NF 2 Gene

The official name of this gene is “neurofibromin 2 (merlin).”

NF2 is the gene's official symbol. The NF2 gene is also known by other names.

Biallelic inactivation of the NF2 gene has been established as the principal underlying genetic event in patients with sporadic and syndrome-associated vestibular schwannoma (VS). Two independent teams contemporaneously identified the NF2 gene located on chromosome 22 at 22q12.2 in 1993, which codes for the tumor suppressor protein Merlin, also called schwannomin

The NF2 gene provides instructions for the production of a protein called merlin, also known as schwannomin [1) 2)].

To date, few studies have used whole-exome sequencing, mate-pair analysis, and RNA-seq to profile genome-wide alterations in sporadic VS. Using high-throughput deep sequencing, “two-hit” alterations in the NF2 gene were identified in every tumor and were not present in peripheral blood supporting that all events were somatic. Type of NF2 gene alteration and accessory mutations outside the NF2 locus may predict phenotypic expression and clinical course [3)].

1)
Rouleau GA, Merel P, Lutchman M, et al. Alteration in a new gene encoding a putative membrane-organizing protein causes neurofibromatosis type 2. Nature 1993;363:515–21.
2)
Trofatter JA, MacCollin MM, Rutter JL, et al. A novel moesin-, ezrin-, radixin-like gene is a candidate for the neurofibromatosis 2 tumor suppressor. Cell 1993;72:791–800.
3)
Carlson ML, Smadbeck JB, Link MJ, Klee EW, Vasmatzis G, Schimmenti LA. Next Generation Sequencing of Sporadic Vestibular Schwannoma: Necessity of Biallelic NF2 Inactivation and Implications of Accessory Non-NF2 Variants. Otol Neurotol. 2018 Aug 13. doi: 10.1097/MAO.0000000000001932. [Epub ahead of print] PubMed PMID: 30106846.