Case Report

Peters anomaly with post axial polydactyly, ocular hypertelorism, a low nasal bridge, retrognathia, undescended testis, microphthalmia, and club foot in an Indian neonate: A case report

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Abstract

A case of Peters anomaly with bilateral post axial polydactyly, convex soles, ocular hypertelorism, a low nasal bridge, retrognathia, undescended testis, microphthalmia and club foot was examined in a neonatal Indian baby girl who had been delivered in the hospital and admitted to the newborn unit. She died aged five days. There were no cases of Peters anomaly recorded in India according to a literature search. In addition, available data point to the majority of the principal associations in Peters anomaly to be genitourinary anomalies, making this case a rare one in its isolated collection of musculoskeletal associations. A Indian baby girl of who was born through a Cesarean section presented in the new born unit of our hospital with bilateral corneal opacities, bilateral polydactyly, camptodactyly and club foot. This is a rare case of Peters anomaly and its association with Patau syndrome makes it special.

Key words

Peters anomaly, Polydactyly, Ocular hypertelorism, Retrognathia, Undescended testis, Micropthalmia, Club foot.

Introduction

Peters anomaly is one of the main causes of congenital corneal opacities. It is a rare form of anterior chamber development, either sporadic or inherited [1, 2], that presents as corneal opacity from birth with the opaque cornea obstructing the pupil and thus causing visual loss. In addition, there is anterior chamber dysgenesis with connection between the cornea and the iris and/or the lens in some cases. We have described here a case of newborn male who delivered by lower segment Cesarean section (LSCS) with low birth weight (LBW) and admitted in view of respiratory distress, the patient was managed by oxygen, IV fluids and antibiotics. Physical examination suggested congenital clubfoot and convex soles, ocular hypertelorism, a low nasal bridge, retrognathia, undescended testis, microphthalmia. USG abdomen suggested bilateral ballotable kidneys and bilateral undescended testis.

Case report

The patient was male, newborn, low birth weight (LBW), born via lower segment Cesarean section (LSCS) the fourth child of a non-consanguineous couple. Antenatal care was started at the fourth month of pregnancy, and the mother perceived the first fetal movements during the fifth month. The mother was 36 years old when she became pregnant. She reported the use of oral contraceptive pills during conception, and anemia and urinary tract infection were detected and treated during antenatal care. The child was born at term, from a LSCS delivery, and weighed 2,200 g. At birth, he was cyanotic, icteric, and cried weakly. The initial clinical examination detected polydactyly in the left hand, congenital clubfoot and convex soles, ocular hypertelorism, a low nasal bridge, retrognathia, undescended testis, microphthalmia with low set ears and sunken eyeballs. The child was admitted in view of respiratory distress started oxygen via hood, IV fluids, IV antibiotics (magnex, amikacin). Polydactyly was corrected on the child's third day of life. Jaundice was managed with phototherapy and the left foot was immobilized. The infant was kept in an incubator for 19 days. Muscular hypertonia was observed during the first days of life. 2D ECHO suggested of SITUS SOLITUS with levocardia with large patent ductus arteriosus (PDA) of 4 mm and small secendum ASD. The cardiac pathologies were initially treated with medication (loop diuretics and potassium-sparing diuretics). Patient had intermittent cyanosis and oxygen was continued. Due to persistent distress and decreased platelet counts, antibiotics stepped up to Meropenem. Patient had recurrent bleeding and received Packed cell transfusion. Fluconazole was started i/v/o Fungal sepsis, feeds started gradually. Ophthalmological reference was done which suggested of corneal opacification in right eye and coloboma. Left eye showed microcornea and cataractous lens. USG eye suggested complete closed funnel retinal detachment. Chromosomal studies suggested abnormal male karyotype with 47 XY +13 chromosomal pattern in all cells examined result consistent with patau syndrome. The parents were evaluated and the chromosomal studies were normal. The child gradually was weaned off from oxygen and blood parameters were improved and thus patient was discharged after proper counselling to parents (**Photo – 1 to 4**).

Discussion

Peters anomaly is a developmental disorder of the cornea which can occur either sporadically or inherited. Inherited cases are either autosomal dominant or recessive [1]. It is among the causes of congenital clouding and/or corneal opacity. Sclerocornea, tears in the Descemet membrane secondary to birth trauma, ulcers, congenital

glaucoma, congenital hereditary endothelial damage and various metabolic derangements are other causes of congenital corneal opacity [2]. It is important to differentiate Peters anomaly from other causes of corneal opacity. In Peters anomaly, the corneal opacity is either central or paracentral and it is usually without vascularization [3-5].

Photo – 1: Polydactyly in left hand.

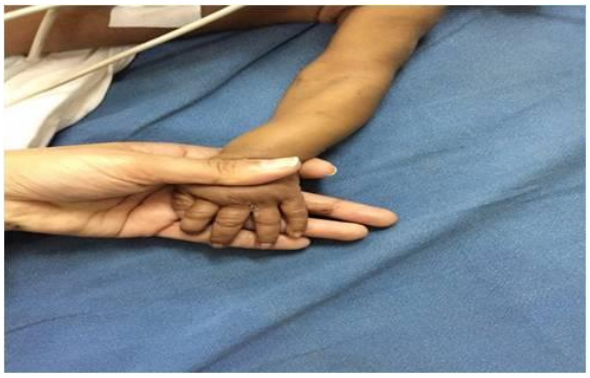

Photo – 2: Club foot and convex soles.

Two forms (Types I and Type II) of Peters anomaly are recognized. Type I occurs in the majority of the cases and type II occurs less often [2]. In the type I anomaly, the lens may or may not be cataractous and does not adhere to the cornea. Type II is usually associated with a cataractous lens. The lens in this type also adheres to the cornea [2-4]. Normally mutations in the PAX6 gene are associated with defects in the development of ocular tissues [5]. Thus, some cases of Type II Peters anomaly, which often tends to occur bilaterally, may be associated with PAX6 gene mutations [4-6]. This type is also associated with more systemic and ocular malformations [4].

Photo – 3: Retrognathia and cleft palate.

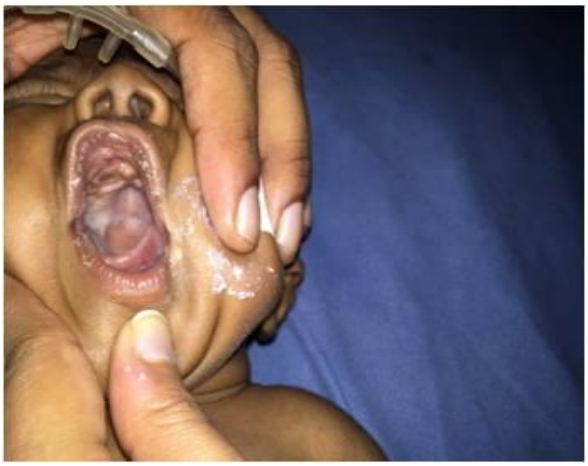

Photo – 4: Low set ears and hypertelorism.

Peters anomaly has unknown etiology but environmental along with genetic factors are thought to play a role in its genesis [5]. The critical step in the development of Peters anomaly occurs in the first trimester during the formation of the anterior chamber [5].

Gender predilection for Peters anomaly has not been shown by any studies. A study by Bhandari, et al., showed equal distribution in both sexes [6]. More cases tend to occur bilaterally [6] and these are more prone to have

systemic associations [6-9]. However, both types of Peters anomaly may be either unilateral or bilateral. Systemic associations occurring with Peters anomaly are congenital heart disease, neurologic defects, genitourinary abnormalities, external ear abnormalities and cleft lip and palate [4]. Various chromosomal abnormalities like trisomy 13-15, ring chromosome 21, Norrie disease, partial deletion of chromosome 11 q, mosaic trisomy 9 and the 49XXXXY syndrome have also been associated with Peters anomaly [4].

Treatment for Peters anomaly involve keratoplasty. The success rate, which relies on strict amblyopia therapy and requires prolonged follow-up, varies but is better with isolated unilateral cases [6, 7].

Our case has its unique characteristics with a normal anterior chamber and lens, and the absence of adherence between the cornea and the lens it seems to be type I Peters anomaly. However, its bilaterality and multiple associations argue for a unique association not recorded elsewhere in literature. Furthermore, as noted above, the systemic associations with Peters anomaly tend to be genitourinary, neurologic, cardiac and orofacial [4, 6]. The associated patau syndrome in this case makes it a special one and points to a possible new syndrome.

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