DIAGNOSTIC CRITERIA FOR *NF2*-RELATED SCHWANNOMATOSIS

2022 UPDATE

CHILDREN'S TUMOR FOUNDATION
ENDING NF THROUGH RESEARCH

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*

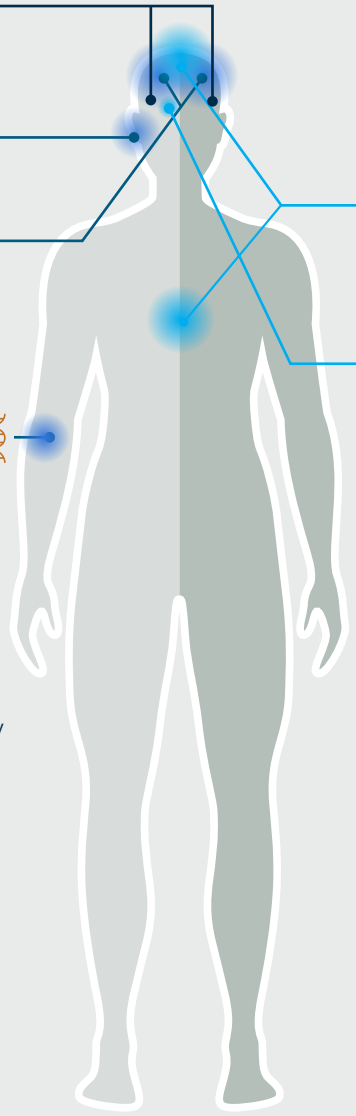
Mosaicism

Mosaicism is confirmed for NF2-related schwannomatosis by **either** of the following:

» Clearly less than 50% pathogenic variant allele fraction in blood or saliva

OR

» Pathogenic variant not detected in clinically unaffected tissue but shared pathogenic variant in two or more anatomically unrelated tumors



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)

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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**

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