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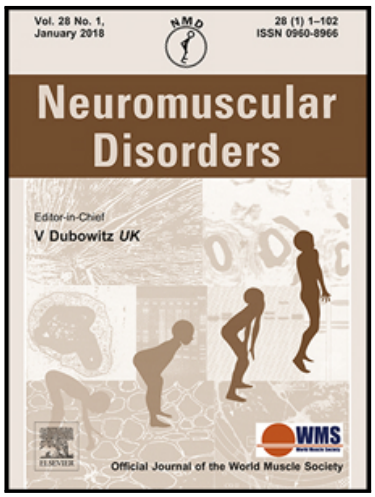

A new severe mutation in the SLC5A7 gene related to congenital myasthenic syndrome type 20

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HIGHLIGHTS:

- New case of congenital myasthenic syndrome type 20. Only 7 patients published.
- New mutations not described previously,
- Very severe form of disease, and poor response to therapy diagnosis achieved early by repetitive stimulation.