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Late Presentation Of Isolated Unilateral Axenfeld Rieger Syndrome- A Case Report

Dr. Tanvi Gupta[1], Dr. Ajita Sasidharan[2]
[1](Glaucoma Department, Sankara Eye Hospital, Coimbatore, India)
[2](Glaucoma Department, Sankara Eye Hospital, Coimbatore, India)

Abstract
Axenfeld Rieger syndrome is a rare developmental disorder which is usually bilateral with frequent systemic involvement. An atypical case with late prentation and unilateral features of Axenfeld Rieger Syndrome i.e. posterior embryotoxon, peripheral anterior synechiae of 1-2 clock hours and corectopia in the direction of the synechiae without glaucoma or systemic involvement is presented here.

Key Word*: Axenfeld Rieger Syndrome, posterior embryotoxon, corectopia, unilateral ARS*

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I. Introduction

Axenfeld – Rieger Syndrome (ARS) is a very rare autosomal dominant disorder with incidence of 1:200000 over the world. [1] It is a neural crest cell derived anterior segment dysgenesis. This disorder is attributed to mutations in genes, most commonly PITX2 and FOXC1 genes.[2] It involves abnormalities of ocular and systemic structures ranging from heart and brain to cranio-facial structures and several other disorders like dental and peri-umbilical abnormalities.[3] There is a 50% risk of a secondary glaucoma[4] in these patients due to high insertion of iris into the angle and under- development of trabecular meshwork and Schlemm's canal. [5]

Our aim here is to describe a case of unilateral Axenfeld- Rieger syndrome diagnosed incidentally in an elderly lady who came to us with blurring of vision due to lenticular opacities in both her eyes.

II. Case Report

A 50 year old female presented to us with chief complaint of gradual painless progressive diminution of vision in both eyes since 6 months. No significant ocular history, no history of systemic disease and no relevant family history was given by the patient. Her uncorrected visual acuity was 20/200 improving to 20/120 with pin hole in right eye and 20/120 not improving with pinhole in left eye.

On slit lamp examination her right eye showed a posterior embryotoxon from 3'0 clock to 10'0 clock, slight inferior displacement of pupil, and inferior peripheral anterior synechiae 2 clock hours [Figure 1]. The anterior chamber was shallow (Van Herick Grade two) in both eyes and there was NS2 grade cataract in both eyes. Intra-ocular pressure measured by Goldmann Applanation Tonometer was 20 mmHg in right eye and 14 mm Hg in left eye.

On gonioscopy, all angles were occludable and there was a 1-2 clock hours PAS in inferior angle in right eye. [Figure 2(a)]. Fundus examination showed discs with a vertical cup disc ratio of 0.3. Macula was within normal limits in botheyes and foveal reflex was dull.

Axial length was 21.00 mm in right eye and 21.10 mm in left eye and IOL power calculated was 26.00 in right eye. On specular microscopy, cell count was 2428 cells/mm^2 in right eye and 2769 cells/mm^2 in left eye. Endothelial cells looked normal with clear margins and no light and dark areas in the cells. [Figure 2 (b) and 2(c)]. Intercanthal distance,inter-pupillary distance and outer canthal distance were within normal limits [Figure 3(b)].

Patient had normal intelligence and hearing and was communicative. General appearance was normal and there was no facial dysmorphism [Figure 3(a)]. On general examination, her dentition was normal and there was no redundant per-umbilical skin [Figure 3(c)]. Electrocardiography (ECG) showed no abnormal findings [Figure 4].

A provisional diagnosis of isolated unilateral Axenfeld Rieger Syndrome in right eye was made. ARS is usually a bilateral entity, but in our patient it was present unilaterally and other eye was completely normal. Patient was operated for the cataract uneventfully by Small Incision Cataract Surgery (temporal section). Intra-ocular lens was placed in capsular bag. Patient obtained a uncorrected visual 20/80 improving with pinhole to 20/60 in right eye on the next day of surgery.

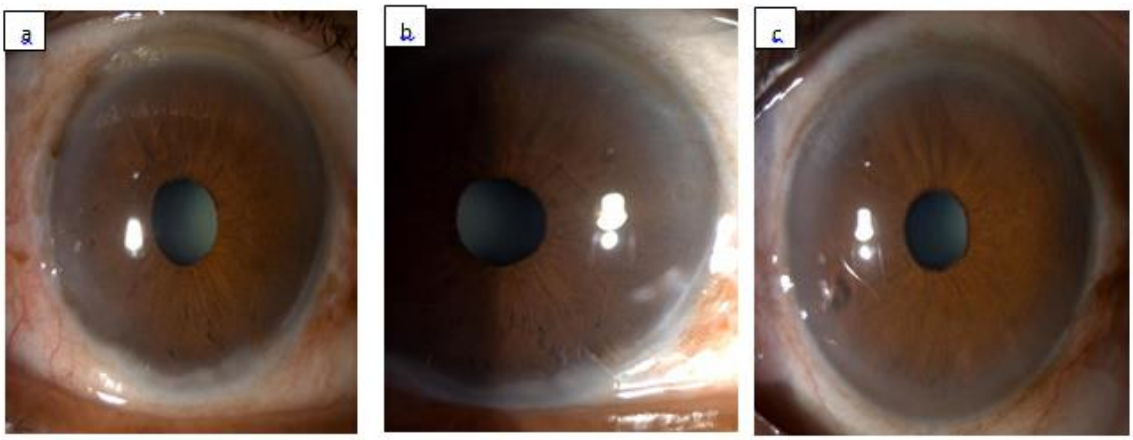

Figure 1: Anterior segment photo of a) right eye showing posterior embryotoxon, PAS, mild corectopia b) right eye highlighting the PAS and posterior embryotoxon c) left eye with no abnormal findings

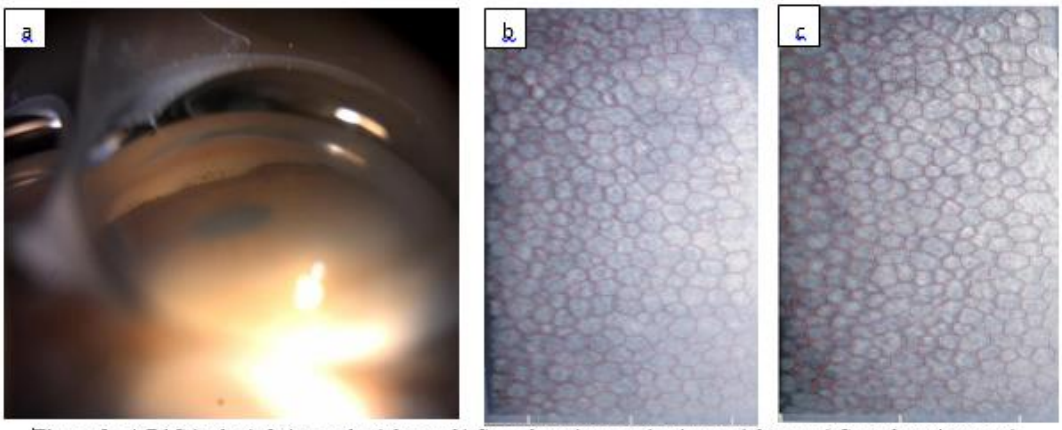

Figure 2: a) PAS in the inferior angle right eye b) Specular microscopic picture right eye c) Specular microscopic picture left eye

III. Discussion

Axenfeld Rieger Syndrome is a condition which arises due to abnormal neural crest cell migration and differentiation with changes in peripheral cornea, iris and anterior chamber angle with or without systemic abnormalities.[1,6] Most common systemic abnormalities include mild facial dysmorphism (maxillary hypoplasia, hypertelorism, broad flat nose, mandibular prognathism), abnormal dentition (microdontia, hypodontia and oligodontia or cone- shaped teeth, crowded teeth) and redundant periumbilical skin.[1,4,7,8] There were no such systemic findings in our patient.

The ocular involvement in ARS is usually bilateral, but it may be asymmetric and, rarely, unilateral. In our patient

Figure 3: a) and b) Front view of face c) periumbilical skin

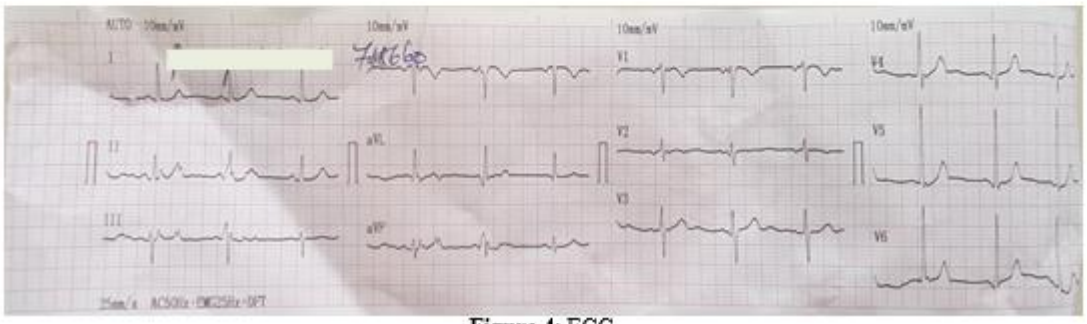

Figure 4: ECG

however, ARS presented unilaterally. Shields[9] postulated that the developmental arrest late in gestation results in primordial endothelium being retained over parts of the iris and anterior chamber angle. Posterior embryotoxon is produced due to abnormal activity of this layer in the angle and its contraction causes iris stromal thinning, corectopia, distortion of pupil, ectropion uvea and hole formation.. In a typical ARS, iridocorneal adhesions bridge angle from peripheral iris to at or near the prominent ridge. Corectopia if present occurs in the direction of these strands and hole formation and iris thinning occurs in the quadrant away from direction of corectopia.[4]

Posterior embryotoxon is seen in 8-15% of normal population, and may not be seen in ARS.[9] However when posterior embryotoxon is identified in a patient with an anterior segment disorder, the first consideration should be ARS. Absence of other corneal abnormalities, such as megalocornea, sclerocornea and corneal opacity can help indistinguishing ARS from other anterior segment disorders.[10]

Our case showed a post embryotoxon in 7 clock hours from 3'0 clock to 10'0 clock and mild corectopia with the pupil displaced slightly inferiorly towards the PAS in the right eye only. Left eye did not show any abnormal findings.

Age of presentation in ARS is commonly childhood or early adulthood but it can occur rarely in late adulthood too. In a case series of 24 patients by Shields et al, average age at which patient was seen by the authors was 26.2 years (range 6 weeks- 71 years) and 58% of them developed glaucoma.[4] In another study by Reis et al done with 128 ARS patients , average age of patients was 20 years (range 2 weeks- 66 years) and 69 percent of them developed glaucoma.

84.5 percent had systemic findings.[8] In our case, the ARS has been detected in late adulthood at 50 years of age whichis uncommon. Patient did not develop any glaucoma and there were no systemic abnormalities.

A close differential diagnosis of this case would be Iridocorneo-endothelial syndrome as it presents unilaterally, there is no family history in this patient and she has presented in late adulthood. Changes in the iris and peripheral anterior synechiae maybe seen in both the conditions. However posterior embryotoxon is a feature of ARS and is rarely seen in ICE syndrome[11.] Secondly, specular microscopic features of ICE syndrome which include loss of hexagonality, pleomorphism in shape and size and darkening areas within the cells are also not there in this patient.[12]

Genetic counselling can help in risk assessment and prevention of this syndrome. Patients with ARS can

have multiple ocular and systemic problems. They need neurodevelopmental evaluation, screening echocardiogram, brain imaging,and hearing tests and thus an interdisciplinary approach to manage effectively.

IV. Conclusion

Axenfeld Rieger syndrome is a bilateral developmental disorder which can be associated with various systemic anomalies. Iris abnormalities may progress with continued contraction of membrane and these patients may develop glaucoma in late adulthood too. They need to be followed-up regularly for early diagnosis and treatment of glaucomawith IOP monitoring and optic nerve head examination.

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