GLOSSARY

Chromosome
Chromosomes are found in each cell in the body. Each human cell normally contains 46 total chromosomes – organised in two sets of 23 chromosomes – that come in two types: sex chromosomes and autosomal chromosomes. Each cell in the human body contains these chromosomes which contain genetic material (genes) that make up an individual's DNA (deoxyribonucleic acid). Sex chromosomes determine gender. In the final of the 23 sets of chromosomes, females have two X chromosomes, while males have an X and a Y chromosome; in some intersex people, there are variations in the configuration of the 23rd chromosome set. Phenotypes are produced by multiple chromosomes acting together.

Cryptorchidism
Cryptorchidism refers to the condition in which the testes fail to descend into the scrotum and are retained within the abdomen or inguinal canal.

Clitoroplasty, clitoridectomy
Clitoridectomy is the surgical excision of the clitoris. Until the 1960s clitoridectomy was the principal surgical procedure used to manage enlargement of the clitoris in intersex. Clitoroplasty is a surgical procedure to alter the physiology of the clitoris, and includes procedures in which part of the erectile tissue of the clitoris is removed (clitoral reduction) or relocated (clitoral recession) to reduce the apparent size of the clitoris.

Cloacal Extrophy
Cloacal Extrophy is a condition in which an infant has the bladder and a portion of the intestines exposed outside the abdomen. In males the penis is either flat and short or sometimes split. In females the clitoris is split and there may be two vaginal openings. Frequently the intestine is also short and the anus may not be open.

Dysgenesis
Dysgenesis refers to abnormal organ development during embryonic growth and development of a foetus. Gonadal and adrenal dysgenesis are two of the more common types of dysgenesis. Gonadal dysgenesis may result in a streak gonad.

Endocrinology
Endocrinology is a medical specialisation dealing with the body's production, use and response to hormones.

Genitoplasty
Genitoplasty is the surgical alteration of external genitalia, and is a procedure sometimes performed on individuals with ambiguous genitalia. The two essential elements of feminising genitoplasty are clitoral reduction/recession (clitoroplasty, see above) and vaginoplasty (see below).
**Genotype**

A person's genotype describes all of the genetic information that is encoded in his or her chromosomes (for example 46,XY or 46XX, among others). It also refers to the genetic information carried by a pair of genes (one from each parent) which controls a particular characteristic.

**Germ cell tumour**

Germ cells are those embryonic cells that have the potential to develop into gonads. Germ cell tumours are tumorous growths based in those cells, and can be cancerous or non-cancerous.

**Gonad**

Gonads are reproductive glands; the term can refer to either testicles or ovaries. Gonads in foetuses develop into either testes or ovaries depending on the chromosomal constitution of the foetus. In some intersex people, gonads do not differentiate fully into one type or the other.

**Streak gonad**

Streak gonads consists of fibrous tissue without any germ cells, and therefore are unable to function.

**Gonadectomy**

A gonadectomy is the removal of an ovary or testis. In some intersex cases, gonadectomy is undertaken if the testes are inconsistent with the sex of assignment. In some CAIS individuals the testes are intra-abdominal or contained in inguinal herniae (a protrusion of the abdominal cavity).

**Histology**

Histology is the science dealing with the microscopic identification of specific cells and tissue.

**Hypospadias**

Hypospadias is a development disorder affecting the urethra. In the male, it is a developmental anomaly in which the urethra opens on the underside of the penis or on the perineum. In females hypospadias is a developmental anomaly in which the urethra opens into the vagina.

**Immunohistochemistry**

Immunohistochemistry is a medical diagnostic tool. Histochemistry is the study of the chemistry of organic tissue through observing chemical reactions. Immunohistochemistry is a form of histochemistry which relies on the principle of certain antibodies binding specifically to certain receptors (antigens) in biological tissue; these reaction patterns can then be assessed. Immunohistochemistry is widely used to detect specific structures in tissues and in the diagnosis of abnormal cells such as those found in tumours.
Inguinal

Inguinal refers to the region of the groin. In the male foetus the inguinal canals are a pair of openings that connect the abdominal cavity with the scrotum. An inguinal hernia is a protrusion through the lower abdominal wall.

Intra-abdominal

Intra-abdominal refers to the area of the body in which the ovaries and uterus are found. In some intersex conditions, the position of the testes is intra-abdominal rather than scrotal.

Karyotype

A karyotype refers to the number and structure of chromosomes in the nucleus of a cell; that is, the complete set of chromosomes in an individual. The karyotype is usually identical in all the cells of an organism (but not in some rare types of intersex). The standard human karyotype contains 22 pairs of autosomal chromosomes and one pair of sex chromosomes (46 chromosomes in total). The standard karyotype for females is denoted as 46,XX whereas the standard male karyotype is expressed 46,XY.

Labiaplasty

Labiaplasty is a surgical procedure to modify, usually by reducing the size of, the labia, the folds of flesh and skin that surround the female genitals.

Neoplastic

Neoplasty is any abnormal growth of new tissue.

Prophylactic

A prophylactic is an agent or procedure that prevents the development of a condition or a disease.

Phenotype

Phenotype refers to the complete observable characteristics of an individual, including anatomical, physiological, biochemical and behavioural traits, as determined by the interaction of both genetic makeup and environmental factors.

Scarification

Scarification is the creation of scar tissue following surgical procedures.

Scrotal

In relation to the position of the testes, scrotal testes are in the scrotum. Testes can in some intersex variations be intra-abdominal or inguinal.
**Vaginoplasty**

Vaginoplasty is a surgical procedure to create a vaginal canal. Some intersex conditions such as Complete Androgen Insensitivity Syndrome may cause individuals to develop a blind vaginal pouch that averages 2.5 to 3.0 cm in depth, compared to an average of 10-12 cm depth for non-CAIS individuals. Some individuals in these circumstances will undergo vaginoplasty.

**Key references**


**Oxford Reference Online Premium Collection**

