Special Issue

Molecular Mechanisms, Pathophysiology and Phenotypes of Mitochondrial Disorders

Message from the Guest Editor

Mitochondrial diseases are the most common inheritable metabolic diseases resulting from defects in oxidative phosphorylation. They are caused by mutations of nuclear or mitochondrial DNA in genes involved in mitochondrial function. While some mitochondrial disorders only affect a single organ (e.g., the eye in Leber hereditary optic neuropathy), many involve multiple organ systems and often present with prominent neurologic features. Understanding the phenotypic diversity and elucidating the molecular mechanisms at the basis of these diseases has, however, proved challenging. With this Special Issue, we intent to explore the molecular basis, the clinical spectrum, the diagnostic approach and the treatment advances of these devastating disorders.

Guest Editor

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