Glossary Of Common Terms Found In Clinical Reports

OMPRN
Ontario Molecular Pathology Research Network

This glossary serves as a helpful reference tool for understanding some of the terms within your clinical report. Please note that this is not a comprehensive list, and certain definitions may be simplified. To understanding your report completely, we encourage you to talk to your doctor. Additional resources that can help you understand your report can be found on the OMPRN website (scan the QR code to visit).

Term	Definition (terms in bold are also defined in this glossary)
Allele	One of two or more versions of a gene or **DNA** sequence at a particular region on a **chromosome**. A person has two alleles for each gene, one **inherited** from each parent.
Amino Acid	An amino acid is the basic molecule that serves as the building blocks for proteins. There are 20 different amino acids. Some amino acids can be synthesized in the body; however, others (essential amino acids) must be obtained from a person's diet.
Autosomal	Is associated with any of the 22 numbered pairs of **chromosomes** found in most human cells. Autosomal chromosomes are numbered 1-22. The sex chromosomes (X and Y chromosomes) determine whether an individual is male or female and are not considered autosomal chromosomes.
Benign	Not cancer. Benign tumours may grow larger but do not spread to other parts of the body. Also called non-malignant.
Carcinoma	Cancer that begins in a part of the skin or in the layer of cells that covers internal organs.
Carrier	An individual who carries an **allele** that is linked to a disease.
Chromosome	A structure found inside the nucleus of a cell. A chromosome is made up of proteins and **DNA** organized into genes. Each cell normally contains 23 pairs of chromosomes.
DNA	The molecule inside cells that contains the genetic information needed for a person and most other organisms to develop and grow and is passed from one generation to the next. DNA is made up of two strands that twist into the shape of a spiral ladder called a double helix. Each strand has a backbone that is made up of sugar and phosphate molecules that attach to one of four bases: adenine (A), thymine (T), guanine (G), and cytosine (C). Also called deoxyribonucleic acid. *See Images 1 and 2.*
DNA Methylation	DNA methylation works by adding a chemical modification to **DNA**. Typically, this group is added to specific places on the DNA, where it blocks the proteins that attach to DNA to "read" the gene. This chemical group can be removed through a process called demethylation. Typically, methylation turns genes "off" and demethylation turns genes "on."
DNA Mutation	A mutation is a change in the **DNA** sequence. A mutation is also called a "**variant.**"
DNA Sequencing	Sequencing DNA means determining the order of the four chemical building blocks - called "bases" - that make up the **DNA** molecule. The sequence tells scientists the kind of genetic information that is carried in a particular DNA segment.
Dominant	A person receive two versions of each gene, known as **alleles**, from each parent. If the alleles of a gene are different, one allele may be preferentially expressed compared to the other – this is the dominant gene. The effect of the other allele, called **recessive**, is masked.
Epigenetics	Is the study of how your behaviors and environment can cause changes that affect the way your genes work. Unlike genetic changes, epigenetic changes may be reversible and do not change your **DNA sequence**, but they can change how your body reads a DNA sequence. **DNA methylation** is an example of an epigenetic change.
Exons	Exons are the genome's protein-coding regions and are collectively known as the exome.
Fluorescence In-situ Hybridization (FISH)	FISH is a molecular pathology technique used to detect the presence and location of specific **DNA** and **RNA** sequences within cells and tissue.
Flow Cytometry	Is a lab test used to analyze the characteristics of cells or tiny particles. During the process, a sample of cells or particles is suspended in fluid and injected into a flow cytometer machine. Approximately 10,000 cells can be analyzed and processed by a computer in less than one minute.
Frameshift Mutation	A type of insertion or deletion of DNA bases in a gene that affects how the gene functions in cells. Also called frameshift **variant**.
Genome	The complete set of **DNA** (genetic material) in an organism. *See Image 1*
Genotype	The genotype represents the genetic blueprint of an organism. It encompasses the entire selection of genetic information **inherited** from one's ancestors.

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Germline	Germline DNA refers to tissue derived from reproductive cells (egg or sperm) that become incorporated into the **DNA** of every cell in the body of the offspring. A germline mutation may be passed from parent to offspring. Also called constitutional DNA.
Heterozygous	Refers to having inherited different versions (**alleles**) of a gene from each biological parent. Thus, an individual who is heterozygous for a gene has two different versions of that gene.
Histology	The study of tissues and cells under a microscope.
Histopathology	Is the diagnosis and study of diseases of tissues and involves examining tissues and/or cells under a microscope. Pathologists are experts in histopathology.
Homologous	Refers to having inherited the same versions (**alleles**) of a gene from each biological parent. Thus, an individual who is homozygous for a gene has two identical versions of that gene.
In silico analysis	In silico studies are biological experiments carried out entirely on a computer, or via a computer simulation
Inherited	Refers to a trait or variants encoded in **DNA** and passed from parent to offspring during reproduction.
Intronic region	Introns are sections of an **RNA** transcript, or the **DNA** encoding it, that are cut out before the RNA molecule makes a protein. The sections of DNA (or RNA) that code for proteins is called **exons**.
Malignant	A term used to describe cancer. Malignant cells grow in an uncontrolled way and can invade nearby tissues and spread to other parts of the body through the blood and lymphatic system.
Nucleotide	A molecule that is the basic building block of the nucleic acids **DNA** and **RNA**. A nucleotide is made up of a nitrogen-containing base (adenine, guanine, thymine, and cytosine in DNA, and adenine, guanine, uracil, and cytosine in RNA), a phosphate group, and a sugar molecule (deoxyribose in DNA, and ribose in RNA). DNA and RNA are polymers made up of many nucleotides.
Phenotype	The phenotype constitutes the visible aspects of a person resulting from the instruction in their **genome**. In essence, the **genotype** serves as the underlying script, dictating what should be expressed at the phenotype (visible) level.
Promotor region	A region of **DNA** upstream of a gene that tells the cell when the gene should be expressed in a process called transcription. The resulting transcription produces an **RNA** molecule.
Proteomics	The study of the structure and function of proteins, including the way they work and interact with each other inside cells.
Recessive	Individuals **inherit** two versions of each gene, known as **alleles**, from each parent. In the case of a recessive trait, the alleles of the trait-causing gene are the same, and both (recessive) alleles must be present to express the trait. A recessive allele does not produce a trait at all when only one copy is present. This contrasts to a **dominant** trait, which requires that only one of the two alleles be present to express the trait.
RNA	RNA stands for ribonucleic acid. It is a type of nucleic acid found in all the cells of your body. RNA can act as a messenger that instructs a cell how to produce a protein. Certain RNAs can also perform cellular functions on their own.
Somatic	Cells that are not part of the **germline** are called somatic cells, and any changes to the **DNA** that occur within these cells are known as somatic variants. Somatic variants are not present in every cell in the body and are not passed from parent to child.
Variant	Any change in the **DNA** sequence of a cell. Variants may be caused by mistakes during cell division, or they may be caused by exposure to DNA-damaging agents in the environment. Variants can be harmful, beneficial, or have no effect. If they occur in cells that make eggs or sperm, they can be inherited; if variants occur in other types of cells, they are not inherited. Certain variants may lead to cancer or other diseases. A variant is sometimes called a mutation.

Image 1: The structure and organisation of the human genome.

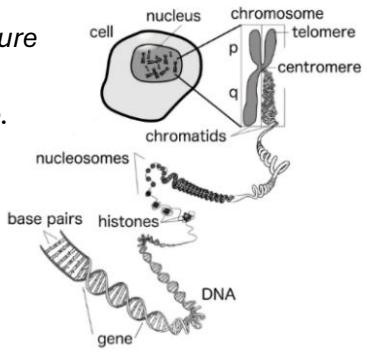

Image 2: The structure of DNA and RNA.