Congenital hypertrophy of the retinal pigment epithelium

Group pigmentation; bear tracks

DESCRIPTION

Congenital hypertrophy of the retinal pigment epithelium (CHRPE – 'chirpy') is a benign condition where the pigmentary lesions may be unilateral or bilateral, solitary or multifocal, with the latter sometimes being in the form of smaller patches that are grouped together, giving the impression of animal paw-prints (bear tracks) in the fundus. Lesions of an atypical appearance may be a marker for familial adenomatous polyposis (FAP), most commonly in Gardener's syndrome, an autosomal dominant condition where the colon polyps have a 100 per cent chance of becoming malignant. In CHRPE, the retinal pigment epithelial (RPE) cells are larger and contain more melanin granules than normal. Adenocarcinoma developing from CHRPE has been reported but is considered a rare occurrence.

SYMPTOMS

There are no symptoms associated with CHRPE.

SIGNS

A typical CHRPE is flat and black or grey, with a sharply defined border, contrasting with the slate-grey, ill-defined appearance of a choroidal naevus. They can range from very small to many disc diameters in size. Frequently, a depigmented halo lies just inside the edge of the lesion. Further depigmented areas or lacunae may develop within the lesion with time. This is sometimes referred to as a sunburst effect. Some CHRPEs are conspicuously lacking in pigment, just having a pigmented edge. The multifocal, so-called bear-track form of CHRPE is often confined to a specific area within the fundus. CHRPE that are associated with FAP are described as atypical, being oval or even fishtail-like in shape, bilateral, more isolated in position rather than grouped together and variable in size up to approximately a disc diameter. CHRPE block choroidal fluorescence on fluorescein angiography.

PREVALENCE

Multifocal CHRPE are fairly common and occur in about 1 per cent of the population. Solitary lesions in one eye are less common.

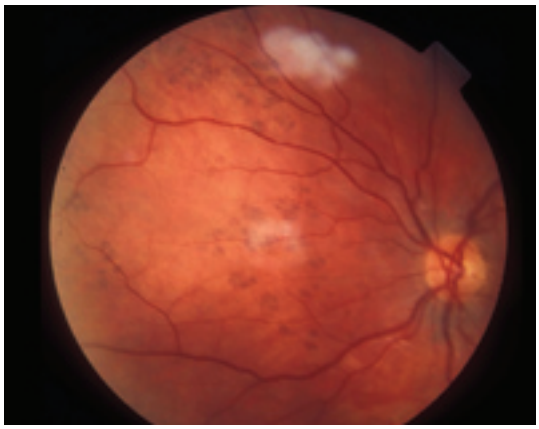

FIGURE 1. 'Bear-track' deposits of CHRPE situated nasal to the left optic disc

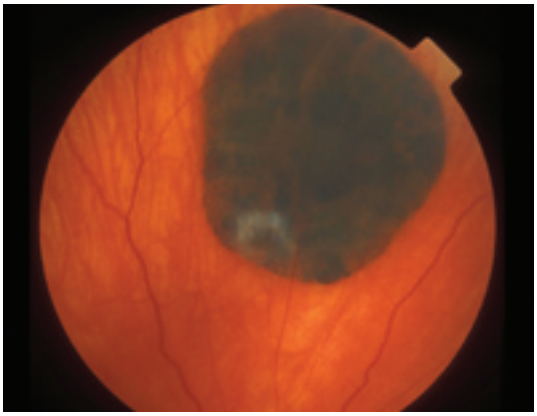

FIGURE 2. Large CHRPE in the superior fundus

CHRPE associated with FAP are rare and are seen in about one in 100,000 people.

DIFFERENTIAL DIAGNOSIS

Other pigmented lesions of the fundi include: melanoma – choroidal, choroidal naevus, combined hamartoma of the RPE, choroidal metastases from malignant melanoma elsewhere in the body, retinal adenocarcinoma, chorioretinal scarring (RPE hyperplasia).

MANAGEMENT

Imaging

Routine examination and serial photography is all that is usually required.

Advice

There is no treatment for CHRPE. The possibility of adenocarcinoma arising from a CHRPE is rare. The practitioner should keep in mind the potential systemic association with FAP, albeit uncommon, and refer patients with suspicious lesions or a suggestive case history to their general practitioner for evaluation. Single lesions in one eye, or those forming grouped pigmentation are not considered to be associated with FAP.

The full series of these articles will be available in the book *Posterior Eye Disease and Glaucoma A-Z* by Bruce AS, O'Day J, McKay D and Swann P. £39.99 For further information click on the Bookstore at www.opticianonline.net.

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