Short communication

Choroidal neovascularization secondary to choroideremia

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A B S T R A C T

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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N e o v a s c u l a r i z a c i ó n   c o r o i d e a   s e c u n d a r i a   a   c o r o i d e r e m i a

R E S U M E N

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 choroideremia 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 choroideremia 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.
and decreased visual acuity (VA) in both eyes, more intense in the left eye (LE). In the initial visit, right eye (RE) VA was 20/25 and 20/100 in LE. Anterior pole ophthalmological examination was normal. Funduscopy revealed atrophy areas in RPE, the retina and the choroids, alternating with RPE hypertrophia areas. In the LE, the patient exhibited macular hemorrhage compatible with CNV (Fig. 1), confirmed by means of fluorescein angiography (FA) (Figs. 2 and 3) and optic coherence tomography (OCT, OCT Topcon 3D OCT-2000, Spectralis OCT Heidelberg Engineering) (Fig. 4). Campimetry evidenced concentric bilateral visual field decrease. Electrophysiological tests demonstrated markedly decreased photopic response in both eyes. The certainty diagnostic was obtained through genetic study which detected hemizygosis deletion of exons 6 and 7 in the sequence encoding the REP-1 gene.

The patient was not treated due to missing subsequent checkups.

Six months later, the patient visited the practice again. On this occasion, Re: VA was of 20/25 while LE VA exhibited an improvement up to 20/25 which is maintained to date. The subretinal liquid disappeared, which is compatible with spontaneous resolution of the neovascular membrane which also explained the visual acuity improvement (Fig. 5).

Conclusions

Choroideremia is a rare X-linked retinal degeneration. Typically, it expresses in males as a progressive atrophy of RPE and choroids photoreceptors. It is produced by a mutation or deletion in the choroideremia gene which encodes REP-1 protein. The physiopathology 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 choroids atrophy. Accordingly, it could be recommendable in these cases to await spontaneous resolution. However, present treatment would be mainly antiangiogenics therapy to avoid fibrosis secondary to exudation.

Conflict of interests

No conflict of interests has been declared by the authors.

References