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Multimodal Imaging of Congenital Retinal Pigment Epithelial Hypertrophy: A Case Report

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Abstract **Case Report**

Introduction: Congenital hypertrophy of the retinal pigment epithelium is a benign, asymptomatic congenital hamartoma characterized by flat, well-defined pigmented lesions on the fundus. It has three clinical forms: solitary, multiple and atypical. ***Objectives and Methods***: We report the case of a 58-year-old female patient whose fundus examination revealed a juxtapapillary pigmented lesion. Fluorescein angiography showed the lesion to be hypofluorescent. Its tomographic appearance is distinctive, with thinning of the neurosensory retina opposite the lesion, and thickening of the retinal pigment epithelium in pigmented areas and thinning in lacunar areas. After a 12-month follow-up, the lesion remained stable. ***Discussion***: Congenital hypertrophy of the retinal pigment epithelium is a benign ocular condition characterized by single or multiple pigmented retinal lesions. Histologically, it consists of a single-cell layer of hypertrophied retinal pigment epithelium cells, densely filled with large, round macromelanosomes. Although this is a benign, asymptomatic condition that is usually diagnosed incidentally, further evaluation may be necessary to rule out associated syndromes and differential diagnoses, particularly in the case of multiple or atypical lesions. Patient education regarding the potential risk of growth, choroidal neovascularization, or exceptionally malignant transformation to adenocarcinoma is necessary. ***Conclusion*:** Congenital hypertrophy of the retinal pigment epithelium is a rare and benign condition, generally requiring no treatment, but regular long-term follow-up is mandatory.

Keywords: Hypertrophy, Retinal Pigment Epithelium, Atypical, Multifocal, Follow-Up.

INTRODUCTION

Congenital hypertrophy of the retinal pigment epithelium is a benign, asymptomatic congenital hamartoma characterized by flat, well-defined pigmented lesions on the fundus [1]. It occurs in three forms: solitary (unifocal), multiple (multifocal) and atypical [1].

In some cases, it may be associated with an autosomal dominant cancer syndrome, familial adenomatous polyposis, Gardner's syndrome or Turcot's syndrome, almost all of whose patients develop colorectal carcinoma in middle age, in the absence of treatment [2].

This association is all the more likely as the lesions present a number of clinical features that need to be distinguished, in particular the atypical form, thus necessitating a thorough multidisciplinary systemic evaluation with gastroenterologists [2].

It's an asymptomatic condition that doesn't usually cause visual disturbances, and the diagnosis is usually made incidentally.

Nevertheless, further evaluation may be necessary to rule out associated syndromes and differential diagnoses, particularly in the case of multiple or atypical lesions.

Although this condition is benign and generally requires no treatment, patient education concerning the potential risk of growth, nodular transformation, choroidal neovascularization or, exceptionally, malignant transformation, is essential, hence the importance of regular long-term follow-up [2].

OBSERVATION

The patient was 58 years old, melanodermic, with no previous pathological history, and was consulting for a change in her optical correction.

Visual acuity was 10/10 with optical correction in both eyes.

Ophthalmological examination of the left eye was strictly normal. On the right, anterior segment examination and intraocular pressure measurement were normal, and the photomotor reflex was present and symmetrical.

Fundus examination revealed a single, strongly pigmented, juxta-papillary lesion, occupying the superior and inferotemporal parts, totally flat, with sharp margins and a vast area of chorioretinal atrophy (figure 1).

Fundus photography is useful for documentation and follow-up.

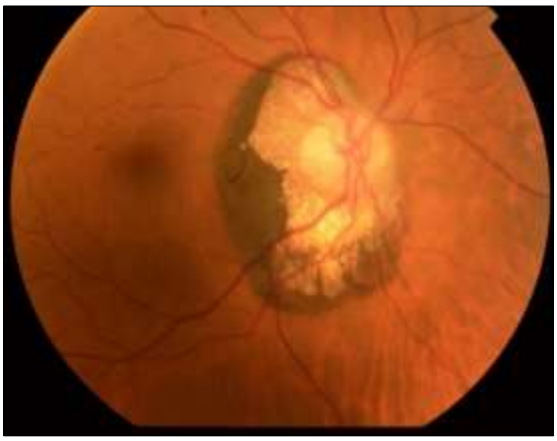

Figure 1: Fundus photograph showing a single pigmented, juxta-papillary lesion, totally flat, with sharp margins and a wide area of chorioretinal atrophy

On angiography, the lesion remained hypofluorescent throughout the agiographic sequences, with a window effect in the atrophic zone (figure 2).

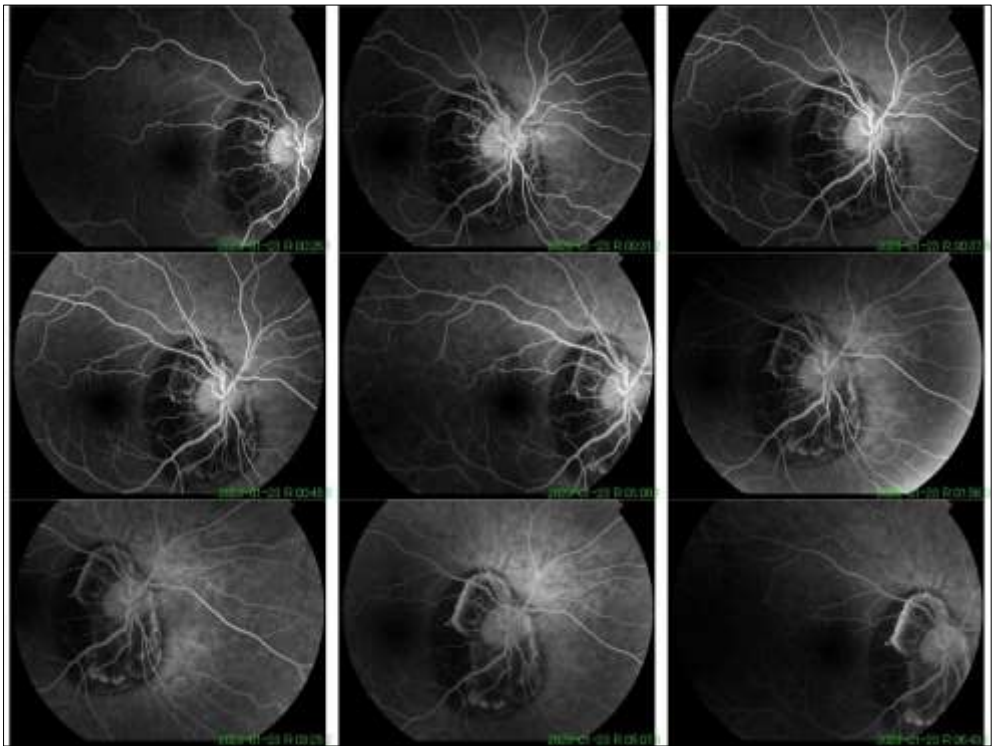

Figure 2: Agiographic sequences of the right eye showing the hypofluorescent character of the lesion in early, intermediate and late time with a window effect at the level of the atrophic zone

On OCT, the neurosensory retina is thinned opposite the lesion, the retinal pigment epithelium is thickened in pigmented areas and thinned in lacunar areas (Figure 3).

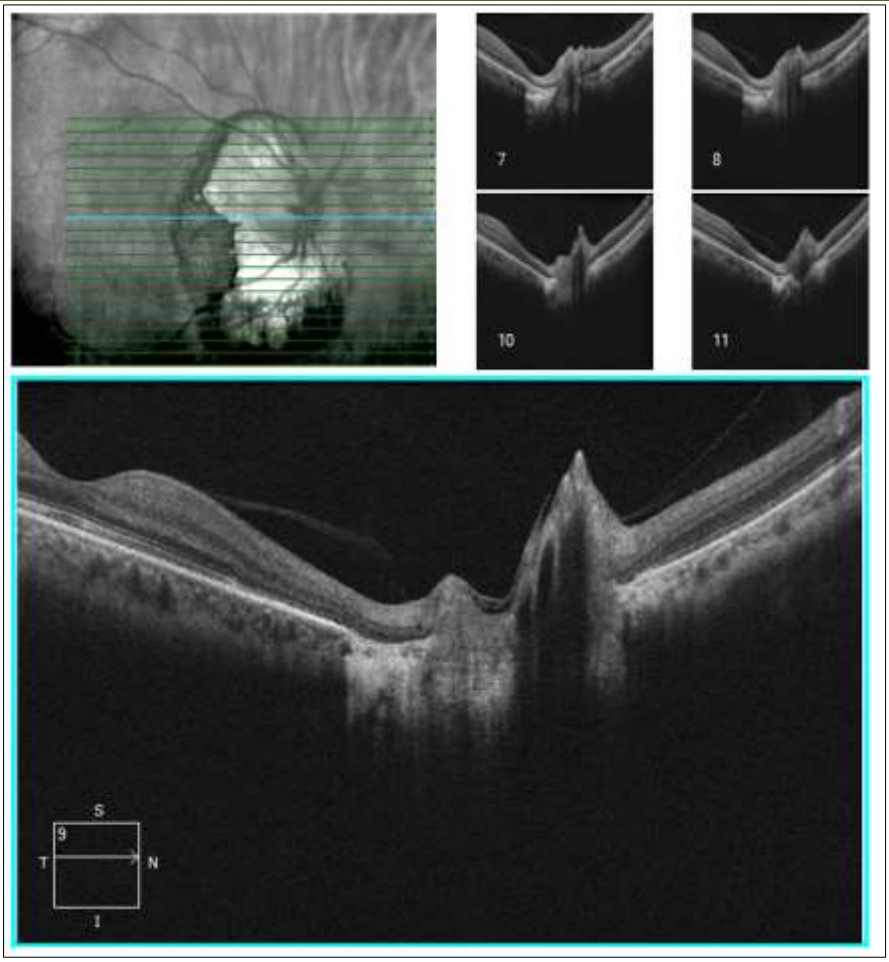

Figure 3: OCT b-scan of the juxtapapillary lesion showing thinning of the neurosensory retina opposite the lesion, and thickening of the retinal pigment epithelium in pigmented areas and it is thinned in lacunar areas

Despite this being a single lesion, a general examination in cooperation with the gastroenterologists came back normal.

The patient is still under surveillance.

DISCUSSION

Congenital hypertrophy of the retinal pigment epithelium is a benign ocular condition characterized by flat, well-defined retinal pigmented lesions [1].

It is a congenital hamartoma of the retinal pigment epithelium. Its prevalence has been estimated at 1.2% [1].

Histologically, it consists of a single-cell layer of hypertrophied retinal pigment epithelium cells, densely filled with large, round macromelanosomes [1].

These lesions are usually unilateral and solitary, although multiple or bilateral cases have been reported. In fact, it presents in three forms: solitary (unifocal), multiple (multifocal) and atypical.

These lesions are usually discovered incidentally during routine ophthalmological examinations, often in asymptomatic individuals.

The solitary form, the most common, typically consists of a single, flat, round, hyperpigmented retinal lesion. Its color can vary from light gray to brown or black, and its edges can be smooth or scalloped [2].

They are generally located at the equatorial level, with a predominance in the upper-temporal quadrant, but can be localized throughout the fundus. Macular involvement is rare. In our case, the lesion is juxta papillary.

Size varies from 100 µm to several papillary diameters. The lesion may be surrounded by a depigmented marginal halo or contain multiple hypopigmented lacunae. These hypopigmented areas tend to enlarge slowly over time.

In the multifocal form, lesions are arranged in clusters. Each cluster may comprise up to 30 lesions, varying in size from 100 to 300 µm [2].

In the atypical form, lesions are generally smaller in diameter (50-100 µm) than solitary lesions, and present clinically as multiple oval, fusiform, comma-shaped lesions distributed haphazardly across the fundus [2].

Bilateral lesions are observed in 78% of patients.

These atypical lesions may be associated with familial adenomatous polyposis, Gardner's syndrome or Turcot's syndrome, almost all of whose patients develop colorectal carcinoma in middle age, in the absence of treatment [3].

Further evaluation and a general multidisciplinary examination are therefore necessary to rule out these associated syndromes.

Lesions are generally hypoautofluorescent due to their high melanin content. Non-pigmented halos or lacunae may be hyperautofluorescent.

On fluorescein angiography, they are hypofluorescent, generally blocking the underlying choroidal fluorescence, except in areas of lacunae or depigmented halos, which may appear hyperfluorescent [4].

Optical coherence tomography reveals retinal thinning and photoreceptor loss over lesions, with thinning or absence of RPE in lacunar areas [4].

The electroretinogram (ERG), electro-oculogram (EOG), A-scan and B-scan ultrasound do not contribute to the evaluation of congenital hypertrophy of the retinal pigment epithelium.

This benign condition generally requires no treatment, but long-term follow-up is necessary.

Patient education regarding the potential risk of growth, nodular transformation, choroidal neovascularization, occurrence of retinal vascular anomalies (capillary rarefaction, microaneurysms, chorioretinal anastomosis), neovascular membranes or, exceptionally, malignant transformation into adenocarcinoma [5, 6].

CONCLUSION

Congenital hypertrophy of the retinal pigment epithelium is a rare and benign condition, generally requiring no treatment, but regular long-term follow-up is mandatory.

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