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A Rare Case Series on Axenfeld - Rieger Syndrome in Mother and Daughter without Glaucoma

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Abstract: ***Description of clinical findings of a rare case of axenfeld reiger syndrome spotted ina mother and daughter, with normal IOP. Rare autosomal dominant pattern Axenfeld Reiger syndrome seen in a 31 year old mother - Both eyes having microcornea, normal intraocular pressure with Right eye cornealopacity, iridogoniodysgenesis with polymegathism and pleomorphism on specular microscopy and left eye iridogoniodysgenesis with Dshapedpupil, dental findings showing hypoplastic maxilla and malocclusion which was dentally rehabilitated.8 year old daughter has both eyes normal IOP, right eye posterior embryotoxon and correctopia and left eye posterior embrotoxon, atrophic iris andcorrectopia with dental findings of malocclusion and hypoplastic maxilla.***

Keywords: iridogoniodysgenesis, axenfeld – reiger syndrome, Glaucoma, Glaucoma syndromes, iris abnormalities

1. Introduction

Failure of the embryonic mesenchyme to differentiate into corneal endothelium, iris, angle structures, and crystalline lens leads to a spectrum of abnormalities collectively called Axenfeld - Rieger anomaly.

It is bilateral, having autosomal dominant inheritance, associated genes - FOXC1, PITX2. Axenfeld anomaly - an anteriorly displaced and prominent Schwalbe's line (posterior embryotoxon) with some peripheral iris processes. Rieger's anomaly – mid peripheral iris adhesions to the cornea, iris abnormalities, which can include marked hypoplasia of the anterior iris stroma as well as pupillary abnormalities, such as distortion of the pupil, polycoria, and corectopia. Microcornea or macrocornea may be there.

Fifty per cent of those with Axenfeld - Rieger cases develop glaucoma in time probably related to the anterior segment dysgenesis Many are associated with skeletal and dental abnormalities.

2. Materials and Methods

The pair of patients were identified in the clinic during their routine eye checkup, vision and refraction was taken with snellens chart at 6 meters, slit lamp biomicroscopy done with gonioscopy examination and imaging of the same, gross clinical photos were taken

The mothers past records were also documented.

3. Case Description

Hereby presenting two case reports of a mother and daughter diagnosed with Axenfeld reiger anomaly who had presented to the ophthalmology clinic for routine eye examination.

Case - 1 - Mother

A 31 year old female presented with right examination eye loss of vision since childhood associated with whitish discolouration of the right eye. She had no history of any past treatments taken for the same, although she had consulted eye doctors without relief. She has past history of dental malocclusion for which she had taken treatment 12 years ago. No other significant systemic diseases or treatments taken in then past She had a flat bridged nose in examination. On examination she had right eye absent perception of light and right eye best corrected vision with snellens chart at 6 meter was 6/12 with a spherical power of - 3.25 and cylinder of - 2.00 at 125 degree. Near vision of right eye with snellens chart at 33cm was more than N36 in right eye and N6 (plano) in left eye. intraocular pressure measured with Goldmans applanation tonometer was 15 mmhg in left eye and right eye couldnt be measured due to the scar. Right eye on slit lamp showed adherent leucoma involving roughly temporal half of the cornea, with vascularisation with involvement of visual axis, and iris was adherent on to the scar showing no pupil in the visible area, explaining her vision loss. USG b scan was done which showed normal findings with anechoic vitreous and an attached retina. Ultrabiomicroscopy of right eye showed thick irregular cornea, thin atrophuic iris, marked zonular stretching from 6 - 11 o clock, with normal lens, posterior capsule and ciliary body. Left eye showed a temporal posterior embryotoxon, clear cornea, iris atrophy, d shaped pupil and a clear lens. Fundus examination on indirect ophthalmoscopy with 20 d condensing lens showed a normal fundus with 0.3 disc diameter, normal sized disc, pink in colour with healthy neuroretinal rim and normal macula. Specular microscopy of left eye shows reduced endothelial cell count, marked pleomorphism and polymegathism with few guttatea.

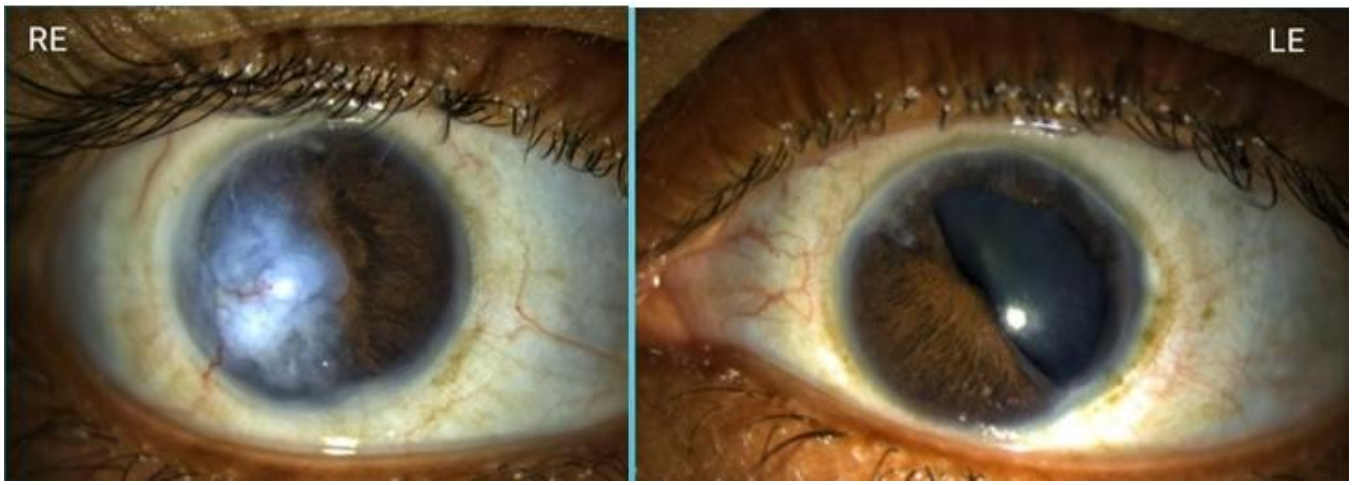

Figure 1 and 2: Gross slit lamp image of right and left eye of the mother

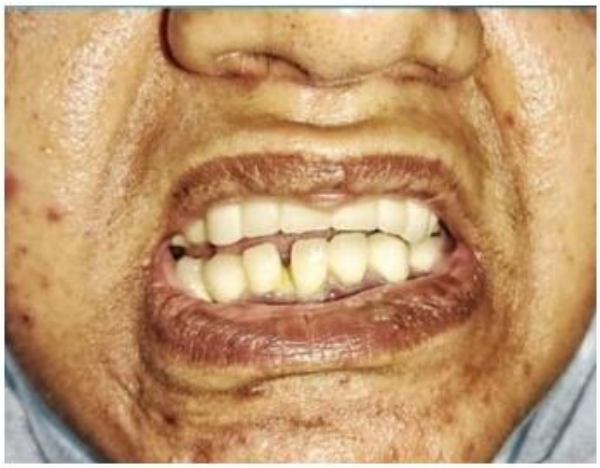

Figure 3: Dental profile of the mother and depressed nasal bridge

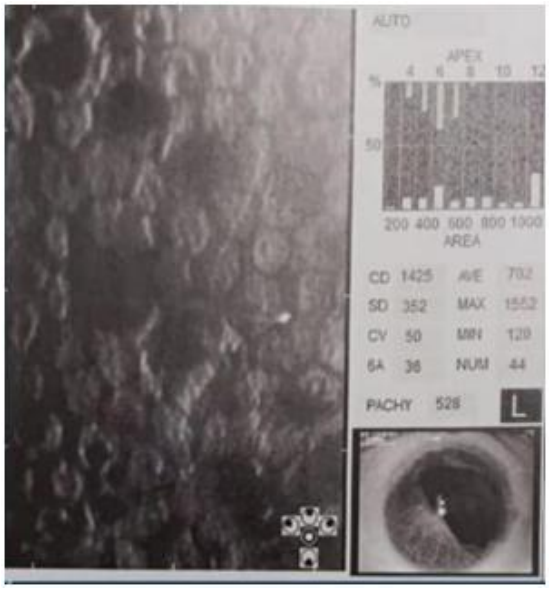

Figure 4: Specular microscopy of left eye of mother

Case 2 - Daughter

7 year old female presented with both eye dimunition of vision. She had no other ocular complaints. Systemic diseases or any treatment history was not present. She had dental malocclusion, hypoplastic maxilla and abnormal bite. Tested at 6m with snellens visual acuity chartHer best corrected vision was right eye 6/12 (- 4.25 DS and - 0.5DC at 160 degree) and left eye 6/9 (- 5/5 DS and - 1.75 Dc at 180 degree), Near vision of both eyes with snellens chart at 33cm was N6 (plano). Here we can see she has high degree of astigmatism with myopia. IOP checked with goldmanns applanation tonometer was right eye 13 mmhg and left eye 14 mmhg. On examination, right eye showed posterior embryotoxon temporally, temporal correctopia, iris atrophic pach, There was no view of fundus, although USG Bscan done showed normal findings. Left eye showed temporal and nasal posterior embryotoxon, iris atrophy, strands of iris directed temporally. Fundus examination showed 0.2 CDR Cup with normal size, pink colour and healthy neuroretinal rim. Both eye gonioscopy shows iris strands adherent over schwalbes line

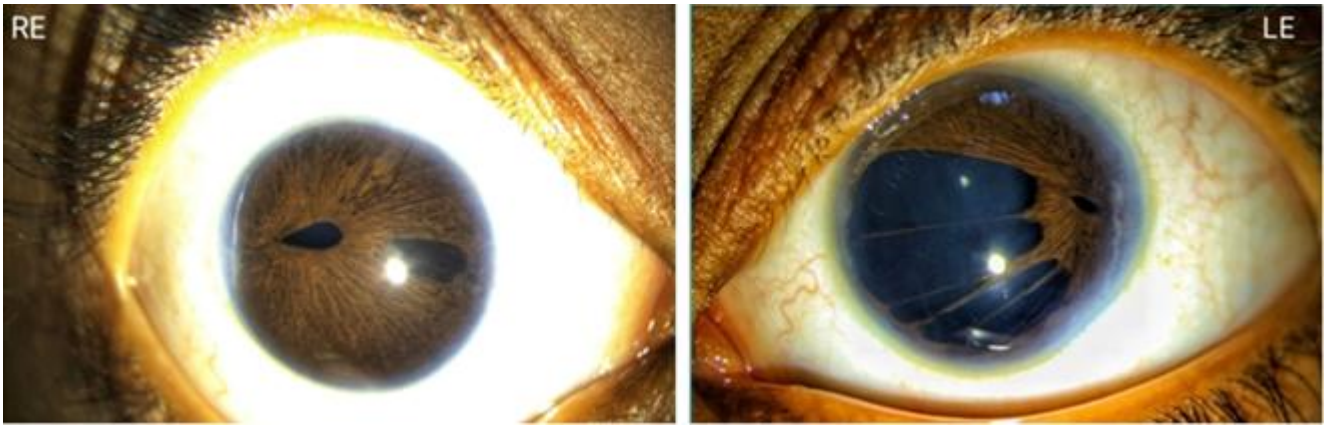

Figure 5, 6: Gross slit lamp image of right and left eye of daughter

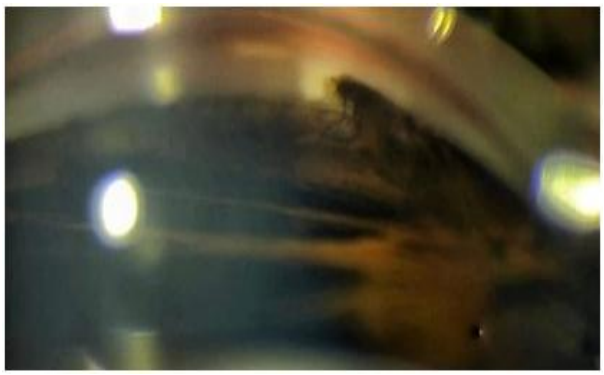

Figure 7: Gonioscopy image of angles with iris processes and strands of iris

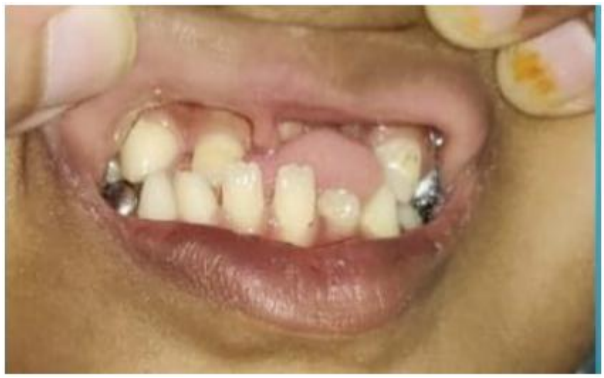

Figure 8: Dental malocclusion in daughter

4. Discussion

Axenfeld reiger syndrome being a congenital disorder, remains rather difficult to treat definitively. Although the above two cases had normal intraocular pressure, if high it is ultimately treated with glaucoma surgery. The cosmetic unappeal is definite and in some cases like above, vision loss due to sequel as like adherent leucomas cause morbidity. The patients were counselled of the genetic nature of the disease, the need to regularly monitor intraocular pressure and routine checkups to salvage remnant vision.

5. Conclusion

The goal was to detect and describe the signs and symptoms in the pair, and to educate the patient the nature of the disease, inheritance pattern and need to monitor IOP, and watch out for signs in the child for better visual prognosis.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient (s) has/havegiven his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identitv. But anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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