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Case Report

Unlike the Textbook Triad: A Rare Case of Aicardi Syndrome

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ABSTRACT

Aicardi syndrome (AS) is an X-linked inherited disorder characterized by infantile spasms, chorioretinal lacunae, and agenesis or hypogenesis of the corpus callosum. Since the description of the first case of Aicardi syndrome in 1965 not many have contributed to the list, hence preserving its rarity. The purpose of this case report was to demonstrate the spectrum of MRI findings and follow up in the course of Aicardi syndrome.
Key words: Corpus callosum; agenesis; Aicardi; microcephaly; infant

INTRODUCTION

Aicardi syndrome (AS) was first described by Jean Aicardi in 1965 and is characterized by agenesis or hypogenesis of the corpus callosum, chorioretinal lacunae, and early infantile spasms. Most of AS patients develop normally until 3 months of age, and then, epileptic seizures and developmental delay start. It is an X-linked dominant genetic disorder and lethal in males and, therefore, almost exclusively observed in females. Almost all patients are females; however, XXY male patients have been reported. [1]

CASE PRESENTATION

A 2 month old female infant was referred to Department of Radio-diagnosis, Kasturba Medical College, and Mangalore with anophthalmia, microcephaly and global developmental delay. A syndrome association was suspected clinically. No other morphological abnormalities were evident on clinical examination. Antenatal history was uneventful. No history of maternal infection during pregnancy. Child cried at birth which was a term delivery. Blood and urine reports were within normal limits and CSF testing did not yield any clues to diagnosis. No chromosomal disease pattern was discovered on analysis. Detailed birth and development history was elicited from parents. No episode of seizures had occurred although they confirmed delayed milestones compared to a normal infant. Plain MRI study of brain and orbits on a Siemens Magnetom Avanto 1.5T system revealed partial agenesis of corpus callosum and unilateral optic nerve hypoplasia. Screening of spine showed no abnormalities. Follow up showed infantile spasms at age of 9 months.

IMAGING FINDINGS

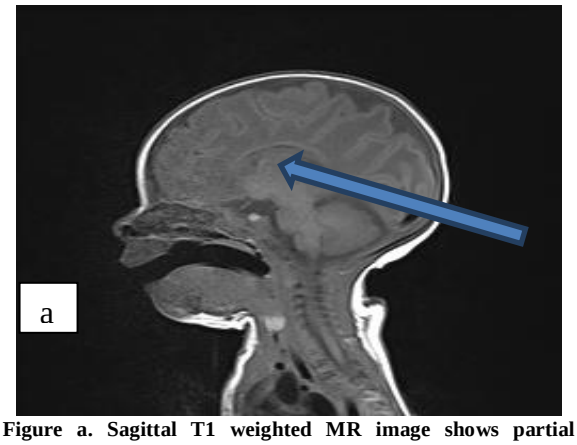

Figure a. Sagittal T1 weighted MR image shows partial agenesis of corpus callosum.

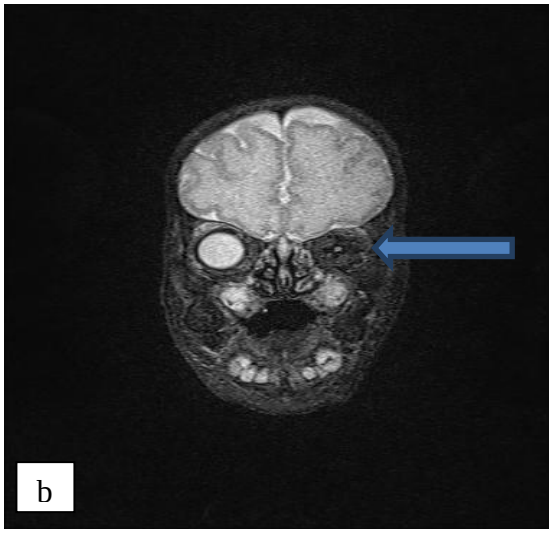

Figure b. Coronal T2 weighted MR image showing left anophthalmia.

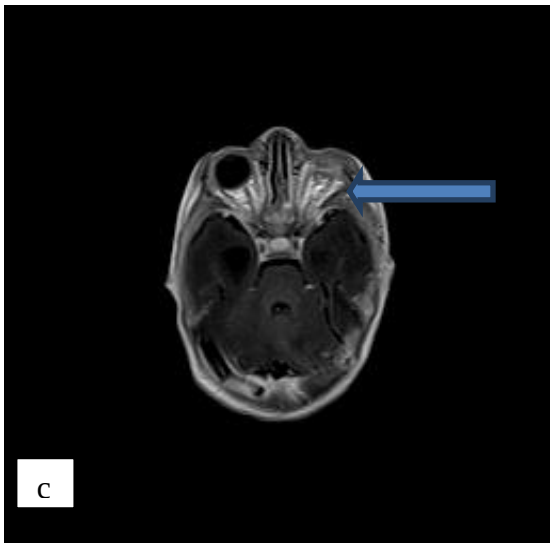

Figure c. Axial T1 image on right showing left optic nerve hypoplasia.

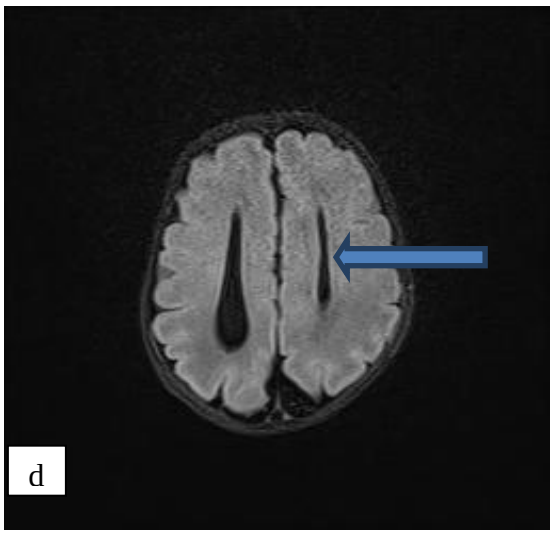

Figure d. Axial FLAIR image shows parallel orientation of lateral ventricles.

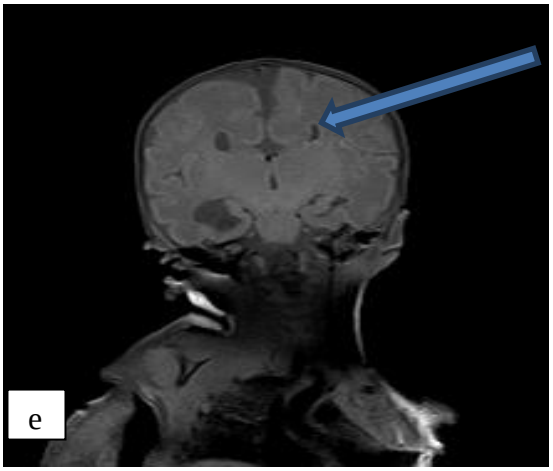

Figure e. Coronal T1 weighted MR image showing Viking/devil horns sign (arrow)-feature of agenesis of corpus callosum.

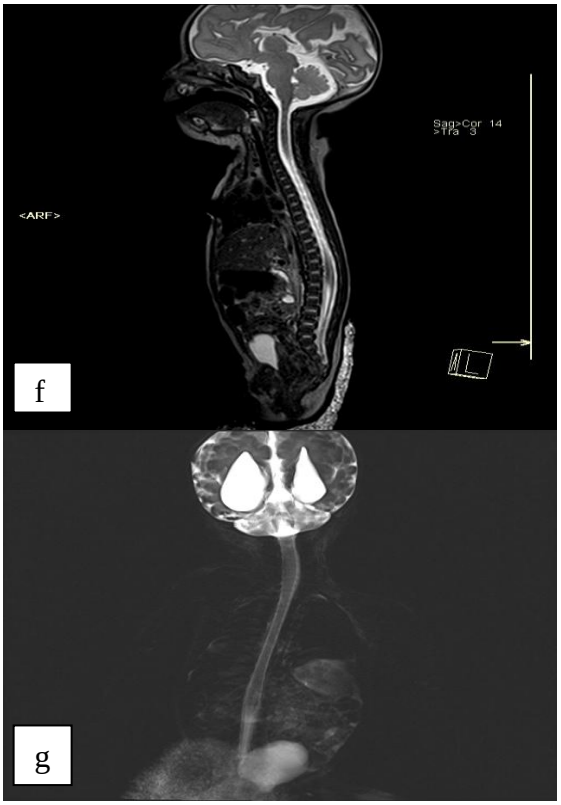

Figure f & g. screening of spine for vertebral anomalies and MR myelogram were normal.

DISCUSSION

The diagnosis of Aicardi syndrome is based exclusively on established clinical diagnostic criteria as follows:

Modified diagnostic criteria have been proposed [Sutton et al 2005, adapted from Aicardi 1999]: [1,3]

- The presence of all three classic features (classic triad) is diagnostic for Aicardi syndrome.

- The existence of two classic features plus at least two other major or supporting features is strongly suggestive of the diagnosis of Aicardi syndrome. [1,3]

Classic triad

- Agenesis of the corpus callosum
- Distinctive chorioretinal lacunae
- Infantile spasms. [4]

Major features

- Cortical malformations (mostly polymicrogyria)
- Periventricular and subcortical heterotopia
- Cysts around third cerebral ventricle and/or choroid plexus
- Optic disc/nerve coloboma or hypoplasia. [4]

Supporting features

- Vertebral and rib abnormalities
- Microphthalmia
- "Split-brain" EEG
- Gross cerebral hemispheric asymmetry
- Vascular malformations or vascular malignancy. [4]

MRI is the modality of choice to evaluate brain, orbits and spine. It also can point to aetiology of developmental delay such as old hypoxic injury and assess degree of myelination and prognostication. The corpus callosum is the brain's largest commissural tract connecting the cerebral hemispheres. It consists of genu, rostrum, body and splenium. Line drawn between anterior and posterior commissure is defining for routine axial MR plane. Classically mentioned pathognomonic chorioretinal lacunae were not identified on ophthalmologic examination in our case but presence of other findings like partial agenesis of corpus callosum, unilateral optic nerve hypoplasia and infantile spasms are strongly suggestive of Aicardi. While total agenesis points to an early insult, partial indicates insult was later on. A parallel course of lateral ventricles on axial images and devil's horns on coronal sections is seen in partial agenesis as seen in our case (Figure f & g).

Survival period is variable. Life-long management by a team of paediatric neurologist with ophthalmological and physiotherapy consultation is needed. Management of infantile spasms and medically refractory epilepsy is essential.

TEACHING POINT

Corpus callosum dysgenesis is associated with classical MR features and could have associated abnormalities like polymicrogyria and heterotopia. Identifying multiple anomalies in brain and spine could point to particular syndromes, encourage genetic counselling where necessary and avoid secondary complications.

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