

## Bilingual Glossary of Exome Sequencing Terms English- Spanish

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

Term	Definition	Equivalent or Paraphrase
abnormal (adj)	Different from what is considered normal, average or expected. For example, a gene sequence that is different than that found in most people.	no normal/que no es normal
benign (adj)	In genetics, refers to DNA changes that do not create health consequences, i.e. genetic variants that do not cause harm.	benigno/a
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 provide DNA for genetic sequencing.	prueba de sangre
buccal swab (n)	A way to collect DNA from the cells on the inside of a person's cheek.	muestra bucal
carrier (n)	A person who carries a genetic variant in one of the two copies of a particular gene, regardless of whether they are at risk of having the related condition. Often used when describing a deleterious variant.	portador/a

cell (n)	<p>A small (microscopic) structure that forms the basic building block of every known living organism.</p> <p>Cells are the building blocks that make up our bodies, and different cells have different functions. A nerve cell is a little different from a skin cell, which is different from a white blood cell, which is different a muscle cell, for example.</p> <p>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.</p>	Célula
chromosome (n)	<p>Thread-like structures located inside the nucleus of cells. In humans, there are 23 pairs of chromosomes, for a total of 46 chromosomes. Each chromosome is made of proteins and a single molecule of DNA, which carries genetic information.</p> <p>When the sperm fertilizes the egg, each chromosome from the male joins the matching chromosome from the female, and now the fertilized egg has, again 46 chromosomes – 23 pairs.</p> <p>If we stretch out the DNA of a chromosome, it would look like a long ladder that is twisted into a spiral. The “rungs” on this so-called “ladder” are various combinations of four chemical molecules called adenine, thymine, guanine and cytosine. These are often referred to as A, T, G, and C for short. These letters spell out the genetic code.</p>	cromosoma
chromosome abnormalities (n)	<p>A problem in the chromosome that can lead to health or developmental issues. For example, both of a pair of chromosomes can come from only one parent, with none from the other, or a chromosome can be missing, or a fragment of a chromosome can be duplicated or deleted.</p>	anormalidades en los cromosomas

chromosome test/karyotype (n)	A test that looks at the number and types of chromosomes in a cell.	pruebas de los cromosomas / prueba de cariotipo
clinically significant (adj)	If a test result is “clinically significant,” the result indicates a medical problem that can impact a person’s health.	clínicamente significativos
condition (e.g. “genetic condition”) (n)	A chronic (long-term) health issue.	condición (por ej., “condición genética”)
de novo (adj)	Genetic changes that are not inherited but occur for the first time in a given person during cell division.	mutación que aparece por primera vez en una familia
deleterious mutation/disease-causing mutation (n)	A change/variant in a person’s DNA that has the potential to disrupt the function of the gene, causing a medical condition or higher risk of a given disease. Also can be called “pathogenic” or “likely pathogenic”.	mutación que causa enfermedad mutación dañina/perjudicial/nociva
deletion (n)	A missing section of genetic information (DNA).	deleción
DNA (deoxyribonucleic acid) (n)	<p>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.</p> <p>DNA can be thought of as the instructions for how our body grows, develops and functions.</p>	ADN (ácido desoxirribonucleico)

DNA marker (also called a “genetic marker”) (n)	A readily recognizable genetic trait, gene, or DNA segment.	marcador de ADN (también llamado “marcador genético”)
DNA sequence (n)	The exact order of the chemicals that make up a DNA molecule.	secuencia de ADN
DNA sequencing (n)	The laboratory technique used to determine the exact order of the chemicals that make up a DNA molecule. This is one type of genetic testing.	secuenciación de ADN
dominant (adj)	A genetic trait in which one copy of the gene is sufficient for a trait to be expressed; dominant traits are inherited from one parent. In a dominant genetic condition, if one copy of the gene has a variant, the person will be affected with the condition.	dominante
double helix (n)	The DNA of a chromosome is in the shape of a spiral, which can be visualized as a long, twisted ladder. This shape is referred to as a “double helix.”	doble hélice
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 genetic material.	duplicación
early age of onset (n)	A diagnosis at an early age. For example, in cancer which more often happens in later life, early onset is before age 50.	El cáncer aparece cuando la persona es joven
egg (n)	The reproductive cells of a woman. When fertilized by sperm, the egg will grow into a baby.	óvulo

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.	causas o factores del medioambiente
exome sequencing (n)	A genetic test which looks specifically at the exons in a person's DNA sequence: the part of the gene that encodes the proteins produced by the gene. The exome makes up about 2% of the whole genome (all the genetic material in the body). Exome sequencing can provide information about a variety of genetic conditions.	secuenciación del exoma
exon (n)	The part of the genome that codes for proteins. The majority of deleterious variants occur in exons.	exón
familial (adj)	Belonging to a family; e.g. a familial trait is a trait that is shared among family members and may be due to genetic or environmental factors or both.	familiar
family history (n)	The medical history of the members of a family.  A genetic clinician usually draws a family tree to diagram a family's medical history.	historia médica familiar
fragment (n)	A small piece; an incomplete part of a whole.	fragmento
gene (n)	A specific sequence of DNA that determines specific traits in an individual.	gen
gene copy (n)	In human cells, DNA is arranged in 23 pairs of chromosomes, for a total of 46 chromosomes. These chromosomes contain all	copia de gen

	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.	
normal gene copy (n)	A normal copy of a gene is one that is found most frequently in the population. Also called the “working copy.”	copia normal del gen
altered gene copy (n)	Having a piece of genetic information missing from a gene.	copia alterada del gen
gene deletion (n)	Having a piece of genetic information missing from a gene.	delección genética
gene panel testing (n) or “panel testing”	Genetic testing that looks at two or more specific genes known to be associated with particular conditions for which a patient may be at risk.	análisis de varios genes a la vez
gene variant (n)	A change from a typical DNA sequence. A genetic variant can be benign, deleterious or of uncertain significance. The term “variant” is increasingly being used instead of the term “mutation”.	variante genética
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>	generación

genetic alphabet (n)	A group name for four chemical bases, or “nucleotides,” that form the basis of all genetic material. They are adenine (A), thymine (T), cytosine (C), and guanine (G).	alfabeto genético
gene sequence (n)	Precise order of nucleotides (chemical letters) within a gene or DNA molecule. (see genetic alphabet)	secuencia del gen
genetic (adj)	Having to do with inherited traits.	genético/a
genetic counseling (n)	A discussion with a genetic counselor. 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.	asesoramiento genético o consejería genética
genetic counselor (n)	A healthcare professional with a specialized graduate degree who works with people undergoing genetic testing. Genetic counselors provide information about genetic conditions, help patients understand their chances of being affected by a genetic condition or having a child with a genetic condition, and help them make informed decisions about testing and treatment. Genetic counselors also provide emotional support to patients and families.	asesor/a o consejero/a genético
genetic discrimination (n)	The act of refusing to provide, or charging more for, insurance, employment or any service based on an individual’s genetic test result.	discriminación genética
genetic factors (n)	Specific aspects of a person’s genetic make-up that influence that person’s health and development.	factores genéticos

genetic information (n)	The information encoded in genes, which tells every cell in a body how to grow, what to do and how to divide to replicate (copy) itself.	información genética
genetic material (n)	All the parts of a cell that carry genetic information. Genetic material could include genes, parts of genes, a group of genes, a DNA molecule, a fragment of DNA, a group of DNA molecules, or the entire set of genetic instructions.	material genético
genetic predisposition (n)	An increased chance of a person developing a certain trait or disease based on that person's particular genetic makeup.	predisposición genética
geneticist (n)	A doctor or scientist who studies genetics.	doctor de genética genetista
genetic test (n)	A laboratory test designed to determine if a person has a gene variant or a typical DNA sequence.	análisis genético
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 variant found in a family member or when one specific variant is known to cause a genetic condition.	análisis en un punto específico del genoma
individual gene testing (n)	Individual gene testing: genetic analysis of the entirety of one specific gene.	análisis de un solo gen
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.	análisis de varios genes a la vez



microarray (n)	Microarray: genetic analysis that looks for unexpected numbers of specific genes (two copies of every gene code is usually expected)	chip de ADN
exome sequencing (n)	Exome testing: A genetic test which looks specifically at the exons in a person's DNA sequence: the part of the gene that encodes the proteins produced by the gene. The exome makes up about 2% of the whole genome (all the genetic material in the body). Exome sequencing can provide information about a variety of genetic conditions.	secuenciación del exoma
genome sequencing (n)	A genetic test which looks specifically at the entire person's DNA sequence (not only the exons like in exome sequencing): Genome sequencing can provide information about a variety of genetic conditions.	secuenciación del genoma
genetic trait (n)	A characteristic within a family that is passed down from parent to child genetically.	rasgo genético o característica genética
general population (n)	<p>"Most people."</p> <p>For example, if you have the same risk of getting cancer as the general population, that means that you have the same chance of getting cancer as everyone else. This is in contrast to a "high risk population" who has a greater chance of getting cancer than everyone else.</p>	población general
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</p>	análisis de línea germinal

	words, the cells from which the eggs and sperm come. After the egg and sperm come together to form a baby, these cells will then divide and multiply and will form the entire body.	
hereditary (adj)	Passed down from parent to child.	hereditario
hereditary material (n)	Genetic material that is passed down from parent to child.	material hereditario
individual gene testing (n) or single gene testing (n)	Genetic analysis of one specific gene.	análisis de un solo gen
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.	patrón hereditario
inherited (adj)	Passed down from parent to child.	heredado/a
insertion (n)	An extra segment of DNA added at a place where it is not usually found.	inserción
Likely pathogenic (adj)	Clinically, likely pathogenic variants are treated as if they are pathogenic or disease causing. Variants are classified as likely pathogenic if there is some evidence, that a variant is the definite cause for symptoms.	Probablemente patológico, es decir que probablemente puede causar una enfermedad
malignant (adj)	In cancer, this term means that the cancer cells or tumor are harmful and have the potential of spreading to other tissues or parts of the body.	maligno/a
molecule (n)	The smallest unit of a chemical compound that still has the	molécula

	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.	
multifactorial (adj)	Due to a combination of genetic and non-genetic (environmental, hormonal, etc.) risk factors that act together to determine risk.	varios factores/ multifactorial
mutation (n)	A change in a gene, which can be deleterious (disease causing) or benign (non-disease-causing).  (In Spanish, the word is sometimes confused with “mutilation.”)	mutación
packets of genetic information (n)	A phrase genetic counselors or geneticists use to describe genes or chromosomes.	paquetes de información genética
pattern (n)	A repeating arrangement or sequence; for example, the pattern of cancers in a family tree.	patrón
pedigree (n)	A family tree that can be used to trace the inheritance of specific genetic traits.	árbol genealógico
penetrance (n)	The probability that a specific genetic trait will be expressed if a person carries a deleterious mutation or variant. “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.	penetrancia

prognosis (n)	The most likely outcome of a disease process.	pronóstico
proliferation (cell proliferation) (n)	The controlled process by which a cell multiplies.  Cancer arises when the process of cell division becomes uncontrolled.	proliferación o multiplicación (proliferación de células)
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.	proteína
random (adj)	Happening in an unpredictable way.	al azar
recessive (adj)	A genetic trait in which both copies of a gene are needed for the trait to be expressed; recessive traits are inherited from both biological parents, each one contributing one copy of the gene in question.  In cancer genetics, recessive inheritance refers to conditions in which both copies of a particular gene pair need to be altered to develop the disease.	recesivo/a
red flag (n)	A warning sign or a clue.	señal de alerta
risk (n)  at risk (adj)	The chance that something will happen; for example, in cancer genetics, the chance of getting cancer.  Has some possibility of getting a disease or medical condition.	riesgo  corre riesgo

at average risk (adj)	Has the same possibility of getting a disease or medical condition as the general population.	riesgo promedio o riesgo normal
at high risk (adj)	Has a greater possibility of getting a disease or medical condition than the general population.	riesgo alto
at higher risk than 'X' (adj)	Has a greater possibility of getting a disease or medical condition than "X".	riesgo mayor que 'X'
risk factor (n)	A circumstance or element that increases the risk of getting a disease or medical condition.	factor de riesgo
runs in the family (v)	Is passed down from parents to children to grandchildren.	hereditario/a
saliva (spit) test (n)	A genetic test done by collecting saliva (spit) instead of blood.	prueba de saliva
screen (v)	To test a person for a particular common health problem, even though the person has no symptoms to suggest that they have or are a carrier of the particular disease.	hacer una prueba de detección
screening test (n)	A test that looks for a common health problem even though the patient has no symptoms to suggest they have or are a carrier of the particular disease or condition.	prueba de detección
sporadic (adj)	Random, once in a while.  In genetics, sporadic cancers are those caused by random	esporádico/a

	chance or unknown factors in the environment.	
statistically significant (adj)	Refers to the likelihood that a relationship between two factors is linked by something other than chance. If a relationship is statistically significant, it is not caused by chance.	estadísticamente significativo
syndrome (n)	<p>A group of symptoms or features that consistently occur together or a condition characterized by a set of associated features.</p> <p>For example, a hereditary cancer syndrome refers to risk for a group of cancers all caused by a particular gene mutation. BRCA1 and BRCA2 deleterious variants increase risk for breast and ovarian cancers. This is called Hereditary Breast and Ovarian Cancer Syndrome. See also, Lynch Syndrome.</p>	síndrome
test result (n)		resultado del análisis genético
negative (adj)	A negative result on a genetic test means that the laboratory did not find the specific genetic change(s) that the test was designed to identify. Also called “normal”.	negativo
true negative (n)	When an individual is tested to for a specific genetic change that is present in other family members, a negative result can be considered a “true negative.” This means that the individual does not have the hereditary condition that others in the family have.	negativo verdadero
positive (adj)	A positive result on a genetic test means that the laboratory did find a specific genetic change that is associated with a genetic	positivo

variant of uncertain significance (n)	condition. In cancer genetics, that means that the patient has an increased risk of getting the cancer(s) associated with the gene. Also called “abnormal”.  This is an unclear result. This happens when the genetic test finds a change in your DNA but scientists and doctors do not know enough about what it means. Your doctor will recommend care based on you and your family’s health history. 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.	variante de significado incierto
polymorphism (n)	Everyone has some degree of commonly occurring genetic changes that are not associated with disease. If the test finds this sort of change, it reports a “polymorphism.”	polimorfismo
trait (n)	A characteristic.	Rasgo o característica
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.	error en la escritura/error tipográfico
variant (n)	A version of something that differs from the norm. So, a genetic variant is a change to the usual genetic sequence.	variante

## Examples of genes that are often checked in exome sequencing

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



## Family relationships

NOTE: For Genetic Counselors, it is very important to know if a relative is a blood relative or a relative by marriage.

Term	Definition	Equivalent or Paraphrase
parent (n)	Mother or father.	padre(s)
mother (n)	For genetic purposes, the woman whose egg was fertilized and grew to be you.	madre(s)
mother-in-law (n)	Your husband or wife's mother.	suegra
step-mother (n)	Your father's wife who is not your biological mother.	madrstra
adoptive mother (n)	A woman who is not your biological mother but who accepted legal responsibility for and raised you.	madre adoptive
godmother (n)	A woman chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.	madrina
father (n)	For genetic purposes, the man whose sperm fertilized the egg that grew to be you.	padre
father-in-law (n)	Your husband or wife's father.	suegro
step-father (n)	Your mother's husband who is not your biological father.	padrastra
adoptive father (n)	A man who is not your biological father but who accepted legal responsibility for and raised you.	padre adoptive

godfather (n)	A man chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.	padrino
aunt (n)	Your mother or father's sister or sister-in-law.	tía
maternal aunt (n)	Your mother's sister or sister-in-law.	tía materna
paternal aunt (n)	Your father's sister or sister-in-law.	tía paterna
cousin (n)	Usually understood to be a first cousin.	primo/a
first cousin (n)	Your aunt or uncle's child.	primo/a
second cousin (n)	Your aunt or uncle's grandchild.	hermano/a
grandmother (n)	Your mother or father's mother.	abuela
maternal grandmother (n)	Your mother's mother.	abuela materna
paternal grandmother (n)	Your father's mother.	abuela paterna
grandfather (n)	Your mother or father's father.	abuelo
maternal grandfather (n)	Your mother's father.	abuelo materno
paternal grandfather (n)	Your father's father.	abuelo paterno
uncle (n)	Your mother or father's brother or brother-in-law.	tío
maternal uncle (n)	Your mother's brother or brother-in-law.	tío materno

paternal uncle (n)	Your father's brother or brother-in-law.	tío paterno
great aunt (n)	Your mother or father's aunt.	tía abuela
maternal great aunt (n)	Your mother's aunt.	tía abuela o materna tía
paternal great aunt (n)	Your father's aunt.	abuela paterna
great uncle (n)	Your father or mother's uncle.	tío abuelo
maternal great uncle (n)	Your mother's uncle.	tío abuelo o materno tío
paternal great uncle (n)	Your father's uncle.	abuelo paterno
great grandparents (n)	The parents of any of your grandparents.	bisabuelos
great grandmother (n)	The mother of any of your grandparents.	bisabuela
great grandfather (n)	The father of any of your grandparents.	bisabuelo
siblings (n)	Sisters and brothers.	hermanas y hermanos
twins (n)	Two siblings born at the same time.	gemelos/as mellizos/as
identical twins (n)	A twin that developed from the same egg and sperm, meaning that they are genetically identical.	gemelos/as idéntico/as mellizo/as idéntico/as

fraternal twins (n)	Twins who developed from a different egg and sperm, meaning that they are genetically different and have the same amount of shared genes as any sibling.	genelos fraternos (gemelos no idénticos)
triplets (n)	Three siblings born at the same time.	trillizos/as
sister (n)	A sibling who is a girl.	hermana
sister-in-law (n)	Your brother's wife.	cuñada
half-sister (n)	A girl who is either the child of your father with a different mother, or the child of your mother with a different father.	media hermana
step-sister (n)	The daughter of your step-mother or step-father and therefore not biologically related to you.	hermanastra
brother (n)	A sibling who is a boy.	hermano
brother-in-law (n)	Your sister's husband.	cuñado
half-brother (n)	A boy who is either the child of your father with a different mother, or the child of your mother with a different father.	medio hermano
step-brother (n)	The son of your step-mother or step-father and therefore not biologically related to you.	hermanastro
niece (n)	Your sibling's daughter.	sobrina
nephew (n)	Your sibling's son.	sobrino
spouse (n)	Your husband or wife.	cónyuge

wife (n)	The woman to whom you are married.	esposa
husband (n)	The man to whom you are married.	esposo
children (n)	Genetically speaking, the people who are produced from your egg or sperm.	hijos
daughter (n)	Your child who is a girl.	hija
daughter-in-law (n)	Your son's wife.	nuera
step-daughter (n)	Your spouse's daughter who is not your biological child.	hijastra
adoptive daughter (n)	A girl for whom you have accepted legal responsibility and raised even though she is not your biological child.	hija adoptiva
goddaughter (n)	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.	ahijada
son (n)	Your child who is a boy.	hijo
son-in-law (n)	Your daughter's husband.	yerno
step-son (n)	Your spouse's son who is not your biological child.	hijastro
adoptive son (n)	A boy for whom you have accepted legal responsibility and raised even though he is not your biological child.	hijo adoptivo
godson (n)	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.	ahijado

ancestor (n)	A person from whom you are descended, usually more remote than a grandparent.	Ancestor o antepasado
fiancé(e) (n)	The person whom you have promised to marry.  Man: fiancé Woman: fiancée	novio novia
domestic partner (n)	The person with whom you are living and have an intimate relationship, but to whom you are not married.	pareja de union libre
divorced (adj)	Having ended a marriage.	divorciado/a
engaged (adj)	Having promised to marry someone.	comprometido/a
widow (n)	A woman whose husband has died.	viuda
widower (n)	A man whose wife has died.	viudo
relative (n)	A person in your family.	Pariente o familiar
blood relation (n)	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 step-mother; your daughter but not your adopted daughter.	pariente de sangre o pariente consanguíneo
next-of-kin (n)	Your closest living blood relation.	pariente más cercano