Common Abbreviations used in Molecular Pathology

OMPRN
Ontario Molecular Pathology Research Network

1. **CNV**
   Copy Number Variation
2. **CTC**
   Circulating Tumour Cells
3. **ctDNA**
   Circulating Tumour DNA
4. **DNA**
   Deoxyribonucleic Acid
5. **FFPE**
   Formalin-Fixed Paraffin-Embedded
6. **FISH**
   Fluorescence in-situ Hybridization
7. **IHC**
   Immunohistochemistry
8. **LOH**
   Loss of Heterozygosity
9. **MSI**
   Microsatellite Instability
10. **NGS**
    Next Generation Sequencing
11. **PCR, qPCR**
    Polymerase Chain Reaction
    Quantitative Polymerase Chain Reaction
12. **RNA**
    Ribonucleic Acid
13. **SNP, SNV**
    Single Nucleotide Polymorphism
    Single Nucleotide Variant
14. **TCGA**
    The Cancer Genome Atlas
15. **TMB**
    Tumor Mutational Burden
16. **VAF**
    Variant Allele Frequency

1

CNV

Copy Number Variation

CNV refers to a type of genetic variation where the number of copies of a particular segment of DNA varies from one individual to another. These variations can range from small deletions or duplications to larger structural changes in the genome.

2

CTC

Circulating Tumour Cells

CTCs are tumor cells that become dislodged from the primary tumor. These cells enter the bloodstream and if they land in favourable sites, they could form metastases.

3

ctDNA

Circulating Tumour DNA

ctDNA is found in the bloodstream and refers to DNA that comes from cancerous cells and tumors. Most DNA is inside a cell's nucleus. As a tumor grows, cells die and are replaced by new ones. The dead cells get broken down and their contents, including DNA, are released into the bloodstream.

4

DNA

Deoxyribonucleic Acid

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.

5

FFPE

Formalin-Fixed Paraffin-Embedded

FFPE refers to a method of preserving tissue samples for histological analysis. Tissue samples are fixed in formalin, dehydrated, and embedded in paraffin wax, allowing them to be stored for long periods while preserving cellular structure.

6

FISH

Fluorescence in-situ Hybridization

FISH is a molecular pathology technique used to detect the presence and location of specific DNA and RNA sequences within cells and tissue.

7

IHC

Immunohistochemistry

IHC is a molecular pathology technique used to visualize specific proteins in tissue samples using antibodies that bind to the target protein.

8

LOH

Loss of Heterozygosity

LOH refers to the loss of one allele at a specific genetic locus, resulting in a region of DNA becoming homozygous. LOH can occur through various mechanisms, including deletion, mutation, or chromosomal rearrangement, and is often associated with tumor suppressor gene inactivation.

9

MSI

Microsatellite Instability

MSI is a condition characterized by the accumulation of errors in microsatellite DNA sequences, which are short repetitive sequences scattered throughout the genome.

10

NGS

Next Generation Sequencing

A high-throughput method used to determine a portion of the nucleotide sequence of an individual's genome. This technique utilizes DNA sequencing technologies that are capable of processing multiple DNA sequences in parallel. Also called Pass Parallel Sequencing.

11

PCR / qPCR

Polymerase Chain Reaction
Quantitative Polymerase Chain Reaction

PCR is a laboratory technique for creating millions or billions of copies of a specific piece of DNA.

12

RNA

Ribonucleic Acid

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.

13

SNP / SNV

Single Nucleotide Polymorphism
Single Nucleotide Variant

SNPs and SNVs are variations in a single nucleotide at a specific position in the genome.

14

TCGA

The Cancer Genome Atlas

TCGA is a comprehensive database that catalogues genomic, transcriptomic, and epigenomic data from thousands of cancer patients across different cancer types. It serves as a valuable resource for cancer research and personalized medicine initiatives.

15

TMB

Tumor Mutational Burden

TMB is a measure of the total number of mutations present in the DNA of tumor cells. It is used as a biomarker to assess the overall mutational load of a tumor and may correlate with response to immunotherapy and prognosis in certain cancers.

16

VAF

Variant Allele Frequency

VAF is the proportion of sequencing reads that contain a specific genetic variant relative to the total number of reads covering that position. It provides information about the abundance of a variant within a sample.