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A novel mutation of AFG3L2 might cause dominant optic atrophy in patients with mild intellectual disability

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Titre	A novel mutation of AFG3L2 might cause dominant optic atrophy in patients with mild intellectual disability
Type de publication	Article de revue
Auteur	Charif, Majida [1], Roubertie, Agathe [2], Salime, Sara [3], Mamouni, Sonia [4], Goizet, Cyril [5], Hamel, Christian [6], Lenaers, Guy [7]
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Mots-clés	AFG3L2 [8], dominant mutation [9], OPA1 [10], Optic nerve [11], retinal ganglion neurons [12]
Résumé en anglais	Dominant optic neuropathies causing fiber loss in the optic nerve are among the most frequent inherited mitochondrial diseases. In most genetically resolved cases, the disease is associated to a mutation in OPA1, which encodes an inner mitochondrial dynamin involved in network fusion, cristae structure and mitochondrial genome maintenance. OPA1 cleavage is regulated by two m-AAA proteases, SPG7 and AFG3L2, which are, respectively involved in Spastic Paraplegia 7 and Spino-Cerebellar Ataxia 28. Here, we identified a novel mutation c.1402C>T in AFG3L2, modifying the arginine 468 in cysteine in an evolutionary highly conserved arginine-finger motif, in a family with optic atrophy and mild intellectual disability. Ophthalmic examinations disclosed a loss of retinal nerve fibers on the temporal and nasal sides of the optic disk and a red-green dyschromatopsia. Thus, our results suggest that neuro-ophthalmological symptom as optic atrophy might be associated with AFG3L2 mutations, and should prompt the screening of this gene in patients with isolated and syndromic inherited optic neuropathies.
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