Short communication

Macular hole and Alport’s syndrome

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A B S T R A C T

Case report: We present the clinical cases of two male patients aged 38 and 39 years, diagnosed with Alport’s syndrome (AS), who suffered a bilateral macular hole (MH) and a giant unilateral MH with retinal thinning in the other eye, respectively.

Discussion: AS is a genetic disorder characterized by mutation of genes encoding type IV collagen, the main component of the internal limiting membrane (ILM), a structure identified in basal membrane of the retinal pigment epithelium–Brüch’s membrane complex. This alteration can influence the predisposition to MHs.

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Agujero macular y síndrome de Alport

R E S U M E N

Casos clínicos: Se presentan los casos clínicos de dos varones de 38 y 39 años, diagnosticados de síndrome de Alport, que presentaron respectivamente un agujero macular bilateral y un agujero unilateral gigante con adelgazamiento retiniano en el otro ojo.

Discusión: El síndrome de Alport es un desorden genético caracterizado por la mutación de genes que codifican el colágeno tipo IV, principal componente de la membrana limitante interna, estructura identificada en el complejo membrana basal del epitelio pigmentario de la retina–membrana de Brüch. Esta alteración puede condicionar la predisposición a la aparición de agujeros maculares.

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Introduction

AS is a hereditary disease that appears in one out of every 5000–50,000 newborns. There are three genetic forms, the most frequent being that linked to dominant chromosome X (80–85%), originated by mutations in the COL4A5 gene which causes an alteration in the collagen type IV of the cochlea basal membranes, renal glomerulus and ocular structures, giving rise to neurosensory hypoacusia and nephropathy (hematuria, nephrotic proteinuria, etc.) with evolution to early kidney failure and with more frequent presentation in males. Ocular
alterations appear in 92% of cases, the most characteristic findings being anterior lenticone (12–47%) and dot-spot retinopathy (85%). At the retinal level, the existence of a MH associated to the AS is an infrequent finding.

Case reports

Case 1

Male, 38, hypertense, with neurosensory hypoacusia, referred due to low visual acuity (VA). The familial anamnesis registered three deaf-mute brothers and a fourth one with poor vision.

Uncorrected VA was of 0.3 in the right eye (RE) and 0.2 in the left eye (LE). Anterior lenticone was detected and the ocular fundus evidenced the existence of a MH in both eyes, surrounded by a perimacular ring of whitish spots (Fig. 1). Optic coherence tomography (OCT) with Stratus OCT-3 (Carl Zeiss Meditec, Dublin, AC) verified the presence of MH in stage IV, with basal diameter of 1347 μm in RE and of 1409 in LE, with hyperreflectiveness in internal retina layers and cystic changes in adjacent tissue (Fig. 2). A systemic study, including renal biopsy, led to the diagnostic of AS. The patient is currently in hemodialysis due to severe renal insufficiency, maintaining the characteristics of the MH.

Case 2

Male, 39, diagnosed of AS with ocular alterations (anterior lenticone and dot-spot retinopathy), who visited due to poor VA in RE. Two years earlier the patient had been intervened for cataract without complications. OCT prior to surgery detected diminished bilateral macular thickness, with internal

Fig. 1 – Case 1 patient ocular fundus appearance. Macular hole can be seen in both eyes with ring-shaped retinopathy with whitish spots around it.

Fig. 2 – Macular optic coherence tomography of case 1 patient showing a full thickness macular hole with hyper-reflectiveness in the internal retina layers and cystic changes in adjacent tissue.
Fig. 3 – Case 2 patient ocular fundus appearance, showing dot-spot retinopathy (in the form of whitish-yellowish dots) from the macular area up to the mid-periphery. Note the absence of macular hole in both eyes and the presence of ring-shaped retinopathy with whitish dots surrounding it.

Fig. 4 – Macular optic coherence tomography of case 2 patient, showing diminished retinal thickness in the macular area, with hyper-reflective areas in the internal layers. Thickness reduction is more marked in the macular temporal area.

Retina layer thinning mainly in the temporal macular region (Figs. 3 and 4). VA was of 0.01 in the RE and 0.7 in the LE. In the ocular fundus of the RE, a full thickness MH was detected and verified with OCT. The MH had a diameter of 1400 μm (Fig. 5). The patient declined surgery and an expectant attitude was observed. At present, the MH subsists with a diameter of 2900 μm and significant retinal tissue loss and cystic alterations. The LE exhibits similar progressive thinning in the macular temporal area with thicknesses of 164 μm (Figs. 6 and 7).

Fig. 5 – Ocular fundus appearance of case 2 patient after the appearance of the macular hole in the right eye.
Discussion

MH is an anomaly infrequently associated to AS. Its pathogeny is unknown but it is related to alterations of the basal membrane collagen chains (ILM and retina pigment epithelium basal membrane–Bruch membrane–choroidal complex).

There are several theories about the reason for the deterioration of collagen that leads to the development of MH in AS. Shaikh et al.¹ postulated alterations in the vitreoretinal interface in the ILM, with focal tractions generating retina detachment and MH. Navarro et al.² affirmed that a degenerated internal retina surface is vulnerable to rupture, to which Müller cell proliferation is added to produce the vitreo- retinal traction which would predispose to the development of MH. The significant difficulty in peeling the ILM during MH surgeries in patients with AS suggests the involvement of this structure in pathogeny.³

Gupta and Kumar⁴ proposed as a cause the coalescence of cystic micro-cavities (formed by the passage of fluid through a damaged Bruch membrane due to high arterial pressure or associated renal failure).

The development of OCT is a big development in the analysis of said lesions. Usui et al. demonstrated retinal thickness reduction mainly in the temporal macular area with internal layers involvement. Savige et al.⁵ established that patients with dot-spot retinopathy exhibit perifoveal hyper-reflectiveness in the ILM/nervous fiber layer (NFL), indicating that the mean thickness of the retina is diminished particularly in the external and internal temporal macular area (corresponding to the lozenge or lack-lustre macular reflex), which have a potential capacity to develop MH.

In the above cases we have presented 2 patients with dot-spot retinopathy with central involvement. Both exhibited an MH in which OCT revealed a hyper-reflectiveness pattern in the ILM/NFL layer with retinal thinning, mainly in the external temporal macular area. The second patient exhibited said thinning prior to the development of MH and apparently is developing this alteration in the LE. As the pathogeny of this process is unknown, we consider OCT to be an essential instrument for the evolutionary study of these patients, mainly in significant macular thinning as a step prior to the formation of an MH.

Fig. 6 – Macular optic coherence tomography of case 2 patient, showing full thickness macular hole with loss of internal retinal tissue and formation of adjacent cysts. The left eye evidences retinal thickness reduction at the expense of the internal layers.

Fig. 7 – Macular optic coherence tomography of case 2 patient, showing foveal region thickness and marked thickness reduction in the macular temporal area.
Conflict of interests

No conflict of interests has been declared by the authors.

REFERENCES


