CFEH Facebook Case #108

A 25 year old Caucasian female was referred due to an unusual optic nerve appearance. A disc photo, fundus auto fluorescence and OCT image of her left eye are below. Visual field testing showed an enlargement of the blind spot in the left field. Her vision was 6/6 in each eye and the right eye was unremarkable. What is the most likely diagnosis for this anomaly and what are the potential complications?
ANSWER

Congenital optic disc pit.

Congenital optic disc pits are hypo-pigmented oval or round excavations in the optic nerve head which can be bilateral in 10-15% of cases. Whilst typically seen temporally on the disc, they can be found at any location either centrally or at the edge of the disc. Acquired pits may be associated with high myopia or glaucoma. As neither of these are not present in this case, the pit is most likely congenital in nature.

Optic disc pits can be associated with macular oedema, schisis or neurosensory detachment of the macula – termed optic disc pit maculopathy. Maculopathy will typically occur in the third to fourth decade of life, reducing acuity to 6/21 or worse. While spontaneous regression can occur, most cases have a poor prognosis with a gradual worsening of vision. The literature varies in estimating the prevalence of maculopathy but it is reported to be in the range of 25-75% of patients with optic disc pits.