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CASE REPORT

A MALE PATIENT WITH AICARDI SYNDROME: A RARE CASE REPORT

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ABSTRACT

Aicardi syndrome a rare genetic disorder. It is even rarer in male as it is an x-linked disorder. It is characterized by infantile spasm, agenisis of corpus callosum and chorioretinal lacunae. Here we, present a case where a male patient presented with features of Aicardi syndrome at the age of 30 years.

INTRODUCTION

Aicardi syndrome is a rare genetic disorder first described by Dr Jean Dennis Aicardi and characterized by infantile spasms, agenesis of corpus callosum and chorioretinal lacunae[1].Aicardi Syndrome is theorized to be caused by defect in the X-Chromosome [2]. It is usually fatal in male. Few cases have been reported in male but most of them were 'XXY'. The estimated incidence in the United States is 1 in 105,000 live births[3]. Here we present a case of a male who presented with features compatible with Aicardi syndrome.

Case report

A 30yrs old male, born of nonconsanguinous marriage presented with multiple episodes of focal seizure with secondary generalization. He had first episode of convulsion at the age of 11 months, since then he had multiple episodes of convulsions. Each episode was preceded by aura. Each episode lasted for around 1-2 days with each convulsions lasting for around 2 minutes with patient being drowsy in between. There was history of infantile spasm. Mother, also, gave history suggestive of moderate mental retardation due to which he couldn't complete his primary schooling. No history of involvement of any cranial nerve. On the day of admission, patient was stuporous, pupils were equal in size with equally reacting to light, plantar bilateral extensor, vitals were stable, other systemic examination were essentially normal. He was put on inj. Levetiracetam 1gm loading dose, then 500 mg i.v. BD. After 1 day of admission patient became conscious. His speech was dysarthric. Ophthalmological examination revealed peripapillary chorioretinal lacunae with pale optic disc. There was no neurocutaneous marker. His initial investigations revealed, Hb 13gm%, leukocyte 10500/cumm, $N_{65}L_{31}M_3E_1$, platelet 165000/cumm, FBS 88mg/dl, urea 48 mg/dl, creatinine 0.76 mg/dl, Na 141 meq/l, K 4.0 meq/l, calcium 9mg/dl, LFT within normal limit . MRI brain showed encephalomalacia with gliosis or schizencephaly with focal atrophy in right parieto-occipital lobe, agenesis of splenium and adjacent body of corpus callosum (Image 1a & 1b). Interictal EEG was normal.

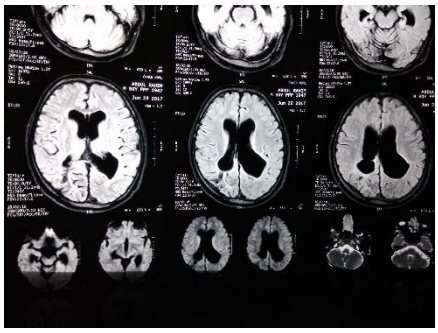

Image: MRI brain 1a

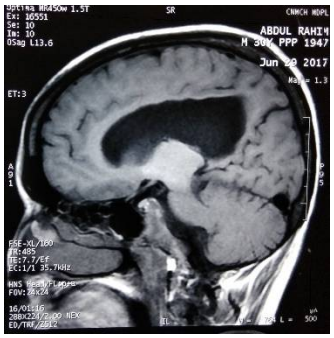

Image: MRI brain 1b

The patient was discharged in stable condition with advice of, tablet levetiracetam 500 mg BD. Since discharge patient is on regular follow up and he did not have any episode of convulsion till date.

DISCUSSION

Aicardi syndrome (AS) is an x-linked dominant syndrome characterized by the classical triad[1] of Partial or complete absence of the corpus callosum, "lacunae" of the retina, infantile spasm. But with advancement of time and investigation, larger spectrum of the disease has been recognized with full and partial triad and it had been found that higher functioning AS individuals do exist[4]. Other associated features include microcephaly, polymicrogyria, porencephalic cysts and enlarged cerebral ventricles due to hydrocephalus, iris coloboma, skeletal deformity, basal ganglia calcification, gastrointestinal structural abnormalities. The mutation appears to be de novo[5,6]. Most patients die early with average age of death 8.8 years with oldest patient being of 32 years of age[7]. Only few male cases have been reported till date[8].

Our patient is a 30 years old male who presented with repeated episodes of convulsion since childhood with history of infantile spasm and chorio retinal lacunae. The patient being male increases the uniqueness of the case.

CONCLUSION

Aicardi syndrome is a rare disorder, even rarer in male with most patients die early in their life but here we, present a case where a male patient presented with features of Aicardi syndrome at the age of 30 years.

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