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Brain imaging in adult mitochondrial disorders

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Since the central-nervous-system (CNS) is the second most frequently affected organ in mitochondrial disorders (MIDs) and since MIDs are increasingly recognized, it is important to know about the morphological CNS abnormalities on imaging in these patients. The study aims at summarizing and discussing current knowledge and recent advances concerning CNS imaging abnormalities in adult MIDs. The most relevant CNS abnormalities in adult MIDs on imaging include white and grey matter lesions, stroke-like lesions as the morphological equivalent of stroke-like episodes, cerebral atrophy, calcifications, optic atrophy, and lactic acidosis. Since these CNS lesions may go along with or without clinical manifestations, it is important to screen all MID patients for cerebral involvement. Some of these lesions may remain unchanged for years whereas others may be dynamic, either in the sense of progression or regress. Typical dynamic lesions are stroke-like lesions and grey matter lesions. Clinically relevant imaging techniques for visualization of CNS abnormalities in MIDs are the computed tomography, magnetic-resonance-imaging, MR-spectroscopy, SPECT, PET, and angiography. CNS imaging in adult MIDs is important for diagnosing and monitoring CNS involvement. It also contributes to the understanding of the underlying pathomechanisms that lead to CNS involvement in MIDs.

Biography

Josef Finsterer received his MD and he is a Prof. of Neurology, from the University of Vienna, Austria. Since his training as a Clinical Neurologist and Electro physiologist at the Neurological Krankenhaus Rosenhügel 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, his research interests focus on genetics, orphan diseases, and cardiac involvement in genetic conditions.

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