

CFEH Facebook Case #76

A 50 year old Caucasian female was referred to CFEH for examination with 6/6 visual acuity in each eye. She takes medication for systemic hypertension, but otherwise reports good general health. There was distortion on Amsler grid when tested with each eye. As both eyes had a similar appearance, only the left images are included. What macular condition does this patient have?



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The clinical findings are consistent with a diagnosis of fundus flavimaculatus.

The fleck-like yellow lesions surrounding both maculae spare the fovea and correspond to areas of mixed stipled hyper and hypoautofluorescence. OCT imaging shows the lesions to be causing disruption to the RPE and ISe line, with some focal areas of subretinal/ RPE deposits.

Fundus flaviamaculatus is part of the same disease spectrum as Stargardt's disease but has a later age of onset and slower progression. Both are related to a defect of the ABCA4 gene.

The flecks seen in fundus flavimaculatus can take many different shapes, including round, pisciform, butterfly or spear-like. They initially appear yellow-white in colour and are well defined, although over time they change in appearance becoming grey, fuzzy and ill-defined. The flecks may also be found at variable levels of the retina although they are typically at the level of the RPE (type 1) or the outer nuclear layer (type 2).