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Case Report

Axenfeld Rieger Syndrome-A Rare Syndrome Revisited

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Abstract

Axenfeld Rieger Syndrome is an anterior segment dysgenesis accompanied by systemic abnormalities. We report a case of 17 years old male with iris coloboma in right eye and anteriorly displaced Schwalbe's line in both eyes. Intraocular pressure was raised in both eyes. Patient was diagnosed case of ventricular septal defect (VSD) with patent ductus arteriosus (PDA) for which he was operated six years back. On physical examination he had scoliosis and on Oro dental examination maxillary hypoplasia was seen. Patient was diagnosed as Axenfeld Rieger Syndrome with glaucoma and was treated with antiglaucoma drugs. This article aims to discuss the clinical features and management of this rare congenital anomaly.

Keywords: Axenfeld Rieger Syndrome; Patent Ductus Arteriosus (PDA); Ventricular Septal Defect (VSD)

Introduction

Axenfeld Rieger Syndrome is a bilateral developmental disorder of eyes with autosomal dominant or sporadic inheritance. In 1920 Axenfeld described posterior embryotoxon of the cornea [1]. Reiger described anterior segment abnormalities with iris atrophy, iris hole formation, corectopia. In Rieger syndrome there is Rieger anomaly with extraocular malformations like hypodontia, microdontia, maxillary hypoplasia, broad nasal bridge, telecanthus, hypertelorism, paraumbilical skin and hypospadias [1].

Case Report

We report a case of 17 years male with complaints of diminution of vision right eye for six years. There was no history of redness, photophobia, watering, and trauma. Patient was operated for VSD with PDA six years back. Family, drug, personal, and socioeconomic history were not significant. On general physical examination patient had low set ears, long thin digits with syndactyly of both hands and both feet shows overriding of toes. There was prominence of left infrascapular region due to scoliosis (Figure 1 and 2). On auscultation of cardiovascular system S1S2 was normal.

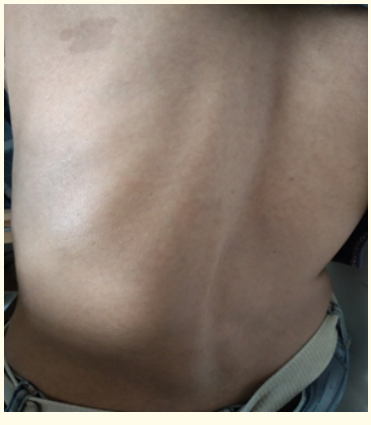

Figure 1: Showing scoliosis.

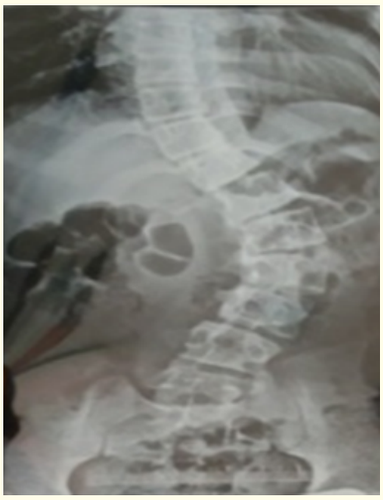

Figure 2: X ray PA view showing scoliosis.

On ocular examination visual acuity in right eye was 6/36 and left was 6/9 with pin hole. Pupillary reaction was normal in both eyes and ocular movements were also normal. On anterior segment examination iris coloboma with ciliary staphyloma was present in right eye. There was megalocornea with horizontal diameter 14 mm and vertical diameter 13.5 mm in both eyes. Posterior embryotoxon was present in both eyes (Figure 3 and 4). On Gonioscopy prominence of Schwalbe ring with insertion of the pectinate strands which were running from the anterior surface of the iris to this ring was seen bilaterally.

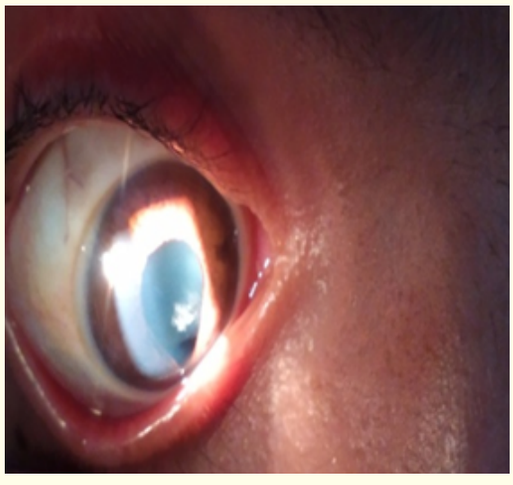

Figure 3: Posterior embryotoxon of the right eye.

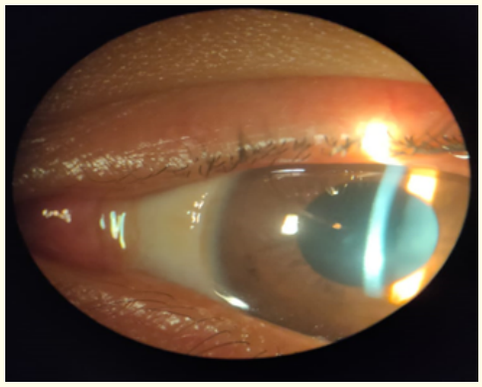

Figure 4: Posterior embryotoxon in left eye.

Axial length of right eye was 27 mm and left eye was 29 mm. Intraocular pressure was 22.7 mm of hg in right eye and 24 mm of hg in left eye. Central corneal thickness in right eye was 593 mm with corrected IOP 18.7 mm of hg and in left eye was 599 mm with corrected IOP20.2 mm of hg. Patient was started on antiglaucoma drug timolol.

Posterior segment examination of fundus shows tilted disc with optic cup disc ratio 0.6 in both eyes. OCT of right eye shows thinning of retinal nerve fibre layer in all quadrants and OCT of left eye shows thinning of retinal nerve fibre layer in inferior quadrant (Figure 5).

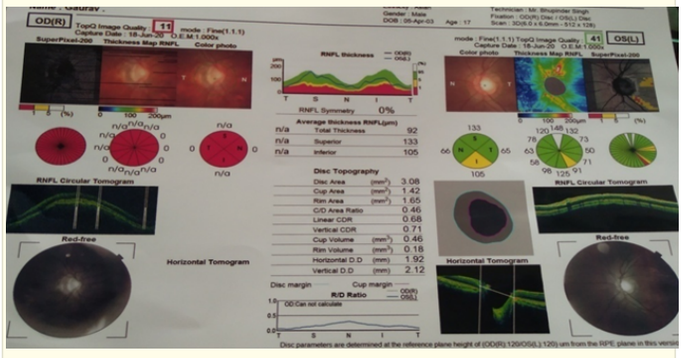

Figure 5: OCT of both eyes.

Perimetry of both eyes shows paracentral scotomas (Figure 6 and 7).

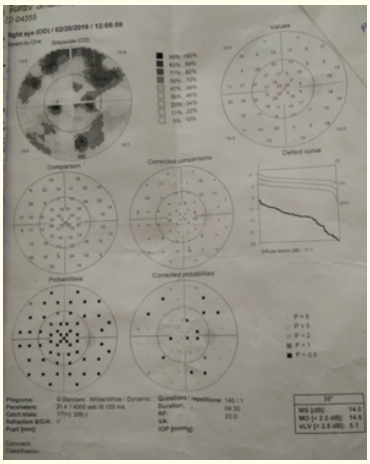

Figure 6

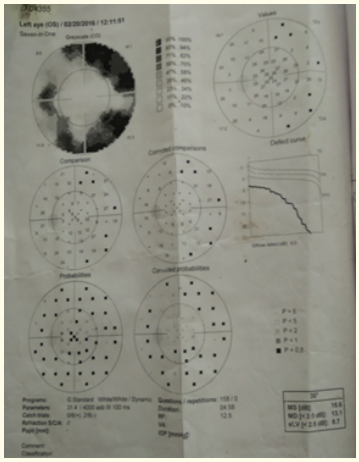

Figure 7

Discussion

Posterior embryotoxon (PE) is a congenital abnormality in which a grey white, line is present on the posterior surface of the cornea, near the limbus. It is an anteriorly displaced Schwalbes line [3]. PE is seen in association with anterior segment abnormalities, like Peters anomaly, Axenfeld–Riegers syndrome, Alagille's syndrome. The genetic mutations in PITX2 and FOXC1 are associated with Axenfeld-Rieger syndrome and anterior segment dysgenesis [3,4].

In Axenfeld-Rieger syndrome ocular features are bilateral and include mild stromal thinning of iris, severe iris atrophy with hole formation, ectropion uveae, corectopia, iridocorneal adhesions. The pathophysiology of Axenfeld-Rieger syndrome is that developmental arrest of anterior segment structures derived from neural crest cells occurs. The neural crest cells also give rise to mesenchyme related to forebrain, pituitary gland, bones, cartilages of the upper face and dental papillae, spinal ganglia, walls of aortic arches, long bones and melanocytes [5,6].

Schlemm's canal is rudimentary or absent and there is incomplete maturation of trabecular meshwork which leads to inadequate drainage of aqueous resulting in glaucoma [7]. Glaucoma appears in childhood or early adulthood due to an associated angle anomaly or secondary synechial angle closure. Glaucoma is managed medically with antiglaucoma drugs. If medical therapy fails to control intraocular pressure surgery like trabeculectomy and aqueous shunts may be required [8].

Conclusion

It is therefore imperative to carry out detailed systemic examination and essential investigations along with ophthalmological examination in patients with posterior embryotoxon. It is also of paramount importance to screen these patients for glaucoma in early childhood.

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