

Disorders of sexual development: What you need to know

Disorders of sexual development (DSD) are a series of conditions that affect a person's genitals or reproductive organs. In this handout, you will learn about DSD, including the different types of DSD, symptoms, diagnosis and treatment.

WHAT ARE DISORDERS OF SEXUAL DEVELOPMENT (DSD)?

Babies who are born with incompletely developed external genitals or with external genitals which do not match their internal organs are said to have **disorders of sexual development (DSD)**. DSD are considered **congenital problems** (problems present at birth) of development. DSD can have many causes.

WHAT ARE THE SYMPTOMS OF DSD?

Babies born with DSD can have the following symptoms:

- **Genitals that can look different at birth.** They may look like boys who have a poorly developed or unfinished penis or like girls with a large clitoris. They may also look like typical girls who have a sex gland that turns out to be a testicle.
- **No outside sign of genitals at all.**
- **Problems with the adrenal glands.** These children could become sick about a week after birth because they cannot make enough important adrenal hormones called cortisol and aldosterone. These hormones control how the body responds to stress and stores salt and water.
- **Other congenital issues with development.** There are rarely serious problems in the rest of the body. This can range from kidney problems, brain development problems and problems with development of the arms, legs, hands or feet.

WHAT ARE THE CAUSES OF DSD?

There are many causes of DSD. Not all causes have yet been found. They can include:

- Problems with the adrenal glands
- Problems making various male hormones or the body's inability to respond to these hormones
- Genetic problems with the development of the sex glands (either ovary in most girls or testicle in most boys)

HOW DO DOCTORS DIAGNOSE DSD?

Doctors can diagnose DSD in one or more of the following ways:

- **Physical exam.** In the beginning, doctors will do a careful physical exam of your child's body and genitals.
- **Family history.** The doctors will ask the mother and father about other children in the family with similar problems and any exposures to chemicals before birth.
- **Check the development of your baby's chromosomes (pieces of genetic material).** Chromosomes are usually checked with a blood test or a cheek scraping (when doctors gently swipe a cotton swab against the inside of your baby's cheek and check it under a microscope).
- **Blood tests** to check different blood chemicals related to adrenal gland development and development of the sex glands. This can help identify conditions like congenital adrenal hyperplasia (CAH), which may require rapid treatment.
- **Imaging tests** to check the anatomy of the outside and inside genitals. This can include X-rays, ultrasounds or a special exam by a trained doctor.
- **Genetic testing** can check all the genes in the body and see if any are likely to have caused DSD. Results from genetic testing can take days to months to come back, depending on the test type or results.

HOW DO DOCTORS TREAT DSD?

Treatment for DSD is based on what is causing the condition. You and your family are an important part of your baby's care team. Doctors will talk with you about your baby's condition and their sex of rearing. The **sex of rearing** is the sex assigned to your baby before they go home. The care team can help you make the decision about your baby's sex. This does not have to be a permanent decision. As babies grows up, they can learn how to express their gender identity.

Many babies grow up to be comfortable with their sex of rearing. However, some babies with DSD decide they might be more comfortable with a different gender when they are older. Emotional and psychological support from you and your baby's care team is important no matter what decisions are made.

Depending on the reason for DSD, some people may not be fertile because their bodies cannot make eggs or sperm. However, this does not interfere with the ability to lead a good life, to marry (if wished), adopt children (if wished) or have children by other means. If you have questions or concerns about your baby's fertility when they are older, please ask the care team.

About your baby's care at MGHfC

When babies with DSD are born, it takes a well-organized team of doctors and other medical providers to understand why DSD has happened and provide the best care possible. In some cases, people with DSD receive diagnoses long after birth. At MassGeneral Hospital for Children (MGHfC), our team-based, family-centered approach can help manage your baby's condition and help them have the best possible life. Throughout their life, your baby might see many specialists, including endocrinologists (hormone doctor), geneticists (genetics doctors), specialized surgeons (who can help treat physical genital differences), psychiatrists, psychologists (mental health doctors) and social workers. At MGHfC, you and your family are also important members of the care team.

The sex determined by the chromosome is also important. Babies who have XX chromosomes usually have different reasons for DSD than those who have XY chromosomes. Based on your baby's test results, they can help develop a care plan.

What are chromosomes and how do they work?

Chromosomes are pieces of genetic material that contain all the information needed to control development and body function throughout life. Most girls have 2 X chromosomes (XX) and 46 chromosomes in total. Most boys have one X and one Y chromosome (XY) with 46 chromosomes in total. The X and Y chromosomes are called sex chromosomes. The other chromosomes are called autosomes. If there is a problem in how the sex chromosomes or other autosomes develop, doctors can often find these through genetic testing.

WHERE CAN I LEARN MORE ABOUT DSD?

- **The care team at MGHfC**

- **Accord Alliance**

www.accordalliance.org

An organization dedicated to helping people and families with DSD. They have an excellent handbook for parents which is available online.

- **Cares Foundation**

www.caresfoundation.org

Dedicated to helping people and families with congenital adrenal hyperplasia.

- **Magic Foundation**

www.magicfoundation.org

Help and information for many different disorders of the endocrine glands including congenital adrenal hyperplasia.

- **Androgen insensitivity and DSD Support group**

www.aisdsd.org

Patient, family support group with medical advisors dedicated to helping people with various disorders of sexual differentiation

Pediatric Endocrine and Diabetes Center

Mass General Hospital for Children

55 Fruit Street, Suite 6C

Boston, MA 02114

www.massgeneralforchildren.org/endocrine

For more information please call

617-726-2909

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