Case Report

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A Case Report of Axenfeld-Rieger Anomaly

Ophthalmology Section

PRACHI SINGH[1], RIKA SINGH[2], SUPREET BALLUR[3]

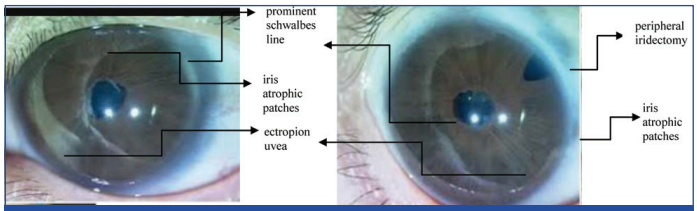
ABSTRACT

Axenfeld-Rieger (AR) syndrome is an inherited Autosomal Dominant (AD) disorder with ocular, dental, and craniofacial defects, whereas AR anomaly presents only with ocular changes. Hereby, the authors present a case report of 40-year-old male patient who reported to the Outpatient Department (OPD) with complaints of diminution of vision in Both Eyes (BE) for three years. There was a significant family history. Visual acuity was limited to hand movements in both eyes. Both eyes showed prominent Schwalbe's line, iris atrophic patches, and ectropion uvea. Intraocular Pressure (IOP) was high in the Right Eye (RE) and within normal limits in the Left Eye (LE). No craniofacial or dental anomalies were detected. Gonioscopy revealed a closed angle with broad peripheral anterior synechiae in both eyes. Fundus examination revealed a Cup Disc Ratio (CDR) of 0.5 in the RE and 0.9 in the LE. AR anomaly is a rare case. Long-term follow-up of these cases is as important as early diagnosis and treatment to prevent blindness.

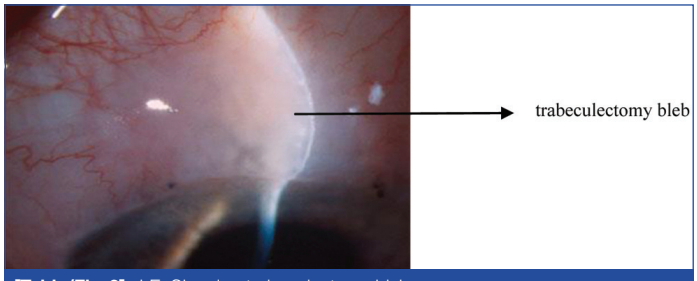
Keywords: Anterior segment dysgenesis, Intraocular pressure, Posterior embryotoxon, Secondary glaucoma

CASE REPORT

A 40-year-old male patient presented to the Ophthalmology OPD with a complaint of progressive vision loss in both eyes over the past three years. The patient reported no other ocular issues. Five years ago, the patient underwent trabeculectomy in the left eye. There is a positive and significant family history, as the patient's father and son have similar vision complaints. The Best Corrected Visual Acuity (BCVA) was assessed as hand movements in both eyes. Upon slit lamp examination, the right and left eyes [Table/Fig-1,2] exhibited prominent Schwalbe's line, iris atrophic patches, and ectropion uvea.



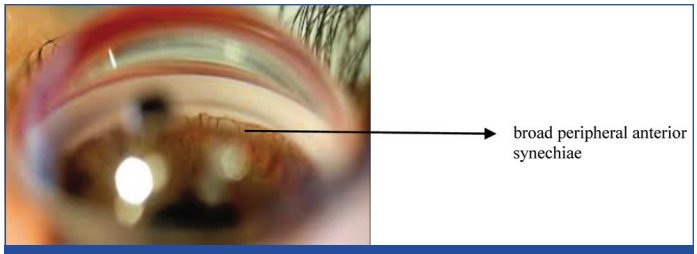
[Table/Fig-1]: Right Eye (RE).
[Table/Fig-2]: Left Eye (LE). (Images from left to right).



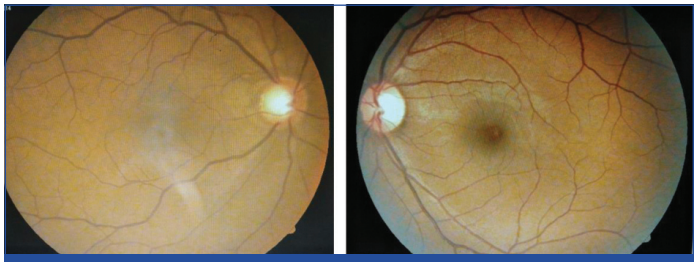
[Table/Fig-3]: LE: Showing trabeculectomy bleb.

The IOP measured by Goldmann's Applanation Tonometer was 43 mmHg in the right eye and 16 mmHg in the left eye. Upon examination, the left eye exhibited a trabeculectomy bleb [Table/Fig-3] and peripheral iridectomy [Table/Fig-2].

Gonioscopy on the right eye using Goldmann's three-mirror lens showed a closed angle with broad peripheral anterior synechiae [Table/Fig-4]. Fundus examination via indirect ophthalmoscopy revealed a Cup Disc Ratio (CDR) of 0.5 in the right eye and 0.9 in the left eye [Table/Fig-5,6], respectively.



[Table/Fig-4]: Closed angle with broad peripheral anterior synechiae (RE).



[Table/Fig-5]: Right Eye (RE) fundus with CDR 0.5.
[Table/Fig-6]: Left Eye (LE) fundus with CDR 0.9. (Images from left to right).

The patient sought opinions from a neurophysician and a dentist to rule out myotonic dystrophy and dental anomalies such as hypodontia, microdontia, and maxillary hypoplasia. No craniofacial or dental anomalies were detected. After ruling out other potential differential diagnosis, the patient was diagnosed with an AR anomaly. The patient was then initiated on antiglaucoma medication, receiving Timolol 0.5% in combination with Dorzolamide 2% eye drops twice daily in the right eye and sustained-release Acetazolamide 250 mg tablets twice daily.

Once the IOP was reduced to 20 mmHg in the right eye, and following the necessary work-up and consent, trabeculectomy was performed in the right eye under local anaesthesia. One week postoperatively, the IOP was 16 mmHg in the right eye and 14 mmHg in the left eye. The patient was followed-up after one month, but there was no observed visual improvement in both eyes.

DISCUSSION

The failure of normal development of the anterior segment of the eye is known as anterior segment dysgenesis, and it is associated with an increased risk of glaucoma [1,2]. Anterior segment dysgenesis

encompasses a group of rare autosomal dominant conditions, including posterior embryotoxon, AR syndrome, Peter's anomaly, and Aniridia [3]. Several different gene mutations, encoding for transcriptional regulators, have been described. These mutations specify the migration and differentiation of mesenchymal progenital cells of neural crest origin into distinct anterior segment tissues. The interplay between PITX2 and FOXC1 explains the phenotypic variability and genetic heterogeneity of anterior segment dysgenesis [1,4]. AR syndrome is an inherited autosomal dominant disorder with high penetrance and variable expressibility. The characteristic features include posterior embryotoxon, iridocorneal strands, iris changes, and associated systemic signs such as oligodontia, microdontia, cranial, and facial defects [4].

The Axenfeld-Rieger Anomaly (ARA) anomaly presents only with ocular changes [5]. Shields has postulated that both ocular and systemic phenotypes result from the disruption of migration and/or differentiation processes of the neural crest-derived tissues, such as the corneal endothelium, the anterior chamber angle structures, and the iris. The developmental anomaly may involve a high insertion of the anterior uvea in the trabecular meshwork or Schlemm's canal, or broad iridocorneal adhesions [6].

Bilateral developmental ocular abnormalities account for the main features, which may be asymmetrical. Other features include umbilical cord anomalies, hypoplastic mid-face, and agenesis of certain teeth (usually maxillary incisors). Posterior embryotoxon is present in 95% of cases of Alagille syndrome, which is characterised by vertebral defects, cardiopulmonary malformations, and paucity of intrahepatic bile ducts [7]. Axenfeld-Rieger syndrome can also present without a posterior embryotoxon [8]. There are a few case reports of AR syndrome [3,9]. There are only a few cases of Axenfeld or only Rieger's anomaly [4]. However, the present is a rare case report of a combination of both Axenfeld and Rieger's anomalies presenting with only ocular findings and a significant family history. If extraocular developmental abnormalities, especially in the teeth, facial bones, and periumbilical skin, are seen with AR anomaly, then it will be termed as AR syndrome [10].

Axenfeld-Rieger anomaly presents with ocular features such as ectropion uveae, corectopia, iris stromal hypoplasia, full-thickness iris defects, severe iris atrophy, and extensive peripheral anterior synechiae. In 50% of cases, it has been observed that glaucoma develops during early childhood or early adulthood, either due to associated angle anomalies or secondary synechial angle closure. Other ocular structures that are altered include a small or absent Schlemm's canal, aberrant development of the trabecular meshwork, and altered extracellular matrix [2]. Initially, elevated IOP is managed medically; subsequently, surgery is required. In early-onset glaucoma cases of ARA, surgery provides a satisfactory visual outcome [11].

CONCLUSION(S)

The Axenfeld-Rieger anomaly is a condition that can lead to gradual and irreversible visual loss, necessitating early diagnosis and intervention. We emphasise long-term follow-up in such cases. ARA is a blinding condition that represents the tip of the iceberg. In such potentially blinding anomalies, early diagnosis, intensive intervention, and long-term follow-up can lead to better visual outcomes.

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PARTICULARS OF CONTRIBUTORS:
1. Senior Resident, Department of Ophthalmology, Rohilkhand Medical College, Bareilly, Uttar Pradesh, India.
2. Professor, Department of Periodontology, Rohilkhand Dental College, Bareilly, Uttar Pradesh, India.
3. Consultant, Department of Cardiothoracic and Vascular Surgery, Satya Sai Heart Institute, Ahmedabad, Gujarat, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:
Prachi Singh,
F-7, Rohilkhand Medical College Campus, Staff Quarters,
Pilibhit Bypass Road, Bareilly-243006, Uttar Pradesh, India.
E-mail: singh1406prachi@gmail.com

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