Mitochondrial medicine: the future is now

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The mitochondrial cytopathies are complex disorders that show phenotypic and genetic heterogeneity. Over the last decade, advances in mitochondrial research has earned it a "seat at the table" and its own specialty of medicine: "mitochondrial medicine". Considered the "powerhouses of the cell" and the major driver of cellular ATP, we can now appreciate the expanded role of the mitochondria in health and disease as an evolving and multidisciplinary area of research\[1,2\]. The scope of inherited and genetic mitochondrial disorders ranges from infancy onset global disease to organ specific disorders in adults such as myopathy and cardiac and liver disorders\[3\]. Secondary mitochondrial dysfunction has also been shown to play a role in many adult neurodegenerative conditions\[4\].

This Special Issue about mitochondrial medicine focuses on the complexities of clinical phenotypes, diagnosis, genetics, genomics, and pharmacogenomics which may drive individualized and personalized treatment. Although genomic testing is standard in terms of the clinical panels that are available, the field is still expanding\[5\]. Low heteroplasmy in accessible tissues and variants of unknown significance still require functional studies to clarify their meaning and contribution to the phenotype and/or biochemical alterations in patients\[6\].

In relation to disease, the emerging bioenergetic mitochondrial model suggests that mitochondrial defects contribute to the development of age-, stage-, and stress-related diseases by altering complex cellular and physiological functions\[7\].

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DECLARATIONS

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REFERENCES