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Short communication

Atypical presentation of Best Disease☆

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ABSTRACT

Clinical case: A 43-year-old man was treated for reduced visual acuity, initially attributed to strabismic amblyopia. On fundus examination, bilateral neurosensory detachments (NSD) were observed in posterior pole, surrounded by deposits of lipofuscin. His 3-year-old son was also examined and circumscribed NSD was observed with the presence of pseudohypopyon in OD and a fibrosis scar in OS. The Arden ratio was decreased in electrooculography (EOG) in both patients, and genetic studies revealed a single mutation of the BEST1 gene.
Discussion: The existence of extensive bilateral NSD may be an unusual form of presentation of Best disease. Family history, EOG, and genetic study supported this diagnosis.

Presentación atípica de la enfermedad de Best

Palabras clave:
Enfermedad de Best
Distrofia macular viteliforme de Best
Desprendimiento neurosensorial de la retina

RESUMEN

Caso clínico: Varón de 43 años atendido por agudeza visual reducida, inicialmente atribuida a ambliopía estrábica. Funduscópicamente se observaban extensos desprendimientos neurosensoriales (DNS) bilaterales en el polo posterior, delimitados por acumulación de lipofucsina. Un hijo suyo de 3 años fue también explorado y, en el fondo de ojo, presentaba DNS circunscritos con presencia de pseudohipopion en el ojo derecho y fibrosis cicatricial en el ojo izquierdo. Para ambos, el índice de Arden estaba disminuido en la electrooculografía, y el estudio genético reveló una misma mutación del gen BEST1.
Discusión: La existencia de amplios DNS bilaterales puede ser una forma inusual de presentación de la enfermedad de Best. La historia familiar, la electrooculografía y el estudio genético avalaron este diagnóstico.

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Introduction

Best's vitelliform macular dystrophy (BVMD), also known as Best disease (BD), is a progressive maculopathy from dominant autosomal inheritance with incomplete penetrance and highly variable clinic expressions, even among relatives.[1–3] Presentation is generally bilateral, with onset during childhood,[1–3] and is characterized by a typical single subretinal round or oval-shaped yellowish-orange subretinal aggregate located in the macula,[1–4] with initial appearance in egg yolk form (vitelliform). This lesion could evolve towards progressive degradation of vitelliform material and the appearance of retinal pigment epithelium (RPE) atrophy of the compromised area.[1–5] This final atrophic/cicatricial stage could involve acute subretinal hemorrhages, sometimes associated to choroidal neovascularization[1–3] and less frequently to the appearance of macular holes.[1] Even so, visual impairment is not generally abrupt but slow and insidious up to the more advanced stages.[2] The significant phenotypic variation is due mainly to the evolution stages of this dystrophy but also to genetic heterogeneity.[1,2] One case of adult BD is described with atypical presentation and another case of the same family that was an important diagnostic clue.

Clinic case report

Male, 43, who visited due to poor vision, initially attributed to strabismic amblyopia. Decimal best corrected visual

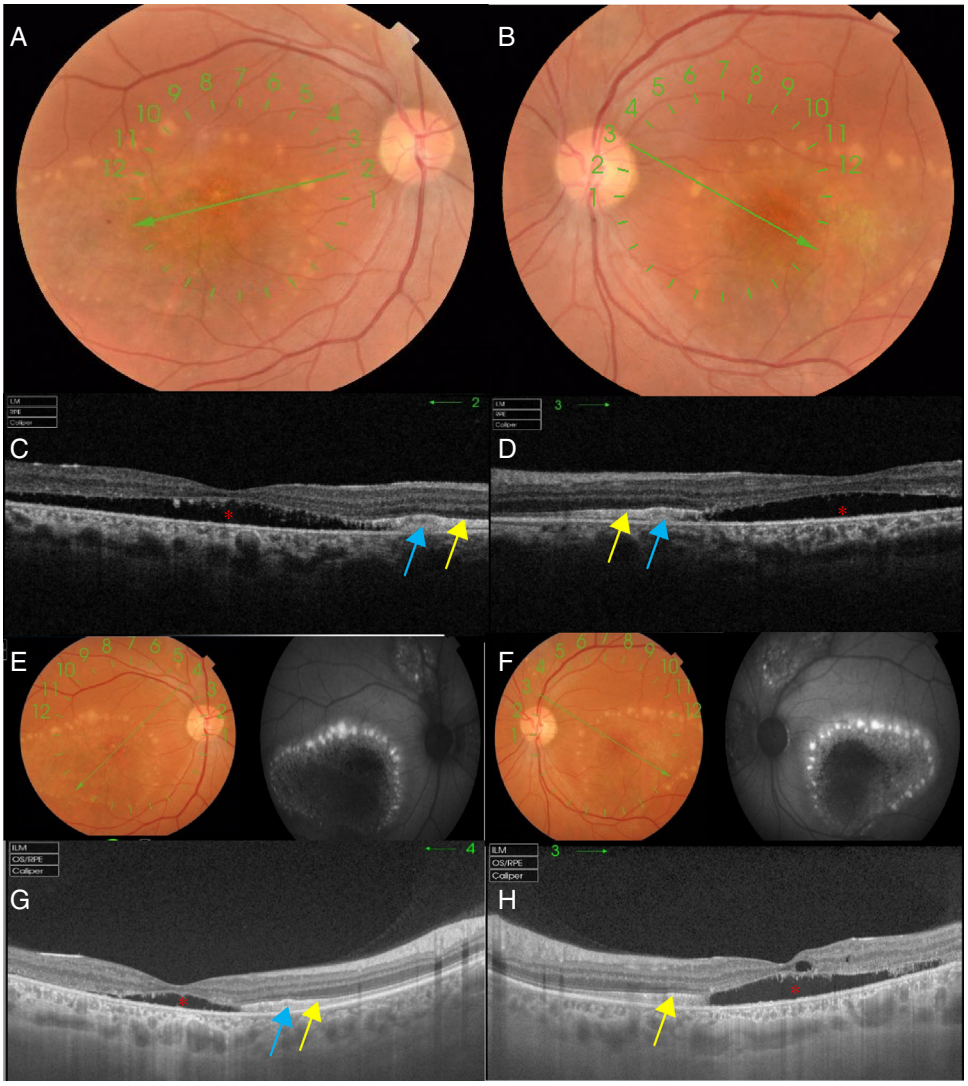

Fig. 1 – Baseline examination of the father. Ocular fundus of RE (A) and LE (B): neurosensory detachment evidenced by lipofuscin granules and of the superior peripapillary isolated aggregates. Macular OCT of RE (C) and LE (D): central serous detachment (red asterisks), accumulation of hypo-reflective material (lipofuscin) over retinal pigment epithelium (RPE) (blue arrows), thickened and hypo-reflectiveness strip between RPE and ellipsoid line (yellow arrows). Evolution. Ocular fundus and autofluorescence of RE (E) and LE (F): less extended NSD. Hyperfluorescence of lipofuscin deposits. Macular OCT of RE (G) and LE (H): flatter NSD (red asterisks) with peripheral accumulation of hypo-reflective material over the RPE (blue arrow), increased hyper-reflective photoreceptor strip (yellow arrow) and intraretinal cyst in LE.