. Parents also note high stress related to reconciling what they perceive to be conflicting information about their child’s DSD [2]. To promote optimal decision making and child/family adjustment, both the health care team and families must have a shared, accurate understanding of both biological sex features and gender identity, as well as how they differ. [4]. To that end, this article provides 1) a concise overview of the construct of gender identity, 2) proposes terminology and definitions to promote consistent and accurate communication (see Table 1), and 3) reviews psychosocial contextual factors to enhance conversations about gender with families.

Gender identity review

DSD refers to conditions in which the *biological sex features* that typically distinguish males from females are discordant or have developed atypically. Biological sex features are usually consistent with a person’s *gender identity* [i.e. their sense of themselves as a boy/man or girl/woman (if binary)]. In other words, most children with a penis (biological sex feature) think of themselves as a boy (gender identity).

Gender identity is a “lived experience” – only a person themselves can know what their gender identity is. Thus, the “boy” or “girl” label given prenatally or at birth (and celebrated at gender reveal parties), does not actually reflect a gender identity – it is the “best guess” (and usually a really good guess, based typically on genital appearance) that parents and health care providers have as to a person’s future gender identity. The formation of gender identity is the result of complex interactions between biological and social factors, and relies on cognitive development [5, 6]; by age 2 years children understand gender labels, by age 3 years children typically will label themselves a particular gender (“I am a girl!”), and by age 4 years the gender label is typically being expressed consistently. Gender atypical behavior is often first recognized in early childhood, and can range from interests/activities culturally atypical for a particular gender (e.g., a boy wearing a dress) to expressed desire to be a

¹Both *sex assigned at birth* and *gender assigned at birth* are terms that have been used within DSD. Most recently, *sex assigned at birth* has been favored; this recognizes that gender identity cannot actually be assigned by others.

different gender or insistence that one is actually a different gender [7]. The best predictor of gender identity is sex assignment at birth.

In many societies, there has been a paradigm shift in gender identity expression in recent years. In the US, 15 states offer non-binary ID options, and legislation has recently been proposed that adds a 3rd gender option to US passports [8]. 35% of youth categorized as “Gen Z” report personally knowing someone who uses gender-neutral pronouns, and nearly 60% support the availability of non-binary gender options on form or online profiles. Of note, these percentages are higher compared with older generations; e.g., for Millennials, 25% reported knowing someone who uses gender-neutral pronouns, and 50% supported non-binary options on forms/online profiles [9]. Thus, increased exposure to and acceptance of a range of gender identity expression may continue into future generations.

Gender identity has been conceptualized as a multidimensional construct including aspects such as *gender typicality* (whether behaviors/interests conform to gender stereotypes), *gender contentedness* (the degree to which a person feels glad to be their gender), and *felt pressure for gender differentiation* (i.e., pressure to conform to stereotypes)[6]. These concepts are distinct, albeit related constructs [10], and are important to distinguish for patients and, in particular, families. In DSD, children may express a high level of *gender atypicality*, while at the same time having high *gender contentedness* (e.g. a child with CAH who strongly aligns with a female gender identity but likes to do more boy-typical activities). The multi-dimensional model of gender identity has recently been extended to include the potential for determining the degree to which an individual feels similar or dissimilar to both genders [6]. Both gender contentedness and gender typicality have been shown to be related to positive adjustment for children; felt pressure for gender differentiation is noted to have a particularly negative impact on adjustment, particularly in the context of gender discontentment or atypicality. Several measures assessing children’s gender identity have been developed, including the Parent-report Gender Identity Questionnaire for Children (for children 2.5 to 12 years of age) [11], the Gender Identity Interview for Children (for children 2.5 to 12 years)[12], and the Multidimensional Gender Identity Scale (for children 8 years of age and older) [13].

Careful consideration of sex assignment based on prediction of future gender identity is important, but it must be realized that it is impossible to predict with certainty so parents must be prepared to respond when gender identity does not develop as assigned. Ideally this can be recognized before gender dysphoria occurs. The rate of gender dysphoria is higher in several DSD diagnoses compared with the general population [14]. Gender dysphoria is diagnosed when there is 1) “marked incongruence between one’s experienced/expressed gender and assigned gender, of at least 6 months’ duration”, and 2) this incongruence is associated with clinically significant distress [15]. In prospective studies of children with gender dysphoria in the general population (i.e., non-DSD samples), gender dysphoria remits in the majority of children [16]. Youth with gender dysphoria present with high rates of anxiety, depression and suicidal ideation [17]. Youth with non-binary gender identities (presumably initially raised with a binary gender assignment) also demonstrate poor mental health outcomes [18]. “Not choosing” a gender until the child declares their own gender identity is a path recently taken by some families [19]– it is unclear what the psychosocial

ramifications of this approach will be given the rarity of the experience. Protective factors that mitigate the risk conferred by gender variance include healthy self-esteem and positive relationships with parents and peers [20].

Consideration of gender must consider the family influences on gender identity [21]. In addition to genetic transmission of biological factors from parent to child, the family provides formative gender-related experiences to a child. For example, parents provide a model for gender typical behavior (through their own interests and activities), create opportunities for gendered behavior through their control of the environment, shape the gendered behavior of their children through attention and consequences, and verbally share their own beliefs about gender with their children. Siblings may also play an important influential role in gender identity development and gender-related parenting (e.g., less gender-stereotypical parenting in families with same-sex siblings vs both sons and daughters). And, as noted previously, family acceptance and support of gender variance (when it is present) appears to be important for positive adjustment [20]. More broadly, parental self-efficacy (a parent's belief that they can influence their child toward successful outcomes) and parental coping both positively predict better child adjustment [22].

Other cultural factors that contextualize sex assignment (both initial assignment and openness to change) include societal views on gender, which can change over time [23]. In most societies, expressed gender plays a role in resource allocation, including income and inheritance. Gender also often impacts division of labor, rules of conduct, educational attainment, health, political empowerment, and initiation of and freedom in relationship building (particularly, marriage) [24, 25]. The influence of gender on these arenas likely depends on a number of factors, e.g. the rigidity of the gender binary in a society; societal variations in these gendered facets impact the gender ideologies that parents and health care providers bring to the discussion of gender assignment. For example, Joseph and colleagues [26] observed family preference for male sex assignment in their case series of DSD in India, interpreting this preference in light of the social advantages male gender confers in that society, and difficulty in arranging marriage for infertile girls. It is also important to understand whether there is an understood "third" (or more) gender category in the family's culture – while binary sex is the dominant sex categorization across societies, many cultures have specific terms recognizing people who identify outside the binary (e.g., Indian "hijras", Hawaiian and Tahitian "m h ", Samoan "fa'afafine", Indigenous North American "two-spirits"). Families from these cultures may be interpreting information related to their child's DSD through the lens of their cultural experience of "intersex." These culturally recognized categories differ widely in the roles and social status that they hold in their respective societies, and are often quite stigmatizing; the assumptions that families from these cultures bring into the DSD context must be voiced and understood [26–28]. Even within one nation, geographical differences in gender ideology may exist. For example, in a recent large US study, gender diverse youth in rural settings had higher rates of gender-related harassment relative to youth in urban settings (50.6% vs 41.4%, respectively). Of note, the positive influence of protective factors such as supportive families and teachers was similar across geographical locations [29].

Religious background also may posit a strong influence on a family's perspective related to sex assignment and change. In some faith traditions, gender influences access to spiritual activities, and some religions have very strong negative views on sex assignment change [24, 25, 30]. Furthermore, some religions include systems of law that will influence sex reassignment. For example, families from Islamic countries may be influenced by their awareness of *fatwas* (formal legal opinions from experts in Islamic law) relating to sex assignment and reassignment [31].

Clinical implications

Every attempt should be made to determine a specific etiologic diagnosis that, when possible, is verified by molecular genetics, evidence of fetal androgen exposure, and extent of internal and external reproductive system virilization. Even then, the fact remains that there are variations and complexities even within a specific diagnosis. When there are sufficient diagnostic information and outcome data to guide sex assignment at birth, health care providers must provide these data to family and advocate strongly for a sex assignment that is most likely to align with future gender identity. However, the limitations of outcome data must be acknowledged. And, for any specific child and family, psychosocial contextual factors may be so distinctive that consideration of an alternative sex assignment at birth may be warranted. Families deserve to have 1) complete and accurate counseling of sex versus gender, 2) an understanding of the gender identity outcomes relevant to their child's DSD, and 3) their personal and cultural gender ideologies understood by the health care team. It is also critical that medical teams use language that is understandable to and respectful of individuals and families, and to seek clarity on what families actually mean when using their terminology [32]. From this mutual understanding, clear and consistent communication related to gender concerns can enhance decision making and optimize quality of life outcomes individuals and families.

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This is the 2nd article in a series regarding care of children born with a DSD for whom gender assignment at birth is a primary consideration due to ambiguous genitalia. This article provides a concise overview of some of the important psychosocial facets of gender and gender identity. Our goal in presenting this information is for affected individuals, their families, and multidisciplinary care providers to develop a shared and accurate language related to gender identity, and for health care teams to have enhanced knowledge in order to provide culturally sensitive family-centered care.

Table 1:

Gender terms

Broad term	Features	Examples	Notes
Biological "sex"	Chromosomes	46,XX 46,XY 46,XX with deletions or redundant sections 46,XY with deletions 47,XXY, etc 47,XXY	Currently, karyotype sex is deemphasized since genetic direction of sexual differentiation is at the genetic level of the autosome as well as sex chromosomes, as illustrated by 46XX males and 46XY females who developed as such by gene expression, independent of sex chromosomes
	Internal anatomy	Gonads, uterus, fallopian tubes, vagina, urethra, vasa deferentia, prostate, ejaculatory ducts	Internal duct development is largely a result of the presence of functioning testes that secrete AMH to inhibit female structures in a 46,XY DSD individual. Internal anatomy can be altered through hormonal or surgical intervention. In some cases, anatomy needs to be altered in order to improve functioning.
	External anatomy (e.g., genitalia)	Labia majora/ scrotum Clitoris/Penis Empty scrotum	The development of the embryonic structures into labia majora or scrotum and a clitoris or penis is dependent upon the amount of testosterone during the first trimester of fetal life. Lack of testes in the scrotum may mean that they are undescended, missing or that gonads are ovaries. External anatomy can be altered through hormonal or surgical intervention. In some cases, anatomy needs to be altered in order to improve functioning. In some cases, surgery is not necessary for function but is chosen by individuals or families solely to change appearance.
	Hormonal activity	Androgens Estrogens Other gonadal hormones	All have specific functions, although androgens hold a major role in both internal and external reproductive system development.
	Gender identity	A sense a person has about themselves as to whether they are a boy/man, girl/woman or some other gender category	Typically consistent with gender assigned at birth and typically expressed consistently by 4 years of age. Gender can be experienced in a binary fashion (e.g. experienced as being a boy fully and completely), or can be more fluid (e.g., experienced as having a sense of being both a boy and a girl). Many people with gender dysphoria or who are transgender will report that their gender identity has actually been consistent since childhood but they did not feel comfortable sharing this information; more recently, there have been cases of "late-onset" gender dysphoria in which there is a dramatic shift in gender identity. In some DSD conditions, gender identity may shift specifically during pubertal development (e.g., 5 α -reductase 2 deficiency)
Gender	Gender role	Behaviors, personality traits and features of appearance that are typically linked with a particular gender	Gender roles are greatly influenced by factors within family and peer systems (e.g., parental preferences, favored peers' activities) as well as the larger sociocultural context (e.g., gender stereotypes). As sociocultural influences change over time, so, too, can gender roles (e.g., the increase in women athletes since Title IX).
	Gender/sex assigned at birth	The "guess" that parents and medical providers make about which gender a child will identify with later in life, typically based on fetal/infant anatomy/physiology	Both phrases are used within DSD: "Sex assigned at birth" is a phrase that is most commonly used within LGBTQ communities, and has been more recently gained favor within DSD.
	Gender dysphoria	Significant distress related to one's gender identity not corresponding with the gender assigned at birth.	In some cases, gender dysphoria resolves by a person making socio-cultural, hormonal or surgical changes that allow them to more fully express themselves and/or be accepted in a way consistent with their gender identity (e.g., as seen in the transgender population); in some instances, gender dysphoria remits over time.
Other	Intersex		Some affected individuals prefer the term "Intersex" rather than "DSD" or a diagnosis specific term (e.g., "CAIS"). "Intersex" can refer to the co-occurrence of biological/somatic features typically associated with males and females (e.g.,

Broad term	Features	Examples	Notes
			a person with 46,XY karyotype who also has a uterus). For other individuals, “intersex” also connotes a psychosocial identity, as does other gender identity categories (e.g., “gender fluid”).

Note: There are numerous terms to describe a myriad of gender categories. These terms are constantly changing. When individuals or families reference a specific gender term, asking for clarification of what the term means to them is advisable. Many websites offer lists of terms and definitions (e.g., <https://www.apa.org/pi/lgbt/programs/safe-supportive/lgbt/key-terms.pdf>).