Congenital Paramyotonia
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A 22-year-old woman with family history of muscle myotonia presented with a 15-year history of difficulty opening eyes and grip-release myotonia in both hands. Physical examination showed typical percussion myotonia of the tongue, thenar eminence, and grip-release myotonia (Figure 1). Patient had myotonic discharges and myopathic low-amplitude and short duration motor-unit potentials on EMG. CMAP amplitude decreased significantly after short/long time exercise and cold tests. Whole-exome sequencing identified the patient with a heterozygous mutation in SCN4A gene (c.4765 G>A; p.V1589M), a finding that was consistent with a diagnosis of congenital paramyotonia.¹ Treatment with lamotrigine, and the patient was slight abatement of muscle myotonia.² Congenital paramyotonia is a rare genetic neuromuscular disorder characterized by muscles difficulty relax after contraction,
leading to muscle stiffness and loss of control during movements, particularly more
easily triggered in cold environments and after exercise. The percussion myotonia of
the tongue, thenar eminence, and grip-release myotonia can’t be ignored, which are
useful signs for clinician in diagnosis and differential diagnosis.

Photographs and text from: Chunyan Cao and Ganqin Du, Department of Neurology,
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References
autosomal dominant myotonia associated with the V1589M missense mutation in the muscle
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Figure 1. (A) Myotonia of the Tongue. (B) Thenar eminence myotonia. (C) Grip-release myotonia. (D) Sanger sequencing: SCN4A(c.4765G>A, p.V1589M).

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