Special Issue

Natural History in Neurogenetic Disorders: Molecular Characterization in the NGS Era

Message from the Guest Editor

The breakthrough of next-generation sequencing (NGS) technologies has significantly changed the diagnostic approach to inherited neurological disorders. NGS techniques allow for rapid and inexpensive large-scale genomic analysis, creating unprecedented opportunities to integrate genomic data into the clinical phenotyping and management of subjects with neurogenetics diseases. In particular, a deep-phenotyping approach is essential for correctly interpreting genetic variants and reevaluating patients who lack a conclusive genetic diagnosis. Some examples include mitochondrial diseases, neuromuscular disorders, hereditary spastic paraplegia and hereditary ataxias. Furthermore, next-generation sequencing techniques provide powerful ways of interrogating the epigenome and, in combination with other omics technologies, could contribute to identifying new disease mechanisms. For this Special Issue, we are looking for original research articles and state-of-the-art reviews on the molecular characterization of neurogenetic disorders.

Guest Editor

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