

## CASE REPORT

Egypt. J. Med. Hum. Genet. Vol. 10, No. 1, May, 2009

### Abnormal presentation of Peters' anomaly in a family with microcornea cataract syndrome

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#### ABSTRACT

A case of Peters' anomaly with bilateral crease on ear lobule and hypospadias was reported in a family with microcornea, cataract syndrome. The eyes had all the features of Peters' Anomaly. The combination of these findings has not been reported in literature to our knowledge, making this case a unique one.

#### Key Words:

Peters' anomaly, persistent papillary membrane, ear lobule crease, hypospadias, microcornea, cataract syndrome.

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#### INTRODUCTION

Peters' anomaly consists of a central corneal leukoma, absence of the posterior corneal stroma and descemet membrane, and a variable degree of iris and lenticular attachments to the central aspect of the posterior cornea<sup>1</sup>. 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<sup>2</sup>. It occurs as an isolated ocular abnormality or in association with other ocular defects; for example, Peters' anomaly with cataract and cataract-microcornea syndrome.

Peters' plus syndrome is an infrequently described entity that combines anomalies in the anterior chamber of the eye with

other multiple congenital anomalies and a developmental delay. Major symptoms are extremely variable, anterior chamber anomalies, cupid bow of the upper lip, cleft lip and palate, short stature, broad hands and feet and variable mental delay. The syndrome follows an autosomal recessive pattern of inheritance. The etiology is unknown, but may involve abnormal neural crest development.<sup>3</sup>

#### CASE REPORT

An infant boy 2.5 months old, first offspring of a non consanguineous marriage. He was born at full term by vaginal delivery. The birth weight was 2.9 kg. The mother 26 years old was on aldomet during pregnancy for hypertension. She has sclerocornea, microcornea and cataract. Pedigree analysis revealed the presence of microcornea, cataract and convergent squint in the maternal father, microcornea and cataract in a maternal uncle and aunt and no other affected family members (Figure. 1).

On examination, the weight was 6 Kgs (90<sup>th</sup> centile). Facial examinations revealed bilateral central corneal opacity, the left corneal opacity was larger than that on the right side (Figure. 2), bilateral ear lobule crease and preauricular pits (Figure. 3). He had wide forehead, broad nasal bridge and long philtrum. Also he had short fingers, bilateral clinodactyly of little finger (Figure. 4) and bilateral simian creases in both hands (Figure. 5). Genital examination revealed hypospadias and bilateral descended normal sized testes. Abdominal examination was clinically free and this was confirmed by abdominal ultrasonography. Chest and cardiac examinations were free. He had no neurological manifestations. Ophthalmologic examination of the eyes revealed iridocorneal contact from persistent papillary membrane (Peters' anomaly). The lenses were clear bilaterally. Fundus abnormality was excluded, and congenital glaucoma was ruled out. Various laboratory parameters were normal. Echocardiography showed no apparent significant cardiac lesion. Peripheral blood cytogenetic analysis revealed normal male karyotype.



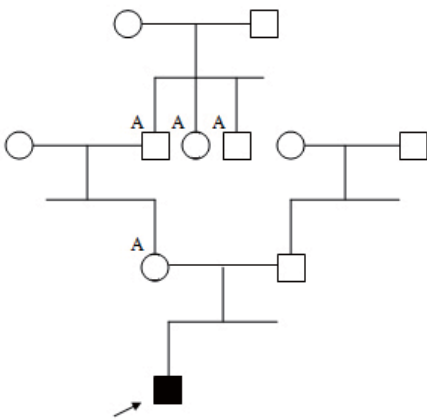
Fig. 2: Bilateral central corneal opacity.



Fig. 3: Ear lobule crease, pre auricular pits.



Fig. 4: Short fingers and clinodactyly of little finger.



A: Microcornea and cataract.

Fig. 1: Pedigree of the family.



**Fig. 5:** Simian crease.

## DISCUSSION

Peters' Anomaly occurs with various ophthalmological findings, mainly iridolenticular contact, keratolenticular contact, cataracts, glaucoma and variations in the globe volume<sup>4</sup>. A variant of Peters' anomaly has been described involving the lens. This has been termed as Peters' anomaly with corneolenticular contact or cataract<sup>5</sup>. By analysis of the pedigree of the family of our case, it revealed four members of the family with microcornea cataract syndrome suggesting most probably autosomal dominant inheritance. Microcornea-cataract syndrome (OMIM 116150) is characterized by the association of congenital cataract and microcornea, without any other systemic anomaly or dysmorphism. Many families showing microcornea-cataract syndrome have been described. The inheritance pattern in these families has been reported as autosomal dominant<sup>6</sup>. Although the autosomal dominant inheritance pattern, our case presented only with Peters' anomaly without microcornea and cataract. Salmon<sup>7</sup>, suggested that the embryological origins of microcornea, sclerocornea and Peters' anomaly are

similar on the basis of the analysis of a family with microcornea-cataract syndrome in which six individuals had sclerocornea and Peters anomaly.

Some investigators<sup>2</sup>, have divided Peters' anomaly into 2 types: A mesodermal or neuroectodermal form (Type I), which shows no associated lens changes and a surface ectodermal form (Type II), which does. In our case Peters' anomaly was present bilaterally as an isolated finding in both eyes as there was no cataract (Type I).

Also in our case there was no glaucoma. Yang et al.<sup>8</sup> reviewed the long-term outcome in 19 patients with Peters' anomaly. Severe glaucoma was a prominent feature among their patients. Despite combined medical and surgical interventions, glaucoma could be controlled in only 32% of eyes with glaucoma associated with Peters' anomaly. Visual results were poor due to uncontrolled glaucoma, amblyopia, neurologic impairment and other anterior and posterior segment anomalies that might accompany Peters' anomaly.

The combination of bilateral ear lobule crease, pre auricular pits and hypospadias with Peters' anomaly has not been reported in literature to our knowledge. We presume that there no previous cases reported with Peters' anomaly associated with ears abnormalities or hypospadias. Mayer<sup>9</sup> and Heon et al.<sup>10</sup> 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. Cases with 4p-, mosaic trisomy 9 and trisomy 13 were also seen in these series. Trauboulsi and Maumenee<sup>11</sup>, reported a se-

ries of 29 cases with a similar incidence of mental retardation and associated malformations.

## REFERENCES

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1. Peters A. Ueber angeborene defektbildung der descemetischen membran. *Klin.Mbl.Augenheilk.* 1906; 44: 27-40, 105-19.
2. Kliegman RM, Behrman RE, Jenson HB, Stanton BF. Abnormalities of cornea. In: Kliegman RM, Behrman RE, Jenson HB, Stanton BF, editors. *Nelson textbook of pediatrics.* 18<sup>th</sup> ed.: Saunders; 2007. p. 2591-3.
3. Maillette de Buy Wenniger Prick, L. J., Hennekam RC. The Peters' plus syndrome: A review. *Ann. Genet.* 2002; 45 (2): 97-103.
4. Bateman JB, Maumenee IH, Sparkes RS. Peters anomaly associated with partial deletion of the long arm of chromosome 11. *Am. J. Ophthalmol.* 1984; 97(1): 11-5.
5. Harley RD. *Pediatric ophthalmology.* 2<sup>nd</sup> ed.: WB Saunders Co.; 1983.
6. Willoughby CE, Shafiq A, Ferrini W, Chan LL, Billingsley G, Priston M, et al. CRYBB1 mutation associated with congenital cataract and microcornea. *Mol. Vis.* 2005;11:587-93.
7. Salmon JF, Wallis CE, Murray AD. Variable expressivity of autosomal dominant microcornea with cataract. *Arch. Ophthalmol.* 1988; 106 (4): 505-10.
8. Yang LL, Lambert SR, Lynn MJ, Stulting RD. Surgical management of glaucoma in infants and children with Peters' anomaly: Long-term structural and functional outcome. *Ophthalmology* 2004; 111 (1): 112-7.
9. Mayer UM. Peters' anomaly and combination with other malformations (series of 16 patients). *Ophthalmic Paediatr. Genet.* 1992; 13(2): 131-5.
10. Heon E, Barsoum Homsy M, Cevrette L, Jacob JL, Milot J, Polemeno R, et al. Peters' anomaly. The spectrum of associated ocular and systemic malformations. *Ophthalmic Paediatr. Genet.* 1992; 13 (2): 137-43.
11. Traboulsi EI, Maumenee IH. Peters' anomaly and associated congenital malformations. *Arch. Ophthalmol.* 1992; 110 (12): 1739-42.