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Central nervous system involvement in mitochondrial disorders

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Statement of the Problem: Central nervous system (CNS) disease is increasingly recognized as a manifestation of mitochondrial disorders (MIDs). However, the broad range of clinical CNS manifestation is still underestimated. This review aims at summarizing and discussing previous and recent findings concerning the cerebral manifestations of MIDs.

Methodology & Theoretical Orientation: MIDs literature review was conducted.

Findings: MIDs frequently present as mitochondrial multiorgan disorder syndrome (MIMODS) either already at onset or later in the course. After the muscle, the brain is the second most organ which is frequently affected in MIMODS. Cerebral manifestations of MIDs are variable and may present with or without a lesion on imaging or functional studies but there can be imaging/functional lesions without clinical manifestations. The most well-known cerebral manifestations of MIDs include stroke-like episodes, epilepsy, headache, ataxia, movement disorders, hypopituitarism, muscle weakness, psychiatric abnormalities, nystagmus, white and grey matter lesions, atrophy, basal ganglia calcification, and hypometabolism on FDG-PET. For most of the MIDs only symptomatic therapy is currently available. Symptomatic treatment should be supplemented by vitamins, co-factors, and antioxidants.

Conclusion & Significance: Cerebral manifestations of MIDs need to be recognized and appropriately managed since they strongly determine the outcome of MID patients.

Biography

Josef Finsterer received his MD and is a Professor of Neurology from the University of Vienna, Austria. Since his training as a Clinical Neurologist and Electrophysiologist at the Neurological Krankenhaus Rosenhuegel and the Ludwig Boltzmann Institute for Epilepsy and Neuromuscular Disorders, he is involved in the management of neuromuscular disorders, particularly muscular dystrophies and metabolic myopathies. In addition to neuromuscular disorders, research interests focus on genetics, orphan diseases, and cardiac involvement in genetic conditions.

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