Teaching Video NeuroImage: Carbamazepine Improves Gait Initiation in Autosomal Recessive Myotonia Congenita

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A 60-year-old man presented with muscle stiffness. He had tended to fall since childhood. His parents were first cousins, and his relatives had no symptoms. Examination showed myotonia in ocular, hand, and limb muscles, followed by improvement with repeated activity (warm-up phenomenon; Video 1). Cold exposure did not aggravate myotonia. His lower legs showed muscle hypertrophy. Genetic tests showed a homozygous p.M560T mutation in the *CLCN1* gene, which encodes skeletal muscle chloride channel 1, consistent with autosomal recessive myotonia congenita (Becker disease).[1] Although treatment with mexiletine or phenytoin did not improve his symptoms, carbamazepine (350 mg/d) did, especially the walking disturbance (Video 2). Although the warm-up phenomenon is sometimes also observed in sodium channel myotonia,[2] it is characteristic of myotonia congenita and an important clue for the diagnosis.

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Name	Location	Contribution
Yasuhiro Fuseya, MD, PhD	Department of Molecular and Cellular Physiology, Kyoto University, Kyoto, Japan	Managed the patient and drafted the manuscript and video
Nana Ishikawa, MD	Akahoshi Neurology Clinic, Osaka, Japan	Managed the patient
Ryogen Sasaki MD, PhD	Department of Neurology, Kuwana City Medical Center, Mie, Japan	Performed a genetic test
Hirofumi Yamashita, MD, PhD	Department of Neurology, Japanese Red Cross Wakayama Medical Center, Wakayama, Japan	Managed the patient and drafted the manuscript and video

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