National Society of Genetic Counselors

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Genetic Testing

Glossary Terms

Amniocentesis:

Amniocentesis: a test done by taking fluid from the amniotic sac during pregnancy to test for conditions in the fetus.

Allele:

Allele: one of the possible forms of a gene.

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.

Buccal Sample:

Buccal sample: a way to collect DNA from the cells on the inside of a person's cheek, either through a saliva sample or cheek swab.

Carrier Testing/Screening:

Carrier testing/screening: a medical test that is typically performed for family planning which helps intended parents know if they carriers for certain inherited genetic conditions, and if they have carrier conditions in common, whether their children could be at-risk.

CGC (Certified Genetic Counselor):

CGC (Certified Genetic Counselor): a credential held by genetic counselors in the United States that is received after passing an examination that demonstrates their skills and knowledge as a healthcare professional.

Chorionic Villus Sampling:

Chorionic villus sampling (CVS): a test done by taking cells from the placenta in early pregnancy to detect genetic conditions in the fetus.

Chromosome:

Chromosome: DNA is packaged into structures called chromosomes, found in the cells of living things. Humans have 46 chromosomes that come in 23 pairs. The first are numbered 1-22, and are called "autosomes". The last chromosomes pair are called sex chromosomes: females usually have XX chromosomes, and males usually have XY.

DNA:

DNA: the building block of life contained in each of our cells that carries our genetic information.

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.

Direct-to-Consumer Testing:

Direct-to-consumer testing: also known as at-home DNA testing or DTC testing, these are tests that are marketed directly to consumers and enables them to purchase various types of genetic tests and receive the results without the involvement of a personal physician or insurance company.

Exome:

Exome: the portion of DNA which contains the genes, which is approximately 1-2% of all our DNA.

Gene:

Gene: a segment of DNA which contains a specific instruction for the body. This tells the body how to make a protein, which is a building block of the body. Genes are inherited and also determine characteristics of an individual, such as eye color or whether or not they have certain genetic diseases.

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: 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.

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.

Personalized Medicine:

Personalized medicine: term used to describe an emerging healthcare approach that tailors disease prevention and/or treatment based on an individual's genes, environment, lifestyle, and other factors.

Preimplantation Genetic Testing (PGT):

Preimplantation genetic testing (PGT): screening test performed during in vitro fertilization (IVF). There are three types: PGT-M, PGT-SR, and PGT-A, each involving testing of an embryo for certain genetic conditions (such as cystic fibrosis or Down syndrome). This type of testing was formerly called preimplantation genetic diagnosis (PGD).

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)

Tumor/Somatic Testing:

Tumor/somatic testing: testing that is done on cells from a tumor or cancer to look for DNA changes. Knowing the DNA profile of a tumor can help doctors tailor treatment. Most DNA changes found in a tumor are not inherited and not present in a person's healthy cells.

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.