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BRAIN IMAGING IN PEDIATRIC MITOCHONDRIAL DISORDERS

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Objectives: Since the central-nervous-system (CNS) is the second most frequently affected organ in mitochondrial disorders (MIDs) and since pediatric MIDs are increasingly recognized, it is important to know about the morphological CNS abnormalities on imaging in these patients. The review aims at summarizing and discussing current knowledge and recent advances concerning CNS imaging abnormalities in pediatric MIDs.

Methods: Systematic literature review.

Results: The most relevant CNS abnormalities in pediatric 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 lactacidosis. 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 pediatric MIDs are the computed tomography, magnetic-resonance-imaging, MR-spectroscopy, SPECT, PET, and angiography.

Conclusions: CNS imaging in pediatric 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 has graduated at the University of Vienna, specialisation in Neurology, Habilitation in Neurology about automated EMG analysis. Having expertise and specialisation in the field of neuromuscular disorders and its multisystem implications, he is currently working as a Neurologist in the General Hospital Rudolfstiftung, Vienna and is giving lectures about neurological aspects in comparative medicine at the Messerli Institute of the Veterinary University of Vienna.

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