Case Report of Anterior Segment Dysgenesis

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Case Report

Abstract: Reporting a paediatric case with anterior segment dysgenesis. The slit lamp evaluation of a 4 yr old male child showed features of bilateral microphthalmos, microcornea, central corneal opacity with alternating esotropia and horizontal jerky nystagmus. Also showed iris atrophy with corectopia. In addition to these left eye had iridocorneal adhesions and cataractous lens. The child had facial features like beak shaped nose, pre auricular tag, microdontia, hypodontia with high arched palate. ECHO showed PDA with left to right shunt. Intraocular pressure was normal. Thus reporting a case of anterior segment dysgenesis with combined features of Riegers syndrome and Peters anomaly.

***Keywords*:** Anterior segment dysgenesis, Axenfeld–Rieger syndrome, Peters anomaly, Microphthalmos, Microcornea, Iridocorneal adhesions, Glaucoma.

Introduction

Anterior segment dysgenesis (ASD) is a failure of the normal development of the tissues of the anterior segment of the eye. It leads to anomalies in the structure of the mature anterior segment, associated with an increased risk of glaucoma and corneal opacity. This congenital anomaly was first described by Albert Peters, a German ophthalmologist. Several different gene mutations have been identified underlying these anomalies with the majority of ASD genes encoding transcriptional regulators. The emerging view is that the genes PITX2 and FOXC1 act in concert to specify a population of mesenchymal progenitor cells, mainly of neural crest origin, as they migrate anteriorly around the embryonic optic cup. These same genes then regulate mesenchymal cell differentiation to give rise to distinct anterior segment tissues. Thus variation in the gene dosage and in the normal level of transcription factor activity causes a range of anterior segment anomalies.

Case Report

4 year old male child was brought to eye OPD with history of decreased vision since birth. Child follows light. On Hirschberg test it showed 15 degree esotropia. Bilateral horizontal jerky nystagmus was noticed. Slit lamp examination showed B/L conjunctivalisation of cornea, microphthalmos, microcornea and central corneal opacity.

Corneal Measurements

	Horizontal	**Vertical**
Right Eye	9 mm	8 mm
Left Eye	8 mm	8 mm

Iris showed patches of atrophy. Left eye had iridocorneal adhesions and hence anterior chamber depth was irregular. Left eye pupil showed correctopia. Lens showed cataractous changes. Fundus was not visualised due to corneal opacity and lenticular changes. Thus B scan was done and showed no retinal detachment or vitreous disturbances.

Intraocular pressure- right eye – 14mm Hg, left eye – 17 mm Hg (Schiotz tonometry)

General examination

Face: beak shaped nose. **Ear:** pre auricular tag. Left tragus not fully developed. **Oral cavity:** microdontia, hypodontia, high arched palate. All the routine blood investigations- Hb, TC, DC, ESR were normal. ECHO shows PDA (left to right shunt) From the above findings, a provisional diagnosis of anterior segment dysgenesis which showed both features of Riegers syndrome and Peters anomaly was made.

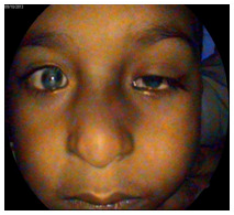

Figure 1: Bilateral corneal opacity

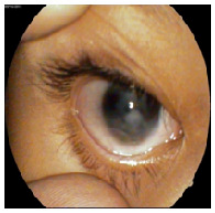

Figure 2: Right eye corneal opacity

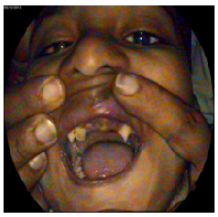

Figure 3: Microdontia with hypodontia

Discussion

Axenfeld–Rieger syndrome: It is a spectrum of disorders consisting of - (a) *Axenfeld anomaly*, (b) *Rieger anomaly* and (c) *Rieger syndrome*.

***Axenfeld anomaly*:** Characterized by Posterior embryotoxon with attachment of strands of peripheral iris tissue.

Rieger anomaly: Characterised by Posterior embryotoxon, Iris stromal hypoplasia, Ectropion uveae, Corectopia and full-thickness iris defects. Glaucoma develops in about 50% of cases, usually during early childhood or early adulthood due to an associated angle anomaly or secondary synechial angle closure

Rieger syndrome: characterized by – Dental anomalies consisting of hypodontia (few teeth) and microdontia (small teeth). Facial anomalies include maxillary hypoplasia, broad nasal bridge, telecanthus and hypertelorism. Other anomalies include redundant paraumbilical skin and hypospadias. Hearing loss, hydrocephalus, cardiac and renal anomalies and congenital hip dislocation are rare.

Peters anomaly

Most cases are sporadic, although AR inheritance and chromosomal defects have been described. It is bilateral in 80% of cases.

Signs: Central corneal opacity of variable density. Underlying defect involving the posterior stroma, Descemet membrane and endothelium with or without iridocorneal or lenticulocorneal adhesions

Ocular associations occasionally present include Axenfeld–Rieger anomaly, aniridia, microphthalmos, persistent fetal vasculature and retinal dysplasia.

Glaucoma occurs in about 50% of cases as a result of an associated angle anomaly in which there is incomplete development of the trabecular meshwork and Schlemm canal. Elevation of IOP is usually evident in infancy but may occasionally develop in childhood or even later.

Systemic associations include craniofacial anomalies, central nervous system anomalies, fetal alcohol syndrome, chromosome abnormalities, and 'Peters plus' syndrome (short-limbed dwarfism, cleft lip/palate and learning difficulties).

Conclusion

To conclude Anterior segment dysgenesis can present with overlapping features of Axenfeld- Riegers syndrome and Peters anomaly. Thus it is not only important for a complete ocular examination to prevent gradual and irreversible visual loss, but also the emphasis is on detailed systemic examination for accurate diagnosis and adequate treatment.

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