

GLOSSARY



N.B. Some definitions contain words in italics (like this). You will find these words defined elsewhere in the glossary.

A

Allele: one of two or more alternative forms of a *gene* that exist at a specific gene location on a chromosome and which give rise to alternative inherited characteristics. For example, many different alleles are known at the location of the gene associated with cystic fibrosis; some pairs of alleles give rise to the disease, while other pairs do not.

Amino acids: simple organic units in living things which link together to make *proteins*. There are 20 amino acids which are commonly found in the proteins of living organisms.

Amniocentesis: the removal during pregnancy of a small quantity of the amniotic fluid surrounding the *fetus* in the womb. This fluid contains cells from the fetus, and analysis of these cells can give doctors specific information about how the fetus is developing (for example, whether it has Down's syndrome).

Autosome: any *chromosome* which is not a *sex chromosome*. There are 22 pairs of autosomes in the nucleus of each human cell and two sex chromosomes, making 23 pairs (or 46 chromosomes) in total.

Autosomal: affecting the autosomes (i.e. all *chromosomes* except the *sex chromosomes*).

C

Carrier: a person who has one faulty and one working copy of the gene for a recessive *genetic disorder* or for a characteristic. Carriers of most *recessive* disorders are themselves unaffected because the working copy of the gene overrides the faulty copy, but they can pass a copy of the faulty gene on to their children.

Chromosomal condition/ disorder: when a whole *chromosome* (rather than a single *gene*) is missing or perhaps parts of whole chromosomes are re-arranged, giving rise to specific conditions or disorders (eg Down's Syndrome).

Chromosome: A thread-like structure made of DNA which is found in the *nucleus* of animal and plant cells. Most human cells contain 46 *chromosomes* (23 pairs), but eggs and sperm (the *sex cells*) contain only 23 unpaired chromosomes. There are thought to be approximately 100,000 pairs of genes spread out along the 46 chromosomes which make up the human genome.

GLOSSARY

D

Dermatologist: a person specialising in the diagnosis and treatment of skin diseases.

DNA: deoxyribonucleic acid. *Chromosomes* are made up of long strands of DNA and *genes* are segments of this DNA. DNA molecules are made up of two sugar-phosphate strands held together by pairs of organic bases and twisted into the shape of a double helix. The order of the four bases (or nucleotides) - adenine (A), cytosine (C), guanine (G) and thymine (T) - determines the order in which *amino acids* are joined together to make the many tens of thousands of proteins your body needs to develop and work properly.

Dominant: If a faulty *gene* is dominant, it will show an effect even though there is a working copy of the gene on the other *chromosome*. Huntington's disease is an example of a dominantly-inherited *genetic disorder*. A person only needs to inherit a faulty gene for Huntington's from one parent to develop the disease.

E

Embryo: a developing organism. In humans, the word embryo is used until about eight weeks after *fertilisation*. [See foetus.]

Enzyme: a *protein* which helps chemical reactions to take place in cells.

Eugenics: an attempt to influence human evolution through programmes aimed at encouraging the passing on of 'desirable' characteristics and preventing the transmission of 'undesirable' ones.

F

Fertilisation: When a sperm cell penetrates the outer layer of an egg cell and joins with it to form a new life. Fertilisation can take place inside the body (as a result of the sperm meeting the egg after sexual intercourse has taken place) or sometimes in a laboratory (where it is known as *in vitro* fertilization, or IVF. Some people refer to babies conceived in this way as 'test-tube babies'.)

Foetus: in humans, the developing *embryo* in the womb from about eight weeks after *fertilisation* until birth.

G

Gamete: a *sex cell* (i.e. sperm or egg cell) containing 23 unpaired *chromosomes*.

Gene: a segment of DNA which carries coded instructions for *amino acids*, the building blocks from which *proteins* are made. Passed on from one generation to the next in the *chromosomes*, genes are responsible for determining our inherited characteristics.

GLOSSARY

G

Gene Therapy: an experimental technique in which doctors introduce working copies of a missing or faulty gene into living cells in the hope that the new genes will help the affected cells to produce the protein they need to function properly.

Genetic Code: segments of DNA (or *genes*) contain coded instructions for the production of *amino acids* and therefore of *proteins*. [See DNA].

Genetic Condition/Disorder: a condition resulting from the genetic make-up of that individual. The word 'disorder' implies unwanted consequences (for example disability or disease) arising from the genes which one has inherited.

Genome: all the genetic material of an organism. Scientists have mapped the human genome (i.e. to found the relative positions of all the *genes* on each *chromosome*).

Germ Cell: a *sex cell* (i.e. sperm or egg cell).

Germ-line gene therapy: this is currently against the law. Germ-line gene therapy involves replacing or repairing faulty genes in *germ cells* and would therefore have an effect on the next generation.

H

Helix: see *DNA*

Human Genome: see *Genome*

I

Infertile: a person who is infertile cannot have children. This could be because he/she does not produce enough healthy sperm /egg cells to achieve fertilisation, or perhaps the tubes down which the sperm/egg travel are blocked or damaged.

In vitro fertilisation (IVF): see *Fertilisation*.

M

Metabolic disorder: when a person is born with a vital *enzyme* missing or not working properly and a metabolic blockage occurs. The blockage usually means that chemicals are unable to get through to where they are needed and so build up abnormally on one side of the blockage, which can have very serious consequences for health.

GLOSSARY

M

Miscarriage: a pregnancy which ends before the *fetus* is 24 weeks old. About 1 in 6 pregnancies ends in early miscarriage. (i.e., before the 12th week of pregnancy). If a woman has a miscarriage, her baby usually dies, although sometimes it can be saved by a special care unit in hospital.

Mutation: a change in the structure of *DNA*. Such changes can be passed on to the next generation. A few mutations are beneficial, but others can cause *genetic disorders*.

N

‘Nature v. nurture’: the debate about the degree to which human psychology and behaviour are influenced by genes and/or environmental factors is sometimes referred to as the ‘nature versus nurture’ debate. It is likely that both nature (ie, our genetic inheritance) and nurture (i.e. the way we are brought up, the food we eat, our expectations etc.) influence our behaviour, but calculating the precise influence of each is a hugely complex task.

Neurological: a neurological *genetic condition* is an inherited disorder affecting the nervous system.

Nucleus: the part of the cell which contains the *chromosomes*.

Nurture: see ‘nature v nurture’

O

Orphan diseases: rare diseases which could be treated but for which no treatment is likely to be developed. This is because drug companies are unwilling to invest huge sums of money in developing new treatments for a limited market. Of the 4500 or so known genetic disorders, those which affect relatively small numbers of people could be classed as orphan diseases.

Orthopaedic clinic: a place where doctors study, and try to cure, problems arising from disease and injury to bones, particularly in childhood.

Ovum: another word for the female *sex cell* (egg).

P

Pattern of inheritance: see *Dominant*, *Recessive* and *X-linked*.

Pre-implantation diagnosis: a technique being developed to help families affected by specific *genetic disorders* who wish to avoid passing on a faulty copy of the gene which causes the disorder to their children. Eggs are taken from the woman and fertilised *in vitro*. At a very early stage in the development of each fertilised egg, a single cell is removed and its genetic material (*DNA*) analysed. Only *embryos* which do not contain copies of the faulty gene are re-implanted in the woman’s womb where it is hoped that one embryo will go on to develop into a baby. Pre-implantation diagnosis (PID) is currently available in only a few countries around the world and for a limited number of *genetic conditions*.

Prenatal tests: tests carried out when a woman is pregnant to check if the *fetus* is developing normally.

GLOSSARY

P

Proteins: molecules made up of many *amino acids*. Proteins are an important part of all living cells and play vital roles in the chemical reactions which enable organisms to function.

R

Recessive: If a faulty *gene* is recessive, it will usually show little or no effect unless the same recessive gene is faulty in **both** of a pair of *chromosomes*. Sickle cell anaemia is an example of a recessively-inherited disorder. A person who inherits **two** faulty copies of the gene (one from each parent) will have sickle-cell anaemia. A person who only inherits one faulty copy of the gene will be a *carrier* and could pass the faulty gene onto any children he/she may have.

Replicate: to make an exact copy of something. *Chromosomes* (which are made up of very long strands of *DNA*) replicate before cell division takes place.

S

Screening: genetic screening is when doctors test everyone within a population or subset of that population (ie, all women of childbearing age, or all male newborn babies) to see if they have a particular *gene* or predisposition towards a specific *genetic disorder*. The people being screened may have no family history of the disorder or reason to suspect that they might have this gene or predisposition. [See *Testing*].

Sex cell: another way of describing the sperm (male sex cell) or egg (female sex cell). [See also *Germ cell*.]

Sex chromosomes: these are responsible for determining the sex of an individual. In humans they are known as X and Y *chromosomes*; females have two X chromosomes in most body cells, while males have an X and a Y chromosome.

Sex determination: the process whereby the inheritance of the *sex chromosomes* decides the sex of an *embryo*.

Sex-linked: see *X-linked*.

GLOSSARY

T

Termination: another word for abortion (i.e. the ending of a pregnancy by medical means).

Testing : genetic testing is when doctors test an individual with a known family history of a particular *genetic disorder* to see whether that person has, or does not have, a copy of the faulty *gene* associated with that disorder. [see *screening*]

Translocation: when parts of *chromosomes* transfer themselves to other chromosomes which are not of the same pair. This can lead to problems and abnormalities in the developing *fetus*.

U

Unaffected carrier: see *carrier*

X

X chromosome: the female *sex chromosome*.

X-linked: refers to the inheritance of a particular characteristic or disorder from a gene carried on the X (or *female sex*) *chromosome*. Also known as *sex-linked* inheritance.

XYY male: a male with an extra Y *chromosome* in most of his body cells.

Y

Y chromosome: the male *sex chromosome*.

Z

Zygote: a fertilised egg.