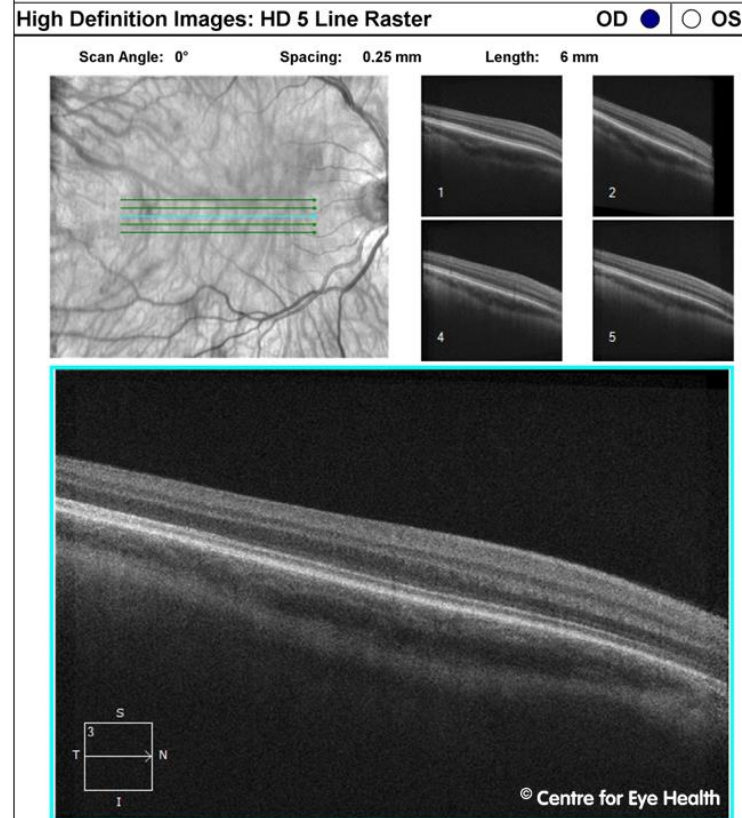
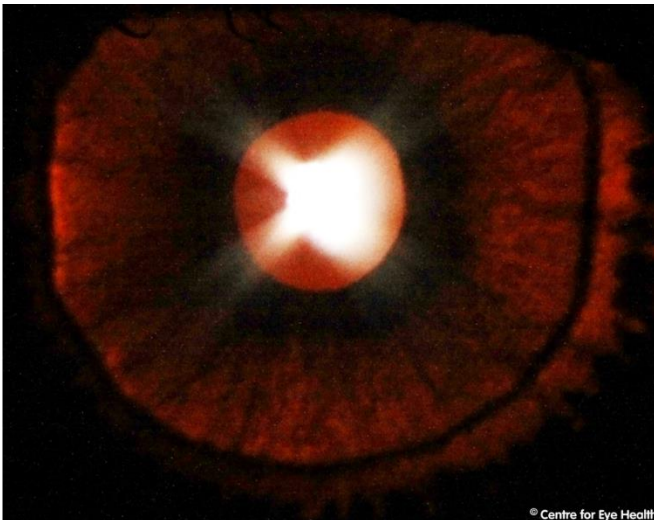




CFEH Facebook Case #116

A 36 year old female presented for assessment. Her visual acuity is 6/38+ OD and 6/24- OS, she has nystagmus and mentions she is hyper-sensitive to bright lights. Anterior eye and macular OCT images are below (note that both eyes had a similar appearance). What systemic condition does this patient have?



ANSWER

Ocular Albinism.

The anterior eye images show reduced pigmentation of both the iris and the lashes, most notable on the lower lid where eye makeup is absent. In the second image, retro-illumination defects of the iris may be clearly seen and the macular OCT scan shows an absence of the normal foveal pit and reflex (foveal hypoplasia). These signs, along with the reduced visual acuity and noted nystagmus are characteristic of ocular albinism.

Other signs and symptoms that may be associated with ocular albinism include refractive errors, prominent choroidal vasculature, and optic disc hypoplasia. 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.