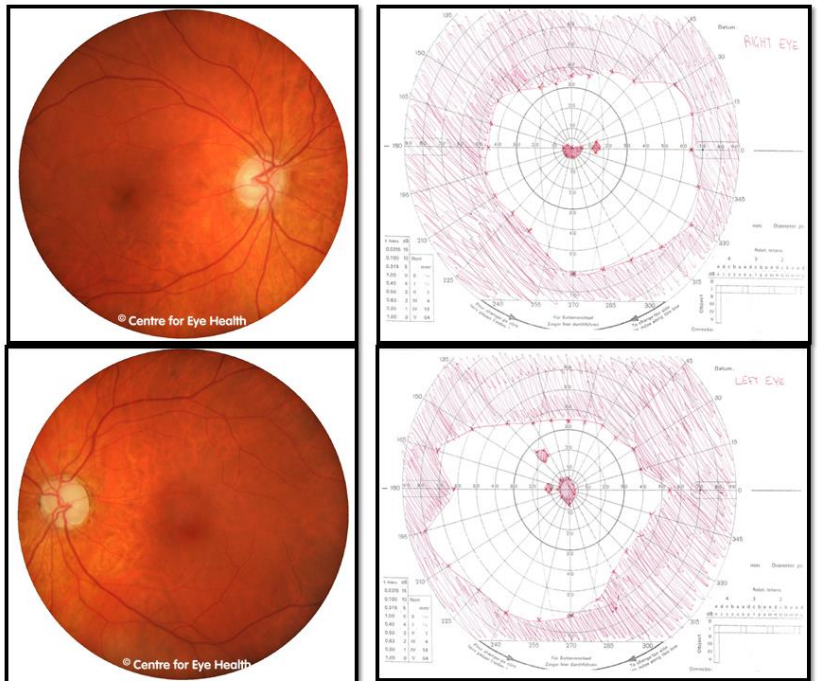
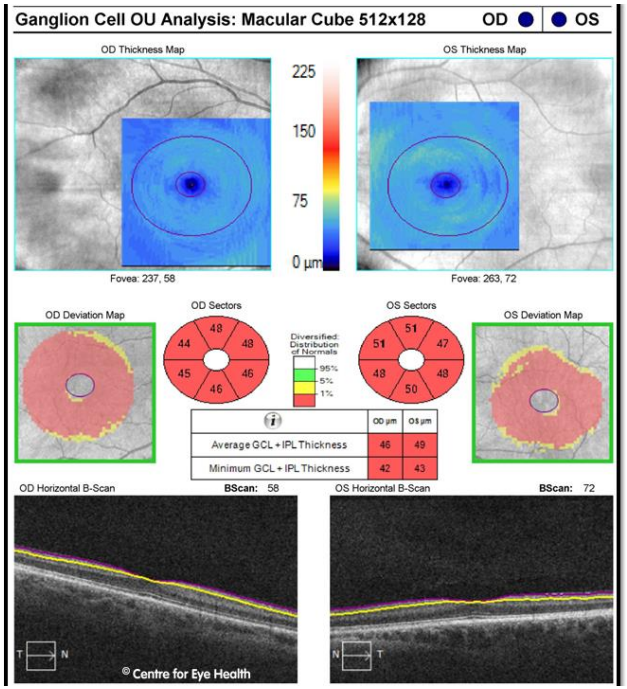
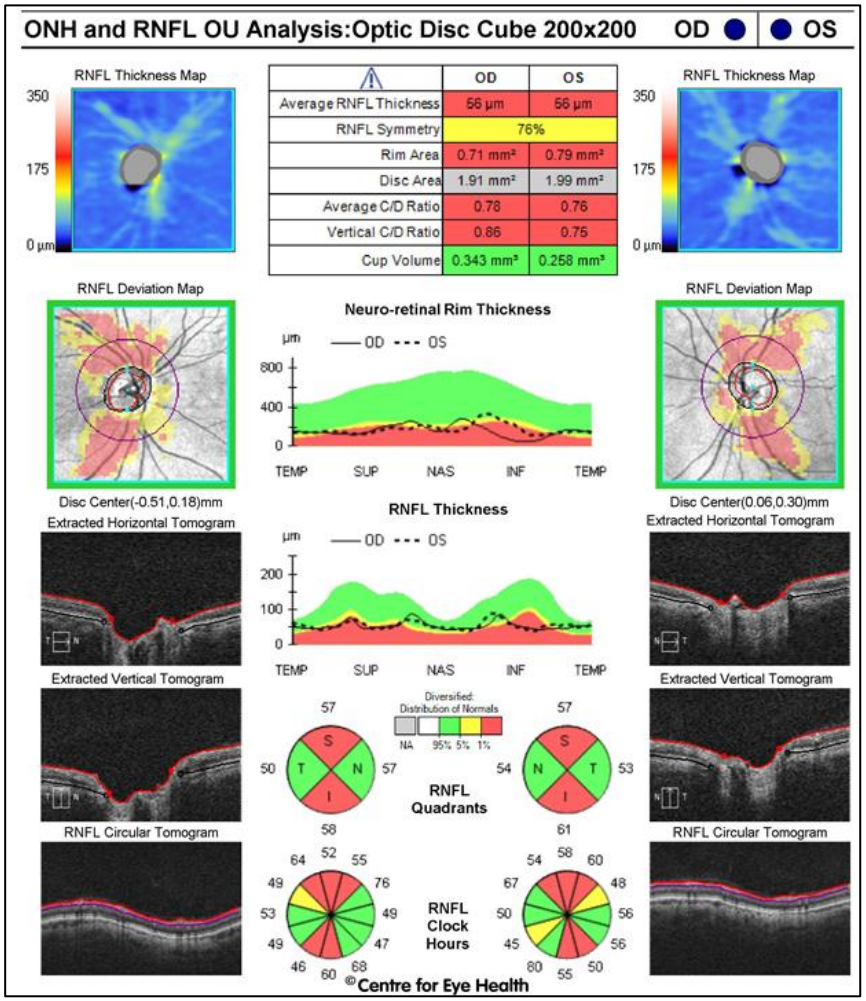
CFEH facebook Cases

CFEH Facebook Case #24

A 53 year old Caucasian male presented complaining of increased peripheral blur over the last couple of years. He has had poor central vision for many years. His father, brother and uncle all have all suffered progressive and severe vision loss. His general health is good, and the only medications taken are for hypertension. Entering acuity is counting fingers at 3 meters in both eyes, and this was not improved with a pinhole. Below are the retinal photos, fields (recorded with Goldmann kinetic perimeter), Cirrus RNFL and GC analysis. What condition does this patient suffer from?







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LEARNING FOR VISION

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NEW SOUTH WALES
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ANSWER

Leber's Hereditary Optic Neuropathy (LHON).

LHON usually affects men with onset between ages 10 and 30 years of age. There are often no symptoms until an acute, severe painless loss of vision is experienced with acuity less than 6/60 and central or cecocentral field loss. This loss of vision is usually permanent, however a small percentage (10-20%) do recover some vision.

Approximately a quarter of all cases are bilateral at onset, however unilateral cases will typically also start to show symptoms in the other eye 2-3 months after the first eye becomes symptomatic. The fundus can either appear entirely normal (20% cases), or can show pseudopapilloedema, peripapillary telangiectasia and tortuosity of retinal arterioles. Fundus findings may be subtle but can preceed vision loss. Atrophy of the optic discs can be seen within 6 weeks of the acute vision loss, a finding that is accompanied by a dense central scotoma.

Pathologically, there is damage to the retinal ganglion cells, axonal degeneration and demyelination then atrophy from the optic nerve back to the Lateral Geniculate Nucleus. The RPE and photoreceptor layers are spared.

LHON arises from a mutation in the mitochondrial DNA, affecting the genes that code for NADH Dehydrogenase protein which is required for normal mitochondrial phosphorylation. The mutation is inherited only from the mother.

Non-symptomatic carriers of the gene mutation have been found to show a temporal thickening of the retinal nerve fibre layer (Savini et al. 2005), and may also show a subtle impairment of optic nerve function, including reduced contrast sensitivity, altered colour vision and subnormal electrophysiology results (Sadun et al. 2006)

No treatment has been shown to be effective, although new medical and gene therapies remain under investigation. Genetic counselling and referral to low vision services are recommended.