DISTROFIA MACULAR ANULAR BENIGNA CONCÉNTRICA

BENIGN CONCENTRIC ANNULAR MACULAR DYSTROPHY

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ABSTRACT

Case report: A case of benign concentric annular macular dystrophy is described. A 32-year-old woman presented with loss of quality in visual acuity. Ophthalmologic examination, fluorescein angiogram, electrophysiologic tests and visual field measurements were performed.

Discussion: It is very important to include in the differential diagnosis other dystrophies which present a «bull’s eye» pattern on fluorescein angiography, given that preservation of relatively good visual acuity is a special feature of this disease (Arch Soc Esp Oftalmol 2007; 82: 373-376).

Key words: Benign concentric annular macular dystrophy, «bull’s eye» maculopathy, fluorescein angiography, electrophysiological tests, IMPG1 gene.

RESUMEN

Caso clínico: Presentamos un caso clínico de distrofia macular anular benigna concéntrica. Se trata de una mujer de 32 años que se presenta con pérdida de calidad visual. Se realiza exploración oftalmoscópica, angiografía fluoresceínica, pruebas electrofisiológicas y campimetría.

Discusión: Es de gran importancia realizar un cuidadoso diagnóstico diferencial con otras distrofias que presenten un patrón angiográfico en «ojo de buey», dado que la conservación de una relativamente buena agudeza visual es una característica señalada de esta enfermedad.

Palabras clave: Distrofia macular anular benigna concéntrica, maculopatía en «ojo de buey», angiografía fluoresceínica, pruebas electrofisiológicas, gen IMPG1.
INTRODUCTION

Benign concentric annular macular dystrophy is a highly unusual disease first described by Deutman in 1974 (1) as an autosomic dominant disorder. Usually, visual acuity is well preserved and the eye fundus reveals a concentric and bilateral defect along the macular pigment epithelium whose angiographic translation is a ring-shaped hyperfluorescence. There may also exist alterations in the visual field, electroretinograph and color perception.

Recently, benign concentric annular macular dystrophy has been associated to an apparent mutation of the IMPG1 gene (interphotoreceptor matrix proteoglycane 1), which resides in the chromosome 6.

CASE REPORT

A 32-year-old woman without a relevant history reports loss of visual quality during the day. Her visual acuity without optic correction records 1 for both eyes. During the exploration of the posterior pole, we observed in both eyes an alteration of the retinal pigment epithelium (RPE) in the macular region and isolated pigment clumps in the peripheral retina (figs. 1 and 2). The Humphrey 30-2 computed perimetry reveals a paracentral scotoma in both eyes (figs. 3 and 4). The fluorescein angiography (FAG) reveals a bilateral hyperfluorescence by window effect compatible with the bull’s eye angiographic image in both eyes (figs. 5 and 6). The chromatic vision test was normal for both eyes. The electrophysiological tests performed, electroretinogram (ERG) and electrooculogram (EOG), showed no alterations. Follow-up of the patient did not reveal any changes in the different exploratory tests performed so far.
DISCUSSION

Benign concentric annular macular dystrophy is an autosomic dominant disease with a highly variable clinical expression (1). Its most characteristic feature is the RPE’s concentric alteration in the macular region, whose angiographic translation is a hyperfluorescence of circular morphology resulting from the window effect corresponding to the RPE atrophy area, providing the characteristic «bull’s eye macula» image. The preservation of a relatively good visual acuity is a marked feature in this disease. The visual field defect most frequently described are central and paracentral scotomas which, in order to be accurately distinguished, will require a wide field perimetry. As for color vision, the existence of defects in the blue-yellow axis is predominant, though not constant (1).

The electrophysiological profile for this dystrophy is highly variable, while the ERG and EOG are normal, as in the present case, suggesting a macular focal defect rather than a general defect. Due to the non-selective dysfunction of cones and rods, it is tempting to locate this disease’s primary defect in the retinal pigment epithelium (2).

The «bull’s eye» angiographic image, family history and hereditary pattern are the most relevant data when determining a differential diagnosis among different clinical conditions (1). Retinopathies caused by synthetic antimalaria drugs require a history of intake of chloroquine or hydroxychloroquine. In cone dystrophies (heterogeneous hereditary pattern; dominant, recessive or linked to the X chromosome), certain symptoms and clinical signs such as photophobia, severe defect in color vision and above all an extremely altered photopic electroretinogram, aid in diagnosis. Stargardt’s disease (generally with an recessive autonomous hereditary pattern) shows at an early age a marked reduction in visual acuity, although in the initial stages the eye fundus may appear normal despite the severe loss of visual acuity, although dif-

Fig. 4: Left eye Humphrey 30-2 computed perimetry: paracentral scotoma.

Fig. 5: Right eye fluorescein angiography: hyperfluorescence due to the window effect compatible with the bull’s eye angiographic image.

Fig. 6: Left eye fluorescein angiography: hyperfluorescence due to the window effect compatible with the bull’s eye angiographic image.
ferential diagnosis is determined with the characteristic choroidal angiographic silence and the altered EOG (2). The fenestrated sheen macular dystrophy shows certain characteristic glows (3) and central areolar dystrophy (dominant autonomous hereditary pattern) progresses with good initial visual acuity and worsens with the emergence of a well-defined atrophy without the ring pattern and central involvement (3). Reverse retinitis pigmentosa shows a significant initial involvement in the ERG and EOG (2,3).

When determining a detailed differential diagnosis, it is important to refer to family history, especially the parents’ ophthalmologic exploration, since this genetic disease is dominant and autosomic, except for de novo mutations, and thus we can expect at least one parent, although asymptomatic, to show certain signs of this disease. It is also worth noting the significance of molecular genetic studies to make differential diagnoses in the future (4).

Thanks to the relatively good prognosis for this dystrophy, it is necessary to perform an accurate diagnosis allowing to distinguish it from other diseases, and to do so it is essential to perform electrophysiological testing (ERG and EOG) and FAG (3).

Although the term «benign» was used by Deutman due to the fact that vision did not change, later on certain cases progressed to a more generalized atrophy involving cones and rods, gradual reduction in visual acuity and dyschromatopsia (2,5).

REFERENCES