CASE REPORT

Congenital hypertrophy of the retinal pigment epithelium, a casual discovery

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ABSTRACT

Introduction: congenital hypertrophy of the retinal pigment epithelium is a congenital hamartoma of the retinal pigment epithelium. It is seen as a typically benign pigmented fundus lesion. Sometimes, in its single-solitary presentation, it can be confused with a malignant pigmented lesion of the fundus.
Patient information: two patients attended the retina clinic of the "Arnaldo Milián Castro" Hospital for different diseases. Per protocol, they underwent routine examination and were diagnosed, by chance, with congenital hypertrophy of the retinal pigment epithelium in its unique-solitary variant.
Conclusions: congenital hypertrophy of the retinal pigment epithelium in its single-solitary form of presentation is not associated with any other ocular or systemic disease. Its diagnosis is casual, in a routine examination. The essential thing is not to confuse it with other malignant pigmented lesions of the fundus. Follow-up with color retinography and periodic observation are recommended.
Key words: congenital hypertrophy of the retinal pigment epithelium; diagnosis

RESUMEN

Introducción: la hipertrofia congénita del epitelio pigmentario de la retina es un hamartoma congénito del epitelio pigmentario de la retina. Se observa como una lesión pigmentada del fondo de ojo típicamente benigna. En ocasiones, en su forma de presentación única-solitaria, puede confundirse con una lesión pigmentada maligna del fondo de ojo.
Información del paciente: dos pacientes acudieron a la Consulta de retina del Hospital "Arnaldo Milián Castro" por diferentes enfermedades. Por protocolo se les realizó el examen de rutina y se les diagnosticó, de forma casual, una hipertrofia congénita del epitelio pigmentario de la retina en su variante única-solitaria.
Conclusiones: la hipertrofia congénita del epitelio pigmentario de la retina en su forma de presentación única-solitaria no se asocia con alguna otra enfermedad ocular o sistémica. Su diagnóstico es casual, en un examen de rutina. Lo esencial es no confundirla con otras lesiones pigmentadas malignas del fondo de ojo. El seguimiento con retinografía a color y la observación periódica son lo recomendado.
Palabras clave: hipertrofia congénita del epitelio pigmentario de la retina; diagnóstico

INTRODUCTION

Congenital hypertrophy of the retinal pigment epithelium (CHRPE) is a congenital hamartoma of the retinal pigment epithelium (RPE). It is usually seen as a typically benign and generally asymptomatic pigmented fundus lesion that presents in three clinical variants: single-solitary lesion, grouped lesions - known as bear tracks - and an atypical variant.[1,2]
It has an estimated prevalence of 1.2% and is usually diagnosed as an incidental finding on routine examination.[1]
The atypical variant (multiple or bilateral lesions) is closely associated with familial polyposisadenomatosis - Gardner's syndrome; the incidence is reported to range from 58% to 92%.[3]
Occasionally, a single large solitary lesion of CHRPE may be mistaken for a malignant fundus lesion.[1]
Although the diagnosis is primarily clinical, in settings where diagnostic tools are not available for the study of doubtful cases, knowledge of their clinical appearance is of vital importance to prevent misdiagnosis that may create confusion in patients. We present two cases with CHRPE in its single-solitary variant found in a routine ophthalmologic examination.

PATIENT INFORMATION

Case 1

Black, male, 37-year-old patient referred to the Retina Clinic with a diagnosis of central serous chorioretinopathy in his left eye for examination of the peripheral retina of both eyes.
No personal or family pathologic history of interest was collected.
In the ophthalmologic examination she had a visual acuity without crystals of 20/20 in her right eye and 20/40 in her left eye (Snellen scale). With positive crystals she achieved a best corrected visual acuity of 20/30 in her left eye.
Ocular pressure (Goldman tonometer) was 16 mm/Hg in both eyes.
Anterior segment biomicroscopy showed no findings of interest in either eye, posterior segment biomicroscopy with 90 diopter air lens revealed normal appearing optic disc, vessels and macula in her right eye. In his left eye, an optic disc with defined borders and normal coloration was found. In the macula there was an image of serous retinal detachment with somewhat cloudy subretinal fluid of approximately two disc diameters (DD) involving the fovea and extending nasally, suggestive of central serous chorioretinopathy.
Indirect binocular ophthalmoscopy was performed with scleral depression in both eyes. In his left eye there were no findings of interest. In her right eye, a solitary, pigmented, flat, flat lesion of approximately five to six DD, without associated subretinal fluid, with scalloped edges and areas of atrophy on its surface, was found in the superior temporal quadrant, towards the equator and medial periphery (Figure 1).
A combined A -and B- mode ocular ultrasound was performed on her right eye to corroborate the flat appearance of the lesion and to rule out the presence of signs of a malignant pigmented fundus lesion (Figure 2).

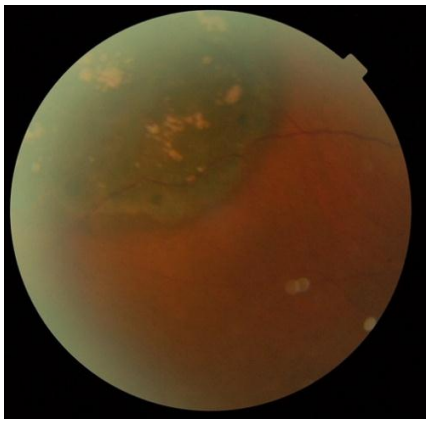

Figure 1. Fundus photograph showing a flat, solitary, pigmented lesion with scalloped edges, depigmented halo in normal retina contiguous to the edges of the lesion and patches of atrophy on its surface

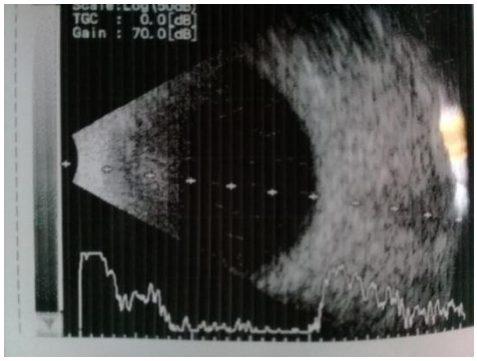

Figure 2. Combined ocular ultrasound, A and B mode; no gross lesion or mass effect observed

It was interpreted as CHRPE in its single-solitary variant; a chance finding in a routine examination.

Case 2

White, female patient, 48 years of age, who came to the emergency department because she "saw a little fly and lightning" in her right eye, with 24 hours of evolution. Symptomatic acute posterior vitreous detachment was suspected and she was referred to the Retina Clinic for peripheral retinal examination.

No personal or family pathologic history of interest was collected.

In the ophthalmologic examination performed she presented a visual acuity without crystals of 20/20 in both eyes (Snellen scale).

Ocular pressure (Goldman tonometer) was 14 mm/Hg in both eyes.

Anterior segment biomicroscopy showed no findings of interest in either eye.

Posterior segment biomicroscopy with 90 diopter air lens showed normal appearing optic disc, vessels and macula in both eyes and indirect binocular ophthalmoscopy with scleral depression in both eyes to complete the physical examination as a protocol for peripheral retinal examination.

In her right eye she presented with an ongoing posterior vitreous detachment with no holes or retinal tear.

In her left eye, an extensive, solitary, pigmented lesion, approximately 13 to 15 DD, almost occupying a quadrant, flat, with no associated subretinal fluid,

irregular borders and patchy areas of atrophy on its surface, was found in the nasal sector between the equator and the mid-periphery (Figure 3).
A combined ocular ultrasound, mode A and B (Figure 4), was performed on her left eye to corroborate the flat aspect of the lesion and to rule out the presence of signs of a malignant pigmented lesion of the fundus.

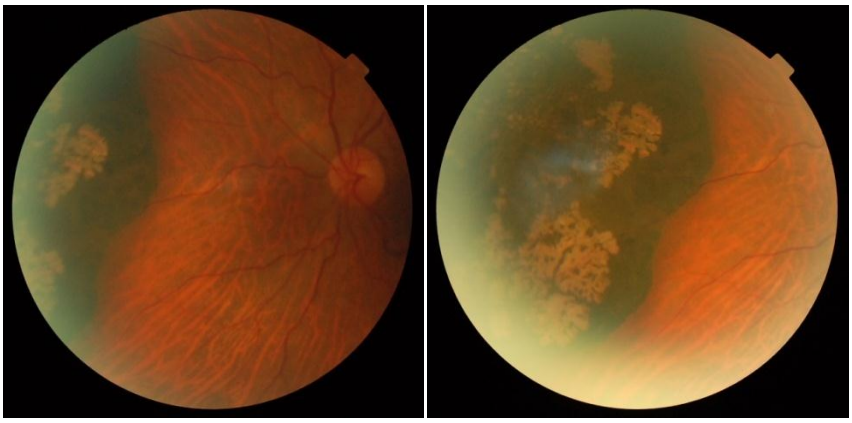

Figure 3. Fundus photograph showing an extensive, flat, solitary, pigmented lesion with irregular borders and patches of atrophy on its surface

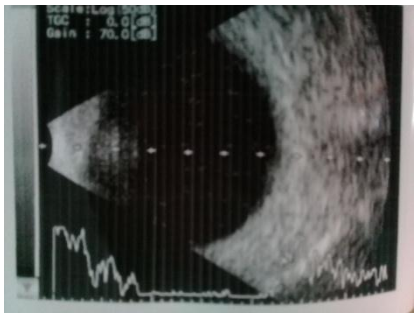

Figure 4. Combined A- and B-mode ocular ultrasound; no gross lesion or mass effect seen

It was interpreted as CHRPE in its single-solitary variant; a chance finding in a routine examination.

DISCUSSION

CHRPE is a benign pigmented fundus lesion with a characteristic clinical appearance. It is usually asymptomatic, presents before the age of 30 years, and is often discovered on routine ophthalmologic examination. It is usually located in the equator and mid-periphery of the retina; however, on rare occasions, it may be found in the posterior pole and involve the optic disc and macula. When they involve the fovea, visual acuity is usually affected.[4,5]
It may present as a solitary lesion, as grouped lesions (bear tracks) or atypical variant (multiple or bilateral lesions). The single-solitary variant of HCEPR is the most common form of presentation.[1,5]
Solitary CHRPE is pigmented, flat, rounded, with smooth and scalloped borders. The color can vary from light gray to dark black. There is a typical demarcation between CHRPE and the normal RPE adjacent to the lesion that has the appearance of a depigmented halo. The retina and vessels involved in

the lesion generally show a normal appearance.[1,6] Some lesions may progress to vascular alterations including obliteration of capillaries and large vessels, microaneurysmal changes, retinochoroidal anastomoses, and neovascularization.[6,7]

Gaps of hypopigmentation and atrophy of the RPE may be found on the surface of the lesion. These areas of hypopigmentation have a tendency to grow slowly with the passage of time.[6]

The size can range from 100 µm to several DD and even occupy an entire quadrant of the fundus in some cases. Lesions can be found in any quadrant, but the superior temporal is the most affected. Solitary lesions show a slow and gradual growth in up to 46-83% of cases after three or more years of follow-up.[5,6]

The risk of malignancy is very low and extremely rare; however, there are reports of highly pigmented nodular lesions arising from HCEPR that developed their own blood supply with associated subretinal fluid and developed into malignant epitheliomas demonstrated by histopathologic analysis.[8]

Single-solitary CHRPE has not been associated with any other ocular or systemic disease.[9] In our experience it can be found incidentally on routine examination; this is confirmed by the circumstances under which these two reported cases were diagnosed.

Diagnosis is mainly clinical, without the use of any diagnostic tool in most cases.[1] When necessary, fundus autofluorescence, fluorescein angiography and optical coherence tomography are the most useful.[1,8,10] Color retinography is essential for documentation and follow-up of these lesions.[1]

Some authors propose[1] that ocular ultrasound mode A or B (or both) does not contribute or contribute to the diagnosis of CHRPE in its single-solitary variant. Others[8,10] use it to confirm the flat nature of the lesion and to rule out the mass effect or overlying lesion. In this investigation it was used in a confirmatory manner and it was understood that, despite the fact that solitary HCEPR has a characteristic clinical appearance, ocular ultrasound -in the absence of other diagnostic means- can be especially useful in extensive lesions with absence of some of its typical features that may generate some confusion.

The relevance of correctly diagnosing an HCEPR in its single-solitary presentation lies in the ability of the specialist not to confuse it with other pigmented lesions of the fundus such as choroidal nevi, melanocytomas and malignant melanoma of the choroid.[1,8,10]

Expectant management, without the need for therapeutic interventions, is the rule. Periodic observation is recommended for all patients.[8,11] In exceptional cases undergoing malignant transformation, external radiation, brachytherapy plaques and photocoagulation are available options for treatment.[8]

CHRPE is a benign pigmented fundus lesion with a characteristic clinical appearance. The single-solitary form of presentation is the most common variant and is not associated with any other ocular or systemic disease. It is usually diagnosed on routine examination. What is essential in the diagnosis is not to confuse it with other malignant pigmented fundus lesions. Follow-up with color retinography and periodic observation are recommended.

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CONFLICT OF INTEREST

The authors declare that they have no conflict of interest.