A diagnosis of SMARCB1-related schwannomatosis can be made when a patient meets one of the following:

» At least one pathologically confirmed schwannoma or hybrid nerve sheath tumor AND a SMARCB1 pathogenic variant in an unaffected tissue such as blood or saliva

» A common SMARCB1 pathogenic variant in two anatomically distinct schwannomas or hybrid nerve sheath tumors

Note: diagnosis requires surgical specimen to confirm tumor histology

Mosaicism

Mosaicism is confirmed for LZTR1-related or SMARCB1-related schwannomatosis by either of the following:

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

OR

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

Note: diagnosis requires surgical specimen to confirm tumor histology

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