Short communication

Best's vitelliform macular dystrophy associated with choroidal neovascularization

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ABSTRACT

Case report: We report the case of a child with a sudden loss of vision of the left eye. Ophthalmoscopic examination revealed vitelliform lesions in both foveal centers, as well as an adjacent hemorrhage in his left eye. Fluorescein angiography confirmed the presence of a neovascular membrane in his left eye. The electrooculogram showed disease. According to complementary studies the patient was diagnosed with Best's disease associated with choroidal neovascularization.

Discussion: The diagnosis of Best's vitelliform macular dystrophy is often a casual finding as visual acuity tends to remain stable for long periods of time. A sudden deterioration in vision may suggest complications, such as choroidal neovascularization.

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Distrofia macular vitelliforme de Best asociada a neovascularización coroidea

RESUMEN

Caso clínico: Se presenta el caso de un niño con pérdida súbita de visión en ojo izquierdo. El examen fundoscópico revela una lesión foveal vitelliforme bilateral, y una hemorragia adyacente en ojo izquierdo. La angiografía con fluoresceína confirma la presencia de una membrana neovascular en ojo izquierdo. El electrooculograma resulta patológico. Tras completar el estudio, es diagnosticado de enfermedad de Best asociada a neovascularización coroidea.

Discusión: El diagnóstico de enfermedad de Best puede ser casual dado que la agudeza visual suele permanecer estable. Una pérdida súbita de visión ha de sugerirnos la aparición de complicaciones tales como neovascularización coroidea.

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Introduction

Best’s disease or Best’s vitelliform macular dystrophy (BVMD) is one of the most frequent macular dystrophies. It is generally found accidentally as good vision is maintained for many years. This paper presents an atypical case of BVMD associated to the neovascular membrane (NVM), with sudden loss of vision.

Clinical case

A male aged 9, Caucasian, visited the practice referring sudden visual acuity (AV) loss in the left eye (LE). His parents being adoptive, they were unable to refer relevant history. Corrected AV was of 0.7 in the right eye (RE) and 0.4 in LE. Biomicroscopy, intraocular pressure and pupil reflexes were within normal ranges. The ocular fundus assessment revealed a subretinal yellowish macular lesion having a vitelliform shape in both eyes. In addition, the LE exhibited adjacent hemorrhage (Figs. 1 and 2). Considering the suspicion of NVM in said eye, fluorescein angiography was performed which confirmed the suspicion (Fig. 3). Autofluorescence was also studied and hyperautofluorescent images were observed, matching the lipofuscin deposits (Figs. 4 and 5). Spectral domain optic coherence tomography (OCT) revealed a hyper-reflective injury due to the subretinal material as well as adjacent fluid (Fig. 6). The study was completed with electrophysiological tests, which confirmed the suspected diagnosis due to the pathological results of the standard electrooculogram (EOG), with Arden’s coefficient being of 1.34 in the RE and 1.49 in the LE. An unusual result was the EI pattern stimulus electoretinogram (ERG) which was also altered probably due to the presence of NVM.

An observation control approach was decided, with AV remaining stable after one month. The tendency toward
the self-limitation of choroidal NVM in young patients was described by Ho and Glaser, probably due to the hyper function of the retina pigment epithelium (RPE) in the area of the injury and to the production of neovascularization inhibitors. On the other hand, prospective studies with definitive results on the possible adverse effects of antiangiogenic injection in young patients have not yet been made.

Discussion

BVMD is an autosomic dominant inheritance maculopathy with variable expressiveness and penetration, related to mutations of the bestrophin gene located in chromosome 11 (11q131). The product of said gene is bestrophin-1, a transmembrane protein the expression of which is associated to chlorine channels in the EPR cells. The ionic flow alteration would explain both the electrophysiological findings and the accumulation of lipofuscin. Accordingly, a reduced luminous peak of EOG is typical. It seems that this peak reflects the depolarization of the basal RPE membrane due to an increase in the conductivity of chlorine, so that a poor function of these channels would give rise to an Arden quotient below 1.5. EOG can be useful to detect patients who exhibit said mutation without funduscopy involvement. The detection of carriers is important because they can transmit the disease to the following generations.

BVMD is typically bilateral and appears in childhood. Patients frequently exhibit yellowish lesions due to accumulation of lipofuscin which deposits excessively due to the inability of RPE to digest the external segments of photoreceptors in the context of an ionic imbalance.

Classically, it has been classified in 6 phenotypic stages:

1. Pre-vitelliform stage: altered EOG can be detected although the funduscopy appears normal.
2. Vitelliform stage: a subretinal yellowish lesion can be seen, similar to the yolk of an egg.
3. Pseudohypopion: the material deposits forming a level with respect to the LSR situated in the upper area.
4. Vitelliruptive stage: the lesion becomes more uneven and the alteration of the RPE becomes more evident.
5. Atrophy: the pigment disappears leaving a central atrophy area in the RPE with ensuing visual loss.
6. Subretinal neovascularization: on some occasions NVM can develop causing a sudden AV reduction. Said complication is infrequent and can appear in late stages, in contrast with the instant case.

In what concerns supplementary tests, a marked hyperautofluorescence of the vitelliform material is characteristic although in advanced stages hypofluorescence predominates due to RPE atrophy. OCT allows the identification of the deposit site and the existence of subretinal fluid or RPE atrophy. FA is not routinely performed unless NVM is suspected.

Despite the typical and striking macular alterations in BVMD patients, its diagnosis is accidental as they maintain good vision. For this reason, any AV reduction should suggest the appearance of complications, such as NVM, fibrosis or macular atrophy. However, as stated above, said injuries generally appear in late stages, presentation at the onset being atypical.
Conflicts of interests

No conflict of interest has been declared by the authors.

References


