3 Communicating with Clients Affected by Diverse Sex Development

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I like to listen. I have learned a great deal from listening carefully. Most people never listen.

—Ernest Hemingway

"Is it a boy or a girl?" This is by far the most likely first question ever asked about a human being. With prenatal testing becoming increasingly routine these days, the answer does not even have to wait until the baby arrives. But, what happens when the expected straightforward response to the question is not available? What do the reactions, often characterized by shock and confusion, tell us about some shared assumptions that we have about sex and gender? How are affected individuals and families to make sense of these situations, and how can clinicians discuss them in more helpful ways?

The stability of dimorphic sex categories in our society obscures the fact that all human embryos begin life with a common set of reproductive and genital structures. Sex differentiation typically begins at about 6 weeks of embryonic life, and a sex-undifferentiated fetus gradually assumes the anatomical structures and appearance of what we think of as male or female. The tissues that develop into the testes, penis, and scrotum in males are initially the same as those that develop into the ovaries, womb, vagina, clitoris, and labia in females.

A number of genetic conditions can disrupt regular embryonic sex differentiation and development, so that the outcomes may not clearly correspond to one of the two mutually exclusive physical sex categories recognized by most contemporary societies. That is to say, the usual markers of sex—karyotype, gonads, and genitalia—are not entirely male-typical or female-typical. The term "intersex" is often

used to refer to these developmental outcomes (Hughes 2002). Further back in time, the term "hermaphroditism" prevailed (see Dreger et al. 2005). Numbers depend, of course, on inclusion criteria. Using a very broad definition, the prevalence of live births affected by atypical sex characteristics has been estimated to be as high as 1 in 200 (Blackless et al. 2000).

Diverse sex development is not limited to babies presenting genital ambiguity that renders gender assignment difficult. It is, for example, relatively common for men and boys to be born with the urethral opening at the base or on the shaft of the penis rather than at the tip. More rarely, some men have a single testis, and some have testes that are "vanishing." Some men present for fertility investigations only to be told that they have uterine and ovarian structures and are potentially capable of menstruation. Some girls do not menstruate in their teens. Clinical investigations may identify in some girls "ovarian insufficiency," in other girls healthy ovaries but no womb, cervix, or vagina, and, in still others, abdominal testes and a male-typical chromosomal karyotype. For reasons not well understood, some boys and girls do not go into puberty spontaneously, and some present signs of puberty in early childhood. There is actually no need to look to people with identified genetic variations for atypical sex characteristics. In the general population, for example, many healthy men have breasts and many healthy women do not; furthermore many healthy men have very little facial or body hair and many healthy women have a lot.

Advances in biotechnology have contributed to the identification of an increasing number of genetic links to atypical sex characteristics. A few years ago, an international group of experts revised and standardized the nomenclature, to make future biomedical research more expedient. The result was the first and only international consensus statement.

> An umbrella term, "disorders of sex development," was put forward as a way to denote a broad range of diagnoses; that is, to include all "congenital conditions in which development of chromosomal, gonadal or anatomical sex is atypical" (Hughes, Houk, Lee, Ahmed, & LWPES/ESPE Consensus Group 2006).
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> In parallel with increasing interests in molecular biology, the range of conditions covered by the term is widening to include sex-chromosome aneuploidies that may not be associated with anomalies of the external genitalia or gonadal failure (e.g., XXY or XO mosaicism).

All terms used to describe body differences are potentially pejorative, and all of them can be expected to cause offence. It is unsurprising that "disorders of sex development" has been greeted with mixed reactions (Hughes et al. 2006 [Responses]; Davis, 2011). For the purpose of this chapter, we refer to these same conditions as *diverse sex development* (DSD), to acknowledge the fact that many affected individuals consider their bodies different rather than disordered.

"Sex" is often equated with the biological and is expressed in dimorphic terms, whereas "gender" has been introduced to denote the psychosocial and behavioral. Biological factors are generally believed to contribute to gender identity, but the importance of biology is positioned very differently across disciplines. "Gender" is a concept that is meant to transcend binary categorization. But, just like "sex," it, too, is expressed in dimorphic terms. "Sex" and "gender" are thus often used interchangeably. Our restrictive language is unable to do justice to human diversity in body, identity, and behavior. Experts in gender studies say that it is no accident that we have been strongly taught to assume that body, identity, and sexuality are binary and mutually exclusive when they are best thought of as continuous. These distortions contribute to clinical communication challenges in the DSD field. The majority of people, regardless of age, race, class, religion, disability, and sexuality, take for granted that there are two types of bodies—male and female. The fact that most bodies tend to be more or less male-typical or more or less female-typical is given little thought. It is therefore not surprising that bodies with both male-typical and female-typical features are greeted with surprise.

In recent years, medical management of these bodies has been the subject of intense debates, triggered not least by dissatisfied and aggrieved adult service users. The debates raise profound issues about the purpose of maintaining sex and gender dichotomies in the face of bodies and identities that challenge them; about the ethics of normalizing genital surgery on infants and children with body differences; about medical and parental power; about informed consent; and about how best to assist people caught between the benefits and costs of normative embodiment and resistance to it (Liao and Boyle, 2004). Given such uncertainties, the importance of optimal clinical communication should be obvious.

In this chapter, suggestions for improved clinical communications are offered, with the tacit acknowledgment that these interventions would have limited values without a collective effort to interrogate (hetero)normative embodiment. Our suggestions are informed by the work that we have done with and for people with a range of DSD diagnoses, with clinical examples drawn mainly though not exclusively on women with XY conditions. We believe that our recommendations for sensitive exploration that is founded on continued scrutiny of our own inevitably value-laden perceptions are relevant to a much broader range of clinical situations that challenge people's sense of maleness and femaleness (e.g., consulting to women post-mastectomy). Molecular research of atypical sex characteristics may separate people into categories, but the communication challenges for care providers in different specialties and for differently labeled service users have much in common.

BACKGROUND

Diagnosis of genetic conditions associated with DSD may be made at any age, sometimes along with other body differences or health problems. When the external

genitalia appear ambiguous (e.g., the clitoris is sufficiently enlarged to appear like a small penis, or the labia are sufficiently fused to give a scrotal appearance), diagnosis of the underlying condition is often made in infancy. When the external genitalia look typical but the internal genitalia do not (e.g., absent or small vagina and/or uterus, presence of intra-abdominal testes in a girl), the underlying condition may not be identified until adolescence (e.g., following investigations for primary amenorrhea). In the case of childhood presentation, most individuals would have come under medical management. The pediatric specialists most likely to assume clinical responsibilities are endocrinologists and urologists.

> When genital differences are detected, virtually all children with a known female karyotype are assigned female, and feminizing genital surgery usually ensues; children with a known male karyotype whose penis is absent or very small are also likely to be assigned female (Creighton & Liao 2004).

DSD conditions are too numerous to mention. Therefore, only two of the better known conditions are briefly described here. Androgen insensitivity syndrome (AIS) affects only fetuses with male-typical karyotype (XY). The fetus produces typical amounts of androgens but lacks receptors to respond to them. In its "complete" form (CAIS), the infant typically presents female external genitalia, hence the condition may not be diagnosed until adolescence, usually prompted by absence of menstruation. In its partial form (PAIS), the individual is born with genitalia that are virilized to a greater or less extent. People with AIS do not have ovaries, womb, or cervix, and instead have testicular gonads and a vagina that is shorter than average. Until relatively recently, concealment of the diagnosis from CAIS and other XY females was typical, in the interest of psychological adjustment. Case Study 1 represents this type of scenario in a client with an XY female condition known as pure XY gonadal dysgenesis or Swyer syndrome. Health professionals in ~~genetic~~ may be required not only to help newly diagnosed clients assimilate complex medical information and process its impact on their sense of self, they may also need to take special care to eliminate further communication mishaps in older clients working through the emotional burden of having been deceived (see Box 3.1).

A second, better known DSD diagnosis is congenital adrenal hyperplasia (CAH), a metabolic condition that affects XX and XY babies. In this situation, the fetus is exposed to unusually high levels of prenatal androgens. An XX infant would have a womb, upper vagina, ovaries, and fertility potential and also present partially virilized external genitalia. Fertility potential has primacy in gender assignment. Therefore, most XX babies with CAH are assigned female.

In most cases of ambiguous genitalia, surgery is carried out to "feminize" the genitals, even for children who are known to be genetically male but whose penis is believed to be too small for them to be successfully reared as males. The greater emphasis on female sex assignment has partly been influenced by the lesser technical difficulty of

Box 3.1 Case Study 1

In 2011, a 50-year-old woman contacted the Androgen Insensitivity Syndrome Support Group (AISSG) ~~U.K.~~ in some distress. She had been seen by her family doctor at age 14, when periods had not started, and, at age 17, laparoscopic investigations identified "streak ovaries" (streaks of undeveloped and inactive gonadal tissue typical for Swyer syndrome) and she was told that she could never have children. When training as a nurse, she had bloods taken and was told that she had "an extra male chromosome." The implications were not explained to her (she subsequently found "XY karyotype not disclosed" written in her medical notes). At 21 she had a "gonadectomy," with the same family doctor citing a cancer risk as the reason. She then attended an endocrine clinic once a year and started on hormone replacement therapy (HRT).

The identification of defects of the aorta and near blood vessels in 2010 led her to consult a gynecologist to discuss stopping HRT. This clinician confirmed the diagnosis of Swyer syndrome, which means she has a uterus and might have been able to sustain a pregnancy via in vitro fertilization and egg donation. She has missed out on potential fertility treatment and genetic counseling.

In her extended dialogue with AISSG, she said, "I had pieces of the jigsaw but the lack of a name did mean that I couldn't put it all together." She referred to "the existential crisis I have been in, as to who I now am, with this new info."

constructing genitals that can be penetrated than genitals that can penetrate (Chase, 1998a). Such surgery is usually carried out in the first 2 months of life and no later than 2 years, with the intention of providing secure gender identity and psychological adjustment and of relieving parents of what is assumed to be an intolerable burden of uncertainty (Liao & Boyle 2004). Following gender assignment and surgical sexing of the genitals, it has been traditional practice to recommend secrecy or at least incomplete disclosure to the child or siblings, again in the interest of psychological adjustment.

"Feminizing" surgery is without exception invasive and may include any or all of the following: reduction of the external clitoris (the visible part), refashioning of the labia, or reconstruction of the vagina. Children cannot consent to the elective surgery. Furthermore, research with adults suggests that surgery often has to be repeated, and that, contrary to pediatric claims, appearance and functional and sexual outcomes are unsatisfactory in adults (Creighton, Minto, & Steele 2001; Crouch et al. 2008; Minto, Liao, Conway, & Creighton, 2003). Normalizing genital surgery is avoided in male-assigned babies, because doctors are much less confident about phalloplasties on babies and young children, and tissues are preserved for procedures in adolescence and adulthood.

It may come as a surprise that the availability of (the small number of) designated adult DSD services is a relatively recent provision in the United Kingdom.

Until a decade or so ago, clients discharged from pediatric services were lost to follow-up, and little was known about how they fared in the adult world. Any effort to examine the effects of the interventionist surgical care protocol has tended to focus on whether the individual changes gender later on. The implicit assumption was that if people did not reassign gender in adulthood, then the interventions were justified. So confident were doctors about childhood normalizing genital surgery that its social, emotional, and sexual benefits and harms were left unexamined for several decades.

Criticisms levied at the medical profession in recent years have focused on the withholding of diagnostic information, nonconsensual surgery, the absence of follow-up evaluations, and inadequate psychological support (e.g. Anonymous 1994; Chase 1998b; Simmonds 2004). It is now recognized that the best care is multidisciplinary and one that incorporates consumer views. Improved access to psychological care for people with so-called disorders of sex development has been proposed since the mid-1990s. However, service delivery—its theoretical frameworks, service priorities, and methods—have never been coherently articulated (Liao 2007; Liao & Simmonds, in press).

PREPARING FOR THE CONSULTATION

Because of the stigma attached to DSD, service users can be expected to be highly variable in their relationship to the diagnosis. Some service users would find it aversive to ask for directions to "the DSD Clinic." The name of the clinic reflects the user-centeredness of the service. Choice exists in naming the clinic for example after a user advocate or the name of the local area.

DSD diagnoses are numerous but each is rare. Therefore, it is unrealistic to expect clinic staff to be familiar with the details of DSD conditions. Training for ancillary staff, such as receptionists, radiographers, and outpatient nurses, should focus on basic etiquette, such as not openly discussing the purpose of the appointment and calling out or displaying the client's clinic number rather than his or her full name. Some service users may choose to use a pseudonym. This choice is often made in the interest of partners and/or family members who prefer to remain anonymous. Having to share a waiting area with pregnant women, or where walls are plastered with baby photographs, can be painful for some DSD clients, as can being asked about their last period or cervical smear test by an intake nurse in front of other people.

Box 3.2 contains an excerpt from an article that appeared in the *British Medical Journal* by an anonymous woman with CAIS who has this to say about how poor communications added to her "despair."

Box 3.2

I was admitted to hospital for adhesions and a nurse asked me if I had previously had a penis. I was seen by a psychiatrist in his office in a maternity hospital with a heavily pregnant doctor participating. I was seen by a psychosexual counsellor who tried to take me apart and reconstruct my psyche, a process which nearly caused a mental breakdown. (Anonymous 1994)

The team needs to decide on who the best person is to welcome the client or family at their first visit. This person may be a nurse, a psychologist, or a service coordinator. A few minutes spent in preparing the client or family for the consultation can help the client or family to reach a state of relative calm by the time they meet the medical consultant. Inquiry about the journey, about hopes and concerns regarding the appointment, about the most important questions at the forefront of their minds, can help the client or family to feel that they are attending a service that is sufficiently developed to anticipate their preoccupations. The first contact person can also offer the client or family information about the clinic and individual team members, and the order of events and their scheduling.

Photographs of team members in the waiting room can help to "humanize" the service immediately.

DSD is seldom discussed in everyday life, so ~~that~~ the tertiary center that clients attend may be the only safe place where all aspects of their conditions can be freely and openly discussed. Care providers should not underestimate the high value placed by service users on a robust professional alliance.

COMMUNICATING IN THE CONSULTATION: TALKING TO PARENTS ABOUT THEIR CHILD'S DSD

Adults with DSD conditions suggest that parents need to "come to terms" with the diagnoses themselves in order to help their children (Simmonds 2004). When presented with a child with non-normative sex development, however, parents can be expected to feel extremely confused and emotionally vulnerable. "Coming to terms" may require sustained support for coping with losses and fears, and for examining our taken-for-granted beliefs about "normal sexuality" and "normal life."

Health professionals' behaviors can be expected to be highly influential on their client's sense-making of the situation. Every clinician meeting the parents for

the first time should begin with congratulating the parents on the arrival of their baby and then engage them in the usual exchanges about their baby and about being parents (again). When there is a delay in gender assignment, it is important to listen to the parents' reactions without rushing to placate them (see Box 3.3).

It can be reassuring for parents to know that, although they had not come across these situations, the situations are not so uncommon. The message that DSD is at least as common as twins or red hair may be easier to relate to than prevalence in numerical terms. The professional and reliable presence of an understanding and expert team will help to contain emotions.

There may be further investigations to implement. If not urgent, then the client or family should be allowed sufficient time to absorb the information in order to make an informed choice. Medical preoccupations may not be in synch with what is occupying the client's mind. Here are some example questions to elicit information on how the parents are processing the event:

- These are some very clinical words, what do they mean to you? What effects do these words have on you?
- Who is able to support you; are there others whose support you need?
- How do you see this affecting how you feel about yourself? And your child?
- In what way will your relationships change, in what way will they remain the same?
- How would you like other people to understand this condition? What words would you use? Can we help?
- How would you describe the way you're feeling right now? What can we do to help?

Box 3.3

A set of guidelines (DSD Guidelines 2006) provides an example wording, which we have modified here:

"Your baby was born with a kind of variation that happens more often than you hear about. Our team works with these situations regularly. We are doing a series of tests to find out whether your baby is probably going to feel more like a boy or a girl. We expect to have more information for you within [realistic timeframe]. Although you may not feel ready to send out a birth announcement with the baby's gender and name, there are some choices to consider, such as letting close friends and families know that you have a healthy baby who is full of personality, or send photographs to show how lovely your baby is, or let them know how best to support you."

Gender Assignment in Infancy

Aside from the diagnostic information and prognostic projections, parental preference for gender assignment may be informed by familial, cultural, and religious values, and whether there are already boys and girls within the family. Other parents want to know the "true sex" of their child and whether their child will grow up to be a happy individual.

Parents may be reassured that the same conditions that enable children to thrive are exactly the same as those that enable children with DSD to thrive.

Parents may ask if their child would be "normal" in relationships. This type of question offers opportunities for the team to help the parents to "unpack" what "normal" means, and what they value most about relationships. Implicit in questions about normal relationships is whether their child will be homosexual. Some parents may feel unsure about asking this question, perhaps because they are afraid to hear what they think might be the answer, or perhaps because they worry about offending the clinician to whom they are talking. It is important to anticipate the concern and assist the clients to find the words, so that the issues can be openly discussed. It is not possible to predict any child's future sexuality, and the majority of gay people do not have a DSD condition. Whichever direction their sexuality takes, young people will benefit from the same support and acceptance.

Perhaps the most important message to offer parents is that they are not alone, that other families have worked with the same situation. Today, parents can potentially access Internet support from all over the world, or telephone or face-to-face contact with peer ~~forums~~.

Feminizing Genital Surgery for Female-Assigned Children

Feminizing surgery does not improve the child's health, therefore it is a difficult choice for parents to make on behalf of their daughter. Good intentions of care providers are not a replacement for thorough exploration that gives equal weight to immediate surgery and delaying surgery until the child can participate in the decision process. Care providers must avoid subtly pressuring parents to make a decision before they have had enough time to bond with their child and to digest all of the information or consult with support groups if they wish. Clinicians should be prepared to discuss the pros and cons of potential interventions, answer any questions, and even refer for a second opinion if the parents are finding it too difficult to reach a balanced decision that they are comfortable with.

Whatever parents choose, they do so in the interest of the child. Those who choose surgery may be motivated by the expectation that surgery will normalize

their daughter's genital appearance and that she will be spared any ridicule or rejection. They may believe that it will encourage a female gender identify. They may also believe that their concerns about sharing information with babysitters, helpers, and teachers about the child's diagnosis will be removed. It is important to be clear about how realistic these expectations are. Normalizing surgery does not eliminate the diagnosis. Their psychological engagement with the issues will continue to be required.

Discussion of the short-term risks and long-term uncertainties will help parents make what is a stressful and perhaps traumatic decision that they will have to live with. Pediatric surgeons are generally more optimistic about surgery than are adult care providers. One reason for the schism is that the former will follow-up infants and children for short periods—before puberty and engagement in sexual relationships—whereas the latter see postpubertal adolescents and adults who have been operated on as children, often repeatedly and with poor outcomes. It is recommended that pediatric doctors inform parents that surgery is highly unlikely to be one-off, and that there is a debate about satisfactory outcomes in terms of appearance and function in adulthood.

For the currently small proportion of parents who decide to defer surgery until their child can give consent, psychological input for communication skills development ~~are~~ needed. The parents may need to educate not just the affected child but also siblings and the extended family or wider community; in other words, they will need help in developing a more constructive, systemic approach for education about non-normative sex development in the child.

Intimate Examinations and Photography

> Children who are operated on are especially vulnerable to repeated genital examinations and photography because doctors are compelled to monitor and evidence results. Some pediatricians have alluded to the "repeated psychological insult caused by frequent genital examinations and operations" (Jaaslekainen, ~~Tiitimen, &Voutilainen~~, 2001), and some gynecologists and psychologists have warned about the emotional distress caused by medical photography (Creighton, Alderson, Brown, & Minto 2002). Genital surgery for ambiguous genitalia is intended to normalize gender identity and sexuality but, ironically, the attendant scrutiny that follows surgery is a direct challenge to the psychological justification for it.

Small developmentally appropriate steps can be taken to reduce the shame and embarrassment associated with intimate examinations and photography:

- The presence of any observer must be rationalized with a tacit assumption that families will feel under a great deal of pressure to comply with doctors whose

good will is important to them. The family should always have the benefit of the slightest doubt.
- Invite the child to bring his or her favorite toy or object of comfort.
- Give time for the family to settle.
- Involve the child rather than distract him or her to avoid possible dissociation under stress that could lead to future psychological problems.
- Invite child and parents to ask any questions concerning the necessity and nature of the examinations and photography.
- Offer choices to provide a sense of control (e.g., invite the child to choose a gown, whether he or she would prefer the doctor to stand to the right or left, and so on).
- Start with other parts of the body (e.g., feet, fingers, ears). Invite the child to do the same for his or her doll or teddy bear likewise, to find out how doll or teddy is doing.
- Invite the child to use the stethoscope to check if the doctor is well.
- In case of photography, provide a toy camera for the child to take pictures of the adults present.
- In nonurgent situations, offer a genuine choice as to whether the child or family would prefer to have the examination or photography at the next appointment.

The existence of a protocol for intimate examination and photography and the care that goes into it reflects the team's collective awareness of psychological risks and respect for emotional safety for the child. Every photography occasion should have a rational basis—what is it for, what should be the timing, how should the client or family be prepared to maximize perceived control, dignity, and respect?

Counseling Parents About Risks of the Same Diagnosis in Future Pregnancies

DSD conditions differ in the extent to which a known genetic component is implicated. In an X-linked recessive condition such as AIS, there is a 1 in 4 chance in a given pregnancy of an XY boy, an XY AIS child, an XX girl or an XX carrier girl. A range of mutations on the androgen receptor (AR) gene has been identified, and most, if not all, cases of CAIS can be explained by receptor defects. However, the majority of PAIS individuals exhibit no defect in androgen-receptor binding, suggesting that other genetic defects are involved. It is also possible in PAIS for the same genetic defect to express itself as differing genital appearances. CAIS and PAIS may thus be caused by different defects at the genetic/cellular level and are thought not to occur in the same family (Ahmed et al. 2000). Also, in about a half to one-third of cases, AIS arises from a de novo mutation. In some of the other XY female DSDs, such as Swyer syndrome, 5-alpha-reductase deficiency, and Leydig cell hypoplasia, a role of ~~genetic~~ in the condition has not been established with any certainty.

At a time when the AR gene and its mutations were being actively researched (up to the late 1990s), families in the United Kingdom would have to persuade a research team to carry out carrier testing as a favor on the back of their research, there being no National Health Service (NHS) testing service available. This was a long and frustrating process for some families, as illustrated in Box 3.4, which was provided by a family who welcomed genetic testing but were unable to access the service.

The Androgen Insensitivity Syndrome Support Group (AISSG) has received inquiries from parents of newborn babies with AIS who are angry that they have only now discovered that older female relatives had kept to themselves facts about their own AIS (with or without knowledge of its inherited nature) for many years. Conversely, there have been enquiries from older adult women who have been prompted, by the birth of an AIS baby into the wider family (in an era when parents are more likely to have received truthful disclosure and to be more active in seeking out information), to look into the reason for their own failure to menstruate and consequent infertility. There have also been cases in which a woman seeking AIS carrier testing, at the time when this was done in a research laboratory, was surprised and somewhat alarmed to discover that the researchers had been aware of AIS in her

Box 3.4 Case Study 2

In 2012, Androgen Insensitivity Syndrome Support Group (AISSG) ~~U.K.~~ was contacted by a 36 year-old woman with complete androgen insensitivity syndrome (CAIS). She was diagnosed at 12 months of age, following a left inguinal hernia, but did not find out about her diagnosis until she was 27 years old.

She has an older brother and two older ~~XX-female~~ sisters. Her genetic testing to confirm CAIS had helped her sisters to get tested for carrier status. She explained, "My middle sister, who already had a girl [now aged 12] and two boys, is not a carrier. However my oldest sister is a carrier…I think her IVF was out of necessity (she was 40) and not due to her being a carrier for AIS. Her daughter [aged 18 months] was conceived from a donor egg so there is no chance of her being affected by AIS."

In relation to genetic risks, this woman said: "because I didn't know [about the diagnosis], neither did my sisters…who could then have been carriers, so then my [middle] sister's daughter could have had it too. And again, when my sister wanted to find out in advance [if she might be a carrier], the consultant she saw was very anti finding out and told her that her daughter had a right not to know and that even if she turned out to be a carrier he would not test her daughter."

When asked if her story could be used in this chapter, she said: "I would be extremely happy for you to use my story in any way which you think it might bring an end to all this nonsense of doctors/parents keeping critical medical information from clients and their families."

family members for many years, even though she was the first to have sought such testing.

In a different scenario, invitation to genetic testing could fill the client with dread of having to disclose and discuss DSD within the family. A collaborative approach that safeguards informed consent means plenty of time and opportunities to help clients explore the potential implications of genetic testing for family relationships, as well as education and support for potential carriers to appreciate the implications (including the communication challenges with the next generation).

Advice to Parents About Talking to the Growing Child

Far from protecting the child, narratives by affected adults and psychological research suggest that lying, evasive answers, and refusal to discuss DSD is potentially psychologically harmful to the affected growing child.

It is imperative that parents are open and honest about their child's DSD, and clinicians should take it upon themselves to lead by example and communicate in no uncertain terms that DSD is nothing to be ashamed of. However, although openness and honesty signals to the child that he or she is fully accepted without shame, parents differ in how resourced they are for meeting the communication challenges. A central concern for parents is what message to give to the child about what he or she should do with the information. Thus, disclosure about DSD may feel like a floodgate that has to be either fully closed or fully opened.

In tolerating and accepting clients' distress, even if it feels overwhelming at times, clinicians are nurturing the clients' capacity for the emotionally charged and risky task of talking to their child and other family members about DSD. In sharing information about DSD, parents are more likely to be able to access social support and end their isolation. Conversely, if parents are poorly cared for by the clinical team, their capacity for open communication may be diminished and, in time, their fear and shame may be magnified.

COMMUNICATING WITH THE ADOLESCENT AND ADULT CLIENT

The following excerpt from the British Medical Journal article mentioned earlier illustrates very well our rigid dimorphic view of gender that is so unhelpful to people with DSD. There are no easy solutions to this problem, but a supported exploration of the identity issues provides a space where clients can begin to develop their own responses.

I am chromosomally male, a pseudo-hermaphrodite. These two phrases pervaded my rational thought. I did not think I was female. I did not think I was male. I did not know what I was. (Anonymous 1994)

Diagnosis

As mentioned earlier, some DSD diagnoses are reached during adolescence, a time when people tend to be highly sensitive about social norms in bodies and their appearances. Affected adolescents and adults are likely to be devastated when told of their diagnosis. Their self-evaluations may be compromised by dominant stories of "normality" and "abnormality" about sex and sexuality in the social world, stories that position their situation as "deviant."

Any of the following diagnostic information, delivered in a clinical language, can be shocking: absent vagina, absent menstruation, infertility, chromosomes, heredity, lifelong medication, hirsutism, weight gain, and short stature. An important first task then, is to develop a shared language with clients. Questions such as the following may be helpful for starting a discussion:

- What words would you prefer to use?
- What's the most challenging aspect of your situation to put into words?

Clinicians can be overly focused on medical information, when it is the meaning that determines reactions. Communication of the diagnostic and treatment information in plain language needs to be followed by empathic engagement with the young person's emotional reactions, which needs to be followed by the development of richer vocabularies drawn on alternative discourses to address the social, emotional, and sexual aspects of DSD.

Questions such as the following may be helpful for exploring the client's understanding of the condition and, should they be necessary, treatment options:

- Can you tell me the name of the diagnosis? What kind of information have you been given about this?
- What have you been told about the investigations that are happening and what they are for?
- What is your understanding of how the tests fit with your overall treatment plan?
- How would you describe the way you're feeling right now?

The following questions may be helpful for extending the dialogue:

- What effects do these long words have on you?
- How would you like other people to understand this?
- If you were to brainstorm different words, what might you come up with?

Decision About Genital (Re)constructive Surgery

Where a gender boundary is blurred by characteristics deemed to belong to the "opposite sex", individuals can become extremely preoccupied with fixing the problem. Many women with DSD diagnoses speak of feeling like outsiders and feeling unentitled to relationships until they have had surgery, such as vaginoplasty, to remove the obstacles for "normal sex" (Boyle et al. 2005). When clients seek medical treatment, they may also be seeking "normality" in identity, relationships, and sexual practices. Health professionals feel under a great deal of pressure to ensure that diagnostic information simultaneously comes with offers of some form of a normality solution. But, although this may help to contain doctor and client anxiety, such action inadvertently comes with the subtext that body differences are unacceptable, indeed inconceivable—even for a minute. It is this subtext that is ultimately unhelpful.

Decision to undergo surgery may be fear-based—to avoid the need to explain about DSD. It is possible to further the client's understanding of the benefits and limitations of any treatment, for example, by asking:

- Can you repeat to me in your own words what you have been told about the treatment—what is the procedure called, what does it involve, how long does the operation take, what is the recovery period?
- What do you understand to be the potential benefits and risks—immediately and in the longer term?
- Which aspects of your life are dependent on the operation? Which aspects are more up to you?
- Who else can help you reach a balanced decision not just about treatment but the best timing for you?
- What do you think the operation may leave unaddressed? What else might be needed? What steps would you take? Who else can help?

Fertility Issues

In many cases of XY female DSD conditions, there is a fundamental infertility because of interrupted gonadal development and lack of a uterus; however in those with Swyer syndrome, the presence of a primitive uterus can sometimes, with hormone treatment, lead to menstruation and pregnancy using in vitro fertilization (IVF) and a donor egg. The case study in Box 3.5 illustrates an acute need for better communication about ~~genetic~~.

Genetic Research

In the current race toward molecular discoveries, scientists and clinicians may not have developed sufficient appreciation of the potentially far-reaching implications

Box 3.5 Case Study 3

A "female with partial androgen insensitivity syndrome (PAIS)" who has two affected sisters, all of whom were said to have inherited the condition from their father, recently contacted Androgen Insensitivity Syndrome Support Group (AISSG) ~~U.K.~~ One of the sisters had two children, including a son with PAIS. The inquirer had been trying unsuccessfully to have children for 3 years. Both she and husband had been fully tested and received the "all-clear." She said, "I never mentioned the genetic condition to my doctors as I didn't think it made a difference at this point. However, I am now wondering if this could affect my chances of conception?"

When it was pointed out that AIS is usually passed on via the mother (if inherited, as opposed to being a de novo mutation) and that women with PAIS do not have ovaries or uterus, and do not menstruate, so cannot conceive, the inquirer replied:

"My dad has the PAIS. As it is his X that is affected, he passed on his bad X (so to speak) to me and my two sisters. Us three sisters have all been told by a genetic counsellor that we all have 50% good X and 50% bad X and have all had a blood test to confirm this. My nephew, now aged 8, inherited the bad X off my sister, who has also been confirmed and diagnosed as having PAIS; my nephew has had countless operations to try and correct the hypospadias on his penis. My sister with PAIS has had children and I have had scans and have all internal and external female organs. We have been told that as carriers ~~as~~ PAIS, females are only affected by having less pubic hair. It only came to light that me and my two sisters had this when my nephew was born and tested. We have definitely got PAIS and all have a 1 in 2 chance of passing this on to our children."

An XY male fathered by a man with androgen insensitivity could not have inherited his father's X chromosome, and thus would neither inherit nor carry the gene for the syndrome. A genetic female conceived in such a way would have inherited her father's X chromosome, and would thus become a carrier. A female carrier has a 50% chance of passing the affected androgen receptor (AR) gene to her children. If the affected child is a genetic female, she too will be a carrier; and an affected 46,XY child will have AIS. Therefore, it was explained to this woman that she and her sisters must have been ~~XX females~~ who are *carriers* of the faulty gene inherited from their PAIS father; that they did not *have* PAIS, because only people with an XY karyotype could have PAIS. They could not be both a carrier of, and be affected by, AIS—it had to be one or the other. It was recommended that they seek advice about any possible effect on the fertility of carriers.

The case study illustrates the complexity of ~~genetic~~ to lay people, and the importance of developing creative and multimedia means of communication, rather than relying solely on verbal exchanges and literacy. Level of understanding needs to be carefully explored in genetic counseling, which offers opportunities for previous misconceptions to be corrected and for clients to be signposted to seek appropriate advice on important issues such as fertility.

that genetic testing can have for some clients and their relationships with self and others. We have met clients who felt torn by having to discuss their DSD, which was a family taboo, but for whom such discussion was necessary to involve generations of family members in genetic research as a token of gratitude to their doctors. Genetic testing can be a driver for first-time disclosure within the extended family. Although this can be a positive process, the client needs to be ready and must be doing this for him- or herself. Where there is a sibling with the same diagnosis, one individual's decision will profoundly affect the other, who may view the process in hostile terms. We have come across significant conflict between affected siblings because of genetic testing. Professional psychological support is typically not considered necessary.

Because people affected by DSD have historically been poorly served by their care providers, genetic researchers whose work is less than likely to benefit the affected individual must work within the strictest ethical and professional governance frameworks.

SUMMARY

Bodies that challenge our erroneous binary categorizations of sex and gender have always existed. They have come under professional and scientific scrutiny for only a short period of time, relative to the thousands of years of their recorded history. The socially sanctioned project of bioclassification and normalization of atypically sexed bodies is replete with controversies and schisms. At one end of the polarity, bio banks are being built to study molecular causes of "abnormal" reproductive and genital development, mainly to improve understanding of "normal" development. At the other end, the very notion of "normality" is understood as a social construct that suits specific temporal and cultural locations. Somewhere within this spectrum of knowledge production sits medical management of atypically sexed bodies.

Space does not permit a full exposition of research and practice ethics in the field. Nevertheless, we hope we have provided sufficient context to argue for a swing toward a client-centered approach to clinical management and research and have signposted to the critical importance of robust communication skills in such a service model. Elsewhere, we argue that there is sufficient negative user feedback to warrant the development of in-depth communication training for clinicians in the field (Liao & ~~Simmonds in~~ press). Meanwhile, here is a summary list of points that we think clinicians should bear in mind in terms of communicating about DSD:

- Always privilege emotional safety, dignity, and respect for the client.
- Be aware of one's own personal values and assumptions relating to "normality" in sex and sexuality and their influence on professional practice.
- Role model shame-free communication about DSD that enables rather than diminishes the client.

- Speak in relative rather than absolute terms (e.g., "variations" and "difference" instead of "normal" and "abnormal," "shorter than average" instead of "short," "unwanted hair growth" instead of "hirsutism").
- Tailor communication to the age, developmental stage, and current emotional well-being of the client.
- Check understanding, expectations, and feelings relating to aspects of the diagnosis and any procedures.
- Encourage balanced decision making in relation to elective procedures.
- Provide information about user support organizations.

Routine collection of anonymous client feedback is a tangible way to involve clients in ongoing service improvement. It offers them opportunities to express their appreciation of the team and the organization.

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