Teaching Video NeurolImages: Epileptic spasms and characteristic ophthalmologic findings

A diagnostic conundrum

Chaithanya Reddy, MD, Muhammed Basil, MBBS, Priyanka Madaan, DM, Lokesh Saini, DM, Sameer Vyas, DM, Simar Rajan Singh, MS, Deeksha Katoch, MS, and Jitendra Kumar Sahu, DM

Neurology® 2020;95:e2602-e2604. doi:10.1212/WNL.0000000000010424

Correspondence

Dr. Madaan

doc.priyanka72@gmail.com

Figure 1 Fundus and EEG findings

RetCam images of right (A) and left eye (B) show multiple, ovoid, hypopigmented areas (arrowheads) around the optic disc, suggesting chorioretinal lacunae. Note an optic disc coloboma (asterisk; B), with hyperpigmented edge. Ten-second EEG epoch (C; pediatric montage; sensitivity: 100 μ V; sweep speed: 30 mm/s) shows high-amplitude slow waves with bursts of polyspike-wave discharges followed by attenuation, consistent with modified hypsarrhythmia.

A 3-month-old girl presented with developmental delay and clustered asymmetric epileptic spasms (video). She was first-born to nonconsanguineous healthy parents with a smooth perinatal transition. Examination revealed normal head size, central hypotonia, and characteristic fundus findings (figure 1, A and B). EEG revealed hypsarrhythmia (figure 1C). Peculiar fundus and neuroimaging findings (figure 2) confirmed the diagnosis of Aicardi syndrome (AS). Epileptic spasms failed to respond to adrenocorticotropin and vigabatrin. This report

MORE ONLINE

► Video

► Teaching slides

links.lww.com/WNL/B178

From the Pediatric Neurology Unit (C.R., P.M., L.S., J.K.S.) and Department of Pediatrics (M.B.), Advanced Pediatrics Centre, Department of Radiodiagnosis and Imaging (section of Neuroimaging and Interventional Radiology) (S.V.), and Advanced Eye Centre (S.R.S., D.K.), PGIMER, Chandigarh, India.

Go to Neurology.org/N for full disclosures. Funding information and disclosures deemed relevant by the authors, if any, are provided at the end of the article.

Figure 2 Brain MRI

Axial T2-weighted (A–C) and T1-weighted sagittal (D), coronal (E), and axial (F) images showing an extraaxial cyst in posterior fossa (white arrows) with cerebellar hypoplasia. Note colpocephaly, nonvisualization of corpus callosum, midline dorsal interhemispheric cyst (asterisks), nodular periventricular heterotopia in right parietal region (black arrows), and polymicrogyria in left frontal lobe (oval markers).

emphasizes the utility of skillful neuroophthalmologic assessment in children with epileptic spasms.

AS is an X-linked dominant, callosal agenesis syndrome with a dismal prognosis.¹ Besides AS, callosal agenesis has been associated with many polymalformative syndromes, which should be considered with screening.²

Acknowledgment

The authors thank the patient's parents for images. The patient was a part of project supported by the Council of Scientific and Industrial Research, New Delhi.

Study funding

No targeted funding reported.

Disclosure

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

Appendix Authors

Name	Location	Contribution
Bandi V. Chaithanya Reddy, MBBS, MD	PGIMER, Chandigarh, India	Patient management, literature review, initial draft manuscript preparation, final approval of the version to be published
Muhammed Basil, MBBS	PGIMER, Chandigarh, India	Patient management, literature review, final approval of the version to be published
Priyanka Madaan, MD, DM	PGIMER, Chandigarh; CSIR, New Delhi, India	Patient management, analysis of EEG, literature review, critical review of manuscript for important intellectual content, final approval of the version to be published
Lokesh Saini, MD, DM	PGIMER, Chandigarh, India	Patient management, critical review of the manuscript, final approval of the version to be published

Continued

Appendix *(continued)*

Name	Location	Contribution
Sameer Vyas, MD, DM	PGIMER, Chandigarh, India	Analysis of the radiologic data, critical review of manuscript, final approval of the version to be published
Simar Rajan Singh, MBBS, MS	PGIMER, Chandigarh, India	Analysis of RetCam images, patient management, final approval of the version to be published
Deeksha Katoch, MBBS, MS	PGIMER, Chandigarh, India	Analysis of RetCam images, patient management, final approval of the version to be published
Jitendra Kumar Sahu, MD, DM	PGIMER, Chandigarh, India	Patient management, critical review of manuscript for important intellectual content, final approval of the version to be published

References

1. Aicardi J, Lefebvre J, Lerique-Koechlin A. A new syndrome: spasms in flexion, callosal agenesis, ocular abnormalities. *Electroencephalogr Clin Neurophysiol* 1965;19:609–610.
2. Reddy C, Bhattacharya D, Madaan P, Saini L. Corpus callosum agenesis with interhemispheric cyst: a neuroimage to remember. *BMJ Case Rep* 2019;12:e231375.

Neurology®

Teaching Video NeuroImages: Epileptic spasms and characteristic ophthalmologic findings: A diagnostic conundrum

Chaithanya Reddy, Muhammed Basil, Priyanka Madaan, et al.

Neurology 2020;95:e2602-e2604 Published Online before print July 20, 2020

DOI 10.1212/WNL.00000000000010424

This information is current as of July 20, 2020

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/95/18/e2602.full
References	This article cites 2 articles, 0 of which you can access for free at: http://n.neurology.org/content/95/18/e2602.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 Pediatric http://n.neurology.org/cgi/collection/all_pediatric Infantile spasms http://n.neurology.org/cgi/collection/infantile_spasms 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

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 © 2020 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.