Axenfeld-Rieger Syndrome with Secondary Glaucoma

Case History
-68 year old Caucasian male
-No ocular/visual complaints. Presented for follow up appointment for intraocular pressure check.

- Ocular history:
  1. Axenfeld-Rieger Syndrome OU
  2. Secondary glaucoma OU
  3. History of angle closure
  4. Optic disc pallor OS
  5. History of macular edema OS secondary to ACIOL
  6. Pseudophakia OU

- Systemic history:
  1. Nicotine and alcohol dependence
  2. Macrocytosis
  3. General health deterioration
  4. Coronary arteriosclerosis
  5. Chronic obstructive lung disease
  6. Barrett’s esophagus
  7. Essential hypertension
  8. Hypothyroidism

- Ocular meds:
  1. Latanoprost QHS OU
  2. Cosopt BID OU
  3. Brimonidine BID OU
  4. Pilocarpine QID OD only

Pertinent findings
Visual acuities with spectacle correction were 20/50 in the right eye and count fingers at 2 feet in the left.
Confrontation fields showed superior constriction in the right eye and patient was unable in the left. Pupils had no reaction to light in both eyes with anisocoria and correctopia OU. Slit lamp examination revealed an intact flat plate graft superiorly in the right eye covering an Ahmed valve at 12 o’clock in the left. An anterior chamber intraocular lens was present in both eyes. The iris of each eye showed atrophy inferiorly, and patent peripheral iridotomies superior temporally. Intraocular pressures via Goldmann applanation tonometry were 20 mmHg in both eyes. No fundus exam was performed at this visit.

Differential diagnosis
- ICE (iridocorneal endothelial) syndrome
- Peter’s anomaly
- Congenital ectropion uveae
- Aniridia
- Ectopia lentis et pupilae

Diagnosis
Axenfeld-Rieger Syndrome is diagnosed based on clinical features, including iris and pupillary abnormalities in the presence of an anteriorly displaced schwalbe’s line. Patients may also have systemic irregularities, such as dental abnormalities, craniofacial dysmorphism, and redundant umbilical skin.

Discussion
Axenfeld-Rieger Syndrome is a rare condition with a genetic component, likely autosomal dominant, with ocular manifestations and possible systemic associations. The underlying pathophysiology of the condition is related to incomplete development of neural crest cells in multiple areas of the body, including the anterior chamber, teeth, and bones of the face. Incomplete development of neural crest cells in the trabecular meshwork and/or schlemm’s canal leads to an inadequate drainage system for the aqueous, resulting in glaucoma. Patients are usually diagnosed
with Axenfeld-Rieger syndrome in childhood, but glaucoma may not present until adulthood. Patients usually present with a classic anteriorly displaced Schwalbe’s line, known as posterior embryotoxin, along with correctopia and iris atrophy. Posterior embryotoxin may be detected on slit lamp examination, or it may only visible during gonioscopy. Despite anterior segment anomalies, most patients maintain adequate vision, unless vision is lost due to glaucoma. Unfortunately, glaucoma in patients with Axenfeld-Rieger Syndrome can be debilitating and difficult to control.

Treatment/management
Typically, the only aspect of Axenfeld-Rieger syndrome that requires treatment is secondary glaucoma. This type of glaucoma can be very debilitating and difficult to manage (1). Initial treatment includes topical glaucoma medications. Aqueous suppressants may be more effective than miotics in these patients. Unfortunately, topical therapy alone often fails due to the incomplete development of the trabecular meshwork. Thus, surgical interventions that provide an alternate aqueous drainage route, such as such as trabeculectomy or the implantation of aqueous shunts, must be considered (2). In general, trabeculectomies tend to be less successful in patients with congenital glaucoma due to angle malformations (3). According to one study, there may be no difference in surgical outcome between Ahmed valve implantation and a repeat trabeculectomy in patients with congenital glaucoma and a prior failed trabeculectomy with Mitomycin C (4). However, Patients that undergo a repeat trabeculectomy may be at higher risk for post-surgical complications, such as endophthalmitis (4).

References:


Conclusion
Axenfeld-Rieger Syndrome is a rare bilateral condition, usually diagnosed in childhood, which may or may not present with systemic associations. Glaucoma occurs in approximately half of these patients, but it may not present until adulthood. Therefore, all patients with Axenfeld-Rieger Syndrome should be followed closely regardless of a diagnosis of glaucoma. In patients that develop secondary glaucoma, maximum topical therapy may not be efficacious, so surgical interventions should be considered.