GENETICS FORUM: Glossary of genetic terminology

Adult-onset condition:	A change in a person's DNA that increases his/her risk for developing a specific health condition later in life, often in middle age (40s-60s).
Carrier Status:	An individual has a DNA change for a health condition but does not display symptoms. In order to have the health condition, an individual must have inherited DNA changes from both parents. An individual having one normal gene and one DNA change does not have the disease. Two carriers may produce children with the health condition.
Carrier screening:	A type of genetic testing performed on people who display no symptoms for a genetic disorder but may be at risk for passing it on to their children.
Cell:	The basic building block of living things found in all of the tissues and organs in your body.
Chromosome:	An organized section of DNA found in all of person's cells. Humans have 23 pairs of chromosomes. Each parent gives one chromosome to each pair so that a child gets half of their chromosomes from their mother and half from their father.
DNA:	(Deoxyribonucleic Acid) The chemical name for the molecule that carries genetic instructions in all living things.
DNA Change:	This refers to a change in a DNA sequence, often called a mutation or variant. These changes can be present when a person is born or can happen during their lifetime. Changes can result from DNA copying mistakes made during cell division, exposure to radiation, chemicals, or infection by viruses.
Dominant:	A DNA change that is almost always expressed, even if only one copy is present.
Genome sequencing:	A laboratory technique used to determine the exact sequence of DNA, including any changes in a DNA sequence.
Gene:	The basic physical unit of inheritance. Genes are passed from parents to children and contain the information needed to specify personal characteristics. Humans have approximately 20,000 genes on their chromosomes.

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Genetic Information Nondiscrimination Act (GINA)	Federal legislation that makes it unlawful to discriminate against individuals on the basis of their genetic information in regard to health insurance and employment.
Genetics:	The study of genes and heredity (see **Heredity**) in an organism.
Genetic testing:	A laboratory method that looks at DNA changes in your genes. Genetic tests are used to diagnose genetic conditions, identify carriers and individuals at increased risk. Genetic test results can be used to make decisions about screening, management of care and potential response to medications.
Genetic variation:	Every person has a unique set of DNA. We share much of our DNA with other humans and with every living thing. The small set of DNA changes that differ between humans is called genetic variation. This is what gives you your traits (see **Trait**).
Genome:	All of a person's genetic information.
Genomics:	The study of the entire genome of an organism.
Heredity:	The transmission of personal characteristics from one generation to the next through the genes.
Human Genome Project:	An international project that mapped and sequenced the entire human genome. It was completed in April 2003.
Inherited:	Passed down from parents to children through generations. The genes present in the parents are passed down to their children through egg or sperm cells.
Medically actionable:	This is when a change in a person's DNA increases the risk for developing a specific health condition, where actions can be taken to prevent, delay, or reduce symptoms of the health condition.
Nucleotide:	The building blocks of DNA that contain one of the letters A, T, G or C.
Pathogenic:	This is when a DNA change causes a health condition.

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Pharmacogenetics:	The study of the interaction of an individual's genetic makeup and response to a drug.
Protein:	Proteins are required for the structure, function, and regulation of the body's cells, tissues, and organs; each protein has unique functions. Examples are hormones, enzymes, and antibodies.
Recessive:	Two DNA changes are needed in the same gene one from your mom and one from your dad. For a male, if one copy is present on the X chromosome (see **Chromosome**) he gets from his mother this is also recessive.
Reference genome:	This is a representative set of DNA sequences. When a new person has their genome sequenced, it is compared to a reference genome to see what is unique to that person.
Risk:	In the area of genetics, this is the probability that an individual will develop a particular health condition. Both genes and environment influence risk.
Secondary findings:	These are health conditions that you did not know you have a higher chance of developing. They are discovered unintentionally and are unrelated to the reason the genetic test is being performed. These are sometimes called "incidental findings".
Targeted treatment:	This is when medication or medical procedures are given differently to people who have the same health condition but different DNA changes.
Trait:	A specific characteristic like height, eye color, personality, or abilities. This also includes personal risk for a health condition. Traits are determined by genes or by the environment or, more commonly, by interactions between genes and the environment.
Variant of unknown significance (VUS):	Scientists and doctors do not know what many DNA changes that can be found by genome sequencing mean. A VUS is a DNA change that may or may not lead to a health condition or other harmful trait (see **Trait**) but at this time, we do not know enough information about the DNA change to predict what it means.

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