Toe-extension myotonia in myotonic dystrophy type 1

Myotonia is a cardinal symptom of adult-onset myotonic dystrophy type 1 (DM1). Myotonia, delayed relaxation following contraction, is most prominent in the hand and forearm muscles. Two sisters with genetically confirmed DM1 exhibited toe-extension myotonia (video on the *Neurology*® Web site at Neurology.org) in contrast to toe-flexion myotonia that can occur in DM1. There was a typical warm-up phenomenon. A variant phenotype of Charcot-Marie-Tooth-like polyneuropathy¹ was not present. Mutations in the *CLCN1* gene encoding the skeletal muscle chloride channel, in addition to abnormal splicing, can enhance myotonia in DM type 2.² However, in our patients sequencing of the *CLCN1* gene did not show mutations.

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Supplemental data
at Neurology.org

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