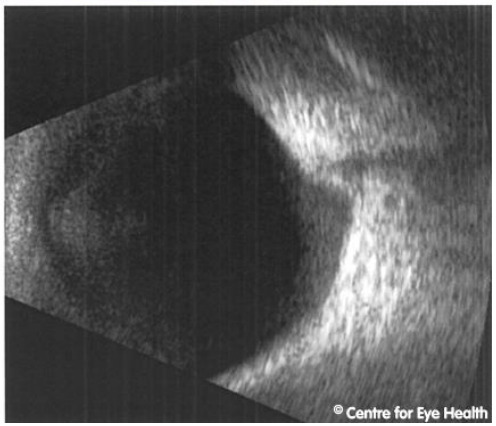
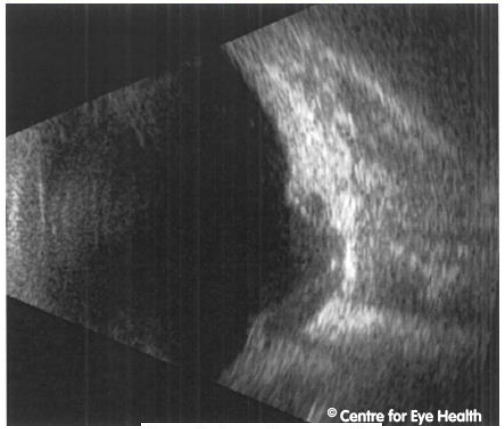
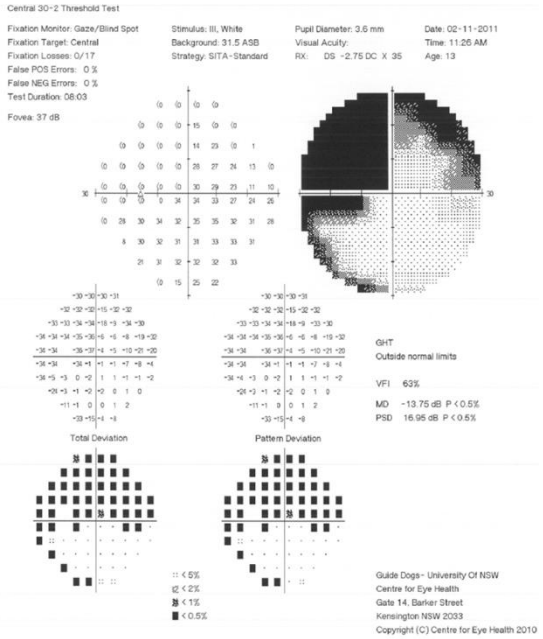
CFEH facebook Cases

CFEH Facebook Case #22

A 17 year old Indian female presented for examination. She had a right esotropia, best corrected vision of 6/15- in the left eye and light perception only in the right eye. Optomap images and B-scan ultrasound are included for both eyes. Visual fields were not possible on the right eye, but the results for the left eye are included. What is your diagnosis?







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ANSWER

A coloboma.

A typical coloboma forms as a result of the embryonic fissure failing to close completely during development. During normal development, the fusion of the embryonic fissure begins at around 33-40 days post conception. A coloboma occurs when there is a fault in this closure process, with the location of the anomaly determined by the location of the closure fault. The size of the coloboma can be variable depending on how much of the fissure fails to fuse. Typically colobomas are found inferior-nasally.

Colobomas can occur in the retina/choroid, the optic nerve and/or the iris and if a coloboma is found in one location, a complete examination should be performed as it may also be present in additional locations. Optic disc colobomas cause the disc to appear enlarged and excavated, often more so in the vertical meridian. Location is usually at the inferior aspect of the disc so the inferior neuro-retinal rim is typically either thin or absent, and normal disc tissue is confined to a small superior wedge. Visual acuity is often decreased and there is an associated superior scotoma.

In this case, the patient has a large coloboma in both eyes. The optic disc is completely involved in the right eye and partially in the left. The excavation of both can be appreciated by looking at the B-scan ultrasound. There was an associated iris coloboma inferiorly in the right eye. The left eye still had some macula function as the edge of the coloboma is adjacent to the macula rather than encompassing it as it does in the right eye (fig 2 below).

Colobomas may show either autosomal dominant or autosomal recessive inheritance or may be an isolated anomaly. Another cause may be teratogens – agents that disturb the normal development of an embryo or foetus. These include exposure to thalidomide in the first trimester and possibly LSD ingestion, although the evidence for this is not conclusive. Various other genetic disorders also have coloboma as a rare manifestation, however the correlation is, again, lacking in conclusive evidence.

For more information on this condition and other neuro retinal rim anomalies, a video lecture on this topic is available through Learning for Vision – click here