

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

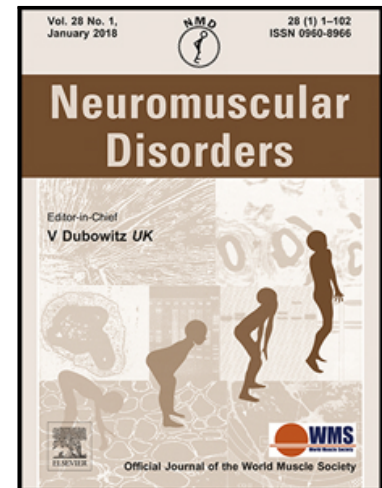
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PII: S0960-8966(18)30025-7
DOI: [10.1016/j.nmd.2018.07.002](https://doi.org/10.1016/j.nmd.2018.07.002)
Reference: NMD 3581

To appear in: *Neuromuscular Disorders*

Received date: 11 January 2018
Revised date: 24 June 2018
Accepted date: 13 July 2018

Please cite this article as: Sophelia Hoi-Shan Chan , Nens van Alfen , Inger Johanne Thuestad , Janice Ip , Angel On-Kei Chan , Christopher Mak , Brian Hon-Yin Chung , Aad Verrips , Erik-Jan Kamsteeg , A recurrent de novo DYNC1H1 tail domain mutation causes spinal muscular atrophy with lower extremity predominance, learning difficulties and mild brain abnormality, *Neuromuscular Disorders* (2018), doi: [10.1016/j.nmd.2018.07.002](https://doi.org/10.1016/j.nmd.2018.07.002)



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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

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