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Early Diagnosis Research of Inherited Neuropathies

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Message from the Guest Editors

The diagnosis of inherited neuropathies has been largely improved in the last decade. However, numerous patients are still undiagnosed, inducing a lack of adapted clinical care without therapeutic hopes. While single-nucleotide variants and small indel mutations are now easily detected, structural variations, for instance, are still often underdiagnosed. In this issue, we would like to report new technologies, but also new bioinformatics tools developed recently and verified experimentally, to improve the diagnosis of inherited neuropathies. Structural variations, which are new kinds of mutations currently described to be responsible for neuropathies, could be an interesting aspect of this issue. Articles presenting new strategies supported by experimental results or preclinical applications will be considered. This Special Issue will help the diagnosis of neuropathic patients to be improved, and we believe it could serve as a reference for all inherited diseases.



