



# Help Me Understand Genetics

## Genetic Consultation

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# Genetic Consultation

## 1 What is a genetic consultation?

A genetic consultation is a health service that provides information and support to people who have, or may be at risk for, genetic conditions. During a consultation, a genetics professional meets with an individual or family to discuss genetic risks or to diagnose, confirm, or rule out a genetic condition.

Genetics professionals include medical geneticists (doctors who specialize in genetics) and genetic counselors (certified healthcare workers with experience in medical genetics and counseling). Other healthcare professionals such as nurses, psychologists, and social workers trained in genetics can also provide genetic consultations.

Consultations usually take place in a doctor's office, hospital, genetics center, or other type of medical center. These meetings are most often in-person visits with individuals or families, but they are occasionally conducted in a group. Additionally, genetic consultations may be carried out using telemedicine (also known as telehealth), in which the meeting is done from a distance using computers, cameras, videoconferencing tools, or the telephone.

### **For more information about genetic consultations:**

MedlinePlus offers a list of links to information about genetic counseling (<https://medlineplus.gov/geneticcounseling.html>).

MedlinePlus also offers information about telehealth (<https://medlineplus.gov/telehealth.html>).

Additional background information is provided by the National Human Genome Research Institute in its Frequently Asked Questions About Genetic Counseling (<https://www.genome.gov/FAQ/Genetic-Counseling>).

Information about genetic counseling, including the different types of counseling, is available from the National Center for Biotechnology Information (NCBI) in the booklet Making Sense of Your Genes: A Guide to Genetic Counseling (<https://www.ncbi.nlm.nih.gov/books/NBK115508/>).

The National Society of Genetic Counselors discusses what happens during an appointment (<https://www.aboutgeneticcounselors.org/Prepare-for-an-Appointment/What-Happens-During-an-Appointment>).

The Centre for Genetics Education also offers an introduction to genetic counseling (<https://www.genetics.edu.au/publications-and-resources/facts-sheets/fact-sheet-6-genetic-counselling>).

## 2 Why might someone have a genetic consultation?

Individuals or families who are concerned about an inherited condition may benefit from a genetic consultation. The reasons that a person might be referred to a genetic counselor, medical geneticist, or other genetics professional include:

- A personal or family history of a genetic condition, birth defect, chromosomal disorder, or hereditary cancer.
- Two or more pregnancy losses (miscarriages), a stillbirth, or a baby who died.
- A child with a known inherited disorder, a birth defect, intellectual disability, or developmental delay.
- A woman who is pregnant or plans to become pregnant at or after age 35. (Some chromosomal disorders occur more frequently in children born to older women.)
- Abnormal test results that suggest a genetic or chromosomal condition.
- An increased risk of developing or passing on a particular genetic disorder on the basis of a person's ethnic background.
- People related by blood (for example, cousins) who plan to have children together. (A child whose parents are related may be at an increased risk of inheriting certain genetic disorders.)
- A person received results from direct-to-consumer genetic testing and they want to discuss the implications of the results.

A genetic consultation is also an important part of the decision-making process for genetic testing. A visit with a genetics professional may be helpful even if testing is not available for a specific condition, however.

### **For more information about the reasons for having a genetic consultation:**

An overview of indications for a genetics referral (<https://www.ncbi.nlm.nih.gov/books/NBK115554/>) is available from The Genetic Alliance booklet "Understanding Genetics: A Guide for Patients and Professionals."

The Centers for Disease Control and Prevention (CDC) Office of Public Health Genomics has a list of reasons why people go for genetic counseling ([https://www.cdc.gov/genomics/gtesting/genetic\\_counseling.htm](https://www.cdc.gov/genomics/gtesting/genetic_counseling.htm)).

The National Society of Genetic Counselors details the reasons to see a genetic counselor, including pregnancy and family planning (<https://www.aboutgeneticcounselors.org/Reasons-to-See-a-Genetic-Counselor/Pregnancy-and-Family-Planning>), cancer (<https://www.aboutgeneticcounselors.org/Reasons-to-See-a-Genetic-Counselor/Cancer>), childhood conditions (<https://www.aboutgeneticcounselors.org/Reasons-to-See-a-Genetic-Counselor/Childhood-Conditions>), and at-home genetic testing (<https://www.aboutgeneticcounselors.org/Reasons-to-See-a-Genetic-Counselor/At-Home-Genetic-Testing>).

### 3 What happens during a genetic consultation?

A genetic consultation provides information, offers support, and addresses a patient's specific questions and concerns. To help determine whether a condition has a genetic component, a genetics professional asks about a person's medical history and takes a detailed family health history (a record of health information about a person's immediate and extended family). The genetics professional may also perform a physical examination and recommend appropriate medical tests.

If a person is diagnosed with a genetic condition, the genetics professional provides information about the diagnosis, how the condition is inherited, the chance of passing the condition to future generations, and the options for testing and treatment.

During a consultation, a genetics professional will:

- Interpret and communicate complex medical information.
- Help each person make informed, independent decisions about their health care and reproductive options.
- Respect each person's individual beliefs, traditions, and feelings.

A genetics professional will NOT:

- Tell a person which decision to make.
- Coerce a couple about whether or not to have children.
- Recommend that a woman continue or end a pregnancy.
- Tell someone whether to undergo testing for a genetic disorder.

#### **For more information about what to expect during a genetic consultation:**

The National Society of Genetic Counselors offers information about what to expect from a genetic counseling session (<https://www.aboutgeneticcounselors.org/Prepare-for-an-Appointment/What-Happens-During-an-Appointment>), what happens during a prenatal genetic counseling appointment (<https://www.aboutgeneticcounselors.org/Reasons-to-See-a-Genetic-Counselor/Pregnancy-and-Family-Planning>), and insurance coverage for genetic counseling (<https://www.aboutgeneticcounselors.org/Prepare-for-an-Appointment/Insurance-Billing-and-Protecting-Your-Privacy>).

Information about the role of genetic counselors and the process of genetic counseling (<https://www.ncbi.nlm.nih.gov/books/NBK115552/>) are available from the Genetic Alliance publication "Understanding Genetics: A Guide for Patients and Professionals."

## 4 How can I find a genetics professional in my area?

To find a genetics professional in your community, you may wish to ask your doctor for a referral. If you have health insurance, you can also contact your insurance company to find a medical geneticist or genetic counselor in your area who participates in your plan.

Several organizations have tips for finding a healthcare professional:

- The Genetic and Rare Diseases Information Center, a service of the National Institutes of Health, provides a guide to finding specialists in particular genetic and rare conditions.
- The Tuberous Sclerosis Alliance provides advice on finding and choosing a doctor. Although this advice is written for adults with tuberous sclerosis, much of it applies to people with any chronic health condition.

Additional resources for locating a genetics professional in your community are available online:

- The National Society of Genetic Counselors (NSGC) offers a searchable directory of genetic counselors in the United States and Canada. You can search by location, name, area of practice/specialization, and/or ZIP Code.
- The American Board of Genetic Counseling (ABGC) provides a searchable directory of certified genetic counselors worldwide. You can search by practice area, name, organization, or location.
- The Canadian Association of Genetic Counsellors (CAGC) has a searchable directory of genetic counselors in Canada. You can search by name, distance from an address, province, or services.
- The American College of Medical Genetics and Genomics (ACMG) has a searchable database of medical genetics clinic services in the United States.
- The National Cancer Institute provides a Cancer Genetics Services Directory, which lists professionals who provide services related to cancer genetics. You can search by type of cancer or syndrome, location, and/or provider name.

If you have a health condition that has not been diagnosed, you may be interested in the Undiagnosed Diseases Network. They have information about how to apply for this multicenter research study.

## 5 What is the prognosis of a genetic condition?

The prognosis of a genetic condition includes its likely course, duration, and outcome. When health professionals refer to the prognosis of a disease, they may also mean the chance of recovery; however, most genetic conditions are life-long and are managed rather than cured.

Disease prognosis has multiple aspects, including:

- How long a person with the disorder is likely to live (life expectancy)
- When signs and symptoms develop and whether they worsen (and how quickly) or are stable over time
- Quality of life, such as independence in daily activities
- Potential for complications and associated health issues

The prognosis of a genetic condition depends on many factors, including the specific diagnosis and an individual's particular signs and symptoms. Sometimes the associated genetic variant, if known, can also give clues to the prognosis. Additionally, the course and outcome of a condition depends on the availability and effectiveness of treatment and management approaches. The prognosis of very rare diseases can be difficult to predict because so few affected individuals have been identified. Prognosis may also be difficult or impossible to establish if a person's diagnosis is unknown.

The prognoses of genetic disorders vary widely, often even among people with the same condition. This variability is likely caused by a combination of genetic, environmental, and lifestyle factors, many of which can be difficult to identify. Some genetic disorders cause physical and developmental problems that are so severe they are incompatible with life. These conditions may cause a miscarriage of an affected embryo or fetus, or an affected infant may be stillborn or die shortly after birth. People with less severe genetic conditions may live into childhood or adulthood but have a shortened lifespan due to health problems related to their disorder. Genetic conditions with a milder course may be associated with a normal lifespan and few related health issues.

The prognosis of a disease is based on probability, which means that it is likely but not certain that the disorder will follow a particular course. Your healthcare provider is the best resource for information about the prognosis of your specific genetic condition. He or she can assess your medical history and signs and symptoms to give you the most accurate estimate of your prognosis.

### **Learn more about the prognosis of genetic conditions:**

MedlinePlus Genetics provides an additional list of resources that can help you locate a genetics professional (<https://medlineplus.gov/genetics/understanding/consult/findingprofessional/>) in your area.

A discussion of the prognosis of disorders with a neurological basis (<https://www.ninds.nih.gov/healthandinformation/20140813a/genetics/genetics.html>)

ih.gov/Disorders/All-Disorders) is available from the National Institute of Neurological Disorders and Stroke (NINDS).

The National Cancer Institute (NCI) provides an overview of cancer prognosis (<https://www.cancer.gov/about-cancer/diagnosis-staging/prognosis>).

Local and national support and advocacy groups are also excellent resources for information about specific genetic conditions, including disease prognosis. Patient support resources related to specific genetic conditions can be identified through the Genetic Alliance's Disease InfoSearch (<https://www.diseaseinfosearch.org/>).



## 6 How are genetic conditions diagnosed?

A doctor may suspect a diagnosis of a genetic condition on the basis of a person's physical characteristics and family history, or on the results of a screening test.

Genetic testing is one of several tools that doctors use to diagnose genetic conditions. The approaches to making a genetic diagnosis include:

- **A physical examination:** Certain physical characteristics, such as distinctive facial features, can suggest the diagnosis of a genetic disorder. A geneticist will do a thorough physical examination that may include measurements such as the distance around the head (head circumference), the distance between the eyes, and the length of the arms and legs. Depending on the situation, specialized examinations such as nervous system (neurological) or eye (ophthalmologic) exams may be performed. The doctor may also use imaging studies including x-rays, computerized tomography (CT) scans, or magnetic resonance imaging (MRI) to see structures inside the body.
- **Personal medical history:** Information about an individual's health, often going back to birth, can provide clues to a genetic diagnosis. A personal medical history includes past health issues, hospitalizations and surgeries, allergies, medications, and the results of any medical or genetic testing that has already been done.
- **Family health history:** Because genetic conditions often run in families, information about the health of family members can be a critical tool for diagnosing these disorders. A doctor or genetic counselor will ask about health conditions in an individual's parents, siblings, children, and possibly more distant relatives. This information can give clues about the diagnosis and inheritance pattern of a genetic condition in a family.
- **Laboratory tests, including genetic testing:** Molecular, chromosomal, and biochemical genetic or genomic testing are used to diagnose genetic disorders. Other laboratory tests that measure the levels of certain substances in blood and urine can also help suggest a diagnosis.

Genetic testing is currently available for many genetic conditions. However, some conditions do not have a genetic test; either the genetic cause of the condition is unknown or a test has not yet been developed. In these cases, sequencing the entire genome may result in locating the responsible genetic variant. Additionally, a combination of the approaches listed above may be used to make a diagnosis. Even when genetic testing is available, the tools listed above are used to narrow down the possibilities (known as a differential diagnosis) and choose the most appropriate genetic tests to pursue.

A diagnosis of a genetic disorder can be made anytime during life, from before birth to old age, depending on when the features of the condition appear and the availability of testing. Sometimes, having a diagnosis can guide treatment and management decisions. A genetic diagnosis can also suggest whether other family members may be affected by or at risk of a specific disorder. Even when no treatment is available for a particular condition, having a diagnosis can help people know what to expect and may help them

identify useful support and advocacy resources.

### **For more information about diagnosing genetic conditions:**

MedlinePlus Genetics provides additional information about genetic testing (<https://medlineplus.gov/genetics/understanding/testing/>) (<https://medlineplus.gov/genetics/understanding/inheritance/familyhistory/>).

The National Center for Biotechnology Information (NCBI) provides an in-depth guide called Understanding Genetics (<https://www.ncbi.nlm.nih.gov/books/NBK132142/>), which includes a chapter about how genetics professionals diagnose many types of genetic disorders.

The Centers for Disease Control and Prevention (CDC) offers a fact sheet about the diagnosis of birth defects (<https://www.cdc.gov/ncbddd/birthdefects/diagnosis.html>), including information about screening and diagnostic test.

The American College of Medical Genetics offers practice guidelines, (<https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/Practice-Guidelines.aspx>) including diagnostic criteria, for several genetic disorders. These guidelines are designed for geneticists and other healthcare providers.

GeneReviews (<https://www.ncbi.nlm.nih.gov/books/NBK1116/>), a resource from the University of Washington and the NCBI, provides detailed information about the diagnosis of specific genetic disorders as part of each peer-reviewed disease description.

The Undiagnosed Diseases Network (<https://undiagnosed.hms.harvard.edu/>) is a research study that helps people with diseases that have not been diagnosed. They have information about applying to participate (<https://undiagnosed.hms.harvard.edu/apply/>) in this study.

The Genetic and Rare Diseases Information Center (GARD), a service of the National Institutes of Health, also provides tips for the undiagnosed (<https://rarediseases.info.nih.gov/guides/pages/24/tips-for-the-undiagnosed>).

## 7 How are genetic conditions treated or managed?

Many genetic disorders result from gene changes that are present in essentially every cell in the body. As a result, these disorders often affect many body systems, and most cannot be cured. However, approaches may be available to treat or manage some of the associated signs and symptoms.

For a group of genetic conditions called inborn errors of metabolism, which result from genetic changes that disrupt the production of specific enzymes, treatments sometimes include dietary changes or replacement of the particular enzyme that is missing. Limiting certain substances in the diet can help prevent the buildup of potentially toxic substances that are normally broken down by the enzyme. In some cases, enzyme replacement therapy can help make up for the enzyme shortage. These treatments are used to manage existing signs and symptoms and may help prevent future complications. An example of an inborn error of metabolism is phenylketonuria (PKU).

For other genetic conditions, treatment and management strategies are designed to improve particular signs and symptoms associated with the disorder. These approaches vary by disorder and are specific to an individual's health needs. For example, a genetic disorder associated with a heart defect might be treated with surgery to repair the defect or with a heart transplant. Conditions that are characterized by defective blood cell formation, such as sickle cell disease, can sometimes be treated with a bone marrow transplant. Bone marrow transplantation can allow the formation of normal blood cells and, if done early in life, may help prevent episodes of pain and other future complications.

Some genetic changes are associated with an increased risk of future health problems, such as certain forms of cancer. One well-known example is familial breast cancer related to mutations in the *BRCA1* and *BRCA2* genes. Management may include more frequent cancer screening or preventive (prophylactic) surgery to remove the tissues at highest risk of becoming cancerous.

Genetic disorders may cause such severe health problems that they are incompatible with life. In the most severe cases, these conditions may cause a miscarriage of an affected embryo or fetus. In other cases, affected infants may be stillborn or die shortly after birth. Although few treatments are available for these severe genetic conditions, health professionals can often provide supportive care, such as pain relief or mechanical breathing assistance, to the affected individual.

Most treatment strategies for genetic disorders do not alter the underlying genetic mutation; however, a few disorders have been treated with gene therapy. This experimental technique involves changing a person's genes to prevent or treat a disease. Gene therapy, along with many other treatment and management approaches for genetic conditions, are under study in clinical trials.

### **Find out more about the treatment and management of genetic conditions:**

GeneReviews (<https://www.ncbi.nlm.nih.gov/books/NBK1116/>), a resource from the

University of Washington and the National Center for Biotechnology Information (NCBI), provides detailed information about the management of specific genetic disorders as part of each peer-reviewed disease description.

The Genetic and Rare Diseases Information Center, a service of the National Institutes of Health, provides this video with suggestions for finding information about treatment ([https://www.youtube.com/watch?v=by4nQriQcKs&list=PLtOMdJ\\_3bSznIDTV\\_tD2qOKLraENN9PLv&index=3](https://www.youtube.com/watch?v=by4nQriQcKs&list=PLtOMdJ_3bSznIDTV_tD2qOKLraENN9PLv&index=3)) for genetic and rare conditions.

MedlinePlus provides additional information related to the treatment of genetic conditions:

- Inborn Errors of Metabolism (<https://medlineplus.gov/ency/article/002438.htm>)
- Bone Marrow Transplantation (<https://medlineplus.gov/bonemarrowtransplantation.html>)
- Palliative care (<https://medlineplus.gov/palliativecare.html>) (also known as supportive care)
- Gene Therapy (<https://medlineplus.gov/genetics/understanding/therapy/>)
- Precision Medicine (<https://medlineplus.gov/genetics/understanding/precisionmedicine/definition/>)

The Fetal Treatment Center at the University of California, San Francisco describes stem cell treatments for inherited diseases (<https://fetus.ucsf.edu/stem-cells/>).

ClinicalTrials.gov (<https://clinicaltrials.gov/>), a service of the National Institutes of Health, provides easy access to information on clinical trials. You can search for specific trials or browse by condition ([https://www.clinicaltrials.gov/ct2/search/browse?brwse=cond\\_alpha\\_all](https://www.clinicaltrials.gov/ct2/search/browse?brwse=cond_alpha_all)), trial sponsor ([https://www.clinicaltrials.gov/ct2/search/browse?brwse=spns\\_cat](https://www.clinicaltrials.gov/ct2/search/browse?brwse=spns_cat)), location ([https://www.clinicaltrials.gov/ct2/search/browse?brwse=locn\\_cat](https://www.clinicaltrials.gov/ct2/search/browse?brwse=locn_cat)), or treatment approach (for example, drug interventions ([https://www.clinicaltrials.gov/ct2/search/browse?brwse=intr\\_cat](https://www.clinicaltrials.gov/ct2/search/browse?brwse=intr_cat))).