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Genetics of Mitochondrial Diseases: State of the Art

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Deadline for manuscript submissions:
closed (30 September 2022)

Message from the Guest Editor

Mitochondrial diseases (MDs) have a minimum prevalence of around 1:5000. In pediatric patients, these pathologies are typically devastating with a poor prognosis. MDs are genetically and clinically heterogeneous, which presents an important hindrance to their clinical diagnosis, understanding and treatment. To date, more than 330 nuclear-encoded and most mtDNA-encoded genes have been implicated in these pathologies. The number of new disease genes has increased dramatically with the wider use of high thruput sequencing, and its recent integration with other omics approaches will undoubtedly lead to a further increase in the diagnostic yield for patients with MDs.

This special issue welcomes review and research articles addressing recent advances in the field of mitochondrial medicine from diagnosis to new treatment strategies; articles reporting on novel mitochondrial disease genes and animal models expanding our understanding of mitochondrial diseases will also be considered.

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Message from the Editor-in-Chief

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