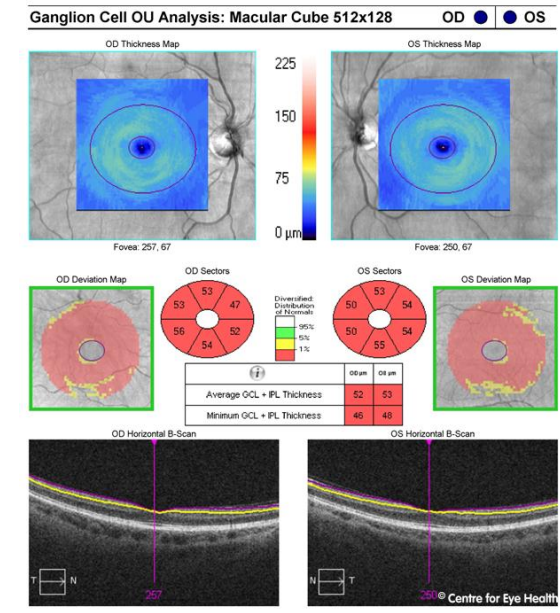
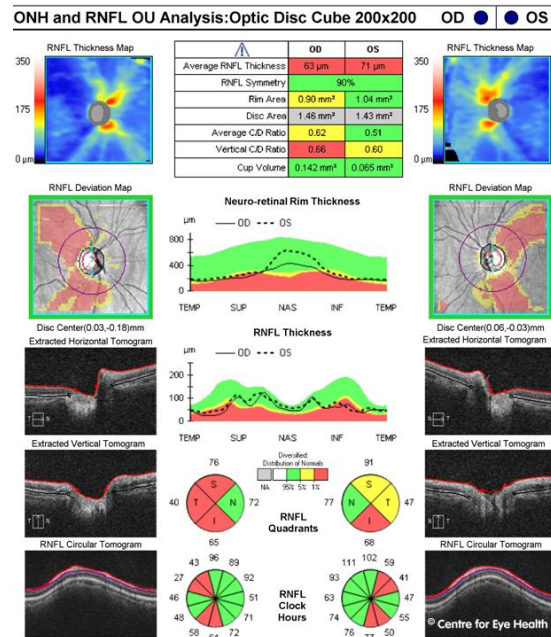
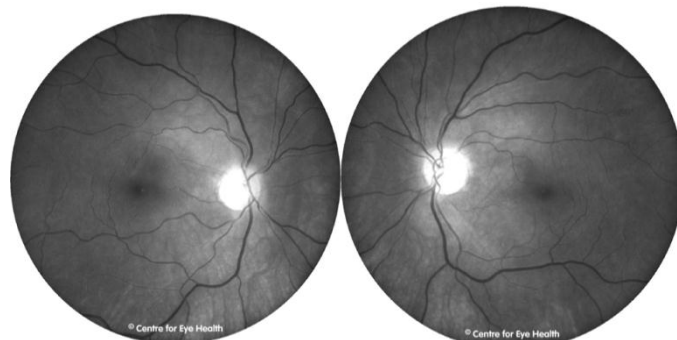
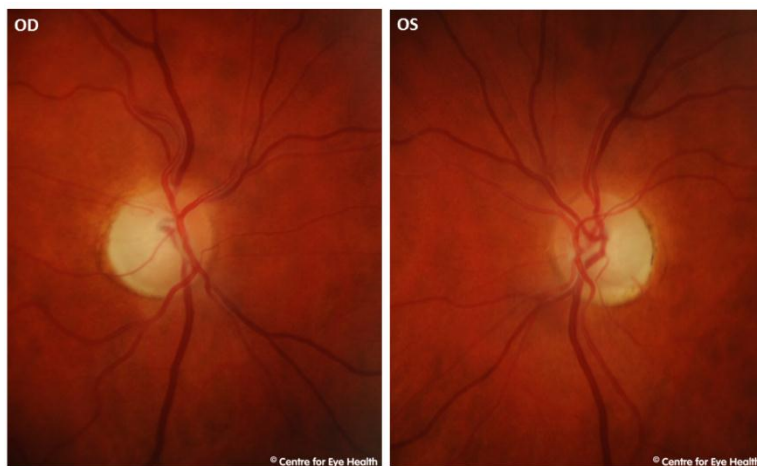


## CFEH Facebook Case #48

A 42 year old Caucasian female presented with a history of progressive vision loss in both eyes, in particular at night which she reported as having started in her teenage years. She is asthmatic and a smoker. Her father had 'a problem with his optic nerves'. Pinhole acuities were 6/19-2 OU, contrast sensitivity was reduced and colour vision testing showed a clear tritan defect in both eyes. Visual fields were unreliable despite repeated attempts. What is the most likely diagnosis for this patient?



Proudly brought to you by  
**LEARNING & VISION**

Proudly brought to you by

**LEARNING FOR VISION**



# ANSWER

Dominant optic atrophy (DOA), also known as dominantly inherited juvenile optic atrophy (DIJOA) or autosomal inherited optic atrophy.

Both discs show temporal pallor with shallow “saucerised” cupping of the inferior, superior and temporal neuro retinal rims. These findings are characteristic of optic atrophy. There is diffuse thinning of the RNFL as seen on red-free, more notable in the left eye. The supero- and infero-temporal quadrants show marked thinning compared with the normative database on Cirrus, and ganglion cell analysis shows marked thinning that is symmetrical between the two eyes.

DOA is an inherited bilateral optic neuropathy. Presentation is usually in the first or second decade of life, however it can present later in life (as in this case). Presenting signs and symptoms include a slow, progressive, insidious decrease of vision. The degree of loss can vary significantly both between and within families, with vision usually in the range of 6/9 – 6/30.

Inheritance is autosomal dominant with penetrance of 43-100%, depending on the family. The main gene responsible is OPA1, this being the affected gene in 75% of cases. 20% of cases of DOA are “syndromic”, meaning they have associated systemic signs, including neurosensory deafness, myopathy, peripheral neuropathy, stroke, multiple sclerosis or progressive external ophthalmoplegia.