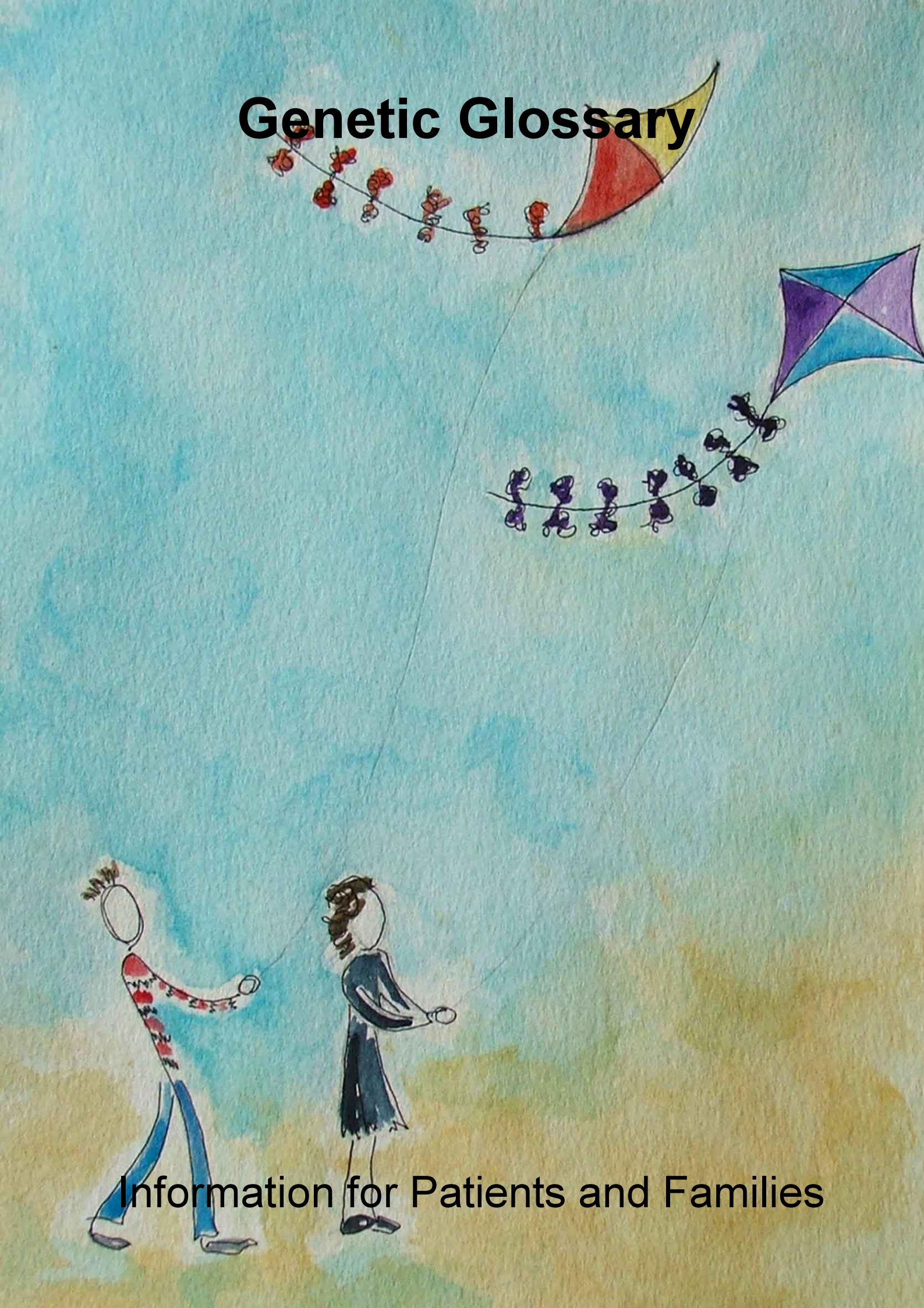
Genetic Glossary



Information for Patients and Families

amniocentesis. A test that is used to take a sample to test an unborn baby's **gene**s or **chromosomes**. The baby is surrounded by fluid in the **womb**. The fluid contains a few of the babies 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.

autosomal. 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**.

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. The changed **gene** is dominant over the normal **gene**.

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**.

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, but carries one fault copy of a **gene**.

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.

chromosomes. Thread-like structures which can be seen under the microscope and contain the **genes**. The usually number of chromosomes in humans is 46. One set of 23 chromosomes we inherit from our mother and one set of 23 chromosomes we inherit from our father.

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

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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**.

de novo. Phrase from the Latin language, meaning "from new". Used to describe a **translocation** which is "new" in a child, i.e. both the child's parents have normal **chromosomes.**

duplication. The abnormal repetition of a sequence of genetic material in a **gene** or **chromosome**.

egg. The mother's contribution to the **cell** which will grow 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. Earliest 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 **cell**s that will develop into a baby. It is possible for very early embryos to grow outside the mothers womb.

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.

gene. Information needed for the body to work, stored in a chemical form 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.

genetic counsellor. A specialist who gives information and support to people who are concerned about a condition which may have a genetic basis.

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 please 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**.

inversion. An alteration in the sequence of **genes** along a particular **chromosome**.

karyotype. A description of the **chromosome** structure of an individual including the number of **chromosomes** 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**.

nuchal translucency test. An **ultrasound scan** of the back of the baby's neck, where there is normally a fluid-filled space early 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 (or after-birth). The placenta lies against the 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.

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

reciprocal translocation. A reciprocal **translocation** occurs when two fragments break off from two different **chromosomes** and swap places.

ring chromosome. Term used when the ends of a **chromosome** have joined together in a ring shape.

robertsonian translocation. A Robertsonian **translocation** occurs when one **chromosome** becomes attached to another.

sex chromosomes. The X **chromosome** and the Y **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.

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**.

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.

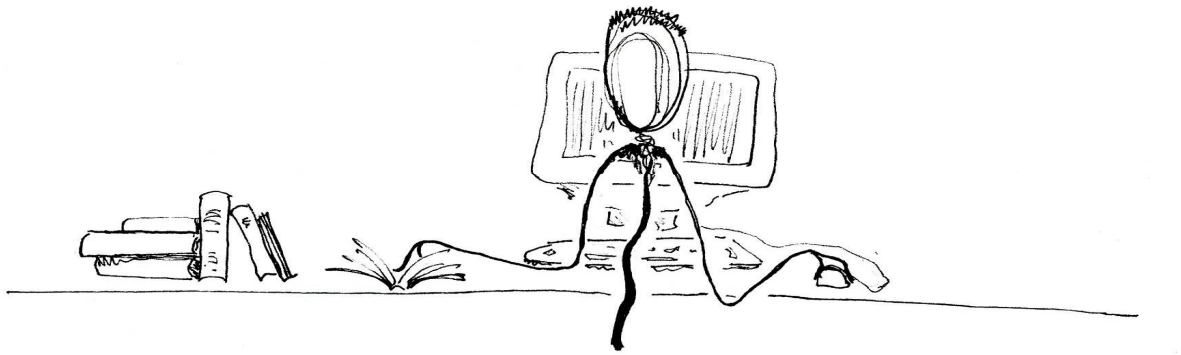
X chromosome. One of the **sex chromosomes**. Females have two X **chromosomes**. Males usually have one X and one Y **chromosome**.

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.

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 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 have one **Y chromosome** and one **X chromosome**. Females have two **X chromosomes**.



Other genetic glossaries can be found at the following addresses:

Genetic Interest Group
www.gig.org.uk/glossary.htm

Genetics Education Centre, Kansas University
www.kumc.edu/gec/gossnew.html

National Human Genome Research Institute
www.genome.gov/glossary.cfm

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