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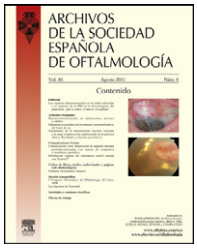

Short communication

Macular hole and Alport's syndrome☆

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ABSTRACT

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.

Agujero macular y síndrome de Alport

Palabras clave:
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Agujero macular
Tomografía de coherencia óptica

RESUMEN

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.

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

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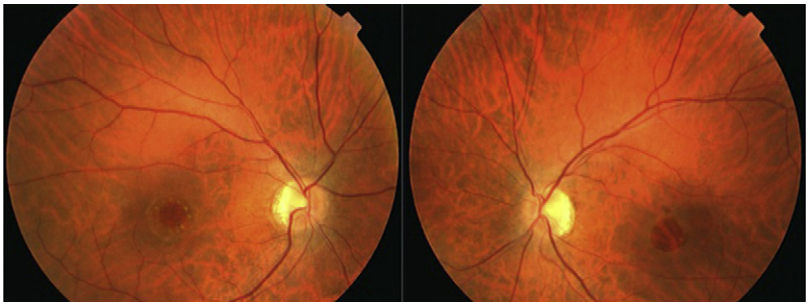

Fig. 1 – Case 1 patient ocular fundus appearance. Macular hole can be seen in both eyes with ring-shaped retinopathy with whitish spots around it.

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

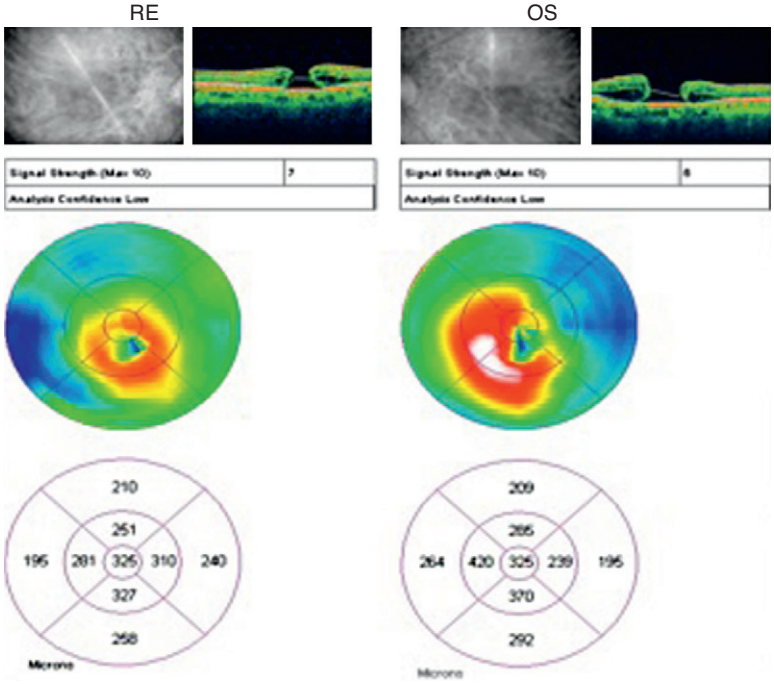

Fig. 2 – Macular optic coherence tomography of case 1 patient showing a full thickness macular hole with hyper-reflectiveness in the internal retina layers and cystic changes in adjacent tissue.