Congenital nasolacrimal duct cyst/dacryocystocele: an argument for a genetic basis

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Abstract
Introduction: Embryogenesis of a congenital nasolacrimal duct (NLD) cyst is attributed to the failure of the Hasner membrane of the NLD system to canalize. Prenatal diagnosis of congenital NLD cysts supports the argument for a developmental error, with a postnatal prevalence of 0%. The role of a genetic basis for this malformation has never been ascribed. We present a set of monozygotic twins with bilateral congenital NLD cysts as an argument for a genetic basis of this entity.

Method: Case report and literature review.

Results: We present two cases of bilateral congenital NLD cysts occurring in a set of monozygotic twins. Patients were delivered at 37 weeks via cesarean section. The pregnancy was complicated by pre-term labor at 33 weeks requiring administration of terbutaline and betamethasone. At presentation, twin A had bilateral eye discharge, erythema and swelling medial to the medial canthi as well as nasal obstruction. Computed tomography (CT) showed classic bilateral cystic masses in the inferior meatus. The diagnosis of bilateral infected congenital dacryocystocele was made. Twin B initially presented with only bilateral eye discharge and CT showed a dilated frontal sinus (Figure 1B). Twin B subsequently developed early signs of bilateral dacryocystocele the following day. Both patients underwent lacrimal probing and endoscopic marsupialization of the dacryocystocele. Biopsies were consistent with dacryocystocele.

Conclusion: Dacryocystocele is a common presentation of unresolved neonatal NLD obstruction. This case report in a set of identical twins is an argument for a genetic basis for the formation of this lesion.

Introduction:
Congenital nasolacrimal duct (NLD) obstruction is thought to be a prenatal developmental failure to canalize the NLD system. The lacrimal system begins to form in the 5th week of fetal development. Formation of a lumen in the lacrimal duct occurs in the 10th week of development, which coincides with ciliation of the inferior meatal lining. Through canalization of the lacrimal duct, communication with the nasal inferior meatus is completed from the 6th fetal month to beyond term. If this normal developmental process fails, a thin membranous membrane barrier can persist at the lower end of the NLD, occurring in about 5-6% of full-term newborns. Congenital dacryocystocele is an uncommon condition in which cystic swelling of the lachrymal sac accompanies obstruction of the lacrimal drainage system both above and below the sac. Although the upper part of the system may be anatomically obstructed, most often there is simply an unusually competent valve of Rosenmüller that prevents reflux of accumulated fluid from the sac. Persistent obstruction at the level of the valve of Hasner and secondary functional obstruction at valve of Rosenmüller lead to the formation of a dacryocystocele [1]. A congenital dacryocystocele presents during the first few weeks of life as a benign, bluish-gray mass in the inferomedial canthus. There may be associated blushing of mucus at the lower end of the NLD into the nasolacrimal duct, resulting in epiphora and possibly respiratory distress of a newborn, when the cyst significantly compromises the airway. Diagnosis of dacryocystocele with intranasal cyst is made through physical examination, clinical history, nasal endoscopy and imaging. Neonatal patients with acute dacryocystitis and cellulitis should give rise to the suspicion of the existence of an intra nasal cyst. Bilateral nasal endoscopy is essential in the workup of infants presenting with clinical findings suspicious for a dacryocystocele, and often will show a cystic mass in inferior meatus. Computed tomography (CT) helps to provide details of the anatomy of the nasolacrimal duct and cyst. Treatment of epiphora associated with simple nasolacrimal duct obstruction is typically conservative, as the majority of patients can experience spontaneous resolution. Management of dacryocystocele remains somewhat controversial. Harris advocated conservative medical management with the majority of these subjects not requiring surgical intervention. The majority view recommends initial conservative management with attempted manual decompression and massage of the nasolacrimal sac for uninfected dacryocystocele before pursuing combined NLD probing and endoscopic nasal mucuplasty [2]. If the condition does not resolve spontaneously, with obvious local inflammatory changes usually develops within the first few weeks of life in most cases. Many surgeons will recommend lacrimal probing no later than one month of age in persistent cases. Epidemiologically, congenital dacryocystocele is relatively uncommon and is estimated to occur in 1 in 3884 births [3]. It has been reported to be more common in female [4,5] and non-Hispanic white infants [4]. Although familial cases have been described sporadically [5,4,9,10], it is generally accepted as an isolated developmental error. We suggest a genetic basis of this disorder through presenting our case series.

Case Presentation:
We present two cases of bilateral congenital dacryocystoceles occurring in a set of identical twins. Two 23-week-old female patients presented to the emergency department at our institution for evaluation of epiphora, edema and nasal discharge associated with swelling of the medial canthal regions. These ex-37-week-old girls were delivered by cesarean section, and the pregnancy was complicated by pre-term labor at 33 weeks requiring terbutaline and betamethasone given to the patients’ mother. The mother was treated with amoxicillin-clavulanate for a urinary tract infection near the time of the delivery, but otherwise had an uneventful pregnancy. The twins had an uneventful post-delivery course in the newborn nursery for observation and were discharged to home. Twin A was seen by a primary care provider one day prior to admission and treated with cephalaxin and polymyxin B sulfate/triamcinolone sulfate ophthalmic solution for bilateral eye discharge. At presentation, twin A had bilateral eye discharge, erythema and swelling medial to the medial canthi as well as nasal obstruction. CT showed classic bilateral cystic masses in the inferior meatus (Figure 1A). The diagnosis of bilateral infected congenital dacryocystoceles was made. Twin B initially presented with only bilateral eye discharge and CT confirmed a dilated frontal sinus (Figure 1B). Twin B subsequently developed early signs of bilateral infected dacryocystoceles the following day. Both patients underwent lacrimal probing and endoscopic marsupialization of the bilateral nasolacrimal cysts, with biopsies consistent with dacryocystoceles (Figure 2A through 5). They were discharged from the neonatal intensive care unit within 24 hours.

Discussion:
Congenital NLD obstruction is found relatively frequently and an incidence of 3% to 7% has been reported [6]. The majority spontaneously resolves during the first few weeks of life [7], experience at our institution suggests that the existence of the two entities has a sine qua non relationship. Although there is limited evidence of heritable factors in play for congenital dacryocystocele in the literature, our case series of a set of identical twins with bilateral dacryocystoceles advances the notion that this congenital development disorder has a genetic basis. Potential inherited predisposition to dacryocystocele formation is suggested by increased female prevalence (3:9 times more commonly than males) and an increased incidence in Caucasian and Hispanic populations [1]. However, the increased prevalence in females could be attributed to an anatomically narrower nasolacrimal duct. Traquair indicated a pattern of familial inheritance in 15% of patients in a large study of the etiology of dacryocystitis [8]. Further literature review showed 3 cases of familial nasolacrimal duct obstruction; however two of the cases were associated with syndromes that resulted in nasolacrimal duct obstruction and were not related to isolated congenital duct obstruction.

References:
5. Shandy RS, Durar, Congenital nasolacrimal duct obstruction resulting from lacrimal puncta agenesis. [10]. The third case involved non-twin female siblings who were both diagnosed with unilateral right dacryocystoceles. Given the limited data, this study concluded that the likely cause is a sporadic embriologic event [1].
6. The basis for an argument for a genetic basis for this case series lies in the bilaterality of the lesions occurring in a set of monozygotic twins. One possible explanation for why a genetic predisposition has not been better elucidated is that there is significant underreporting of this finding due to the large percentage of spontaneous resolution [1]. It stands to reason that there could be a genetic predisposition, which has not been established in a large majority of cases, as multiple members of a familial group with subclinical findings would never be evaluated or studied.

Conclusion:
Congenital dacryocystocele is a form of nasolacrimal duct obstruction that can be a challenging clinical entity in the ophthalmology and otolaryngology practice that deals with neonates. There have been few reported suggestive of familial predisposition. We report identical twin siblings with bilateral congenital dacryocystoceles that lends credence to a genetic basis.