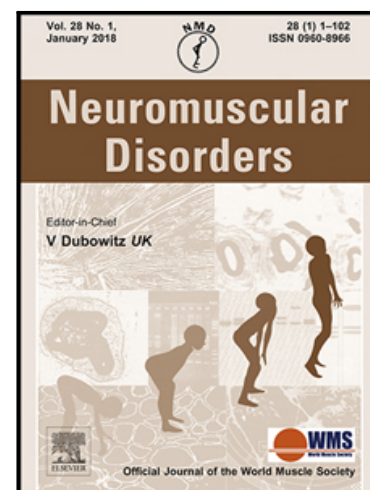


ECEL1 gene related contractural syndrome: long-term follow-up and update on clinical and pathological aspects

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Highlights:

- Recessive mutations in the *ECEL1* gene cause heterogeneous contractural phenotypes
- We describe 7 novel patients from 4 unrelated families with *ECEL1* gene mutations
- We provide long-term clinical follow-up for 5 patients with *ECEL1* gene mutations
- We observed myopathic features, core-like areas and nCAM positive fibres on histopathology
- We provide novel pathomechanical insights into *ECEL1* gene related conditions

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