

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

Dr. Guido Primiano

Fondazione Policlinico Universitario Agostino Gemelli IRCCS,
Dipartimento di Neuroscienze, Università Cattolica del Sacro Cuore,
00168 Rome, Italy

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Editorial Office
MDPI, Grosspeteranlage 5
4052 Basel, Switzerland
Tel: +41 61 683 77 34
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Message from the Editor-in-Chief

The International Journal of Molecular Sciences (*IJMS*, ISSN 1422-0067) is an open access journal, which was established in 2000. The journal aims to provide a forum for scholarly research on a range of topics, including biochemistry, molecular and cell biology, molecular biophysics, molecular medicine, and all aspects of molecular research in chemistry. *IJMS* publishes both original research and review articles, and regularly publishes special issues to highlight advances at the cutting edge of research. We invite you to read recent articles published in *IJMS* and consider publishing your next paper with us.

Editor-in-Chief

Prof. Dr. Maurizio Battino

Department of Odontostomatologic and Specialized Clinical Sciences, Sez-Biochimica, Faculty of Medicine, Università Politecnica delle Marche, Via Ranieri 65, 60100 Ancona, Italy

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