Genetically determined optic neuropathies

Titre	Genetically determined optic neuropathies
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Auteur	Milea, Dan [1], Amati-Bonneau, Patrizia [2], Reynier, Pascal [3], Bonneau, Dominique [4]
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Résumé en anglais	Purpose of review: The present review focuses on recent advances in the knowledge of hereditary optic neuropathies resulting from retinal ganglion cell degeneration, mostly due to mitochondrial dysfunctions.Recent findings: Autosomal dominant optic atrophy, the most common hereditary optic neuropathy, appears to have a more variable clinical presentation than previously thought. Acute visual loss, reversible visual loss, or visual loss associated with extraocular symptoms (deafness, extraocular ophthalmoplegia, multiple sclerosis-like disease) are infrequent, though possible presentations. In Leber's hereditary optic neuropathy, recent findings suggest that the large variability of the clinical expression could be modulated by several factors: genetic (downregulation of the OPA1 gene), environmental (smoking), and anatomic (predisposition of the optic nerve head to axonal loss). Globally, hereditary optic neuropathies may represent an underestimated cause of unexplained and sporadic optic atrophy. Recent advances, such as the discovery of a new gene involved in autosomal recessive optic neuropathy, reinforce the central role played by mitochondrial dysfunctions in the pathogenesis of optic neuropathies. Summary: Hereditary optic neuropathies may have a heterogenous presentation and serve as a paradigm for neurodegenerative diseases affecting mitochondrial structure, plasticity, and function.
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Liens

[1] http://okina.univ-angers.fr/d.milea/publications

[2] http://okina.univ-angers.fr/patrizia.bonneau/publications
[3] http://okina.univ-angers.fr/pascal.reynier/publications
[4] http://okina.univ-angers.fr/d.bonneau/publications
[5] http://okina.univ-angers.fr/publications/ua316
[6] http://dx.doi.org/10.1097/WCO.0b013e3283347b27