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Comprehensive Case Analysis: Peters - Plus Syndrome in a 3 - Year - Old Male with Phenotypic Down's Syndrome

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Abstract: ***Peters plus syndrome is an autosomal recessive disorder characterized by anterior segment dysgenesis with systemic manifestations. A case report of a 3 year old male child presented with Peters plus syndrome having bilateral corneal opacity, left eye buphthalmos and phenotypic downs syndrome. Written informed consent was obtained from Patient's parents to publish the report.***

Keywords: Peters plus syndrome, peters anomaly, anterior segment dysgenesis, gross developmental delay, keratoplasty

1. Introduction

Peters - plus syndrome presents with ocular features, systemic malformations, and variable degrees of developmental delay. The term was first described by Dr Alfred Peters in 1906. Incidence is rare. Ocular abnormalities typically involve the anterior chamber known as Peters anomaly is anterior segment dysgenesis syndrome characterized by corneal opacity. Peters anomaly Type 1 is associated with iridocorneal adhesion while peters anomaly type 2 is associated with keratolenticular Adhesions. Ocular features with systemic association is known as Peters plus syndrome. Growth retardation, short stature, developmental delayand brachydactyly appear to be present in most of the patients, whereas external ear anomalies, cleft lip and/or palate, and cardiac and genitourinary malformations are less common. (1) It occurs due to defective B3GALTL gene encoding glycosyltransferase which play a crucial role in embryogenesis. (1, 2) Here we report a Male child affected by peters plus syndrome.

2. Case Report

A 3 year old male, Preterm vaginal delivery by birth with gross developmental delay was brought to tertiary care centre with complaints of opacity in both eyes since 6 months, it was insidious in onset. He was presented with complaints of watering in left eye. There was no history of trauma. Mother gave history of Gestational diabetes mellitus antenatally and was on diabetic diet.

On examination both eyes were not following to light and was uncooperative on torch light examination. Examination under Anesthesia was done under GA and Hordeolumexternum noted in medial aspect of Right Eye lower lid. A sausage shaped leucomatous corneal opacity, deep stromal vascularisation, iridocorneal contact from 1 o clock to 7 o clock hours, nondilating pupil, clear lens was noted in Right Eye. Central visualised Fundus was within normal limits.

Moderate proptosis was noted in left eye. Large Central leucomatous corneal opacity with thick fleshy mass sparing 1 mm of peripheral clear corneawas noted. There was deep stromal vascularisation and peripheral anterior synechiea were noted in left Eye.

Horizontal corneal diameter of right eye was 10 mm and of left eye was 10.8 mm.

IOP of right eye using I care was found to be 14 mmHg and of left eye to be 36 mm Hg.

On B scan deep cup was noted in left eye. Retina was attached.

Systemic manifestations included round face with long philtrum and low set ears, short limbs with broad distal extremities was examined by Paediatrician. He had gross developmental delay. Child hadhaematuria.

Pediatric endocrine opinion was taken and diagnosed as phenotypic downs syndrome with hypothyroidism. USG abdomen was suggestive of cystitis while 2D Echo was normal and diagnosis of Peters plus syndrome was made.

Penetrating keratoplasty is planned for right eye once the patient becomes fit for general anesthesia. Heis started on topical anti glaucoma medication eye drops timolol for buphthalmos in left eye.

Patient is currently on tab thyroxine 25microgram one tablet in morning for hypothyroidism. patient is now under close followup for the same.

3. Discussion and Conclusion

Peters plus syndrome also known as Krause kivlin syndrome an autosomal recessive disorder with homozygous pattern with B3GLCT mutation. It is characterized by eye

abnormalities with systemic manifestations Extraocular manifestations may include congenital heart disease, genitourinary Anomaly and conductive hearing loss. (3)

The most common® anterior chamber defect is Peters' anomaly, consisting of central corneal clouding and iridolenticulocorneal adhesions. Peters' anomaly may be classified as type I, a mild form, or type II, a more severe form associated with lens abnormalities including cataracts, congenital glaucoma, and a poorer visual prognosis. The eye involvement is usually bilateral. (4)

Patients rarely presents with iris coloboma.

Peters plus syndrome should be suspected in individuals with anterior chamber anomalies of the eye (usually bilateral but in some cases unilateral), with or without any of the following

- Short limbs with broad distal extremities
- Characteristic facial features including an exaggerated cupid's bow of the upper lip, short palpebral fissures, and ear anomalies
- Cleft lip, palate
- Variable developmental delay and intellectual disability. (4)

On Histopathology examination there is an absence of Descemetmembrane and endothelium at the site of corneal opacity. The chance of survival of children affected with peters plus syndrome is much less due to systemic manifestations of the syndrome. Genotypic testing can be done to diagnose the condition, but largely it is diagnosed clinically.

Parents are to be counselled regarding bad prognosis prior to initiating treatment (5)

Pupillectomy is to be done in case of clear peripheral cornea till definite management is done, to prevent amblyopia.

The usual management of Peters plus syndrome ispenetrating keratoplasty for severe bilateral corneal opacification before age three to six months to prevent amblyopia. Keratoprosthesis can be considered in case of failed graft.

Buphthalmos can be managed conservatively tilltrabeculotomy ortrabecuectomyis done. Selective Endothelialectomyis a new alternative for treatment of Peters syndome. (6)

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