

**Synonym(s):** Von Recklinghausen's Disease  
Condensed from [Neurofibromatosis Fact Sheet](#)

**Table of Contents (click to jump to sections)**

[What is Neurofibromatosis?](#)

[Is there any treatment?](#)

[What is the prognosis?](#)

[What research is being done?](#)

[Organizations](#)

[Related NINDS Publications and Information](#)

[Publicaciones en Español](#)

[Additional resources from MEDLINEplus](#)

### **What is Neurofibromatosis?**

The neurofibromatoses are genetic disorders of the nervous system that primarily affect the development and growth of neural (nerve) cell tissues. These disorders cause tumors to grow on nerves and produce other abnormalities such as skin changes and bone deformities. Although many affected persons inherit the disorder, between 30 and 50 percent of new cases arise spontaneously through mutation (change) in an individual's genes. Once this change has taken place, the mutant gene can be passed on to succeeding generations. Scientists have classified the disorders as neurofibromatosis type 1 (NF1) and neurofibromatosis type 2 (NF2). NF1 is the more common type of the neurofibromatoses. In diagnosing NF1, a physician looks for changes in skin appearance, tumors, or bone abnormalities, and/or a parent, sibling, or child with NF1. Symptoms of NF1, particularly those on the skin, are often evident at birth or during infancy and almost always by the time a child is about 10 years old. NF2 is less common. NF2 is characterized by bilateral (occurring on both sides of the body) tumors on the eighth cranial nerve. The tumors cause pressure damage to neighboring nerves. To determine whether an individual has NF2, a physician looks for bilateral eighth nerve tumors and similar signs and symptoms in a parent, sibling, or child. Affected individuals may notice hearing loss as early as the teen years. Other early symptoms may include tinnitus (ringing noise in the ear) and poor balance. Headache, facial pain, or facial numbness, caused by pressure from the tumors, may also occur.

### **Is there any treatment?**

Treatments for both NF1 and NF2 are presently aimed at controlling symptoms. Surgery can help some NF1 bone malformations and remove painful or disfiguring tumors; however, there is a chance that the tumors may grow back and in greater numbers. In the rare instances when tumors become malignant (3 to 5 percent of all cases), treatment may include surgery, radiation, or chemotherapy. For NF2, improved diagnostic technologies, such as MRI, can reveal tumors as small as a few millimeters in diameter, thus allowing early treatment. Surgery to remove tumors completely is one option but may result in hearing loss. Other options include partial removal of tumors, radiation, and if the tumors are not progressing rapidly, the conservative approach of watchful waiting. Genetic testing is available for families with documented cases of NF1 and NF2. New (spontaneous) mutations cannot be confirmed genetically. Prenatal diagnosis of familial NF1 or NF2 is also possible utilizing amniocentesis or chorionic villus sampling procedures.

### **What is the prognosis?**

In most cases, symptoms of NF1 are mild, and patients live normal and productive lives. In some cases, however, NF1 can be severely debilitating. In some cases of NF2, the damage to nearby vital structures, such as other cranial nerves and the brainstem, can be life-threatening.

### **What research is being done?**

Several years ago, research teams located the exact position of the NF1 gene on chromosome 17. The product of the NF1 gene is a large and complex protein called neurofibromin. One portion of this protein is similar to a family of proteins called GAP (guanosine triphosphatase-activating protein). Scientists have demonstrated that GAP proteins play a significant role in tumor suppression in certain cancers. The similarity of the NF1 protein to GAP proteins suggests that the NF1 protein may have a similar switching role in the development of neurofibromas. Scientists theorize that defects in the gene may lessen or inhibit the normal output of its protein and allow the irregular cell growth that may lead to tumor development. Intensive efforts have led to the identification of the NF2 gene on chromosome 22. The NF2 gene product is a tumor suppressor protein. Basic studies in molecular genetics may lead one day to nonsurgical or pharmacologic treatments aimed at retarding or suppressing tumors associated with the neurofibromatoses. The Interinstitute Medical Genetics Research Program at the NIH Clinical Center conducts NF2 family history research. Using specimens from some of the families, scientists have isolated and sequenced the NF2 gene and have described two different patterns of clinical features in NF2 patients. Investigators are continuing to study these patterns to see if they correspond to specific types of gene mutations.

[Select this link](#) to view a list of studies currently seeking patients.

## Organizations

### Children's Tumor Foundation

95 Pine Street  
16th Floor  
New York, NY 10005  
[info@ctf.org](mailto:info@ctf.org)  
<http://www.ctf.org>  
Tel: 800-323-7938 212-344-6633  
Fax: 212-747-0004

### National Cancer Institute (NCI)

National Institutes of Health, DHHS  
6116 Executive Boulevard, Ste. 3036A, MSC 8322  
Bethesda, MD 20892-8322  
[cancergovstaff@mail.nih.gov](mailto:cancergovstaff@mail.nih.gov)  
<http://cancer.gov>  
Tel: 800-4-CANCER (422-6237) 800-332-8615 (TTY)

### Neurofibromatosis, Inc. (NF Inc.)

P.O. Box 18246  
Minneapolis, MN 55418  
[nfinfo@nfinc.org](mailto:nfinfo@nfinc.org)  
<http://www.nfinc.org>  
Tel: 301-918-4600 800-942-6825

### Acoustic Neuroma Association

600 Peachtree Parkway  
Suite 108  
Cumming, GA 30041  
[info@anausa.org](mailto:info@anausa.org)  
<http://www.anausa.org>  
Tel: 770-205-8211 877-200-8211  
Fax: 770-205-0239/877-202-0239

### International RadioSurgery Association

3002 N. Second Street  
Harrisburg, PA 17110  
[office1@irsa.org](mailto:office1@irsa.org)  
<http://www.irsa.org>  
Tel: 717-260-9808  
Fax: 717-260-9809

## Related NINDS Publications and Information

- [Neurofibromatosis Fact Sheet](#)

Neurofibromatosis fact sheet compiled by the National Institute of Neurological Disorders and Stroke (NINDS).

- [NEUROBIOLOGY OF DISEASE IN CHILDREN: Neurofibromatosis](#)

NEUROBIOLOGY OF DISEASE IN CHILDREN

- [Defining the Future of Neurofibromatosis Research](#)

Summary of an NINDS workshop, "Defining the Future of Neurofibromatosis Research," May 4-5, 2000.

## Publicaciones en Español

- [Las Neurofibromatosis](#)

Prepared by:

Office of Communications and Public Liaison  
National Institute of Neurological Disorders and Stroke  
National Institutes of Health  
Bethesda, MD 20892

NINDS health-related material is provided for information purposes only and does not necessarily represent endorsement by or an official position of the National Institute of Neurological Disorders and Stroke or any other Federal agency. Advice on the treatment or care of an individual patient should be obtained through consultation with a physician who has examined that patient or is familiar with that patient's medical history.

All NINDS-prepared information is in the public domain and may be freely copied. Credit to the NINDS or the NIH is appreciated.

Last updated December 13, 2007