Case Report

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Radiology Section

Aicardi Syndrome: A Rare Cause of Infantile Spasms

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ABSTRACT

Infantile spasms may occur in isolation or as part of multiple neurodevelopmental syndromes. We report an eight-month-old female infant presenting with infantile spasms, developmental delay, and characteristic features of Aicardi syndrome. Electroencephalogram (EEG) showed asynchronous multifocal epileptiform abnormalities, while brain Magnetic Resonance Imaging (MRI) demonstrated corpus callosum agenesis, midline cysts, cortical malformations, and posterior fossa anomalies. Ophthalmoscopy confirmed chorioretinal lacunae. Brain MRI and EEG remain crucial complementary modalities for early diagnosis of infantile spasms and underlying aetiologies. This case reinforces consideration of Aicardi syndrome in female infants presenting with spasms and characteristic ocular findings.

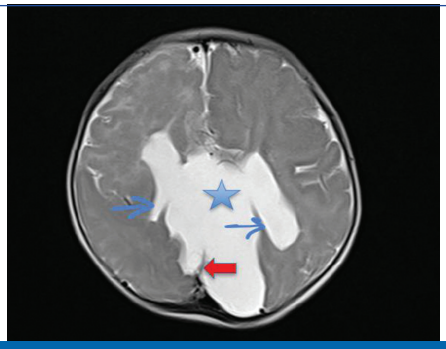
Keywords: Corpus callosum agenesis, Dandy Walker malformation, Holoprosencephaly MRI, Neurodevelopmental disorder

CASE REPORT

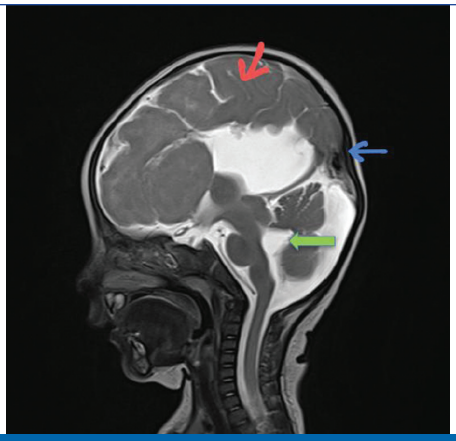
An eight-month-old female infant presented with global developmental delay and clusters of infantile spasms (clusters of flexor and extensor spasms) occurring 3-4 times per day for the past two months. She was born preterm at 33 weeks by vaginal delivery, with no delayed cry or Neonatal Intensive Care Unit (NICU) stay. There was a history of parental consanguinity but no family history of seizures and developmental anomalies. There was no history of antenatal maternal exposures or infections. No antenatal scans were available with the patient. No genetic testing or formal counselling was undertaken, as the patient presented as an unscheduled walk-in at 33 weeks' gestation.

On clinical examination, the infant demonstrated global developmental delay involving the gross motor, social, and visual domains, with absent visual fixation. No dysmorphic features were noted.

MRI brain showing absence of the septum pellucidum and corpus callosum, with parallel orientation of the lateral ventricles [Table/Fig-1], colpocephaly, and radiating gyri on sagittal images [Table/Fig-2], consistent with callosal agenesis. A Cerebrospinal Fluid (CSF)-intense cystic lesion was noted in the parieto-occipital region, communicating freely with the body and trigones of moderately dilated lateral ventricles, representing a Barkovich type I interhemispheric cyst [Table/Fig-1]- a typical midline cyst associated with Aicardi syndrome. The falx cerebri was present only anteriorly, with interdigitating gyri in the frontal region.

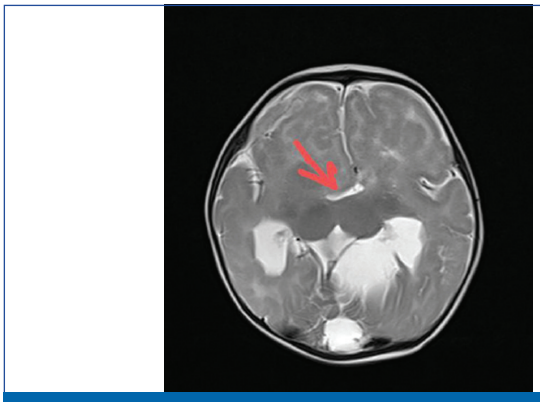


[Table/Fig-1]: Axial T2-weighted image shows Barkovich type 1 interhemispheric cyst (asterix); Dilated and parallel-arranged lateral ventricles (blue arrow); Residual posterior falx cerebri (red arrow),

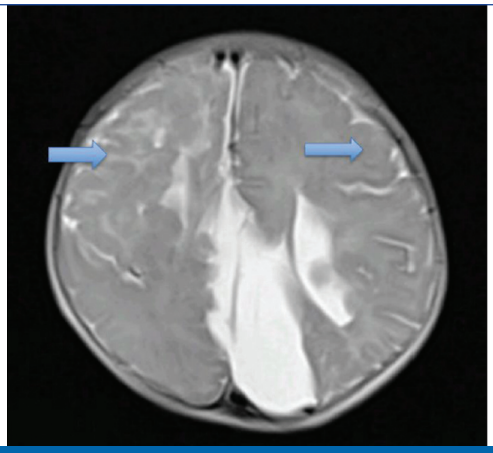


[Table/Fig-2]: Sagittal T2-weighted image shows corpus callosum agenesis (orange arrow); tenting of tentorium cerebelli (blue arrow); mildly dilated 4th ventricle (green arrow).

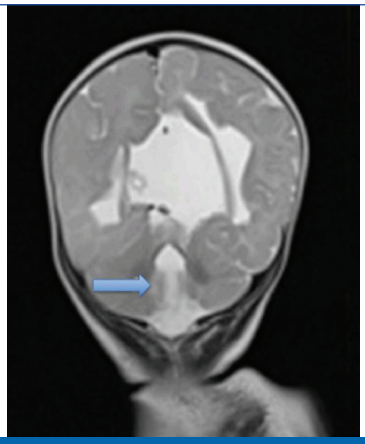
The thalami and cerebral peduncles were fused in the midline, suggestive of semilobar holoprosencephaly [Table/Fig-3]. Bilateral hippocampi were hypoplastic. Periventricular nodular heterotopia was seen along both lateral ventricles. Polymicrogyria, in the form of closely packed small dysplastic gyri, was present in both frontal and occipital lobes [Table/Fig-4]. The infratentorial compartment showed near complete absence of the cerebellar vermis [Table/Fig-5], with only a small superior vermian remnant, along with superior tenting of the tentorium and torcular–lambdoid inversion.



[Table/Fig-3]: Axial T2-weighted image shows fused thalami (orange arrow).

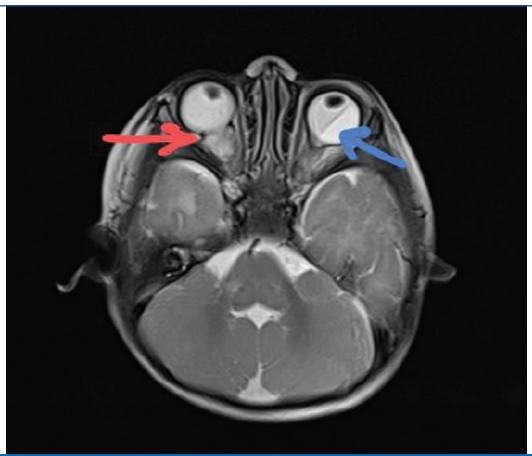


[Table/Fig-4]: Axial T2 section showing polymicrogyria.



[Table/Fig-5]: Coronal T2-weighted image shows near complete absence of cerebellar vermis (arrow).

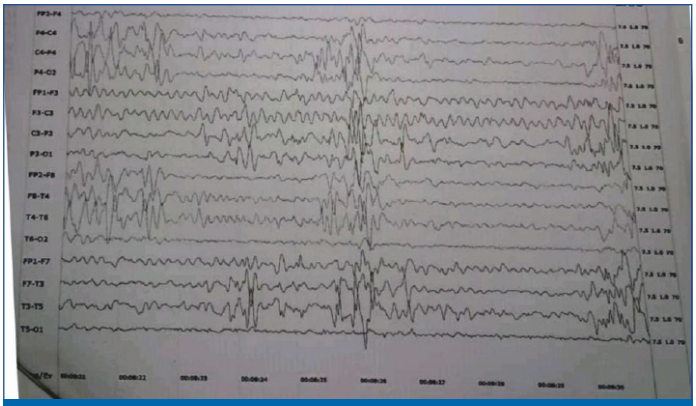
Evaluation of the orbits revealed bilateral globe abnormalities, including left-sided retinal detachment, defects in the posterior coats, and herniation of vitreous, findings consistent with chorioretinal coloboma [Table/Fig-6]. These ocular features were subsequently confirmed on ophthalmoscopy, supporting the diagnosis of Aicardi syndrome.



[Table/Fig-6]: Axial T2 weighted image shows bilateral Coloboma (Red arrow); Left-sided Retinal Detachment (Blue arrow).

EEG revealed frequent paroxysmal polypots throughout the entire recording, confirming the presence of infantile spasms [Table/Fig-7].

The patient was initiated on antiepileptic therapy comprising oral valproate (20-40 mg/kg/day), vigabatrin (150 mg/kg/day in two divided doses), and levetiracetam (20-60 mg/kg/day in two divided doses), along with physiotherapy support. The patient was discharged within 48 hours. At the 1-month follow-up, the parents reported a significant reduction in seizure frequency (upto <7 per week).



[Table/Fig-7]: EEG showing asynchronous multifocal epileptiform abnormalities with burst suppression.

DISCUSSION

Aicardi syndrome is marked by the classic triad of distinct symptoms: agenesis of the corpus callosum, chorioretinal lacunae, and infantile spasms [1]. Characteristic MRI features in patients with Aicardi syndrome include identifying partial or complete agenesis of the corpus callosum and cortical migration anomalies [2]. These imaging findings are pivotal in distinguishing Aicardi syndrome from other neurological disorders with overlapping symptoms [2,3].

Differential diagnoses considered in this case included Dandy-Walker malformation, congenital infections, and other syndromic or isolated forms of corpus callosum agenesis. Dandy-Walker malformation was excluded as the posterior fossa was not enlarged. Congenital infections, particularly TORCH-related etiologies, were ruled out due to the absence of intracranial calcifications, white matter changes, microcephaly, or chorioretinitis. Other causes of corpus callosum agenesis were also considered; however, the findings as described in our case were highly specific for Aicardi syndrome.

Hopkins B et al., provide a comprehensive review of the imaging findings, such as intracranial cysts, posterior fossa anomalies and the abnormalities in the ventricular system often accompanying corpus callosum agenesis. Their work aids in understanding the typical and atypical imaging features, structural brain anomalies that contribute to the clinical symptoms of the syndrome [4]. Imaging plays an integral role in both diagnosis and understanding the pathological underpinnings of the disorder, guiding clinical management [2,5]. MRI helped delineate the structural anomalies in the brain, including the cortical migration defects. Additionally, EEG supported the imaging findings by confirming the epileptic pattern, thus aiding the diagnostic confidence. Altogether, a multimodality approach informs prognosis, guides management and assists in counselling the families regarding severity and expected outcomes.

Management of Aicardi syndrome, as reflected in our case, involves early intervention with Antiepileptic Drugs (AEDs), which has been shown to potentially mitigate seizure frequency and severity, contributing positively to developmental outcomes [6,7]. However, the variability in response to AEDs highlights the need for personalised treatment strategies and suggests potential areas for further research into optimal management protocols [6].

Considering the genetic aetiology of the syndrome, often linked to de novo mutations and predominantly affecting females [8], genetic counselling is vital for providing family support and future planning. Multi-disciplinary collaborative care strategy is crucial for addressing the comprehensive needs of patients with Aicardi syndrome [9]. Guadagni MG et al., and Wang JH et al., described simple patient case reports associating infantile spasms with corpus callosum agenesis and other parallel core diagnostic features as seen in our case [10,11]. Smith CD et al., used larger cohorts that demonstrated the supratentorial additional findings similar to our case to be commonly associated with corpus callosum agenesis [12]. Pomar L et al., and Gacio S et al., have demonstrated

that the presence of corpus callosum agenesis, midline cysts, polymicrogyria, and cerebellar vermian hypoplasia represents a discernible neuroimaging phenotype of Aicardi syndrome across different stages of development, additionally describing the presence of posterior fossa anomalies [13,14]. Further research is needed to explore the underlying genetic mechanisms and to optimise therapeutic strategies for this complex disorder [7].

CONCLUSION(S)

This case highlights the importance of considering Aicardi syndrome in female infants presenting with infantile spasms, developmental delay, and characteristic ocular findings. A unique aspect of this case is the co-existence of extensive supratentorial malformations with posterior fossa anomalies, along with chorioretinal defects, emphasising the wide phenotypic variability in Aicardi Syndrome. This case underscores the need for vigilant clinical suspicion and thorough neuroimaging assessment in infants with early-onset epileptic spasms to ensure prompt initiation of treatment.

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