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Teaching NeuroImages: When the teeth are the clue to the etiology of an epileptic encephalopathy

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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]

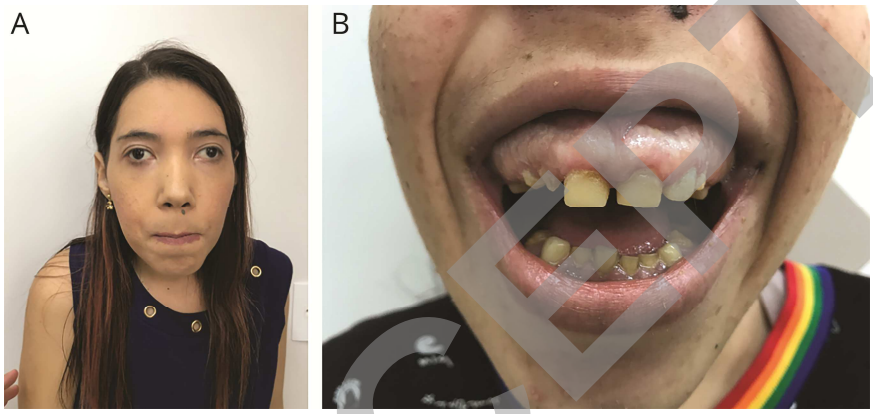
Appendix 1: Authors

Victor Hugo Pantoja Leão	São Paulo – Brazil	Drafted the article and revised it critically for important intellectual content. Approved the final version to be published.
Marcelo de Melo Aragão	São Paulo – Brazil	Drafted the article and revised it critically for important intellectual content. Approved the final version to be published.
Ricardo Silva Pinho	São Paulo – Brazil	Drafted the article and revised it critically for important intellectual content. Approved the final version to be published.
Marcelo Rodrigues Masruha	Espírito Santo – Brazil	Drafted the article and revised it critically for important intellectual content. Approved the final version to be published.

Figure title: Kohlschutter-Tönz syndrome

Figure legend:

Figure – 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.



Teaching Slides-http://links.lww.com/WNL/B205

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