Multiple Sclerosis–Like Disorder in Opa1-Related Autosomal Dominant Optic Atrophy

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Résumé en anglais	Autosomal dominant optic atrophy (ADOA) is a progressive ophthalmologic disorder caused in two-thirds of the cases by a mutation in the optic atrophhy 1 (IPA1) gene, a nuclear gene encoding a mitochondrial protein. We report a patient in whom an OPA1 mutation was responsible for a bilateral optic atrophy associated with multiple sclerosis-like (MSL) features. In addition, biochemical studies performed on fibroblasts from this patient showed a significant mitochondrial coupling defect associated with reduced ATP production and respiratory function in comparison to controls.
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