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

Author(s):
Yasuhiro Fuseya, Doctor of medicine degree\textsuperscript{1,2}; Nana Ishikawa, Doctor of medicine degree\textsuperscript{3}; Ryogen Sasaki, Doctor of medicine degree\textsuperscript{4}; Hirofumi Yamashita, Doctor of medicine degree\textsuperscript{5}

Neurology\textsuperscript{®} Published Ahead of Print articles have been peer reviewed and accepted for publication.

This manuscript will be published in its final form after copyediting, page composition, and review of proofs. Errors that could affect the content may be corrected during these processes.
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). Although treatment with mexiletine or phenytoin did not improve his symptoms, carbamazepine (350 mg/day) did, especially the walking disturbance (Video 2). Whereas the warm-up phenomenon is sometimes also observed in sodium channel myotonia, it is characteristic of myotonia congenita and an important clue for the diagnosis.
## Appendix 1. Authors

<table>
<thead>
<tr>
<th>Name</th>
<th>Location</th>
<th>Contribution</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yasuhiro Fuseya MD, PhD</td>
<td>Department of Molecular and Cellular Physiology, Kyoto University, Kyoto, Japan</td>
<td>managed the patient, drafted the manuscript and video</td>
</tr>
<tr>
<td>Nana Ishikawa MD</td>
<td>Akahoshibi Neurology Clinic, Osaka, Japan</td>
<td>managed the patient</td>
</tr>
<tr>
<td>Ryogen Sasaki MD, PhD</td>
<td>Department of Neurology, Kuwana City Medical Center, Mie, Japan</td>
<td>performed a genetic test</td>
</tr>
<tr>
<td>Hirofumi Yamashita MD, PhD</td>
<td>Department of Neurology, Japanese Red Cross Wakayama Medical Center, Wakayama, Japan</td>
<td>managed the patient, drafted the manuscript and video</td>
</tr>
</tbody>
</table>


REFERENCES


Video legends

Video 1 Carbamazepine(-): The patient has myotonia during gait initiation, with the warm-up phenomenon and also grip and eyelid myotonia.

Video 2 Carbamazepine(+): The myotonia during gait initiation improves dramatically with carbamazepine, and it suggests that myotonia may improve with medications that block voltage-gated sodium channels. Muscle hypertrophy is observed in his lower legs (latter part).
Teaching Video NeuroImage: Carbamazepine Improves Gait Initiation in Autosomal Recessive Myotonia Congenita
Yasuhiro Fuseya, Nana Ishikawa, Ryogen Sasaki, et al.
Neurology published online October 21, 2021
DOI 10.1212/WNL.0000000000013010

This information is current as of October 21, 2021