ISSN: 2754-4990

Journal of Ophthalmology Research Reviews & Reports

SCIENTIFIC Research and Community

Case Report

Open Access

# Axenfeld-Rieger Syndrome: A Case Report with Literature Review

**M Zakoun*, S Belghmaidi, J Hakam, I Hajji and A Moutaouakil**

Ophtalmology Department, Mohammed VI university Hospital, Marrakesh, Morocco

***Corresponding author**
M Zakoun, Ophtalmology Department, Mohammed VI university Hospital, Marrakesh, Morocco. E-Mail: zakoun.maria@gmail.com

**Received:** October 26, 2021; **Accepted:** November 01, 2021; **Published:** November 08, 2021

## Introduction

Axenfeld–Rieger syndrome (ARS) is a rare autosomal dominant disorder that has both systemic and ocular anterior segment dysgenesis. The ocular manifestations include posterior embryotoxon, iris and anterior angle abnomalies with a high risk of glaucoma and blindness. The systemic manifestations can include craniofacial abnomalies such as maxillary hypoplasia, hypodontia, oligodontia and microdont.

## Case Report

We report the case of an 8 years old boy with no personal or family history, who was referred for redness and bilateral progressive decrease of the visual acuity. An ophthalmic examination was performed. The best corrected visual acuity was 2/10 on the rigt eye and 4/10 on the left eye. The intraocular pressure was respectively 28mmhg and 26 mmgh in both eyes. The white to white corneal diameter was 10.5mm on the left eye and 11mm on the right eye.

The slit lamp examination showed in both eyes a clear cornea with posterior embryotoxon , stromal iris hypoplasia with corectopia and polycoria. The gonioscopy showed anterior synechia in both eyes and the fundus examination found an advanced excavated optic disc in both eyes.

A general exam was performed and revealed dental abnormalities such as superior hypodontia and microdontia and cone-shaped teeth. There were no anomalies in the periumbilical skin or other systemic features.

The diagnostic of axenfeld Rieger syndrome with secondary glaucoma was made. First, a combined beta blockers and carbonic anhydrase inhibitors drug was prescribed to reach a controlled IOP. Then, a trabeculotomy was performed with good outcomes.

## Discussion

Axenfeld-Rieger syndrome is a spectrum of rare disorders characterized by anomalous development of the neural crest–derived anterior segment structures, including the anterior chamber angle, the iris, and the trabecular meshwork and systemics features. Mutations in the FOXC1and PITX2genes were associated with ARS in an autosomal dominant inherited manner [1].

The ocular involvement in ARS is usually bilateral, rarely asymmetric and exceptionally unilateral. It includes anomalies in the cornea (posterior embryotoxon), iris (corectopia or polycoria associated with anterior iris stromal atrophy), and angle structures that can result in secondary glaucoma in approximately 50% of affected patients and can lead to blindness [2-3].

Systemic developmental abnormalities in ARS includes cardiac malformations, non-involution of periumbilical skin, craniofacial dysmorphy (hypertelorism, telecanthus, maxillary hypoplasia),dental malformations (microdontia, hypodontia, oligodontia, anodontia). Anal stenosis, hypospadias, pituitary gland abnormalities, arachnoid cysts, growth retardation can also be seen [4-5].

The management of the ocular findings in ARS should be proposed whenever a secondary glaucoma is diagnosed. In that case, a conventional glaucoma surgery like trabeculectomy and trabeculotomy is required. In fact, Mandal and Pehere confirmed the safety and effectiveness of the combination of trabeculotomy and trabeculectomy for ARS children with early-onset of glaucoma. The maxillary and oral anomalies need orthondontic surgery and intensive restorative care with implantology [4-6].

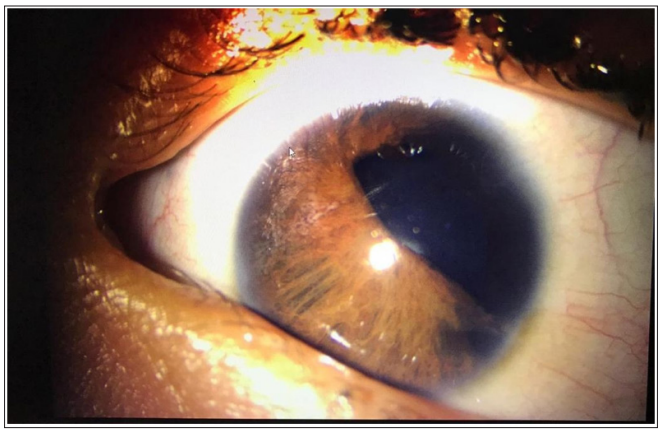

**Figure 1:** Right eye with polycoria, iris atrophy and posterior embryotoxon

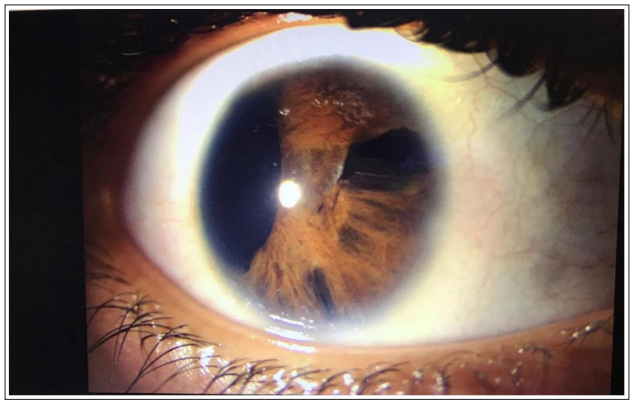

**Figure 2:** Left eye with polycoria, corectopia, iris atrophy, posterior embryotoxon

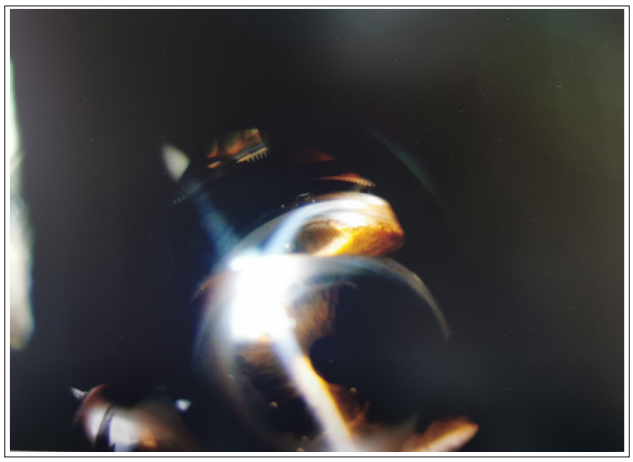

**Figure 3:** Gonioscopy showing goniosynechiae and angle dysgenesis

## Conclusion

The aim of this case report is to show the interest of an early diagnosis and management of Axenfield Rieger Syndrome in order to avoid secondary glaucoma and blindness.

## References

1. Agarwal, P Jain, K Sandesh S, & Chopra, S (2020) Axenfeld–Rieger Syndrome: Rare Case Presentation and Overview. Journal of Maxillofacial and Oral Surgery 19: 364-369.
2. Honkanen R A, NishimuraDY, Swiderski RE, Bennett S R, Hong S,et al (2003) A family with Axenfeld–Rieger syndrome and Peters anomaly caused by a point mutation (Phe112Ser) in the FOXC1 gene. American journal of ophthalmology 135: 368-375.
3. Puthalath A S, Agrawal A, Rana R, Samanta, R (2020) A case of Axenfeld-Rieger syndrome (ARS) with asymmetric ocular phenotypes and left glaucomatous optic atrophy. BMJ Case Reports 13: 237224.
4. Chekhchar M, Charadi A, Achibane A, Hajji I, Moutaouakil A (2019) Axenfeld-Rieger syndrome: A case report Syndrome. Journal Français d'Ophtalmologie.
5. Seifi M, Walter MA (2018) Axenfeld-Rieger syndrome. Clinical Geneticsm 93: 1123-1130.
6. Song W, Hu X (2017) the rare Axenfeld–Rieger syndrome with systemic anomalies: A case report and brief review of literature. Medicine 96: 7791.