MYPG | GLOSSARY OF TERMS

-A-	
Allele	An allele is one of two or more versions of a gene. An individual inherits two alleles for each gene, one from each parent.
Alteration	A change resulting in something that is different from the original.
Amino acids	A set of 20 different molecules used to build proteins. Proteins consist of one or more chains of amino acids called polypeptides. The sequence of the amino acid chain causes the polypeptide to fold into a shape that is biologically active. The amino acid sequences of proteins are encoded in the genes.
Amplification	Increase in the number of copies of a gene sequence. Cancer cells sometimes produce multiple copies of genes in response to signals from other cells or their environment.
-C-	
CancerComplete	This test uses a more in-depth approach to look at part of the genome which includes 20,766 genes in the cancer cells. This testing provides information about particular genes in tumor cells and how they are different from normal cells.
CancerSelect	This test looks at 120 well-characterized cancer genes and the genes that affect how a person responds to medications.
Chromosome	Organized package of DNA found in the nucleus of the cell. Humans have 23 pairs of chromosomes--22 pairs of numbered chromosomes, called autosomes, and one pair of sex chromosomes, X and Y. Each parent contributes one chromosome to each pair so that offspring get half of their chromosomes from their mother and half from their father.
Circulating tumor DNA	Small pieces of genetic information from the cancer or tumor that can be found in blood or other bodily fluids outside of the actual tumor.
Clinically actionable	Describes a mutation that has a known therapy or treatment to target it.
Clinical trial	A type of research study that tests how well new medical approaches work in people. These studies test new methods of screening, prevention, diagnosis, or treatment of disease.
-D-	
Deletion	Type of mutation involving the loss of genetic material. It can be small, involving a single missing DNA base pair, or large, involving a piece of a chromosome.
Diagnostic	Having to do with the process of identifying a disease or sub-type of a disease.
DNA	A chemical name for the molecule that carries genetic instructions in all living things. The DNA molecule consists of two strands that wind around one another to form a shape known as a double helix. Each strand has a backbone made of alternating sugar (deoxyribose) and phosphate groups. Attached to each sugar is one of four bases--adenine (A), cytosine (C), guanine (G), and thymine (T). The two strands are held together by bonds between the bases; adenine bonds with thymine, and cytosine bonds with guanine. The sequence of the bases along the backbones serves as instructions for assembling protein and RNA molecules.
DNA Sequencing	DNA sequencing is a laboratory technique used to determine the exact sequence of bases (A, C, G, and T) in a DNA molecule. The DNA base sequence carries the information a cell needs to assemble protein and RNA molecules. DNA sequence information is important to scientists investigating the functions of genes. The technology of DNA sequencing was made faster and less expensive as a part of the Human Genome Project.
-E-	
Enzyme	A protein that speeds up chemical reactions in the body.
Exome	The part of the genome that corresponds to the complete complement of exons of an organism or cell.
Exon	A portion of a gene that codes for amino acids.

-F-	
FFPE (formalin-fixed paraffin-embedded)	A method of saving and storing a tissue sample (like tumor) that has been removed from the body.
Frameshift	A frameshift mutation is a type of mutation involving the insertion or deletion of a nucleotide in which the number of deleted base pairs is not divisible by three. “Divisible by three” is important because the cell reads a gene in groups of three bases. Each group of three bases corresponds to one of 20 different amino acids used to build a protein. If a mutation disrupts this reading frame, then the entire DNA sequence following the mutation will be read incorrectly.
-G-	
Gene	Basic physical unit of inheritance. Genes are passed from parents to offspring and contain the information needed to specify traits. Genes are arranged, one after another, on structures called chromosomes. A chromosome contains a single, long DNA molecule, only a portion of which corresponds to a single gene. Humans have approximately 20,000 genes arranged on their chromosomes.
Genome	The entire set of genetic instructions found in a cell. In humans, the genome consists of 23 pairs of chromosomes, found in the nucleus, as well as a small chromosome found in the cells’ mitochondria. These chromosomes, taken together, contain approximately 3.1 billion bases of DNA sequence.
Germ line	The sex cells (eggs and sperm) that are used by sexually reproducing organisms to pass on genes from generation to generation. Egg and sperm cells are called germ cells, in contrast to the other cells of the body that are called somatic cells.
-I-	
Insertion	A type of mutation involving the addition of genetic material. An insertion mutation can be small, involving a single extra DNA base pair, or large, involving a piece of a chromosome.
-L-	
Ligand	A molecule that binds to another molecule, used especially to refer to a small molecule that binds specifically to a larger molecule.
-M-	
Mutation	A change in a DNA sequence. Mutations can result from DNA copying mistakes made during cell division, exposure to ionizing radiation, exposure to chemicals called mutagens, or infection by viruses. Germ line mutations occur in the eggs and sperm and can be passed on to offspring, while somatic mutations occur in body cells and are not passed on.
-N-	
Nonsense	A nonsense mutation is the substitution of a single base pair that leads to the appearance of a stop codon where previously there was a codon specifying an amino acid. The presence of this premature stop codon results in the production of a shortened, and likely nonfunctional, protein.
Nonsynonymous coding	A nonsynonymous substitution results in a change in amino acid.
-O-	
Oncogene	Mutated gene that contributes to the development of a cancer. In their normal, unmutated state, oncogenes are called proto-oncogenes, and they play roles in the regulation of cell division. Some oncogenes directly cause a cell to divide. Other oncogenes can prevent other protective measures from working properly, and the cancer cell continues to divide.
Overexpression	Excessive expression of a gene by producing too much of its effect or product, which may lead to the development of cancer when occurs in specific genes
-P-	
Pathway	The sequence of usually enzyme-catalyzed reactions by which one substance is converted into another.
Plasma	The fluid portion of the blood that carries the red blood cells, white blood cells, and platelets.
PCR Polymerase chain reaction (PCR):	A laboratory technique used to amplify DNA sequences. The method involves using short DNA sequences called primers to select the portion of the genome to be amplified. The temperature of the sample is repeatedly raised and lowered to help a DNA replication enzyme copy the target DNA sequence. The technique can produce a billion copies of the target sequence in just a few hours.
Prognostic mutation	A change in the genetic information of the cancer cells that indicates the chances the cancer can be treated successfully; may suggest the likely course or outcome of the disease.

Protein	Important class of molecules found in all living cells. A protein is composed of one or more long chains of amino acids, the sequence of which corresponds to the DNA sequence of the gene that encodes it. Proteins play a variety of roles in the cell, including structural (cytoskeleton), mechanical (muscle), biochemical (enzymes), and cell signaling (hormones).
-R-	
Rearrangement	A structural alteration in a chromosome, usually involving breakage and reattachment of a segment of chromosome material, resulting in an abnormal configuration; examples include inversion and translocation.
-S-	
Sequencing	The process of determining the order and components of a piece of DNA.
Serum	Watery portion of blood left over after red blood cells, white blood cells, platelets and clotting factors have been removed.
SNP	Single nucleotide polymorphisms (SNPs) are a type of polymorphism involving variation of a single base pair.
Somatic	A somatic cell is any cell of the body except sperm and egg cells. Somatic cells are diploid, meaning that they contain two sets of chromosomes, one inherited from each parent. Mutations in somatic cells can affect the individual, but they are not passed on to offspring.
Synonymous	(also called a silent substitution) is the substitution of one base for another in an exon of a gene coding for a protein, such that the produced amino acid sequence is not altered.
-T-	
Targeted therapy	Regarding cancer treatment, targeted therapies block the growth and spread of cancer by interfering with specific molecules involved in tumor growth and progression. The molecules are considered the “targets.”
Therapeutic mutation	A change in the genetic information of the cancer cell that indicates what treatments will or will not be effective.
Translocation	A type of chromosomal abnormality in which a chromosome breaks and a portion of it reattaches to a different chromosome. Chromosomal translocations can be detected by analyzing karyotypes of the affected cells.
Tumor suppressor gene	A tumor suppressor gene directs the production of a protein that is part of the system that regulates cell division. The tumor suppressor protein plays a role in keeping cell division in check. When mutated, a tumor suppressor gene is unable to do its job, and as a result uncontrolled cell growth may occur. This may contribute to the development of a cancer.
-W-	
Wild-type	The normal gene or allele, the opposite of mutant.
Whole genome sequencing	Testing the entire set of genetic instructions of an organism.

Definitions obtained from:

The National Cancer Institute, part of the National Institutes of Health: http://www.cancer.gov/cancertopics

The National Human Genome Research Institute, part of the National Institutes of Health: http://www.genome.gov/glossary/

The Genetics Home Reference, part of the National Institutes of Health: http://ghr.nlm.nih.gov/glossary

The Merriam-Webster Dictionary: http://www.merriam-webster.com/dictionary