Nearly a year ago, an international consensus was reached regarding the revision of diagnostic criteria for Neurofibromatosis type 2 and schwannomatosis, along with a proposal to update the nomenclature (Scott R. Plotkin et al. "Updated diagnostic criteria and nomenclature for neurofibromatosis type 2 and schwannomatosis: An international consensus recommendation," Genetics in Medicine (2022) 24, 1967–1977). This guideline was endorsed by ERN GENTURIS in August 2022.

Historically, neurofibromatosis encompassed three distinct forms, classifying them as **Neurofibromatosis type 1, Neurofibromatosis type 2, and schwannomatosis**. However, it became evident that the defining feature of Neurofibromatosis Type 2 is schwannomas, not neurofibromas. Today, we unequivocally understand that patients with NF2 never exhibit neurofibromas among their clinical manifestations. It was evident that continuing to label these patients as having Neurofibromatosis type 2 would be inaccurate and potentially misleading for both patients and healthcare professionals, increasing the risk of incorrect diagnoses.

An international consortium of healthcare experts, led by Drs. Scott Plotkin, Gareth Evans, and Eric Legius, and supported by the Children's Tumor Foundation, meticulously reviewed the existing diagnostic criteria for Neurofibromatosis Type 2 and schwannomatosis. One of the initial revelations was that both conditions shared a common clinical presentation, namely schwannomas. The phenotypes of these diseases fall along a spectrum, lacking clear-cut subtypes based on clinical characteristics.

The authors of the study proposed the term "schwannomatosis" under the updated criteria to better capture the overlapping clinical features of these related conditions. Furthermore, they suggested classifying the type of schwannomatosis based on the gene harboring a pathogenic variant (PV) identified through molecular analysis. Consequently, in the revised nomenclature, NF2 would be referred to as "*NF2*-related schwannomatosis" while schwannomatosis would be categorized as either "*SMARCB1*-related schwannomatosis" (for patients with germline PV in *SMARCB1*), "*LZTR1*-related schwannomatosis" (for patients with germline PV in *LZTR1*), "22q-related schwannomatosis" (for patients with multiple schwannomas sharing common molecular findings on chromosome 22q), "schwannomatosis-not otherwise specified (NOS)" (for patients displaying clinical features of NF2/schwannomatosis without molecular analysis), or "schwannomatosis-not elsewhere classified (NEC)" (for patients in whom molecular analysis of blood and tumors failed to detect a PV). Therefore, it is imperative that patients suspected of having NF2 or schwannomatosis undergo comprehensive molecular genetic testing, which may involve multiple tissues, including available tumor tissue.

This group of specialists firmly believes that the name change from NF2 to schwannomatosis will significantly reduce misdiagnosis as NF1 and underscore the clinical and genetic overlap between NF2 and schwannomatosis. The renaming is grounded in the fact that neurofibromas do not occur in NF2, rendering the term "neurofibromatosis" inaccurate. Maintaining the label of NF2 as a neurofibromatosis only fosters confusion, even among medical professionals, particularly in relation to NF1.

The new nomenclature changed the names of all the schwannomatosis conditions to reflect the underlying gene, thus: ***NF2*-related schwannomatosis, *SMARCB1*-related schwannomatosis, *LZTR1*-related schwannomatosis, 22q-related schwannomatosis and schwannomatosis not otherwise specified (NOS)** (https://www.ctf.org/images/uploads/documents/DiagnosticCriteria_v1.pdf).

The patient advocacy group - NF Patients United has already discussed the new nomenclature with European patient representatives through a virtual meeting.

Now is the time to **embrace this new nomenclature within ERN GENTURIS**. In the coming weeks, we will update the consensus nomenclature on our website, and **we encourage all GENTURIS members to advocate for the adoption of this nomenclature in their respective countries.** Members of ERN GENTURIS Thematic Group 1 are available to support you throughout the process of updating the nomenclature for this group of pathologies.

Statement prepared by ERN GENTURIS Thematic Group 1 core team