22q-RELATED SCHWANNOMATOSIS

Previously classified as “schwannomatosis without identified mutation in blood” for patients with multiple schwannomas with common molecular findings on chromosome 22q

A diagnosis of 22q-related schwannomatosis can be made when an individual does not meet criteria for NF2-related schwannomatosis, SMARCB1-related schwannomatosis, or LTZR1-related schwannomatosis, and has both of the following molecular features:

» Loss of heterozygosity (LOH) of the same chromosome 22q markers in two anatomically distinct schwannomas or hybrid nerve sheath tumors AND

» A different NF2 pathogenic variant in each tumor which cannot be detected in unaffected tissue such as blood or saliva

Note: diagnosis requires at least two surgical specimens

GENETIC ANALYSIS FOR SCHWANNOMATOSIS:

» Genes involved with schwannomatosis are not yet fully understood

» Genetic analysis is required for the diagnosis of a specific type of schwannomatosis (except for NF2-related, or NOS). In most cases, it will NOT be possible to diagnose the type of schwannomatosis based on clinical criteria alone

» Genetic analysis ALONE is not sufficient to diagnose all types of schwannomatosis; diagnosis requires a clinical feature, such as schwannoma

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