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Case Report

Bilaterally displaced Schwalbe's line: An unusual presentation of Axenfeld Rieger syndrome

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ABSTRACT

Purpose: To report a case on unusual presentation of Axenfeld Reiger (A-R) syndrome.
Observation: Axenfeld Rieger syndrome commonly presents with prominent Schwalbe's line near the limbus. Presentation with displaced Schwalbe's line is rarely reported. We report 2 cases who presented with bilaterally displaced Schwalbe ring as strange cord like structure in anterior chamber with iris tissue adhesions.
Conclusion: Axenfeld – Reiger syndrome is an autosomal dominant disease with ocular findings involving cornea (megalocornea), iris (iris atrophy,correctopia) and angle (anterior insertion of iris, prominent Schwalbe line). We report unsual case of detached Schwalbe's line with iris tissue adhesions in anterior chamber A-R syndrome.

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1. Introduction

Schwalbe's line is prominently placed near the limbus in the posterior cornea and usually visible on gonioscopy. This ring is often associated with congenital anomalies, such as corectopia, polycoria and in Axenfeld- Rieger (A –R) syndrome.[1,2] Axenfeld Reiger syndromne is an Autosomal Dominant disorder with mutation in PITX2 and FOXC1 genes which encode for different transcription factors. The disease spectrum involves Axenfeld anomaly – if the changes rae confined to the angle only, Rieger anomaly if there are iris as well as angle anomaly and Axenfeld Reiger syndrome whne there are systemic abnormalities also present along with the ocular abnormalities.

ARS is a multisystem abnormality. Systemic abnormalities mainly involve the maxilla, dental (hypodontia, microdontia, diastemia), umblical (umblical hernia), cardiac, Ocular findings mainly include cornea (megalocornea), iris (iris atrophy and correctopia), angle (anterior insertion of iris and prominent schwalbe's line). Burian et al. has described varied locations and presentations of the line on gonioscopic and external examinations.[3]

We describe atypical presentation of A –R syndrome in two pediatric cases who presented with complete displacement of Schwalbe's line in the anterior chamber with iris adhesions.

2. Case Report

2.1. Case 1

A 11year-old male child presented to our department with gradual painless decrease of vision in both eyes. He was found to have hyperopia (+4.0 D both eyes) with best corrected visual acuity of 20/40 in right eye and

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20/30 in left. Stereopsis was 400 second of arc. The past medical and family ocular history were unremarkable. On external examination, he had telecanthus and 30 PD esotropia on primary gaze. Slit lamp biomicroscopy showed normal corneal diameters (11mm) with cornea clear and cord like structures traversing anterior chamber in both eyes (Figure 1). These structures had iris strands attached and were more prominent post dilatation. Central corneal thickness was 520mm in right and 510mm in the left. Intraocular pressure (IOP) was 20mm Hg in right eye and 14 mm Hg in left eye by applanation tonometry. Fundus examination revealed 0.4 cup disc ratio in both eyes.

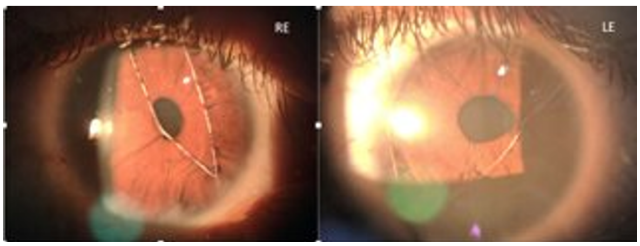

Fig. 1: Slit lamp image of anterior segment of both eyes showing displaced Schwalbe's line with attached iris strands

The patient had normal hearing and intelligence as per his age group, with normal development. The child was prescribed proper refractive correction and was advised 3 monthly regular follow up for detection of glaucoma.

2.2. Case 2

A 15 year- old male was referred to our pediatric ophthalmology department for abnormal structures visualized on slit lamp. There was no history of any ocular disorder or family illness. His best corrected visual acuity was 20/50 in both eyes with refractive correction of +7.0 in right eye and +6.5 -1.00 at 160 in the left eye. On slit lamp examination, corneal diameters were normal (12.5mm both eyes) and white cord like structures with iris adhesions were noticed in the anterior chamber of both eyes (Figure 2). Central corneal thickness and pressure in right eye was 500mm, 20 mm Hg respectively and 510 mm and 24 mm Hg in the left. Examination showed 0.5 cup in both eyes.

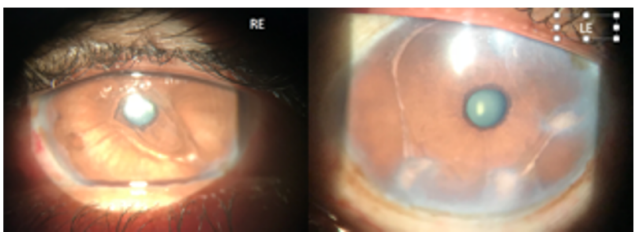

Fig. 2: Slit lamp biomicroscopy revealing Schwalbe's line in the anterior chamber

On gonioscopy, angles were open with prominent Schwalbe's line along the angle circumference, with cord-like structure taking its origin at the level of Schwalbe's line.

The child was prescribed glasses and kept on regular follow up for development of glaucoma.

3. Discussion

Axenfeld Rieger syndrome is an autosomal dominant disorder caused by defection migration or differentiation of neural crest cells during embryonic developement.[2] Sixty percent cases have mutations in PITX2(4q25) and FOXc1 (6p25) genes that code for transcription factors.[1] In general, mutation in PITX2 gene is more frequently seen on patients with ocular and systemic anomalies, whereas mutation in FOXC1 is found in patients with only ocular findings. There are only a few reports related to presentation of A-R syndrome with displaced Schwalbe's line in the anterior chamber. Espana and colleagues described bilateral displaced Schwalbe's line in a 37- year old who presented with megalocornea and glaucoma.[4] Parikh et al. reported 17- year old with megalocornea, iris tissue adhesions to bilaterally displaced Schwalbe's line. They hypothesised that common origin of Schwalbe's line and iris stroma from the neural crest cells could explain the iris strands attachment to detached Schwalbe's line.

Our series reports 2 patients with sporadic A-R syndrome, who presented with normal corneal diameters, and bilaterally displaced Schwalbe's line in the anterior chamber with attached iris tissue. Though none of the patients have developed glaucoma till followed up, close follow up is important every 3 monthly for timely diagnosis and management of glaucoma.

4. Conclusion

Axenfeld-Reiger syndrome is a multisystem anomaly which requires multidisciplinary approach to treatment. The patient needs to be followed up for development of glaucoma.

5. Source of Funding

None.

6. Conflict of Interest

None.

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