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PHYSIOTHERAPY

AICARDI SYNDROME – A RARE CASE REPORT

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Abstract

Aicardy syndrome is a rare genetic disorder identified by French Neurologist ,Dr.Jean Aicardy in 1965.The number of identified cases of children with Aicardy syndrome is very difficult to assess accurately, and it affects only females and in very rare cases ,males with Klinefelters syndrome. Aicardy syndrome is characterized by the absence of corpus callosum, either partial or complete, infantile spasms, lesions or lacunae of the retina of the eye that are very specific to this disorder, microcephaly, and procencephalic cysts. We present a case report of Aicardy syndrome which has come to our hospital OPD, the patient was 7 year old girl, having severe spasm during flexion, and scoliosis. The patient was mentally retarded, with a dental abscess, and cleft palate. Once the patient was identified with Aicardy syndrome, she has subjected to detailed evaluation for other abnormalities.

Keywords: Corpus callosum, Microcephaly, Cleft palate, Scoliosis

Introduction

Aicardi syndrome is a rare genetic disorder identified by the French Neurologist, Dr. Jean Aicardi in 1965.The number of identified cases of children with Aicardi syndrome is very difficult to asses accurately. Aicardi syndrome is a rare genetic malformation syndrome characterized by the partial or complete absence of a key structure in the brain called corpus callosum, the presence of retinal abnormalities [1], and seizures in the form of infantile spasms. Aicardy syndrome is theorized to be caused but a defect on the X chromosome as it has thus far only been observed in girls or in boys with Klinefelters syndrome [2]. Symptoms typically appear before a baby reaches about 5 months of age. Almost all reported cases of Aicardy syndrome have been in females. Currently, there are 500 reported cases world wide ,with only one case of siblings affected with condition. There fore it is believed this disorder is a new mutation and not all of the family members may carry the defective gene [3]. The few males that have been identified with Aicardy syndrome have proved to have 47 chromosomes including an XXY sex chromosome compliment. Aicardy syndrome appears to be lethal in normal males who have only one X chromosome.Aicardy syndrome appears to be inherited in an X –linked dominant pattern due to mutant gene on the X chromosome that is lethal in XY males [4]. All cases of Aicardy syndrome are thought to be due to new mutations. No person with Aicardy syndrome is known to have transmitted the X –linked gene responsible for the syndrome to the next generation. The characteristic features of Aicardi syndrome are partial or complete absence of the corpus callosum in the brain, eye abnormalities known as lacunae of the retina that are quite specific to this disorder and the development of seizures called infantile spasms [5]. Other types of defects of the brain such as microcephaly, prosencephalic cysts, and enlarged cerebral ventricles due to hydrocephalus are common in Aicardi syndrome. Treatment of Aicardi syndrome primarily involves management of seizures and continuing intervention programs for developmental delays. Almost all people reported with Aicardi syndrome have experienced developmental delay of a significant degree, typically resulting in moderate to found mental retardation.

Case Report

A seven year old girl was admitted in our hospital, with quadriplegia, spasticity with marked involvement of left lower and she was mentally retarded. On spiral CT scan we diagnosed the absence of corpus callosum. Collected the medical history of the patient, after the 3rd month of birth the parents have noticed the upward movement of eye ball and limb convulsions. At third month also the child did not achieved social smile she was not following light tracking. From third month to till 6th year child was under many medications. At present history there is marked developmental delay, mental retardation and seizure. The child does not have head control, trunk control, head movements possible only supine with assistance. Limb resistance with rigidness in movements to exercise, absence of all reflexes .In detailed examination we found the shoulder was protracted and elevated, elbow was flexed with supinated forearm and extended wrist. Right thoracic cage was convexed and pelvis was tilted anteriorly.

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Discussion

Treatment of Aicardi syndrome primarily involves management of seizures and early continuing intervention programs for developmental delays. Treatment is indicated as follows in patients with Aicardi syndrome; use conventional epilepsy therapies for the many possible seizures manifestations, infantile spasm requires specific interventions and is typically unresponsive to conventional anticonvulsants [6]. This seizure type may be especially recalcitrant to therapy. Guidelines for the medical treatment of infantile spasms have been established by the American Academy of Neurology and the Child Neurology Society. Adrenocorticotropic hormone is effective for some patients and should be considered. Vigabatrin, a more recently introduced therapy for infantile spasm, blocks gamma –aminobutyric acid (GABA)-T, an enzyme that breaks down GABA the major inhibitory neurotransmitter in the brain [7]. Although concerns have been raised about possible ophthalmologic sequelae after using vigabatrin, it has been effective for infantile spasms without the serious life threatening adverse effects of ACTH. If retinal disruption from the congenital insult is deemed vigabatrin. Profound mental retardation, immobilization, seizures and scoliosis may contribute to cardiopulmonary dysfunction. Patients have a shortened life span and commonly die from pulmonary infections [8]. Consultation with a neurologist is probably needed during the first tear of life. A pediatric ophthalmologist is best able to confirm retinal lacunae, consult an orthopedic, pulmonary or gastroenterologist if complications arise from scoliosis, pulmonary function, or feeding or aspiration difficulties [9]. No information has been published on cortical resection or the use of vagus nerve stimulation for seizures in Aicardy syndrome. The patients are not advised to be specific for their dietary supplements, but if they use ketogenic diet to control seizures associated with this condition, specifically infantile spasms, may be indicated [10]. Initiation of a detailed discussion with family before therapy about the multiple medications available and potential complications of each and individualize treatment to best suit the patient and the capabilities and wishes of the family[11].Currently no cure for Aicardy Syndrome. A symptomatic treatment may be recommended to manage the seizures, usually in the form of anti-seizure medicines. A few intervention programs to manage mental retardation are also administered. Physical therapy is also recommended to aid the child in her development. The life expectancy for an affected infant is dependent on how severe the condition-is.

Figure 1 (a) Axial MRI T2-weighted image at level thalami, and (b) coronal MRI T2-weighted image at level of the occipital lobes. The left choroid plexus is large and contains large high signal round lesions (arrows). There is absence of the corpus callosum

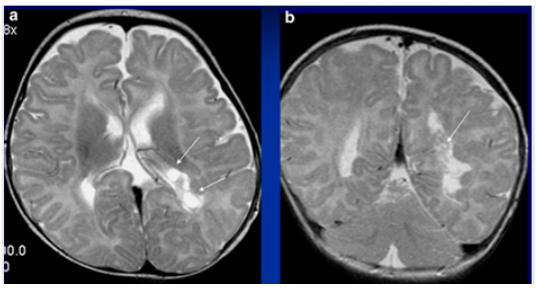
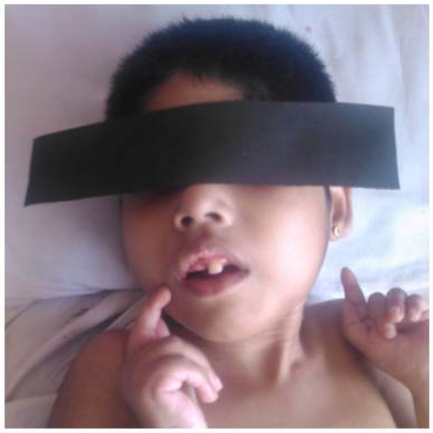


Figure 2: A seven year old girl with quadriplegia, spasticity with marked involvement of left lower and mentally retarded



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