



A lifetime of specialist care

Glossary of genetics terms

Alleles

Alternative forms of a gene at the same position on a chromosome (locus). For instance, at the ABO gene locus on chromosome 9, there are three main alleles in the population - A, B and O. An individual inherits two alleles at the ABO blood group locus - one from each parent. The two alleles together are responsible for a person's ABO blood group type, which is very important to know for blood transfusions. The different possible combinations of alleles give rise to the A, B, AB and O blood groups.

It is not usual to talk about "alleles" when discussing genetic conditions with patients. Health professionals usually refer to the "usual form of the gene" and the "gene containing a change which has caused the condition." The term "allele" may well be seen on molecular genetics reports which describe, in technical language, a variant which may have been found.

Carrier

A healthy person who is usually asymptomatic, but who has a heterozygous gene alteration for an autosomal recessive (ie having one usual and one altered copy of a particular gene) or X-linked recessive disorder, or a balance structural rearrangement of their chromosomes.

Carrier testing

Testing to determine an individual's carrier status in someone who is already known to be at an increased risk of being a carrier for a specific inherited disorder, usually because a relative is known to be a carrier or has the condition.

Carrier testing for family members will be offered and coordinated by the ICC team.

Chromosome

Chromosomes are thread-like structures found in the nucleus of a cell. Each chromosome is made up of DNA tightly coiled many times around proteins called histones that support its structure. In humans there are usually 46 (23 pairs). One of each pair of chromosomes is inherited from a person's mother, the other of the pair from the father. Gametes (sperm and egg cells) have 23 chromosomes (one of each pair). Each chromosome is made up of DNA tightly coiled many times around proteins called histones that support its structure.

One pair of chromosomes, the sex chromosomes (XX in women and XY in men), is involved in determining the sex of an individual. The remaining 22 pairs of non-sex chromosomes (numbered 1 to 22) are known as the autosomes. Genes are arranged in a specific order on the chromosomes.

Consanguineous

Where partners share at least one common ancestor ("blood relatives").

Copy number variation (CNV)

Differences in the number of copies of a particular gene or segment of DNA between individual genomes, due to gains or losses of around one thousand to several million base-pairs. These have been discovered by comparing genomes between people using comparative genomic hybridization arrays.

Copy number variations which include coding regions, and thus alter the number of copies of a gene present, are also sometimes referred to as changes in gene dosage.

Copy number variants may be inherited or may have occurred for the first time in the person whose genome is being examined. Some appear to cause no medical problems, but some CNVs have been associated with susceptibility or resistance to disease.

De novo mutation

New mutation. A new mutation which has occurred due to an error in the copying of genetic material or an error in cell division.

This may result in a family member being the first member in a family to have a genetic condition (autosomal dominant, X-linked or chromosomal) as a result of a variant in a gamete (egg or sperm) of one of the parents or in the fertilized egg itself or developing fetus.

DNA

DNA (Deoxyribonucleic acid) is the molecule that encodes genetic information (the code itself is based on the sequence of four different chemicals, or bases, known as A (adenine), C (cytosine), G (guanine) and T (thymine)).

Family tree

A diagram showing how members of a family are related to each other.

Information from a family tree (medical pedigree) can be important in making diagnoses and assisting clinical management. Taking and drawing a genetic family history may involve asking for information on many members of a family, or may be targeted to detect a family pattern of a particular condition, according to the clinical question.

Gene

The basic biological unit of inheritance. A gene consists of a segment of deoxyribonucleic acid (DNA) which usually codes for a protein, a component of a protein or regulatory elements. Humans have about 23,000 protein coding genes. The genes are arranged in linear order on the chromosomes.

Genetic counselling

The process by which information on genetic disorders is given to a family and supporting them in making their own decisions.

Genetic heterogeneity

The production of apparently identical disorders by different genetic mechanisms or by genes at different loci (position on a chromosome).

Heterozygote (Heterozygous)

Referring to an individual who has two different alleles, usually one normal and the other altered, at a particular locus for a particular trait or condition.

Homozygote (Homozygous)

Refers to an individual who has two identical alleles at a particular locus for a particular trait or condition.

Inheritance

Transmission from parent to child through genes.

Mutation/pathogenic variant/altered gene

The term mutation may be used to refer to any alteration in a particular gene, but should only be used to describe an alteration known to be causing a disease. Laboratory reports will refer to any genetic alteration as a variant, and one which is thought to cause disease as a pathogenic variant. In clinical practice you may also hear the term "altered gene" in place of the technical terms.

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 show complete penetrance if clinical symptoms are present in all individuals who have the disease-causing mutation. Some conditions are described as having reduced or incomplete penetrance. This means that clinical symptoms are not always present in individuals who have the disease-causing mutation. Many cardiac conditions demonstrate reduced penetrance.

Presymptomatic testing

Testing of an asymptomatic individual in whom the discovery of a gene mutation, or results of other investigations, indicates a high likelihood of their developing a specific diagnosis at some point in the future.