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A Rare Case Of Peter's PlusAnomaly In An Infant

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Abstract
Peter's Plus syndrome is an infrequent genetic disease , characterised by low height , peculiar facial appearance, eye anomalies and mental retardation. We report here a rare case of bilateral corneal opacity in 5 month old infant with ocular features of Peter's anomaly. Our case is of Peter's plus anomaly as it is associated with acyanotic heart disease and dysmorphic facial features.

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I. Introduction

This congenital anomaly was first described by Albert Peters, a German ophthalmologist (1862-1938)[1] .Peter's anomaly is rare and severe condition affecting both the eyes in more than half of the cases. It is defect in the endothelium and Descemet's membrane resulting in corneal clouding. Untreated case of Peter's anomaly can lead to amblyopia or congenital blindness. This condition is one of a group of disorders or differential diagnosis of congenital corneal opacities. Prevalence of Peter's anomaly is < 1 / 1000000 [2]. Previous observations points to a genetic etiology .[3]

II. Case

A case of five month old female infant (post term birth)was referred from paediatric department. The birthweight of child was 2.4kgs. Baby was delivered by C- section in our hospital.Cry after birth was present . No history of non-consanguineous marriage.No history of hospitalisation in post natal period . The child presented to us with bilateral corneal opacity. There is no one from the maternal and paternal side presenting similar complaint. No one in family showed any congenital abnormalities. There was history of the antenatal symptoms of UTI in 6^{th} month of pregnancy.

Examination was done after sedating the child. Diffuse light examination ,Microscopic examination ,direct ophthalmoscopy andindirect ophthalmoscopy examinations were performed. A complete pediatric work up was done. Parental informed consent to participate in this study was obtained.

III. Results

Ocular examination of anterior segment showed normal lids and conjunctiva.

Both eye Corneal opacity was present, pupillary reaction could not be assessed with diffuse illumination. On microscopic examination bilateral central corneal leucomatous opacity was seen . The opacity is not extending completely to the periphery. Clear area is present between periphery and central corneal opacity . Opacity was more in left eye than right eye. Vertical and Horizontal corneal diameter measured by callipers were 10mm in both eyes.

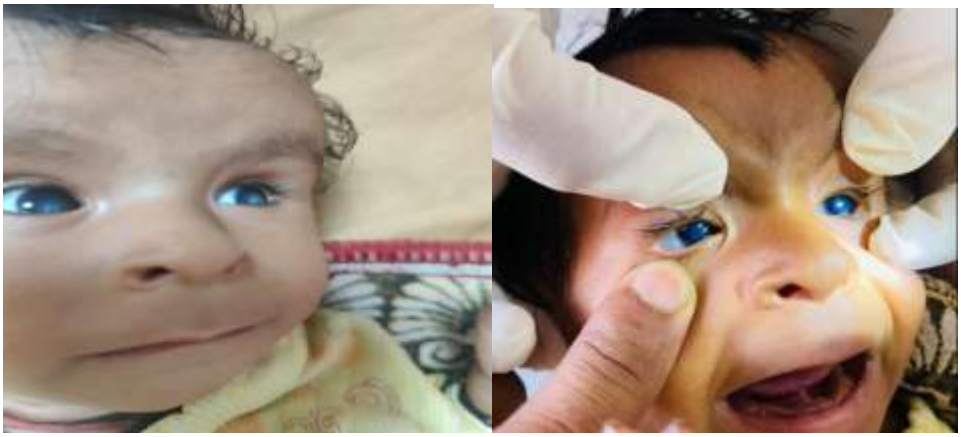

Figure 1 and 2 : Showing bilateral central corneal leucomatous opacities with clear periphery

Anterior Chamber depth was normal.
IOP was done by Schiotz Tonometry and was found to be 10.2 mmHg in RE and 12.2 mmHg in LE .

Distant Direct ophthalmoscopy showed normal fundal glow. On Indirect ophthalmoscopy , media was observed to be hazy ,disc was within normal limit rest details were not clearly seen.
Systemic examination revealed that patient has dysmorphic facial features.

Figure 3 : showing dysmorphic facial features

Patient was evaluated for TORCH infection, but it came out to be negative. Upper abdominal sonography results were also normal. On 2 D Echo patient was diagnosed with
- Acyanotic Congenital Heart Disease
-large size OS ASD with L-R shunt
-Spontaneously closed perimembranous VSD
- LPA Ostial narrowing present.

IV. Discussion

Peters plus syndrome (PPS) is a combination of congenital Peter's anomaly and systemic abnormalities. Ocular findings consist predominantly anterior segment abnormalities without posterior segment involvement

The characteristic facial features of Peters plus syndrome include a prominent forehead; small, malformed ears; narrow eyes; a long area between the nose and mouth (philtrum); and a pronounced double curve of the upper lip (Cupid's bow). The neck may also be broad and webbed. A cleft lip with or without a cleft palate is present in about half of the people with this condition.

Developmental milestones, such as walking and speech, are delayed in most children with Peters plus syndrome. Most affected individuals also have intellectual disability that can range from mild to severe, although some have normal intelligence. The severity of physical features does not predict the level of intellectual disability.

Less common signs and symptoms of Peters plus syndrome include heart defects, structural brain abnormalities, hearing loss, and kidney or genital abnormalities.

Peter's anomaly is a rare condition in which there is opacification or clouding of the cornea leading to amblyogenic effect on a developing infant. Hallmark of Peter's anomaly is central defect in Descemet's

membrane and corneal endothelium along with thinning & opacification of corresponding area of corneal stroma.[4,5,6,7] .

Mutations in the *B3GLCT* gene cause Peters plus syndrome. The *B3GLCT* gene provides instructions for making an enzyme called beta 3-glucosyltransferase (B3Glc-T), which is involved in the complex process of adding sugar molecules to proteins (glycosylation). Glycosylation modifies proteins so they can perform a wider variety of functions. Most mutations in the *B3GLCT* gene lead to the production of an abnormally short, nonfunctional version of the B3Glc-T enzyme, which disrupts glycosylation. It is unclear how the loss of functional B3Glc-T enzyme leads to the signs and symptoms of Peters plus syndrome, but impaired glycosylation likely disrupts the function of many proteins, which may contribute to the variety of features.[9]
Disease is inherited in Autosomal recessive pattern .

The critical event is 1st trimester of pregnancy, during the formation of the anterior chamber. Premature infants are at highest risk . Studies have shown deficiency of heparansulphate and fetal alcohol syndrome can lead to abnormal neural crest development in utero.

During embryogenesis structures of anterior segment are formed separately. If anterior segment development is abnormal then it can lead to attachment of cornea either to iris or to lens. This anterior segment dysgenesis causes Peter's anomaly. [8]
There are following subtypes of Peter's anomaly. [4]

A. Peter's anomaly I-the defect is in iris, corneal endothelium and Descemet's membrane leading mild to moderate corneal opacity.

B. Peter's anomaly II-lens, though developed normally, is pushed forward against the cornea leading to loss of descemet membrane causing severe corneal opacity that may involve the entire cornea.
C. Peter's anomaly can also be associated with AxenfeldRieger syndrome.

Corneal opacification leads low vision early in life and causes amblyopia. Other conditions can be present such asglaucoma, cataract, and microphthalmia. Mostly Peter's anomaly is bilateral and such individuals may exhibit strabismus[8] .

In Peter's Plus syndrome anterior segment examination will reveal a central leucoma on the cornea with loss of endothelium and Descemet's membrane. Iris strands are attached to the opacified cornea. In type II Peter's lens is attached to the cornea. Diagnosis can be done by genetic testing.

Differential diagnosis of congenital corneal opacifications are (STUMPED):
- Sclerocornea,
- Tears in Descemet's (eg. Congenital Glaucoma)
- Ulcers
- Metabolic disorders
- Peter's anomaly
- Endothelial dystrophy
- Dermoid

Treatments for Peter's Plus syndrome is to aim for visual maturation. Full thickness penetrating keratoplasty is the current standard indicated in Peter's for infants. The success rate of penetrating keratoplasty was significantly higher in patients with Peter's anomaly type I (87.5%), as compared to patients with Peter's anomaly type II (14.2%)[3] .

Combined surgery (cataract extraction and penetrating keratoplasty can be done in presence of opacified lens and cornea lens adhesion (i.e. Triple Procedure).Peter's Plus syndrome patient should be regularly monitored for raised intraocular pressure as glaucoma can be associated with it & if found raised appropriate medical or surgical management should be instituted.

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References

[1] Peters A. Über AngeboreneDefektbildung Der DescemetschenMembran. KlinMonblAugenheilkd. 1906;44:27–40.
[2] The Portal For Rare Diseases And Orphan Available At Http://Www.Orpha.Net/Consor/Cgibin/Disease_Search.Php?Lng=En&Data_Id=2590&Disease=Peters-Anomaly&Search=Disease_Search_Simple.
[3] Bhandari R, Ferri S, Whittaker B, Liu M, LazzaroDr. Peters Anomaly: Review Of The Literature. Cornea. 2011 Aug. 30(8):939-44. [Medline].
[4] ParulIchhpujani, Suresh Kumar Gupta, Surinder Singh Pandav,9.3.3 Congenital And Developmental Glaucoma: Clinical Profile, Postgraduate Ophthalmology Volume 1, First Edition, Jaypee Brothers Medical Publishers (P) Ltd, 2012;866-19
[5] Epstein Dl. Diagnosis And Management Of Lens Induced Glaucoma,Ophthalmology 1982;89:227.
[6] VerhoeffFh, Lemoine An. EndophthalmitisPhacoanaphylactica. In:Transaction Of The International Congress Of Ophthalmologist. Washington, Dc: William F Fell 1922;234

[7] Marak Ge, Font Ri, Rao Na. Abrogation Of Tolerance To Lens Protein. I. Effect Of Lipopolysaccharide, Opthalmic Res1979;11:192
[8] Strabismus And Amblyopia In Bilateral Peters Anomalynajjar D.M., Christiansen S.P., Bothun E.D., Summers C.G.(2006) Journal Of Aapos, 10 (3) , Pp. 193- 197.