

Special Issue

Recent Advances in Neurofibromatosis Type 1: From Molecular Insights to Novel Therapies

Message from the Guest Editors

Neurofibromatosis type 1 (NF1) is a complex genetic disorder caused by pathogenic variants in the NF1 gene, leading to a wide spectrum of clinical manifestations, including benign and malignant tumors. While research on genotype-phenotype correlations is still limited, recent studies are increasingly focused on understanding these relationships to enhance patient management. Significant progress has been made in targeted therapies, such as MEK inhibitors, which have demonstrated promising results in reducing plexiform neurofibroma volumes in clinical trials. Furthermore, ongoing investigations are exploring the role of mast cells and fibroblasts in the tumor microenvironment, as well as novel agents targeting the Ras/MAPK pathway. Advanced imaging techniques and emerging biomarkers are also improving early diagnosis and monitoring of malignant transformations, such as in malignant peripheral nerve sheath tumors (MPNSTs). This Special Issue aims to highlight the latest scientific discoveries, from molecular research to innovative clinical interventions, to better understand the disease and improve patient outcomes and quality of life.

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