Glossary of terms relating to genetic healthcare (English)

alleles	Alternative forms of a gene at the same locus (position on a chromosome). A person inherits one allele from the father and one from the mother
aneuploidy	An alteration in the number of chromosomes within a cell, for example 45 or 47 chromosomes instead of the usual 46. An example is trisomy 21
assisted reproduction	Any procedure or medication used to increase the chances of a woman conceiving a child
autosomal recessive	A pattern of inheritance where a condition is only present if both copies (one from mother and one from father) of a particular gene have a pathogenic variant
BRCA	BRCA is the term used to refer to Breast Cancer genes. There are two known BRCA genes that are involved in breast and ovarian cancer: BRCA1 and BRCA2
cardiomyopathy	A chronic disease of the heart muscle
carrier	A person who has one pathogenic variant and one normal copy of a gene associated with an autosomal recessive condition. Carriers do not usually display any symptoms
cascade testing	The process of testing family members, by starting with the closest (1st degree) relatives, then testing 2nd degree relatives through each affected 1st degree relative, and so on through the family
chromosomal abnormality	An alteration in one or more chromosomes causing structural changes, additional or deleted sections of DNA

chromosome	A tightly wound single thread of DNA, along with structural proteins which is efficient for storage in the nucleus.
complex consanguinity	Consanguinity is a term meaning being related to someone by sharing an ancestor. Complex consanguinity occurs when two people are related to the same ancestor in more than one way, through multiple consanguineous partnerships in the family
congenital heart defect	A problem with the development of the heart that is present from birth
consanguinity	Being related to someone by sharing an ancestor. The more closely related you are, the more DNA you will share in common
cytogeneticist	A scientist who analyses the number and structure of chromosomes to detect abnormalities
de novo	An alteration in a gene that is present for the first time in one family member as a result of a mutation in a gamete (egg or sperm) from one of the parents or in the fertilized egg itself. Also called a new mutation
direct-to-consumer	Tests that are sold or marketed to customers, usually online without the involvement of a healthcare professional
dominant	Used to describe a characteristic or condition that appears in an individual who is a heterozygous for a specific gene (ie a person who has one normal and one altered gene)
Down syndrome	A condition causing distinctive physical features and learning disabilities caused by the presence of part or all of an extra copy of chromosome 21
Edwards syndrome	A very serious condition caused by the presence of an additional copy of chromosome 18, usually resulting in death before one year of life. Also called trisomy 18
familial	Pertaining to the family group. Often used to describe an inherited condition
familial cancer syndrome	A condition that is due to a pathogenic gene variant, which results in members of a family being more likely to develop certain cancers due to gene alterations passed from parent to child
first-degree	Relatives that share 50% of their genes with an individual, such as parent, child or full sibling
gamete	A cell containing half the genetic information needed for fertilisation to occur. Gametes are sperm and ova

gene	The basic biological unit of inheritance. A gene consists of the segment of deoxyribonucleic acid (DNA) which usually makes a specific protein or component of a protein. Humans have about 20,000 genes. The genes are arranged in linear order on the chromosomes
gene mutation	An alteration in a gene which is present in less than 1% of the population and may have negative consequences for health or development.
genetic	Involving, resulting from or relating to a gene or many genes
genetic condition	A condition caused by alteration(s) in a specific gene
genetic counselling	The process by which information on genetic disorders is given to a family, discussion of their options and support for decision making
genetic services	Specialist healthcare services for patients or families at risk of or affected by a genetic condition
genetic testing	Genetic testing is a type of medical test that identifies changes in chromosomes or genes. The results of a genetic test can confirm or rule out a suspected genetic condition or help determine a person's chance of developing or passing on a genetic disorder
hereditary	Transmitted or capable of being transmitted from parent to child
heterozygous	When referring to a particular trait or condition, this means an individual who has one normal and one altered copy of a particular gene
homozygous	When referring to a particular trait or condition, an individual who has either two normal or two altered copies of a particular gene
incidence	The number of new occurrences of a condition or disease in the general or a specified population over a given period of time
index	The person who is first identified in the family as having a particular condition, as in index patient or index case
inheritance	The passing of familial elements from one generation to another
inherited	Passed on from parent to child through genes
inherited pre-disposition	Refers to a situation where a person is more likely to develop a condition than the average person, due to alterations in genes passed on from parent to child
inversion	The rearrangement of the order of DNA material along a specific chromosome

karyotype	The complete set of chromosomes in an individual. A normal female karyotype is 46, XX and the normal male karyotype is 46, XY
locus	The precise physical site or location of a specific gene on a chromosome
micro-satellite instability	Areas of frequent mutation caused by a defect in DNA repair mechanisms
miscarriage	Delivery of a non-viable fetus before 24 weeks of pregnancy
neural tube defect	Problems associated with disruption of the development of the brain, spine or spinal cord in early pregnancy. Examples are spina bifida and anencephaly
non-invasive prenatal diagnosis	A form of testing in pregnancy, where the fetus can be tested for a genetic condition using a maternal blood sample. There is no risk to the fetus. Sometimes also called non-invasive prenatal testing. Abbreviated to NIPD or NIPT
pathogenic gene variant	An alteration in a gene that causes a particular condition
penetrance	The proportion of individuals with a mutation causing a particular disorder who exhibit clinical symptoms of that disorder. A condition (most commonly inherited in an autosomal dominant manner) is said to have complete penetrance if clinical symptoms are present in all individuals who have the disease-causing variant , and to have reduced or incomplete penetrance if clinical symptoms are not always present in individuals who have the disease-causing variant
pre-implantation genetic diagnosis	A modified version of in vitro fertilisation. A cell from each embryo is tested to determine whether it has inherited the specific genetic condition before implanting only those embryos that have not inherited it, also called PGD
prenatal diagnosis	Testing done during pregnancy to see whether the fetus is or will be affected by a particular condition
prenatal test	Testing done during pregnancy to see whether the fetus is affected by a particular condition
reciprocal translocation	Where one part of chromosomal material exchanges places with another piece on a different chromosome
Robertsonian translocation	Rearrangement or attachment between chromosomes 13, 14, 15, 21 or 22. These chromosomes have very small arms at one end and these are lost, causing two chromosomes to fuse at the centromere

second-degree	Grandparent, grandchild, aunt, uncle, niece or nephew
stillbirth	Birth of a baby who does not ever take a breath, delivered after 24 weeks of pregnancy
teratogen	A substance or other agent which causes the malformation of an embryo or fetus
termination of pregnancy	Ending the pregnancy voluntarily due to medical issues or parental decision making. Also known as an induced abortion or interruption of pregnancy
trait	A characteristic that is determined by genes passed from parent to child
trisomy	Occurs when an extra copy of a particular chromosome is present in the cell nucleus, making three copies in total instead of the usual two copies
X-linked	A pattern of inheritance where a condition is caused by a pathogenic alteration in a gene on the X chromosome. These conditions are likely to be extremely serious or lethal in males as they have only one copy of the X chromosome