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AICARDI SYNDROME: CASE REPORT

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ABSTRACT
Aicardi syndrome is a congenital disorder characterized by severe psychomotor retardation, corpus callosum agenesis, chorioretinal lacunae, and early-onset infantile spasms. The prognosis is generally poor for children with the classical form. We reporta case of Aicardi syndrome characterized by corpus callosum hypoplasia, chorioretinal lacunae, and seizures.

KEYWORDS: Aicardi Syndrome; hypoplasia ofcorpus callosum; seizures.

INTRODUCTION

Aicardi Syndrome is a rare genetic disorder described in 1965.[1] The main manifestations include multiple cranial, ocular, and skeletal malformations. It is theorized to be caused by defect in the X-Chromosome.[2]

A significant number of Aicardi syndrome girls are of normal birth and of normal development until three months of age when infantile spasms begin. Infantile spasms are the first symptoms that urge the clinicians to prescribe further investigations.[3]

CASE REPORT

A 29-day- old girl was admitted for evaluation of abnormal movements. The girl was born after a normal pregnancy and delivery with an Apgar score 6 ^{1} min, 8 ^{5} mins to $G_4P_4L_3D_1$, 36-year-old mother with history of parental consanguinity. Birth weight was 3500 g. The family history was contributory to death of a sibling at 1 year 11 months of age who started to develop abnormal movements at age of 3 months, and spasticity.

At day 10 of life, patient started to have episodes of chocking while feeding and perioral cyanosis, she was diagnosed with gastroesophygeal reflux for which she was started on Ranitidine.

At 29 days of life, she presented to our care with abnormal movements in the upper and lower extremities, uprolling of eyes, and lip smacking as well as chocking like episodes that didn't resolve on medication and spasticity.

Upon physical examination, patient had hyper extended neck, spastic extremities, arched position of the body when held supine, weak Moro reflex, poor suck reflex, and poor swallowing, intact deep tendon reflexes.

Investigations showed normal Serum Calcium, Electrolytes, TSH and CPK. Brain MRI showed hypoplasia of the corpus callosum (Figure.1), EEG showed presence of low voltage activity located to both temporal area and predominant on the left side and furthermore it showed absence of spindles. Funduscopic examination showed bilateral pallor of optic nerve disks and patches of chorioretinitis (lacunea). Neonatal screening was normal. Our patient was started on antiepileptic medications (Phenobarbital and Phenytoin).

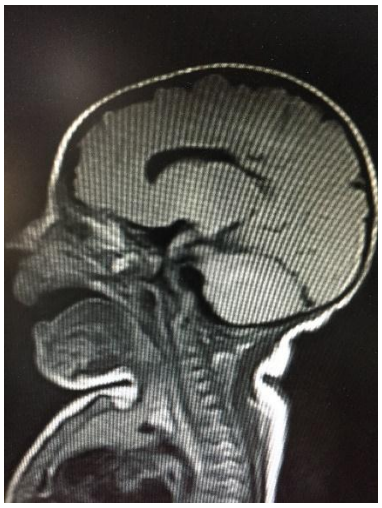

Figure 1: MRI brain that showed hypoplasia of corpus callosum.

DISCUSSION

Aicardi described a combination of agenesis of the corpus callosum and infantile spasms with ocular abnormalities, in 1965, which comprises the syndrome that now bears his name.[1]

It is a rare genetic disease, 853 cases are estimated in the US and over 4000 worldwide.[4]

It appears to be inherited in an X-linked dominant pattern owing to a mutant gene on the X chromosome that is lethal in XY male, however, it can also occur in males with Klinfelter Syndrome (47, XXY).

The main pitfall in the diagnostic procedure is the eye examination, since the chorioretinal lacunae may be confused with findings in cases of intrauterine infection, especially in children/infants.[5]

A significant number of Aicardi syndrome girls are of normal birth and of normal development until three months of age when infantile spasms begin. Infantile spasms are the first symptoms that urge the clinicians to prescribe further investigations.[3]

Cerebral malformations like polimicrogyria, grey matter heterotopias, intraventricular cysts, poroencephalic cysts and chorid plexus papillomas coexist at times. Occasional finding are: abnormalities of ribs and spine, cleft palate and ocular anomalies.[3]

Psychomotor retardation is usually severe; neuromotor retardation is commonly present with lack of motor and language skills.[6]

American researchers have recently defined the typical facial features of the affetcted females: prominent maxilla, upturned nasal tip, decreased angle of the nasal bridge, sparse lateral eyebrows.[7]

Several prognostic factors have been considered. Patients having retinal lesions five times the size of the optic disc diameter show poorer cognitive outcome.[8] Better prognosis was observed in those patients with fewer ocular anomalies, particularly if one eye was spared.[9] Indeed, unilateral localization of the retinal anomalies and the absence of macroscopic migration disordersto be the basis of the favorable cognitive development observed in their patient.

An increased incidence of dermatologic vascular malformations, pigmentary lesions, intracranial tumors like choroid plexus papillomas, gastrointestinal complications and hormonal imbalance leading to precocious puberty or delayed puberty have also been reported.[10]

Available data do not support a direct relationship between the severity of brain malformations and cognitive development; for example, partial versus complete agenesis of the corpus callosum[8] and cortical heterotopias appear not to influence visual functions and psychomotor performance.

In conclusion, the absence of a reliable genetic or biochemical marker results in the diagnosis of Aicardi Syndrome still based on clinical and neuroradiological grounds.

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