A 48-year-old man presented with episodic paraplegia and stiffness of hands, face, and tongue, along with eyelid myotonia when exposed to cold temperature, which he had since childhood. Eyelid myotonia was evoked either by exposure to cold weather (video on the Neurology® Web site at www.neurology.org and figure 1) or by forceful eye closure. Myotonia was elicited with percussion of the tongue and thenar eminence. EMG showed myotonic discharge at room temperature (figure 2). Creatine kinase was mildly elevated to 293 U/L. The genetic study showed missense mutation (R1448C) in the voltage-gated sodium channel, type IV, alpha subunit (SCN4A). The patient’s daughter had the same genetic mutation and similar symptoms.
Teaching Video NeuroImages: Cold-induced eyelid myotonia
Yi-Jen Wu and Chou-Ching Lin
Neurology 2013;80:e81
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