



**EDUCATIONAL OBJECTIVE:** Readers will utilize genetic counselors for patients who have a personal or family history of a hereditary condition

**JESSICA L. MESTER, MS, CGC**  
 Certified Genetic Counselor, Genomic  
 Medicine Institute, Cleveland Clinic

**ALLISON H. SCHREIBER, MS, CGC**  
 Certified Genetic Counselor, Genomic Medicine  
 Institute, Cleveland Clinic

**ROCIO T. MORAN, MD**  
 Genomic Medicine Institute and Pediatric  
 Institute, Cleveland Clinic

# Genetic counselors: Your partners in clinical practice

## ABSTRACT

As our understanding of the human genome has grown, so too has the need for health care providers who can help patients and families understand the implications of these new discoveries for their health care. Increasingly, genetic counselors are working in partnership with physicians to provide a continuum of care from risk assessment to diagnosis. In this article, we explain the process of genetic counseling and its value for patients who have a personal or family history of a hereditary condition.

## KEY POINTS

The sequencing of the human genome has provided valuable information about the genetic causes of many conditions, but it has also uncovered tremendous complexities.

Genetic counselors are master’s-trained allied health care professionals with specific expertise in identifying and educating patients at risk for inherited conditions.

Genetic testing should not be ordered without informed consent and without appropriate counseling before and after the test.

Huntington disease, which is inherited in an autosomal dominant manner, illustrates the need for genetic counseling before predictive testing.

The National Society of Genetic Counselors ([www.nsgc.org](http://www.nsgc.org)) and the American Board of Genetic Counseling ([www.abgc.net](http://www.abgc.net)) provide searchable databases of genetic counselors.

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**S**UPPOSE A NEW PATIENT walks into your office for a routine physical examination. As part of your discussion, you ask about her family history. She relates that her grandmother and uncle had colon cancer.

You know that colon cancer can be hereditary, but you are unsure whether this patient’s family history is significant. You know genetic testing can be ordered, but you only have 15 minutes with the patient and you are unsure which test is appropriate and how it can be ordered. What should you do next?

With advances in genetics and genomics have come expectations that health care providers understand and apply these discoveries to patient care. Identification of a genetic diagnosis can lead to personalized treatment and intensive screening, which can reduce the patient’s risk of contracting the disease in question or dying of it.<sup>1,2</sup> But genetic testing may also take patients on an emotional journey as they adjust to learning new information about themselves and the health care implications such a diagnosis may have for themselves and their family members.

Genetic counseling is an important component of risk assessment and testing. With increasing demands and shorter appointment times, physicians are finding it harder to provide comprehensive risk assessment and genetic counseling.<sup>3-5</sup> Just as “physician extenders” have helped streamline various aspects of health care, genetic counselors can serve as partners to physicians, from helping determine which testing to consider to helping guide follow-up care after test results are received.

Genetic counselors can help not only patients who have a personal or family history

of a hereditary condition, but also their physicians and family members. This article will explain the process of genetic counseling and testing, highlight complexities through case examples, and provide a brief review outlining which patients should be referred for genetic counseling.

## ■ WHAT IS GENETIC COUNSELING?

The National Society of Genetic Counselors defines genetic counseling as “the process of helping people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease.”<sup>6</sup>

The process includes:

- Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence
- Education about inheritance, testing, management, prevention, resources, and research
- Counseling to promote informed choices and adaptation to the risk or condition.<sup>6</sup>

## ■ WHAT HAPPENS DURING A COUNSELING SESSION?

The goals and outcomes of a successful genetic counseling session (TABLE 1) reflect the need for genetic counselors to not only give patients enough information to understand what is being discussed, but also to monitor their emotional responses and respond to their needs for support.<sup>7</sup> The components of a typical genetic counseling session include:

- Contracting (reviewing why the patient is here)
- Reviewing the patient’s personal medical history
- Documenting relevant diagnoses in the family history
- Educating about the condition in question and relevant basic information about genetics
- If testing is indicated, educating about what the test will and will not tell the patient
- If test results are being discussed, discussing the implications of the results for the patient’s management and the utility of testing for relatives
- Identifying additional sources of support

TABLE 1

### Goals and outcomes of a successful genetic counseling session

- Collect family history in a detailed format
- Educate the patient about the condition and patterns of inheritance
- Provide immediate and long-term psychosocial support
- Provide guidance about what the diagnosis means for the future
- Facilitate family communication
- Aid in decision-making

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and education for patients, such as disease-specific support groups

- Making sure the patient understands the information provided
- Monitoring the patient’s emotional and psychological reactions and responding appropriately.

Before the visit, which may last from 30 minutes to several hours, the genetic counselor reviews the patient’s available medical information, performs a literature search covering relevant topics, and prepares supporting educational resources such as visual aids. After the visit, the genetic counselor contacts the patient to discuss the results of any tests ordered, makes sure the follow-up plan is clear, and arranges return visits if these are indicated. Studies have shown that these nonbillable patient-related activities take at least as much time as the actual patient visit.<sup>8,9</sup>

## ■ EVIDENCE THAT GENETIC COUNSELING IS BENEFICIAL

Although genetic counseling may be time-consuming, its benefits to patients have been proven in a number of studies.

**Improved patient knowledge.** Three controlled trials found a significant increase in knowledge about cancer genetics in patients who received genetic counseling as part of their clinical services.<sup>10–12</sup> Additionally, a large prospective multicenter study found a continued significant increase in cancer ge-

**Visits may last from 30 minutes to several hours**

netics knowledge in women who had received genetic counseling for inherited breast cancer risk 1 year earlier.<sup>13</sup>

**More accurate perception of risk.** A meta-analysis of three studies found a significant increase in the accuracy of breast cancer risk perceptions among women who had received genetic counseling.<sup>14</sup>

**Improved psychosocial outcomes.** Anxiety was reduced in 82% of parents who received genetic counseling after screening of their newborn was positive for hemoglobinopathy trait.<sup>15</sup> And 1 year after genetic counseling, parents of patients with psychotic disorders reported reduced anxiety as a result of an increased understanding of accurate recurrence risks.<sup>16</sup>

**Improved risk-reducing behaviors.** Increased genetic counseling support led to improved communication and increased contact with genetics services for at-risk family members.<sup>17</sup> Genetic counseling also led to higher rates of mammography, clinical breast examination, and breast self-examination.<sup>18</sup>

### ■ WHO ARE GENETIC COUNSELORS?

Genetic counselors are allied health professionals with a master's degree and with specific expertise in identifying and educating patients at risk for inherited conditions. They are certified through the American Board of Genetic Counseling. Genetic counseling is a licensed profession in many states,<sup>19</sup> and licensure legislation is pending in several others.

### ■ HOW GENETIC COUNSELORS FACILITATE DIFFICULT COMPONENTS OF GENETIC TESTING

Genetic counselors can serve as complementary practitioners who possess the time and expertise to discuss some of the more complex components of the genetic testing process, further discussed here.

#### **Making sure that testing is appropriate and that the right test is ordered**

Let us revisit our introductory scenario—a patient presents to your office and relates a family history of colon cancer. What would you do if she then says, “I know there’s a gene for

colon cancer; I want that test today so I can know if I’m at risk.” You get the sense that the patient is anxious and determined to get this testing done today. Which of the following would you do?

- Say “OK,” enter “colon cancer gene” in your hospital’s laboratory ordering system, and pray that the results are normal.
- Remember that a representative from a genetic testing company came by your office and left sample collection kits. Say “OK,” draw the patient’s blood in the tubes provided, check off testing for “comprehensive colorectal genetics panel,” and pray the results are normal.
- Tell the patient: “Most colon cancers are not necessarily caused by an inherited syndrome. However, a detailed analysis of your family history seems warranted. There are many genes that can play a role in inherited colon cancer risk, and I want to make sure the right test is done for the right person in your family. I’m going to refer you to a genetic counselor who can take a detailed family history and discuss the risks and benefits of genetic testing with you.” You make the referral and within 1 or 2 weeks, your patient is seen for genetic counseling.

#### **If you chose ‘colon cancer gene’ testing**

The phlebotomy and laboratory personnel at your facility are likely unsure what kind of sample to draw and where it should be sent. As of this writing, at least 14 genes have been associated with a risk of colorectal cancer, and testing for these genes is available through dozens of laboratories across the country.

In this scenario, your hospital does not have sufficient information to follow through on your orders, and someone pages you to discuss it. However, you are in the midst of a busy clinic and are not able to return the page promptly, so the laboratory informs the patient that it cannot draw her blood for testing today. The patient leaves feeling angry and upset.

#### **If you chose commercial genetic testing**

You may have just ordered testing for four of the genes known to cause Lynch syndrome,

Although genetic counseling may be time-consuming, it has shown benefit

an inherited condition predisposing to colon, uterine, and a few other cancer types. While testing like this may be labeled as “comprehensive,” it may not include all disorders associated with colon cancer. Such shotgun approaches to patient care without consideration of family history can often lead to ordering genetic testing that may be not only medically unnecessary, but also not reimbursable by insurance companies.

Continuing with the case above, the patient’s insurance company determines that testing is not medically necessary, and she is billed for the entire cost of more than \$4,400. Her results are normal, and she feels reassured that she is not at increased risk of colon cancer.

A year later, the patient phones you to say that her uncle had genetic testing with positive results. She sends you the letter she received along with the genetic counselor’s clinic note—the uncle’s mutation is in a completely different gene from the ones you tested. While she was previously told she was at low risk, the appropriate site-specific genetic test (average cost range \$185–\$450) to target the specific mutation is positive, and she is at increased risk of colon cancer, but is now able to pursue increased screening to reduce her risks of developing and dying from this disease.

If the patient had not been made aware of her uncle’s results, she may not have received this screening. If she were diagnosed with later-stage colon cancer after developing symptoms, she may feel you are liable for this diagnosis based on her perception that she was not at risk according to the previously negative genetic testing results ordered by you. After learning about her family history and that the right test was not ordered for her, the patient pursues legal action.

### If you chose genetic counseling

If you chose to refer the patient for genetic counseling, congratulations! Your patient is seen for risk assessment and genetic counseling.

As part of the genetic counseling session, a comprehensive family history identifies the patient’s uncle who was diagnosed with colon cancer. He was previously seen for genetics assessment and was found to have a mutation in the *APC* gene, predisposing him to familial adenomatous polyposis. Site-specific testing,

which the genetic counselor is able to get covered by the patient’s insurance through a letter of medical necessity, reveals that your patient shares her uncle’s mutation. As indicated by national guidelines, you refer the patient to a gastroenterologist for medical management, which will significantly reduce her chances of developing and dying of colorectal cancer.

It is preferable to see the family member at highest risk for an inherited condition—usually, but not always the affected relative—for genetic consultation first. During the consultation the genetic counselor would decide which syndrome, if any, is the best fit for the family.

If the affected relative tests positive, targeted and less costly testing for the specific mutation identified (ie, site-specific testing) can then be offered to family members to provide a yes-or-no answer as to their risk status.

If the relative most likely to be gene-positive tests negative, no genetic testing would be recommended for family members, as the genetic cause of the cancer in the family is unknown. In this situation, family members may be advised to pursue the same screening measures as those with a positive gene test due to their strong family history.

### ■ INFORMED CONSENT FOR GENETIC TESTING

Genetic testing consists of much more than a simple blood draw. Obtaining informed consent for genetic testing is a crucial step in the testing process, as the results can be complex and often affect multiple family members. When *predictive* genetic testing is being discussed, special conversations need to take place to make sure that decisions are well informed. Genetic counselors can facilitate these discussions and guide patients and families through the decision-making process.

### Example: Huntington disease

The need for genetic counseling before predictive testing is best illustrated by Huntington disease, a progressive neurodegenerative disorder with typical onset in the third or fourth decade of life. Over the disease course, patients experience decreases in motor control (leading to the aptly named “Huntington chorea”), cognitive decline, and changes in

**Genetic testing consists of much more than a simple blood draw**

psychiatric state. Ultimately, most patients die 15 to 20 years after the onset of symptoms. Treatment is palliative and symptom-based.

Huntington disease is inherited in an autosomal dominant manner, meaning that each child of an affected person has a 50% risk of inheriting the gene change responsible for this condition and of eventually developing the disease. It is caused by an expansion within the *HD* gene; this expansion may grow with successive generations, leading to earlier onset of symptoms.<sup>20</sup>

The availability of predictive testing—which enables people who are at risk but who are without symptoms to find out their genetic status—ultimately leads each at-risk person to ask herself or himself, Do I want to know? Studies have found that only 15% to 67% of offspring of parents with Huntington disease (offspring are at 50% risk of the disease) elected to be tested, and in one longitudinal study, this rate of “uptake” decreased over time.<sup>21,22</sup> However, any estimates of uptake may be falsely elevated, given the likelihood that those not wishing to consider testing may not feel the need for a clinical visit focused on this subject.

After predictive testing became available, an increased risk of suicide in persons at risk of Huntington disease was documented.<sup>23,24</sup> In view of this risk and the careful decision-making support that people at risk need, predictive testing guidelines were developed by a committee of medical experts and members of Huntington disease family organizations.<sup>25</sup> As part of the guidelines, a multivisit pretesting process was established that includes extensive education and counseling. Delay of testing is recommended when contraindications are identified, such as evidence of coercion or a serious psychiatric condition. Most genetic testing companies offering predictive testing require a signature from the ordering clinician verifying that pretest counseling has been completed; some also include a provision that the ordering clinician will relay results to the patient in person.

More than 15 years after these guidelines were adopted, a study of suicide risk in at-risk persons continued to find rates higher than in the general population, but lower than in earlier studies.<sup>26</sup> Whether this careful pretest

counseling protocol is directly related to a possible decrease in suicide risk has yet to be established, but its successful use in patients undergoing predictive Huntington disease testing has led to its adoption in other neurodegenerative diseases such as Alzheimer disease and Parkinson disease.

### ■ EXPLAINING POSITIVE GENETIC TESTING RESULTS

If genetic testing identifies a mutation, genetic counselors can help patients understand the implications of the results for themselves and for their relatives. Some patients become quite inquisitive, and the genetic counseling session morphs into a graduate-level discussion of genes, DNA, disease pathways, genetic-environmental interactions, availability of gene therapy, and clinical trials. The genetic counselor also makes the patient aware of other resources, such as disease-specific support groups, which may be developed by patients and families to provide support and practical knowledge.

In some cases, attention turns to at-risk relatives, and the genetic counselor may role-play with the patient to rehearse ways to share information with them. Genetic counselors may give patients a letter to distribute to family members with a copy of the patient's test results, briefly explaining the condition identified and how relatives may find a genetic counselor in their area for their own risk assessment.

### ■ WHAT ABOUT GENETIC DISCRIMINATION?

Genetic discrimination is addressed in many genetic counseling sessions.

As defined by the National Human Genome Research Institute, genetic discrimination is “prejudice directed against people who have or may have a genetic disease.”<sup>27</sup>

In May 2008, the Genetic Information Nondiscrimination Act (GINA) was signed into law, providing some legal protections against genetic discrimination for patients undergoing predictive genetic testing. The law applies to most employers and health insurers but does not protect against discrimination by life or disability insurers. When discussing

**Predictive testing guidelines for Huntington disease have been developed by medical experts and members of HD family organizations**



genetic testing, genetic counselors ensure that patients are aware of their rights and protections.

GINA would not be relevant for a patient who has a medical condition that may affect his or her insurability. For example, someone with thyroid cancer who is found to have an underlying gene mutation may still be denied any type of insurance coverage on the basis of his or her personal cancer diagnosis. However, should that person's son who has not been diagnosed with cancer opt to undergo predictive testing, GINA would provide protection against employment and health insurance discrimination, as described above.

### ■ DIRECT-TO-CONSUMER GENETIC TESTING

As DNA technology has become increasingly complex, so has the task of understanding new tests and their clinical relevance to patients.

In the last several years, more companies have begun to offer direct-to-consumer genetic testing, which may be ordered without the involvement of a health care professional. While some companies hire or work closely with genetic counselors to conduct pretest and posttest genetic counseling, others do not, and preliminary research has found that only a minority of primary care physicians feel prepared to answer patients' questions about direct-to-consumer genetic testing.<sup>28</sup>

Genetic counselors stay abreast of emerging technologies and are prepared to answer questions from patients who are considering or have already undergone such testing and from physicians who may wonder if a patient's direct-to-consumer genetic testing results should affect his or her management.

Direct-to-consumer genetic testing will be discussed in depth in a future article in this series.

### ■ EXPLAINING 'NORMAL' (NEGATIVE) GENETIC TEST RESULTS

When testing results are normal, patients are educated about the meaning of "normal" results, the residual risk, and screening that might be appropriate in each person's situation.

Sometimes a normal result does not mean the patient is not at risk for disease—for most diseases, genetic testing is not perfect and can-

not identify a mutation in every at-risk family. Patients who have a family history of certain conditions may still face a higher risk despite normal test results. In these situations it is imperative that the family continue to adhere to follow-up recommendations even with normal test results.

### Example: Marfan syndrome

Marfan syndrome is an autosomal dominant connective tissue disorder that, if unrecognized, is associated with significant morbidity and mortality. People with Marfan syndrome are at increased risk of aortic aneurysms, which can rupture spontaneously, leading to sudden death.

Although at least 70% of patients with Marfan syndrome have a mutation in *FBN1*, other patients meeting the clinical diagnostic criteria do not. Despite a normal genetic test result, they should adhere to the same screening guidelines as a person who tests positive.<sup>29</sup>

This concept—that screening should still be done despite a normal "Marfan test"—may be difficult for patients to grasp without a discussion of the imperfect sensitivity of genetic testing and of their real ongoing risks. Even more difficult to understand is the idea that the patient's family members should also be screened as though they have the disease, given that the family's mutation is unknown and predictive testing cannot be conducted.

Further complicating matters, other disorders such as Loeys-Dietz and vascular Ehlers-Danlos syndrome can mimic Marfan syndrome by causing aortic aneurysms, but management recommendations for them are very different.<sup>30,31</sup>

The appropriate genetic diagnosis for patients with aortic aneurysms can be facilitated by referring them to genetic counselors, who can identify appropriate testing. In this way, physicians can personalize medical management and give screening recommendations specific to the genetic disorder present.

### ■ EXPLAINING UNCERTAIN RESULTS

There are three possible results for most genetic tests—positive (a pathogenic or disease-causing mutation was found), negative (normal), and the frustrating "variant of uncertain significance" (VUS).

**Sometimes, a 'normal' result does not mean the patient is not at risk for the disease**

**TABLE 2**

**Common reasons for referrals for genetic counseling**

**Adults**

Diagnosed adult-onset genetic conditions, such as Huntington disease, myotonic dystrophy, polycystic kidney disease  
 Family history of a genetic condition  
 Abnormal genetic test results

**Cardiovascular**

Cardiomyopathy  
 Thoracic aortic aneurysm at a young age  
 Two or more family members with the same cardiovascular condition  
 Family history of sudden cardiac death  
 Known genetic mutation in the family

**Cancer**

Early age at cancer diagnosis  
 Multiple generations affected by cancer  
 Multiple primary cancers in an individual  
 Clustering of genetically related cancers (eg, colon and uterine cancers)  
 Rare cancers (eg, male with breast cancer)  
 Known genetic mutation the family

**Preconception**

Previous child with a genetic condition, birth defect, or mental retardation  
 Family history of a genetic condition  
 Ethnicity-based screening  
 Multiple pregnancy losses  
 Consanguinity

**Prenatal**

Abnormal maternal serum screen  
 Anomalies on ultrasonography  
 Advanced maternal age  
 Exposure to teratogenic infections, drugs, or potentially mutagenic agents during pregnancy  
 One or both partners with abnormal carrier screening

**Pediatric**

Congenital anomaly (birth defect)  
 Dysmorphic features  
 Developmental delay, intellectual disability, autism  
 Abnormal genetic test result  
 Families concerned about recurrence risk

function and cause disease, a VUS that is clearly unknown poses a challenge not only to patient management, but also to family members seeking personal risk assessments.

Knowledge of how or if specific variants relate to disease is emerging. In time, some variants become reclassified as either disease-causing mutations or benign polymorphisms. However, careful consideration needs to be given to how to explain the abnormal result to the patient and to at-risk family members, as well as to how to explain the clinical implications of the VUS.

**Example: Hereditary breast and ovarian cancer syndrome**

People with hereditary breast and ovarian cancer syndrome face a lifetime risk of breast cancer of up to 87% and a risk of ovarian cancer of up to 44%. Most families with this syndrome have an inherited change in either the *BRCA1* or *BRCA2* gene.<sup>32,33</sup> Given these risks, prophylactic mastectomy and oophorectomy are among the management options for mutation-positive patients. In the absence of clear genetic counseling, a patient with a VUS might see the “abnormal” test result and believe herself to be mutation-positive and thus at very high cancer risk.

An important role for the genetic counselor is to clarify the pathogenicity of a particular VUS. When a VUS is found, genetic counselors search for information about the variant by reviewing the medical literature, discussing it with the testing laboratory, arranging for family studies when appropriate, and contacting researchers whose work focuses on the gene in question.

Failure to properly research a particular VUS can lead to unnecessary and risky surgical procedures, as well as to falsely labelling relatives as being at risk. Until a VUS is reclassified as a disease-causing mutation, testing for it should not be offered to family members (unless through a research study), nor should medical management be based solely on the results of a particular VUS. In time, a VUS may be reclassified as either a benign polymorphism or a disease-causing mutation, and the genetic counselor will recontact the patient and physician with updated information and recommendations.

A VUS result means that an abnormality was detected in the gene, but that there are insufficient data about whether the abnormality alters the function of the gene in question and, thus, leads to disease. Since some gene variants are known to be common in the general population and not linked to disease and others are known to definitely alter a gene’s

## WHOM SHOULD I REFER?

Genetic counseling is available for patients and families in diverse settings within health systems. The six primary areas of practice are general, cardiovascular, cancer, preconception, prenatal, and pediatrics.

Patients with a personal or family history of a hereditary condition can benefit from genetic counseling regardless of whether they would be considered appropriate for genetic testing.<sup>34</sup>

At current count, there are 4,424 genetic disorders for which the underlying cause has been identified.<sup>35</sup> Individually, each disorder is rare, but when they are considered as a whole, they affect a significant minority of the general population. It is estimated that before age 25 years, 53 (5.3%) of every 1,000 people will be diagnosed with a disease that has an important genetic component.<sup>36</sup> From 20% to 30% of infant deaths are related to a genetic disorder,<sup>37,38</sup> and 22% of unaffected adults have a family history of cancer significant enough to warrant a genetics referral.<sup>39</sup> See **TABLE 2** for a list of common indications for referral.

## HOW CAN I FIND GENETIC COUNSELING SERVICES?

The National Society of Genetic Counselors ([www.nsgc.org](http://www.nsgc.org)) and American Board of Ge-

netic Counseling ([www.abgc.net](http://www.abgc.net)) both provide searchable databases of registered genetic counselors.

## KNOWLEDGE CONTINUES TO EXPAND

Genetic knowledge continues to expand, and testing is becoming available for a growing number of medical conditions. Appropriate identification of individuals with and at risk for genetic disorders through the use of genetic testing and screening is a cornerstone of personalized medicine, with the ultimate goal of improving patient outcomes. However, in this era of value-based medicine and fewer health care dollars, genetic testing must be used in a way that maximizes its clinical impact with a careful fiscal approach.

Genetic counselors are specially trained health care professionals with expertise in genetic and genomic medicine who work in collaboration with physicians to guide patients through the complexities of heritable conditions and emerging technologies. They are trained to personalize, interpret, and communicate complex science into data that will assure best outcomes for patients and their families. Developing a partnership with the genetic counselors in your area can provide multiple benefits to your patients as well as to your own practice. ■

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**ADDRESS:** Jessica Mester, MS, Certified Genetic Counselor, Genomic Medicine Institute, NE50, Cleveland Clinic, 9500 Euclid Avenue, Cleveland, OH 44195; e-mail [mesterj@ccf.org](mailto:mesterj@ccf.org).