



GLOSSARY (EXPLANATION OF TERMS)

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).

Estrogens - 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.

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, related to being a boy, girl, man or woman in our society.

Gender assignment - When a child is born with a DSD and his or her sex is unclear, the child is given a "gender assignment," which means the parents decide whether to raise the child as a boy or a girl with the help of the Medical Team

Gender role - A part that a person plays as a boy, girl, man, or woman in our society.

Genital folds - No matter how they end up in terms of *sex development*, all embryos 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 embryos 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 embryos prior to *sex development*, gonadal ridges consist of tissue that develops into *gonads* (*ovaries*, *testes*, *ovotestes*, or *gonadal 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.

Intersex - Intersex is a term sometimes used to refer to the condition of having a sex anatomy that is not considered standard for a male or a female.

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 an embryo.

Mullerian ducts - Present in all embryos 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.

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 - Sex usually specifically refers to a person's physical anatomy as female, male or *intersex*.

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.

Sexual orientation - This refers to whether an individual is sexually attracted to men or women or both. If a person is identified as a man and is attracted to a man, he is said to be homosexual. If a person is identified as a woman and is attracted to men, she is said to be heterosexual.

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 embryos 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 embryos 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.

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