

ARTICLE



Management of congenital nasolacrimal duct obstruction in down syndrome

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BACKGROUND: Congenital nasolacrimal duct obstruction (CNLDO) is common in Down Syndrome (DS), and more difficult to treat. Our purpose was to describe the management of CNLDO in paediatric patients with DS.

METHODS: Retrospective cohort study. Medical chart review of all DS patients diagnosed with CNLDO at the Division of Ophthalmology at the Children's Hospital of Philadelphia during a 12-year period (2009–2020). Main outcome measures included: Surgical interventions, primary probing outcome, rate of dacryocystorhinostomy (DCR) and/or conjunctivodacryocystorhinostomy (CDCR), and overall success.

RESULTS: 126 patients (236 eyes) were included, mean age of 1.8 ± 2.1 years (range 0.1–11.3 y), 110 (87%) had bilateral CNLDO. Mean follow-up time was 41 months. 84 patients (67%) underwent at least one surgical intervention; the mean number of surgical interventions in this group was 1.8 ± 1.4 per patient (range, 1–6). The most common primary intervention was probing ($n = 74$, 88%), mostly ($n = 57$, 68%) with monocalicular silicone intubation. Probing with silicone intubation had a higher success rate compared to probing alone ($P = 0.002$). Twenty (24% of 84) patients underwent DCR/CDCR during the follow-up period. Complete resolution was achieved in 123 patients (98%).

CONCLUSIONS: CNLDO in Down syndrome is associated with high rates of bilateral obstructions and with less favourable surgical outcomes. Many patients ultimately require a more robust surgical intervention such as DCR or CDCR. The use of monocalicular stent in initial probing was associated with a higher success rate, and would appear to be appropriate in all CNLDO-DS cases.

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INTRODUCTION

Down syndrome (DS), also called Down's syndrome, is the most common chromosomal anomaly associated with intellectual disability and is characterized by a variety of clinical features [1]. It has an approximate incidence of 1 of 800 births worldwide [1]. Common ophthalmic findings include refractive disorders, iris abnormalities, strabismus, retinal abnormalities, and cataract [2–6]. The most common oculo-facial findings are upward slanting of the palpebral fissure, epicanthal folds, blepharitis, ectropion, and epiblepharon [5, 7, 8]. The lacrimal drainage system is often affected, with a reported incidence of congenital nasolacrimal duct obstruction (CNLDO) as high as 30% [2, 7, 9, 10] and appears to be more difficult to treat [8, 11]. While the overall success of tear duct surgeries in children around the age of one is approximately 90% [12–14], the optimal surgical approach in DS is both unclear and apparently less successful [11].

In this study, we describe the surgical and medical management of all paediatric DS patients diagnosed with CNLDO treated at a single ophthalmology department between 2009–2020. Our purpose was to describe the types, and number of lacrimal interventions and to evaluate the surgical outcome.

METHODS

A single-centre retrospective medical chart review was performed for all children diagnosed with CNLDO who were treated by the department of ophthalmology at The Children's Hospital of Philadelphia (CHOP) over a 12-year period (2009–2020). The diagnosis of CNLDO was based on the history, ocular examination, and dye disappearance test. The collected data included patients' demographics, history of lacrimal surgical interventions done prior to examination at CHOP, and subsequent management, including medical and surgical interventions. All procedures began with a probing. When recorded in the chart, stenosis was graded manually (on a scale of 1–3) by the lacrimal surgeon.

Main outcome measures were types and number of surgical interventions, outcome of primary probing, dacryocystorhinostomy (DCR) and/or conjunctivodacryocystorhinostomy (CDCR) rate, and overall success rate (i.e. complete resolution of CNLDO symptoms).

The study was conducted in compliance with the United States Health Insurance Portability and Accountability Act and adhered to the tenets of the Declaration of Helsinki. Approval of this retrospective medical chart review was obtained from the local institutional review board of the CHOP.

Statistical analysis

In bilateral cases, only one eye (right) was included in the statistical analysis to prevent the effect of inter-eye correlation. The Chi-square test

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Table 1. Surgical intervention in 84 paediatric patients with down syndrome and congenital nasolacrimal obstruction.

| Patients characteristics | Number of intervention | | | | | |
|---|------------------------|------------|---------------------|-----------|---------------------|---------------------|
| | 1 | 2 | 3 | 4 | 5 | 6 |
| No. of patients (n, % of 84) | n = 84,100% | n = 32,38% | n = 14,20% | n = 9,11% | n = 6,7% | n = 5,6% |
| Age at procedure, years (mean ± SD) | 3.0 ± 2.4 | 5.1 ± 3.3 | 4.8 ± 2.3 | 9.5 ± 5.8 | 8.3 ± 4.0 | 8.7 ± 4.3 |
| Procedure type (n, % of patients) | | | | | | |
| Probing and irrigation | 17,20% | 3,9% | 0,0% | 0,0% | 0,0% | 0,0% |
| Monocanalicular stent intubation | 57,68% | 10,31% | 1,7% | 0,0% | 0,0% | 0,0% |
| Balloon dacryoplasty | 6,7% | 9,28% | 5,36% | 0,0% | 0,0% | 0,0% |
| w stent | 5,6% | 8,25% | 5,36% | | | |
| w/o stent | 1,1% | 1,3% | 0,0% | | | |
| Dacryocystorhinostomy (DCR) | 3,4% | 10,31% | 6,42% | 9,100% | 4,67% | 1,20% |
| Endoscopic | 2,3% | 7,22% | 4 ^a ,28% | 8,89% | 3 ^a ,50% | 1 ^a ,20% |
| External | 1,1% | 3,9% | 2,14% | 1,11% | 1,17% | 0,0% |
| Conjunctivodacryocystorhinostomy (CDCR) | 0,0% | 0,0% | 0,0% | 0,0% | 2,33% | 4,80% |

^aWith intraoperative topical mitomycin c (MMC) in 1 case.

was used to calculate proportional differences between categorical groups; the Yates' continuity correction was applied to analyze 2 × 2 tables. Independent samples Mann-Whitney test was used to compare means of NLD stenosis grade with binary variables. Independent samples t-test was used to compare mean number of interventions with binary variables. Binary logistic regression was used to find associations with surgical intervention and with DCR/CDCR rates. Statistical analysis was carried out using SPSS (version 26, SPSS Inc., Chicago, IL). All results are presented as mean ± standard deviation.

RESULTS

A total of 126 patients (236 eyes) were included, of which 70 (56%) were females. 110 (87%) had bilateral CNLDO; among the 16 unilateral obstructions, 11 (69%) were on the right side and 5 (31%) on the left. Mean (±SD) age was 1.8 ± 2.1 years (range 0.1–11.3 y). Mean follow-up time was 41.2 ± 45.0 months.

Ten patients (8%) had a history of previous lacrimal surgery prior to presentation at our institution. Additional ocular history included ptosis (*n* = 3), congenital nystagmus (*n* = 3), strabismus (*n* = 2), retinopathy of prematurity (*n* = 2), bilateral entropion (*n* = 1), bilateral retinal scar (*n* = 1), dermoid cyst (*n* = 1), and bilateral congenital cataract (*n* = 1).

Surgical Interventions

Eighty-four (67%) patients underwent at least one surgical intervention; 32 (38%) of these patients underwent at least additional one procedure (range, 1–6). The average age at presentation was higher in children who underwent surgical intervention (2.0 vs. 1.2 years in the conservative management group, *P* = 0.03, independent *t*-test). The mean number of surgical interventions was 1.8 ± 1.4 (Table 1). The probability of undergoing multiple procedures increased with the number of interventions: 32/84 (38%) of patients who underwent at least one surgical intervention underwent at least an additional procedure, 14/32 (44%) of patients who underwent at least two procedures underwent another, 9/14 (64%) of third, 6/9 (67%) of fourth, and almost all (5/6, 83%) patients who underwent at least five surgical interventions. After three failed interventions, DCR/CDCR was performed in all patients. Overall, 20 (24%) patients underwent DCR and/or conjunctivodacryocystorhinostomy with placement of lacrimal bypass tube (Lester Jones or Cox type tube, CDCR). Patients with bilateral NLDO were more likely to undergo at least one surgical intervention compared with patients with unilateral obstructions (AOR 4.8, *P* = 0.007, binary logistic regression), while gender and age at presentation were not found to be related.



Fig. 1 A 1.8-year-old patient with Down syndrome, bilateral congenital nasolacrimal duct obstruction, and right lacrimal sac fistula noted as a small dimple under the right medial canthus (arrow). The patient underwent bilateral probing, monocanalicular stent intubation into the lower puncta, and excision of the right lacrimal fistulous tract, with complete resolution of symptoms. Parent consent was received for the use of this figure.

Age at presentation, gender, and bilaterality were not associated with DCR/CDCR (binary logistic regression).

Lacrimal disorders

Upper lacrimal system disorders including punctal/canalicular/common canalicular disorders were observed in 25 (11%) eyes of 17 (13%) patients. These underwent more procedures than patients with a normal upper lacrimal system (2.2 vs. 1.1 respectively, *P* = 0.04, independent samples *t* test).

Three patients (2.3%) had lacrimal fistulae (Fig. 1). Two of these had right-side fistula and bilateral NLDO, both resolved after a single procedure of probing and irrigation (P&I) with stent placement and fistulectomy. The third case had bilateral fistulae and NLDO, with only mild symptoms; no procedure was performed, and the condition gradually improved over time.

Initial surgical intervention

Mean age at first intervention was 3.0 ± 2.4 years (range, 0.7–11.8 y). The most common primary interventions were probing with monocanalicular silicone intubation (*n* = 57, 68%) followed by probing alone (*n* = 17, 20%). Balloon dacryoplasty was performed in 6 cases (7%), and dacryocystorhinostomy (DCR) in 3 (4%) cases, endoscopic in 2 cases, and external in one (Fig. 2). Intraoperatively, stenosis grade as appreciated by the surgeon when probing was performed, was similar between males and females, was not correlated with initial procedure success rate, or with DCR/CDCR rates (*P* > 0.05 for all, independent samples Mann-Whitney test).

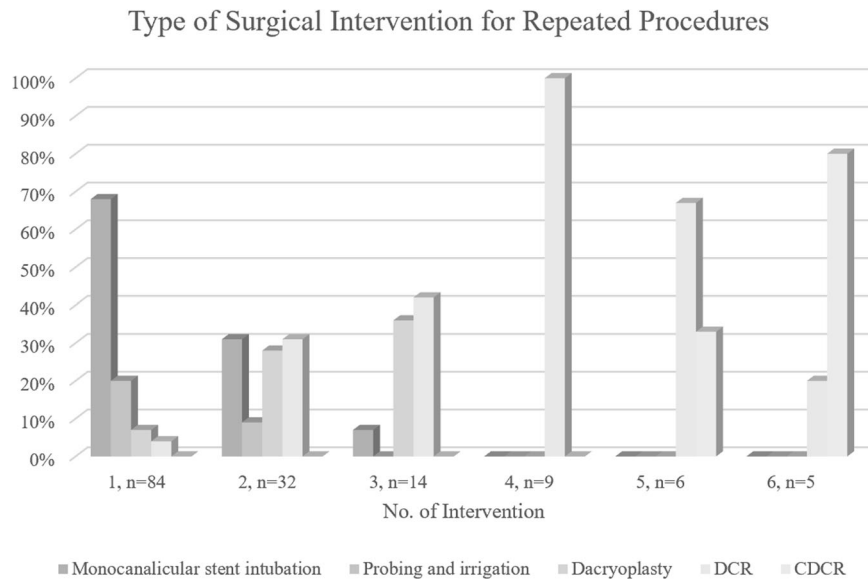


Fig. 2 Type of surgical intervention per procedure number in 84 paediatric patients with Down syndrome and congenital nasolacrimal duct obstruction. Probing with monocanalicular stent intubation was the most common procedure in the first intervention, while dacryocystorhinostomy (DCR) was most common between the third and fifth procedure; conjunctivodacryocystorhinostomy (CDCR) was most common in 5 patients who underwent six lacrimal procedures.

Table 2. Outcome of initial probing in 74 paediatric patients with down syndrome and congenital nasolacrimal obstruction.

| | Overall | Monocanalicular stent intubation | Probing and irrigation ^a | P value ^b |
|--|-----------|----------------------------------|-------------------------------------|----------------------|
| Number of patients | n = 74 | n = 57 | n = 17 | – |
| Age at probing, years (mean ± SD) | 2.8 ± 2.2 | 2.8 ± 2.3 | 2.5 ± 1.1 | NS ^{c,d} |
| Gender F:M | 39:35 | 28:29 | 11:6 | NS ^{c,e} |
| Success rate (n, % of patients) | 44, 59% | 40, 70% | 4, 24% | 0.002 ^e |
| Underwent DCR/CDCR ^f (n, % of patients) | 15, 20% | 7, 12% | 8, 47% | 0.005 ^e |
| Number of surgical interventions (mean ± SD) | 1.4 ± 1.3 | 1.4 ± 0.8 | 2.9 ± 1.8 | 0.003 ^d |

Higher success rates were observed when monocanalicular stent intubation was performed.

^aWithout monocanalicular silicone stent intubation.

^bComparing probing with stent vs. probing without stent.

^cNon-significant (>0.05).

^dIndependent samples *t*-test.

^eChi-square with Yates' continuity correction.

^fDCR dacryocystorhinostomy, CDCR Conjunctivodacryocystorhinostomy.

Initial intervention success rate

The mean age of the 74 patients who had probing with or without intubation as their initial intervention was 2.8 ± 2.2 years. The procedure was successful in 44 (59%) patients, while 28 (38%) underwent at least one additional procedure at a later stage, and surgical intervention was recommended for the additional two patients. Overall, 15 (20%) of the patients who underwent initial probing ultimately underwent DCR/CDCR during their follow-up period, all with complete resolution of their symptoms.

When comparing the 57 (68%) patients in whom monocanalicular intubation was performed to the 17 (32%) in which probing alone was performed, a significantly higher success rate was observed in the stent group ($n = 40/57$, 70% vs. $n = 4/17$, 24% respectively, $P = 0.002$, chi-square), and a significantly lower rates of DCR/CDCR during follow-up ($n = 7/57$, 12% vs. $8/17$, 47%, $P = 0.005$, chi-square). The average number of surgical interventions during follow-up period was lower in the stents group (1.4 vs. 2.9 respectively, $P = 0.003$, independent samples *t*-test) (Table 2).

Symptoms resolved after initial balloon dacryoplasty in 3 cases ($n = 3/6$, 50%) and in 2 of those who had a DCR ($n = 2/3$, 67%).

Final surgical intervention

Mean age at last intervention was 4.7 ± 4.0 y (range, 0.7–20.7 y). These included probing with monocanalicular stent intubation ($n = 44$, 52%), probing alone ($n = 7$, 9%), balloon dacryoplasty ($n = 13$, 15%), DCR ($n = 15$, 18%) mostly endoscopic ($n = 14$, 17%), and conjunctivodacryocystorhinostomy (CDCR) ($n = 4$, 5%).

Complications and outcome

Postoperative complications included corneal abrasion from monocanalicular stent ($n = 3$), punctal pyogenic granuloma ($n = 2$), keratitis ($n = 1$), corneal ulcer ($n = 1$), and preseptal cellulitis ($n = 1$), all resolved with conservative treatment. Resolution of NLDO symptoms was achieved in 123 (98%) patients. The mean follow-up time in the conservative management group ($n = 42$) was 25.8 ± 25.2 months; all but one patient were symptoms free at the end of follow-up ($n = 41/42$, 98%). The mean follow-up period in the intervention group was 48.8 ± 50.6 months; the symptoms resolved in all but two patients ($n = 82/84$, 98%). One of the these underwent P&I with stent intubation, and the other had P&I with stent intubation followed

by balloon dacryoplasty. In the three patients with persistent tearing (bilateral in two cases, unilateral in one), surgical intervention was recommended.

DISCUSSION

In this study, we evaluated the management of 126 paediatric patients (236 eyes) with DS and CNLDO. Two-thirds of the patients underwent at least one surgical intervention, with an average of 1.8 procedures per patient (range, 1–6). Patients with bilateral CNLDO were more likely to undergo surgical intervention. Initial probing was successful in 59% of patients. The success rate of initial probing with silicone intubation was significantly higher (70%), associated with fewer DCR/CDCR (12%), and overall had less procedures (1.4 on average per patient). Overall, 24% of these patients underwent DCR and/or CDCR during the follow-up period. Final success was achieved in 98% of patients.

As expected, the patients in the conservative group were younger than those in the interventional group. Although it may be difficult to predict if symptoms will resolve spontaneously; conservative management is commonly advised in patients younger than one year of age, and/or in children with minor symptoms [12].

The higher rate and complexity of CNLDO in DS are in part related to the unique facial morphology and abnormal persistence of Hasner's membrane or bony obstruction along the distal portion of the nasolacrimal duct (NLD) [2, 8, 11, 15]. The high rate of bilateral obstructions (87%) demonstrated in this study is in line with previous reports [8, 16–18]. In addition, various anatomic anomalies were reported in DS, and a decreased lacrimal pump function in these children has been demonstrated [2, 8, 11, 15]. For example, NLD in DS is more often complicated by anomalies of the upper lacrimal drainage system, as seen in 13% of our patients [8]. Congenital lacrimal fistulae are also more frequent in DS, as observed in 2.3% of our patients, remarkably higher than in the general population (1:2000) [19, 20].

Based on the diverse lacrimal pathology in DS, it is not surprising that we found significantly lower success rates of initial probing (59%) as compared to the 78%–97% success rate of in the general paediatric population [14, 21–23]. This rate is influenced by the use of monocalicular stent intubation, age at intervention, bilaterality, and complex obstructions. Of note, the mean age at first probing in this study was 2.7 years, relatively older than recommended for initial intervention, which might influence the outcome, as demonstrated in previous studies [22, 24, 25]. Nonetheless, 59% remains a low success rate even after taking this factor into consideration. Of note, when DCR is indicated, external approach may be more frequently utilized in DS due to thicker nasal bones [26].

The overall less favourable outcome in CNLDO-DS surgical interventions was demonstrated in this study by high failure rates of initial probing and balloon dacryoplasty, the high number of lacrimal procedures per patient, and the high rate of DCR/CDCR. This supports the results of previous studies which reported lower surgical outcome in DS-NLDO, and raises the question regarding the optimal initial treatment in this unique group of patients. Lueder et al. [11] demonstrated a higher success rate with balloon dacryoplasty compared with probing as an initial procedure (71% vs. 29%) in a series of 15 DS patients; this was not demonstrated in the current study, where success rate was 50% for balloon dacryoplasty and 59% in the probing group. Coats et al. [8] described surgical outcome in 22 children (38 eyes) with DS after 2 years of age, compared with 44 non-DS children (59 eyes), and reported a slightly lower success rate of initial probing and intubation in DS (76% vs. 81%), yet concluded that treatment of NLDO-DS has a success rate comparable to that for control patients of a similar age. In Coats' study, 7 (18%) eyes underwent more than 1 procedure, compared with 38% in our cohort. On the

other hand, on their series of 17 DS and 104 non-DS patients after failed probing, Lim et al. [18] reported similar success rates for probing with silicone intubation in DS, and concluded that NLD silicone intubation is a successful procedure for DS. These studies indicate that the initial surgical approach may be debatable, as no single intervention proved superiority over the other. Despite the worse outcome, the overall success rate (98%) is encouraging.

Limitations of this study stem from its retrospective design, thus precluding recommendation of a definitive treatment scheme.

In conclusion, CNLDO in DS is associated with higher rates of bilateral obstructions and with less favourable surgical outcomes after initial surgical intervention compared with the general paediatric population. More patients will ultimately require a more robust surgical intervention as DCR or CDCR. Overall, almost all patients achieve complete resolution. The use of monocalicular stent intubation in initial probing was associated with a higher success rate and is suggested to be selected as the primary procedure in all cases.

SUMMARY

What was known before

- Congenital nasolacrimal duct obstruction (CNLDO) is more common in Down syndrome (DS), and generally believed to be more difficult to treat in comparison with the general paediatric population.
- Only a few small case series discuss the management of these patients, and the surgical approach is debatable.

What this study adds

- We present a retrospective cohort of 126 paediatric patients (236 eyes) with DS and CNLDO, the largest cohort reported to date.
- Higher rates of bilateral obstructions and less favourable surgical outcomes were observed, compared with the general paediatric population. More patients underwent a more robust surgical intervention such as DCR or CDCR.
- Most importantly, the use of monocalicular stent intubation in initial probing was associated with significantly higher success rate and is advised in all DS cases.

DATA AVAILABILITY

The data that support the findings of this study are available from the corresponding author, DLP, upon reasonable request.

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AUTHOR CONTRIBUTIONS

DLP and ST designed the concept of this work and drafted this manuscript. JAK, WRK and KER critically reviewed the manuscript. All authors have read and approved the final manuscript.

COMPETING INTERESTS

The authors declare no competing interests.

ADDITIONAL INFORMATION

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