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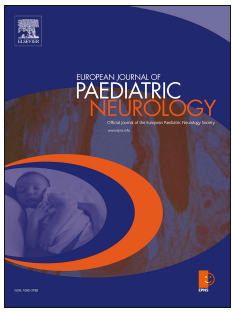

A novel homozygous MFN2 mutation associated with severe and atypical CMT2 phenotype

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A novel homozygous MFN2 mutation associated with severe and atypical CMT2 phenotype

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Abbreviations: CMT, Charcot-Marie-Tooth disease; CMT2A, Charcot-Marie-Tooth type 2A; MFN2, mitofusin 2; NCV, nerve conduction velocity; EMG, electromyography; CMAP, compound motor action potential; CTDP1, congenital cataracts facial dysmorphism; CCFDN, congenital cataracts facial dysmorphism and neuropathy; SIL1, Marinesco-Sjögren syndrome.