

Case Report

CONGENITAL HYPERTROPHY OF RETINAL PIGMENT EPITHELIUM (CHRPE) WITH TYPICAL 'BEAR TRACK' PRESENTATION

Grizzly bear attacks

Amit S Nene¹, Pushpanjali Ramteke¹, Smitesh Shah²

ABSTRACT

Familial adenomatous polyposis (FAP) is an autosomal dominant condition caused by a mutation in APC tumor suppressor gene (chromosome 5q21). Affected patients have nearly 100 percent progression to colorectal cancer. Solitary congenital hypertrophy of retinal pigment epithelium (CHRPE) and its multi focal variant, congenital grouped pigmentation (bear tracks), are isolated ophthalmic entities and are not associated with FAP and Gardner syndrome. Patients found to have solitary or multifocal CHRPE on ophthalmoscopy can be counselled that they do not have an increased risk of colon cancer compared with that of the general population.

Keywords: CHRPE – FAP - colorectal cancer - bear track - Gardner syndrome

Case report

A 54-year old female presented in our OPD for routine eye check-up with no specific complaints. On examination best corrected visual acuity was 20/20 N6 with fundus evaluation showing multiple pigmented lesions which were bilateral, sharply circumscribed and variably sized.

Discussion

Familial adenomatous polyposis (FAP) is an autosomal dominant condition caused by a mutation in APC tumor suppressor gene (chromosome 5q21).[1] Affected patients have variable number of adenomatous polyps, mostly in the large intestine, that have nearly 100 percent progression to colorectal cancer with extracolonic manifestations such as desmoid tumours, osteomas, dental anomalies, and soft-tissue tumours. Solitary CHRPE and its multi focal variant, congenital grouped pigmentation (bear tracks), are isolated ophthalmic entities and are not associated with FAP and Gardner

syndrome. It is characterized by multifocal flat, well-demarcated hyperpigmented lesions in a grouped configuration with a larger lesion surrounded by several smaller ones resembling the footprint of an animal, or 'bear tracks'. [2] Figures [A,B,C]. RPE lesions seen with FAP are ovoid, multifocal, bilateral with irregular borders, and haphazardly distributed with peripheral location often with a white tail of depigmentation at margins. Differential diagnosis include choroidal nevus, melanoma, and chorioretinal scar. Choroidal nevi are usually flat and fairly round, have ill-defined borders, grey in color, with presence of surface drusen, while choroidal melanoma is most often greater than 2 mm and has sentinel blood vessel. Patients found to have solitary or multifocal CHRPE on ophthalmoscopy can be counselled that they do not have an increased risk of colon cancer compared with that of the general population.

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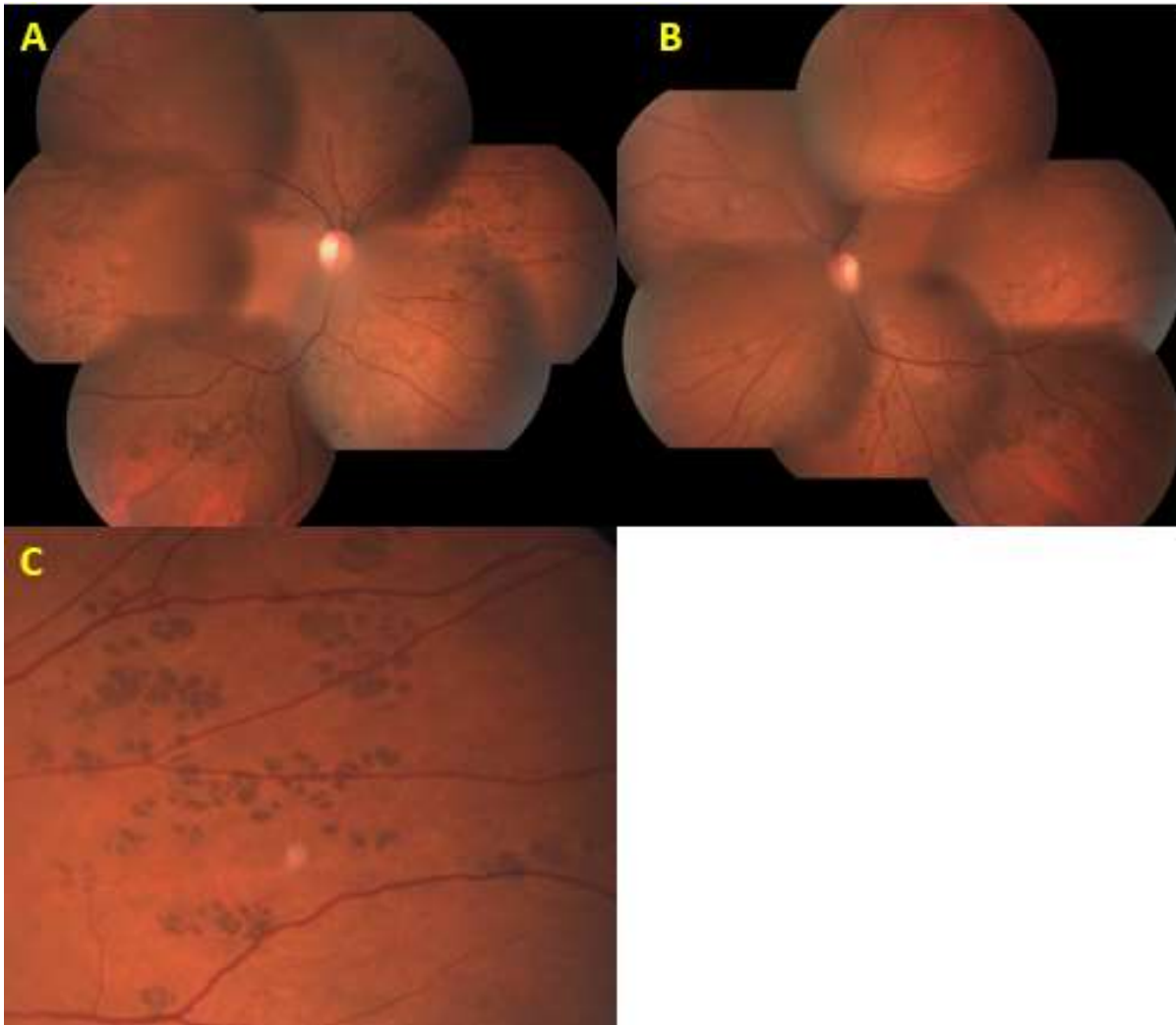


Figure A, B & C: shows multifocal flat, well-demarcated, hyperpigmented lesions of the retina in a grouped configuration with a larger lesion surrounded by several smaller ones resembling the footprint of an animal, or "bear tracks", near the equatorial region

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Received Date: October 12, 2018
Accepted Date: November 26, 2018
Published Online: March 5, 2019

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