Genetic Testing and HDGC

PATIENT RESOURCES
HDGC

BACKGROUND

What Are Genes?

Genes are a biological material that provide instructions for how our bodies look and function. Genes are made up of a chemical called deoxyribonucleic acid, or DNA.


Genetic Testing

Gene Variants


Genetic Counselor

What Are Gene Variants?
DNA is made up of four building-block chemicals, labeled with the letters A, T, C and G. These letters of DNA need to be in a particular order for genes to work properly. Gene variants, or mutations, occur when the letters are out of order, added or missing.

A gene variant that contributes to the development of a disease is known as a *pathogenic or likely pathogenic variant (PLP)*

What is Genetic Testing?
Genetic testing is a type of medical test that looks at the order of the letters in your DNA to determine what gene variants you carry. This may help determine any diseases or disease risks that you have.

What is a Genetic Counselor?
Genetic counselors have training in medical genetics and counseling to guide patients as they look for more information about how inherited diseases might affect them or their relatives.

A genetic counselor will help guide you through the process of getting a genetic test and interpreting its results. The National Society of Genetic Counselor's website has a tool for finding a genetic counselor that you can access **here**.

You may also be able to meet with a doctor, nurse or physician assistant who specializes in cancer genetics, if that resource is available in your hospital or cancer center.

Refer to "**Questions to Ask Your Genetic Counseling Provider**" for some questions you may want to ask your genetic counselor before having any genetic tests.

Who Would Benefit From Genetic Testing?

You may benefit from genetic testing if you have been diagnosed with diffuse gastric cancer (DGC). If you have not had cancer but you have a blood relative (parent, sibling, grandparent, aunt, uncle or cousin) that has been diagnosed with diffuse gastric cancer, then you may benefit from genetic testing. Having cases of intestinal gastric cancer and other types of cancers in the family, like breast cancer, can also be a reason to seek genetic testing.

Genetic Testing for Hereditary Gastric Cancer

Diffuse gastric cancer can run in families if there is a pathogenic or likely pathogenic (PLP) variant in the *CDH1* or *CTNNA1* gene. There are other genes that are known to increase the risk of developing intestinal gastric cancer. These genes are *APC*, *BMPR1A*, *EPCAM*, *MLH1*, *MSH2*, *MSH6*, *PMS2*, *SMAD4*, *STK11* and *TP53*. There is also early evidence to suggest that the genes *ATM*, *BRCA1*, *BRCA2*, *CHEK2* and *PALB2* may increase the risk of gastric cancer.

If you or a close blood relative have been diagnosed with gastric cancer then genetic testing may be right for you.

Gene panel testing involves testing for more than one gene at the same time. All genetic testing is performed on a blood, cheek swab or saliva sample to look for gene variants.

Legal Protections Against Genetic Discrimination

The Genetic Information Nondiscrimination Act (GINA) was signed into federal law in 2008. GINA prohibits health insurers and most employers from discrimination against individuals based on genetic information, including the results of genetic tests and family history information. More information about GINA can be found by visiting www.ginahelp.org.

I'VE DONE GENETIC TESTING AND HAVE A *CDH1* PLP VARIANT. NOW WHAT?

If you have a PLP variant, you'll want to consider your cancer risk management options. Information on your options can be found in "**What is Hereditary Diffuse Gastric Cancer (HDGC)**." In addition, it is important to consider what this means for your family members. Though everyone has two copies of the *CDH1* gene, one from their father and one from their mother, it only takes one PLP variant in *CDH1* for an individual to be at risk for cancer. This type of genetic inheritance is called an *autosomal dominant pattern*. This pattern means that parents, children and siblings of an individual with a *CDH1* variant have a 50 percent chance of having that same variant in their genes.

See "**Speaking With Your Family**" for some recommendations on how to discuss your genetic testing results with family members. Any relative 18 years old or older is eligible for genetic testing. Anyone considering genetic testing should receive appropriate education and counseling.

Reproductive options for individuals with a pathogenic variant in *CDH1*

If you are considering having children, you may be wondering if there are ways to avoid passing a PLP gene variant on to your children. Prior to becoming pregnant, it is recommended to meet with a genetic counselor who specializes in fertility to discuss prenatal diagnoses, the use of a donor or preimplantation genetic testing.

Preimplantation genetic testing (PGT) allows people who carry a PLP gene variant to have children without that variant. The PGT process begins with in vitro fertilization (IVF).

It is important to keep in mind that PGT can be an emotional and costly process that requires a commitment of time. Decisions around having children can be complex for individuals and couples. For information and support surrounding these issues, please visit **https://sharinghealthygenes.com/**.