Axenfeld-Rieger Syndrome-A rare case

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Abstract

This is a rare case of Axenfeld-Rieger syndrome. Axenfeld-Rieger(A-R) syndrome is an autosomal dominant disease. This is an unusual presentation of combination of both Axenfeld and Rieger's anomaly with systemic associations.

Key words : Axenfeld-Rieger Syndrome, posterior embryotoxon, secondary glaucoma.

Introduction

Axenfeld-Rieger syndrome is an autosomonal dominant disease. They belong to a group of mesodermal dysgenesis, also called as Iridocorneal dysgenesis. These are a group of congenital disorders which involve the cornea, iris and anterior chamber angle. They are frequently associated with congenital glaucoma. These conditions occur as a result of an abnormality in fetal neural crest cells. Common ocular findings in Axenfeld-Rieger's syndrome involve cornea (prominent schwalbe's line), iris abnormalities like (mild atroply to severe degenerative changes like hole formation, corectopia and ectropionuveae) and angle abnormalities like (anterior insertion of iris, tissue strands bridging the angle from peripheral iris to prominent schwalbe's line). Nearly 50% of subjects present with secondary glaucoma[1].

Case report

A 12 year old boy presented to our OPD for routine eye checkup. His visual acuity for distance was 6/9 unaided in both eyes with no pin hole improvement and for near it was N_6. On slit lamp examination, right eye showed prominent schwalbe's line (Posterior embryotoxon), corectopia, and diffuse iris stromal hypoplasia (Figure 1). Intraocular pressure (IOP) in the right eye=29 mmHg with applanation tonometer. Left eye had similar findings and IOP was 25mmHg with applanation tonometer.

Gonioscopy revealed open angles in both eyes with anterior insertion of iris into trabecular meshwork, prominent iris processes and broad based synechiae at places and prominent schwalbe's line. Fundus examination was normal.

Patient was not co-operativefor assessment of visual fields. Topical Brimonidine eye drops one drop twice a day was started initially.

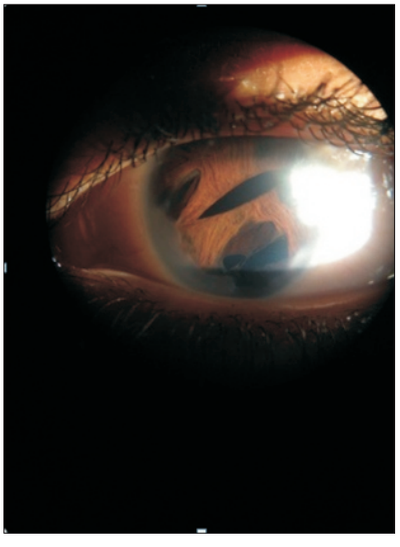

Figure 1. Diffuse stromal iris atrophy withpseudopolycoria.

Other associations were paraumbilical skintag (Figure 2), sensorineural deafness, pituitary abnormalities, dental anomalies likehypodontia (few teeth)/anodontia and microdontia (small teeth) (Figure 3, Figure 4). Facial anomalies include hypoplasia of maxilla broad nasal bridge, telecanthus and hypertelorism (increased inter orbital distance) were present.

Family history was significant. Patient's younger sister presented with similar findings in right eye while the left eye anterior segment was normal. Other associations were paraumbilical skin tags, sensorineural deafness and pituitary abnormalities.

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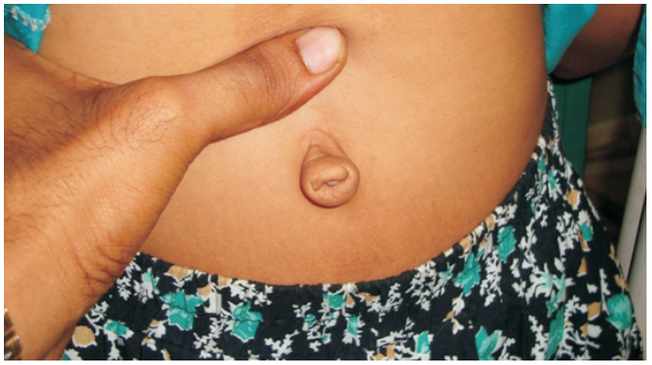

Figure 2. Paraumbilical skin tags

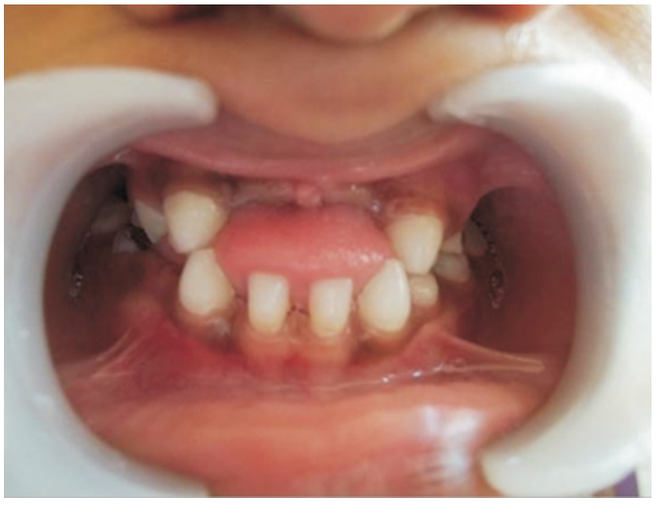

Figure 3. Anodontia in the upper jaw and microdontia in the lower jaw

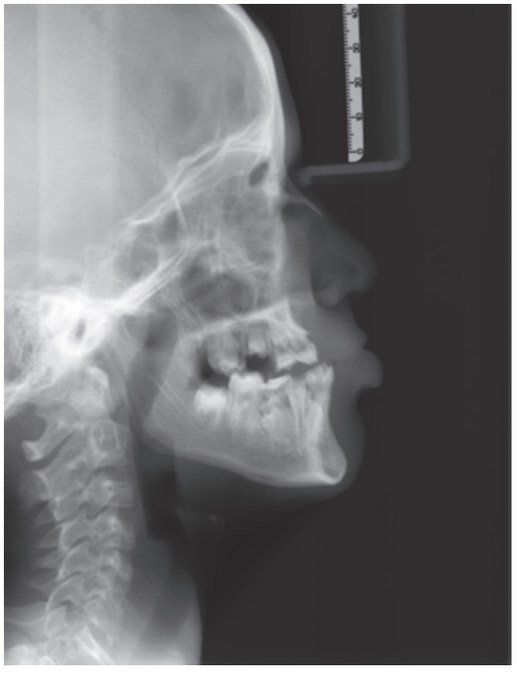

Figure 4. Dental anomalies

Discussion

Patients displaying Axenfeld-Rieger (A-R) syndrome are generally asymptomatic. The condition is diagnosed based upon findings from routine biomicroscopic and gonioscopic evaluation. Historically, this condition was incorporated under the broader heading of anterior chamber cleavage syndromes and included Axenfeld's anomaly, Axenfeld's syndrome, Rieger's anomaly, and Rieger's syndrome[2].Impaired neural crest cell migration and differentiation during embryonic development are considered important in the pathogenesis of A-R syndrome [3].

There are few case reports of Axenfeld-Rieger syndrome. There are only few Axenfeld or only Rieger's anomaly. But this is a rare case report of combination of both Axenfeld and Rieger's anamoly presenting with ocular and non-ocular findings of Axenfeld-Rieger's syndrome. Glaucoma must be a concern in every patient presenting with this disorder. In fact, when glaucoma does occur, it can be quite severe.

Patient's IOP is well controlled with Brimonidine eye drops. We have been periodically monitoring IOP, visual fields and fundus. Any slightest IOP/visual disturbance, then surgical management should be considered, but in our case, after 3 months, patient was lost to follow up.

References

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