

Glossary

- **Genome:** An individual's entire collection of DNA
- **Genetic Disease:** A disease caused by a DNA change(s)
- **Rapid Whole Genome Sequencing:** Decoding the entire genome's DNA molecules
- **Genomic Medicine:** Medicine that is informed by genome information
- **Nucleotide:** a single DNA letter (base)
- **Incidental Findings:** Genetic findings unrelated to a patient's presentation
- **DNA:** deoxyribonucleic acid, a string of nucleotides
- **Variant:** a DNA change from the (normal) reference genome sequence
- **Phenotype:** clinical features of a patient with a disease
- **Pathogenic:** A disease causing variant
- **Chromosome:** A deoxyribonucleic acid molecule in the nucleus of an individual
- **Autosomal:** Located on a chromosome that is not a sex chromosome
- **Recessive:** A disease expressed in offspring only when inherited from both parents
- **Proband:** A patient with a disorder
- **Trio:** The patient and his/her parents