National Society of Genetic Counselors

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Pediatric/Children

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.

Birth Defect:

Birth defect: a physical difference that is present at or before birth.

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.

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.

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.

Congenital:

Congenital: a term that describes a condition that is present at birth.

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.

Dominant:

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

Dysmorphic:

Dysmorphic: a term used to describe physical features, particularly facial, that are not usually found in an individual of the same age or ethnic background.

Exome:

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

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.

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.

Gene Therapy:

Gene therapy: a treatment in which a non-working gene is replaced with a working copy of the gene in order to prevent or correct a known genetic disorder.

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.

Geneticist:

Geneticist: a doctor who has completed a genetics residency or fellowship and has also passed the medical board certification/examination for genetics. A clinical geneticist evaluates patients at risk for inherited diseases and treats inherited disorders.

Genome:

Genome: All of someone's DNA. The genome includes all of a person’s genes, as well as the non-coding areas of DNA found in-between the genes.

Genotype:

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

Germline:

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.

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.

Mitochondrial Inheritance:

Mitochondrial inheritance: genes located in the mitochondria that are passed from the mother to the child, the father does not pass these down.

Multifactorial Conditions:

Multifactorial conditions: medical conditions caused by multiple genetic contributions in addition to the environment.

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.

Phenotype:

Phenotype: describes the features you have as a result of your DNA.

Rare Disease:

Rare disease: a disease that affects less than 200,000 Americans at any given time. Note that different countries may use different numbers to describe a rare disease or disorder.

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.

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)

Sporadic:

Sporadic: usually refers to a condition that happened for the first time in a person, which wasn't inherited from their parents.

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.


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