Interpreting for Cancer Genetics

Bilingual Glossary of Cancer Genetics Terms English –

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

Term	Definition	Equivalent or Paraphrase
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.	
benign (adj)	Something mild that does not threaten health or life.	
	In cancer genetics, "benign" means "not cancerous."	
blood test (n)	A test in which blood is drawn (usually from the patient's arm) and sent to a laboratory for analysis.	
	In cancer genetics, blood tests provide cells for genetic sequencing.	
breast cancer (n)	Cancer of the breast tissue. This cancer is more common in women, but it can affect men as well.	
buccal swab (n)	A way to collect DNA from the cells on the inside of a person's cheek.	
ca125 blood test (n)	A blood test used to look for early signs of ovarian cancer in women with a high cancer risk.	
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.	
cell (n)	A small (microscopic) structure that forms the basic building block of every known living organism.	
chromosome (n)	Thread-like structures located inside the nucleus of cells. In humans, there are	

Term	Definition	Equivalent or Paraphrase
	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.	
colonoscopy	A diagnostic test in which a flexible tube with a tiny camera on the end is inserted into the colon (large intestine) through the rectum in order to see the condition of the inside of the colon, or to take a small sample of tissue, or to cut out polyps (small growths).	
condition (e.g. "genetic condition") (n)	A chronic (long-term) health issue.	
consanguinity (n)	When parents are blood relatives to each other.	
DCIS (ductal carcinoma in situ) (n)	A pre-cancerous growth in the breast, which is still within the milk ducts, but has the potential of growing and spreading to other parts of the breast tissue. Sometimes called Stage 0 breast cancer.	
DTC (direct-to-consumer genetic testing) (n)	Genetic testing that is marketed directly to people at home through the internet or the TV etc. instead of through a medical provider.	
deleterious mutation/ disease-causing mutation (n)	A change in a person's DNA that may cause a medical condition. In cancer genetics, having a disease-causing mutation may increase the <i>chance</i> of getting cancer but does not mean that a person will <i>definitely</i> get cancer.	
DNA (deoxyribunucleic 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 banking (n)	The secure, long-term storage of a person's genetic material. DNA banking allows families to have access to a deceased family member's DNA. When new genetic testing techniques are developed, doctors can test the banked DNA and use genetic information from the deceased person to tailor medical treatment for living descendants.	

Term	Definition	Equivalent or Paraphrase
DNA marker (also called a "genetic marker") (n)	A readily recognizable genetic trait, gene, or DNA segment.	
DNA mutation (n)	A change in the typical order of the chemicals that make up the DNA.	
	Mutations or variants are often compared to misspelled words because chemicals that make up the DNA sequence are not in the expected order.	
DNA sequence (n)	The exact order of the chemicals that make up a DNA molecule.	
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.	
dominant (adj)	A genetic trait in which one copy of the gene is sufficient for a trait to be expressed; dominant traits can be inherited from a single parent.	
	In cancer genetics, dominant inheritance refers to conditions in which having only one altered copy of a particular gene pair is sufficient to cause an increased risk for cancer.	
duplication (n)	A duplication occurs when part of a chromosome is copied abnormally, resulting in extra genetic material.	
early age of onset (n)	Cancer diagnosed at an early age, usually before the age of 50.	
egg (n)	The reproductive cells of a woman. When fertilized by sperm, the egg will grow into a baby.	
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.	
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.	

Term	Definition	Equivalent or Paraphrase
family history (n)	The medical history of the members of a family.	
to 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.	
fragment (n)	A small piece; an incomplete part of a whole.	
gene (n)	A specific sequence of DNA that determines specific traits in an individual.	
gene copy (n)	In human cells, DNA is arranged in 23 pairs of chromosomes, for a total of 46 chromosomes. These chromosomes contain all the genes that make up the DNA. One chromosome of each pair is inherited from each parent. As such, individuals have two copies of every gene, one inherited from the mother and one inherited from the father.	
normal gene copy (n) altered gene copy (n)	A normal copy of a gene is one that is found most frequently in the population. Also called the "working copy."	
	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."	
gene deletion (n)	Having a piece of genetic information missing from a gene.	
generation (n)	The people who constitute a single step in a line of descent from an ancestor; a group of people born and living more or less at the same time.	
	Example: You, your brothers and sisters, all your spouses and your cousins are in the same generation. Your parents, your aunts and uncles and all their spouses form a previous generation. Your grandparents, their siblings and spouses from an even earlier generation. Your children and nieces and	

Term	Definition	Equivalent or Paraphrase
	nephews form a later generation.	
genetic (adj)	Having to do with inherited traits.	
genetic counseling (n)	A discussion with a genetic counselor about the basic concepts of genetics, genetic conditions, the chances of being affected by a genetic condition or having a child with a genetic condition, and genetic testing and treatment.	
genetic counseling intern (n)	A genetic counseling student who has not yet completed his/her academic studies and is now practicing under the supervision of a more experienced counselor in preparation for providing genetic counseling services independently after obtaining his/her graduate degree.	
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.	
genetic discrimination (n)	The act of refusing to provide, or charging more for, insurance or any service based on an individual's genetic make-up.	
genetic factors (n)	Specific aspects of a person's genetic make-up that influence that person's health and development.	
genetic information (n)	The information encoded in genes, which tells every cell in a body how to grow, what to do and how to reproduce.	
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.	
genetic predisposition (n)	An increased chance of a person developing a certain trait or disease based on that person's particular genetic makeup.	

Term	Definition	Equivalent or Paraphrase
genetic trait (n)	A characteristic within a family that is passed down from parent to child genetically.	
geneticist (n)	A doctor or scientist who studies genetics.	
genetic test (n) single gene test (n) gene panel test (n)	A laboratory test designed to determine if a person has a gene mutation or a typical DNA sequence. Single gene test: analysis of one particular gene. Gene panel test: analysis of several genes that have been shown to be associated with a particular condition; <i>Example:</i> breast cancer gene panel.	
general population (n)	"Most people." For example, if you have the same risk of getting cancer as the general population, that means that you have the same chance of getting cancer as everyone else. This is in contrast to a "high risk population" who has a greater chance of getting cancer than everyone else.	
germline testing (n)	Germline testing refers to the analysis of a person's DNA, which he or she inherited from his or her parents. In biology and genetics, the germline is the group of cells that will pass on the genetic material to children, in other words, the cells from which the eggs and sperm come. After the egg and sperm come together to form a baby, these cells will then divide and multiply and will form the entire body. Germline testing is different from testing the DNA of someone's tumor, which may have a mutation that happened during the person's lifetime and was not inherited from his or her parents.	
hereditary (adj)	Passed down from parent to child.	

Term	Definition	Equivalent or Paraphrase
hereditary material (n)	Genetic material that is passed down from parent to child.	
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.	
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	
inherited (adj)	Passed down from parent to child.	
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.	
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.	
metastasis (n)	The spreading of cancer from one organ to another.	
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.	
multifactorial (adj)	Due to a combination of genetic and non-genetic (environmental, hormonal, etc.) risk factors that act together to determine risk.	
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."	
oncologist (n)	A physician who specializes in diagnosing and treating cancer.	

Term	Definition	Equivalent or Paraphrase
oophorectomy	The surgical removal of one or both ovaries.	
ovarian cancer (n)	Cancer of the ovaries, the organs in a woman that release eggs.	
packets of genetic information (n)	A phrase genetic counselors use to describe genes or chromosomes.	
pattern (n)	A repeating arrangement or sequence; for example, the pattern of cancers in a family.	
pedigree (n)	A family tree that can be used to trace the inheritance of specific genetic traits.	
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.	
predisposition to cancer	Having a change in one of number of specific genes (a genetic mutation) that creates a higher than normal risk of a person developing cancer.	
prognosis (n)	The most likely outcome of a disease process.	
proliferation (cell proliferation) (n)	The controlled process by which a cell multiplies. Cancer arises when the process of cell division becomes uncontrolled.	
prophylactic mastectomy (n)	A mastectomy is the surgical removal of the breast. "Prophylactic" means something done to prevent disease. So a prophylactic mastectomy is the surgical removal of a healthy breast to prevent breast cancer. Prophylactic mastectomy is an option for individuals with a very high breast cancer risk, for example, women with a BRCA mutation.	
protein (n)	A molecule made up of chains of amino acids. Proteins do most of the work in cells and are required for the structure, function, and regulation of the body's	

Term	Definition	Equivalent or Paraphrase
	tissues and organs. Genes determine how specific amino acids are put together to form a specific protein.	
random (adj)	Happening in an unpredictable way.	
recessive (adj)	A genetic trait in which both copies of a gene are needed for the trait to be expressed; recessive traits are inherited from both parents, each one contributing one copy of the gene in question.	
	In cancer genetics, recessive inheritance refers to conditions in which both copies of a particular gene pair need to be altered to develop the disease.	
recurrence (n)	When something happens again.	
	In cancer, a return of the disease after treatment and after a period of time during which the cancer cannot be detected.	
red flag (n)	A warning sign or a clue.	
risk (n)	The chance that something will happen; in the case of cancer genetics, the chance of getting cancer.	
at risk (adj)	Has some possibility of getting cancer.	
at average risk (adj)	Has the same possibility of getting cancer as the general population.	
at high risk (adj)	Has a greater possibility of getting cancer than the general population.	
at higher risk than 'X" (adj)	Has a greater possibility of getting cancer than "X".	
risk factor (n)	A circumstance that increases the risk of getting cancer.	
runs in the family (e.g. cancer runs in the	Is passed down from parents to children to grandchildren.	

Term	Definition	Equivalent or Paraphrase
family) (v)		
saliva (spit) test (n)	A genetic test done by collecting saliva (spit) instead of blood.	
screen (v)	To test a person for a particular common health problem, even though the person has no symptoms to suggest that they have the problem.	
screening test (n)	A test that looks for a common health problem even though the patient has no symptoms to suggest they have the particular disease.	
sperm (n)	The reproductive cells of the man. When sperm fertilize a woman's egg, a baby develops.	
sporadic (adj)	Random, once in a while.	
	In genetics, sporadic cancers are those caused by random chance or unknown factors in the environment.	
statistically significant (adj)	Not caused by chance.	
syndrome (n)	A group of symptoms or features that consistently occur together or a condition characterized by a set of associated features.	
	For example, a hereditary cancer syndrome refers to risk for a group of cancers all caused by a particular gene mutation. BRCA1 mutations increase risk for breast and ovarian cancers. This is called Hereditary Breast and Ovarian Cancer Syndrome. See also, Lynch Syndrome.	
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.	
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	

Term	Definition	Equivalent or Paraphrase
	"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.	
uninformative negative (n)	Sometimes a family has a strong history of a particular cancer, but no genetic change related to that cancer has been found in that family. An "uninformative negative" result means that a specific test was negative, but is not conclusive because the "family" mutation has not being identified, and/or there may be changes that the test could not detect in the gene being analyzed, or in other genes that were not tested.	
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.	
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.	
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."	
trait (n)	A characteristic.	
tumor (n)	An abnormal growth of cells; a tumor can be benign (not harmful) or malignant (harmful, cancer).	
tumor testing (n)	An analysis of the DNA of someone's tumor. This analysis is meant to identify changes that a person might have acquired in his/her tumor cells that are not	

Term	Definition	Equivalent or Paraphrase
	present in the rest of his/her body cells and were not inherited from his/her parents. This testing is often done to help make treatment decisions.	
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.	
ultrasound (n)	An imaging method that uses high frequency sound waves to create a picture of something inside the body.	
variant (n)	A version of something that differs from the norm. So, a genetic variant is a change to the usual genetic sequence.	

Examples of genes that are often checked in genetic testing

Term	Definition	
BRCA 1 BRCA 2	"The Breast Cancer Genes."	
	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.	
MLH1 MSH2	"The Colon Cancer Genes"	
MSH6 PMS2	These genes are involved in fixing mistakes that occur when DNA replicates. An abnormality in one of these genes can lead to Lynch Syndrome, a condition linked to increased risk of cancer in the colon, rectum, uterus, ovaries, as well as other organs.	
ATM	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.	
	A mutation in the ATM gene can lead to increased risks for cancer or a condition called ataxia-telangiectasia a	

Term	Definition
	degenerative neurological condition that causes severe disability.
PALB2	Another gene in which mutations can lead to breast cancer and possibly other cancers.

Family relationships

Term	Definition		
parent	Your mother or father.		
mother	For genetic purposes, the woman whose egg was fertilized and grew to be you.		
mother-in-law	Your husband or wife's mother.		
step-mother	Your father's wife who is not your biological mother.		
adoptive mother	A woman who is not your biological mother but who accepted legal responsibility for and raised you.		
godmother	A woman chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.		
father	For genetic purposes, the man whose sperm fertilized the egg that grew to be you.		
father-in law	Your husband or wife's father.		
step-father	Your mother's husband who is not your biological father.		
adoptive father	A man who is not your biological father but who accepted legal responsibility for and raised you.		
godfather	A man chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church.		
aunt	Your mother or father's sister or sister-in-law. ¹		

¹ NOTE: For Genetic Counselors, it is very important to know if a relative is a blood relative or a relative by marriage.

Term	Definition
maternal aunt	Your mother's sister or sister-in-law. ²
paternal aunt	Your father's sister or sister-in-law. ³
uncle	Your mother or father's brother or brother-in-law.4
maternal uncle	Your mother's brother or brother-in-law. ⁵
paternal uncle	Your father's brother or brother-in-law. ⁶
cousin	Usually understood to be a first cousin.
first cousin	Your aunt or uncle's child.
second cousin	Your aunt or uncle's grandchild.
grandparent	Your parent's father or mother.
grandmother	Your mother or father's mother.
maternal grandmother	Your mother's mother.
paternal grandmother	Your father's mother.
grandfather	Your mother or father's father.
maternal grandfather	Your mother's father.
paternal grandfather	Your father's father.

² ibid ³ ibid ⁴ ibid ⁵ ibid ⁶ ibid

Term	Definition
great aunt	Your mother or father's aunt.
maternal great aunt	Your mother's aunt.
paternal great aunt	Your father's aunt.
great uncle	Your father or mother's uncle.
maternal great uncle	Your mother's uncle.
paternal great uncle	Your father's uncle.
great grandparents	The parents of any of your grandparents.
great grandmother	The mother of any of your grandparents.
great grandfather	The father of any of your grandparents.
siblings	The children of your father and mother.
twins	A sibling born at the same time as you.
identical twins	A twin that developed from the same egg and sperm as you, meaning that you are genetically identical.
fraternal twins	A twin who developed from a different egg and sperm than you, meaning that you are genetically different and have the same amount of shared genes as any sibling.
triplets	Two siblings born at the same time as you.
sister	A sibling who is a girl.
sister-in-law	Your brother's wife.
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.

Term	Definition
step-sister	The daughter of your step-mother or step-father and therefore not biologically related to you.
brother	A sibling who is a boy.
brother-in-law	Your sister's husband.
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.
step-brother	The son of your step-mother or step-father and therefore not biologically related to you.
niece	Your sibling's daughter.
nephew	Your sibling's son.
spouse	Your husband or wife.
wife	The woman to whom you are married.
husband	The man to whom you are married.
children	Genetically speaking, the people who are produced from your egg or sperm.
daughter	Your child who is a girl.
daughter-in-law	Your son's wife.
step-daughter	Your spouse's daughter who is not your biological child.
adoptive daughter	A girl for whom you have accepted legal responsibility and raised even though she is not your biological child.
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.

Term	Definition		
son	Your child who is a boy.		
son-in-law	Your daughter's husband.		
step-son	Your spouse's son who is not your biological child.		
adoptive son	A boy for whom you have accepted legal responsibility and raised even though he is not your biological child.		
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.		
ancestor	A person from whom you are descended, usually more remote than a grandparent.		
fiancé(e)	The person whom you have promised to marry.		
	Man: fiancé		
	Woman: fiancée		
domestic partner	The person with whom you are living and have an intimate relationship, but to whom you are not married.		
divorced	Having ended a marriage.		
engaged	Having promised to marry someone.		
widow	A woman whose husband has died.		
widower	A man whose wife has died.		
relative	A person in your family.		
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 step-mother; your daughter but not your adopted daughter.		

Term	Definition	
next-of-kin	Your closest living blood relation.	

Common interpreting errors

Source speech	Interpreted as	More accurate rendition
"Cancer just happens by chance."	"Cancer happens suddenly." "Cancer happens because you are unlucky."	"Sometimes we don't know what causes cancer."
cancer genes	cancer cells	genes that may cause cancer when altered (mutated)
"You are at higher risk for developing cancer."	getting cancer	There is a greater possibility that you might get cancer
"These genes protect us against cancer."	"They protect us, like the immune system They kill cancer cells"	"These genes help us to not get cancer."
colonoscopy	scan	Colonoscopy

Analogies

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

Examples:

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

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

"In the DNA there are molecule strings - like in this picture, each letter is a different molecule string."

"So a mutation is <u>like</u> a broken gene."

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

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

"It's like chapters in a book, and like misspellings."

"Our cells are like libraries and our chromosomes are like books."

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

"Inside every cell are genes, which are <u>like</u> instruction books for our bodies; they tell our bodies how to grow and develop."

"It is kind of <u>like</u> if you a reading a book = and you notice that a word is spelled wrong; that is what this genetic test looks for."

Genetic counselors may mix the analogy in with more literal descriptions:

Example:

"We know that there are two in particular that when they don't work right, when there's a typo in the instruction manual, they can cause a high risk for breast and ovarian cancer."

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

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

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

"So a mutation is **like** an unexpected change in the order of the letters."

"It's like chapters in a book, and like misspellings."

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

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

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

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

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