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A CLASSIC CASE OF AICARDI'S SYNDROME

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ABSTRACT

Aicardi's syndrome is an X-linked congenital disorder characterized by a constellation of anomalies mostly affecting the central nervous system, the most important one being the dysgenesis of corpus callosum, in a female neonate typically presenting with infantile spasms. MR imaging is the investigation of choice in detecting the anomalies and evaluating the extent of the abnormalities associated with this syndrome. In this report, we present a typical case of Aicardi's syndrome with classic imaging findings thus confirming the efficacy of MR in evaluation of the condition.

Key words: Aicardi's syndrome, Corpus callosum, MR imaging, Congenital.

CASE REPORT

A 54 day old female neonate presented with periods of infantile spasms and was referred for MRI. She was born following normal vaginal delivery and the parents noticed spasms since 1 month.

Findings

Figure 1:Axial T1 weighted image shows complete agenesis of corpus callosum with aninterhemispheric cyst not communicating with lateral ventricles and third ventricle. A cyst involving right choroid plexus and an isointense lesion involving left choroid plexus, possibly papilloma.

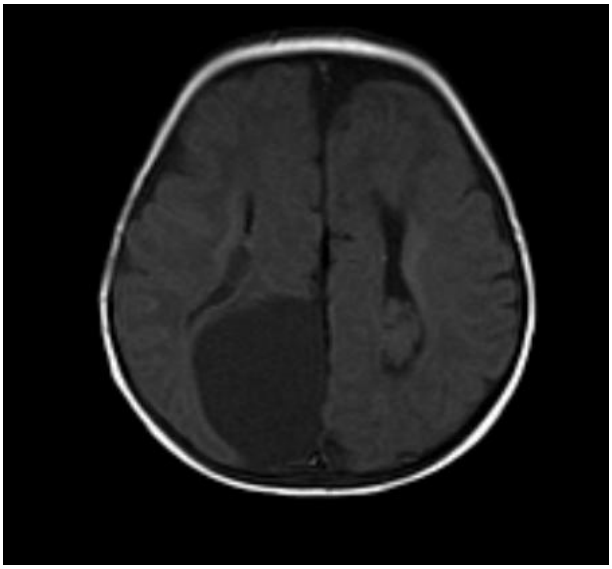

Figure 1

F**igure 2: Sagittal T2 weighted image shows complete agenesis of corpus callosum**.

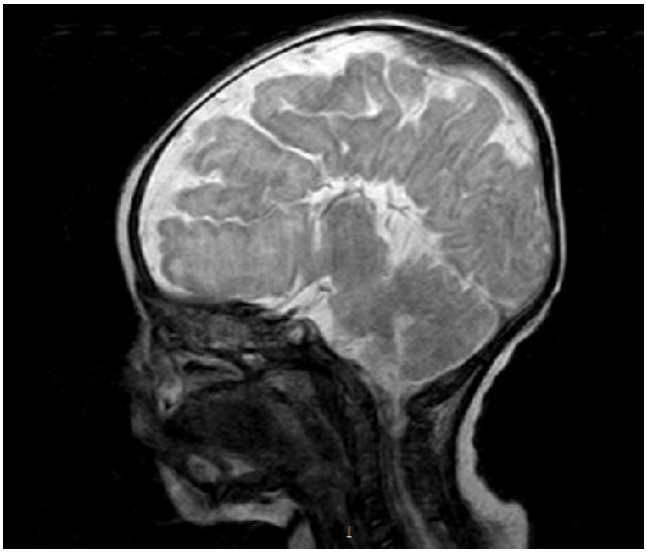

Figure 2

Figure 3: Axial T2 weighted image shows complete agenesis of corpus callosum with interhemispheric cyst. A cyst involving right choroid plexus and papilloma involving left choroid plexus. Polymicrogyria involving bilateral frontal cortices. Left sided ventriculomegaly. Subcortical and subependymal nodular heterotopia in relation to left lateral ventricle.

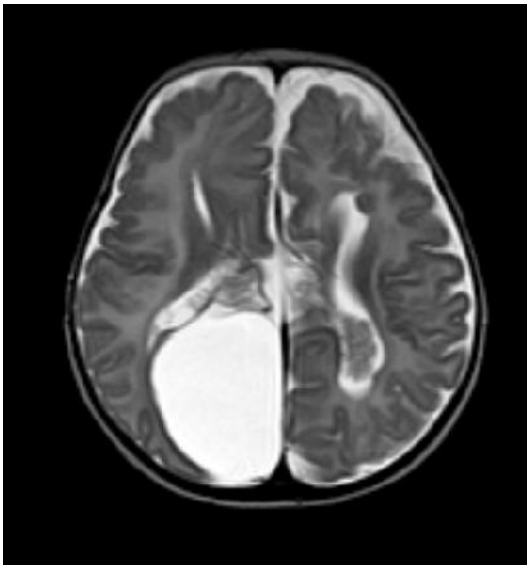

Figure 3

Figure 4: Axial T2 weighted image involving the orbit shows right coloboma.

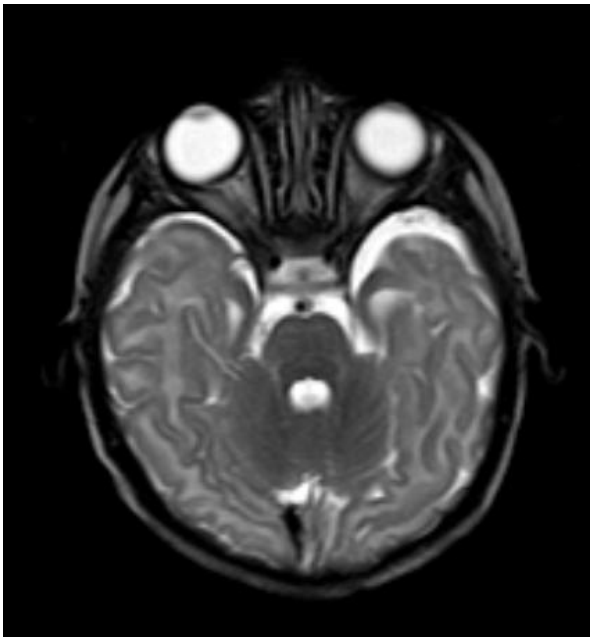

Figure 4

Figure 5: Axial T2 weighted image showing bilateral cerebellar heterotopias.

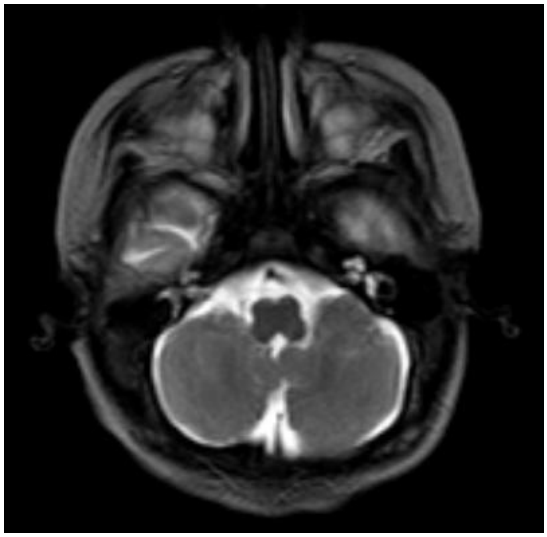

Figure 5

Figure 6: Coronal T2 FLAIR image showing complete agenesis of corpus callosum with interhemispheric cyst. Right choroid plexus cyst. Left choroid plexus papilloma.Left ventriculomegaly with subependymal nodular heterotopia.

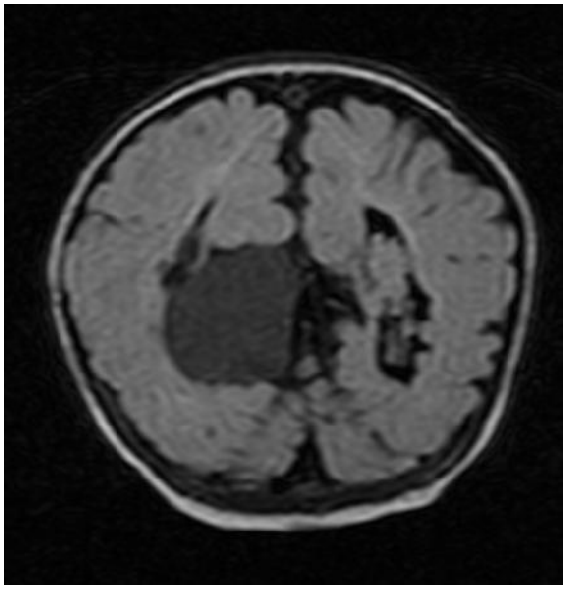

Figure 6

Considering the clinical history and the constellation of findings the final diagnosis of Aicardi syndrome was made.

DISCUSSION

Aicardi's syndrome was first described by Aicardi et al in 1965 as an X-linked dominant disorderclassically characterized by a triad of infantile spasms, callosaldysgenesis, and distinctive chorioretinal lacunae resulting from spontaneous balanced translocation of X chromosome[1, 5].

It is a rare neurodevelopmental disorder and the constellation of abnormalities include congenital chorioretinal lacunae, corpus callosum dysgenesis, seizures, polymicrogyria, cerebral heterotopias,choroid plexus cysts and papillomas.[2,3,4]

Hundred percent associations is seen withpolymicrogyria that was predominantly frontal and perisylvian and often associated with underopercularization. Periventricular and subcortical nodular heterotopias may also be present. Posterior fossa abnormalities like cerebellar heterotopias and tectal enlargement are also found are some cases of Aicardi's syndrome. [2, 3, 4]

CONCLUSION

It can be seen thatimaging in multiple planes is essential in detecting and assessing the extent of the intracranial and intraorbital abnormalities seen in Aicardi's syndrome. The extent of the abnormalities determines the prognosis and so multiplanar MR imaging is the investigative tool optimally suited for such purposes. This report thus confirms the adequacy of MR imaging in demonstrating the brain parenchymal lesions, intraventricular lesions and orbital anomalies seen with Aicardi's syndrome.

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