

CFEH Facebook Case #15

A 44 year old female was referred to CFEH for a macular assessment regarding an unusual fundus appearance. The patient feels as though her night vision is getting worse although remarked that it was improved by spectacles. Family ocular history includes optic neuritis (mother).

Best corrected acuities were 6/4.8 OD and 6/7.6+2 OS with a low hyperopic script. Pupils were equal, round and reactive to light in both eyes. Amsler Grid was unremarkable in the right eye, and described as being "wobbly" in the left. Contrast sensitivity was within the normal range in both eyes.

Retinal photography, Optomap, fundus autofluorescence and OCT line scans are below. Only the left eye images are shown here for simplicity, however both eyes were similar in appearance for all imaging.

What would be your diagnosis and management of this patient?

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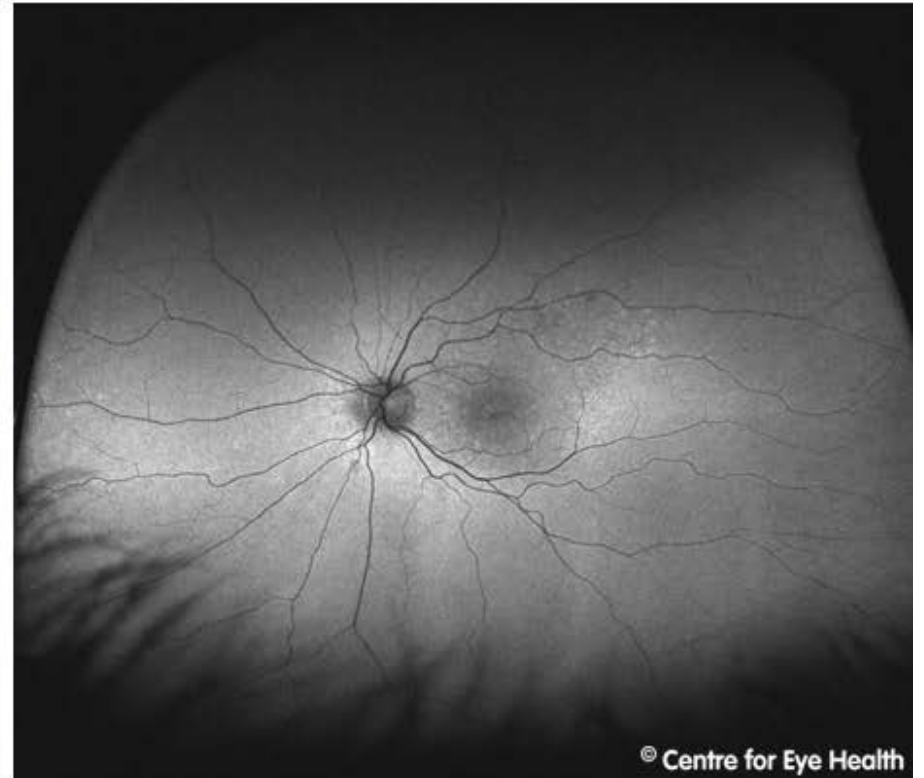


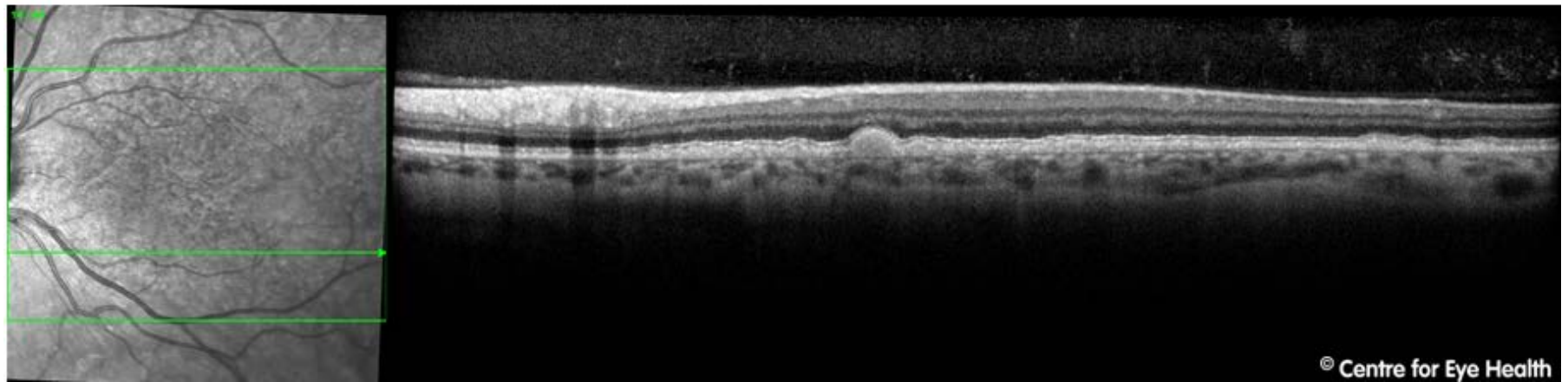
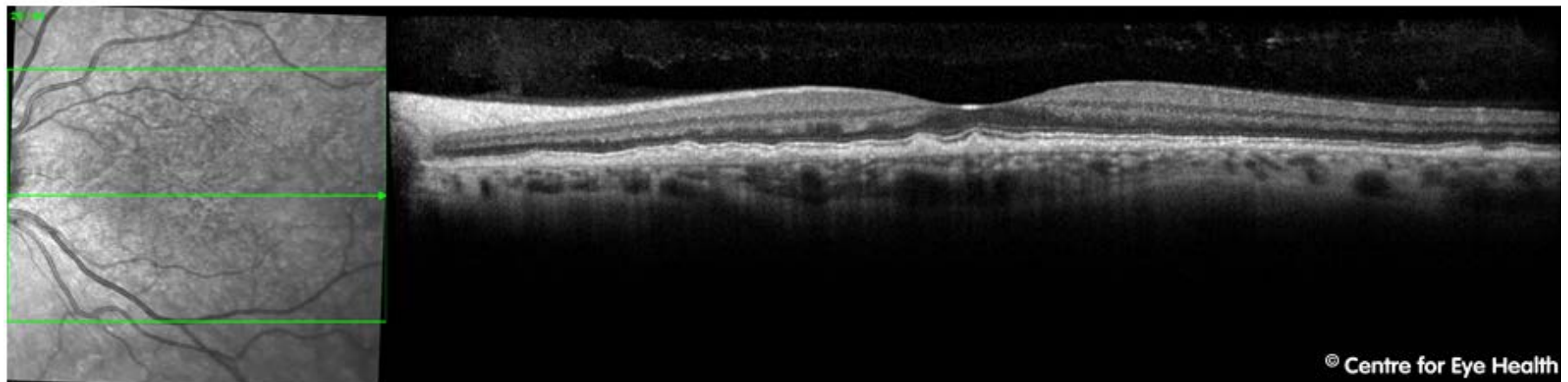
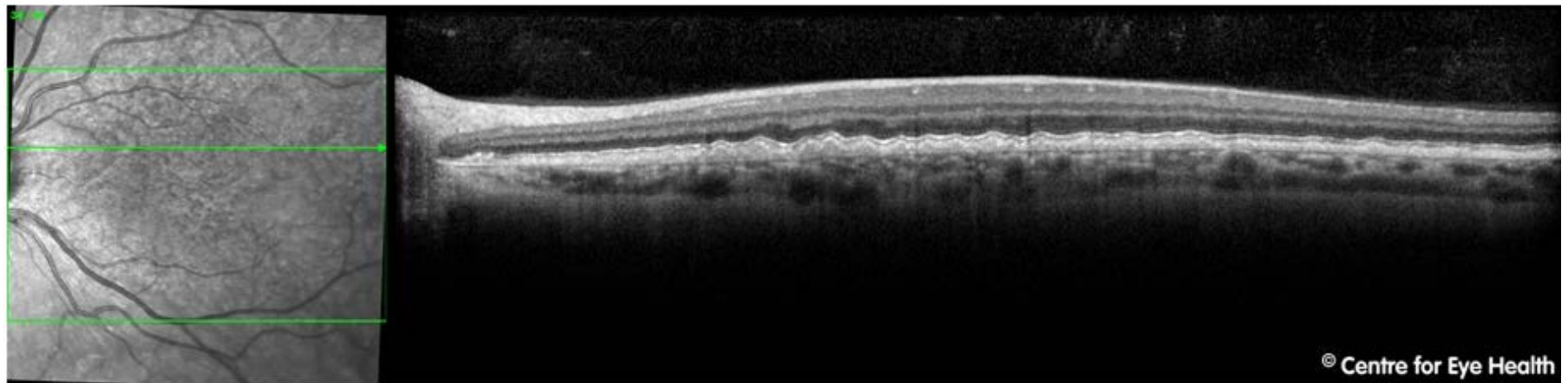
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Familial dominant drusen

Retinal photography and the optomap image show diffuse drusen throughout the posterior pole, nasal to the disc, affecting the macula. Fundus autofluorescence shows a pattern of speckled hyper and hypo-autofluorescence. The OCT line scans show drusenoid elevations of the RPE.

This clinical picture suggests the diagnosis of familial dominant drusen, also known as Malattia leventinese, Doyne honeycomb retinal dystrophy or autosomal dominant radial drusen. It is unclear if these names represent one disorder or a spectrum of disorders. Malattia leventinese has been specifically defined as familial dominant drusen when the drusen are centred on the fovea in a radiating pattern.

Familial dominant drusen typically present in the 2nd-3rd decade of life with patients remaining asymptomatic until the 4th decade of life when metamorphopsia and/or decreased vision can begin. The condition is typified by bilateral, relatively symmetrical drusen that are prominent on the temporal side, but also seen outside the arcades and nasal to the optic nerve. The peripheral retina normally remains free of drusen. Diagnosis is confirmed if the patient reports family members with a similar pattern of drusen.

As the condition progresses drusen become confluent and pigment hyperplasia develops. Subsequent geographic atrophy and choroidal neovascularization can cause significant vision loss. Patients should self-monitor their vision at home with an amsler grid and undertake regular review, including OCT imaging. If choroidal neovascularization develops, referral to an ophthalmologist for review is required.

A 2012 study using a small sample size found treatment with Argon green laser may have a beneficial effect on patients with familial dominant drusen, however further assessment and follow up is still needed.