

Disorders of Sex Development

What are disorders of sex development?

Disorders of sex development (DSDs) are conditions in which the reproductive organs and genitals do not develop normally. People with DSDs have a mix of male and female sexual characteristics.

There are different types of DSDs:

- A person may be genetically female (they have two X sex chromosomes), but have genitals that appear more masculine. They may, for example, have an enlarged clitoris that resembles a penis and their labia (lips surrounding the vagina) may be fused together. The most common cause for this type of DSD is a metabolic condition called congenital adrenal hyperplasia (CAH), which also affects other aspects of health.
- Another type of DSD occurs when a person is genetically male (having one X and one Y sex chromosome), but the genitals appear more female. This type of DSD is most often caused by complete or partial androgen insensitivity syndrome (AIS), which causes the body to respond abnormally to male hormones.
- People with male, female, or mixed chromosomes can be born with both ovarian and testicular tissue. In this type of DSD, the genitals can appear female, male or a mix of both.
- Sometimes sexual organs are normally formed as either male or female, but growth and sexual development are disrupted at puberty. Individuals with this type of DSD have an abnormal chromosome arrangement, such as a missing or extra X chromosome. Females with a DSD called Turner syndrome, for example, are missing an X chromosome and are usually infertile and of shorter than average height.

What causes disorders of sex development?

DSDs occur because of variations in the genes that control sexual development or because a person has an underlying condition that affects how the body responds to sex hormones. For example, people with a DSD caused by congenital adrenal hyperplasia (CAH), do not produce an enzyme required to make two hormones, cortisol and aldosterone. As a result the body produces too much androgen (male sex hormone), resulting in male sex characteristics. If the DSD is caused by androgen insensitivity syndrome, the body does not process androgen in a normal way.

How are disorders of sex development diagnosed?

If sexual organs are ambiguous at birth, a child may receive a diagnosis right away. Other types of DSD do not become apparent until puberty. Partial androgen insensitivity syndrome, for example, is usually diagnosed at birth because the genitals appear different. In complete androgen insensitivity syndrome, the genitals look normal for a female, so the condition may not be picked up until a girl fails to menstruate.

When a DSD is suspected, tests are used to confirm the diagnosis. These include chromosome analyses, blood tests to measure hormone levels, and imaging studies to examine internal sex organs.

How are disorders of sex development treated?

It is important that a child with a DSD be carefully evaluated at a multidisciplinary clinic that specializes in these disorders. Pediatric endocrinologists, urologists, psychiatrists and other specialists work together to present treatment options. These may include medications, such as supplemental hormones, to encourage pubertal development and manage other growth and development problems. Surgical reconstruction of the genitalia may also be an option, although it is not necessary in all cases. With proper diagnosis and treatment, most individuals with DSDs live normal lives.

Disorders of Sexual Differentiation (DSD) Clinic

At UCSF we have a unique multidisciplinary clinic that provides care to families of children with ambiguous genitalia. Our experts include urologists, endocrinologists, reproductive endocrinologists, geneticists, child psychologists, a mental health nurse practitioner, nurses and social work professionals. The team meets with families to discuss diagnosis, management, goals and questions, and concerns surrounding genital surgery. The entire DSD team meets on the third Tuesday of every month to collaborate with individual families to provide a management plan for each child's medical and psychological needs. Angelique Champeau, CPNP is the clinic coordinator. Please contact us 415 353-2083 to schedule an appointment.

Support groups, such as the Cares Foundation (<http://www.caresfoundation.org>) also provide help and support to young people, adults and families who are affected by CAH.

See the next page for contact information.

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Appointments & Location

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