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ORIGINAL INVESTIGATION

Lacrimal drainage system involvement in Peters anomaly: clinical features and outcomes

Nandini Bothra, Abhimanyu Sharma, and Mohammad Javed Ali

Govindram Seksaria Institute of Dacryology, L.V. Prasad Eye Institute, Hyderabad-34, India

ABSTRACT

Purpose: To present first of its kind series on the clinical features and outcomes of lacrimal drainage disorders in Peters anomaly and Peters plus syndrome.

Methods: A retrospective chart review was performed of all consecutive patients who were known cases of Peters anomaly or Peters plus anomaly and were diagnosed with associated congenital lacrimal drainage disorders. The study period was from June 2016 to Dec 2020. All these patients underwent examination under anaesthesia for a detailed assessment of lacrimal drainage anomalies. Where indicated, they were treated with probing, intubation, or in refractory patients with a dacryocystorhinostomy. The anatomical and functional outcomes were assessed.

Results: Of the 282 patients with Peters anomaly, 4 (1.4%) patients had associated lacrimal drainage system anomalies while of the 16 Peters plus anomaly children, 3 (18.75%) had associated lacrimal drainage system anomalies. A total of 12 lacrimal drainage systems of 12 eyes of 7 patients of Peters anomaly were found to be involved. Upper or lower punctal agenesis were noted in 3 eyes. Three eyes had complex congenital nasolacrimal duct obstruction (CNLDO), two of which had a bony NLD block and one had a misdirected nasolacrimal duct through the inferior turbinate. One eye had a diffuse NLD stenosis without a CNLDO. Following appropriate management, at a mean follow-up of 25.7 months (range: 3–48 months), all the eyes except one (91%, 10/11) demonstrated anatomical and functional success.

Conclusion: Lacrimal drainage involvement was more common in Peters plus syndrome. Multiple proximal and distal lacrimal drainage segment anomalies were noted in all the variants of Peters anomaly; however, Peters plus syndrome was noted to usually involve both the segments.

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CNLDO; DCR; lacrimal; Peters anomaly; syndrome

Introduction

Peters anomaly was initially described by German ophthalmologist Alfred Peters as a syndrome of shallow anterior chamber, synechiae between iris and cornea, central corneal leukoma, and a defect in the Descemet membrane.[1,2] The incidence of ocular malformations in new-born range from 3.3 to 6.0 per 10,000 births[3] and Peters anomaly accounts for the largest proportion of the anterior segment dysgenesis.[4] The inheritance of this disorder can be sporadic (most common), autosomal dominant via homeobox genes PAX6, FOXC1, and PITX2 or autosomal recessive (via CYP1B1).[5] The genes PAX6, FOXC1, and PITX2 are involved in development of the anterior segment of the eye and CYP1B1 gene encodes a member of the cytochrome p450 superfamily of enzymes which are involved in the metabolism of a signalling molecule essential for development of cornea.[5]

Peters anomaly are of three types: Type 1 is incomplete separation of the cornea and iris with the corneal opacity; Type 2 is incomplete separation of cornea and lens with corneal opacity; and Peters plus syndrome, characterized by Peters anomaly in association with systemic malformations such as cleft lip/palate, short stature, abnormal ears, or mental retardation.[2,5] Type 2 is more severe with poorer visual prognosis.

Peters anomaly is also associated with mal-development of the lacrimal drainage system and to the best of authors knowledge, there exists only one such report in the literature.[6] The current case series presents exclusively the developmental lacrimal drainage abnormalities seen in patients with Peters and Peters plus anomalies.

Methods

The study adhered to the Declaration of Helsinki and was approved by the Institutional Ethics Committee. A retrospective chart review was performed of all consecutive patients who were known cases of Peters anomaly or Peters plus anomaly and were diagnosed with associated congenital lacrimal drainage disorders. The study period was from June 2016 to Dec 2020. Of the 282 patients with Peters anomaly, 4 (1.4%) patients had associated lacrimal

CONTACT Mohammad Javed Ali javed@lvpei.org Govindram Seksaria Institute of Dacryology, L.V. Prasad Eye Institute, Hyderabad-34, India

drainage system anomalies while of the 16 Peters plus anomaly children, 3 (18.75%) had associated lacrimal drainage system anomalies. All these patients underwent examination under anaesthesia for a detailed assessment of lacrimal drainage anomalies. All the lacrimal anomalies could be assessed clinically with the help of nasal endoscopy and did not require additional radiological investigations. Where indicated, they were treated with probing, intubation, or in refractory patients with a dacryocystorhinostomy. The anatomical and functional outcomes were assessed. The anatomical success was defined as patent lacrimal drainage system on irrigation and functional success was defined as resolution of epiphora, normal tear meniscus height, and fluorescein dye disappearance test.

Results

A total of 12 lacrimal drainage systems of 12 eyes of 7 patients of Peters anomaly were found to be involved (Table 1). The mean age at presentation was 12.64 months (range: 0.5 months–24 months) and male to female ratio was 4:3. Of the 7 patients, two patients had Peters anomaly type 1, two had Type 2, and three had Peters plus syndrome. Five patients had bilateral involvement of their lacrimal drainage apparatus. The complaints of epiphora and visibly high tear meniscus height with discharge and matted lashes were the reasons for the Dacryology service referral.

Upper or lower punctal agenesis were noted in three eyes (1 patient of peter's plus syndrome), one of which was noted to involve both the upper and lower punctum (Figure 1a,b). One eye also showed upper and lower canalicular stenosis. Distal lacrimal drainage system involvement was more common as compared to the proximal lacrimal passages. The majority finding (4/7, 57.14%) in the lower lacrimal passage was a congenital nasolacrimal duct obstruction (CNLDO)[7] (Figure 1c,d), which was managed by irrigation and probing under endoscopy guidance. One eye showed the presence of diffuse nasolacrimal duct stenosis without associated CNLDO, which was managed by a 4-week intubation by the Monoka-Crawford stent. Three eyes had a complex CNLDO,[7] two of which had a bony NLD block (Peter's plus syndrome) and one had a misdirected nasolacrimal duct through the inferior turbinate. All these eyes with complex CNLDO underwent endoscopic dacryocystorhinostomy as per standard protocols.[8,9] Overall, the surgical interventions were performed for 11 of the 12 eyes. One eye with both upper and lower punctal agenesis was observed with an intention for a possible conjunctivodacryocystorhinostomy at a later date. At a mean follow-up of 25.7 months (range: 3–48 months), all the eyes except one (91%, 10/11)

Table 1.: Summary of lacrimal drainage abnormalities in Peters anomaly.

Pt no	Age (months)	Sex	Peters anomaly	Upper lacrimal drainage apparatus	Lower lacrimal drainage apparatus	Lacrimal intervention	Follow-up (months)	Success
1	0.5	Male	Peters plus syndrome	OU LP agenesis	OU membranous obstruction at lower end of NLD	OU irrigation and probing	36	Yes (A+,F+)
2	24	Female	Peters type 2	OU UP agenesis	OD NLD stenosis	OD Crawford monoka intubation	36	No (A+, F-)
3	9	Male	Peters type 1	–	OD membranous obstruction at lower end of NLD	OD irrigation and probing	3	Yes (A+,F+)
4	1	Male	Peters type 2	–	OS membranous obstruction at lower end of NLD	OS irrigation and probing	9	Yes (A+,F+)
5	24	Male	Peters plus syndrome	–	OU bony stop at sac-NLD junction	OU endoscopic DCR	24	Yes (A+,F+)
6	6	Female	Peters plus syndrome	OD UP + LP agenesis OS UC + LC canalicular stenosis	OS misdirected NLD thru the inferior turbinate OS lacrimal sac diverticulum	OS endoscopic DCR	48	Yes (A+,F+)
7	24	Female	Peters type 1	–	OU membranous obstruction at the lower end of the NLD	OU irrigation + probing	24	Yes (A+,F+)

Pt no, patient number; UP, upper punctum; LP, lower punctum; UC, upper canaliculus; LC, lower canaliculus; NLD, nasolacrimal duct; DCR, dacryocystorhinostomy; A, anatomical success; F, functional success.

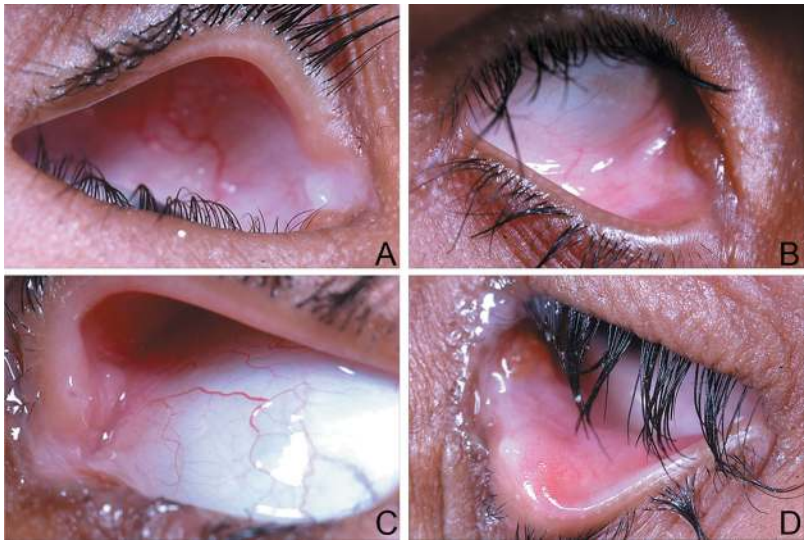

Figure 1. Lacrimal drainage involvement in Peters plus syndrome: external photograph of the right eye showing upper (a) and lower (b) punctal agenesis along with microphthalmos. The left eye showed matting of eyelashes (c and d) with a normal upper punctum (c) and regurgitation of mucoid discharge from the lower punctum on lacrimal sac compression (d), indicative of nasolacrimal duct obstruction.

demonstrated anatomical and functional success. The one exception was the patient with diffuse nasolacrimal duct stenosis who improved anatomically, but the tear meniscus height continued to remain elevated and was lost to follow-up later.

Discussion

The embryology of the lacrimal system is such that the morphology of the lacrimal drainage system is well developed in the embryonic stage itself (9th week, 23rd Carnegie stage). The lateral portion of the lacrimal cord is differentiated into superior and inferior canaliculus proximally and lacrimal sac distally and the medial part of the cord continues caudally and lateral to the inferior meatal lamina. After the 10th week, the canalization of the lacrimal cord and development of the surrounding tissue occurs.[10] Development of face and has maxillary region as also the lacrimal drainage system is related to the first and second branchial arches. Any disruption in this development can lead to abnormality in the development of the lacrimal system.[11]

The genes associated with Peters anomaly are mainly homeo-box containing genes, which direct the formation of numerous parts of the body during early embryonic development including the craniofacial development.[5,12] Homeobox-containing genes encode DNA-binding proteins that regulate gene expression and control various aspects of morphogenesis and cell differentiation.[12] Thus, mutations can lead to abnormal development of the craniofacial region leading to anomalies of the lacrimal drainage system.

Children with Peters anomaly can have visual deprivation which may lead to development of sensory deprivation amblyopia and hence poor visual prognosis. Therefore, early intervention is the key to successful outcome.[2] Lacrimal drainage anomalies, thus, if present, ideally need to be addressed before any planned intraocular intervention to avoid any potential risk of endophthalmitis.

Lacrimal drainage system involvement in association with Peters anomaly has been described in two sisters till date. Both the patients had bilateral nasolacrimal duct aplasia with recurrent attacks of dacryocystitis. In view of the associated arrhinia, a bypass was not possible and hence a bilateral lacrimal sac extirpation was performed.[6] In comparison, the present series of 12 eyes showed multiple proximal and distal lacrimal drainage developmental defects most of which could be successfully managed by probing or intubation or dacryocystorhinostomy. In conclusion, lacrimal drainage involvement is not uncommon in Peters anomaly with Peters syndrome showing both proximal and distal segment involvement. Physicians encountering cases of Peters anomaly should keep in mind the need for lacrimal drainage assessment and management, as appropriate, to reduce the burden of morbidity.

Disclosure statement

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References

1. Peters A. Ueber angeborene Decectbildung der Descemetschen Membran. *Klin Monbl Augenheilkd*. 1906;44:27–40.
2. Bhandari R, Ferri S, Whittaker B, Liu M, Lazzaro DR. Peters anomaly: review of the literature. *Cornea*. 2011;30:939–944. doi:10.1097/ICO.0b013e31820156a9.
3. Kupfer C, Kuwabara T, Stark WJ. The histopathology of Peters anomaly. *Am J Ophthalmol*. 1975;80:653–660. doi:10.1016/0002-9394(75)90397-9.
4. Ni W, Wang W, Hong J, Zhang P, Liu C. A novel histopathologic finding in the Descemet's membrane of a patient with Peters anomaly: a case-report and literature review. *BMC Ophthalmol*. 2015;15:139. doi:10.1186/s12886-015-0131-y.
5. Salik I, Gupta A, Tara A, Zaidman G, Barst S. Peters anomaly: a 5-year experience. *Paediatr Anaesth*. 2020;30:577–583. Epub. doi:10.1111/pan.13843.
6. Ruprecht KW, Majewski F. Familiäre Arhinie mit Petersscher Anomalie und Kiefermissbildungen, ein neues Fehlbildungssyndrom? [Familiary arhinia combined with Peters' anomaly and maxilliar deformities, a new malformation syndrome (author's transl)]. *Klin Monbl Augenheilkd*. 1978;172:708–715.
7. Kushner BJ. The management of nasolacrimal duct obstruction in children aged between 18 months and 4 years. *J Aapos*. 1998;2:57–60. doi:10.1016/S1091-8531(98)90112-4.
8. Bothra N, Vasanthapuram VH, Ali MJ. Infantile endoscopic dacryocystorhinostomy: indications, anatomical considerations, and outcomes. *Ophthalmic Plast Reconstr Surg*. 2020;36(4):e100–e103. Epub. doi:10.1097/IOP.0000000000001615.
9. Bothra N, Naik MN, Ali MJ. Outcomes in pediatric powered endoscopic dacryocystorhinostomy: a single-center experience. *Orbit*. 2019;38:107–111. doi:10.1080/01676830.2018.1477808.
10. Ali MJ, Kakizaki H. Embryology of the lacrimal drainage system. In: Ali MJ, ed. *Principles and Practice of Lacrimal Surgery*. 2nd ed. Singapore: Springer; 2018:9–18.
11. Ali MJ, Paulsen F. Syndromic and nonsyndromic systemic associations of congenital lacrimal drainage anomalies: a major review. *Ophthalmic Plast Reconstr Surg*. 2017;33:399–407. doi:10.1097/IOP.0000000000000923.
12. Mark M, Rijli FM, Chambon P. Homeobox genes in embryogenesis and pathogenesis. *Pediatr Res*. 1997;42:421–429. doi:10.1203/00006450-199710000-00001.