Teaching Video NeuroImages: Epileptic spasms and characteristic ophthalmologic findings

A diagnostic conundrum

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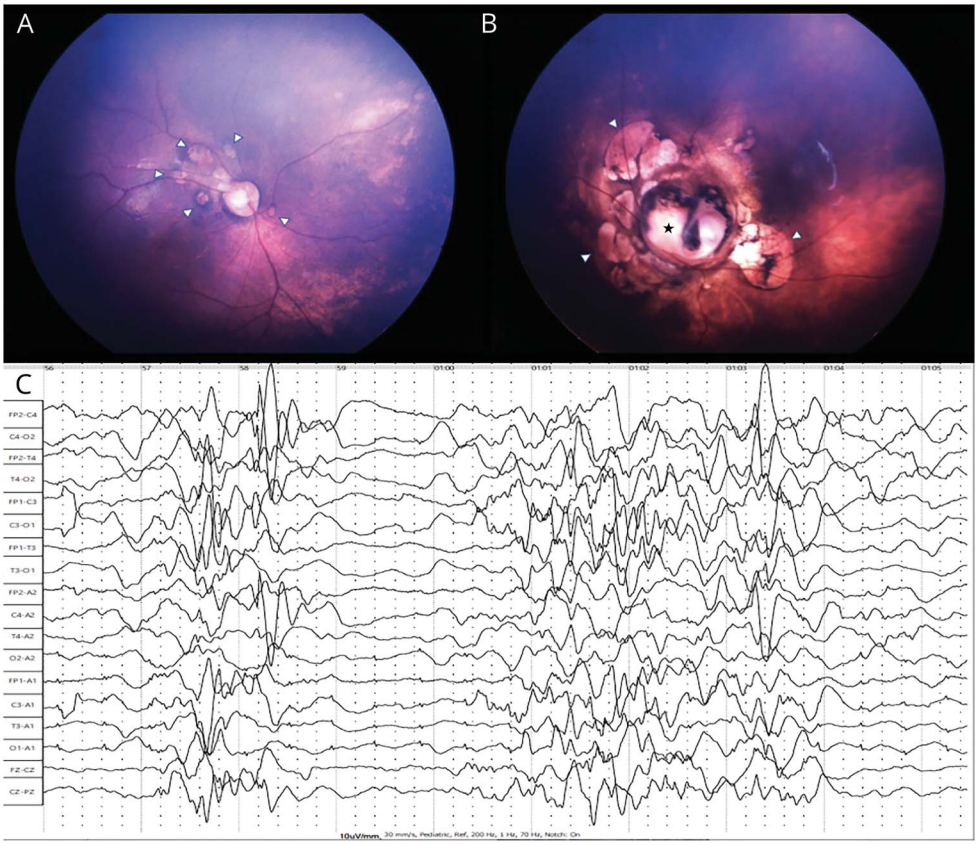
Figure 1 Fundus and EEG findings



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 μV; 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

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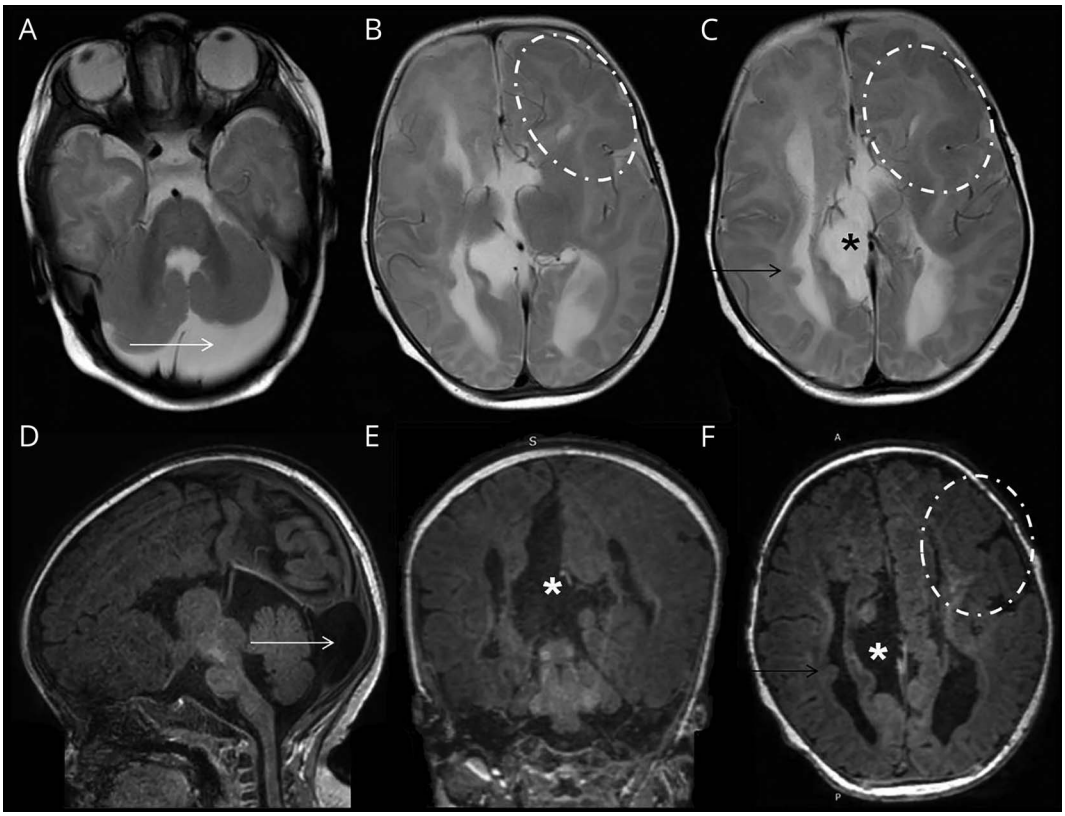
Figure 2 Brain MRI



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]

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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

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