Centre for Eye Health

CFEH facebook Cases

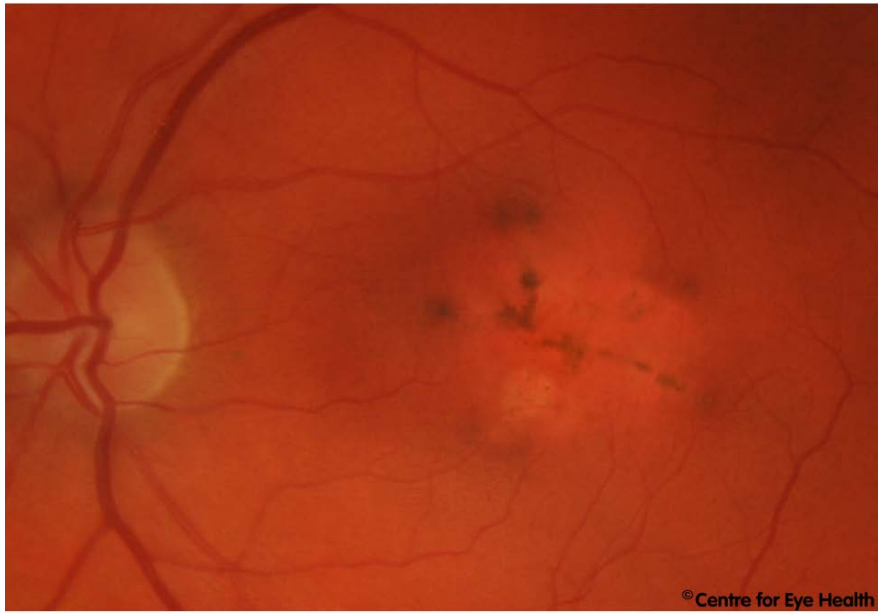
CFEH Facebook Case #5

A 50 year old Caucasian male was referred by his optometrist to CFEH for a macular assessment. He was seen elsewhere 6 years previous for new onset floaters and has no pertinent family ocular history. He currently takes Seroquel, Citalopram, Valium and Lipitor.

Best corrected acuities were 6/7.6 OD and 6/7.6-1 OS. Testing with an Amsler grid revealed distortion in both eyes.

Fundus photos, red-free photos, fundus autofluorescence and OCT images are below. As both eyes are similar, only the left eye is shown. What would be your diagnosis?

A quick reference to help differentially diagnosing this condition is the CFEH Chair-side reference "Non AMD Macular Conditions". A copy can be downloaded by clicking here. See our other Chairside references at http://www.centreforeyehealth.com.au/chairside-referrences



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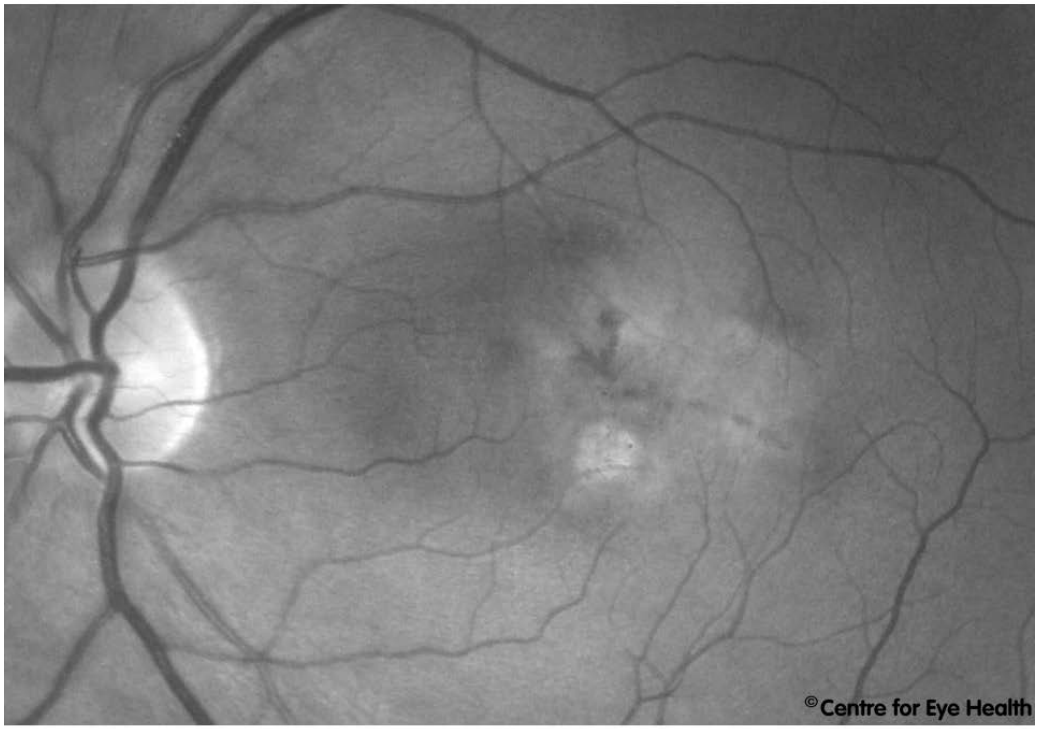

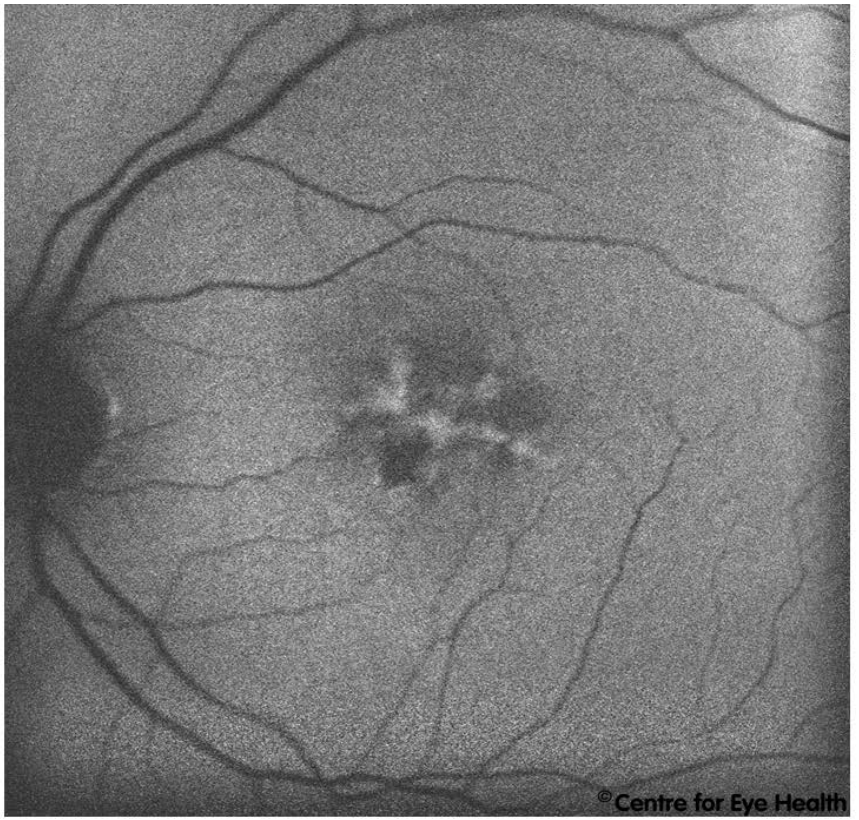

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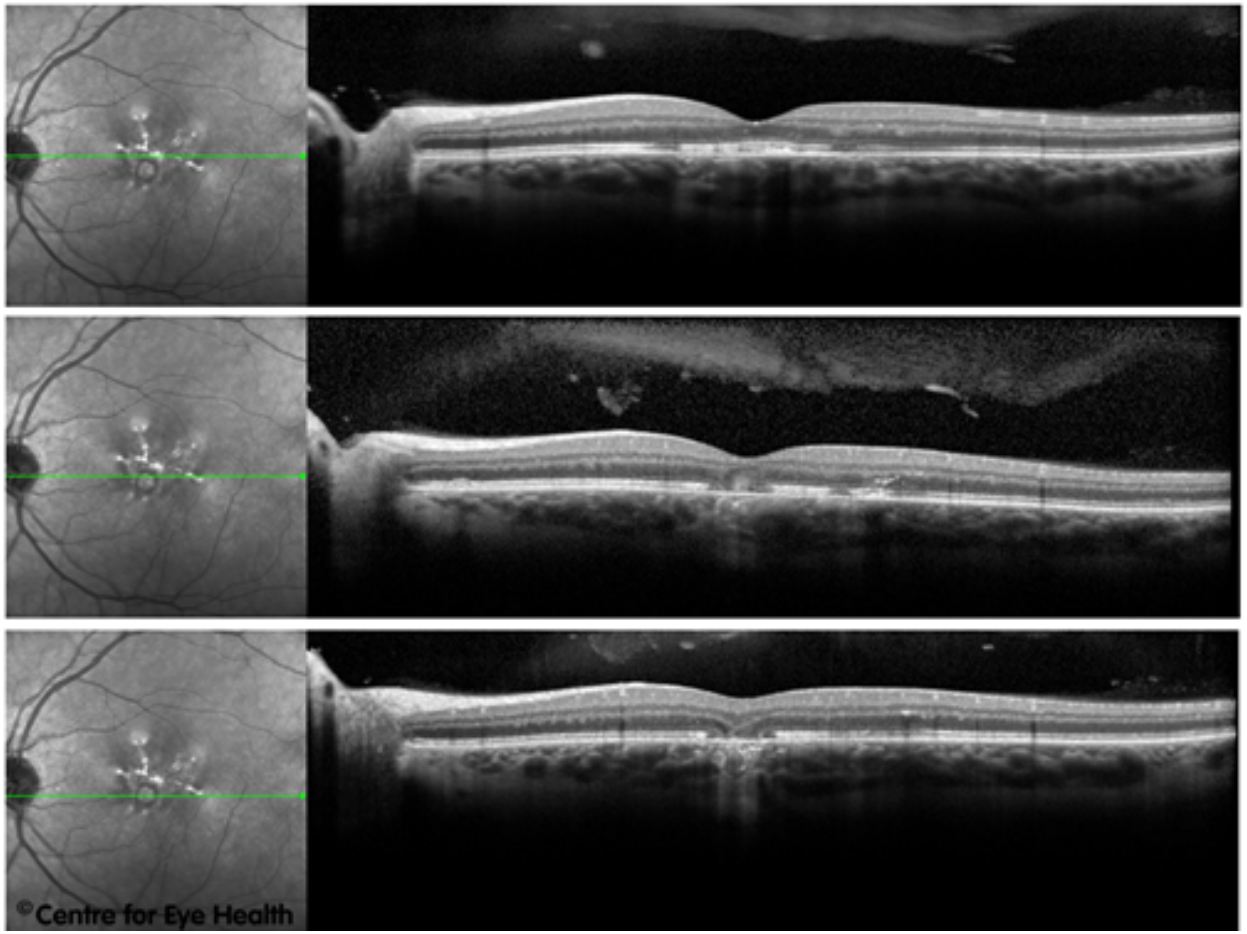

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ANSWER

Pattern dystrophy.

Funduscopy and retinal photography show both hyper and hypo pigmentary changes at both maculae. Fundus autofluorescence shows a pattern of hyper fluorescence at the macula. The OCT images show signs of RPE pigment migration (most notable in the middle image shown) and atrophy of the outer retinal layers, including the photoreceptors (most notable in the middle and lower images shown). The middle image also shows a hyper-reflective sub-retinal lesion that is typical of a pattern dystrophy.

Pattern dystrophy is characterised by the development of "patterns" of yellow, orange or grey pigment at the macula. It is typically a bilateral, autosomal dominant condition associated with mutations in the RDS/peripherin gene and has an age of onset ranging from 30-50 years. Presenting symptoms typically include a mild loss of best corrected visual acuity and metamorphopsia, both of which have been noted in this case, however some cases may remain asymptomatic.

There are 5 main "pattern" presentations possible and these are categorised according to the distribution of the pigment deposits:

1. **Adult-onset foveomacular vitelliform dystrophy – is characterised by yellow, round, bilateral subfoveal lesions that typically have a central pigmented spot and may be slightly raised.**

2. **Butterfly dystrophy (as in this case) – bilateral butterfly-shaped pigmentation at the level of the RPE. Usually there is an area of total RPE and photoreceptor layer loss.**

3. **Reticular dystrophy – appears as a reticular network of pigmented lines criss-crossing over the posterior pole with pigmented "knots" at the intersection of these lines.**

4. **Fundus pulverulentus – the rarest presentation, characterised by a coarse pigment mottling of the RPE in the macular area. This condition can be difficult to differentiate from other maculopathies.**

5. **Multifocal dystrophy – characterised by irregular yellow-white flecks scattered throughout the fundus, similar to those seen in fundus flavimaculatus. Associated macular lesions can range from yellow/grey deposits through to areas of severe chorioretinal atrophy. Multifocal dystrophy does not have the "dark choroid" seen in fundus flavimaculatus on fluorescein angiography.**

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ANSWER

The prognosis depends on which type of Pattern dystrophy is present and the age of onset of symptoms. This patient has butterfly dystrophy and can be expected to maintain relatively normal acuity for much of his life, however atrophic lesions can develop with age, which would adversely affect this acuity.

To download the CFEH Chair-side reference guides to help with your differential diagnosis of this and other macular condtions, please http://www.centreforeyehealth.com.au/chairside-referrences/

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