ARCH SOC ESP OFTALMOL. 2017;xxx(xx):xxx–xxx

ARCHIVOS DE LA SOCIEDAD ESPAÑOLA DE OFTALMOLOGÍA

www.elsevier.es/oftalmologia

Short communication

Posterior microphthalmos, retinitis pigmentosa and optic disc drusen with white dots. A case report☆,☆☆

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ARTICLE INFO

Article history:
Received 21 November 2016
Accepted 2 January 2017
Available online xxx

Keywords:
Retinitis pigmentosa
Microphthalmia
Optic nerve drusen
Foveoschisis
White dots

ABSTRACT

Objective: To present the case of a patient with a posterior microphthalmos-optic disc drusen-retinitis pigmentosa syndrome associated, for the first time, with white dots in the posterior pole.

Methodology: The posterior microphthalmos, retinitis pigmentosa and optic disc drusen syndrome was described for the first time in literature in 1991. Later, it was associated with a pattern of foveal thickening and/or foveoschisis. Different forms of mutations on chromosomes 11 and 14 have been identified as being responsible for the appearance of this syndrome, but the inheritance pattern is unknown.

Discussion: The case is reported of a 37 year-old man, with no personal or family history of interest, diagnosed with this syndrome in association with white dots in the posterior pole. Such a morphological association has never been published before in literature.

Conclusion: The posterior microphthalmos, retinitis pigmentosa and optic disc drusen syndrome is a very rare entity, and has never been described associated with white dots in the posterior pole. More case reports are needed to establish clear patterns of both the disease and inheritance.

☆ Please cite this article as: Plaza P, Iturralde O, Abascal C. Síndrome del microftalmos posterior-drusas papilares-retinosis pigmentaria asociado a puntos blancos. Caso clínico. Arch Soc Esp Oftalmol. 2017. http://dx.doi.org/10.1016/j.oftal.2017.01.003

☆☆ This study was presented as a paper in the 37th Congress of the Northern Ophthalmological Association held in Bilbao on November 27–28, 2015.

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Síndrome del microftalmos posterior-drusas papilares-retinosis pigmentaria asociado a puntos blancos. Caso clínico

Palabras clave:
Retinitis pigmentaria
Microftalmos
Drusas del nervio óptico
Foveosquisis
Puntos blancos

RESUMEN

Objetivo: Presentar el caso de un paciente con un síndrome del microftalmos posterior-drusas papilares-retinosis pigmentaria asociado por primera vez a puntos blancos en el polo posterior.

Material: El síndrome del microftalmos posterior-drusas papilares-retinosis pigmentaria fue descrito por primera vez en la literatura en 1991. Posteriormente se asoció a un patrón de engrosamiento foveolar o foveosquisis. Distintas formas de mutaciones en los cromosomas 11 y 14 han sido identificadas como responsables de la aparición de este síndrome, sin que se haya podido esclarecer, hasta el momento, un patrón de herencia determinado.

Discusión: Presentamos el caso de un varón de 37 años, sin antecedentes personales ni familiares de interés, con este síndrome asociado a puntos blancos en el polo posterior. Esta asociación morfológica nunca ha sido descrita en la literatura.

Conclusión: El síndrome del microftalmos posterior-drusas papilares-retinosis pigmentaria es una entidad muy poco común y nunca ha sido descrita asociada a puntos blancos en el polo posterior. Es necesaria más casuística para establecer patrones claros tanto de la enfermedad como en su herencia.

Introduction

The posterior microphthalmos, retinitis pigmentosa and optic disc drusen syndrome was first described by Buys and Pavlin in 1999.[1] Since then it has been reported 4 times. Patients with said syndrome require close follow-up because visual acuity could diminish due to the 3 syndrome components. To this we must add possible foveoschisis, which has been associated to the same syndrome by several authors.[2,3]

The case of a 37-year-old male exhibiting the 3 characteristics that define said syndrome is presented. In addition, said patient exhibited white dots in the posterior pole, which is submitted as a possible new characteristic of the syndrome. The present case is scientifically relevant due to the rarity of said syndrome and because it is the first to be reported in association with white dots in the posterior pole.

Clinical case

Male, 37, operated for convergent strabismus in childhood and in follow-up due to high hypermetropia. The patient did not refer relevant family history and had no systemic diseases.

Examination revealed 15° endotropia in primary gaze position with a visual acuity of 20/40 with (+14) in right eye and 20/25 with (+13.50; −0.75 at 90°) in left eye. Biomicroscopy examination was normal. Ocular fundus revealed papillary drusen that blurred the papillary contour, whitish dots in posterior pole, apparently anodyne macula and clear vitreous (Fig. 1). The periphery exhibited bone spicules beyond the vascular arcades. The pattern was bilateral and symmetrical.

In autofluorescence, the posterior pole white dots matched hyperfluorescent material depositions in the retina pigment epithelium (Fig. 2). Optical coherence tomography (Cirrus, Heidelberg, Zeiss, Jena, Germany) showed bilateral foveal thickening with a central thickness of 398 μm in right eye and 417 μm in the left eye. The right eye exhibited parafoveal spongelike pattern (Fig. 3).

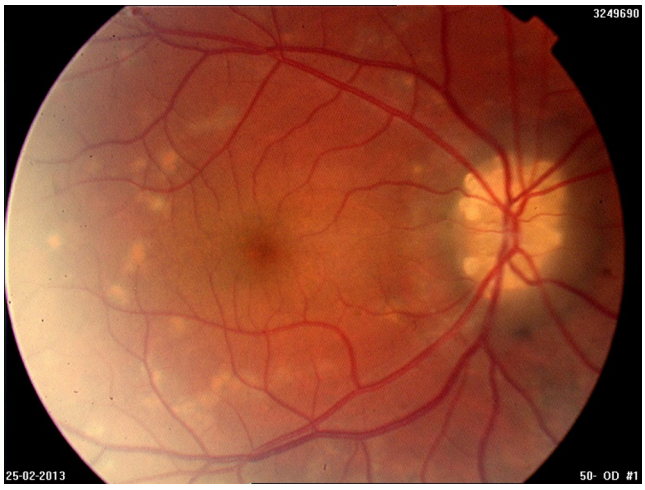

Fig. 1 – Right eye retinograph, showing drusen in the optic nerve and whitish dots in posterior pole. Left eye retinograph exhibited the same pattern.

Angiography showed a broad window effect that remained stable in time (Fig. 4). Biometry revealed an axial diameter of 16.54 mm in right eye and 16.63 mm in left eye. Corneal diameters were 11.0 mm in the right eye and 11.9 mm in the contralateral eye. The right eye anterior chamber had a depth of 2.75 mm and the left eye depth was 2.83 mm. Sita-Fast 24-2 campimetry (Humphrey, Humphrey 3, Zeiss, Jena, Germany) revealed increased bilateral blind spot, with arc-shaped