Gene Screen BC

What is …

This category includes explanations of the science of genomics and the technology being used to take us into new areas of scientific discovery as well as explanations of genetic diseases. Some examples with brief definitions are listed below

Array Comparative Genomic Hybridization (aCGH or aGH)
Array comparative genomic hybridization is a technique used to detect genomic copy number variations or very small duplications or deletions in different chromosomes, known as **Copy Number Variants** (CNV's). CNVs can be pathogenic for certain conditions, but most often are benign polymorphisms that may be associated with common diseases.

Epigenetics
Genes can be influenced by the environment, which means our lifestyle can impact the expression of our genes. Epigenetics is the field that studies the relationship between our environment and our genes. It involves the study of changes in phenotype (appearance) or gene expression caused by mechanisms other than changes in the underlying DNA sequence

Gene Sequencing
This is a technique used to determine the order of the nucleotide bases (adenine, guanine, cytosine, and thymine) within the DNA molecule that covers the entire region of a gene. It can be used to produce sequence data in a wide variety of clinical and research applications. Today's gene sequencing technology is very fast, powerful and the cost is declining rapidly.

Gene Therapy
Gene therapy is a technique for correcting defective genes responsible for disease development. Researchers may use one of several approaches for correcting faulty genes. A normal gene may be inserted into a nonspecific location (using a virus or some other vector) within the genome to replace a nonfunctional gene. This approach is most common. An abnormal gene could be swapped for a normal gene through homologous recombination. The abnormal gene could be repaired through selective reverse mutation, which returns the gene to its normal function. The regulation (the degree to which a gene is turned on or off) of a particular gene could be altered.

Genotyping
Genotyping is a technique used to analyze genetic differences between individuals or cells and is used for doing genetic linkage and association studies in human families and populations.

Genomic imprinting
This is when a gene or part of a chromosome is "turned off" depending on which parent it was inherited from. During the development of the sperm or egg some genes or parts of chromosomes are "paternally stamped" when inherited from the father and some are "maternally stamped" when inherited from the mother.

MetaGenomics
Metagenomics is the study of genetic material recovered directly from environmental samples, including genetic material of bacteria and viruses from human tissues. This relatively new field of genetic research enables studies of organisms that are not easily cultured in a laboratory as well as studies of organisms in their natural environment.

What is…… (Continued)

Multifactorial Causation
Multifactorial causation refers to determination of a phenomenon by many interacting factors. For example **multifactorial inheritance** is the type of hereditary pattern seen when there is more than one genetic factor involved and, sometimes, when there are also environmental factors participating in the causation of a condition.

Polygenic Inheritance
Polygenic inheritance refers to the inheritance of quantitative traits which are influenced by multiple genes, not just one. It also looks at the role of environment in someone's development. As many traits are spread across a continuum, rather than being divided into black and white differences, polygenic inheritance helps explain the way in which these traits are inherited and focused. A related concept is **Pleiotropy**, an instance where one gene influences multiple traits.

Rare Diseases
A rare disease is defined as having a frequency of less than 1 in 20,000. There are over 7,000 known rare diseases and many more are undiagnosed. Collectively 1 in 10 Canadians are affected in some way.

Uniparental Disomy
Uniparental disomy (UPD) arises when an individual inherits two copies of a chromosome pair from one parent and no copy from the other parent.

X-Inactivation
X-inactivation (also called lyonization) is a process by which one of the two copies of the X chromosome present in females is inactivated.