



Neurofibromatosis type 2 is a genetic predisposition to develop growths and tumors in different parts of the body. These slow growing tumors most commonly occur in the central nervous system, with the average age of onset in young adulthood (between 18-24 years old). Almost all people with NF2 will develop bilateral vestibular schwannomas (benign tumors that develop in the inner ear on both sides that can affect balance and hearing) by the age of 30. Complications from other central nervous system tumors can include changes in vision, weakness or numbness in the arms and legs, or fluid buildup in the brain. Some people with NF2 can also develop cataracts (a clouding of the lens of the eye) in either one or both of their eyes, and can occur as early as childhood.

Causes

We have over 20,000 different genes in the body. These genes are like instruction manuals for how to build a protein, and each protein has an important function that helps to keep our body working how it should. The *NF2* gene makes a protein called merlin. Merlin works with other proteins to control how quickly the body's cells multiply (called a tumor suppressor protein). Merlin is made in the body's central nervous system. If someone has a pathogenic (or harmful) variant in one of their *NF2* genes, then their body does not make as much merlin as it should. Without enough merlin, there is not proper control over how fast our cells grow, which can lead them to multiply more cell growth than there should be. This unregulated cell growth can lead to tumors and the other signs and symptoms that we associate with NF2.

Pathogenic variants in the *NF2* gene causes NF2, and are inherited in an [autosomal dominant](#) pattern, meaning that children of a carrier each have a 50% risk to inherit the pathogenic variant and associated health risks. Notably, women and men both have the *NF2* genes and have the same chances to inherit and pass down variants in this gene. Therefore, both sides of the family are important when assessing inherited risk. About 50% (or 1 in 2 people) with NF2 will also have a parent with NF2. The other 50% have no family history and will be the first ones in their family with this condition.

It is estimated that approximately 1 in 33,000 people have NF2.

Genetic Testing for NF2

Genetic testing for pathogenic variants in *NF2* is currently available, but there are a few different ways to approach testing:

- [Single site analysis](#): Testing specific to a known pathogenic variant in the family



- Full gene [sequencing](#) and [rearrangement analysis](#): Comprehensive testing to search for all currently detectable pathogenic variants in the gene
- [Gene panels](#): Newer, more broadly based gene tests that would include not only the *NF2* gene, but other genes known or suspected to be associated with these types of tumors.

Diagnosing NF2

A diagnosis of NF2 can be complex and therefore may be best conducted by a medical provider who is familiar with NF2. A diagnosis can usually be made based on a review of medical records (clinical diagnosis) and genetic testing for pathogenic variants in the *NF2* gene.

NF2 is suspected in a person with any of the following:

- Bilateral vestibular schwannomas (formerly known as acoustic neuromas); benign tumors of the cranial nerves that control hearing and are associated with hearing loss, ringing of the ears, and trouble balancing
- Meningioma
- Neuropathy such as a facial palsy, foot drop or hand drop
- Schwannomas of the spine
- Hamartoma (benign tumor) of the retina
- Neurofibromas

If you or your family meet any of the above criteria, a meeting with a pediatric or adult genetics specialist can be helpful to further determine if someone has NF2.

Medical Management for NF2

Medical management for someone with NF2 should be overseen by a medical provider who is familiar with NF2 . Because NF2 affects people in very different ways, medical management should be specific to an individual's needs. The frequency of each screening tool is tailored to an individual's age. Some options for surveillance include:

- Regular neurological exams and physicals
- MRI of the brain
- Hearing evaluations
- Eye exams

More personalized recommendations will take into account your personal or family history,



and should be discussed with your medical provider.

Click [here](#) to learn more about scheduling a genetic counseling appointment for questions about hereditary cancer predisposition.

Resources

[The Washington University Neurofibromatosis Center](#)

[Children's Tumor Foundation](#)

[Neurofibromatosis Network](#)