Facts About Genetic Testing

What is a genetic test?

A genetic test determines the DNA sequence of a certain region of the human genome. This region could be a whole gene, a portion of a gene, or other areas thought to regulate genes. The test will look for certain changes in the sequence that are known to have consequences on the function of a gene. These tests can be used to 1) diagnose a disease or other trait, 2) determine if a person is a carrier of a mutation that could lead to disease in their children, and 3) predict if a disease or trait that is not yet detectable by other medical tests may occur in the future.

How are the tests performed?

Most genetic tests are performed on a DNA sample. Because every tissue in the body is made of cells that contain DNA, any tissue can be used as a source of DNA. However, blood is the most common source because it can easily be obtained in large quantities; typically 5-10 ml (1-2 teaspoons) are taken for a test. Some laboratories will allow cheek cells to be submitted for genetic testing. In this case, the cells are usually collected by rubbing the inside of the mouth with a small brush.

After the DNA is obtained from a blood or other tissue sample, a variety of different methods can be used to look for mutations in your genes. Which method is used often depends on the size of the gene and the types of mutations that are typically found in that gene. Sometimes the whole coding sequence of the gene is examined. This is similar to reading a page in a book to look for spelling errors in all of the words. Other times, methods are used that only look for the most common mutations. In this case, the test is similar to looking at a page only for a specific misspelled word and ignoring all of the other words. This latter approach is often taken if the gene's role in disease is mostly due to only a small number of known mutations. Some laboratories may combine these methods by first screening for common mutations and then, in certain circumstances, examining the whole coding sequence of the gene.

Why should I have genetic testing?

Genetic testing can help identify the cause of the hearing loss and may help predict whether it will worsen. It can also help with treatment decisions because the successful use of hearing aids and/or cochlear implants may depend on the cause of the hearing loss. In addition, if a genetic test is positive it can reduce the need to perform the many other clinical tests that are used to find other causes of the hearing loss. Furthermore, if a genetic test for nonsyndromic hearing loss is positive, it can assure the family that no other problems associated with syndromic forms of hearing loss will develop. Testing can also help predict the likelihood that future children or other persons in the family will develop hearing loss.

Who can help me decide if I should have a genetic test?

Often your primary care physician, pediatrician, or ear specialist (otolaryngologist or audiologist) can help you decide if having a genetic test is appropriate. However, the field of genetics is developing so quickly that it is often useful to go to a genetics specialist, such as a genetic counselor, clinical geneticist, or genetic testing lab to get the most up-to-date information about genetic testing for hearing loss.

Where do I go for genetic testing?

There are many places that offer genetic testing for hearing loss. Your doctor will assist you in finding a place. If your doctor is unfamiliar with available genetic tests for hearing loss, he or she can visit the GeneTests website (www.genetests.org) and search the Laboratory Directory for "deafness" or the gene name (e.g. "connexin 26'). Many of the locations that perform genetic testing have voluntarily registered at this site. If none of these sites are nearby, your doctor can often mail your blood sample to one of these sites.

How much does a genetic test cost and how long will it take to get results?

The cost and turn-around-time of a genetic test may vary depending on the lab and the methods used for testing. A typical range of cost might be $300-$2000 and a typical range of time to get the results might be 2-6 weeks. If the test only screens for common mutations or if the gene that is being tested is small (like connexin 26) the test may be less expensive and the results may be obtained more quickly. However, if the gene is large and the test examines the entire coding sequence of the gene, it may be more expensive and may take longer to perform. Insurance companies will often pay for genetic tests, but you should check with your company before your doctor orders the test.

Can the results of genetic tests harm me?

Genetic information has been used in the past for discriminatory purposes; however, most states now have laws that prevent employers and insurance companies from discriminating against people on the basis of their genetic makeup. You can find out if such laws exist in your state by going to http://www.genome.gov/10001621.