



Hereditary optic neuropathies share a common mitochondrial coupling defect

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Résumé en anglais	<p>Hereditary optic neuropathies are heterogeneous diseases characterized by the degeneration of retinal ganglion cells leading to optic nerve atrophy and impairment of central vision. We found a common coupling defect of oxidative phosphorylation in fibroblasts of patients affected by autosomal dominant optic atrophy (mutations of OPA1), autosomal dominant optic atrophy associated with cataract (mutations of OPA3), and Leber's hereditary optic neuropathy, a disorder associated with point mutations of mitochondrial DNA complex I genes. Interestingly, the energetic defect was significantly more pronounced in Leber's hereditary optic neuropathy and autosomal dominant optic atrophy patients with a more complex phenotype, the so-called plus phenotype. <i>Ann Neurol</i> 2008</p>
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