Short communication

Choroidal neovascularization secondary to choroideremia

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ABSTRACT

Case report: The case is presented of a 30-year-old man, with night blindness and decreased visual acuity (VA) in both eyes, but more significant in the left eye (LE) of 20/100. Lesions consistent with choroideremia and LE macular hemorrhage were observed in the fundus. CNV was confirmed by OCT. A definitive diagnosis was obtained by genetic study. No treatment was given as the patient did not return. At 6 months there was a regression of CNV with VA 20/25 in the LE.

Conclusions: CNV associated with choroideremia is uncommon. Treatment would antiangiogenic therapy, however spontaneous resolution is possible.

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Neovascularización coroidea secundaria a coroideremia

RESUMEN

Caso clínico: Varón de 30 años, con nictalopía y disminución de agudeza visual (AV) en ambos ojos, mayor en el ojo izquierdo (OI) de 20/100. En el fondo de ojo se observan lesiones compatibles con coroideremia y en OI, una hemorragia macular. Se confirma una NVC mediante OCT. El diagnóstico de certeza se obtuvo por estudio genético. No se realiza tratamiento porque el paciente no acude. A los 6 meses presenta regresión de la NVC con AV 20/25 en OI.

Conclusiones: La NVC asociada a coroideremia es infrecuente. El tratamiento sería la terapia antiangiogénica; sin embargo, es posible la resolución espontánea.

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Introduction

Choroideremia is an infrequent X-linked retinal degeneration. Typically, it appears in males in the form of progressive atrophy of photoreceptors, retina pigment epithelium (RPE) and choroids. It is caused by a mutation or deletion in the choroideremia gene which encodes the Rab escort isoform 1 protein (REP-1). The physiopathology of how this mutation gives rise to RPE atrophy is not well known. Some cases report choroidal neovascularization (CNV) which worsens the visual prognostic.

Case report

Male, 30, with familial history of poor vision, referred to our hospital due to night blindness, poor peripheral vision.

Fig. 1 – Left eye background showing areas of atrophy.

Fig. 2 – FA left eye in early times.

Fig. 3 – FA left eye in later times.

Fig. 4 – OCT left eye showing subretinal fluid.
Conclusions

Choroideremia is a rare X-linked retinal degeneration. Typically, it expresses in males as a progressive atrophy of RPE and choroid photoreceptors. It is produced by a mutation or deletion in the choroidemia gene which encodes REP-1 protein. The pathophysiopathology of this degeneration is not clear. A genetic study confirming the clinical presumption is essential for a definitive diagnostic, which also provides the possibility of identifying bearers and offering adequate genetic counseling for possible prenatal diagnostics.

There are very few cases in the literature describing neovascular membranes in the context of this disease and said cases refer to initial or intermediate stages probably due to the limited growth capacity of the membrane when surrounded by restricting RPE and choroid atrophy. Accordingly, it could be recommendable in these cases to await spontaneous resolution. However, present treatment would be mainly antiangiogenic therapy to avoid fibrosis secondary to exudation.

Conflict of interests
No conflict of interests has been declared by the authors.

References