



National Coordinating Center  
for the Regional Genetics Networks

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

**Glossary of Pediatric Genetics Terms**

**English – Russian**

**2023 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>.

### NCC Funding Acknowledgment

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

### The Cross Cultural Health Care Program

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

<https://xculture.org>

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

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## Preface

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

## How to Use the Glossary

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

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

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## Glossary of Terms

### English – Language

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

| Term  | Definition   | Translation   |
|---|--|---|
| <b>Abnormal (adj)</b>   | Different from what is considered normal, average, or expected, e.g., a gene sequence that is different than that found in most people.  | Аномальный  |
| <b>Abortion (n)</b>   | The deliberate ending of a pregnancy by the removal of an embryo or fetus from the womb.<br><br>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.”   | Аборт   |
| <b>ACMG 59 (n)</b>  | 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 | АКМГ 59   |
| <b>Adaptive skills (n)</b>  | 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.  | Адаптивные навыки   |
| <b>Adoption (n)</b>   | The process of accepting legal responsibility for someone else’s child to raise as your own.   | Усыновление /удочерение                                     |
| <b>American College of Medical Genetics and Genomics (ACMG) (n)</b> | 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.                                   | Американский колледж медицинской генетики и геномики (АКМГ) |
| <b>Amniocentesis (n)</b>  | 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.   | Амниоцентез   |

| Term                           | Definition   | Translation                |
|--------------------------------|--|----------------------------|
| <b>Amniotic fluid (n)</b>      | 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.   | Амниотическая жидкость     |
| <b>Anencephaly (n)</b>         | A neural tube defect that results in insufficient brain growth in a fetus. Babies with anencephaly usually die soon after birth or are stillborn.  | Анэнцефалия                |
| <b>Benign (adj)</b>            | Something that does not threaten health or life. May refer to a type of change to the DNA that does not create health consequences.  | Доброкачественный          |
| <b>Biochemical testing (n)</b> | Blood tests to identify elevated levels of certain naturally occurring substances/chemicals in the body that are linked to genetic conditions.   | Биохимическое тестирование |
| <b>Birth canal (n)</b>         | The passageway from the uterus through the cervix, the vagina, and the vulva through which a baby passes during the birth process.   | Родовые пути               |
| <b>Birth defect (n)</b>        | 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.” | Врожденный порок           |
| <b>Blood test (n)</b>          | A test in which blood is drawn (usually from the patient’s arm) and sent to a laboratory for analysis.<br><br>In genetics, blood tests may provide cells for genetic sequencing, or blood samples may be used to assess things like protein or hormone levels.                                     | Анализ крови               |
| <b>Brace (n)</b>               | A medical appliance that provides support to joints or bones, to weak muscles, or to strained ligaments.   | Бандаж                     |
| <b>Buccal swab (n)</b>         | A way to collect DNA from the cells on the inside of a person's cheek.   | Буккальный мазок           |
| <b>Carrier (n)</b>             | 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.   | Носитель                   |
| <b>Cell (n)</b>                | 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.                                    | Клетка                     |



| Term   | Definition   | Translation   |
|--|--|---|
| <b>Cervix (n)</b>                                | The narrow lower part of the uterus that connects to the vagina.   | Шейка матки   |
| <b>Chorionic Villus Sampling (CVS) (n)</b>       | A procedure during pregnancy in which a sample of cells from the placenta is removed to check for possible genetic abnormalities.  | Проба ворсинчатого хориона (ПВХ) / Биопсия ворсин хориона |
| <b>Chromosome (n)</b>                            | 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. | Хромосома   |
| <b>Chromosome test / Karyotype (n)</b>           | A test that looks at the number and arrangement of chromosomes in a cell.  | Хромосомный тест / кариотип                               |
| <b>Cleft lip (n)</b>                             | 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.                 | Расщелина (незаращение) верхней губы                      |
| <b>Cleft palate (n)</b>                          | 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.                                | Расщелина (незаращение) нёба.                             |
| <b>Clinically significant (adj)</b>              | If a test result is “clinically significant,” the result indicates a medical problem that can impact a person’s life.  | Клинически значимый                                       |
| <b>Cognition (n)</b>                             | The processes involved in thinking, learning, understanding, and remembering.  | Когнитивная деятельность                                  |
| <b>Condition (e.g., “genetic condition”) (n)</b> | A long-term medical health issue.  | Заболевание (например, «наследственное заболевание»)      |
| <b>Consanguinity (n)</b>                         | When parents are blood relatives to each other.  | Кровное родство   |
| <b>Consultation model (n)</b>                    | 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.                                 | Консультационная модель                                   |
| <b>Continuing care model (n)</b>                 | 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.                          | Модель постоянного ухода                                  |

| Term  | Definition   | Translation   |
|---|--|---|
| <b>Cystic fibrosis (n)</b>                                    | 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.   | Муковисцидоз  |
| <b>De novo (adj)</b>  | Genetic changes that are not inherited but that occur during cell division.  | De novo/ Впервые обнаруженный   |
| <b>Deleterious mutation/<br/>Disease-causing mutation (n)</b> | A change in a person’s DNA that may cause a medical condition.   | Патогенная мутация (генетический вариант)/ Мутация (генетический вариант), вызывающая заболевание |
| <b>Deletion (n)</b>   | Having a section of genetic information (DNA) missing.   | Делеция   |
| <b>Developmental assessment (n)</b>                           | An evaluation of how an individual patient’s intellectual, emotional, and social development compare with others of the same age.  | Оценка развития   |
| <b>Diagnostic test (n)</b>                                    | 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. | Диагностический тест  |
| <b>DNA (Deoxyribonucleic Acid) (n)</b>                        | 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.  | ДНК (дезоксирибонуклеиновая кислота)  |
| <b>DNA marker (also called a “Genetic marker”) (n)</b>        | A readily recognizable genetic trait, gene, or DNA segment.  | ДНК-маркер (также называемый «генетический маркер»)   |

| Term                                     | Definition   | Translation                     |
|--|--|---------------------------------|
| <b>DNA mutation (n)</b>                  | <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>         | Мутация ДНК                     |
| <b>DNA sequence (n)</b>                  | The exact arrangement of the chemicals that make up a section of DNA.  | Последовательность ДНК          |
| <b>DNA sequencing (n)</b>                | 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.  | Секвенирование ДНК              |
| <b>Dominant (adj)</b>                    | A genetic trait in which one copy of the gene is sufficient for a trait to be expressed. In a dominant genetic condition, if one copy of the gene has a mutation, the person will be affected with the condition.  | Доминантный                     |
| <b>Donor egg or sperm (n)</b>            | 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.                         | Донорская яйцеклетка или сперма |
| <b>Down syndrome (Trisomy 21) (n)</b>    | <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>                             | Синдром Дауна (трисомия 21)     |
| <b>Duplication (n)</b>                   | 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.  | Дупликация                      |
| <b>Edwards syndrome (Trisomy 18) (n)</b> | 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. | Синдром Эдвардса (трисомия 18)  |
| <b>Egg (n)</b>                           | The reproductive cells of a woman. When fertilized by sperm, the egg will grow into an embryo. Also called an "ovum," plural "ova."  | Яйцеклетка                      |

| Term                                       | Definition   | Translation                  |
|--|--|------------------------------|
| <b>Embryo (n)</b>                          | An unborn mammal, between conception and 8 weeks of gestation.   | Эмбрион                      |
| <b>Environmental causes or factors (n)</b> | 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. | Факторы окружающей среды     |
| <b>Exome sequencing (n)</b>                | A genetic test in which a patient's exons are sequenced and studied.   | Секвенирование экзома        |
| <b>Exon (n)</b>                            | The part of the genome that codes for proteins. The majority of deleterious mutations occur in exons.  | Экзон                        |
| <b>Fallopian tube (n)</b>                  | The tube that connects an ovary to the uterus.   | Фаллопиева труба             |
| <b>False negative (n)</b>                  | 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.   | Ложноотрицательный результат |
| <b>False positive (n)</b>                  | 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.   | Ложноположительный результат |
| <b>Familial (adj)</b>                      | 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.  | Семейный                     |
| <b>Family history (n)</b>                  | The medical history of the members of a biological family.   | Семейный анамнез             |
| <b>Fertilization (n)</b>                   | The joining of an egg and sperm to create the first cell that will develop into an embryo, then fetus, then baby.  | Оплодотворение               |
| <b>Fetal surgery (n)</b>                   | Surgery conducted on a fetus while it is still in the uterus.  | Фетальная хирургия           |
| <b>Fetus(n)</b>                            | An unborn mammal, between 8 weeks of gestation to birth.   | Плод                         |

| Term                                 | Definition  | Translation                                     |
|--------------------------------------|---|---|
| <b>Flip a coin (v)</b>               | 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. | Подбросить монету                               |
| <b>First trimester screening (n)</b> | 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.  | Скрининг первого триместра                      |
| <b>Fragile X syndrome (n)</b>        | 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.  | Синдром ломкой Х-хромосомы                      |
| <b>Fragment (n)</b>                  | A small piece; an incomplete part of a whole.   | Фрагмент  |
| <b>Gene (n)</b>                      | A specific sequence of DNA that codes for one or many functions within the cell and body.   | Ген   |
| <b>Gene copy (n)</b>                 | 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.  | Копия гена                                      |
| <b>Normal gene copy (n)</b>          | A normal copy of a gene is one that is found most frequently in the population. Also called the “working copy.”   | Нормальная копия гена                           |
| <b>Altered gene copy (n)</b>         | 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.”   | Измененная копия гена                           |
| <b>Gene deletion (n)</b>             | Having a piece of genetic information missing from a gene.  | Делеция гена                                    |
| <b>Gene panel testing (n)</b>        | Genetic testing that looks at 2 or more specific genes known to be associated with particular conditions.   | Тестирование генетической панели (панели генов) |
| <b>Gene variant (n)</b>              | A change from the typical DNA sequences. A genetic variant can be benign, deleterious or of uncertain   | Вариант гена                                    |

| Term                              | Definition   | Translation                |
|-----------------------------------|--|----------------------------|
|                                   | significance. Also called “mutation,” although “genetic variant” is becoming the more common usage.  |                            |
| <b>General population (n)</b>     | <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>   | Общая популяция            |
| <b>Generation (n)</b>             | <p>The people who constitute a single step in a line of descent from an ancestor; a group of people born and living more or less at the same time.</p> <p>Example: You, your brothers, and sisters, all your spouses and your cousins are in the same generation. Your parents, your aunts, and uncles and all their spouses form a previous generation. Your grandparents, their siblings, and spouses form an even earlier generation. Your children and nieces and nephews form a later generation.</p> | Поколение                  |
| <b>Genetic alphabet (n)</b>       | 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).   | Генетический алфавит       |
| <b>Genetic counseling (n)</b>     | 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.  | Генетическая консультация  |
| <b>Genetic counselor (n)</b>      | 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.                 | Генетический консультант   |
| <b>Genetic discrimination (n)</b> | 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.  | Генетическая дискриминация |

| Term                                | Definition  | Translation                                     |
|-------------------------------------|---|---|
| <b>Genetic factors (n)</b>          | Specific aspects of a person's genetic make-up that influence that person's health and development.   | Генетические факторы                            |
| <b>Genetic information (n)</b>      | The instructions encoded in DNA, which tells every cell in a body how to grow, what to do and how to reproduce.   | Генетическая информация                         |
| <b>Genetic material (n)</b>         | 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. | Генетический материал                           |
| <b>Genetic predisposition (n)</b>   | An increased chance of a person developing a certain trait or disease based on that person's particular genetic makeup.   | Наследственная предрасположенность              |
| <b>Genetic test (n)</b>             | A laboratory test designed to determine if a person has a gene mutation or a typical DNA sequence.  | Генетический тест                               |
| <b>Single site gene testing (n)</b> | 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.                          | генетическое тестирование по одной точке        |
| <b>Individual gene testing (n)</b>  | Individual gene testing: genetic analysis of the entirety of one specific gene.   | Тестирование отдельных генов                    |
| <b>Gene panel test (n)</b>          | Gene panel test: analysis of several genes that have been shown to be associated with a particular condition, e.g., breast cancer gene panel.   | Тестирование панели генов (генетической панели) |
| <b>Microarray (n)</b>               | Microarray: genetic analysis that looks for unexpected numbers of specific genes (two copies of every gene code is usually expected).   | Микроматричный анализ                           |
| <b>Exome sequencing (n)</b>         | Exome testing: a genetic test in which the exons of a patient's genetic code are sequenced and studied.   | Секвенирование экзома                           |
| <b>Genome sequencing (n)</b>        | Genome sequencing: a genetic test in which the entire genetic code (the genome) of a patient is mapped out and studied.   | Секвенирование генома                           |
| <b>Genetic trait (n)</b>            | A characteristic within a family that is passed down from parent to child through their DNA.  | Генетический признак                            |
| <b>Geneticist (n)</b>               | A doctor or scientist who studies genetics.   | Генетик   |

| Term   | Definition  | Translation                        |
|--|---|------------------------------------|
| <b>Genetics (adj)</b>                          | The science of how an organism’s genes interact with the environment to produce certain traits.   | Генетический                       |
| <b>Genetics (n)</b>                            | 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.  | Генетика                           |
| <b>Genome (n)</b>                              | The entire genetic code of an individual.   | Геном                              |
| <b>Genome sequencing (n)</b>                   | A genetic test in which the entire genetic code of a patient is mapped out and studied.   | Секвенирование генома              |
| <b>Germline testing (n)</b>                    | <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>   | Тестирование зародышевой линии     |
| <b>Guardianship (n)</b>                        | A legal arrangement, supervised by a court, in which one person takes responsibility for a minor or an adult with a disability.   | Попечительство/<br>Опекунство      |
| <b>Helix (n)</b>                               | Something that is spiral in form. DNA is often described as being a “double helix.”   | Спираль                            |
| <b>Hereditary (adj)</b>                        | Passed down from parent to child.   | Наследственный                     |
| <b>Hereditary material (n)</b>                 | Genetic material that is passed down from parent to child.  | Наследственный материал            |
| <b>Huntington’s disease (n)</b>                | 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. | Болезнь Хантингтона                |
| <b>Individualized Education Plan (IEP) (n)</b> | 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.   | Индивидуальный план обучения (ИПО) |



| Term  | Definition   | Translation                               |
|---|--|---|
| <b>In Vitro Fertilization (IVF) (n)</b>     | <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> | Экстракорпоральное оплодотворение (ЭКО)   |
| <b>Individual gene testing (n)</b>          | Genetic analysis of the entirety of one specific gene.   | Тестирование отдельного (единичного) гена |
| <b>Infertility (n)</b>                      | The inability to have children.  | Бесплодие                                 |
| <b>Informed consent (n)</b>                 | 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.  | Информированное согласие                  |
| <b>Inheritance pattern (n)</b>              | 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.   | Тип наследования                          |
| <b>Inherited (adj)</b>                      | Passed down from parent to child.  | Унаследованный                            |
| <b>Insertion (n)</b>                        | An extra segment of DNA added in at a place where it is not usually found.   | Вставка                                   |
| <b>Intellectual disability (n)</b>          | 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.   | Интеллектуальное нарушение                |
| <b>Klinefelter's syndrome (47, XXY) (n)</b> | 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.   | Синдром Клайнфельтера (47, XXY)           |
| <b>Lynch syndrome (n)</b>                   | An inherited genetic disorder that increases a person's risk of getting cancer of the colon, rectum, uterus, ovaries, and other cancers.   | Синдром Линча                             |
| <b>Marker chromosome (n)</b>                | A small extra fragment of a chromosome detected when doing a chromosome test like a karyotype. Marker chromosomes can sometimes cause health or  | Маркерная хромосома                       |

| Term   | Definition   | Translation  |
|--|--|--|
|  | development problems, depending on how much and what genetic material is contained within.   |  |
| <b>Maternal Serum Screening (MSS or Maternal serum alpha-fetoprotein test) (n)</b> | 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.  | Скрининг материнской сыворотки (СМС или анализ материнской сыворотки на альфа-фетопротеин) |
| <b>Metastasis (n)</b>  | The spreading of cancer from one organ to another.   | Метастаз   |
| <b>Microarray (n)</b>  | 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. | Микроматричный анализ  |
| <b>Miscarriage (n)</b>   | The spontaneous (not intentional) loss of a pregnancy. See "abortion." Also called pregnancy loss.   | Выкидыш  |
| <b>Molecule (n)</b>  | 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.   | Молекула   |
| <b>Mosaicism (n)</b>   | A condition in which some, but not all, cells in a sample show a genetic difference.   | Мозаицизм  |
| <b>Motor Abilities (n)</b>   | The ability to move and use one's muscles.   | Двигательная способность / Моторика  |
| <b>Multifactorial (adj)</b>  | Due to a combination of genetic and non-genetic (environmental, hormonal, etc.) risk factors that act together to determine risk.  | Многофакторный   |
| <b>Mutation (n)</b>  | A change in a gene, usually deleterious. See "genetic variant."  | Мутация  |
| <b>Non-Invasive Prenatal Testing (NIPT) (n)</b>                                    | 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.  | Неинвазивное пренатальное тестирование (НИПТ)  |

| Term                                      | Definition   | Translation                          |
|---|--|--------------------------------------|
| <b>Nuchal thickening (n)</b>              | 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. | Утолщение воротникового пространства |
| <b>Open Neural Tube Defect (ONTD) (n)</b> | 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.  | Незаращение нервной трубки (ННТ)     |
| <b>Ova (n)</b>                            | See "egg."   | Яйцеклетка                           |
| <b>Ovary (n)</b>                          | The organ in a woman that stores and releases eggs. There are normally two.  | Яичник                               |
| <b>Packets of genetic information (n)</b> | A phrase genetic counselors use to describe genes or chromosomes.  | Пакеты генетической информации       |
| <b>Pathogenic (adj)</b>                   | Disease causing.   | Патогенный                           |
| <b>Pedigree (n)</b>                       | A family tree that can be used to trace the inheritance of specific genetic traits.  | Родословная                          |
| <b>Penetrance (n)</b>                     | 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.                                | Пенетрантность                       |
| <b>Placenta (n)</b>                       | The organ that develops together with an embryo in a pregnant woman's uterus to nourish the embryo through the umbilical cord.   | Плацента                             |
| <b>Presymptomatic (adj)</b>               | Before symptoms appear.  | Предсимптоматический                 |
| <b>Prognosis (n)</b>                      | The most likely outcome of a disease process.  | Прогноз                              |
| <b>Protein (n)</b>                        | 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.  | Белок                                |

| Term                                 | Definition   | Translation                     |
|--------------------------------------|--|---------------------------------|
| <b>Random (adj)</b>                  | Happening in an unpredictable way.   | Случайный                       |
| <b>Recessive (adj)</b>               | 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.<br><br>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. | Рецессивный                     |
| <b>Red flag (n)</b>                  | A warning sign or a clue.  | Настораживающий признак         |
| <b>Replicate (v)</b>                 | To copy.   | Реплицироваться                 |
| <b>Reproductive history (n)</b>      | 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.   | Репродуктивный анамнез          |
| <b>Ring chromosome (n)</b>           | An abnormal formation of a chromosome in which the ends of two of the chromosome's arms have linked together to form a ring.   | Кольцевая хромосома             |
| <b>Risk (n)</b>                      | The chance that something will happen; in the case of prenatal genetics, the chance that the child will have a genetic condition.  | Риск                            |
| <b>At risk (adj)</b>                 | Has some possibility of having a genetic condition.  | Находящийся в группе риска      |
| <b>At average risk (adj)</b>         | Has the same possibility of having a genetic condition as the general population.  | Средняя степень риска           |
| <b>At high risk (adj)</b>            | Has a greater possibility of having a genetic condition than the general population.   | Высокая степенью риска          |
| <b>At higher risk than "X" (adj)</b> | Has a greater possibility of having a genetic condition than "X."  | С более высоким риском, чем «X» |
| <b>Risk factor (n)</b>               | A circumstance that increases the risk of having a genetic condition.  | Фактор риска                    |
| <b>Runs in the family</b>            | Passed down from parents to children to grandchildren.   | Передается по наследству        |
| <b>Saliva (spit) sample (n)</b>      | A way to collect DNA from the cells in saliva (spit).  | Образец слюны                   |

| Term                               | Definition  | Translation                             |
|------------------------------------|---|---|
| <b>Screen (v)</b>                  | 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.  | Проводить скрининг                      |
| <b>Screening test (n)</b>          | <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> | Скрининг-тест                           |
| <b>Semen (n)</b>                   | 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.  | Семя                                    |
| <b>Seminal fluid (n)</b>           | See “semen.”  | Семенная жидкость                       |
| <b>Sensory abilities (n)</b>       | The ability to see, hear, touch, taste, and smell.  | Способности восприятия                  |
| <b>Sickle cell disease (n)</b>     | 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.   | Серповидно-клеточная болезнь            |
| <b>Single site analysis (n)</b>    | A genetic test that looks at only the part of a gene that is known to be associated with a certain disease.   | Анализ одного участка                   |
| <b>Somatic genetic changes (n)</b> | 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.  | Соматические генетические изменения     |
| <b>Special education (n)</b>       | Education that is tailored to individuals with developmental delays or other learning disabilities.   | Коррекционно-развивающее обучение (КРО) |

| Term                                   | Definition   | Translation              |
|--|--|--------------------------|
| <b>Sperm (n)</b>                       | The reproductive cells of the man. When sperm fertilize a woman's egg, a baby develops.  | Сперма                   |
| <b>Sperm donation (n)</b>              | 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.  | Донорство спермы         |
| <b>Spina bifida (n)</b>                | 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. | Расщепление позвоночника |
| <b>Statistically significant (adj)</b> | 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.                     | Статистически значимый   |
| <b>Supportive seating (n)</b>          | Seating, such as a wheelchair, that helps an individual function in their environment and that prevents worsening of their condition.  | Поддерживающее сиденье   |
| <b>Teratogen (n)</b>                   | Something that has the potential to disturb the normal development of an embryo or fetus.  | Тератоген                |
| <b>Termination of pregnancy (n)</b>    | See "abortion."  | Прерывание беременности  |

| Term   | Definition  | Translation  |
|--|---|--|
| <p><b>Test result (n)</b></p> <p><b>Negative (adj)</b></p> <p><b>True negative (n)</b></p> <p><b>Positive (adj)</b></p> <p><b>Variant of uncertain significance (n)</b></p> <p><b>Polymorphism (n)</b></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>Результат теста</p> <p>Отрицательный</p> <p>Истинно отрицательный</p> <p>Положительный</p> <p>Вариант неопределенного значения</p> <p>Полиморфизм</p> |
| <p><b>Testes (n)</b></p>   | <p>The organs in a man that create and release sperm. Also called “testicles.” Singular testis.</p>   | <p>Яички</p>   |
| <p><b>Toxic exposure (n)</b></p>   | <p>Contact with something that is harmful or poisonous.</p>   | <p>Токсическое воздействие</p>   |
| <p><b>Trait (n)</b></p>  | <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>  | <p>Признак</p>   |

| Term                     | Definition   | Translation               |
|--------------------------|--|---------------------------|
| <b>Translocation (n)</b> | 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. | Транслокация              |
| <b>Typo (n)</b>          | A mistyped word; shortened slang for “typographical error.”<br>In genetic counseling “a typo” is commonly used as an analogy to refer to mutations or variants in a gene.  | Опечатка                  |
| <b>Ultrasound (n)</b>    | An imaging method that uses high frequency sound waves to create a picture of something inside the body, such as a fetus or baby.  | УЗИ                       |
| <b>Uterus (n)</b>        | 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.”   | Матка                     |
| <b>Vagina (n)</b>        | The tube-like part of the female reproductive system that extends from the cervix to the outside of the body.  | Влагалище                 |
| <b>Variant (n)</b>       | A version of something that differs from the norm. So, a genetic variant is a change to the usual genetic sequence.  | Вариант                   |
| <b>Vas deferens (n)</b>  | The tubes that lead from the testes to the urethra, through which sperm is ejaculated.   | Семявыводящий проток      |
| <b>X-linked (adj)</b>    | A trait that is influenced or determined by a gene on the “X” chromosome.  | Сцепленный с X-хромосомой |



## 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               |
|------------------------|---|---------------------------|
| <b>Parent</b>          | Your mother or father.  | Родитель                  |
| <b>Mother</b>          | For genetic purposes, the woman whose egg was fertilized and grew to be you.  | Мать                      |
| <b>Mother-in-law</b>   | Your husband or wife's mother.  | Теща/свекровь             |
| <b>Stepmother</b>      | Your father's wife who is not your biological mother.   | Мачеха                    |
| <b>Adoptive mother</b> | A woman who is not your biological mother but who accepted legal responsibility for and raised you.                         | Приемная мать             |
| <b>Godmother</b>       | A woman chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church. | Крестная мать             |
| <b>Father</b>          | For genetic purposes, the man whose sperm fertilized the egg that grew to be you.   | Отец                      |
| <b>Father-in-law</b>   | Your husband or wife's father.  | Тесть/свекор              |
| <b>Stepfather</b>      | Your mother's husband who is not your biological father.  | Отчим                     |
| <b>Adoptive father</b> | A man who is not your biological father but who accepted legal responsibility for and raised you.                           | Приемный отец             |
| <b>Godfather</b>       | A man chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.   | Крестный отец             |
| <b>Aunt</b>            | Your mother or father's sister or sister-in-law.  | Тетя                      |
| <b>Maternal aunt</b>   | Your mother's sister or sister-in-law.  | Тетя по материнской линии |
| <b>Paternal aunt</b>   | Your father's sister or sister-in-law.  | Тетя по отцовской линии   |
| <b>Uncle</b>           | Your mother or father's brother or brother-in-law.  | Дядя                      |
| <b>Maternal uncle</b>  |   | Дядя по материнской линии |

| Term                             | Definition   | Translation                                |
|----------------------------------|--|--|
| <b>Paternal uncle</b>            | Your mother's brother or brother-in-law.<br>Your father's brother or brother-in-law. | Дядя по отцовской линии                    |
| <b>Cousin</b>                    | Usually understood to be a first cousin.   | Двоюродный брат/сестра                     |
| <b>First cousin</b>              | Your aunt or uncle's child.  | Двоюродный брат/сестра                     |
| <b>First cousin once removed</b> | Your aunt or uncle's grandchild or your first cousin's child                         | Двоюродный племянник/двоюродная племянница |
| <b>Second cousin</b>             | The children of first cousins are second cousins to each other.                      | Троюродный брат/сестра                     |
| <b>Grandparent</b>               | Your parent's father or mother.  | Дедушка/бабушка                            |
| <b>Grandmother</b>               | Your mother or father's mother.  | Бабушка                                    |
| <b>Maternal grandmother</b>      | Your mother's mother.  | Бабушка по материнской линии               |
| <b>Paternal grandmother</b>      | Your father's mother.  | Бабушка по отцовской линии                 |
| <b>Grandfather</b>               | Your mother or father's father.  | Дедушка                                    |
| <b>Maternal grandfather</b>      | Your mother's father.  | Дедушка по материнской линии               |
| <b>Paternal grandfather</b>      | Your father's father.  | Дедушка по отцовской линии                 |
| <b>Great aunt</b>                | Your mother or father's aunt.  | Двоюродная бабушка                         |
| <b>Maternal great aunt</b>       | Your mother's aunt.  | Двоюродная бабушка по материнской линии    |
| <b>Paternal great aunt</b>       | Your father's aunt.  | Двоюродная бабушка по отцовской линии      |
| <b>Great uncle</b>               | Your father or mother's uncle.   | Двоюродный дедушка                         |
| <b>Maternal great uncle</b>      | Your mother's uncle.   | Двоюродный дедушка по материнской линии    |
| <b>Paternal great uncle</b>      | Your father's uncle.   | Двоюродный дедушка по отцовской линии      |
| <b>Great grandparents</b>        | The parents of any of your grandparents.   | Прадедушка и прабабушка                    |
| <b>Great grandmother</b>         | The mother of any of your grandparents.  | Прабабушка                                 |

| Term                     | Definition   | Translation                       |
|--------------------------|--|-----------------------------------|
| <b>Great grandfather</b> | The father of any of your grandparents.  | Прадедушка                        |
| <b>Siblings</b>          | The children of your father and mother.  | Братья и сестры                   |
| <b>Half siblings</b>     | 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.     | Неполнородные братья и сестры     |
| <b>Stepsiblings</b>      | 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.                    | Сводные братья и сестры           |
| <b>Twins</b>             | Two siblings born at the same time.  | Близнецы                          |
| <b>Identical twins</b>   | Twins who developed from the same egg and sperm, meaning that they are genetically identical. Also called monozygotic twins.   | Однояйцевые близнецы              |
| <b>Fraternal twins</b>   | 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. | Разнояйцевые близнецы             |
| <b>Triplets</b>          | Three siblings born at the same time.  | Тройняшки                         |
| <b>Sister</b>            | A sibling who is a girl.   | Сестра                            |
| <b>Sister-in-law</b>     | Your brother's wife or your husband's sister.  | Невестка/золовка/свояченица       |
| <b>Half sister</b>       | A girl who is either the child of your father with a different mother, or the child of your mother with a different father.  | Единоутробная/единокровная сестра |
| <b>Stepsister</b>        | The daughter of your stepmother or stepfather and therefore not biologically related to you.   | Сводная сестра                    |
| <b>Brother</b>           | A sibling who is a boy.  | Брат                              |
| <b>Brother-in-law</b>    | Your sister's husband or your wife's brother.  | Зять/шурин/свояк/деверь           |

| Term                     | Definition  | Translation                     |
|--------------------------|---|---------------------------------|
| <b>Half brother</b>      | A boy who is either the child of your father with a different mother, or the child of your mother with a different father.                              | Единоутробный/единокровный брат |
| <b>Stepbrother</b>       | The son of your stepmother or stepfather and therefore not biologically related to you.   | Сводный брат                    |
| <b>Niece</b>             | Your sibling's daughter.  | Племянница                      |
| <b>Nephew</b>            | Your sibling's son.   | Племянник                       |
| <b>Spouse</b>            | Your husband or wife.   | Супруг/супруга                  |
| <b>Wife</b>              | The woman to whom you are married.  | Жена                            |
| <b>Husband</b>           | The man to whom you are married.  | Муж                             |
| <b>Children</b>          | Genetically speaking, the people who are produced from your egg or sperm.   | Дети                            |
| <b>Daughter</b>          | Your child who is a girl.   | Дочь                            |
| <b>Daughter-in-law</b>   | Your son's wife.  | Жена сына                       |
| <b>Stepdaughter</b>      | Your spouse's daughter who is not your biological child.  | Падчерица                       |
| <b>Adoptive daughter</b> | A girl for whom you have accepted legal responsibility and raised even though she is not your biological child.   | Приемная дочь                   |
| <b>Goddaughter</b>       | 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. | Крестница                       |
| <b>Son</b>               | Your child who is a boy.  | Сын                             |
| <b>Son-in-law</b>        | Your daughter's husband.  | Муж дочери                      |
| <b>Stepson</b>           | Your spouse's son who is not your biological child.   | Пасынок                         |
| <b>Adoptive son</b>      | A boy for whom you have accepted legal responsibility and raised even though he is not your biological child.   | Приемный сын                    |
| <b>Godson</b>            | A boy for whom you have accepted spiritual responsibility within a ceremony of  | Крестник                        |

| Term                    | Definition   | Translation               |
|-------------------------|--|---------------------------|
|                         | the Catholic church. He may or may not be biologically related to you.   |                           |
| <b>Ancestor</b>         | A person from whom you are descended, usually more remote than a grandparent.  | Предок                    |
| <b>Fiancé(e)</b>        | The person whom you have promised to marry.<br><br>Man: fiancé<br><br>Woman: fiancée   | Жених/невеста             |
| <b>Domestic partner</b> | The person with whom you are living and have an intimate relationship, but to whom you are not married.  | Гражданский(ая) супруг(а) |
| <b>Divorced</b>         | Having ended a marriage.   | В разводе                 |
| <b>Engaged</b>          | Having promised to marry someone.  | Помолвлен/помолвлена      |
| <b>Widow</b>            | A woman whose husband has died.  | Вдова                     |
| <b>Widower</b>          | A man whose wife has died.   | Вдовец                    |
| <b>Relative</b>         | A person in your family.   | Родственник/родственница  |
| <b>Blood relation</b>   | 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. | Кровный родственник       |
| <b>Next of kin</b>      | Your closest living blood relation.  | Ближайший родственник     |

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