

---

## Neurology Publish Ahead of Print

DOI: 10.1212/WNL.000000000207823

### Teaching NeuroImages: The Putaminal Eye, A Highly Characteristic Imaging Feature of MEGDEL Syndrome

**Author(s):**

Raiya Altamimi, MD<sup>1</sup>; hesham aldhalaan, M.D.<sup>1</sup>; Ehab Tous<sup>1</sup>; Manal Nicolas-Jilwan, MD<sup>1</sup>

**Corresponding Author:**

Manal Nicolas-Jilwan, manaljilwan@hotmail.com

**Affiliation Information for All Authors:** 1. King Faisal Specialist Hospital & Research Centre;

**Equal Author Contribution:**

**Contributions:**

Manal Nicolas-Jilwan: Drafting/revision of the manuscript for content&comma; including medical writing for content; Major role in the acquisition of data; Study concept or design; Analysis or interpretation of data; Additional contributions (in addition to one or more of the above criteria)

Raiya Altamimi: Drafting/revision of the manuscript for content&comma; including medical writing for content; Analysis or interpretation of data; Additional contributions (in addition to one or more of the above criteria)

hesham aldhalaan: Drafting/revision of the manuscript for content&comma; including medical writing for content; Additional contributions (in addition to one or more of the above criteria)

Ehab Tous: Drafting/revision of the manuscript for content&comma; including medical writing for content; Additional contributions (in addition to one or more of the above criteria)

**Figure Count:**

1

**Table Count:**

0

**Search Terms:**

[ 91 ] All Genetics, [ 120 ] MRI, [ 227 ] All Pediatric

**Acknowledgment:**

**Study Funding:**

The authors report no targeted funding.

**Disclosure:**

The authors report no relevant disclosures.

**Preprint DOI:**

**Received Date:**

2023-04-06

**Accepted Date:**

2023-07-10

**Handling Editor Statement:**

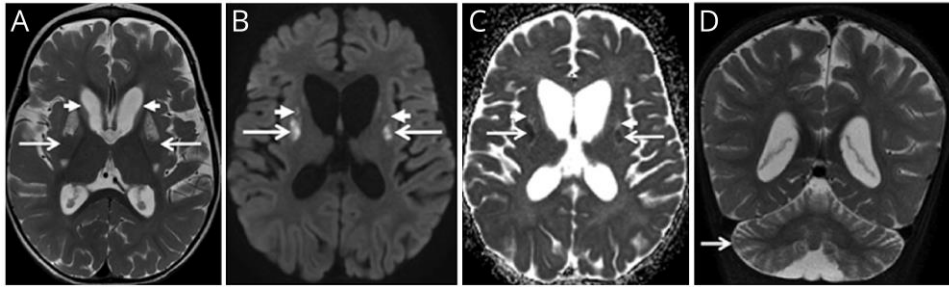
Submitted and externally peer reviewed. The handling editor was Resident and Fellow Deputy Editor Ariel Lyons-Warren, MD, PhD.

ACCEPTED

A 22 month-old female product of a consanguineous marriage presented with recurrent nausea, diarrhea, elevated liver enzymes, metabolic acidosis and developmental regression. Urine analysis revealed elevated 3-methylglutaric acid and 3-methylglutaconic acid. Brain MRI showed bilateral basal ganglia injury (Figure 1A-C) and cerebellar atrophy (Figure 1D). Auditory brainstem response assessment demonstrated bilateral profound sensorineural hearing loss. DNA analysis revealed pathogenic homozygous variant of the *SERAC1* gene, diagnostic of MEGDEL syndrome (3-Methylglutaconic aciduria, Deafness, Encephalopathy, Leigh-like syndrome).

MEGDEL syndrome is an infantile onset syndrome characterized by dystonia, deafness, progressive spasticity, developmental delay or regression, and 3-methylglutaconic aciduria. The causative *SERAC1* gene encodes a phosphatidylglycerol remodeler, essential for mitochondrial function and intracellular cholesterol trafficking. When imaging at the appropriate stage (1 to 4 years of age), the pattern of basal ganglia injury on T2-weighted images sparing the middorsal putamina, called “putaminal eyes,” is pathognomonic<sup>1</sup>. Life expectancy is unknown. Some patients die in infancy.

## Figure 1 Classic MRI brain findings in MEGDEL



Axial T2-weighted image (A) showing symmetrical T2 hyperintensity with atrophy of the caudate nuclei (short arrows) and T2 hyperintensity of the putamina with sparing of their middorsal portions (long arrows). Axial diffusion image (B) and ADC map (C) showing mixed diffusion characteristics in the areas of injury: restricted diffusion (long arrows, sites of active injury) and facilitated diffusion (short arrows). Coronal T2-weighted image (D) showing cerebellar atrophy (arrow).

## References

1. Wortmann SB, van Hasselt PM, Barić I, et al. Eyes on MEGDEL: distinctive basal ganglia involvement in dystonia deafness syndrome. *Neuropediatrics*. 2015;46(2):98-103.

ACCEPTED

# Neurology®

## Teaching NeuroImages: The Putaminal Eye, A Highly Characteristic Imaging Feature of MEGDEL Syndrome

Raiya Altamimi, hesham aldhalaan, Ehab Tous, et al.  
*Neurology* published online August 21, 2023  
DOI 10.1212/WNL.0000000000207823

**This information is current as of August 21, 2023**

<b>Updated Information &amp; Services</b>	including high resolution figures, can be found at: <a href="http://n.neurology.org/content/early/2023/08/21/WNL.0000000000207823.citation.full">http://n.neurology.org/content/early/2023/08/21/WNL.0000000000207823.citation.full</a>
<b>Subspecialty Collections</b>	This article, along with others on similar topics, appears in the following collection(s): <b>All Genetics</b> <a href="http://n.neurology.org/cgi/collection/all_genetics">http://n.neurology.org/cgi/collection/all_genetics</a> <b>All Pediatric</b> <a href="http://n.neurology.org/cgi/collection/all_pediatric">http://n.neurology.org/cgi/collection/all_pediatric</a> <b>MRI</b> <a href="http://n.neurology.org/cgi/collection/mri">http://n.neurology.org/cgi/collection/mri</a>
<b>Permissions &amp; Licensing</b>	Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at: <a href="http://www.neurology.org/about/about_the_journal#permissions">http://www.neurology.org/about/about_the_journal#permissions</a>
<b>Reprints</b>	Information about ordering reprints can be found online: <a href="http://n.neurology.org/subscribers/advertise">http://n.neurology.org/subscribers/advertise</a>

*Neurology*® is the official journal of the American Academy of Neurology. Published continuously since 1951, it is now a weekly with 48 issues per year. Copyright © 2023 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.

