PATHOGENOMONIC CHORIORETINAL LACUNAR LESIONS IN AN INFANT WITH AICARDI’S SYNDROME

CORIORRETINOPATÍA LACUNAR COMO PRESENTACIÓN DE SÍNDROME DE AICARDI EN EL LACTANTE

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ABSTRACT

Clinical case: We report the case of an 81-day-old female infant who was brought to the Emergency Department because of a seizure. At 20 weeks of gestational age she was diagnosed to have agenesis of the corpus callosum, with this being confirmed later by magnetic resonance imaging. Ophthalmological examination of the fundus showed peripapillary chorioretinal lesions (lacunar chorioretinopathy) in both eyes.

Discussion: Chorioretinal lacunar and retinal pigment epithelial abnormalities are the basis for the diagnosis of this syndrome (Arch Soc Esp Oftalmol 2007; 82: 311-314).

Key words: Lacunar chorioretinopathy, epilepsy, agenesis of the corpus callosum, infantile spasms, Aicardi syndrome.

RESUMEN

Caso clínico: Se presenta el caso de una niña de 2 meses y 3 semanas de vida que ingresa desde Urgencias por crisis convulsivas. Fue diagnosticada en la semana 20 de gestación de agenesia de cuerpo calloso, posteriormente confirmada en ecografía cerebral postnatal. En la exploración del fondo de ojo, se observaron lagunas retinianas (coriorretinitis lacunar) peripapilares en «sacabocados» en ambos ojos. La resonancia magnética craneal muestra agenesia completa del cuerpo calloso.

Discusión: Las lesiones atróficas coriorretinianas con alteración del epitelio pigmentario de la retina, son fundamentales en el diagnóstico del síndrome de Aicardi.

Palabras clave: Coriorretinitis lacunar, epilepsia, agenesia de cuerpo calloso, espasmos infantiles, síndrome de Aicardi.
INTRODUCTION

Aicardi’s Syndrome is a dominant genetic disorder linked to the X chromosome. It was first described in 1965 by neurologist Jean Aicardi1. It is extremely difficult to determine the exact number of children suffering from Aicardi’s syndrome, but it is estimated that between 300 and 500 cases have been described across the world.

It is characterized by the typical triad (1):
1. Ophthalmologic anomalies (the most specific being lacunar chorioretinopathy)
2. Agenesis of callous body (partial or full).
3. Infantile spasms.

Most cases are diagnosed before five months of age. Children develop normally until they turn three months old, the time when they begin to suffer infantile spasms, which are frequently asymmetrical and preceded by a tonic or clonic seizure limited to the area where spasms are predominant. Other neurological alterations that may be observed include hemiparesis, microcephaly, mental and motor retardation.

In most female infants, the electroencephalogram (EEG) reveals asynchrony and suppression of discharges during the first weeks of age, which signals the absence of interconnection between hemispheres.

The most specific finding is a more or less extensive unilateral or bilateral lacunar chorioretinopathy. In fifty percent of cases, a coloboma of the optic disk may be present. Other ocular alterations include microphthalmia, retinal pigment deposits, hypertelorism, absence of pupillary reflexes, strabismus or persistence of pupillary membranes.

The brain magnetic resonance is the most informative imaging test; it reveals in most cases malformations of the central nervous system (SNC) accompanying the agenesis of the callous body.

Most of the time, the existence of the classic triad helps to diagnose this syndrome. However, in rare cases, especially when no agenesis of the callous body is detected, the finding of at least two of the new main criteria (see Table I) should be enough to establish a diagnosis (2).

CASE REPORT

The case of a full term baby girl is described herein.

Pregnancy progressed normally with normal cytological results in the amniocentesis test, performed 16 weeks into pregnancy. On the 20th week, an agenesis of the callous body was diagnosed, later confirmed by the postnatal brain ultrasound.

Two months and three weeks later, the baby is admitted in the emergency room suffering from epileptic crises consisting of asymmetrical spasms with trunk and head flexion, lower extremities flexion or extension, head rotation and four-second duration ocular deviation conjugated upwards and to the right and repeating every 6-8 second interval and up to 90 spasms.

Hemogram, coagulation and general serum biochemistry were normal. EEG revealed upon admission an asymmetrical record with irritative activity and poor organization on the right hemisphere.

The ophthalmologic assessment revealed the existence of a bilateral lacunar chorioretinitis (images adjacent to the papilla). The remaining ophthalmologic study yielded no pathological findings.

In view of these clinical findings, Aicardi’s syndrome was suspected, and subsequently a brain magnetic resonance was requested. It revealed a full agenesis of the callous body (figs. 1 and 2) with associated anomalies: heterotopia of the grey matter along the right side ventricle (figs. 3 and 4), polymicrogyria and cyst of the choroid plexus along the right side ventricular atrium (fig. 5).

The costovertebral x-ray did not reveal abnormalities.

Table I. Diagnosis criteria for Aicardi’s Syndrome

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<thead>
<tr>
<th>Classic Triad:</th>
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<tr>
<td>Infantile spasms</td>
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<tr>
<td>Agenesis of the callous body</td>
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<tr>
<td>Lacunar chorioretinitis</td>
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<tr>
<td>New main criteria</td>
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<tr>
<td>Cortical malformations</td>
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<tr>
<td>Periventricular heterotopias</td>
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<td>Third ventricle/choroid plexus cysts</td>
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<tr>
<td>Choroid plexus papillomas</td>
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<td>Optic disk coloboma</td>
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<tr>
<td>New main criteria</td>
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<tr>
<td>Bone anomalies (vertebrae/ribs)</td>
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<tr>
<td>Microphthalmia (and other ocular alterations)</td>
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<td>Asymmetrical thickening of the brain hemisphere</td>
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<td>«Splint brain» in EEG</td>
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Aicardi’s syndrome is characterized by the association of ophthalmologic anomalies, epileptic crises (this manifestation is noticed more prematurely) which are difficult to bring under therapeutic control (3) or infantile spasms and partial or full agenesis of the callous body.

Its incidence and prevalence are lower and affect the female gender (it is fatal in males and has only been described in children with trisomy 46 XXY).
Among the most frequent ophthalmologic manifestations, we find round gaps corresponding to the chorioretinal atrophy, the key sign in the eye fundus diagnosis (fig. 6). Large coloboma papillas or associated staphylomas may also be found.

Less frequent are the associated strabismus, microphthalmy, persistent pupillary membranes and hypertelorism. (4), all of them considered as minor findings when diagnosing this syndrome (Table I).

Indirect binocular ophthalmoscopy in certain neuropediatric diseases is very useful. The study of the eye fundus allows discovering certain childhood diseases and avoiding in many cases complex diagnosis tests, the high cost associated to them and a more accurate approach to the pathology reported by the patient.

Other clinical manifestations associated with these patients are vertebral anomalies, generalized hypotony, the existence of telangiectasias, head and thorax deformities, facial asymmetries (5), cutaneous hemangiomas, hypoplasia of the fifth finger, low implantation of ears or psychomotor retardation significantly limiting the activity of patients.

There is no remedial treatment. Aicardi’s syndrome presents a very unfavorable neuroevolutionary prognosis, although some cases may present a moderate intellectual affection.

REFERENCES