



# My46 Trait Profile

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## Neurofibromatosis type 2

Other Names: Bilateral acoustic neurofibromatosis

*Neurofibromatosis type 2 is a genetic condition characterized by growths that develop around the hearing and balance nerves in the inner ears. It is caused by mutations in the NF2 gene that makes a protein called merlin.*

### Characteristics of Neurofibromatosis type 2

Neurofibromatosis type 2 (NF2) is a genetic condition characterized by bilateral vestibular schwannomas (i.e., growths or tumors that develop around the hearing and balance nerves within both inner ears). These tumors usually develop in teenagers or young adults. Symptoms include hearing loss, tinnitus (i.e., ringing in the ear), and loss of balance or dizziness. Since these symptoms are often subtle and non-specific, vestibular schwannomas may go undiagnosed for some time. Hearing tests and imaging of the head are needed to identify these tumors. Other common features of NF2 are cataracts (i.e., clouding of the lens) and tumors in other parts of the brain or spinal cord (e.g., meningiomas and gliomas). Intelligence is usually normal in NF2.

### Diagnosis/Testing

NF2 can be diagnosed either by meeting specific clinical criteria or by genetic testing for a change or mutation in a gene called NF2. Individuals with bilateral vestibular schwannomas meet clinical criteria for a diagnosis of NF2. Individuals with a unilateral (i.e., one-sided) vestibular schwannoma might also have NF2, but additional features must be present for the clinical diagnosis of NF2 to be made. The NF2 gene makes a protein called merlin (also called schwannomin), which functions to help keep cells from growing in an uncontrolled way. Mutations in the NF2 gene are thought to interfere with this function.

Genetic testing does not detect mutations in all individuals with NF2. In individuals with a clinical diagnosis of NF2 with no family history of the condition, genetic testing is less likely to identify a mutation than in individuals with NF2 with a family history of the condition.

### Management/Surveillance

Management and surveillance of NF2 often includes regular physical exams, eye exams, hearing evaluations, and imaging studies of the head. Surgery or a special type of directed radiation therapy (i.e., stereotactic radiosurgery) to remove a vestibular schwannoma is required in some individuals with NF2.

### Mode of inheritance

NF2 is inherited in an autosomal dominant pattern. This means inheriting one NF2 mutation is enough for an individual to be affected and show signs of NF2. The mutation can be inherited from an affected parent or it can occur brand new (de novo) in an affected child.

### Risk to family members

The risk to family members depends on whether or not the individual with NF2 has a parent affected with NF2. If a parent also has NF2, the risk of having a child with NF2 is 50% with each pregnancy. If a parent does not have NF2, the

risk of future pregnancies being affected is very low.

### Special considerations

None

### Resources

Childrens Tumor Foundation: Ending Neurofibromatosis Through Research

<http://www.ctf.org>

Genetics Home Reference: Neurofibromatosis type 2

<http://ghr.nlm.nih.gov/condition/neurofibromatosis-type-2>

Neurofibromatosis, Inc.

<http://www.nfinc.org>

### References

[Blakeley, J. et al. \(2012\)](#). "Consensus Recommendations for Current Treatments and Accelerating Clinical Trials for Patients with Neurofibromatosis Type 2." American Journal of Medical Genetics 158A(1): 24-41.

Evans DG. (Updated 2011 August 18). Neurofibromatosis 2. In: GeneReviews at GeneTests Medical Genetics Information Resource (database online). Copyright, University of Washington, Seattle. 1997-2013. Available at <http://www.ncbi.nlm.nih.gov/books/NBK1201/>. Accessed [01/23/2013].

[Hoa, M. et al. \(2012\)](#). "Neurofibromatosis 2." Otolaryngologic Clinics of North America 45(2): 315-332.

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