OPA1-associated disorders: Phenotypes and pathophysiology

Titre	OPA1-associated disorders: Phenotypes and pathophysiology
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Résumé en anglais	The OPA1 gene, encoding a dynamin-like mitochondrial GTPase, is involved in autosomal dominant optic atrophy (ADOA, OMIM #165500). ADOA, also known as Kjer's optic atrophy, affects retinal ganglion cells and the axons forming the optic nerve, leading to progressive visual loss. OPA1 gene sequencing in patients with hereditary optic neuropathies indicates that the clinical spectrum of ADOA is larger than previously thought. Specific OPA1 mutations are responsible for several distinct clinical presentations, such as ADOA with deafness (ADOAD), and severe multi-systemic syndromes, the so-called “ADOA plus” disorders, which involve neurological and neuromuscular symptoms similar to those due to mitochondrial oxidative phosphorylation defects or mitochondrial DNA instability. The study of the various clinical presentations of ADOA in conjunction with the investigation of OPA1 mutations in fibroblasts from patients with optic atrophy provides new insights into the pathophysiological mechanisms of the disease while underscoring the multiple physiological roles played by OPA1 in energetic metabolism, mitochondrial structure and maintenance, and cell death. Finally, OPA1 represents an important new paradigm for emerging neurodegenerative diseases affecting mitochondrial structure, plasticity and functions.
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Titre abrégé	OPA1-associated disorders

Liens
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