National Coordinating Center
for the Regional Genetics Networks

The National Coordinating Center for the Regional Genetics Networks

Glossaries of Genetics Terms

English – French

2023 Edition

Compiled and edited by:
Cynthia E. Roat, MPH

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

This page intentionally left blank.

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.

NCC Funding Acknowledgment

This project is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) under Cooperative Agreement #UH9MC30770 from 6/2020-5/2024 for $800,000 per award year. This information or content and conclusions are those of the author and should not be construed as the official position or policy of, nor should any endorsements be inferred by HRSA, HHS or the U.S. Government.

Additional Acknowledgments

Cynthia E. Roat, MPH

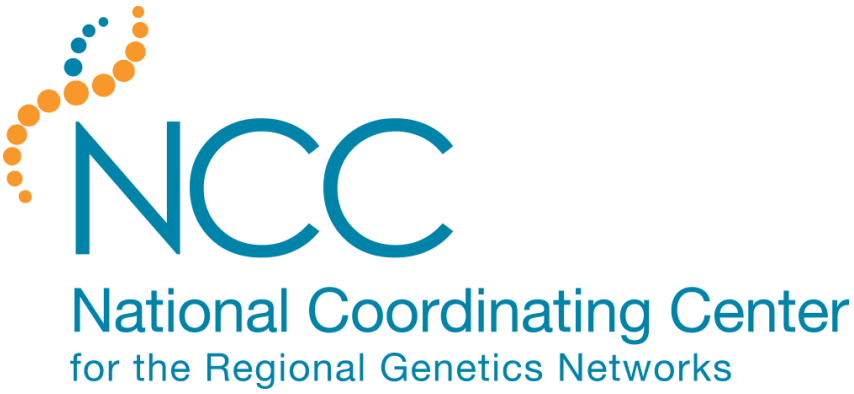
Cindy Roat is an international consultant in language access in health care and patient navigation. She provides training and consulting for health care interpreters, providers, administrators, and patient navigators. Cindy compiled and edited this glossary for the NCC.

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/

Table of Contents



The National Coordinating Center for the Regional Genetics Networks

Glossary of Autism Spectrum Disorder (ASD) Genetics Terms

English – French

2022 Edition

Compiled and edited by:
Cynthia E. Roat, MPH

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

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 Autism Spectrum Disorders (ASD) is provided to help enable access to ASD 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 is a list of references used for the definitions and places where you can find more information.

Glossary of Terms

English – French

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

Term	Definition	Translation
A-B-C analysis (n)	An approach to understanding behavior by examining the Antecedent (the cause), the Behavior, and the Consequence (the result).[1]	Analyse ABC
Abstract language (n)	Any vocabulary or phrases with meanings that are not clearly stated. Examples include idioms like, "it's raining cats and dogs" or "break a leg" where the intended meaning does not match the literal meaning. There are also many examples of this in books and poetry where the author may describe something using abstract language like "it felt like a weight on my chest."[5]	Langage abstrait
Adaptive behavior or adaptive skills (n)	Includes communication, self-care, home living, social skills, community use, self-direction, health and safety, functional academics, leisure, and work. These are skills that help the person be successful in their environment and are learned skills rather than innate abilities.[2]	Comportement adaptatif ou capacités d'adaptation
Age of majority (n)	The age established under state law when an individual is no longer a minor and has the right to make certain legal decisions without consent.[1]	Âge de la majorité
Anxiety disorder (n)	A pattern of constant worry or tension under many different circumstances.[1]	Trouble anxieux
Applied Behavior Analysis (ABA) (n)	ABA is the name of a professional field that uses principles of learning to increase performance of socially desirable behaviors. ABA practitioners carefully observe and measure behaviors and the context in which they occur in order to individualize teaching plans to improve specific behaviors. ABA is commonly used as one component in interventions for Autism Spectrum Disorders.[2]	Analyse comportementale appliquée (ACA)
Apraxia (n)	See "Dyspraxia."[2]	Apraxie

Term	Definition	Translation
Attention Deficit Hyperactivity Disorder (ADHD) (n)	A disorder that shows up in the areas of inattention, hyperactivity, and impulsiveness. It is evidenced by frequent shifting from one activity or focus to another, having difficulty organizing and completing tasks correctly, impulsive response, or failure to follow rules. It may occur with or without hyperactivity which includes behaviors such as excessive running, talking, fidgeting, and/or restlessness.[2]	Trouble de déficit de l'attention avec hyperactivité (TDAH)
Audiologist (n)	A specialist who evaluates for hearing loss as a potential cause or contributor to developmental delay, and designs/supports interventions to minimize the impact of hearing loss when it is found.[2]	Audiologiste
Augmentative and Alternative Communication (AAC) (n)	Any method of communicating without speech, such as by signs, gestures, picture boards, or electronic or non-electronic devices. These methods can help individuals who are unable to use speech or who need to supplement their speech to communicate effectively.[2]	Communication améliorée et alternative (CAA)
Autism Spectrum Disorder (ASD) (n)	A neurodevelopmental disorder. ASD symptoms are typically evident before a child is 3 years of age. The symptoms range from mild to severe – and individuals often have varied skills levels in different domains of functioning. Autism affects the person's overall development in 2 primary areas: 1. *social communication*, or the way a person uses gestures, body language, and language to communicate and interact socially and relate with others. 2. the presence of *restricted, repetitive patterns of behavior, interests, or activities* (e.g., repetitive motor movements, echolalia/repeating speech, idiosyncratic phrases, extreme distress at small changes/difficulty with transitions, strong attachment to unusual objects/topics, adverse or intense responses to sensory input).[2]	Trouble du spectre de l'autisme (TSA)
Aversive (adj)	An unwanted stimulus designed to change an individual's behavior through punishment. These should rarely be used in the treatment of autism.[1]	Aversif
Behavior Intervention Plan (BIP) (n)	A plan to improve an individual's behavior, created based on the results of a Functional Behavior Assessment.[1]	Plan d'intervention en matière de comportement (PIC)
Behavioral disorder (n)	A condition in which behavior significantly deviates from acceptable norms.[1]	Trouble du comportement
Biomarker (n)	An indicator of a certain biological state.[1]	Marqueur biologique
Bipolar disorder (n)	A brain disorder that causes unusual shifts in mood, energy, activity levels, and the ability to carry out day-to-day tasks; also known as manic-depressive illness.[1]	Trouble bipolaire

Term	Definition	Translation
Board Certified Behavior Analyst (BCBA) (n)	A professional certified to provide ABA therapy by the Behavior Analyst Certification Board (BACB).[1]	Analyste du comportement certifié par le conseil
Body language (n)	A form of non-verbal communication that is an important part of social communication. It includes things like gestures (head nodding or shaking), personal space, facial expressions, and eye contact.[5]	Langage corporel
Case manager (n)	A professional from a school or service agency such as the Department of Developmental Disabilities who serves as a direct contact for families and helps gather resources, team members and ideas.[1]	Gestionnaire de soins
Catatonia (n)	A state in which a person does not move and does not respond to others.[1]	Catatonie
Challenging behaviors (n)	Behaviors that are destructive and harmful to the individual or others (e.g., hitting, biting, running away), that prevent learning and cause others to label or isolate the individual for being odd or different.[1]	Comportements problématiques
Civil commitment (n)	A legal process in which an individual experiencing a mental health crisis is ordered into treatment against his or her will, including to a hospital.[1]	Procédure civile d'internement
Cognitive behavioral therapy (n)	Type of therapy designed to help improve an individual's inappropriate or challenging behaviors by replacing the negative thoughts that cause these behaviors with positive thoughts.[1]	Thérapie cognitive du comportement
Cognitive development (n)	How children learn to think, interpret information, make decisions, and solve problems. Areas of cognitive development include verbal reasoning (using language), non-verbal reasoning (using visual/spatial information), as well as executive control/functioning.[2]	Développement cognitif
Communication (n)	The developmental area that involves skills which enable people to understand (receptive communication) and share (expressive communication) thoughts and feelings. Waving goodbye, smiling, nodding, making eye-contact, using spoken language, following directions, and reading and writing are examples of communication.[2]	Communication
Communication disorder (n)	Difficulty with understanding and/or expressing messages.[2] Communication disorders include problems with hearing, with making speech sounds (articulation), with having a clear voice (voice disorders), stuttering (fluency disorders), difficulty learning, knowing, and using grammar (language disorders), difficulty using language to get things done (social communication or pragmatic language disorders), and using language to learn (language-based learning disabilities such as dyslexia).[2]	Trouble de la communication
Comorbid (adj)	Pertaining to a disease or disorder that occurs simultaneously with another.[1]	Comorbide

Term	Definition	Translation
Compulsion (n)	The drive to do something in particular or in a particular way, such as the need to straighten all the forks at the dinner table.[1]	Compulsion
Conservatorship (n)	The legal right given to a person to be responsible for the assets and finances of a person deemed fully or partially incapable of providing these necessities for himself or herself.[1]	Tutelle
Crisis plan (n)	A document that outlines in specific detail the necessary strategies and steps that must be taken when a crisis occurs.[1]	Plan de crise
Data analysis (n)	The process of thoroughly inspecting information related to challenging behaviors in order to draw out useful information and conclusions that may result in strategies to improve behavior.[1]	Analyse de données
De-escalation (n)	The process of stopping a challenging behavior or crisis from intensifying and calming the situation.[1]	Désescalade
Depression (n)	A mood disorder in which feelings of sadness, anger, or frustration interfere with everyday life for an extended period of time.[1]	Dépression
Developmental assessment (n)	A structured evaluation of a child's development in one or more areas including cognitive, language, motor, social/emotional, and adaptive. Professionals that conduct developmental assessments include developmental behavioral pediatricians, psychologists, speech language pathologists, occupational therapists, physical therapists, educators, and audiologists. Types of tests that are used during developmental assessments include interview, observation, questionnaires, standardized tests, and non-standardized tests.[2]	Évaluation du développement
Developmental delay (n)	The term used to describe the condition of an infant or young child who is not achieving new skills in the typical time frame and/or is exhibiting behaviors that are not appropriate for his or her age. Some children who are developmentally delayed eventually have a specific diagnosis of a particular developmental disability. Other children with delays catch up with their typically developing peers.[2]	Retard du développement
Developmental Disability (DD) (n)	A severe chronic disability that is attributed to a physical or mental impairment, other than the sole diagnosis of mental illness, or to a combination of mental and physical impairments, is manifested before the individual attains the age of 22, is likely to continue indefinitely, results in the inability to live independently without external support or continuing and regular assistance, reflects the need for a combination and sequence of special, interdisciplinary, or generic care, treatment, or other services that are planned and coordinated for that individual.[2]	Déficience développementale (DD)

Term	Definition	Translation
Developmental pediatrician (n)	Also known as a developmental-behavioral pediatrician, these professionals have training and experience to assess and treat a wide range of medical and psychosocial aspects of a child's developmental and behavioral difficulties. Their expertise may make them a good choice for children with complicated medical or developmental problems.[2]	Pédiatre du développement
Differential diagnosis (n)	Distinguishing between two or more diseases with similar symptoms to identify which is causing distress or challenging behavior.[1]	Diagnostic différentiel
Disruption (n)	An event that causes an unplanned deviation from a situation.[1]	Perturbation
Down syndrome, also known as trisomy 21 (n)	The most common and readily identifiable chromosomal condition associated with intellectual disability. Children with Down syndrome typically have developmental delays, but this can range from mild to severe. Common physical signs of Down syndrome include decreased muscle tone; short neck; flattened facial profile and nose; small head, ears, and mouth; upward slanting eyes; white spots on the colored part of the eye (called Brushfield spots); wide, short hands with short fingers; a single, deep, crease across the palm of the hand; a deep groove between the first and second toes.[2]	Syndrome de Down aussi connu sous le nom de trisomie 21
Dual diagnosis (n)	The identification of an additional mental health disorder individuals with developmental disabilities. [1]	Diagnostic mixte
Dysarthria (n)	A term used to describe the impact on speech production of muscle weakness and/or reduced muscle control due to neural damage.[2]	Dysarthrie
Dyspraxia (n)	A condition characterized by a difficulty with planning and performing coordinated movements although there is no apparent damage to muscles. Dyspraxia can impact any motor system and will be described based on the motor system affected (e.g., upper limb dyspraxia, speech/verbal dyspraxia). The term "apraxia" is often used as a synonym for dyspraxia.[2]	Dyspraxie
Early intervention (n)	Programs or services designed to meet the developmental needs of infants and toddlers (birth to three years old) and their families.	Intervention précoce
Echolalia or Echophrasia (n)	The immediate and involuntary repetition of words or phrases just spoken by others, often a symptom of autism or some types of schizophrenia. Also called echophrasia.[3]	Écholalie ou échophrasie
Elopement (n)	A situation in which an individual leaves a safe place, a caretaker, or supervised situation, either by "bolting," wandering or sneaking away.[1]	Fugue
Epilepsy (n)	A brain disorder in which a person has repeated seizures (episodes of disturbed brain activity or convulsions) over time.[1]	Épilepsie
Escalating (v)	Increasing or worsening rapidly.[1]	Aggraver

Term	Definition	Translation
Evidence-Based Practice (EBP) (n)	A teaching strategy or practice that has been proven through research studies to improve skills or behaviors for a certain population.[5]	Pratique fondée sur des preuves
Executive control / executive functioning (n)	A group of skills that helps people plan, organize, control behavior, focus on multiple streams of information at the same time, self-monitor, and revise action plans as necessary. Acquiring these skills is one of the most important tasks of the early childhood years.[2]	Contrôle exécutif/fonctionnement exécutif
Extinction (n)	A response used to eliminate a behavior that involves ignoring a mild behavior when it is used for attention.[1]	Extinction
Extinction burst (n)	Short term response to extinction in which there is a sudden and temporary increase in the response's frequency, followed by an eventual decline.[1]	Inversement de l'extinction
Face blindness (n)	An impairment in the recognition of faces.[1]	Agnosie des visages
Fading (v)	Gradually reducing the number of prompts or types of prompts to encourage more independence for the learner.[5]	Désétayage
Fecal digging (v)	The process in which an individual puts his fingers into his rectum.[1]	Fouille rectale
Fecal smearing (v)	The process in which feces are spread on property or the individual himself.[1]	Frottis fécal (Scatolie)
Fetal Alcohol Spectrum Disorder (FASD) (n)	Children whose mothers drank during pregnancy, and who have developmental impairment may be diagnosed with one of several FASDs. The developing brain is the organ most vulnerable to prenatal alcohol exposure, but the range of impact of prenatal alcohol exposure includes both physical and neurodevelopmental impairment. These can include growth difficulty, minor facial anomalies, general developmental delays, seizures, learning disabilities, ADHD, executive functioning and processing challenges, as well as mental health or behavioral challenges. The most severe FASD is Fetal Alcohol Syndrome (FAS – see FAS), but other FASDs are far more common.[2]	Ensemble des troubles causés par l'alcoolisation fœtale (ETCAF)
Fetal Alcohol Syndrome (FAS) (n)	A permanent birth defect syndrome caused by maternal drinking during pregnancy. FAS is characterized by growth deficiency; a cluster of 3 minor facial abnormalities including a thin upper lip, a smooth philtrum (i.e., the groove between nose and upper lip); small eyes; and significant abnormalities in brain development. FAS is the leading preventable cause of intellectual disability and other neurodevelopmental conditions.[2]	Syndrome d'alcoolisme fœtal (SAF)
Function of behavior (n)	The purpose or reason behind a specific behavior for an individual.[1]	Fonction du comportement

Term	Definition	Translation
Functional Behavior Analysis (FBA) (n)	The process of systematically determining the function of behaviors, usually inappropriate, that are displayed by people. Behaviors are defined, measured, and analyzed in terms of what happened before and after their occurrence. Over time the events before and after the behavior occurs are systematically changed in order to determine the function of the behavior for the person displaying it.[2]	Analyse fonctionnelle du comportement (AFC)
Functional communication (n)	Effective and appropriate communication that an individual uses across his daily activities to meet his or her needs.[1] Can be verbal or non-verbal.	Communication fonctionnelle
Generalization (n)	The ability to use a target skill or behavior across the intervention timespan or timeframe, setting, and individuals (e.g., teachers, peers, parents). For example, when teaching a child to greet others, a teacher may initially implement the intervention when the student first enters classroom. Although the child may begin to independently greet others in the context of entering the classroom, she may not be able to do so when coming home from or entering a different room in the school. If she can greet others across settings and individuals, then she has engaged in generalization of the skill.[5]	Généralisation
Gestures (n)	Body and hand movements used to communicate. Examples include pointing, waving, opening arms to demonstrate something is “big,” banging a fist on a table to emphasize a point or show anger, etc.[5]	Gestes
Guardianship (n)	The legal right given to a person to be responsible for the food, health care, housing, and other necessities of a person deemed fully or partially incapable of providing these necessities for himself or herself.[1]	Tutelle
Hypersensitivity (n)	Overly intense or exaggerated response to sensation. It may include defensive responses (like covering ears in anticipation of a sound, pulling away quickly when touched lightly) or disliking or avoiding of certain situations that involve an undesirable sensory experience (like avoiding public restrooms because the toilet flushing is aversive).[5]	Hypersensibilité
Hyposensitivity (n)	Under exaggerated response to sensation. May include lack of response entirely or a reduced or slower response than expected. A child with hyposensitivity may not stop working and turn around when their name is called, may not seem to notice that another child ran into them, may smile, or laugh several seconds after you tickle them, may cry several seconds after getting hurt or not at all, etc.[5]	Hyposensibilité

Term	Definition	Translation
Idiosyncratic language/ vocabulary (n)	Using language in unusual ways that may not be obvious to the communication partner. This includes using scripts from previous conversations or media applied to different contexts like saying "C is for cookie" to express that they like something. Or "Dora loves waterfall!" to request to take a bath/shower.[5]	Langage/vocabulaire idiosyncratique
Incontinence (n)	The (usually) involuntary passing of feces or urine, generally not into a toilet or diaper.[1]	Incontinence
Individualized Education Program (IEP) (n)	A written statement of a child's current level of development (abilities and impairments) and an individualized plan of instruction, including the goals, the specific services to be received, the people who will carry out the services, the standards, and timelines for evaluating progress, and the amount and degree to which the child will participate with non-handicapped peers at school. The IEP is developed by the child's parents and the professionals who evaluated the child. It is required by the Individuals with Disabilities Education Act (IDEA) for all children in special education, ages three years and up.[2]	Programme éducatif individualisé (PEI)
Individualized Family Service Plan (IFSP) (n)	A written plan describing the infant's or toddler's current level of development, the family's strengths and needs related to enhancement of the infant's or toddler's development, goals for the child and the other family members (as applicable), including the criteria, procedures and time lines used to evaluate progress (the IFSP should be evaluated and adjusted at least once a year and reviewed at least every six months), and the specific early intervention services needed to meet the goals (including the frequency and intensity and method of delivering services, the projected date of initiating services and the anticipated duration of services). The IFSP is developed and implemented by the child's parents and a multidisciplinary early intervention team (IFSP Team). The Individualized Family Service Plan is required by the Individuals with Disabilities Education Act (IDEA) for all infants and toddlers receiving early intervention services.[2]	Plan de service familial individualisé
Individuals with Disabilities Education Act (IDEA) (n)	The federal law reauthorized in 2004 that amends the Education for All Handicapped Children Act (Public Law 94-142). Part C of the law focuses on services to infants and toddlers who are at-risk or have developmental disabilities.[2]	Individuals with Disabilities Education Act (IDEA) (loi sur l'éducation des personnes handicapées)
Intellectual disability (n)	Characterized by significantly impaired intellectual functioning, existing concurrently with related limitations in two or more of the following applicable adaptive skill areas: communication, self-care, home living, social skills, community use, self-direction, health and safety, functional academics, leisure, and work.[2]	Handicap intellectuel / Déficience intellectuelle

Term	Definition	Translation
Interdisciplinary evaluation team (n)	A group of health care professionals from diverse disciplines who form a team to work collaboratively in conducting a cohesive patient evaluation.[2]	Équipe d'évaluation interdisciplinaire
Intervention (n)	A strategy or process put in place in order to improve or modify an individual's behavior e.g., medication, Applied Behavior Analysis).[1]	Intervention
Involuntary commitment (n)	A legal process in which an individual experiencing a mental health crisis is ordered into treatment against his or her will, including to a hospital.[1]	Internement forcé
Joint attention (n)	A set of early social communication skills used to show and/or share interest about an outside object or event with a communication partner. They include initiating joint attention where the child shifts their gaze and/or gestures toward an object with the goal of getting another person to notice it. They also include responding to gaze shifts and gestures from other people so that they can learn what others find interesting.[5]	Attention conjointe
Language delay (n)	A disorder in which a learner's ability to understand and/or use language is behind what would be expected based on their age.[5]	Retard du langage
Language disorder (n)	In children, this could mean trouble getting their meaning or message across to others (expressive language disorder), or understanding messages coming from others (receptive language disorder), as well as difficulty using language to get things done (pragmatic language/social communication disorder).[2]	Trouble du langage
Learning disability (n)	A disorder that affects how a person learns and understands primary skills such as reading, writing and math.[2]	Trouble d'apprentissage
Least Restrictive Environment (LRE) (n)	The educational setting that permits a child with disabilities to derive the most educational benefit while participating in a regular educational environment to the maximum extent appropriate. It is presumed that a child with a disability will be educated in the general education classroom, with appropriate supports, unless the IEP Team deems another setting as more appropriate. LRE is a requirement under the IDEA.[2]	Milieu le moins restrictif possible
Maladaptive behavior (n)	A type of behavior that is often used to reduce anxiety, but whose result does not provide adequate or appropriate adjustment to the environment or situation.[1]	Comportement inadapté
Medicaid (n)	A government program that provides healthcare coverage for low-income families and individuals with disabilities in the United States.[1]	Medicaid
Medical home (n)	A team-based healthcare delivery model led by a physician that provides comprehensive and continuous medical care to patients.[1]	Soins à domicile

Term	Definition	Translation
Mental health hold (n)	Involuntary hospitalization due to a mental health crisis.[1]	Internement pour raison de santé mentale
Motor skill (n)	The learned ability to perform movements, such as holding the body upright to sit, using the hands to manipulate small items, scooping food onto a spoon and bringing it to the mouth, and moving the lips and tongue to articulate different sounds. Fine motor skills involve use of the small muscles of the body while gross motor skills are associated with large muscle movements.[2]	Habileté motrice
Neurodevelopmental conditions (n)	Problems affecting the development of the central nervous system or brain, resulting in delayed or unusual motor, speech, social or learning deficits.	Affections neurodéveloppementales
Nonverbal communication (n)	Any form of or attempt at unspoken or "physical" communication. Examples are temper tantrums, gestures, pointing and leading another person to a desired object.[2]	Communication non verbale
Nutritionist (n)	Nutrition services are provided by Registered Dietitian Nutritionists (RDNs). The nutritionist evaluates the child's growth, energy intake, and nutritional status to make sure they are getting appropriate nutrients from their diet. A nutritionist often will work with a feeding therapist (e.g., speech or occupational therapist), to develop a plan when feeding problems are suspected.[2]	Nutritionniste
Obsession (n)	A repetitive thought or feeling dominated by a particular idea, image, or desire, such as a person who only wants to talk about elevators.[1]	Obsession
Obsessive Compulsive Disorder (OCD) (n)	An anxiety disorder in which people have unwanted and repeated thoughts, feelings, ideas, or sensations (obsessions) that make them feel driven to do something (compulsions).[1]	Trouble obsessionnel compulsif (TOC)
Occupational Therapist (OT) (n)	A skilled healthcare provider that assists people across the lifespan participate in the things they want and need to do through the therapeutic use of everyday activities (occupations). Occupational therapy benefits individuals who have physical, developmental, or cognitive limitations. With children, treatment is geared toward development of fine motor skills (e.g., writing, cutting), gross motor skills (e.g., climbing stairs, jumping), self-care (e.g., dressing, eating), and play. OT is the primary profession that addresses sensory processing differences.[2]	Ergothérapeute
Ototoxic (n)	Damaging to the ears, causing sound sensitivities, dizziness, or balance issues.[1]	Ototoxique
Overcorrection (n)	A punishment mechanism for a challenging behavior that involves requiring an individual to engage in repetitive behavior to an excessive extent in an attempt to prevent the behavior from reoccurring.[1]	Correction excessive

Term	Definition	Translation
Pediatric Autoimmune Neuropsychiatric Disorders Associated with Streptococcal infections (PANDAS) (n)	A subset of children and adolescents who have Obsessive Compulsive Disorder (OCD) and/or tic disorders, and in whom symptoms worsen following infections such as strep throat and scarlet fever.[1]	Troubles neuropsychiatriques pédiatriques auto-immuns associés aux infections streptococciques (PANDAS)
Phenylketonuria (PKU) (n)	An inherited disorder that increases the levels of a substance called phenylalanine in the blood. Phenylalanine is found in food proteins and in some artificial sweeteners. If PKU is not treated, phenylalanine can build up to harmful levels in the body, causing intellectual disability and other serious health problems such as seizures, delayed development, behavioral problems, and psychiatric disorders.[2]	Phénylcétonurie (PCU)
Physical Therapist (PT) (n)	A healthcare team member who treats conditions that limit the ability to move and perform functional activities of daily life. Physical therapists provide services to improve body structures (e.g., muscles), enhance functional activities (e.g., walking), and promote improved participation in daily life. Goals for children may include developing improved strength, range of motion, coordination, balance, and acquisition of new motor skills.[2]	Physiothérapeute (PHT)
Pica (n)	An eating disorder that involves eating things that are not food (e.g., dirt, plastic).[1]	Pica
Picture Exchange Communication Systems (PECS) (n)	A unique augmentative/alternative communication intervention package that involves teaching an individual to give a picture of a desired item to a "communicative partner," and goes on to teach discrimination of pictures and how to put them together in sentences.[1]	Systèmes de communication par échange d'images (PECS)
Polypharmacy (n)	The use of multiple medications by a patient.[1]	Polypharmacie
Positive Behavior Supports (PBS) (n)	An approach to helping people improve their difficult behavior by understanding what is causing it, and then developing strategies to increase positive behaviors.[1]	Soutien comportemental positif (SCP)
Post-Traumatic Stress Disorder (PTSD) (n)	An anxiety disorder that can occur after witnessing or experiencing a traumatic event.[1]	Trouble de stress post-traumatique (TSPT)
Prematurity (n)	A premature birth is a birth that takes place more than three weeks before a baby is due. Normally, a pregnancy lasts about 40 weeks, so a premature birth is one occurring before the start of the 37th week of pregnancy.[2]	Prématurité
Prompting (v)	Any help given to a learner to assist in using a specific skill or behavior. Prompts can come in many different forms including visual, verbal, gestural, models, and partial or full physical.[5]	Assister/guider/inciter

Term	Definition	Translation
Psychologist (n)	Someone with a PhD or PsyD, trained in the evaluation, study and/or treatment of psychiatric and cognitive disorders. A school psychologist is a professional trained in psychology and education who collaborates with children, educators, parents, and other professionals to create healthy and supportive learning environments for students based on careful evaluation of cognitive, executive, and adaptive skills.[2]	Psychologue
Psychosis (n)	A loss of contact with reality that usually includes delusions and hallucinations.[1]	Psychose
Psychotropic (adj)	A medication or intervention that affects brain activity, behavior, or perception.[1]	Psychotrope
Puberty (n)	The process of physical changes that occur when a child's body matures into an adult.[1]	Puberté
Red flags (n)	Behaviors that cause caregivers concern in an area of a child's development. They warn you to stop, look, and think, and then observe and document.[2]	Signaux d'alarme
Reinforce (v)	To strengthen with additional material or support.[1]	Renforcer
Reinforcement strategies (n)	Methods used to promote or increase positive behavior by providing motivating reinforcers, such as praise, a favorite toy, a cookie, or a preferred activity. Also called a "reward" or an "incentive."	Stratégies de renforcement
Resilience (n)	An ability to recover from or adjust easily to change or a difficult situation.[1]	Résilience
Respite care (n)	A service that provides short-term breaks that can relieve stress, restore energy, and promote balance for caregivers.[1]	Soins de répit
Restraints (n)	Physical restrictions immobilizing or reducing the ability of an individual to move their arms, legs, body, or head freely. This can be someone holding a person so they cannot move, or it can be an item that restrains their body such as a device that holds their hands or bodies down.[1]	Attaches
Restricted or repetitive behaviors or interests (n)	Topics or tangible items that individuals with autism pursue with great intensity and focus for long durations of time.[4]	Comportements ou intérêts restreints ou répétitifs
Reward (n)	A prize, token, or preferred activity given to an individual for good behavior, designed to promote the same behavior in the future.[1]	Récompense
Risk factor (n)	Conditions that increase the likelihood of aggression.[1]	Facteur de risque
Ritual (n)	A repetitive behavior that a person appears to use in a systematic way in order to promote calm or prevent anxiety, such as arranging all the pillows in a certain way before being able to settle in to sleep.[1]	Rituel
Rumination (n)	The practice of (voluntarily or involuntarily) spitting up partially digested food and re-chewing it, then swallowing again or spitting it out. Rumination often seems to be triggered by reflux or other gastrointestinal concerns.[1]	Rumination

Term	Definition	Translation
Schizophrenia (n)	A chronic, severe, and disabling brain disorder that makes it hard for individuals to think clearly and tell the difference between what is real and not real.[1]	Schizophrénie
Screening test or tool (n)	An evaluation tool to identify children who are at-risk for having or developing a developmental disability. This is different from a diagnostic tool that is used to determine if a person has, or does not have, a neurodevelopmental disability.[2]	Test ou outil de dépistage
Seclusion (n)	A situation in which an individual is isolated in a room in response to a behavior they have exhibited.[1]	Isolement
Sedating (v)	Calming, sleep-inducing, or numbing an individual experiencing challenging behaviors or struggling during difficult situations.[1]	Administrer un sédatif
Seizure disorder (n)	A seizure disorder, which can also be called epilepsy, is a disorder in which brain activity is disturbed and causes seizures. There are many types of seizures. Some involve uncontrollable jerking movements or losing awareness, but some can just look like a staring spell.[5]	Trouble convulsif/épileptique
Self-advocacy (n)	The ability of an individual to communicate his or her wants and concerns and make his or her own decisions.[1]	Autonomie sociale
Sensory avoidance (n)	Blocking or staying away from something that is painful or bothersome.[1]	Évitement sensoriel
Sensory defensiveness (n)	A tendency to react negatively or with alarm to sensory input which is generally considered harmless or non-irritating.[1]	Défense sensorielle
Sensory input (n)	Any source that creates sensation and activates one or more of the senses -vision, smell, sound, taste, and touch.[1]	Indices sensoriels
Sensory processing disorder (n)	Refers to difficulty detecting, organizing, or responding to sensory information received and interpreted in the brain via all seven senses and that interferes with participation in daily life, development, behavior, and social interactions.[2]	Trouble du traitement sensoriel
Sensory-seeking behavior (n)	Behaviors caused by a need for additional stimulation of certain senses as a way of maintaining attention or achieving a calmer state.[1]	Comportement de recherche de sensation
Sleep apnea (n)	A usually chronic, common disorder in which an individual has one or more pauses in breathing or shallow breaths up to 30 or more times per hour during sleep, and results in daytime sleepiness.[1]	Apnée du sommeil
Sleep disturbances (n)	Sleep disturbances can include a variety of issues including difficulties falling asleep or staying asleep, being on different sleep rhythms (e.g., awake at night), or needing increased amounts of sleep.[5]	Troubles du sommeil

Term	Definition	Translation
Social communication disorder (n)	Applies to children who have deficits in the social use of language, but do not have the restricted interests or repetitive behavior commonly found in autism spectrum disorders.[2]	Trouble de la communication sociale
Social reciprocity (n)	The primary component of healthy social development – acts of kindness – interactions. It is about "joint attention" where there is purposeful language including body language and pretend play. Lack of social reciprocity is a key red flag, whether the child is not engaged in the world around him. This concern should not be ignored.[2]	Réciprocité sociale
Social work or social worker (n)	Social work practice is aimed at assisting individuals, groups, or communities to enhance or restore their capacity for social functioning and creating societal conditions favorable to reach their goals. The primary mission of the social work profession is to enhance human well-being and help meet the basic human needs of all people, with particular attention to the needs and empowerment of people who are vulnerable, oppressed, and living in poverty.[2]	Assistance sociale ou assistant(e) social(e)
Special Needs Parent Advocate (n)	An advocate for parents of children with special needs who helps ensures that the child's rights and needs are met in school and in the community.[1]	Porte-parole de parents d'enfants ayant des besoins éducatifs spécifiques
Specific learning disability (n)	A disorder that manifests itself with a deficit in areas such as attention, reasoning, processing, memory, communication, reading, writing, spelling, calculation, coordination, social competence, and emotional maturity. Often identified by a discrepancy between school performance and the performance expected based on overall intelligence.[2]	Trouble d'apprentissage particulier
Speech generating device or Voice output technology (n)	A technological device that helps people who are unable to use speech to express their needs and exchange information with other people.[1]	Système de synthèse vocale ou technologie de reproduction vocale
Speech sound disorder (n)	Speech disorders in which some speech sounds in a child's native language are not produced, not produced correctly, or are not used correctly.[2]	Trouble de la phonation
Speech-language pathologist (n)	A clinician who assesses, diagnoses, treats, and helps prevent speech, language, cognitive, communication, voice, swallowing, fluency, and other related disorders.[2]	Orthophoniste
Standardized test (n)	A test administered and scored in a consistent or standard manner. It is administered in controlled conditions that specify where, when, how, and for how long children respond to the questions. In standardized tests, the questions, conditions for administering, scoring procedures, and interpretations are consistent. A well designed standardized test provides an assessment of an individual's mastery of a domain of knowledge or skill.[2]	Test standardisé

Term	Definition	Translation
Staring spells (n)	Occasions when an individual is in a trance staring into space, which can often signal seizure activity.[1]	Épisodes de regard fixe
Stereotypy (n)	Repetitive or ritualistic movements such as body rocking or crossing and uncrossing of legs.[1]	Stéréotypie
Stimulation (n)	Excitement or activity triggered by a stimulus either internally or externally.[1]	Stimulation
Supplemental Security Income (SSI) (n)	A Federal income supplement program designed to help aged, blind, and disabled people who have little or no income, and provides cash to meet basic needs for food, clothing, and shelter.[1]	Supplemental Security Income (SSI) (Allocation supplémentaire de revenu de sécurité sociale)
Tangibles (n)	Items or rewards that can be touched, such as a toy or piece of candy.[1]	Éléments matériels
Time delay (n)	A prompting procedure that fades prompts during activities by having longer delays before a prompt is provided.	Délai de temporisation
Tourette syndrome or Tourette's syndrome (n)	A neurological disorder characterized by tics, or repetitive, stereotyped, involuntary movements and vocalizations.[1]	Syndrome Gilles de la Tourette
Tracking scales (n)	A document or other tool used to track information such as changes in an individual's behaviors, side effects of medications, school performance, etc.[1]	Échelles de suivi
Transition plan / transition services (n)	Part of IDEA, transition services means a coordinated set of activities for a child with a disability that is results-oriented and focused on improving the academic and functional achievement of the child with a disability in order to facilitate the child's movement from school to post-school activities, including postsecondary education, vocational education, integrated employment (including supported employment), continuing and adult education, adult services, independent living, or community participation. Transition services will be based on the individual child's needs, taking into account the child's strengths, preferences, and interests.[2]	Plan de transition/services de transition
TRICARE (n)	The health care program for Uniformed Service members, retirees, and their families worldwide.[1]	TRICARE (assurance médicale pour les militaires, leurs familles et retraités)
Visual cues (n)	An image or symbol presented to the learner that either prompts him to complete the target behavior/skill or assists in supporting the understanding of a particular activity, instruction, or direction. Visual cues can be used as prompts or supports.[5]	Indices visuels
Visual schedules (n)	A display that shows the activities of a day or steps of an activity to support the learner in moving between activities or through activities. The displays can be made from objects, pictures, photos, or written text, and may be created for a full day, part of a day, or just the next two activities.[5]	Programme visuel

Term	Definition	Translation
Vocational rehabilitation (n)	A state and federally funded program designed to help people with disabilities become employed and to help those already employed perform more successfully through training, counseling, and other support methods.	Réinsertion professionnelle
Voice output technology or Voice output communication aid or Speech generating device (n)	A technological device that helps people who are unable to use speech to express their needs and exchange information with other people.[1]	Technologie de reproduction vocale ou communication de reproduction vocale ou système de synthèse vocale
Wraparound (n)	An integrated, multi-agency, community-based planning process designed to build teams of providers, family members and natural supports to help keep complex youth in their homes and communities.[1]	Processus Wraparound

References

[1] The definition for these terms were taken from "Challenging Behaviors Glossary" published in 2012 by Autism Speaks. This glossary is part of a downloadable toolkit available at: https://www.autismspeaks.org/tool-kit/challenging-behaviors-tool-kit

[2] The definitions for these terms were taken from "Glossary of Terms Related to Neurodevelopmental Disabilities" by John Thorne, PhD, CCC-SLP. The "Glossary of Terms Related to Neurodevelopmental Disabilities" was adapted by John Thorne, PhD, CCC-SLP, from a number of in-depth resources (12/16) including:
http://www.inclusivechildcare.org/inclusion_glossary.cfm
http://www.ddrcco.com/resources-and-training/glossary-of-developmental-disability-terms.php
http://autismnow.org/at-home/learn-and-understand-autism/autism-and-developmental-disabilities-glossary/
http://www.gennextmsp.org/wp-content/uploads/2015/05/ECS-Glossary-May2015.pdf
http://www.ncca.biz/Aistear/pdfs/PrinciplesThemes_ENG/Glossary_ENG.pdf
http://www.johnson-center.org/downloads/pdfs/What_is_a_Standardized_Test.pdf

[3] The definition was taken from The American Heritage® Stedman's Medical Dictionary. Copyright ©2002,2001, 1995 by Houghton Mifflin Company. Published by Houghton Mifflin Company.

[4] DSM-IV, 1994.

[5] Excerpted from Steinbrenner, J., Sam, A., Chin, J., Morgan, W., & AFIRM for Paras Team. (2019). *Introduction to ASD.* FPG Child Development Institute, University of North Carolina. Retrieved from https://afirm.fpg.unc.edu/introduction-asd

More information is available at:

- Autism Society
  Founded in 1965 by Dr. Bernard Rimland, Dr. Ruth Sullivan and many other parents of children with autism, the Autism Society is the leading source of trusted and reliable information about autism. Through its national network, the Autism Society has spearheaded numerous pieces of federal, state and local legislation, including the 2006 Combating Autism Act, the first federal autism-specific law. The Autism Society's website is one of the most visited websites on autism in the world and our on-line resource database, Autism Source™, and National Contact Center (800-3-AUTISM) provide information and service referrals to thousands of people each year. Look online at https://www.autism-society.org/.

- Autism Speaks
  Autism Speaks was founded in February 2005 by Bob and Suzanne Wright, grandparents of a child with autism. Recognizing the need for a powerful voice Bernie Marcus donated $25 million to help financially launch the organization.

Building upon the legacy of three leading autism organizations, Autism Coalition for Research and Education (ACRE), the National Alliance for Autism Research (NAAR) and Cure Autism Now (CAN), who merged with the organization, Autism Speaks has made extraordinary advancements in the autism community.

Chief among these are increased global awareness of autism, better understanding of the breadth of the autism spectrum, and advocacy to increase research and access to care and support.

Today, Autism Speaks is dedicated to advancing research into causes and better treatments for autism spectrum disorders and related conditions both through direct funding and collaboration.

Look online at https://www.autismspeaks.org/.

- Centers for Disease Control and Prevention (CDC)
  CDC works 24/7 to protect America from health, safety, and security threats, both foreign and in the U.S. Whether diseases start at home or abroad, are chronic or acute, curable or preventable, human error or deliberate attack, CDC fights disease and supports communities and citizens to do the same.

  CDC increases the health security of our nation. As the nation's health protection agency, CDC saves lives and protects people from health threats. To accomplish our mission, CDC conducts critical science and provides health information that protects our nation against expensive and dangerous health threats, and responds when these arise.

  Look online at:
  https://search.cdc.gov/search/index.html?query=autism%20spectrum%20disorder

National Coordinating Center
for the Regional Genetics Networks

The National Coordinating Center for the Regional Genetics Networks

Glossary of Cancer Genetics Terms

English – French

2023 Edition

Compiled and edited by:
Cynthia E. Roat, MPH

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

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 cancer genetics is provided to help enable access to cancer genetics 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 cancer genetics terms is a page with Examples of Genes That are Often Checked in Genetic Testing, and 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. There is also a page on Common Interpreting Errors, and a page on Analogies. Genetic counselors often use analogies to explain complex ideas in genetics.

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
Benign (adj)	Something mild that does not threaten health or life. In cancer genetics, "benign" means "not cancerous."	Bénin(e)
Blood test (n)	A test in which blood is drawn (usually from the patient's arm) and sent to a laboratory for analysis. In cancer genetics, blood tests provide cells for genetic sequencing.	Analyse sanguine
Breast cancer (n)	Cancer of the breast tissue. This cancer is more common in women, but it can affect men as well.	Cancer du sein
Buccal swab (n)	A way to collect DNA from the cells on the inside of a person's cheek.	Frottis buccal
CA-125 blood test (n)	A blood test used to look for early signs of ovarian cancer in women with a high cancer risk.	Analyse sanguine du CA-125
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)	A small (microscopic) structure that forms the basic building block of every known living organism.	Cellule
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 proteins and a single molecule of DNA, which carries genetic information.	Chromosome
Colonoscopy (n)	A diagnostic test in which a flexible tube with a tiny camera on the end is inserted into the colon (large intestine) through the rectum in order to see the condition of the inside of the colon, or to take a small sample of tissue, or to cut out polyps (small growths).	Coloscopie
Condition (e.g., "genetic condition") (n)	A chronic (long-term) health issue.	Trouble/affection (par ex., "affection génétique")
Consanguinity (n)	When parents are blood relatives to each other.	Consanguinité

Term	Definition	Translation
DCIS (ductal carcinoma in situ) (n)	A pre-cancerous growth in the breast, which is still within the milk ducts, but has the potential of growing and spreading to other parts of the breast tissue. Sometimes called Stage 0 breast cancer.	Carcinome canalaire in situ (CCIS)
DTC (direct-to-consumer genetic testing) (n)	Genetic testing that is marketed directly to people at home through the internet or the TV etc. instead of through a medical provider.	Tests génétiques offerts directement aux consommateurs (DTC)
Deleterious mutation/ Disease-causing mutation (n)	A change in a person's DNA that may cause a medical condition. In cancer genetics, having a disease-causing mutation may increase the chance of getting cancer but does not mean that a person will definitely get cancer.	Mutation délétère/mutation entraînant une maladie
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 banking (n)	The secure, long-term storage of a person's genetic material. DNA banking allows families to have access to a deceased family member's DNA. When new genetic testing techniques are developed, doctors can test the banked DNA and use genetic information from the deceased person to tailor medical treatment for living descendants.	Banque d'ADN
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 order of the chemicals that make up the DNA. 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
DNA sequence (n)	The exact order of the chemicals that make up a DNA molecule.	Séquence de l'ADN
DNA sequencing (n)	The laboratory technique used to determine the exact order of the chemicals that make up a DNA molecule. This is one type of genetic testing.	Séquençage de l'ADN

Term	Definition	Translation
Dominant (adj)	A genetic trait in which one copy of the gene is sufficient for a trait to be expressed; dominant traits can be inherited from a single parent. In cancer genetics, dominant inheritance refers to conditions in which having only one altered copy of a particular gene pair is sufficient to cause an increased risk for cancer.	Dominant
Duplication (n)	A duplication occurs when part of a chromosome is copied abnormally, resulting in extra genetic material.	Duplication
Early age of onset (n)	Cancer diagnosed at an early age, usually before the age of 50.	
Egg (n)	The reproductive cells of a woman. When fertilized by sperm, the egg will grow into a baby.	Ovule
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
Familial (adj)	Belonging to a family; e.g., a familial trait is a trait that is shared among 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 family.	Antécédent familial
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
Fragment (n)	A small piece; an incomplete part of a whole.	Fragment
Gene (n)	A specific sequence of DNA that determines specific traits in an individual.	Gène

Term	Definition	Translation
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.	Copie de gène
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."	Copie d'un gène normal
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."	Copie d'un gène modifié
Gene deletion (n)	Having a piece of genetic information missing from a gene.	Délétion d'un gène
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.	Population générale
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.	Génération
Genetic (adj)	Having to do with inherited traits.	Génétique
Genetic counseling (n)	A discussion with a genetic counselor 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.	Consultation génétique

Term	Definition	Translation
Genetic counseling intern (n)	A genetic counseling student who has not yet completed his/her academic studies and is now practicing under the supervision of a more experienced counselor in preparation for providing genetic counseling services independently after obtaining his/her graduate degree.	Interne/stagiaire en consultation génétique
Genetic counselor (n)	A healthcare professional with a specialized graduate degree who works with people undergoing genetic testing. 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.	Conseiller en génétique
Genetic discrimination (n)	The act of refusing to provide, or charging more for, insurance or any service based on an individual's genetic make-up.	Discrimination génétique
Genetic factors (n)	Specific aspects of a person's genetic make-up that influence that person's health and development.	Facteurs génétiques
Genetic information (n)	The information encoded in genes, which tells every cell in a body how to grow, what to do and how to reproduce.	Informations génétiques
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)	A laboratory test designed to determine if a person has a gene mutation or a typical DNA sequence.	Test génétique
Single gene test (n)	Single gene test: analysis of one particular gene.	Test génétique sur un point spécifique
Gene panel test (n)	Gene panel test: analysis of several genes that have been shown to be associated with a particular condition; Example: breast cancer gene panel.	Test d'un panel de gènes

Term	Definition	Translation
Genetic trait (n)	A characteristic within a family that is passed down from parent to child genetically.	Caractère génétique
Geneticist (n)	A doctor or scientist who studies genetics.	Généticien(ne)
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. After the egg and sperm come together to form a baby, these cells will then divide and multiply and will form the entire body. Germline testing is different from testing the DNA of someone's tumor, which may have a mutation that happened during the person's lifetime and was not inherited from his or her parents.	Analyse germinale
Hereditary (adj)	Passed down from parent to child.	Héréditaire
Hereditary material (n)	Genetic material that is passed down from parent to child.	Matériel héréditaire
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)
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
Malignant (adj)	In cancer, this term means that the cancer cells or tumor are harmful and have the potential of spreading to other tissues or parts of the body.	Malin (masculin)/Maligne (féminin)

Term	Definition	Translation
Metastasis (n)	The spreading of cancer from one organ to another.	Métastase
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
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, which can be deleterious (disease causing) or benign (non-disease-causing). In Spanish, the word is sometimes confused with "mutilation."	Mutation
Oncologist (n)	A physician who specializes in diagnosing and treating cancer.	Oncologue
Oophorectomy	The surgical removal of one or both ovaries.	Ovariectomie
Ovarian cancer (n)	Cancer of the ovaries, the organs in a woman that release eggs.	Cancer ovarien ou Cancer des ovaires
Packets of genetic information (n)	A phrase genetic counselors use to describe genes or chromosomes.	Paquets d'information génétique
Pattern (n)	A repeating arrangement or sequence; for example, the pattern of cancers in a family.	Modèle ou Profil
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
Predisposition to cancer	Having a change in one of number of specific genes (a genetic mutation) that creates a higher than normal risk of a person developing cancer.	Prédisposition au cancer
Prognosis (n)	The most likely outcome of a disease process.	Pronostic

Term	Definition	Translation
Proliferation (cell Proliferation) (n)	The controlled process by which a cell multiplies. Cancer arises when the process of cell division becomes uncontrolled.	Prolifération (prolifération des cellules)
Prophylactic mastectomy (n)	A mastectomy is the surgical removal of the breast. "Prophylactic" means something done to prevent disease. A prophylactic mastectomy is the surgical removal of a healthy breast to prevent breast cancer. Prophylactic mastectomy is an option for individuals with a very high breast cancer risk, for example, women with a BRCA mutation.	Mastectomie prophylactique (n)
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.	Protéine
Random (adj)	Happening in an unpredictable way.	Aléatoire
Recessive (adj)	A genetic trait in which both copies of a gene are needed for the trait to be expressed; recessive traits are inherited from both parents, each one contributing one copy of the gene in question. In cancer genetics, recessive inheritance refers to conditions in which both copies of a particular gene pair need to be altered to develop the disease.	Récessif
Recurrence (n)	When something happens again. In cancer, a return of the disease after treatment and after a period of time during which the cancer cannot be detected.	Récurrence Rechute Récidive
Red flag (n)	A warning sign or a clue.	Signal d'alarme

Term	Definition	Translation
Risk (n)	The chance that something will happen; in the case of cancer genetics, the chance of getting cancer.	Risque
At risk (adj)	Has some possibility of getting cancer.	À risque
At average risk (adj)	Has the same possibility of getting cancer as the general population.	À risque moyen
At high risk (adj)	Has a greater possibility of getting cancer than the general population.	À haut risque
At higher risk than "X" (adj)	Has a greater possibility of getting cancer than "X."	À risque plus élevé que « X »
Risk factor (n)	A circumstance that increases the risk of getting cancer.	Facteur de risque
Runs in the family (e.g., cancer runs in the family) (v)	Is passed down from parents to children to grandchildren.	Avoir des antécédents dans la famille
Saliva (spit) test (n)	A genetic test done by collecting saliva (spit) instead of blood.	Échantillon de salive
Screen (v)	To test a person for a particular common health problem, even though the person has no symptoms to suggest that they have the problem.	Dépister
Screening test (n)	A test that looks for a common health problem even though the patient has no symptoms to suggest they have the particular disease.	Test de dépistage
Sperm (n)	The reproductive cells of the man. When sperm fertilize a woman's egg, a baby develops.	Sperme
Sporadic (adj)	Random, once in a while. In genetics, sporadic cancers are those caused by random chance or unknown factors in the environment.	Sporadique
Statistically significant (adj)	Not caused by chance.	Important sur le plan statistique

Term	Definition	Translation
Syndrome (n)	A group of symptoms or features that consistently occur together or a condition characterized by a set of associated features. For example, a hereditary cancer syndrome refers to risk for a group of cancers all caused by a particular gene mutation. BRCA1 mutations increase risk for breast and ovarian cancers. This is called Hereditary Breast and Ovarian Cancer Syndrome. See also, Lynch Syndrome.	Syndrome

Term	Definition	Translation
Test result (n)		Résultat d'analyse
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.	Négatif
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.	Vrai négatif
Uninformative negative (n)	Sometimes a family has a strong history of a particular cancer, but no genetic change related to that cancer has been found in that family. An "uninformative negative" result means that a specific test was negative but is not conclusive because the "family" mutation has not been identified, and/or there may be changes that the test could not detect in the gene being analyzed, or in other genes that were not tested.	(Résultat) négatif non informatif
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.	Positif
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.	Mutation d'importance incertaine
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."	Polymorphisme
Trait (n)	A characteristic.	Trait
Tumor (n)	An abnormal growth of cells; a tumor can be benign (not harmful) or malignant (harmful, cancer).	Tumeur

Term	Definition	Translation
Tumor testing (n)	An analysis of the DNA of someone's tumor. This analysis is meant to identify changes that a person might have acquired in his/her tumor cells that are not present in the rest of his/her body cells and were not inherited from his/her parents. This testing is often done to help make treatment decisions.	Dépistage des tumeurs
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.	Ultrason/ échographie
Variant (n)	A version of something that differs from the norm. So, a genetic variant is a change to the usual genetic sequence.	Mutation

Examples of Genes That are Often Checked in Genetic Testing

Term	Definition	Translation
BRCA 1 **BRCA 2**	"The Breast Cancer Genes." The tumor suppressor genes that in mutated form tend to be associated with an increased risk of certain cancers and especially breast and ovarian cancers.	BRCA 1 BRCA 2
MLH1 MSH2 MSH6 PMS2	"The Colon Cancer Genes" These genes are involved in fixing mistakes that occur when DNA replicates. An abnormality in one of these genes can lead to Lynch Syndrome, a condition linked to increased risk of cancer in the colon, rectum, uterus, ovaries, as well as other organs.	MLH1 MSH2 MSH6 PMS2
ATM	The ATM gene helps create a protein located in the cell nucleus that controls the rate at which cells grow and divide. The ATM protein also helps cells recognize damaged DNA strands. A mutation in the ATM gene can lead to increased risks for cancer or a condition called ataxia-telangiectasia a degenerative neurological condition that causes severe disability	ATM
PALB2	Another gene in which mutations can lead to breast cancer and possibly other cancers.	PALB2

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

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

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)
Domestic partner	The person with whom you are living and have an intimate relationship, but to whom you are not married.	Compagne/compagnon

Term	Definition	Translation
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

Common Interpreting Errors

Source speech	Interpreted as	More accurate rendition
"Cancer just happens by chance."	"Cancer happens suddenly." "Cancer happens because you are unlucky. "	"Sometimes we don't know what causes cancer."
Cancer genes	Cancer cells	Genes that may cause cancer when altered (mutated)
"You are at higher risk for developing cancer."	Getting cancer	There is a greater possibility that you might get cancer
"These genes protect us against cancer."	"They protect us, like the immune system . . . They kill cancer cells . . ."	"These genes help us to not get cancer."
Colonoscopy	Scan	Colonoscopy

Analogies

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

Examples:

"Genes are **like** an instruction book or an instruction manual. And in a book, there is a certain sequence to a story."

"**It's as if** you were reading through a long book and looking for one typo, for a letter that is mistaken."

"In the DNA there are molecule strings – **like** in this picture, each letter is a different molecule string."

"So a mutation is **like** a broken gene."

"So a mutation is **like** an unexpected change in the order of the letters."

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

"It's **like** chapters in a book, and like misspellings."

"Our cells are **like** libraries and our chromosomes are like books."

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

"Inside every cell are genes, which are **like** instruction books for our bodies; they tell our bodies how to grow and develop."

"It is kind of **like** if you a reading a book = and you notice that a word is spelled wrong; that is what this genetic test looks for."

Genetic counselors may mix the analogy in with more literal descriptions:

Example:

"We know that there are two in particular that when they don't work right, **when there's a typo in the instruction manual**, they can cause a high risk for breast and ovarian cancer."

Some analogies may not work with certain patients, due to linguistic or experiential differences.

Analogies that focus on genes as "letters in a book" may make no sense to speakers of character-based written languages such as Chinese, Japanese, and Korean.

"**It's as if** you were reading through a long book and looking for one typo, for a letter that is mistaken."

"So a mutation is **like** an unexpected change in the order of the letters."

"It's **like** chapters in a book, and like misspellings."

"It is kind of **like** if you a reading a book and you notice that a word is spelled wrong; that is what this genetic test looks for."

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

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

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

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

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

Compiled and edited by:
Cynthia E. Roat, MPH

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

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.

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
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

Term	Definition	Translation
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.”	Déficience 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
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)

Term	Definition	Translation
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
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

Term	Definition	Translation
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 an increased risk 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
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

Term	Definition	Translation
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 *duplication* occurs when part of a chromosome is copied abnormally, resulting in extra *genetic* 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
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

Term	Definition	Translation
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
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

Term	Definition	Translation
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.	Copie de gène
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."	Copie d'un gène normal
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."	Copie d'un gène modifié
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)	"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.	Population générale
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.	Génération
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).	Alphabet génétique

Term	Definition	Translation
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.	Consultation génétique
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.	Conseiller en génétique
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.	Discrimination génétique
Genetic factors (n)	Specific aspects of a person's genetic make-up that influence that person's health and development.	Facteurs génétiques
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.	Informations génétiques
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

Term	Definition	Translation
Genetic test (n)	A laboratory test designed to determine if a person has a gene mutation or a typical DNA sequence.	Test génétique
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.	Test génétique sur un point spécifique
Individual gene testing (n)	Individual gene testing: genetic analysis of the entirety of one specific gene.	Test d'un gène individuel
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.	Test d'un panel de gènes
Microarray (n)	Microarray: genetic analysis that looks for unexpected numbers of specific genes (two copies of every gene code is usually expected).	Puce à ADN
Exome sequencing (n)	Exome testing: a genetic test in which the exons of a patient's genetic code are sequenced and studied.	Séquençage de l'exome
Genome sequencing (n)	Genome sequencing: a genetic test in which the entire genetic code (the genome) of a patient is mapped out and studied.	Séquençage du génome
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
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

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.	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)	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.	Fécondation/Fertilisation In Vitro (FIV)
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

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.	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 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 *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.	Protéine
Random (adj)	Happening in an unpredictable way.	Aléatoire
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.	Récessif

Term	Definition	Translation
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
Risk (n)	The chance that something will happen; in the case of prenatal genetics, the chance that the child will have a genetic condition.	Risque
At risk (adj)	Has some possibility of having a genetic condition.	À risque
At average risk (adj)	Has the same possibility of having a genetic condition as the general population.	À risque moyen
At high risk (adj)	Has a greater possibility of having a genetic condition than the general population.	À haut risque
At higher risk than "X" (adj)	Has a greater possibility of having a genetic condition than "X."	À risque plus élevé que « X »
Risk factor (n)	A circumstance that increases the risk of having a genetic condition.	Facteur de risque
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
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

Term	Definition	Translation
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."	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
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

Term	Definition	Translation
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
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.	Résultat d'analyse Négatif
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.	Vrai négatif
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.	Positif
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.	Mutation d'importance incertaine
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."	Polymorphisme
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)	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.	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

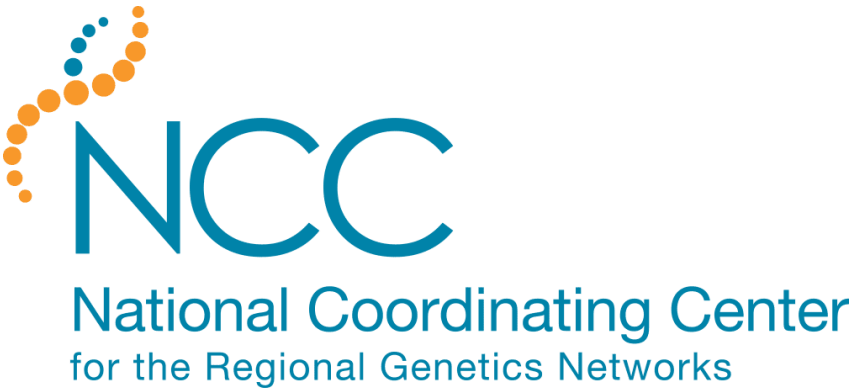
Term	Definition	Translation
Cousin	Usually understood to be a first cousin.	Cousin(e)
First cousin	Your aunt or uncle's child.	Cousin germain/ cousine germaine
First cousin once removed	Your aunt or uncle's grandchild or your first cousin's child	Cousin(e) issu(e) de germain/ petit(e) cousin(e)
Second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

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

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)
Domestic partner	The person with whom you are living and have an intimate relationship, but to whom you are not married.	Compagne/compagnon

Term	Definition	Translation
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

This page intentionally left blank.



The National Coordinating Center for the Regional Genetics Networks

Glossary of Prenatal Genetics Terms

English – French

2022 Edition

Compiled and edited by:
Cynthia E. Roat, MPH

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

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 Prenatal Genetics is provided to help enable access to prenatal 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 prenatal genetics terms are:

- 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.
- Examples of analogies used in genetics. Genetic counselors often use analogies to explain complex ideas in genetics. An analogy explains a new concept by comparing it to something more common.

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
Adoption (n)	The process of legally taking someone else's child into your family to raise as your own child.	Adoption
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
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. E.g., when discussing cancer, "benign" means "not cancerous."	Bénin(e)
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

Term	Definition	Translation
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.”	Déficience congénitale
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
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 has a genetic mutation in one of their two copies of a particular gene that is associated with a genetic condition. The other copy of the gene does not have a mutation, so that person usually does not have any medical issues related to the gene change.	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
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 types of chromosomes in a cell.	Analyse chromosomique/caryotype

Term	Definition	Translation
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
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
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
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
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 specific problem. Screening tests often only report whether a patient is at an increased risk for the medical problem in question, whereas diagnostic tests report whether the problem is actually present.	Test de diagnostic

Term	Definition	Translation
DNA (Deoxyribonucleic Acid) (n)	The material that carries the genetic information of a cell. It provides the instructions used in the development, functioning and reproduction of the organism of which it is a part. If you could stretch out the DNA of a chromosome and look at it through a microscope, it would look like a long ladder that is twisted into a spiral. The 'sides' of the ladder are made up of alternating phosphate and sugar groups. The 'rungs' are various combinations of two nitrogen bases: Adenine-Thymine and Cytosine-Guanine. Individual sections of DNA that code for specific traits/functions are called genes.	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
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

Term	Definition	Translation
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 duplication occurs when part of a chromosome is copied abnormally, resulting in extra genetic 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
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 among 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

Term	Definition	Translation
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
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
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.	Population générale
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.	Génération

Term	Definition	Translation
Genetics (adj)	The science of how an organism's genes interact with the environment to produce certain traits.	Génétique
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.	Consultation génétique
Genetic counseling intern (n)	A genetic counseling student who has not yet completed his/her academic studies and is now practicing under the supervision of a more experienced counselor in preparation for providing genetic counseling services independently after obtaining his/her graduate degree.	Interne/stagiaire en consultation génétique
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.	Conseiller en génétique
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.	Discrimination génétique
Genetic factors (n)	Specific aspects of a person's genetic make-up that influence that person's health and development.	Facteurs génétiques
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.	Informations génétiques
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)	A laboratory test designed to determine if a person has a change to their DNA.	Test génétique

Term	Definition	Translation
Genetic trait (n)	A characteristic within a family that is passed down from parent to child through their DNA.	Caractère génétique
Genetic 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.	Mutation génétique
Geneticist (n)	A doctor or scientist who studies genetics.	Généticien(ne)
Hereditary (adj)	Passed down from parent to child.	Héréditaire
Hereditary material (n)	Genetic material that is passed down from parent to child.	Matériel héréditaire
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.	Fécondation In Vitro (FIV)
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 libre et éclairé/ consentement informé
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)	Having 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)

Term	Definition	Translation
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 in the second trimester, between 14-20 weeks.	Dépistage par le sérum maternel (analyse du taux d'alpha-foœtoprotéine sérique maternelle)
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
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." In French, the word is sometimes confused with "mutilation."	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

Term	Definition	Translation
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
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 du tube neural
Ova (n)	See “egg.”	Ovules
Ovary (n)	The organ in a woman that stores and releases eggs. There are normally two.	Ovaire
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 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.	Protéine
Random (adj)	Happening in an unpredictable way.	Aléatoire

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.	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
Risk (n)	The chance that something will happen; in the case of prenatal genetics, the chance that the child will have a genetic condition.	Risque
At risk (adj)	Has some possibility of having a genetic condition.	À risque
At average risk (adj)	Has the same possibility of having a genetic condition as the general population.	À risque moyen
At high risk (adj)	Has a greater possibility of having a genetic condition than the general population.	À haut risque
At higher risk than 'X" (adj)	Has a greater possibility of having a genetic condition than "X".	À risque plus élevé que « X »
Risk factor (n)	A circumstance that increases the risk of having a genetic condition.	Facteur de risque
Runs in the family (e.g., sickle cell disease runs in the family) (v)	When a certain condition or trait seems to appear in many members of a single family	Avoir des antécédents dans la famille (par ex, la famille a des antécédents de maladie drépanocytaire)

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)	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., 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'.	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
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
Sperm (n)	The reproductive cells of the man. When sperm fertilize a woman's egg, a baby develops.	Sperme
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

Term	Definition	Translation
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
Test results (n) **negative (adj)**	A negative result on a diagnostic genetic test means that the laboratory did not find the specific genetic change that the test was designed to identify. A “screen negative” result on a screening test means that the person’s risk of having whatever the test was designed to find is lower than the risk for most people.	Résultats d’analyse Négatif
Positive (adj)	A positive result on a diagnostic genetic test means that the laboratory did find a specific genetic change that is associated with a hereditary condition (a deleterious mutation). A “screen positive” result on a screening test means that the person’s risk of having whatever the test screened for is significantly higher than expected. That still does not mean that the person definitely has that condition.	Positif
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. Many of these variants will eventually be reclassified as either a “positive” or “negative” result. Most become “negative” and are thought to represent natural variation between individuals.	Mutation d’importance incertaine
Polymorphism (n)	Everyone has some degree of commonly occurring genetic changes that are not associated with medical problems. If the test finds this sort of change, it reports a “polymorphism.”	Polymorphisme
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)	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.	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
Cousin	Usually understood to be a first cousin.	Cousin(e)
First cousin	Your aunt or uncle's child.	Cousin germain/ cousine germaine
First cousin once removed	Your aunt or uncle's grandchild or your first cousin's child	Cousin(e) issu(e) de germain/ petit(e) cousin(e)
Second cousin	The children of first cousins are second cousins to each other.	Cousin(e) issu(e) de germain/ cousin(e) au deuxième degré
Grandparent	Your parent's father or mother.	Grand-parent
Grandmother	Your mother or father's mother.	Grand-mère
Maternal grandmother	Your mother's mother.	Grand-mère maternelle
Paternal grandmother	Your father's mother.	Grand-mère paternelle
Grandfather	Your mother or father's father.	Grand-père
Maternal grandfather	Your mother's father.	Grand-père maternel
Paternal grandfather	Your father's father.	Grand-père paternel
Great aunt	Your mother or father's aunt.	Grand-tante
Maternal great aunt	Your mother's aunt.	Grand-tante maternelle
Paternal great aunt	Your father's aunt.	Grand-tante paternelle
Great uncle	Your father or mother's uncle.	Grand-oncle
Maternal great uncle	Your mother's uncle.	Grand-oncle maternel
Paternal great uncle	Your father's uncle.	Grand-oncle paternel
Great grandparents	The parents of any of your grandparents.	Arrière-grands-parents
Great grandmother	The mother of any of your grandparents.	Arrière-grand-mère
Great grandfather	The father of any of your grandparents.	Arrière-grand-père

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

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)
Domestic partner	The person with whom you are living and have an intimate relationship, but to whom you are not married.	Compagne/compagnon

Term	Definition	Translation
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

Analogies

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

Examples:

"Genes are **like** an instruction book or an instruction manual. And in a book, there is a certain sequence to a story."

"**It's as if** you were reading through a long book and looking for one typo, for a letter that is mistaken."

"So, a mutation is **like** an unexpected change in the order of the letters."

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

"It's **like** chapters in a book, and like misspellings."

"Our cells are **like** libraries and our chromosomes are like books."

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

"Inside every cell are genes, which are **like** instruction books for our bodies; they tell our bodies how to grow and develop."

"It is kind of **like** you a reading a book and you notice that a word is spelled wrong; that is what this genetic test looks for."

Some analogies may not work with certain patients, due to linguistic or experiential differences.

Analogies that focus on genes as "letters in a book" may make no sense to speakers of character-based written languages such as Chinese, Japanese, and Korean.

"**It's as if** you were reading through a long book and looking for one typo, for a letter that is mistaken."

"So a mutation is **like** an unexpected change in the order of the letters."

"It's **like** chapters in a book, and like misspellings."

"It is kind of **like** if you a reading a book and you notice that a word is spelled wrong; that is what this genetic test looks for."

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

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

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

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

This page intentionally left blank.

Index

A

B

C

D

E

F

G

H

I

J

K

L

M

N

O

P

R

S

T

U

V

W

X