Molecular screening of 980 cases of suspected hereditary optic neuropathy with a report on 77 novel OPA1 mutations

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Résumé en anglais	We report the results of molecular screening in 980 patients carried out as part of their work-up for suspected hereditary optic neuropathies. All the patients were investigated for Leber's hereditary optic neuropathy (LHON) and autosomal dominant optic atrophy (ADOA), by searching for the ten primary LHON-causing mtDNA mutations and examining the entire coding sequences of the OPA1 and OPA3 genes, the two genes currently identified in ADOA. Molecular defects were identified in 440 patients (45% of screened patients). Among these, 295 patients (67%) had an OPA1 mutation, 131 patients (30%) had an mtDNA mutation, and 14 patients (3%), belonging to three unrelated families, had an OPA3 mutation. Interestingly, OPA1 mutations were found in 157 (40%) of the 392 apparently sporadic cases of optic atrophy. The eOPA1 locus-specific database now contains a total of 204 OPA1 mutations, including 77 novel OPA1 mutations reported here. The statistical analysis of this large set of mutations has led us to propose a diagnostic strategy that should help with the molecular work-up of optic neuropathies. Our results highlight the importance of investigating LHON-causing mtDNA mutations as well as OPA1 and OPA3 mutations in cases of suspected hereditary optic neuropathy, even in absence of a family history of the disease. © 2009 Wiley-Liss, Inc.
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