Recessive Mutations in RTN4IP1 Cause Isolated and Syndromic Optic Neuropathies

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Résumé en anglais	Autosomal-recessive optic neuropathies are rare blinding conditions related to retinal ganglion cell (RGC) and optic-nerve degeneration, for which only mutations in TMEM126A and ACO2 are known. In four families with early-onset recessive optic neuropathy, we identified mutations in RTN4IP1, which encodes a mitochondrial ubiquinol oxydo-reductase. RTN4IP1 is a partner of RTN4 (also known as NOGO), and its ortholog Rad8 in C. elegans is involved in UV light response. Analysis of fibroblasts from affected individuals with a RTN4IP1 mutation showed loss of the altered protein, a deficit of mitochondrial respiratory complex I and IV activities, and increased susceptibility to UV light. Silencing of RTN4IP1 altered the number and morphogenesis of mouse RGC dendrites in vitro and the eye size, neuro-retinal development, and swimming behavior in zebrafish in vivo. Altogether, these data point to a pathophysiological mechanism responsible for RGC early degeneration and optic neuropathy and linking RTN4IP1 functions to mitochondrial physiology, response to UV light, and dendrite growth during eye maturation.
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