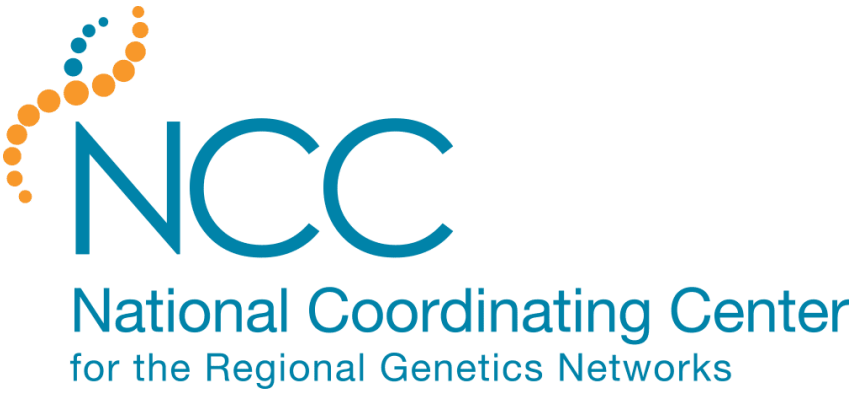

The National Coordinating Center for the Regional Genetics Networks

Glossary of Pediatric Genetics Terms

English – *Add Language of Translation*

2022 Edition

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In collaboration with:
The Cross Cultural Health Care Program (CCHCP)

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Acknowledgments

Mission of the NCC

The National Coordinating Center for the Regional Genetics Networks (NCC) has been funded by the Health Resources and Services Administration (HRSA) since 2004. Our mission is to improve access to genetics for underserved populations. This mission is accomplished through different programs, many of which are educational tools and resources for non-genetics professionals. Learn more at https://nccrcg.org.

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Additional Acknowledgments

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Cindy Roat is an international consultant in language access in health care and patient navigation. She provides training and consulting for health care interpreters, providers, administrators, and patient navigators. Cindy compiled and edited this glossary for the NCC.

The Cross Cultural Health Care Program

The mission of The Cross Cultural Health Care Program (CCHCP) is to serve as a bridge between communities and health care institutions to advance access to quality health care that is culturally and linguistically appropriate. CCHCP provides resources, such as bilingual medical glossaries, as well as medical interpreter, patient navigator, and cultural competence training for individuals and institutions with the goal of systems change. Learn more at: https://xculture.org

CCHCP is supporting and coordinating the translation process of this glossary and is providing the glossary free of charge in its online store: https://xculture.org/store/

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Preface

The seven Regional Genetics Networks (RGNs), the National Genetics Education and Family Support Center (Family Center), and the National Coordinating Center for the Regional Genetics Networks (NCC) mission is to improve access to quality genetic services for medically underserved populations. This bilingual glossary of terms related to Pediatric Genetics is provided to help enable access to pediatric medical services to limited English proficiency populations.

How to Use the Glossary

This glossary is organized alphabetically in English. The first column shows the term in English. The second column has the definition of the term in English. The third column has the translation of the term.

At the end of the translations of the pediatric genetics terms is a glossary of translations for family relationships terms. It is very important for genetic counselors to know if a relative is a blood relative or a relative by marriage.

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Glossary of Terms

English – Language

(n) – noun (v) – verb (adj) – adjective (adv) – adverb

Term	Definition	Translation
Abnormal (adj)	Different from what is considered normal, average, or expected, e.g., a gene sequence that is different than that found in most people.	
Abortion (n)	The deliberate ending of a pregnancy by the removal of an embryo or fetus from the womb. Technically, this is called an "induced abortion" in order to distinguish it from a "spontaneous abortion" which is also called a miscarriage or stillbirth. Also called a "termination of pregnancy" or just "termination."	
ACMG 59 (n)	Fifty-nine genes that the American College of Medical Genetics and Genomics has identified in which variances have a high risk of causing disease – but disease that can be mitigated through early detection or treatment. The ACMG recommends that variances in these genes should be reported to patients even if finding them was not the reason for genetic testing	
Adaptive skills (n)	Skills/abilities needed to perform daily activities, particularly those that allow an individual to live independently and/or work. Examples: brushing teeth, bathing, dressing, food shopping, and taking public transportation.	
Adoption (n)	The process of accepting legal responsibility for someone else's child to raise as your own.	
American College of Medical Genetics and Genomics (ACMG) (n)	A professional membership organization based in the United States that represents the interests of clinical geneticists, clinical laboratory geneticists, and genetic counselors. The ACMG focuses on improving patient care, establishing standards of care and laboratory policy, and educating members about advances in the field.	
Amniocentesis (n)	A procedure by which a sample of amniotic fluid is withdrawn from the amniotic sac. This is usually done by inserting a long needle through the abdominal and uterine walls with the guidance of ultrasound.	

Term	Definition	Translation
Amniotic fluid (n)	The liquid that surrounds a fetus as it develops in the uterus. This liquid contains skin cells that have sloughed off the fetus as well as other fetal cells.	
Anencephaly (n)	A neural tube defect that results in insufficient brain growth in a fetus. Babies with anencephaly usually die soon after birth or are stillborn.	
Benign (adj)	Something that does not threaten health or life. May refer to a type of change to the DNA that does not create health consequences.	
Biochemical testing (n)	Blood tests to identify elevated levels of certain naturally occurring substances/chemicals in the body that are linked to genetic conditions.	
Birth canal (n)	The passageway from the uterus through the cervix, the vagina, and the vulva through which a baby passes during the birth process.	
Birth defect (n)	A problem or physical difference with how the body works that is present at birth. Birth defects can be caused by genetic abnormalities, environmental influences, random chance, or by circumstances related to the birth process. Also called “congenital malformation” or “congenital anomaly.”	
Blood test (n)	A test in which blood is drawn (usually from the patient’s arm) and sent to a laboratory for analysis. In genetics, blood tests may provide cells for genetic sequencing, or blood samples may be used to assess things like protein or hormone levels.	
Brace (n)	A medical appliance that provides support to joints or bones, to weak muscles, or to strained ligaments.	
Buccal swab (n)	A way to collect DNA from the cells on the inside of a person's cheek.	
Carrier (n)	A person who carries a genetic mutation in one of the two copies of a particular gene, regardless of whether they get cancer or not.	
Cell (n)	The smallest (microscopic) functional unit of living organisms. All living things are composed of one or more cells. Within each cell are the structures needed for the cell to process energy, dispose of waste, reproduce, and perform specialized functions.	

Term	Definition	Translation
Cervix (n)	The narrow lower part of the uterus than connects to the vagina.	
Chorionic Villus Sampling (CVS) (n)	A procedure during pregnancy in which a sample of cells from the placenta is removed to check for possible genetic abnormalities.	
Chromosome (n)	Thread-like structures located inside the nucleus of cells. In humans, there are 23 pairs of chromosomes, for a total of 46 chromosomes. Each chromosome is made of a long strand of DNA, which carries genetic information.	
Chromosome test / Karyotype (n)	A test that looks at the number and arrangement of chromosomes in a cell.	
Cleft lip (n)	A congenital malformation that occurs when the upper lip does not form properly, causing an opening in the upper lip that can extend to the nose. This can occur together with a cleft palate or on its own.	
Cleft palate (n)	A congenital malformation where the roof of the mouth (palate) does not form properly, resulting in an opening into the nasal cavity. This can occur together with a cleft lip or on its own.	
Clinically significant (adj)	If a test result is “clinically significant,” the result indicates a medical problem that can impact a person’s life.	
Cognition (n)	The processes involved in thinking, learning, understanding, and remembering.	
Condition (e.g., “genetic condition”) (n)	A long-term medical health issue.	
Consanguinity (n)	When parents are blood relatives to each other.	
Consultation model (n)	The model of care for patients with genetic conditions in which the genetic team acts as consultants, supporting the care being provided principally by the patient’s Primary Care Provider.	
Continuing care model (n)	The model of care for patients with genetic conditions in which the genetic team takes primary responsibility for supervising and coordinating all care related to the patient’s genetic condition.	

Term	Definition	Translation
Cystic fibrosis (n)	A progressive genetic condition that affects the exocrine glands (the glands that make sweat and digestive juices) and causes the production of thick, sticky mucus. This mucus blocks the pancreatic duct, the intestines, and the lungs, leading to persistent respiratory infections.	
De novo (adj)	Genetic changes that are not inherited but that occur during cell division.	
Deleterious mutation/ Disease-causing mutation (n)	A change in a person's DNA that may cause a medical condition.	
Deletion (n)	Having a section of genetic information (DNA) missing.	
Developmental assessment (n)	An evaluation of how an individual patient's intellectual, emotional, and social development compare with others of the same age.	
Diagnostic test (n)	A medical test that determines whether a patient has a particular medical problem. Diagnostic tests are often used when providers have a specific reason to believe that the medical problem may be present. Compare this to screening tests, which may be given routinely even if the provider has no reason to believe the patient has a certain problem. Screening tests often only report whether a patient is at an increased risk for the medical problem in question, whereas diagnostic tests report whether the problem is actually present.	
DNA (Deoxyribonucleic Acid) (n)	The molecule that carries the genetic information of a cell. It provides the instructions used in the development, functioning and reproduction of the organism of which it is a part. The DNA is organized into chromosomes.	
DNA marker (also called a "Genetic marker") (n)	A readily recognizable genetic trait, gene, or DNA segment.	
DNA mutation (n)	A change in the typical sequence of the chemicals that make up the DNA, like the change in the order of letters in a word. Mutations or variants are often compared to misspelled words because chemicals that make up the DNA sequence are not in the expected order.	

Term	Definition	Translation
DNA sequence (n)	The exact arrangement of the chemicals that make up a section of DNA.	
DNA sequencing (n)	The laboratory technique used to determine the exact arrangement of the chemicals that make up a section of DNA. This is one type of genetic testing.	
Dominant (adj)	A genetic trait in which one copy of the gene is sufficient for a trait to be expressed. In a dominant genetic condition, if one copy of the gene has a mutation, the personal will be affected with the condition.	
Donor egg or sperm (n)	An egg (singular) or sperm (plural) donated by one person to be joined under laboratory conditions and implanted in a woman's uterus. The donor egg or sperm may come from the woman or man who will raise any resulting child, or they may come from a third party.	
Down syndrome (Trisomy 21) (n)	A genetic condition in which there are three copies of chromosome number 21 instead of two. Down syndrome causes a distinct facial appearance, intellectual disability, developmental delays, and may be associated with thyroid or heart disease.	
Duplication (n)	Having an extra section of genetic information (DNA). A *duplication* occurs when part of a chromosome is copied abnormally, resulting in extra *genetic* material.	
Edwards syndrome (Trisomy 18) (n)	A genetic condition in which there are three copies of chromosome number 18 instead of two. Trisomy 18 is a very severe condition that causes problems with the brain, the heart, the kidneys, and the digestive tract. Most children affected by trisomy 18 die before or soon after birth.	
Egg (n)	The reproductive cells of a woman. When fertilized by sperm, the egg will grow into an embryo. Also called an "ovum," plural "ova."	
Embryo (n)	An unborn mammal, between conception and 8 weeks of gestation.	
Environmental causes or factors (n)	Causes of illness that come from a person's surroundings, not from their genetic make-up. Environmental causes of illness include infection, trauma, diet, exposure to toxins, etc. Some environmental causes of illness can be avoided or changed, while genetic causes cannot.	

Term	Definition	Translation
Exome sequencing (n)	A genetic test in which a patient's exons are sequenced and studied.	
Exon (n)	The part of the genome that codes for proteins. The majority of deleterious mutations occur in exons.	
Fallopian tube (n)	The tube that connects an ovary to the uterus.	
False negative (n)	A test result that finds no evidence of a condition when the condition actually does exist. For example, a false negative on a pregnancy test finds that the woman is not pregnant when, in fact, she is pregnant.	
False positive (n)	A test result that finds evidence of a condition when the condition does NOT actually exist. For example, a false positive on a pregnancy test finds that the woman is pregnant when, in fact, she is not.	
Familial (adj)	Belonging to a family, e.g., a familial trait is a trait that is shared by family members and may be due to genetic or environmental factors or both.	
Family history (n)	The medical history of the members of a biological family.	
Fertilization (n)	The joining of an egg and sperm to create the first cell that will develop into an embryo, then fetus, then baby.	
Fetal surgery (n)	Surgery conducted on a fetus while it is still in the uterus.	
Fetus(n)	An unborn mammal, between 8 weeks of gestation to birth.	
Flip a coin (v)	A random decision-making tool used in the U.S. While a coin is flipped into the air and caught, a person predicts whether it will fall with the "heads" side up or the "tails" side up. If the coin falls as predicted, the person "wins." This expression is often used as a metaphor for any outcome that is random and has two possible outcomes, and to describe a situation in which each outcome is as likely as the other.	
First trimester screening (n)	A blood test and ultrasound conducted at 10-13 weeks of pregnancy to screen for Down syndrome (trisomy 21), Edwards syndrome (trisomy 18), and certain other fetal problems.	

Term	Definition	Translation
Fragile X syndrome (n)	A genetic condition that affects the FMR1 gene so that it does not function properly. This syndrome causes intellectual disability, behavioral and learning challenges and various physical characteristics that are not life threatening.	
Fragment (n)	A small piece; an incomplete part of a whole.	
Gene (n)	A specific sequence of DNA that codes for one or many functions within the cell and body.	
Gene copy (n) **Normal gene copy (n)** **Altered gene copy (n)**	In human cells, DNA is arranged in 23 pairs of chromosomes, for a total of 46 chromosomes. These chromosomes contain all the genes that make up the DNA. One chromosome of each pair is inherited from each parent. As such, individuals have two copies of every gene, one inherited from the mother and one inherited from the father. A normal copy of a gene is one that is found most frequently in the population. Also called the "working copy." An altered copy of a gene is one that differs from the working copy and is considered to be "abnormal." Also called the "non-working copy."	
Gene deletion (n)	Having a piece of genetic information missing from a gene.	
Gene panel testing (n)	Genetic testing that looks at 2 or more specific genes known to be associated with particular conditions.	
Gene variant (n)	A change from the typical DNA sequences. A genetic variant can be benign, deleterious or of uncertain significance. Also called "mutation", although "genetic variant" is becoming the more common usage.	
General population (n)	"Most people." For example, if you have the same risk of getting cancer as the general population, that means that you have the same chance of getting cancer as everyone else. This is in contrast to a "high risk population" who has a greater chance of getting cancer than everyone else.	

Term	Definition	Translation
Generation (n)	The people who constitute a single step in a line of descent from an ancestor; a group of people born and living more or less at the same time. Example: You, your brothers, and sisters, all your spouses and your cousins are in the same generation. Your parents, your aunts, and uncles and all their spouses form a previous generation. Your grandparents, their siblings, and spouses from an even earlier generation. Your children and nieces and nephews form a later generation.	
Genetic alphabet (n)	A group name for four chemical bases, or "nucleotides," that form the basis of all genetic material. They are adenine (A), thymine (T), cytosine (C), and guanine (G).	
Genetic counseling (n)	A discussion with a medical professional with expertise in genetics about the basic concepts of genetics, genetic conditions, the chances of being affected by a genetic condition or having a child with a genetic condition, and genetic testing and treatment.	
Genetic counselor (n)	A healthcare professional with a specialized graduate degree who works with people who have concerns about genetic conditions in their family. Genetic counselors provide information about genetic conditions, help patients understand their chances of being affected by a genetic condition or having a child with a genetic condition, and help them make informed decisions about testing and treatment. Genetic counselors also provide emotional support to patients and families.	
Genetic discrimination (n)	Occurs when people are treated differently by their employer or insurance company due to their genetic makeup. There are federal and state laws that help protect against genetic discrimination.	
Genetic factors (n)	Specific aspects of a person's genetic make-up that influence that person's health and development.	
Genetic information (n)	The instructions encoded in DNA, which tells every cell in a body how to grow, what to do and how to reproduce.	
Genetic material (n)	All the parts of a cell that carry genetic information. Genetic material could include genes, parts of genes, a group of genes, a DNA molecule, a fragment of DNA, a group of DNA molecules, or the entire set of genetic instructions.	

Term	Definition	Translation
Genetic predisposition (n)	An increased chance of a person developing a certain trait or disease based on that person's particular genetic makeup.	
Genetic test (n)	A laboratory test designed to determine if a person has a gene mutation or a typical DNA sequence.	
Single site gene testing (n)	Single site gene testing: testing at a specific point in the genome. This is usually done to look for a mutation found in a family member or when one specific mutation is known to cause a genetic condition.	
Individual gene testing (n)	Individual gene testing: genetic analysis of the entirety of one specific gene.	
Gene panel test (n)	Gene panel test: analysis of several genes that have been shown to be associated with a particular condition, e.g., breast cancer gene panel.	
Microarray (n)	Microarray: genetic analysis that looks for unexpected numbers of specific genes (two copies of every gene code is usually expected).	
Exome sequencing (n)	Exome testing: a genetic test in which the exons of a patient's genetic code are sequenced and studied.	
Genome sequencing (n)	Genome sequencing: a genetic test in which the entire genetic code (the genome) of a patient is mapped out and studied.	
Genetic trait (n)	A characteristic within a family that is passed down from parent to child through their DNA.	
Geneticist (n)	A doctor or scientist who studies genetics.	
Genetics (adj)	The science of how an organism's genes interact with the environment to produce certain traits.	
Genetics (n)	The field of science and medicine that studies the biologic basis of heredity (how traits are passed from one generation to another) and how the instructions for life are used by all living organisms.	
Genome (n)	The entire genetic code of an individual.	
Genome sequencing (n)	A genetic test in which the entire genetic code of a patient is mapped out and studied.	

Term	Definition	Translation
Germline testing (n)	Germline testing refers to the analysis of a person's DNA, which he or she inherited from his or her parents. In biology and genetics, the germline is the group of cells that will pass on the genetic material to children, in other words, the cells from which the eggs and sperm come.	
Guardianship (n)	A legal arrangement, supervised by a court, in which one person takes responsibility for a minor or an adult with a disability.	
Helix (n)	Something that is spiral in form. DNA is often described as being a "double helix."	
Hereditary (adj)	Passed down from parent to child.	
Hereditary material (n)	Genetic material that is passed down from parent to child.	
Huntington's disease (n)	A neurological disorder that causes physical and cognitive decline. Cognitive symptoms include problems with reasoning and controlling emotions. Physical symptoms include abnormal movements that cannot be controlled by the individual. This disorder is linked to a dominant gene variant, which means that a person inheriting only one copy of the affected gene will develop the disorder.	
Individualized Education Plan (IEP) (n)	A customized plan written and updated yearly for every child in public school special education classes. The IEP addresses current levels of educational performance, goals, and the special education and services that the child will need to meet those goals.	
In Vitro Fertilization (IVF) (n)	The fertilization of an egg by a sperm outside of a woman's body. The process involves extracting eggs from a woman's ovaries, collecting sperm from a man, and combining a sperm and egg in a laboratory dish. The resulting fertilized egg is usually then implanted in a woman's uterus so that it can develop into a baby.	
Individual gene testing (n)	Genetic analysis of the entirety of one specific gene.	
Infertility (n)	The inability to have children.	

Term	Definition	Translation
Informed consent (n)	The process of agreeing to a procedure or course of treatment after understanding what the procedure/treatment entails, the potential risks and benefits associated with it, and the other options available.	
Inheritance pattern (n)	The manner in which a particular genetic trait or disorder is passed from a parent to a child, e.g., autosomal dominant or recessive, X-linked dominant or recessive, or multifactorial.	
Inherited (adj)	Passed down from parent to child.	
Insertion (n)	An extra segment of DNA added in at a place where it is not usually found.	
Intellectual disability (n)	A condition, varying in severity, in which a person has significant impairments in mental abilities, social skills, and core functions of daily living compared to others their age.	
Klinefelter's syndrome (47, XXY) (n)	A genetic condition in which a male has two copies of the X chromosome and one copy of the Y chromosome; compared to the typical chromosome makeup where a male has one X chromosome and one Y chromosome.	
Lynch syndrome (n)	An inherited genetic disorder that increases a person's risk of getting cancer of the colon, rectum, uterus, ovaries, and other cancers.	
Marker chromosome (n)	A small extra fragment of a chromosome detected when doing a chromosome test like a karyotype. Marker chromosomes can sometimes cause health or development problems, depending on how much and what genetic material is contained within.	
Maternal Serum Screening (MSS or Maternal serum alpha-fetoprotein test) (n)	A blood test available to pregnant women that identifies elevated risks for Down syndrome, trisomy 18 and neural tube defects. Usually conducted at in the second trimester, between 14-20 weeks.	
Metastasis (n)	The spreading of cancer from one organ to another.	

Term	Definition	Translation
Microarray (n)	A high-resolution chromosome test that provides more information than a basic karyotype. A microarray measures the amount of chromosome material in a sample and can detect both large changes (e.g., if there is an extra or missing chromosome) as well as small changes (e.g., if there are very small pieces of chromosomes that are extra or missing). It does not look at the visual appearance or arrangement of chromosomes but measures the amount of genetic material.	
Miscarriage (n)	The spontaneous (not intentional) loss of a pregnancy. See "abortion." Also called pregnancy loss.	
Molecule (n)	The smallest unit of a chemical compound that still has the properties of that compound. For example, a molecule of water is made up of two hydrogen atoms and one oxygen atom. Separately, they are just atoms, but when bonded together, they make a water molecule.	
Mosaicism (n)	A condition in which some, but not all, cells in a sample show a genetic difference.	
Motor Abilities (n)	The ability to move and use one's muscles.	
Multifactorial (adj)	Due to a combination of genetic and non-genetic (environmental, hormonal, etc.) risk factors that act together to determine risk.	
Mutation (n)	A change in a gene, usually deleterious. See "genetic variant."	
Non-Invasive Prenatal Testing (NIPT) (n)	A blood test available to pregnant women that identifies elevated risk for certain genetic conditions in the fetus. This test focuses on fragments of DNA from placental cells – which carry the fetus' genetic make-up – that are found in the mother's bloodstream.	
Nuchal thickening (n)	There is a pocket of fluid at the back of the neck of a fetus which can be measured in an ultrasound between 10-14 weeks gestation (called the nuchal translucency). If there is a large amount of fluid at this point, or if later in pregnancy the neck skin itself appears to be thicker, this is associated with a higher risk of chromosome problems and other rare genetic conditions.	

Term	Definition	Translation
Open Neural Tube Defect (ONTD) (n)	A birth defect of the spine, spinal cord, or brain that results from a hole in the spinal column not closing up when appropriate during early fetal development. The two most common types of ONTDs are spina bifida and anencephaly.	
Ova (n)	See “egg.”	
Ovary (n)	The organ in a woman that stores and releases eggs. There are normally two.	
Packets of genetic information (n)	A phrase genetic counselors use to describe genes or chromosomes.	
Pathogenic (adj)	Disease causing.	
Pedigree (n)	A family tree that can be used to trace the inheritance of specific genetic traits.	
Penetrance (n)	The probability that a specific genetic trait will be expressed if a person carries a mutation. “Complete penetrance” means that everyone who carries a particular gene mutation will show the trait related to that altered gene. “Incomplete penetrance” means that only some of the people who have the altered gene will actually show the related trait.	
Placenta (n)	The organ that develops together with an embryo in a pregnant woman’s uterus to nourish the embryo through the umbilical cord.	
Presymptomatic (adj)	Before symptoms appear.	
Prognosis (n)	The most likely outcome of a disease process.	
Protein (n)	A molecule made up of chains of *amino acids*. Proteins do most of the work in cells and are required for the structure, function, and regulation of the body's tissues and organs. Genes determine how specific amino acids are put together to form a specific protein.	
Random (adj)	Happening in an unpredictable way.	

Term	Definition	Translation
Recessive (adj)	A genetic trait in which both copies of a gene need to be altered for the trait to be expressed; recessive traits are typically inherited from both parents, each one contributing one copy of the gene in question. In prenatal genetics, recessive inheritance refers to conditions in which both copies of a particular gene pair need to be altered to develop a specific disease or condition.	
Red flag (n)	A warning sign or a clue.	
Replicate (v)	To copy.	
Reproductive history (n)	The experiences a woman has had related to pregnancy and childbirth. A reproductive history usually includes age at the onset of menses (have a monthly period), age at the beginning of sexual intercourse, age at first conception, number of pregnancies, number of live births, number of miscarriages, number of abortions.	
Ring chromosome (n)	An abnormal formation of a chromosome in which the ends of two of the chromosome's arms have linked together to form a ring.	
Risk (n) **At risk (adj)** **At average risk (adj)** **At high risk (adj)** **At higher risk than "X" (adj)** **Risk factor (n)**	The chance that something will happen; in the case of prenatal genetics, the chance that the child will have a genetic condition. Has some possibility of having a genetic condition. Has the same possibility of having a genetic condition as the general population. Has a greater possibility of having a genetic condition than the general population. Has a greater possibility of having a genetic condition than "X." A circumstance that increases the risk of having a genetic condition.	
Runs in the family	Passed down from parents to children to grandchildren.	
Saliva (spit) sample (n)	A way to collect DNA from the cells in saliva (spit).	

Term	Definition	Translation
Screen (v)	To assess a person's risk for a particular health problem, even if the person has no symptoms to suggest that they have the problem.	
Screening test (n)	A test that looks to detect a particular health problem, even if a patient has no symptoms of that particular condition. Prenatal screening tests aren't looking for genetic changes. They are designed to look at hormones, chemicals, other things that indicate a fetus might have a higher risk of a certain condition. They are not DNA results, do not give a definite answer about whether someone is affected, and do not provide a firm diagnosis. They are usually reported as a risk number (e.g., a 1 in ____ chance), and if this number is above a certain cut-off point, the result will be categorized as "screen positive" or "screen negative."	
Semen (n)	A liquid produced by the testes, the prostate gland, the seminal vesicle, and the bulbourethral gland that carries, nourishes, and protects sperm cells on their way to fertilizing an egg. Also called seminal fluid.	
Seminal fluid (n)	See "semen."	
Sensory abilities (n)	The ability to see, hear, touch, taste, and smell.	
Sickle cell disease (n)	A genetic disorder caused by a mutation in a gene that helps to make hemoglobin, an important part of red blood cells. In sickle cell disease, red blood cells can become sickle-shaped (instead of round as they should be). These affected blood cells do not function properly, and die early, leaving a shortage of red blood cells, and can block blood flow causing pain.	
Single site analysis (n)	A genetic test that looks at only the part of a gene that is known to be associated with a certain disease.	
Somatic genetic changes (n)	Genetic changes in an individual that occur after conception. For example, exposure to the ultraviolet rays in sunlight can cause problems with replication of genetic material in cells, leading to changes in the gene that controls cell growth and then to skin cancer. These changes are somatic genetic changes.	
Special education (n)	Education that is tailored to individuals with developmental delays or other learning disabilities.	
Sperm (n)	The reproductive cells of the man. When sperm fertilize a woman's egg, a baby develops.	

Term	Definition	Translation
Sperm donation (n)	The process through which a man allows his sperm to be collected and used to fertilize the eggs of a woman who is not his sexual partner.	
Spina bifida (n)	A congenital defect of the spine in which part of the spinal cord is exposed through an opening in the bone structure. This may result in nerve damage and some degree of paralysis in the legs.	
Statistically significant (adj)	A measurement of whether the findings of research are meaningful. Refers to the likelihood that a relationship between two factors is linked by something other than chance.	
Supportive seating (n)	Seating, such as a wheelchair, that helps an individual function in their environment and that prevents worsening of their condition.	
Teratogen (n)	Something that has the potential to disturb the normal development of an embryo or fetus.	
Termination of pregnancy (n)	See “abortion.”	

Term	Definition	Translation
Test result (n)		
Negative (adj)	A negative result on a genetic test means that the laboratory did not find the specific genetic change that the test was designed to identify.	
True negative (n)	When a patient is being tested to see if he or she has a specific genetic change that is present in other family members, a negative result can be considered a "true negative." This means that instead of having an increased risk for cancer like other family members, they have the same risk as everyone else in the general population.	
Positive (adj)	A positive result on a genetic test means that the laboratory did find a specific genetic change that is associated with a hereditary cancer syndrome (a deleterious mutation). In cancer genetics, that means that the patient has an increased risk of getting the cancer(s) associated with the gene.	
Variant of uncertain significance (n)	If the laboratory finds a genetic change for which currently there is not enough information to know if this change is problematic or not, it reports a "variant of uncertain significance." Basically, this means that the laboratory found a genetic change, but they don't know what it means. All of these variants will eventually be reclassified as either a "positive" or "negative" result. Most become "negative" and are thought to represent natural variation between individuals.	
Polymorphism (n)	Everyone has some degree of commonly occurring genetic changes that are not associated with cancer. If the test finds this sort of change, it reports a "polymorphism."	
Testes (n)	The organs in a man that create and release sperm. Also called "testicles." Singular testis.	
Toxic exposure (n)	Contact with something that is harmful or poisonous.	
Trait (n)	A characteristic of a person. In genetics, traits are aspects of a person defined or influenced by their genetic code, e.g., eye color, blood type, risk for certain diseases.	

Term	Definition	Translation
Translocation (n)	Translocations are a type of chromosome rearrangement. They can be "balanced," meaning that two chromosomes have pieces that have "swapped" with each other, but there is still the correct amount of chromosome material. Balanced translocations do not usually cause medical or development problems. They can also be "unbalanced" where pieces of chromosomes have traded places, but there is missing or extra chromosome material. Unbalanced translocations will often cause medical and development problems.	
Typo (n)	A mistyped word; shortened slang for "typographical error." In genetic counseling "a typo" is commonly used as an analogy to refer to mutations or variants in a gene.	
Ultrasound (n)	An imaging method that uses high frequency sound waves to create a picture of something inside the body, such as a fetus or baby.	
Uterus (n)	The organ in a woman's lower abdomen in which a fertilized egg develops into an embryo, then a fetus, then a baby. Also called "womb."	
Vagina (n)	The tube-like part of the female reproductive system that extends from the cervix to the outside of the body.	
Variant (n)	A version of something that differs from the norm. So, a genetic variant is a change to the usual genetic sequence.	
Vas deferens (n)	The tubes that lead from the testes to the urethra, through which sperm is ejaculated.	
X-linked (adj)	A trait that is influenced or determined by a gene on the "X" chromosome.	

Family Relationships

For genetic counselors, it is very important to know if a relative is a blood relative or a relative by marriage.

Term	Definition	Translation
Parent	Your mother or father.	
Mother	For genetic purposes, the woman whose egg was fertilized and grew to be you.	
Mother-in-law	Your husband or wife's mother.	
Stepmother	Your father's wife who is not your biological mother.	
Adoptive mother	A woman who is not your biological mother but who accepted legal responsibility for and raised you.	
Godmother	A woman chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.	
Father	For genetic purposes, the man whose sperm fertilized the egg that grew to be you.	
Father-in-law	Your husband or wife's father.	
Stepfather	Your mother's husband who is not your biological father.	
Adoptive father	A man who is not your biological father but who accepted legal responsibility for and raised you.	
Godfather	A man chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.	
Aunt	Your mother or father's sister or sister-in-law.	
Maternal aunt	Your mother's sister or sister-in-law.	
Paternal aunt	Your father's sister or sister-in-law.	
Uncle	Your mother or father's brother or brother-in-law.	
Maternal uncle	Your mother's brother or brother-in-law.	
Paternal uncle	Your father's brother or brother-in-law.	

Term	Definition	Translation
Cousin	Usually understood to be a first cousin.	
First cousin	Your aunt or uncle's child.	
First cousin once removed	Your aunt or uncle's grandchild or your first cousin's child	
Second cousin	The children of first cousins are second cousins to each other.	
Grandparent	Your parent's father or mother.	
Grandmother	Your mother or father's mother.	
Maternal grandmother	Your mother's mother.	
Paternal grandmother	Your father's mother.	
Grandfather	Your mother or father's father.	
Maternal grandfather	Your mother's father.	
Paternal grandfather	Your father's father.	
Great aunt	Your mother or father's aunt.	
Maternal great aunt	Your mother's aunt.	
Paternal great aunt	Your father's aunt.	
Great uncle	Your father or mother's uncle.	
Maternal great uncle	Your mother's uncle.	
Paternal great uncle	Your father's uncle.	
Great grandparents	The parents of any of your grandparents.	
Great grandmother	The mother of any of your grandparents.	
Great grandfather	The father of any of your grandparents.	

Term	Definition	Translation
Siblings	The children of your father and mother.	
Half siblings	Siblings (brothers and sisters) who have either the same mother and different fathers, or the same father but different mothers. Half siblings share some genetic similarity with you.	
Stepsiblings	The children of your stepmother but not your father; or the children of your stepfather but not your mother. Stepsiblings do not share any genetic similarity with you.	
Twins	Two siblings born at the same time.	
Identical twins	Twins who developed from the same egg and sperm, meaning that they are genetically identical. Also called monozygotic twins.	
Fraternal twins	Twins who developed from different eggs and sperm, meaning that they are genetically different and have the same number of shared genes as any other sibling. Also called dizygotic twins.	
Triplets	Three siblings born at the same time.	
Sister	A sibling who is a girl.	
Sister-in-law	Your brother's wife or your husband's sister.	
Half sister	A girl who is the either the child of your father with a different mother, or the child of your mother with a different father.	
Stepsister	The daughter of your stepmother or stepfather and therefore not biologically related to you.	
Brother	A sibling who is a boy.	
Brother-in-law	Your sister's husband or your wife's brother.	
Half brother	A boy who is the either the child of your father with a different mother, or the child of your mother with a different father.	
Stepbrother	The son of your stepmother or stepfather and therefore not biologically related to you.	
Niece	Your sibling's daughter.	
Nephew	Your sibling's son.	

Term	Definition	Translation
Spouse	Your husband or wife.	
Wife	The woman to whom you are married.	
Husband	The man to whom you are married.	
Children	Genetically speaking, the people who are produced from your egg or sperm.	
Daughter	Your child who is a girl.	
Daughter-in-law	Your son's wife.	
Stepdaughter	Your spouse's daughter who is not your biological child.	
Adoptive daughter	A girl for whom you have accepted legal responsibility and raised even though she is not your biological child.	
Goddaughter	A girl for whom you have accepted spiritual responsibility within a ceremony of the Catholic church; she may or may not be biologically related to you.	
Son	Your child who is a boy.	
Son-in-law	Your daughter's husband.	
Stepson	Your spouse's son who is not your biological child.	
Adoptive son	A boy for whom you have accepted legal responsibility and raised even though he is not your biological child.	
Godson	A boy for whom you have accepted spiritual responsibility within a ceremony of the Catholic church. He may or may not be biologically related to you.	
Ancestor	A person from whom you are descended, usually more remote than a grandparent.	
Fiancé(e)	The person whom you have promised to marry. Man: fiancé Woman: fiancée	

Term	Definition	Translation
Domestic partner	The person with whom you are living and have an intimate relationship, but to whom you are not married.	
Divorced	Having ended a marriage.	
Engaged	Having promised to marry someone.	
Widow	A woman whose husband has died.	
Widower	A man whose wife has died.	
Relative	A person in your family.	
Blood relation	A person who is related to you by blood not marriage, e.g., your sister, but not your sister-in-law; your mother but not your stepmother; your daughter but not your adopted daughter.	
Next of kin	Your closest living blood relation.	

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