A 9-month-old girl presented with global developmental delay and refractory generalized seizures. Microcephaly, poor visual fixation, and intermittent dystonic posturing were observed on clinical examination. MRI brain (figure) revealed delayed myelination and restricted diffusion involving optic radiations, cerebral peduncles, red nuclei, globus pallidi, and corticospinal tract. EEG showed background slowing and multifocal epileptiform discharges. Workup revealed a homozygous, likely pathogenic variant in ITPA (c.124+1 G>A) and reduced inosine triphosphate pyrophosphohydrolase (ITPase) activity in skin fibroblasts (0.19 nmol/mg protein × h, controls 6.86 ± 2.51). Imaging pattern of delayed myelination and restricted diffusion is suggestive of ITPase deficiency in a child presenting with early infantile epileptic encephalopathy.1,2

Study Funding
No targeted funding reported.
Disclosure
The authors report no disclosures relevant to the manuscript. Go to Neurology.org/N for full disclosures.

Appendix Authors

<table>
<thead>
<tr>
<th>Name</th>
<th>Location</th>
<th>Contribution</th>
</tr>
</thead>
<tbody>
<tr>
<td>Karthik Muthusamy, MD</td>
<td>Clinical Genomics, Mayo Clinic, Rochester, MN</td>
<td>Designed and conceptualized study, analyzed the data, drafted the manuscript for intellectual content</td>
</tr>
<tr>
<td>Suzanne Boyer, RDN, LD</td>
<td>Clinical Genomics, Mayo Clinic, Rochester, MN</td>
<td>Revised the manuscript for intellectual content</td>
</tr>
<tr>
<td>Marc Patterson, MD</td>
<td>Neurology, Pediatrics and Clinical Genomics, Mayo Clinic, Rochester, MN</td>
<td>Interpreted the data, revised the manuscript for intellectual content</td>
</tr>
<tr>
<td>Jorgen Bierau, PhD</td>
<td>Laboratory Medicine, Maastricht University, the Netherlands</td>
<td>Interpreted the data, revised the manuscript for intellectual content</td>
</tr>
</tbody>
</table>

Appendix (continued)

<table>
<thead>
<tr>
<th>Name</th>
<th>Location</th>
<th>Contribution</th>
</tr>
</thead>
<tbody>
<tr>
<td>Saskia Wortmann, MD, PhD</td>
<td>University Children's Hospital, Paracelsus Medical University (PMU), Salzburg, Austria; Amalia Children's Hospital, Radboud UMC, Nijmegen, the Netherlands</td>
<td>Interpreted the data, revised the manuscript for intellectual content</td>
</tr>
<tr>
<td>Eva Morava, MD, PhD</td>
<td>Clinical Genomics and Laboratory Medicine and Pathology, Mayo Clinic, Rochester, MN</td>
<td>Analyzed the data, revised the manuscript for intellectual content</td>
</tr>
</tbody>
</table>

References
Teaching NeuroImages: Neuroimaging Findings in Inosine Triphosphate Pyrophosphohydrolase Deficiency
Karthik Muthusamy, Suzanne Boyer, Marc Patterson, et al.
Neurology 2021;97;e109-e110 Published Online before print February 16, 2021
DOI 10.1212/WNL.0000000000011719

This information is current as of February 16, 2021

Updated Information & Services
including high resolution figures, can be found at:
http://n.neurology.org/content/97/1/e109.full

References
This article cites 2 articles, 0 of which you can access for free at:
http://n.neurology.org/content/97/1/e109.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 Movement Disorders
http://n.neurology.org/cgi/collection/all_movement_disorders
Developmental disorders
http://n.neurology.org/cgi/collection/developmental_disorders
MRI
http://n.neurology.org/cgi/collection/mri

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