



Genetics Home Reference

Your Guide to Understanding Genetic Conditions

Handbook

Help Me Understand Genetics

Genetic Consultation

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Chapter 5

Genetic Consultation

Table of Contents

What is a genetic consultation?	3
Why might someone have a genetic consultation?	4
What happens during a genetic consultation?	5
How can I find a genetics professional in my area?	6
How are genetic conditions diagnosed?	7
How are genetic conditions treated or managed?	10

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 disorders. 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 or over the telephone.

For more information about genetic consultations:

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

Additional background information is provided by the National Genome Research Institute in its Frequently Asked Questions About Genetic Counseling (<http://www.genome.gov/19016905>).

Information about genetic counseling, including the different types of counseling, is available from the National Society of Genetic Counselors in its booklet Making Sense of Your Genes: A Guide to Genetic Counseling (<http://www.nsgc.org/Portals/0/GuidetoGeneticCounseling.pdf>).

The Centre for Genetics Education also offers an introduction to genetic counseling (<http://www.genetics.edu.au/Information/Genetics-Fact-Sheets/Genetic-Counselling-FS3>).

The National Center for Biotechnology Information (NCBI) provides additional information about genetic consultations (<http://www.ncbi.nlm.nih.gov/projects/GeneTests/static/concepts/primer/primerwhatiscons.shtml>).

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, mental retardation, 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 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:

The National Center for Biotechnology Information (NCBI) provides a detailed list of common reasons for a genetic consultation (<http://www.ncbi.nlm.nih.gov/projects/GeneTests/static/concepts/primer/primerwhoshould.shtml>).

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

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 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 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.
- Advise a couple 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 Center for Biotechnology Information (NCBI) provides a detailed list of topics that are often discussed during a genetics consultation (<http://www.ncbi.nlm.nih.gov/projects/GeneTests/static/concepts/primer/primerwhatiscons.shtml>).

The National Society of Genetic Counselors offers information about what to expect from a genetic counseling session as part of its FAQs About Genetic Counselors and the NSGC (<http://www.nsgc.org/Home/ConsumerHomePage/PatientFAQs/tabid/338/Default.aspx#SEEAGC>).

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

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

- The National Center for Biotechnology Information (NCBI) provides a list of U.S. and international genetics clinics (<http://www.ncbi.nlm.nih.gov/sites/GeneTests/clinic?db=GeneTests>). Clinics can be chosen by state or country, by service, and/or by clinic name. State maps can help you locate a clinic in your area.
- The National Society of Genetic Counselors offers a searchable directory of genetic counselors in the United States (<http://www.nsgc.org/FindaGeneticCounselor/tabid/64/Default.aspx>). You can search by location, name, area of practice/specialization, and/or ZIP Code.
- The National Cancer Institute provides a Cancer Genetics Services Directory (<http://www.cancer.gov/cancertopics/genetics/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.

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 medical 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 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, 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:

Genetics Home Reference provides information about genetic testing (<http://ghr.nlm.nih.gov/handbook/testing>) and the importance of family medical history (<http://ghr.nlm.nih.gov/handbook/inheritance/familyhistory>). Additionally, links to information about the diagnosis of specific genetic disorders are available in each condition summary (<http://ghr.nlm.nih.gov/BrowseConditions>) under the heading “Where can I find information about diagnosis or management of...?”

Genetic Alliance provides an in-depth guide about genetic counseling called Making Sense of Your Genes (http://www.nsgc.org/client_files/GuidetoGeneticCounseling.pdf), which includes information about how genetics professionals diagnose many types of genetic disorders.

This article from Nature Education (<http://www.nature.com/scitable/topicpage/diagnosing-down-syndrome-cystic-fibrosis-tay-sachs-646>) discusses the diagnosis of several well-known genetic conditions.

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

Boston Children’s Hospital provides this brief overview of testing for genetic disorders (<http://www.childrenshospital.org/az/Site2884/mainpageS2884P3.html>).

The American College of Medical Genetics offers practice guidelines (http://www.acmg.net/AM/Template.cfm?Section=Practice_Guidelines&Template=/CM/HTMLDisplay.cfm&ContentID=7350), including diagnostic criteria, for several genetic disorders. These guidelines are designed for geneticists and other healthcare providers.

The Agency for Healthcare Research and Quality’s (AHRQ) National Guideline Clearinghouse has compiled screening, diagnosis, treatment, and management guidelines (<http://guideline.gov/browse/by-topic-detail.aspx?id=48661&ct=1>) for many genetic disorders.

GeneReviews (<http://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 diagnosis of specific genetic disorders as part of each peer-reviewed disease description.

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 compensate for the enzyme shortage. These treatments are used to manage existing signs and symptoms and may help prevent future complications.

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:

Links to information about the treatment of specific genetic disorders are available in each Genetics Home Reference condition summary (<http://ghr.nlm.nih.gov/BrowseConditions>) under the heading “Where can I find information about diagnosis or management of...?”

GeneReviews (<http://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 Agency for Healthcare Research and Quality's (AHRQ) National Guideline Clearinghouse has compiled screening, diagnosis, treatment, and management guidelines (<http://guideline.gov/browse/by-topic-detail.aspx?id=48661&ct=1>) for many genetic disorders.

Information related to the approaches discussed above is available from MedlinePlus:

- Inborn Errors of Metabolism (<http://www.nlm.nih.gov/medlineplus/ency/article/002438.htm>)
- Bone Marrow Transplantation (<http://www.nlm.nih.gov/medlineplus/bonemarrowtransplantation.html>)
- Palliative care (<http://www.nlm.nih.gov/medlineplus/palliativecare.html>) (also known as supportive care)

Genetics Home Reference offers consumer-friendly information about gene therapy (<http://ghr.nlm.nih.gov/handbook/therapy>), including safety, ethical issues, and availability.

ClinicalTrials.gov (<http://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 (http://www.clinicaltrials.gov/ct2/search/browse?brwse=cond_alpha_all), trial sponsor (http://www.clinicaltrials.gov/ct2/search/browse?brwse=spns_cat), location (http://www.clinicaltrials.gov/ct2/search/browse?brwse=locn_cat), or treatment approach (for example, drug interventions (http://www.clinicaltrials.gov/ct2/search/browse?brwse=intr_cat)).



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