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CASE REPORT

RARE ASSOCIATION OF POSTERIOR EMBRYOTOXON WITH MAXILLARY HYPOPLASIA, VENTRICULAR SEPTAL DEFECT, PULMONARY ATRESIA AND PATENT DUCTUS ARTERIOSUS

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ABSTRACT: BACKGROUND: Posterior embryotoxon is a congenital anomaly, considered to be a relatively mild disorder and can occur in 15% of normal eyes. Bilateral posterior embryotoxon associated with maxillary hypoplasia, Ventricular Septal Defect (VSD), Pulmonary Atresia (PA) with Patent Ductus Arteriosus (PDA) is of rare occurrence and hasn't been reported in literature till date. **CASE:** We report a case of 12 year old female who came to us in eye Out Patient Department for routine eye checkup and on detailed ocular examination we found anteriorly displaced Schwalbe's line. On detailed physical examination and investigations she was found to have pan-systolic murmur and continuous machinery murmur. On Echocardiography, the patient was found to have large peri-membranous VSD with PA and PDA. On Oro-dental examination she was found to have maxillary hypoplasia. **CONCLUSION:** The present case is reported due to the rarity and sporadic character of the condition and its rare association with cardiac defect and maxillary hypoplasia.

KEYWORDS: Posterior Embryotoxon, Ventricular Septal Defect, Maxillary Hypoplasia, Neural Crest Cells.

KEYMESSAGE: Any patient with posterior embryotoxon should be thoroughly examined and investigated to rule out other systemic anomalies associated with this condition.

INTRODUCTION: In 1920, Axenfeld first described a gray-white circular line on the posterior surface of the cornea near the limbus in an otherwise normal person.[1] He referred to this abnormality as embryotoxon corneae posterius. This gray-white line was later identified histologically as the prominence of Schwalbe's line.[2]

CASE: A 12 year female attended our Eye Out Patient Department for ophthalmic checkup. She was admitted in paediatric ward for dyspnea on exertion and recurrent chest infections. There was no history of any ocular complaints. Family history and birth history were not contributory. Her weight for age and height for age were 17 kgs (Less than 3rd centile) and 123cms (less than 3rd centile). Her detailed physical examination revealed pan-systolic murmur in left upper para-sternal area associated with thrill and a continuous machinery murmur in pulmonary area in 2nd left intercostal space. Her other systemic examination was normal. On oro-dental examination, she was found to have maxillary hypoplasia (Figure 1).

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Fig. 1: Maxillary hypoplasia

On ocular examination the unaided visual acuity was 6/6 in both eyes. The cornea showed a glassy white annular membrane 1 mm all around the cornea in both eyes (Figure 2-4). Slit Lamp examination revealed it to be situated on the posterior surface of the cornea. Gonioscopy revealed the prominence of Schwalbe's ring with insertion of the pectinate strands which were running from the anterior surface of the iris to this ring. Fundus examination did not reveal any abnormality.

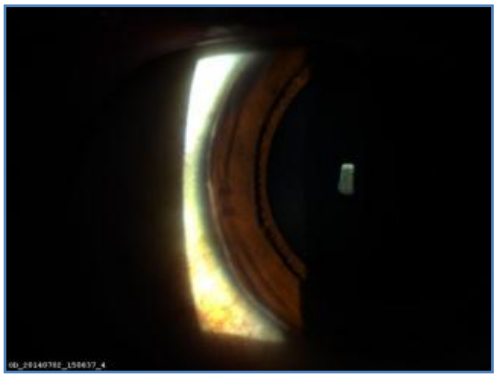

Fig. 2: Right eye showing Posterior Embryotoxon

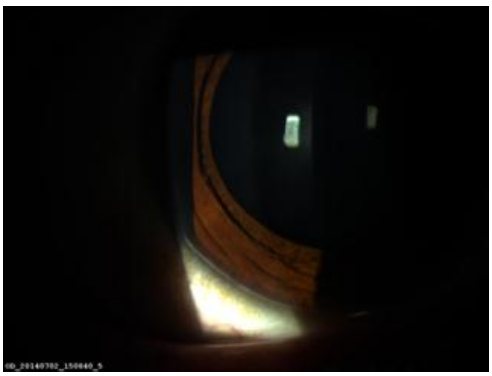

Fig 3: Right eye showing Posterior Embryotoxon

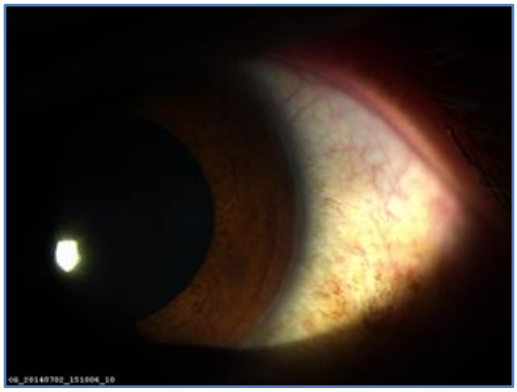

Fig. 4: Left eye showing Posterior Embryotoxon

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Her external ocular examination was normal. She was investigated for glaucoma. Diurnal variation and applanation tonometry did not reveal any change suggestive of the defect.

Her routine laboratory investigations including complete haemogram, liver function tests, renal function tests, lipid profile were all within normal limits. On Echocardiography, the patient was found to have large peri-membranous VSD with PA with PDA. Abdominal ultrasonography, CT scan skull and MRI Spine were normal.

DISCUSSION: Posterior embryotoxon is a congenital anomaly, considered to be a relatively mild disorder and can occur as an isolated defect. However, it can also be detected in association with other ocular and systemic congenital anomalies e.g. Axenfeld-Reiger syndrome, Peters' anomaly, Alagille syndrome and Sclerocornea. Embryologically, posterior embryotoxon is one of the mesenchymal dysgenesis of the anterior ocular segment caused by the abnormal migration of neural crest cells.[3,4] The cardiac and cranial neural crest derivatives are also responsible for the development of craniofacial mesenchyme including bone, cartilage and connective tissues of the face, and the heart outflow tract including aortic arch/pulmonary artery septum, large arteries wall musculoconnective tissue.[5]

As the trabecular meshwork is also derived from the neural crest, it stands to reason that patients with posterior embryotoxon are predisposed to open-angle glaucoma.[6-8] Definite cause of the condition is unknown. However, an autosomal hereditary tendency has been noted with familial occurrence in 2 to 3 Generations.[9-11] The occurrence in this case was sporadic without any other member of the family being affected. It is therefore necessary for the ophthalmologists to do detailed history, examination and necessary investigations to rule out other systemic anomalies associated with posterior embryotoxon.

SUMMARY: This case is unique of its own kind due to rare association of bilateral posterior embryotoxon in the cornea with cardiac anomalies consisting of VSD, PA, PDA and maxillary hypoplasia.

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