Teaching Video NeuroImages: Shake Mom's hand to get the diagnosis

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Figure Inverted V-shaped (tented/fish-shaped) upper lip characteristic of facial weakness in congenital myotonic dystrophy

A term male newborn was admitted to the neonatal intensive care unit for respiratory distress and feeding difficulty. His 19-year-old mother was apparently healthy. The baby had generalized hypotonia, weakness, a weak cry, and a tented upper lip (figure). Maternal examination facilitated diagnosis (videos 1 and 2 on the *Neurology*[®] Web site at www.neurology.org). Genetic testing identified >99 CTG repeats, confirming myotonic dystrophy type 1 (DM1), a trinucleotide repeat disorder involving the *DMPK* gene.¹ Congenital DM1, usually maternally transmitted,² presents antenatally with polyhydramnios and reduced fetal movements. Postnatally it presents with generalized weakness, hypotonia, respiratory compromise, and feeding difficulties.¹ The inverted V-shaped upper lip

represents facial weakness. Because myotonia is absent in newborns, examination of the mother is diagnostic.

AUTHOR CONTRIBUTIONS

Dr. Ghosh collected and organized the data and wrote the first manuscript (including the first draft). Dr. Moodley conceptualized the study, verified the results, and revised the manuscript at all stages. Dr. Indulkar verified the results and revised the manuscript at all stages.

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Supplemental data at
www.neurology.org

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Disclosure: The authors report no disclosures.

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Neurology 2011;77:e114

DOI 10.1212/WNL.0b013e318236ef77

This information is current as of November 7, 2011

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