

## A glossary of terms

Term	Definition
Array CGH	Microarray based genomic comparative hybridisation. This is a technique used to detect chromosome imbalances by comparing patient and control DNA and comparing differences between the two sets.
Clinical exome	Differs per laboratory, but usually involves sequencing of all genes known to be associated with human disease.
<i>De novo</i>	A genetic alteration that has arisen for the first time in an individual (ie was not inherited from either parent).
Exome	The coding portion of the genes. The exome constitutes 1-2 % of the genome.
Gene panel	A collection of genes to be sequenced together, which are usually linked by common biological pathways, or known disease associations.
Genome	The entirety of an individual's genetic material including $\cong$ 20 000 genes and the genetic material between genes.
Genomic technologies	Technologies used to analyse and manipulate genomic information.
Germline mosaicism	When a genetic variant is only present in a proportion of the egg or sperm cells but not in the rest of the cells in the body.
Germline mutation	Where a genetic error occurs in the egg or sperm pre fertilization. Therefore, the genetic mutation is passed onto the offspring.
Homozygous	Both alleles of the gene contain the same variant.
Next generation sequencing	High-throughput DNA sequencing where millions of DNA bases are sequenced in parallel.
Pathogenic (variant or mutation)	A genetic variant associated with disease. Also referred to as a mutation.
Phenotype	The set of observable characteristics of an individual as a result of their genotype/environment.
Somatic mutation	A mutation that occurs in a proportion of the cells in the body except the germ cells (sperm and egg).
Variant (genetic)	A variation in the DNA sequence (from the reference sequence). The majority of genetic variation is not disease causing.
Variant of unknown significance	An alteration to the DNA sequence where it is unclear (on the basis of the available evidence) whether it is disease-causing or not.