International Journal of Early Childhood Special Education (INT-JECSE)
DOI:10.9756/INTJECSE/V14I5.323 ISSN: 1308-5581 Vol 14, Issue 05 2022

Axenfeld Rieger Syndrome and its Manifestations: A Case Report

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Abstract--- Axenfeld Rieger syndrome (ARS) is an uncommon genetic disorder with a range of ocular and systemic manifestations such as dental, craniopharyngeal, and umbilical anomalies. This report aims to introduce a typical case of ARS and present its manifestations. The patient was a 13-year-old woman complaining of blurred vision and occasional eye pain. General Examination of the patient showed impaired development of her teeth and redundant periumbilical skin with an umbilical hernia. Best-corrected visual acuity was 20/25 and 20/30 in the right and left eyes respectively. Slit-lamp examination revealed a thin opacity ridge of the cornea, beside the limbus (posterior embryotoxon), adhesions of the corneal periphery to iris, a severely atrophic iris without crypts, irregular and elongated pupils in both eyes, in addition to pseudopolycoria due to iris atrophy in the right eye. On gonioscopy, peripheral anterior synechiae (PAS) were present in both eyes. Intraocular pressure (IOP) measured using a Goldman tonometer was 17 mm Hg and 16 mm Hg, in the right and left eyes respectively. With timolol-dorzolamide drops every 8 hours and latanoprost at night. On examination of the posterior segment of the eyes, the vitreous was optically clear, the right optic disc was pink and had a sharp margin, and the cup disc ratio was 4/10, the optic disc of the left eye was also pink and had a sharp margin, cup to disc ratio was 5/10. The foveal reflex was good and the retina was unremarkable up to the periphery in both eyes.

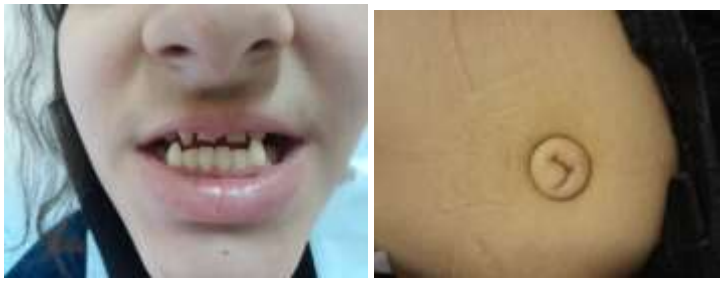
Keywords--- Axenfeld Rieger Syndrome, Manifestations.

I. Introduction

Axenfeld Rieger Syndrome (ARS) is a rare genetic disorder of embryonic development with a prevalence of 1 in 200,000 live births 1. This disease causes ocular involvement with anterior segment anomaly and systemic manifestations such as dental, craniopharyngea, and umbilical malformations 2. This report aims to introduce a typical case of ARS and present its manifestations.

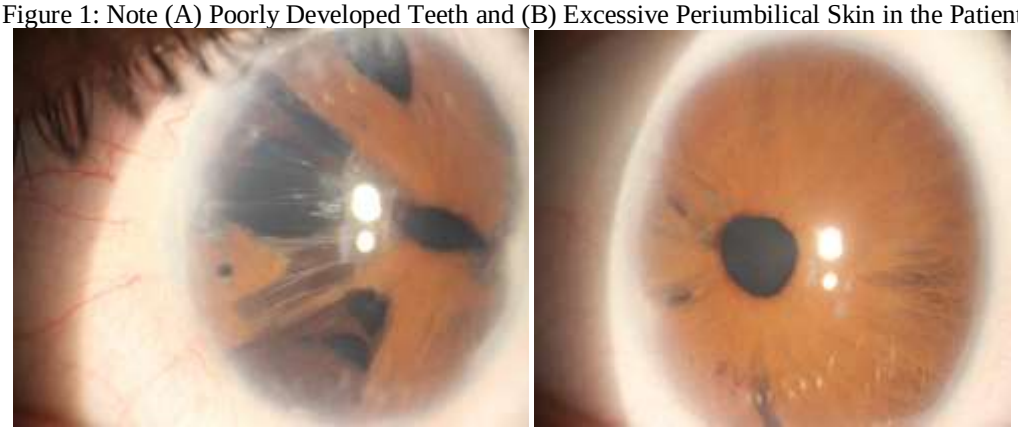
Case Presentation

The patient was a 13-year-old woman complaining of blurred vision and occasional eye pain. There was no history of systemic disease and systemic drug use. General Examination of the patient showed impaired development of her teeth and redundant periumbilical skin with an umbilical hernia (Figure 1). Best-corrected visual acuity was 20/25 and 20/30 in the right and left eyes respectively. Slit-lamp examination revealed a thin opacity ridge of the cornea, beside the limbus (posterior embryotoxon), adhesions of the corneal periphery to iris, a severely atrophic iris without crypts, irregular and elongated pupils in both eyes, in addition to pseudopolycoria due to iris atrophy in the right eye (figure 2,3). On gonioscopy peripheral anterior synechiae (PAS) were present in both eyes.



A B

Figure 1: Note (A) Poorly Developed Teeth and (B) Excessive Periumbilical Skin in the Patient



A B

Figure 2: Biomicroscopic Photographs of the Right (A) and Left (B) Eyes Shows the Mentioned Anterior Segment Anomalies

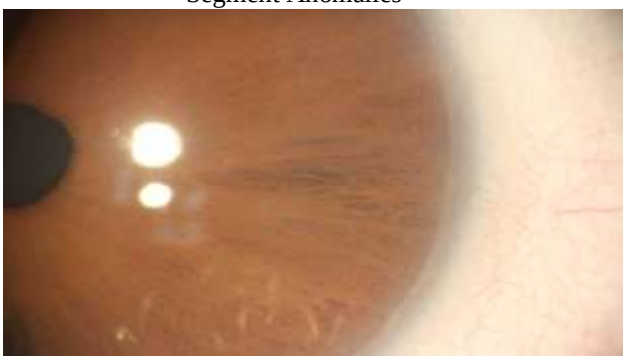


Figure 3: Biomicroscopic Photographs of the Eye Shows the Posterior Embryotoxon

Angle adhesion was evident in anterior segment optical coherence tomography (OCT) imaging (Figure 4). The thickness of the right and left corneas was 571 and 610 microns, respectively. Intraocular pressure (IOP) measured using a Goldman tonometer were 17 mm Hg and 16 mm Hg, in the right and left eyes respectively with timolol-dorzolamide drops every 8 hours and latanoprost at night. On examination of the posterior segment of the eyes, the vitreous was optically clear, the right optic disc was pink and had a sharp margin, and cup to disc ratio was 4/10, the optic disc of the left eye was also pink and had a sharp margin, cup to disc ratio was 5/10. The foveal reflex was good and the retina was unremarkable up to periphery in both eyes. (Figure 5). In order to evaluate glaucoma, the 24-2 central visual field with SITA STANDARD strategy and Macular Ganglion Cell Complex (GCC) analysis were performed. However, there were no glaucomatous changes (Figure 6,7).

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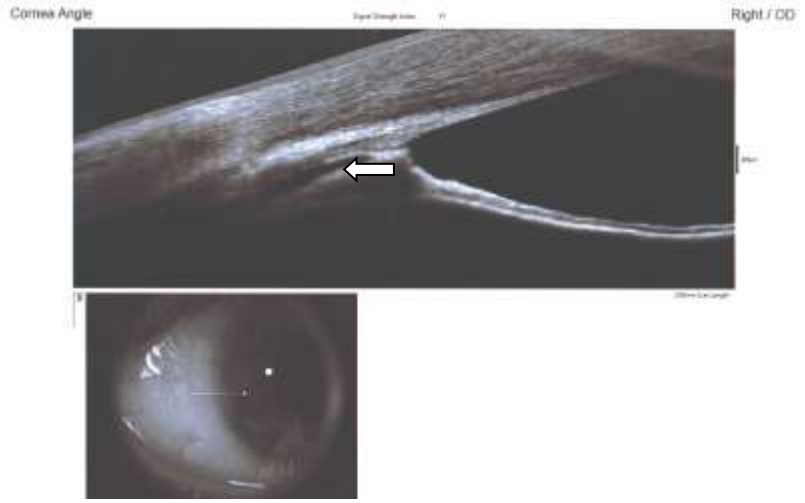
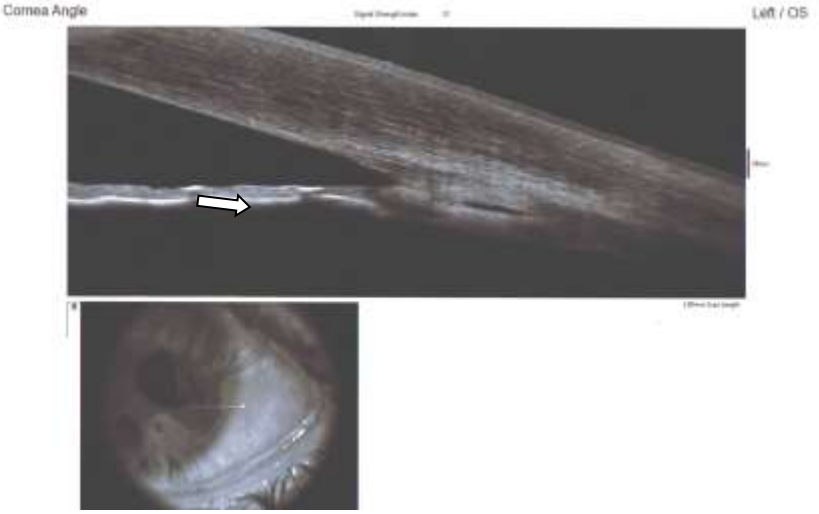


A



B

Figure 4: Anterior Segment Optical Coherence Tomography of the Right (A) and Left(B) Eyes. Performed in the Patient. Note the Angle Developmental Anomaly and Iris Adhesions to Angle Structures (Arrows)

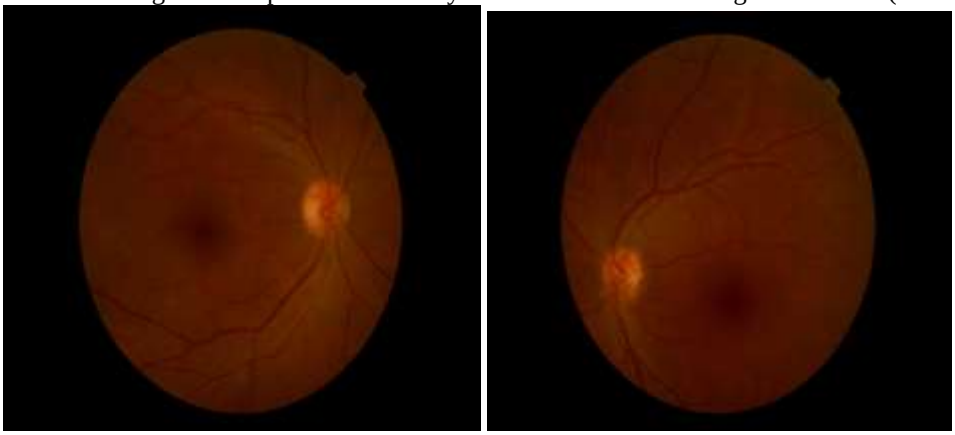


Figure 5: Fundus Photographs of the Right (A) and Left (B) Eyes

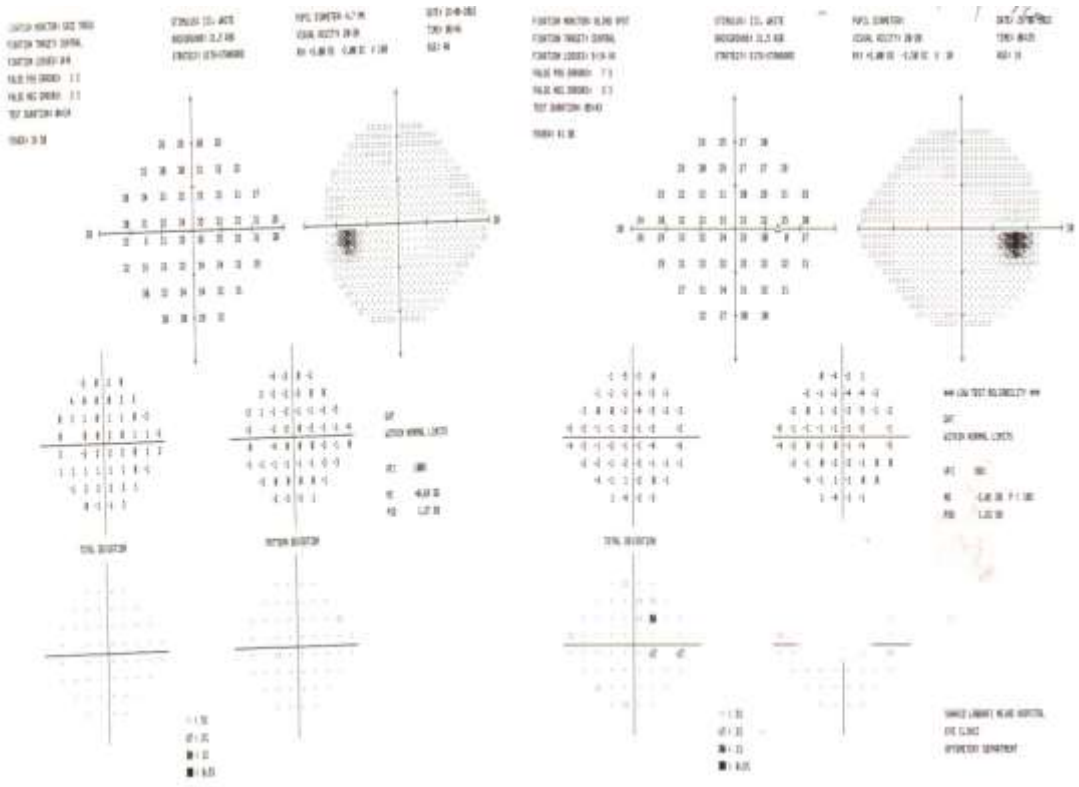


Figure 6: 24-2 Central Visual Fields with SITA STANDARD Strategy Shows no Evidence of no Glaucomatous Damage

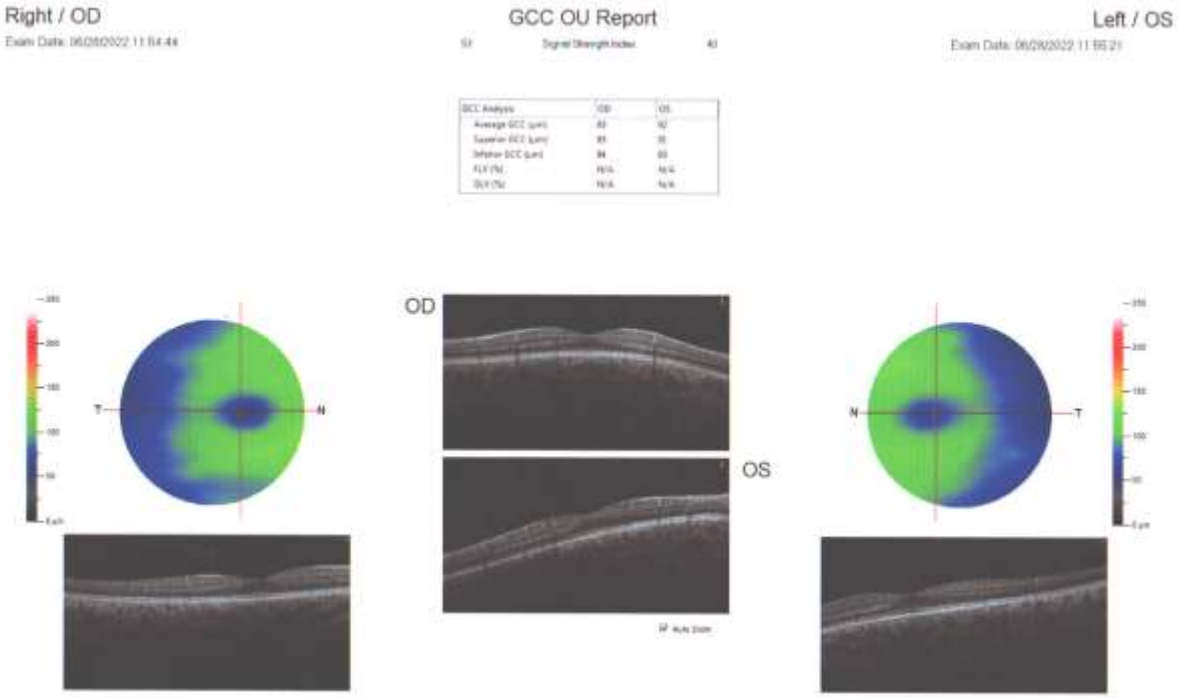


Figure 7: Optical Coherence Tomography Analysis of the Macular Ganglion Cell Complex (GCC) shows no Early Glaucomatous Damage in either Eye of the Patient

II. Discussion

In this study, a 13-year-old woman with Axenfeld Rieger Syndrome (ARS) was reported. ARS is an uncommon disease that most often transmitted as an autosomal dominant inheritance 3. The effective genes for this disease are PITX2 (pituitary homeobox transcription factor 2) and FOXC1 (forkhead box transcription factor C1), but sporadic transmission also occurs 4. In our patient, since the patient's father had similar symptoms, the hereditary transmission was probably involved. The disease is associated with a range of symptoms, from ocular involvement to systemic manifestations of dental, and craniopharyngeal and umbilical malformations 2. Ocular manifestations include corneal involvement, posterior embryotoxon, iridocorneal angle adhesions, irregular-shaped pupils, iris atrophy, ocular hypertension (OHT), and glaucoma 4,5.

Posterior embryotoxon is a corneal abnormality visible with slit-lamp biomicroscopy as thin opacity ridge of the cornea, beside the limbus. It is an anteriorly displacement Schwalbes line (Figur3). Posterior embryotoxon may also be seen in normal eyes without the disease 5,6.

This syndrome usually occurs early in life, but patients usually remain asymptomatic until increased in intraocular pressure and decrease vision 6. Increased intraocular pressure and glaucoma are seen in almost 50% of cases and are one of the causes of decreased vision in these patients 7,8. In most cases, the disease affects the eyes bilaterally 5. Anti-glaucoma drugs are used to control OHT and glaucoma in the early stages 5.6. However, in some cases, the response to the drug is insufficient and requires eye surgery such as trabeculectomy to control eye pressure 9. In our patient, she had an increase in left eye intraocular pressure during periodic examinations, which could not be controlled by adding brimonidine drops to previous drugs, however, trabeculectomy procedure with mitomycin 0.2 mg/ml applied for 2 minutes at the surgical site was performed. After the above surgery, the IOP of the left eye was controlled without antiglaucoma drops. In previous studies, trabeculectomy with anti-fibrotic has been successfully accepted to control intraocular pressure and glaucoma in patients with ARS 10,11.

III. Conclusion

Axenfeld Rieger syndrome is a disease whose complications include increased intraocular pressure, glaucoma, and irreversible vision loss. However periodic eye examinations, IOP measurement and paraclinical tests to diagnose glaucoma are necessary. Due to the inherited transmission of this disease, the first-degree relatives of patients should be subjected to detailed eye examinations for the possible diagnosis of this syndrome.

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