CENTRAL SEROUS CORIORETINOPATHY IN ADULT ONSET FOVEOMACULAR VITELLIFORM DYSTROPHY

CORIORETINOPATÍA SEROSA CENTRAL EN LA DISTROFIA VITELIFORME FOVEOMACULAR DEL ADULTO

PINÓS J¹, SABATER A², NAVARRO C¹, CARBONELL P¹, GONZALVO A¹

ABSTRACT

Case report: The clinical case of a 30 year-old male patient with a bilateral and symmetric adult-onset foveomacular vitelliform dystrophy is presented. The simultaneous onset of a central serous chorioretinopathy (CSCR) with multiple white dots in the proximity of the temporal vascular arcades is documented. Fluorescein angiography showed a combined alteration of both types of photoreceptors, and the acute lesion of the CSCR at the posterior pole of the eye.

Discussion: Adult-Onset Foveomacular Vitelliform Dystrophy is a hereditary condition which results in an alteration of the posterior pole of the eye, but is not usually associated with any acute complications. The onset of a CSCR, as seen in this case, is unusual 2008; 83: 505-508).

Key words: Adult-onset foveomacular vitelliform dystrophy, central serous chorioretinopathy, retinal dystrophy, retinal pattern dystrophy, maculopathy.

RESUMEN

Caso Clínico: Se presenta el caso clínico de un varón de 30 años con una distrofia foveomacular viteliforme del adulto bilateral simétrica, que de forma simultánea desarrolla una coriorretinopatía central serosa (CSCR) y en el que se aprecia la coexistencia de múltiples puntos blancos en la proximidad de las arcadas vasculares temporales. La angiografía fluoresceínica demuestra la alteración mixta de ambos tipos de fotorreceptores, así como la lesión aguda de la CSCR en el polo posterior OI.

Discusión: La distrofia foveomacular del adulto es una alteración hereditaria del polo posterior que no suele evolucionar con complicaciones agudas. La aparición de una coriorretinopatía central no es habitual.

Palabras clave: Distrofia foveomacular viteliforme del adulto, coriorretinopatía serosa central, distrofia retiniana, distrofia retiniana en patrón, maculopatía.
INTRODUCTION

Vitelliform foveomacular dystrophy was first described by Adams in 1883 (1). Subsequently it was individualized by Gass in 1974. Initially, adult vitelliform foveomacular dystrophy was included in the «pattern» dystrophies of the pigmented epithelium. Nowadays it is considered to be a nosological entity in its own right.

This short communication presents a case report in which a healthy patient who visits the practice due to a central serous chorioretinopathy is identified as having a bilateral vitelliform foveomacular dystrophy with whitish points in the proximity of the greater vascular arches.

CASE REPORT

A 30 year-old male who visits the practice due to metamorphopsia in left eye beginning an indeterminate time ago. His visual acuity was normal in both eyes and he did not refer any symptoms.

The anamnesis did not identify any personal history of relevance. No familial history was found in relation to relevant ocular disorders.

- **Gross visual acuity:**
  - Right eye: 1.
  - Left eye: 0,8 difficult with metamorphopsia.

- **Eye fundus:** The eye fundus exhibited the characteristic appearance of vitelliform foveomacular dystrophy. The macular area of both eyes exhibited rounded, slightly elevated, yellowish subretinal injuries of about 1/3 DD (disc diameter), with a more pigmented area in the center and surrounding it. The greater vascular arches show numerous whitish-yellowish spots. These injuries are not confluent (figs. 1 and 2).

- **Fluorescein angiography:** fluorescein angiography (FA) (figs. 3 and 4) showed in initial stages small hyperfluorescent rings in both eyes, around a central hypo-fluorescent point (one subfoveal in the right eye and two in the left eye, one subfoveal and the other slightly displaced in the temporal direction). This typical clinical form is shared in the other dystrophies of the in pattern pigmented epithelium. A few small telangetasias were found in the terminal vessels (figs. 5 and 6).

  Since the early stages a hyper-fluorescent point is observed, situated nasally to the foveola, corresponding to a fugue point of a central serous chorioretinopathy. This injury increases slightly in size along the angiography with the progressive impregnation of the surrounding area. Around it we observed a nearly flat neuroretina detachment, which is difficult to see in the exploration and was detected due to the out-of-focus in the area.

  The numerous white spots, which are well defined and dispersed, close to the temporal arches (above all the superior temporal arch) exhibit small window fluorescent defects associated to the damages of the pigmentary epithelium similar to those of the Fundus Flavimaculatus or Albipunctatus.

At present, the results of electrophysiological tests made on this patient (electroretinogram — ERG— and electro-oculogram —EOG—) yield results within normality. The chromatic vision exploration did not show significant alterations. We
were not able to carry out genetic molecular studies, although these would probably be useful for the differential diagnostic of the entity.

The spontaneous evolution of the case, with disappearance of the neuroepithelial detachment and total resolution of LE metamorphopsia, was complete after 5 months. To date, no relapses have occurred.

**DISCUSSION**

The instant case report has the peculiarity that it combines (probably in a non-causal manner) the typical injuries of the adult vitelliform dystrophy with central serous chorioretinopathy. In addition, the case exhibits multiple white spots corresponding to the mixed alteration of photoreceptors.

The whitish-yellowish symmetric subretinal macular injuries are the diagnostic key for adult vitelliform dystrophy. Sometimes, these injuries are non-symmetric (2).

In some cases said injuries can be confused with the «egg yolk» injuries typically seen in the second stage of Best’s vitelliform dystrophy.

Generally, campimetry does not reveal any findings other than, perhaps, a small relative central scotoma, which can be very difficult to determine.

Electrophysiological studies usually produce normal results for this entity due to the focal nature of the injuries. In some severe cases the EOG may be altered. Arden’s index in EOG can be normal (63.5%) or altered (36.5%). When said index is normal, it is useful for the differential diagnostic with Best dystrophy which is pathological in all cases.

The yellowish egg-yolk injuries are identified by means of Optical Coherence Tomography as a
hyper-reflective area between the pigmented epithelium and the photoreceptor layer. Usually it is possible to see how the highly reflective line of the photoreceptor layer is elevated an detached from the pigmented epithelium. It is differentiated from Best’s vitelliform dystrophy by the size of the macular lesions which are smaller due to the tendency of Best’s dystrophy to shape itself into a pseudohydropopion configuration and because Arden’s index in the EOG is in all cases pathological, whereas in adult vitelliform dystrophy it can be normal (3).

There is no effective treatment for adult vitelliform dystrophy. Laser photocoagulation, corticoids and vitamins A and E have been used without demonstrable effect. The prognosis for the disease is relatively good, as it usually evolves to a slow deterioration of central visual acuity, maintaining useful vision in most cases described in the literature. Rarely, complications may appear (mainly choroidal neovascular membranes). Intravitreous Bevacizumab can be used for treating vitelliform dystrophy (4).

Molecular genetic studies can be useful for the differential diagnostic of these entities, mainly to determine mutations in the Periferine/RDS and VMD2 genes, the latter being the gene of Best’s type of vitelliform macular dystrophy (5).

REFERENCES