

## NASOLACRIMAL DUCT OBSTRUCTION IN DOWN SYNDROME CHILDREN: EVIDENCE FROM KARACHI

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### ABSTRACT

For many families raising a child with Down syndrome, watery eyes may seem like a minor inconvenience. Yet behind every tear may lie an overlooked condition: Nasolacrimal Duct Obstruction (NLDO). This study explores the prevalence, symptoms, healthcare-seeking behaviors, and treatment experiences of children with Down syndrome living in Karachi, Pakistan. Through responses collected from parents and caregivers, the research reveals a story not only of ocular health but also of healthcare accessibility, delayed diagnosis, parental awareness, and resilience. Findings indicate that nearly half of the children experienced persistent watery eyes, while only a minority received specialist consultation. Most families relied on conservative management such as eye massage and eye drops, and very few children underwent surgical intervention. The study highlights the urgent need for early screening, caregiver education, and equitable access to pediatric ophthalmic services.

**Keywords:** Down syndrome, nasolacrimal duct obstruction, ocular health, caregiver experiences, pediatric ophthalmology, qualitative interpretation, thematic findings

### 1. INTRODUCTION:

#### More Than Just Watery Eyes

Imagine a mother wiping tears from her child's eyes several times a day. Not tears of sadness, but tears that never stop flowing. For many children with Down syndrome, this is a daily reality. Down syndrome, one of the most common chromosomal conditions worldwide, is associated with numerous ocular manifestations, including nasolacrimal duct obstruction (NLDO). NLDO occurs when the tear drainage pathway becomes

partially or completely blocked, resulting in persistent tearing, eye discharge, recurrent infections, and discomfort. Children with Down syndrome experience a higher prevalence of NLDO than the general pediatric population due to anatomical and developmental differences affecting the lacrimal drainage system. Despite its frequency, NLDO often remains under recognized, especially in resource-limited settings where access to pediatric ophthalmology services may be restricted. This study sought to understand

not only the prevalence of NLDO but also the lived experiences of affected families in Karachi. Down syndrome (DS), caused by trisomy 21, is the most common chromosomal disorder worldwide and is associated with a broad range of systemic and ophthalmic abnormalities. Ocular manifestations in children with Down syndrome include refractive errors, strabismus, cataracts, keratoconus, blepharitis, and nasolacrimal duct obstruction (NLDO). Among these conditions, NLDO is frequently overlooked despite its significant impact on visual comfort, ocular health, and quality of life (Roizen & Patterson, 2003).

Nasolacrimal duct obstruction occurs when the lacrimal drainage pathway fails to open properly, preventing tears from draining from the eye into the nasal cavity. This obstruction results in persistent tearing (epiphora), recurrent eye discharge, chronic conjunctivitis, and increased susceptibility to ocular infections (Paul, Shepherd, & Gilbert, 2010). Children with Down syndrome have a higher prevalence of NLDO compared to the general pediatric population due to characteristic craniofacial abnormalities and developmental anomalies of the lacrimal drainage system.

#### **Down syndrome and Ocular Abnormalities**

Down syndrome affects approximately 1 in every 700 live births globally and remains one of the leading causes of intellectual disability (Centers for Disease Control and Prevention [CDC], 2023). The syndrome is associated with multiple ophthalmic disorders, with studies reporting ocular abnormalities in more than 60% of affected children (Creavin & Brown, 2009).

The anatomical features commonly observed in Down syndrome, including flattened nasal bridges, narrow nasolacrimal canals, midface hypoplasia, and altered connective tissue development, contribute significantly to the occurrence of NLDO. These structural differences may impair tear drainage and increase the likelihood of obstruction (Fong et al., 2013).

Research indicates that visual impairment in children with Down syndrome can negatively

influence cognitive development, educational performance, communication skills, and social integration. Therefore, early recognition and management of ophthalmic disorders are essential components of comprehensive healthcare for this population (Woodhouse et al., 2014).

#### **Epidemiology of Nasolacrimal Duct Obstruction**

Congenital nasolacrimal duct obstruction is one of the most common ocular conditions in infancy, affecting approximately 5–20% of newborns in the general population (MacEwen & Young, 1991). Most cases resolve spontaneously during the first year of life; however, persistent obstruction often requires medical or surgical intervention. Studies have consistently reported a substantially higher prevalence of NLDO among children with Down syndrome. Lueder (2000) found that lacrimal drainage abnormalities occur more frequently in children with chromosomal disorders, particularly Down syndrome, due to anatomical and developmental factors.

Similarly, Kratky et al. (1996) reported that children with Down syndrome experience significantly higher rates of lacrimal drainage dysfunction than their non-syndromic counterparts. These findings suggest that routine ophthalmic screening should be considered a standard component of Down syndrome healthcare programs.

#### **Pathophysiology of NLDO in Down syndrome**

The pathogenesis of NLDO in children with Down syndrome is multifactorial. Anatomical narrowing of the nasolacrimal duct, delayed canalization during embryonic development, and craniofacial dysmorphism contribute to impaired tear drainage (Mimura et al., 2005).

Furthermore, generalized hypotonia and connective tissue abnormalities commonly associated with Down syndrome may affect the function of the lacrimal pump mechanism, reducing the efficiency of tear drainage (Kim et al., 2017).

Persistent obstruction can lead to chronic tear stagnation, which creates a favorable environment for bacterial growth and recurrent ocular

infections. If left untreated, prolonged inflammation may result in dacryocystitis, conjunctivitis, and visual discomfort (Nelson & Wagner, 2011).

### **Clinical Manifestations and Diagnostic Challenges**

Children with NLDO typically present with excessive tearing, mucous discharge, eyelid crusting, and recurrent conjunctivitis. However, diagnosing NLDO in children with Down syndrome can be challenging because communication difficulties and intellectual disabilities may limit symptom reporting (Bull, 2020).

Several studies have highlighted delays in diagnosis among children with developmental disabilities. Parents and caregivers may attribute excessive tearing to allergies, environmental factors, or minor infections rather than recognizing it as a sign of lacrimal obstruction (Mills & Bodman, 2018).

The diagnosis of NLDO is generally established through clinical examination, fluorescein dye disappearance testing, lacrimal irrigation, and imaging studies when necessary. Early identification remains critical for preventing long-term complications and reducing caregiver burden (Kushner, 2014).

### **Public and Private Healthcare Disparities**

Healthcare access remains an important determinant of ocular health outcomes. In developing countries such as Pakistan, disparities often exist between public and private healthcare sectors regarding diagnostic facilities, specialist availability, waiting times, and treatment accessibility (World Health Organization, 2023).

Children receiving care in public healthcare institutions may face delays in specialist referrals and limited access to pediatric ophthalmology services. Conversely, private healthcare facilities generally offer more rapid diagnostic evaluations and advanced treatment options but may remain inaccessible to low-income families due to financial constraints.

Studies examining healthcare inequalities have demonstrated that socioeconomic factors significantly influence early diagnosis and treatment outcomes in pediatric eye diseases (Marmot et al., 2020). These disparities may be particularly relevant for children with Down syndrome, who frequently require multidisciplinary and long-term healthcare services.

### **Psychosocial Impact on Families**

The burden of chronic ocular disorders extends beyond the affected child and influences family well-being. Persistent tearing, recurrent infections, frequent medical appointments, and surgical interventions can create emotional, financial, and logistical challenges for caregivers (King et al., 2016).

Parents of children with Down syndrome often report increased stress associated with managing multiple health conditions simultaneously. Delays in diagnosis or limited access to specialized care may further exacerbate caregiver anxiety and reduce quality of life (Skotko, Levine, & Goldstein, 2011).

Consequently, family-centered healthcare approaches emphasizing caregiver education, counseling, and support services are essential for improving clinical outcomes and treatment adherence.

Although international literature has documented the association between Down syndrome and nasolacrimal duct obstruction, evidence from Pakistan remains extremely limited. Few studies have explored the prevalence, clinical presentation, and healthcare experiences of children with Down syndrome affected by NLDO. Moreover, comparative investigations examining differences between public and private healthcare sectors are scarce. Understanding these disparities is essential for informing healthcare policy, improving service delivery, and ensuring equitable access to ophthalmic care.

The literature demonstrates that children with Down syndrome are at a significantly increased risk of developing nasolacrimal duct obstruction due to anatomical, developmental, and functional

abnormalities of the lacrimal drainage system. Delayed diagnosis, healthcare access barriers, and limited awareness contribute to ongoing morbidity and reduced quality of life. Given the scarcity of local evidence, research investigating NLDO among children with Down syndrome in Karachi is both timely and necessary. Findings from such studies can support evidence-based screening programs, caregiver education initiatives, and policy interventions aimed at improving ophthalmic care for this vulnerable population.

### 3. Research Methodology

A cross-sectional study was conducted among children with Down syndrome associated with the Karachi Down Syndrome Program (KDSP). Data were collected through structured questionnaires administered to parents and caregivers. The questionnaire explored symptoms, healthcare utilization, treatment patterns, and outcomes related to NLDO.

A total of **300 valid responses** were analyzed. The majority of participants were between 1 and 10 years of age, reflecting the age group in which NLDO is most frequently recognized.

### Thematic Findings

**Theme 1: Tears That Parents Learn to Live With**  
One of the strongest themes emerging from the data was the normalization of symptoms by caregivers.

Nearly **47.3% of children experienced persistent watery eyes**, making it the most frequently reported symptom.

For many families, watery eyes became such a routine occurrence that it was often viewed as a characteristic of the child rather than a medical condition requiring intervention.

"The tears were always there. We thought it was just part of having Down syndrome."

This finding suggests a gap between symptom recognition and understanding of underlying pathology.

### Theme 2: Living with Discomfort Beyond the Tears

NLDO was not limited to excessive tearing.

Many children experienced additional symptoms:

- Frequent eye discharge (18.3%)
- Eye redness (17.7%)
- Recurrent eye infections (13.7%)
- Light sensitivity (13.3%)
- Frequent eye rubbing (13.3%)

These symptoms suggest that NLDO can affect comfort, visual wellbeing, and quality of life, even when caregivers perceive it as a minor issue.

### Theme 3: The Silent Barrier of Delayed Healthcare Access

A particularly important finding was the low rate of specialist consultation.

Although symptoms were common, only **24% of caregivers had consulted an eye specialist** regarding their child's condition.

This theme reflects systemic barriers including:

- Limited awareness of NLDO
- Financial constraints
- Transportation difficulties
- Scarcity of pediatric ophthalmology services

The data suggest that many children remain undiagnosed until symptoms become more severe.

### Theme 4: Parents as Primary Care Providers

Before reaching specialist care, most families attempted home-based management.

Common interventions included:

- Eye massage (32.3%)
- Eye drops (20.3%)

Parents often became the first line of treatment, relying on advice from relatives, community members, or primary care providers.

The significant relationship between gender and utilization of eye massage suggests differences in caregiving patterns or treatment-seeking behaviors among families.

### Theme 5: Surgery as a Last Resort

Another recurring theme was the preference for conservative management.

Only:

- 4% of cases involved discussion of surgery.

- 3% underwent surgical intervention.
- This finding indicates that both families and healthcare providers tend to reserve surgery for persistent or severe cases.
- For most children, management focused on observation, massage, and symptomatic relief.

#### **Theme 6: Hope through Natural Improvement**

Not all stories in the dataset were marked by ongoing symptoms.

Approximately 11% of caregivers reported noticeable improvement after treatment, while some children experienced spontaneous improvement over time.

This theme highlights the importance of monitoring and individualized treatment planning rather than assuming all cases require invasive intervention.

#### **Theme 7: Public and Private Healthcare – Two Different Journeys**

The study identified differences between public and private healthcare experiences.

Children receiving care through private facilities were more likely to receive:

- Earlier diagnosis
- Faster specialist referral
- More comprehensive ophthalmic evaluation

In contrast, public-sector families frequently encountered:

- Delayed consultations
- Resource limitations
- Longer waiting periods

These differences underscore inequities in healthcare access that may influence long-term outcomes.

#### **4. Discussion**

The findings demonstrate that NLDO represents more than an ophthalmic condition. It is intertwined with caregiver awareness, healthcare accessibility, and social determinants of health. Nearly half of the children experienced symptoms suggestive of NLDO, confirming its importance as a common ocular manifestation of Down syndrome. However, the disparity between symptom prevalence and specialist consultation

suggests that many cases remain unrecognized or untreated. This observation aligns with international literature indicating that children with Down syndrome often experience delayed identification of ocular conditions. The predominance of conservative management reflects both clinical practice and practical realities within resource-limited settings.

#### **Implications for Practice**

This study highlights several priorities:

1. Routine ophthalmic screening for all children with Down syndrome.
2. Parent education programs on recognizing NLDO symptoms.
3. Early referral pathways for pediatric ophthalmology services.
4. Training of primary healthcare providers to identify ocular manifestations.
5. Strengthening access to specialized eye care within public hospitals.

#### **5. Conclusion**

**Behind every watery eye is a child, a family, and a story.**

This research demonstrates that nasolacrimal duct obstruction is a common but often underrecognized condition among children with Down syndrome in Karachi. While many families successfully manage symptoms through conservative measures, delayed diagnosis and limited access to specialist care remain significant challenges. Early detection, improved caregiver awareness, and equitable healthcare access are essential for improving quality of life and preventing long-term complications.

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