OPA1-related disorders: Diversity of clinical expression, modes of inheritance and pathophysiology

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Résumé en anglais	Mutations in the Optic Atrophy 1 gene (OPA1) were first identified in 2000 as the main cause of Dominant Optic Atrophy, a disease specifically affecting the retinal ganglion cells and the optic nerve. Since then, an increasing number of symptoms involving the central, peripheral and autonomous nervous systems, with considerable variations of age of onset and severity, have been reported in OPA1 patients. This variety of phenotypes is attributed to differences in the effects of OPA1 mutations, to the mode of inheritance, which may be mono- or bi-allelic, and eventually to somatic mitochondrial DNA mutations. The diversity of the pathophysiological mechanisms involved in OPA1-related disorders is linked to the crucial role played by OPA1 in the maintenance of mitochondrial structure, genome and function. The neurological expression of these disorders highlights the importance of mitochondrial dynamics in neuronal processes such as dendritogenesis, axonal transport, and neuronal survival. Thus, OPA1-related disorders may serve as a paradigm in the wider context of neurodegenerative syndromes, particularly for the development of novel therapeutic strategies against these diseases.
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