International Journal of Medical Ophthalmology

E-ISSN: 2663-8274
P-ISSN: 2663-8266
www.ophthalmoljournal.com
IJMO 2022; 4(2): 37-39
Received: 05-10-2022
Accepted: 07-11-2022

Dr. Shalu Gupta
Assistant Professor, Department of Ophthalmology, Dr. YS Parmar Government Medical College, Nahan, Himachal Pradesh, India

Dr. Mohammad Shahid Khan
Senior Resident, Department of Ophthalmology, Dr. YS Parmar Government Medical College, Nahan, Himachal Pradesh, India

Corresponding Author:
Dr. Mohammad Shahid Khan
Senior Resident, Department of Ophthalmology, Dr. YS Parmar Government Medical College, Nahan, Himachal Pradesh, India

Axenfeld-rieger syndrome: A case report

Dr. Shalu Gupta and Dr. Mohammad Shahid Khan

DOI: https://doi.org/10.33545/26638266.2022.v4.i2a.126

Abstract
Axenfeld–rieger syndrome (ARS) is a rare autosomal dominant genetic condition characterized by anterior segment dysgenesis and systemic abnormalities.

Keywords: Small incision cataract surgery, trabeculectomy, glaucoma, intraocular pressure

Introduction
Axenfeld–rieger syndrome (ARS) is an autosomal dominant genetic condition characterized by anterior segment dysgenesis and systemic abnormalities. In 1920, axenfeld characterized the anomaly when he described posterior embryotoxon and iris strands adherent to the anteriorly displaced schwalbe's line [1]. Rieger described patients with congenital iris abnormalities including iris hypoplasia, corectopia, and polycoria, later referred as Rieger anomaly [2]. Rieger anomaly associated with systemic findings like dental, facial bone defects, umbilical abnormalities or pituitary involvement is known as Rieger Syndrome [3, 4]. The combination of Axenfeld anomaly and Rieger syndrome is known collectively as Axenfeld –Rieger syndrome.

Case report
An eleven years old male child was presented in Eye OPD with complaints of decreased vision and polyopia. On complete eye examination, his corrected visual acuity was 6/24 in right eye and 6/12 in the left eye. On Slit lamp bio-microscopic examination, it was found that he was having polycoria (multiple pupils) in both eyes (Figure 1A & 1B); in gonioscopic examination there were peripheral anterior synechie at 5 o'clock & 7 o'clock in right eye and at 10 O'clock in the left eye. His intraocular pressure was 18.9 in right eye and 19.5 in the left eye, with 5.5 gm weight by schiotz tonometer. Fundus examination was normal in both eyes.

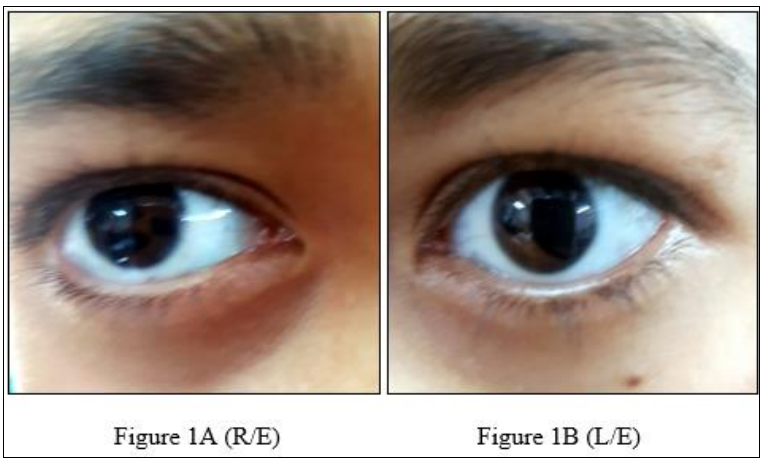

Fig 1(A&B): Child with polycoria

On systemic examination he was having broad nasal bridge, telecanthus, maxillary hypoplasia and mandibular hypognathism (Figure 2).

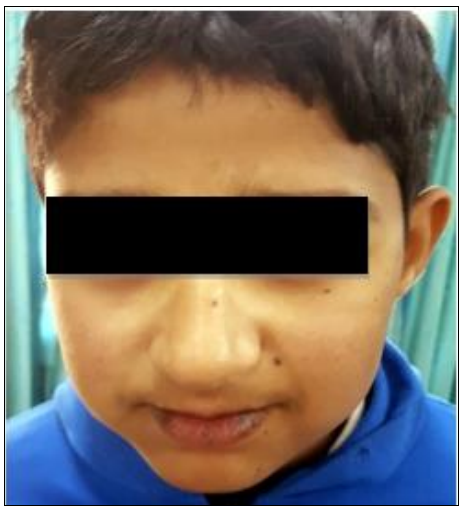

Fig 2: Child with telecanthus & broad nasal bridge

On dental examination he was having oligodontia (fewer than normal teeth) and microdontia (small teeth)[Figure 3A & 3B].

Fig 3 (A & B): Showing dental abnormalities

There was history of regular treatment at skin department for segmental vitiligo (upper abdominal area). On abdominal examination he had protuberant umbilicus with redundant skin folds.

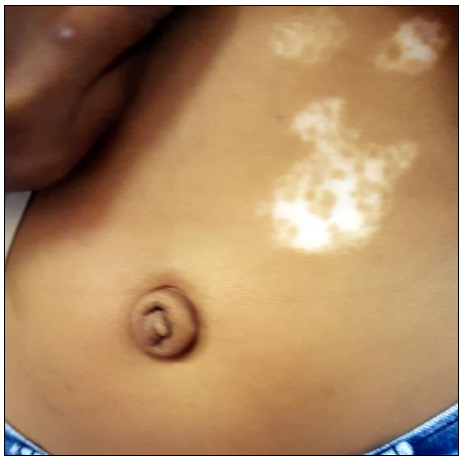

Fig 4: Showing protuberant umbilicus and vitiligo

The family history was not significant. The patient was referred to dental department for dental treatment. The patient was kept at regular follow up for early diagnosis of glaucoma and was referred to higher center for polyopia management.

Discussion

Anterior segment dysgenesis is a failure of normal development of the anterior segment of the eye. The structural anomalies of the mature anterior segment are associated with increased risk of glaucoma [1, 2, 5, 6]. Anterior segment dysgenesis is a group of rare autosomal dominant conditions, but can occur sporadically also [6], including posterior embryotoxon, Axenfeld-Rieger syndrome, Peter's anomaly and aniridia. Posterior embryotoxon may be absent in Axenfeld-Rieger syndrome [7].

ARS shows genetic heterogeneity and mutations of mainly two genes PITX2 and FOXC1 [4]. ARS patients are mainly diagnosed by the ophthalmologists and the diagnosis remains primarily clinical. As systemic changes are occasional findings of ARS, it will be useful to examine the patient for further changes, such as face and tooth abnormalities (Microdontia, hypodontia, oligodontia and adontia), redundant periumbilical skin and facial dysmorphism to facilitate the diagnosis. In case of an ASD, it is important to examine the patient annually with slit lamp examination, including gonioscopy, IOP measurements and fundus examination to assess the retinal nerve fiber layer and optic nerve head, to evaluate for early diagnosis of glaucoma. Autoperimetry (automated measurements of the visual fields) is necessary whenever glaucoma is suspected.

There is no sex predilection. Most cases are diagnosed during infancy or childhood; however glaucoma occurs later in childhood or adulthood due to an associated angle anomaly or secondary synechial angle closure [3, 4]. Disorders of ASD, which should be considered in differential diagnosis of ARS, are IH (iris hypoplasia/ iridogoniodysgenesis syndrome), Peters anomaly and PCG (primary congenital glaucoma). In IH, iris hypoplasia and goniodysgenesis is present, but posterior embryotoxon or iris adhesions are not found. Peters anomaly is an ASD with central absence of the corneal endothelium, descemet's membrane and posterior corneal stroma, leading to central corneal opacity. Cataract may also be present in Peters anomaly [8]. Patients with PCG have buphthalmos, goniodysgenesis and a high IOP. In PCG embryotoxon or iris adhesions are normally not observed.

Conclusion

The ocular and systemic features of Axenfeld-Rieger syndrome are well-described. Our stress is on long-term follow up in cases of abnormal ocular findings observed; the condition can lead to gradual and irreversible visual loss and needs special care and monitoring. The treatment is patient dependent since the severity of ARS varies. Elevation of intraocular pressure is initially managed medically, although surgery may be required afterwards. In addition, if photophobia is present in patients with corectopia and polycoria, they may use contact lenses to cover the holes in the iris. Thus a regular ophthalmological follow up along with genetic counseling to parents is very important for overall development of child.

Acknowledgments

I am thankful to the department of Ophthalmology of our institution for the diagnosis and management of this patient.

Financial Disclosure(s)

The author(s) have no proprietary or commercial interest in any materials discussed in this article.

Funding/Support
The author(s) don't have any government and non-government support.

Conflicts of interest
There are no conflicts of interest.

References
1. Simone Dressler, Philip Meyer-Marcotty, Nicole Weisschuh, Anahita Jablonski-Momeni, Klaus Pieper. Gwendolyn Gramer and Eugen Gramer. Dental and Craniofacial Anomalies Associated with Axenfeld Rieger Syndrome with PITX2 Mutation. Case Reports in Medicine; c2010. Article ID 621984.
2. Shields MB, Buckley E, Klintworth GK, R. Thresher, Axenfeld-Rieger syndrome. A spectrum of developmental disorders, Survey of Ophthalmology. 1985;29(6):387-409.
3. Alward WLM. Axenfeld-Rieger syndrome in the age of molecular genetics, American Journal of Ophthalmology. 2000;130(1):107-115.
4. Phillips JC. Four novel mutations in the PITX2 gene in patients with Axenfeld-Rieger syndrome, Ophthalmic Research. 2002;34(5):324–326.
5. Sowden JC. Molecular and developmental mechanisms of anterior segment dysgenesis. Eye. 2007;21:1310-1318.
6. Hjalt TA, semina EV:current molecular understanding of axenfeld–rieger syndrome. Expert Rev Mol Med 2005;7:1-17.
7. Sim KT, Krri B, Kaye SB: Posterior embryotoxon may not be a forme fruste of Axenfeld –Rieger's syndrome. J AAPOS. 2004;8:504-506.
8. Townsend WM: Congenital corneal leukomas. Am J Ophthalmol. 1974;77: 80-86.

How to Cite This Article
G Shalu, Mohammad SK. Axenfeld-rieger syndrome: A case report. International Journal of Medical Ophthalmology. 2022;4(2):37-39.