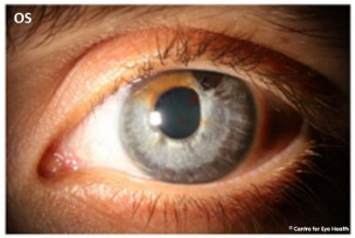
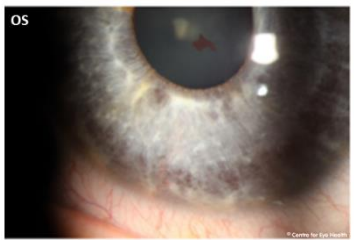
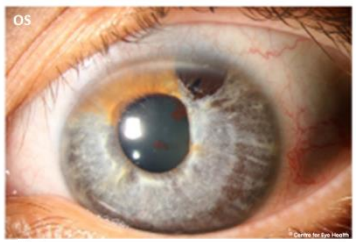
CFEH facebook Cases

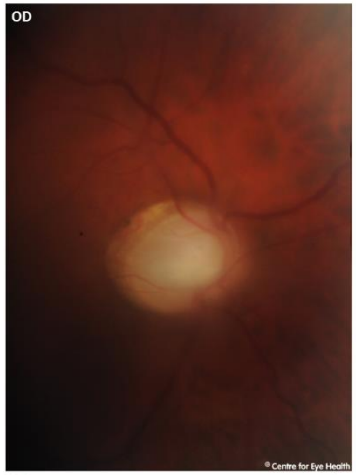
CFEH Facebook Case #44

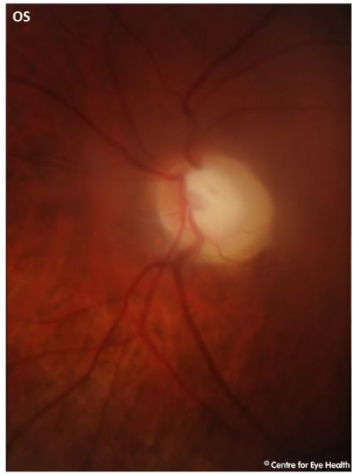
A 26 year old Caucasian male presented for review at CFEH. He was diagnosed in Russia with glaucoma at a few months of age after his older brother was also diagnosed. His father developed glaucoma at 52 years of age. He has had a total of 4 surgeries per eye and is currently on timolol 0.5% b.i.d OU. He reported good general health. His IOP's were 14mmHg and 15mmHg in the right and left eyes respectively with pachymetry readings of 612µm and 608µm. Gonioscopy showed open angles with an atypical, featureless appearance of the angle structures. Peripheral anterior synechiae were noted in the superior angle of both eyes. Imaging results are below. Note that the anterior eye photos were taken when the patient was partially dilated. What is the likely syndrome this patient has?

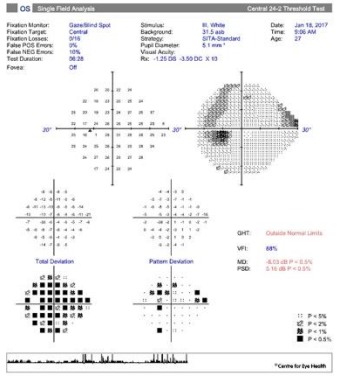


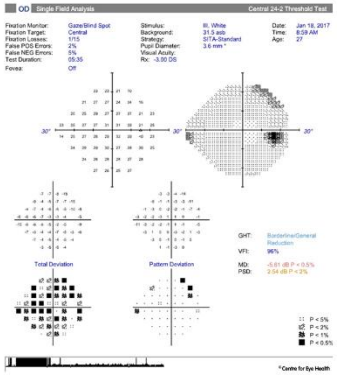


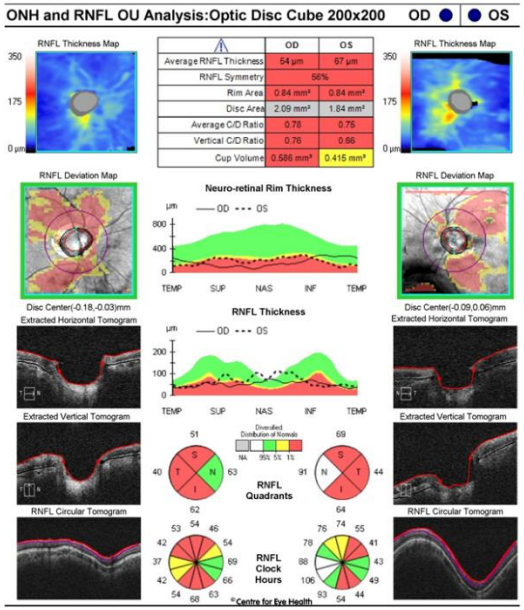












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ANSWER

Axenfeld-Reiger Syndrome.

This syndrome is primarily an eye condition but may have systemic manifestations. It affects 1 in 200 000 people and has an autosomal dominant inheritance pattern.

Ocular manifestations can include iris atrophy, corectopia (displacement of the pupil), polycoria, corneal abnormalities and hypertelorism (an abnormally increased distance between the eyes). This patient has a large corneal diameter, pupil miosis, superior-nasal displacement of both pupils (more prominent before dilation), left pseudopolycoria, posterior embryotoxin and a moth-eaten appearance to the irides. This anterior segment dysgenesis is consistent with Axenfeld-Reiger syndrome. The synechiae noted on gonioscopy are likely to have resulted from prior surgeries. Optic nerve head evaluation showed thinning of the neuro-retinal rim and enlargement of the cup, while Cirrus OCT shows a marked thinning of the RNFL that is characteristic of glaucoma.

Additional systemic manifestations can include:

- A flattened midface with broad flat nasal bridge and prominent forehead
- Microdontia (one or more teeth are smaller than normal)
- Oligodontia (congenital absence of some teeth)
- Redundant periumbilical skin (extra folds of skin around the belly button)
- Heart defect
- Hypospadias
- Anal stenosis
- Pituitary gland abnormalities which can slow growth