



National Coordinating Center
for the Regional Genetics Networks

The National Coordinating Center for the Regional Genetics Networks

Glossaries of Genetics Terms

English – Korean

2022 Edition

Compiled and edited by:
Cynthia E. Roat, MPH

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

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Acknowledgments

Mission of the NCC

The National Coordinating Center for the Regional Genetics Networks (NCC) has been funded by the Health Resources and Services Administration (HRSA) since 2004. Our mission is to improve access to genetics for underserved populations. This mission is accomplished through different programs, many of which are educational tools and resources for non-genetics professionals. Learn more at <https://nccrcg.org>.

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Additional Acknowledgments

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

The Cross Cultural Health Care Program

The mission of The Cross Cultural Health Care Program (CCHCP) is to serve as a bridge between communities and health care institutions to advance access to quality health care that is culturally and linguistically appropriate. CCHCP provides resources, such as bilingual medical glossaries, as well as medical interpreter, patient navigator, and cultural competence training for individuals and institutions with the goal of systems change. Learn more at:

<https://xculture.org>

CCHCP is supporting and coordinating the translation process of this glossary and is providing the glossary free of charge in its online store: <https://xculture.org/store/>

Table of Contents

Glossaries of Genetics Terms	1
Glossary of Autism Spectrum Disorder (ASD) Genetics Terms	5
Preface	6
How to Use the Glossary.....	6
Glossary of Terms	7
References	23
Glossary of Pediatric Genetics Terms	25
Preface	26
How to Use the Glossary.....	26
Glossary of Terms	27
Family Relationships	45
Glossary of Prenatal Genetics Terms	51
Preface	52
How to Use the Glossary.....	52
Glossary of Terms	53
Family Relationships	69
Analogies.....	75
Index	77



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

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

English – Korean

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Preface

The seven Regional Genetics Networks (RGNs), the National Genetics Education and Family Support Center (Family Center), and the National Coordinating Center for the Regional Genetics Networks (NCC) mission is to improve access to quality genetic services for medically underserved populations. This bilingual glossary of terms related to Autism Spectrum Disorders (ASD) is provided to help enable access to ASD medical services to limited English proficiency populations.

How to Use the Glossary

This glossary is organized alphabetically in English. The first column shows the term in English. The second column has the definition of the term in English. The third column has the translation of the term.

At the end of the translations is a list of references used for the definitions and places where you can find more information.

Glossary of Terms
English – Korean

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

Term	Definition	Translation
A-B-C analysis (n)	An approach to understanding behavior by examining the Antecedent (the cause), the Behavior, and the Consequence (the result). ¹	A-B-C 분석
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	추상어
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	적응 행동 또는 적응 기술
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	성년
Anxiety disorder (n)	A pattern of constant worry or tension under many different circumstances. ¹	불안장애
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	응용 행동 분석 (ABA)
Apraxia (n)	See “Dyspraxia.” ²	실행증

Glossary of Autism Spectrum Disorder (ASD) Genetics Terms – Korean

Term	Definition	Translation
Attention Deficit Hyperactivity Disorder (ADHD) (n)	A disorder that shows up in the areas of inattention, hyperactivity, and impulsiveness. It is evidenced by frequent shifting from one activity or focus to another, having difficulty organizing and completing tasks correctly, impulsive response, or failure to follow rules. It may occur with or without hyperactivity which includes behaviors such as excessive running, talking, fidgeting, and/or restlessness. ²	주의력결핍 과잉행동장애 (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	청력학자
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	증강 및 대안 의사 소통 (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	자폐 스펙트럼 장애 (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	혐오성/유해 자극
Behavior Intervention Plan (BIP) (n)	A plan to improve an individual’s behavior, created based on the results of a Functional Behavior Assessment. ¹	행동 중재 계획 (BIP)
Behavioral disorder (n)	A condition in which behavior significantly deviates from acceptable norms. ¹	행동 장애
Biomarker (n)	An indicator of a certain biological state. ¹	바이오마커
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	양극성 장애/조울증

Glossary of Autism Spectrum Disorder (ASD) Genetics Terms – Korean

Term	Definition	Translation
Board Certified Behavior Analyst (BCBA) (n)	A professional certified to provide ABA therapy by the Behavior Analyst Certification Board (BACB). ¹	국제 응용 행동분석학회 (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	신체 언어
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	사례 관리자
Catatonia (n)	A state in which a person does not move and does not respond to others. ¹	긴장증
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	도전적 행동
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	민사적 감금제도
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	인지 행동 치료
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	인지 발달
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	의사소통
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	의사소통 장애
Comorbid (adj)	Pertaining to a disease or disorder that occurs simultaneously with another. ¹	동반이환

Glossary of Autism Spectrum Disorder (ASD) Genetics Terms – Korean

Term	Definition	Translation
Compulsion (n)	The drive to do something in particular or in a particular way, such as the need to straighten all the forks at the dinner table. ¹	강제
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	후견인 제도
Crisis plan (n)	A document that outlines in specific detail the necessary strategies and steps that must be taken when a crisis occurs. ¹	위기 관리 대책
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	데이터 분석
De-escalation (n)	The process of stopping a challenging behavior or crisis from intensifying and calming the situation. ¹	단계적 축소
Depression (n)	A mood disorder in which feelings of sadness, anger, or frustration interfere with everyday life for an extended period of time. ¹	우울증
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	발달 검사
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	발달 지연
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	발달 장애 (DD)

Glossary of Autism Spectrum Disorder (ASD) Genetics Terms – Korean

Term	Definition	Translation
Developmental pediatrician (n)	Also known as a developmental-behavioral pediatrician, these professionals have training and experience to assess and treat a wide range of medical and psychosocial aspects of a child’s developmental and behavioral difficulties. Their expertise may make them a good choice for children with complicated medical or developmental problems. ²	발달 장애 소아과 의사
Differential diagnosis (n)	Distinguishing between two or more diseases with similar symptoms to identify which is causing distress or challenging behavior. ¹	감별 진단
Disruption (n)	An event that causes an unplanned deviation from a situation. ¹	중단
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	다운증후군 또는 21삼체성
Dual diagnosis (n)	The identification of an additional mental health disorder individuals with developmental disabilities. ¹	이중 진단
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	구음 장애
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	통합 운동 장애
Early intervention (n)	Programs or services designed to meet the developmental needs of infants and toddlers (birth to three years old) and their families.	조기 중재
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	반향 언어 또는 음성 모방
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	도피
Epilepsy (n)	A brain disorder in which a person has repeated seizures (episodes of disturbed brain activity or convulsions) over time. ¹	뇌전증

Glossary of Autism Spectrum Disorder (ASD) Genetics Terms – Korean

Term	Definition	Translation
Escalating (v)	Increasing or worsening rapidly. ¹	단계적으로 확대하다
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	근거 기반 실무 (EBP)
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	집행 관리 / 집행 기능
Extinction (n)	A response used to eliminate a behavior that involves ignoring a mild behavior when it is used for attention. ¹	소거
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	소거 격발
Face blindness (n)	An impairment in the recognition of faces. ¹	안면 인식 장애
Fading (v)	Gradually reducing the number of prompts or types of prompts to encourage more independence for the learner. ⁵	줄이다
Fecal digging (v)	The process in which an individual puts his fingers into his rectum. ¹	분변을 파내다
Fecal smearing (v)	The process in which feces are spread on property or the individual himself. ¹	분변을 도말하다
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	태아 알코올 스펙트럼 장애 (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	태아 알코올 증후군 (FAS)
Function of behavior (n)	The purpose or reason behind a specific behavior for an individual. ¹	행위 기능

Term	Definition	Translation
Functional Behavior Analysis (FBA) (n)	The process of systematically determining the function of behaviors, usually inappropriate, that are displayed by people. Behaviors are defined, measured, and analyzed in terms of what happened before and after their occurrence. Over time the events before and after the behavior occurs are systematically changed in order to determine the function of the behavior for the person displaying it. ²	기능적 행동 분석
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.	기능적 의사소통
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	일반화
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	제스처/몸짓
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	후견법인
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	과민성
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	과민성 감퇴

Term	Definition	Translation
Idiosyncratic language/vocabulary (n)	Using language in unusual ways that may not be obvious to the communication partner. This includes using scripts from previous conversations or media applied to different contexts like saying “C is for cookie” to express that they like something. Or “Dora loves waterfall!” to request to take a bath/shower. ⁵	특이한 언어/어휘
Incontinence (n)	The (usually) involuntary passing of feces or urine, generally not into a toilet or diaper. ¹	실금
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	개별화 교육 프로그램 (IEP)
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	개별화 가족 서비스 계획안 (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	특수 교육법 (IDEA)

Glossary of Autism Spectrum Disorder (ASD) Genetics Terms – Korean

Term	Definition	Translation
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	지적 장애
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	전문가 평가단
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	중재
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	비자발적 입원
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	공동 주의
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	언어 발달 지연
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	언어 발달 장애
Learning disability (n)	A disorder that affects how a person learns and understands primary skills such as reading, writing and math. ²	학습 장애
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	최소 제한 환경 (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	부적응 행동

Glossary of Autism Spectrum Disorder (ASD) Genetics Terms – Korean

Term	Definition	Translation
Medicaid (n)	A government program that provides healthcare coverage for low-income families and individuals with disabilities in the United States. ¹	메디케이드
Medical home (n)	A team-based healthcare delivery model led by a physician that provides comprehensive and continuous medical care to patients. ¹	환자 중심 의료 가정
Mental health hold (n)	Involuntary hospitalization due to a mental health crisis. ¹	정신 건강 억류/정신병 억류
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	운동 기능
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.	신경발달 조건
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	비언어적 의사소통
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	영양사
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	강박/집착
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	강박 장애 (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	작업 치료사 (OT)

Glossary of Autism Spectrum Disorder (ASD) Genetics Terms – Korean

Term	Definition	Translation
Ototoxic (n)	Damaging to the ears, causing sound sensitivities, dizziness, or balance issues. ¹	이독성/귀독성
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	과잉 수정
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	연쇄구균 감염 관련 소아기 자가면역성 신경정신과적 질환 (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	페닐케톤뇨증 (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	물리 치료사 (PT)
Pica (n)	An eating disorder that involves eating things that are not food (e.g., dirt, plastic). ¹	이식증/무분별 탐식증
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	그림교환 의사소통 체계 (PECS)
Polypharmacy (n)	The use of multiple medications by a patient. ¹	다제 약물 병용
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	긍정적 행동지원 (PBS)
Post-Traumatic Stress Disorder (PTSD) (n)	An anxiety disorder that can occur after witnessing or experiencing a traumatic event. ¹	외상 후 스트레스 장애 (PTSD)
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	미숙

Glossary of Autism Spectrum Disorder (ASD) Genetics Terms – Korean

Term	Definition	Translation
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	고취시키다
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	심리학자
Psychosis (n)	A loss of contact with reality that usually includes delusions and hallucinations. ¹	정신증
Psychotropic (adj)	A medication or intervention that affects brain activity, behavior, or perception. ¹	항정신성
Puberty (n)	The process of physical changes that occur when a child's body matures into an adult. ¹	사춘기
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	적신호
Reinforce (v)	To strengthen with additional material or support. ¹	강화하다
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."	강화 전략
Resilience (n)	An ability to recover from or adjust easily to change or a difficult situation. ¹	회복력
Respite care (n)	A service that provides short-term breaks that can relieve stress, restore energy, and promote balance for caregivers. ¹	일시적 위탁
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	강박
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	제한적 반복 행동과 관심
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	보상
Risk factor (n)	Conditions that increase the likelihood of aggression. ¹	위험 인자
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	의례

Glossary of Autism Spectrum Disorder (ASD) Genetics Terms – Korean

Term	Definition	Translation
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	반추
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	정신분열증/조현병
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	선별검사 또는 도구
Seclusion (n)	A situation in which an individual is isolated in a room in response to a behavior they have exhibited. ¹	고립
Sedating (v)	Calming, sleep-inducing, or numbing an individual experiencing challenging behaviors or struggling during difficult situations. ¹	진정시키다
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	발작 장애
Self-advocacy (n)	The ability of an individual to communicate his or her wants and concerns and make his or her own decisions. ¹	자기 옹호
Sensory avoidance (n)	Blocking or staying away from something that is painful or bothersome. ¹	감각 회피
Sensory defensiveness (n)	A tendency to react negatively or with alarm to sensory input which is generally considered harmless or non-irritating. ¹	감각 방어
Sensory input (n)	Any source that creates sensation and activates one or more of the senses -vision, smell, sound, taste, and touch. ¹	감각 입력신호
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	감각 처리 장애
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	감각 탐색 행동
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	수면 무호흡증

Glossary of Autism Spectrum Disorder (ASD) Genetics Terms – Korean

Term	Definition	Translation
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	수면 장애
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	사회적 의사소통 장애
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	사회적 호혜성
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	사회 복지 또는 사회 복지사
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	특수 요구 학부모 옹호자
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	특정 학습 장애
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	음성 산출 기기 또는 음성 출력 장비
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	말소리 장애
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	언어 병리학자

Glossary of Autism Spectrum Disorder (ASD) Genetics Terms – Korean

Term	Definition	Translation
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	표준 시험
Staring spells (n)	Occasions when an individual is in a trance staring into space, which can often signal seizure activity. ¹	응시 발작
Stereotypy (n)	Repetitive or ritualistic movements such as body rocking or crossing and uncrossing of legs. ¹	상동증
Stimulation (n)	Excitement or activity triggered by a stimulus either internally or externally. ¹	자극
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	생활 보조금 (SSI)
Tangibles (n)	Items or rewards that can be touched, such as a toy or piece of candy. ¹	유형
Time delay (n)	A prompting procedure that fades prompts during activities by having longer delays before a prompt is provided.	시간 지연
Tourette syndrome or Tourette's syndrome (n)	A neurological disorder characterized by tics, or repetitive, stereotyped, involuntary movements and vocalizations. ¹	뚜렛 증후군
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	추적 스케일
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	전환 계획/전환 서비스
TRICARE (n)	The health care program for Uniformed Service members, retirees, and their families worldwide. ¹	트라이케어

Term	Definition	Translation
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	시각적 단서
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	시각적 스케줄
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.	직업 재활
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	음성 출력 장비 또는 음성 출력 의사소통 보조기 또는 음성 생성 기기
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	랩어라운드

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](https://www.autism-society.org/)
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™](https://www.autism-source.com/), 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](https://www.autismspeaks.org/)
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 Pediatric Genetics Terms

English – Korean

2022 Edition

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



Preface

The seven Regional Genetics Networks (RGNs), the National Genetics Education and Family Support Center (Family Center), and the National Coordinating Center for the Regional Genetics Networks (NCC) mission is to improve access to quality genetic services for medically underserved populations. This bilingual glossary of terms related to Pediatric Genetics is provided to help enable access to pediatric medical services to limited English proficiency populations.

How to Use the Glossary

This glossary is organized alphabetically in English. The first column shows the term in English. The second column has the definition of the term in English. The third column has the translation of the term.

At the end of the translations of the pediatric genetics terms is a glossary of translations for family relationships terms. It is very important for genetic counselors to know if a relative is a blood relative or a relative by marriage.

Glossary of Terms

English – Korean

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

Term	Definition	Translation
Abnormal (adj)	Different from what is considered normal, average, or expected, e.g., a gene sequence that is different than that found in most people.	비정상적
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.”	낙태
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	유전체 분석 임상 진단 서비스 (ACMG 59)
Adaptive skills (n)	Skills/abilities needed to perform daily activities, particularly those that allow an individual to live independently and/or work. Examples: brushing teeth, bathing, dressing, food shopping, and taking public transportation.	적응 기술
Adoption (n)	The process of accepting legal responsibility for someone else’s child to raise as your own.	입양
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.	미국의학 유전학회 (ACMG)
Amniocentesis (n)	A procedure by which a sample of amniotic fluid is withdrawn from the amniotic sac. This is usually done by inserting a long needle through the abdominal and uterine walls with the guidance of ultrasound.	양수 천자

Term	Definition	Translation
Amniotic fluid (n)	The liquid that surrounds a fetus as it develops in the uterus. This liquid contains skin cells that have sloughed off the fetus as well as other fetal cells.	양수
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.	무뇌증
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.	양성
Biochemical testing (n)	Blood tests to identify elevated levels of certain naturally occurring substances/chemicals in the body that are linked to genetic conditions.	생화학적 실험
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.	산도
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.”	선천성 결함
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.	혈액 검사
Brace (n)	A medical appliance that provides support to joints or bones, to weak muscles, or to strained ligaments.	보조기
Buccal swab (n)	A way to collect DNA from the cells on the inside of a person's cheek.	구강 스왑
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.	보인자
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.	세포

Term	Definition	Translation
Cervix (n)	The narrow lower part of the uterus than connects to the vagina.	자궁 경부
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.	용모막 용모 샘플링 (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.	염색체
Chromosome test / Karyotype (n)	A test that looks at the number and arrangement of chromosomes in a cell.	염색체 검사/핵형
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.	구순열
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.	구개열
Clinically significant (adj)	If a test result is “clinically significant,” the result indicates a medical problem that can impact a person’s life.	임상적 유의성
Cognition (n)	The processes involved in thinking, learning, understanding, and remembering.	인지
Condition (e.g., “genetic condition”) (n)	A long-term medical health issue.	조건 (예: ‘유전적 조건’)
Consanguinity (n)	When parents are blood relatives to each other.	혈족
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.	자문 모델
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.	자문 케어 모델

Term	Definition	Translation
Cystic fibrosis (n)	A progressive genetic condition that affects the exocrine glands (the glands that make sweat and digestive juices) and causes the production of thick, sticky mucus. This mucus blocks the pancreatic duct, the intestines, and the lungs, leading to persistent respiratory infections.	낭성 섬유증
De novo (adj)	Genetic changes that are not inherited but that occur during cell division.	신생합성
Deleterious mutation/ Disease-causing mutation (n)	A change in a person’s DNA that may cause a medical condition.	해로운 변이/질병을 일으키는 변이
Deletion (n)	Having a section of genetic information (DNA) missing.	결실
Developmental assessment (n)	An evaluation of how an individual patient’s intellectual, emotional, and social development compare with others of the same age.	발달 검사
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.	진단 검사
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 (데옥시리보핵산)
DNA marker (also called a “Genetic marker”) (n)	A readily recognizable genetic trait, gene, or DNA segment.	DNA 마커(‘유전자 마커’라고도 칭함)
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.	DNA 변이

Term	Definition	Translation
DNA sequence (n)	The exact arrangement of the chemicals that make up a section of DNA.	DNA 염기서열
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.	DNA 염기서열 분석
Dominant (adj)	A genetic trait in which one copy of the gene is sufficient for a trait to be expressed. In a dominant genetic condition, if one copy of the gene has a mutation, the personal will be affected with the condition.	우성
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.	난자 또는 정자 기증자
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.	다운증후군 (21삼체성)
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.	복제
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.	에드워드 증후군 (18삼체성)
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."	난자
Embryo (n)	An unborn mammal, between conception and 8 weeks of gestation.	배아
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.	환경적 원인 또는 요인

Term	Definition	Translation
Exome sequencing (n)	A genetic test in which a patient's exons are sequenced and studied.	엑솜 염기서열 분석
Exon (n)	The part of the genome that codes for proteins. The majority of deleterious mutations occur in exons.	엑손
Fallopian tube (n)	The tube that connects an ovary to the uterus.	난관
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.	가음성
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.	가양성
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.	가족성
Family history (n)	The medical history of the members of a biological family.	내력
Fertilization (n)	The joining of an egg and sperm to create the first cell that will develop into an embryo, then fetus, then baby.	수정
Fetal surgery (n)	Surgery conducted on a fetus while it is still in the uterus.	태아 수술
Fetus(n)	An unborn mammal, between 8 weeks of gestation to birth.	태아
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.	제1삼분기 선별검사
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.	동전 던지가하다

Term	Definition	Translation
Fragile X syndrome (n)	A genetic condition that affects the FMR1 gene so that it does not function properly. This syndrome causes intellectual disability, behavioral and learning challenges and various physical characteristics that are not life threatening.	유약 X 증후군
Fragment (n)	A small piece; an incomplete part of a whole.	절편
Gene (n)	A specific sequence of DNA that codes for one or many functions within the cell and body.	유전자
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.	유전자 복제
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.”	일반적 유전자 복제
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.”	변형된 유전자 복제
Gene deletion (n)	Having a piece of genetic information missing from a gene.	유전자 결실
Gene panel testing (n)	Genetic testing that looks at 2 or more specific genes known to be associated with particular conditions.	유전자 패널 검사
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.	유전자 변이형
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.	일반 인구

Term	Definition	Translation
Generation (n)	<p>The people who constitute a single step in a line of descent from an ancestor; a group of people born and living more or less at the same time.</p> <p>Example: You, your brothers, and sisters, all your spouses and your cousins are in the same generation. Your parents, your aunts, and uncles and all their spouses form a previous generation. Your grandparents, their siblings, and spouses from an even earlier generation. Your children and nieces and nephews form a later generation.</p>	세대
Genetic alphabet (n)	<p>A group name for four chemical bases, or “nucleotides,” that form the basis of all genetic material. They are adenine (A), thymine (T), cytosine (C), and guanine (G).</p>	유전 알파벳
Genetic counseling (n)	<p>A discussion with a medical professional with expertise in genetics about the basic concepts of genetics, genetic conditions, the chances of being affected by a genetic condition or having a child with a genetic condition, and genetic testing and treatment.</p>	유전 상담
Genetic counselor (n)	<p>A healthcare professional with a specialized graduate degree who works with people who have concerns about genetic conditions in their family. Genetic counselors provide information about genetic conditions, help patients understand their chances of being affected by a genetic condition or having a child with a genetic condition, and help them make informed decisions about testing and treatment. Genetic counselors also provide emotional support to patients and families.</p>	유전 상담가
Genetic discrimination (n)	<p>Occurs when people are treated differently by their employer or insurance company due to their genetic makeup. There are federal and state laws that help protect against genetic discrimination.</p>	유전적 차별
Genetic factors (n)	<p>Specific aspects of a person’s genetic make-up that influence that person’s health and development.</p>	유전적 요소
Genetic information (n)	<p>The instructions encoded in DNA, which tells every cell in a body how to grow, what to do and how to reproduce.</p>	유전 정보

Term	Definition	Translation
Genetic material (n)	All the parts of a cell that carry genetic information. Genetic material could include genes, parts of genes, a group of genes, a DNA molecule, a fragment of DNA, a group of DNA molecules, or the entire set of genetic instructions.	유전 물질
Genetic predisposition (n)	An increased chance of a person developing a certain trait or disease based on that person's particular genetic makeup.	유전적 소인 (
Genetic test (n) Single site gene testing (n) Individual gene testing (n) Gene panel test (n) Microarray (n) Exome sequencing (n) Genome sequencing (n)	A laboratory test designed to determine if a person has a gene mutation or a typical DNA sequence. 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. Individual gene testing: genetic analysis of the entirety of one specific gene. Gene panel test: analysis of several genes that have been shown to be associated with a particular condition, e.g., breast cancer gene panel. Microarray: genetic analysis that looks for unexpected numbers of specific genes (two copies of every gene code is usually expected). Exome testing: a genetic test in which the exons of a patient's genetic code are sequenced and studied. Genome sequencing: a genetic test in which the entire genetic code (the genome) of a patient is mapped out and studied.	유전 검사 사립체 유전자 검사 개별 유전자 검사 유전자 패널 검사 마이크로어레이 엑솜 염기서열 분석 게놈 염기서열 분석
Genetic trait (n)	A characteristic within a family that is passed down from parent to child through their DNA.	유전 형질
Geneticist (n)	A doctor or scientist who studies genetics.	유전학자
Genetics (adj)	The science of how an organism's genes interact with the environment to produce certain traits.	유전학적
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.	유전학

Term	Definition	Translation
Genome (n)	The entire genetic code of an individual.	게놈
Genome sequencing (n)	A genetic test in which the entire genetic code of a patient is mapped out and studied.	게놈 염기서열 분석
Germline testing (n)	Germline testing refers to the analysis of a person's DNA, which he or she inherited from his or her parents. In biology and genetics, the germline is the group of cells that will pass on the genetic material to children, in other words, the cells from which the eggs and sperm come.	생식세포 검사
Guardianship (n)	A legal arrangement, supervised by a court, in which one person takes responsibility for a minor or an adult with a disability.	후견법인
Helix (n)	Something that is spiral in form. DNA is often described as being a "double helix."	나선
Hereditary (adj)	Passed down from parent to child.	유전성
Hereditary material (n)	Genetic material that is passed down from parent to child.	유전물질
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.	헌팅턴 병
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.	개별화 교육 계획안 (IEP)
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.	체외수정 (IVF)

Term	Definition	Translation
Individual gene testing (n)	Genetic analysis of the entirety of one specific gene.	개별적 유전자 검사
Infertility (n)	The inability to have children.	난임
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.	사전동의
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.	상속 양식
Inherited (adj)	Passed down from parent to child.	상속된
Insertion (n)	An extra segment of DNA added in at a place where it is not usually found.	삽입
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.	지적 장애
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.	클라인펠터 증후군 (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.	린치 증후군
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.	마커 염색체
Maternal Serum Screening (MSS or Maternal serum alpha-fetoprotein test) (n)	A blood test available to pregnant women that identifies elevated risks for Down syndrome, trisomy 18 and neural tube defects. Usually conducted at in the second trimester, between 14-20 weeks.	모체혈청 선별검사(MSS 또는 모체혈청 알파태아단백 검사)
Metastasis (n)	The spreading of cancer from one organ to another.	전이

Term	Definition	Translation
Microarray (n)	A high-resolution chromosome test that provides more information than a basic karyotype. A microarray measures the amount of chromosome material in a sample and can detect both large changes (e.g., if there is an extra or missing chromosome) as well as small changes (e.g., if there are very small pieces of chromosomes that are extra or missing). It does not look at the visual appearance or arrangement of chromosomes but measures the amount of genetic material.	마이크로어레이
Miscarriage (n)	The spontaneous (not intentional) loss of a pregnancy. See “abortion.” Also called pregnancy loss.	유산
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.	분자
Mosaicism (n)	A condition in which some, but not all, cells in a sample show a genetic difference.	섞임증
Motor Abilities (n)	The ability to move and use one’s muscles.	운동 능력
Multifactorial (adj)	Due to a combination of genetic and non-genetic (environmental, hormonal, etc.) risk factors that act together to determine risk.	다인성
Mutation (n)	A change in a gene, usually deleterious. See “genetic variant.”	변이 (n)
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.	비침습적 산전검사 (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.	후경부 투명대

Term	Definition	Translation
Open Neural Tube Defect (ONTD) (n)	A birth defect of the spine, spinal cord, or brain that results from a hole in the spinal column not closing up when appropriate during early fetal development. The two most common types of ONTDs are spina bifida and anencephaly.	개방성 신경관 결손 (ONTD)
Ova (n)	See “egg.”	난자
Ovary (n)	The organ in a woman that stores and releases eggs. There are normally two.	난소
Packets of genetic information (n)	A phrase genetic counselors use to describe genes or chromosomes.	유전자 정보 패킷
Pathogenic (adj)	Disease causing.	병원체
Pedigree (n)	A family tree that can be used to trace the inheritance of specific genetic traits.	가계도
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.	침투
Placenta (n)	The organ that develops together with an embryo in a pregnant woman’s uterus to nourish the embryo through the umbilical cord.	태반
Presymptomatic (adj)	Before symptoms appear.	증상 발현 전 단계
Prognosis (n)	The most likely outcome of a disease process.	예후
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.	단백질
Random (adj)	Happening in an unpredictable way.	무작위성

Term	Definition	Translation
Recessive (adj)	<p>A genetic trait in which both copies of a gene need to be altered for the trait to be expressed; recessive traits are typically inherited from both parents, each one contributing one copy of the gene in question.</p> <p>In prenatal genetics, recessive inheritance refers to conditions in which both copies of a particular gene pair need to be altered to develop a specific disease or condition.</p>	열성
Red flag (n)	A warning sign or a clue.	적신호
Replicate (v)	To copy.	복제하다
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.	출산 이력
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.	고리 염색체
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>위험</p> <p>위험적</p> <p>평균 위험적</p> <p>고위험적</p> <p>'X' 보다 위험적</p> <p>위험 요인</p>
Runs in the family	Passed down from parents to children to grandchildren.	집안 내력이 있다
Saliva (spit) sample (n)	A way to collect DNA from the cells in saliva (spit).	타액(침) 표본 (n)

Term	Definition	Translation
Screen (v)	To assess a person’s risk for a particular health problem, even if the person has no symptoms to suggest that they have the problem.	선별검사하다
Screening test (n)	<p>A test that looks to detect a particular health problem, even if a patient has no symptoms of that particular condition.</p> <p>Prenatal screening tests aren’t looking for genetic changes. They are designed to look at hormones, chemicals, other things that indicate a fetus might have a higher risk of a certain condition. They are not DNA results, do not give a definite answer about whether someone is affected, and do not provide a firm diagnosis. They are usually reported as a risk number (e.g., a 1 in ____ chance), and if this number is above a certain cut-off point, the result will be categorized as “screen positive” or “screen negative.”</p>	선별검사
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.	정액
Seminal fluid (n)	See “semen.”	정액
Sensory abilities (n)	The ability to see, hear, touch, taste, and smell.	감각 능력
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.	겸형 적혈구병
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.	단일 부위 분석
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.	체세포 유전 변형
Special education (n)	Education that is tailored to individuals with developmental delays or other learning disabilities.	특수 교육

Term	Definition	Translation
Sperm (n)	The reproductive cells of the man. When sperm fertilize a woman's egg, a baby develops.	정자
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.	정자 기증
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.	척추 이분증
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.	통계적 유의성
Supportive seating (n)	Seating, such as a wheelchair, that helps an individual function in their environment and that prevents worsening of their condition.	자세보조용구
Teratogen (n)	Something that has the potential to disturb the normal development of an embryo or fetus.	기형원
Termination of pregnancy (n)	See "abortion."	임신중절

Term	Definition	Translation
<p>Test result (n) Negative (adj)</p> <p>True negative (n)</p> <p>Positive (adj)</p> <p>Variant of uncertain significance (n)</p> <p>Polymorphism (n)</p>	<p>A negative result on a genetic test means that the laboratory did not find the specific genetic change that the test was designed to identify.</p> <p>When a patient is being tested to see if he or she has a specific genetic change that is present in other family members, a negative result can be considered a “true negative.” This means that instead of having an increased risk for cancer like other family members, they have the same risk as everyone else in the general population.</p> <p>A positive result on a genetic test means that the laboratory did find a specific genetic change that is associated with a hereditary cancer syndrome (a deleterious mutation). In cancer genetics, that means that the patient has an increased risk of getting the cancer(s) associated with the gene.</p> <p>If the laboratory finds a genetic change for which currently there is not enough information to know if this change is problematic or not, it reports a “variant of uncertain significance.” Basically, this means that the laboratory found a genetic change, but they don’t know what it means. All of these variants will eventually be reclassified as either a “positive” or “negative” result. Most become “negative” and are thought to represent natural variation between individuals.</p> <p>Everyone has some degree of commonly occurring genetic changes that are not associated with cancer. If the test finds this sort of change, it reports a “polymorphism.”</p>	<p>검사 결과 음성</p> <p>진음성</p> <p>양성</p> <p>불확실성 변이형</p> <p>다형성</p>
Testes (n)	The organs in a man that create and release sperm. Also called “testicles.” Singular testis.	고환
Toxic exposure (n)	Contact with something that is harmful or poisonous.	독성 노출
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>	특성

Term	Definition	Translation
Translocation (n)	Translocations are a type of chromosome rearrangement. They can be “balanced,” meaning that two chromosomes have pieces that have “swapped” with each other, but there is still the correct amount of chromosome material. Balanced translocations do not usually cause medical or development problems. They can also be “unbalanced” where pieces of chromosomes have traded places, but there is missing or extra chromosome material. Unbalanced translocations will often cause medical and development problems.	전좌
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.	돌연변이
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.	초음파
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.”	자궁
Vagina (n)	The tube-like part of the female reproductive system that extends from the cervix to the outside of the body.	질
Variant (n)	A version of something that differs from the norm. So, a genetic variant is a change to the usual genetic sequence.	변이체
Vas deferens (n)	The tubes that lead from the testes to the urethra, through which sperm is ejaculated.	정관
X-linked (adj)	A trait that is influenced or determined by a gene on the “X” chromosome.	X연관

Family Relationships

For genetic counselors, it is very important to know if a relative is a blood relative or a relative by marriage.

Term	Definition	Translation
Parent	Your mother or father.	부모
Mother	For genetic purposes, the woman whose egg was fertilized and grew to be you.	어머니
Mother-in-law	Your husband or wife's mother.	시어머니, 장모님
Stepmother	Your father's wife who is not your biological mother.	새어머니
Adoptive mother	A woman who is not your biological mother but who accepted legal responsibility for and raised you.	양어머니
Godmother	A woman chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.	대모
Father	For genetic purposes, the man whose sperm fertilized the egg that grew to be you.	아버지
Father-in-law	Your husband or wife's father.	시아버지, 장인어른
Stepfather	Your mother's husband who is not your biological father.	새아버지
Adoptive father	A man who is not your biological father but who accepted legal responsibility for and raised you.	양아버지
Godfather	A man chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.	대부
Aunt	Your mother or father's sister or sister-in-law.	고모, 이모
Maternal aunt	Your mother's sister or sister-in-law.	이모
Paternal aunt	Your father's sister or sister-in-law.	고모
Uncle	Your mother or father's brother or brother-in-law.	삼촌, 외삼촌
Maternal uncle	Your mother's brother or brother-in-law.	외삼촌

Term	Definition	Translation
Paternal uncle	Your father's brother or brother-in-law.	삼촌
Cousin	Usually understood to be a first cousin.	사촌
First cousin	Your aunt or uncle's child.	사촌
First cousin once removed	Your aunt or uncle's grandchild or your first cousin's child	사촌의 자녀
Second cousin	The children of first cousins are second cousins to each other.	육촌
Grandparent	Your parent's father or mother.	조부모
Grandmother	Your mother or father's mother.	할머니
Maternal grandmother	Your mother's mother.	외할머니
Paternal grandmother	Your father's mother.	친할머니
Grandfather	Your mother or father's father.	할아버지
Maternal grandfather	Your mother's father.	외할아버지
Paternal grandfather	Your father's father.	친할아버지
Great aunt	Your mother or father's aunt.	조부모의 여자 형제
Maternal great aunt	Your mother's aunt.	할머니의 여자 형제
Paternal great aunt	Your father's aunt.	할아버지의 여자 형제
Great uncle	Your father or mother's uncle.	조부모의 남자 형제
Maternal great uncle	Your mother's uncle.	할머니의 남자 형제
Paternal great uncle	Your father's uncle.	할아버지의 남자 형제
Great grandparents	The parents of any of your grandparents.	증조부모
Great grandmother	The mother of any of your grandparents.	증조모

Term	Definition	Translation
Great grandfather	The father of any of your grandparents.	증조부
Siblings	The children of your father and mother.	형제,자매
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.	이복 형제,자매
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.	의붓 형제,자매
Twins	Two siblings born at the same time.	쌍둥이
Identical twins	Twins who developed from the same egg and sperm, meaning that they are genetically identical. Also called monozygotic twins.	일란성 쌍둥이
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.	이란성 쌍둥이
Triplets	Three siblings born at the same time.	삼둥이
Sister	A sibling who is a girl.	여자 형제
Sister-in-law	Your brother's wife or your husband's sister.	남자 형제의 아내 또는 남편의 여자 형제
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.	이복 여자 형제
Stepsister	The daughter of your stepmother or stepfather and therefore not biologically related to you.	의붓 여자 형제

Term	Definition	Translation
Brother	A sibling who is a boy.	남자 형제
Brother-in-law	Your sister's husband or your wife's brother.	여자 형제의 남편 또는 아내의 남자 형제
Half brother	A boy who is the either the child of your father with a different mother, or the child of your mother with a different father.	이복 남자 형제
Stepbrother	The son of your stepmother or stepfather and therefore not biologically related to you.	의붓 남자 형제
Niece	Your sibling's daughter.	여조카
Nephew	Your sibling's son.	남조카
Spouse	Your husband or wife.	배우자
Wife	The woman to whom you are married.	아내
Husband	The man to whom you are married.	남편
Children	Genetically speaking, the people who are produced from your egg or sperm.	자식
Daughter	Your child who is a girl.	딸
Daughter-in-law	Your son's wife.	며느리
Stepdaughter	Your spouse's daughter who is not your biological child.	의붓딸
Adoptive daughter	A girl for whom you have accepted legal responsibility and raised even though she is not your biological child.	입양딸
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.	대녀

Term	Definition	Translation
Son	Your child who is a boy.	아들
Son-in-law	Your daughter's husband.	사위
Stepson	Your spouse's son who is not your biological child.	의붓아들
Adoptive son	A boy for whom you have accepted legal responsibility and raised even though he is not your biological child.	입양아들
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.	대자
Ancestor	A person from whom you are descended, usually more remote than a grandparent.	조상
Fiancé(e)	The person whom you have promised to marry. Man: fiancé Woman: fiancée	약혼자
Domestic partner	The person with whom you are living and have an intimate relationship, but to whom you are not married.	동거인
Divorced	Having ended a marriage.	이혼
Engaged	Having promised to marry someone.	약혼
Widow	A woman whose husband has died.	과부
Widower	A man whose wife has died.	편부
Relative	A person in your family.	친척
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.	혈연
Next of kin	Your closest living blood relation.	최근친

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

Glossary of Prenatal Genetics Terms

English – Korean

2022 Edition

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**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 – Korean

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

Term	Definition	Translation
Abnormal (adj)	Different from what is considered normal, average, or expected, e.g., a gene sequence that is different than that found in most people.	비정상적
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.”	낙태
Adoption (n)	The process of legally taking someone else’s child into your family to raise as your own child.	입양
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.	양수 천자
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.	양수
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.	무뇌증
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.”	양성
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.	산도

Term	Definition	Translation
Birth defect (n)	A problem or physical difference with how the body works that is present at birth. Birth defects can be caused by genetic abnormalities, environmental influences, random chance, or by circumstances related to the birth process. Also called “congenital malformation” or “congenital anomaly.”	선천성 결함
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.	혈액 검사
Buccal swab (n)	A way to collect DNA from the cells on the inside of a person's cheek.	구강 스왑
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.	보인자
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.	세포
Cervix (n)	The narrow lower part of the uterus than connects to the vagina.	자궁 경부
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.	용모막 용모 샘플링 (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.	염색체
Chromosome test / Karyotype (n)	A test that looks at the number and types of chromosomes in a cell.	염색체 검사/핵형
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.	구순열

Term	Definition	Translation
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.	구개열
Clinically significant (adj)	If a test result is “clinically significant,” the result indicates a medical problem that can impact a person’s life.	임상적 유의성
Condition (e.g., “genetic condition”) (n)	A long-term medical health issue.	조건 (예: ‘유전적 조건’)
Consanguinity (n)	When parents are blood relatives to each other.	혈족
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.	낭성 섬유증
Deleterious mutation/ Disease-causing mutation (n)	A change in a person’s DNA that may cause a medical condition.	해로운 변이/질병을 일으키는 변이
Deletion (n)	Having a section of genetic information (DNA) missing.	결실
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.	진단 검사

Term	Definition	Translation
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 (데옥시리보핵산)
DNA marker (also called a “Genetic marker”) (n)	A readily recognizable genetic trait, gene, or DNA segment.	DNA 마커(‘유전자 마커’라고도 칭함)
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>	DNA 변이
DNA sequence (n)	The exact arrangement of the chemicals that make up a section of DNA.	DNA 염기서열
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.	DNA 염기서열 분석
Dominant (adj)	A genetic trait in which one copy of the gene is sufficient for a trait to be expressed. In a dominant genetic condition, if one copy of the gene has a mutation, the personal will be affected with the condition.	우성
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.	난자 또는 정자 기증자

Term	Definition	Translation
Down syndrome (Trisomy 21) (n)	<p>A genetic condition in which there are three copies of chromosome number 21 instead of two.</p> <p>Down syndrome causes a distinct facial appearance, intellectual disability, developmental delays, and may be associated with thyroid or heart disease.</p>	다운증후군 (21삼체성)
Duplication (n)	<p>Having an extra section of genetic information (DNA). A duplication occurs when part of a chromosome is copied abnormally, resulting in extra genetic material.</p>	복제
Edwards syndrome (Trisomy 18) (n)	<p>A genetic condition in which there are three copies of chromosome number 18 instead of two.</p> <p>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.</p>	에드워드 증후군 (18삼체성)
Egg (n)	<p>The reproductive cells of a woman. When fertilized by sperm, the egg will grow into an embryo. Also called an "ovum," plural "ova."</p>	난자
Embryo (n)	<p>An unborn mammal, between conception and 8 weeks of gestation.</p>	배아
Fallopian tube (n)	<p>The tube that connects an ovary to the uterus.</p>	난관
False negative (n)	<p>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.</p>	가음성
False positive (n)	<p>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.</p>	가양성
Familial (adj)	<p>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.</p>	가족성
Family history (n)	<p>The medical history of the members of a biological family.</p>	내력
Fertilization (n)	<p>The joining of an egg and sperm to create the first cell that will develop into an embryo, then fetus, then baby.</p>	수정

Term	Definition	Translation
Fetal surgery (n)	Surgery conducted on a fetus while it is still in the uterus.	태아 수술
Fetus (n)	An unborn mammal, between 8 weeks of gestation to birth.	태아
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.	제1삼분기 선별검사
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.	동전 던지가하다
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.	유약 X 증후군
Fragment (n)	A small piece; an incomplete part of a whole.	절편
Gene (n)	A specific sequence of DNA that codes for one or many functions within the cell and body.	유전자
General population (n)	<p>“Most people.”</p> <p>For example, if you have the same risk of getting cancer as the general population, that means that you have the same chance of getting cancer as everyone else. This is in contrast to a “high risk population” who has a greater chance of getting cancer than everyone else.</p>	일반 인구
Generation (n)	<p>The people who constitute a single step in a line of descent from an ancestor; a group of people born and living more or less at the same time.</p> <p>Example: You, your brothers, and sisters, all your spouses and your cousins are in the same generation. Your parents, your aunts, and uncles and all their spouses form a previous generation. Your grandparents, their siblings, and spouses from an even</p>	세대

Term	Definition	Translation
	earlier generation. Your children and nieces and nephews form a later generation.	
Genetics (adj)	The science of how an organism’s genes interact with the environment to produce certain traits.	유전학적
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.	유전 상담
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.	유전 상담 인턴
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.	유전 상담가
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.	유전적 차별
Genetic factors (n)	Specific aspects of a person’s genetic make-up that influence that person’s health and development.	유전적 요소
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.	유전 정보
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.	유전 물질
Genetic predisposition (n)	An increased chance of a person developing a certain trait or disease based on that person’s particular genetic makeup.	유전적 소인

Term	Definition	Translation
Genetic test (n)	A laboratory test designed to determine if a person has a change to their DNA.	유전 검사
Genetic trait (n)	A characteristic within a family that is passed down from parent to child through their DNA.	유전 형질
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.	유전적 변이체
Geneticist (n)	A doctor or scientist who studies genetics.	유전학자
Hereditary (adj)	Passed down from parent to child.	유전성
Hereditary material (n)	Genetic material that is passed down from parent to child.	유전물질
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.	체외수정 (IVF)
Infertility (n)	The inability to have children.	난임
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.	사전동의
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.	상속 양식
Inherited (adj)	Passed down from parent to child.	상속된
Insertion (n)	Having an extra segment of DNA added in at a place where it is not usually found.	삽입
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.	지적 장애

Term	Definition	Translation
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.	클라인펠터 증후군 (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.	마커 염색체
Maternal Serum Screening (MSS or maternal serum alpha-fetoprotein test) (n)	A blood test available to pregnant women that identifies elevated risks for Down syndrome, trisomy 18 and neural tube defects. Usually conducted at in the second trimester, between 14-20 weeks.	모체혈청 선별검사(MSS 또는 모체혈청 알파태아단백 검사)
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.	마이크로어레이
Miscarriage (n)	The spontaneous (not intentional) loss of a pregnancy. See "abortion." Also called pregnancy loss.	유산
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.	분자
Mosaicism (n)	A condition in which some, but not all, cells in a sample show a genetic difference.	섞임증
Multifactorial (adj)	Due to a combination of genetic and non-genetic (environmental, hormonal, etc.) risk factors that act together to determine risk.	다인성
Mutation (n)	A change in a gene, usually deleterious. See "genetic variant." In Spanish, the word is sometimes confused with "mutilation."	변이

Term	Definition	Translation
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.	비침습적 산전검사 (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.	후경부 투명대
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.	개방성 신경관 결손 (ONTD)
Ova (n)	See “egg.”	난자
Ovary (n)	The organ in a woman that stores and releases eggs. There are normally two.	난소
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.	침투
Placenta (n)	The organ that develops together with an embryo in a pregnant woman’s uterus to nourish the embryo through the umbilical cord.	태반 (n)
Presymptomatic (adj)	Before symptoms appear.	증상 발현 전 단계
Prognosis (n)	The most likely outcome of a disease process.	예후
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.	단백질

Term	Definition	Translation
Random (adj)	Happening in an unpredictable way.	무작위성
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>	열성
Red flag (n)	A warning sign or a clue.	적신호
Replicate (v)	To copy.	복제하다
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.	출산 이력
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.	고리 염색체
Risk (n)	The chance that something will happen; in the case of prenatal genetics, the chance that the child will have a genetic condition.	위험
At risk (adj)	Has some possibility of having a genetic condition.	위험적
At average risk (adj)	Has the same possibility of having a genetic condition as the general population.	평균 위험
At high risk (adj)	Has a greater possibility of having a genetic condition than the general population.	고위험적
At higher risk than "X" (adj)	Has a greater possibility of having a genetic condition than "X".	'X' 보다 위험적
Risk factor (n)	A circumstance that increases the risk of having a genetic condition.	위험 요인

Term	Definition	Translation
Runs in the family (e.g., sickle cell disease runs in the family) (v)	When a certain condition or trait seems to appear in many members of a single family	집안 내력이 있다 (예: 겸형 적혈구병은 집안 내력이다)
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.	선별검사하다
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>	선별검사
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.	정액
Seminal fluid (n)	See “semen.”	정액
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.	겸형 적혈구병
Sperm (n)	The reproductive cells of the man. When sperm fertilize a woman’s egg, a baby develops.	정자
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.	정자 기증

Term	Definition	Translation
Spina bifida (n)	A congenital defect of the spine in which part of the spinal cord is exposed through an opening in the bone structure. This may result in nerve damage and some degree of paralysis in the legs.	척추 이분증
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.	통계적 유의성
Teratogen (n)	Something that has the potential to disturb the normal development of an embryo or fetus.	기형원
Termination of pregnancy (n)	See “abortion.”	임신중절
<p data-bbox="203 766 365 829">Test result (n) negative (adj)</p> <p data-bbox="251 997 397 1018">Positive (adj)</p> <p data-bbox="251 1386 414 1480">Variant of uncertain significance (n)</p> <p data-bbox="251 1732 446 1764">Polymorphism (n)</p>	<p data-bbox="516 766 1112 955">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.</p> <p data-bbox="516 997 1112 1123">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).</p> <p data-bbox="516 1165 1112 1312">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.</p> <p data-bbox="516 1386 1112 1701">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.</p> <p data-bbox="516 1743 1112 1858">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.”</p>	<p data-bbox="1144 766 1258 850">검사 결과 음성</p> <p data-bbox="1161 997 1226 1018">양성</p> <p data-bbox="1161 1354 1356 1386">불확실성 변이형</p> <p data-bbox="1161 1732 1242 1753">다형성</p>

Term	Definition	Translation
Testes (n)	The organs in a man that create and release sperm. Also called “testicles.” Singular testis.	고환
Toxic exposure (n)	Contact with something that is harmful or poisonous.	독성 노출
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.	특성
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.	전좌
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.	돌연변이
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.	초음파
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.”	자궁
Vagina (n)	The tube-like part of the female reproductive system that extends from the cervix to the outside of the body.	질
Variant (n)	A version of something that differs from the norm. So, a genetic variant is a change to the usual genetic sequence.	변이체
Vas deferens (n)	The tubes that lead from the testes to the urethra, through which sperm is ejaculated.	정관

Term	Definition	Translation
X-linked (adj)	A trait that is influenced or determined by a gene on the “X” chromosome.	X연관

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Family Relationships

For genetic counselors, it is very important to know if a relative is a blood relative or a relative by marriage.

Term	Definition	Translation
Parent	Your mother or father.	부모
Mother	For genetic purposes, the woman whose egg was fertilized and grew to be you.	어머니
Mother-in-law	Your husband or wife's mother.	시아머니, 장모님
Stepmother	Your father's wife who is not your biological mother.	새어머니
Adoptive mother	A woman who is not your biological mother but who accepted legal responsibility for and raised you.	양어머니
Godmother	A woman chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.	대모
Father	For genetic purposes, the man whose sperm fertilized the egg that grew to be you.	아버지
Father-in-law	Your husband or wife's father.	시아아버지, 장인어른
Stepfather	Your mother's husband who is not your biological father.	새아버지
Adoptive father	A man who is not your biological father but who accepted legal responsibility for and raised you.	양아버지
Godfather	A man chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.	대부
Aunt	Your mother or father's sister or sister-in-law.	고모, 이모
Maternal aunt	Your mother's sister or sister-in-law.	이모
Paternal aunt	Your father's sister or sister-in-law.	고모

Term	Definition	Translation
Uncle	Your mother or father's brother or brother-in-law.	삼촌, 외삼촌
Maternal uncle	Your mother's brother or brother-in-law.	외삼촌
Paternal uncle	Your father's brother or brother-in-law.	삼촌
Cousin	Usually understood to be a first cousin.	사촌
First cousin	Your aunt or uncle's child.	사촌
First cousin once removed	Your aunt or uncle's grandchild or your first cousin's child	사촌의 자녀
Second cousin	The children of first cousins are second cousins to each other.	육촌
Grandparent	Your parent's father or mother.	조부모
Grandmother	Your mother or father's mother.	할머니
Maternal grandmother	Your mother's mother.	외할머니
Paternal grandmother	Your father's mother.	친할머니
Grandfather	Your mother or father's father.	할아버지
Maternal grandfather	Your mother's father.	외할아버지
Paternal grandfather	Your father's father.	친할아버지
Great aunt	Your mother or father's aunt.	조부모의 여자 형제
Maternal great aunt	Your mother's aunt.	할머니의 여자 형제
Paternal great aunt	Your father's aunt.	할아버지의 여자 형제
Great uncle	Your father or mother's uncle.	조부모의 남자 형제
Maternal great uncle	Your mother's uncle.	할머니의 남자 형제
Paternal great uncle	Your father's uncle.	할아버지의 남자 형제
Great grandparents	The parents of any of your grandparents.	증조부모

Term	Definition	Translation
Great grandmother	The mother of any of your grandparents.	증조모
Great grandfather	The father of any of your grandparents.	증조부
Siblings	The children of your father and mother.	형제,자매
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.	이복 형제,자매
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.	의붓 형제,자매
Twins	Two siblings born at the same time	쌍둥이
Identical twins	A twin that developed from the same egg and sperm, meaning that they are mostly genetically identical. Also called monozygotic twins.	일란성 쌍둥이
Fraternal twins	A twin who developed from a different egg and sperm than, meaning that he or she is genetically different and has the same number of shared genes as any other sibling. Also called dizygotic twins.	이란성 쌍둥이
Triplets	Three siblings born at the same time	삼둥이
Sister	A sibling who is a girl.	여자 형제
Sister-in-law	Your brother's wife.	남자 형제의 아내 또는 남편의 여자 형제
Half sister	A girl who is the either the child of your father with a different mother, or the child of your mother with a different father.	이복 여자 형제
Stepsister	The daughter of your stepmother or stepfather and therefore not biologically related to you.	의붓 여자 형제

Term	Definition	Translation
Brother Brother-in-law Half brother Stepbrother	A sibling who is a boy. Your sister's husband. A boy who is the either the child of your father with a different mother, or the child of your mother with a different father. The son of your stepmother or stepfather and therefore not biologically related to you.	남자 형제 여자 형제의 남편 또는 아내의 남자 형제 이복 남자 형제 의붓 남자 형제
Niece	Your sibling's daughter.	여조카
Nephew	Your sibling's son.	남조카
Spouse	Your husband or wife.	배우자
Wife	The woman to whom you are married.	아내
Husband	The man to whom you are married.	남편
Children	Genetically speaking, the people who are produced from your egg or sperm.	자식
Daughter Daughter-in-law Stepdaughter Adoptive daughter Goddaughter	Your child who is a girl. Your son's wife. Your spouse's daughter who is not your biological child. A girl for whom you have accepted legal responsibility and raised even though she is not your biological child. A girl for whom you have accepted spiritual responsibility within a ceremony of the Catholic church; she may or may not be biologically related to you.	딸 며느리 의붓딸 입양딸 대녀

Term	Definition	Translation
Son	Your child who is a boy.	아들
Son-in-law	Your daughter's husband.	사위
Stepson	Your spouse's son who is not your biological child.	의붓아들
Adoptive son	A boy for whom you have accepted legal responsibility and raised even though he is not your biological child.	입양아들
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.	대자
Ancestor	A person from whom you are descended, usually more remote than a grandparent.	조상
Fiancé(e)	The person whom you have promised to marry. Man: fiancé Woman: fiancée	약혼자
Domestic partner	The person with whom you are living and have an intimate relationship, but to whom you are not married.	동거인
Divorced	Having ended a marriage.	이혼
Engaged	Having promised to marry someone.	약혼
Widow	A woman whose husband has died.	과부
Widower	A man whose wife has died.	편부
Relative	A person in your family.	친척
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.	혈연
Next of kin	Your closest living blood relation.	최근친

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Analogies

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

Examples:

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

“It’s as if you were reading through a long book and looking for one typo, for a letter that is mistaken.”

“So, a mutation is like an unexpected change in the order of the letters.”

“A gene is like a recipe, and mutations are like changes in that recipe.”

“It’s like chapters in a book, and like misspellings.”

“Our cells are like libraries and our chromosomes are like books.”

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

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

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

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

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

“It’s as if you were reading through a long book and looking for one typo, for a letter that is mistaken.”

“So a mutation is like an unexpected change in the order of the letters.”

“It’s like chapters in a book, and like misspellings.”

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

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

“A gene is like a recipe, and mutations are like changes in that recipe.”

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

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

Index

A

A-B-C analysis	7
Abnormal	27, 53
Abortion.....	27, 53
Abstract language	7
ACMG 59.....	27
Adaptive behavior or adaptive skills.....	7
Adaptive skills	27
Adoption	27, 53
Adoptive daughter.....	48, 72
Adoptive father.....	45, 69
Adoptive mother	45
Adoptive son.....	49
Age of majority	7
Altered gene copy.....	33
American College of Medical Genetics and Genomics (ACMG)	27
Amniocentesis	27, 53
Amniotic fluid	28, 53
Ancestor.....	49, 73
Anencephaly	28, 53
Anxiety disorder	7
Applied Behavior Analysis (ABA)	7
Apraxia.....	7
At average risk	40, 63
At high risk.....	40, 63
At higher risk than	40, 63
At risk.....	40, 63
Attention Deficit Hyperactivity Disorder (ADHD)	8
Audiologist.....	8
Augmentative and Alternative Communication (AAC)	8
Aunt	45, 69
Autism Spectrum Disorder (ASD).....	8
Aversive	8

B

Behavior Intervention Plan (BIP)	8
Behavioral disorder	8
Benign.....	28, 53
Biochemical testing.....	28
Biomarker	8
Bipolar disorder	8
Birth canal.....	28, 53
Birth defect	28, 54
Blood relation	49
Blood Relation	73

Blood test.....	28, 54
Board Certified Behavior Analyst (BCBA).....	9
Body language	9
Brace	28
Brother.....	48, 72
Brother-in-law	48
Buccal swab	28, 54

C

Carrier	28, 54
Case manager	9
Catatonia	9
Cell.....	28, 54
Cervix	29, 54
Challenging behaviors.....	9
Children	48, 72
Chorionic Villus Sampling (CVS).....	29, 54
Chromosome	29, 54
Chromosome test / Karyotype	29, 54
Civil commitment.....	9
Cleft lip.....	29, 54
Cleft palate	29, 55
Clinically significant	29, 55
Cognition	29
Cognitive behavioral therapy	9
Cognitive development.....	9
Communication	9
Communication disorder	9
Comorbid	9
Compulsion.....	10
Condition	29
Condition (e.g., “Genetic condition”)	55
Consanguinity	29, 55
Conservatorship.....	10
Consultation model	29
Continuing care model	29
Cousin	46, 70
Crisis plan.....	10
Cystic fibrosis	30, 55

D

Data analysis.....	10
Daughter	48, 72
Daughter-in-law.....	48
De novo.....	30
De-escalation	10
Deleterious mutation.....	30, 55
Deletion	30, 55
Depression.....	10

Developmental assessment.....	10, 30
Developmental delay.....	10
Developmental Disability (DD)	10
Developmental pediatrician	11
Diagnostic test.....	30, 55
Differential diagnosis.....	11
Disease-causing mutation.....	55
Disruption	11
Divorced.....	49, 73
DNA (Deoxyribonucleic Acid).....	30, 56
DNA marker (also called a “Genetic marker”)	30, 56
DNA mutation.....	30, 56
DNA sequence	31, 56
DNA sequencing	31, 56
Domestic partner.....	49, 73
Dominant.....	31, 56
Donor egg or sperm.....	31, 56
Down syndrome (Trisomy 21)	11, 31, 57
Dual diagnosis.....	11
Duplication.....	31, 57
Dysarthria	11
Dyspraxia	11
<i>E</i>	
Early intervention	11
Echolalia or Echophrasia.....	11
Edwards syndrome (Trisomy 18)	31, 57
Egg.....	31, 57
Elopement	11
Embryo	31, 57
Engaged	49, 73
Environmental causes or factors	31
Epilepsy.....	11
Escalating.....	12
Evidence-Based Practice (EBP)	12
Executive control / executive functioning	12
Exome sequencing.....	32, 35
Exon	32
Extinction.....	12
Extinction burst.....	12
<i>F</i>	
Face blindness	12
Fading	12
Fallopian tube.....	32, 57
False negative	32, 57
False positive	32, 57
Familial.....	32, 57
Family history	32, 57

Father	45, 69
Father-in-law	45
Fecal digging	12
Fecal smearing	12
Fertilization	32, 57
Fetal Alcohol Spectrum Disorder (FASD)	12
Fetal Alcohol Syndrome (FAS)	12
Fetal surgery	32, 58
Fetus	32, 58
Fiancé.....	73
Fiancé(e)	49
First cousin.....	46
First cousin once removed.....	46
First trimester screening.....	32, 58
Flip a coin.....	32, 58
Fragile X syndrome	33, 58
Fragment	33, 58
Fraternal twins.....	47, 71
Function of behavior.....	12
Functional Behavior Analysis (FBA)	13
Functional communication	13
G	
Gene	33, 58
Gene copy	33
Gene deletion	33
Gene panel test	35
Gene panel testing.....	33
Gene variant	33
General population.....	33, 58
Generalization	13
Generation.....	34, 58
Genetic alphabet	34
Genetic counseling	34, 59
Genetic counseling intern.....	59
Genetic counselor.....	34, 59
Genetic discrimination.....	34, 59
Genetic factors	34, 59
Genetic information	34, 59
Genetic material	35, 59
Genetic predisposition.....	35, 59
Genetic test	35, 60
Genetic trait.....	35, 60
Genetic variant	60
Geneticist.....	35, 60
Genetics.....	35, 59
Genome	36
Genome sequencing.....	35, 36
Germline testing	36

Gestures.....	13
Goddaughter.....	48
Godfather	45
Godmother	45
Godson.....	49
Grandfather	46, 70
Grandmother	46, 70
Grandparent	46, 70
Great aunt.....	46, 70
Great grandfather	47, 71
Great grandmother.....	46, 71
Great grandparents	46, 70
Great uncle	46, 70
Guardianship	13, 36
<i>H</i>	
Half brother	48
Half siblings.....	47
Half sister	47, 71
Helix.....	36
Hereditary.....	36, 60
Hereditary material	36, 60
Huntington’s disease	36
Husband.....	48, 72
Hypersensitivity	13
Hyposensitivity	13
<i>I</i>	
Identical twins	47, 71
Idiosyncratic language/ vocabulary	14
In Vitro Fertilization (IVF).....	60
In Vitro Fertilization (IVF).....	36
Incontinence	14
Individual gene testing	35, 37
Individualized Education Plan (IEP).....	36
Individualized Education Program (IEP).....	14
Individualized Family Service Plan (IFSP)	14
Individuals with Disabilities Education Act (IDEA)	14
Infertility	37, 60
Informed consent	37, 60
Inheritance pattern.....	37, 60
Inherited	37, 60
Insertion.....	60
Intellectual disability.....	15, 37, 60
Interdisciplinary evaluation team	15
Intervention	15
Involuntary commitment.....	15

J

Joint attention15

K

Klinefelter's syndrome (47, XXY)37, 61

L

Language delay15

Language disorder15

Learning disability15

Least Restrictive Environment (LRE)15

Lynch syndrome37

M

Maladaptive behavior15

Marker chromosome37, 61

Maternal aunt45, 69

Maternal grandfather46

Maternal grandmother46

Maternal great aunt46

Maternal great uncle46, 70

Maternal Serum Screening (MSS or maternal serum alphetoprotein test)37, 61

Maternal uncle45

Medicaid16

Medical home16

Mental health hold16

Metastasis37

Microarray35, 38, 61

Miscarriage38, 61

Molecule38

Mosaicism38, 61

Mother45, 69

Mother-in-law45, 69

Motor Abilities38

Motor skill16

Multifactorial38, 61

Mutation38

N

Nephew48, 72

Neurodevelopmental conditions16

Next of kin49

Next-Of-Kin73

Niece48, 72

Non-Invasive Prenatal Testing (NIPT)38, 62

Nonverbal communication16

Normal gene copy33

Nuchal thickening38, 62

Nutritionist	16
<i>O</i>	
Obsession.....	16
Obsessive compulsive disorder (OCD)	16
Occupational therapist (OT)	16
Open neural tube defect	39, 62
Ototoxic	17
Ova.....	39, 62
Ovary	39, 62
Overcorrection.....	17
<i>P</i>	
Packets of genetic information.....	39
Parent	45
Paternal aunt	45
Paternal grandfather	46
Paternal grandmother	46
Paternal great aunt.....	46
Paternal great uncle	46
Paternal uncle.....	45
Pathogenic.....	39
Pediatric Autoimmune Neuropsychiatric Disorders Associated with Streptococcal infections (PANDAS)	17
Pedigree.....	39
Penetrance.....	39, 62
Phenylketonuria (PKU).....	17
Physical therapist (PT)	17
Pica	17
Picture Exchange Communication Systems (PECS).....	17
Placenta	39, 62
Polymorphism.....	43, 65
Polypharmacy	17
Positive Behavior Supports (PBS)	17
Post-Traumatic Stress Disorder (PTSD).....	17
Prematurity.....	17
Presymptomatic.....	39, 62
Prognosis	39, 62
Prompting.....	18
Protein	39, 62
Psychologist.....	18
Psychosis.....	18
Psychotropic	18
Puberty	18
<i>R</i>	
Random	39, 63
Recessive	40, 63
Red flag.....	40, 63
Red flags	18

Reinforce	18
Reinforcement strategies	18
Relative	49, 73
Replicate	40, 63
Reproductive history	40, 63
Resilience	18
Respite care	18
Restraints	18
Restricted or repetitive behaviors or interests	18
Reward	18
Ring chromosome	40, 63
Risk	40, 63
Risk factor	18, 40, 63
Ritual	18
Rumination	19
Runs in the family	40, 64
S	
Saliva (spit) sample	40
Schizophrenia	19
Screen	41, 64
Screening test	41, 64
Screening test or tool	19
Seclusion	19
Second cousin	46
Sedating	19
Seizure disorder	19
Self-advocacy	19
Semen	41, 64
Seminal fluid	41, 64
Sensory abilities	41
Sensory avoidance	19
Sensory defensiveness	19
Sensory input	19
Sensory processing disorder	19
Sensory-seeking behavior	19
Siblings	47, 71
Sickle cell disease	41, 64
Single site analysis	41
Single site gene testing	35
Sister	47, 71
Sister-in-law	47
Sleep apnea	19
Sleep disturbances	20
Social communication disorder	20
Social reciprocity	20
Social work or social worker	20
Somatic genetic changes	41
Son	49, 73

Son-in-law49

Special education41

Special Needs Parent Advocate20

Specific learning disability20

Speech generating device20

Speech sound disorder20

Speech-language pathologist20

Sperm.....42, 64

Sperm donation42, 64

Spina bifida42, 65

Spouse48, 72

Standardized test21

Staring spells21

Statistically significant42, 65

Stepbrother48

Stepdaughter48, 72

Stepfather45, 69

Stepmother45, 69

Stepsiblings47, 71

Stepsister47

Stepson49, 73

Stereotypy21

Stimulation21

Supplemental Security Income (SSI)21

Supportive seating42

T

Tangibles21

Teratogen42, 65

Termination of pregnancy42, 65

Test result43, 65

Testes43, 66

Time delay21

To flip a coin58

Tourette syndrome or Tourette’s syndrome21

Toxic exposure43, 66

Tracking scales21

Trait43, 66

Transition plan / transition services21

Translocation44, 66

TRICARE21

Triplets47, 71

Twins47, 71

Typo44, 66

U

Ultrasound44, 66

Uncle45, 70

Uterus44, 66

V

Vagina44, 66
 Variant44, 66
 Variant of uncertain significance43, 65
 Vas deferens44, 66
 Visual cues22
 Visual schedules.....22
 Vocational rehabilitation22

W

Widow49, 73
 Widower49, 73
 Wife48, 72
 Wraparound22

X

X-linked44, 67