CONDITION

Bilateral acoustic neurofibromatosis

Learn about NF2-related schwannomatosis, an inherited condition that can cause tumors affecting hearing, balance, nerves, the brain, and the spine.

WHAT IT IS
Inherited nerve tumor condition

BODY SYSTEM
Nervous system

MAIN SYMPTOMS
Hearing loss, tinnitus, balance problems, facial weakness

WHAT CAUSES IT
Inherited NF2 gene change

HOW IT IS DIAGNOSED
MRI and NF2 genetic testing

MAIN TREATMENT
Monitoring, surgery, radiation, targeted medicine

CAN IT BE CURED?
No, new tumors can appear

IS IT AN EMERGENCY?
Sometimes, seizure lasting several minutes

LONG-TERM OUTLOOK
Variable, with potentially serious neurological complications

Overview

Bilateral acoustic neurofibromatosis is an older name for NF2-related schwannomatosis . It was also previously called neurofibromatosis type 2, or NF2. The newer name better reflects the condition's cause and the types of tumors it produces.

NF2-related schwannomatosis is a genetic condition that increases the likelihood of developing tumors along nerves. Its hallmark is a vestibular schwannoma on each side of the head. These tumors arise from cells that support the nerves responsible for hearing and balance. "Acoustic neuroma" is another common name, although vestibular schwannoma is more medically accurate.

These tumors are usually benign, meaning they are not cancer. Even so, they may cause important health problems by pressing on nearby nerves, the brainstem, or the spinal cord.

Hearing loss, tinnitus, balance difficulties, facial weakness, and other neurological symptoms can develop.

People with this condition may also develop schwannomas on other nerves, meningiomas around the brain or spinal cord, and tumors called ependymomas within the spinal cord. Eye findings, including certain cataracts, may occur as well.

The condition affects each person differently. Some tumors remain stable for years, while others grow or cause symptoms. Regular monitoring helps the care team identify meaningful changes and decide whether observation, surgery, medicine, hearing support, or another treatment is appropriate.

Symptoms

Symptoms depend on which nerves or structures are affected. They may begin gradually and can differ between the two sides of the body.

Vestibular schwannomas commonly cause:

A larger tumor may affect nearby cranial nerves, which connect the brain to the face and head. This can cause facial numbness, facial weakness, swallowing difficulty, or changes in voice.

Tumors elsewhere in the nervous system may cause headaches, seizures, weakness, numbness, persistent pain, or difficulty walking. A spinal tumor may affect an arm or leg or interfere with bladder and bowel control. Peripheral nerve tumors can appear as painful or painless lumps under the skin.

Eye findings can include reduced vision from cataracts or changes involving the retina, the light-sensitive tissue at the back of the eye. In some children and young adults, an eye problem, skin tumor, facial weakness, or nerve problem may appear before hearing symptoms.

Symptoms alone cannot confirm NF2-related schwannomatosis. Many common conditions can also cause hearing changes, tinnitus, headaches, or dizziness. New or persistent symptoms should be assessed rather than assumed to be caused by a tumor.

- Hearing loss in one or both ears
- Ringing, buzzing, or other sounds in the ear, called tinnitus
- Difficulty understanding speech, especially in noisy places
- Unsteadiness, dizziness, or problems with balance
- A feeling of pressure or fullness in an ear

Causes and risk factors

NF2-related schwannomatosis is caused by a harmful change, called a pathogenic variant, in the NF2 gene. This gene provides instructions for making merlin, a protein that helps control cell growth. When merlin does not work normally, certain nerve-supporting cells can grow into tumors.

The condition follows an autosomal dominant inheritance pattern. A person with an NF2 pathogenic variant in their egg or sperm cells has a 1 in 2 chance of passing it to each child. The chance is the same for every pregnancy.

Some people inherit the variant from an affected parent. In others, it arises for the first time early in development. This is called a de novo variant.

A person may also have mosaic NF2-related schwannomatosis . Mosaicism means the variant is present in some cells but not others. Mosaic disease may cause fewer tumors or affect one part of the body more than another. It can also make genetic testing and assessment of inheritance more complicated.

Family history and an NF2 pathogenic variant are the main known risk factors. The condition is not caused by diet, exercise, stress, or something a person did. Tumor growth can also vary among relatives who carry the same genetic change.

Diagnosis

Diagnosis usually involves a clinical evaluation, imaging, hearing tests, and genetic testing. Because the condition is uncommon and can overlap with other tumor-predisposition disorders, assessment at a center experienced in schwannomatosis can be helpful.

The evaluation may include:

MRI uses a magnetic field to create detailed images. It can show vestibular schwannomas, meningiomas, spinal tumors, and other findings. Contrast material may be used to make small tumors easier to see.

Current diagnostic criteria consider tumor type, tumor location, eye findings, family history, and molecular test results. Bilateral vestibular schwannomas strongly support the diagnosis, but a person may meet criteria through another combination of findings.

Genetic testing usually begins with a blood or saliva sample. A negative result does not always rule out the condition, especially when mosaicism is suspected. Testing tumor tissue may identify an NF2 variant that is not detectable in blood. A genetics professional can explain what a positive, negative, or uncertain result means for the person and their relatives.

Doctors may also consider other explanations, such as a single sporadic vestibular schwannoma, other forms of schwannomatosis, or unrelated brain and spinal tumors.

- A detailed personal and family medical history
- A neurological examination
- Magnetic resonance imaging, or MRI, of the brain and internal auditory canals
- MRI of the spine when indicated
- Hearing tests, including an audiogram and speech-understanding tests
- An eye examination
- Genetic counseling and testing

Treatment and management

There is no single treatment plan for everyone with NF2-related schwannomatosis. Decisions are based on symptoms, hearing, tumor location, tumor growth, age, overall health, and the likely effects of treatment.

Care commonly involves specialists in neurology, neurosurgery, ear and skull-base surgery, oncology, medical genetics, audiology, ophthalmology, rehabilitation, and mental health.

Monitoring

A tumor that is small, stable, and not causing major symptoms may be monitored with MRI scans, hearing tests, and clinical examinations. Monitoring avoids treatment-related risks, but it requires regular follow-up so meaningful changes are not missed.

Surgery

Surgery may be considered when a tumor is growing, causing neurological problems, compressing important structures, or threatening useful hearing. The goal may be complete removal, partial removal, pressure relief, or protection of nerve function.

Surgery near the hearing, balance, or facial nerves can affect those functions. The expected benefit and the possibility of hearing loss, facial weakness, balance problems, infection, bleeding, or other complications should be discussed with an experienced surgical team.

Radiation treatment

Focused radiation, including stereotactic radiosurgery, may control selected tumors without an open operation. Radiation is used cautiously in people with NF2-related schwannomatosis because they often have multiple tumors, may need treatment at a young age, and may face long-term radiation effects. It is not suitable for every tumor.

Medicine

Bevacizumab is a medicine that blocks signals involved in blood-vessel growth. It may shrink some rapidly growing vestibular schwannomas and improve or stabilize hearing in some people. Its use for NF2-related schwannomatosis depends on local approval, specialist judgment, and the individual clinical situation.

Bevacizumab can cause significant adverse effects, so treatment requires medical screening and monitoring. Other targeted medicines are being studied in clinical trials. Participation in a trial may be an option for some people but does not guarantee benefit.

Hearing and supportive care

Hearing aids may help when speech understanding remains adequate. Cochlear implants or auditory brainstem implants may be considered when ordinary hearing aids are not effective, depending on the condition of the hearing nerve and inner ear.

Other supportive treatments can include balance therapy, physical or occupational therapy, pain management, speech or swallowing therapy, vision care, and counseling. Captioned communication, assistive listening devices, and sign language can also support independence and communication.

Complications

Hearing loss is one of the most important potential complications. It may progress gradually or change after tumor treatment. Loss of hearing in both ears can affect communication, education, employment, relationships, and personal safety.

Vestibular schwannomas and other tumors can also lead to:

Treatment itself may affect hearing, facial movement, balance, or other nerve functions. These risks are weighed against the harm that an untreated, growing tumor could cause.

Most NF2-associated tumors are benign. Malignant, or cancerous, transformation is uncommon, but any rapidly changing tumor needs specialist review. Emotional strain is also common when a person is managing repeated scans, hearing changes, treatment decisions, or uncertainty about relatives and children.

- Long-term tinnitus or balance problems
- Facial numbness or weakness
- Difficulty swallowing or speaking
- Chronic pain or peripheral nerve damage

- Weakness, altered sensation, or difficulty walking
- Seizures
- Vision problems
- Spinal cord compression
- Buildup of fluid around the brain, called hydrocephalus

Follow up

Long-term follow-up is essential because new tumors can appear and existing tumors can change. The schedule should be individualized according to age, symptoms, previous treatment, tumor burden, and genetic findings.

Routine follow-up may include neurological examinations, hearing and speech-understanding tests, brain MRI, spinal imaging, eye examinations, and review of balance, pain, mobility, communication, and daily function. People receiving medicine or recovering from surgery may need additional testing.

Contact the care team promptly for a noticeable change in hearing, new facial weakness or numbness, worsening balance, persistent vomiting, new weakness, increasing pain, vision changes, or new bladder or bowel problems. Sudden hearing loss should receive urgent medical assessment because prompt evaluation may identify causes other than tumor growth that are time-sensitive.

Seek emergency care for a seizure lasting several minutes, loss of consciousness, sudden severe weakness, inability to walk, severe breathing or swallowing difficulty, or a sudden intense headache with repeated vomiting or confusion.

Genetic counseling can help relatives understand whether testing is appropriate. It can also explain reproductive options without directing a person toward any particular choice. Psychological support, hearing-loss services, school or workplace accommodations, and patient support organizations may be useful parts of ongoing care.

Sources

- Evans DG. "NF2-Related Schwannomatosis." GeneReviews . University of Washington, Seattle. Originally published 1998; updated April 20, 2023.

- Plotkin SR, Messiaen L, Legius E, et al. "Updated diagnostic criteria and nomenclature for neurofibromatosis type 2 and schwannomatosis: An international consensus recommendation." Genetics in Medicine . 2022;24(9):1967-1977.
- Halliday D, Emmanouil B, Pretorius P, et al. "ERN GENTURIS tumour surveillance guidelines for individuals with neurofibromatosis type 2-related schwannomatosis." eClinicalMedicine . 2023;56:101818.
- Goldbrunner R, Weller M, Regis J, et al. "EANO guideline on the diagnosis and treatment of vestibular schwannoma." Neuro-Oncology . 2020;22(1):31-45.