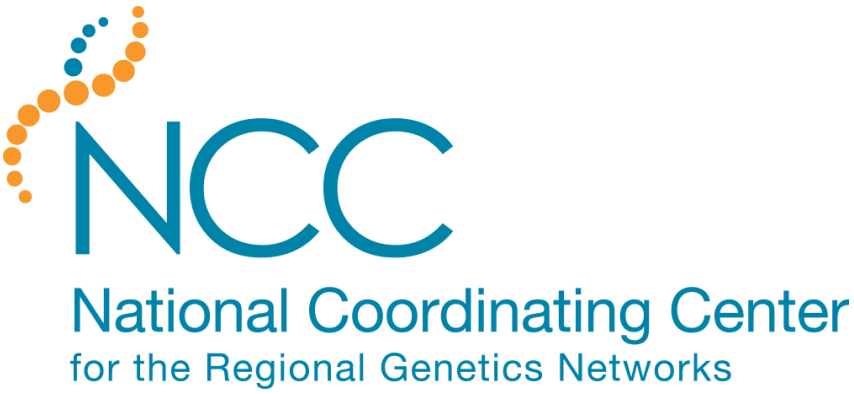

The National Coordinating Center for the Regional Genetics Networks

Glossary of Pediatric Genetics Terms

English – Portuguese

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 – Portuguese

(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.	Anormal
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."	Aborto
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	ACMG 59
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.	Habilidades adaptativas
Adoption (n)	The process of accepting legal responsibility for someone else's child to raise as your own.	Adoção
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.	Colégio Americano de Genética Médica e Genômica (ACMG)
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.	Amniocentese

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.	Líquido amniótico
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.	Anencefalia
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.	Benigno
Biochemical testing (n)	Blood tests to identify elevated levels of certain naturally occurring substances/chemicals in the body that are linked to genetic conditions.	Exames bioquímicos
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.	Canal de parto
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.”	Defeito de Nascimento
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.	Exame de Sangue
Brace (n)	A medical appliance that provides support to joints or bones, to weak muscles, or to strained ligaments.	Tala
Buccal swab (n)	A way to collect DNA from the cells on the inside of a person's cheek.	Esfregaço bucal
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.	Portador
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.	Célula

Term	Definition	Translation
Cervix (n)	The narrow lower part of the uterus than connects to the vagina.	Colo do útero /cérvice/ cérvix
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.	Biópsia de Vilo Corial (BVC)
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.	Cromossomo
Chromosome test / Karyotype (n)	A test that looks at the number and arrangement of chromosomes in a cell.	Exame de cromossomos/cariótipo
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.	Lábio leporino
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.	Palato fendido / fenda palatina
Clinically significant (adj)	If a test result is “clinically significant,” the result indicates a medical problem that can impact a person’s life.	Clinicamente significativo
Cognition (n)	The processes involved in thinking, learning, understanding, and remembering.	Cognição
Condition (e.g., “genetic condition”) (n)	A long-term medical health issue.	Condição (por ex. “condição genética”)
Consanguinity (n)	When parents are blood relatives to each other.	Consanguinidade
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.	Modelo de consulta
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.	Modelo de tratamento contínuo

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.	Fibrose cística
De novo (adj)	Genetic changes that are not inherited but that occur during cell division.	De novo
Deleterious mutation/ Disease-causing mutation (n)	A change in a person's DNA that may cause a medical condition.	Mutação deletéria/ Mutação causadora de doença
Deletion (n)	Having a section of genetic information (DNA) missing.	Deleção
Developmental assessment (n)	An evaluation of how an individual patient's intellectual, emotional, and social development compare with others of the same age.	Avaliação do desenvolvimento
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.	Teste/ exame de diagnóstico
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 (Ácido desoxirribonucleico)
DNA marker (also called a "Genetic marker") (n)	A readily recognizable genetic trait, gene, or DNA segment.	Marcador de DNA (também chamado de "Marcador Genético")
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.	Mutação do DNA

Term	Definition	Translation
DNA sequence (n)	The exact arrangement of the chemicals that make up a section of DNA.	Sequência de 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.	Sequenciamento de DNA
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.	Dominante
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.	Doador de óvulo ou esperma
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.	Síndrome de Down (Trissomia 21)
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.	Duplicação
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.	Síndrome de Edwards (Trissomia 18)
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."	Óvulo
Embryo (n)	An unborn mammal, between conception and 8 weeks of gestation.	Embrião
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.	Causas ou fatores ambientais

Term	Definition	Translation
Exome sequencing (n)	A genetic test in which a patient's exons are sequenced and studied.	Sequenciamento de exomas
Exon (n)	The part of the genome that codes for proteins. The majority of deleterious mutations occur in exons.	Exon
Fallopian tube (n)	The tube that connects an ovary to the uterus.	Trompa de falópio
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.	Falso negativo
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.	Falso positivo
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.	Familiar
Family history (n)	The medical history of the members of a biological family.	Histórico familiar
Fertilization (n)	The joining of an egg and sperm to create the first cell that will develop into an embryo, then fetus, then baby.	Fertilização
Fetal surgery (n)	Surgery conducted on a fetus while it is still in the uterus.	Cirurgia fetal
Fetus(n)	An unborn mammal, between 8 weeks of gestation to birth.	Feto
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.	Triagem pré-natal do primeiro trimester
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.	Cara ou coroa

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.	Síndrome do X frágil
Fragment (n)	A small piece; an incomplete part of a whole.	Fragmento
Gene (n)	A specific sequence of DNA that codes for one or many functions within the cell and body.	Gene
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.	Cópia do gene
Normal gene copy (n)	A normal copy of a gene is one that is found most frequently in the population. Also called the "working copy."	Cópia normal do gene
Altered gene copy (n)	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."	Cópia alterada do gene
Gene deletion (n)	Having a piece of genetic information missing from a gene.	Exclusão do gene
Gene panel testing (n)	Genetic testing that looks at 2 or more specific genes known to be associated with particular conditions.	Testagem de painel do gene
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.	Variante do gene
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.	População em geral

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.	Geração
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).	Alfabeto genético
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.	Aconselhamento genético
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.	Conselheiro genético
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.	Discriminação genética
Genetic factors (n)	Specific aspects of a person's genetic make-up that influence that person's health and development.	Fatores genéticos
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.	Informação genética
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.	Material genético

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.	Predisposição genética
Genetic test (n)	A laboratory test designed to determine if a person has a gene mutation or a typical DNA sequence.	Exame genético
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.	Exame de gene de local único
Individual gene testing (n)	Individual gene testing: genetic analysis of the entirety of one specific gene.	Exame de gene individual
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.	Exame de painel do gene
Microarray (n)	Microarray: genetic analysis that looks for unexpected numbers of specific genes (two copies of every gene code is usually expected).	Microarranjo
Exome sequencing (n)	Exome testing: a genetic test in which the exons of a patient's genetic code are sequenced and studied.	Sequenciamento de exoma
Genome sequencing (n)	Genome sequencing: a genetic test in which the entire genetic code (the genome) of a patient is mapped out and studied.	Sequenciamento de genoma
Genetic trait (n)	A characteristic within a family that is passed down from parent to child through their DNA.	Traço genético
Geneticist (n)	A doctor or scientist who studies genetics.	Geneticista
Genetics (adj)	The science of how an organism's genes interact with the environment to produce certain traits.	Genético(a)
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.	Genética
Genome (n)	The entire genetic code of an individual.	Genoma
Genome sequencing (n)	A genetic test in which the entire genetic code of a patient is mapped out and studied.	Sequenciamento de genoma

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.	Teste da linha germinal
Guardianship (n)	A legal arrangement, supervised by a court, in which one person takes responsibility for a minor or an adult with a disability.	Tutoria
Helix (n)	Something that is spiral in form. DNA is often described as being a "double helix."	Hélice
Hereditary (adj)	Passed down from parent to child.	Hereditário(a)
Hereditary material (n)	Genetic material that is passed down from parent to child.	Material hereditário
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.	Doença de Huntington
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.	Plano de Educação Individualizada (PEI)
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.	Fertilização In Vitro (FIV)
Individual gene testing (n)	Genetic analysis of the entirety of one specific gene.	Testagem de genes individuais
Infertility (n)	The inability to have children.	Infertilidade

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.	Consentimento informado
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.	Padrão hereditário
Inherited (adj)	Passed down from parent to child.	Herdado(a)
Insertion (n)	An extra segment of DNA added in at a place where it is not usually found.	Inserção
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.	Deficiência intelectual
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.	Síndrome de Klinefelter (47, XXY)
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.	Síndrome de Lynch
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.	Cromossomo marcador
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.	Exame de Alfa-Fetoproteína do Soro Materno (AFPSM)
Metastasis (n)	The spreading of cancer from one organ to another.	Metástase

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.	Microarranjo
Miscarriage (n)	The spontaneous (not intentional) loss of a pregnancy. See "abortion." Also called pregnancy loss.	Aborto espontâneo
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.	Molécula
Mosaicism (n)	A condition in which some, but not all, cells in a sample show a genetic difference.	Mosaicismo
Motor Abilities (n)	The ability to move and use one's muscles.	Habilidades motoras
Multifactorial (adj)	Due to a combination of genetic and non-genetic (environmental, hormonal, etc.) risk factors that act together to determine risk.	Multifatorial
Mutation (n)	A change in a gene, usually deleterious. See "genetic variant."	Mutação
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.	Testagem Pré-Natal Não Invasiva (TPNNI)
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.	Translucência Nucal Aumentada

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.	Defeitos Abertos do Tubo Neural (DTN)
Ova (n)	See “egg.”	Óvulos
Ovary (n)	The organ in a woman that stores and releases eggs. There are normally two.	Ovário
Packets of genetic information (n)	A phrase genetic counselors use to describe genes or chromosomes.	Pacotes de informação genética
Pathogenic (adj)	Disease causing.	Patogênico
Pedigree (n)	A family tree that can be used to trace the inheritance of specific genetic traits.	Linhagem
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.	Penetrância
Placenta (n)	The organ that develops together with an embryo in a pregnant woman’s uterus to nourish the embryo through the umbilical cord.	Placenta
Presymptomatic (adj)	Before symptoms appear.	Pré-sintomático
Prognosis (n)	The most likely outcome of a disease process.	Prognóstico
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.	Proteína
Random (adj)	Happening in an unpredictable way.	Aleatório

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.	Recessivo
Red flag (n)	A warning sign or a clue.	Sinal de Alerta
Replicate (v)	To copy.	Replicar
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.	Histórico reprodutivo
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.	Cromossomo em Anel
Risk (n)	The chance that something will happen; in the case of prenatal genetics, the chance that the child will have a genetic condition.	Risco
At risk (adj)	Has some possibility of having a genetic condition.	Em risco
At average risk (adj)	Has the same possibility of having a genetic condition as the general population.	Em risco moderado
At high risk (adj)	Has a greater possibility of having a genetic condition than the general population.	Em alto risco
At higher risk than "X" (adj)	Has a greater possibility of having a genetic condition than "X."	Em risco mais alto que "X"
Risk factor (n)	A circumstance that increases the risk of having a genetic condition.	Fator de risco
Runs in the family	Passed down from parents to children to grandchildren.	É de família
Saliva (spit) sample (n)	A way to collect DNA from the cells in saliva (spit).	Amostra de saliva (cuspe)

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.	Triagem
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."	Teste de triagem/ exame de rastreamento
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.	Sêmen
Seminal fluid (n)	See "semen."	Líquido Seminal
Sensory abilities (n)	The ability to see, hear, touch, taste, and smell.	Habilidades sensoriais
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.	Doença das células falciformes
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.	Análise de local único
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.	Alterações genéticas somáticas
Special education (n)	Education that is tailored to individuals with developmental delays or other learning disabilities.	Educação especial
Sperm (n)	The reproductive cells of the man. When sperm fertilize a woman's egg, a baby develops.	Esperma

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.	Doação de esperma
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.	Espinha bífida
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.	Estatisticamente significativo
Supportive seating (n)	Seating, such as a wheelchair, that helps an individual function in their environment and that prevents worsening of their condition.	Assento de apoio
Teratogen (n)	Something that has the potential to disturb the normal development of an embryo or fetus.	Teratogênico
Termination of pregnancy (n)	See “abortion.”	Interrupção da gravidez/gestação

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.	Resultado do teste
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.	Verdadeiro negativo
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.	Positivo (adj)
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.	Variante de significado desconhecido
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."	Polimorfismo
Testes (n)	The organs in a man that create and release sperm. Also called "testicles." Singular testis.	Testículos
Toxic exposure (n)	Contact with something that is harmful or poisonous.	Exposição tóxica
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.	Traço

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.	Translocação
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.	Erro tipográfico
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.	Ultrassom
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."	Útero
Vagina (n)	The tube-like part of the female reproductive system that extends from the cervix to the outside of the body.	Vagina
Variant (n)	A version of something that differs from the norm. So, a genetic variant is a change to the usual genetic sequence.	Variante
Vas deferens (n)	The tubes that lead from the testes to the urethra, through which sperm is ejaculated.	Canal deferente
X-linked (adj)	A trait that is influenced or determined by a gene on the "X" chromosome.	Vinculado a X

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.	Pai ou mãe
Mother	For genetic purposes, the woman whose egg was fertilized and grew to be you.	Mãe
Mother-in-law	Your husband or wife's mother.	Sogra
Stepmother	Your father's wife who is not your biological mother.	Madrasta
Adoptive mother	A woman who is not your biological mother but who accepted legal responsibility for and raised you.	Mãe adotiva
Godmother	A woman chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.	Madrinha
Father	For genetic purposes, the man whose sperm fertilized the egg that grew to be you.	Pai
Father-in-law	Your husband or wife's father.	Sogro
Stepfather	Your mother's husband who is not your biological father.	Padrasto
Adoptive father	A man who is not your biological father but who accepted legal responsibility for and raised you.	Pai adotivo
Godfather	A man chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.	Padrinho
Aunt	Your mother or father's sister or sister-in-law.	Tia
Maternal aunt	Your mother's sister or sister-in-law.	Tia do lado materno
Paternal aunt	Your father's sister or sister-in-law.	Tia do lado paterno
Uncle	Your mother or father's brother or brother-in-law.	Tio
Maternal uncle	Your mother's brother or brother-in-law.	Tio do lado materno
Paternal uncle	Your father's brother or brother-in-law.	Tio do lado paterno

Term	Definition	Translation
Cousin	Usually understood to be a first cousin.	Primo
First cousin	Your aunt or uncle's child.	Primo de primeiro grau
First cousin once removed	Your aunt or uncle's grandchild or your first cousin's child	Primo de segundo grau
Second cousin	The children of first cousins are second cousins to each other.	Primo de terceiro grau
Grandparent	Your parent's father or mother.	Avô
Grandmother	Your mother or father's mother.	Avó
Maternal grandmother	Your mother's mother.	Avó do lado materno
Paternal grandmother	Your father's mother.	Avó do lado paterno
Grandfather	Your mother or father's father.	Avô
Maternal grandfather	Your mother's father.	Avô do lado materno
Paternal grandfather	Your father's father.	Avô do lado paterno
Great aunt	Your mother or father's aunt.	Tia-avó
Maternal great aunt	Your mother's aunt.	Tia-avó do lado materno
Paternal great aunt	Your father's aunt.	Tia-avó do lado paterno
Great uncle	Your father or mother's uncle.	Tio-avô
Maternal great uncle	Your mother's uncle.	Tio-avô do lado materno
Paternal great uncle	Your father's uncle.	Tio-avô do lado paterno
Great grandparents	The parents of any of your grandparents.	Bisavós
Great grandmother	The mother of any of your grandparents.	Bisavó
Great grandfather	The father of any of your grandparents.	Bisavô

Term	Definition	Translation
Siblings	The children of your father and mother.	Irmãos
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.	Meio-irmãos
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.	Irmãos de madrasta ou padrasto Irmãos postiços
Twins	Two siblings born at the same time.	Gêmeos
Identical twins	Twins who developed from the same egg and sperm, meaning that they are genetically identical. Also called monozygotic twins.	Gêmeos idênticos
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.	Gêmeos fraternos
Triplets	Three siblings born at the same time.	Trigêmeos
Sister	A sibling who is a girl.	Irmã
Sister-in-law	Your brother's wife or your husband's sister.	Cunhada
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.	Meio-irmã
Stepsister	The daughter of your stepmother or stepfather and therefore not biologically related to you.	Irmã de madrasta ou padrasto/ irmã postiça
Brother	A sibling who is a boy.	Irmão
Brother-in-law	Your sister's husband or your wife's brother.	Cunhado
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.	Meio-irmão
Stepbrother	The son of your stepmother or stepfather and therefore not biologically related to you.	Irmão de madrasta ou padrasto/ irmão postiço

Term	Definition	Translation
Niece	Your sibling's daughter.	Sobrinha
Nephew	Your sibling's son.	Sobrinho
Spouse	Your husband or wife.	Esposo(a)
Wife	The woman to whom you are married.	Mulher
Husband	The man to whom you are married.	Marido
Children	Genetically speaking, the people who are produced from your egg or sperm.	Filhos
Daughter	Your child who is a girl.	Filha
Daughter-in-law	Your son's wife.	Nora
Stepdaughter	Your spouse's daughter who is not your biological child.	Enteada
Adoptive daughter	A girl for whom you have accepted legal responsibility and raised even though she is not your biological child.	Filha adotiva
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.	Afilhada
Son	Your child who is a boy.	Filho
Son-in-law	Your daughter's husband.	Genro
Stepson	Your spouse's son who is not your biological child.	Enteado
Adoptive son	A boy for whom you have accepted legal responsibility and raised even though he is not your biological child.	Filho adotivo
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.	Afilhado
Ancestor	A person from whom you are descended, usually more remote than a grandparent.	Ancestral

Term	Definition	Translation
Fiancé(e)	The person whom you have promised to marry. Man: fiancé Woman: fiancée	Noivo(a)
Domestic partner	The person with whom you are living and have an intimate relationship, but to whom you are not married.	Parceiro doméstico
Divorced	Having ended a marriage.	Divorciado(a)
Engaged	Having promised to marry someone.	Estar noivo(a)
Widow	A woman whose husband has died.	Viúva
Widower	A man whose wife has died.	Viúvo
Relative	A person in your family.	Parente
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.	Relação consanguínea/ laço sanguíneo
Next of kin	Your closest living blood relation.	Parentes próximos

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