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New Insight into the Molecular Genetics of Neurodevelopmental Disorders

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Message from the Guest Editors

Neurodevelopmental disorders (NDDs) are a group of rare diseases mainly of genetic origin, resulting from abnormal brain development and characterized by impaired cognition, communication, adaptive functioning deficits, and impaired psychomotor skills. NDDs which include rare genetic syndromes, intellectual disability (ID), autism spectrum disorder (ASD) and epilepsy, have proven to be genetically and phenotypically heterogeneous raising unexpected difficulties to the phenotype-driven approach to diagnosis. The advances in genetic testing achieved through Next Generation Sequencing (NGS) have shown an enormous improvement in discovering genes associated with NDDs. However, interpreting the pathogenetic role of variants of unknown significance requires rigorous evaluation of multiple lines of evidence, including the functional effect of the variants at the protein, cellular, or model organism levels. With a better understanding of the molecular pathomechanisms and phenotypic spectra, patient management can be improved and targeted therapeutics may become more available.

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