CONDITION

Neurofibromatosis

Neurofibromatosis is a group of genetic conditions that cause tumors to grow along nerves.

WHAT IT IS
Group of genetic conditions causing nerve-related tumors

BODY SYSTEM
Nervous system, skin, eyes, and bones

MAIN SYMPTOMS
Skin changes, nerve tumors, pain, hearing or balance problems

WHAT CAUSES IT
Inherited or new gene changes

HOW IT IS DIAGNOSED
Clinical criteria, imaging, hearing tests, and genetic testing

MAIN TREATMENT
Monitoring, symptom control, surgery, and targeted medicines

CAN IT BE CURED?
No - the gene change remains lifelong

IS IT AN EMERGENCY?
Sometimes - sudden weakness or severe neurological symptoms

LONG-TERM OUTLOOK
Often manageable with lifelong, type-specific monitoring

Overview

Neurofibromatosis is a group of genetic conditions that can cause tumors to grow on or around nerves. These tumors are usually benign, meaning they are not cancer. Their effects depend on where they grow and which type of neurofibromatosis you have.

The main forms are:

The name "neurofibromatosis type 2" is still widely used, but specialists now call it NF2-related schwannomatosis . This reflects updated knowledge about the different genetic forms of schwannomatosis.

Symptoms range from mild to life-changing, even within the same family. Some people mainly have visible skin changes. Others develop hearing loss, pain, problems with movement, or

complications from tumors pressing on nearby structures.

Early recognition matters because regular checks can find some complications before they cause lasting problems.

- Neurofibromatosis type 1 (NF1): Often affects the skin, eyes, bones, and nerves. It can cause café-au-lait spots, freckling, and neurofibromas, which are usually benign nerve tumors.
- NF2-related schwannomatosis: Often causes tumors called schwannomas on the nerves responsible for hearing and balance. It may also cause other brain, spinal, or nerve tumors.
- Other forms of schwannomatosis: Usually cause schwannomas on nerves elsewhere in the body. Long-term nerve pain can be a major problem.

Symptoms

NF1 often first appears in childhood. Flat, light-brown skin patches called café-au-lait spots may be present at birth or appear during the first years of life. Freckles may develop in the armpits or groin.

Neurofibromas usually appear later. These may be soft bumps on or under the skin. Most do not hurt, although they can itch, catch on clothing, or affect appearance.

A plexiform neurofibroma grows along a group of nerves and may extend through nearby tissue. It can cause swelling, pain, weakness, or a difference in the size of a limb or part of the face.

Other possible features of NF1 include:

NF2-related schwannomatosis often causes symptoms in the teenage years or adulthood, although it can begin earlier. Tumors on the hearing and balance nerves are called vestibular schwannomas . They may cause gradual hearing loss, ringing in the ears, dizziness, or poor balance.

Other tumors can affect the brain, spinal cord, or nerves elsewhere in the body. Possible symptoms include numbness, weakness, facial movement problems, headaches, seizures, or changes in vision.

Other forms of schwannomatosis commonly cause ongoing or repeated pain. A tumor pressing on a nerve may also cause tingling, numbness, or weakness. Some people have only one part of the body affected, particularly when the gene change is present in only some cells.

- Learning, attention, speech, or coordination difficulties

- Headaches or seizures
- Curving of the spine or other bone changes
- High blood pressure
- Vision problems from a tumor along the optic nerve
- Small, harmless spots on the coloured part of the eye

Causes and risk factors

Neurofibromatosis is caused by a change in a gene that normally helps control cell growth. When that gene does not work as expected, certain cells around nerves can grow into tumors.

NF1 is caused by a change in the NF1 gene. NF2-related schwannomatosis involves the NF2 gene. Other forms of schwannomatosis may involve genes such as SMARCB1 or LZTR1 , although genetic testing does not find a cause in everyone.

These conditions can be inherited. A person with an NF1 or NF2-related gene change generally has a 1 in 2 chance of passing it to each child. The severity in a child cannot be predicted from the parent's symptoms.

Many people are the first in their family to have the condition. This happens when a new gene change develops in an egg, sperm, or early embryo. It is not caused by anything a parent did during pregnancy.

Sometimes the gene change is mosaic , meaning it is present in only some of the body's cells. Mosaic neurofibromatosis may affect one area of the body or cause fewer symptoms. It can also make genetic testing and inheritance estimates more complex.

Diagnosis

Diagnosis usually combines your medical history, family history, examination, and tests. The exact process depends on the suspected type.

NF1 can often be diagnosed using established clinical criteria. These include a particular combination of café-au-lait spots, freckling, neurofibromas, eye findings, bone changes, certain tumors, family history, or an NF1 gene change.

Skin patches alone do not always mean NF1. Other genetic conditions, including Legius syndrome, can cause similar markings without neurofibromas.

If NF2-related schwannomatosis or another schwannomatosis is suspected, testing may

include:

A genetic result can confirm the diagnosis and help identify the subtype. However, a negative blood test does not always rule it out. Some gene changes are difficult to detect, and mosaic changes may not be present in blood.

A biopsy is not routinely needed for every tumor. It may be considered if imaging or symptoms suggest that a tumor is behaving unusually.

- MRI scans of the brain, spine, or affected nerves
- Detailed hearing and balance tests
- Eye examinations
- Neurological examinations of strength, feeling, and movement
- Genetic testing of blood, saliva, or sometimes tumor tissue

Treatment and management

There is no treatment that removes the underlying gene change. Care focuses on monitoring, treating troublesome tumors, protecting nerve function, and supporting daily life.

Regular follow-up is tailored to the type of neurofibromatosis, your age, existing tumors, and symptoms. It may include skin and neurological examinations, blood pressure checks, eye tests, hearing tests, and MRI scans. Children with NF1 may also need monitoring of growth, development, learning, and bone health.

Many small, painless tumors do not need treatment. A tumor may be removed if it causes pain, weakness, pressure on an organ, repeated injury, or major concerns about appearance. Surgery is planned carefully because nerve tumors may be closely attached to functioning nerves.

Treatment may also include:

Targeted medicines can slow or shrink certain tumors by blocking signals involved in cell growth. For example, a medicine called a MEK inhibitor may be considered for some symptomatic plexiform neurofibromas in NF1 that cannot be safely removed. Bevacizumab may help selected people with growing vestibular schwannomas and hearing loss related to NF2. These treatments require specialist assessment and monitoring.

Radiotherapy is considered cautiously, particularly in NF1, because radiation may raise the long-term risk of new tumors in the treated area. Decisions about surgery, medicine, or radiotherapy are best made by a specialist team familiar with neurofibromatosis.

- Pain management, including treatment aimed at nerve pain
- Physiotherapy or occupational therapy
- Hearing aids or hearing implants
- Educational and psychological support
- Treatment for high blood pressure, seizures, or bone problems
- Surgery for selected brain, spinal, or peripheral nerve tumors

Complications

Most neurofibromas are benign. However, a plexiform neurofibroma in NF1 can rarely change into a cancer called a malignant peripheral nerve sheath tumor .

Possible warning signs include a lump that grows quickly, becomes hard, causes new persistent pain, or leads to weakness or numbness. These changes need prompt assessment, but they do not automatically mean cancer.

NF1 can also be associated with tumors along the optic pathway, which carries visual information from the eye to the brain. Other complications may include high blood pressure, narrowed blood vessels, seizures, scoliosis, fragile bones, or learning difficulties.

NF2-related schwannomatosis may lead to hearing loss, balance problems, facial weakness, or pressure on the brain or spinal cord. Associated tumors can include meningiomas, which arise from the lining around the brain and spinal cord, and ependymomas, which arise from cells lining fluid-filled spaces in the nervous system.

Other forms of schwannomatosis can cause chronic nerve pain, reduced movement, sleep problems, and difficulty working or carrying out everyday tasks.

The specific risks are different for each type, so follow-up should match the confirmed or suspected diagnosis.

Prevention

Neurofibromatosis cannot usually be prevented. An inherited or newly occurring gene change is not caused by lifestyle, diet, stress, or something a parent did.

If you have neurofibromatosis or a relevant gene change, genetic counselling can explain the chance of passing it on. It can also discuss testing options before or during pregnancy, including preimplantation genetic testing with in vitro fertilisation. These are personal choices,

and testing cannot predict exactly how severely a child would be affected.

Monitoring does not prevent the condition, but it may reduce harm by finding treatable complications earlier. Keeping scheduled eye, hearing, blood pressure, developmental, and imaging checks is therefore important.

Living with it and what to expect

Neurofibromatosis is lifelong, but its course is different for each person. Existing skin markings usually remain. New neurofibromas may appear over time in NF1, particularly around puberty or pregnancy, although the number and impact are difficult to predict.

Some tumors remain stable for years. Others grow and require treatment. Symptoms such as pain, hearing loss, or weakness may develop gradually, so long-term comparison of examinations and scans can be more useful than a single test.

Many people with NF1 have mild to moderate symptoms and live independently. Learning or attention difficulties may need support during school and sometimes at work. Visible tumors can affect confidence even when they are medically harmless.

People with NF2-related schwannomatosis may need ongoing hearing support and repeated treatment for tumors. Other forms of schwannomatosis may require long-term pain care. Mental health support can be valuable when pain, appearance changes, hearing loss, or uncertainty affect wellbeing.

Tell new healthcare professionals about your diagnosis, particularly before surgery or imaging. Keeping a record of previous scans, genetic results, operations, hearing tests, and tumor changes can make future care easier.

When to get medical help

Arrange a prompt medical assessment if you notice:

Use emergency services for sudden weakness on one side, sudden inability to walk, a seizure lasting several minutes or repeated seizures without recovery, loss of consciousness, or a sudden severe headache with neurological symptoms.

Routine appointments remain important even when you feel well. They are used to check blood pressure, vision, hearing, development, and known tumors rather than to respond to an emergency.

- A tumor that is rapidly enlarging, hardening, or becoming persistently painful

- New or worsening weakness, numbness, balance problems, or facial movement changes
- New hearing loss, ringing in one ear, or repeated dizziness
- A change in vision, including reduced or double vision
- Persistent headaches, repeated vomiting, or new seizures
- New bladder or bowel control problems
- A child losing skills or developing clear learning, growth, or movement concerns

Common questions

Is neurofibromatosis cancer?

No. Neurofibromatosis is a genetic condition, and most tumors linked to it are benign. Some people with NF1 have an increased risk of particular cancers, so a rapidly changing or persistently painful tumor should be checked promptly.

Will my child inherit neurofibromatosis?

If you have a gene change that can be inherited, each pregnancy generally has a 1 in 2 chance of receiving it. Mosaic forms may have a different chance. Genetic counselling can give advice based on your test results and family history.

Do café-au-lait spots turn into tumors?

No. Café-au-lait spots are areas of increased skin pigment and do not become neurofibromas or cancer. They can be one clue to NF1, but a few spots can also occur in people without the condition.

Can neurofibromas be removed?

Yes, selected neurofibromas can be removed with surgery or other procedures. Removal may be considered for pain, loss of function, repeated irritation, suspected cancer, or appearance concerns. Some tumors can return or new ones can form elsewhere.

Does neurofibromatosis affect life expectancy?

Many people, especially those with mildly affected NF1, live well into adulthood and have active lives. Life expectancy can be reduced by serious vascular problems or malignant tumors in some people with NF1, and by complex nervous-system tumors in severe NF2-related schwannomatosis. Monitoring aims to identify these problems early.

Can I exercise if I have neurofibromatosis?

Many people can exercise normally. Activities may need adjustment if you have weak bones,

balance problems, a spinal tumor, significant pain, or recent surgery. Advice should be based on your known complications rather than the diagnosis alone.

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