



Neurofibromatosis

Neurofibromatosis is a genetic disorder that can affect the spinal cord.

Q: What is Neurofibromatosis?

Neurofibromatosis (NF) refers to a group of genetic conditions in which tumors grow on nerves and on or underneath the skin. The tumors are typically non-cancerous.

Q: What causes Neurofibromatosis?

NF can be caused by an abnormal gene inherited from a parent or a new genetic alteration occurring at conception.

Q: What are the different types of Neurofibromatosis?

- **Neurofibromatosis type 1 (NF1)**, also known as von Recklinghausen disease, is the most common type of NF, affecting about 1 in 3,000 people. Children with NF1 are usually diagnosed by the age of 10.
- **Schwannomatosis (SWN)**, including **NF2-related Schwannomatosis**, is a rare type of neurofibromatosis. NF2-related Schwannomatosis is the most common type of Schwannomatosis and affects about 1 in 25,000 people. Symptoms can emerge at any time but typically appear during the teenage years and up until the age of 30.

Q: What are the symptoms of NF1?

Symptoms of NF1 may include:

- Flat, light brown spots on the skin (also known as “café au lait” spots)
- Freckles in the underarm or groin
- Soft bumps (fibromas) that grow on nerves on or under the skin
- Large bumps (plexiform neurofibromas) that grow on multiple nerves at once
- Small (and harmless) growths on the iris

About 15 – 20 % of children with NF1 will also develop tumors on their optic pathway. In some cases, these tumors will not need any treatment; when vision is affected, they might be treated with chemotherapy. Children living with NF1 may also have some learning challenges, ADHD or speech delays.

Q: How is NF1 diagnosed?

Doctors will check for skin changes, including visible growths, unusual freckles and café au lait spots; perform imaging tests to identify tumors; and assess any vision and/or neurological issues. Sometimes genetic testing may be done to confirm a diagnosis. To receive a diagnosis of NF1, a child must have two or more of the following symptoms:

- A minimum of six café au lait spots on the skin
- Freckles in the underarm or groin area
- One or more small bumps growing on or under the skin and/or one or more large bumps growing on or under the skin
- A tumor in the cells around the optic nerve
- Tiny bumps in the iris
- Abnormalities in the eye's blood vessels
- Severe scoliosis or other bone abnormalities
- An NF1 gene mutation or a parent with the mutation

Q: What are long-term health issues that may be caused by NF1?

Complications of NF1 include scoliosis, hypertension, congenital heart defects, hardening of the arteries that lead to the kidneys, narrowing of blood vessels to the brain and diminished visual spatial abilities. Women with NF1 have a higher risk of developing breast cancer before the age of 50.

Q: Can NF1 tumors become cancerous?

Most NF1 tumors are benign, but individuals with NF1 may develop malignant peripheral nerve sheath tumors (MPNST). Because this tumor grows and spreads quickly, it is important to identify and diagnose in its early stages. NF1 has also been associated with a higher risk of developing other cancers, including breast cancer, sarcomas and gastrointestinal tumors. Plexiform neurofibromas, which typically affect the spine, can also turn into MPNST.

Q: What are the symptoms of NF2-related Schwannomatosis?

Slow growing tumors located on the nerves that affect hearing and balance and along and inside the spinal cord are symptoms of NF2-related Schwannomatosis. Complications vary but may include:

- Hearing loss or ringing in the ears
- Problems with balance
- Weakness in facial muscles
- Difficulty swallowing
- Vision problems, including cataracts
- Weakened arm and leg muscles on both sides of the body

Q: How is NF2-related Schwannomatosis diagnosed?

Doctors will perform a physical exam, neurologic assessment and imaging tests to identify and map tumor growth. To receive an NF2-SWN diagnosis, there must be tumors growing on both sides of the body on nerves that carry signals of balance and hearing and/or tumors growing inside the spinal cord and/or in the membranes that surround it.

Q: Can NF2-related Schwannomatosis tumors turn into cancer?

NF2-related Schwannomatosis tumors only rarely become cancerous.

Q: What is the treatment for Neurofibromatosis?

There is no cure for Neurofibromatosis, but symptoms can be managed. Regular physicals and eye exams will be important for children diagnosed with NF1; in addition, possible learning difficulties should be closely monitored. Surgery may be needed to remove tumors that are causing pressure or pain. To treat NF2-related Schwannomatosis, doctors will manage pain and neurological problems, including vision problems, hearing loss and weakened gait. Surgery or radiation may be needed in order to mitigate and stem hearing loss.

Q: Which doctors will I need to see to treat Neurofibromatosis?

Diagnosing and managing symptoms of both NF1 and NF2-related Schwannomatosis will require a multidisciplinary healthcare team comprised of geneticists, neurologists, medical oncologists, neuro-oncologists and ophthalmologists. Some hospitals, including Boston Children's and the Cleveland Clinic, have units dedicated to neurofibromatosis. Check your region's teaching and larger research hospitals to see if they offer specialty clinics for children or adults.

Q: What other resources are available for families affected by Neurofibromatosis?

The Neurofibromatosis Network, a non-profit organization founded in 1988, advocates

for NF research, offers educational webinars and helps families connect with other community members. Visit its website for at <https://www.nfnetwork.org/> for more information.

Q: Are there clinical trials specific to neurofibromatosis?

Yes. Clinical trials related to neurofibromatosis may be found through a database maintained by the U.S. National Library of Medicine at www.ClinicalTrials.gov.

In addition, the Neurofibromatosis Consortium was established by the Department of Defense Neurofibromatosis Research Program to pursue clinical trials for the treatment of neurofibromatosis (NF) complications in children and adults. For more information visit its website at <http://www.uab.edu/nfconsortium>.

Sources: National Institutes of Neurological Disorders and Stroke, Memorial Sloan Kettering Hospital, Mayo Clinic, Seattle Children's, American Cancer Society, National Library of Medicine, University of Alabama at Birmingham Heersink School of Medicine.

Need to talk to someone?

Our Information Specialists are available to answer your questions.

Call toll-free 1-800-539-7309 Mon-Fri, 7 am - 8 pm EST.

Or schedule a call or ask a question online at

<https://www.ChristopherReeve.org/Ask>.

Resources for Neurofibromatosis:

Children's Tumor Foundation

<https://www.uab.edu/nfconsortium/>

697 Third Avenue, Suite 418

New York, NY 10017

Phone: 212-344-6633

The Children's Tumor Foundation, established in 1978, funds research to treat and cure neurofibromatosis.

National Institutes of Neurological Disorders and Stroke: Neurofibromatosis

<https://www.ninds.nih.gov/health-information/disorders/neurofibromatosis?search-term=neurofibromatosis>

Neurofibromatosis Consortium

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