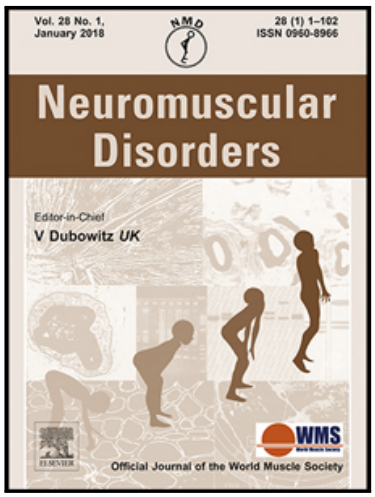

A recurrent de novo DYNC1H1 tail domain mutation causes spinal muscular atrophy with lower extremity predominance, learning difficulties and mild brain abnormality

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HIGHLIGHTS

- De novo missense mutation c.751C>T in DYNC1H1 gene is a pathogenic mutation causing SMALED
- The mutation has a high phenotypic-genotypic correlation in 4 unrelated patients
- Leg muscle MRI findings are highly specific in DYNC1H1-related SMALED
- Muscle biopsy findings are variable and non-specific so has limited use in DYNC1H1-related SMALED