



New Perspectives in Molecular Diagnosis of Neuromuscular Disorders

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Message from the Guest Editor

Neuromuscular disorders (NMDs) affects the peripheral nervous system and muscle, consequently leading to a significant disability in children as well as in adults.

Over the past decade, research has expanded our understanding of the molecular bases of NMDs. However, the causes remain unknown for many patients, also with clear clinical and histological evidence of disease. The genetic and phenotypic heterogeneity and the lack of segregation data in sporadic cases complicate the detection of causative genetic variants in individual families. In addition, some NMD genes, such as TTN, NEB, RYR, and DMD, are the largest human genes with a wide mutational spectrum, including single base substitutions, small indel, and intragenic deletion/duplication of one or more exons.

In this Special Issue, we will publish reviews and original papers covering recent advances and perspectives in the molecular diagnosis of NMDs, in research and clinical practice and the latest developments that are underway in NGS.





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Message from the Editor-in-Chief

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