NF2-RELATED SCHWANNOMATOSIS

A diagnosis of NF2-related schwannomatosis can be made when a patient has one of the following:

» Bilateral vestibular schwannomas (VS)
» An identical NF2 pathogenic variant* in at least two anatomically distinct NF2-related tumors (schwannoma, meningioma, and/or ependymoma)
» When either two Major OR one Major and two Minor criteria are present as follows:

**MAJOR CRITERIA**

» Unilateral vestibular schwannoma (VS)
» First-degree relative other than a sibling with NF2-related schwannomatosis
» Two or more meningiomas
  (Note: single meningioma qualifies as a minor criterion)
» NF2 pathogenic variant* in an unaffected tissue such as blood or saliva

* When the variant is present at significantly less than 50%, the diagnosis is mosaic NF2-related schwannomatosis

**MINOR CRITERIA**

Can count more than once of each type (e.g., two schwannomas = two minor criteria)

» Ependymoma; schwannoma (Note: if the major criterion is unilateral vestibular schwannomas, at least one schwannoma must be dermal in location)

Can count only once

» Juvenile subcapsular or cortical cataract; retinal hamartoma; epiretinal membrane in a person aged less than 40 years; meningioma
  (Note: multiple meningiomas qualify as a major criteria; meningioma cannot be used as both a major and minor criterion)

**ADDITIONAL GENETIC CRITERIA:**

» Genetic analysis may identify pathogenic NF2 variants in blood in 66%-90% of individuals
» Genetic analysis is not REQUIRED for diagnosis. It will be possible to diagnose NF2-related schwannomatosis based on clinical criteria without genetic analysis
» Genetic analysis with family history will be sufficient to diagnose NF2-related schwannomatosis (no requirement to have tumors)

More information including a link to the 2021 and 2022 publications with updates to the diagnostic criteria for all types of neurofibromatosis and schwannomatosis can be found at ctf.org/criteria.

Learn more about all types of neurofibromatosis and schwannomatosis on the Children’s Tumor Foundation website at: ctf.org