Available Online at http://www.recentscientific.com

ISSN: 0976-3031

International Journal of Recent Scientific Research
Vol. 6, Issue, 3, pp.3000-3001, Marach, 2015

International Journal of Recent Scientific Research

RESEARCH ARTICLE

AICARDI SYNDROME. A RARE CASE REPORT WITH REVIEW OF LITERATURE

Mir Sumsam Ali Khurram, C.N. Prasad and Syed Muzammil Hassan

Department of Pediatrics, Deccan College of Medical Sciences, Hyderabad, T.S., India

ARTICLE INFO

Article History:

Received 2nd, February, 2015
Received in revised form 10th, February, 2015
Accepted 4th, March, 2015
Published online 28th, March, 2015

Key words:

Aicardi Syndrome, agenesis of corpus callosum, infantile spasm, mental retardation.

ABSTRACT

Aicardi Syndrome is a rare neuro developmental disorder identified by Jean Aicardi in 1965 Characterized by infantile spasm, agenesis of corpus callosum and ocular abnormalities. We report a case of 18 months old male child with Aicardi Syndrome, who presented with Seizure, mental retardation and characteristic eye lesion in which a diagnosis was made on C.T. Scan or MRI Scan of Brain.

INTRODUCTION

Aicardi Syndrome is a rare genetic disorder described by Jean Aicardi, a French neurologist in 1965 (1) Multiple cranial, ocular, and Skeletal malformations are main manifestations of the patients. Aicardi Syndrome is theorized to be caused by defect in the X-Chromosome (2). It is observed more commonly in females than the males. We report a case of 18 months old male child, who was recently managed at Owaisi Hospital and research centre a part of Deccan College of Medical Sciences, Hyderabad, Telangana State, India.

Case Report

An 18 month old male child presented to us with seizure of the complex partial type and myoclonic type since the age of 8 months. He was born by C-Section at term to a primigravida mother with history of delayed cry.

The parents were consanguineous marriage and there was no antenatal illness. At present there is marked developmental delay, mental retardation and seizure. On examination, the child had hypotonia, microphthalmia of right eye (Fig:1) Spastic Limb, normal deep tendon reflexes and bilateral extensor plantar response. Investigation revealed leucocytosis. Serum Calcium and Serum Electrolytes were normal, X-Ray of Chest, Spine and skill were normal. Funduscopic examination showed bilateral, variable size, discrete, two luci of pale areas with surrounding pigmentation.

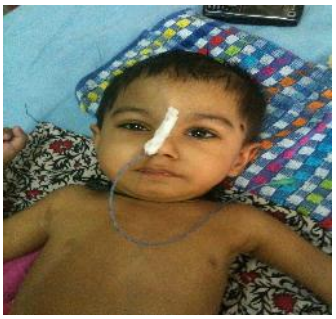

Figure 1 Cinical photograph showing micro opthalmia of right eye.

EEG showed characteristic pattern of hypsarrhythmia. C.T. & M.R.I. Scan of brain (Fig: 2) and (Fig: 3) showed ventriculomegaly and agenesis of corpus callosum.

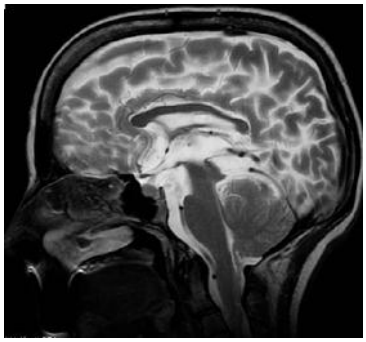

Figure 2 C.T. Scan of brain shows Ventriculomegaly and partial agenesis of corpus callosum.

*Corresponding author: **Mir Sumsam Ali Khurram**
Department of Pediatrics, Deccan College of Medical Sciences, Hyderabad, T.S., India

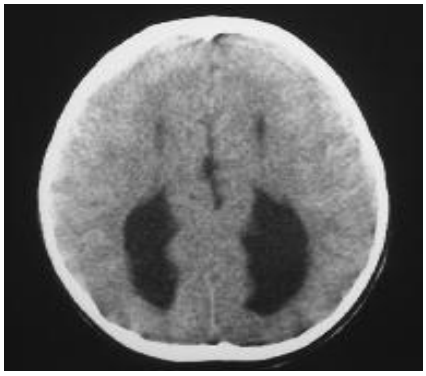

Figure 3 C.T. Scan of brain showing ventriculomegaly and agenesis of corpus callosum suggestive of aicardi syndrome.

Karyotyping showed normal 46XX pattern. The child was treated with broad spectrum antibiotics and antiepileptic drugs and discharged on oral eption and levipil.

DISCUSSION

The present case had the salient features of Aicardi syndrome viz agenesis of corpus callosum, chorioretinal lacunae and infantile spasm. It may be associated with other anomalies (3). The accurate etiology is not identified. Aicardi syndrome 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. All the cases are sporadic, not promising due to new mutation (5, 7). The analysis of Aicardi syndrome is based on clinical facial appearance including the pathognomonic chorioretinal lacunae identified on ophthalmic examination, brain magnetic resonance imaging finding and skeletal finding. Management of Aicardi Syndrome requires antiepileptic drugs for seizure control. Improve outcome with vigabatrin have been reported. Seizure start early in Aicardi Syndrome and have always has been myoclonic in type. The eye assessment of the patient usually shows unilateral or bilateral ocular anomalies.

Funduscopic assessment shows bilateral, variable sized discrete, two loci pale area with surrounding pigmentedl(chorioretinal lacunae). This findings had been observed in our case. The patient presented with early onset infantile spasm, severe mental retardation, delayed milestone, loss of speech, microphthalmia and coloboma of optic disc (4). The most common abnormalities together with microgyria, polygyria, hydrocephalus, hemivertebrae, fusion of vertebrae, spinabifida may be connected with Aicardi Syndrome (6, 8). None of above findings was found in our case. Most affected children have 46xx karyotype. Only two cases have reported in literature in males, one of them had 47xxy and other had 46xx karyotype.

This disorder is lethal in hemizygous male, the location of the gene in Aicardi Syndrome is XP22.3area (9). The prognosis of the patient depending on the severity of the symptoms and life expectancy limited from months to few years. In our case a positive prognosis could be expected, because of infantile spasm have not residenrial since birth. A constructive prognosis is predictable in our case, and because the agenesis of corpus callosum is partial and chorioretinal lacunae are relatively small in size (10).

CONCLUSION

We are presented a case with Aicardi Syndrome, who manifested the infantile spasm, mental retardation, and funduscopic examination could be a great help in the diagnosis of this rare genetic disorder.

References

1. Aicardi J, Lefebra J, Lerique Koechlin A. 1965. A new syndrome spasm in flaxions, callosal agenesis, ocular abnormalities Electroencephalogr clin Neurophysio., 19:609-610
2. Bertoni JM, Von Loh S, Allen RJ. 1979. The Aicardi Syndrome: Report of 4 cases and review of literature Ann Neural., 5:475 – 482
3. De Jang JGY, Deiieman JW, Houben M, Manschot WK, de Minjer A. 1976. Callosum, infantile spasm, ocular anomalies (Aicardi Syndrome). Clinical and pathological findings. Neurology, 26:1152-1158
4. Font RL, Marines HM, Cartwright JrJ, Bausecman SC. 1991. Aicardi Syndrome: A Clinicopathologic case report including electron microscopic observations. Opthalmol., 98:1727-1731
5. Nadia RS, Joshi A, Vaidya SD, Manavi AB, Kelkar SR. 1985. The Aicardi Syndrome neuron India. 33:149 – 164
6. Park BR, Jung HJ, GO CJ. 1982. Sedys A case of Cardi's Syndrome. *J Korean Pediatric SOC*.,12:1285 –1288
7. Phillips HE, Carter AP, Kennedy Jr JL, Rosman NP, O, Conner JF. 1978. Aicardi Syndrome, Radiological manifestations Radiology, 127:435–465
8. Ropaers HH, Zuffardi O, Bianchi E, Tiepolo L. 1982. Agenesis of Corpos Callosum, ocular and skeletal anomalies. (X linked dominant Aircardi Syndrome in a girl with balanced X/3 translocation Humgenet, 61:364–368
9. Singhi PD, Gupta A, Agarwal A. 1991. Aicardi Syndrome Indian pediatr., 23:153 – 1516
10. Xacoub M, Missaouin, Tabari B, Ghorbel M, Tlilik, Selm H, Essouss A. 2003. Aicard Syndrome with favorable outcome Arch Paedi Atr., 10:530–532

How to cite this article:

Mir Sumsam Ali Khurram *et al*., Aicardi Syndrome. A rare case Report with review of Literature. *International Journal of Recent Scientific Research Vol. 6, Issue, 3, pp.3000-3001, March, 2015*

****