

Special Issue

Neurofibromatosis Type 1 and NF2-Related Schwannomatosis: Advances from Basic Research to Current Therapies

Message from the Guest Editors

Neurofibromatosis type 1 (NF1) and NF2-related schwannomatosis are rare genetic tumor predisposition syndromes characterized by multiple benign and occasionally malignant tumors of the nervous system. NF1 is caused by pathogenic variants in the NF1 gene encoding neurofibromin, leading to dysregulated RAS/MAPK signaling, while NF2-related schwannomatosis results from NF2 gene mutations affecting the tumor suppressor merlin. These disorders present with diverse clinical features, often causing neurological deficits and reduced quality of life. Although recent advances, such as MEK inhibitors for NF1-related plexiform neurofibromas and bevacizumab for NF2-related vestibular schwannomas, have improved management, curative therapies remain elusive. This Special Issue will cover recent progress from basic molecular mechanisms to translational and clinical studies, including novel therapeutic strategies, preclinical models, biomarker discovery, surgical and nonsurgical interventions, genotype–phenotype correlations, and the development of targeted drugs. Our aim is to foster a deeper understanding and improved treatment of these challenging disorders.

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Deadline for manuscript submissions

31 July 2026



International Journal of Molecular Sciences

an Open Access Journal
by MDPI

Impact Factor 4.9
CiteScore 9.0
Indexed in PubMed



mdpi.com/si/251854

*International Journal of
Molecular Sciences*
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