

Congenital Hyperplasia of the Retinal Pigment Epithelium & Familial Adenomatous Polyposis

You may have been referred to the eye clinic because your doctor or optician has noticed a dark patch at the back of your eye called a “CHRPE”. CHRPE stands for Congenital Hyperplasia of the Retinal Pigment Epithelium. These are areas where the pigmented cells of the retina grew more than usual during development – giving rise to dark, flat areas on the retina. People with CHRPE are born with them, although they are usually diagnosed later in life during a routine eye exam.

Is CHRPE dangerous?

CHRPE lesions in themselves are not dangerous for your eye or general health. However, there is a type of abnormality very similar to CHRPE which can be linked with a condition called **Familial Adenomatous Polyposis (FAP)**. There are particular features of these abnormalities which distinguish them from typical CHRPE, including the number, shape and appearance of them. If there is a suspicion that you have the type of abnormality associated with FAP, your doctor will refer you for further investigations. People with these FAP-related abnormalities usually know of a family member with the same condition. In contrast, **if you have typical CHRPE, there is no need for any further investigations as they are not linked to any other conditions.**

Types of CHRPE/FAP lesions

Typical CHRPE come in two forms – unifocal or multifocal. Unifocal CHRPE appear as a single round and flat, darkly pigmented area with smooth borders (Fig 1). Multifocal CHRPE appear with several patches where one larger area is surrounded by several small areas, giving an appearance like a bear paw print – these are sometimes called “bear tracks” (Fig. 2). These types of CHRPE are usually just present in one eye, although there are cases where they are in both. Bear tracks and unifocal CHRPE are not associated with FAP.

In contrast, “CHRPE” abnormalities associated with FAP tend to appear in both eyes, and are randomly distributed across the retina. They are usually smaller and more oval shaped than typical CHRPE, and have irregular borders with one edge being depigmented and shaped like a comma or fish tail (Fig 3).



Figure 1. Typical CHRPE

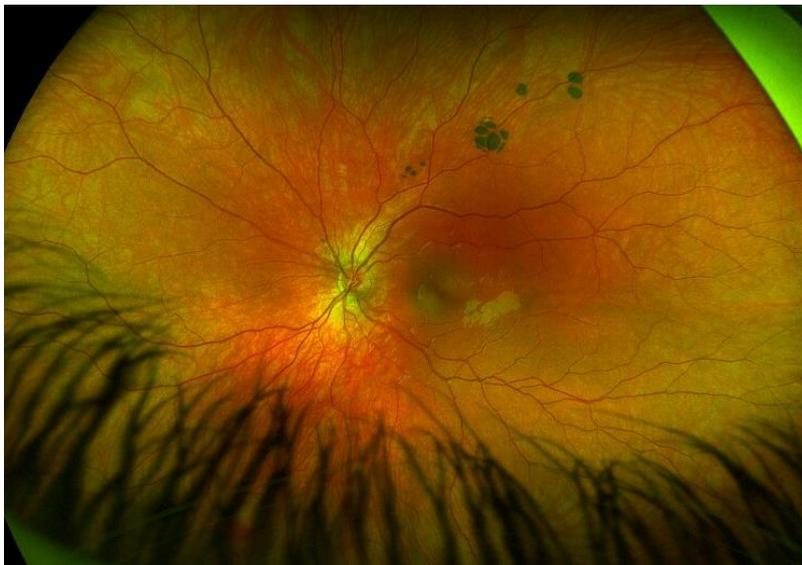


Figure 2. "Bear tracks" (multifocal CHRPE)



Figure 3. Pigmented lesions associated with FAP

What is FAP? Including what further investigations are required

FAP stands for Familial Adenomatous Polyposis. This is a condition where the large intestine (colon) is prone to developing benign growths called polyps. However, these polyps can become cancerous and so it is important to be referred to a bowel specialist for assessment. Therefore, if you have the abnormalities in your retina linked with this condition, you will be referred to see a specialist. FAP is caused by a mutation in the “APC” (adenomatous polyposis coli) gene. If suspected, you will have a blood test for this gene and be referred to a clinical geneticist to help confirm or exclude the diagnosis.

Do I need any treatment for my eyes?

CHRPE lesions, including the type associated with FAP, do not require any ocular treatment and do not need monitored by the hospital eye department. We would recommend you attend your local optician on a yearly basis.