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Genetics Glossary

Introduction

This glossary was adapted from the National Coordinating Center for the Regional Genetics Networks (NCC) (supported by Cooperative Agreement #UH9MC30770). Additional terms and definitions were added by the Washington State Prenatal Genetics Task Force. This glossary is intended to assist medical professionals with prenatal genetic terminology and help provide definitional clarity in their conversations with their patients.

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. Some of the terms have examples and considerations for a further explanation. Each term will have the parts of speech, such as noun (n), adjective (adj), and verb (v).

Term	Definition
Abnormal (adj)	Different from what is considered normal, average, or expected. Examples and considerations: 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. Examples and considerations: Technically, this is called an “induced abortion” to tell the difference from a “spontaneous abortion” which is also called a miscarriage or stillbirth. Also called a “termination of pregnancy” or just “termination.”
Advanced maternal age (n)	A phrase used to refer to a pregnancy in a woman who is 35 years or older.
Adoption (n)	The process of legally taking someone else’s child into your family to raise as your own child.

Amniocentesis (n)	A procedure by which a sample of amniotic fluid (see amniotic fluid), is withdrawn from the amniotic sac, a thin walled sac. This is usually done by inserting a long needle through the abdominal and uterine walls, and guided by the image from an ultrasound.
Amniotic fluid (n)	The liquid that surrounds a fetus as it develops in the uterus. This liquid contains skin cells that have been shed off the fetus as well as other fetal cells.
Anencephaly (n)	A neural tube (the embryonic brain and spine) defect that results in insufficient brain growth in a fetus. Examples and considerations: Babies with anencephaly usually die soon after birth or are stillborn.
Aneuploidy (n)	The occurrence of an extra or missing copy of a chromosome. Also called a chromosal condition or disorder. See Trisomy and Monosomy.
Aneuploidy screening (n)	This is a screening test done to assess the risk of having a baby with an aneuploidy. Aneuploidy screening can be done by drawing a sample of blood from the pregnant patient measuring the thickness of the fluid under the baby's neck by ultrasound, or analyzing the fetal DNA in the maternal blood. The test is usually done after 10 weeks of pregnancy. See Aneuploidy. Examples and considerations: Aneuploidy can cause conditions like Down's syndrome, Edward's syndrome, Patau's syndrome, Turner syndrome, Klinefelter syndrome, and other syndromes.
Assisted Reproductive Technology (ART) (n)	Refers to a range of techniques for enhancing fertility, such as in vitro fertilization, in which both the egg and the sperm are manipulated. See IVF.
Autosome (n)	Any chromosome that is not a sex chromosome. Of the 23 pairs of chromosomes in humans, 22 pairs are autosomes. See Chromosome, Sex chromosome.
Benign (adj)	Something that does not threaten health or life. Examples and considerations: May refer to a type of change to the DNA that does not create health consequences. e.g., when discussing cancer, “benign” means “not cancerous.”
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.

	Examples and considerations: Birth defects can be caused by genetic abnormalities, environmental influences, random chance, or by circumstances related to the birth process. Also known as, “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. Examples and considerations: In genetics, blood tests may provide cells for genetic sequencing, or blood samples may be used to assess things like protein or hormone levels.
Buccal swab (n)	A way to collect DNA from the cells on the inside of a person's cheek.
Carrier (n)	A person who has a genetic mutation in one of their two copies of a particular gene that is associated with a genetic condition. The other copy of the gene does not have a mutation, so that person usually does not have any medical issues related to the gene change.
Carrier screening (n)	A genetic test used to determine if a healthy person is a carrier of a recessive genetic disease. It provides information about an individual's reproductive risk and their chances of having a child with a genetic disease. See Recessive, Carrier, and Genetic test.
Cascade screening (n)	A systematic process for the identification of individuals within a family at risk for a hereditary condition. - The screening begins with finding a pathogenic or likely pathogenic variant through broad-based testing (such as full gene or multigene panel testing) in one family member, usually affected with the condition. - Then, testing just for the specific family variant is extended to at-risk biological relatives. This process is repeated as more affected individuals or pathogenic variant carriers are identified. Cascade screening is sometimes referred to as cascade testing.
Cell (n)	The smallest (microscopic) functional unit of living organisms. Examples and considerations: All living things are composed of one or more cells. Within each cell are the structures called organelles that are subunits needed for the cell to process energy, dispose of waste, reproduce, and perform specialized functions.

Cell-free DNA testing 9N)	Prenatal cell-free DNA testing is a non-invasive test done after the 10th week of pregnancy to examine the fetal DNA that is naturally present in the maternal bloodstream. The test determines if a woman has a higher chance of having a fetus with aneuploidy and also identifies the sex of the fetus. This test is sometimes called cell-free DNA screening and Non-Invasive Prenatal Testing or Screening. See Non-Invasive Prenatal Testing.
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 (n)	A test that looks at the number and types of chromosomes in a cell. Also called, karyotype test.
Cleft lip (n)	A birth defect 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 birth defect 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)	A test result indicating a medical problem that can impact a person's life.
Condition (n)	A long-term medical health issue (e.g. genetic condition)
Congenital	Physical differences with how the body works or a condition that is present before or at birth.
Consanguinity (n)	When parents are blood relatives to each other.
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.
Deleterious mutation/ Disease-causing mutation (n)	A change in a person's DNA that may cause a medical condition. This is sometimes also called a *pathogenic variant*, referring to the disease-causing nature of the mutation.

Deletion (n)	Having a section of genetic information (DNA) missing.
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. Examples and considerations: Compare this to screening tests, which may be given routinely, even if the provider has no reason to believe the patient has a specific 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.
Diploid (adj)	Referring to a cell containing two complete sets of chromosomes, one from each parent. In humans, that number is 46.
DNA (Deoxyribonucleic Acid) (n)	The material 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. Examples and considerations: If you could stretch out the DNA of a chromosome and look at it through a microscope, it would look like a long ladder that is twisted into a spiral. The 'sides' of the ladder are made up of alternating phosphate and sugar groups. The 'rings', are various combinations of two nitrogen bases: Adenine-Thymine and Cytosine- Guanine. Individual sections of DNA that code for specific traits/functions are called genes.
DNA marker (n)	A readily recognizable genetic trait, gene, or DNA segment. Also called a gnetiic marker.
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. Examples and considerations: Mutations or variants are often compared to misspelled words because chemicals that make up the DNA sequence are not in the expected order.
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. Examples and considerations: In a dominant genetic condition, if one copy of the gene has achange in the DNA sequence, the person 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. This condition 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) Examples and considerations: A duplication occurs when part of a chromosome is copied abnormally, resulting in extra genetic material.
Early Imaging Ultrasound (n)	An ultrasound that can be performed as early as the seventh week of pregnancy. It detects fetal heartbeat, measures the size of the fetus, and confirms gestational age of the fetus. Also called a first trimester ultrasound or dating ultrasound. See Ultrasound.
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. Examples and considerations: Most children affected by trisomy 18 die before or soon after birth.
Egg (n)	The reproductive cells of a female. When fertilized by sperm, the egg will grow into an embryo. Also called an "ovum," plural "ova."
Egg donor/source (n)	Refers to the fertile woman who donates an egg, or oocyte, to another woman to help her conceive. It is a part of assisted reproductive technology, or ART. The egg donor will be the biological mother sof any child born through this donation. See ART.
Embryo (n)	An unborn mammal, between conception and 8 weeks of gestation.

Exome (n)	The sequence of all the regions of DNA in a genome that code for all the protein a body makes. Examples and considerations: In humans, the exome is about 1.5% of the genome.
Expanded carrier screening (n)	Evaluates an individual's carrier state for multiple conditions at once and regardless of ethnicity.
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 does exist. Examples and considerations: 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. Examples and considerations: For example, a false positive on a pregnancy test finds that the woman is pregnant when, in fact, she is not.
Familial (adj)	Occurring within members of a family. Examples and considerations: A familial trait is a trait that is shared among 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.
FISH (Fluoresence in Hybridization) (n)	A test that can visualize and map the genetic material in an individual's cells, including specific genes or portions of genes. Examples and considerations: This test may be used for understanding a variety of chromosomal abnormalities and other genetic mutations.

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.
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. Examples and considerations: Genetic counselors may use this to represent an example in genetics. For example, the patient has a 50% chance that a mutation will be passed on every time the patient has a child like flipping a coin.
Fragile X syndrome (n)	A genetic condition that affects the FMR1 gene so that it does not function properly. This condition 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.
General population (n)	"Most people" Examples and considerations: 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, Versus,a "high risk population". The high risk population has a greater chance of getting cancer than everyone else.
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. Examples and considerations: 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.
Genetics (adj)	The scientific study of genes and heredity - of how certain qualities or traits are passed from parent to offspring as a result of changes in DNA sequence. See Gene and Hereditary.

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 counseling intern (n)	A genetic counseling student who has not yet completed their academic studies and is now practicing under the supervision of a more experienced counselor in preparation for providing genetic counseling services independently after obtaining their graduate degree.
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. Examples and considerations: 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.
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 change to their DNA.
Genetic trait (n)	A characteristic within a family that is passed down from parent to child through their DNA.
Genetic variant (n)	A change from the typical DNA sequences. A genetic variant can be benign, deleterious or of uncertain significance. Examples and considerations:

	Also called *mutation*, although *genetic variant* is becoming the more common usage.
Geneticist (n)	A doctor or scientist who studies genetics.
Genome (n)	The entire set of DNA instructions found in a cell
Genotype (n)	The genetic makeup of a cell or an individual. Examples and considerations: The term is used with reference to a specific characteristic that is decided based on the genetic makeup.
Genotyping (n)	The technology that detects small genetic differences that can lead to observable physical differences in traits (See Phenotype)
Gestational Carrier (n)	A woman bearing a genetically unrelated child for another person or couple. Examples and considerations: Also called a gestational surrogate. Typically, in vitro fertilization is used to fertilize the intended parent's egg, and then the resulting embryo is placed in the gestational carrier's uterus.
Haploid (adj)	Refers to the presence of a single set of chromosomes in an organism's cells. Only the egg and sperm cells are haploid. In humans, that number is 23.
Hemoglobinopathies (n)	A term for a group of inherited blood disorders and diseases that primarily affect red blood cells.
Hereditary (adj)	Passed down from parent to child.
Hereditary material (n)	Genetic material that is passed down from parent to child.
Heteroplasmy (n)	Describes the situation in which two or more mtDNA (mitochondrial DNA) variants exist within the same cell. See mitochondrial DNA.
Intracytoplasmic Sperm Injection (ICSI)	A technique used during in vitro fertilization (IVF) where a single sperm is injected directly into the egg for the purpose of fertilization.
Integrated Screen (n)	A two-part prenatal screening test combining first and second trimester screening results. It requires blood drawings and an ultrasound that assesses the risk of a baby being born with Down syndrome, trisomy 18, and open neural tube defects (ONTDs).
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.
Infertility (n)	The inability to have children.

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 way 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)	Having 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 impairments in mental abilities, social skills, and core functions of daily living compared to others their age.
Karyotype (n)	An individual's complete set of chromosomes. The term also refers to a laboratory-produced image of a person's chromosomes isolated from an individual cell and arranged in numerical order. Examples and considerations: A karyotype may be used to look for abnormalities in chromosome number or structure.
Klinefelter's syndrome (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. Examples and considerations: Klinefelter's syndrome is often diagnosed only in adulthood, and adversely affects testicular development and male fertility. It is also referred to as (47, XXY) (n).
Marker chromosome (n)	A small extra fragment of a chromosome found when doing a chromosome test like a karyotype. Examples and considerations: 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 prenatal screening 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. The test measures the level of four pregnancy-related proteins in the pregnant patient's blood which are made by the fetus and the placenta. Also known as a quad a screen.

Meiosis (n)	A type of cell division in sexually reproducing organisms that reduces the number of chromosomes in the daughter cells from diploid to haploid. See Haploid and Diploid.
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 and small changes. It does not look at the visual appearance or arrangement of chromosomes but measures the amount of genetic material. Examples and considerations: The chromosome material in a sample may indicate the following: • Large changes: e.g. if there is an extra or missing chromosome • Small changes: (e.g. if there are very small pieces of chromosomes that are extra or missing
Microdeletion (n)	Are chromosomal deletions that are too small to be detected by light microscopy using conventional cytogenetic (a branch of genetics) methods. Specialized testing is needed to identify these deletions. Also known as submicroscopic deletions. ——————————————— Examples and considerations: Some microdeletions can cause adverse health outcomes, while others can be benign.
Microduplication (n)	Are chromosomal duplications that are too small to be detected by light microscopy using conventional cytogenetics methods. Specialized testing is needed to identify these duplications. Also known as submicroscopic duplications.
Miscarriage (n)	The spontaneous (not intentional) loss of a pregnancy. See “abortion.” Also called pregnancy loss.
Mitosis (n)	A type of cell division by which a diploid cell replicates its chromosomes and then segregates them, producing two identical diploid nuclei in preparation for cell division. See Diploid.
Mitochondrial DNA (n)	Also called mtDNA, it is the DNA within a cell, inside a subunit (or organelle) of the cell called a mitochondria. This DNA is different from the DNA inside the nucleus of a cell. Mitochondrial DNA is in the form of a circulat chromosome, and it helps the organelle convert energy from food into a form of energy the cell can use.
Molecule (n)	The smallest unit of a chemical compound that still has the properties of that compound. ——————————————— Examples and considerations:

	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.
Monosomy (n)	Refers to the condition in which only one chromosome from a pair is present in cells rather than the two copies usually found in each cell. See Aneuploidy.
Mosaicism (n)	A condition in which some, but not all, cells in a sample show a genetic difference. Examples and considerations: It is caused by an error in cell division (mitosis). This results in some cells having the normal number of 46 chromosomes, and other cells having more (47) or fewer (45) chromosomes. Mosaicism can cause several types of disorders.
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 harmful. Examples and considerations: See “genetic variant.”
Nondisjunction (n)	Occurs when chromosomes do not separate properly during cell division. This produces cells with imbalanced chromosome numbers.
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 pregnant patient’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.
Nuchal Translucency (NT) scan	An NT scan is a screening test during the first trimester of pregnancy that measures the size of the clear tissue, called the nuchal translucency at the back of the fetus's neck. It helps determine risk of congenital conditions like Down syndrome in the fetus. See First trimester screening.
Oocyte (n)	An immature egg or ovum. It is produced by the ovary. See Egg, Ova, Ovary.

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. Examples and considerations: 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 ovaries.
Pathogenic variant (n)	A genetic alteration that increases an individual’s susceptibility or predisposition to a certain disease or disorder. When such a variant (or mutation) is inherited, development of symptoms is more likely, but not certain. Also called deleterious mutation, disease-causing mutation, predisposing mutation, and susceptibility gene mutation. See Deleterious mutation.
Penetrance (n)	The probability that a specific genetic trait will be expressed if a person carries a mutation. Examples and considerations: • “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.
Perinatal (n)	Pertaining to the period immediately before and after birth. For statistical purposes, the perinatal period is defined as the period from the 28th week of pregnancy to the end of the 1st week after birth.
Placenta (n)	The organ that develops together with an embryo in a pregnant woman’s uterus to nourish the embryo through the umbilical cord.
Preimplantation (adj)	Of, involving, or being an embryo before uterine implantation
Prenatal (adj)	Refers to a time period or action existing, performed, or used before birth. Also called antenatal.
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. Examples and considerations: 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)	To happen in an unpredictable way.
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. Examples and considerations: 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)	The chance that something will happen; in the case of prenatal genetics, the chance that the child will have 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."
Risk factor (n)	A circumstance that increases the risk of having a genetic condition.
Runs in the family (v)	When a certain condition or trait seems to appear in many members of a single family Examples and considerations: For example, sickle cell disease runs in the family.
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.

	Examples and considerations: • Prenatal screening tests aren't looking for genetic changes. • Designed to look at hormones, chemicals, and other factors that indicate a fetus might have a higher risk of a certain condition. • They are not DNA results and do not give a definite answer about whether someone is affected, • Screening tests do not provide a firm diagnosis. They are usually reported as a risk number (e.g., 1 in X chances), 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."
Sensitivity (n)	Refers to a test's ability to identify an individual with disease as testing positive for the disease (i.e., true positive)
Sequencing (n)	A test that determines the order of the four chemical building blocks - called "bases" - that make up the DNA molecule. The sequence can give information on the genetic information that is carried in a particular DNA segment.
Sex chromosome (n)	A chromosome that determines the sex of the individual. Sex chromosomes are one pair of the total 23 pairs of chromosomes in humans. There are 2 sex chromosomes, X and Y, that in combination determine the sex of an individual. Males are XY and females are XX.
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. Examples and considerations: The 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.
Specificity (n)	Refers to a test's ability to identify an individual without disease as testing negative for the disease. Examples and considerations: A test showing results as a true negative.

Sperm (n)	The reproductive cells of the male. Examples and considerations: When sperm fertilize a woman's egg, a baby develops.
Spermatocyte (n)	An immature male germ cell that develops into sperm. It is produced by the testes.
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.
Sperm source/donor (n)	Refers to the donation of sperm by a man with the intention that it be used in the artificial insemination or other fertility treatment of one or more women who are not his sexual partners in order that they may become pregnant by him. Where pregnancies go to full term, the sperm donor will be the biological father of every child born through this donation. See ART.
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.
Targeted carrier screening (n)	Carrier screening for diseases that are targeted at traditionally high-risk populations. Examples and considerations: Such as screening individuals of Ashkenazi Jewish descent for Tay–Sachs disease.
Teratogen (n)	A factor that has the potential to disturb the normal development of an embryo or fetus.
Termination of pregnancy (n)	See "abortion."
Test results (n): Negative (adj)	A negative result on a diagnostic genetic test means that the laboratory did not find the specific genetic change that the test was designed to identify. A "screen negative" result on a screening test means that the person's risk of having whatever the test was designed to find is lower than the risk for most people.

Test results (n): Positive (adj)	A positive result on a diagnostic genetic test means that the laboratory did find a specific genetic change that is associated with a hereditary condition (e.g. a deleterious mutation). A positive result on a diagnostic test means the person most likely has the condition detected by the test. A “screen positive” result on a screening test means that the person’s risk of having whatever the test screened for is significantly higher than expected. It does not mean that the person has that condition.
Trisomy (n)	Refers to the condition in which there are three copies of a chromosome instead of the usual complement of two of each chromosome found in each cell. See Aneuploidy.
Trisomy 13 (Patau's Syndrome) (n)	A rare, serious genetic disorder caused by having an additional copy of chromosome 13 in some or all the body's cells. It's also called trisomy 13. This is a severe condition affecting development and health outcomes. See Trisomy.
Turner's Syndrome (X0) (n)	Turner syndrome is a condition that affects only females, and results when one of the X chromosomes (sex chromosomes) is missing or partially missing. Turner syndrome can cause a variety of medical and developmental problems. See Sex chromosome.
Quad screen (n)	See Maternal Serum Screening.
Vanishing Twin Syndrome (VTS) 9N)	A miscarriage that causes a pregnancy involving twins to become a pregnancy involving one baby. It occurs when one of the embryos detected during an ultrasound stops developing. VTS can’t be treated or prevented.
Variant of uncertain significance (VUS) (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.” ——————————————— Examples and considerations: This means that the laboratory found a genetic change, but they don’t know what it means. Many 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.
Whole Exome Sequencing (WES) (n)	A type of genetic test that sequences the entire exome. It can be used to identify underlying causes of certain symptoms or disease. (See Exome)
Whole Genome Sequencing (WGS)	A genetic test that sequences and determines the order of bases in the genome.It can be used to identify underlying causes of certain symptoms or disease.(See Genome)
Polymorphism (n)	Everyone has some degree of commonly occurring genetic changes that are not associated with medical problems. 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. Examples and considerations: Eye color, blood type, risk for certain diseases are examples of traits.
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. Examples and Considerations: 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. Examples and considerations: For example, 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.
47XXX (n)	Also called Trisomy X or Triple X syndrome, is an aneuploidy in which a female has an extra X chromosome. Symptoms range from mild to moderate seizures and developmental disabilities.

47XYY (n)	Also known as Jacobs syndrome, is an aneuploidy in which a male has an extra Y chromosome. There are usually few symptoms such as being taller than average and an increased risk of learning disabilities. See Aneuploidy.

Family Relationship Terms

The family relationship terms are meant to be used as guidance and inform genetic counselors about biological relationships (i.e., if a relative is a blood relative).

Family Relationships

Term	Definition
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 Maternal uncle Paternal uncle	Your mother or father's brother or brother-in-law. Your mother's brother or brother-in-law. Your father's brother or brother-in-law.
Cousin First cousin First cousin once removed Second cousin	Usually understood to be a first cousin. Your aunt or uncle's child. Your aunt or uncle's grandchild or your first cousin's child The children of first cousins are second cousins to each other.
Grandparent	Your parent's father or mother.
Grandmother Maternal grandmother Paternal grandmother	Your mother or father's mother. Your mother's mother. Your father's mother.
Grandfather Maternal grandfather Paternal grandfather	Your mother or father's father. Your mother's father. Your father's father.
Great aunt Maternal great aunt Paternal great aunt	Your mother or father's aunt. Your mother's aunt. Your father's aunt.
Great uncle Maternal great uncle Paternal great uncle	Your father or mother's uncle. Your mother's 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.
Siblings Half siblings Stepsiblings	The children of your father and mother. 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.

	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 Identical twins Fraternal twins	Two siblings born at the same time. A twin that developed from the same egg and sperm, meaning that they are mostly genetically identical. Also called monozygotic twins. A twin who developed from a different egg and sperm than, meaning that he or she is genetically different and has the same number of shared genes as any other sibling. Also called dizygotic twins.
Triplets	Three siblings born at the same time
Sister Sister-in-law Half sister Stepsister	A sibling who is a girl. The sister of one's husband or wife. A girl who is either the child of your father with a different mother, or the child of your mother with a different father. The daughter of your stepmother or stepfather and therefore not biologically related to you.
Brother Brother-in-law Half brother Stepbrother	A sibling who is a boy. The brother one one's husband or wife. A boy who is either the child of your father with a different mother, or the child of your mother with a different father. The son of your stepmother or stepfather and therefore not biologically related to you.
Niece	Your sibling's daughter.
Nephew	Your sibling's son.
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 Daughter-in-law	Your child who is a girl. Your son's wife.

Stepdaughter Adoptive daughter Goddaughter	Your spouse's daughter who is not your biological child. A girl for whom you have accepted legal responsibility and raised even though she is not your biological child. A girl for whom you have accepted spiritual responsibility within a ceremony of the Catholic/Christian church; she may or may not be biologically related to you.
Son Son-in-law Stepson Adoptive son Godson	Your child who is a boy. Your daughter's husband. Your spouse's son who is not your biological child. A boy for whom you have accepted legal responsibility and raised even though he is not your biological child. A boy for whom you have accepted spiritual responsibility within a ceremony of the Catholic/ Christian 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
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.

Analogies

Genetic counselors often use analogies to explain complex ideas in genetics. An analogy explains a new concept by comparing it to something more common. A clue that an analogy is being used are the words "**It's as if**" and "**It's like**".

Example(s)
"Genes are **like** an instruction book or an instruction manual. And in a book, there is a certain sequence to a story."
"**It's as if** you were reading through a long book and looking for one typo, for a letter that is mistaken."
"A mutation is **like** an unexpected change in the order of the letters."
"A gene is **like** a recipe, and mutations are like changes in that recipe."
"It's **like** chapters in a book, and like misspellings."
"Our cells are **like** libraries and our chromosomes are like books."
"**Like** a flip of a coin." (to explain that there is a 50% chance that a mutation will be passed on every time the patient has a child).
"It is kind of **like** you a reading a book and you notice that a word is spelled wrong; that is what this genetic test looks for."

Some analogies may not work with certain patients, due to linguistic or experiential differences.
Analogies that focus on genes as "letters in a book" may make no sense to speakers of character-based written languages such as Chinese, Japanese, and Korean.

Example(s)
"**It's as if** you were reading through a long book and looking for one typo, for a letter that is mistaken."
"A mutation is **like** an unexpected change in the order of the letters."
"It's **like** chapters in a book, and like misspellings."
"It is kind of **like** if you a reading a book and you notice that a word is spelled wrong; that is what this genetic test looks for."

Analogies that focus on genes as a recipe may not work for patients who do not cook with written recipes.

Example(s)
A gene is **like** a recipe, and mutations are like changes in that recipe."

Analogies that discuss random chance in terms of a coin toss may not work for patients from cultures where people don't flip coins to make decisions.

Example(s)
"**Like** a flip of a coin" (to explain that there is a 50% chance that a mutation will be passed on every time the patient has a child)

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