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Congenital Hypertrophy of the Retinal Pigment Epithelium: Following the Bear Tracks

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Introduction

Congenital hypertrophy of the retinal pigment epithelium (CHRPE) is a well-recognized benign ocular finding, characterized by flat, pigmented lesions.[1] CHRPE lesions are typically unilateral and solitary but can also present as multiple or bilateral occurrences. They are relatively common, asymptomatic, and usually diagnosed incidentally during routine eye examinations. While generally benign, CHRPE or CHRPE-like lesions may warrant further evaluation to rule out systemic disorders such as familial adenomatous polyposis (FAP) or other differential diagnoses, especially in atypical or multiple lesions.

Case Report

A 19-year-old male was referred to our retina service after his optometrist noted multiple pigmented lesions in the peripheral retina of both eyes during a routine eye examination. The patient was asymptomatic, with a best corrected visual acuity of 6/6 in both eyes. Fundus examination revealed multiple flat, round, pigmented lesions with well-demarcated margins arranged in clustered distributions throughout the mid to far periphery of the retina of both eyes, consistent with a "bear track" pattern. These lesions were homogenously hypoautofluorescent.

Although no clear features suggested systemic associations, the gastroenterology service was consulted for evaluation given the widespread extent of the lesions.

Etiology

CHRPE has long been recognized as a benign pigmented lesion presenting in the midperiphery of the fundus.[2] Historically, it has been referred to as "benign melanoma of the RPE[3]" and "hypertrophy with hyperpigmentation of the retinal pigment epithelium (RPE),[4]" terms that describe the phenotypic characteristics of the lesion. Upon identification of its congenital etiology, the term "congenital hypertrophy of RPE" was later adopted.[5]

Although lesions described under the CHRPE terminology can appear similar clinically, they can vary substantially in pathophysiology and management. Currently, there is no formal consensus on the definition of CHRPE; however, three variants have been proposed: solitary, grouped, and atypical.[6]

Solitary: Solitary or unifocal CHRPE refers to solitary and unilateral lesions that develop sporadically and do not have any systemic associations. This form is typically considered to be true CHRPE lesions.[6]

Grouped: Grouped or multifocal CHRPE is usually unilateral and consists of multiple lesions clustered within a quadrant of the retina. These lesions are termed "bear tracks" due to their similarity to animal tracks. Grouped CHRPE is nonhereditary and benign, with no systemic associations.[1]

Atypical: Atypical or multiple CHRPE lesions are usually numerous and bilateral, and have a strong association with hereditary colonic polyposis and colon cancer. To reduce diagnostic confusion with benign CHRPE, multiple studies have proposed various terminology, including "pigmented ocular fundus lesions of FAP[7]," "RPE hamartomas associated with familial adenomatous polyposis,[1]" and "multiple retinal pigment epithelial hamartomata.[8]"

FAP is an autosomal dominant condition defined by the development of >100 synchronous colonic polyps that progress to colon cancer in nearly all cases.[9] FAP arises from germline mutations in the APC tumour suppressor gene located on chromosome 5q21. Up to one-third of newly diagnosed cases occur in individuals with no identified family history and are thought to arise from de novo mutations.[9] FAP associated with extracolonic features—including desmoid tumours, epidermoid cysts, osteomas, dental abnormalities, skin cancers, and CHRPE—are termed Gardner syndrome.[1,9-11] Atypical CHRPE has also been closely linked to APC gene mutations that give rise to medulloblastoma in Turcot syndrome.[12]

Given the potentially fatal nature of untreated FAP, identifying clinical features that distinguish atypical CHRPE from its benign counterparts is essential.

Epidemiology

The prevalence rate of CHRPE in the general population is not well defined in the literature but has been estimated to be 1.2%, with solitary CHRPE comprising the vast majority of cases.[13] Conversely, atypical CHRPE has been reported in up to 78–90% of FAP patients and in approximately 38% of their siblings.[16] The estimated prevalence of FAP ranges from 1 in 8,300 to 1 in 37,000.[9]

Histopathology

Most solitary and grouped CHRPE lesions demonstrate hypertrophy of the RPE monolayer, with cells appearing vertically elongated and columnar compared to normal cuboidal RPE cells.[16] These cells are densely packed with large, round pigment granules known as macromelanosomes, in contrast to wedge-shaped granules seen in normal RPE. The underlying Bruch's membrane may appear thicker due to enlargement of the RPE basement membranes, while the remainder of Bruch's membrane, choriocapillaris, and choroid remain normal. The inner retina and retinal vasculature also remain unaffected.[6] All lesions are characteristically flat.

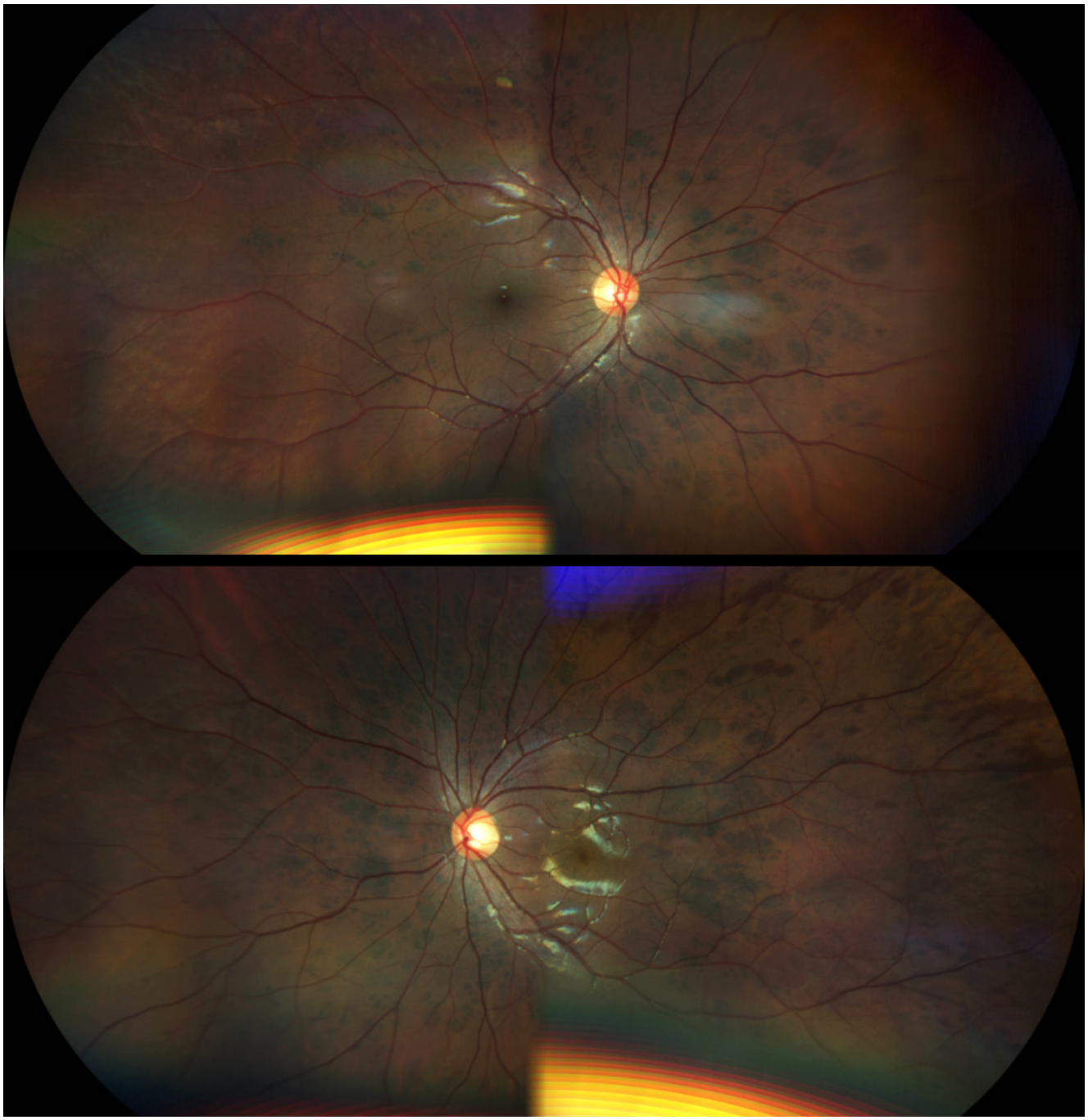

Figure 1. Montage of colour fundus images of the right (**top**) and left (**bottom**) eyes, demonstrating multiple pigmented lesions in both eyes; *patient data and images courtesy of Freddy Lee, MD and Amit Mishra, MD, FRCSC.*

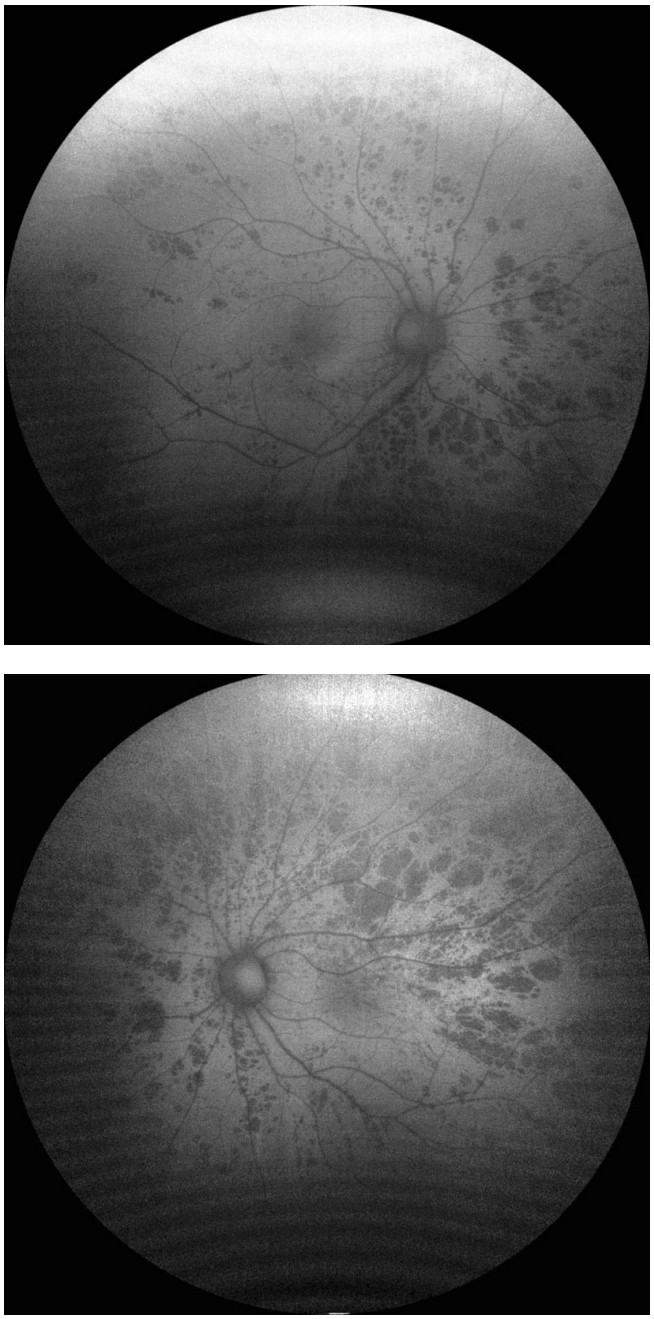

Figure 2. Fundus autofluorescence images of the right (**top**) and left (**bottom**) eyes, indicating hypoautofluorescence of the lesions in both eyes; *patient data and images courtesy of Freddy Lee, MD and Amit Mishra, MD, FRCSC.*

In contrast, atypical lesions associated with FAP demonstrate RPE hypertrophy and hyperplasia, full-thickness retinal involvement, and associated retinal vascular changes. These histopathologic features classify atypical lesions as hamartomas of the RPE.[16]

Clinical Findings

Solitary CHRPE

Solitary lesions are typically flat, round, and hyperpigmented, with well-demarcated margins separating them from the adjacent normal RPE. They may be associated with a surrounding halo or inner "punched-out" lacunae that appear depigmented.[16] The overlying retina and vasculature are typically normal; however, retinal vascular changes including blood vessel obliteration, microaneurysms, and neovascularization may be observed in some lesions.

Solitary CHRPE lesions are most commonly found in the equatorial region, within the superotemporal[16] and inferotemporal[2] quadrants. Rarely, lesions can be found in the peripapillary region or in the macula. Foveal involvement or macular neovascularization are associated with decreased vision.[6,17] The mean lesion diameter is approximately 4.7 mm but can vary from 100 µm to lesions occupying an entire retinal quadrant. Solitary lesions can demonstrate slow and benign enlargement in 46–83% of cases.[16]

Grouped CHRPE

Grouped or multifocal CHRPE consists of clusters of flat, pigmented lesions that are well demarcated. Each cluster contains 3–30 lesions, which are generally smaller than solitary lesions and vary in size from 100–300µm.[1] Lesions tend to increase in size toward the fundus periphery. Grouped CHRPE can resemble animal footprints ("bear tracks") and is typically confined to a single quadrant, though several clusters can be present in one eye.[6,16] In contrast to solitary lesions, grouped CHRPE lesions lack hypopigmented halos and lacunae.

Rarely, clusters of lesions can appear depigmented and albinotic, which are termed "polar bear tracks" and are not associated with gastrointestinal malignancy.[6]

Atypical CHRPE

Atypical CHRPE lesions associated with FAP are distinguished from their benign counterparts by characteristic clinical features. Unlike solitary CHRPE, atypical lesions are bilateral and numerous, with less well-demarcated margins. Compared to grouped CHRPE, they lack a specific distribution pattern, and instead display irregular and depigmented borders, with variable morphologies including pisciform (fish-shaped), oval, comma, or comet configurations.[1,6,16] Atypical CHRPE lesions are typically smaller (50–100µm) and exhibit varying pigmentation patterns of brown, black, and light grey. Notably, these lesions commonly display aberrant features such as retinal invasion, proliferation of glial cells, capillaries, RPE, pigmented satellite lesions, and mottled RPE changes.[1,16]

The presence of atypical CHRPE is strongly associated with Gardner syndrome; however, the absence of these lesions has no predictive or diagnostic value in the diagnosis of FAP.

Diagnostic Procedures

The diagnosis of CHRPE is clinical, and additional investigations are generally not necessary. Fundus images are helpful for documentation and longitudinal monitoring of lesions. In cases where the diagnosis is uncertain, ancillary testing may be beneficial in distinguishing lesions from other conditions in the differential diagnosis.

Fundus autofluorescence

On fundus autofluorescence imaging, CHRPE lesions display complete hypoautofluorescence. Surrounding depigmented halos or lacunae are generally isoautofluorescent or slightly hyperautofluorescent.[18]

Optical coherence tomography (OCT)

OCT reveals hyperreflectivity and thickening of the RPE, corresponding to the hypertrophy of the RPE, as well as retinal thinning and photoreceptor loss overlying the lesions. Lacunae are associated with RPE loss and increased transmission of light into the underlying tissues.[19]

Fluorescein angiography

On fluorescein angiography, the hypertrophic RPE blocks visualization of the underlying choroidal vasculature, which appears as hypofluorescent lesions. Depigmented halos or lacunae are typically hyperfluorescent, consistent with window defects. CHRPE lesions do not demonstrate leakage; however, subtle microvascular changes, including capillary nonperfusion, neovascularization, and capillary microaneurysms may be observed.[20]

OCT angiography (OCT-A)

The utility of OCT-A is generally limited due to segmentation issues related to retinal thinning and RPE thickening. When interpretable, OCT-A typically shows normal retinal and choroidal vasculature in CHRPE.[6]

A-scan or B-scan ultrasonography

Ultrasonography is non-diagnostic in CHRPE as lesions are flat and may not be readily detected. However, it can be helpful for measuring lesion thickness to rule out differential diagnoses.[6]

Electrophysiology

Electrophysiologic testing, including electroretinography and electro-oculography, is typically normal and non-contributory for the diagnosis of CHRPE.[6]

Differential Diagnoses

Choroidal nevus

Choroidal nevi typically have margins that are not well defined with a feathery appearance and lack associated lacunae. Pigmentation may range from light to dark brown, and overlying drusen or mottling may be present.

Choroidal melanoma

Choroidal melanoma is almost always elevated, with less homogenous pigmentation and demarcated margins. Growth is usually observed in all three dimensions.

Melanocytomas

Melanocytomas can resemble CHRPE clinically but are distinguished by their elevated profile and feathery margins.

Congenital simple RPE hamartoma

Clinically, these lesions appear as distinct, solitary nodules that protrude into the neurosensory retina and are most commonly located in the macular region.[1]

Combined hamartoma of the retina and RPE

Combined hamartoma of the retina and RPE are characterized by an ill-defined grey retinal mass with tortuous and straightened retinal vessels.[1]

Other pigmented fundus lesions such as focal pigmentation from trauma, inflammation, or drug toxicity, can be differentiated based on their irregular shape, associated findings, and the patient's clinical history to suggest other acquired conditions.

Management

The vast majority of patients with solitary or grouped CHRPE remain asymptomatic. These lesions are benign and do not require active intervention. The prognosis is excellent, and patients should be counselled to continue routine ophthalmic follow-up with regular eye examinations.[1]

In contrast, patients suspected of having atypical CHRPE associated with FAP require further evaluation by gastroenterology and adherence to established screening protocols to enable prompt detection of colon polyps and malignancy. Patients with a diagnosis of FAP should undergo flexible sigmoidoscopy or colonoscopy every 1–2 years, starting at 10–12 years of age or earlier if symptoms such as diarrhea, rectal bleeding, or abdominal pain develop.[9] Given that colonic adenocarcinoma is inevitable in FAP, ophthalmologists should maintain a high index of suspicion and a low threshold for identifying atypical CHRPE and initiating a referral for gastroenterology assessment.

Complications

Complications related to CHRPE are exceedingly rare and are associated with its benign growth behaviour. In some cases, CHRPE lesions can enlarge slowly, with reports of foveal extension that may affect visual acuity.[17] Nodular growth has been reported in solitary CHRPE, which can acquire a retinal feeding artery and draining vein and result in exudative retinal detachment.[1,6] CHRPE can also be complicated by choroidal neovascularization or cystoid macular edema, resulting in decreased vision.[1,21]

Conclusions

CHRPE is a common, benign finding that is often identified incidentally and referred for ophthalmologic assessment. Unfortunately, the absence of a consensus on a definition or classification for CHRPE may contribute to challenges in identifying atypical lesions associated with FAP. Although certain clinical features can help distinguish atypical CHRPE from its benign counterparts, there is considerable overlap in "high-risk" characteristics, such as ovoid shape and bilaterality, among solitary and grouped CHRPE, which can confound definitive clinical diagnoses.

Given that atypical CHRPE can indicate inevitable progression to colon cancer, referral to gastroenterology may be prudent in cases such as ours, in which lesions do not demonstrate classical features of solitary or grouped CHRPE but also lack clearly defined atypical features.

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As with any case study, the results should not be interpreted as a guarantee or warranty of comparable results. Individual results may vary depending on the patient's circumstances and condition.

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