Clinical Bioinformatics: Unlocking Genomics in Healthcare – Glossary

The glossary below provides a list of key terms used throughout the course. You do not need to read them all now; we'll be linking back to the main glossary step wherever these terms appear, so you may refer back to this list if you are unsure of the terminology being used.

Term	Definition
Autosomal dominant	**Describes inheritance pattern of a genetic condition where one copy of the variant gene is inherited from one of the patients**
Autosomal recessive	**Describes inheritance pattern of a genetic condition where two copies of the variant gene are inherited (one from each parent)**
Cloud computing	**The practice of using a network of remote servers hosted on the Internet to store, manage, and process data**
Evolutionary Sequence Conservation	**Sequence similarity is used as evidence of structural and functional conservation, and evolutionary relationships between sequences.**
ExAc	**Exome Aggregation Consortium is a coalition of investigators seeking to aggregate and harmonize exome sequencing data from a variety of large-scale sequencing projects**
Exome	**Is the entire protein coding sequence of the genome**
Gene panels	**Sets of 10-100s of genes used to identify variants in the human genome linking to specific phenotypes or conditions**
Genotype	**In its broadest sense is the genetic characteristics of an individual, when**

	referring to a particular trait it describes the variant forms of a gene that are carried by an organism
Incidental Finding (IF)	Unexpected genetic change found during sequencing of the genome
In Silico	Perform using computer modelling or simulation
Linux	Command-line based computer operating system
Locus specific Database (LSBD)	A database describing variants found at particular gene loci
Mis-sense mutation	A single base pair change that will cause the formation of an alternate amino acid at that position in the sequence
Multiple Sequence Alignment (MSA)	Multiple Sequence Alignment is generally the alignment of two or more biological sequences (protein or nucleic acid) of similar length. From the output, homology can be inferred and the evolutionary relationships between the sequences studied
Next Generation Sequencing (NGS)	The process by which millions of fragments of DNA can be sequenced in parallel from the same sample.
Nonsense variant	A single base change in the nucleotide sequence that causes the formation of a stop codon either forming a truncated protein or non-sense mediated decay of the transcript
Nonsynonymous variant	A single base change in nucleotide sequence that changes the codon leading to the formation of an alternate amino acid
Phenotype	The observed characteristics or traits of an individual
Pfam	Database of protein domain families
Reference genome sequence (human)	A digital sequence assembled from sequencing the DNA from a number of donors
Sense variant	A single base change in nucleotide sequence that encodes the same amino acid, as several codons encode for the same amino acid

Single nucleotide polymorphism (SNP)	A position in the genome where alternate bases are found
Single Nucleotide Variant (SNV)	A position in the genome where an alternate base is found in the test genome relative to the reference genome
Synonymous variant	a single base change in nucleotide sequence that encodes the same amino acid, as several codons encode for the same amino acid
Splice-site	The position of two base pairs at the intron/exon boundary by which the process of splicing occurs to produce the mature mRNA transcript
Variant of Unknown Significance	A variation in a genetic sequence whose association with disease risk is unknown
Whole exome sequencing (WES)	Sequencing of exons only within a genome by NGS
Whole genome sequencing (WGS)	Sequencing of the entire genome by NGS
X-linked	Describes the inheritance pattern of a genetic condition that is inherited on the X chromosome, hence males will definitely inherit the disorder as they only have one X chromosome whereas females may show milder symptoms of the condition depending on which genetic disorder it is.