

healthsmart!

Genetic Counseling and Testing: What You Need to Know

Genetic Counseling

Genetic testing can be a powerful tool in assessing your overall health. The results will allow you to take charge of your health and start making important decisions. However, the process can be overwhelming and not everyone wants to know that he or she is at risk for a disease. That is why the first step is to see a genetic counselor or geneticist. Your primary care physician may refer you to a genetic counselor or a geneticist, or you may refer yourself.

Genetic counselors have specialized graduate degrees and experience in medical genetics and counseling. A geneticist is a medical doctor who has completed a residency or fellowship in clinical genetics. Both counselors and geneticists are experts trained in assessing your family's health history and helping you decide whether to take a genetic test. Counselors and geneticists will help you weigh the advantages and disadvantages of testing, but they will not make your decision for you or be directive in what they say.

Questions you may want to ask your genetic counselor or geneticist include:

- What will the test results mean for my overall health?
- Could my children or siblings have this condition?
- What special care does a person with this condition need?
- Will the condition eventually improve?

- What causes this condition?
- How accurate are the tests for this condition?
- How much will the test cost?
- Are there any clinical trials that may benefit me?
- Are there any support groups for this condition?
- Where can I go for a second opinion?
- Will results of my test affect my job or ability to be insured for health care?

Getting started

The counseling and testing process varies for adults, children and expectant mothers undergoing prenatal testing. The first step, however, is always to obtain a referral to a genetic counselor from your family practitioner. Before administering the test, your counselor will review informed consent policies and other information about the genetic test with you. At that point the counselor will refer you to a geneticist, the doctor who will administer the test.

After counseling, if you have decided you want to move forward with testing, your geneticist will schedule the testing

Genetic Testing

What does a genetic test involve?

By analyzing your DNA, RNA, chromosomes and proteins, genetic testing can usually detect whether you have a specific disorder. Genetic testing allows doctors to pinpoint missing or abnormal genes. Often these missing or abnormal genes can result in genetic disorders. Abnormalities may occur spontaneously,

be inherited from your parents or result from exposure to radiation or toxic chemicals. Everyone, regardless of age or sex, is eligible for genetic testing.

Test results

Genetic testing cannot tell if you will definitely develop a disease, it can only determine whether you are a carrier for a particular disease. Being a carrier does not necessarily mean the symptoms will show up. Also, positive test results do not predict the severity of the disease. Symptoms for most genetic disorders can range from mild to critical (severe).

Even if you have been tested in the past, it may be beneficial to retest for a disorder. Because of the many advances in genetic testing, tests performed even as early as two years ago may not be as accurate as today's tests. Ask for your primary physician or geneticist/genetic counselor's recommendation.

Advantages of genetic testing

- Genetic testing will allow you to make choices about your health.
- Learning that you do not have a genetic disorder will provide peace of mind.
- Learning that you do have a genetic disorder will allow your physician to begin potential treatment as soon as possible if you wish.
- You can take other steps to protect your health.
- The information can be used to plan aspects of your future such as your career and your family.

Disadvantages of genetic testing

- The test can be expensive and may not be covered by insurance.
- Genes are only one part of the puzzle—many cancers and disorders are caused by other factors too. A negative result does not mean you definitely will not develop the disease.
- A positive result will not predict the severity of the disorder. It also does not mean you will develop the disease. Genetic tests only show whether you have a mutation that has been shown to be associated with the disease. The probability of developing the disease may be high or low, depending on the specific abnormality.

- Some tests fail to detect all of the mutations that can cause a disease.
- There is the possibility that information about your genetic tests may be acquired by your insurance company and employer and used to deny you health coverage or employment.

Prenatal genetic testing

Genetic testing often surfaces as a topic during pregnancy. For example, serum screening is a type of prenatal test that uses a blood sample from the mother to identify the unborn child's potential for developing disorders such as Down syndrome. The screening is available to any woman between 15 and 21 weeks of pregnancy. Amniocentesis is another common prenatal test, usually performed between 16 and 18 weeks of pregnancy. During amniocentesis, amniotic fluid from around the fetus is removed and examined for genetic disorders such as Down syndrome and cystic fibrosis. Discussion of prenatal genetic testing should begin with your obstetrician.

Newborn screenings

As of Dec 6, 2005, 29 newborn tests are mandated in Kentucky. For instance, phenylketonuria (PKU) is a test done in all 50 states to make sure newborns can process a substance called phenylalanine. Phenylalanine is found in almost all food and can build up in the bloodstream, causing brain damage if not processed. Tests such as these are done because treatments are currently available. If tests are positive, you and your doctor are notified. Tests may be redone or new test ordered. The Kentucky newborn screening program has contracts with UK HealthCare and other medical centers to provide special consultations if your child has one of the disorders.

An emerging trend

Home genetic testing kits are growing in popularity. Sold on the Internet and in retail stores, the tests require users to send a sample of their genetic material to a lab for analysis. However, according to a report by the General Accountability Office, the investigating arm of Congress, these test kits offer almost no useful guidance and are often misleading. Other concerns include:

- Quality control. Are the labs testing the materials reliable? What protocols are they following?
- Who interprets the results? How are the results explained to the consumer?

Genetic testing in children

Children are also candidates for genetic testing. In many cases, children with previously undiagnosed learning problems and delayed development are tested to confirm a disorder. However, learning the results of a test may be a highly emotional or confusing experience for the parents and/or child. Your counselor may advise against performing the test. When considering a genetic test on a child, it is helpful for parents or caregivers to ask:

- Is it in the child's best interest to have the test?
- Is it possible to delay the test until the child is mature enough to decide whether he or she wants it?

Genetic testing in adults

Often, adults will be tested for the following purposes:

- Diagnostic testing – Your physician can confirm or rule out a suspected diagnosis. Often, a genetic test may be conducted for unexplained symptoms. The results will help your physician develop a plan for your care.
- Carrier testing – Healthy individuals can be tested to see whether they are at risk for passing the disease on to their children. For example, carrier tests are often offered to couples who have a family history of cystic fibrosis and are planning on having a child.
- Presymptomatic testing – Presymptomatic tests are offered to healthy patients who may be at a high risk for future problems due to a strong family history of a disorder. They help anticipate problems later in life. The tests predict the likelihood of developing a genetic disorder in adulthood, such as Huntington's disease. While cancers (especially breast, ovarian and colon) and heart disease are complex and have both genetic and environmental causes, genetic testing can indicate a predisposition for these diseases.

What are common genetic disorders?

Over 1,300 gene tests are currently available. Genetic disorders frequently tested include:

- Cystic fibrosis – Cystic fibrosis is tested by examining 32 different mutations in a particular gene. The disorder affects the normal movement of salt in and out of cells that line the lungs and pancreas. This produces a thick, sticky mucus which leads to frequent lung infections.

- Down syndrome – Down syndrome is the most common genetic birth defect, often resulting in multiple abnormalities. It is caused by an extra chromosome and results in some degree of mental retardation, heart defects and problems with vision and hearing. Down syndrome is tested for by taking a blood sample and performing a chromosome analysis.
- Neurofibromatosis I – Neurofibromatosis I is caused by mutations in the NF1 gene. This gene makes a protein called neurofibromin, which is responsible for keeping cells from growing and dividing rapidly. Mutations in the NF1 gene cause tumors to form on nerves throughout the body.
- Sickle cell disease – Sickle cell disease is caused by a red blood cell disorder. It is characterized by pain episodes, serious infections and damage to vital organs. Hemoglobin electrophoresis, a test that measures the different types of hemoglobin (hemoglobins are protein-iron compounds in the blood that carry oxygen to the cells and carbon dioxide away from the cells to the lungs) in the blood, is used to diagnose the disease. Sickle cell primarily affects African-Americans and people of Arabian, Asian, Caribbean, Indian, Mediterranean, and South and Central American descent.
- Tay-Sachs disease – Tay-Sachs disease is caused by the absence of a vital enzyme called hexosaminidase-A (Hex-A). Without Hex-A, a fatty substance builds up in cells, especially in the brain's nerve cells. This ongoing buildup causes progressive cell damage. Tests for the disease measure the amount of Hex-A in the blood or in white blood cells.

In the next five years, the following genetic tests are likely to be developed and reach the market:

- Expansion and refinement of currently available tests (e.g., for cystic fibrosis or Fragile X Syndrome)
- Earlier, less invasive pregnancy screening through PAP techniques. This technique involves taking cells from the fetus, genetically identifying them and screening for abnormalities such as Down syndrome and cystic fibrosis.
- Maturity-onset diabetes in the young (a rare form of diabetes type 2 in children)
- Familial Alzheimer's disease (early-onset)
- Parkinson's disease (early-onset)

Source: *The Future of Genetic Testing - Health Tech*

Insurance and genetic testing

The degree to which genetic testing is covered by insurance varies with each individual company and state. Test prices can range from about \$60 up to \$3,000. Although the test itself may be expensive, the results might allow you to avoid other costly tests to confirm a diagnosis.

Genetic testing at UK HealthCare

UK HealthCare has two geneticists and one genetic counselor on staff. They can identify clinical trials that could potentially provide a way to treat or manage genetic disorders. Call 859-257-1000 or 1-800-333-8874 to schedule an appointment.

UK HealthCare shares its genetic counseling expertise across eastern Kentucky. Outreach clinics meet in the following county health departments:

- Ashland – call **606-329-9444** ext. **2236**, meets six times per year
- Corbin – call **606-528-5613**, meets six times per year
- Somerset – call **606-679-4416**, meets six times per year
- Pineville – call **606-248-7170**, meets twice per year
- Paintsville – call **606-789-2590**, meets four times per year
- Pikeville – call **606-437-5500**, ext. **328**, meets six times per year
- Whitesburg – call **606-633-2945**, meets twice per year

Resources

Locate a genetic counselor – National Society of Genetic Counselors

www.nsgc.org

What to expect from a genetics consultation – Mountain States Genetics Regional Collaborative Center

www.mostgene.org/dir/expect.htm

What is genetic testing? – National Institutes of Health

www.accessexcellence.org/AE/AEPC/NIH/

An introduction to genetics and genetic testing – KidsHealth

www.kidshealth.org/parent/system/medical/genetics.html

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Call UK Health Connection at **(859) 257-1000** or toll free **(800) 333-8874** to make an appointment or request a referral. Visit us online at **www.ukhealthcare.uky.edu**.

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