ATYPICAL MACULAR COLOBOMA: A CASE REPORT

COLOBOMA MACULAR ATÍPICO: A PROPÓSITO DE UN CASO

LÓPEZ-GARCÍA JC¹, BUESA-GÓMEZ J², PARDO-SAIZ A¹, CALLIZO-TOMÁS J³

ABSTRACT

Clinical case: This was a 17-month-old boy who had macular retinochoroidal lesions in both eyes following maternal varicella during pregnancy.

Discussion: The scars were suggestive of congenital chorioretinal infection, but because of negative serology and the clinical picture, we believe the problems are atypical macular colobomata (Arch Soc Esp Oftalmol 2006; 81: 713-716).

Key words: Macular coloboma, macular coloboma, macular dysplasia, atypical macular coloboma, congenital maculopathy, macular chorioretinal scar.

INTRODUCTION

An atypical macular coloboma is an infrequent group of colobomae located in the macula instead of in the inferior or inferior-nasal region. It is difficult to explain this anomaly from the embryological viewpoint because it does not appear to originate due to defects in the closure of the fetal slit. This short communication presents a case of a child with bilateral involvement in which the lesions look similar to the chorioretinal scars caused by infections in the intrauterine environment. Serology must be utilized to exclude an infectious cause.

CASE REPORT

A male baby 17 months old, referred by his pediatrician for strabismus. He exhibited endotropy (15 degrees) and a small hypertropy in the right eye.
(RE), with the left eye (LE) being dominant. Visual acuity (VA) is low in the RE (the patient becomes irritated when the LE is covered) and acceptable in the LE (patient has gaze in focus and picks up small objects). The light reflex on the cornea leads us to suspect an eccentric fixation in both eyes (fig. 1).

The baby had suffered feverish convulsions. The family history includes the mother having varicella while being pregnant (between week 16 and 18) and the mother’s uncle having the Alport syndrome. The anterior pole is normal. In skiascopy, the neutral point is +2 in both eyes. Indirect ophthalmoscopy revealed oval lesions in both maculae, predominantly in the temporal half, suggesting long-evolving retinochoroiditis, with normal periphery. The lesions in the RE are larger (fig. 2), highly pigmented and with whitish gylial tissue in the center. The temporal arches are curved towards the pathological area, and temporal parapapillary atrophy is also found. The LE features a well defined chorioretinal atrophy, with pigmented edges (fig. 3), normal retinal vessels, with some choroidal vessels over the sclera with pigment spots. A scleral ectasia can be seen under the retina. The baby did not exhibit nystagmus or other ocular or systemic lesions. The cranium X-ray is normal.

As intrauterine ocular infection was suspected, serology was requested for TORCH (acronym for Toxoplasma, Rubeola, Cytomegalovirus and Herpes), syphilis and lymphocyte choriomeningitis virus for child and mother. Both were negative for toxoplasma (both IgG 0.14 UI/ml, for enzyme immunoassay, EIA). Cytomegalovirus, child negative, mother positive with low titles (IgG 2,1 Index, for EIA). Herpes type I, child negative, mother positive with low titles (IgG 2 Index, for EIA). Herpes type II, child negative, mother positive with low titles (IgG 2.39 Index e IgM negative, for EIA). Syphilis, RPR mother and child negative. Rubeola, both positive due to being vaccinated (child IgG 59.3 UI/ml, mother IgG 113.7 UI/ml, for EIA). Lymphocyte choriomeningitis, both positive but with low non-significant titles (IgG 1/32, with indirect immunofluorescence).

The above results discarded infectious etiology because the child’s antibodies are not high, leading us to suspect a bilateral atypical macular coloboma. As there are hereditary cases (1), eye fundus is examined in his parents and siblings, with normal results for all. The problem can also be associated to hypercalciuria and hypomagnesemia (2), but in this case the calcium/creatinine and magnesemia index are within normal levels.

Fig. 1: RE endotropy, fixed with LE in eccentric manner.

Fig. 2: Composite image of two RE retinographies. Large area of macular chorioretinal atrophy, with pigment accumulations.

Fig. 3: LE retinography. Atrophic lesion in temporal macula. Arrows point at scleral ectasia.
Subsequently, we followed the evolution up to age 3, the VA in RE is of perception and projection of light and in the LE of 0.2. In visual evoked potentials, with stimulation based on luminous eyeglasses, we found a clear asymmetry in detriment of the RE. The possibility of penalizing with patches was dismissed due to the large size of the RE coloboma. It would be interesting to perform an ocular echography but it was decided to wait until the patient grew a little to ensure his cooperation.

**DISCUSSION**

Differential diagnostics must consider congenital retinochoroiditis and retinal dystrophies.

Presumed diagnostics involves intrauterine infection due to toxoplasma (frequently bilateral macular scar) or congenital varicella, discarded as a result of serology. In addition, the child has no clinical signs of infection by the lymphocyte choriomeningitis virus (microcephalia, hydrocephalia, intracranial calcifications and neurological sequels) and the low antibodies count are not indicative of recent infection. Analysis results for rubeola was positive due to vaccination. Scleral ectasia is present in the atypical macular coloboma and not in old infectious retinochoroiditis. Retinal dystrophy can also be discarded because it constitutes a congenital condition which appears in the first decades of life. Besides, this child does not evidence evolution of the lesions. At this age, patients do not cooperate for electroretinogrammes.

It is difficult to explain the localization of this type of coloboma, since it is not in the usual inferonasal location. Some authors believe this could be due to a rotation of the fetal slit, or there might have been more than one fetal slit and the failed closure of the secondary slit would be the cause of the condition (3). Other authors do not believe this to be a true coloboma but an anomaly in the development of the macular structures. Thus, Mann suggest calling these lesions «macular displasia» (4). Gil-Gibernau prefers the term «macular agenesia or aplasia» (2). Pian et al (5) consider that these are due to a faulty differentiation of the arched arrays in the horizontal line, temporal to the fovea.

In any case, all the above explanations are only hypotheses and, taking into account our current knowledge, it is convenient to maintain this clinical entity under its current denomination.

**REFERENCES**