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

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ABSTRACT

Embryogenesis of a congenital nasolacrimal duct (NLD) cyst is attributed to the failure of the Hasner membrane of the NLD system to cannulate. Prenatal diagnosis of congenital NLD cysts supports the argument for a developmental error, with a postnatal prevalence of 6%. 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. A case report and literature review were performed. 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 preterm 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 dacryocystoceles was made. Twin B initially presented with only bilateral eye discharge and CT showed a dilated NLD system. Twin B subsequently developed early signs of bilateral dacryocystoceles the following day. Both patients underwent lacrimal probing and endoscopic marsupialization of the dacryocystoceles. Biopsies were consistent with dacryocystocele. 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.*

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Congenital nasolacrimal duct (NLD) obstruction is thought to be a prenatal developmental failure to cannulate the NLD system. The lacrimal system begins to form in the 5th week of fetal development. Formation of a lumen in the lacrimal cord occurs in the 10th week of development, which coincides with cavitation of the inferior meatal lumen. Through canalization of the lacrimal cord, 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 ~5–6% of full-term newborns.

Congenital dacryocystocele is an uncommon condition in which cystic swelling of the lacrimal 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 Rosenmuller 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 Rosenmuller 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 bulging of mucosa at the lower end of the NLD into the nasal cavity, resulting in epiphora and possibly respiratory dysfunction 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 coexistence of an intranasal 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 NLD and cyst.

Treatment of epiphora associated with simple NLD obstruction is typically conservative, because the majority of patients can experience spontaneous resolution. Management of dacryocystoceles 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

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the nasolacrimal sac for uninfected dacryocystoceles before pursuing combined NLD probing and endoscopic nasal marsupialization.[2] If the condition does not resolve spontaneously, infection 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 1 month of age in persistent cases.

Epidemiologically, congenital dacryocystoceles is relatively uncommon and is estimated to occur in 1 of 3884 births.[3] It has been reported to be more common in female[4,5] and non-Hispanic white patients.[4] Although familial cases have been described sporadically,[1,4,6,7] 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-day-old female infants presented to the emergency department at our institution for evaluation of erythema, edema, and ocular discharge associated with swelling of the medial canthal regions. These ex-37-week female infants were delivered by cesarean section, and the pregnancy was complicated by preterm 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 postdelivery course in the newborn nursery for observation and were discharged to home. Twin A was seen by a primary care provider 1 day before admission and was treated with cephalexin and polymyxin B sulfate/trimethoprim sulfate ophthalmic ointment 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 (Fig. 1 *A*). The diagnosis of bilateral infected congenital dacryocystoceles was made. Twin B initially presented with only bilateral eye discharge and CT confirmed a dilated NLD system (Fig. 1 *B*). Twin B subsequently developed early signs of bilateral infected dacryocystoceles the following day. Both patients underwent lacrimal probing and endoscopic marsupialization of the bilateral nasal cysts, with biopsy specimens consistent with dacryocystoceles (Fig. 2, *A–D*). They were discharged from the neonatal intensive care unit within 24 hours.

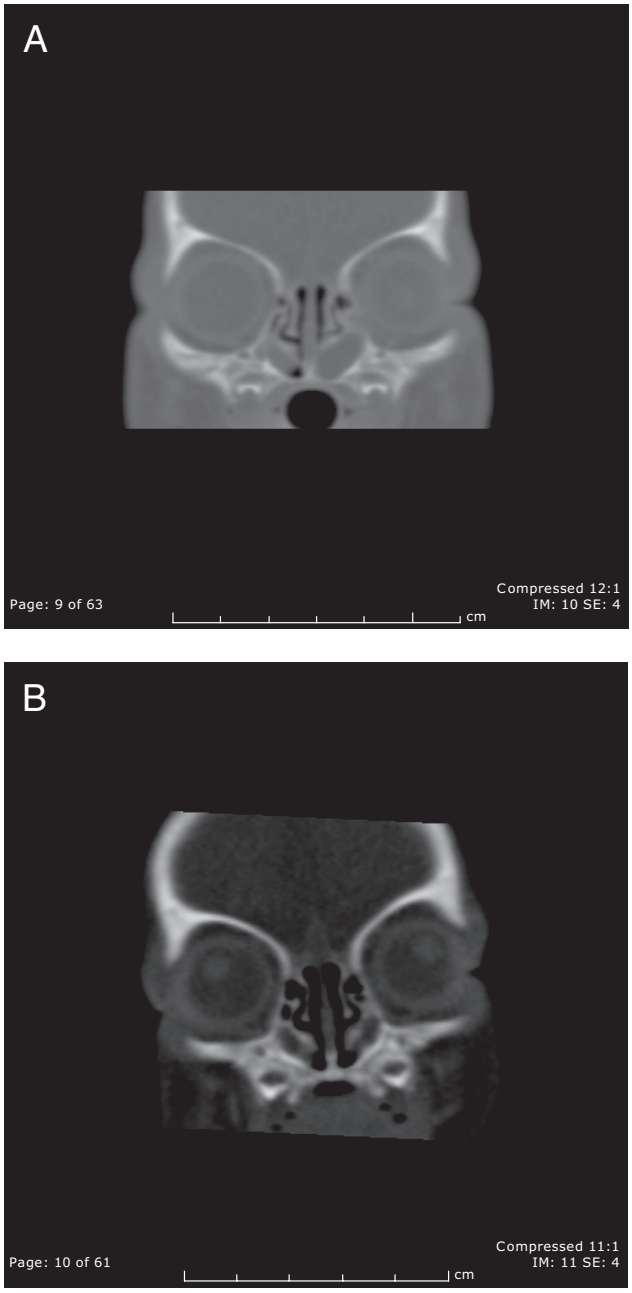

Figure 1. *Coronal computed tomography (A) of twin A and (B) of twin B showing bilateral cystic masses in the inferior meatus and a dilated nasolacrimal duct system.*

DISCUSSION

Congenital NLD obstruction is found relatively frequently and an incidence of 35–73% has been reported.[8] The majority spontaneously resolves during the 1st few weeks of life. Although a congenital dacryocystocele accompanied with a NLD cyst is reported to occur in 0.1% of infants during the 1st year of life,[9] 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 ad-

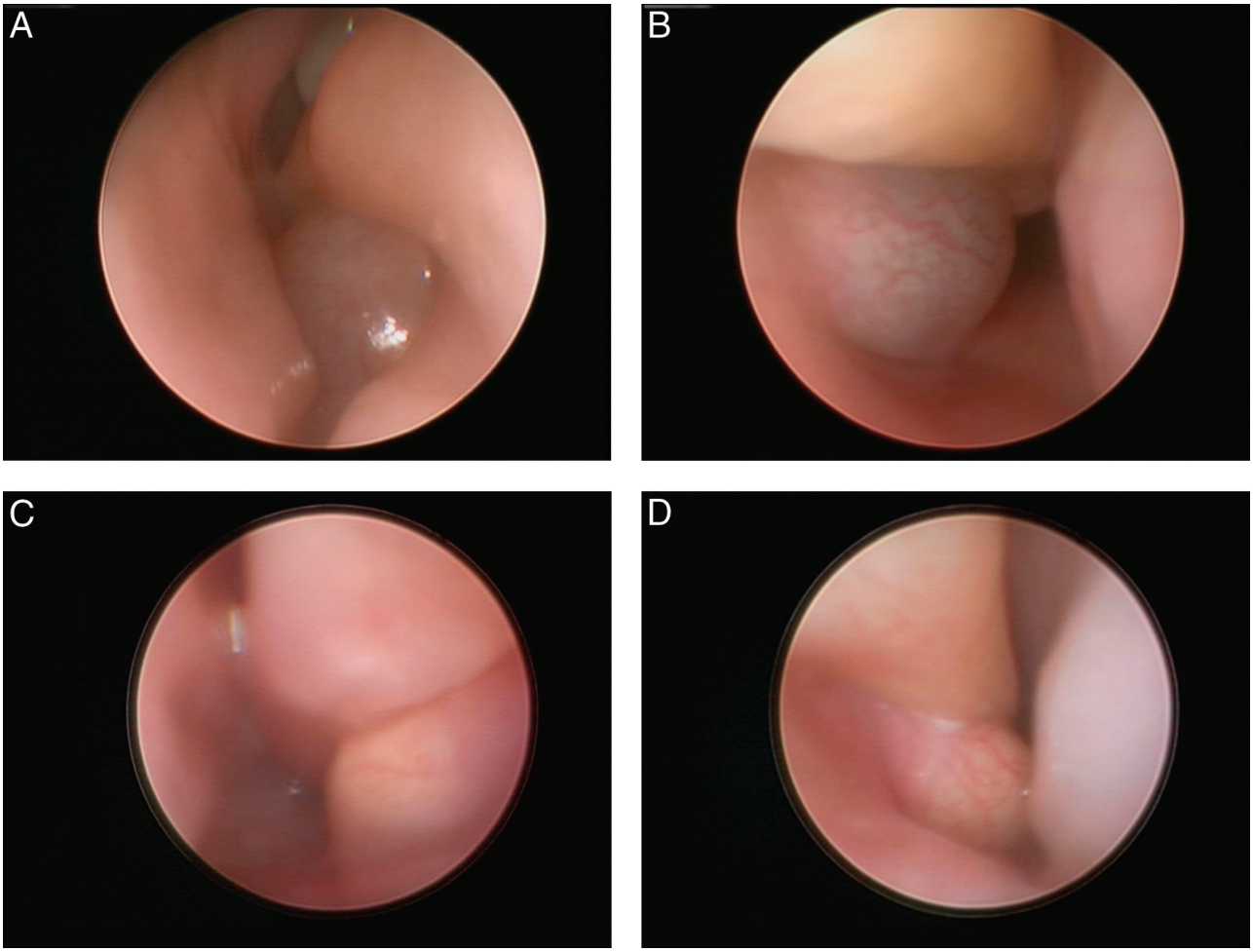

Figure 2. *Nasal endoscopy showing a cystic mass in the (A) left inferior and the (B) right inferior meatus of twin A. Nasal endoscopy showing a cystic mass in the (C) left inferior and the (D) right inferior meatus of twin B.*

vances the notion that this congenital development disorder has a genetic basis.

Potential inherited predisposition to dacryocystocele formation is suggested by increased female prevalence (three to nine times more commonly than male prevalence) and an increased incidence in white and Hispanic populations.[1] However, the increased prevalence in female subjects could be attributed to an anatomically narrower NLD. Traquair indicated a pattern of familial inheritance in 11% of patients in a large study of the etiology of dacryocystitis.[10] Further literature review showed three cases of familial NLD obstruction; however, two of the cases were associated with syndromes that resulted in NLD obstruction and were not related to isolated congenital duct obstruction. The first case involved a mother and daughter with branchio-oculo-facial syndrome with resulting congenital NLD obstruction.[6] The second case was a brother and sister with congenital NLD obstruction resulting from lacrimal puncta agenesis.[7] The third case involved nontwin female siblings who were both diagnosed with unilateral right dacryocystoceles. Given the limited data, this study concluded that the likely cause is a sporadic embryologic event.[1]

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 because of the large percentage of spontaneous resolution.[1,11–14] 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 NLD obstruction that can be a challenging clinical entity in the ophthalmology and otolaryngology practice that deals with neonates.[15–18] There have been few reported cases suggestive of familial predisposition. We report iden-

tical twin siblings with bilateral congenital dacryocystoceles that lends credence to a genetic basis.

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