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 ROGDI1 and SLC13A5 mutations cause KTS, but in ROGDI-associated KTS, the onset of seizures is rarely found in the neonatal period.2

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

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

References

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

This information is current as of September 4, 2020

Updated Information & Services
including high resolution figures, can be found at:
http://n.neurology.org/content/96/1/e157.full

References
This article cites 2 articles, 1 of which you can access for free at:
http://n.neurology.org/content/96/1/e157.full#ref-list-1

Subspecialty Collections
This article, along with others on similar topics, appears in the following collection(s):
All Epilepsy/Seizures
http://n.neurology.org/cgi/collection/all_epilepsy_seizures
All Genetics
http://n.neurology.org/cgi/collection/all_genetics
All Pediatric
http://n.neurology.org/cgi/collection/all_pediatric

Permissions & Licensing
Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at:
http://www.neurology.org/about/about_the_journal#permissions

Reprints
Information about ordering reprints can be found online:
http://n.neurology.org/subscribers/advertise