Teaching NeuroImages: When the Teeth are the Clue to the Etiology of an Epileptic Encephalopathy

Victor Hugo Pantoja Leão, MD, Marcelo de Melo Aragão, MD, MSc, Ricardo Silva Pinho, MD, PhD, and Marcelo Rodrigues Masruha, MD, PhD

Neurology® 2021;96:e157-e158. doi:10.1212/WNL.0000000000010758

Correspondence
Dr. Masruha
marcelomasruha@gmail.com

Figure Kohlschutter-Tönz Syndrome

(A) Facial gestalt showed hypotonic impression and divergent strabismus, without overt dysmorphic features. (B) Oral examination revealed generalized yellowish-brown discoloration and pitting of all the surfaces of the teeth, suggestive of enamel hypoplasia.

A 24-year-old woman with severe intellectual disability presented with refractory epilepsy since the first day of life. She had global developmental delay and remarkable abnormalities in both primary and secondary teeth (figure). Brain MRI was normal. A whole exome sequencing revealed *SLC13A5* compound heterozygous mutations C>T at chr17:6.606.350, p.Gly219Arg, and G>A at chr17:6.590.909, p.Pro505Leu.

Kohlschutter-Tönz syndrome (KTS) is a rare autosomal recessive disease characterized by epileptic encephalopathy, intellectual disability, and amelogenesis imperfecta. Both *ROGDI*[1] and *SLC13A5* mutations cause KTS, but in *ROGDI*-associated KTS, the onset of seizures is rarely found in the neonatal period.[2]

MORE ONLINE

→**Teaching slides**
links.lww.com/WNL/B205

Study Funding

No targeted funding reported.

Disclosure

The authors report no disclosures relevant to the manuscript. Go to Neurology.org/N for full disclosures.

From the Department of Neurology and Neurosurgery (V.H.P.L., M.d.M.A., R.S.P., M.R.M.), Escola Paulista de Medicina, Brazil; and Instituto de Neurociência do Espírito Santo (M.R.M.), Brazil.

Go to Neurology.org/N for full disclosures.

Appendix Authors

Name	Location	Contribution
Victor Hugo Pantoja Leão, MD	São Paulo, Brazil	Drafted the article and revised it critically for important intellectual content and approved the final version to be published.
Marcelo de Melo Aragão, MD, MSc	São Paulo, Brazil	Drafted the article and revised it critically for important intellectual content and approved the final version to be published.
Ricardo Silva Pinho, MD, PhD	São Paulo, Brazil	Drafted the article and revised it critically for important intellectual content and approved the final version to be published.
Marcelo Rodrigues Masruha, MD, PhD	Espírito Santo, Brazil	Drafted the article and revised it critically for important intellectual content and approved the final version to be published.

References

1. Riemann D, Wallrafen R, Dresbach T. The Kohlschutter-Tonz syndrome associated gene Rogdi encodes a novel presynaptic protein. Sci Rep 2017;7:15791.
2. Schossig A, Bloch-Zupan A, Lussi A, et al. SLC13A5 is the second gene associated with Kohlschutter-Tonz syndrome. J Med Genet 2017;54:54–62.

Teaching NeuroImages: When the Teeth are the Clue to the Etiology of an Epileptic Encephalopathy

Victor Hugo Pantoja Leão, Marcelo de Melo Aragão, Ricardo Silva Pinho, et al.

Neurology 2021;96;e157-e158 Published Online before print September 4, 2020
DOI 10.1212/WNL.0000000000010758