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Optometry
NEW SOUTH WALES
AUSTRALIAN CAPITAL TERRITORY

ANSWER

Familial Dominant Drusen (Also known as Doyne's honeycomb dystrophy or Malattia Levantine)

This condition was covered a couple of months ago in our Facebook case presentations (case 15). However, this is a vastly different and more advanced case.

Familial dominant drusen is typified by a bilateral, relatively symmetrical distribution of drusen, most prominent temporal to the macula, but also seen outside the vascular arcades and nasal to the optic nerve head. The peripheral retina normally remains free of drusen.

Historically, there were two conditions defined in the early literature as separate entities: Doyne's honeycomb dystrophy described a presentation similar to that seen in this patient, while Malattia Leventinese specifically referred to drusen radiating temporally from the fovea. Further studies later revealed that the two conditions both occur due to a single defect in the gene that codes for the protein fibrillin 3, and were in fact different expressions of the same disease.

As both this and case 15 illustrate, the phenotype is extremely variable. This patient has an advanced form of the disease with large, coalescent drusen, RPE irregularities and disruption of the outer retinal layers. Some patients with this condition may develop choroidal neovascularisation, however in this case there was no obvious evidence of current exudative changes.