

Genetic Counseling: What You Never Knew You Needed to Know

By Meg Bradbury, MS, CGC, MSHS

Genetic counselors are trained healthcare professionals with a degree in genetics and counseling which enables them to provide personalized guidance to people who may need to make decisions about their own, or their family's, health. These counselors have received training primarily from accredited programs focusing on coursework in psychology, statistics, counseling, developmental anatomy, biochemistry, genetics, and clinical research, as well as rotations of clinical training. After the training these newly-minted genetic counselors often go on to complete an examination and an evaluation of their clinical portfolio offered by the American Board of Genetic Counselors (ABGC) to ensure that standards have been met to provide competent genetic counseling services. Genetic counselors have expertise in genetic testing, research, disease management and prevention, risk assessment, and how inherited diseases and conditions such as bleeding disorders affect families. Currently there are more than 4,000 certified genetic counselors in the US.

With so much to learn about genetic counselors and how they work within the bleeding disorders community, we've put together a comprehensive list of questions, and even provided the answers! You may not have known what you needed to know but that's about to change!

What is the role of a genetic counselor?

Often, genetic counselors serve as medical advocates who use their expertise to assist clients and families to understand the natural history, inheritance patterns, and inheritance risk of bleeding disorders. They use the family's history and information about the disorder to determine the recurrence risk and the benefits or limitations of genetic testing. Genetic counselors, because of their training, have an ability to adapt to the needs of the patient and work as a liaison with the other clinicians by helping to identify and provide resources and support. They often serve as a bridge between medical specialties and are able to provide a clear understanding about complex medical information. Genetic counselors can aid patients and families to absorb those thorny medical concepts and figure out what options might be best integrated into their own care.

What does a genetic counseling session look like?

Typically, genetic counseling sessions last between 30-60 minutes and comprise a number of elements including contracting, information gathering, risk assessment, education, psycho-social assessment/counseling, privacy concerns, and case management. Ideally, a relationship with a genetic counselor should involve a number of meetings and include pre- and post-test sessions. One of the goals in pre-test counseling is to develop a testing strategy and help the patient or family to figure out the best options for them.

As a woman in a family with a bleeding disorder, what special considerations should I know about?

Women who are at-risk for being a carrier should complete genetic testing prior to conception or early on in their pregnancy. This allows the family to take additional steps during the delivery process, such as having factor on hand to prevent hemorrhaging or complications during birth for either the mother or the newborn. Prenatal testing might be of interest for the parents and a genetic counselor would discuss some of these tests such as chorionic villus sampling (CVS) and amniocentesis (Amnio). However, these tests are only an option if a familial pathogenic genetic variant is known.

For women who are not yet pregnant there are also pre-conception options to discuss with a genetic counselor such as in vitro fertilization (IVF) and pre-implantation genetic diagnosis (PGD). For IVF, fertilization of the egg with sperm occurs within a laboratory to form a blastula, or embryo, and then at the early cell stage of development PGD can be used to identify which embryos carry a pathogenic variant that causes a bleeding disorder. The parents can choose to implant only those without a pathogenic variant, or to prepare to raise a child with a bleeding disorder.

What is genetic testing and how is it done?

As many within the community may know, testing for hemophilia includes the measuring of how much factor VIII and factor IX protein is circulating in an individual's blood. This

result can be diagnostic in males but in females who may be carriers factor level alone may not be informative. Women were previously thought to be asymptomatic carriers but have now been shown, in some cases, to be symptomatic and receive a diagnosis of hemophilia. Genetic testing looks specifically at genes, such as factor VIII and factor IX, that can carry a pathogenic variant and lead to hemophilia A or hemophilia B, respectively. Having the results from this gene test allows for molecular confirmation of the clinical diagnosis and it can therefore be used for targeted genetic testing in other at-risk family members. This can be of particular importance for carriers in which factor level might not be revealed.

Genetic confirmation of the diagnosis can be useful to an entire extended family, and prompt testing of other individuals at risk for being carriers and also for future reproductive planning and prenatal diagnosis. A molecular or genetic diagnosis can also be used for enrollment in clinical trials or research studies. Results can take several weeks to be reported back to the clinician.

Genetic test results can be dense and complicated, but a genetic counselor is specifically trained to help interpret and translate this complex information in comprehensive clinical notes. These notes can be used as a resource to speak with other providers and help explain risk, diagnosis, and genetic results. Some genetic results have been associated with certain clinical presentations and may provide information about disease progression or severity, or inhibitor risk. This type of information in medicine is called genotype-phenotype correlation. Genotype refers to the gene pattern instruction in an individual and phenotype refers to the clinical presentation, like factor levels. Not all variants identified in a gene are considered disease-causing or pathogenic. For example, some variants or changes found within our genes are considered benign (variant likely benign, or VLB) while others may be considered variants of uncertain significance (VUS), a status about which we know little at this time. Genetic counselors are trained to help interpret these genetic results in relation to the diagnosis and how they may relate to the family. Often genetic counselors have this discussion about possible result outcomes prior to testing in order for patients and families to be prepared for whatever the result may be. Genetic counselors can also help develop letters to other family members who may have questions or be interested to know their own risk.

Are genetic counseling services covered by insurance?

Genetic counseling is typically paid for by health insurance and in many cases insurance also pays for genetic testing when it is recommended by a clinician. It is important to check with

your health insurance company to verify coverage and benefits including out-of-pocket deductibles before beginning the process. Depending on the type of insurance and the company's policies, you may also need a referral from your provider. Insurance companies have different policies and may vary in coverage for genetic testing. Some companies require meeting with a genetic counselor prior to genetic testing. Genetic counselors can work with families to figure out who should be tested first in the family for the more targeted testing which can be more cost effective. Support organizations and/or research studies may also offer genetic testing. It is important to discuss these opportunities with your clinical team to determine how these may be appropriate for you or your family.

How to find a genetic counselor?

Genetic counselors work in a variety of settings including hematology clinics, genetic testing laboratories, hospitals, research studies, clinical trials, insurance companies, or through telemedicine in which you can connect with a genetic counselor remotely from any location, including your home. As clinicians, genetic counselors may also practice within your local hemophilia treatment centers (HTCs), seeing individuals or entire families, or they might specialize in particular areas, like prenatal, pediatrics, neurology or oncology.

Looking for a genetic counselor in your area?

1. Contact your local HTC and ask to schedule an appointment with the genetic counselor.
2. The National Society of Genetic Counselors (NSGC) has a service resource entitled, *Find a Genetic Counselor Directory*. This directory was developed to assist physicians, patients and other genetic counselors in locating genetic counseling services, and for students curious about the profession. Visit the NSGC website at www.nsgc.org and use the search function to search by "Types of Specialization" in which you can select 'Hematology' as an option.

About the Author

Meg Bradbury is a Lead/Senior Genetic Counselor at GeneDx in the Neurogenetics Department. She graduated from the genetic counseling program at University of Wisconsin-Madison in 2005. Prior to GeneDx, Meg worked as a genetic counselor at Children's National Medical Center at a multidisciplinary neuromuscular disorder clinic. She went on to get her Masters in Clinical and Translational Research at George Washington. She is a member of the National Society of Genetic Counselors. She is a board member of the Hemophilia Association of the Capital Area (HACA) and the Genetics Work Group of the American Thrombosis and Hemostasis Network (ATHN)/National Hemophilia Program Coordinating Center (NHPCC). ■■

