Genomics & Precision Health

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Glossary of Terms

Allele

An allele is the version of the gene that is present. Each person has two alleles for each gene, one from each parent. If the alleles of a gene are the same, the person is homozygous for the gene. If the alleles are different, the person is heterozygous for the gene.

Chromosome

DNA is packaged into small units called chromosomes. A chromosome contains a single, long piece of DNA with many different genes. Every human cell contains 23 pairs of chromosomes. There are 22 pairs of numbered chromosomes, called autosomes, and one pair of sex chromosomes, which can be XX or XY. Each pair contains two chromosomes, one from each parent, which means that children get half of their chromosomes from their mother and half from their father.

Copy number variation (CNV)

A copy number variation (CNV) is when the number of copies of a gene or other section of DNA is different between people.

DNA

Deoxyribonucleic acid (DNA) contains the genetic instructions in all living things. DNA is made up of two strands that wind around each other and looks like a twisting ladder (a shape called a double helix). A DNA strand has four different bases arranged in different orders. These bases are T (thymine), A (adenine), C (cytosine), and G (guanine). DNA is “read” by the order of the bases, that is by the order of the Ts, Cs, Gs, and As. The specific order, or sequence, of these bases determines the exact information carried in each gene (for example, instructions for making a specific protein). DNA has the same structure in every gene and in almost all living things.

DNA Methylation

DNA methylation is a chemical addition to a piece of DNA that turns it on or off.

DNA Mutation

A mutation is a change in a DNA sequence. DNA mutations in a gene can change what protein is made. Mutations present in the eggs and sperm (germline mutations) can be passed on from parent to child, while mutations that occur in body cells (somatic mutations) cannot be inherited.

Dominant

Dominant diseases can be caused by only one copy of a gene with a DNA mutation. If one parent has a disease, each child has a 50% chance of inheriting the mutated gene.

Environmental Factors

Environmental factors can include exposures related to where we live as well as behaviors such as smoking and exercise and cultural factors such as foods that we eat.

Epigenetics

Epigenetics is the study of changes in phenotype caused by something other than changes in the underlying DNA sequence (for example, DNA methylation).

Gene

A gene is a part of DNA that carries the information needed to make a protein. People inherit one copy of each gene from their mother and one copy from their father. The genes that a person inherits from his or her parents can determine many things. For example, genes affect what a person will look like and whether the person might have certain diseases.

Gene Expression

Gene expression refers to the process of making proteins using the instructions from genes. Changes in gene expression can affect how much of a protein is made, as well as when the protein is made.

Genomics
Genomics refers to the study of all of the genetic material in an organism.

Genotype
The genotype of a person is her or his genetic makeup. It can also refer to the alleles that a person has for a specific gene.

Metabolites
Metabolites are the chemicals that are produced by the cells in the body when they break down sugars, fats, and proteins to make energy.

Phenotype
Phenotype is how a person looks (on the outside and inside the body) due to his or her genes and the environment (for example, having a certain eye color, being a specific blood type, or being a certain height). Phenotype also can refer to how a person's body functions, for example, whether he or she has a certain disease.

Protein
A protein is made up of building blocks called amino acids. The main role of DNA is to act as the instructions for making proteins. It is actually proteins that make up most of the structures in our bodies and perform most of life's functions. For example, proteins make up hair and skin. Proteins in our eyes change shape in response to light so we can see. Proteins in our bodies break down food. Proteins are made in cells and are the major parts of cells, which are the vital working units of all living things.

Recessive
For recessive diseases, both copies of a gene must have the DNA mutation for a person to have one of these diseases. If both parents have one copy of the mutated gene, each child has a 25% chance of having the disease, even though neither parent has it. In such cases, each parent is called a carrier of the disease. They can pass the disease on to their children, but do not have the disease themselves.

Single nucleotide polymorphism (SNP)
Single nucleotide polymorphisms (SNPs) are changes at a single DNA base that are present among at least 1% of people in at least one population. For example, at a given DNA location, some people will have one base (e.g., adenine), while other people will have a different base (e.g., guanine). The SNP that is more common among a given group of people is called the major allele and the one that is less common is called the minor allele.

Related Links

- Birth Defects
- Developmental Disabilities
- Genomics Family Health History
- Genomics
- Epigenetics

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