SCIENTIFIC REPORT

Results of lacrimal assessment in patients with congenital clinical anophthalmos or blind microphthalmos

M P Schittkowski, R F Guthoff

Br J Ophthalmol 2007;**91**:1624–1626. doi: 10.1136/bjo.2007.120121

Aim: To report clinical findings relating to the lacrimal system in congenital clinical anophthalmos and severe blind microphthalmos.

Methods: A retrospective (up to 2003) and prospective (2004 onwards) study of the notes of 60 consecutive patients treated surgically with highly hydrophilic self-inflating expanders for congenital anophthalmos or severe blind microphthalmos between 1997 and 2006. The lacrimal drainage system was always probed and irrigated under general anaesthesia before any other procedure was started.

Results: Nine patients were excluded due to possible misdiagnosis because of previous lid or orbit surgery elsewhere or due to missing data. The analysis therefore included 23 girls and 28 boys aged between 1 and 90 months (median age: 4 months). Twenty-three patients presented with unilateral and 18 with bilateral anophthalmos, and 10 had unilateral microphthalmos; consequently, 102 orbits (of which, 69 were with probable pathology) were available for assessment. In unilateral cases, the lacrimal system on the normal side was never affected. On the anophthalmic or microphthalmic side, the lacrimal system was normal in 17 orbits only (24.6%). The most frequent finding was canalicular stenosis (40 orbits; 58%). Common canaliculus stenosis was observed in 5 orbits (7.3%) and nasolacrimal duct obstruction in 7 orbits (10.1%). There were no cases of punctal anomaly.

Conclusions: In congenital clinical anophthalmos the lacrimal system is affected in up to 78% of cases, mostly due to canalicular stenosis. Even if there is no clear evidence of an embryological connection, this association is certainly not a random finding.

In a case report published in 2003, Oguz *et al*[8] suggested that congenital nasolacrimal duct occlusion might represent a "possible new association" with clinical anophthalmos. The aim of the present study was to substantiate this hypothesis by analysing the notes for all patients ever treated in our hospital with hydrogel expanders for congenital clinical anophthalmos or blind microphthalmos.[12 13]

MATERIAL AND METHODS

Patients

Our analysis included the notes for all 60 consecutive patients who were surgically treated (at least once) with highly hydrophilic self-inflating expanders for congenital anophthalmos and severe microphthalmos over a 10-year period from 1997 to 2006.

The study was performed retrospectively up to 2003; consequently, all patients without lacrimal assessment *before* first-time lid or orbit surgery were excluded to avoid misdiagnosing iatrogenic lacrimal pathology as congenital. From 2004 onwards, the study was continued prospectively: all patients underwent lacrimal assessment prior to first-time surgery. Patients who had previously undergone surgery or had even been probed elsewhere were also excluded from this study.

Lacrimal assessment

Before any lid or orbit surgery was started, the lacrimal drainage system was investigated under general anaesthesia. The findings were assessed, as recommended in Jane Olver's *Colour Atlas of Lacrimal Surgery*:[10]

- First, we confirmed that all four puncta were present. Aplasia, stenosis or membranous occlusion was ruled out. The relative position of the upper and lower puncta to the lid and to each other was checked.
- Second, the lacrimal system was probed and irrigated. The punctum was gently dilated using a regular punctal dilator. A 2-ml syringe with standard saline solution and a Bangerter cannula (manufacturer: Geuder AG, Heidelberg) were used for further probing and irrigation. This cannula has a blunt tip and a side-aperture to permit liquid outflow. The cannula was inserted first vertically and then horizontally following the anatomy of the canaliculus.
- In cases of stenosis, the cannula was marked at that point before entry to the punctum. After cannula retraction, the distance between the marked point and the tip was measured in millimetres and documented as the length of the canaliculus before the stenosis.
- If the cannula could be advanced into the sac, irrigation was then performed. Success was confirmed by placing a small suction catheter into the nose, where fluid flow could be observed immediately after irrigation from above. If any doubt remained, fluorescein dye was used additionally to confirm findings obtained previously.

RESULTS

Fifty-one out of 60 patients were included in our analysis: 9 were not eligible because they fulfilled the exclusion criteria referred to above. The study included 23 girls and 28 boys aged between 1 and 90 months (median age: 4 months). Twenty-three patients presented with unilateral and 18 with bilateral anophthalmos, and 10 had unilateral microphthalmos. In summary, 102 orbits were available for assessment, including 69 orbits with probable pathology and 33 orbits in unilateral disease that were probably healthy.

In unilateral disease, the lacrimal system on the normal side was never affected, and there were not even any cases of typical congenital nasolacrimal duct obstruction (CNLDO). On the anophthalmic or microphthalmic side, the lacrimal system was normal in 17 orbits only (24.6%). The most frequent finding was canalicular stenosis, which was encountered in 37 orbits (53.6%) in both canaliculi and in 3 orbits (4.4%) in one canaliculus (see fig 1). Typically, the blockage was detected

Abbreviation: CNLDO, congenital nasolacrimal duct obstruction

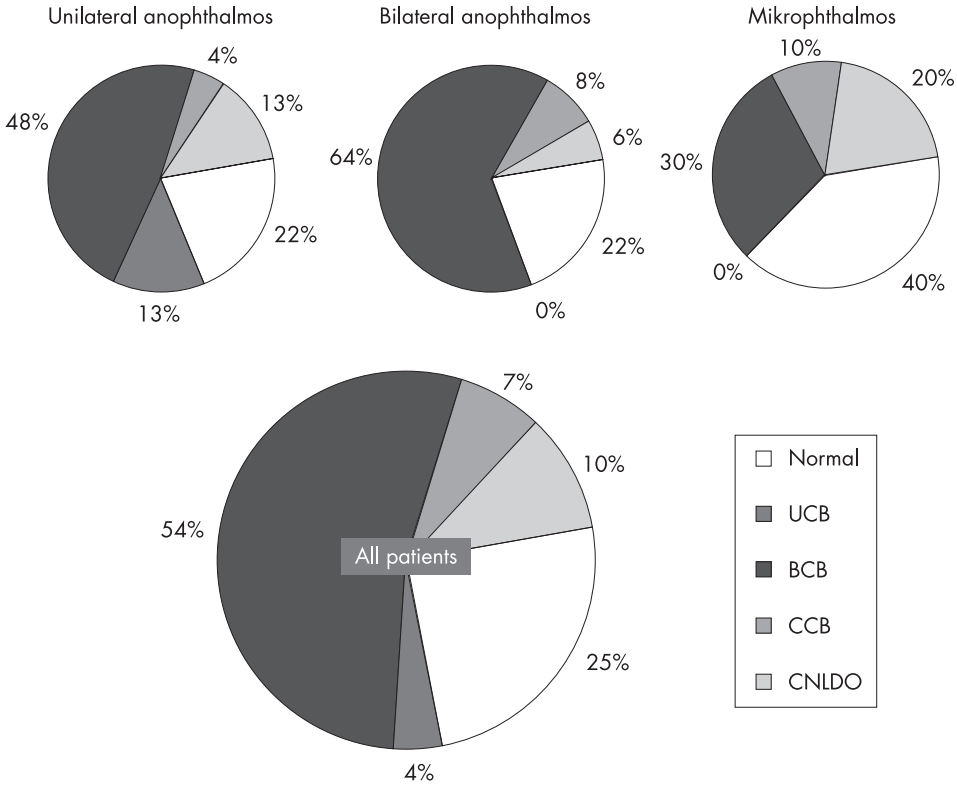

Figure 1 Results of lacrimal assessment in congenital anophthalmos and microphthalmos (UCB, uni-canalicular block; BCB, bi-canalicular block; CCB, common canalicular block; CNLDO, congenital nasolacrimal duct obstruction).

after 4 mm (range: 1–8 mm, standard deviation: 1.85 mm). Common canaliculus stenosis was found in 5 orbits (7.3%). Nasolacrimal duct obstruction was recorded in 7 orbits (10.1%), and we have been able to restore patency in all these cases.

Minor differences were noted, depending on the underlying disease (fig 1): patients with microphthalmos were less affected than those with anophthalmos. In anophthalmos, more children were found to have bi-canalicular stenosis in bilateral than in unilateral disease.

Punctal absence or atresia was not observed. There were no instances of supernumerary puncta or canaliculi, anomalies of punctal shape or position, or punctal ectopia.

DISCUSSION

Congenital disorders

To our knowledge, this is the first report describing systematic lacrimal assessment in congenital anophthalmos or blind microphthalmos. The lacrimal system was found also to be involved in as many as 78% of our patients with congenital clinical anophthalmos or blind microphthalmos. Obstruction of the lacrimal system was principally due to canalicular stenosis. Although there is no clear evidence of an embryological connection, this association is certainly not a random finding.

Embryology

The lacrimal passages develop along the line of the cleft between the lateral nasal and the maxillary processes.[3] Beneath the surface of this line, a solid rod of ectodermal cells can be found in the 4-week-old embryo. In the 6-week-old fetus, the cord detaches from the surface and becomes surrounded by mesenchymal tissue (fig 2). Also beginning in week 6, a second epithelial cell cord appears to emerge from the nasal cavity. The two cords elongate and approximate but remain separate before fusing later.

Canalisation begins with the disintegration of the central cells in the cord in Month 3 and is thought to be complete in Month 4 (fig 3).[3] The process starts at the upper (ocular) end and progresses downwards, while the same phenomenon unfolds in the second epithelial outgrowth from the nasal cavity. Final unification usually takes place at the end of Month 6 but may be delayed for months after birth.

The lacrimal puncta open between Month 6 and 7 of gestation, together with eyelid separation.[3] Since we were unable to detect any punctal pathology, it may be concluded that all the pathology reported in our study had to have developed before that time. This insight is important when discussing any accompanying pathology.

Congenital deformities

Developmental anomalies of the lacrimal passages are uncommon, and they follow the lines suggested by their embryological history:[3]

- Punctal absence and atresia: Although canalicular budding is normal, there is a dehiscence failure of the overlying conjunctival epithelium. The persistent fine membrane may be perforated by a minute orifice. It is very rare for there to be no clinical indication of the site of the punctum. If the pathology is isolated, it more often affects the inferior puncta, but usually all four puncta are involved.[2]
- Punctal ectopia: Anomalies in punctal shape or position have seldom been reported. Some instances of medial displacement have been described in the literature.[2]
- Supernumerary puncta and canaliculi: Duke-Elder has summarised a number of different clinical case histories reported in the literature to date;[2] these had their origin in an outspreading of multiple buds from the embryological lacrimal sac.
- Canalicular stenosis: By contrast with the conditions mentioned above, canalicular stenosis was encountered in our patient population where it was the most common lacrimal pathology. Canalicular stenosis might be due to a failure in budding or in canalisation of the solid rod.[2] Because budding starts from the lacrimal sac and not from the lids, the latter cause is more likely, especially if the amount of canaliculus that can be probed is significant.
- Nasolacrimal duct atresia: The failure of canalisation is characterised by a "persistent membrane formed by two

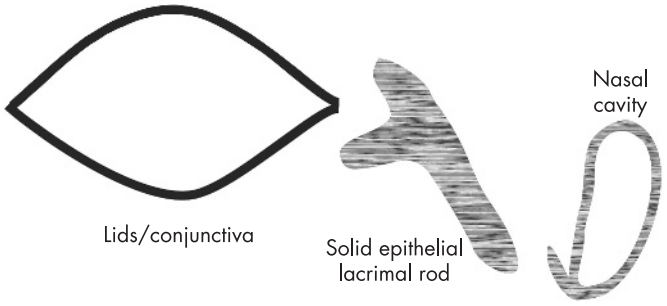

Figure 2 Schematic diagram illustrating the development of the lacrimal passages in a 6-week-old embryo (modified from Duke-Elder).[3] Note the bud-like outgrowth of the canaliculi from the lacrimal cord and the formation of a second cord starting from the primitive nasal cavity.

layers of cells from the lining of the nasolacrimal duct and from the nasal mucosa''.[9]

Anophthalmos is the most severe malformation of the eye, followed by blind microphthalmos. Gundlach *et al*[5] suggested that this anomaly is a developmental field defect, as defined by Opitz.[11] According to Gundlach and Pfeifer,[4] it is located in the diacephalic region, which is the border zone between the frontonasal and the posterolateral regions of the face. Because this stretches from the temple and traverses the orbit as far as the lateral part of the nose, it also influences the formation of the lacrimal passages. This theory may help to explain the association of the pathologies discovered in this study, and why—if the anomaly is seen as a process in this developmental field—the globe or the lacrimal system is not harmed in isolation.

Therapy

Aside from addressing an academic issue, we also need to consider whether this study reveals any practical implications for therapy. In CNLDO, it is indisputable that functional epiphora, and in particular the risk of dacryocystitis, constitutes an indication for probing if spontaneous resolution fails to materialise as the nose matures. The only matter for debate is the patient's age at which manipulation of the system should commence.[6 7 9] In our patients, simultaneous probing was successful in all 7 orbits affected by CNLDO.

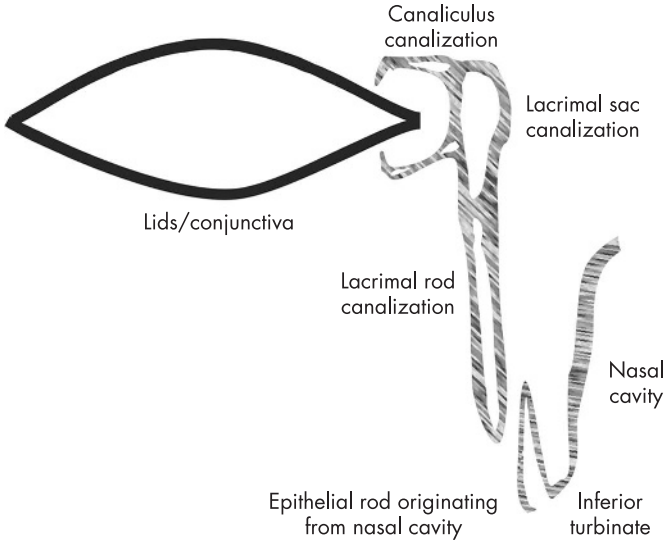

Figure 3 Further development and canalisation of the lacrimal rod up to Month 4 (modified from Duke-Elder).[3] Note that canalisation commences in multiple locations with isolated cavities that eventually unite to form a continuous central lumen.

Our patients tended to present with common canaliculus stenosis or, more frequently, stenosis of both the inferior and superior canaliculus. Because of these findings, dacryocystitis is never to be expected because of the absence of any reservoir for fluid and bacteria. The only symptom that might potentially be experienced is epiphora, and even our oldest patients never had that complaint.

According to Wang *et al*,[14] canalicular agenesis may occur in conjunction with CNLDO and is therefore complicated to treat. In adults, bypass surgery with a Lester–Jones tube (LJT) is recommended.[15] The long-term results are promising, especially in patients with canalicular obstruction,[16] and this particular technique has also been used in children.[1 17 18]

Welham and Hughes reported that the LJT success rate would be highest in congenital anomalies (94%) compared with functional problems (50%), infections (88%) or trauma (89%).[17] Where surgery is performed, an open technique is preferable so that retrograde canalicular surgery can be attempted.[18]

To date, we believe that there is no need for surgery if there are no clinical symptoms and no complaints at all; this conclusion will be revisited as our patient population is followed up over the coming years.

Authors' affiliations
M P Schittkowski, R F Guthoff, University Eye Department, Rostock University, Rostock, Germany

Competing interests: None declared.

Correspondence to: Dr Michael P Schittkowski, University Eye Department, Rostock University, Doberaner Strasse 140, D-18055 Rostock, Germany; michael.schittkowski@med.uni-rostock.de

Accepted 1 June 2007
Published Online First 13 June 2007

REFERENCES

1 **Cahill KV**, Burns JA. Management of epiphora in the presence of congenital punctal and canalicular atresia. *Ophthal Plast Reconstr Surg* 1991;**7**:167–72.
2 **Duke-Elder S**. The lacrimal apparatus. In: Duke-Elder S, ed. *System of ophthalmology. Vol. III Normal and abnormal development. Pt. 2 Congenital deformities*, St. Louis: CV Mosby, 1963:911–41.
3 **Duke-Elder S**, Cook C. The lacrimal passages. In: Duke-Elder S, ed. *System of ophthalmology. Vol. III Normal and abnormal development. Pt. 1 Embryology*, St. Louis: CV Mosby, 1963:241–6.
4 **Gundlach KKH**, Pfeifer G. Classification of facial malformations. *Int J Oral Surg*, 1981;**10**(Suppl 1), 267–72.
5 **Gundlach KKH**, Guthoff RF, Hingst V, *et al*. Expansion of the socket and the orbit for congenital clinical anophthalmia. *Plast Reconstr Surg* 2005;**116**:1214–22.
6 **Kashkouli MB**, Beigi B, Parvaresh MM, *et al*. Late and very late initial probing for congenital nasolacrimal duct obstruction: what is the cause of failure? *Br J Ophthalmol* 2003;**87**:1151–3.
7 **Katowitz JA**, Michael GW. Timing of initial probing and irrigation in congenital nasolacrimal duct obstruction. *Ophthalmology* 1987;**94**:698–705.
8 **Oguz H**, Ozturk A, San I. Congenital nasolacrimal duct occlusion with clinical anophthalmos: a possible new association. *Ophthalmic Genet* 2003;**24**:181–5.
9 **Olver J**. Paediatric lacrimal surgery. In: Olver J, ed. *Colour atlas of lacrimal surgery*. Oxford: Butterworth-Heinemann, 2002:69–89.
10 **Olver J**. Lacrimal assessment. In: Olver J, ed. *Colour atlas of lacrimal surgery*. Oxford: Butterworth-Heinemann, 2002:39–67.
11 **Opitz JM**. Editorial comment: The developmental field concept. *Am J Med Genet* 1985;**21**:1.
12 **Schittkowski MP**, Gundlach KKH, Guthoff RF. Treatment of congenital clinical anophthalmos with high hydrophilic hydrogel expanders. *Ophthalmologe* 2003;**100**:525–34.
13 **Schittkowski MP**, Guthoff RF. Injectable self inflating hydrogel pellet expanders for the treatment of orbital volume deficiency in congenital microphthalmos: preliminary results with a new therapeutic approach. *Br J Ophthalmol*, 2006;**90**:1173–7.
14 **Wang JK**, Lai PC, Liao SL. Punctual and canalicular agenesis presented with congenital nasolacrimal duct obstruction. *Graefes Arch Clin Exp Ophthalmol* 2002;**240**:960–1.
15 **Welham RA**. Canalicular obstruction and the Lester–Jones tube: what to do when all else fails. *Trans Ophthalmol Soc UK* 1973;**93**:623–32.
16 **Welham RA**, Guthoff R. The Lester–Jones tube: a 15-year follow-up. *Graefes Arch Clin Exp Ophthalmol* 1985;**223**:106–8.
17 **Welham RA**, Hughes SM. Lacrimal surgery in children. *Am J Ophthalmol* 1985;**99**:27–34.
18 **Yuen SJ**, Oley C, Sullivan TJ. Lacrimal outflow dysgenesis. *Ophthalmology* 2004;**111**:1782–90.