Abstract: Axenfeld Rieger anomaly is a developmental disorder of the structures of neural crest origin, both ocular and extraocular. 3 case reports of Axenfeld Rieger anomalies and its association with glaucoma were discussed here.

Keyword: Axenfeld Rieger syndrome, posterior embryotoxon, glaucoma, anterior chamber cleavage syndrome, correctopia

Introduction: Axenfeld Rieger anomaly is a developmental disorder of the structures of neural crest origin, both ocular (bilateral involvement of the anterior segment) and extraocular (teeth and facial bones). It has an overall incidence of 1 in 2000 people worldwide. It is an autosomal dominant disorder with a variety of chromosomal aberrations including deletions in the region of chromosome 4q25, partial trisomy 16q, chromosome 13 deletion, or partial duplication of the short arm of chromosome 2.

Case Report 1: Six year old child born of non-consanguinous marriage was brought by his parents with complaints of defective vision and headache since childhood. On general examination, no evidence of any congenital anomalies found and systemic examination was absolutely normal. On ocular examination, in the right eye, visual acuity was 6/24 with +7.00D sph 6/6, IOP by applanation was 28 mm of Hg, anterior segment-posterior embryotoxon present. Fundus examination was normal with CDratio of 0.3. Automated perimeter showed field defects in the superior arcuate areas. In the left eye, visual acuity was 6/24 with +8.00D sph 6/6, IOP by applanation was 22 mm of Hg, anterior segment-posterior embryotoxon present. Fundus examination was normal with CDratio of 0.3. Automated perimeter showed field defects in the paracentral areas. UBM BE showed narrow angles with PAS. He was diagnosed as a case of axenfeld anomaly BE. He was initially started on betaxalol and then as the field changes progressed in the right eye, travoprost was added. Left eye was stable, under medical control. As the field defects were progressing along with raised IOP in the right eye, RE Trabeculotomy with Trabeculectomy was done. Post-operatively, after 2 months, IOP by applanation was 18 mm of Hg in the right eye and 16 mm of Hg in the left eye. IOP in the right eye was under surgical control and in the left eye was under medical control.

Case Report 2: Ten year old child born of non-consanguinous marriage has come with her parents with complaints of defective vision since childhood. General and systemic examination was normal. Ocular examination in the right eye showed visual acuity of 6/9 ph 6/6, cornea haziness with posterior embryotoxon, IOP of 30 mm of Hg, CDratio of 0.7 to 0.8, narrow angles with multiple iris processes in gonioscopy. Left eye showed visual acuity of 6/6, corneal haziness with posterior embryotoxon, correctopia, IOP of 30 mm of Hg, CDratio of 0.8, narrow angles with multiple iris processes in gonioscopy. Automated perimeter showed fixation defects in both eyes. UBM BE showed narrow angles with peripheral anterior synechiae (PAS) and multiple iris processes. He was diagnosed as a case of Rieger’s anomaly BE. Patient was started on betaxalol and dorzolamide eye drops for both eyes. IOP was under medical control now.

Case Report 3: Twenty one year old female came with h/o defective vision LE since childhood, headache on & off since 6 months, pain BE since 15 days. She had an orthognathic surgery for prognathism done 2 yrs ago. On general examination, hypodontia, microdontia, umbilical hernias were seen. Systemic examination was normal. In the right eye, visual acuity was 6/12 ph 6/9, IOP was 34 mm of Hg, anterior chamber showed variable depth, pupil was eccentric, reacting to light normally. CDratio was 0.8, angles were narrow in gonioscopy. In the left eye, visual acuity was CF CF, IOP was 36 mm of Hg, anterior chamber showed variable depth, pupil was eccentric, correctopic, reacting to light normally. Fundus view was very hazy, angles were narrow with PAS formation in gonioscopy.
B scan showed low intense echoes seen in vitreous cavity s/o vitreous degenerations. UBM RE showed normal central anterior chamber depth and shallow peripheral anterior chamber depth d/t broad based 360 degree PAS and LE showed superior half of iris developed with stretch holes and inferior half of iris developed as stump like pattern with broad based PAS. He was diagnosed as Axenfeld Rieger syndrome BE. Patient was started on timolol and dorzolamide eye drops for both eyes and IOP is under medical control.

**DISCUSSION**

Axenfeld Rieger syndrome is a spectrum of disorders including a) Axenfeld anomaly, b) Rieger anomaly, c) Rieger syndrome. Gene loci has been mapped to 4q25 (PITX2), 6p25 (FKHL7) and 13q14 (RIEG2). The following features are shared by all the three groups:

- Bilateral developmental ocular anomalies which may not be necessarily symmetrical.
- Frequent family history with autosomal dominant inheritance.
- No gender predilection.
- Associated glaucoma.

**Axenfeld Anomaly**

Posterior embryotoxon with attachment of strands of peripheral iris tissue.

**Rieger Anomaly**

Posterior embryotoxon
- Iris stromal hypoplasia
- Ectropion uveae
- Corectopia and full thickness iris defects
- Broad based nasal bridge, telecanthus, and hypertelorism.

**Axenfeld Rieger Syndrome**

Rieger anomaly and the following features are present:
- Dental-hypodontia (few teeth) and microdontia (small teeth),
- Facial-maxillary hypoplasia,
- Broad based nasal bridge,
- Hypertelorism.

**MANAGEMENT**

Infancy-guarded filtration surgery. Childhood & young adulthood-medical therapy tried, but surgical therapy (shunt procedures, trabeculotomy with trabeculectomy, trabeculectomy with antimetabolites) needed later.

**CONCLUSION**

Axenfeld Rieger anomaly and syndrome is a developmental disorder associated with congenital glaucoma and a frequent family history with autosomal dominant inheritance. It is a very rare disorder with an incidence of 1 in 2000 people, with 50% of them associated with congenital glaucoma. Management of these cases are also proved to give successful results. Such cases, who were managed successfully in our hospital were discussed. So when any patient with systemic anomaly reports, it is wiser to rule out congenital glaucoma in these patients, thereby reducing the blindness rate in our society.

**REFERENCES:**

gonioscopy of patient 2- narrow angles with PAS.

patient 2 UBM-narrow angles with PAS

patient 3-UBM-narrow angles with iris stump

patient 3 profile picture-microdontia, hypodontia.

patient 3- umbilical hernia.

patient 3-corectopia with iris atrophy