Reversible optic neuropathy with OPA1 exon 5b mutation

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Titre	Reversible optic neuropathy with OPA1 exon 5b mutation
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Résumé en anglais	A new c.740G>A (R247H) mutation in OPA1 alternate spliced exon 5b was found in a patient presenting with bilateral optic neuropathy followed by partial, spontaneous visual recovery. R247H fibroblasts from the patient and his unaffected father presented unusual highly tubular mitochondrial network, significant increased susceptibility to apoptosis, oxidative phosphorylation uncoupling, and altered OPA1 protein profile, supporting the pathogenicity of this mutation. These results suggest that the clinical spectrum of the OPA1-associated optic neuropathies may be larger than previously described, and that spontaneous recovery may occur in cases harboring an exon 5b mutation. Ann Neurol 2008
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