Axenfeld-Rieger Syndrome

- rare spectrum of developmental disorders
- abnormalities of both ocular and extraocular structures derived from the neural crest
- bilateral
- secondary glaucoma because of arrested angle development in about 50%
- rare, AD inherited disorder
- developmental arrest occurs late in gestation results in primordial endothelium being retained over parts of the iris and anterior chamber angle
- Contraction of this primordial monolayer causes iris stromal thinning, corectopia, and hole formation.
- With contraction, the anterior uvea is hindered from posterior migration, which results in a high insertion of the iris into the anterior chamber angle
- A significant association with chromosome 4 has been shown, but no sex predilection has been found
- The anterior segment structures affected are primarily of neural-crest derivation.
- The extraocular defects most frequently seen involve the teeth and facial bones
- anteriorly displaced Schwalbe's line (posterior embryotoxin), which appears as a white ring on the posterior cornea near the limbus.
- It tends to be more common temporally if it is not seen over 360°.
- The anteriorly displaced Schwalbe's line occurs in 8% to 15% of the general population and may not always be present with A-R syndrome.
- gonioscopy, exhibits posterior embryotoxin and iridocorneal adhesions that are broad to threadlike in nature.
- These iridocorneal adhesions may extend anteriorly to Schwalbe's line and obscure the scleral spur and trabecular meshwork.
- Iris defects that range from stromal thinning to actual hole formations, corectopia, and ectropion uveae, may occur.
- Microdontia (peg-like incisors), hypodontia (decreased number of evenly spaced teeth), and anodontia (focal absence of teeth) are noted most commonly
- maxillary hypoplasia and a lower lid that protrudes.
- Telecanthus and hypertelorism
- primary empty-sella syndrome has been documented
- PATHOLOGY → peripheral cornea characteristically exhibits an anteriorly displaced Schwalbe's line. This posterior embryotoxon shows a cellular monolayer with basement membrane that covers dense collagen. The iridocorneal strands tend to be iris stroma mixed with the above-mentioned cellular monolayer. This cellular membrane also may extend over the iris surface, which distorts the iris and creates iris stromal thinning, and results in actual hole formation and corectopia as it contracts.
- Medical therapy is recommended prior to the initiation of surgical intervention.
- Medications that decrease aqueous output (b-blockers, a-agonists, and carbonic anhydrase inhibitors) have proved more beneficial than those that affecting outflow (pilocarpine).
- Glaucoma most often occurs in children and young adults
• Surgical intervention may be goniotomy, or trabeculectomy.
• The procedure of choice in A-R syndrome is trabeculectomy with the adjunctive use of antimetabolites.
• If the initial surgical treatment fails, seton procedures may have to be utilized.