A RARE CASE OF AXENFELD-RIEGER SYNDROME

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ABSTRACT: Axenfeld-Rieger syndrome is an uncommon autosomal dominant condition characterized by anterior segment dysgenesis and systemic abnormalities. We report a case of Axenfeld Rieger syndrome associated with secondary developmental glaucoma who remained undiagnosed since childhood. Twenty-one year old male patient presented with progressive dimness of vision and coloured halos. Slit lamp examination showed scleral thinning and megalocornea in both the eyes and iris hypoplasia in the right eye only. Posterior embryotoxon, few tissue bands, high insertion of the iris roots and anterior synechiae were seen on gonioscopy. Intraocular pressure was 32mmHg in the right eye and 38mmHg in the left eye. Glaucomatous disc changes were observed in both eyes on fundus examination. Systemic features such as maxillary hypoplasia, oligodontia, microdontia, taurodontism, micrognathia and redundant periumbilical skin were observed.

KEYWORDS: Axenfeld-Rieger Syndrome, Anterior segment Dysgenesis, Systemic abnormalities, Developmental glaucoma.

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INTRODUCTION: Axenfeld-Rieger syndrome is thought to have an autosomal dominant pattern of inheritance and is associated with a high incidence of glaucoma. It is characterized by ocular anterior segment anomalies such as prominent anteriorly displaced Schwalbe's line, occasional megalocornea, tissue strands in the anterior chamber angle, iris defects ranging from mild stromal thinning to marked atrophy with hole formation, corectopia and ectropion uveae. The systemic anomalies are developmental defects of the teeth, facial bones and other associated abnormalities like redundant periumbilical skin.

CASE REPORT: A twenty-one year old male patient reported to the ophthalmology outpatient department with complains of gradual progressive diminution of vision more in the right eye and coloured halos in both eyes on and off since childhood. The patient had visited several doctors in the past for the same complaints. He was prescribed refractive correction which he did not use regularly. He did not give history of any medical treatment.

There was no history of redness, watering, photophobia, diplopia or any other ophthalmic complains.

Patient gives history of beedi smoking since five years. No significant family history present.

On systemic examination, maxillary hypoplasia, microdontia, oligodontia, micrognathia and redundant periumbilical skin were noted.

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On ophthalmic examination,best corrected vision in the right eye was 20/80 and left eye was 20/40p.Superior scleral thinning was observed in both the eyes. Megalocornea with a horizontal white to white diameter of 14 mm was measured in both eyes with vernier calliper. Posterior embryotoxon was seen in both eyes.Left eye showed a superior ring shaped stromal opacity as seen on slit lamp examination. Central corneal thickness was 544 microns and 668 microns in the right eye and left eye respectively. Both eyes showed deep anterior chamber with no contents. Anterior iris stromal hypoplasia was seen at 3o'clock position in the right eye. A-Scan indicated an axial length of 27 mm in both eyes. Intraocular pressure (IOP) was 32 mmHg in the right eye and 38 mmHg in the left eye by applanation tonometry. Gonioscopy revealed prominent Schwalbe's line, anterior insertion of the iris, anterior synechiae at places in both eyes and a few tissue bands in the angle in the superior quadrant of the left eye. Fundus examination showed 0.8 cup to disc ratio with superior neuroretinal rim thinning and bayonetting vessels in both eyes. 30-2 single field analysis SITA standard perimetry shows right eye superior and inferior arcuate field defects and left eye early temporal field changes.

Based on the above findings, our diagnosis was Axenfeld-Rieger Syndrome associated with developmental glaucoma.

The patient was started on eye drops travoprost and eye drops containing a combination of brimonidine and timolol and spectacle glasses were prescribed. After two weeks, the IOP was 14 mmHg in the right eye, 22 mmHg in the left eye by applanation tonometry and the patient was symptomatically relieved.

The patient remains under close follow up.

DISCUSSION: A developmental arrest of neural crest cells occurring late in gestation plays an important role in the pathological mechanism of Axenfeld Rieger syndrome.[1,2] There is abnormal retention of the primordial endothelial layer on portions of the iris and the anterior chamber angle which leads to the iridocorneal tissue strands whereas contraction of the tissue layer on the iris leads to iris defects.[3] Maldevelopment of the cells also leads to high insertion of the iris roots due to which there is incomplete maturation of the trabecular meshwork and Schlemm canal leading to glaucoma.[1] The neural crest cells give rise to corneal stroma, Schwalbe's line, iris stroma, trabecular meshwork and Schlemm's canal and also the bones and cartilages of the upper face and dental papillae.[1,2] More than half of the patients with Axenfeld Rieger syndrome develop glaucoma.[1] One of the main ocular features seen is posterior embryotoxon and also tissue strands from the peripheral iris to the Schwalbe's line.[4]

CONCLUSION: Axenfeld-Rieger syndrome is an uncommon condition that holds a risk of gradual and irreversible vision loss due to glaucoma and corneal opacity. All the ocular and non-ocular abnormal features that were seen were present due to maldevelopment of the neural crest cells. Therefore a thorough examination should be done of all the structures derived from neural crest cells. Patients with iris hypoplasia should be investigated for glaucoma and a long term follow up should be kept.

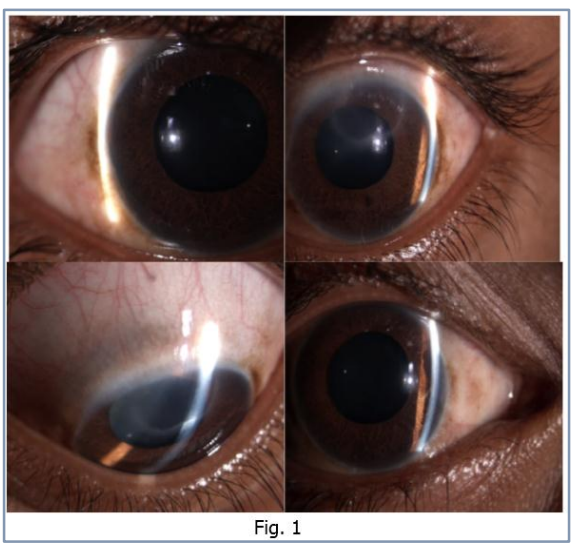

Fig. 1

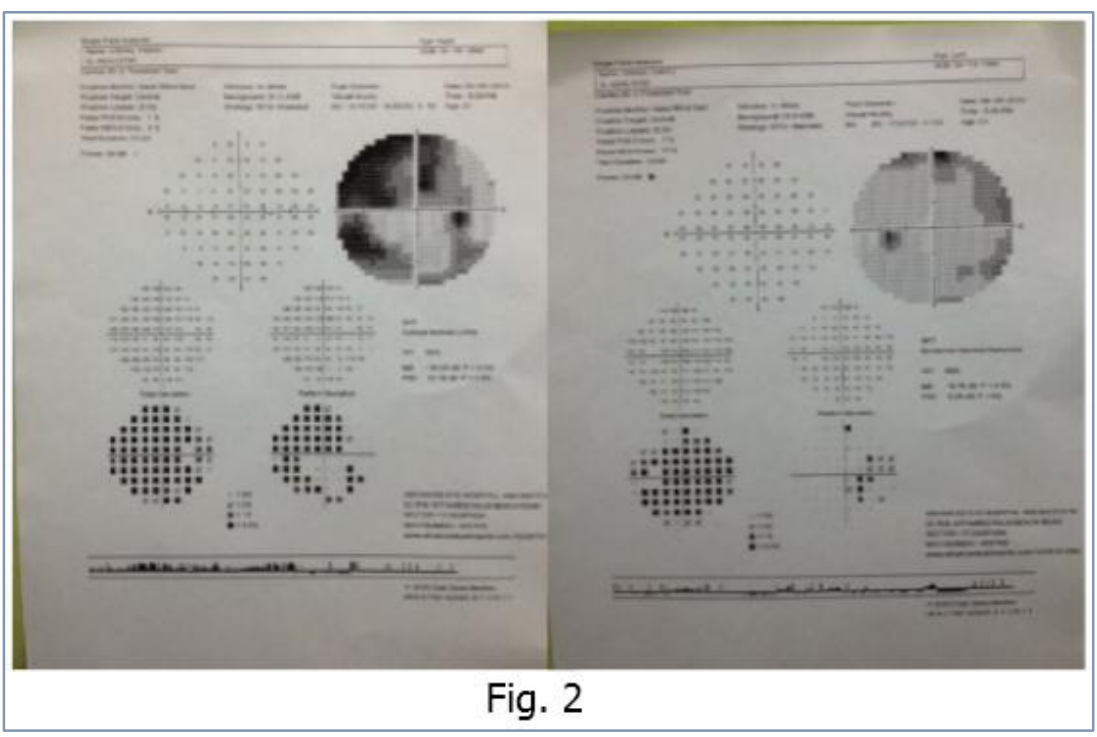

Fig. 2

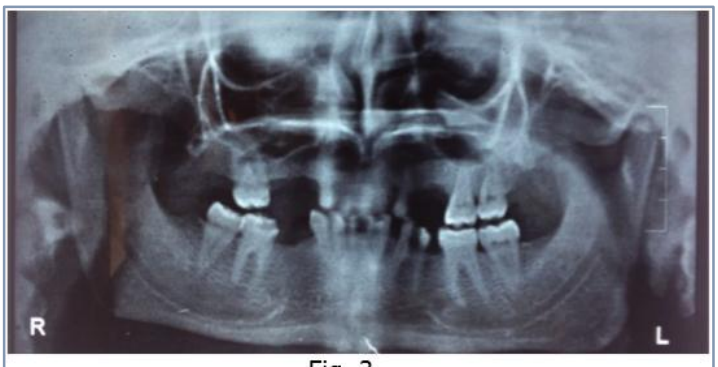

Fig. 3

REFERENCES:

1. Shields MB, Buckley E, Klintworth GK, Thresher R. Axenfeld-Rieger syndrome. A spectrum of developmental disorders. Surv Ophthalmol. 1985: 29: 387-409. [PubMed]
2. Ozeki H, Shirai S, Ideka K, Ogura Y. Anomalies associated with Axenfeld Rieger Syndrome. Graefes Arch Clin Exp Ophthalmol 1999:237:730-4.
3. Shields MB. Axenfeld-Rieger syndrome: A theory of mechanism and distinctions from the iridocorneal endothelial syndrome. Trans Am Ophthalmol Soc.1983: 81: 736-84. [PubMed]
4. Sim KT, Karri B, Kaye SB. Posterior embryotoxon may not be a forme fruste of Axenfeld-Rieger's Syndrome. J AAPOS 2004: 8: 504-6. [PubMed]