



Genetics Home Reference

Your Guide to Understanding Genetic Conditions

Handbook

Help Me Understand Genetics

Genetic Consultation

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Chapter 5

Genetic Consultation

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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/19016939>).

An overview of genetic counseling (http://genome.wellcome.ac.uk/doc_WTD020928.html) is available from the Wellcome Trust.

The Centre for Genetics Education also offers an introduction to genetic counseling (<http://www.genetics.com.au/pdf/factsheets/fs03.pdf>).

GeneTests from the University of Washington provides additional information about genetic consultations (<http://www.genetests.org/servlet/access?id=8888891&key=Bjk2m9R0Ueq3H&fcn=y&fw=CpPj&filename=/concepts/primer/primerwhatiscons.html>).

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:

GeneTests from the University of Washington provides a detailed list of common reasons for a genetic consultation (<http://www.genetests.org/servlet/access?id=8888891&key=Bjk2m9R0Ueq3H&fcn=y&fw=Lcuu&filename=/concepts/primer/primerwhoshould.html>).

An overview of indications for a genetics referral (http://www.geneticalliance.org/ksc_assets/pdfs/manual/chapter_6.pdf) 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:

GeneTests from the University of Washington provides a detailed list of topics that are often discussed during a genetics consultation (<http://www.genetests.org/servlet/access?id=8888891&key=mzpjVQaID3TY6&fcn=y&fw=jAzO&filename=/concepts/primer/primerwhatiscons.html#whathappens>).

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/consumer/faq_consumers.cfm). Scroll down to question 5, "This is my first visit to a genetic counselor. What can I expect from this visit?"

Information about the role of genetic counselors and the process of genetic counseling (http://www.geneticalliance.org/ksc_assets/pdfs/manual/chapter_5.pdf) 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:

- GeneTests from the University of Washington provides a list of genetics clinics around the United States (<http://www.genetests.org/servlet/access?id=8888891&key=vDM247XLm0tl8&fcn=y&fw=GJBG&filename=/clinicsearch/searchclinic.html>) and international genetics clinics (http://www.genetests.org/servlet/access?submit=International&id=8888891&db=genestar&fcn=clinicsearch_intl&key=vDM247XLm0tl8). You can also access the list by clicking on “Clinic Directory” at the top of the GeneTests home page. Clinics can be chosen by state or country, by service, and/or by specialty. 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/resource/link.cfm>). 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://cancer.gov/search/genetics_services/), which lists professionals who provide services related to cancer genetics. You can search by type of cancer or syndrome, location, and/or provider name.



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