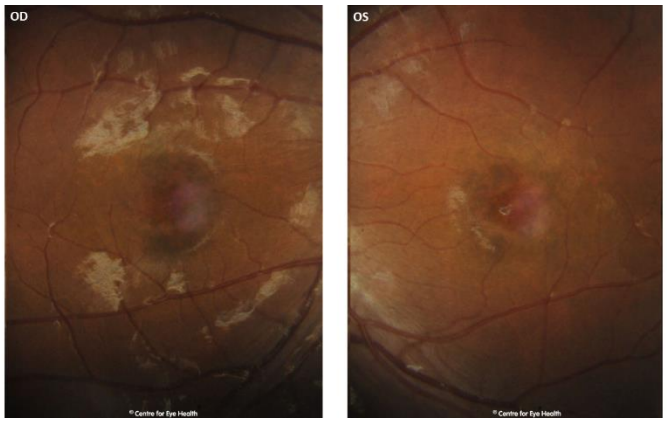
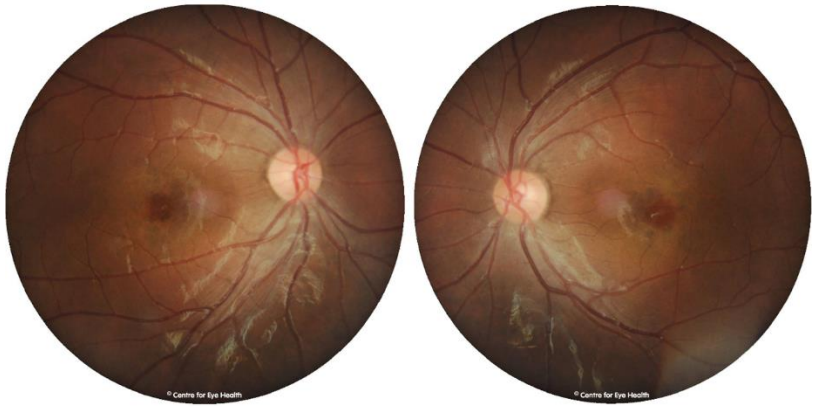
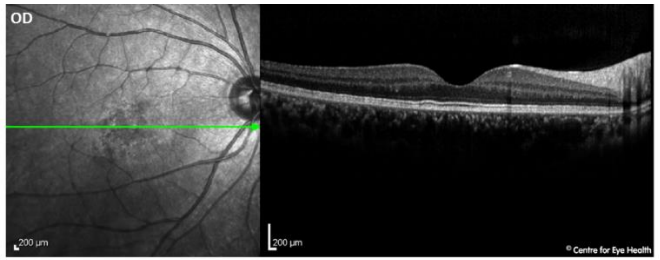
CFEH facebook Cases

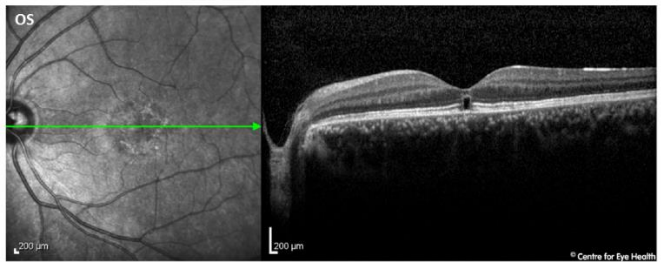
CFEH Facebook Case #45

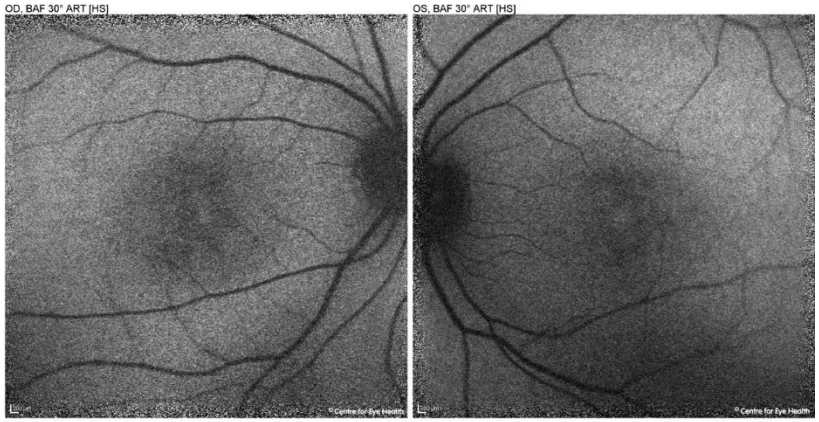
A 21 year old Asian female was referred for a macular assessment. She was diagnosed with type 1 diabetes at the age of 14, which is being managed by regular insulin injections. Around the same time she started suffering from tinnitus which progressed to a profound loss of hearing in high ranges. Her father had late onset diabetes but there was no other reported relevant family medical or ocular history. VA's were 6/6 OD and 6/7.5-2 OS. Imaging results are below. What is the rare condition that this patient is likely to have?











Proudly brought to you by

LEARNING FOR VISION

Centre for Eye Health

Optometry NEW SOUTH WALES AUSTRALIAN CAPITAL TERRITORY

Proudly brought to you by LEARNING FOR VISION

Centre for Eye Health | Optometry NEW SOUTH WALES AUSTRALIAN CAPITAL TERRITORY

ANSWER

Maternally Inherited Diabetes and Deafness (MIDD)

This condition is caused by a point mutation in the mitochondrial DNA and is thought to be responsible for between 0.5 and 2.8% of diabetes. It is characterised by maternally inherited diabetes (85% penetrance) and impaired hearing, usually in the high frequencies. The hearing loss is sensorineural and originating from the cochlear rather than the cranial nerve. MIDDS is often accompanied by a pattern macular dystrophy.

The macular dystrophy is characterised by both pigmented retinal lesions and atrophy of the RPE and/or choroid. The pigmented lesions range from appearing small and found at the macula through to radiating patterns surrounding the macula or optic nerve. Visual symptoms can include vision loss, photophobia, night vision problems and scotomas. The OCT images from this patient show some mottling of the RPE and a left micro-hole at the macula with some mottled hyper-autofluorescence, in particular at the fovea.

The age of onset of MIDD is typically between 25 and 35 years of age however it can range from 11 years to 68 years. Onset is usually insidious (such as in Type 2 diabetes), however about 20% of cases have an acute presentation with ketoacidosis occurring in around 8% of cases.

The mitochondrial mutation causing this condition is also responsible for other systemic manifestations, including MEALS (mitochondrial encephalomyopathy, lactic acid and stroke-like episodes); psychiatric disorders (depression, schizophrenia and phobias); myopathy of the proximal limb muscles; endocrine disease, renal disease and rapid onset heart failure in young adults. MIDD increases the risk of developing these conditions.

It was recommended this patient undergo electrophysiological testing to confirm a diagnosis of macular dystrophy. She was also referred to a retinal specialist for confirmation of a diagnosis.