A 57 year old Asian female was referred to CFEH for investigation of a pigmented lesion at right the optic nerve head. As the left eye was unremarkable, this case will focus on the right eye only. Best corrected acuity was 6/6, pupils were equal and reactive to light, with no relative afferent pupillary defect. The patient’s medical, ocular and family history were all unremarkable. What is your diagnosis and management of this patient?
ANSWER

Melanocytoma of the optic nerve head.

This patient has an irregularly pigmented dark lesion that is located partly on the disc, extending over the superior margin and onto the retina. The disc margins appear somewhat undefined. The OCT line scans show a dome-shaped elevation at the superior aspect of the disc, with posterior optical shadowing. The b-scan ultrasound shows a small area of hyper-reflectivity at the right optic nerve head, indicating an elevated and probably solid lesion.

A melanocytoma of the optic disc is a typically benign pigmented tumour. It is a variant of a naevus and can also involve the uveal tract (iris, ciliary body and choroid). They usually present as an isolated entity, with no proved systemic associations, and rarely cause visual impairment, however associated complications may occasionally occur. These complications can include disc oedema, intraretinal oedema, subretinal fluid, retinal haemorrhage and exudation, central retinal vein occlusions, neuroretinitis and malignant transformation (1-2% of cases).

Melanocytoma has equal incidence in all races with a slight predilection for females. It is typically unilateral although bilateral cases have been reported in children. Lesions are typically small with average measurements of 2mm in diameter and 1mm in apical thickness.

Recommended management involves 6-12 monthly review (depending on the level of risk or concern over malignancy). Examination should include stereoscopic examination, photography, OCT and automated perimetry.