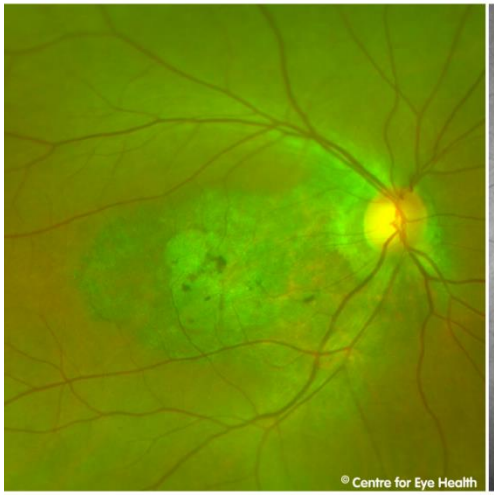
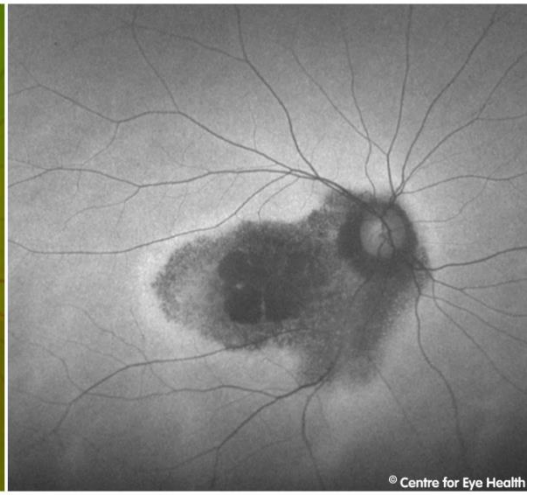
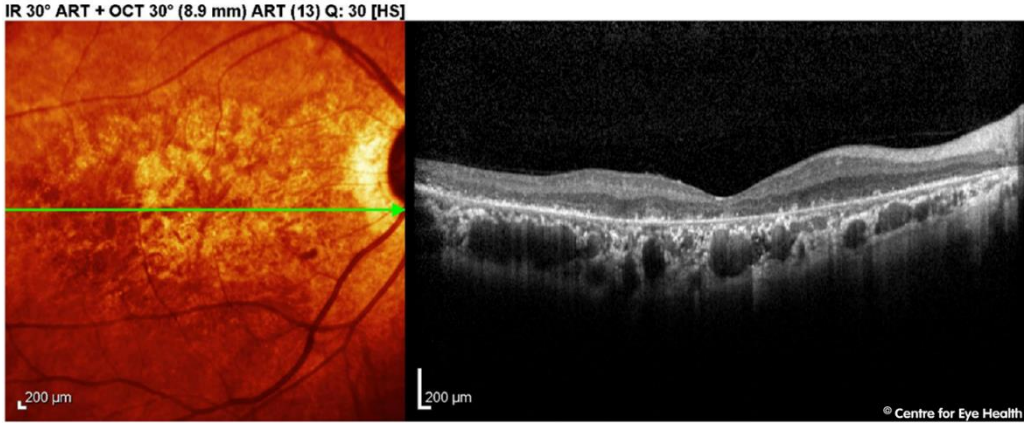
CFEH facebook Cases

CFEH Facebook Case #104

A 66 year old Asian female presented for a retinal assessment. Best corrected vision is less than 6/60 and the patient reported having had blurry vision since she was 16 years old. Both eyes have a similar appearance so only the right eye is shown below. What is the cause of this patient's vision loss?







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ANSWER

This is an advanced presentation of Stargardt Disease – a macular dystrophy caused by a mutation in the ABCA4 gene. It is characterised by progressive bilateral central vision loss, usually beginning in childhood or early adolescence.

Stargardt disease is characterised by abnormal levels of lipofuscin and loss of the outer retinal layers. A reduction in lipofuscin density and RPE atrophy (as seen in the OCT image) have caused the hypo-autofluoresence noted in this patient.

In earlier presentations when this condition is typically diagnosed, you would expect to see yellow-white retinal flecks which show a mixture of hyper and hypo-autofluorescence as in the images below.