CONDITION

Differences of sex development

Differences of sex development are inborn variations in sex chromosomes, hormones, reproductive organs, or genital anatomy.

WHAT IT IS
Congenital variations in chromosomes, hormones, gonads, or reproductive anatomy

BODY SYSTEM
Reproductive and hormone systems

MAIN SYMPTOMS
Atypical genitals, delayed puberty, absent periods, fertility difficulties

WHAT CAUSES IT
Genetic, chromosome, hormone-production, or hormone-response differences

HOW IT IS DIAGNOSED
Examination, hormone tests, genetic tests, and imaging

MAIN TREATMENT
Individualised monitoring, hormone care, emotional support, sometimes surgery

CAN IT BE CURED?
Sometimes - some effects treatable, underlying variation usually remains

IS IT AN EMERGENCY?
Sometimes - vomiting and dehydration may signal adrenal crisis

LONG-TERM OUTLOOK
Most people live healthy, full lives with tailored care

Overview

Differences of sex development, often shortened to DSD, are inborn variations in how sex characteristics develop. These characteristics include chromosomes, hormones, gonads and reproductive anatomy. Gonads are the organs that usually develop into ovaries or testes.

DSD is an umbrella term covering many variations with different causes and health effects. Some are noticed at birth. Others are found during puberty, fertility testing or unrelated medical care.

Some people use the word "intersex." Others prefer "variation of sex characteristics," a specific diagnosis or no label. Older records may use "disorder of sex development." Language is personal and should reflect the person's preference.

Having a DSD does not necessarily mean someone is unwell. Some types can affect hormones, puberty, fertility or future health risks. Rarely, a newborn may develop a dangerous shortage of adrenal hormones.

Sex characteristics do not determine gender identity, which is a person's internal sense of their gender.

Symptoms

Signs depend on the specific variation and the person's age. Some people have no symptoms or need no treatment.

Possible signs in a newborn include:

Certain forms of congenital adrenal hyperplasia (CAH) may cause poor feeding, repeated vomiting, dehydration, unusual sleepiness or poor weight gain in the first weeks of life. CAH affects adrenal hormones needed for blood pressure, salt balance and the response to stress.

Signs in childhood or adolescence may include:

In adults, a DSD may be found during investigation of absent or irregular periods, low hormone levels, sexual difficulties or infertility. These concerns also have many other possible causes.

- Genital anatomy that differs from usual male or female patterns
- A penis that is smaller than expected or has an opening underneath
- Testes that cannot be felt in the scrotum
- Fused or enlarged genital tissue
- A difference between prenatal chromosome results and genital appearance
- Puberty starting earlier, later or differently than expected
- Periods not starting
- Breast development or facial and body hair differing from expectations
- Unexpected genital changes at puberty
- Groin swelling caused by a gonad in a hernia
- Concerns about height or growth

Causes and risk factors

Sex development before birth involves many genes and hormones. A difference at any stage

can affect chromosomes, gonad development, hormone production, hormone response or reproductive anatomy.

Examples include:

Some DSDs are inherited from one or both biological parents. Others result from a new genetic or chromosome change. Testing does not always identify a single cause.

DSDs are generally not caused by anything a parent did during pregnancy, parenting, sexual activity or gender identity. Family history can increase the likelihood of certain inherited types, but many people have no known family history.

- Sex chromosome variations: A chromosome pattern other than the usual XX or XY pattern, or different patterns in different cells
- Congenital adrenal hyperplasia: An inherited enzyme difference affecting adrenal hormones; some forms increase androgen production
- Androgen insensitivity syndrome: The body responds partly or not at all to androgens
- Differences in gonad development: Ovarian or testicular tissue develops differently or produces different hormone levels
- Differences in hormone conversion: The body does not convert one sex hormone into another in the usual way

Diagnosis

There is no single test for every DSD. Diagnosis may combine physical findings, hormone tests, imaging and genetic information. The care team may include endocrinologists, genetic specialists, urologists, gynaecologists, psychologists and specialist nurses.

For a newborn, the first priority is checking for problems needing prompt treatment. Blood tests may measure body salts, blood sugar and adrenal hormones, particularly if CAH is possible.

Further assessment may include:

Genetic testing may identify a diagnosis, but results can take time or remain inconclusive. If a baby is medically stable, decisions about naming or sex assignment should be discussed carefully rather than rushed.

Older children and adults should receive age-appropriate, honest information and be involved in decisions. Privacy is important, but secrecy can cause distress and make future care harder.

- Examination and medical history

- Blood or urine hormone tests
- Chromosome analysis and genetic testing
- Ultrasound or other imaging of internal reproductive organs
- Tests of adrenal or gonad function

Treatment and management

Treatment depends on the diagnosis, health needs and the person's goals. Not everyone needs medical treatment.

People with an adrenal hormone shortage may need lifelong hormone replacement. They may also need extra medicine during serious illness, injury or surgery, when the body would normally produce more stress hormones.

Hormones may be offered to support puberty, bone strength, menstrual health or general wellbeing when the ovaries or testes do not produce enough. Choices may change as a child grows and becomes more involved in decisions.

Surgery is not required for every DSD. It may be considered for blocked urine flow, repeated infections, painful anatomy, a hernia or an identified tumour risk. Some procedures are mainly intended to change appearance.

Irreversible, non-urgent procedures require careful discussion of benefits, limitations, sexual sensation, fertility and whether the decision can safely wait until the person can participate.

Other care may include:

Care should respect the person's privacy, language, gender identity and right to participate in decisions.

- Emotional and psychological support
- Help with body image, relationships or sexual wellbeing
- Fertility assessment and reproductive counselling
- Peer support
- Monitoring of hormones, growth, bones or gonadal tissue

Complications

Possible complications vary by diagnosis, and many people experience few or none.

Some forms of CAH can cause an adrenal crisis: a severe shortage of essential adrenal hormones. This may cause dehydration, low blood pressure, low blood sugar and dangerous changes in body salts.

Low sex hormone levels can affect puberty and weaken bones over time. Monitoring and hormone treatment may reduce this risk.

Fertility may be reduced because of differences in gonads, reproductive organs or hormones, but infertility is not universal. Some people conceive without treatment; others use assisted reproduction or different routes to parenthood.

Some DSDs increase the risk of tumours in gonadal tissue. Risk varies greatly according to the diagnosis, age, gonad location and genetic findings. Monitoring or surgery may be discussed when a specific risk is identified.

Stigma, secrecy, unwanted attention and repeated examinations can affect mental health. Previous surgery may cause scarring, reduced sensation, pain, or urinary and sexual difficulties. Care should address emotional and social wellbeing as well as physical health.

Prevention

Most differences of sex development cannot be prevented because they arise before birth through genetic, chromosome or hormone-pathway variations.

If an inherited cause is identified, genetic counselling can explain how it may be passed through a family and discuss testing or reproductive options. It provides information and support; it does not mean anyone is responsible.

Early recognition cannot prevent a DSD, but it can identify urgent hormone problems and ensure appropriate follow-up.

Living with it and what to expect

The long-term course varies. Some people need only an initial assessment and occasional review. Others need lifelong hormone replacement or regular monitoring of growth, bones, adrenal function or gonadal tissue.

Different life stages may bring new questions. Puberty can raise issues involving hormones, body changes and gender identity. In adulthood, sexual health, fertility, relationships and pregnancy may become more important.

A diagnosis may become more precise as genetic testing improves. This usually means

medicine can better explain the original variation, not that the person's body has changed.

Most people with a DSD attend school, work, exercise, form relationships and lead full lives. Some have ongoing medical needs; others mainly need reliable information and access to support.

A planned move from children's to adult services should preserve medical records, review medicines and monitoring, and identify who will provide future hormone, reproductive or psychological care.

When to get medical help

Seek emergency care for a baby or person with possible adrenal hormone problems who has:

Sudden severe lower abdominal or groin pain also needs urgent assessment. In some people, this may mean gonadal tissue has twisted and lost its blood supply.

Arrange prompt medical assessment if a newborn's genital appearance differs from expected patterns, even if the baby seems well. Clinicians can check hormones and body salts without assuming there is an emergency.

Seek routine medical advice if puberty is not progressing as expected, periods have not started, groin swelling develops, or there are fertility or hormone concerns. Support is also available if medical visits, body concerns, disclosure or identity uncertainty are causing distress.

- Repeated vomiting or severe diarrhoea
- Poor feeding with unusual sleepiness or weakness
- Signs of dehydration, such as very few wet nappies or little urine
- Fainting, confusion, collapse or difficulty waking
- A known adrenal condition and inability to keep emergency medicine down

Common questions

Is DSD the same as intersex?

The terms overlap, but they are not identical for everyone. DSD is commonly used in healthcare. Some people identify as intersex; others prefer a specific diagnosis or another term. Use the language the person chooses.

Is a difference of sex development a disease?

Not necessarily. DSD describes variations in sex characteristics. Some require treatment because they affect hormones or other health needs. Others do not cause illness.

Can doctors tell a baby's sex right away?

Not always. Hormone, chromosome, genetic and imaging results may be needed. Unless there is an urgent medical problem, careful assessment is safer than rushing major decisions.

Does DSD determine someone's gender identity?

No. Chromosomes, genital anatomy and hormone patterns do not reliably predict gender identity. A child's identity develops over time and should be listened to and supported.

Can someone with a DSD have children?

Some people conceive without assistance. Others have reduced fertility or cannot produce eggs or sperm. This depends on the gonads, internal reproductive anatomy and diagnosis. Fertility assessment can provide individual information.

Does everyone with a DSD need surgery?

No. Surgery may help with certain medical or functional problems, but it is not routine. Non-urgent procedures that permanently change anatomy should include discussion of whether they can wait until the person can participate in the decision.

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