Aicardi syndrome: when to suspect the unexpected

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Epileptic Disorders

CASE STUDY

We report a 30-year-old woman who started experiencing infantile spasms at the age of two months; global developmental delay was noted shortly afterwards.

Brain MRI revealed multiple malformations of cortical development and agenesis of the corpus callosum (*figures 1, 2*), while EEG showed multiple independent spike foci and asymmetric slow background activity.

Aicardi syndrome was suspected and subsequently confirmed when chorioretinal lacunae were identified on fundus examination.

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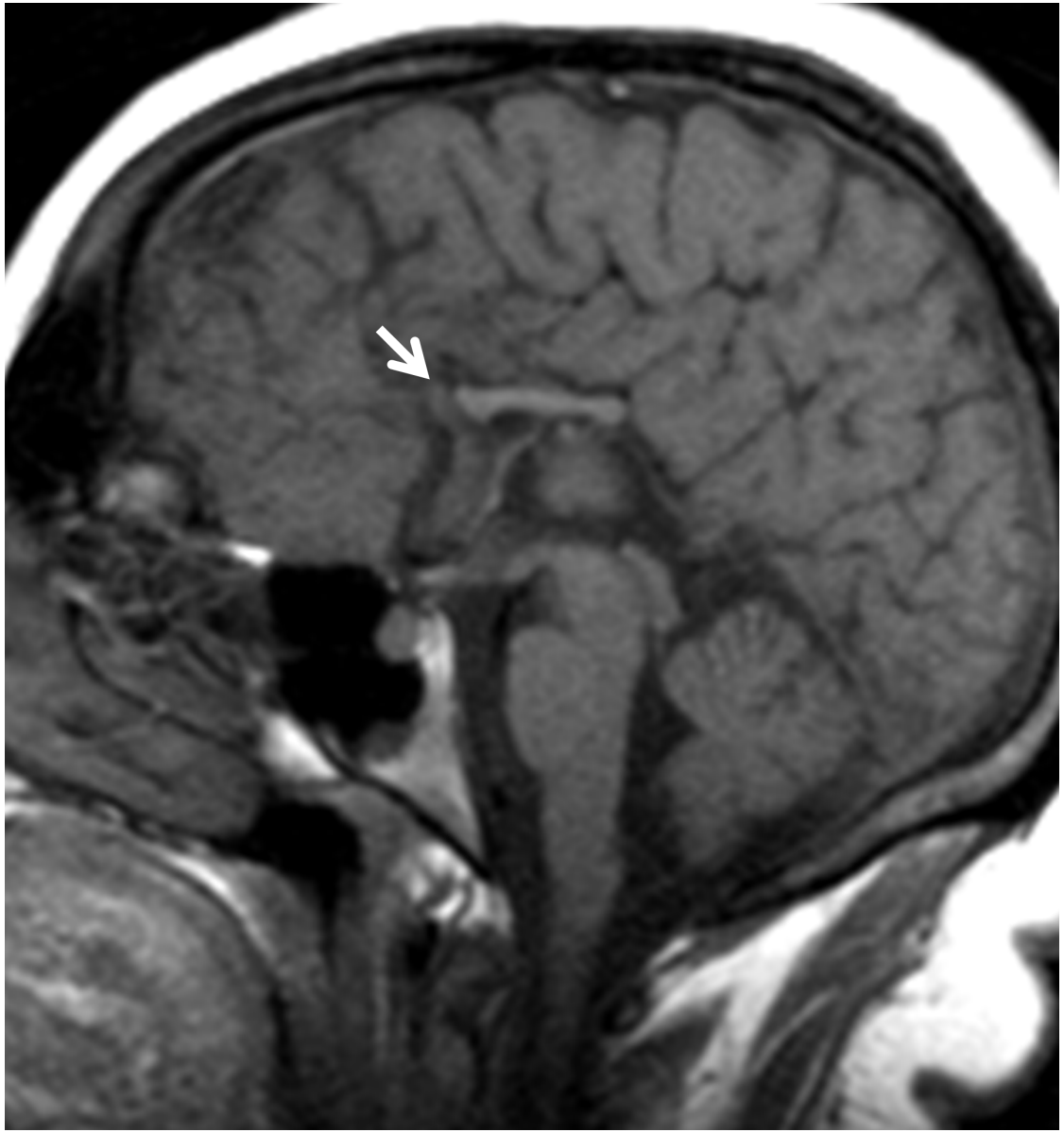

Figure 1.
T1-weighted sagittal brain MRI demonstrating partial agenesis of the corpus callosum;
The genu and rostrum structures are absent (white arrow), while the body and a small portion of the anterior splenium are present.

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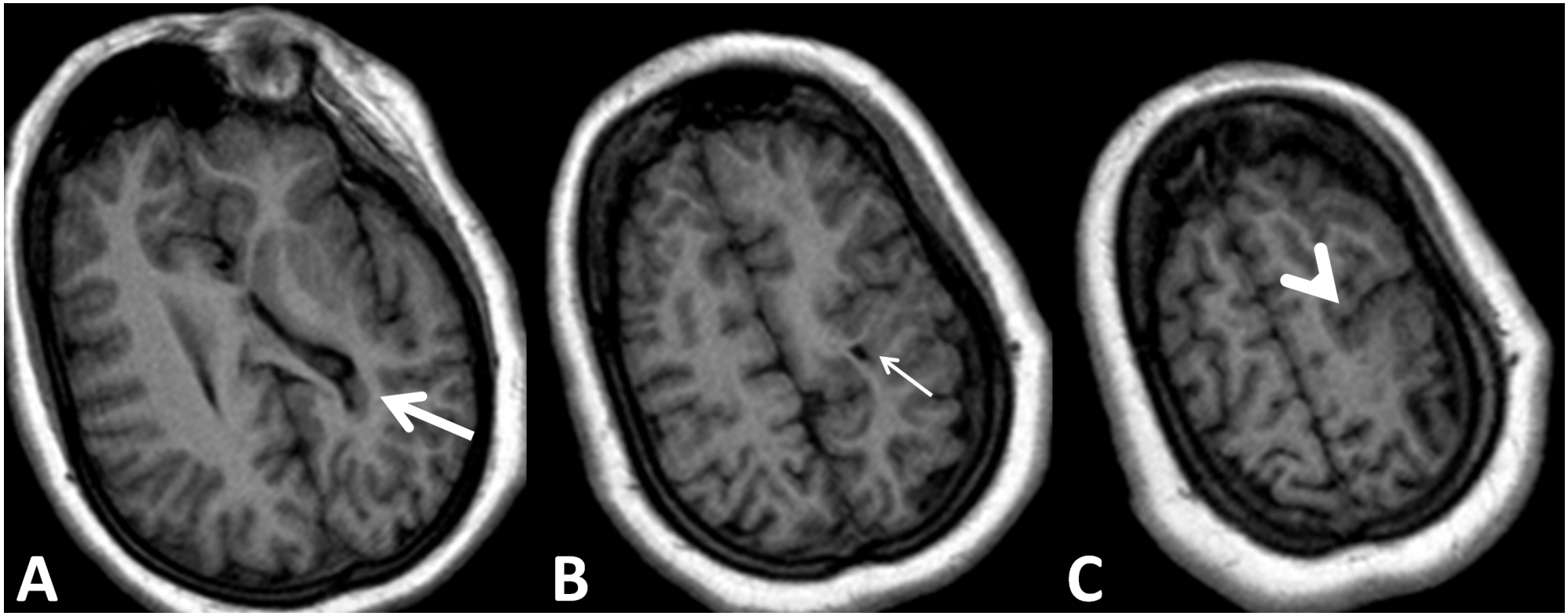

Figure 2. T1-weighted axial brain MRI showing **heterotopic gray grey matter** along the left lateral ventricle compatible with **periventricular or subependymal heterotopia** (arrow in **A),** and a **closed-lip schizencephaly** (small arrow in **B),** lined by **polymicrogyric cortex and additional, more frontal polymicrogyria** involving the posterior left frontal lobe **(arrowhead in C).**

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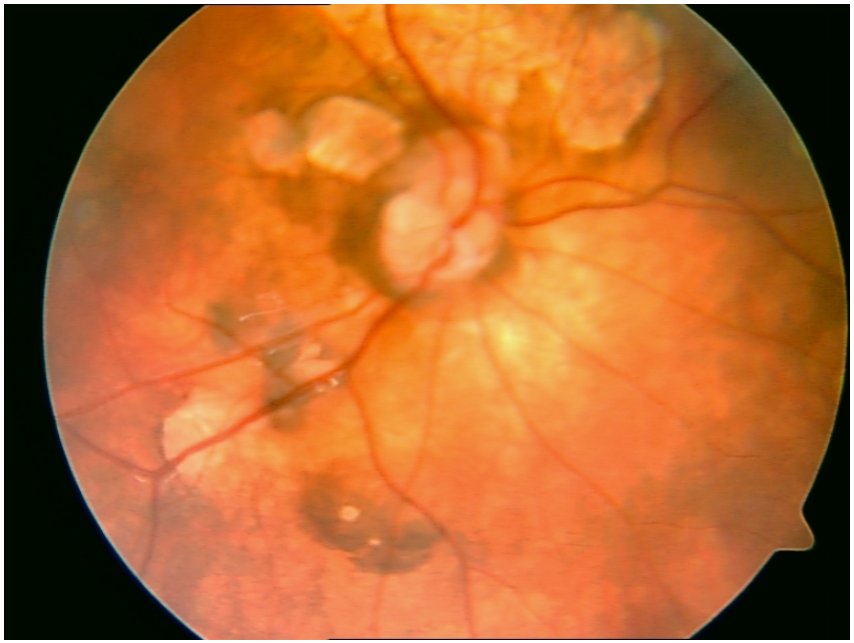

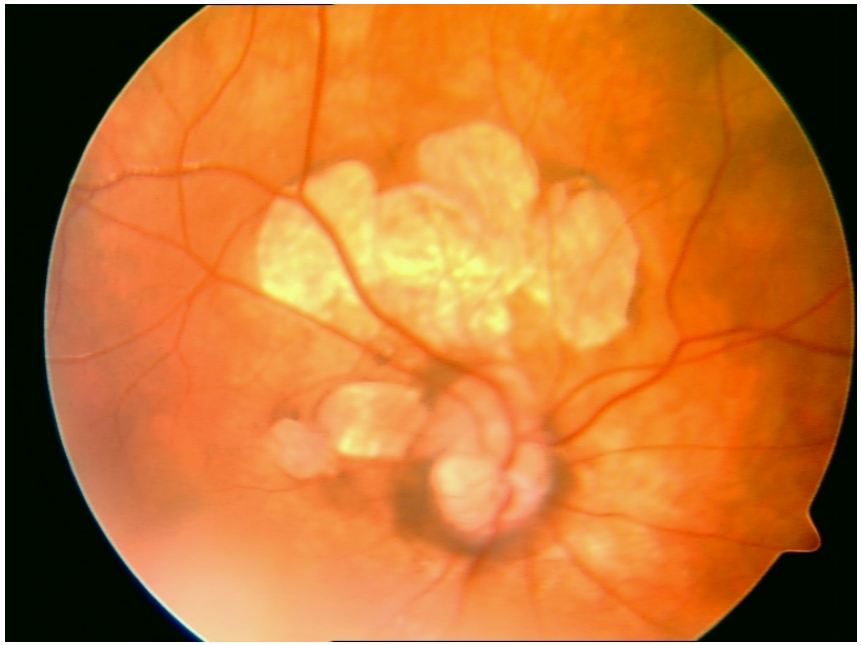

The **choroidoretinal lacunae** are considered pathognomonic for AS.
They are virtually always multiple although of variable extent and generally bilateral. They present at funduscopy as rounded whitish or pinkish areas ranging in size from 1/10th to several disc diameters. They are on the same plane as the retina so that vessels do not bend on crossing their borders.

NB: *Figures representing chroidoretinal lacunae and comment provided by the Editor-in-Chief of Epileptic Disorders for didactic purposes.*
They do not exactly represent those observed to the patient reported by the authors.

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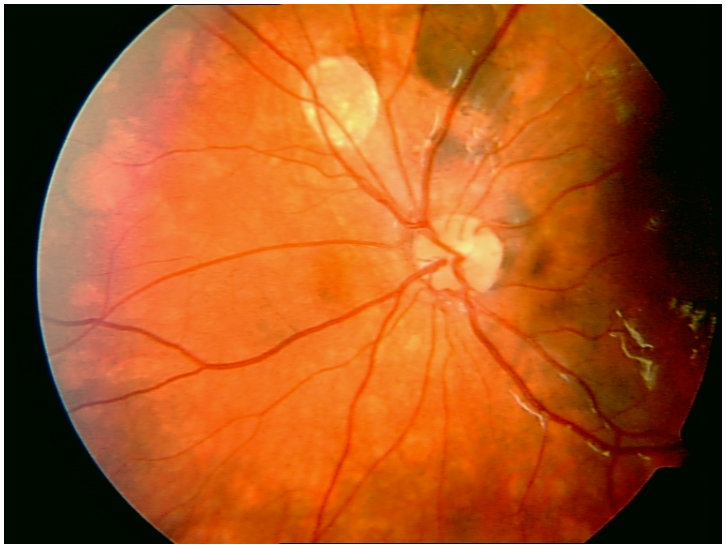

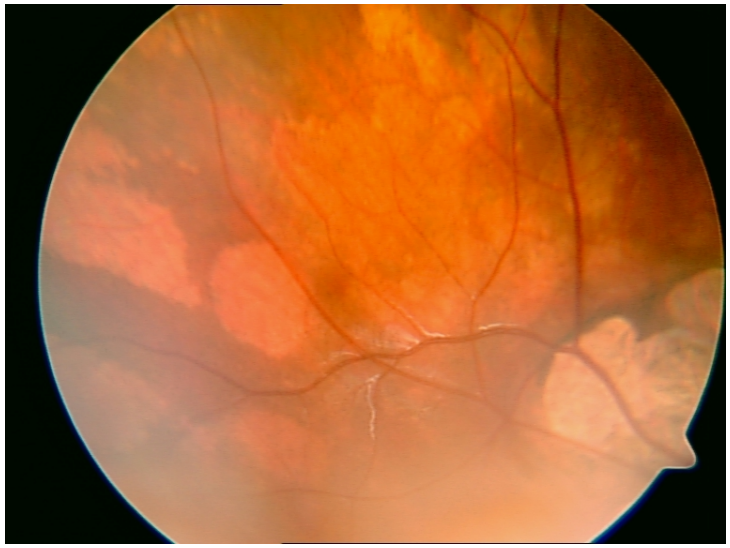

Pigment deposits are frequently present at their periphery or even in the central part and may increase with age. However, the size of the lacunae does not change, even at examinations several years apart. The largest lacunae tend to cluster around the disc, whereas small pinkish lesions tend to be more peripheral. The eye pathology does not feature any degenerative or inflammatory lesion. There is thinning of the choroid and sclera in the areas corresponding to the lacunae and the pigment epithelium is hypopigmented or depigmented with degeneration of the rods and cones.

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Take Home Message

Aicardi syndrome should be suspected also in adult patients who present a history of infantile spasms and/or intractable epilepsy and agenesis of the corpus callosum, either partial or complete.

Following clinical suspicion, ophthalmologic examination should be performed to confirm this diagnosis (Aicardi *et al*., 1965; Aicardi, 2005; Hopkins *et al*., 2008).

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