

A 60-year-old man with myotonia during gait initiation

Teaching Video NeuroImages

Neurology

Resident & Fellow Section



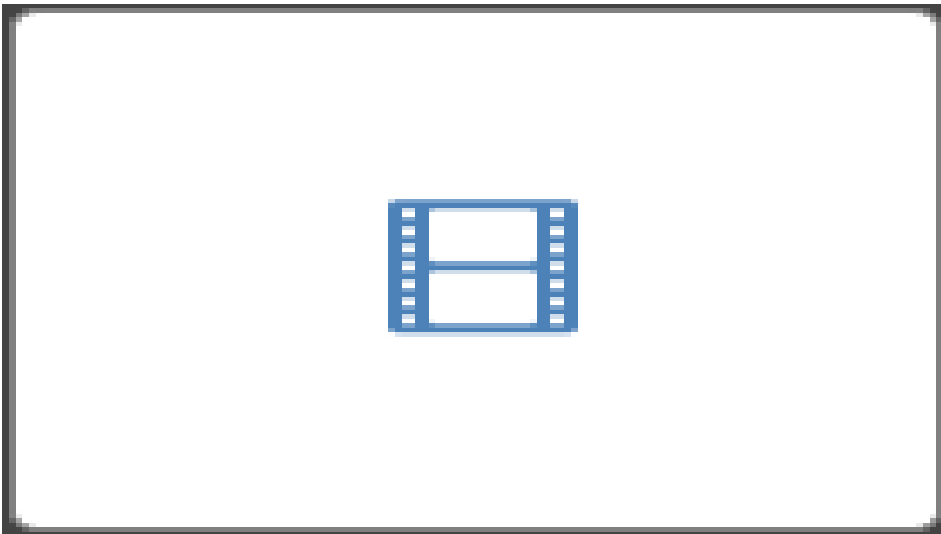
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Vignette

- A 60-year-old man presented with muscle stiffness.
- He had tended to fall since childhood, and his parents were first cousins.
- Examination showed myotonia in ocular, hand, and limb muscles, followed by improvement with repeated activity.
- Cold exposure did not aggravate myotonia.
- His lower legs showed muscle hypertrophy.

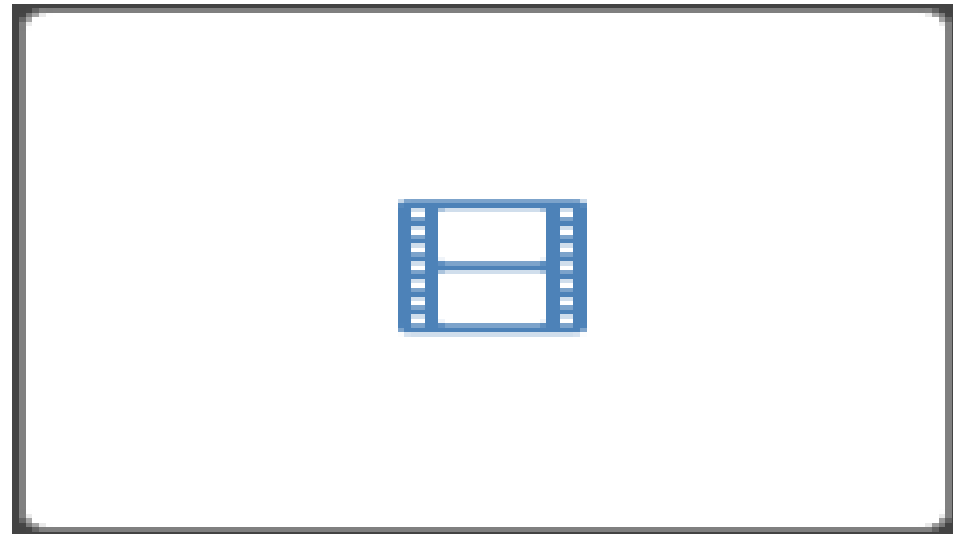
Video

Video 1



carbamazepine(-)

Video 2



carbamazepine(+)

Carbamazepine improves gait initiation in autosomal recessive myotonia congenita

- Genetic tests showed a homozygous c.1679 T>C (p.M560T) variation in the *CLCN1* gene, consistent with autosomal recessive myotonia congenita (Becker disease)¹.
- Carbamazepine (350 mg/day) improved the myotonia, especially the walking disturbance. This suggests that myotonia may improve with medications that block voltage-gated sodium channels.
- Though the warm-up phenomenon is sometimes also observed in sodium channel myotonia²—in which cold exposure often worsens myotonia—it is characteristic of myotonia congenita and is an important clue for the diagnosis.



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