Novel gene mutation in an atypical late-onset mitochondrial form of multifocal dystonia

Titre	Novel gene mutation in an atypical late-onset mitochondrial form of multifocal dystonia
Type de publication	Article de revue
Auteur	Bris, Céline [1], Rouaud, Tiphaine [2], Desquiret-Dumas, Valérie [3], Gueguen, Naïg [4], Goudenège, David [5], Barth, Magalie [6], Bonneau, Dominique [7], Amati-Bonneau, Patrizia [8], Lenaers, Guy [9], Reynier, Pascal [10], Lebre, Anne-Sophie [11], Procaccio, Vincent [12]
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Résumé en anglais	Mitochondrial complex I, the largest component of the mitochondrial respiratory chain, comprises 44 subunits of which 7 are encoded by the mitochondrial genome and the remainder by the nuclear genome. Isolated complex I deficiencies represent a major contribution within the group of respiratory chain defects. We report an atypical case carrying a homozygous NDUFS4 missense mutation, with late-onset multifocal dystonia, in contrast to expected clinical phenotypes due to other NDUFS4 mutations, which have been constantly reported to be responsible for Leigh syndrome of early onset and death.
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