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CHOROID PLEXUS PAPILLOMA AND AICARDI SYNDROME

Case report

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ABSTRACT - A case of Aicardi syndrome associated with a choroid plexus papilloma of the third and both lateral ventricles in a 15 months old baby girl is repported. The tumor was completely removed via three craniotomies. Reports of the literature with the association of choroid plexus papilloma and Aicardi syndrome are rare. We suggest that children diagnosed with Aicardi syndrome should routinely undergo imaging studies of the brain, such as computed tomography or magnetic resonance.

KEY WORDS: Aicardi syndrome, choroid plexus papilloma, hydrocephalus, corpus callosum agenesis.

Papiloma do plexo coróideo e síndrome de Aicardi: relato de caso

RESUMO - Relatamos o caso de uma criança com 15 meses de idade, portadora da síndrome de Aicardi associada a tumores nos ventrículos laterais e terceiro ventrículo (papilomas) que foram retirados cirurgicamente através de três craniotomias. A ocorrência de papiloma do plexo coróideo associada à sindrome de Aicardi é raramente descrita na literatura. Sugerimos que as crianças portadoras da síndrome de Aicardi sejam rotineiramente submetidas a estudo radiológico do encéfalo através da tomografia computadorizada ou ressonância nuclear.

PALAVRAS-CHAVE: síndrome de Aicardi, papiloma do plexo coróideo, hidrocefalia, agenesia do corpo caloso.

Aicardi[1] described a syndrome characterizided by corpus callosum agenesis, myoclonic seizures and visual disturbances in 1965. After the original report, Aicardi (1986) described other changes that are usually associated with the clinical presentation, such as skeletal malformation and development delay. This disease was reported only in girls. Since Aicardi description[1], several reports have included other manifestations to the clinical picture, as cytomegalic inclusion[2], scoliosis and hemivertebrae[3], cleft palate[4], craniofacial asymmetry[5], hearing anomalies[5], tumors of the central nervous system (CNS)[2-7], choroid plexus cysts[7]. The association of Aicardi syndrome and choroid plexus papilloma has been reported in the literature (ten cases having already been described[2-7]). This association is rare, nevertheless it may be regarded as an integral part of the disease[2,3].

In this paper we present the case of a child diagnosed with Aicardi syndrome and choroid plexus papilloma in multiple location (lateral and third ventricles) treated by several surgeries.

CASE

Baby girl LVD was born in April 1994, at term by vaginal delivery, after a healthy pregnancy. She was the second daughter of a young non-consanguineous couple, with no history of neurologic disorders. It was already noted that she presented on strabismus, abnormal eye movements and seizures of the myoclonic type. A computed tomography (CT) showed diffuse dilation of the ventricles (Fig 1). She was partially controlled with anticonvulsivant drugs. On 15 months, it was observed that the head circumference increased beyond expectation. Another CT was performed and the patient referred to the pediatric neurosurgery service with the diagnosis of congenital hydrocephalus.

On admission in June 1995, the child showed evident signs of development delay, macrocephaly, with increased and bulging anterior fontanel, fundoscopic changes (colobomas) and infantile spasms (West syndrome). The quality of the CT was not adequate, but it showed ventricular dilation and suggested choroid plexus enhancement. Magnetic resonance image (MRI) (Figs 2 and 3) showed contrast enhancing lesions inside the both lateral and third ventricles, suggesting choroid plexus tumors. In July 1995, a right parietal craniotomy was performed, with radical re-

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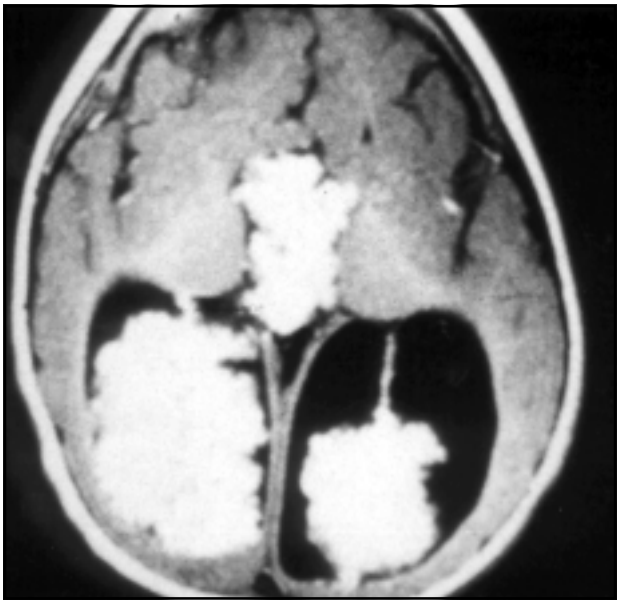
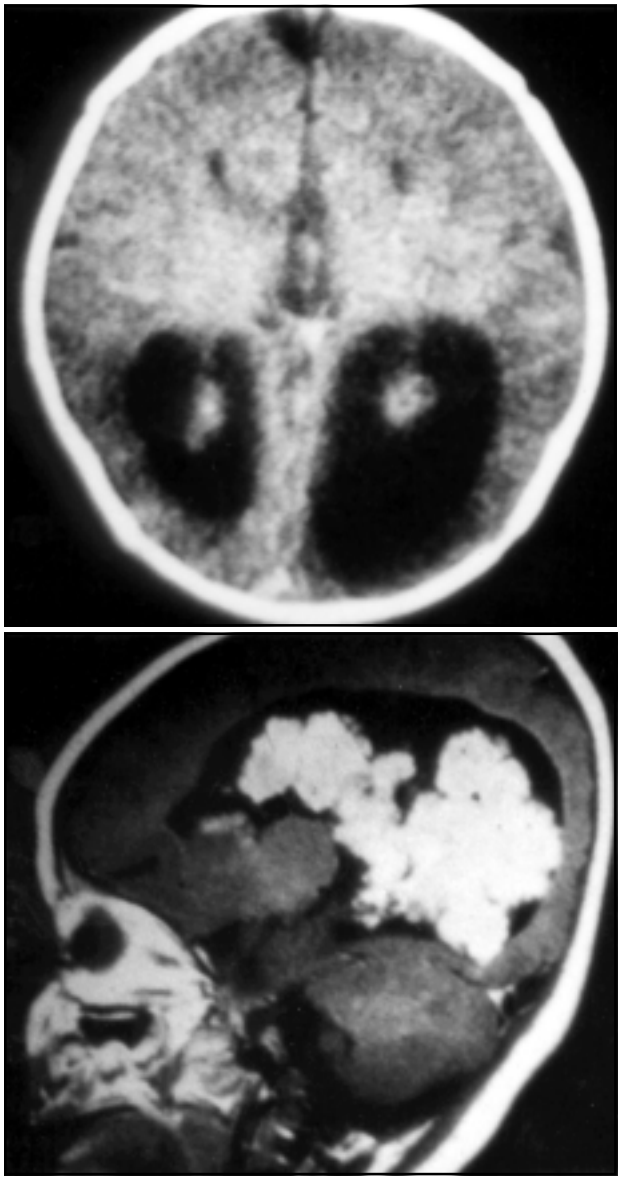


Fig 1. CT (without contrast) performed at one month of age, showing ventricular dilation.

moval of the lesion inside the right lateral ventricle. One month later, she was again operated on, a left parietal craniotomy performed with radical excision of the lesion inside the left lateral ventricle. In October of the same year, a right frontal craniotomy was performed and radical removal of the third ventricle lesion was achieved. It turned out easier on account of the corpus callosum agenesis. The pathological examination always disclosed choroid plexus papilloma. Ventricular drainage was necessary in December 1995 and a ventriculo-peritoneal shunt then was inserted. Follow-up in October 2001, six years after diagnosis, showed a child with the characteristic cognitive deficit of Aicardi syndrome and good seizure control with the help of medication.



Figs 2 and 3. Axial and sagittal T1 weighted MRI following gadolinium administration show the presence of tumoral lesions inside both lateral and third ventricles, and agenesis of the corpus callosum.

DISCUSSION

It appears that the Aicardi disease is a hamartoma[5], as neurofibromatosis and tuberous sclerosis, diseases that show a common association of heterotopias and intracranial tumors[5]. The clinical/radiological manifestations of the Aicardi syndrome[1], initially described as a triad of infantile spasms, total or partial agenesis of the corpus callosum and retinal changes, now include periventricular and subcortical heterotopias, choroid plexus cysts and tumors, colobomas, skeletal changes of vertebrae and ribs, microphthalmia, cerebral hemispheres asymmetry, cortical gyrus changes, tumors outside of CNS and cranialfacial alteration. A review of the literature[2-7] revealed ten cases in which the disease manifested in association with choroid plexus papilloma. These numbers should increase when children with Aicardi syndrome are routinely submitted to TC and/or MRI. The findings may be incidental, with no manifestation of increased intracranial pressure.

The patient here reported presented seizures of difficult control with medication, development delay and clinical evidence of hydrocephalus. The diagnosis of the tumors was made only after MRI. In the majority of cases reporting association of Aicardi disease and choroid plexus papilloma, the tumor was located in only one ventricle[2,4,6,7]. Tackibana et al.[3] described a case in which the tumors occupied the right lateral and the third ventricles. Hamano et al.[5] reported a case in which the CT showed tumors in three ventricles, however there were no pathological studies of these tumors.

The case presented here is apparently the first one to report a child with Aicardi disease and tumors in three ventricles (both lateral and third), that caused intracranial hypertension and were treated surgically with pathological diagnosis of choroid plexus papilloma. The patient is still alive, after more than six years after the diagnosis.

We conclude that all children with symptoms suggestive of Aicardi disease should undergo radiological studies of the brain, namely CT or MRI, and have their intracranal pressure clinically monitored to evaluated the need for surgical removal of the lesions.

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