CASE REPORT

PETER'S TYPE II ANOMALY ASSOCIATED WITH CONGENITAL HEART DEFECT: RARE CASE REPORT

Darshan Kumar U. Kansara[1], Sunil G. Biradar[2]

HOW TO CITE THIS ARTICLE:
Darshan Kumar U. Kansara, Sunil G. Biradar. "Peter's Type II Anomaly Associated with Congenital Heart Defect: Rare Case Report". Journal of Evidence based Medicine and Healthcare; Volume 2, Issue 10, March 09, 2015; Page: 1565-1568.

ABSTRACT: PURPOSE: Reporting a case of Peters' type II with congenital heart defect a rare form of anterior segment dysgenesis in which abnormal cleavage of the anterior chamber occurs. It is characterized by central corneal opacification secondary to defects in a posterior corneal stroma, Descemet's membrane, and endothelium. Majority of cases are sporadic, bilateral, and associated with other systemic malformations. **MATERIAL AND METHOD:** A18 year old female presented with a history of bilateral cloudy corneas and diminution of vision since early childhood. Visual acuity was RE 6/60, LE 6/60 with PH improvement in BE 6/36. Slit lamp examination showed BE micro cornea with central and paracentral macular grade opacity with lenticulo-irido-corneal adhesions. Iris was found to be fused with the posterior surface of the peripheral cornea with poor formation of the anterior chamber with coloboma of iris. Lens was cataractous. Fundus examination in the left eye showed no abnormality. She was diagnosed as Peters' type II anomaly. Systemic examination revealed congenital heart defects. Echocardiography showed congenital bicuspid aortic valve, grade 3 aortic regurgitation. **RESULT:** Patient require penetrating keratoplasty with combined procedure (Cataract extraction with trabeculectomy). **CONCLUSION:** Early detection of the disease will help in early treatment, with good vision development to combat amblyopia.
KEYWORDS: peters' type II anomaly, anterior segment dysgenesis, microcornea, congenital heart defect.

INTRODUCTION: Peters' anomaly is a rare form of anterior segment dysgenesis in which abnormal cleavage of the anterior chamber occurs. Peters' anomaly may be caused by incomplete migration and differentiation of the precursor cells of the central corneal endothelium and descemet membrane or a defective separation between the primitive lens and cornea during embryogenesis.[1] It is defined as a congenital central corneal opacity with corresponding defects in the posterior corneal stroma, Descemet's membrane, and endothelium leading to Peter's type I. Peters' type II in addition will have lens abnormalities and tend to be bilateral. 60% of those with Peters' anomaly are bilateral. In both forms, opacification of the cornea leads to an amblyogenic effect on a developing infant and carries high risk early onset glaucoma. Peters' anomaly may be associated with other ocular or systemic abnormalities like microphthalmos, colobomata, retinal and optic nerve dysplasia, microcornea, developmental delay, central nervous system defects, craniofacial abnormalities, seizure disorders, genitourinary malformations, and cardiopulmonary defects.[2] Sporadic, autosomal dominant, and autosomal recessive forms have been described.[3]
A small number of cases have been described with mutations in PAX6 gene, RIEG1 gene, and other genes.

CASE REPORT: An 18 year female presented with a history of bilateral cloudy corneas and diminution of vision since early childhood. Parents noticed gradually progressing corneal opacity and diminution of vision in both eyes for distance and for near.

Her ocular examination revealed Visual acuity was RE 6/60, LE 6/60 with PH improvement in BE 6/36. Near visual acuity in BE was N 24.

Slit lamp examination showed BE microcornea (8.4 X 8.6 mm) with central and paracentral macular grade opacity with lenticulo-irido-corneal adhesion (Figure 1 & 2). Whole of the cornea was involved leaving peripheral 2 mm of cornea clear. Bilateral multiple temporal and nasal iridocorneal adhesions could be appreciated with iris colobomapostessorsynechiae (Figure 3). Poorly formed anterior chamber and cataractouslens. IOP measured by applanation tonometry was 20 mm Hg in RE and 18 mm Hg in LE. Gonioscopic examination of the angle revealed ill-defined angle structure over 3/4 of the angle circumference (Figure 4). Fundus examination in the left eye showed no abnormality. A diagnosis of Peter's type II anomaly was made. Systemic examination of respiratory, central nervous system was normal but cardiac examination revealed congenital heart defects. Echocardiography showed congenital bicuspid aortic valve and grade 3 aortic regurgitation.

DISCUSSION: Peters' Anomaly occurs with various ophthalmological findings, mainly iridolenticular contact, keratolenticular contact, cataracts, glaucoma and variations in the global volume.[4]A variant of Peters' anomaly has been described involving the lens. This has been termed as Peter's type II or Peters' anomaly with corneo lenticular contact or cataract.[1]

Most cases of Peter's anomaly are sporadic, although reports of parental consanguinity and more than one affected sibling support an autosomal recessive or irregularly dominant mode of inheritance in some cases. Our patient is sporadic.

Systemic abnormalities include congenital heart defects, hydrocephalus, and renal dysgenesis.[5]

Mesenchymal cells which differentiate into corneal endothelium, stroma, iris, and aqueous outflow structure have been proved to be derived from neural crest cells by histochemicalmethod.[6] Bahn et al suggested that corneal endothelial disorders, Peter's anomaly, congenital glaucoma, posterior embryotoxon, Axenfeld's anomaly, Rieger's anomaly, and sclerocornea result from abnormal neural crest cell migration.[7] Thus, in most cases of Peter's anomaly and of Axenfeld's anomaly the pathogenesis is related to abnormal migration of neural crest cells.

Bilateral Peters' anomaly has a stronger association (71.8%) with systemic malformation.[8] In another study by Mayer.[9] and Heon ET al.[10] published two series of cases with Peters' anomaly. About 60% of cases had non-ocular abnormalities, 25% of the cases had cardiac defects, 30% had CNS abnormalities and an additional 20% had developmental delay. In our case, bilateral micro cornea with central and paracentral opacity with cataract and congenital heart defect was seen.

Patient will require penetrating keratoplasty with combined procedure (Cataract extraction with trabeculectomy) with guarded visual prognosis.

CONCLUSION: Peter's anomaly is a rare disease in ophthalmic practice; however, its early detection and treatment are important to reduce the risk of visual impairment. We believe that removal of the opacity by penetrating keratoplasty and cataract surgery to obtain clear visual axis and improved visual function.

ACKNOWLEDGMENTS: I am very thankful to my patient for kind co-operation. I am very grateful to my staff for their constant support and as well as support from Shri B.M. Patil Medical College and research institute, Bijapur.

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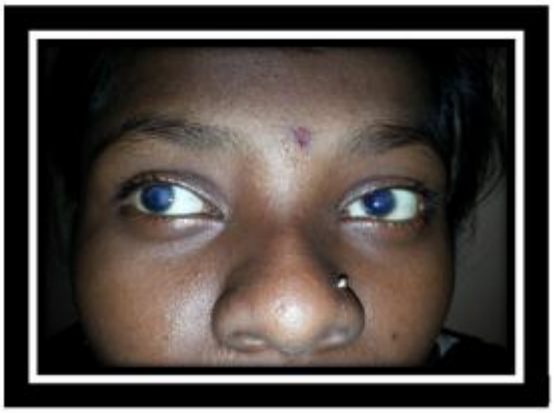

Fig. 1: Bilateral corneal opacity

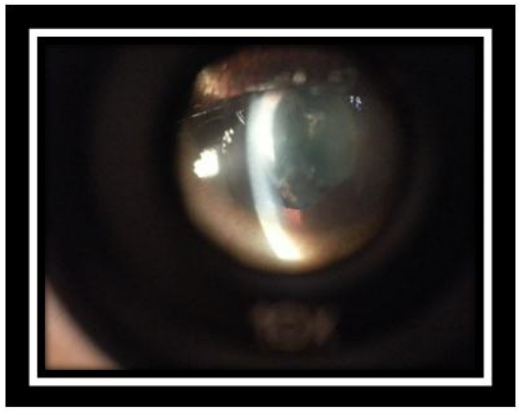

Fig. 2: Lenticulo-irido-corneal adhesions

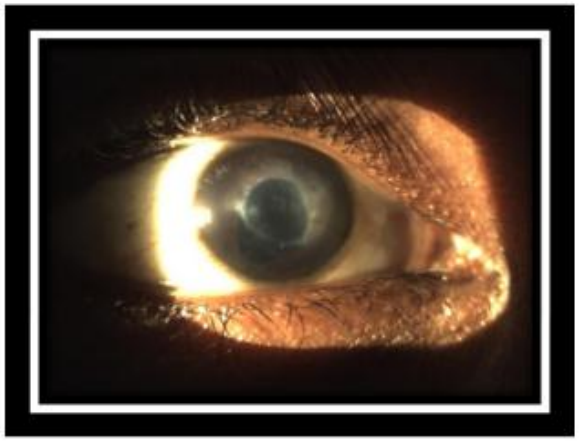

Fig. 3: Iris coloboma with synechiae

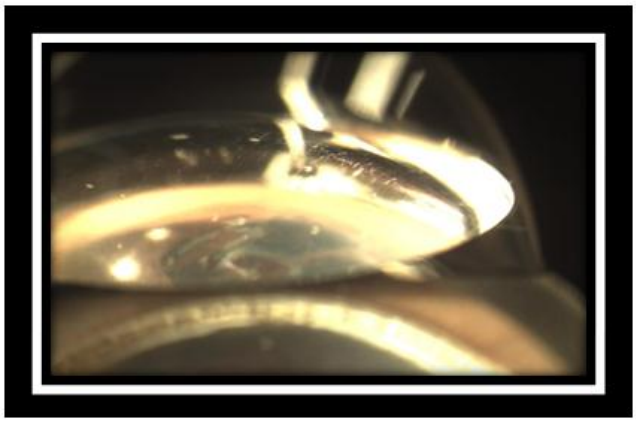

Fig. 4: Ill-defined angle structure on gonioscopy examination

AUTHORS:

1. Darshan Kumar U. Kansara
2. Sunil G. Biradar

PARTICULARS OF CONTRIBUTORS:

1. Post Graduate, Department of Ophthalmology, Shri B. M. Patil Medical College & Research Centre, Bijapur.
2. Professor, Department of Ophthalmology, Shri B. M. Patil Medical College & Research Centre, Bijapur.

NAME ADDRESS EMAIL ID OF THE CORRESPONDING AUTHOR:

Dr. Darshan U. Kansara,
B-27, NRI PG Hostel,
Shri B. M. Patil Medical College
& Research Centre,
Bijapur- 556103.
E-mail: kdarshan83@yahoo.com

Date of Submission: 26/02/2015.
Date of Peer Review: 27/02/2015.
Date of Acceptance: 02/03/2015.
Date of Publishing: 09/03/2015.