Genetics Glossary

**Adenine**: One of four chemical base pairs that make up DNA

**Autosomal dominant**: The mode of inheritance where an individual receives a mutation from one parent. This single mutation is sufficient to cause disease.

**Autosomal recessive**: The mode of inheritance where an individual receives a mutation in the same gene from both parents and develops associated symptoms/cancers.

**Base pairs**: Base pairs are what DNA is made of.

**BRCA PRO**: A breast and ovarian cancer risk assessment/mutation probability model

**Cell**: The basic unit of life. The human body is made up of trillions of different types of cells.

**Chromosome**: Genes are packaged into structures called chromosomes.

**CLAUS**: A breast cancer risk assessment model

**Cytosine**: One of four chemical base pairs that make up DNA

**DNA (deoxyribonucleic acid)**: DNA is what genes are made of.

**Environment**: Anything that we are exposed to

**GAIL**: A breast cancer risk assessment model

**Gene/genetics**: The “blueprint” for life. Genes are inherited from our parents, and are essentially instructions for the body.

**Genetic counselor**: Someone who is board certified and able to analyze your risk for hereditary conditions

**Genetic testing**: Either a blood or saliva test that is used to look for mutations in our genes

**GINA**: The Genetic Information Nondiscrimination Act of 2008 is a federal law that protects individuals from genetic discrimination in health insurance and employment. Genetic discrimination is the misuse of genetic information.

**Guanine**: One of four chemical base pairs that make up DNA

**Hereditary/inherited**: Something that is “passed down” from a parent to a child
**HIPPA**: Federal Health Insurance Portability and Accountability Act of 1996. This law establishes national standards to protect individuals' medical records and other personal health information.

**Mammogram**: A specialized medical imaging device that uses a low-dose X-ray system to see inside the breasts and look for cancer

**Management**: Follow-up care and/or treatment

**Next Generation Sequencing**: A relatively newer method of performing genetic analysis. This method can be used to analyze several genes at the same time.

**(Breast) MRI**: A form of breast cancer screening that uses magnetic fields, radio waves, and a computer to produce detailed pictures of the structures within the breast

**Mutation**: A change in a gene that is typically harmful

**Negative**: One of three possible results one can receive after genetic testing. This implies that no disease-causing mutation was identified in any of the genes analyzed.

**Panel**: Also known as “next generation sequencing,” a panel is a cost and time-effective method of analyzing multiple genes at the same time.

**Polyp**: Abnormal growths of tissue that can be found in any organ and can be either benign or precancerous

**Positive**: One of three possible results one can receive after genetic testing. This implies that a disease-causing mutation was detected with testing.

**Predisposition**: Having a higher risk

**Protein**: The product of a gene

**Risk assessment**: The process used by genetic counselors of looking at risk factors and family history to estimate a person’s risk for having a mutation or for getting cancer

**Single-site test**: Genetic testing that looks specifically for a mutation that is already present/known in the family. The remaining gene and other genes are not analyzed.

**Sporadic**: Something that happens randomly

**Tamoxifen**: A medication used for treatment of breast cancer. This can also be considered in unaffected individuals to help reduce the risk for developing future breast cancers.
**Thymine**: One of four chemical base pairs that make up DNA

**Tyrer-Cuzick (IBIS)**: A breast cancer risk assessment/mutation probability model

**Variant of uncertain significance**: One of three possible results that one can receive from genetic testing. This result implies that there is currently not enough information about this particular change in the gene to understand its role in causing disease.