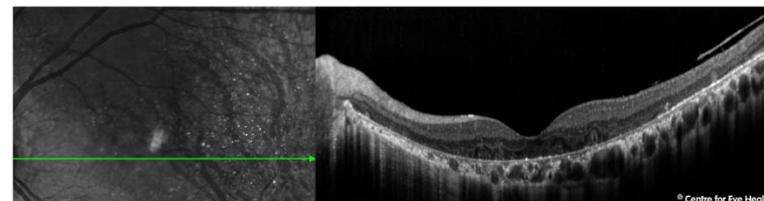
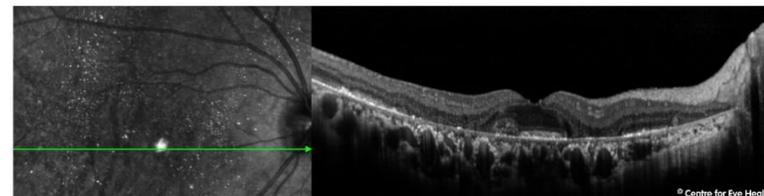
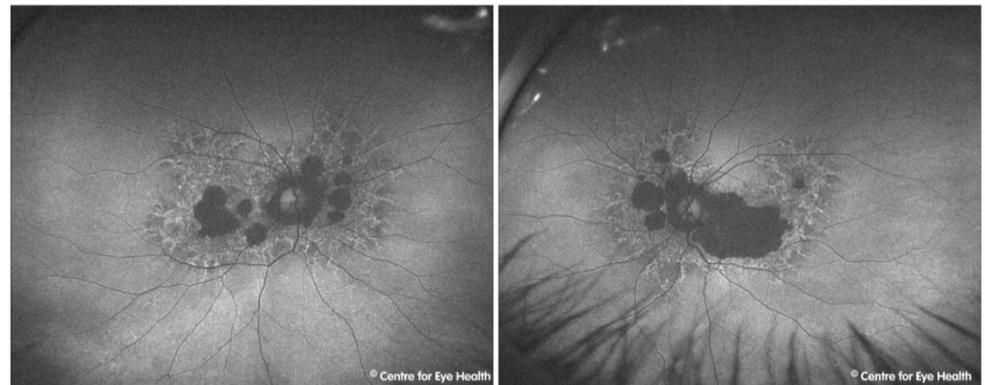
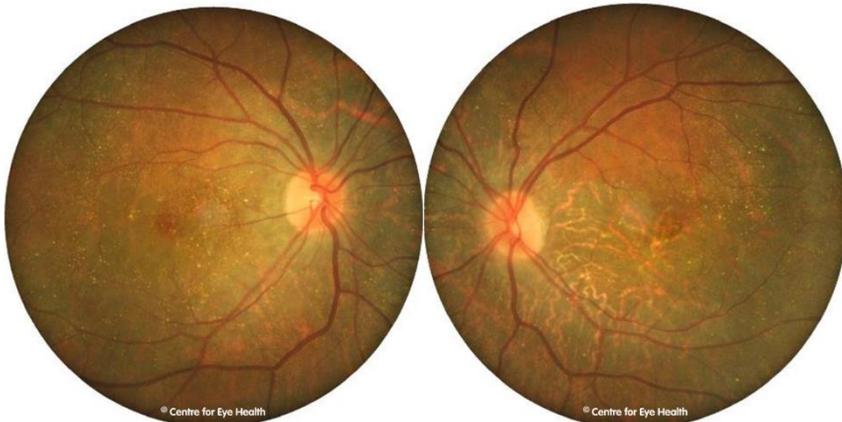




CFEH Facebook Case #46

A 58 year old Asian female was referred for investigation of numerous small white spots around the posterior pole in both eyes. Best corrected visual acuity was 6/7.5⁺ OD and 6/7.5⁻² OS. Testing with an Amsler grid showed distortion throughout the grid in both eyes, as well as areas where the gridlines were blurred. Contrast sensitivity was reduced at 1.48 units OD and 1.32 units OS (normal range 1.52–1.92 units). What is the most likely diagnosis for this patient?



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ANSWER

Bietti Crystalline Dystrophy.

Bietti crystalline dystrophy is characterised by the presence of numerous glistening yellow crystalline deposits distributed throughout the fundus and occasionally also present in the superficial cornea near the limbus. OCT investigations show these deposits may be located throughout all retinal layers but are most commonly found adjacent to the inner side of the RPE. The deposits are less apparent in areas of pigment epithelial atrophy.

A characteristic feature of Bietti crystalline dystrophy is the presence of circular hypo-reflective areas within the outer nuclear layer, termed tubular degeneration or tubular formation. These areas are typically found where there is damage to the RPE and rarely seen where the RPE is intact. In this case, the deposits are predominantly found in the outer retinal layers. There is loss, attenuation and disruption of the ISe zone, outer retinal layers and choriocapillaris and tubular degeneration can be identified (circled in red on OCT image below).

Fundus autofluorescence highlights the multiple areas of geographic atrophy in this patient. These areas appear densely hypo-autofluorescent with speckled hypo-autofluorescence surrounding them as well as radiating branches of hyper-autofluorescence.

Bietti crystalline dystrophy usually shows an autosomal recessive inheritance pattern and is most common in East Asian populations, particularly those from China and Japan. Presentation is usually in the 4th or 5th decade of life when symptoms start to develop. Presenting symptoms are typically a slow progressive vision loss and nyctalopia. As the condition progresses there is a diffuse atrophy of the choriocapillaris and decrease in size and number of the crystals. The atrophic areas progressively become confluent and extend into the peripheral retina with the end result being diffuse chorioretinal atrophy.

The rate of progression and severity of this condition is quite variable. This patient was referred to a retinal specialist.

