National Society of Genetic Counselors

aboutgeneticcounselors.com

Cardiology

Glossary Terms

Autosomal:

Autosomal: a trait or condition caused by a gene on an "autosome", meaning chromosomes 1 through 22. In other words, a characteristic not connected with the X or Y chromosome. Examples of autosomal genetic conditions include: Cystic Fibrosis and Sickle Cell Anemia.

Benign Variant:

Benign variant: a DNA change that does not affect a gene's function or cause a health problem. We all have many benign variants that make us unique from each other that do not cause medical issues.

Cardiomyopathy:

Cardiomyopathy: a condition in which part or all of the heart muscle is thicker than normal. This may cause heart rhythm problems, breathlessness, and chest pain, but some people are symptom-free. Certain types of this condition have a known genetic cause.

Diagnostic Testing:

Diagnostic testing: a medical test that provides information that allows a physician to diagnose a condition.

DNA Testing:

DNA testing: a form of genetic test that specifically analyzes DNA. This type of testing may be used to identify DNA changes that can cause disease, or it may be done to learn other genetic information about an individual, such as paternity or ancestry.

Dominant:

Dominant: a form of a gene that, when present, overpowers the alternate (recessive) form. This is also referred to as the dominant allele.

Familial:

Familial: a condition where there is a strong family history, but there is not an identified genetic cause.

Family History:

Family history: Information about health and medical issues in relatives. Family history is one of the most important tools a genetic counselor uses to identify genetic risk. Patterns of specific health issues in an individual's family history are useful to determine the risk of disease and appropriate genetic testing options.

Genetic Counselor:

Genetic Counselor: a healthcare professional who has training in genetics and counseling who works alongside doctors, nurses, or other members of your healthcare team. Genetic counselors assess your medical history and family history to determine if you are at risk for a genetic or hereditary condition and discuss the implications of available genetic testing options.

Genetic Risk:

Genetic risk: the contribution our genes play in the chance that we will develop a certain illness or condition.

Genetic Testing:

Genetic testing: analyzing a person's genetic material (i.e. chromosomes or genes) in order to identify changes which could lead to genetic disease or predisposition to disease.

Genotype:

Genotype: describes your genetic makeup. This term is usually used when referring to a particular trait.

Germline Testing:

Germline testing: testing typically performed on a blood or saliva sample to look for inherited genetic changes or changes that someone has from birth, which can be passed down from one generation to the next.

Genetic Information Nondiscrimination Act (GINA):

Genetic Information Nondiscrimination Act (GINA): a federal law passed in 2008 that protects people from their genetic information being misused and impacting their health insurance coverage or employment.

Hereditary/Inherited:

Hereditary/Inherited: a condition or disease that can be passed down from one generation to the next. Inherited conditions are determined by genetic factors.

Informed Consent:

Informed consent: the process of learning key details about a medical treatment (such as a drug,

surgery or test) to be able to decide whether or not to have the treatment or test.

Likely Pathogenic Variant:

Likely pathogenic variant: a DNA change we think likely causes a gene not to work and may lead to a health problem. Usually, scientists and doctors are 90% sure these cause disease, but not enough evidence has been collected to be 100% sure yet.

Mutation (Genetic Mutation):

Mutation (genetic mutation): a change in DNA from its original state. Some mutations do not cause any problems, while others result in disease; however, this term is most often used when an individual has a genetic change that causes a health condition.

Pathogenic Variant:

Pathogenic variant: a change in the DNA that is known to cause a gene not to work and may lead to a health problem.

Pedigree:

Pedigree: a family tree using established symbols. Genetic counselors use pedigrees to review the family history and look for patterns that might suggest a risk for genetic disorders or health-related conditions in the family.

Recessive:

Recessive: a form of a gene that is masked by the alternate (dominant) form of the gene and is only

expressed when the person has two of these non-dominant (recessive) copies. This is also referred to as the recessive allele.

Sequencing:

Sequencing: a test that reads through the DNA code letter by letter to look for changes that can cause health problems. This test can be done on a single gene, a panel of genes, or on even more of the DNA (genome or exome).

X-linked:

X-linked: trait or condition caused by a gene on the X-chromosome.

SNP (Single Nucleotide Polymorphism) Testing:

SNP (single nucleotide polymorphism) testing: testing that looks for single letter changes within the DNA. (This is different than sequencing an entire gene or genome)

Variant:

Variant: a change in your DNA instructions (like a change in the spelling of a word) that is different than what most people have. It may or may not change the way the gene works.

VUS (Variant of Uncertain/Unknown Significance):

VUS (Variant of Uncertain/Unknown Significance): a change in the DNA instructions that scientists and doctors are unsure yet if it causes a health problem.