

Scottish Differences of Sex Development

SDSD

National Managed Clinical Network

Glossary of Terms

Information leaflet

This document has been prepared by Public Services Delivery Scotland (PSD Scotland) on behalf of the Scottish Differences of Sex Development (SDSD) network. Accountable to Scottish Government, PSD Scotland works at the heart of the health service providing national strategic services to the rest of NHS Scotland and other public sector organisations to help them deliver their services more efficiently and effectively. The SDSD network membership is made up of a multidisciplinary group of healthcare professionals who provide care to children and adults born with a difference in sex development or anomaly of the genitalia. The network is supported by a PSD Scotland Programme Team to drive improvement.

Endorsed September 2025
Review September 2028
NSD610-015.02

These are some terms you may hear in a discussion about your child's DSD. If at any time you hear a term that you don't understand, ask a doctor or other medical professional to explain it.

Adrenal glands - the adrenal glands, located above the kidneys, they are responsible for secreting various hormones, including androgens.

Androgens - are hormones (chemical messengers) made mostly by the testes but also made to a lesser extent in the adrenal glands, and in the ovaries.

The two major types of androgens involved in sex development are Testosterone and Dihydrotestosterone.

Bifid scrotum - A scrotum that is separated by a deep cleft or groove into two parts.

Chordee - A curving of the penis.

Chromosomes - These are the rod-like structures within our cells that are the packages for our genes. Each cell in your body has a full set of your chromosomes.

The chromosomes contain the body's genes; we have 46 chromosomes in each cell: that includes 22 pairs of chromosomes (one of each pair from each parent) In addition to those 22 pairs, we have two additional chromosomes, and these are called the "sex chromosomes". Sex chromosomes are designated by the letters X and Y.

Most females have two X chromosomes, and so we say they have the Karyotype 46,XX; the number 46 tells you they have 46 chromosomes total, and the "XX" tells you that two of those chromosomes are X chromosomes.

Most males have one X chromosome and one Y chromosome, and so we say they have the karyotype 46,XY (46 chromosomes, including one X and one Y).

However, there are many other patterns of chromosomes. Some people have an extra X or Y, some are missing an X, some females have a Y chromosome and some males have two X's.

Diagnosis - This usually means the name of a cause of a DSD.

Dihydrotestosterone - A "strong" *androgen* (male sex *hormone*).

Foetus – Refers to unborn offspring that has developed from an embryo to a stage where its major organs and body systems are formed and it is primarily focused on growth and maturation. This transition from embryo to foetus officially occurs around the eighth or ninth week of development after conception, with the foetal stage continuing until birth.

Gender - While "sex" usually refers to a person's physical anatomy, the term "gender" usually refers to mental, social and cultural characteristics, regardless of anatomy.

Genital folds - All foetuses have genital folds early in the womb, prior to sex development. These folds later develop into the labia majora in most girls and the scrotum in most boys. Children with a DSD sometimes have external genital structures that look in-between labia and scrotum.

Genital tubercle - Present in all foetuses in early development, the genital tubercle is a structure of the external genitalia that develops into the phallus (in other words, the clitoris or penis).

Gonadal ridges - Common in all foetuses prior to *sex development*, gonadal ridges consist of tissue that develops into *gonads* (*ovaries*, *testes*, *ovotestes*, or g*onadal streaks*).

Gonadal streaks - Gonadal streaks are poorly developed gonad tissue present in place of *testes* or in place of *ovaries* in some people with a DSD.

Gonadectomy -"Gonadectomy" means to perform surgery to remove the sex glands (*ovaries*, *testes* or *ovotestes*).

Gonads - “Gonads” is a general term for the sex glands. The term “gonad” can refer to an *ovary*, a *testis* (testicle), an *ovotestis* or a streak gonad.

Gynecomastia - Gynecomastia refers to the enlargement of a man’s breasts, usually due to a *hormone* imbalance or to hormone treatments.

Hormones - Hormones are molecules that carry messages from one group of cells to another through the blood, stimulating (“waking up”) some body processes and inhibiting (“shutting down”) others.

Intrauterine – Refers to an early developing baby inside the womb.

Karyotype - A karyotype shows the chromosomes in a cell. A karyotype is used to see what kinds of chromosomes a person has. It is created by taking a blood or tissue sample from a person.

The result is usually reported as the number and type of a person’s chromosomes, such as 46,XX (the individual has 22 pairs of matched chromosomes and two X chromosomes; 46,XY (the individual has 22pairs of matched chromosomes), one X chromosome and one Y.

Mosaic karyotype - A person is said to have a “mosaic karyotype” when he or she has one kind of *karyotype* in some of his or her cells, and a different karyotype in other cells. An example is when a person is said to have a 45,X/46,XX karyotype; that means he or she has 45,X in some cells, and 46,XX in other cells. Mosaicism happens because sometimes cells divide incorrectly early in the life of a foetus.

Mullerian ducts - Present in all foetuses in early development, in most females the Mullerian ducts develop into the *uterus*, fallopian tubes and the upper part of the vagina.

Mullerian inhibiting substance - Also called anti-Mullerian hormone. A *hormone* normally produced by the *testes* in the early stages of male fetal development that prevents the

Mullerian ducts from developing into the fallopian tubes, *uterus* and upper part of the vagina.

Multidisciplinary clinic - If a *multidisciplinary team* meets together on a regular basis to provide “one" visit for families with a DSD, that is called a multidisciplinary clinic. The advantage of this is that it often simplifies medical care and generally ensures that your child’s doctors and other members of the medical team are talking with each other and working together.

Oestrogens - are hormones (molecules, or chemical messengers) mainly produced in the ovaries, but also produced to a lesser extent in the adrenal glands and testes. They are responsible for breast development. They are also responsible for helping to regulate the menstrual cycle.

Ovaries - The ovaries are the female *gonads* (sex glands) located in the lower abdomen of most girls and women; usually one on either side of the *uterus*.

Ovotestes - Ovotestes are *gonads* (sex glands) containing both ovarian and testicular tissue.

Paediatric endocrinologist - A paediatric endocrinologist is a children’s doctor who specialises in the endocrine system, commonly known as the hormonal system.

Paediatric urologist - A paediatric urologist is a children’s doctor specialising in the reproductive organs (sex organs) and the organs of the urinary system.

Secondary sex characteristics - These are changes that typically occur at the time of puberty. They can include body hair growth, change in pitch of voice, genital growth, breast development, muscle development and growth of the Adam’s apple.

Sex development - This is the term for the step-by-step changes that relate to the biological (physical) features of a person’s sex. The development of sex begins at conception with the combining of sex *chromosomes* from the mother’s egg and the father’s sperm. Sex development continues in the womb with the prenatal development of the internal sex organs (including the *gonads*) and the external sex organs (like the penis, clitoris, labia and scrotum).

For most people sex development continues naturally, little by little, through all stages of life. This is most noticeable at puberty (which brings many changes: fat distribution; voice pitch; hairline; pubic, underarm and body hair; genital and nipple appearance; breast development; skin - oil and texture; body odour) and at menopause.

Testes -The testes (also called testicles) are the male-typical *gonad* (sex gland), usually located in a scrotum

Testicles - See testes.

Testosterone -Testosterone is the main male sex *hormone* that is produced in the *testes.*

Urethral folds - Present in all foetuses early in development, the urethral folds typically develop into the labia minora in females and the urethra and the shaft of the penis in males.

Uterus (Womb) - This is the organ that typically connects with the vagina in females. It is where babies develop and is also the source of menstrual flow.

Wolffian ducts - Present in all foetuses in early development. The Wolffian ducts typically develop in males into the *vas deferens*, the *epididymis* and the *seminal vesicles*.

The **vas deferens** is the passageway that carries sperm from the epididymis to the ejaculatory duct.

The **epididymis** is an organ located on the *testes* that has passageways (ducts) that carry sperm from the testes to the vas deferens.

The **seminal vesicles** are glands that produce the fluid component of semen.

Further Information

This leaflet will be available in other languages (e.g. Punjabi, Urdu, Polish) or formats (e.g. larger print) on request.

Contact: nss.sdsd@nhs.scot

Website: www.nn.nhs.scot/sdsd

©PSD Scotland 2026. You can copy or reproduce the information in this resource for use within NHS Scotland and for non-commercial educational purposes under creative commons CC BY-NC 4.0 Deed | Attribution Non-Commercial 4.0 International | Creative Commons. Use of this document for commercial purposes is permitted only with the written permission of PSD Scotland.