



Teaching Video *NeuroImages*: Gelastic cataplexy as the first neurologic manifestation of Niemann-Pick disease type C



José Luiz Pedroso, MD,
PhD
Eduardo Ferracioli Fusão,
MD
Carol Ladeia-Frota, MD
Juliana Harumi Arita,
MD
Orlando G. Barsottini,
MD, PhD
Marcelo Rodrigues
Masruha, MD, PhD
Luiz Celso Pereira
Vilanova, MD, PhD

A 10-year-old boy presented to our hospital with a 3-year history of fall attacks triggered by laughing, leading to a generalized loss of muscle tone without loss of consciousness (video). One year later, motor delayed skills started. Examination showed ataxia, moderate cognitive impairment, and vertical gaze palsy. EEG revealed diffuse slowing and disorganization of background rhythms. Molecular analysis disclosed heterozygosity p.P1007A and p.A1035V mutations, diagnostic of Niemann-Pick disease type C (NPC).

NPC is a lysosomal storage disorder caused by intracellular accumulation of cholesterol and glycosphingolipids. Neurologic manifestations include ataxia,

dementia, seizures, and supranuclear vertical gaze palsy.¹ Gelastic cataplexy is rarely described as the first neurologic symptom.² Children with very frequent cataplexy should be tested early for NPC.

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Correspondence & reprint
requests to Dr. Pedroso:
jlpedroso.neuro@gmail.com

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From the Department of Neurology, Universidade Federal de São Paulo, Brazil.

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