

DNA Testing for Neurofibromatosis 2

Genes and DNA Testing

Genes are the inherited material in a cell that determines how the cell will function. Small changes in genes can lead to different disorders and diseases throughout the body. Recent developments have now identified the genes and changes responsible for certain conditions. It is now possible to test individuals at risk for certain disorders to see if they carry these altered genes. Currently, genetic testing is available for NF2.

What is Neurofibromatosis Type 2?

Neurofibromatosis 2, or NF2, is a genetic disorder primarily affecting the nervous system. People with NF2 develop tumors affecting hearing and balance, called vestibular schwannomas (previously known as acoustic neuromas). Other tumors may develop in the brain and along the spinal cord or peripheral nerves. While the location of these tumors often causes problems, they usually are *benign*.

NF2 is caused by a change in a gene on chromosome 22. Half the persons with NF2 are born to unaffected parents. The other half are born to a parent who also has NF2. NF2 is a fully penetrant disease. Everyone who carries a change in the gene will develop vestibular schwannomas, usually by the age of 22.

In the past, people with NF2 were diagnosed in the teenage years. With improved imaging technology and more awareness of the disorder, an increasing number of young people below the age of 12 are being diagnosed. Specialized MRI scans that can visualize through the bony canal to the ear are needed when NF2 is suspected. Individuals benefit from advances in earlier detection and treatment of the disorder. Genetic testing now offers a potential means of earlier diagnosis in children of persons with NF2.

What can a genetic test tell me?

A genetic test can tell if the child of a person with NF2 also has NF2. Although NF2 patients do not usually have symptoms until adulthood, if children are detected *before* onset of symptoms, more treatment options are available for vestibular tumors. Genetic testing can potentially save money, reduce the burden of repeated clinical exams and definitely diagnose children with NF2, *before* the onset of symptoms. Currently, using SSCP and DNA sequencing, genetic changes can be detected in 60 to 66% of persons with bilateral vestibular schwannomas. If an individual is found to have a genetic alteration, family members can then be screened for the same change. In families with two or more persons affected with NF2, linkage analysis may be more appropriate than looking for alterations in the gene itself.

If no change in the NF2 gene is detected, *this does not guarantee that you do not have NF2*. If an experienced NF physician has already clinically diagnosed you, the results of a gene test will not change this diagnosis or tell you how severe the symptoms may be. Once you decide to have a gene test for NF2, your physician needs to arrange for a blood or tumor sample to be sent to one of the laboratories offering testing services, along with a detailed family medical history. The detection rate may be higher if a tumor sample is used. Tumors can be retrieved from hospital archives, even if surgery was performed years ago. Tumor sample from deceased individuals can also be used.

Preimplantation diagnosis and prenatal genetic testing are available to those individuals at risk for having a child with NF2 in whom a genetic alteration has been found.

How do I find out more about genetic testing?

To find out more about gene testing for NF2 contact your physician. Your physician may refer you to a genetic counselor to discuss more about testing and its limitations. You, your physician and counselor can contact the following laboratories offering NF2 testing in the United States.

Medical Genomics Laboratory
720 20th Street S
KAUL Building-Suite 330
Birmingham, AL 35294
205-934-5562
Email: mgl@genetics.uab.edu

Athena Diagnostics, Inc.
Worcester, Massachusetts
800-394-4493 ext. 3021

Massachusetts General Hospital
Neurogenetics DNA
Diagnostic Laboratory
Charlestown, Massachusetts
617-726-5721
617-724-6057

Things to consider...

Genetic testing is not for everyone. Having a test is a very private decision. While the test may be very useful, it does have its limitations and complications. The results of the genetic test can not tell you how severe NF2 will be. A negative test does not mean that you do not have NF2. Furthermore, testing cannot change a clinical diagnosis of NF2.

In order to make your decision, you may want to discuss with your physician if the test is medically necessary and if it will change the course of treatment in any way. Make sure you are psychologically prepared for the results of testing. Your physician and genetic counselor can discuss with you in detail the testing and possible impact on your life. Neither lab offering testing accepts insurance and the test is costly. Keep in mind that NF2 is considered a “pre-existing” condition and may influence health, life and disability insurance coverage. Remember to take the time to make the best decision for you about genetic testing.

For more information on NF2, several organization provide additional assistance

Neurofibromatosis, Inc.
P.O. Box 66884
Chicago, IL 60666
Phone: 630-627-1115
Fax: 630-627-1117
Email: nfinfo@nfinc.org
Website: www.nfinc.org

Acoustic Neuroma Association
600 Peachtree Pkwy, Suite 108
Cumming, Georgia 30041-6899
Phone: 770-205-8211
Fax: 770-205-0239
Email: ANUSA@aol.com
<http://anusa.org>

Association of Late Onset Deafened Adults (ALDA)
P.O. Box 641763
Chicago, IL 60664-17639
TTY: 708-445-0860

House Ear Institute
2100 West Third Street
Los Angeles, CA 90057
Phone: 213-483-4431 Fax: 213-483-8789
TDD: 213-484-2642
Email: webmaster@hei.org Website: www.hei.org