Case Report

# Axenfeld-Rieger Syndrome: A Rare Case

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**Abstract**

*A typical case of Axenfeld-Rieger syndrome (ARS) with clinical features of bilateral posterior embryotoxon, correctopia, pseudopolycoria and segmental iris hypoplasia is presented. Gonioscopy revealed anteriorly displaced Schwalbe's line and bands of iris extending across the iridocorneal angle to the trabecular meshwork. Fundus was within normal limits in both the eyes. Systemic examination revealed hypodontia, microdontia, prominent lower lip and redundant peri-umbilical skin. There was no associated cardiovascular or other associated systemic association. Axenfeld-Rieger syndrome is a multi-system anomaly requiring a multi-disciplinary approach to management. The patient needs to be regularly followed-up for development of glaucoma*

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***Keywords:*** *Axenfeld-Rieger syndrome, microdontia, pseudopolycoria, redundant peri-umbilical skin*

## Introduction

Axenfeld-Rieger syndrome (ARS) is an autosomal dominant genetic condition characterized by anterior segment dysgenesis and systemic abnormalities. In 1920, Axenfeld described posterior embryotoxon and iris strands adherent to the anteriorly displaced Schwalbe's line, so it bears his name.[1] It is a rare congenital, mostly bilateral condition. It is caused by mutation in one of several different genes, the most common of which are the PITX2 and FOXC1 genes, encoding for different transcription factors. In general, mutation in the PITX2 gene is more frequently seen in patients with ocular and systemic anomalies, whereas mutation in FOXC1 was found in patients with only ocular findings. The spectrum of disease involves Axenfeld anomaly (if changes are confined to the angle only), Rieger anomaly (if there are iris as well as angle abnormalities), and Axenfeld-Rieger syndrome (if there are associated non-ocular features such as maxillary, dental, cardiac or umbilical abnormalities). ARS is a multi-system anomaly requiring a multi-disciplinary approach to management. The patient needs to be regularly followed-up for early diagnosis and treatment of glaucoma. Early diagnosis is important to protect the eyes from glaucomatous damage.

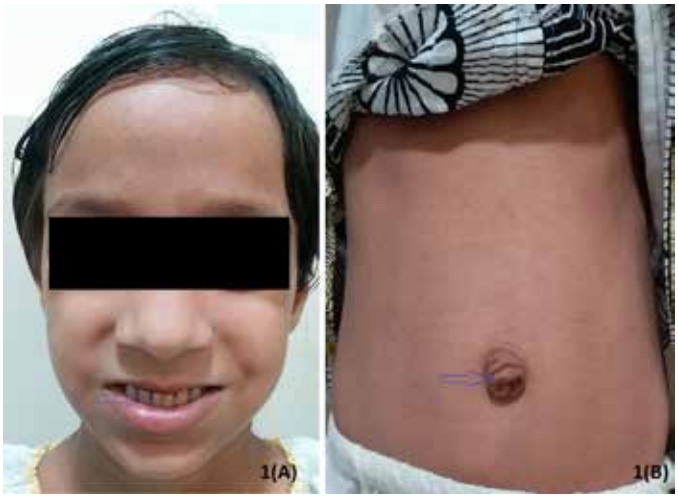


***Figure 1:*** *1(A) showing microdontia, diastema and prominent lower lip and 1(B) showing redundant peri-umbilical skin.*

## Case Summary

A 7 years old girl presented to us with chief complaint of painless, progressive diminution of vision since few months, associated with photophobia and watering. The child was born full term vaginal delivery with no neonatal intensive care unit admission. Her visual acuity at presentation was 6/12 in right eye improving to 6/6 with pinhole, and 6/18 in left eye improving to 6/9 with pinhole on Snellen's chart. She had normal hearing and intelligence as per her age group, with normal developmental growth.

There was no family history of similar complaints in the siblings. Her general examination revealed that the teeth were small (microdontia) (Figure 1a), with wide spaces between them (diastema), hypodontia, prominent lower lip and redundant peri-umbilical skin (Figure 1b). Detailed cardiovascular system examination revealed normal electrocardiogram (Figure 5b) and normal echocardiography. Her intraocular pressure by applanation tonometry was 22 mm of Hg and 18 mm of Hg in right and left eye respectively. On detailed anterior segment examination, cornea was clear in both the eyes. = Her central corneal thickness was 563 microns and 532 microns in right and left eye respectively. Specular microscopy (Figure 2) demonstrated endothelial cell density of 2351 cells/ $mm^2$ and 2776 cells / $mm^2$ and hexagonal cell percentage as 77% and 58% in right and left eye respectively. Iris showed eccentric pupil (correctopia), segmental iris hypoplasia (Figure 3a) and pseudopolycoria (Figure 3b). Anterior segment optical coherence tomography revealed similar picture of iris hypoplasia and peripheral iridocorneal adhesions (Figure 3c). Gonioscopy (Figure 4) revealed anteriorly displaced.

Schwalbe's line with iridocorneal adhesion bands of iris extending across the iridocorneal angle to the trabecular meshwork. Fundus (Figure 5a) was within normal limits with no signs of glaucomatous changes at present.

On the above findings, a diagnosis of Axenfeld-Rieger syndrome was made. The child was prescribed proper


***Figure 2:*** *Specular microscopy: Endothelial cell density of 2351cells/mm2 and 2776 cells/mm2 and hexagonal cell percentage as 77% and 58% in right and left eye respectively*

refractive correction and was advised 3 monthly regular follow-up for the detection of glaucoma.

## Discussion

The spectrum of disease involves Axenfeld anomaly (if changes are confined to the angle only), Rieger anomaly (if there are iris as well as angle abnormalities), and Axenfeld-Rieger syndrome (if there are associated nonocular features such as maxillary, dental, cardiac or umbilical).

A clear to white line in the peripheral cornea due to anterior displacement of Schwalbe's line, termed posterior embryotoxon, is the hallmark of Axenfeld-Rieger syndrome. It was present in our patient over the entire circumference of the cornea though it can be confined to a few clock hours only. Posterior embryotoxon is most commonly seen in

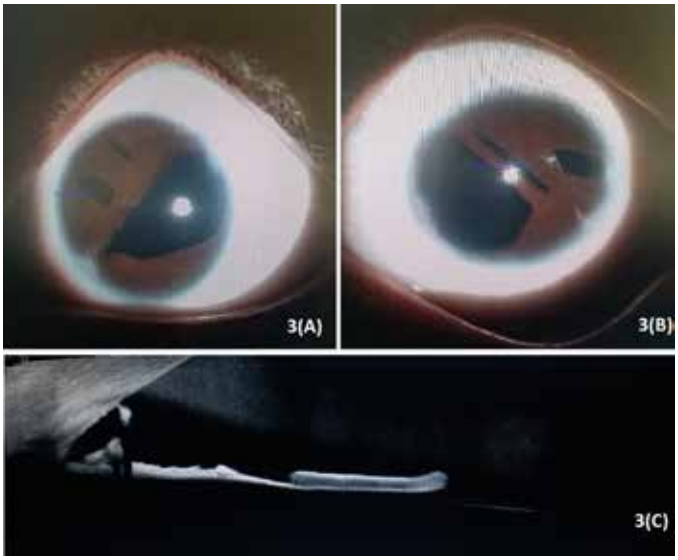


***Figure 3:*** *(A) Segmental iris hypoplasia, (B) Pseudopolycoria, (C)- A S OCT demonstrating iris hypoplasia and peripheral iris iridocorneal adhesions*

the temporal cornea. It is important to note that posterior embryotoxon is not universal and is neither specific to this syndrome as can also be seen in otherwise around 15 percent of normal population.[2] The corneal endothelium was normal as revealed by specular microscopy; differentiating it from Iridocorneal endothelial syndrome. Our patient also had iris hypoplasia, displacement of the pupil from the central position (corectopia). In eyes with corectopia, the pupil is displaced towards the area of iris process adhesions which was also present in our patient and confirmed by gonioscopic examination. Our patient also had extra holes in the iris (psedopolycoria), in the quadrant opposite to the corectopia. The iris changes are usually stationary, but have been reported to progress in some patients.[3] The iridocorneal angle on gonioscopy revealed anteriorly displaced Schwalbe's line and bands of iris extending across the iridocorneal angle to the trabecular meshwork. These strands are called iris processes. These are not the fine processes seen in normal iridocorneal angles. They can range from single strand to broad sheets covering several clock hours. These strands are different from synechiae, as they do not close the angle. Glaucoma develops in about 50% of patients with Axenfeld-Rieger syndrome.[4] The risk of developing glaucoma is not correlated with the extent of iris processes but is related to the height of the iris insertion in the iridocorneal angle. Patients with more anterior iris insertions have a higher risk of developing glaucoma.[5] Our patient had intraocular pressure of 22 mm of Hg and 18 mm of Hg in right and left eye respectively, but as the fundus was within normal limits with no signs of glaucomatous changes presently; so was kept on 3 monthly follow-up. Our patients also had dental anomalies as few teeth (hypodontia) and small teeth (microdontia). Patients also had redundant periumbilical skin. This may be mistaken with an umbilical hernia in some patients. The most important differential diagnosis of Axenfeld - Rieger syndrome is the iridocorneal endothelial

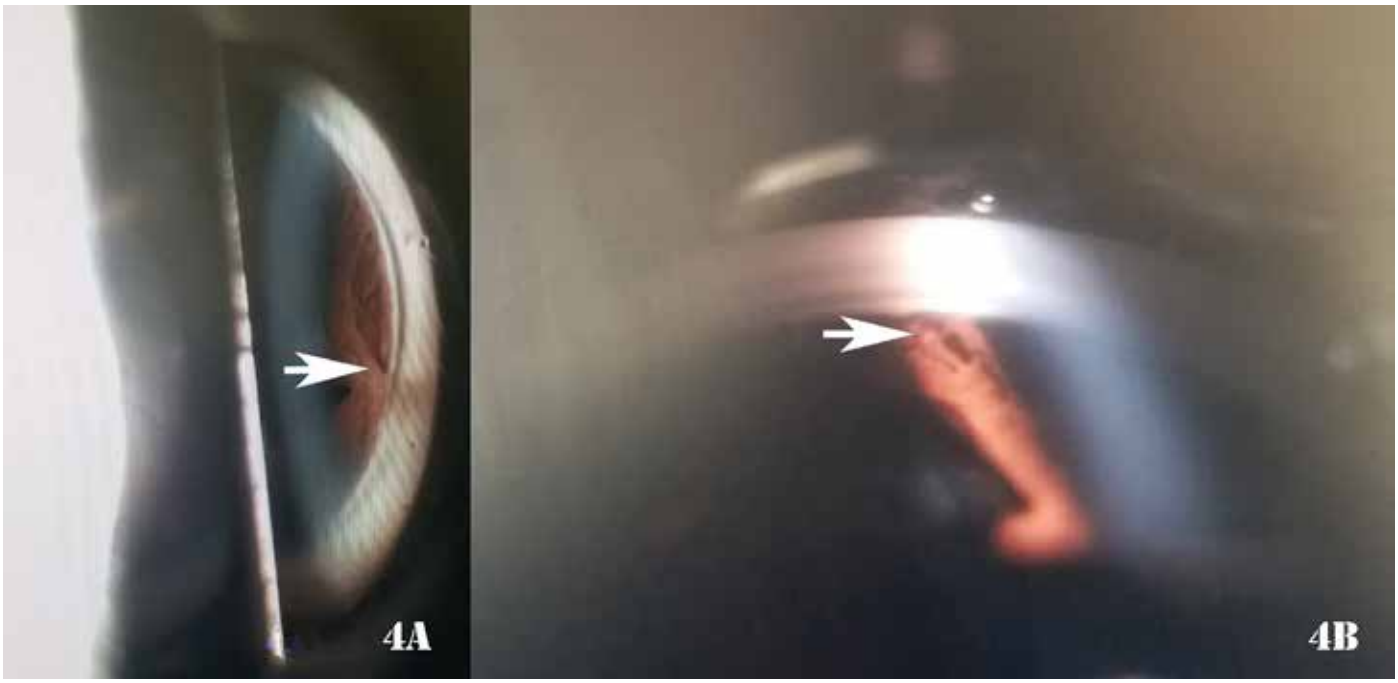


*Figure 4: (4A),(4B) Gonioscopy of both eyes demonstrating bands of iris extending across the iridocorneal angle to the trabecular meshwork.*

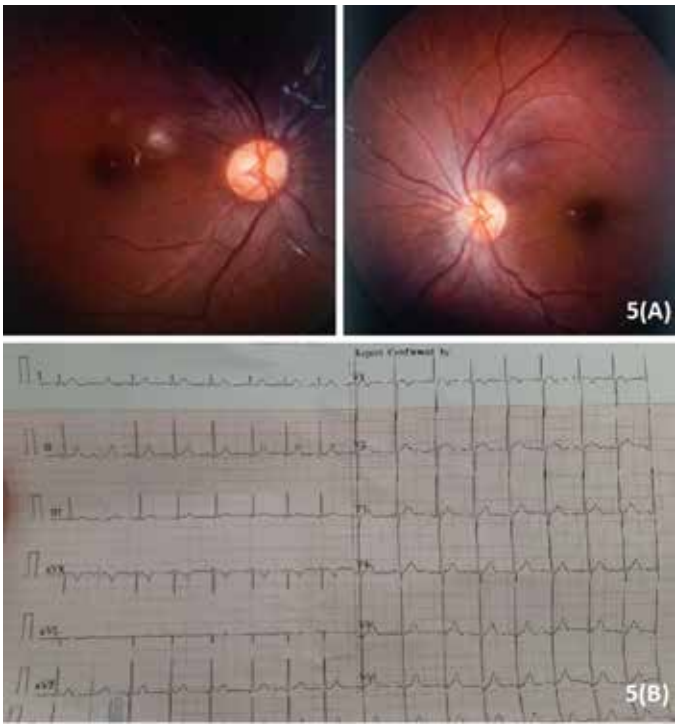


***Figure 5:*** *(5A) Colored fundus photograph showing normal optic disc and central fundus (5B) Normal Echocardiography of patient*

(ICE) syndrome. Both syndromes can have corectopia and polycoria and both may cause glaucoma. The iris atrophy in the ICE syndrome can look very much like the iris hypoplasia in Axenfeld-Rieger syndrome. In both diseases, broad sheets of iris may cover the iridocorneal angles but in Axenfeld-Rieger syndrome these are iris processes, while in the ICE syndrome they are broad synechiae. Despite these similarities, both can be easily identified. Axenfeld-Rieger syndrome is almost always bilateral, while ICE syndrome is nearly always unilateral. Axenfeld-Rieger syndrome is present since birth, while ICE syndrome typically develops during adulthood. Posterior embryotoxon is seen in majority of cases of Axenfeld-Rieger syndrome and but is not a feature of ICE syndrome.

## Conclusion

Axenfeld-Rieger syndrome is a multi-system anomaly requiring a multi-disciplinary approach to management. The patient needs to be regularly followed-up for the development of glaucoma.

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