Neurofibromatosis

Overview

By Mayo Clinic Staff

Neurofibromatosis is a genetic disorder that causes tumors to form on nerve tissue. These tumors can develop anywhere in your nervous system, including your brain, spinal cord and nerves. Neurofibromatosis is usually diagnosed in childhood or early adulthood.

The tumors are usually noncancerous (benign), but sometimes can become cancerous (malignant). Symptoms are often mild. However, complications of neurofibromatosis can include hearing loss, learning impairment, heart and blood vessel (cardiovascular) problems, loss of vision, and severe pain.

Neurofibromatosis treatment aims to maximize healthy growth and development and to manage complications as soon as they arise. When neurofibromatosis causes large tumors or tumors that press on a nerve, surgery can help ease symptoms. Some people may benefit from other therapies, such as stereotactic radiosurgery or medications to control pain.

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Neurofibromatosis

Symptoms and causes

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Symptoms

There are three types of neurofibromatosis, each with different signs and symptoms.

Neurofibromatosis 1

Neurofibromatosis 1 (NF1) usually appears in childhood. Signs are often evident at birth or shortly afterward, and almost always by age 10. Signs and symptoms are often mild to moderate, but can vary in severity.

Signs and symptoms include:

- **Flat, light brown spots on the skin (cafe au lait spots).** These harmless spots are common in many people. Having more than six cafe au lait spots is a strong indication of NF1. They are usually present at birth or appear during the first years of life and then stabilize.

- **Freckling in the armpits or groin area.** Freckling

usually appears by ages 3 to 5. Freckles are smaller than cafe au lait spots and tend to occur in clusters in skin folds.

- **Tiny bumps on the iris of your eye (Lisch nodules).** These harmless nodules can't easily be seen and don't affect your vision.

- **Soft bumps on or under the skin (neurofibromas).** These benign tumors usually develop in or under the skin, but can also grow inside of the body. Sometimes, a growth will involve multiple nerves (plexiform neurofibroma).

- **Bone deformities.** Abnormal bone growth and a deficiency in bone mineral density can cause bone deformities such as a curved spine (scoliosis) or bowed lower leg.

- **Tumor on the optic nerve (optic glioma).** These tumors usually appear by age 3, rarely in late childhood and adolescence, and almost never in adults.

- **Learning disabilities.** Impaired thinking skills are common in children with NF1, but are usually mild. Often there is a specific learning disability, such as problem with reading or mathematics. Attention-deficit/hyperactivity disorder (ADHD) is also common.

- **Larger than average head size.** Children with NF1 tend to have a larger than average head size due to increased brain volume.

- **Short stature.** Children with NF1 often are below average in height.

Neurofibromatosis 2

Neurofibromatosis 2 (NF2) is much less common than NF1. Signs and symptoms of NF2 usually result from the development of benign, slow-growing tumors (acoustic neuromas) in both ears. Also known as vestibular schwannomas, these tumors grow on the nerve that carries sound and balance information from the inner ear to the brain.

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Diagnosis

Signs and symptoms generally appear in the late teen and early adult years, and can vary in severity. Signs and symptoms can include:

- Gradual hearing loss
- Ringing in the ears
- Poor balance
- Headaches

Sometimes NF2 can lead to the growth of schwannomas in other nerves of the body, including the cranial, spinal, visual (optic) and peripheral nerves. Signs and symptoms of these schwannomas can include:

- Numbness and weakness in the arms or legs
- Pain
- Balance difficulties
- Facial drop
- Vision problems or the development of cataracts

Schwannomatosis

This rare type of neurofibromatosis usually affects people after the age of 20. Schwannomatosis causes tumors to develop on skull (cranial), spinal and peripheral nerves — but not on the nerve that carries sound and balance information from the inner ear to the brain. Because tumors don't usually grow on both hearing nerves, schwannomatosis doesn't cause the hearing loss experienced by people with NF2.

Schwannomatosis causes chronic pain, which can occur anywhere in your body. Other symptoms include:

- Numbness or weakness in various parts of your body

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Locations, travel & lodging
Clinical trials
Research
Costs & insurance
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- Loss of muscle

When to see a doctor

See your doctor if you or your child develops signs or symptoms of neurofibromatosis. The tumors associated with neurofibromatosis are often benign and slow growing. So although it's important to obtain a timely diagnosis, the situation isn't an emergency.

Causes

Neurofibromatosis is caused by genetic defects (mutations) that either are passed on by a parent or occur spontaneously at conception. The specific genes involved depend on the type of neurofibromatosis:

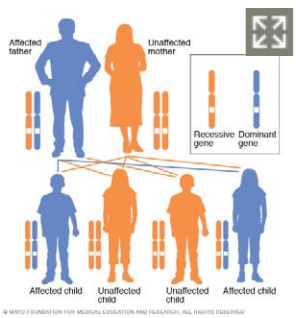
- **NF1.** The NF1 gene is located on chromosome 17. This gene normally produces a protein called neurofibromin that helps regulate cell growth. The mutated gene causes a loss of neurofibromin, which allows cells to grow uncontrolled.

- **NF2.** The NF2 gene is located on chromosome 22, and produces a protein call merlin. The mutated gene causes a loss of merlin, leading to uncontrolled cell growth.

- **Schwannomatosis.** So far, two genes are known to cause schwannomatosis.

Risk factors

The biggest risk factor for neurofibromatosis is a family history of the disorder. About half of people with NF1 and NF2 inherited the disease. NF1 and NF2 that isn't inherited results from new gene mutations.



Autosomal dominant inheritance

NF1 and NF2 are both autosomal dominant disorders, which means that any child of a parent with the disorder

has a 50 percent chance of inheriting the genetic mutation. **pattern**

The inheritance pattern for schwannomatosis is less clear. Researchers currently estimate that the risk of inheriting schwannomatosis from an affected parent is about 15 percent.

Complications

Complications of neurofibromatosis vary, even within the same family. Generally, complications result from tumor growth distorting nerve tissue or pressing on internal organs.

NF1 complications

Complications of NF1 include:

- **Neurological problems.** Learning and thinking difficulties are the most common neurological problem associated with NF1. Uncommon complications include epilepsy and buildup of excess fluid in the brain.
- **Concerns with appearance.** Visible signs of neurofibromatosis — such as extensive cafe au lait spots, numerous neurofibromas in the facial area or large neurofibromas — can cause anxiety and emotional distress, even if they're not medically serious.
- **Skeletal problems.** Some children have abnormally formed bones, which can result in bowing of legs and fractures that sometimes don't heal. NF1 can cause curvature of the spine (scoliosis) that may need bracing or surgery. NF1 is also associated with decreased bone mineral density, which increases your risk of weak bones (osteoporosis).
- **Vision problems.** Occasionally in children, an optic glioma can develop, affecting vision.
- **Problems during times of hormonal change.** Hormonal changes associated with puberty, pregnancy or menopause might cause an increase in

neurofibromas. Most women with NF1 have healthy pregnancies but will likely need monitoring by an obstetrician familiar with the disorder.

- **Cardiovascular problems.** People with NF1 have an increased risk of high blood pressure and, rarely, blood vessel abnormalities.

- **Breathing problems.** Rarely, plexiform neurofibromas can put pressure on your airway.

- **Cancer.** An estimated 3 to 5 percent of people with NF1 develop cancerous tumors. These usually arise from neurofibromas under the skin or from plexiform neurofibromas. People with NF1 also have a higher risk of other forms of cancer, such as breast cancer, leukemia, brain tumors and some types of soft tissue cancer.

- **Benign adrenal gland tumor (pheochromocytoma).** This noncancerous tumor secretes hormones that raise your blood pressure. A pheochromocytoma is generally surgically removed.

NF2 complications

Complications of NF2 include:

- Partial or total deafness

- Facial nerve damage

- Vision problems

- Small benign skin tumors (skin schwannomas)

- Weakness or numbness in the extremities

- Multiple benign brain tumors or spinal tumors requiring frequent surgeries (meningiomas)

Schwannomatosis complications

The pain caused by schwannomatosis can be debilitating and

may require surgical treatment or management by a pain specialist.

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Your doctor will start with a physical examination and review of your medical history and family medical history.

NF1 often can be diagnosed based on physical examination. Your doctor may use a special lamp to check your skin for cafe au lait spots. A physical examination and family history are also important for a diagnosis of NF2.

Your doctor also might recommend:

- **Eye exam.** An eye doctor can detect Lisch nodules and cataracts.
- **Ear exam.** Tests such as audiometry, electronystagmography and brainstem auditory evoked response can help assess hearing and balance problems in people with NF2.
- **Imaging tests.** X-rays, CT scans or MRIs can help identify bone abnormalities, tumors in the brain or spinal cord, and very small tumors. An MRI might be used to help identify optic gliomas. Imaging tests are also often

used to monitor NF2 and schwannomatosis.

- **Genetic tests.** Tests to identify NF1and NF2 are available and can be done prenatally. Ask your doctor about genetic counseling. Genetic tests for schwannomatosis are limited.

For a diagnosis of NF1, you must have at least two signs of the condition. If your child has only one sign and no family history of NF1, your doctor will likely monitor the child for the development of any additional signs. A diagnosis of NF1 is usually made by age 4.

Genetic testing may help establish the diagnosis.

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Diagnosis

Neurofibromatosis

Treatment

By Mayo Clinic Staff

Neurofibromatosis can't be cured, but treatments are available for your signs and symptoms. Generally, the sooner you or your child is under the care of a doctor trained in treating neurofibromatosis, the better the outcome.

Monitoring

If your child has NF1, your doctor is likely to recommend yearly age-appropriate checkups to:

- Assess your child's skin for new neurofibromas or changes in existing ones
- Check for signs of high blood pressure
- Evaluate your child's growth and development — including height, weight and head circumference — according to growth charts available for children with NF1
- Check for signs of early puberty

- Evaluate your child for any skeletal changes and abnormalities
- Assess your child's learning development and progress in school
- Obtain a complete eye examination

Contact your doctor promptly if you notice any changes in signs or symptoms between visits. It's important to rule out the possibility of a cancerous tumor and to obtain appropriate treatment at an early stage.

Surgery and other procedures

Your doctor might recommend surgery or other procedures to treat severe symptoms or complications of neurofibromatosis.

- **Surgery to remove tumors.** Symptoms can be relieved by removing all or part of tumors that are compressing nearby tissue or damaging organs. If you have NF2 and have experienced hearing loss, brainstem compression or tumor growth, your doctor might recommend surgery to remove acoustic neuromas that are causing your problems. Complete removal of schwannomas in people with schwannomatosis can ease pain substantially.
- **Stereotactic radiosurgery.** This procedure delivers radiation precisely to your tumor and doesn't require an incision. Stereotactic radiosurgery might be an option to remove acoustic neuromas if you have NF2. Stereotactic radiosurgery can help preserve your hearing.
- **Auditory brainstem implants and cochlear implants.** These devices might help improve your hearing if you have NF2 and hearing loss.

Cancer treatment

Malignant tumors and other cancers associated with neurofibromatosis are treated with standard cancer therapies, such as surgery, chemotherapy and radiation therapy. Early diagnosis and treatment are the most important factors

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Treatment

resulting in good outcome.

Pain medications

Managing pain is an important part of treatment for schwannomatosis. Your doctor might recommend:

- Gabapentin (Neurotin) or pregabalin (Lyrica) for nerve pain
- Tricyclic antidepressants such as amitriptyline
- Serotonin and norepinephrine reuptake inhibitors such as duloxetine (Cymbalta)
- Epilepsy medications such as topiramate (Topamax) or carbamazepine (Carbatrol, Tegretol)

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