Unusual presentation in Axenfeld-Rieger syndrome

Introduction:

A-R syndrome (Axenfeld-Rieger syndrome) is considered to be autosomal dominant in inheritance, 60% of cases have been linked to mutations in the transcription factors PITX2(4q25), FOXc1(6p25) and unidentified genes on 13q14, 16q24 and chromosome 11.

Usual ocular findings involve the cornea (megalocornea and prominent Schwalbe's line), iris (mild atrophy to severe degenerative changes like hole formation, correctopia and ectropion uvea) and angle (anterior insertion of iris, tissue strands bridging the angle from the peripheral iris to the prominent Schwalbe's line). Glaucoma appears in nearly 50% of the subjects with A-R syndrome. Schwalbe's line in A-R syndrome usually appears on the slit-lamp as a prominent and anteriorly displaced white line in the posterior cornea near the limbus. It may be incomplete (usually temporal) or complete (360 degrees).

Recently Espana et al reported unusual bilateral detached Schwalbe's line in the anterior chamber in a patient of A-R syndrome.[3] We report another unusual case of detached Schwalbe's line with attached iris tissue strands in the anterior chamber in A-R syndrome.

Report:

A fourteen-year old child presented with gradual painless decrease in vision in both eyes since one year and redness in the left eye since the last three days. He was known case of glaucoma for 6 months and was using Timolol 0.5% and Pilocarpine 2% eye drops in both eyes.

His best-corrected visual acuity was 20/50 in the right eye and 20/40 in the left eye. On examination, the corneas were enlarged in size, the horizontal diameter measuring 14 mm

in the right eye and 13 mm and in the left eye. Haab's striae and posterior embryotoxon were seen in both eyes. (Figure 1) The left eye also showed blepharitis and marginal keratitis. The most striking feature in the slit-lamp examination was the presence of a white cord-like structure traversing the anterior chamber in both eyes (more prominent in the left eye), with adhesions to the iris tissue along its course. (Figure 2 and Figure 3) IOP (Intraocular pressures) were 22 mm in the right eye and 18 mm in the left eye by applanation tonometry. Gonioscopy in both eyes revealed a prominent Schwalbe's line along a part of the angle circumference, with the cord-like structure taking its origin at the level of Schwalbe's line. (Figure 4) The angles were open in both eyes but showed anterior insertion of the iris into the trabecular meshwork with prominent iris processes and broad-based synechiae at places. (Figure 5) Fundus examination revealed small discs with 0.9 cupping, inferior notch and superior rim thinning in the right eye; left eye showed 0.8 cupping with superior notch. Based on the above findings, our diagnosis was Axenfeld-Rieger syndrome with secondary developmental glaucoma. The cord-like structure probably represented the detached Schwalbe's line. Humphrey visual fields revealed superior and inferior arcuate defects in the right eye and inferior arcuate defect in the left eye. The patient was advised Timolol 0.5% in both eyes and Latanoprost 0.005% in the right eye. He was also given the usual treatment for marginal keratitis, to which he responded well. The IOP, however, were not adequately controlled with medications alone in the right eye and we performed a trabeculectomy in the right eye. Post-operatively, the child had satisfactory IOP control. At 6-month post-operatively his IOP in right eye was 10 mm Hg and 14 mm Hg in left eye. His left eye IOP is controlled on medication alone.

DISCUSSION

Espana et al recently reported a somewhat similar clinical picture in a 37-year old man with bilateral detached Schwalbe's line in the anterior chamber in a patient of Axenfeld-Rieger syndrome. Our patient had iris tissue strands attached to this structure and gonioscopic findings clearly showing its origin at the level of Schwalbe's line. To our knowledge, this is the first ever case report describing detached Schwalbe's line with

attached to the iris tissue strands in the anterior chamber in a case of Axenfeld-Rieger syndrome.

Impaired neural crest cell migration and differentiation during embryonic development are considered important in the pathogenesis of Axenfeld-Rieger syndrome. Different anterior segment structures like the corneal stroma, Schwalbe's line, iris stroma and trabecular meshwork have their genesis from these neural crest cells. The common origin of Schwalbe's line and iris stroma from the neural crest cells could possibly explain the presence of the detached Schwalbe's line with attached iris tissue strands in the anterior chamber.

The hallmark of this case was the presence in the anterior chamber of this strange cord-like structure, the nature of which was not immediately apparent. This peculiar picture of a worm- or thread-like structure in the anterior chamber can be worth remembering as rare clinical sign in A-R syndrome while examining patients with developmental glaucoma, particularly in paucity of tell-tale clinical signs.