**X chromosome.** One of the **sex chromosomes.** Females usually have two **X chromosomes.** Males usually have one **X** and one **Y chromosome.**

**X-linked conditions.** A genetic condition caused by a **mutation** (change) in a **gene** on the **X chromosome.** X linked conditions include hemophilia, Duchenne muscular dystrophy and fragile X syndrome. For more information see the **X-linked Inheritance leaflet.**

**XX.** This represents the usual **sex chromosomes** of a female. Females usually have two **X chromosomes.** One **X chromosome** is inherited from each parent.

**XY.** This represents the **sex chromosomes** of a male. Males usually have one **X chromosome** and one **Y chromosome.** A male inherits his **X chromosome** from his mother and his **Y chromosome** from his father.

**Y chromosome.** One of the **sex chromosomes.** Males usually have one **Y chromosome** and one **X chromosome.** Females usually have two **X chromosomes.**

Other genetic glossaries can be found at the following addresses:
- [www.geneticalliance.org.uk/glossary.htm](http://www.geneticalliance.org.uk/glossary.htm)
- [www.kumc.edu/gec/glossary.html](http://www.kumc.edu/gec/glossary.html)
- [www.genome.gov/glossary.cfm](http://www.genome.gov/glossary.cfm)

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Genetic Glossary

amniocentesis. A test that is used to take a sample to test an unborn baby’s genes or chromosomes. The baby is surrounded by fluid in the womb. The fluid contains a few of the baby’s skin cells. A small sample of the fluid is taken with a thin needle, through the skin of the mother’s abdomen (tummy or belly). The fluid is sent to a laboratory for testing. For more information see the Amniocentesis Test leaflet.

autosomal. Involving the autosomes.

autosomal dominant genetic conditions. These are conditions whereby a person needs only to inherit one changed copy (mutation) of the gene in order to be affected by the condition, or become affected by the condition later in life. The changed gene is dominant over the normal gene. For more information see the Dominant Inheritance leaflet.

autosomal recessive genetic conditions. These are conditions whereby a person has to inherit two changed copies (mutation) of the gene (a changed copy from each parent) to be affected by the condition. A person who has only one copy of the changed gene will be an unaffected carrier. For more information see the Recessive Inheritance leaflet.

autosomes. We have 23 pairs of chromosomes. Pairs number 1 to 22 are called autosomes and look the same in men and women. Pair number 23 are different in men and women and are called the sex chromosomes.

balanced translocation. A translocation in which no chromosome material is lost or gained, but it is rearranged. A person with a balanced translocation is not usually affected by it.

carrier. A person who is generally not affected with the condition (at that moment), but carries one fault copy of a gene. In the

chromosome. The sex chromosomes control whether a person is male or female. Females have two X chromosomes. Males have one X and one Y chromosome.

sex-linked condition. See X-linked condition.

smear test. A test recommended for all women, to check for abnormality in the cells at the opening of the womb.

sperm. The father’s contribution to the cell which will grow to form a new baby. Each sperm contains 23 chromosomes; one from each pair in the father. The sperm joins with an egg to make a complete cell. A baby develops from this first cell.

translocation. Rearrangement of chromosome material. Arises when a piece of a chromosome is broken off and attaches to another. For more information see the Chromosome Translocations leaflet.

ultrasound scan. A painless test that uses sound waves to create images of the growing baby during the mother’s pregnancy. It may be performed by passing the head of the scanner across the skin of the abdomen (tummy/belly) or from within the vagina.

unbalanced translocation. A translocation in which the chromosome rearrangement has some extra chromosome material or has some missing chromosome material, or both extra and missing material. May arise in the child of a parent with a balanced translocation. For more information see the Chromosome Translocations leaflet.

uterus. Medical term for the womb.

vagina. The connection from the womb to the outside of a woman, the birth canal.

womb. The part of a woman’s body in which a baby grows during pregnancy.
in pregnancy. If the baby has a congenital condition (such as Down syndrome), the size of the space may be abnormal.

Ovary / ovaries. Organs in a woman’s body that produce eggs.

placenta. An organ which lies against the inside wall of the womb in a pregnant woman. The baby gets its nourishment from the placenta. The placenta grows from the fertilised egg so usually has the same genes as the baby.

positive result. A test result which shows that the person tested does have the change (mutation) in the gene.

predictive testing. A genetic test for a condition that may or will occur later in life. When the genetic test is for a condition that will almost certainly develop the condition in the future, the test is sometimes called a presymptomatic test.

prenatal diagnosis. Test during a pregnancy for the presence or absence of a genetic condition in the baby.

presymptomatic test. See predictive test.

reciprocal translocation. A reciprocal translocation occurs when two fragments break off from two different chromosomes and swap places. For more information see the Chromosome Translocations leaflet.

ring chromosome. Term used when the ends of a chromosome have joined together in a ring shape. For more information see the Chromosome Changes leaflet.

robertsonian translocation. A Robertsonian translocation occurs when one chromosome becomes attached to another. For more information see the Chromosome Translocations leaflet.

sex chromosomes. The X chromosome and the Y chromosome.

carrier (of a chromosome translocation). A person who has a balanced translocation whereby no chromosome material is lost or gained and is not usually affected by it.

cell. The human body is made up of millions of cells, which act like building blocks. Cells in different parts of the body look different and do different things. Every cell (except for eggs in women and sperm in men) contains two copies of each gene.

chorionic villus sampling, CVS. A test that is done during pregnancy to take cells in order to test the baby’s genes or chromosomes for specific genetic conditions. A small number of cells are taken from the developing placenta and sent to a laboratory for testing. For more information see the Chorionic Villus Sampling leaflet.

conception. The joining of an egg and a sperm to make the first cell of a new baby.

deletion. The omission of a part of the genetic material; the term can be used to describe either a missing area of a gene or a chromosome. For more information see the Chromosome Changes leaflet.

de novo. Phrase from the Latin language, meaning “from new”. Used to describe a gene or chromosome which is “new”, i.e. both that person’s parents have normal genes or chromosomes.
DNA. A chemical substance which makes up the genes, and which contains the information needed for the body to work.

duplication. The abnormal repetition of a sequence of genetic material in a gene or chromosome. For more information see the Chromosome Changes leaflet.

egg. The mother’s contribution to the cell which will go on to make a new baby. The egg contains 23 chromosomes; one from each pair in the mother. The egg joins with a sperm to make a complete cell. A baby develops from this first cell.

embryo. An early stage of human development. The embryo develops from the first cell in the very early stages of pregnancy. It arises after fertilisation of an egg by a sperm. It does not look like a baby yet, but is made up of the cells that will develop into a baby.

family tree. A diagram to show the people in your family who do and do not have the genetic condition, and how they are related to you and to each other.

fetus. The period when the baby develops from the end of the embryo phase to a newborn i.e. from the 9th week after fertilization until birth.

gene. Information needed for the body to work, stored in a chemical form (DNA) on chromosomes.

genetic. Caused by genes, concerning genes.

genetic condition. A condition or disease caused by an abnormality in a gene or chromosome.

-genetic counselling. Information and support for people who are concerned about a condition which may have a genetic basis. (Is this term used in your country? If not it might be better to leave out this entry.)

genetic counsellor. A specialist who gives information and support to people who are concerned about a condition which may have a genetic basis. (Is this term used in your country? If not it might be better to leave out this entry.)

-genetic test. A test which can help identify if there is a change in a particular gene or chromosome. It is usually a blood or tissue test. For more information see the What is a Genetic Test? leaflet.

hereditary condition. One that is inherited (passed down through families).

insertion. The introduction of additional genetic material into a gene or chromosome. For more information see the Chromosome Changes leaflet.

inversion. An alteration in the sequence of genes along a particular chromosome. For more information see the Chromosome Changes leaflet.

karyotype. A description of the chromosome structure of an individual including the number of chromosomes, the type of sex chromosomes (XX or XY) and any variation from the normal pattern.

miscarriage. Early end to a pregnancy, before the baby can survive outside the womb.

mutation. A change in a gene. Sometimes when a gene is changed, its information is altered so it does not work properly. This may cause a genetic condition.

-negative result. A test result which shows that the person tested does not have the change (mutation) in the gene.

nuchal translucency test. An ultrasound scan of the back of the baby’s neck, where there is normally a fluid-filled space early