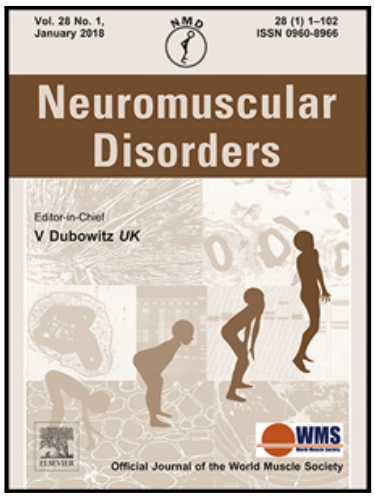

A Novel AIFM1 Mutation in a Chinese Family with X-linked Charcot-Marie-Tooth disease type 4

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Highlights

- This is the first reported CMTX4 case in China.
- A novel missense mutation (c.513G>A, p.Met171Ile) was identified in *AIFM1* gene.
- Chracteristic calf MRI finding might be a feature of CMTX4 in early stage.
- Abnormal mitochondrial morphology and accumulation is a pathological feature.