ISSN- O: 2458 - 868X, ISSN–P: 2458 – 8687
National Library of Medicine - ID: 101731606

International Journal of Medical Science and Innovative Research (IJMSIR)
IJMSIR : A Medical Publication Hub
Available Online at: www.ijmsir.com
Volume – 9, Issue – 4, July – 2024 , Page No. : 03 – 08

A case report on Aicardi Syndrome

[1]Dr. Shiksha Dahiya, Post Graduate Resident, Department of Radio-diagnosis, Sree Balaji Medical College and Hospital, Chennai, Tamilnadu.

[2]Dr. G.Murugan, Professor and HOD, Department of Radio-diagnosis, Sree Balaji Medical College and Hospital, Chennai, Tamilnadu.

Corresponding Author: Dr. Shiksha Dahiya, Post Graduate Resident, Department of Radio-diagnosis, Sree Balaji Medical College and Hospital, Chennai, Tamilnadu.

Citation this Article: Dr. Shiksha Dahiya, Dr. G.Murugan, "A case report on Aicardi Syndrome", IJMSIR - July - 2024, Vol – 9, Issue - 4, P. No. 03 – 08.

Type of Publication: Case Report

Conflicts of Interest: Nil

Abstract

Aicardi syndrome is a genetic disorder linked to the X chromosome, predominantly affecting females. Diagnosis requires the presence of three key features: absence of the corpus callosum, infantile spasms, and chorioretinal lacunae. This case study details the presentation of a female newborn delivered at 36weeks gestation, who presented to the emergency department with arm and leg stiffness. An electroencephalogram confirmed generalized hypsarrhythmia, characteristic of infantile spasms. Magnetic resonance imaging revealed complete absence of the corpus callosum. Subsequent ophthalmoscopic examination identified multiple hypopigmented lesions in the retina consistent with chorioretinal lacunae. These findings collectively confirmed the diagnosis of Aicardi syndrome. Treatment commenced with anticonvulsant therapy and physiotherapy. This report underscores the clinical and radiological criteria crucial for suspecting and diagnosing this rare genetic disorder.

Keywords: Aicardi syndrome, Neurodevelopmental disorders, Agenesis of the corpus callosum, Chorioretinal lacunae, infantile spasms.

Introduction

Aicardi syndrome (AS) is a rare genetic neurodevelopmental disorder primarily found in females, occurring at a rate of approximately one case per 110,000 live births [1, 2]. AS is characterized by a triad of abnormalities including agenesis of the corpus callosum, infantile spasms, and chorioretinal lacunae [1, 3, 4]. Moreover, it can present with polymicrogyria, per ventricular heterotopia, choroid plexus cysts, abnormalities in the cerebellum, enlargement of the cisterna magna, and malformations of the cost vertebral region[1,4].Diagnosis of AS is challenging due to its rarity, typically requiring a multidisciplinary approach involving neuroimaging, ophthalmological examination, and pediatric neurology assessment [3]. Currently, there is no cure for AS management focuses on antiepileptic medications, physiotherapy, and dietary interventions [5].

Kroner et al. reported approximately 853 cases of AS in the United States and around 4000 cases globally, indicating a disease that is inadequately documented in the medical literature [6]. Therefore, this article aims to present and discuss the neurological and neuroradiological findings observed in a newborn diagnosed with AS.

Case Description

We present a case involving a female infant delivered via elective cesarean section at 36 weeks of gestation to a 35-year-old mother who underwent multiple ultrasound assessments throughout pregnancy, initially suspecting Dandy-Walker syndrome. The baby weighed 2050 g, measured 48 cm in height, and had an APGAR score of 8/10. There were no reported congenital diseases in the family history.

At 18 days old, the infant began exhibiting symptoms including arm and leg stiffness, accompanied by neck hyperextension occurring 15-20 times daily. During the physical examination, the patient appeared alert with typical newborn reflexes, bilateral iris colobomas resembling those of a cat's eye, mild scoliosis, and absence of ribs 7-9.

Due to the occurrence of infantile spasms and bilateral iris colobomas, magnetic resonance imaging (MRI) was performed. The MRI revealed agenesis of the corpus callosum, a multiseptated interhemispheric cyst communicating with the ventricular system, and bilateral coloboma cysts (see Figure 1)

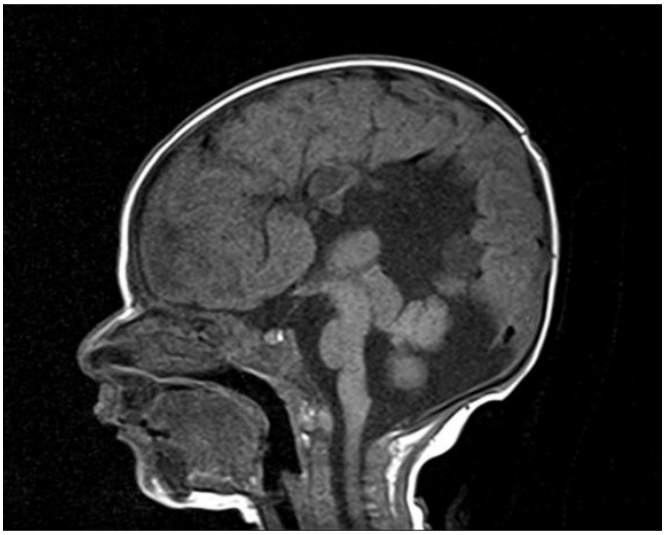

Fig. 1; Brain MRI. T1-weighted image shows lo agenesis of corpus callosum and a multiseptated interhemispheric cyst.

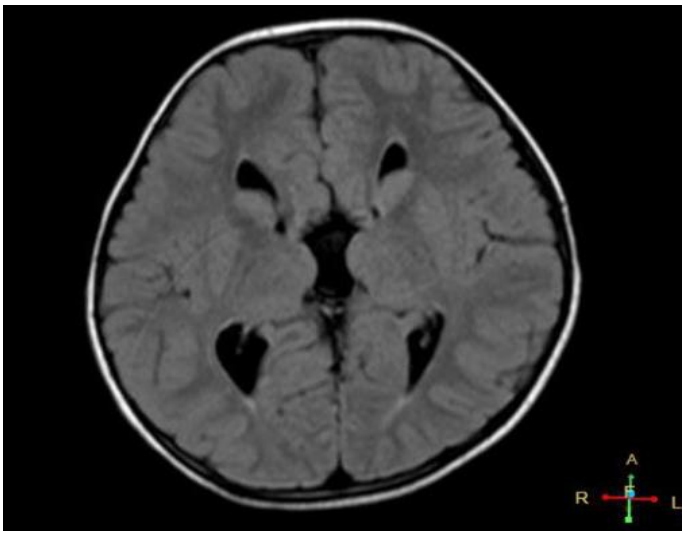

Fig.2: T2-weighted image shows a left coloboma cyst located in the posterior portion of the bilateral eyeball.

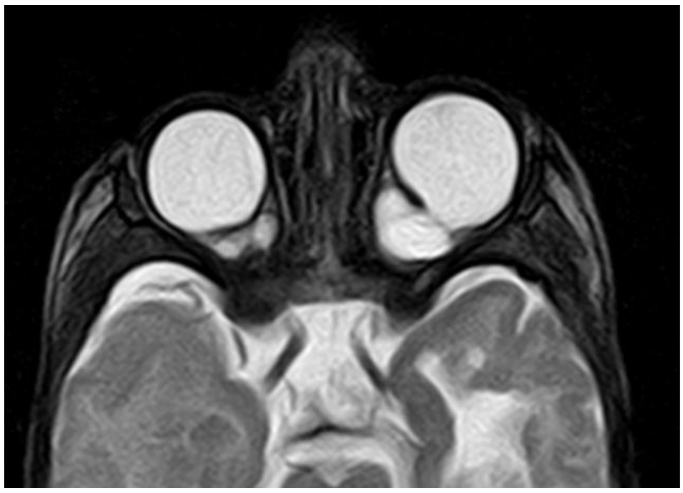

Fig. 3: Axial T1W image shows “racing car sign” characteristic of corpus callosum agenesis.

Based on the MRI results, the patient was referred to the pediatric neurology department. An electroencephalogram (EEG) was subsequently performed, showing frequent paroxysmal activity observed throughout the recording. This pattern consisted of widespread high-amplitude slow waves predominantly observed in the left hemisphere, which confirmed the diagnosis of infantile spasms.(see Figure 4)

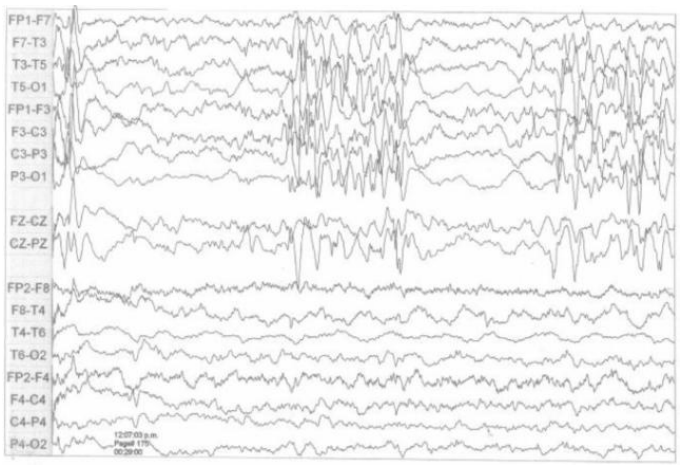

Fig. 4: An abnormal EEG revealed frequent paroxysmal activity across the entire recording, marked by generalized polypots (high voltage and slow waves primarily in the left hemisphere), there by confirming the diagnosis of infantile spasms.

Because of the infantile spasms and agenesis of the corpus callosum, suspicion of AS was raised. Consequently, the patient was referred to the ophthalmology department, where an ophthalmoscopy revealed multiple hypopigmented chorioretinal lesions consistent with chorioretinal lacunae (see Figure 5). With the confirmation of corpus callosum agenesis, infantile spasms, and chorioretinal lacunae, the classic triad indicative of AS was fulfilled, leading to a definitive diagnosis.

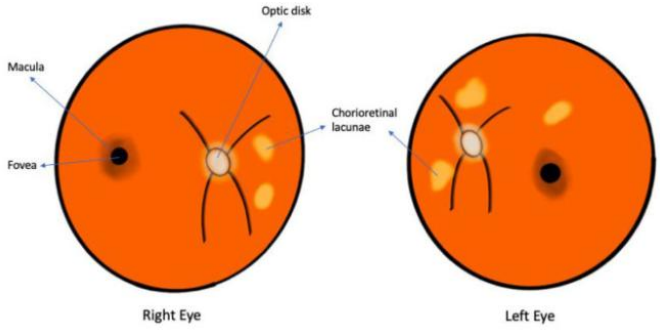

Fig. 5: Illustration depicting the patient's ophthalmoscopy. Bilateral chorioretinal lacunae are shown.

Treatment commenced with lamotrigine, valproic acid, levetiracetam, along with physiotherapy sessions and regular appointments with pediatric neurology. Currently, the patient continues to experience infantile spasms, albeit at a reduced frequency of 10-12 episodes per day.

Discussion

AS is an X-linked dominant genetic disorder predominantly affecting females [7]. The condition typically involves one of the two X chromosomes in females (XX), allowing for survival, whereas in males (XY), the mutation is usually lethal due to the affected X chromosome [8]. However, AS has been reported in males with Klinefelter syndrome (XXY), possibly due to the presence of an additional X chromosome providing survival [9]

AS commonly presents within the first three months of life with infantile spasms, characterized by frequent episodes of rapid muscle contractions and upper limb hyperextension [5]. In this case, the patient experienced between 12 and 20 infantile spasms daily, persisting despite anti seizure therapy initiation.

AS poses a diagnostic challenge due to its rarity, necessitating a multidisciplinary approach for confirmation. Some patients may exhibit interhemispheric cysts, which can mimic Dandy-Walker syndrome during prenatal ultrasound assessments, as observed in this instance [3]. Another critical differential diagnosis is Lennox- Gastaut syndrome, which shares clinical features such as tonic seizures resembling infantile spasms of AS [3].

Accurate diagnosis of AS requires the presence of the classic triad: agenesis of the corpus callosum, infantile spasms, and chorioretinal lacunae. Prenatal identification of this triad is not feasible; thus, clinical suspicion is essential for early detection, particularly in cases involving brain cysts [4]. Complete agenesis of the corpus callosum, as seen in this patient, is reported in approximately 72% of AS cases, with partial agenesis occurring in the remaining 28% [10].

In addition to the classic triad, this patient presented with a multiseptated interhemispheric cyst communicating with the ventricular system, a common finding in AS affecting nearly 50% of patients. These cysts, originating from the choroid plexus, can vary in size and, in rare cases, lead to hydrocephalus if they compress the aqueduct [8].

Imaging plays a pivotal role in AS diagnosis, with features like agenesis of the corpus callosum and choroid plexus cysts strongly suggestive of the condition [8]. Early suspicion based on MRI findings facilitated the patient's referral to pediatric neurology for definitive diagnosis. Typical EEG patterns in AS include disorganized basal activity and hypsarrhythmia, characterized by independent paroxysms between hemispheres due to corpus callosum agenesis, known as "split-brain" phenomenon [12].

Treatment of AS primarily involves anticonvulsant medications, although therapeutic response can be limited, necessitating adjunct therapies such as the ketogenic diet. Studies suggest varying efficacy of the ketogenic diet, particularly beneficial for patients without initial infantile spasms [13].

Patients with AS frequently present with congenital abnormalities like vertebral anomalies and rib abnormalities, requiring ongoing physical therapy and orthopedic monitoring [8]. In this case, mild scoliosis necessitated physical therapy to manage potential future progression due to muscle imbalance from absent ribs [14].

AS has a grave prognosis, with a median survival age of approximately 18.5 years. Respiratory complications, often related to hypersecretion, are the leading cause of mortality [5, 15]. Factors associated with a better prognosis include late-onset infantile spasms, partial corpus callosum agenesis, and smaller chorioretinal lacunae [16, 17].

Conclusions

Diagnosing AS necessitates a multidisciplinary approach involving ophthalmology, radiology, and pediatric neurology to identify the classic triad and confirm the disease accurately. Clinical suspicion of AS should be considered in all infants exhibiting stiffening of the arms and legs, prompting mandatory MRI and electroencephalogram assessments to establish the diagnosis. Additionally, screening for costovertebral anomalies is essential in all patients diagnosed with AS.

Patient consent

Verbal and signed consent was obtained from the patient concerned. The study was conducted anonymously.

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