



National Coordinating Center
for the Regional Genetics Networks

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

Glossary of Pediatric Genetics Terms

English – Tagalog

2022 Edition

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

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Acknowledgments

Mission of the NCC

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

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

The Cross Cultural Health Care Program

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

<https://xculture.org>

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

Table of Contents

Preface.....	5
How to Use the Glossary.....	5
Glossary of Terms.....	7
Family Relationships.....	25
Index.....	31



Preface

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

How to Use the Glossary

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

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

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Glossary of Terms

English – Tagalog

(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.	Hindi-normal
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.”	Pagpapalaglag
ACMG 59 (n)	Fifty-nine genes that the American College of Medical Genetics and Genomics has identified in which variances have a high risk of causing disease – but disease that can be mitigated through early detection or treatment. The ACMG recommends that variances in these genes should be reported to patients even if finding them was not the reason for genetic testing	ACMG 59
Adaptive skills (n)	Skills/abilities needed to perform daily activities, particularly those that allow an individual to live independently and/or work. Examples: brushing teeth, bathing, dressing, food shopping, and taking public transportation.	Mga kasanayan sa pag-angkop
Adoption (n)	The process of accepting legal responsibility for someone else’s child to raise as your own.	Pag-ampon
American College of Medical Genetics and Genomics (ACMG) (n)	A professional membership organization based in the United States that represents the interests of clinical geneticists, clinical laboratory geneticists, and genetic counselors. The ACMG focuses on improving patient care, establishing standards of care and laboratory policy, and educating members about advances in the field.	American College of Medical Genetics and Genomics (ACMG)
Amniocentesis (n)	A procedure by which a sample of amniotic fluid is withdrawn from the amniotic sac. This is usually done by inserting a long needle through the abdominal and uterine walls with the guidance of ultrasound.	Amniocentesis

Glossary of Pediatric Genetics Terms – Tagalog

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.	Panubigan
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.	Anencephaly o depekto sa utak
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.	Hindi-nakapipinsala
Biochemical testing (n)	Blood tests to identify elevated levels of certain naturally occurring substances/chemicals in the body that are linked to genetic conditions.	Pagsusuri ng biochemical
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.	Daanan ng bata sa panganganak
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.”	Depekto sa pagsilang
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.	Pagsusuri sa dugo
Brace (n)	A medical appliance that provides support to joints or bones, to weak muscles, or to strained ligaments.	Brace
Buccal swab (n)	A way to collect DNA from the cells on the inside of a person's cheek.	Pag-swab sa bunganga
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.	Tagapagdala
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.	Selula

Glossary of Pediatric Genetics Terms – Tagalog

Term	Definition	Translation
Cervix (n)	The narrow lower part of the uterus than connects to the vagina.	Kuwelyo ng matris
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.	Chorionic Villus Sampling (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.	Chromosome
Chromosome test / Karyotype (n)	A test that looks at the number and arrangement of chromosomes in a cell.	Pagsusuri ng Chromosome / Karyotype
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.	Bungi
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.	Biyak na ngalangala
Clinically significant (adj)	If a test result is “clinically significant,” the result indicates a medical problem that can impact a person’s life.	Klinikal na napakahalaga
Cognition (n)	The processes involved in thinking, learning, understanding, and remembering.	Kaalaman
Condition (e.g., “genetic condition”) (n)	A long-term medical health issue.	Kondisyon (hal., “genetic na kondisyon”)
Consanguinity (n)	When parents are blood relatives to each other.	Kadugo o magkamag-anak
Consultation model (n)	The model of care for patients with genetic conditions in which the genetic team acts as consultants, supporting the care being provided principally by the patient’s Primary Care Provider.	Modelo ng konsultasyon
Continuing care model (n)	The model of care for patients with genetic conditions in which the genetic team takes primary responsibility for supervising and coordinating all care related to the patient’s genetic condition.	Modelo ng pagpapatuloy ng pangangalaga

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.	Cystic fibrosis
De novo (adj)	Genetic changes that are not inherited but that occur during cell division.	De novo
Deleterious mutation/ Disease-causing mutation (n)	A change in a person’s DNA that may cause a medical condition.	Pagbura ng mutasyon/ Sakit na nagdudulot ng mutasyon
Deletion (n)	Having a section of genetic information (DNA) missing.	Pagbura
Developmental assessment (n)	An evaluation of how an individual patient’s intellectual, emotional, and social development compare with others of the same age.	Pagsusuri sa pag-develop
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.	Diagnostic na pagsusuri
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 (Deoxyribonucleic Acid)
DNA marker (also called a “Genetic marker”) (n)	A readily recognizable genetic trait, gene, or DNA segment.	DNA marker (tinatawag rin na “Genetic marker”)
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.	Mutasyon ng DNA
DNA sequence (n)	The exact arrangement of the chemicals that make up a section of DNA.	Sekwensiya ng DNA

Term	Definition	Translation
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.	Pag-sekwensiya ng DNA
Dominant (adj)	A genetic trait in which one copy of the gene is sufficient for a trait to be expressed. In a dominant genetic condition, if one copy of the gene has a mutation, the personal will be affected with the condition.	Nangingibabaw
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.	Nagbigay ng itlog o semilya
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.	Down syndrome (Trisomy 21)
Duplication (n)	Having an extra section of genetic information (DNA). A <i>duplication</i> occurs when part of a chromosome is copied abnormally, resulting in extra <i>genetic</i> material.	Pagkopya
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.	Edwards syndrome (Trisomy 18)
Egg (n)	The reproductive cells of a woman. When fertilized by sperm, the egg will grow into an embryo. Also called an "ovum," plural "ova."	Itlog
Embryo (n)	An unborn mammal, between conception and 8 weeks of gestation.	Binhi
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.	Mga sanhi o salik sa kapaligiran
Exome sequencing (n)	A genetic test in which a patient's exons are sequenced and studied.	Pag-sekwensiya ng exome

Glossary of Pediatric Genetics Terms – Tagalog

Term	Definition	Translation
Exon (n)	The part of the genome that codes for proteins. The majority of deleterious mutations occur in exons.	Exon
Fallopian tube (n)	The tube that connects an ovary to the uterus.	Tubong palopyan
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.	Negatibong mali
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.	Maling positibo
Familial (adj)	Belonging to a family, e.g., a familial trait is a trait that is shared by family members and may be due to genetic or environmental factors or both.	Familial o sa pamilya
Family history (n)	The medical history of the members of a biological family.	Kasaysayan ng pamilya
Fertilization (n)	The joining of an egg and sperm to create the first cell that will develop into an embryo, then fetus, then baby.	Pagpapabunga
Fetal surgery (n)	Surgery conducted on a fetus while it is still in the uterus.	Operasyon ng sanggol na ipinagbubuntis
Fetus(n)	An unborn mammal, between 8 weeks of gestation to birth.	Ipinagbubuntis na sanggol
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.	Pagpitik ng barya
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.	Pagpapasuri sa unang trimester
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.	Fragile X syndrome

Term	Definition	Translation
Fragment (n)	A small piece; an incomplete part of a whole.	Maliit na piraso
Gene (n)	A specific sequence of DNA that codes for one or many functions within the cell and body.	Gene
<p data-bbox="201 422 501 453">Gene copy (n)</p> <p data-bbox="250 680 501 711">Normal gene copy (n)</p> <p data-bbox="250 812 501 844">Altered gene copy (n)</p>	<p data-bbox="518 422 1122 642">In human cells, DNA is arranged in 23 pairs of chromosomes, for a total of 46 chromosomes. These chromosomes contain all the genes that make up the DNA. One chromosome of each pair is inherited from each parent. As such, individuals have two copies of every gene, one inherited from the mother and one inherited from the father.</p> <p data-bbox="518 680 1122 774">A normal copy of a gene is one that is found most frequently in the population. Also called the “working copy.”</p> <p data-bbox="518 812 1122 907">An altered copy of a gene is one that differs from the working copy and is considered to be “abnormal.” Also called the “non-working copy.”</p>	<p data-bbox="1138 422 1435 453">Kopya ng gene</p> <p data-bbox="1162 680 1435 743">Normal na kopya ng gene</p> <p data-bbox="1162 812 1435 875">Nabagong kopya ng gene</p>
Gene deletion (n)	Having a piece of genetic information missing from a gene.	Pagkabura ng gene
Gene panel testing (n)	Genetic testing that looks at 2 or more specific genes known to be associated with particular conditions.	Pagsusuri sa panel ng gene
Gene variant (n)	A change from the typical DNA sequences. A genetic variant can be benign, deleterious or of uncertain significance. Also called “mutation”, although “genetic variant” is becoming the more common usage.	Variant ng gene
General population (n)	<p data-bbox="518 1276 688 1308">“Most people.”</p> <p data-bbox="518 1346 1122 1541">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>	Pangkalahatang populasyon

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>	Henerasyon
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>	Genetic na alpabeto
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>	Pagpapayo sa genetic
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>	Tagapayo sa genetic
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>	Diskriminasyon sa genetic
Genetic factors (n)	<p>Specific aspects of a person’s genetic make-up that influence that person’s health and development.</p>	Mga salik sa genetic
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>	Impormasyon sa genetic
Genetic material (n)	<p>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.</p>	Genetic na materyal

Term	Definition	Translation
Genetic predisposition (n)	An increased chance of a person developing a certain trait or disease based on that person's particular genetic makeup.	Genetic na predisposition o tendensiya
Genetic test (n)	A laboratory test designed to determine if a person has a gene mutation or a typical DNA sequence.	Pasusuri sa genetic
Single site gene testing (n)	Single site gene testing: testing at a specific point in the genome. This is usually done to look for a mutation found in a family member or when one specific mutation is known to cause a genetic condition.	Single site gene na pagsusuri
Individual gene testing (n)	Individual gene testing: genetic analysis of the entirety of one specific gene.	Indibidawal ng pagsusuri ng gene
Gene panel test (n)	Gene panel test: analysis of several genes that have been shown to be associated with a particular condition, e.g., breast cancer gene panel.	Pagsusuri sa gene panel
Microarray (n)	Microarray: genetic analysis that looks for unexpected numbers of specific genes (two copies of every gene code is usually expected).	Microarray
Exome sequencing (n)	Exome testing: a genetic test in which the exons of a patient's genetic code are sequenced and studied.	Pag-sekwensiya ng exome
Genome sequencing (n)	Genome sequencing: a genetic test in which the entire genetic code (the genome) of a patient is mapped out and studied.	Pag-sekwensiya ng genome
Genetic trait (n)	A characteristic within a family that is passed down from parent to child through their DNA.	Genetic na katangian
Geneticist (n)	A doctor or scientist who studies genetics.	Dalubhasa sa genetics
Genetics (adj)	The science of how an organism's genes interact with the environment to produce certain traits.	Agham tungkol sa genetics
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.	Pag-aaral tungkol sa genetics
Genome (n)	The entire genetic code of an individual.	Genome
Genome sequencing (n)	A genetic test in which the entire genetic code of a patient is mapped out and studied.	Pag-sekwensiya ng genome

Term	Definition	Translation
Germline testing (n)	<p>Germline testing refers to the analysis of a person's DNA, which he or she inherited from his or her parents.</p> <p>In biology and genetics, the germline is the group of cells that will pass on the genetic material to children, in other words, the cells from which the eggs and sperm come.</p>	Pagsusuri sa germline
Guardianship (n)	A legal arrangement, supervised by a court, in which one person takes responsibility for a minor or an adult with a disability.	Tagapangalaga
Helix (n)	Something that is spiral in form. DNA is often described as being a "double helix."	Pilipit
Hereditary (adj)	Passed down from parent to child.	Namamana
Hereditary material (n)	Genetic material that is passed down from parent to child.	Namamanang katangian
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.	Huntington's disease
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.	Individualized Education Plan (IEP)
In Vitro Fertilization (IVF) (n)	<p>The fertilization of an egg by a sperm outside of a woman's body.</p> <p>The process involves extracting eggs from a woman's ovaries, collecting sperm from a man, and combining a sperm and egg in a laboratory dish. The resulting fertilized egg is usually then implanted in a woman's uterus so that it can develop into a baby.</p>	In Vitro Fertilization (IVF)
Individual gene testing (n)	Genetic analysis of the entirety of one specific gene.	Indibidawal ng pagsusuri ng gene
Infertility (n)	The inability to have children.	Pagkabaog

Term	Definition	Translation
Informed consent (n)	The process of agreeing to a procedure or course of treatment after understanding what the procedure/treatment entails, the potential risks and benefits associated with it, and the other options available.	May kabatirang pagsang-ayon
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.	Pattern na namana
Inherited (adj)	Passed down from parent to child.	Minana
Insertion (n)	An extra segment of DNA added in at a place where it is not usually found.	Pagsingit
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.	Intelektwal na kapansanan
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.	Klinefelter's syndrome (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.	Lynch syndrome
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.	Marker chromosome
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.	Maternal Serum Screening (MSS o Maternal serum alpha-fetoprotein na pagsusuri)
Metastasis (n)	The spreading of cancer from one organ to another.	Pagkalat

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.	Microarray
Miscarriage (n)	The spontaneous (not intentional) loss of a pregnancy. See “abortion.” Also called pregnancy loss.	Nakunan
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.	Molecule
Mosaicism (n)	A condition in which some, but not all, cells in a sample show a genetic difference.	Mosaicis
Motor Abilities (n)	The ability to move and use one’s muscles.	Kakayahan sa Paggalaw
Multifactorial (adj)	Due to a combination of genetic and non-genetic (environmental, hormonal, etc.) risk factors that act together to determine risk.	Multifactorial
Mutation (n)	A change in a gene, usually deleterious. See “genetic variant.”	Mutasyon
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.	Non-Invasive Prenatal Testing (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.	Nuchal thickening

Glossary of Pediatric Genetics Terms – Tagalog

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.	Open Neural Tube Defect (ONTD)
Ova (n)	See “egg.”	Ova o itlog
Ovary (n)	The organ in a woman that stores and releases eggs. There are normally two.	Obaryo
Packets of genetic information (n)	A phrase genetic counselors use to describe genes or chromosomes.	Mga impormasyon tungkol sa genetic
Pathogenic (adj)	Disease causing.	Pathogenic o nagdudulot ng sakit
Pedigree (n)	A family tree that can be used to trace the inheritance of specific genetic traits.	Pedigree
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.	Pag-penetrance
Placenta (n)	The organ that develops together with an embryo in a pregnant woman’s uterus to nourish the embryo through the umbilical cord.	Inunan
Presymptomatic (adj)	Before symptoms appear.	Wala pang sintomas
Prognosis (n)	The most likely outcome of a disease process.	Posibleng kalalabasan
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.	Protina
Random (adj)	Happening in an unpredictable way.	Hindi tiyak

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>	Recessive
Red flag (n)	A warning sign or a clue.	Babala
Replicate (v)	To copy.	Pag-kopya
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.	Kasaysayan sa reproduktibo
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.	Ring chromosome
Risk (n)	The chance that something will happen; in the case of prenatal genetics, the chance that the child will have a genetic condition.	Panganib
At risk (adj)	Has some possibility of having a genetic condition.	Nanganganib
At average risk (adj)	Has the same possibility of having a genetic condition as the general population.	Sa katamtamang antas ng panganib
At high risk (adj)	Has a greater possibility of having a genetic condition than the general population.	Nasa mataas na panganib
At higher risk than "X" (adj)	Has a greater possibility of having a genetic condition than "X."	Nasa mas mataas na panganib kaysa sa "X"
Risk factor (n)	A circumstance that increases the risk of having a genetic condition.	Salik ng panganib
Runs in the family	Passed down from parents to children to grandchildren.	Nasa lahi ng pamilya
Saliva (spit) sample (n)	A way to collect DNA from the cells in saliva (spit).	Sample ng laway (dura)

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.	Pag-screen
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>	Screening na pagsusuri
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.	Binhi
Seminal fluid (n)	See “semen.”	Fluid ng binhi
Sensory abilities (n)	The ability to see, hear, touch, taste, and smell.	Kakayahan sa pandama
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.	Sickle cell na sakit
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.	Single site na pag-analysis
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.	Somatic genetic na mga pagbabago
Special education (n)	Education that is tailored to individuals with developmental delays or other learning disabilities.	Espesyal na edukasyon
Sperm (n)	The reproductive cells of the man. When sperm fertilize a woman’s egg, a baby develops.	Semilya

Glossary of Pediatric Genetics Terms – Tagalog

Term	Definition	Translation
Sperm donation (n)	The process through which a man allows his sperm to be collected and used to fertilize the eggs of a woman who is not his sexual partner.	Donasyon ng semilya
Spina bifida (n)	A congenital defect of the spine in which part of the spinal cord is exposed through an opening in the bone structure. This may result in nerve damage and some degree of paralysis in the legs.	Spina bifida
Statistically significant (adj)	A measurement of whether the findings of research are meaningful. Refers to the likelihood that a relationship between two factors is linked by something other than chance.	Istatistika na napakahalaga
Supportive seating (n)	Seating, such as a wheelchair, that helps an individual function in their environment and that prevents worsening of their condition.	Pang-suporta sa pag-upo
Teratogen (n)	Something that has the potential to disturb the normal development of an embryo or fetus.	Teratogen
Termination of pregnancy (n)	See “abortion.”	Pagpapalaglag sa pinagbubuntis

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

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.	Pagsasaayos
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.	Typo
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.	Ultrasound
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.”	Matris
Vagina (n)	The tube-like part of the female reproductive system that extends from the cervix to the outside of the body.	Ari ng babae
Variant (n)	A version of something that differs from the norm. So, a genetic variant is a change to the usual genetic sequence.	Variant
Vas deferens (n)	The tubes that lead from the testes to the urethra, through which sperm is ejaculated.	Vas deferens
X-linked (adj)	A trait that is influenced or determined by a gene on the “X” chromosome.	X-linked

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.	Magulang
Mother	For genetic purposes, the woman whose egg was fertilized and grew to be you.	Ina
Mother-in-law	Your husband or wife's mother.	Biyenang-babae
Stepmother	Your father's wife who is not your biological mother.	Madrasta
Adoptive mother	A woman who is not your biological mother but who accepted legal responsibility for and raised you.	Inang umampon
Godmother	A woman chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.	Ninang
Father	For genetic purposes, the man whose sperm fertilized the egg that grew to be you.	Ama
Father-in-law	Your husband or wife's father.	Biyenang-lalaki
Stepfather	Your mother's husband who is not your biological father.	Amain
Adoptive father	A man who is not your biological father but who accepted legal responsibility for and raised you.	Amang umampon
Godfather	A man chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.	Ninong
Aunt	Your mother or father's sister or sister-in-law.	Tiyahin
Maternal aunt	Your mother's sister or sister-in-law.	Tiyahin sa panig ng ina
Paternal aunt	Your father's sister or sister-in-law.	Tiyahin sa panig ng ama
Uncle	Your mother or father's brother or brother-in-law.	Tiyuhin
Maternal uncle	Your mother's brother or brother-in-law.	Tiyuhin sa panig ng ina
Paternal uncle	Your father's brother or brother-in-law.	Tiyuhin sa panig ng ama

Term	Definition	Translation
Cousin	Usually understood to be a first cousin.	Pinsan
First cousin	Your aunt or uncle's child.	Unang pinsan
First cousin once removed	Your aunt or uncle's grandchild or your first cousin's child	Anak ng pinsan
Second cousin	The children of first cousins are second cousins to each other.	Pangalawang pinsan
Grandparent	Your parent's father or mother.	Lolo at lola
Grandmother	Your mother or father's mother.	Lola
Maternal grandmother	Your mother's mother.	Lola sa panig ng ina
Paternal grandmother	Your father's mother.	Lola sa panig ng ama
Grandfather	Your mother or father's father.	Lolo
Maternal grandfather	Your mother's father.	Lolo sa panig ng ina
Paternal grandfather	Your father's father.	Lolo sa panig ng ama
Great aunt	Your mother or father's aunt.	Tiyahin sa tuhod
Maternal great aunt	Your mother's aunt.	Tiyahin sa tuhod sa panig ng ina
Paternal great aunt	Your father's aunt.	Tiyahin sa tuhod sa panig ng ama
Great uncle	Your father or mother's uncle.	Tiyuhin sa tuhod
Maternal great uncle	Your mother's uncle.	Tiyuhin sa tuhod sa panig ng ina
Paternal great uncle	Your father's uncle.	Tiyuhin sa tuhod sa panig ng ama
Great grandparents	The parents of any of your grandparents.	Lolo at lola sa tuhod
Great grandmother	The mother of any of your grandparents.	Lola sa tuhod
Great grandfather	The father of any of your grandparents.	Lolo sa tuhod

Term	Definition	Translation
Siblings	The children of your father and mother.	Mga kapatid
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.	Mga kapatid sa ama o ina
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.	Mga kapatid na anak ng madrasta o amain
Twins	Two siblings born at the same time.	Kambal
Identical twins	Twins who developed from the same egg and sperm, meaning that they are genetically identical. Also called monozygotic twins.	Identical na kambal
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.	Fraternal na kambal
Triplets	Three siblings born at the same time.	Triplets
Sister	A sibling who is a girl.	Kapatid na babae
Sister-in-law	Your brother's wife or your husband's sister.	Hipag
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.	Kapatid na babae sa ama o ina
Stepsister	The daughter of your stepmother or stepfather and therefore not biologically related to you.	Kapatid na babae sa madrasta o amain
Brother	A sibling who is a boy.	Kapatid na lalaki
Brother-in-law	Your sister's husband or your wife's brother.	Bayaw
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.	Kapatid na lalaki sa ama o ina
Stepbrother	The son of your stepmother or stepfather and therefore not biologically related to you.	Kapatid na lalaki sa madrasta o amain
Niece	Your sibling's daughter.	Pamangkin na babae
Nephew	Your sibling's son.	Pamangkin na lalaki

Term	Definition	Translation
Spouse	Your husband or wife.	Asawa
Wife	The woman to whom you are married.	Misis
Husband	The man to whom you are married.	Mister
Children	Genetically speaking, the people who are produced from your egg or sperm.	Mga anak
Daughter	Your child who is a girl.	Anak na babae
Daughter-in-law	Your son's wife.	Manugang na babae
Stepdaughter	Your spouse's daughter who is not your biological child.	Anak na babae sa dating asawa
Adoptive daughter	A girl for whom you have accepted legal responsibility and raised even though she is not your biological child.	Inamong anak na babae
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.	Inaanak na babae
Son	Your child who is a boy.	Anak na lalaki
Son-in-law	Your daughter's husband.	Manugang na lalaki
Stepson	Your spouse's son who is not your biological child.	Anak na lalaki sa dating asawa
Adoptive son	A boy for whom you have accepted legal responsibility and raised even though he is not your biological child.	Inamong anak na lalaki
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.	Inaanak na lalaki
Ancestor	A person from whom you are descended, usually more remote than a grandparent.	Ninuno
Fiancé(e)	The person whom you have promised to marry. Man: fiancé Woman: fiancée	Kasintahan

Term	Definition	Translation
Domestic partner	The person with whom you are living and have an intimate relationship, but to whom you are not married.	Ka-live in
Divorced	Having ended a marriage.	Diborsiyado
Engaged	Having promised to marry someone.	May kasunduang pakasal
Widow	A woman whose husband has died.	Biyuda
Widower	A man whose wife has died.	Biyudo
Relative	A person in your family.	Kamag-anak
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.	Kadugo
Next of kin	Your closest living blood relation.	Pinakamalapit na kamag-anak

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Index

A

Abnormal	7
Abortion.....	7
ACMG 59.....	7
Adaptive skills	7
Adoption	7
Adoptive daughter.....	28
Adoptive father.....	25
Adoptive mother	25
Adoptive son.....	28
Altered gene copy.....	13
American College of Medical Genetics and Genomics (ACMG)	7
Amniocentesis	7
Amniotic fluid	8
Ancestor.....	28
Anencephaly	8
At average risk	20
At high risk.....	20
At higher risk than	20
At risk.....	20
Aunt	25

B

Benign.....	8
Biochemical testing.....	8
Birth canal.....	8
Birth defect.....	8
Blood relation	29
Blood test.....	8
Brace.....	8
Brother.....	27
Brother-in-law	27
Buccal swab	8

C

Carrier.....	8
Cell.....	8
Cervix	9
Children	28
Chorionic Villus Sampling (CVS)	9
Chromosome	9
Chromosome test / Karyotype	9
Cleft lip.....	9
Cleft palate	9
Clinically significant	9
Cognition	9
Condition	9

Consanguinity	9
Consultation model	9
Continuing care model	9
Cousin	26
Cystic fibrosis	10
<i>D</i>	
Daughter	28
Daughter-in-law	28
De novo.....	10
Deleterious mutation.....	10
Deletion	10
Developmental assessment.....	10
Diagnostic test	10
Divorced.....	29
DNA (Deoxyribonucleic Acid).....	10
DNA marker (also called a “Genetic marker”)	10
DNA mutation.....	10
DNA sequence	10
DNA sequencing	11
Domestic partner	29
Dominant	11
Donor egg or sperm.....	11
Down syndrome (Trisomy 21)	11
Duplication.....	11
<i>E</i>	
Edwards syndrome (Trisomy 18)	11
Egg	11
Embryo	11
Engaged	29
Environmental causes or factors	11
Exome sequencing	11, 15
Exon	12
<i>F</i>	
Fallopian tube	12
False negative	12
False positive	12
Familial.....	12
Family history	12
Father	25
Father-in-law	25
Fertilization.....	12
Fetal surgery	12
Fetus	12
Fiancé(e)	28
First cousin.....	26
First cousin once removed.....	26
First trimester screening.....	12
Flip a coin.....	12
Fragile X syndrome	12
Fragment	13

Fraternal twins.....27

G

Gene13
 Gene copy13
 Gene deletion13
 Gene panel test15
 Gene panel testing.....13
 Gene variant13
 General population.....13
 Generation.....14
 Genetic alphabet14
 Genetic counseling14
 Genetic counselor.....14
 Genetic discrimination.....14
 Genetic factors14
 Genetic information14
 Genetic material14
 Genetic predisposition.....15
 Genetic test15
 Genetic trait.....15
 Geneticist.....15
 Genetics15
 Genome15
 Genome sequencing.....15
 Germline testing16
 Goddaughter.....28
 Godfather25
 Godmother25
 Godson.....28
 Grandfather26
 Grandmother26
 Grandparent26
 Great aunt.....26
 Great grandfather26
 Great grandmother.....26
 Great grandparents26
 Great uncle26
 Guardianship16

H

Half brother27
 Half siblings.....27
 Half sister27
 Helix16
 Hereditary.....16
 Hereditary material16
 Huntington’s disease16
 Husband.....28

I

Identical twins27
 In Vitro Fertilization16

Individual gene testing	15, 16
Individualized Education Plan (IEP).....	16
Infertility	16
Informed consent	17
Inheritance pattern.....	17
Inherited	17
Intellectual disability.....	17
<i>K</i>	
Klinefelter’s syndrome (47, XXY)	17
<i>L</i>	
Lynch syndrome.....	17
<i>M</i>	
Marker chromosome	17
Maternal aunt.....	25
Maternal grandfather	26
Maternal grandmother	26
Maternal great aunt	26
Maternal great uncle	26
Maternal Serum Screening (MSS or maternal serum alphafetoprotein test)	17
Maternal uncle	25
Metastasis.....	17
Microarray	15, 18
Miscarriage	18
Molecule	18
Mosaicism.....	18
Mother.....	25
Mother-in-law.....	25
Motor Abilities	18
Multifactorial	18
Mutation	18
<i>N</i>	
Nephew	27
Next of kin	29
Niece.....	27
Non-Invasive Prenatal Testing (NIPT)	18
Normal gene copy.....	13
Nuchal thickening	18
<i>O</i>	
Open neural tube defect	19
Ova.....	19
Ovary	19
<i>P</i>	
Packets of genetic information.....	19
Parent	25
Paternal aunt	25
Paternal grandfather	26
Paternal grandmother	26

Paternal great aunt.....	26
Paternal great uncle	26
Paternal uncle.....	25
Pathogenic.....	19
Pedigree.....	19
Penetrance.....	19
Placenta	19
Polymorphism.....	23
Presymptomatic.....	19
Prognosis	19
Protein	19

R

Random	19
Recessive	20
Red flag.....	20
Relative	29
Replicate	20
Reproductive history	20
Ring chromosome.....	20
Risk.....	20
Risk factor	20
Runs in the family	20

S

Saliva (spit) sample	20
Screen	21
Screening test	21
Second cousin	26
Semen	21
Seminal fluid	21
Sensory abilities.....	21
Siblings.....	27
Sickle cell disease.....	21
Single site analysis	21
Single site gene testing.....	15
Sister	27
Sister-in-law	27
Somatic genetic changes	21
Son.....	28
Son-in-law	28
Special education.....	21
Sperm.....	21
Sperm donation	22
Spina bifida	22
Spouse	28
Statistically significant	22
Stepbrother	27
Stepdaughter	28
Stepfather	25
Stepmother.....	25
Stepsiblings.....	27
Stepsister	27

Stepson	28
Supportive seating	22
<i>T</i>	
Teratogen	22
Termination of pregnancy	22
Test result	23
Testes.....	23
Toxic exposure	23
Trait	23
Translocation	24
Triplets.....	27
Twins.....	27
Typo	24
<i>U</i>	
Ultrasound	24
Uncle.....	25
Uterus	24
<i>V</i>	
Vagina	24
Variant	24
Variant of uncertain significance	23
Vas deferens	24
<i>W</i>	
Widow	29
Widower	29
Wife	28
<i>X</i>	
X-linked.....	24