Making a Gut Decision

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Introduction:

A 54-year-old man was referred to the retina clinic for findings of abnormal fundus lesions on routine examination. The patient admitted he had not had an eye examination for most of his adult life, until now, as he had noticed gradual difficulty with near vision in both eyes while reading. Otherwise, he had no acute symptoms. The patient worked outdoors in the sun for many years and had a history of cutaneous malignant melanoma of an upper extremity, status post resection and chemotherapy over 12 years ago, in remission. He was not in touch with any of his immediate family and did not know of any family history of major eye problems.

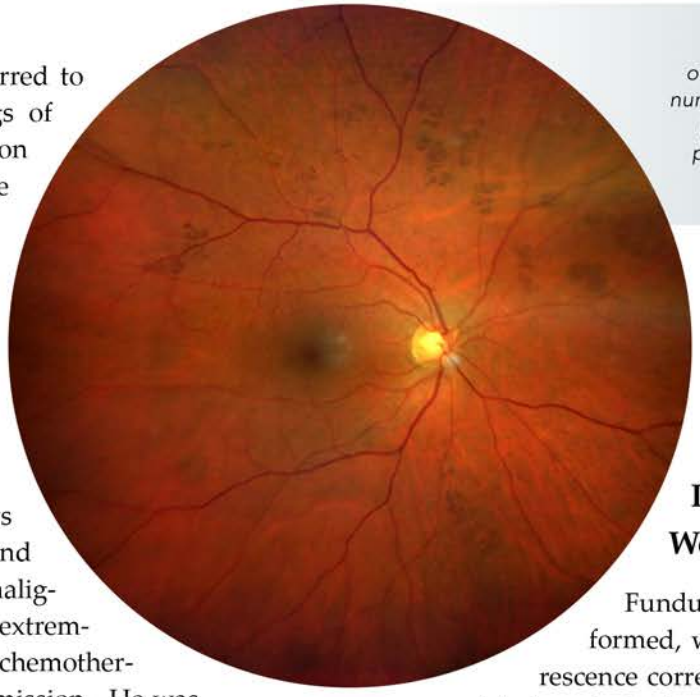
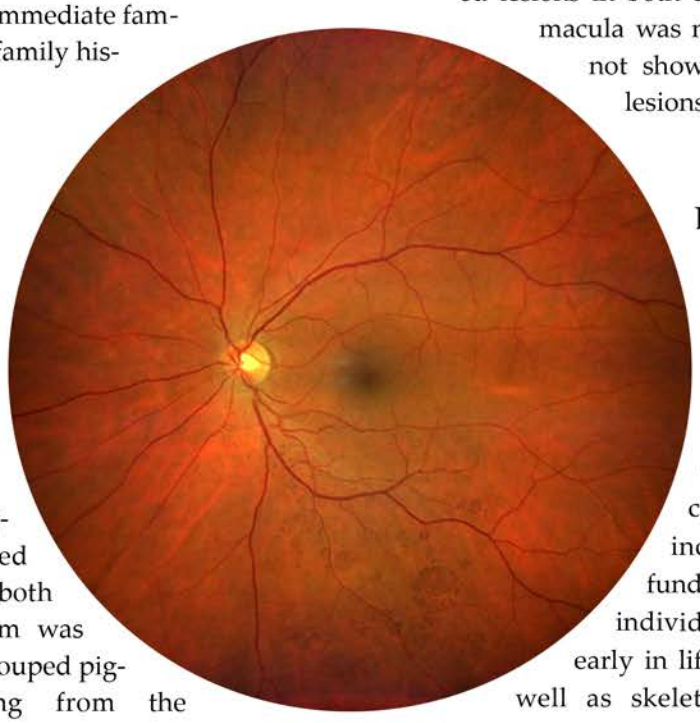


Figure 1 – Fundus photographs of the right and left eyes showing numerous grouped, flat pigmented lesions extending from the peripapillary region to the periphery, partly involving the macula.

Exam:

Best corrected distance visual acuity was 20/20 in both eyes, intraocular pressure and pupil testing were normal bilaterally, and visual fields were full to confrontation finger counting bilaterally. Anterior segment examination revealed mild nuclear sclerosis in both eyes. Dilated fundus exam was notable for numerous flat, grouped pigmented lesions extending from the peripapillary region to the periphery in both eyes, partly involving the macula (Figure 1).



Imaging and Further Workup:

Fundus autofluorescence was performed, which showed hypoautofluorescence corresponding with the pigmented lesions in both eyes (Figure 2). OCT of the macula was normal in both eyes and did not show any abnormality over the lesions.

Discussion:

This patient was referred due to concern for Gardner syndrome, a life-threatening condition of familial adenomatous polyposis (FAP) along with extracolonic manifestations, including pigmented ocular fundus lesions (POFLs). In FAP, individuals develop colon cancer early in life (by the fourth decade), as well as skeletal hamartomas, osteomas,

desmoid tumors, cutaneous cysts, and other tumors [1, 2]. These concerning POFLs, more recently termed retinal pigment epithelium (RPE) hamartomas associated with FAP (RPEH-FAP), are multiple, bilateral solitary pigmented lesions randomly dispersed in multiple quadrants and without sectoral distribution [1]. They are usually round or ovoid with pisiform, comma, or comet configurations, with irregular borders and sometimes areas of depigmentation similar to lacunae in congenital hypertrophy of the RPE (CHRPE) [1, 2]. They often occur in the posterior pole or near vortex veins and histologically may represent RPE hypertrophy or hyperplasia [1].

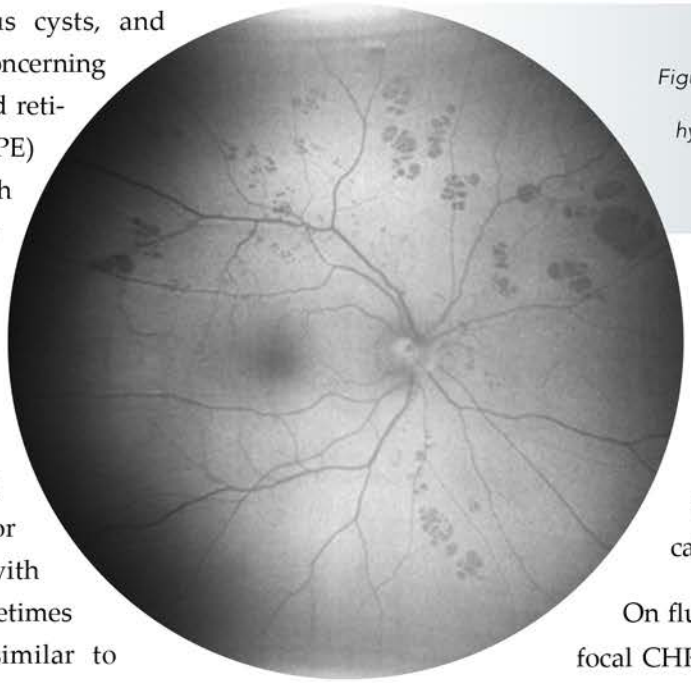
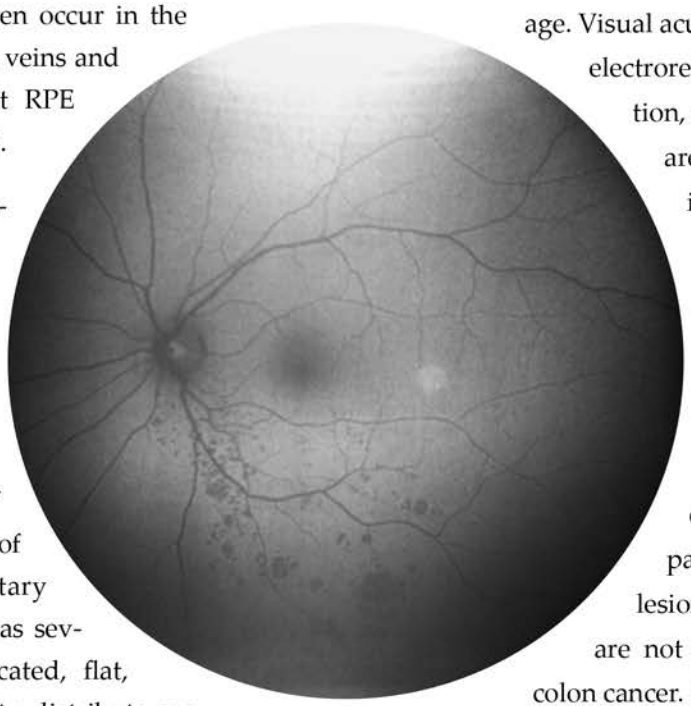


Figure 2 – Fundus autofluorescence of both eyes showing hypopigmentation corresponding to the pigmented lesions.

However, this patient's presentation was more consistent with a classic presentation of an alternate diagnosis, multifocal CHRPE, or congenital grouped pigmentation, sometimes referred to as "bear tracks." This is considered a variant of the more common solitary CHRPE lesion and presents as several groups of well-demarcated, flat, slate-gray lesions that tend to distribute sectorally, with larger lesions located more peripherally. It is not common to see lacunae or depigmented haloes surrounding each of these lesions [1]. On light and electron microscopy of these lesions, RPE cells contain increased numbers of large pigment granules, but not notable hypertrophy or hyperplasia of the RPE [3]. Multifocal CHRPE lesions tend to remain stable, and the condition is sporadic and non-hereditary [1]. Sometimes they are clinically depigmented and called congenital grouped albinotic spots, or "polar bear tracks," however there does not seem to be a difference in natural history of these compared with multifocal CHRPE [1, 4-6].



On fluorescein angiography, multifocal CHRPE shows blocked choroidal fluorescence over the lesions, with no leakage. Visual acuity, color vision, visual fields, electroretinogram (ERG), dark adaptation, and electrooculogram (EOG) are typically within normal limits [7]. Notably, multifocal CHRPE or "bear tracks" have no increased association with FAP, as several studies have demonstrated, despite their similarity to the RPEH-FAP lesions discussed above [1, 2, 8]. Thus, patients with typical bear track lesions as our patient in this case are not at increased risk of familial colon cancer. Some authors have, however, recommended special consideration for patients with another type of pigmented lesion, torpedo maculopathy, which may bear strong resemblance to lesions seen in FAP / Gardner syndrome. Due to overlap in clinical appearance, these authors suggest screening patients with torpedo maculopathy for mutations in the FAP gene (adenomatous polyposis coli) as well as for colonic polyps or other related tumors and recommending dilated fundus examination of their relatives [9].

Management:

In light of the above, this mid-fifty patient with classic bear tracks and absence of family history of familial cancer syndrome was not felt to have an increased risk of colon cancer. He was advised, however, to continue routine health maintenance including evaluation by his primary care physician and age-appropriate screening for colorectal cancer.

Bear Tracks

References:

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