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Diagnosis of Neurogenetic Disorders: Contribution of Next Generation Sequencing and Deep Phenotyping

Guest Editor:

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Message from the Guest Editor

The contribution of genomic variants to the aetiopathogenesis of both paediatric and adult neurological disease is increasingly recognised. The use of next generation sequencing has led to the discovery of novel neurodevelopmental disorders, and provided insight into the aetiopathogenesis of common adult neurological diseases. Correctly classifying the pathogenicity of genomic variants from amongst the large number of variants identified by next generation sequencing is recognised as perhaps the major challenge facing the field. Deep phenotyping techniques can aid variant interpretation by correctly classifying individuals as affected or unaffected for segregation studies. The lack of information on the clinical phenotype of novel genetic subtypes of neurological disease creates limitations for Genetic Counselling. Both deep phenotyping and qualitative studies can capture the clinical and patient's perspective on a disease and provide valuable information.

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Message from the Editor-in-Chief

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