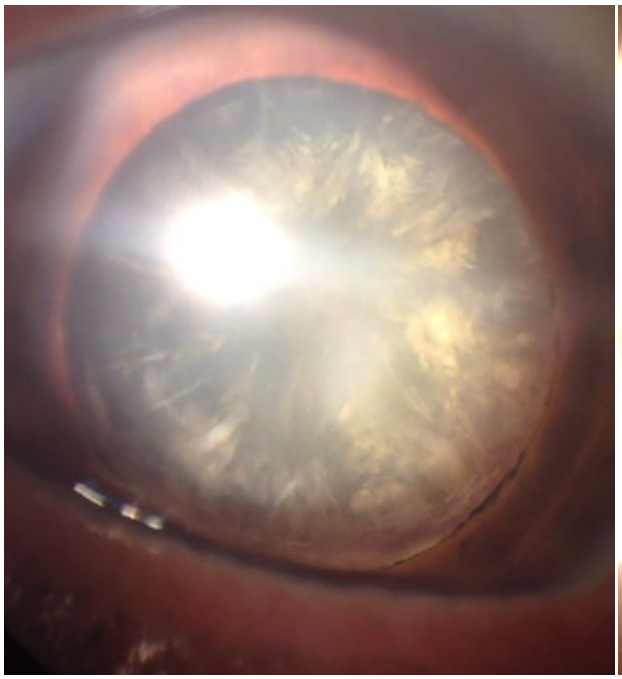
MYSTERY IMAGE

BLINK



Asaf Achiron, MD, Tel Aviv University, Israel.

WHAT IS THIS MONTH'S MYSTERY CONDITION?
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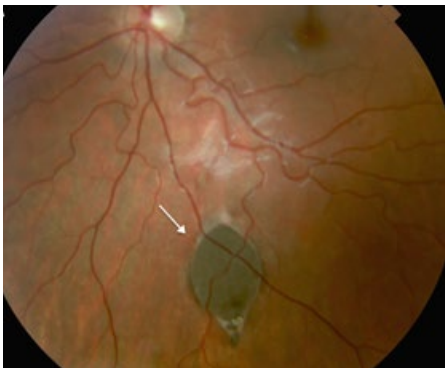
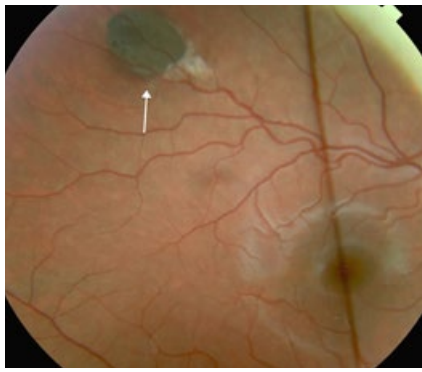
LAST MONTH'S BLINK

Congenital Hypertrophy of RPE in Familial Adenomous Polyposis





A 17-year-old girl presented with bilateral lesions found on funduscopic examination. She had a history of familial adenomatous polyposis (FAP) and denied having any ocular symptoms. Her BCVA was 20/20 in both eyes. The 5 lesions in her left eye and 1 in her right eye were determined to be congenital hypertrophy of retinal pigment epithelium (CHRPE), a hereditary thickening of the retinal pigment epithelium (RPE) layer of the retina.

CHRPE lesions are asymptomatic, flat, and hyperpigmented and vary in color (they may be gray, brown, or black), and they have smooth or scalloped margins that are well-demarcated from the rest of the RPE, which appears normal. CHRPE lesions that are multifocal and bilateral are associated with FAP.[1] This is an autosomal dominant disease that presents with ≥ 100 polyps in the colon in the patient's late 20s or early 30s; it is linked to mutation of the tumor suppressor gene adenomatous polyposis coli. These polyps ultimately develop into colon cancer through the adenoma-carcinoma sequence.

CHRPE lesions are benign and require no treatment. The patient underwent prophylactic total colectomy and remains without ocular symptoms.

1 Meyer CH, Gerding H. In: Ryan SJ et al., eds. *Retina. 5th ed.* London: Saunders; 2013:2209-2213.

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