



National Coordinating Center
for the Regional Genetics Networks

**The National Coordinating Center for the
Regional Genetics Networks**

Glossary of Pediatric Genetics Terms

English – French

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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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 – French

(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.”	Avortement
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.	Capacités d’adaptation
Adoption (n)	The process of accepting legal responsibility for someone else’s child to raise as your own.	Adoption
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.	American College of Medical Genetics and Genomics (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.	Amniocentèse

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.	Liquide amniotique
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.	Anencéphalie
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.	Bénin(e)
Biochemical testing (n)	Blood tests to identify elevated levels of certain naturally occurring substances/chemicals in the body that are linked to genetic conditions.	Tests biochimiques
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.	Filière pelvi-génitale/ canal utérin/ canal de naissance
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.”	Déficiência congénitale/ anomalie de naissance
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.	Analyse sanguine
Brace (n)	A medical appliance that provides support to joints or bones, to weak muscles, or to strained ligaments.	Orthèse/ attelle
Buccal swab (n)	A way to collect DNA from the cells on the inside of a person's cheek.	Frottis buccal
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.	Porteur/porteuse
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.	Cellule

Term	Definition	Translation
Cervix (n)	The narrow lower part of the uterus than connects to the vagina.	Col utérin
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.	Prélèvement de villosités choriales (PVC)
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
Chromosome test / Karyotype (n)	A test that looks at the number and arrangement of chromosomes in a cell.	Analyse chromosomique/caryotype
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.	Fente labiale/Bec-de-lièvre
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.	Fente palatine
Clinically significant (adj)	If a test result is “clinically significant,” the result indicates a medical problem that can impact a person’s life.	Important sur le plan clinique
Cognition (n)	The processes involved in thinking, learning, understanding, and remembering.	Cognition
Condition (e.g., “genetic condition”) (n)	A long-term medical health issue.	Trouble/affection (par ex., « affection génétique »)
Consanguinity (n)	When parents are blood relatives to each other.	Consanguinité
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.	Modèle de consultation
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.	Modèle de soins continus

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 kystique
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.	Mutation délétère/mutation entraînant une maladie
Deletion (n)	Having a section of genetic information (DNA) missing.	Délétion
Developmental assessment (n)	An evaluation of how an individual patient’s intellectual, emotional, and social development compare with others of the same age.	Évaluation du développement
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 <u>an increased risk</u> for the medical problem in question, whereas diagnostic tests report whether the problem is actually present.	Test de diagnostic
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.	ADN (acide désoxyribonucléique)
DNA marker (also called a “Genetic marker”) (n)	A readily recognizable genetic trait, gene, or DNA segment.	Marqueur d’ADN (aussi nommé « marqueur génétique »)
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.	Mutation de l’ADN

Term	Definition	Translation
DNA sequence (n)	The exact arrangement of the chemicals that make up a section of DNA.	Séquence de l'ADN
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.	Séquençage de l'ADN
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.	Dominant
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.	Don d'ovule ou de sperme
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.	Syndrome de Down (trisomie 21)
Duplication (n)	Having an extra section of genetic information (DNA). A <i>duplication</i> occurs when part of a chromosome is copied abnormally, resulting in extra <i>genetic</i> material.	Duplication
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.	Syndrome d'Edwards (trisomie 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."	Ovule
Embryo (n)	An unborn mammal, between conception and 8 weeks of gestation.	Embryon
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.	Causes environnementales ou facteurs environnementaux

Term	Definition	Translation
Exome sequencing (n)	A genetic test in which a patient’s exons are sequenced and studied.	Séquençage de l’exome
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.	Trompe de Fallope
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.	Faux négatif
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.	Faux positif
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.	Familial
Family history (n)	The medical history of the members of a biological family.	Antécédent familial
Fertilization (n)	The joining of an egg and sperm to create the first cell that will develop into an embryo, then fetus, then baby.	Fécondation
Fetal surgery (n)	Surgery conducted on a fetus while it is still in the uterus.	Chirurgie fœtale
Fetus(n)	An unborn mammal, between 8 weeks of gestation to birth.	Fœtus
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.	Dépistage au cours du premier trimestre
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.	Tirer à pile ou face

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.	Syndrome de l’X fragile
Fragment (n)	A small piece; an incomplete part of a whole.	Fragment
Gene (n)	A specific sequence of DNA that codes for one or many functions within the cell and body.	Gène
<p data-bbox="204 611 363 642">Gene copy (n)</p> <p data-bbox="250 869 493 900">Normal gene copy (n)</p> <p data-bbox="250 999 493 1031">Altered gene copy (n)</p>	<p data-bbox="518 611 1097 835">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.</p> <p data-bbox="518 869 1097 968">A normal copy of a gene is one that is found most frequently in the population. Also called the “working copy.”</p> <p data-bbox="518 999 1097 1098">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.”</p>	<p data-bbox="1138 611 1292 642">Copie de gène</p> <p data-bbox="1159 869 1419 900">Copie d’un gène normal</p> <p data-bbox="1159 999 1419 1031">Copie d’un gène modifié</p>
Gene deletion (n)	Having a piece of genetic information missing from a gene.	Délétion d’un gène
Gene panel testing (n)	Genetic testing that looks at 2 or more specific genes known to be associated with particular conditions.	Analyse d’un panel de gènes
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.	Variant génétique
General population (n)	<p data-bbox="518 1472 688 1503">“Most people.”</p> <p data-bbox="518 1535 1105 1728">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.</p>	Population générale

Term	Definition	Translation
Generation (n)	<p>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.</p> <p>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.</p>	Génération
Genetic alphabet (n)	<p>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).</p>	Alphabet génétique
Genetic counseling (n)	<p>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.</p>	Consultation génétique
Genetic counselor (n)	<p>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.</p>	Conseiller en génétique
Genetic discrimination (n)	<p>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.</p>	Discrimination génétique
Genetic factors (n)	<p>Specific aspects of a person’s genetic make-up that influence that person’s health and development.</p>	Facteurs génétiques
Genetic information (n)	<p>The instructions encoded in DNA, which tells every cell in a body how to grow, what to do and how to reproduce.</p>	Informations génétiques

Term	Definition	Translation
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.	Matériel génétique
Genetic predisposition (n)	An increased chance of a person developing a certain trait or disease based on that person's particular genetic makeup.	Prédisposition génétique
Genetic test (n) Single site gene testing (n) Individual gene testing (n) Gene panel test (n) Microarray (n) Exome sequencing (n) Genome sequencing (n)	<p>A laboratory test designed to determine if a person has a gene mutation or a typical DNA sequence.</p> <p>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.</p> <p>Individual gene testing: genetic analysis of the entirety of one specific gene.</p> <p>Gene panel test: analysis of several genes that have been shown to be associated with a particular condition, e.g., breast cancer gene panel.</p> <p>Microarray: genetic analysis that looks for unexpected numbers of specific genes (two copies of every gene code is usually expected).</p> <p>Exome testing: a genetic test in which the exons of a patient's genetic code are sequenced and studied.</p> <p>Genome sequencing: a genetic test in which the entire genetic code (the genome) of a patient is mapped out and studied.</p>	<p>Test génétique</p> <p>Test génétique sur un point spécifique</p> <p>Test d'un gène individuel</p> <p>Test d'un panel de gènes</p> <p>Puce à ADN</p> <p>Séquençage de l'exome</p> <p>Séquençage du génome</p>
Genetic trait (n)	A characteristic within a family that is passed down from parent to child through their DNA.	Caractère génétique
Geneticist (n)	A doctor or scientist who studies genetics.	Généticien(ne)
Genetics (adj)	The science of how an organism's genes interact with the environment to produce certain traits.	Génétique
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.	Génétique

Term	Definition	Translation
Genome (n)	The entire genetic code of an individual.	Génome
Genome sequencing (n)	A genetic test in which the entire genetic code of a patient is mapped out and studied.	Séquençage du génome
Germline testing (n)	<p>Germline testing refers to the analysis of a person’s DNA, which he or she inherited from his or her parents.</p> <p>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.</p>	Analyse germinale
Guardianship (n)	A legal arrangement, supervised by a court, in which one person takes responsibility for a minor or an adult with a disability.	Tutelle
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.	Héréditaire
Hereditary material (n)	Genetic material that is passed down from parent to child.	Matériel héréditaire
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.	Maladie 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.	Plan éducatif individualisé
In Vitro Fertilization (IVF) (n)	<p>The fertilization of an egg by a sperm outside of a woman’s body.</p> <p>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.</p>	Fécondation/Fertilisation In Vitro (FIV)

Term	Definition	Translation
Individual gene testing (n)	Genetic analysis of the entirety of one specific gene.	Test d'un gène individuel
Infertility (n)	The inability to have children.	Infertilité/Infécondité
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.	Consentement éclairé
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.	Transmission héréditaire
Inherited (adj)	Passed down from parent to child.	Hérité(e)
Insertion (n)	An extra segment of DNA added in at a place where it is not usually found.	Insertion
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.	Handicap intellectuel
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.	Syndrome 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.	Syndrome 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.	Chromosome marqueur
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.	Dépistage par le sérum maternel (analyse du taux d'alpha-fœtoprotéine sérique maternelle)
Metastasis (n)	The spreading of cancer from one organ to another.	Métastase

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.	Puce à ADN
Miscarriage (n)	The spontaneous (not intentional) loss of a pregnancy. See “abortion.” Also called pregnancy loss.	Fausse couche
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.	Molécule
Mosaicism (n)	A condition in which some, but not all, cells in a sample show a genetic difference.	Mosaïcisme
Motor Abilities (n)	The ability to move and use one’s muscles.	Capacités motrices
Multifactorial (adj)	Due to a combination of genetic and non-genetic (environmental, hormonal, etc.) risk factors that act together to determine risk.	Multifactoriel(le)
Mutation (n)	A change in a gene, usually deleterious. See “genetic variant.”	Mutation
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.	Test de dépistage prénatal non invasif
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.	Épaississement de la clarté nucale

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.	Anomalie de la moelle épinière par défaut de soudure
Ova (n)	See “egg.”	Ovules
Ovary (n)	The organ in a woman that stores and releases eggs. There are normally two.	Ovaire
Packets of genetic information (n)	A phrase genetic counselors use to describe genes or chromosomes.	Paquets d’information génétique
Pathogenic (adj)	Disease causing.	Pathogène
Pedigree (n)	A family tree that can be used to trace the inheritance of specific genetic traits.	Arbre généalogique
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.	Pénétrance
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.	Présymptomatique
Prognosis (n)	The most likely outcome of a disease process.	Pronostic
Protein (n)	A molecule made up of chains of <i>amino acids</i> . 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.	Protéine
Random (adj)	Happening in an unpredictable way.	Aléatoire

Term	Definition	Translation
Recessive (adj)	<p>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.</p> <p>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.</p>	Récessif
Red flag (n)	A warning sign or a clue.	Signal d’alarme
Replicate (v)	To copy.	Répliquer
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.	Histoire reproductive
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.	Chromosome en anneau
<p>Risk (n)</p> <p>At risk (adj)</p> <p>At average risk (adj)</p> <p>At high risk (adj)</p> <p>At higher risk than “X” (adj)</p> <p>Risk factor (n)</p>	<p>The chance that something will happen; in the case of prenatal genetics, the chance that the child will have a genetic condition.</p> <p>Has some possibility of having a genetic condition.</p> <p>Has the same possibility of having a genetic condition as the general population.</p> <p>Has a greater possibility of having a genetic condition than the general population.</p> <p>Has a greater possibility of having a genetic condition than “X.”</p> <p>A circumstance that increases the risk of having a genetic condition.</p>	<p>Risque</p> <p>À risque</p> <p>À risque moyen</p> <p>À haut risque</p> <p>À risque plus élevé que « X »</p> <p>Facteur de risque</p>
Runs in the family	Passed down from parents to children to grandchildren.	Avoir des antécédents dans la famille
Saliva (spit) sample (n)	A way to collect DNA from the cells in saliva (spit).	Échantillon de salive

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.	Dépister
Screening test (n)	<p>A test that looks to detect a particular health problem, even if a patient has no symptoms of that particular condition.</p> <p>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.”</p>	Test de dépistage
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.	Semence/sperme
Seminal fluid (n)	See “semen.”	Liquide séminal/sperme
Sensory abilities (n)	The ability to see, hear, touch, taste, and smell.	Capacités sensorielles
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.	Maladie drépanocytaire/ drépanocytose
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.	Analyse sur un point spécifique
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.	Mutation génétique somatique
Special education (n)	Education that is tailored to individuals with developmental delays or other learning disabilities.	Éducation spécialisée
Sperm (n)	The reproductive cells of the man. When sperm fertilize a woman’s egg, a baby develops.	Sperme

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.	Don de sperme
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.	Spina bifida
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.	Important sur le plan statistique
Supportive seating (n)	Seating, such as a wheelchair, that helps an individual function in their environment and that prevents worsening of their condition.	Dispositif d'assise de soutien
Teratogen (n)	Something that has the potential to disturb the normal development of an embryo or fetus.	Tératogène
Termination of pregnancy (n)	See "abortion."	Interruption de grossesse

Term	Definition	Translation
<p>Test result (n) Negative (adj)</p> <p>True negative (n)</p> <p>Positive (adj)</p> <p>Variant of uncertain significance (n)</p> <p>Polymorphism (n)</p>	<p>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.</p> <p>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.</p> <p>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.</p> <p>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.</p> <p>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.”</p>	<p>Résultat d’analyse Négatif</p> <p>Vrai négatif</p> <p>Positif</p> <p>Mutation d’importance incertaine</p> <p>Polymorphisme</p>
Testes (n)	The organs in a man that create and release sperm. Also called “testicles.” Singular testis.	Testicules
Toxic exposure (n)	Contact with something that is harmful or poisonous.	Exposition à des substances toxiques
Trait (n)	<p>A characteristic of a person.</p> <p>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.</p>	Trait

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.	Translocation
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.	Faute de frappe/coquille
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.	Ultrason/ échographie
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.”	Utérus
Vagina (n)	The tube-like part of the female reproductive system that extends from the cervix to the outside of the body.	Vagin
Variant (n)	A version of something that differs from the norm. So, a genetic variant is a change to the usual genetic sequence.	Mutation
Vas deferens (n)	The tubes that lead from the testes to the urethra, through which sperm is ejaculated.	Canal déférent
X-linked (adj)	A trait that is influenced or determined by a gene on the “X” chromosome.	Lié(e) au chromosome 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.	Parent
Mother	For genetic purposes, the woman whose egg was fertilized and grew to be you.	Mère
Mother-in-law	Your husband or wife’s mother.	Belle-mère
Stepmother	Your father’s wife who is not your biological mother.	Belle-mère (par alliance)
Adoptive mother	A woman who is not your biological mother but who accepted legal responsibility for and raised you.	Mère adoptive
Godmother	A woman chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.	Marraine
Father	For genetic purposes, the man whose sperm fertilized the egg that grew to be you.	Père
Father-in-law	Your husband or wife’s father.	Beau-père
Stepfather	Your mother’s husband who is not your biological father.	Beau-père (par alliance)
Adoptive father	A man who is not your biological father but who accepted legal responsibility for and raised you.	Père adoptif
Godfather	A man chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.	Parrain
Aunt	Your mother or father’s sister or sister-in-law.	Tante
Maternal aunt	Your mother’s sister or sister-in-law.	Tante maternelle
Paternal aunt	Your father’s sister or sister-in-law.	Tante paternelle
Uncle	Your mother or father’s brother or brother-in-law.	Oncle
Maternal uncle	Your mother’s brother or brother-in-law.	Oncle maternel
Paternal uncle	Your father’s brother or brother-in-law.	Oncle paternel

Term	Definition	Translation
<p>Cousin</p> <p>First cousin</p> <p>First cousin once removed</p> <p>Second cousin</p>	<p>Usually understood to be a first cousin.</p> <p>Your aunt or uncle’s child.</p> <p>Your aunt or uncle’s grandchild or your first cousin’s child</p> <p>The children of first cousins are second cousins to each other.</p>	<p>Cousin(e)</p> <p>Cousin germain/ cousine germaine</p> <p>Cousin(e) issu(e) de germain/ petit(e) cousin(e)</p> <p>Cousin(e) issu(e) de germain/ cousin(e) au deuxième degré</p>
<p>Grandparent</p>	<p>Your parent’s father or mother.</p>	<p>Grand-parent</p>
<p>Grandmother</p> <p>Maternal grandmother</p> <p>Paternal grandmother</p>	<p>Your mother or father’s mother.</p> <p>Your mother’s mother.</p> <p>Your father’s mother.</p>	<p>Grand-mère</p> <p>Grand-mère maternelle</p> <p>Grand-mère paternelle</p>
<p>Grandfather</p> <p>Maternal grandfather</p> <p>Paternal grandfather</p>	<p>Your mother or father’s father.</p> <p>Your mother’s father.</p> <p>Your father’s father.</p>	<p>Grand-père</p> <p>Grand-père maternel</p> <p>Grand-père paternel</p>
<p>Great aunt</p> <p>Maternal great aunt</p> <p>Paternal great aunt</p>	<p>Your mother or father’s aunt.</p> <p>Your mother’s aunt.</p> <p>Your father’s aunt.</p>	<p>Grand-tante</p> <p>Grand-tante maternelle</p> <p>Grand-tante paternelle</p>
<p>Great uncle</p> <p>Maternal great uncle</p> <p>Paternal great uncle</p>	<p>Your father or mother’s uncle.</p> <p>Your mother’s uncle.</p> <p>Your father’s uncle.</p>	<p>Grand-oncle</p> <p>Grand-oncle maternel</p> <p>Grand-oncle paternel</p>
<p>Great grandparents</p>	<p>The parents of any of your grandparents.</p>	<p>Arrière-grands-parents</p>
<p>Great grandmother</p>	<p>The mother of any of your grandparents.</p>	<p>Arrière-grand-mère</p>
<p>Great grandfather</p>	<p>The father of any of your grandparents.</p>	<p>Arrière-grand-père</p>

Term	Definition	Translation
<p>Siblings</p> <p>Half siblings</p> <p>Stepsiblings</p>	<p>The children of your father and mother.</p> <p>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.</p> <p>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.</p>	<p>Germaids/enfants de même parents</p> <p>Demi-fratrie</p> <p>Frère ou sœur par alliance</p>
<p>Twins</p> <p>Identical twins</p> <p>Fraternal twins</p>	<p>Two siblings born at the same time.</p> <p>Twins who developed from the same egg and sperm, meaning that they are genetically identical. Also called monozygotic twins.</p> <p>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.</p>	<p>Jumeaux</p> <p>Jumeaux identiques</p> <p>Jumeaux dizygotes</p>
<p>Triplets</p>	<p>Three siblings born at the same time.</p>	<p>Triplets</p>
<p>Sister</p> <p>Sister-in-law</p> <p>Half sister</p> <p>Stepsister</p>	<p>A sibling who is a girl.</p> <p>Your brother’s wife or your husband’s sister.</p> <p>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.</p> <p>The daughter of your stepmother or stepfather and therefore not biologically related to you.</p>	<p>Sœur</p> <p>Belle-sœur</p> <p>Demi-sœur</p> <p>Sœur par alliance</p>
<p>Brother</p> <p>Brother-in-law</p> <p>Half brother</p> <p>Stepbrother</p>	<p>A sibling who is a boy.</p> <p>Your sister’s husband or your wife’s brother.</p> <p>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.</p> <p>The son of your stepmother or stepfather and therefore not biologically related to you.</p>	<p>Frère</p> <p>Beau-frère</p> <p>Demi-frère</p> <p>Frère par alliance</p>
<p>Niece</p>	<p>Your sibling’s daughter.</p>	<p>Nièce</p>
<p>Nephew</p>	<p>Your sibling’s son.</p>	<p>Neveu</p>

Term	Definition	Translation
Spouse	Your husband or wife.	Conjoint
Wife	The woman to whom you are married.	Épouse
Husband	The man to whom you are married.	Époux
Children	Genetically speaking, the people who are produced from your egg or sperm.	Enfants
Daughter	Your child who is a girl.	Fille
Daughter-in-law	Your son's wife.	Belle-fille
Stepdaughter	Your spouse's daughter who is not your biological child.	Belle-fille (par alliance)
Adoptive daughter	A girl for whom you have accepted legal responsibility and raised even though she is not your biological child.	Fille adoptive
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.	Filleule
Son	Your child who is a boy.	Fils
Son-in-law	Your daughter's husband.	Beau-fils
Stepson	Your spouse's son who is not your biological child.	Beau-fils (par alliance)
Adoptive son	A boy for whom you have accepted legal responsibility and raised even though he is not your biological child.	Fils adoptif
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.	Filleul
Ancestor	A person from whom you are descended, usually more remote than a grandparent.	Ancêtre
Fiancé(e)	The person whom you have promised to marry. Man: fiancé Woman: fiancée	Fiancé(e)

Term	Definition	Translation
Domestic partner	The person with whom you are living and have an intimate relationship, but to whom you are not married.	Compagne/compagnon
Divorced	Having ended a marriage.	Divorcé(e)
Engaged	Having promised to marry someone.	Fiancé(e)
Widow	A woman whose husband has died.	Veuve
Widower	A man whose wife has died.	Veuf
Relative	A person in your family.	Membre de la famille/ Parent (AF)
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.	Parent par le sang
Next of kin	Your closest living blood relation.	Plus proche parent

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