List of Common Genetic Terms and Acronyms

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AAP	American Academy of Pediatrics
ACA	Patient Protection and Affordable Care Act
ACMG	American College of Medical Genetics and Genomics- Professional organization that sets standards of practice for medical geneticists as well as standards and guidelines for laboratory genetics
ACOG	American College of Obstetrics and Gynecology- Professional organization that develops standards of care during pregnancy, which includes recommendations about prenatal screening for possible genetic conditions
ACT	ACMG Action sheets and their accompanying algorithms are a great resources for health care providers looking for information on genetic conditions (identified through newborn screening and beyond) to help inform clinical decision making (ACMG)
ADA	Americans with Disabilities Act
AFP	Alpha FetoProtein- a substance made by a developing fetus which can be measured in a pregnant woman's blood or amniotic fluid. Unusually high or low values for the length of pregnancy may suggest the baby has a condition such as spina bifida or Down sydrome or certain other conditions
Allele	The alternative forms of genes which occur at the same site on a chromosome and which determine alternative forms of a trait
AMA	American Medical Association
Amino Acids	A set of 20 different molecules of a particular type which make up all the proteins in the human body or other living things
Autosomal dominant disorder	Produced by a single mutated dominant allele, even though its corresponding allele is normal
Autosomal recessive disorder	Develop in persons who receive two copies of the mutant gene, one from each parent who is a carrier
Carrier	For some genetic disorders, it takes two genes for a person to have the disorder. A carrier is a person who has only one gene for a disorder. Carriers usually do not have symptoms or have only mild symptoms. They often do not know that they have a gene for a disorder (ACOG)
Carrier Testing	a type of genetic test that can tell you whether you carry a gene for certain genetic disorders. When it is done before or during pregnancy, it allows you to find out your chances of having a child with a genetic disorder (ACOG)
CDC	Center for Disease Control and Prevention
Chromosome	The cellular structure that stores and transmits genetic information
CNV/Copy number variation	A segment of DNA in which copy number differences have been found by comparison of two or more genomes. The segment may range from one kilobase to several megabases in size. The variation is usually due to deletion or duplication
Comorbidity	The term can indicate either a condition existing simultaneously, but independently with another condition or a related medical condition
Compound heterozygosity	Heterozygosity for two different mutant alleles of a gene, often the case for autosomal recessive disorders
CPT	Current Procedural Terminology
CSHN	Children with special health care needs
CVS	Chorionic Villus Sampling
De novo	A genetic alteration that is present for the first time in one family member as a result of a variant (or mutation) in a germ cell (egg or sperm) of one of the parents, or a variant that arises in the fertilized egg itself during early embryogenesis. Also called de novo variant, new mutation, and new variant.
Deletion	The loss of a portion of a chromosome as a result of chromosome breakage
DNA	Deoxyribonucleaic acid- a large molecule that carries the genetic information necessary for the replication of cells and for production of proteins which are essential for growth, the building of new tissue, and the repair of injured or broken -down tissue
Dominant allele	A gene that is expressed, regardless of whether its counterpart allele on the other chromosome is dominant or recessive
EMR	Electronic medical record
Enzyme	A protein that facilitates a specific chemical reaction
Epigenetics	Heritable changes to DNA structure that do not alter the underlying DNA sequence, eg, DNA methylation
Exome	The approximately 1% of the human genome that comprises all exons and therefore the entire protein-coding region of the genome
F2F	Family-to-Family Health Information Centers
FISH	Flourescent In-Situ Hybridization- A specialized lab approach used to study chromosomes
Gene	A unit of inheritance, a working subunit of DNA. Each of the body's 50,000-100,000 genes contains the code for a specific product, typically, a protein such as an enzyme
Gene Editing	a group of technologies that give scientists the ability to change an organism's DNA. These technologies allow genetic material to be added, removed, or altered at particular locations in the genome (GHR)
Gene Therapy	an experimental technique that uses genes to treat or prevent disease. In the future, this technique may allow doctors to treat a disorder by inserting a gene into a patient’s cells instead of using drugs or surgery (GHR)
Genetic Testing	a type of medical test that identifies changes in chromosomes, genes, or proteins. The results of a genetic test can confirm or rule out a suspected genetic condition or help determine a person’s chance of developing or passing on a genetic disorder. More than 1,000 genetic tests are currently in use, and more are being developed (GHR)
Genome	All the genetic material in the chromosomes of a particular organism
Genotype	An individuals specific set of alleles for any given set of genes, whether they are dominant or recessive
Heterozygous	Having different alleles for one or more genes in homologous chromosome segments
Homozygous	Having identical alleles for one or more genes in homologous chromosome segments

HRSA	The Health Resources and Services Administration (HRSA), an agency of the U.S. Department of Health and Human Services, is the primary federal agency for improving health care to people who are geographically isolated, economically or medically vulnerable.
IDEA	Individuals with Disabilities Education Act
Marker	A detectable genetic variant
Methylation	The attachment of a methyl group to DNA. In vertebrates, this typically occurs at CpG sites (cytosine-phos-phate-guanine sites) in the DNA sequence, resulting in the conversion of cytosine to 5-methylcytosine
Monogentic Disorders	Disorder caused by one or more mutations in a single gene, eg, cystic fibrosis (mutations in the CFTRgene). Such disorders are also sometimes referred to Mendelian diseases
Mutation	A change in the number, arrangement, or molecular sequence of a gene
NBS	Newborn screening
NCC	In 2017, the Maternal and Child Health Bureau of the Health Resources and Services Administration (MCHB/HRSA), Genetic Services Branch (GSB) awarded grants to establish the seven Regional Genetics Networks (RGNs), a National Coordinating Center (NCC), and the National Genetics Education and Family Support Center (NGEFSC) as part of on-going efforts to improve the health of medically underserved by promoting the translation of genetic medicine into public health and health care services.
NIH	National Institute of Health
NORD	National Organization for Rare Disorders
NTD	Neural Tube Defect- refers to the incomplete closing of the neural tube (which develops into the spinal cord) during development of the embryo early in pregnancy
PCP	Primary care provider
Pedigree	A diagram showing a genetic family history and biological relationship among members of a family, often for several generations
Penetrance	A term indicating the likelihood that a given gene will acutally result in a genetic disorder or disease
Pharmacogenetics	A branch of genetics which deals with the genetic variability in individual responses to drugs and drug metabolism
Phenotype	The observable properties (structural and functional) of an organism, produced by the interaction between the organism's genotype and the environment in which it finds itself
Polymorphism	A chromosome or DNA variant that is observed in natural populations. A gene locus is defined as polymorphic if a rare allele has a frequency of 0.01 (1%) or more
Recessive Allele	A gene that is expressed only when its counterpart allele on the matching chromosome is also recessive
SNP	Single Nucleotide Polymorphism- DNA sequence variations that occur when a single nucleotide (adenine, thymine, cytosine, or guanine) in the genome sequence is altered; usually present in at least 1% of the population (NIH/NCI)
Structural Variant	Structural genomic variation includes any genetic variant that alters chromosomal structure, including inversions, translocations, duplications and deletions. Duplications and deletions, collectively known as CNVs (see copy number variation) are the most common form of structural variation in the human genome
Translocation	A type of chromosomal abnormality in which a chromosome breaks and a portion of it reattaches to a different chromosomal location (NIH/NCI)
Trisomy	The presence of an extra chromosome, resulting in a total of three copies of that chromosome instead of the normal 2 copies (e.g., trisomy 21, or Down syndrome) - NIH/NCI
VUS- Variant of Unknown Significance	An allele, or variant form of a gene, which has been identified through genetic testing, but whose significance to the function or health of an organism is not known. See links below for more information:
	https://www.ashg.org/education/csertoolkit/uncertainresults.html
	https://www.mountainstatesgenetics.org/wp-content/uploads/sites/257/2018/10/Variants-of-Uncertain-Significance-Final-Oct-2018.pdf
X-linked inheritance	Any gene found on the X chromosome or traits determined by such genes