

# Clinical Guidelines

## for the Management of Disorders of Sex Development in Childhood



CONSORTIUM ON THE MANAGEMENT  
OF DISORDERS OF SEX DEVELOPMENT

Funded by the California Endowment and Arcus Foundation

Provided as a service of Accord Alliance, a project of the Tides Center

# **Clinical Guidelines**

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OF DISORDERS OF SEX DEVELOPMENT

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# CLINICAL GUIDELINES FOR THE MANAGEMENT OF DISORDERS OF SEX DEVELOPMENT IN CHILDHOOD

Consortium on the Management of Disorders of Sex Development

Published August 16, 2006

First Edition

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ISBN: 0-9773201-1-1

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Published by Accord Alliance, a project of the Tides Center, a 501(c)(3) tax-exempt organization

531 Route 22 East #244

Whitehouse Station NJ 08889

[www.accordalliance.org](http://www.accordalliance.org)

## Acknowledgments

This handbook is the result of an extraordinary collaboration among clinicians who care for people with disorders of sex development (DSDs), parents of children with DSDs, adults with DSDs, and other patient advocates. We are truly grateful for the assistance and support that has been provided to us in this work, and we hope that readers will use the associated website ([www.dsdguidelines.org](http://www.dsdguidelines.org)) to give us feedback about it.

We are grateful for the participation of David Cameron, Peter Trinkl, and Esther Morris Leidolf in this project. However, they would like to make it known that they do not support the term “Disorders of Sex Development.”

We would like to thank Yvonne Day ([www.ydaydesigns.com](http://www.ydaydesigns.com)) for design assistance and Bob Stayton ([www.sagehill.net](http://www.sagehill.net)) for technology assistance with DocBook.

This document has been made possible by generous grants from The California Endowment ([www.calendow.org](http://www.calendow.org)) and the Arcus Foundation ([www.arcusfoundation.org](http://www.arcusfoundation.org)) provided to the Intersex Society of North America ([www.isna.org](http://www.isna.org)).

Web-browseable and downloadable versions (both with clickable links) of *Clinical Guidelines for the Management of Disorders of Sex Development in Childhood* and *Handbook for Parents* are available at no cost from [www.dsdguidelines.org](http://www.dsdguidelines.org).

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# Chapter 1 Introduction

## PURPOSE OF THIS DOCUMENT

The purpose of these guidelines is to assist health care professionals in the provision of diagnosis, treatment, education, and support to children born with disorders of sex development (DSDs) and to their families. While debates about the best way to care for patients with DSDs continue,<sup>1-7</sup> this handbook offers individuals and institutions a model aimed at minimizing the potential for harm to patients and their families. It is therefore also designed to reduce the potential for liability and to improve patient follow-up.

These guidelines begin with the commonly-held assumption that the goal of DSD treatment is the long-term physical, psychological, and sexual well-being of the patient.<sup>8-15</sup> This approach is therefore termed “**patient-centered.**”

What follows is largely an ideal treatment scenario involving a well-organized, well-trained, and institutionally-supported multidisciplinary health care team. The authors of this document hope that it will be used to refine, to foster, and even to create such ideal scenarios, but also recognize that the reality for many health care professionals may be challenging.

It is critical, therefore, to emphasize that **it is more important to focus on providing patient-centered care than to create and maintain a multidisciplinary team precisely as described** in the following.<sup>16</sup> Although dedicated multidisciplinary teams which focus on patients’ psychosocial well-being may be more likely than alternatives to provide optimal care, **a dedicated multidisciplinary team is neither a guarantor of nor a necessity of patient-centered care for DSDs.**<sup>17</sup>

The intended audience for this handbook is health care professionals who provide care to pediatric patients with DSDs. Nonetheless, we expect that this handbook will also be read by health professional students, educators, parents of children with DSDs, and adults with DSDs. For this reason, this handbook occasionally repeats points which would be obvious to the specialist clinician. The authors

do not intend to be condescending to the specialist, but rather to be helpful to those readers less familiar with the specialist's work.

## DEFINITION OF DSDs

Disorders of sex development (DSDs) are defined as conditions involving the following elements.

- congenital development of ambiguous genitalia (e.g., 46,XX virilizing congenital adrenal hyperplasia; clitoromegaly; micropenis)
- congenital disjunction of internal and external sex anatomy (e.g., Complete Androgen Insensitivity Syndrome; 5-alpha reductase deficiency)
- incomplete development of sex anatomy (e.g., vaginal agenesis; gonadal agenesis)
- sex chromosome anomalies (e.g., Turner Syndrome; Klinefelter Syndrome; sex chromosome mosaicism)
- disorders of gonadal development (e.g., ovotestes)

DSDs consequently include anomalies of the sex chromosomes, the gonads, the reproductive ducts, and the genitalia. Note that the term “intersex” is avoided here because of its imprecision. See DSDs and Related Concerns [page 5] for a list of DSDs and condition-specific concerns.

## DEFINITION OF PATIENT-CENTERED CARE FOR DSDs

Patient-centered care means remaining clearly focused on the well-being of individual patients.<sup>18</sup> In the case of DSDs this specifically involves the following principles.

1. Provide **medical and surgical care when dealing with a complication that represents a real and present threat** to the patient's physical well-being.<sup>19-20</sup>
2. Recognize that what is normal for one individual may not be what is normal for others;<sup>18,21-23</sup> care providers should **not seek to force the patient into a social norm** (e.g., for phallic size or gender-typical behaviors) that may harm the patient.<sup>24-28</sup>
3. Minimize the potential for the patient and family to feel ashamed, stigmatized, or overly obsessed with genital appearance; avoid the use of stigmatizing terminology (like “pseudo-hermaphroditism”)<sup>29</sup> and medical photography;<sup>30</sup> **promote openness (the opposite of shame) and positive connection with others**, avoid a “parade of white coats” and repetitive genital exams, especially those involving measurements of genitalia.<sup>31</sup>

4. Delay elective surgical and hormonal treatments until the **patient can actively participate in decision-making** about how his or her own body will look, feel, and function;<sup>32-35</sup> when surgery and hormone treatments are considered, health care professionals must ask themselves whether they are truly needed for the benefit of the child or are being offered to allay parental distress;<sup>27,36-39</sup> mental health professionals can help assess this.
5. **Respect parents by addressing their concerns and distress** empathetically, honestly, and directly; if parents need mental health care, this means helping them obtain it.
6. **Directly address the child's psychosocial distress**<sup>40-41</sup> (if any)<sup>42</sup> with the efforts of psychosocial professionals and peer support.
7. Always **tell the truth** to the family and the child;<sup>43-44</sup> answer questions promptly and honestly, which includes being open about the patient's medical history and about clinical uncertainty where it exists.

## METHODOLOGY

These guidelines have been produced by a consortium consisting mainly of: (1) clinical specialists with experience helping patients with DSDs; (2) adults with DSDs; and (3) family members (especially parents) of children with DSDs. These guidelines are therefore unique in that they benefit from the experiences of the three groups (clinicians, patients, and parents) that comprise the DSD clinical triad. A number of the contributors have been in meaningful communication with dozens or even hundreds of individuals and families with personal experience of DSDs. They drew on those relationships in this work.

While this document benefits from the large literature (medical, sociological, and autobiographical) available on DSDs, consultation with these three groups has helped to address the substantial evidentiary gaps in the medical literature on DSD treatment (e.g., the lack of evidence regarding long-term outcomes of current medical and surgical options). It has also provided **an important three-way consensus surrounding the patient-centered care philosophy at the core of these guidelines.**

The authors seek continued dialogue with those with personal and clinical DSD experience in order to refine these recommendations. This handbook represents only the first of numerous anticipated editions. Feedback may be provided by using the “contact” link available at [www.dsdguidelines.org](http://www.dsdguidelines.org).

**Note:** Readers of this clinical handbook are encouraged to utilize the companion “Handbook for Parents” available through [www.dsdguidelines.org](http://www.dsdguidelines.org). A pair of handbooks is also planned for adults with DSDs and for the clinicians who care for them.

## HOW DO DSDs HAPPEN?

Sex development in the human is the result of a complex interaction of an individual's genes and his or her environment. DSDs by definition occur when an individual's sex development takes a path different from that of the typical female or typical male.<sup>11,45-47</sup>

**Typical male development** begins with a 46,XY chromosomal complement. By the 6<sup>th</sup> week after conception, the embryo has developed two bipotential gonads. The *SRY* gene (located on the Y chromosome) causes production of the *SRY* transcription factor which, in conjunction with genes not located on the Y chromosome, induces the gonads to differentiate into testes. Although the embryo initially forms both the Wolffian (male) and Mullerian (female) ducts, the testes produce Mullerian-inhibiting substance (MIS), causing the Mullerian ducts to regress. By about the 12<sup>th</sup> week, testosterone produced by the testes cause the Wolffian ducts to form the spermatic ducts. The external genitalia are sexually undifferentiated until the 7<sup>th</sup> week. Androgens produced by the testes cause the external genitalia to develop into the typical male; the proto-phallus becomes a penis, the labioscrotal folds fuse in the midline to form the scrotum, and the urethra migrates distally to a male-typical position. At puberty, testicular production of testosterone contributes to further sex differentiation. Sex differentiation in the brain appears to be the result of hormonal differentiation and social factors.

**Typical female development** begins with a 46,XX chromosomal complement. Unless the *SRY* and *TDF* genes are present, the bipotential gonads develop into ovaries, the Wolffian ducts atrophy, and the Mullerian ducts develop into the uterus, Fallopian tubes, cervix, and the upper portion of the vagina. Because ovaries do not produce androgens, the proto-phallus becomes a clitoris, the labioscrotal folds become the labia, and the urethra maintains a female-typical position. At puberty, the ovaries produce estrogen and so contribute to further sex differentiation. Sex differentiation in the brain appears to be the result of hormonal differentiation and social factors.

**Causes of DSDs** include chromosomal and genetic anomalies, in utero exposure to sex hormones ingested or produced by the mother, and random developmental variation. Because sex development involves many points of differentiation—only a few of which are mentioned above—the potential for DSDs exists at many stages of human development. Ascertaining the etiology of a DSD helps prepare patients and parents for what is to come, aids in deciding gender assignment, highlights potential problems like adrenal crisis or gonadoblastomas, and may help determine likelihood of a subsequent child being affected. Also, some family support groups are open only to patients with particular diagnoses. Nevertheless, it is not always possible to pinpoint the exact cause of a given patient's sexual variation. An excellent animation of sex development is available at [www.sickkids.ca/childphysiology/cpwp/Genital/genitaldevelopment.htm](http://www.sickkids.ca/childphysiology/cpwp/Genital/genitaldevelopment.htm) For more information, see [www.hopkinschildrens.org/specialties/categorypages/intersex/](http://www.hopkinschildrens.org/specialties/categorypages/intersex/).

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**NOTE:** The challenges of clinical diagnosis for some DSDs along with the variability in the presentation of some DSDs mean that clinically DSDs are sometimes identified by etiology and sometimes by phenotype. As a consequence, some of the following categories may overlap; for example, a patient may have sex-chromosome mosaicism and ovotestes. This list is not meant to be exhaustive of all conditions that might be considered DSDs, nor is it meant to be exhaustive of diagnosis-specific concerns.

All DSDs have the potential to cause psychosocial distress for patients and their families, particularly if genital anatomy is atypical. Regardless of etiology or phenotype, parents' and patient's psychosocial concerns should be promptly addressed by qualified mental health professionals.

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## DSDs AND RELATED CONCERNS

**17-beta reductase deficiency (XX or XY)**—Appearance female but can't make estrogen or testosterone; consequently no pubertal changes. Monitor undescended testes for malignancies. Risk of adrenal insufficiency.

**46,XY 3-beta-hydroxysteroid dehydrogenase (HSD) deficiency**—Usually lethal; risk of severe adrenal deficiency. Endocrine management necessary for maintenance of health and fertility.

**5-alpha reductase (5-AR) deficiency**—Evidence suggests substantial variation in gender identity outcomes.<sup>48</sup> If patient raised as girl, decisions need to be made before puberty about management of masculinizing puberty (e.g., patient may elect orchidectomy).

**Complete Androgen Insensitivity Syndrome (CAIS)**—Raise as girls. Infertile with current technologies. Undescended testicular tissue presents increased risk of malignancy after puberty; counsel patient to consider orchidectomy following puberty. (Delay until puberty allows patient to experience a natural feminizing puberty and to choose orchidectomy.) Vagina may be shorter than average; if patient wishes to lengthen her vagina, she may elect pressure dilation or secondarily surgical intervention.

**Partial Androgen Insensitivity Syndrome (PAIS)**—Initial gender assignment should take into account that higher degrees of prenatal androgen exposure may direct the brain to develop in a more classically-masculine fashion. (It is probable that the more virilized the genitalia, the more likely the brain has been masculinized.) Testosterone injections can be used to test responsiveness. Note that, if the patient is being raised as a girl, the testes will cause some pubertal virilization; leuprolide (marketed as Lupron) may be used to delay puberty so that patient's decision-making

## DSDs AND RELATED CONCERNS (cont.)

about orchidectomy is not rushed. If patient wishes to lengthen her vagina, she may elect pressure dilation or secondarily surgical intervention. If the patient is being raised as a boy, offer the patient hormone therapy at puberty.

**aphallia**—If normal testes, assume the brain has been masculinized. Evidence exists that, if assigned as girls, a notable percentage of these children transition later to become boys. May be fertile; preserve fertility. Any urethro-rectal communication must be repaired to avoid infection and kidney damage.

**clitoromegaly**—Test for CAH, PAIS, etc. Test mother for virilizing condition.

**46,XY cloacal exstrophy**—Complex disorder with variable presentation; long-term survival approximately 70%. In the past many of these children were raised as girls; a notable number of these children have transitioned to boys. Removal of healthy testes should not be performed without patient's consent as it eliminates fertility.

**46,XX congenital adrenal hyperplasia (CAH)**—CAH is potentially life-threatening. Until it has been ruled out, prompt diagnosis and treatment should be considered in all children with genital ambiguity. In cases of CAH, endocrine management is necessary for maintenance of health and fertility and to prevent premature puberty. Menses require drainage opening (separate from urinary system) to avoid pain and infection. Internalized vagina may be a source of urine pooling and infection if left uncorrected.

**gonadal dysgenesis (partial and complete)**—Dysgenetic gonads present substantially elevated risk for malignancies. Infertile. Karyotype varies.

**hypospadias**—Test for CAH, PAIS, etc. If associated with chordee, there may be significant pain with erection. Increased risk of urinary tract infections. Location of meatus may interfere with sperm delivery (fertility); patient may decide to address this with artificial insemination or surgery. Patient may also spray urine or need to urinate in a seated position; see general note above on psychosocial concerns. Hypospadias in conjunction with cryptorchidism increases likelihood of underlying DSD.

**Kallman Syndrome**—Raise in concordance with chromosomal sex. Males potentially fertile. Anosmia (absence of sense of smell).

**47,XXY (Klinefelter Syndrome)**—Genitalia typically unambiguously male, although testes may be small. Gynecomastia common at puberty. Likelihood of azoospermia; reproductive technologies

**DSDs AND RELATED CONCERNS** (cont.)

may be used to enhance fertility. Some learning disabilities are associated with 47,XXY; address with help from learning disability specialists.

**Mayer, Rokitansky, Kuster, Hauser Syndrome (also known as MRKH, Mullerian agenesis, vaginal agenesis)**—Ovaries present with uterus absent, misshapen, or small; associated with kidney and spine anomalies in a minority of patients. Patient may elect pressure dilation or secondarily surgical augmentation if she seeks increased vaginal length.

**46,XY micropenis**—In the past many experts counseled raising these children as girls. Evidence suggests these children can do well as boys and that, if assigned as girls, they may transition later to become boys. Causes variable; one cause of 46,XY micropenis is congenital pan-hypopituitarism, which in males is commonly associated with potentially lethal hypoglycemia, the result of growth hormone and ACTH deficiencies. Unless the pan-hypopituitarism is corrected, this hypoglycemia is typically unresponsive to most standard interventions.

**ovary and testis, and/or ovotestes (historically called true hermaphroditism)**—Testicular tissue presents an increased risk for malignancies. (Note: Use of the term “true hermaphroditism” unnecessarily frightens parents and patients; explain that this is the official term in the medical literature, but that it is a misnomer.)

**Persistent Mullerian Duct Syndrome**—Risk of cryptorchidism and associated complications. Increased risk of infertility.

**progestin-induced virilization**—History of virilizing hormone exposure is limited to prenatal life, so virilization will not progress.

**sex-chromosome mosaicism (e.g., 45,X/46,XY)**—Genotypes and phenotypes vary; may appear ambiguous or may appear unambiguously male or female. Monitor for gonadal malignancies.

**Swyer Syndrome (another name for 46,XY gonadal dysgenesis)**—See gonadal dysgenesis, above.

**45,X (Turner Syndrome)**—High phenotypic variability. Genitalia typically unambiguously female. Usually infertile except through egg donation and in vitro fertilization. Short stature. Association with cardiac and renal anomalies. Heightened risk of non-verbal learning disabilities; address with help from learning disability specialists.



## Chapter 2 Treatment Guidelines

As stated in Chapter 1 *Introduction* [page 1], the following guidelines represent an idealized treatment scenario; not all health care professionals can achieve what is described below given the real constraints of the institutions in which they function. Health care professionals are encouraged to spend their energies on developing local and networked systems that consistently provide **patient-centered care** (see the section called “Definition of Patient-Centered Care for DSDs” [page 2]) rather than attempting to reproduce exactly what is outlined below.

### MULTIDISCIPLINARY TEAM APPROACH

The multidisciplinary team can play a critical role in creating a climate of commitment to the health and welfare of children born with DSDs, as well as to their families.<sup>49</sup> It can make possible the provision of excellent care that has as its goal the long-term physical and psychological well-being of individuals with DSDs and of their families.<sup>40</sup> Although many children born with DSDs are healthy and require little medical management, having families connected with multidisciplinary teams as early as possible may ensure that familiar, expert care givers will be available when psychological, surgical, or medical needs do arise. Additionally, the challenges brought on by the environment of a developing child and family will require ongoing assessment and possible changes to established treatment goals.

Integrated team care allows focus on psychosocial concerns while providing continuity of care in fields that may at some point be needed (e.g., gynecology). It also allows substantial learning among team members and provides a critical mass of families useful for providing local peer support.

In many major medical centers, the multidisciplinary clinic model is just beginning to be used for DSDs. Presently this model is being used effectively for craniofacial anomalies<sup>50-52</sup> as well as other conditions like pediatric diabetes.<sup>53-55</sup> Those attempting to establish multidisciplinary teams and clinics for the management of DSDs may look to colleagues working with similar teams and clinics for ideas and personnel. Such colleagues can provide invaluable advice about arranging space and meetings, obtaining reimbursement for multidisciplinary care, and fostering peer support. To state the obvious:

Having a team in place does not mean the group is truly functioning as a team. Only through regular communication about cases, outreach, new findings, etc., can a team become more than the sum of its parts.

## **TEAM COMPOSITION**

The issues surrounding DSDs are multidimensional requiring cooperation from a number of disciplines in order to provide effective diagnosis, treatment, and support. A multidisciplinary DSD team typically consists of members from each of the following disciplines (in alphabetical order):

- Child Psychology/Psychiatry
- Genetics and Genetic Counseling
- Gynecology
- Nursing
- Pediatric Endocrinology
- Pediatric Urology
- Social Work
- and others as needed.

See Table 2.1 “Division of responsibilities” [page 11] for the division of responsibilities among team members.

**Table 2.1. DIVISION OF RESPONSIBILITIES**

<b>SPECIALTY (alphabetical order)</b>	<b>RESPONSIBILITIES</b>
<b>Child Psychology and/or Child Psychiatry</b>	Diagnosis and management of mental health needs of child; formal evaluations of child's cognitive status to assess child's ability to participate in decision-making; referral for mental health needs of parents; assessment of parent/child relationship and facilitation of healthy parent/child relationship.
<b>Genetics and Genetic Counseling</b>	Diagnosis of genetic etiologies; genetic counseling of parents, mature child, and other concerned family members.
<b>Nursing and Social Work</b>	Coordinate care and provide practical help to patients and parents. Provide educational help and connect families with resources and support services. Keep team apprised of special concerns of the family. If they have the specific training and experience, they may also supplement the mental health services provided by the team's psychologists and psychiatrists.
<b>Pediatric Endocrinology</b>	Diagnosis and management of endocrine disorders, including those involving development of secondary sex characteristics, fertility, and adrenal and pituitary function.
<b>Pediatric Gynecology</b>	Assessment of female sexual anatomy. May work with urologist when surgery is considered.
<b>Pediatric Urology</b>	Diagnosis and management of urologic concerns; provision of surgical services when necessary and requested. (Note that at some institutions <b>general pediatric surgeons</b> provide surgical care.)
others as needed	For example: Parents may occasionally need and want professional <b>adult mental health services</b> , especially in moments of crisis. If there are language differences, a trained <b>medical interpreter</b> will be needed (see When English is not the Family's First Language [page 20]). The team may also need to consult with a <b>clinical ethicist</b> , especially when opinions differ regarding what constitutes the child's best interests. Some teams have also found it useful to have a <b>biochemist</b> as a regular member.

## TEAM OBJECTIVES

The goals of the team include the following.

1. Provide integrated care for patients, including initial support to parents.
2. Develop and implement an integrated consultation system and multidisciplinary clinic with regular case conferences.
3. Provide continuing education for team members regarding DSD issues through regular meetings and in-service programs (e.g., journal club, guest presentations, grand rounds).
4. Implement long-term follow-up of patients and their families to evaluate outcomes, to ensure quality care, and to advance team learning.
5. Connect with and educate community pediatricians and other health care professionals.
6. Reach out to parents and adults with DSDs to grow a local peer support network.

## TEAM LEADER, TEAM COORDINATOR, AND TEAM LIAISON

The efforts of team members need to be managed in a timely, organized, and consistent manner. To ensure this, it is often useful to designate individuals to play the following roles:

- **Team Leader** The team leader, a physician, functions as the formal institutional head of the team; he or she assembles the team and oversees the team's efforts.
- **Team Coordinator** The team coordinator, often a nurse practitioner or social worker, keeps team members connected (including by organizing consultations, case conferences, and team educational opportunities) and ensures timeliness and continuity in case management.
- **Team Liaison** The team liaison (sometimes a nurse practitioner, social worker, genetic counselor, or psychologist) serves as the point person for the family, ensuring that parents are kept informed of all care and treatment options and linking the family with resources and support services.

These roles need not be mutually exclusive or divided in precisely this way. Some teams find that the work of the liaison is best shared by the leader (a physician) and the coordinator (e.g., a social worker), having the leader function as the liaison for major consultations (e.g., the initial meeting with the parents) and the coordinator function as liaison for routine care.

## PROVIDING CONSISTENCY

Providing a consistent liaison from the team to the family allows the family to feel anchored and builds a relationship of trust. It is also helpful to have a liaison in place for when differences of opinion arise within the team. In that case, those opinions can be relayed to the family by the liaison who also explains the logic behind each opinion. He or she can then arrange individual consultations for further explanation of the divergent opinions as needed. This way the emphasis remains on facts and logic rather than on disagreement between persons or personalities, and the group also avoids undermining the team approach. Families learn of clinical disagreement in a consciously supportive environment rather than inadvertently through a series of conflicting consultations.

## TREATMENT PROTOCOLS FOR THE MANAGEMENT OF NEWBORNS WITH DSDs

The discovery of a DSD combined with the stress of birth puts parents in an especially vulnerable position.<sup>56-57</sup> It is critical, therefore, that attending health care professionals at the team's medical center and at referring obstetric and neonatal units be trained to respond in a timely and calm manner that reassures the parents. The following steps outline recommended procedures; these are summarized in flowchart form in Figure 2.1 "Protocol Summary" [page 21].

### 1. **OB/Gyn or other attending health care professional reassures parents and contacts**

**multidisciplinary team.** Upon recognition of a DSD at the time of delivery or anytime thereafter, the attending health care professional first lets the parents or care givers know exactly what they have noticed that is unexpected about the development of the child. (It can be helpful to also tell the parents that these kinds of variations in sex anatomy are not as uncommon as parents might expect.)<sup>39,58-59</sup> The attending assures the parents that the health care professionals are going to attend to the needs of the parents and the child by consulting with health care professionals familiar with these concerns, and assures the parents that they will remain involved and informed in the process.

At this time the attending health care professional contacts the person on the multidisciplinary team designated as the primary contact, and the attending also contacts the family's pediatrician to inform him or her of the finding of an apparent DSD.

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**NOTE:** Because CAH can result in adrenal salt-losing crisis and shock, CAH should be considered in all children with genital ambiguity. Until it has been ruled out, clinicians must attend to prompt diagnosis and treatment of possible CAH.

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## A POUND OF PREVENTION: OUTREACH TEACHING

Parents and mental health care professionals report that over-reactions, unnecessary panic, and drama on the part of staff in labor and delivery constitute a lingering source of fear, anger, and depression.<sup>49,60-61</sup> For example, several parents have been terrified and confused by having a nurse in the delivery room tell them “your baby is a hermaphrodite.” **It is therefore critical that all attending health care professionals behave in a thoughtful and calm manner.** Team members should also be aware that many parents referred from other hospitals may have already been unintentionally traumatized by naive or insensitive comments, and may need immediate help from mental health professionals to address those experiences. The best way to avoid this kind of problem is to do outreach teaching at local institutions so that referring obstetric and neonatal personnel understand the basics of DSD management. This handbook can be helpful in that endeavor.

- 2. Team liaison meets family.** As soon as possible, a team liaison meets the parents/family of the child and explains the referral to the multidisciplinary team. If possible, the meeting occurs with the family’s pediatrician present so that the pediatrician can remain fully involved in the child’s care. This meeting can happen as an outpatient visit if the child is medically stable.

The liaison making first point of contact with the parents will typically find it necessary to provide initial supportive counseling to the family. If the designated liaison is not trained to provide counseling, a mental health professional so trained accompanies the liaison. It is likely that at this point the parents will ask numerous questions. It is important that the liaison indicate that more information will be forthcoming as it becomes available, and that it be acknowledged that waiting is hard. The liaison also informs the parents that tests and an examination will be necessary for the baby, and explains those tests and their purposes, writing down the information for the family.

The liaison also offers reassurance to the parents that they are not alone and that the medical center staff has worked with similarly affected children and their parents. The liaison at this time may also advise the parents of the presence of a peer/parent support person and arrange a contact by phone or in person if the parents so choose. Many parents will find it extremely comforting and calming to meet other parents who have been through a similar situation, and to see that children born with DSDs can grow up to be happy, healthy, socially functional adults.<sup>57</sup>

- 3. Team coordinator contacts multidisciplinary team members.** As soon as the team leader feels it likely a case conference will be needed, the team coordinator contacts members of the multidisciplinary team in order to set up a time for a case conference.

**4. A small number of appropriate representatives of the medical team examine the child with the parents and family pediatrician present.**

**a. Setting the stage for the examination**

- i. Prepare for the examination and follow-up discussion.** This includes having educational materials ready and having a comfortable room reserved for the follow-up discussion, one that will support a full and confidential conversation. Audio recording equipment may also be prepared so that the parents can be offered audio recordings of the discussion. (See below, item labeled “Sit down together and discuss findings” [page 17].)
- ii. Parents participate.** The child’s parents are present for the examination. Whenever possible, it is recommended that both parents and the family pediatrician be present. Parents are also encouraged to bring a trusted friend or relative with them for support (though note that keeping the number of supporters small can help avoid the parents feeling overwhelmed).
- iii. Minimize personnel.** When medical personnel substantially outnumber the family during an examination, the family often feels overwhelmed, frightened, and silenced.<sup>61</sup> Therefore it is recommended that the minimum number of medical personnel be present whenever examining an infant in the presence of her or his family, and whenever examining a patient who is aware of the exam, especially when the exam involves genital inspection.<sup>62</sup> Members of the medical team strive in this way to promote an environment that ensures privacy and dignity, and minimizes any sense of freakishness or panic.
- iv. When teaching.** In teaching settings, attending physicians take care to (a) limit in number trainees present at examinations and meetings with the family by including only those charged with responsibility for care; (b) actively model and discuss patient-centered care with trainees involved in care. This handbook may be useful as a teaching tool in such circumstances.

## PRIVACY, CONFIDENTIALITY, AND CAREFUL USE OF LANGUAGE HELP REDUCE FEELINGS OF FREAKISHNESS

Because DSDs involve sexual ambiguity, there's a large potential for individuals and families dealing with DSDs to feel freakish and ashamed. One MD who is also a parent of children with DSDs suggests that the team social worker or nurse carefully go over HIPAA (Health Insurance Portability and Accountability Act, [www.hhs.gov/ocr/hipaa/](http://www.hhs.gov/ocr/hipaa/)) rules periodically with the family, so that they know their rights. It can also be a good idea to have the team leader occasionally remind the team of how especially important confidentiality and privacy are in cases of DSDs. Health care professionals can also avoid accidentally contributing to feelings of freakishness by avoiding the use of frightening and imprecise language (like "pseudo-hermaphrodite" and "intersex").

Out of a desire to please their physicians or to avoid confrontation, patients may give consent to group examinations or repeated unnecessary examinations even though it upsets them. One psychiatrist who has worked with adults with DSDs says, "I have counseled patients who routinely consented to such examinations but dreaded and felt humiliated by them. One patient in particular reported that she was shown off to every pediatric endocrinologist who visited her MD's institution, leading to a sense that she was her MD's 'most unusual' (freakish) case. Because patients in general do not like to say 'no' to their doctors, doctors should avoid putting them in such situations."

- v. **Demeanor.** Disclosing information regarding a child's DSD is a highly sensitive matter. The approach and demeanor of the team members when interacting with the parents is crucial to the mental and emotional well-being of the family. Members of the team model honest, calm, sensitive, patient, and reassuring behavior that signals that the child is valued and lovable, and that the child is a source of pride rather than shame.
- vi. **No NICU.** If no emergency health concerns exist, the baby is not admitted to the NICU but remains with the parents or is placed in the nursery like other healthy children at the institution. (Sometimes infants born with ambiguous genitalia are placed in NICUs even though they have no life-threatening conditions. This unnecessarily heightens the anxiety felt by family members.)<sup>57,60,62</sup> Health care professionals thus attempt to promote attachment of the parents with their baby and to lessen the potential for interference with parent-child bonding whenever possible.

**b. Examining the infant**

- i. **Introductions.** Representatives of the team introduce themselves to family members and the family pediatrician and explain their roles in the child's care.
- ii. **Use of language.** Representatives of the team explain the basics of how the exam will proceed, encouraging the parents to ask questions and raise concerns as they arise. Those doing the examination make sure to explain each procedure as they perform it. They also educate the parents about the child's genital anatomy by talking about the names for the various structures as they are touched and by explaining any unfamiliar medical terminology. The team members periodically check with the parents to see if they have any questions as the exam proceeds. They model calm, reassuring openness and valuing of the child. This can be furthered by speaking gently to the baby using his or her name (if one has been given) or by using gender-neutral language. Phrases like "you're a good baby," "you're doing so well, little one," or "almost done now, sweet baby" promote connectedness to the baby during the exam, which is reassuring and instructive to the parents.<sup>59-61</sup>
- iii. **Parents dress and hold child.** When the examination is complete, the parents are encouraged to dress and hold their baby during the discussion.
- iv. **Sit down together and discuss findings.** The parents and the team members sit at the same level so that everyone can talk face-to-face. The team may offer to audio record the discussion so the parents can listen later again when they are more relaxed. See Chapter 4 *Scripts for Talking with Parents* [page 37], about how to answer frequently asked questions, including how to explain the process and timeline for gender assignment.) The team members advise the parents about what tests will be ordered and what they are intended to find out. When parents ask questions about sex development or complicated terminology, the team members use diagrams or models, draw pictures, and write down terms, names of procedures and team members, etc. (See Chapter 4 *Scripts for Talking with Parents* [page 37] and Chapter 5 *Core References and Resources* [page 41] for material useful when educating families.) Make sure the family can read the handwriting and are given the material to keep.
- v. **Provide contact information.** The meeting leader leaves parents with a written copy of the names and titles of those who attended the meeting, and provides contact information for the team leader, team coordinator, and/or team liaison (depending on the team's policy for who will function as chief contact at this point).
- vi. **Continuity of support.** Family members are likely to find the next few days of waiting for test results particularly stressful, so a designated liaison keeps in close contact with them to be sure they feel supported. The liaison encourages the family to hold, feed, bathe, dress,

and cuddle the baby, and to perform any other activities that promote a sense of normalcy and bonding.

- 5. Team members order genetic, endocrinologic, radiologic, etc. tests as appropriate.** Tests are promptly ordered to try to ascertain (1) the etiology of the child's condition; (2) relevant details of the child's anatomy and physiology. Findings from these tests will aid in deciding initial gender assignment and in planning for the long-term care of the child. They will also help parents feel more certain and less confused or anxious.
- 6. Multidisciplinary case conference.** The multidisciplinary team meets for a case conference as soon as feasible.

  - The team's liaison presents background on the child and family, including noting whether the family has a strong gender assignment preference.
  - Members of the medical team who conducted the initial examination communicate findings.
  - Available test results are reported.
  - Team members discuss the meaning of the results of examination and tests.
  - Team discusses initial gender assignment recommendation(s), considering particularly which gender the child is likely to identify with when he or she is older.
  - Team discusses treatment options.
  - Team plans short-term follow-up and sketches out long-term follow-up.
  - The team liaison promptly provides an oral and written summary of the meeting to the parents.
- 7. Representatives of the team meet with the family.** As soon as possible after the case conference, appropriate representatives of the team meet with the family and, if feasible, the family's pediatrician, to talk with them about the diagnosis, findings relevant to gender assignment decision-making, treatment options, and plans for follow-up. Parents are offered in advance the opportunity to have a supportive family member or friend join them. Again, the team should consider audio recording the conversation for the parents so they can listen again later. The team representatives give the parents the information available regarding gender identity outcomes in similar cases and let the parents know at what point it makes sense for the parents to settle on the initial gender assignment. (See the section called "Gender Assignment" [page 25].) The representatives of the team allow plenty of time and actively invite questions and concerns from

the parents. (See Chapter 4 *Scripts for Talking with Parents* [page 37], about how to answer frequently asked questions.) They also talk with the family about the strengths of the child and family in order to put the DSD in appropriate context.

Sometimes in an effort to provide education, health care professionals have shown parents relevant articles from medical journals or textbooks. However, it is critical that parents not be unnecessarily frightened with typical medical text photographs (i.e., nude adults with eyes blacked-out). The team's liaison(s) instead tries to introduce parents to adults with DSDs, so that the parent can see that these children grow up to have a good quality of life. If this is not feasible, liaisons may provide positive self-supplied photographs and names of affected adults along with contact information for support groups. (The companion "Handbook for Parents" contains such photographs.)

Shortly after the meeting, the parents are provided with a written record or summary of the discussion as well as an audio recording if available. Families find it helpful to have information about diagnosis, treatment options, and prognosis repeated in subsequent conversations, so that they can fully absorb the information and ask more questions about it.

- 8. Provide long-term, multidisciplinary patient-centered care.** The team provides appropriate, case-specific, long-term, multidisciplinary care (preferably through a multidisciplinary clinic) observing the principles of patient-centered care outlined on the section called "Definition of Patient-Centered Care for DSDs" [page 2]. Psychosocial needs are attended by appropriately trained members of the team. Practitioners will find that some families may require very little assistance; others will need more. Team members especially attend to their long-term responsibility to the patient, and work to ensure that the child is consistently treated in a way that is maximally respectful of his or her body and spirit.

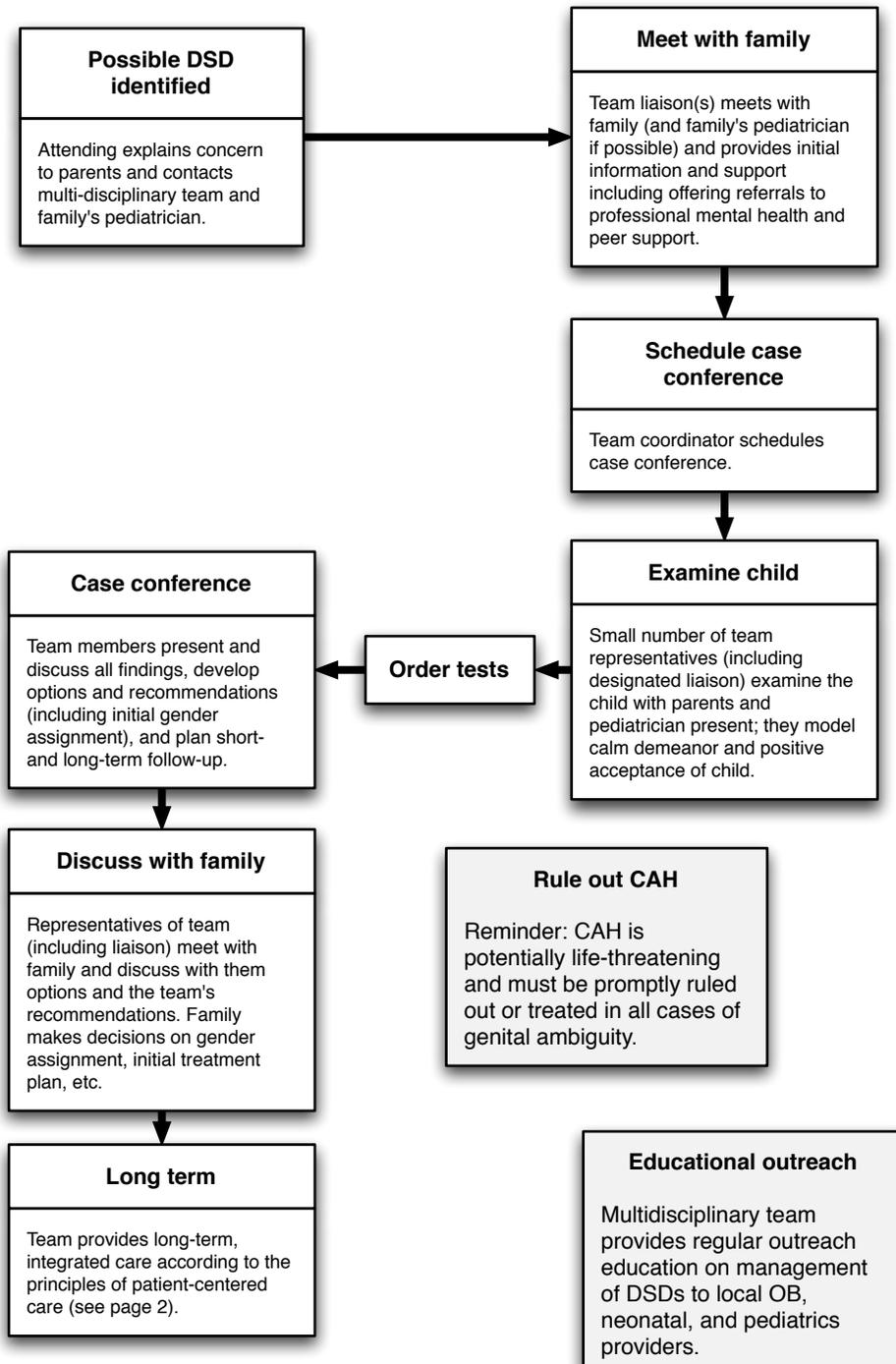
## WHEN ENGLISH IS NOT THE FAMILY'S FIRST LANGUAGE

As we've stressed throughout this handbook, communicating frankly and sensitively with parents and other family members of children with DSDs is especially important. Doing so when a language barrier exists is even more important and more challenging. Language barriers add to the fear and confusion already experienced by families and their importance should not be underestimated; parents who can't communicate with health care providers are literally placing their child's life into the hands of a person with whom they cannot effectively communicate.<sup>63</sup>

**It is a parent's right to receive all information about their child's DSD and any treatments that may be considered in a manner that is clear and understandable to them. Obtaining informed consent means that health care professionals must offer this information in the primary language of the parents.**<sup>32</sup>

For all these reasons, the team providing care should immediately contact a medical interpreter to facilitate open and clear communication with the family. It is imperative that a *trained* medical interpreter be used rather than a family member or non-specialist interpreter. Using a trained medical interpreter means that the information—even when highly technical—is translated correctly. Additionally, a trained medical interpreter, unlike some other interpreters, will also be accustomed to speaking about body parts that might be taboo in some cultures (i.e., sex and reproductive organs) and can do so in a way that is physiologically accurate and culturally sensitive. Thus, a medical interpreter ensures that patients and families get the most accurate information about a child's DSD and all the available treatment options.

The California Academy of Family Physicians' toolkit "Addressing Language Access Issues in Your Practice" offers tips, guidelines, case studies, and recommendations to help your practice better serve all of your patients. The toolkit can be downloaded free from [www.dsdguidelines.org/language\\_access](http://www.dsdguidelines.org/language_access).



**Figure 2.1. PROTOCOL SUMMARY**

## PRENATAL DIAGNOSIS OF DSDs

Improvements in prenatal screening mean that increasingly fetuses are being diagnosed in utero with DSDs.<sup>64</sup> Professionals treating DSDs must therefore be prepared to help in these situations. This may include the following activities.

- educating the parents about what is known regarding the metabolic and psychosocial aspects of the DSD (this may include genetic counseling)
- informing parents about the services available to support children with DSDs and their families
- providing parents with general DSD and condition-specific support group contact information, and encouraging them to talk with other parents and affected adults (meeting an adult with the condition often soothes parents' fear of the unknown)
- offering parents professional mental health support if they are in distress
- doing educational outreach to local prenatal service providers so that they know how to connect expectant parents with DSD-specific health care professionals

Parents can also be offered the companion “Handbook for Parents” (available through [www.dsdguidelines.org](http://www.dsdguidelines.org)). That handbook includes photographs of families and adults with DSDs, words of comfort from other parents, and thoughtful letters from affected adults to parents.

## TREATMENT PROTOCOLS FOR THE MANAGEMENT OF CHILDREN DIAGNOSED WITH DSDs AFTER THE NEWBORN PERIOD

In many cases, DSDs are diagnosed later than the newborn period, sometimes even in adulthood. When DSDs are diagnosed in childhood after the newborn period, the case may be handled via the same steps outlined above and in Figure 2.1 “Protocol Summary” [page 21], with the following differences:

1. Consultation is generally less urgent; assuming no emergency metabolic concerns exist, the team can take more time in ordering tests, conducting the case conference, and advising the family. They should nevertheless be aware that often the family will experience considerable anxiety during the waiting periods. The team psychologist or social worker can help with this, as can peer support volunteers.
2. If the child is old enough to be aware of the examination, even more care should be taken in minimizing the number of examining medical personnel and in treating the child and parents with privacy and dignity. In this case, the examiner should continually engage the child during

the examination, using age-appropriate explanations to describe exactly what the examiner is doing, whether it is a visual exam, physical exam, or an exam using any medical device. A bathrobe brought from home can provide comfort and warmth in the examining room.

3. If the child is old enough to be aware of the increased medical attention, the mental health and social work professionals on the team should help the parents explain to the child what is happening to the extent the child can understand. These explanations should be honest, reassuring, and loving. Pictures often help children understand bodily issues. (See Chapter 5 *Core References and Resources* [page 41].) Children should be encouraged to ask questions and voice fears and concerns, and be reassured that they are not defined by their diagnoses.
4. When the child is mature enough, peer support should be provided. The team coordinator can help arrange informal and formal peer support connections, including family-to-family support.
5. The parents should be offered help in dealing with school personnel and other care givers who may need to be apprised of the situation; for help with this, see the companion “Handbook for Parents” (available through [www.dsdguidelines.org](http://www.dsdguidelines.org)).
6. If the child is approaching the age of puberty or is experiencing puberty, a gynecologist or adult urologist may be asked to do the examination and therefore to function as a member of the multidisciplinary team. Additional assessments that evaluate the environment of the child’s community are critical during puberty. Any evidence that the child feels targeted, bullied, or in other ways socially or emotionally at-risk must be identified and interventions provided.
7. In a small number of cases, the team will find that the child’s initial gender assignment may not accord with the self-identity of the child. In this instance, assuming follow-up care confirms an incorrect gender assignment, the team should provide support to the child and family during the child’s social gender transition. The need for psychosocial support for the parents in this situation cannot be overestimated;<sup>65</sup> when a child’s social identity changes, so does a parent’s. Though the child (and oftentimes his or her peers) will likely find the social transition a relief,<sup>17,65</sup> the parents will often find it a substantial source of distress. In such cases, parents should be connected with the mental health care they need for support and psychoeducation, and the gender transition should be managed by a health professional experienced in this area, even if it means traveling to another town.



## Chapter 3 Background and Elaboration

### GENDER ASSIGNMENT

Gender identity development is the result of a complex interaction between genes and environment.<sup>22,48,66-70</sup> It is impossible to predict with complete confidence what gender **any** child will eventually come to identify with.<sup>8,71</sup> Like all other children, children with DSDs are given an initial gender assignment as boys or girls. But team members should be aware—and advise parents in relevant instances—that children with certain DSDs are more likely than the general population to feel that the gender assignment given to them at birth was incorrect.<sup>72-76</sup>

Gender assignment is a social and legal process **not requiring medical or surgical intervention**. (See the section called “Timing of Surgeries” [page 28] and the section called “Timing of Hormonal Therapy” [page 30].) The role of health care professionals in initial gender assignment is to obtain and help interpret test results concerning the etiology and prognosis of the child’s DSD and concerning the status of the child’s anatomy and physiology (e.g., hormone production, hormone receptors, gross anatomy), so as to inform the parents’ decision about gender assignment.

Thus, initial gender assignment (boy or girl) is made by the parents after the parents have been fully informed about the results of tests and what is known about gender identity development in patients with similar conditions. Because the parents will be the primary care givers for the child, and because they are the legal decision-makers for the child, it is critical that their sense of the situation be taken seriously and that they actively participate in the initial gender assignment. The psychiatrist or psychologist on the team may evaluate the care givers in terms of their education, cognitive capacity, coping skills, etc., as relevant to their ability to understand the DSD and to nurture a child with a DSD. Such factors may, in some cases, have bearing on the gender assignment recommended by the team.

On rare occasions, a child with a DSD may approach puberty without having clearly expressed a gender identity. Consider, for example, a child with histologically normal testes, 46,XY, and partial virilization who is being raised as a girl but who is approaching puberty without a clearly expressed gender identity.

In such cases, leuprolide (marketed as Lupron) may be used to delay puberty while psychologists and psychiatrists help the child explore feelings and options.

Throughout the provision of services to the family, it is critical that the team is supportive of atypical anatomy and behavior in children with DSDs, and that they model this acceptance for family members. Recall that one of the principles of patient-centered care for patients with DSDs is what is normal for one patient may not be what counts as normal for other patients. For instance, gender-atypical behavior is not a reason to encourage gender reassignment in the absence of the patient's desire for gender reassignment.<sup>72</sup> Flexibility is key to making patients and parents feel valued and accepted; i.e., it is key to reducing a sense of stigma. Parents should not be blamed when children express behavior atypical for their gender assignment. They should be supported in recognizing that each child is unique and lovable in his or her own right.

## WHAT DOES THE Y CHROMOSOME MEAN?

A lot of unintended harm happens when people assume a Y chromosome makes a person a boy or man, and the lack of a Y chromosome makes a person a girl or woman. One physician educator had the challenging experience of trying to calm a 23-year old patient who had just been told by a resident that she was "really a man" because he had diagnosed her as having a Y chromosome and Complete Androgen Insensitivity Syndrome.

It is true that in typical male development, the *SRY* gene on the tip of the Y chromosome helps to send the embryo down the masculine pathway. But more than the *SRY* is needed for sex determination and differentiation; for example, women with CAIS have the *SRY* gene, but lack androgen receptors. In terms of hormone effects on their bodies (including their brains), women with CAIS have had much less "masculinization" than the average 46,XX woman, because their cells do not respond to androgens.

Moreover, the *SRY* gene can be translocated onto an X chromosome (so that a 46,XX person may develop along a typical masculine pathway), and many of the genes involved in sex differentiation lie on the autosomes, not on the chromosomes (including *WT-1* on 11, *SOX9* on 17, and *SF-1* on 9). Genes involved in sex differentiation can code for atypical proteins and thus intermediate level of function. For these reasons, some professionals now prefer the term "molecular sex" to "chromosomal sex" or "genetic sex." "Molecular sex" seems to better capture the complexity.

And beyond the genes, a person's sex development can be significantly influenced by environmental factors (including the maternal uterine environment in which the fetus developed). So it is simply incorrect to think that you can tell a person's sex just by looking at whether he or she has a Y chromosome.

## PSYCHOSOCIAL SUPPORT

In the past, many DSDs were recognized as psychosocial challenges but were not addressed with professional psychosocial resources.<sup>77</sup> This has been changing, as it has in the care of other pediatric disorders, such as cleft lip and palate and childhood diabetes.<sup>53</sup> Though many families will adjust well without major professional psychosocial support, particularly if given the message by their pediatricians that their child is acceptable and lovable, those who need it should be provided access and encouragement to use those resources.

Peer support, both formal (in a clinical setting) and informal (through personal connections), often provides at little or no cost a critical form of care for patients and parents.<sup>34,62,78-79</sup> The multidisciplinary team system may provide the critical mass of affected families necessary for good, local peer support. Clinicians can also talk with colleagues at other institutions to connect families that might benefit from speaking with each other, and can develop good working relationships with support groups so that liaisons from the support groups are “on call” to help. Peer support may also be provided through internet groups and through the offering of telephone numbers from family to family (with their permission). At the same time, team members will be able to help families access more formal mental health support when needed.

### TACKLING SHAME

When they leave the clinic, patients and families go out into the world and often encounter direct or indirect messages that they should be ashamed of themselves because the child has a DSD. Health care professionals can help families by regularly providing opportunities to talk about and process reactionary shame. One social worker who helps families with birth anomalies writes, “In my experience, if time is not dedicated to talking about the almost automatic development of shame, and how to reframe this experience, then it continues to flourish. This then leaves everyone at risk for creating a plan that is directed by shame-based decisions even when we least expect it. It’s important to avoid the impression that all negative reactions can somehow be stopped with surgery or other medical care. Shame can best be dealt with when we talk about it. The idea is to talk about it out loud and directly. Shame can be an isolating and terrifying thing, especially when it is either all we talk about or something we never talk about.”

**Meeting others who share similar challenges has been consistently identified by adults and families affected by DSDs as the single most powerful therapeutic experience.** It is not uncommon to hear adults with DSDs and parents of children with DSDs say that connecting with a peer supporter saved them from a sense of growing desperation born out of a mounting shame.<sup>35,59,61-62</sup>

## TIMING OF SURGERIES

It is rare that surgeries will be needed in the neonatal period. Naturally, operations performed to end an imminent threat to the patient's health should be scheduled as quickly as possible, and explained to the parents and, if of an appropriate age, to the child. Examples include operations to create a urinary opening where none exists in a newborn, or to remove malignant tissue.

Genital cosmetic surgeries are sometimes offered to relieve parental distress,<sup>80</sup> but parental distress should instead be addressed directly through peer support and competent mental health care. In providing this care for parents, teams show respect for parents and their children.<sup>38,81</sup> A combination of reassurance and education will help to reduce the family's early negative reactions to the condition while allowing them to honestly discuss their concerns and questions.<sup>38,82</sup>

Past practice favored the use of surgery to reinforce initial gender assignment.<sup>19,83-86</sup> This included operations aimed at making genitalia look more cosmetically normal and the removal of gonadal tissue at odds with the initial gender assignment. For the following reasons, the emerging approach<sup>2,36,87</sup> calls for delaying elective surgeries until the patients themselves can participate in decision-making:

1. Gender assignment is an imperfect art;<sup>48,88-91</sup> a small but significant number of patients with DSDs will develop a gender identity at odds with their initial gender assignment, and some will grow to feel and express nontraditional gender identities.<sup>67,73-74</sup> It is best to let patients decide for themselves what anatomical features accord with their self identities. Professional counseling by a mental health professional can help patients make these decisions.<sup>92</sup>
2. In both autobiographical accounts and outcome studies, a significant number of former patients have reported diminished sexual sensation, sexual dysfunction, or chronic pain following genital operations, including operations (e.g., "nerve-sparing") thought by their advocates to be low risk.<sup>15,26,37,61,72,93-100</sup> Surgery to construct a neovagina carries a risk of neoplasia.<sup>101-102</sup> Because all surgeries carry risk, and because sexual sensation and function is vital not only to an individual's enjoyment of sexuality but to his/her capacity for forming and maintaining intimate relationships and pair-bonds, it is preferable to allow patients to decide for themselves which risks to assume.
3. Operations designed to normalize genital appearance may undermine the multidisciplinary team's central message to the parents that the child is unconditionally acceptable and lovable.<sup>34-35</sup>
4. There is a consistent and growing body of evidence that children raised with "ambiguous" sex anatomy are at no greater risk for psychosocial problems than the general population.<sup>23,28,61,103-109</sup> Meanwhile, there is surprisingly little published evidence to the contrary.<sup>110</sup> As a consequence, there is a lack of demonstrated need for early cosmetic genital surgeries. Interventions have tended to be based on fears about "worst case scenarios," not demonstration of medical need.

5. Allowing a patient to make decisions about elective care signals to the patient a fundamental valuing of his or her autonomy and personhood.<sup>32</sup>

Healthy, functioning gonadal tissue should remain in place unless the patient, fully advised of risks and options, requests it be removed. Improving reproductive technologies may make it possible for patients now considered infertile (e.g., women with CAIS) someday to contribute to procreation (e.g., through sperm aspiration, IVF, and surrogacy). Removal of healthy gonadal tissue leads to loss of potential fertility and the loss of the benefits of endogenous hormones (e.g., prevention of osteoporosis; many women with CAIS report a loss of libido and sense of well-being after gonadectomy), and should therefore happen at the will of the fully informed patient. Note that gonadal tumor risk varies with etiology; the risk is highest in PAIS, and lowest (< 5%) in CAIS and ovotestes.<sup>111-113</sup>

There is a lack of agreement on the recommended age for various treatments and this inevitably influences the ability of affected children to participate in decision-making. A formal assessment of the child's cognitive status by a child psychologist or psychiatrist can assist in determining the extent to which the child is capable of participating in the decision-making process.

## HELPING PARENTS THINK ABOUT ELECTIVE SURGERY

Parents pick up signals from their children's pediatricians about what they should do, sometimes picking up signals pediatricians don't intend.<sup>114-115</sup> The best thing pediatricians can do is to be explicit with parents (and, as age appropriate, patients) about the choices available and the evidence regarding outcomes for each.

If there's no urgent need for surgery—as is the case for many DSD surgeries—clinicians can make vivid the option of waiting by introducing the family to adults who have lived without the intervention being considered and to other parents who have decided to hold off and let the child decide. So, when working with parents considering clitoral reduction surgery for their infant daughter, one pediatric urologist has introduced those parents to another set of parents who chose against infant clitoral reduction surgery—and to the daughter herself (now an adolescent). This way the parents facing a decision have had the opportunity to meet a child who will make the elective surgery decision for herself, and to learn from her and her parents how to manage the psychosocial challenges of raising a girl with a noticeable clitoris.

Pediatric urologists and surgeons know well that sometimes parents seek early elective surgeries for their child with a DSD because they think it will spare the child the psychological trauma of having them done later. This is the case sometimes, for example, with infant orchidectomies chosen on behalf of girls with Complete Androgen Insensitivity Syndrome. But many adults with DSDs have said that they were not spared the psychological trauma of the surgery because it hit them full-on when they came to know about the surgery. Some, in fact, have felt the psychological trauma of surgery was amplified by having had surgeries done without their consent. This in turn increased their sense of betrayal and disenfranchisement, an outcome unintended by their parents and doctors, who made the decision with the best of intentions.

## TIMING OF HORMONAL THERAPY

Endocrinologic therapy administered to avoid an imminent threat to the patient's health should be provided and explained to the parents and, if of an appropriate age, to the child. The most obvious example would be treatments for classical CAH.

For the following reasons, the emerging approach calls for delaying elective sex hormone treatments until the patients themselves can participate in decision-making:

1. In general, elective sex hormone treatments can wait until the child is approaching the age of puberty when the patient can and should participate in informed decision-making. The administration of sex hormones can result in physiologic and behavioral changes discordant with the developing self-identity of the patient.<sup>116-117</sup> For this reason, it is best to have qualified mental

health professionals assess the patient's identity and maturity and suggest options. (Psychiatrists and psychologists can do this assessment and then work with the endocrinologist to suggest options.) Allowing a patient to make decisions about elective care also signals to the patient a fundamental valuing of his or her autonomy and personhood.

2. Exogenous hormones carry risks; for example, testosterone treatments can result in prostatic hypertrophy, reduced fertility, changes in libido, acne, male pattern balding, and high blood pressure. It is best to wait and to allow patients to decide for themselves which risks to assume and when to alter or cease elective treatments.

## REDUCING STIGMA RELATED TO MEDICAL CARE

Adults with DSDs report having been inadvertently subjected to contexts productive of stigma within medical institutions where they were treated as children.<sup>62,97</sup> It is crucial that multidisciplinary teams avoid directly or indirectly stigmatizing patients with DSDs and their families.<sup>2</sup> The following practices should be observed.

- Use verbal and body language that signals openness and acceptance of the child and the parents; be calm, reassuring, and honest<sup>43</sup> (including being honest about uncertainty).
- Use gender-neutral language when gender is uncertain, but **never use objectifying language such as “it”**; instead employ phrases like “your baby” or “your child” or “your little one.”<sup>49,60</sup>
- Encourage parent/child attachment and bonding.
- Keep in mind that, while medically indicated, close inquiry about parental history inadvertently produces feelings of parental responsibility and guilt about the DSD; avoid unnecessarily repeating these questions and explain the medical reason for such questions.
- Talk with team members and the family about the child and family's strengths,<sup>118</sup> so that the challenges posed by the DSD are kept in perspective.
- Provide the patient and family routine opportunities to talk about both their hopes and fears (i.e., provide proactive support).
- Talk **with** the child as soon as he or she is capable of understanding that he or she is being talked **about**; this way patients don't accidentally feel like objects in the room.
- Avoid imparting the message that the child is a curiosity or freak by reducing to a minimum the number of health care professionals who physically attend to the child and family.<sup>61</sup>
- Avoid whenever possible repeated genital exams, especially those involving measurement.

- Treat gender atypical traits of the child as acceptable for that child.<sup>119</sup>
- Limit medical photography<sup>30</sup> to what is absolutely necessary for the patient's care and (as HIPAA requires) do not allow photographs to leave the patient's chart; adults with DSDs have reported lifelong harm from finding their own nude pictures in medical texts or on the internet, and from being unable to obtain copies of photographs that they know should have been kept in their records.
- Make sure the child knows the truth as he or she grows up; this will prevent the harmful situation where he or she learns the truth in a confusing and inadequate way, for example, by a slip of the tongue or a glance at the medical chart.<sup>44</sup>
- Let the patient know he or she is not wholly defined by his or her diagnosis.
- Provide access to peer support and counseling.<sup>79</sup>

Health care professionals should keep in mind that families look to them for guidance; they should monitor their words, movements, and actions carefully to be sure they are fully supporting the patient and family in their unique experience.

Encouraging parents to talk with others, right from the start, will forestall closeting and the growth of shame in the family. For example, families who have been through gender changes suggest that their experiences were made easier by letting trusted family and friends know early on about their children's DSDs.

## EMPOWERING CHILDREN WITH DSDs

A number of adults with DSDs recall being greatly affected, in both positive and negative ways, by how their specialist doctors worked with them. One psychologist who has had extensive contact with children and adults with DSDs offers these thoughts: "Particularly crucial is the child's need to be specifically acknowledged and asked for feedback about an experience, permission for an exam, etc. One adult I spoke to recently talked about what an enormous difference it made for her when a doctor made a joke at his own expense, which made him vulnerable and left her feeling 'seen' as an individual. Another wished someone had told her it was okay to yell and be angry when something hurt. Simply explaining what you're doing will certainly help, but my contacts seemed to need something more, help in finding a voice, permission to become empowered."

## TELLING THE TRUTH

Telling patients the truth about their medical histories and conditions promotes a trusting doctor-patient relationship, signals openness (the opposite of shame), reduces the sense of stigma, and enables patients to understand the health and quality-of-life benefits of adherence to medical care. Patient-centered care by definition cannot occur in the absence of truth-telling. Truth-telling needs to be geared to the child's maturity and is most likely to succeed when it is accomplished in the context of a trusting patient-doctor relationship, one in which an atmosphere of trust has been fostered. In such circumstances, children feel comfortable asking questions and answering questions.

When clinicians are telling the truth to patients, the parents should be present so that there is no misunderstanding. The explanations should be geared to the developmental level of the child and the questions being asked. Full disclosure of the medical facts should typically occur at the latest by age 16 in cooperation with the parents and mental health members of the team. When a patient goes to obtain his or her records at age 18, he or she should find no surprises.

## WHAT'S THE TRUTH?

Telling patients and parents the truth doesn't just mean avoiding outright lies; it also means not withholding critical information like karyotype, diagnosis, and crucial facts of a medical history. When clinicians try to protect patients through euphemisms or by withholding information, they may inadvertently harm the patient and the doctor-patient relationship.

Many physicians were trained in the past to refer to the testes of patients with AIS as "gonads," believing that full knowledge of their AIS and testes would interfere with AIS patients' psychosexual development.<sup>43</sup> This led to much hardship for girls and women with AIS who found out the truth by accident.<sup>44</sup> One peer supporter with AIS relates the following stories: "One girl told me her chart was left in the room, and so she peeked at it and thought it read 'Patient does not know she has AIDS' (when actually it said 'patient does not know she has AIS'). Another told me she was kibitzing with an intern about sex chromosomes, not knowing she had XY chromosomes, when the intern misread the situation and suddenly blurted out to her, 'Ha! Well you have XY!' A third overheard staff commenting 'Hey, we've got a hermaphrodite in Exam Room 2!'" Some introductory biology and genetics classes have included a lab in which buccal smears are performed, revealing the students' karyotypes. Some patients have been permanently alienated from health care professionals and emotionally scarred by these kinds of experiences.

Parents who want providers to withhold information from their children may need support for their own concerns. They should also be advised that once children turn 18, they will be entitled to their full medical records, and that, even before that age, children have a way of picking up on family secrets and lies. Parents should be offered professional counseling in how to speak the truth with their children.

## THE IMPORTANCE OF SEXUAL WELL-BEING

Although it often makes adults uncomfortable to think about the future sexual lives of children, it is very important that attention be paid to ensuring that patients with DSDs grow up to feel sexually healthy, mentally and physically. In the past, too much attention has been paid to genital appearance and gender identity at the expense of the patient's sexual health.<sup>26,120-121</sup> Sexual health is central to an adult's over-all well-being; it is critical to the establishment of short- and long-term intimate relationships and pair bonding, but also more generally to the establishment of a positive sense of self.<sup>122</sup>

Patients have reported that genital surgeries (with resultant scars), gonadectomies, exogenous hormones, being lied to or misled by health care professionals, and repeated genital examinations have contributed to sexual dysfunction and the cascade effects that come from sexual dysfunction (especially difficulties establishing and maintaining partnerships). Clinicians should use the techniques mentioned above

(including actions designed to reduce stigma and maximize patient decision-making) to reduce threats to patients' sexual well-being.



## Chapter 4 Scripts for Talking with Parents

Listed below are examples of questions commonly asked by parents who have just recently become aware of their child's DSD, and answers clinicians can provide. (Substitute the child's name for "your baby" or "your child" if a name has been given.)

**Q: Is my child a boy or a girl?**

**A:** Your question is very important. We wish we could tell you right this minute, but we really can't tell yet. We will have more information after we conduct some tests. It's hard for parents to wait for these test results so we will try to update you every day, and you can call [give contact person's name] at anytime. Although your baby has a condition you probably haven't heard much about, it isn't that uncommon. We've encountered this before, and we'll help you through this time of confusion. As soon as the tests are completed, we will be able to talk with you about the gender in which it makes most sense to raise your child, and we'll give you a lot more information, too, since quite a lot is known about these variations and we are learning more each day. We want to reassure you that our focus is on supporting you and your child in this time of uncertainty.

**Q: What if you assign the wrong sex?**

**A:** After we have back the information we are collecting, we will talk with you more specifically about choosing a gender assignment. We suggest you choose the gender assignment your baby is most likely to identify with as your baby grows up and becomes his or her own person. We can't guarantee we'll get it right. Most of the time we do. But if your baby grows up and as a child or a teenager decides that her or his assigned gender—the one you are going to give your baby now—doesn't fit for your baby anymore, we will be able to help your child, and all of you, adjust to this.

If this unlikely event happens, what other families have told us is that it's important to respect the gender identity your child expresses. It is much too hard to live being seen as a girl if you're a boy, or vice versa. We here at the hospital will keep in touch with you; we're going to provide you care as long as you need us. In the unlikely event your child wishes to change gender, we will all work together to help you and your child make the gender transition. Remember, we have peer counselors, other parents and adults with similar experiences you can talk to. These are people who've been down the same road as you.

We're not saying it's easy, but sometimes it does happen and parents tell us that with counseling, support, and talking to other parents and their kids, everyone gets through it. And your child will always be your child, and you'll always be your child's parent. Gender won't matter to that. (When a child is physically healthy, more reassurance may be provided regarding the physical health of the child. This often helps parents put the DSD in perspective.)

Tell me, what are your thoughts now about your child's gender? (Remember that parental feelings about a child's gender identity are a critical factor in gender assignment. The parents will be the ones to live with the gender assignment and thus will actively contribute to the child's experience of gender development.)

**Q: Can you perform surgery to make my child's genitals look and function normally?**

A: If your child requires surgery for matters of physical health, of course we'll recommend and provide that surgery. Otherwise we're going to recommend delaying genital surgeries until your child is old enough to participate in such a decision. Surgical treatments carry specific risks and the long-term outcomes of today's genital surgeries in children have not been well studied. Waiting to make decisions about surgery has many potential benefits and few downsides. A few examples of benefits are that your child's health will not be affected and the direct benefit of surgery can be better understood when he or she is older. On the other hand, the risks taken under any circumstances when surgery is performed must not be underestimated. Having surgery can also be stressful and traumatic. When surgery is done, it's important that the patient have a chance to understand to the best of his or her ability what can and cannot be accomplished. This can be even more important when the benefit of the surgery is individual and related to appearance.

**Q: What do we tell our friends and family while we wait for the gender assignment?**

A: This is important. We strongly recommend being open and honest about your child's situation. Even if you don't intend to, lying or withholding information will create a sense of shame and secrecy. Though it can be awkward to talk with family and friends about a child's sex development, being honest signals that you are not ashamed—because you have nothing to be ashamed of—and it also allows others to provide you with the love and support you may

need. Isolating yourself at this time will probably make you feel unnecessarily stressed and lonely. Talking about it will help you feel connected with others.

In the beginning of this process, you may feel overwhelmingly emotional when you talk about your child's situation. The team will give you many opportunities to talk about your reactions and to come up with a way to share this information with family and friends. It is our experience that parents are proud of their children and do not intend to act as if they are ashamed or embarrassed by their conditions. But when they find themselves not able to openly or honestly talk about their child, over time it can magnify feelings of shame for the child. More importantly, children diagnosed with DSDs also develop feelings of shame if the topic is avoided, simplified, or continually redefined. We understand that developing these skills and establishing your comfort level will take time and support. Providing help for you in a way that is tailored to your needs and the needs of your child and family is what the team is all about.

So here is what you can tell people: Our baby was born with a kind of variation that happens more often than you hear about. Our doctors are doing a series of tests to figure out whether our baby is probably going to feel more like a boy or a girl. We expect to have more information from them within [specify realistic timeframe], and then we'll send out a birth announcement with the gender and the name we've chosen. Of course, as is true with any child, the various tests the doctors are doing are not going to tell us for sure who our baby will turn out to be. We're going to go on that journey together. We appreciate your love and support and we're looking forward to introducing you to our little one in person soon.

It also helps to let your friends and family know whether your baby is healthy or whether there are some health concerns. Finally, take some pictures of your baby's face and share those pictures with others!

We think you'll encounter what other parents we've worked with have experienced, that family and friends usually have many questions and lots of advice. You can have us meet with family if you like. Also, you might find it helpful to talk with one of our parent peer counselors. We know that this isn't an easy road to walk down at first, but you're not the first to walk down this road, and you won't walk it alone. We want to help you.

**Q: Is my child going to be gay?**

**A:** Many parents wonder about this. There really isn't any way to predict **any** child's future sexual orientation, and your baby is no different. What we do know is that your child is always going to be your child, and that this child is very lovable. The most important thing you can do is to take care of yourself and to provide this child love and honesty, and to have faith that he or she is going to do well in the world.

## ADDRESSING HOMOSEXUALITY

The historical record suggests that clinicians treating patients with DSDs have sometimes been motivated by a desire to avoid the appearance of homosexuality in those patients. Parents have sometimes shared that motivation. It is understandable that in the era of anti-sodomy laws and vice squads, physicians and parents were concerned for the well-being of patients who appeared to be homosexual.

Today specialists treating DSDs generally agree that a patient's sexual orientation should not be the measure of clinical success. Being gay, lesbian, or bisexual is a less common but nonetheless healthy outcome.

That said, clinicians and parents should keep in mind that patients (especially children) will often get the message from those around them that they should feel doubly guilty or ashamed if they were born with a DSD and are gay, lesbian, or bisexual. Out of this guilt or shame, they may assent to therapies they do not really want or need because the child may attempt to compensate for these "failures" or disappointments to parents by being overly compliant with the perceived wishes of family or care givers. Having a psychologist, psychiatrist, or social worker develop a good rapport with the patient may help avoid this problem. Mental health professionals can help assess whether patients are assenting to care for problematic reasons.

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## SUPPORT GROUP INFORMATION

Disclaimer: Although all of the following groups have useful information and contacts to share, this list should not be construed as a blanket endorsement of their work. Furthermore, the list is not exhaustive, and support group contact information changes frequently: websites may be more accurate than mailing addresses; consult [www.accordalliance.org](http://www.accordalliance.org) for the latest information.

### **Androgen Insensitivity Syndrome Support Group (AISSG)**

PO Box 2148 Duncan, OK 73534-2148

[www.aissgusa.org](http://www.aissgusa.org)

### **Androgen Insensitivity Syndrome Support Group Canada (East)**

#206, 115 The Esplanade Toronto, Ontario M5E 1Y7 Canada

email: [sallie AT ican DOT net](mailto:sallie@ican.net) (English)

email: [orquideequebec AT yahoo DOT ca](mailto:orquideequebec@yahoo.ca) (French)

### **Androgen Insensitivity Syndrome Support Group Canada (West)**

#17, 3031 Williams Road Richmond, B.C. V7E 1H9 Canada

email: [lesnick AT shaw DOT ca](mailto:lesnick@shaw.ca)

### **CARES Foundation** Congenital Adrenal Hyperplasia Education & Support

189 Main Street, 2nd floor Millburn, NJ 07041

[www.caresfoundation.org](http://www.caresfoundation.org)

**Hypospadias & Epispadias Association (HEA)**

240 W. 44th St. Suite 1A New York, NY 10036

[www.heainfo.org](http://www.heainfo.org)

**Klinefelter Syndrome and Associates**

11 Keats Court Coto de Caza, CA 92679

[www.genetic.org/ks/](http://www.genetic.org/ks/)

**The Magic Foundation**

6645 W. North Avenue Oak Park, IL 60302

[www.magicfoundation.org](http://www.magicfoundation.org)

**MRKH Organization**

P.O. Box 301494 Jamaica Plain, MA 02130

[www.mrkh.org](http://www.mrkh.org)

**Turner Syndrome Society of the U.S.**

14450 TC Jester, Suite 260 Houston, TX 77014

[www.turner-syndrome-us.org](http://www.turner-syndrome-us.org)

**XY Turner**

Box 5166 Laurel, MD 20726

[www.xyxo.org](http://www.xyxo.org)

Please visit the Accord Alliance website ([www.accordalliance.org](http://www.accordalliance.org)) for the latest information.



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