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Peters anomaly, a rare cause of anterior chamber dysgenesis

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Abstract

Background: Peters Anomaly (PA) is a rare form of developmental malformation involving the anterior segment of the eye as well as other body organs. Anterior chamber dysgenesis is characterized by corneal opacity and various other anomalies which can result in amblyopia of varying degree or even blindness. Early recognition and treatment may help to prevent complications in these patients.

Case presentation: We report an 8 month old female infant who at birth was noted to have dysmorphic facial features, bilateral corneal opacities, left eye proptosis , left megalocornea and bilateral cataracts . Dysmorphic features included depressed nasal bridge, mid facial hypoplasia, low set ears andhypertelorism. There was mild generalised hypotonia otherwise normal systemic examination. On day 2 of life , she was referred to ophthalmology at a tertiary referral centre for further advice and management. Her microarray showed loss of approx. 2.4 Mb in short arm of chromosome 6 at band 6p25.3-p25.2 between base pairs 163083 and 2527433. This loss includes FOXC1(OMIM 601090) gene which has strong association with abnormal eye development and Peters anomaly. Her other tests including urine for CMV,TORCH screening and homocysteine levels, were all reported normal. She went on to have bilateral corneal grafts and lens extraction. Currently she can perceive light but there are on-going concerns about her vision. Her development to date is appropriate.

Conclusion: Role of genetic testing is becoming more and more significant in confirmation of rare diseases. We recommend genetic testing to be considered earlier while investigating the cause of Peters anomaly.

Keywords: Pediatrics, Reactive oxygen species, Anticarcinogenicity, Antimutagenicity.

Introduction

Peters anomaly is a rare form of anterior segment ocular dysgenesis, which can also be associated with additional systemic defects. This can be unilateral or bilateral. It can be isolated or associated with systemic malformations. In one study bilateral involvement of the eyes in thisanomaly were associated with a higher rate of systemic malformations (71.8%) when compared to unilateral involvement (36.8%) [1]. The aetiology of Peters anomaly remains uncertain, but the most likely causes are related to genetic, infectious, traumatic and toxic factors. The majority of cases of Peters anomaly lack a genetic diagnosis [2]. The exact mechanism is not completely understood [3]. Migratory disorders of neural crest cells from 4 to 7 weeks of gestation may be responsible for various ocular anomalies. Mutations in Collagen type IV alpha-1 (COL4A1) and Beta-1,3-galactosyltransferase-like (B3GALTL) have been reported in anterior segment dysgenesis. Corneal opacity obstructs the visual axis, leading to sensory deprivation, amblyopia, and severe visual impairment. A range of possible treatment strategies exists, though the effectiveness of each of them depends on how the disease occurs and whether it is identified in early or advanced stages.Earlier the diagnosis, the higher the possibility of a successful interventions.

Case report

We report an 8 month old female infant with Peters anomaly who presented at birth with bilateral corneal opacities and other eye anomalies .She was born at 39+6/40 weeks of gestation to 28 years old primigrivida by vacuum extraction due to delayed 2nd stage of labour. APGAR score was 9 and 10 at one and five minutes. She did not require any resuscitation. Mother was rubella non immune, denied

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smoking or taking alcohol or drugs during pregnancy. Anomaly scan was normal and pregnancy was uneventful. Membranes were ruptured 44 hours before delivery and three doses of antibiotics were given to mother. She spiked temperature up to 38.5c during last few hours of delivery .At birth bay was noted to have dysmorphic features which included depressed nasal bridge, mid facial hypoplasia, low set ears, hypertelorism, bilateral cloudy corneas, left eye proptosis, left megalocornea and bilateral absent red reflex. Rest of systemic examination was unremarkable except for mild generalized hypotonia. She was admitted to special care baby unit for septic work up,intravenous antibiotics and further evaluation. On day 2 of life she was seen by ophthalmologist at a tertiary referral centre and a provisional diagnosis of Peters anomaly was suggested.

Along with bilateral corneal opacities, she also had cataracts and left raised intraocular pressure. She was put on topical steroids and latanoprost .After counselling with both parents bloods were sent for genetics and other tests. Her renal ultrasound scan was normal and cranial ultrasound scan showed mildly prominent posterior horns but no hydrocephalus. Urine for CMV was negative ,TORCH screening and homocysteine levels were normal. Her microarray revealed a loss of aprox 2.4 Mb in short arm of chromosome 6 at band 6p25.3-p25.2 between basE pairs 163083 and 2527433.This loss includes number of genes including the FOXC1(OMIM 601090) gene which has strong association with abnormal eye development and Peters anomaly. Both parents were also tested for same deletion but they were phenotypically normal.

She got left cataract extraction and corneal transplant was done in both eyes. She also developed adrenal suppression due to exogenous steroids therapy and is on replacement therapy by endocrinology.

She is regularly attending our clinic along with ophthalmology, endocrinology, and physiotherapy input.

She perceives light and her outcome regarding vision is guarded. He development otherwise is normal .

Discussion

Peters anomaly was first described by Dr Alfred Peters in 1906 [4].it is a rare cause of anterior segment dysgenesis of the orbit. The exact prevalence of Peters anomaly is unknown. This condition is one of a group of disorders known as congenital corneal opacities, which affect 3 to 6 individuals per 100,000 [5]. Aetiology of this anomaly remains uncertain, but the most likely causes are related to genetic, infectious, traumatic and toxic factors. This disease mostly appears sporadically, however, both recessive and irregular dominant inheritance has been described. Mutations in any of four genes, FOXC1, PAX6, PITX2, or CYP1B1, disrupts development of the anterior segment of the eye. There are two types of Peters anomaly, which are distinguished by their signs and symptoms. Peters anomaly type I is characterized by an incomplete separation of the cornea and iris and mild to moderate corneal opacity. Type II is characterized by an incomplete separation of the cornea and lens and severe corneal opacity that may involve the entire cornea [6]. Our patient has type II Peters anomaly, associated with bilateral cataract and left glaucoma. Peters plus syndrome includes short disproportionate stature, developmental delay, dysmorphic facial features, cardiac, genito-urinary, and central nervous system malformation. These systemic findings are seen in up to 60% of patients. Our patient has midfacial hypoplasia, depressed nasal bridge, low set ears and hpertelorism but no other system involvement .Treatment depends upon degree of anterior chamber involvement ,associated other eye problems and systemic involvement. Both medical and surgical treatment along with multidisciplinary care is advised. At the moment our patient is getting medical treatment to reduce intraocular pressure and has undergone bilateral lens extraction. She is regularly attending our clinic along with ophthalmology, endocrinology, physiotherapy and genetics. Her development up till now is appropriate but close follow up of her development is ongoing.

Conclusion

Peter anomaly sometimes may have genetic basis and hence genetic testing should be considered earlier while planning investigations. Its earlier recognition and management may have better outcome in terms of vision and overall development.

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