



National Coordinating Center
for the Regional Genetics Networks

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

Glossaries of Genetics Terms

English – Chinese

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

Glossaries of Genetics Terms	1
Glossary of Autism Spectrum Disorder (ASD) Genetics Terms	5
Preface.....	7
How to Use the Glossary.....	7
Glossary of Terms	9
References	29
Glossary of Cancer Genetics Terms	31
Preface	33
How to Use the Glossary.....	33
Glossary of Terms	35
Examples of Genes That are Often Checked in Genetic Testing	47
Family Relationships	48
Common Interpreting Errors	55
Analogies.....	56
Glossary of Pediatric Genetics Terms	57
Preface	59
How to Use the Glossary.....	59
Glossary of Terms	61
Family Relationships	81
Glossary of Prenatal Genetics Terms	89
Preface	90
How to Use the Glossary.....	90
Glossary of Terms	91
Analogies.....	114
Index	117



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

**Glossary of
Autism Spectrum Disorder (ASD) Genetics
Terms**

English – Chinese

2022 Edition

Compiled and edited by:
Cynthia E. Roat, MPH

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

Glossaries of Autism Spectrum Disorder (ASD) Terms – Chinese

This page intentionally left blank.



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.

Glossaries of Autism Spectrum Disorder (ASD) Terms – Chinese

This page intentionally left blank.

Glossary of Terms

English – Chinese

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

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
A-B-C analysis (n)	An approach to understanding behavior by examining the Antecedent (the cause), the Behavior, and the Consequence (the result). ¹	理性情緒行為分析	理性情绪行为分析
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	抽象語言	抽象语言
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	適應行為 / 適應技巧	适应行为或适应技巧
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	成年年齡	成年年龄
Anxiety disorder (n)	A pattern of constant worry or tension under many different circumstances. ¹	焦慮症	焦虑症
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	應用行為分析 (ABA)	应用行为分析 (ABA)
Apraxia (n)	See “Dyspraxia.” ²	失用症	失用症

Glossaries of Autism Spectrum Disorder (ASD) Terms – Chinese

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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	專注力不足過度活躍症 / 注意力缺陷多动障碍 (ADHD)	注意力缺陷多动障碍 (ADHD)
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	聽力學家	听力学家
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	輔助溝通系統 (AAC)	增强和替代沟通 (AAC)
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. <i>social communication</i> , or the way a person uses gestures, body language, and language to communicate and interact socially and relate with others. 2. the presence of <i>restricted, repetitive patterns of behavior, interests, or activities</i> (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	自閉症譜系障礙 (ASD)	自闭症谱系障碍 (ASD)
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	厭惡感的	厌恶感的
Behavior Intervention Plan (BIP) (n)	A plan to improve an individual’s behavior, created based on the results of a Functional Behavior Assessment. ¹	行為介入治療計畫 (BIP)	行为干预治疗计划 (BIP)

Glossaries of Autism Spectrum Disorder (ASD) Terms – Chinese

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
Behavioral disorder (n)	A condition in which behavior significantly deviates from acceptable norms. ¹	行為障礙	行为障碍
Biomarker (n)	An indicator of a certain biological state. ¹	生物標記	生物标志
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	躁鬱症	躁郁症
Board Certified Behavior Analyst (BCBA) (n)	A professional certified to provide ABA therapy by the Behavior Analyst Certification Board (BACB). ¹	國際認證行為分析師 (BCBA)	国际认证行为分析师 (BCBA)
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	肢體語言	肢体语言
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	個案經理	个案经理
Catatonia (n)	A state in which a person does not move and does not respond to others. ¹	緊張症	紧张症
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	具挑戰性行為	具挑战性行
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	民事拘押	民事拘禁
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	認知行為治療	认知行为疗法
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	認知發展	认知发展

Glossaries of Autism Spectrum Disorder (ASD) Terms – Chinese

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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	溝通	沟通
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	溝通障礙	沟通障碍
Comorbid (adj)	Pertaining to a disease or disorder that occurs simultaneously with another. ¹	合併症的	合并症的
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	強迫症	强迫症
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	託管(照管)權	监管权
Crisis plan (n)	A document that outlines in specific detail the necessary strategies and steps that must be taken when a crisis occurs. ¹	危機應對計畫	危机应对计
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	數據分析	数据分析
De-escalation (n)	The process of stopping a challenging behavior or crisis from intensifying and calming the situation. ¹	逐步降級	逐步降级
Depression (n)	A mood disorder in which feelings of sadness, anger, or frustration interfere with everyday life for an extended period of time. ¹	憂鬱症	抑郁症

Glossaries of Autism Spectrum Disorder (ASD) Terms – Chinese

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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	成長發展評估	发育评估
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	發展遲緩	发育迟缓
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	發展障礙 (DD)	发育障碍 (DD)
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	行為發展兒科 醫生	发育儿科医生
Differential diagnosis (n)	Distinguishing between two or more diseases with similar symptoms to identify which is causing distress or challenging behavior. ¹	鑑別診斷	鉴别诊断
Disruption (n)	An event that causes an unplanned deviation from a situation. ¹	中斷	中断

Glossaries of Autism Spectrum Disorder (ASD) Terms – Chinese

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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	唐氏綜合症, 也被稱為 21 號 - 三染色體	唐氏綜合症, 也称为 21 号三染色体
Dual diagnosis (n)	The identification of an additional mental health disorder individuals with developmental disabilities. ¹	合併診斷	合并诊断
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	構音障礙	构音障碍
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	失用症	运动障碍
Early intervention (n)	Programs or services designed to meet the developmental needs of infants and toddlers (birth to three years old) and their families.	早期介入	早期干预
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	模仿言語 / 仿語	仿语或模仿言语
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	潛逃	潜逃
Epilepsy (n)	A brain disorder in which a person has repeated seizures (episodes of disturbed brain activity or convulsions) over time. ¹	癲癇症	癫痫症
Escalating (v)	Increasing or worsening rapidly. ¹	升級	升级
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	循證實踐 (EBP)	循证实践 (EBP)

Glossaries of Autism Spectrum Disorder (ASD) Terms – Chinese

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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	管控/執行功能	管控/执行功能
Extinction (n)	A response used to eliminate a behavior that involves ignoring a mild behavior when it is used for attention. ¹	消除（不良行為）	消除（不良行为）
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	消除爆發行為	消除爆发行为
Face blindness (n)	An impairment in the recognition of faces. ¹	臉盲症	面盲症
Fading (v)	Gradually reducing the number of prompts or types of prompts to encourage more independence for the learner. ⁵	漸減	渐减
Fecal digging (v)	The process in which an individual puts his fingers into his rectum. ¹	指挖糞便	指挖粪便
Fecal smearing (v)	The process in which feces are spread on property or the individual himself. ¹	糞便塗抹	粪便涂抹
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	胎兒酒精譜系障礙 (FASD)	胎儿酒精谱系障碍 (FASD)
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	胎兒酒精綜合症 (FAS)	胎儿酒精综合症 (FAS)
Function of behavior (n)	The purpose or reason behind a specific behavior for an individual. ¹	功能性行為	功能性行为

Glossaries of Autism Spectrum Disorder (ASD) Terms – Chinese

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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	功能性行為分析學 (FBA)	功能性行為分析 (FBA)
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.	功能性溝通	功能性沟通
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	一般化	普遍化
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	手勢	手势
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	監護權	监护权
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	過度敏感 / 超敏反應	高度敏感

Glossaries of Autism Spectrum Disorder (ASD) Terms – Chinese

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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	低度敏感 / 低靈敏度	低度敏感
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	異常語言 / 詞彙	异常语言/词汇
Incontinence (n)	The (usually) involuntary passing of feces or urine, generally not into a toilet or diaper. ¹	失禁	失禁
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	個別化教育計劃 (IEP)	个别化教育计划 (IEP)

Glossaries of Autism Spectrum Disorder (ASD) Terms – Chinese

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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	個別化家庭服務計劃 (IFSP)	个别化家庭服务计划 (IFSP)
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	殘疾人士教育法案 (IDEA)	殘障人士教育法 (IDEA)
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	智力障礙	智力障碍
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	跨學科評估團隊	跨学科评估团队
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	介入治療	干预治疗
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	非自願或強制監管（治療）	非自愿治疗

Glossaries of Autism Spectrum Disorder (ASD) Terms – Chinese

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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	共享式注意力	共享式注意力
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	語言遲緩	语言延迟
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	語言障礙	语言障碍
Learning disability (n)	A disorder that affects how a person learns and understands primary skills such as reading, writing and math. ²	學習障礙	学习障碍
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	最少限制的環 境 (LRE)	最少限制的环 境 (LRE)
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	適應不良的行 為	适应不良的行 为
Medicaid (n)	A government program that provides healthcare coverage for low-income families and individuals with disabilities in the United States. ¹	醫療補助計畫 (俗稱白卡)	医疗补助计划
Medical home (n)	A team-based healthcare delivery model led by a physician that provides comprehensive and continuous medical care to patients. ¹	團隊式醫療護 理	团队式医疗护 理
Mental health hold (n)	Involuntary hospitalization due to a mental health crisis. ¹	強制性滯留	强制性滞留

Glossaries of Autism Spectrum Disorder (ASD) Terms – Chinese

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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	運動技能	运动技能
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.	神經發育障礙	神经发育障碍
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	非語言溝通/身體語言	非语言交流
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	營養師	营养师
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	執念	执念
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	強迫症 (OCD)	强迫症 (OCD)
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	職能治療師 (OT)	职业治疗师 (OT)
Ototoxic (n)	Damaging to the ears, causing sound sensitivities, dizziness, or balance issues. ¹	耳毒性	耳毒性

Glossaries of Autism Spectrum Disorder (ASD) Terms – Chinese

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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	過度更正	过度校正
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	鏈球菌感染導致兒童自體免疫神經精神異常, 暱稱熊貓症 (PANDAS)	链球菌感染导致儿童自身免疫性神经精神疾病 (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	苯丙酮尿症 (PKU)	苯丙酮尿症 (PKU)
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	物理治療師 / 治療師 (PT)	物理治疗师 (PT)
Pica (n)	An eating disorder that involves eating things that are not food (e.g., dirt, plastic). ¹	異食癖	异食癖
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	圖片交換溝通系統 (PECS)	图片交换沟通系统 (PECS)
Polypharmacy (n)	The use of multiple medications by a patient. ¹	多重用藥	多重用药
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	正向行為支持 (PBS)	积极行为支持 (PBS)
Post-Traumatic Stress Disorder (PTSD) (n)	An anxiety disorder that can occur after witnessing or experiencing a traumatic event. ¹	創傷後壓力綜合症 (PTSD)	创伤后应激障碍 (PTSD)

Glossaries of Autism Spectrum Disorder (ASD) Terms – Chinese

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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 37 th week of pregnancy. ²	早產	早产
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	提示	提示
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	心理學家	心理学家
Psychosis (n)	A loss of contact with reality that usually includes delusions and hallucinations. ¹	精神病（香港稱之為思覺失調）	精神病
Psychotropic (adj)	A medication or intervention that affects brain activity, behavior, or perception. ¹	精神藥物的	精神药物的
Puberty (n)	The process of physical changes that occur when a child's body matures into an adult. ¹	青春期	青春期
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	危險訊號	危险信号
Reinforce (v)	To strengthen with additional material or support. ¹	加強	巩固
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."	加強策略 / 強化策略	强化策略
Resilience (n)	An ability to recover from or adjust easily to change or a difficult situation. ¹	抗逆力 / 快速恢復的能力 / 還原能力 / 恢復力	应变能力
Respite care (n)	A service that provides short-term breaks that can relieve stress, restore energy, and promote balance for caregivers. ¹	短期暫替療護 / 喘息服務 / 臨時護理 / 住宿暫託服務	喘息照护
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	約束 / 限制	约束

Glossaries of Autism Spectrum Disorder (ASD) Terms – Chinese

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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	限制或重複性行為或興趣	限制或重复性行为或兴趣
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	獎勵	奖励
Risk factor (n)	Conditions that increase the likelihood of aggression. ¹	風險因素	风险因素
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	常規 / 習慣	常规
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	反芻	反刍
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	精神分裂症 (日本和台灣也稱之為思覺失調)	精神分裂症
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	篩檢測試或工具	筛检试验或工具
Seclusion (n)	A situation in which an individual is isolated in a room in response to a behavior they have exhibited. ¹	隔離	隔离
Sedating (v)	Calming, sleep-inducing, or numbing an individual experiencing challenging behaviors or struggling during difficult situations. ¹	鎮靜	镇静
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	癲癇障礙	癫痫症
Self-advocacy (n)	The ability of an individual to communicate his or her wants and concerns and make his or her own decisions. ¹	自我倡導	自我倡导
Sensory avoidance (n)	Blocking or staying away from something that is painful or bothersome. ¹	感覺規避 / 感官迴避	感官回避
Sensory defensiveness (n)	A tendency to react negatively or with alarm to sensory input which is generally considered harmless or non-irritating. ¹	感覺防禦	感觉防御

Glossaries of Autism Spectrum Disorder (ASD) Terms – Chinese

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
Sensory input (n)	Any source that creates sensation and activates one or more of the senses -vision, smell, sound, taste, and touch. ¹	感官輸入	感官輸入
Sensory processing disorder (n)	Refers to difficulty detecting, organizing, or responding to sensory information received and interpreted in the brain via all seven senses <u>and</u> that interferes with participation in daily life, development, behavior, and social interactions. ²	感覺處理障礙	感官处理障碍
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	感覺尋求行為	感官寻求行为
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	睡眠窒息症	睡眠窒息症
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	睡眠障礙	睡眠障碍
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	社交溝通障礙	沟通障碍
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	社交互動	社会互动
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	社工	社会工作或社会工作者
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	為特殊需要兒童家長的倡導	特殊需要家长代言人

Glossaries of Autism Spectrum Disorder (ASD) Terms – Chinese

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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	特殊學習障礙	特定学习障碍
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	語音生成裝置	语音生成装置 或语音输出技术
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	語音障礙	语音障碍
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	語言病理學家	语言病理学家
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	標準化測驗	标准化测验
Staring spells (n)	Occasions when an individual is in a trance staring into space, which can often signal seizure activity. ¹	凝視呆滯	凝视呆滞
Stereotypy (n)	Repetitive or ritualistic movements such as body rocking or crossing and uncrossing of legs. ¹	重複性行為 / 刻板行為	重复性行为/刻板行为
Stimulation (n)	Excitement or activity triggered by a stimulus either internally or externally. ¹	刺激	刺激
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	聯邦收入補助金 (SSI)	联邦收入辅助金 (SSI)
Tangibles (n)	Items or rewards that can be touched, such as a toy or piece of candy. ¹	有形 / 實體	有形
Time delay (n)	A prompting procedure that fades prompts during activities by having longer delays before a prompt is provided.	延時提示	延时提示
Tourette syndrome or Tourette’s syndrome (n)	A neurological disorder characterized by tics, or repetitive, stereotyped, involuntary movements and vocalizations. ¹	妥瑞綜合症 (俗稱抽動症)	图雷特综合症

Glossaries of Autism Spectrum Disorder (ASD) Terms – Chinese

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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	跟蹤量表	跟踪量表
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	過渡期計劃 / 過渡期服務	过渡计划/过渡服务
TRICARE (n)	The health care program for Uniformed Service members, retirees, and their families worldwide. ¹	軍事醫療護理	军事医疗护理
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	視覺線索	视觉线索
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	視覺時間表	视觉时间表
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.	職業康復/訓練中心	职业康复
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	語音輸出科技或語音輸出溝通輔助器或言語生成裝置	语音输出技术或语音输出沟通辅助或语音生成设备

Glossaries of Autism Spectrum Disorder (ASD) Terms – Chinese

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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	包圍式服務方案	全納式服務

Glossaries of Autism Spectrum Disorder (ASD) Terms – Chinese

This page intentionally left blank.

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>



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

**Glossary of
Cancer Genetics Terms**

English – Chinese

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.



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.

This page intentionally left blank.

Glossary of Terms

English – Chinese

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

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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.	異常的	异常的
Benign (adj)	Something mild that does not threaten health or life. In cancer genetics, “benign” means “not cancerous.”	良性的	良性的
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.	驗血	验血
Breast cancer (n)	Cancer of the breast tissue. This cancer is more common in women, but it can affect men as well.	乳腺癌	乳腺癌
Buccal swab (n)	A way to collect DNA from the cells on the inside of a person's cheek.	口腔黏膜測試	口腔黏膜检测
CA-125 blood test (n)	A blood test used to look for early signs of ovarian cancer in women with a high cancer risk.	CA-125 驗血 癌抗原 125 血液檢測	CA-125 验血 癌抗原 125 血液检测
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.	基因攜帶者	携带者
Cell (n)	A small (microscopic) structure that forms the basic building block of every known living organism.	細胞	细胞
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.	染色體	染色体
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).	結腸鏡檢查 (俗稱照大腸) 結腸鏡檢查 (俗稱大腸鏡)	结肠镜检查 (简称肠镜)

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
Condition (e.g., “genetic condition”) (n)	A chronic (long-term) health issue.	病症 (例如「遺傳病」)	病症 (例如“遗传病”)
Consanguinity (n)	When parents are blood relatives to each other.	血緣	血缘
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.	DCIS (乳腺導管原位癌) (導管原位癌)	DCIS (乳腺导管原位癌) (导管原位癌)
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.	DTC (直接面向消費者的基因檢測)	DTC (直接面向消费者的基因检测)
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.	有害突變 / 致病性突變	有害突变/ 致病性突变
DNA (deoxyribonucleic acid) (n)	The molecule that carries the genetic information of a cell. It provides the instructions used in the development, functioning and reproduction of the organism of which it is a part. The DNA is organized into chromosomes.	DNA (脫氧核糖核酸)	DNA (脱氧核糖核酸)
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.	DNA 資料庫 (基因資料庫)	DNA 资料库 (基因资料库)
DNA marker (also called a “genetic marker”) (n)	A readily recognizable genetic trait, gene, or DNA segment.	脫氧核糖核酸標記(也稱為遺傳標記)	脱氧核糖核酸标记 (也称为“遗传标记”)
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.	脫氧核糖核酸突變	脱氧核糖核酸突变
DNA sequence (n)	The exact order of the chemicals that make up a DNA molecule.	脫氧核糖核酸序列	脱氧核糖核酸序列

Term	Definition	Traditional/ 繁體中文	Simplified/ 简体中文
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.	脫氧核糖核酸定序	脱氧核糖核酸测序
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.	顯性的/顯著的	显性的
Duplication (n)	A duplication occurs when part of a chromosome is copied abnormally, resulting in extra genetic material.	複製	复制
Early age of onset (n)	Cancer diagnosed at an early age, usually before the age of 50.	疾病早发性 (癌症早發)	疾病早发性 (癌症早发)
Egg (n)	The reproductive cells of a woman. When fertilized by sperm, the egg will grow into a baby.	卵子	卵子
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.	環境原因或因素	环境原因或因素
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.	家族性的	家族性的
Family history (n)	The medical history of the members of a family.	家族史	家族史
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.	拋幣而定 (廣東話俗語「擲銀仔」)	掷硬币
Fragment (n)	A small piece; an incomplete part of a whole.	碎片	片段

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
Gene (n)	A specific sequence of DNA that determines specific traits in an individual.	基因	基因
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.	基因復制	基因拷贝
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.”	正常基因復制	正常基因拷贝
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.”	變異基因復制	变异基因拷贝
Gene deletion (n)	Having a piece of genetic information missing from a gene.	基因刪除	基因缺失
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.	一般人口	一般人群
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 form an even earlier generation. Your children and nieces and nephews form a later generation.	一代人 / 同一代人	一代人
Genetic (adj)	Having to do with inherited traits.	遺傳性的	遗传性的
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.	遺傳諮詢	遗传咨询

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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.	遺傳諮詢實習生	遗传咨询实习生
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.	遺傳諮詢師/遺傳顧問	遗传咨询师
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.	基因歧視	基因歧视
Genetic factors (n)	Specific aspects of a person's genetic make-up that influence that person's health and development.	遺傳因素	遗传因素
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.	遺傳資訊	遗传信息
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.	遺傳物質	遗传物质
Genetic predisposition (n)	An increased chance of a person developing a certain trait or disease based on that person's particular genetic makeup.	遺傳傾向性	遗传倾向性

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
Genetic test (n)	A laboratory test designed to determine if a person has a gene mutation or a typical DNA sequence.	基因檢測	基因检测
Single gene test (n)	Single gene test: analysis of one particular gene.	單一基因檢測	单一基因检测
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.	基因組測試	基因组检测
Genetic trait (n)	A characteristic within a family that is passed down from parent to child genetically.	遺傳特徵	遗传特征
Geneticist (n)	A doctor or scientist who studies genetics.	遺傳學家	遗传学家
Germline testing (n)	<p>Germline testing refers to the analysis of a person's DNA, which he or she inherited from his or her parents.</p> <p>In biology and genetics, the germline is the group of cells that will pass on the genetic material to children, in other words, the cells from which the eggs and sperm come. After the egg and sperm come together to form a baby, these cells will then divide and multiply and will form the entire body.</p> <p>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.</p>	殖源基因檢查	生殖系检测
Hereditary (adj)	Passed down from parent to child.	遺傳性的	遗传性的
Hereditary material (n)	Genetic material that is passed down from parent to child.	遺傳物質	遗传物质

Term	Definition	Traditional/ 繁體中文	Simplified/ 简体中文
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.	知情同意書	知情同意书
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.	遺傳模式	遗传模式
Inherited (adj)	Passed down from parent to child.	遺傳的	遗传的
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.	林奇氏症候群	林奇综合症
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.	惡性的	恶性的
Metastasis (n)	The spreading of cancer from one organ to another.	轉移	转移
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.	分子	分子
Multifactorial (adj)	Due to a combination of genetic and non-genetic (environmental, hormonal, etc.) risk factors that act together to determine risk.	多因素的	多因素的
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."	突變	突变
Oncologist (n)	A physician who specializes in diagnosing and treating cancer.	腫瘤科醫生 (腫瘤医师)	肿瘤科医生 (肿瘤医生)
Oophorectomy	The surgical removal of one or both ovaries.	卵巢切除手術	卵巢切除手术
Ovarian cancer (n)	Cancer of the ovaries, the organs in a woman that release eggs.	卵巢癌	卵巢癌

Glossary of Cancer Genetics Terms – Chinese

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
Packets of genetic information (n)	A phrase genetic counselors use to describe genes or chromosomes.	基因資訊 (封包)	遗传信息包
Pattern (n)	A repeating arrangement or sequence; for example, the pattern of cancers in a family.	模式	模式
Pedigree (n)	A family tree that can be used to trace the inheritance of specific genetic traits.	血統/血系	家谱
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.	外顯率	外显率
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.	癌症易患性	癌症易患性
Prognosis (n)	The most likely outcome of a disease process.	病程預測 / 預後	预后
Proliferation (cell proliferation) (n)	The controlled process by which a cell multiplies. Cancer arises when the process of cell division becomes uncontrolled.	增殖 (細胞激增))	增殖 (細胞激增))
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.	預防性乳房切 除術	预防性乳房切 除手术
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.	蛋白質	蛋白质
Random (adj)	Happening in an unpredictable way.	隨機的	随机的

Term	Definition	Traditional/ 繁體中文	Simplified/ 简体中文
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.	隱性的	隐性的
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.	復發性 (復發)	复发性 (复发)
Red flag (n)	A warning sign or a clue.	警告信號	危险信号
Risk (n)	The chance that something will happen; in the case of cancer genetics, the chance of getting cancer.	風險	风险
At risk (adj)	Has some possibility of getting cancer.	有風險的	有风险的
At average risk (adj)	Has the same possibility of getting cancer as the general population.	一般風險的	平均风险的
At high risk (adj)	Has a greater possibility of having a genetic condition than the general population.	高風險的	高风险的
At higher risk than “X” (adj)	Has a greater possibility of getting cancer than the general population. Has a greater possibility of getting cancer than “X.”	風險高於 「X」的	风险高于 “X”的
Risk factor (n)	A circumstance that increases the risk of getting cancer.	風險因素	风险因素
Runs in the family (e.g., cancer runs in the family) (v)	Is passed down from parents to children to grandchildren.	家族遺傳	家族遗传
Saliva (spit) test (n)	A genetic test done by collecting saliva (spit) instead of blood.	唾液（口水） 測試	唾液（口水） 样本
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.	篩檢	筛查
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.	篩檢測試	筛选检测

Glossary of Cancer Genetics Terms – Chinese

Term	Definition	Traditional/ 繁體中文	Simplified/ 简体中文
Sperm (n)	The reproductive cells of the man. When sperm fertilize a woman’s egg, a baby develops.	精子	精子
Sporadic (adj)	Random, once in a while. In genetics, sporadic cancers are those caused by random chance or unknown factors in the environment.	偶發性的	偶发性的
Statistically significant (adj)	Not caused by chance.	有統計意義的	有统计学意义的
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.	綜合症 (症候群)	综合症 (症候群)

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
Test result (n) Negative (adj) True negative (n) Uninformative negative (n) Positive (adj) Variant of uncertain significance (n) Polymorphism (n)	<p>A negative result on a genetic test means that the laboratory did not find the specific genetic change that the test was designed to identify.</p> <p>When a patient is being tested to see if he or she has a specific genetic change that is present in other family members, a negative result can be considered a “true negative.” This means that instead of having an increased risk for cancer like other family members, they have the same risk as everyone else in the general population.</p> <p>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.</p> <p>A positive result on a genetic test means that the laboratory did find a specific genetic change that is associated with a hereditary cancer syndrome (a deleterious mutation). In cancer genetics, that means that the patient has an increased risk of getting the cancer(s) associated with the gene.</p> <p>If the laboratory finds a genetic change for which currently there is not enough information to know if this change is problematic or not, it reports a “variant of uncertain significance.” Basically, this means that the laboratory found a genetic change, but they don’t know what it means. All of these variants will eventually be reclassified as either a “positive” or “negative” result. Most become “negative” and are thought to represent natural variation between individuals.</p> <p>Everyone has some degree of commonly occurring genetic changes that are not associated with cancer. If the test finds this sort of change, it reports a “polymorphism.”</p>	<p>測試結果</p> <p>陰性的</p> <p>真陰性</p> <p>未提供資訊的陰性</p> <p>陽性的</p> <p>意義不明確的變異</p> <p>多態現象 / 多態性 / 多型性</p>	<p>检测结果</p> <p>阴性的</p> <p>真阴性</p> <p>未提供信息的阴性</p> <p>阳性的</p> <p>意义不明确的变异</p> <p>多态性</p>
Trait (n)	A characteristic.	特徵	特征
Tumor (n)	An abnormal growth of cells; a tumor can be benign (not harmful) or malignant (harmful, cancer).	腫瘤	肿瘤

Glossary of Cancer Genetics Terms – Chinese

Term	Definition	Traditional/ 繁體中文	Simplified/ 简体中文
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.	腫瘤檢測 / 腫瘤測試	肿瘤检测 / 肿瘤测试
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.	串錯字	错字
Ultrasound (n)	An imaging method that uses high frequency sound waves to create a picture of something inside the body.	超聲波	超声波
Variant (n)	A version of something that differs from the norm. So, a genetic variant is a change to the usual genetic sequence.	變異	变异

Examples of Genes That are Often Checked in Genetic Testing

Term	Definition	Translation
BRCA 1 BRCA 2	<p>“The Breast Cancer Genes.”</p> <p>The tumor suppressor genes that in mutated form tend to be associated with an increased risk of certain cancers, especially breast and ovarian cancers.</p>	BRCA1 乳腺癌 1 号基因蛋白 BRCA2 乳腺癌 2 号基因蛋白
MLH1 MSH2 MSH6 PMS2	<p>“The Colon Cancer Genes.”</p> <p>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.</p>	MLH1 基因 MSH2 基因 MSH6 基因 PMS2 基因
ATM	<p>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.</p> <p>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.</p>	ATM 肿瘤抑制基因
PALB2	<p>Another gene in which mutations can lead to breast cancer and possibly other cancers.</p>	PALB2 肿瘤抑制基因

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	Traditional 繁體中文	Simplified 簡體中文
Parent	Your mother or father.	父母	父母
Mother	For genetic purposes, the woman whose egg was fertilized and grew to be you.	母親	母亲
Mother-in-law	Your husband or wife's mother.	岳母/婆婆	岳母
Stepmother	Your father's wife who is not your biological mother.	繼母	继母
Adoptive mother	A woman who is not your biological mother but who accepted legal responsibility for and raised you.	養母	养母
Godmother	A woman chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.	教母 契母/ 契媽 (Formal/informal acceptance of kinship in a non-Christian context)	教母
Father	For genetic purposes, the man whose sperm fertilized the egg that grew to be you.	父親	父亲
Father-in-law	Your husband or wife's father.	岳父/公公 老爺 / 家公	岳父
Stepfather	Your mother's husband who is not your biological father.	繼父	继父
Adoptive father	A man who is not your biological father but who accepted legal responsibility for and raised you.	養父	养父
Godfather	A man chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.	契父/ 契爺 (formal/informal acceptance of kinship in a non-Christian context)	教父

Term	Definition	Traditional 繁體中文	Simplified 簡體中文
Aunt	Your mother or father's sister or sister-in-law.	阿姨 (younger aunt) 姨媽 (older aunt)	姨妈/舅妈/姑妈/伯母/叔母
Maternal aunt	Your mother's sister or sister-in-law.	舅母 / 舅媽 (wife of mother's older brother)	姨妈/舅妈
Paternal aunt	Your father's sister or sister-in-law.	姑姑 / 姑媽 (father's older sister/ father's younger sister) 孀孀 (wife of father's younger brother) 伯母 (wife of father's older brother)	姑妈/伯母/叔母
Uncle	Your mother or father's brother or brother-in-law.	小舅 (younger uncle) 大舅 (older unclce)	舅舅/姨夫/伯父/叔叔/姑父
Maternal uncle	Your mother's brother or brother-in-law.	姨丈 (husband of mother's sister)	舅舅/姨夫
Paternal uncle	Your father's brother or brother-in-law.	叔叔/ 叔父(father's younger brother) 伯父 (father's older brother) 姑丈/ 姑爹(husband of father's sister)	伯父/叔叔/姑父

Term	Definition	Traditional 繁體中文	Simplified 簡體中文
Cousin	Usually understood to be a first cousin.	堂(表)兄弟姊妹	堂/表兄弟姐妹
First cousin	Your aunt or uncle's child.		
First cousin once removed	Your aunt or uncle's grandchild or your first cousin's child	堂兄弟姊妹(father's side) 表兄弟姊妹 (mother's side)	堂/表侄子/侄女
Second cousin	The children of first cousins are second cousins to each other.	遠房堂兄弟姊妹 (father's side) 遠房表兄弟姊妹 (mother's side)	远房堂/表侄子/侄女
Grandparent	Your parent's father or mother.	祖父母	外祖父母/祖父母
Grandmother	Your mother or father's mother.	祖母	外祖母/祖母
Maternal grandmother	Your mother's mother.	阿婆	外祖母
Paternal grandmother	Your father's mother.	阿嫲	祖母
Grandfather	Your mother or father's father.	祖父	外祖父/祖父
Maternal grandfather	Your mother's father.	外公/阿公	外祖父
Paternal grandfather	Your father's father.	阿爺/爺爺	祖父
Great aunt	Your mother or father's aunt.	姑婆 / 伯婆 / 妯婆 / 姨婆 / 叔婆	姑/姨祖母
Maternal great aunt	Your mother's aunt.		姨祖母
Paternal great aunt	Your father's aunt.	姑婆 / 伯婆 / 妯婆 / 姨婆 / 叔婆	姑祖母

Term	Definition	Traditional 繁體中文	Simplified 簡體中文
Great uncle	Your father or mother's uncle.	舅公 / 姑公 / 姨公	伯/舅祖父
Maternal great uncle	Your mother's uncle.	伯祖(父) (older brother)	舅祖父
Paternal great uncle	Your father's uncle.	叔公 (younger brother)	伯祖父
Great grandparents	The parents of any of your grandparents.	曾祖父母	曾祖父母
Great grandmother	The mother of any of your grandparents.	曾祖母	曾祖母
Great grandfather	The father of any of your grandparents.	曾祖父	曾祖父
Siblings	The children of your father and mother.	兄弟姊妹	兄弟姐妹
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.	同父異母或同母異父的兄弟姊妹	胞兄弟姐妹
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.	繼兄弟姊妹	继兄弟姐妹
Twins	Two siblings born at the same time.	雙胞胎	双胞胎
Identical twins	Twins who developed from the same egg and sperm, meaning that they are genetically identical. Also called monozygotic twins.	同卵雙胞胎	同卵双胞胎
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.	異卵雙胞胎	异卵双胞胎
Triplets	Three siblings born at the same time.	三胞胎	三胞胎

Term	Definition	Traditional 繁體中文	Simplified 簡體中文
Sister	A sibling who is a girl.	姊妹	姐妹
Sister-in-law	Your brother's wife or your husband's sister.	嫂嫂/嫂子 (older brother's wife) 弟媳 (younger brother's wife)	嫂子/弟妹/大 姑子/小姑子
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.	同父異母姊妹 (same father) 同母異父姐妹 (same mother)	胞姐妹
Stepsister	The daughter of your stepmother or stepfather and therefore not biologically related to you.	繼姊妹	继姐妹
Brother	A sibling who is a boy.	兄弟	兄弟
Brother-in-law	Your sister's husband or your wife's brother.	姊夫 (older sister's) 妹夫 (younger sister's)	姐夫/妹夫/妻 兄/妻弟
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.	同父異母兄弟 (same father) 同母異父兄弟 (same mother)	胞兄弟
Stepbrother	The son of your stepmother or stepfather and therefore not biologically related to you.	繼兄弟	继兄弟
Niece	Your sibling's daughter.	姪女(brother's daughter) 外甥女(sister's daughter)	侄女/外甥女
Nephew	Your sibling's son.	姪子(brother's son) 外甥(sister's son)	侄子/外甥
Spouse	Your husband or wife.	配偶	配偶
Wife	The woman to whom you are married.	妻子	妻子
Husband	The man to whom you are married.	丈夫	丈夫

Term	Definition	Traditional 繁體中文	Simplified 簡體中文
Children	Genetically speaking, the people who are produced from your egg or sperm.	子女	孩子
Daughter	Your child who is a girl.	女兒	女儿
Daughter-in-law	Your son's wife.	媳婦	儿媳
Stepdaughter	Your spouse's daughter who is not your biological child.	繼女	继女
Adoptive daughter	A girl for whom you have accepted legal responsibility and raised even though she is not your biological child.	養女	养女
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.	教女 契女 (Formal/informal acceptance of kinship in a non-Christian context)	教女
Son	Your child who is a boy.	兒子	儿子
Son-in-law	Your daughter's husband.	女婿	女婿
Stepson	Your spouse's son who is not your biological child.	繼子	继子
Adoptive son	A boy for whom you have accepted legal responsibility and raised even though he is not your biological child.	養子	养子
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.	教子 契子/契仔 (Formal/informal acceptance of kinship in a non-Christian context)	教子
Ancestor	A person from whom you are descended, usually more remote than a grandparent.	祖先	祖先
Fiancé(e)	The person whom you have promised to marry. Man: fiancé Woman: fiancée	未婚夫 (妻)	未婚夫/妻

Term	Definition	Traditional 繁體中文	Simplified 簡體中文
Domestic partner	The person with whom you are living and have an intimate relationship, but to whom you are not married.	同居伴侶	同居伴侶
Divorced	Having ended a marriage.	離婚	离婚
Engaged	Having promised to marry someone.	訂婚	订婚
Widow	A woman whose husband has died.	寡婦 / 遺孀	寡妇
Widower	A man whose wife has died.	鰥夫	鳏夫
Relative	A person in your family.	親戚	亲戚
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.	血親	血缘关系
Next of kin	Your closest living blood relation.	近親	直系亲属

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 are 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 are 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).



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

Glossary of Pediatric Genetics Terms

English – Chinese

2022 Edition

Compiled and edited by:
Cynthia E. Roat, MPH

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

This page intentionally left blank.



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.

This page intentionally left blank.

Glossary of Terms

English – Chinese

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

Term	Definition	Traditional/ 繁體中文	Simplified / 简体中文
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.	異常的	异常的
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.”	墮胎	堕胎
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	美國醫學遺傳學和基因組學學院確定的 59 個基因	美国医学遗传学和基因组学学会确定的 59 个基因
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.	適應能力	适应能力
Adoption (n)	The process of accepting legal responsibility for someone else’s child to raise as your own.	領養	收养
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.	美國醫學遺傳學和基因組學學院 (ACMG)	美国医学遗传学和基因组学学会 (ACMG)

Term	Definition	Traditional/ 繁體中文	Simplified / 簡體中文
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.	羊膜穿刺術	羊膜穿刺术
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.	羊水	羊水
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.	無腦畸形	无脑畸形
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.	良性的	良性的
Biochemical testing (n)	Blood tests to identify elevated levels of certain naturally occurring substances/chemicals in the body that are linked to genetic conditions.	生化測試	生化试验
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.	產道	产道
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.”	先天性缺陷	先天性缺陷
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.	驗血	验血
Brace (n)	A medical appliance that provides support to joints or bones, to weak muscles, or to strained ligaments.	支架	支架
Buccal swab (n)	A way to collect DNA from the cells on the inside of a person's cheek.	口腔黏膜測試	口腔黏膜检测

Term	Definition	Traditional/ 繁體中文	Simplified / 簡體中文
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.	基因攜帶者	携带者
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.	細胞	细胞
Cervix (n)	The narrow lower part of the uterus that connects to the vagina.	子宮頸	子宫颈
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.	絨毛膜絨毛取樣 (CVS)	绒毛膜绒毛取样 (CVS)
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.	染色體	染色体
Chromosome test / Karyotype (n)	A test that looks at the number and arrangement of chromosomes in a cell.	染色體檢測 / 核型	染色体检测/核型
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.	兔唇	唇裂
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.	顎裂	腭裂
Clinically significant (adj)	If a test result is “clinically significant,” the result indicates a medical problem that can impact a person’s life.	有臨床意義的/有臨床重要性的	有临床意义的
Cognition (n)	The processes involved in thinking, learning, understanding, and remembering.	認知	认知
Condition (e.g., “genetic condition”) (n)	A long-term medical health issue.	病症 (例如「遺傳病」)	病症 (例如“遗传病”)

Term	Definition	Traditional/ 繁體中文	Simplified / 簡體中文
Consanguinity (n)	When parents are blood relatives to each other.	血緣	血缘
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.	諮詢模式	咨询模式
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.	持續護理模式	持续护理模式
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.	囊性纖維化	囊性纤维化
De novo (adj)	Genetic changes that are not inherited but that occur during cell division.	重新的 / 再次的 / 從頭開始的	重新的
Deleterious mutation/ Disease-causing mutation (n)	A change in a person's DNA that may cause a medical condition.	有害突變 / 致病性突變	有害突变 / 致病性突变
Deletion (n)	Having a section of genetic information (DNA) missing.	缺失	缺失
Developmental assessment (n)	An evaluation of how an individual patient's intellectual, emotional, and social development compare with others of the same age.	發育評估 / 發展評估	发育评估
Diagnostic test (n)	A medical test that determines whether a patient has a particular medical problem. Diagnostic tests are often used when providers have a specific reason to believe that the medical problem may be present. Compare this to screening tests, which may be given routinely even if the provider has no reason to believe the patient has a certain problem. Screening tests often only report whether a patient is at <u>an increased risk</u> for the medical problem in question, whereas diagnostic tests report whether the problem is actually present.	診斷性測試	诊断检测

Term	Definition	Traditional/ 繁體中文	Simplified / 簡體中文
DNA (Deoxyribonucleic Acid) (n)	The molecule that carries the genetic information of a cell. It provides the instructions used in the development, functioning and reproduction of the organism of which it is a part. The DNA is organized into chromosomes.	DNA（脫氧核醣核酸）	DNA（脱氧核糖核酸）
DNA marker (also called a “Genetic marker”) (n)	A readily recognizable genetic trait, gene, or DNA segment.	脫氧核醣核酸標記(也稱為遺傳標記)	脱氧核糖核酸标记（也称为“遗传标记”）
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.	脫氧核醣核酸突變	脱氧核糖核酸突變
DNA sequence (n)	The exact arrangement of the chemicals that make up a section of DNA.	脫氧核醣核酸序列	脱氧核糖核酸序列
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.	脫氧核醣核酸定序	脱氧核糖核酸测序
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 person will be affected with the condition.	顯性的/顯著的	显性的
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.	捐贈卵子或精子	捐赠卵子或精子
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.	唐氏綜合症（三染色體21症）	唐氏综合征（三染色体21症）

Term	Definition	Traditional/ 繁體中文	Simplified / 簡體中文
Duplication (n)	Having an extra section of genetic information (DNA). A <i>duplication</i> occurs when part of a chromosome is copied abnormally, resulting in extra <i>genetic</i> material.	複製	复制
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.	愛德華氏症 (三染色體18症)	爱德华氏症 (三染色体18症)
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.”	卵子	卵子
Embryo (n)	An unborn mammal, between conception and 8 weeks of gestation.	胚胎	胚胎
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.	環境原因或因素	环境原因或因素
Exome sequencing (n)	A genetic test in which a patient’s exons are sequenced and studied.	外顯子組測序	外显子组测序
Exon (n)	The part of the genome that codes for proteins. The majority of deleterious mutations occur in exons.	外顯子	外显子
Fallopian tube (n)	The tube that connects an ovary to the uterus.	輸卵管	输卵管
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.	假陰性	假阴性
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.	假陽性	假阳性
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.	家族性的	家族性的

Term	Definition	Traditional/ 繁體中文	Simplified / 簡體中文
Family history (n)	The medical history of the members of a biological family.	家族史	家族史
Fertilization (n)	The joining of an egg and sperm to create the first cell that will develop into an embryo, then fetus, then baby.	受精	受精
Fetal surgery (n)	Surgery conducted on a fetus while it is still in the uterus.	胎兒手術	胎儿手术
Fetus(n)	An unborn mammal, between 8 weeks of gestation to birth.	胎兒	胎儿
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.	第一孕期篩檢	孕早期筛查
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.	拋幣而定（廣東話俗語「擲銀仔」）	掷硬币
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.	染色體脆弱症	脆性 X 綜合征
Fragment (n)	A small piece; an incomplete part of a whole.	碎片	片段
Gene (n)	A specific sequence of DNA that codes for one or many functions within the cell and body.	基因	基因

Term	Definition	Traditional/ 繁體中文	Simplified / 簡體中文
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.	基因復制	基因拷贝
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.”	正常基因復制	正常基因拷贝
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.”	變異基因復制	变异基因拷贝
Gene deletion (n)	Having a piece of genetic information missing from a gene.	基因刪除	基因缺失
Gene panel testing (n)	Genetic testing that looks at 2 or more specific genes known to be associated with particular conditions.	基因板測試	基因组检测
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.	遺傳變異	遗传变异
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.	一般人口	一般人群

Term	Definition	Traditional/ 繁體中文	Simplified / 簡體中文
Generation (n)	<p>The people who constitute a single step in a line of descent from an ancestor; a group of people born and living more or less at the same time.</p> <p>Example: You, your brothers, and sisters, all your spouses and your cousins are in the same generation. Your parents, your aunts, and uncles and all their spouses form a previous generation. Your grandparents, their siblings, and spouses form an even earlier generation. Your children and nieces and nephews form a later generation.</p>	一代人 / 同一代人	一代人
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).	基因字母表	遗传字母表
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.	遺傳諮詢	遗传咨询
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.	遺傳諮詢師 / 遺傳顧問	遗传咨询师
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.	基因歧視	基因歧视
Genetic factors (n)	Specific aspects of a person’s genetic make-up that influence that person’s health and development.	遺傳因素	遗传因素
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.	遺傳資訊	遗传信息

Term	Definition	Traditional/ 繁體中文	Simplified / 簡體中文
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.	遺傳物質	遗传物质
Genetic predisposition (n)	An increased chance of a person developing a certain trait or disease based on that person's particular genetic makeup.	遺傳傾向性	遗传倾向性
Genetic test (n)	A laboratory test designed to determine if a person has a gene mutation or a typical DNA sequence.	基因檢測	基因检测
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.	單點基因檢測	单点基因检测
Individual gene testing (n)	Individual gene testing: genetic analysis of the entirety of one specific gene.	單一基因檢測/ 個別基因檢測	个体基因检测
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.	基因組測試	基因组检测
Microarray (n)	Microarray: genetic analysis that looks for unexpected numbers of specific genes (two copies of every gene code is usually expected).	微陣列技術/ 基因晶片/ 生物晶片	微阵列
Exome sequencing (n)	Exome testing: a genetic test in which the exons of a patient's genetic code are sequenced and studied.	外顯子組測序	外显子组测序
Genome sequencing (n)	Genome sequencing: a genetic test in which the entire genetic code (the genome) of a patient is mapped out and studied.	基因組測序	基因组测序
Genetic trait (n)	A characteristic within a family that is passed down from parent to child through their DNA.	遺傳特徵	遗传特征
Geneticist (n)	A doctor or scientist who studies genetics.	遺傳學家	遗传学家
Genetics (adj)	The science of how an organism's genes interact with the environment to produce certain traits.	遺傳學的	遗传学的

Term	Definition	Traditional/ 繁體中文	Simplified / 簡體中文
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.	遺傳學	遗传学
Genome (n)	The entire genetic code of an individual.	基因組	基因组
Genome sequencing (n)	A genetic test in which the entire genetic code of a patient is mapped out and studied.	基因組測序	基因组测序
Germline testing (n)	<p>Germline testing refers to the analysis of a person’s DNA, which he or she inherited from his or her parents.</p> <p>In biology and genetics, the germline is the group of cells that will pass on the genetic material to children, in other words, the cells from which the eggs and sperm come.</p>	殖源基因檢查	生殖系检测
Guardianship (n)	A legal arrangement, supervised by a court, in which one person takes responsibility for a minor or an adult with a disability.	監護權	监护权
Helix (n)	Something that is spiral in form. DNA is often described as being a “double helix.”	螺旋	螺旋
Hereditary (adj)	Passed down from parent to child.	遺傳性的	遗传性的
Hereditary material (n)	Genetic material that is passed down from parent to child.	遺傳物質	遗传物质
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.	亨廷頓氏病	亨廷顿氏病
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.	個人化教育計畫 (IEP)	个性化教育计划 (IEP)

Term	Definition	Traditional/ 繁體中文	Simplified / 簡體中文
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.	體外人工受精 (IVF)	体外人工受精 (IVF)
Individual gene testing (n)	Genetic analysis of the entirety of one specific gene.	個人基因檢測	个体基因检测
Infertility (n)	The inability to have children.	不孕症	不孕症
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.	知情同意書	知情同意书
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.	遺傳模式	遗传模式
Inherited (adj)	Passed down from parent to child.	遺傳的	遗传的
Insertion (n)	An extra segment of DNA added in at a place where it is not usually found.	插入	插入
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.	智力障礙	智力障碍
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.	克萊恩費爾特氏綜合徵 / 克氏綜合徵 / 先天性睪丸發育不全 (47, XXY)	克兰费尔特综合征 (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.	林奇氏症候群	林奇综合症

Term	Definition	Traditional/ 繁體中文	Simplified / 簡體中文
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.	標記染色體	标记染色体
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.	孕婦血清篩檢 (MSS 或產婦血清甲胎蛋白測試)	母体血清篩查 (MSS 或母体血清甲胎蛋白检测)
Metastasis (n)	The spreading of cancer from one organ to another.	轉移	转移
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.	微陣列 / 基因晶片 / 生物晶片	微阵列
Miscarriage (n)	The spontaneous (not intentional) loss of a pregnancy. See “abortion.” Also called pregnancy loss.	流產	流产
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.	分子	分子
Mosaicism (n)	A condition in which some, but not all, cells in a sample show a genetic difference.	鑲嵌現象	镶嵌现象
Motor Abilities (n)	The ability to move and use one’s muscles.	運動技能 / 肢體功能 / 動作技能	运动能力
Multifactorial (adj)	Due to a combination of genetic and non-genetic (environmental, hormonal, etc.) risk factors that act together to determine risk.	多因素的	多因素的

Term	Definition	Traditional/ 繁體中文	Simplified / 簡體中文
Mutation (n)	A change in a gene, usually deleterious. See “genetic variant.”	突變	突变
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.	非侵入性 產前篩檢 (NIPT)	无创产前检测 (NIPT)
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.	頸背增厚	颈部增厚
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.	開放性神 經管缺陷 (ONTD)	开放性神经管 缺陷 (ONTD)
Ova (n)	See “egg.”	卵子	卵子
Ovary (n)	The organ in a woman that stores and releases eggs. There are normally two.	卵巢	卵巢
Packets of genetic information (n)	A phrase genetic counselors use to describe genes or chromosomes.	基因資訊 (封包)	遗传信息包
Pathogenic (adj)	Disease causing.	致病的	致病的
Pedigree (n)	A family tree that can be used to trace the inheritance of specific genetic traits.	血統/血系	家谱
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.	外顯率	外显率

Term	Definition	Traditional/ 繁體中文	Simplified / 簡體中文
Placenta (n)	The organ that develops together with an embryo in a pregnant woman's uterus to nourish the embryo through the umbilical cord.	胎盤	胎盘
Presymptomatic (adj)	Before symptoms appear.	症狀前的	症状前的
Prognosis (n)	The most likely outcome of a disease process.	病程預測 / 預後	预后
Protein (n)	A molecule made up of chains of <i>amino acids</i> . Proteins do most of the work in cells and are required for the structure, function, and regulation of the body's tissues and organs. Genes determine how specific amino acids are put together to form a specific protein.	蛋白質	蛋白质
Random (adj)	Happening in an unpredictable way.	隨機的	随机的
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.	隱性的	隐性的
Red flag (n)	A warning sign or a clue.	警告信號	危险信号
Replicate (v)	To copy.	複製	复制
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.	生產病史	生殖史
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.	環狀染色體	环状染色体

Term	Definition	Traditional/ 繁體中文	Simplified / 簡體中文
Risk (n) At risk (adj) At average risk (adj) At high risk (adj) At higher risk than “X” (adj) Risk factor (n)	<p>The chance that something will happen; in the case of prenatal genetics, the chance that the child will have a genetic condition.</p> <p>Has some possibility of having a genetic condition.</p> <p>Has the same possibility of having a genetic condition as the general population.</p> <p>Has a greater possibility of having a genetic condition than the general population.</p> <p>Has a greater possibility of having a genetic condition than “X.”</p> <p>A circumstance that increases the risk of having a genetic condition.</p>	<p>風險</p> <p>有風險的</p> <p>一般風險的</p> <p>高風險的</p> <p>風險高於「X」的</p> <p>風險因素</p>	<p>风险</p> <p>有风险的</p> <p>平均风险的</p> <p>高风险的</p> <p>风险高于“X”的</p> <p>风险因素</p>
Runs in the family	Passed down from parents to children to grandchildren.	家族遺傳	家族遗传
Saliva (spit) sample (n)	A way to collect DNA from the cells in saliva (spit).	唾液（口水） 測試	唾液（口水） 样本
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.	篩檢	筛查
Screening test (n)	<p>A test that looks to detect a particular health problem, even if a patient has no symptoms of that particular condition.</p> <p>Prenatal screening tests aren’t looking for genetic changes. They are designed to look at hormones, chemicals, and other things that indicate a fetus might have a higher risk of a certain condition. They are not DNA results, do not give a definite answer about whether someone is affected, and do not provide a firm diagnosis. They are usually reported as a risk number (e.g., a 1 in ____ chance), and if this number is above a certain cut-off point, the result will be categorized as “screen positive” or “screen negative.”</p>	篩檢測試	筛选检测

Term	Definition	Traditional/ 繁體中文	Simplified / 簡體中文
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.	精液	精液
Seminal fluid (n)	See “semen.”	精液	精液
Sensory abilities (n)	The ability to see, hear, touch, taste, and smell.	感官能力 / 感覺能力	感官能力
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.	鎌狀細胞疾病	镰状细胞病
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.	單點分析	单点分析 (n)
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.	體細胞遺傳變化	体细胞遗传变化
Special education (n)	Education that is tailored to individuals with developmental delays or other learning disabilities.	特殊教育	特殊教育
Sperm (n)	The reproductive cells of the man. When sperm fertilize a woman’s egg, a baby develops.	精子	精子
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.	捐精	捐精
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.	脊柱裂	脊柱裂

Term	Definition	Traditional/ 繁體中文	Simplified / 簡體中文
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.	有統計意義的	有统计学意义的
Supportive seating (n)	Seating, such as a wheelchair, that helps an individual function in their environment and that prevents worsening of their condition.	支援性座椅/ 承托座椅	支持座位
Teratogen (n)	Something that has the potential to disturb the normal development of an embryo or fetus.	畸胎原	致畸原
Termination of pregnancy (n)	See “abortion.”	終止妊娠	终止妊娠

Term	Definition	Traditional/ 繁體中文	Simplified / 簡體中文
<p>Test result (n) Negative (adj)</p> <p>True negative (n)</p> <p>Positive (adj)</p> <p>Variant of uncertain significance (n)</p> <p>Polymorphism (n)</p>	<p>A negative result on a genetic test means that the laboratory did not find the specific genetic change that the test was designed to identify.</p> <p>When a patient is being tested to see if he or she has a specific genetic change that is present in other family members, a negative result can be considered a “true negative.” This means that instead of having an increased risk for cancer like other family members, they have the same risk as everyone else in the general population.</p> <p>A positive result on a genetic test means that the laboratory did find a specific genetic change that is associated with a hereditary cancer syndrome (a deleterious mutation). In cancer genetics, that means that the patient has an increased risk of getting the cancer(s) associated with the gene.</p> <p>If the laboratory finds a genetic change for which currently there is not enough information to know if this change is problematic or not, it reports a “variant of uncertain significance.” Basically, this means that the laboratory found a genetic change, but they don’t know what it means. All of these variants will eventually be reclassified as either a “positive” or “negative” result. Most become “negative” and are thought to represent natural variation between individuals.</p> <p>Everyone has some degree of commonly occurring genetic changes that are not associated with cancer. If the test finds this sort of change, it reports a “polymorphism.”</p>	<p>測試結果 陰性的</p> <p>真陰性</p> <p>陽性的</p> <p>意義不明確 的變異</p> <p>多態現 象 / 多態性 / 多型性</p>	<p>检测结果 阴性的</p> <p>真阴性</p> <p>阳性的</p> <p>意义不明确 的变异</p> <p>多态性</p>
Testes (n)	The organs in a man that create and release sperm. Also called “testicles.” Singular testis.	睪丸	睾丸
Toxic exposure (n)	Contact with something that is harmful or poisonous.	毒性暴露/接觸 有毒物	毒性暴露
Trait (n)	<p>A characteristic of a person.</p> <p>In genetics, traits are aspects of a person defined or influenced by their genetic code, e.g., eye color, blood type, risk for certain diseases.</p>	特徵	特征

Term	Definition	Traditional/ 繁體中文	Simplified / 簡體中文
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.	移位 / 轉移	易位
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.	串錯字	錯字
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.	超聲波	超聲波
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.”	子宮	子宮
Vagina (n)	The tube-like part of the female reproductive system that extends from the cervix to the outside of the body.	陰道	阴道
Variant (n)	A version of something that differs from the norm. So, a genetic variant is a change to the usual genetic sequence.	變異	变异
Vas deferens (n)	The tubes that lead from the testes to the urethra, through which sperm is ejaculated.	輸精管	输精管
X-linked (adj)	A trait that is influenced or determined by a gene on the “X” chromosome.	X 染色體串聯的	X 连锁的

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	Traditional 繁體中文	Simplified 簡體中文
Parent	Your mother or father.	父母	父母
Mother	For genetic purposes, the woman whose egg was fertilized and grew to be you.	母親	母亲
Mother-in-law	Your husband or wife's mother.	岳母/婆婆	岳母
Stepmother	Your father's wife who is not your biological mother.	繼母	继母
Adoptive mother	A woman who is not your biological mother but who accepted legal responsibility for and raised you.	養母	养母
Godmother	A woman chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.	教母 契母/ 契媽 (Formal/informal acceptance of kinship in a non-Christian context)	教母
Father	For genetic purposes, the man whose sperm fertilized the egg that grew to be you.	父親	父亲
Father-in-law	Your husband or wife's father.	岳父/公公 老爺 / 家公	岳父
Stepfather	Your mother's husband who is not your biological father.	繼父	继父
Adoptive father	A man who is not your biological father but who accepted legal responsibility for and raised you.	養父	养父
Godfather	A man chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.	契父/ 契爺 (formal/informal acceptance of kinship in a non-Christian context)	教父

Term	Definition	Traditional 繁体中文	Simplified 简体中文
<p>Aunt</p> <p>Maternal aunt</p> <p>Paternal aunt</p>	<p>Your mother or father's sister or sister-in-law.</p> <p>Your mother's sister or sister-in-law.</p> <p>Your father's sister or sister-in-law.</p>	<p>阿姨 (younger aunt) 姨媽 (older aunt)</p> <p>舅母 / 舅媽 (wife of mother's older brother)</p> <p>姑姑 / 姑媽 (father's older sister/ father's younger sister) 孀孀 (wife of father's younger brother) 伯母 (wife of father's older brother)</p>	<p>姨妈/舅妈/姑 妈/伯母/叔母</p> <p>姨妈/舅妈</p> <p>姑妈/伯母/叔 母</p>
<p>Uncle</p> <p>Maternal uncle</p> <p>Paternal uncle</p>	<p>Your mother or father's brother or brother-in-law.</p> <p>Your mother's brother or brother-in-law.</p> <p>Your father's brother or brother-in-law.</p>	<p>小舅 (younger uncle) 大舅 (older unclce)</p> <p>姨丈 (husband of mother's sister)</p> <p>叔叔/ 叔父 (father's younger brother) 伯父 (father's older brother) 姑丈/ 姑爹 (husband of father's sister)</p>	<p>舅舅/姨夫/伯 父/叔叔/姑父</p> <p>舅舅/姨夫</p> <p>伯父/叔叔/姑 父</p>

Term	Definition	Traditional 繁體中文	Simplified 简体中文
Cousin First cousin	Usually understood to be a first cousin. Your aunt or uncle's child.	堂(表)兄弟姊妹	堂/表兄弟姐妹
First cousin once removed	Your aunt or uncle's grandchild or your first cousin's child	堂兄弟姊妹 (father's side) 表兄弟姊妹 (mother's side)	堂/表侄子/侄女
Second cousin	The children of first cousins are second cousins to each other.	遠房堂兄弟姊妹 (father's side) 遠房表兄弟姊妹 (mother's side)	远房堂/表侄子/侄女
Grandparent	Your parent's father or mother.	祖父母	外祖父母/ 祖父母
Grandmother	Your mother or father's mother.	祖母	外祖母/祖母
Maternal grandmother	Your mother's mother.	阿婆	外祖母
Paternal grandmother	Your father's mother.	阿嫲	祖母
Grandfather	Your mother or father's father.	祖父	外祖父/祖父
Maternal grandfather	Your mother's father.	外公/阿公	外祖父
Paternal grandfather	Your father's father.	阿爺/爺爺	祖父
Great aunt	Your mother or father's aunt.	姑婆 / 伯婆 / 妯娌 婆 / 姨婆 / 叔婆	姑/姨祖母
Maternal great aunt	Your mother's aunt.	姑婆 / 伯婆 / 妯娌 婆 / 姨婆 / 叔婆	姨祖母
Paternal great aunt	Your father's aunt.	姑婆 / 伯婆 / 妯娌 婆 / 姨婆 / 叔婆	姑祖母
Great uncle	Your father or mother's uncle.	舅公 / 姑公 / 姨公	伯/舅祖父
Maternal great uncle	Your mother's uncle.	伯祖(父)(older brother)	舅祖父
Paternal great uncle	Your father's uncle.	叔公(younger brother)	伯祖父

Term	Definition	Traditional 繁體中文	Simplified 簡體中文
Great grandparents	The parents of any of your grandparents.	曾祖父母	曾祖父母
Great grandmother	The mother of any of your grandparents.	曾祖母	曾祖母
Great grandfather	The father of any of your grandparents.	曾祖父	曾祖父
Siblings	The children of your father and mother.	兄弟姊妹	兄弟姐妹
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.	同父異母或同母異父的兄弟姊妹	胞兄弟姐妹
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.	繼兄弟姐妹	继兄弟姐妹
Twins	Two siblings born at the same time.	雙胞胎	双胞胎
Identical twins	Twins who developed from the same egg and sperm, meaning that they are genetically identical. Also called monozygotic twins.	同卵雙胞胎	同卵双胞胎
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.	異卵雙胞胎	异卵双胞胎
Triplets	Three siblings born at the same time.	三胞胎	三胞胎
Sister	A sibling who is a girl.	姊妹	姐妹
Sister-in-law	Your brother's wife or your husband's sister.	嫂嫂/嫂子 (older brother's wife) 弟媳(younger brother's wife)	嫂子/弟妹/大姑子/小姑子
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.	同父異母姊妹 (same father) 同母異父姐妹 (same mother)	胞姐妹
Stepsister	The daughter of your stepmother or stepfather and therefore not biologically related to you.	繼姊妹	继姐妹

Term	Definition	Traditional 繁體中文	Simplified 简体中文
Brother	A sibling who is a boy.	兄弟	兄弟
Brother-in-law	Your sister's husband or your wife's brother.	姊夫(older sister's) 妹夫 (younger sister's)	姐夫/妹夫/妻 兄/妻弟
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.	同父異母兄弟 (same father) 同母異父兄弟 (same mother)	胞兄弟
Stepbrother	The son of your stepmother or stepfather and therefore not biologically related to you.	繼兄弟	继兄弟
Niece	Your sibling's daughter.	姪女(brother's daughter) 外甥女(sister's daughter)	侄女/外甥女
Nephew	Your sibling's son.	姪子(brother's son) 外甥(sister's son)	侄子/外甥
Spouse	Your husband or wife.	配偶	配偶
Wife	The woman to whom you are married.	妻子	妻子
Husband	The man to whom you are married.	丈夫	丈夫
Children	Genetically speaking, the people who are produced from your egg or sperm.	子女	孩子

Term	Definition	Traditional 繁體中文	Simplified 簡體中文
Daughter	Your child who is a girl.	女兒	女儿
Daughter-in-law	Your son's wife.	媳婦	儿媳
Stepdaughter	Your spouse's daughter who is not your biological child.	繼女	继女
Adoptive daughter	A girl for whom you have accepted legal responsibility and raised even though she is not your biological child.	養女	养女
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.	教女 契女 (formal/informal acceptance of kinship in a non-Christian context)	教女
Son	Your child who is a boy.	兒子	儿子
Son-in-law	Your daughter's husband.	女婿	女婿
Stepson	Your spouse's son who is not your biological child.	繼子	继子
Adoptive son	A boy for whom you have accepted legal responsibility and raised even though he is not your biological child.	養子	养子
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.	教子 契子/契仔 (Formal/informal acceptance of kinship in a non-Christian context)	教子
Ancestor	A person from whom you are descended, usually more remote than a grandparent.	祖先	祖先
Fiancé(e)	The person whom you have promised to marry. Man: fiancé Woman: fiancée	未婚夫(妻)	未婚夫/妻

Term	Definition	Traditional 繁體中文	Simplified 簡體中文
Domestic partner	The person with whom you are living and have an intimate relationship, but to whom you are not married.	同居伴侶	同居伴侶
Divorced	Having ended a marriage.	離婚	离婚
Engaged	Having promised to marry someone.	訂婚	订婚
Widow	A woman whose husband has died.	寡婦 / 遺孀	寡妇
Widower	A man whose wife has died.	鰥夫	鳏夫
Relative	A person in your family.	親戚	亲戚
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.	血親	血缘关系
Next of kin	Your closest living blood relation.	近親	直系亲属

This page intentionally left blank.



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

Glossary of Prenatal Genetics Terms

English – Chinese

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 – Chinese

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

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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.	異常的	异常的
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.”	墮胎	堕胎
Adoption (n)	The process of legally taking someone else’s child into your family to raise as your own child.	領養	收养
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.	羊膜穿刺術	羊膜穿刺术
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.	羊水	羊水
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.	無腦畸形	无脑畸形
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.”	良性的	良性的
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.	產道	产道

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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.”	先天性缺陷	先天性缺陷
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.	驗血	验血
Buccal swab (n)	A way to collect DNA from the cells on the inside of a person's cheek.	口腔黏膜測試	口腔黏膜检测
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.	基因攜帶者	携带者
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.	細胞	细胞
Cervix (n)	The narrow lower part of the uterus that connects to the vagina.	子宮頸	子宫颈
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.	絨毛膜絨毛取樣 (CVS)	绒毛膜绒毛取样 (CVS)
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.	染色體	染色体
Chromosome test / Karyotype (n)	A test that looks at the number and types of chromosomes in a cell.	染色體檢測/核型	染色体检测/核型

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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.	兔唇	唇裂
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.	顎裂	腭裂
Clinically significant (adj)	If a test result is “clinically significant,” the result indicates a medical problem that can impact a person’s life.	有臨床意義的 / 有臨床重要性的	有临床意义的
Condition (e.g., “genetic condition”) (n)	A long-term medical health issue.	病症（例如「遺傳病」）	疾病（例如“遗传病”）
Consanguinity (n)	When parents are blood relatives to each other.	血緣	血缘
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.	囊性纖維化	囊性纤维化
Deleterious mutation/ Disease-causing mutation (n)	A change in a person’s DNA that may cause a medical condition.	有害突變 / 致病性突變	有害突变 / 致病突变
Deletion (n)	Having a section of genetic information (DNA) missing.	缺失	缺失
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 <u>an increased risk</u> for the medical problem in question, whereas diagnostic tests report whether the problem is actually present.	診斷性測試	诊断检测

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
DNA (Deoxyribonucleic Acid) (n)	<p>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.</p> <p>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.</p> <p>Individual sections of DNA that code for specific traits/functions are called genes.</p>	DNA (脫氧核糖核酸)	DNA (脱氧核糖核酸)
DNA marker (also called a "Genetic marker") (n)	A readily recognizable genetic trait, gene, or DNA segment.	脫氧核糖核酸標記 (也稱為遺傳標記)	脱氧核糖核酸标记 (也称为“遗传标记”)
DNA mutation (n)	<p>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.</p> <p>Mutations or variants are often compared to misspelled words because chemicals that make up the DNA sequence are not in the expected order.</p>	脫氧核糖核酸突變	脱氧核糖核酸突變
DNA sequence (n)	The exact arrangement of the chemicals that make up a section of DNA.	脫氧核糖核酸序列	脱氧核糖核酸序列
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.	脫氧核糖核酸定序	脱氧核糖核酸测序
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 person will be affected with the condition.	顯性的/顯著的	显性的
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.	捐贈卵子或精子	捐赠卵子或精子

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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.	唐氏綜合症 (三染色體21症)	唐氏綜合症 (三染色体 21 症)
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.	複製	复制
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.	愛德華氏症 (三染色體18症)	爱德华氏症 (三染色体 18 症)
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.”	卵子	卵子
Embryo (n)	An unborn mammal, between conception and 8 weeks of gestation.	胚胎	胚胎
Fallopian tube (n)	The tube that connects an ovary to the uterus.	輸卵管	输卵管
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.	假陰性	假阴性
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.	假陽性	假阳性
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.	家族性的	家族性的
Family history (n)	The medical history of the members of a biological family.	家族史	家族史

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
Fertilization (n)	The joining of an egg and sperm to create the first cell that will develop into an embryo, then fetus, then baby.	受精	受精
Fetal surgery (n)	Surgery conducted on a fetus while it is still in the uterus.	胎兒手術	胎儿手术
Fetus (n)	An unborn mammal, between 8 weeks of gestation to birth.	胎兒	胎儿
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.	懷孕第一期篩檢	孕早期筛查
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.	拋幣而定（廣東話俗語「擲銀仔」）	掷硬币
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.	染色體脆弱症	脆性 X 綜合征
Fragment (n)	A small piece; an incomplete part of a whole.	碎片	片段
Gene (n)	A specific sequence of DNA that codes for one or many functions within the cell and body.	基因	基因
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.	一般人口	一般人群

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
Generation (n)	<p>The people who constitute a single step in a line of descent from an ancestor; a group of people born and living more or less at the same time.</p> <p>Example: You, your brothers, and sisters, all your spouses and your cousins are in the same generation. Your parents, your aunts, and uncles and all their spouses form a previous generation. Your grandparents, their siblings, and spouses form an even earlier generation. Your children and nieces and nephews form a later generation.</p>	一代人 / 同一代人	一代人
Genetics (adj)	The science of how an organism's genes interact with the environment to produce certain traits.	遺傳學的	遗传学的
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.	遺傳諮詢	遗传咨询
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.	遺傳諮詢實習生	遗传咨询实习生
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.	遺傳諮詢師 / 遺傳顧問	遗传咨询师
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.	基因歧視	基因歧视
Genetic factors (n)	Specific aspects of a person's genetic make-up that influence that person's health and development.	遺傳因素	遗传因素

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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.	遺傳資訊	遗传信息
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.	遺傳物質	遗传物质
Genetic predisposition (n)	An increased chance of a person developing a certain trait or disease based on that person's particular genetic makeup.	遺傳傾向性	遗传倾向性
Genetic test (n)	A laboratory test designed to determine if a person has a change to their DNA.	基因檢測	基因检测
Genetic trait (n)	A characteristic within a family that is passed down from parent to child through their DNA.	遺傳特徵	遗传特征
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.	遺傳變異	遗传变异
Geneticist (n)	A doctor or scientist who studies genetics.	遺傳學家	遗传学家
Hereditary (adj)	Passed down from parent to child.	遺傳性的	遗传性的
Hereditary material (n)	Genetic material that is passed down from parent to child.	遺傳物質	遗传物质
In Vitro Fertilization (IVF) (n)	<p>The fertilization of an egg by a sperm outside of a woman's body.</p> <p>The process involves extracting eggs from a woman's ovaries, collecting sperm from a man, and combining a sperm and egg in a laboratory dish. The resulting fertilized egg is usually then implanted in a woman's uterus so that it can develop into a baby.</p>	體外人工受精 (IVF)	体外人工受精 (IVF)
Infertility (n)	The inability to have children.	不孕症	不孕症

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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.	知情同意書	知情同意书
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.	遺傳模式	遗传模式
Inherited (adj)	Passed down from parent to child.	遺傳的	遗传的
Insertion (n)	Having an extra segment of DNA added in at a place where it is not usually found.	插入	插入
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.	智力障礙	智力障碍
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.	克萊恩費爾特氏綜合徵 / 克氏綜合徵 / 先天性睪丸發育不全 (47, XXY)	克兰费尔特综合征 (47, XXY)
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.	標記染色體	标记染色体
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.	孕婦血清篩檢 (MSS 或產婦血清甲胎蛋白測試)	母体血清筛查 (MSS 或母体血清甲胎蛋白检测)

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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.	微陣列技術 / 基因晶片 / 生物晶片	微阵列
Miscarriage (n)	The spontaneous (not intentional) loss of a pregnancy. See “abortion.” Also called pregnancy loss.	流產	流产
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.	分子	分子
Mosaicism (n)	A condition in which some, but not all, cells in a sample show a genetic difference.	鑲嵌現象	镶嵌现象
Multifactorial (adj)	Due to a combination of genetic and non-genetic (environmental, hormonal, etc.) risk factors that act together to determine risk.	多因素的	多因素的
Mutation (n)	A change in a gene, usually deleterious. See “genetic variant.” In Spanish, the word is sometimes confused with “mutilation.”	突變	突变
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.	非侵入性產前篩檢 (NIPT)	无创产前检测 (NIPT)

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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.	頸背增厚	颈部增厚
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.	開放性神經管缺陷 (ONTD)	开放性神经管缺陷 (ONTD)
Ova (n)	See “egg.”	卵子	卵子
Ovary (n)	The organ in a woman that stores and releases eggs. There are normally two.	卵巢	卵巢
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.	外顯率	外显率
Placenta (n)	The organ that develops together with an embryo in a pregnant woman’s uterus to nourish the embryo through the umbilical cord.	胎盤	胎盘
Presymptomatic (adj)	Before symptoms appear.	症狀前的	症状前的
Prognosis (n)	The most likely outcome of a disease process.	病程預測/預後	预后
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.	蛋白質	蛋白质
Random (adj)	Happening in an unpredictable way.	隨機的	随机的

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
Recessive (adj)	<p>A genetic trait in which both copies of a gene need to be altered for the trait to be expressed; recessive traits are typically inherited from both parents, each one contributing one copy of the gene in question.</p> <p>In prenatal genetics, recessive inheritance refers to conditions in which both copies of a particular gene pair need to be altered to develop a specific disease or condition.</p>	隱性的	隐性的
Red flag (n)	A warning sign or a clue.	警告信號	危险信号
Replicate (v)	To copy.	複製	复制
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.	生產病史	生殖史
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.	環狀染色體	环状染色体

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
Risk (n) At risk (adj) At average risk (adj) At high risk (adj) At higher risk than “X” (adj) Risk factor (n)	<p>The chance that something will happen; in the case of prenatal genetics, the chance that the child will have a genetic condition.</p> <p>Has some possibility of having a genetic condition.</p> <p>Has the same possibility of having a genetic condition as the general population.</p> <p>Has a greater possibility of having a genetic condition than the general population.</p> <p>Has a greater possibility of having a genetic condition than “X.”</p> <p>A circumstance that increases the risk of having a genetic condition.</p>	<p>風險</p> <p>有風險的</p> <p>一般風險的</p> <p>高風險的</p> <p>風險高於「X」的</p> <p>風險因素</p>	<p>风险</p> <p>有风险的</p> <p>平均风险的</p> <p>高风险的</p> <p>风险高于“X”的</p> <p>风险因素</p>
Runs in the family (e.g., sickle cell disease runs in the family) (v)	<p>When a certain condition or trait seems to appear in many members of a single family</p>	<p>家族遺傳</p>	<p>家族遗传 (例如镰状细胞病在家族中传播)</p>
Screen (v)	<p>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.</p>	<p>篩檢</p>	<p>筛查</p>

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
Screening test (n)	<p>A test that looks to detect a particular health problem, even if a patient has no symptoms of that particular condition.</p> <p>Prenatal screening tests aren't looking for genetic changes. They are designed to look at hormones, chemicals, other things that indicate a fetus might have a higher risk of a certain condition. They are not DNA results, do not give a definite answer about whether someone is affected, and do not provide a firm diagnosis. They are usually reported as a risk number (e.g., 1 in ____ chance), and if this number is above a certain cut-off point, the result will be categorized as 'screen positive' or 'screen negative'.</p>	篩檢測試	筛选检测
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.	精液	精液
Seminal fluid (n)	See "semen."	精液	精液
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.	鎌狀細胞疾病	镰状细胞病
Sperm (n)	The reproductive cells of the man. When sperm fertilize a woman's egg, a baby develops.	精子	精子
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.	捐精	捐精
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.	脊柱裂	脊柱裂
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.	有統計意義的	有统计学意义的

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
Teratogen (n)	Something that has the potential to disturb the normal development of an embryo or fetus.	畸胎原	致畸原
Termination of pregnancy (n)	See “abortion.”	終止妊娠	终止妊娠
Test results (n)		測試結果	检测结果
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.	陰性的	阴性的
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.	陽性的	阳性的
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.	意義不明確的變異	意义不明确的变
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.”	多元性	多态性
Testes (n)	The organs in a man that create and release sperm. Also called “testicles.” Singular testis.	睪丸	睾丸
Toxic exposure (n)	Contact with something that is harmful or poisonous.	毒性暴露 / 接觸有毒物	毒性暴露

Term	Definition	Traditional/ 繁體中文	Simplified/ 簡體中文
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.	特徵	特征
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.	移位/轉移	易位
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.	串錯字	错字
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	超聲波	超声波
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.”	子宮	子宫
Vagina (n)	The tube-like part of the female reproductive system that extends from the cervix to the outside of the body.	陰道	阴道
Variant (n)	A version of something that differs from the norm. So, a genetic variant is a change to the usual genetic sequence.	變異	变异
Vas deferens (n)	The tubes that lead from the testes to the urethra, through which sperm is ejaculated.	輸精管	输精管
X-linked (adj)	A trait that is influenced or determined by a gene on the “X” chromosome.	X 染色體串聯的	X 连锁的

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	Traditional 繁體中文	Simplified 简体中文
Parent	Your mother or father.	父母	父母
Mother	For genetic purposes, the woman whose egg was fertilized and grew to be you.	母親	母亲
Mother-in-law	Your husband or wife's mother.	岳母/婆婆	岳母
Stepmother	Your father's wife who is not your biological mother.	繼母	继母
Adoptive mother	A woman who is not your biological mother but who accepted legal responsibility for and raised you.	養母	养母
Godmother	A woman chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.	教母 契母/ 契媽 (Formal/informal acceptance of kinship in a non-Christian context)	教母
Father	For genetic purposes, the man whose sperm fertilized the egg that grew to be you.	父親	父亲
Father-in-law	Your husband or wife's father.	岳父/公公 老爺 / 家公	岳父
Stepfather	Your mother's husband who is not your biological father.	繼父	继父
Adoptive father	A man who is not your biological father but who accepted legal responsibility for and raised you.	養父	养父
Godfather	A man chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.	契父/ 契爺 (formal/informal acceptance of kinship in a non-Christian context)	教父

Term	Definition	Traditional 繁体中文	Simplified 简体中文
Aunt	Your mother or father's sister or sister-in-law.	阿姨 (younger aunt) 姨媽 (older aunt)	姨妈/舅妈/姑妈/伯母/叔母
Maternal aunt	Your mother's sister or sister-in-law.	舅母 / 舅媽 (wife of mother's older brother)	姨妈/舅妈
Paternal aunt	Your father's sister or sister-in-law.	姑姑 / 姑媽 (father's older sister/ father's younger sister) 孀孀 (wife of father's younger brother) 伯母 (wife of father's older brother)	姑妈/伯母/叔母
Uncle	Your mother or father's brother or brother-in-law.	小舅 (younger uncle) 大舅 (older unclce)	舅舅/姨夫/伯父/叔叔/姑父
Maternal uncle	Your mother's brother or brother-in-law.	姨丈 (husband of mother's sister)	舅舅/姨夫
Paternal uncle	Your father's brother or brother-in-law.	叔叔/ 叔父 (father's younger brother) 伯父 (father's older brother) 姑丈/ 姑爹 (husband of father's sister)	伯父/叔叔/姑父

Term	Definition	Traditional 繁體中文	Simplified 简体中文
Cousin First cousin	Usually understood to be a first cousin. Your aunt or uncle's child.	堂(表)兄弟姊妹	堂/表兄弟姐妹
First cousin once removed	Your aunt or uncle's grandchild or your first cousin's child	堂兄弟姊妹 (father's side) 表兄弟姊妹 (mother's side)	堂/表侄子/侄女
Second cousin	The children of first cousins are second cousins to each other.	遠房堂兄弟姊妹 (father's side) 遠房表兄弟姊妹 (mother's side)	远房堂/表侄子/侄女
Grandparent	Your parent's father or mother.	祖父母	外祖父母/ 祖父母
Grandmother	Your mother or father's mother.	祖母	外祖母/祖母
Maternal grandmother	Your mother's mother.	阿婆	外祖母
Paternal grandmother	Your father's mother.	阿嫲	祖母
Grandfather	Your mother or father's father.	祖父	外祖父/祖父
Maternal grandfather	Your mother's father.	外公/阿公	外祖父
Paternal grandfather	Your father's father.	阿爺/爺爺	祖父
Great aunt	Your mother or father's aunt.	姑婆 / 伯婆 / 妯娌 婆 / 姨婆 / 叔婆	姑/姨祖母
Maternal great aunt	Your mother's aunt.	姑婆 / 伯婆 / 妯娌 婆 / 姨婆 / 叔婆	姨祖母
Paternal great aunt	Your father's aunt.	姑婆 / 伯婆 / 妯娌 婆 / 姨婆 / 叔婆	姑祖母
Great uncle	Your father or mother's uncle.	舅公 / 姑公 / 姨公	伯/舅祖父
Maternal great uncle	Your mother's uncle.	伯祖(父)(older brother)	舅祖父
Paternal great uncle	Your father's uncle.	叔公(younger brother)	伯祖父

Term	Definition	Traditional 繁體中文	Simplified 簡體中文
Great grandparents	The parents of any of your grandparents.	曾祖父母	曾祖父母
Great grandmother	The mother of any of your grandparents.	曾祖母	曾祖母
Great grandfather	The father of any of your grandparents.	曾祖父	曾祖父
Siblings	The children of your father and mother.	兄弟姊妹	兄弟姐妹
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.	同父異母或同母異父的兄弟姊妹	胞兄弟姐妹
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.	繼兄弟姊妹	继兄弟姐妹
Twins	Two siblings born at the same time.	雙胞胎	双胞胎
Identical twins	Twins who developed from the same egg and sperm, meaning that they are genetically identical. Also called monozygotic twins.	同卵雙胞胎	同卵双胞胎
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.	異卵雙胞胎	异卵双胞胎
Triplets	Three siblings born at the same time.	三胞胎	三胞胎
Sister	A sibling who is a girl.	姊妹	姐妹
Sister-in-law	Your brother's wife or your husband's sister.	嫂嫂/嫂子 (older brother's wife) 弟媳(younger brother's wife)	嫂子/弟妹/大姑子/小姑子
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.	同父異母姊妹 (same father) 同母異父姐妹 (same mother)	胞姐妹
Stepsister	The daughter of your stepmother or stepfather and therefore not biologically related to you.	繼姊妹	继姐妹

Term	Definition	Traditional 繁體中文	Simplified 簡體中文
Brother	A sibling who is a boy.	兄弟	兄弟
Brother-in-law	Your sister's husband or your wife's brother.	姊夫(older sister's) 妹夫 (younger sister's)	姐夫/妹夫/妻 兄/妻弟
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.	同父異母兄弟 (same father) 同母異父兄弟 (same mother)	胞兄弟
Stepbrother	The son of your stepmother or stepfather and therefore not biologically related to you.	繼兄弟	继兄弟
Niece	Your sibling's daughter.	姪女(brother's daughter) 外甥女(sister's daughter)	侄女/外甥女
Nephew	Your sibling's son.	姪子(brother's son) 外甥(sister's son)	侄子/外甥
Spouse	Your husband or wife.	配偶	配偶
Wife	The woman to whom you are married.	妻子	妻子
Husband	The man to whom you are married.	丈夫	丈夫
Children	Genetically speaking, the people who are produced from your egg or sperm.	子女	孩子

Term	Definition	Traditional 繁體中文	Simplified 簡體中文
Daughter	Your child who is a girl.	女兒	女儿
Daughter-in-law	Your son's wife.	媳婦	儿媳
Stepdaughter	Your spouse's daughter who is not your biological child.	繼女	继女
Adoptive daughter	A girl for whom you have accepted legal responsibility and raised even though she is not your biological child.	養女	养女
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.	教女 契女 (formal/informal acceptance of kinship in a non-Christian context)	教女
Son	Your child who is a boy.	兒子	儿子
Son-in-law	Your daughter's husband.	女婿	女婿
Stepson	Your spouse's son who is not your biological child.	繼子	继子
Adoptive son	A boy for whom you have accepted legal responsibility and raised even though he is not your biological child.	養子	养子
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.	教子 契子/契仔 (Formal/informal acceptance of kinship in a non-Christian context)	教子
Ancestor	A person from whom you are descended, usually more remote than a grandparent.	祖先	祖先
Fiancé(e)	The person whom you have promised to marry. Man: fiancé Woman: fiancée	未婚夫(妻)	未婚夫/妻

Term	Definition	Traditional 繁體中文	Simplified 簡體中文
Domestic partner	The person with whom you are living and have an intimate relationship, but to whom you are not married.	同居伴侶	同居伴侶
Divorced	Having ended a marriage.	離婚	离婚
Engaged	Having promised to marry someone.	訂婚	订婚
Widow	A woman whose husband has died.	寡婦 / 遺孀	寡妇
Widower	A man whose wife has died.	鰥夫	鳏夫
Relative	A person in your family.	親戚	亲戚
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.	血親	血缘关系
Next of kin	Your closest living blood relation.	近親	直系亲属

Analogy

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

A-B-C analysis	9
Abnormal	35, 61, 91
Abortion.....	61, 91
Abstract language	9
ACMG 59.....	61
Adaptive behavior or adaptive skills.....	9
Adaptive skills	61
Adoption	61, 91
Adoptive daughter.....	53, 86, 112
Adoptive father.....	48, 81, 107
Adoptive mother	48, 81, 107
Adoptive son.....	53, 86, 112
Age of majority	9
Altered gene copy.....	38, 68
American College of Medical Genetics and Genomics (ACMG)	61
Amniocentesis	62, 91
Amniotic fluid	62, 91
Ancestor.....	53, 86, 112
Anencephaly	62, 91
Anxiety disorder	9
Applied Behavior Analysis (ABA)	9
Apraxia.....	9
At average risk	43, 76, 103
At high risk.....	43, 76, 103
At higher risk than 'X'	43, 76, 103
At risk.....	43, 76, 103
ATM	47
Attention Deficit Hyperactivity Disorder (ADHD)	10
Audiologist.....	10
Augmentative and Alternative Communication (AAC)	10
Aunt	49, 82, 108
Autism Spectrum Disorder (ASD).....	10
Aversive	10

B

Behavior Intervention Plan (BIP)	10
Behavioral disorder	11
Benign.....	35, 62, 91
Biochemical testing.....	62
Biomarker	11
Bipolar disorder	11
Birth canal.....	62, 91
Birth defect.....	62, 92
Blood relation	54, 87, 113

Blood test.....	35, 62, 92
Board Certified Behavior Analyst (BCBA).....	11
Body language	11
Brace	62
BRCA 1	47
BRCA 2	47
Breast cancer	35
Brother.....	52, 85, 111
Brother-in-law	52, 85, 111
Buccal swab	35, 62, 92

C

CA-125 blood test	35
Carrier	35, 63, 92
Case manager	11
Catatonia	11
Cell.....	35, 63, 92
Cervix	63, 92
Challenging behaviors.....	11
Children	53, 85, 111
Chorionic Villus Sampling (CVS).....	63, 92
Chromosome	35, 63, 92
Chromosome test / Karyotype	63, 92
Civil commitment.....	11
Cleft lip.....	63, 93
Cleft palate	63, 93
Clinically significant	63, 93
Cognition	63
Cognitive behavioral therapy	11
Cognitive development.....	11
Colonoscopy	35
Communication	12
Communication disorder	12
Comorbid	12
Compulsion.....	12
Condition (e.g., “Genetic condition”)	36, 63, 93
Consanguinity	36, 64, 93
Conservatorship.....	12
Consultation model	64
Continuing care model	64
Cousin	50, 83, 109
Crisis plan.....	12
Cystic fibrosis	64, 93

D

Data analysis.....	12
Daughter	53, 86, 112
Daughter-in-law.....	53, 86, 112
DCIS (ductal carcinoma in situ).....	36

De novo.....	64
De-escalation	12
Deleterious mutation.....	36, 64, 93
Deletion	64, 93
Depression.....	12
Developmental assessment.....	13, 64
Developmental delay.....	13
Developmental Disability (DD)	13
Developmental pediatrician	13
Diagnostic test	64, 93
Differential diagnosis.....	13
Disease-causing mutation.....	36, 93
Disruption	13
Divorced.....	54, 87, 113
DNA (Deoxyribonucleic Acid).....	36, 65, 94
DNA banking.....	36
DNA marker (also called a “Genetic marker”)	36, 65, 94
DNA mutation.....	65, 94
DNA sequence	36, 65, 94
DNA sequencing	37, 65, 94
Domestic partner.....	54, 87, 113
Dominant.....	37, 65, 94
Donor egg or sperm.....	65, 94
Down syndrome	14
Down syndrome (Trisomy 21)	65, 95
DTC (direct-to-consumer genetic testing)	36
Dual diagnosis.....	14
Duplication.....	37, 66, 95
Dysarthria	14
Dyspraxia	14
E	
Early age of onset	37
Early intervention	14
Echolalia or Echophrasia.....	14
Edwards syndrome (Trisomy 18).....	66, 95
Egg	37, 66, 95
Elopement	14
Embryo	66, 95
Engaged	54, 87, 113
Environmental causes or factors	37, 66
Epilepsy.....	14
Escalating.....	14
Evidence-Based Practice (EBP)	14
Executive control / executive functioning	15
Exome sequencing.....	66, 70
Exon	66
Extinction.....	15
Extinction burst.....	15

F

Face blindness	15
Fading	15
Fallopian tube	66, 95
False negative	66, 95
False positive	66, 95
Familial.....	37, 66, 95
Family history	37, 67, 95
Father	48, 81, 107
Father-in-law	48, 81, 107
Fecal digging	15
Fecal smearing.....	15
Fertilization.....	67, 96
Fetal Alcohol Spectrum Disorder (FASD)	15
Fetal Alcohol Syndrome (FAS)	15
Fetal surgery	67, 96
Fetus	67, 96
Fiancé(e)	53, 86, 112
First cousin.....	50, 83, 109
First cousin once removed.....	50, 83, 109
First trimester screening.....	67, 96
Flip a coin.....	67, 96
Fragile X syndrome	67, 96
Fragment	37, 67, 96
Fraternal twins.....	51, 84, 110
Function of behavior.....	15
Functional Behavior Analysis (FBA)	16
Functional communication	16

G

Gene	38, 67, 96
Gene copy	38, 68
Gene deletion	38, 68
Gene panel test	70
Gene panel testing.....	68
Gene variant	68
General population.....	68, 96
Generalization	16
Generation.....	38, 69, 97
Genetic	38
Genetic alphabet	69
Genetic counseling	38, 69, 97
Genetic counseling intern.....	39, 97
Genetic counselor.....	39, 69, 97
Genetic discrimination.....	39, 69, 97
Genetic factors	39, 69, 97
Genetic information	39, 69, 98
Genetic material	39, 70, 98

Genetic predisposition.....	39, 70, 98
Genetic test	70, 98
Genetic trait.....	70, 98
Genetic variant	98
Geneticist.....	70, 98
Genetics	70, 71, 97
Genome	71
Genome sequencing.....	70, 71
Germline testing	71
Gestures.....	16
Goddaughter.....	53, 86, 112
Godfather	48, 81, 107
Godmother	48, 81, 107
Godson.....	53, 86, 112
Grandfather	50, 83, 109
Grandmother	50, 83, 109
Grandparent	50, 83, 109
Great aunt.....	50, 83, 109
Great grandfather.....	51, 84, 110
Great grandmother.....	51, 84, 110
Great grandparents	51, 84, 110
Great uncle	51, 83, 109
Guardianship	16, 71

H

Half brother	52, 85, 111
Half siblings.....	51, 84, 110
Half sister	52, 84, 110
Helix.....	71
Hereditary.....	71, 98
Hereditary material	40, 71, 98
Huntington's disease	71
Husband.....	52, 85, 111
Hypersensitivity	16
Hyposensitivity	17

I

Identical twins	51, 84, 110
Idiosyncratic language/ vocabulary	17
In Vitro Fertilization (IVF).....	72, 98
Incontinence	17
Individual gene testing	70, 72
Individualized Education Plan (IEP).....	71
Individualized Education Program (IEP).....	17
Individualized Family Service Plan (IFSP)	18
Individuals with Disabilities Education Act (IDEA)	18
Infertility	72, 98
Informed consent	41, 72, 99
Inheritance pattern.....	41, 72, 99

Inherited	41, 72, 99
Insertion.....	99
Intellectual disability.....	18, 72, 99
Interdisciplinary evaluation team	18
Intervention	18
Involuntary commitment.....	18
J	
Joint attention	19
K	
Klinefelter's syndrome (47, XXY)	72, 99
L	
Language delay	19
Language disorder	19
Learning disability.....	19
Least Restrictive Environment (LRE).....	19
Lynch syndrome.....	41, 72
M	
Maladaptive behavior.....	19
Malignant.....	41
Marker chromosome	73, 99
Maternal aunt.....	49, 82, 108
Maternal grandfather	50, 83, 109
Maternal grandmother.....	50, 83, 109
Maternal great aunt	50, 83, 109
Maternal great uncle	51, 83, 109
Maternal Serum Screening (MSS or maternal serum alphafetoprotein test)	73, 99
Maternal uncle	49, 82, 108
Medicaid	19
Medical home.....	19
Mental health hold	19
Metastasis.....	41, 73
Microarray	70, 73, 100
Miscarriage	73, 100
MLH1	47
Molecule	41, 73
Mosaicism.....	73, 100
Mother.....	48, 81, 107
Mother-in-law.....	48, 81, 107
Motor Abilities	73
Motor skill.....	20
MSH2	47
MSH6	47
Multifactorial	41, 73, 100
Mutation.....	41, 74

N

Negative.....	45
Nephew	52, 85, 111
Neurodevelopmental conditions.....	20
Next of kin	54, 87, 113
Niece.....	52, 85, 111
Non-Invasive Prenatal Testing (NIPT)	74, 100
Nonverbal communication	20
Normal gene copy.....	38, 68
Nuchal thickening	74, 101
Nutritionist	20

O

Obsession.....	20
Obsessive compulsive disorder (OCD)	20
Occupational therapist (OT)	20
Oncologist.....	41
Oophorectomy.....	41
Open neural tube defect	74, 101
Ototoxic	20
Ova.....	74, 101
Ovarian cancer.....	41
Ovary	74, 101
Overcorrection.....	21

P

Packets of genetic information.....	42, 74
PALB2	47
Parent	48, 81, 107
Paternal aunt	49, 82, 108
Paternal grandfather	50, 83, 109
Paternal grandmother	50, 83, 109
Paternal great aunt.....	50, 83, 109
Paternal great uncle	51, 83, 109
Paternal uncle.....	49, 82, 108
Pathogenic.....	74
Pattern	42
Pediatric Autoimmune Neuropsychiatric Disorders Associated with Streptococcal infections (PANDAS)	21
Pedigree.....	42, 74
Penetrance.....	42, 74, 101
Phenylketonuria (PKU).....	21
Physical therapist (PT)	21
Pica	21
Picture Exchange Communication Systems (PECS).....	21
Placenta	75, 101
PMS2.....	47
Polymorphism.....	45, 79, 105
Polypharmacy	21

Positive	45
Positive Behavior Supports (PBS)	21
Post-Traumatic Stress Disorder (PTSD).....	21
Predisposition to cancer	42
Prematurity.....	22
Presymptomatic.....	75, 101
Prognosis	42, 75, 101
Proliferation (cell proliferation).....	42
Prompting	22
Prophylactic mastectomy	42
Protein	42, 75, 101
Psychologist	22
Psychosis.....	22
Psychotropic	22
Puberty	22
R	
Random	42, 75, 101
Recessive	43, 75, 102
Recurrence.....	43
Red flag	22, 43, 75, 102
Reinforce	22
Reinforcement strategies	22
Relative	54, 87, 113
Replicate	75, 102
Reproductive history	75, 102
Resilience	22
Respite care	22
Restraints.....	22
Restricted or repetitive behaviors or interests.....	23
Reward.....	23
Ring chromosome	75, 102
Risk.....	43, 76, 103
Risk factor	23, 43, 76, 103
Ritual.....	23
Rumination	23
Runs in the family	43, 76, 103
S	
Saliva (spit) sample	76
Saliva (spit) test	43
Schizophrenia	23
Screen	43, 76, 103
Screening test	43, 76, 104
Screening test or tool	23
Seclusion.....	23
Second cousin	50, 83, 109
Sedating.....	23
Seizure disorder.....	23

Self-advocacy	23
Semen	77, 104
Seminal fluid	77, 104
Sensory abilities	77
Sensory avoidance	23
Sensory defensiveness	23
Sensory input	24
Sensory processing disorder	24
Sensory-seeking behavior	24
Siblings	51, 84, 110
Sickle cell disease	77, 104
Single site analysis	77
Single site gene testing	70
Sister	52, 84, 110
Sister-in-law	52, 84, 110
Sleep apnea	24
Sleep disturbances	24
Social communication disorder	24
Social reciprocity	24
Social work or social worker	24
Somatic genetic changes	77
Son	53, 86, 112
Son-in-law	53, 86, 112
Special education	77
Special Needs Parent Advocate	24
Specific learning disability	25
Speech generating device	25
Speech sound disorder	25
Speech-language pathologist	25
Sperm	44, 77, 104
Sperm donation	77, 104
Spina bifida	77, 104
Sporadic	44
Spouse	52, 85, 111
Standardized test	25
Staring spells	25
Statistically significant	44, 78, 104
Stepbrother	52, 85, 111
Stepdaughter	53, 86, 112
Stepfather	48, 81, 107
Stepmother	48, 81, 107
Stepsiblings	51, 84, 110
Stepsister	52, 84, 110
Stepson	53, 86, 112
Stereotypy	25
Stimulation	25
Supplemental Security Income (SSI)	25
Supportive seating	78
Syndrome	44

T

Tangibles	25
Teratogen	78, 105
Termination of pregnancy	78, 105
Test result	45, 79, 105
Testes	79, 105
Time delay	25
To flip a coin	37, 96
Tourette syndrome or Tourette's syndrome	25
Toxic exposure	79, 105
Tracking scales	26
Trait	45, 79, 106
Transition plan / transition services	26
Translocation	80, 106
TRICARE	26
Triples	51, 84, 110
True negative	45
Tumor	45
Tumor testing	46
Twins	51, 84, 110
Typo	46, 80, 106

U

Ultrasound	46, 80, 106
Uncle	49, 82, 108
Uninformative negative	45
Uterus	80, 106

V

Vagina	80, 106
Variant	46, 80, 106
Variant of uncertain significance	45, 79, 105
Vas deferens	80, 106
Visual cues	26
Visual schedules	26
Vocational rehabilitation	26

W

Widow	54, 87, 113
Widower	54, 87, 113
Wife	52, 85, 111
Wraparound	27

X

X-linked	80, 106
----------------	---------

