

Education & Information

Genetics Information

Genetic Alliance

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Alphabet Soup

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Genetics for Genetics Consumers - Terms and Acronyms

AAP [American Academy of Pediatrics](#), a professional organization. It has developed standards of care for some genetic conditions.

ABCG [American Board of Genetic Counseling](#). It provides certification for master's level genetic counselors.

ABMG [American Board of Medical Genetics](#), a specialty board of the [American Board of Medical Specialists](#). It accredits training programs in medical genetics and certifies medical geneticists (doctorate level practitioners).

ACMG [American College of Medical Genetics](#), a professional organization. It sets standards of practices for medical geneticists and sets standards and guidelines for laboratory genetics.

ACOG [American College of Obstetrics and Gynecology](#), a professional organization. It develops standards of care during pregnancy, which include recommendations about prenatal screening for possible genetic conditions.

ADA [Americans with Disabilities Act](#), a federal civil rights law. It protects people with disabilities and genetic predispositions from discrimination in a number of areas.

AFP Alpha-feto Protein. This is a "substance made by a developing fetus which can be measured in a pregnant woman's blood or amniotic fluid" (Weiss & Mackta, 1996, p.137). Unusually high or low values for the length of the pregnancy may suggest the baby has spina bifida or Down syndrome or certain other conditions.

AG Attorney General. This is the head lawyer for country, state, or federal governments.

Allele Alleles are "the alternative forms of genes which occur at the same site on a chromosome and which determine alternative forms of a trait" (Weiss & Mackta, 1996, p.136).

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 (NCI, 1995).

Amniocentesis "The removal of a sample of amniotic fluid, the fluid surrounding the growing baby (fetus)" (Weiss & Mackta, 1996, p.137).

ART Assisted Reproductive Technology. These are highly sophisticated techniques to help couples who have longstanding difficulties conceiving a baby. It includes in-vitro fertilization.



ASHG [American Society of Human Genetics](#), a professional organization. Members include research scientists as well as clinicians in the field of genetics.

Autosome "Chromosomes other than the sex (X or Y) Chromosomes. Humans have 22 pairs of autosomes, numbered 1 through 22" (Weiss & Mackta, 1996, p.137)

BRCA1 "A gene that normally helps to restrain cell growth" (NCI, 1995, p.25)

BRCA1 breast cancer susceptibility gene "A mutated version of BRCA1, which predisposes a person towards developing breast cancer" (NCI, 1995, p.25).

CAP [College of American Pathologists](#), a professional organization. (Many laboratories are managed by pathologists.) This group sets standards for laboratories, although the standards are not specific to genetic testing. This organization does **proficiency testing** of laboratories to be sure they can get reliable results and interpret their results accurately. This is one part of the review of laboratories to be sure they meet [CLIA](#) standards for federal licensing.

Carrier Testing The goal of these tests is identifying individuals, or couples, who carry a particular recessive gene allele that would result in a particular genetic condition if a child were to inherit copies of this allele from both parents. "Carrier testing is designed for healthy people who have no symptoms of disease, but who are known to be at high risk of being a carrier for the disorder" (NCI, 1995, p.25)

CBA Cost-Benefit Analysis. This is an approach to measuring costs versus benefits in monetary terms in setting public policies regarding health issues.

CDC [Centers For Disease Control and Prevention](#). This is an agency of the federal government that has shown increasing interest in the public health implications of genetics.

CEA Cost-Effectiveness Analysis. This is a way to compare different approaches which might yield a similar health benefit. Measures may include years of life, quality-adjusted life years, the number of people with a condition, or cost per year of life saved.

Chromosome The cellular structure that stores and transmits genetic information. Chromosomes are composed of genes linked together in specific arrangements. The normal number of chromosomes in humans is 46. (Weiss & Mackta, 1996, p.138)

CLIA '88 Clinical Laboratory Improvement Act of 1998. This legislation is the basis for the regulations which must be met for a laboratory to receive a federal license. However, the regulations are not specific to genetic testing.

Clone A group of identical genes, cells, or organisms derived from a single ancestor (NCI, 1995, p.26)



CMM [Centers for Medicare & Medicaid Services](#)

CPT Physicians Current Procedural Terminology. The American Medical Association provides a set of five digit codes for medical procedures to be used for insurance purposes. The codes reflect what system of the body was involved and what service was provided. These codes facilitate processing of insurance claims. There are also associated diagnosis codes.

CRG [Council for Responsible Genetics](#). This is an advocacy group which brings together scientists, educators, journalists, students, and policy makers on issues related to the uses and potential misuses of genetics.

Crossing Over "A phenomenon, also known as recombination, that sometimes occurs during the formation of sperm and egg cells (meiosis); a pair of chromosomes (one from the mother and the other from the father) break and trade segments with one another" (NCI, 1995, p.26)

CVS Chorionic Villus Sampling. This is a diagnostic procedure sometimes used early in pregnancy (at 9 to 11 weeks of growth) to detect certain genetic conditions.

Deletion "The loss of a portion of a chromosome as a result of chromosome breakage" (Weiss & Mackta, 1996, p.138)

DNA Deoxyribonucleic Acid. It is "the substance of heredity; a large molecule that carries the genetic information necessary for the replication of cells and for the production of proteins, which are essential for growth, the building of new tissue, and the repair of injured or broken-down tissue." (Weiss & Mackta, 1996, p.138)

DNA Probe "A specific sequence of single-stranded DNA used to seek out a complementary sequence in other single strands." (Weiss & Mackta, 1996, p.138)

DOE [Department of Energy](#). This part of the federal government is one source of funding for the [Human Genome Project](#).

Dominant Allele "A gene that is expressed, regardless of whether its counterpart allele on the other chromosome is dominant or recessive. Autosomal dominant disorders are produced by a single mutated dominant allele, even though its corresponding allele is normal." (NCI, 1995, p.26)

ELSI Ethical, Legal, and Social Implications/Issues. This originated as a portion of the Human Genome Project that provides funding to examine the broader implications of the identification of genes. This has included the ELSI working group and several task force projects to address some of these concerns plus grants for a variety of independently proposed projects.

Enzyme "A protein that facilitates a specific chemical reaction." (NCI, 1995, p.26)



FDA [Food and Drug Administration](#), a federal agency that oversees the safety of foods, drugs, and cosmetics (1938) and medical devices (1976, 1990).

FISH Fluorescent In-Situ Hybridization. This is a specialized laboratory approach used to study chromosomes.

Gene "A unit of inheritance, a working subunit of DNA. Each of the body's 50,000 to 100,000 genes containst the code for a specific product, typically, a protein such as an enzyme." (NCI, 1995, p.27)

Gene mapping This process is part of the [Human Genome Project](#). Researchers have created a working draft of a schematic map which shows the order and location in which genes, genetic markers, and other landmarks are found along the chromosomes.

GENES [Genetic Network of New York State, Puerto Rico, and the Virgin Islands](#), one of the 10 regional genetics networks.

Genetic Testing Examining a sample of blood or other bodily fluid or tissue for biochemical, chromosomal, or genetic markers that indicate the presence or absence of genetic mutations or genetic conditions, including evidence of carrier status or a predisposition to developing a condition which has multifactorial components.

Gene Therapy This refers to a medical procedure that treats a disorder by replacing the faulty gene.

Genetic Engineering Scientists are altering genetic material to study genetic processes and, potentially, to correct genetic defects.

Genome "All the genetic material in the chromosomes of a particular organism" (NCI, 1995, p.27), such as the set of genes that make up the human organism (the human genome) or those that make up other creatures, such as a fruit fly (the fruit fly genome) or a mouse (the mouse genome).

Genotype This refers to an individual's specific set of alleles for any given set of genes, whether they are dominant or recessive. This might refer to one set of genes or the entire set of genes that a person has.

GLaRGG [Great Lakes Regional Genetics Group](#) - Illinois, Indiana, Michigan, Minnesota, Ohio, Wisconsin, one of ten regional genetics networks.

GPGSN [Great Plains Genetic Service Network](#) - Arkansas, Iowa, Kansas, Missouri, Nebraska, Oklahoma, North Dakota, South Dakota, one of the ten regional genetics networks.

IVF-ET In-Vitro Fertilization with Embryo Transfer. This is an assisted reproductive



technology technique. After the egg and sperm are united in the laboratory, the fertilized egg (embryo) is placed in (transferred to) the uterus of a woman for development.

JCAOH Joint Committee on Accreditation of Hospitals. This organization sets national standards for hospitals, which also impacts standards for laboratories associated with hospitals.

Karotype A karotype is "a systemized array of chromosomes from a single cell, prepared by photography, that demonstrates the number and morphology of the chromosomes" (Weiss & Mackta, 1996, p.139).

LEA Local Education Agency. This is a term used in federal laws relating to education of students with disabilities. It usually means the local school district.

Linkage "The relationship between two genes, or between an identifiable trait and a genetic disorder. Genes that are located relatively close to each other on the same chromosome are said to be linked." (Weiss & Mackta, 1996, p.139)

Linkage Analysis "A gene-hunting technique that traces patterns of heredity in large, high risk families, in an attempt to locate a disease-causing gene mutation by identifying traits that are co-inherited with it" (NCI, 1995, p.28)

LRE Least Restrictive Environment. This is a term used in federal laws relating to education of students with disabilities. It refers to a placement decision about the choice of the most appropriate educational setting for a particular student - the one closest to the regular education environment - as long as appropriate supports or modifications are provided which enable this student with a disability to benefit from this educational setting.

MARHGN [Mid-Atlantic Regional Human Genetics Network](#) -- Delaware, District of Columbia, Maryland, New Jersey, Pennsylvania, Virginia, West Virginia, one of the ten regional genetics networks.

Marker "A detectable genetic variant. Some closely linked markers can be used to deduce the presence or absence of disease-producing genes" (Weiss & Macta, 1996, p.139)

MCHB [Maternal and Child Health Bureau](#), an agency under Title V, Health Resources and Services Administration, within the federal Department of Health & Human Services. This agency is the source of the funding grants for the regional genetics networks and other state and national programs related to genetics and maternal and child health.

MDS Minimum Data Set. This refers to data which must be collected by the regional genetics networks from full service genetics centers. This information is compiled as a national report on genetic services.



MOD [March of Dimes](#). This is a national organization which focuses its literature, activities, and funding on disorders present at birth and a number of genetic conditions.

Molecule "A group of atoms arranged to interact in a particular way; one molecule of any substance is the smallest physical unit of that particular substance." (NCI, 1995, p.28)

MoStGeNe [Mountain States Genetics Network](#) - Arizona, Colorado, Montana, New Mexico, Utah, Wyoming, one of the ten regional genetics networks.

MSAFP Maternal Serum Alpha-Fetoprotein. This refers to alpha-fetoprotein found in the pregnant woman's blood (i.w. maternal serum).

Multifactorial "Caused by many genetic and nongenetic factors" (Weiss & Mackta, 1996, p.139)

Mutation "A change in the number, arrangement, or molecular sequence of a gene" (NCI, 1995, p.28)

NERGG [New England Regional Genetics Group](#) - Connecticut, Maine, Massachusetts, New Hampshire, Rhode Island, Vermont, one of the ten regional genetics networks.

NHGRI [National Human Genome Research Institute](#). This part of the [National Institutes of Health](#) oversees the [Human Genome Project](#).

NIH [National Institutes of Health](#). This is an agency within the federal Department of Health and Human Services. It contains the [National Human Genome Research Institute](#). This agency also funds many other research projects and has sponsored fellowships for doctoral and post-doctoral level trainees in genetics.

NSGC [National Society of Genetic Counselors](#), a professional organization.

NORD [National Organization of Rare Disorders](#). This is a coalition of more than 140 nonprofit voluntary health organizations serving people with rare disorders and disabilities. NORD is committed to the identification, treatment, and cure of rare disorders through programs of education, advocacy, research, and service.

NTD Neural Tube Defect. This refers to an incomplete closing of the neural tube (which develops into the spinal cord) during development of the embryo early in pregnancy. This can lead to such conditions as spina bifida or anencephaly (incomplete development of the brain.)

OGDP [Office of Genomics and Disease Prevention](#). This office at the [Centers for Disease Control and Prevention \(CDC\)](#) provides national public health leadership and builds partnerships with other federal agencies, as well as public health organizations, professional groups, and the private sector.



ORD [Office of Rare Diseases](#). This federal agency within the [National Institutes of Health](#) funds research on rare diseases, those affecting less than 200,000 people in the United States.

Orphan Drug Act of 1983 The federal legislation provided tax incentives and time-limited exclusive marketing to pharmaceutical companies who developed drugs to treat rare disorders.

PacNoRGG [Pacific Northwest Regional Genetics Group](#) - Alaska, Idaho, Oregon, Washington, one of the ten regional genetics networks.

PCR Polymerase Chain Reaction. This is a technique used in molecular biology to study sections of DNA. It increases the amount of DNA available to use in genetic testing or research.

Pedigree A diagram showing a genetic family history and biological relationships among family members, often for several generations.

Penetrance A term indicating the likelihood that a given gene will actually result in a genetic disorder or disease.

PGD Pre-implementation Genetic Diagnosis. This is the testing of a fertilized egg for genetic disorders after in-vitro fertilization, before placing the embryo in the uterus for development.

PHD/PHS Public Health Department/Public Health Service. These are federal and state agencies which provide funding for services linked to public health issues.

Phenotype The physical characteristics into which genes are translated as a result of the interaction of genes and the environment, both before and after birth.

PI Principal Investigator. This term refers to the lead individual in a project or research grant.

PL 94-142 Public Law 94-142, Education of All Handicapped Children Act. This is the original federal law establishing the rights of children with disabilities to receive a free, appropriate public education in the least restrictive setting. It applied to all schools receiving federal funding for students from ages 5 through 20.

PL 101-476 Public Law 101-476, Individuals with Disabilities Education Act. This was the federal law which authorized (legislatively continued) the rights of children with disabilities to a free, appropriate public education. Part B of the law applied to children from ages 3 through 20. Part H provided a broader range of services from birth through age 2.



Predictive gene tests Tests to identify gene mutations that may make a person susceptible to certain diseases or disorders.

Prenatal diagnosis Examining fetal cells taken from the amniotic fluid, the primitive placenta (chorion), or the umbilical cord for biochemical, chromosomal, or gene alterations.

PRIM & R [Public Responsibility in Medicine & Research](#). This is a group that advocates for ethical human and animal research.

PSRGN [Pacific Southwest Regional Genetics Network](#) - California, Hawaii, Nevada, one of the ten regional genetics networks.

PUBS Percutaneous Umbilical Blood Sampling. This is a procedure to take a blood sample from the baby's umbilical cord prenatally.

QA Quality Assurance. This has been applied to controlling quality in laboratory testing to be sure test results are accurate, reliable, and meaningful and interpreted appropriately.

Recessive Allele This is a gene that is expressed only when its counterpart allele on the matching chromosome is also recessive (not dominant). Autosomal recessive disorders develop in persons who receive two copies of the mutant gene, one from each parent who is a carrier.

Recombinant DNA The hybrid DNA produced in the laboratory by joining pieces of DNA from different sources.

Regional Genetics Network [Regional Genetics Network](#) - master list of ten regional United States genetics networks.

RFP Research Funding Proposal. Agencies that grant money announce what types of applications are preferred and outline the steps to be taken by applicants.

RNA Ribonucleic Acid. This is a chemical similar to DNA. The several classes of RNA molecules play important roles in protein synthesis and other cell activities.

Section 504 Federal Civil Rights law, a specific section of Rehabilitation Act of 1973. This protects people with disabilities from discrimination when certain programs receive federal funding.

SERGG [Southeast Regional Genetics Group](#) - Alabama, Florida, Georgia, Kentucky, Louisiana, Mississippi, North Carolina, South Carolina, Tennessee, one of the ten regional genetics networks.

Sex Chromosomes "The chromosomes (X and Y) involved in sex determination. Normal females have two X chromosomes in each cell, and normal males have one X and one Y"



(Weiss & Mackta, 1996, p.139).

SPRANS Special Projects of Regional and National Significance. These are projects related to genetics which are funded by the federal office of Maternal and Child Health. They include funding for the regional genetics networks and their coalition as well as projects with time limitations.

Teratogen A teratogen can be a drug, chemical, physical agent or environmental condition that results in permanent changes in a developing baby due to exposure during the pregnancy. Examples would include thalidomide and German measles.

TEXGENE [Texas Genetics Network](#), one of the ten regional genetics networks.

Translocation "The transfer of all or one part of a chromosome to another location on the same chromosome or to a different chromosome after chromosome breakage." (Weiss & Mackta, 1996, p.139)

Trisomy "Trisomy occurs when there are three members of a given chromosome instead of the normal pair." (Weiss & Mackta, 1996, p.140)

UAP University Affiliated Programs. These programs provide multi-disciplinary services for families with a child with a developmental disability. The programs are affiliated with certain universities.

X-linked Inheritance This refers to any gene found on the X chromosome or traits determined by such genes. It also refers to the specific mode of inheritance of such genes.

References

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