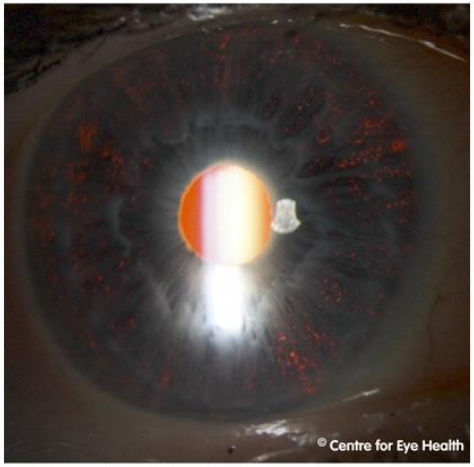
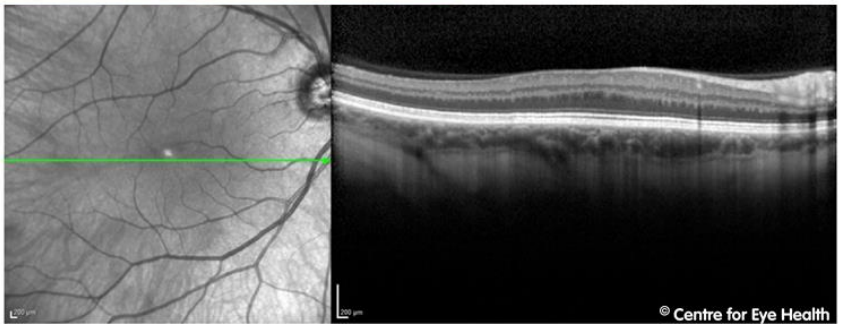
CFEH facebook Cases

CFEH Facebook Case #21

A 54 year old Caucasian female presented to CFEH as her optometrist noted reduced visual acuities. Cover testing showed alternating intermittent exotropia. Entering aided acuities were 6/12+2 OD and 6/9.5 OS. There was no improvement with pinhole. As both eyes have a similar appearance, only images of the right eye are shown. Optomap, anterior eye images and OCT line scans taken through the macula region at the fovea are shown. What is the most likely diagnosis and optometric management of this patient?





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ANSWER

Ocular Albinism.

The anterior eye image shows transillumination defects while the Optomap image suggests reduced retinal pigmentation and the OCT line scans reveal foveal hypoplasia (an absence of foveal reflex and foveal pit). These findings, and the reduced visual acuities suggest a diagnosis of ocular albinism.

Foveal hypoplasia is a common to all types of ocular albinism, however this can be variable in its magnitude and is important in determining visual acuity. The degree of hypopigmentation varies according to the type of albinism. Other associated signs and symptoms can include reduced visual acuity, photophobia, congenital nystagmus, refractive errors, prominent choroidal vasculature, optic disc hypoplasia and transillumination defects

Ocular albinism usually shows an x-linked inheritance pattern, although occasionally can be autosomal recessive. Female carriers of the x-linked type can show partial iris translucency and scattered areas of depigmentation but are otherwise asymptomatic.

As part of the optometric management, an optic nerve head assessment, gonioscopy, visual fields and IOPs should be performed to exclude pigment dispersion syndrome. Management options for ocular albinism include low vision aids, tinted lenses and consideration of genetic counselling.