# Congenital glaucoma

**KIMBERLEY TAN** BSc(Med), MB BS, FRANZCO

**Clear and nonenlarged corneas, nasolacrimal duct obstruction and watery eyes. Would you suspect congenital glaucoma?**

## Case presentation

A six-month-old boy with a complex craniofacial disorder presented to the ophthalmology clinic with a chronic watery eye (Figure 1). He had a small, intermittent, right-sided divergent squint related to his craniofacial disorder, but his visual ability was normal for his age. On examination, his eyes were not enlarged and both corneas were clear.

The patient's lower tear meniscus was visibly enlarged, suggesting a diagnosis of congenital nasolacrimal duct obstruction (NLDO). The application of pressure over his right nasolacrimal sac resulted in the expression of clear fluid from the puncta, which supported the diagnosis.

Given that the patient was not experiencing any complications from the NLDO, conservative therapy consisting of hydrostatic lacrimal sac massage three to four times each day and the occasional course of chloramphenicol eye drops (Chloromycetin Eye Drops, Chlorsig, Minims Chloramphenicol); four times daily for three to four days as required, were prescribed .

Fifteen months later, a formal examination of the boy's eyes was performed under general anaesthesia while he was undergoing airway and bowel surgery. The examination was performed in this way because the patient had proved very difficult to examine while awake.

Dr Tan is an Ophthalmology Staff Specialist at the Sydney Children's Hospital, Randwick, Sydney, and a Visiting Medical Officer at the Royal North Shore Hospital, St Leonards, Sydney, NSW.

The eye examination revealed changes consistent with chronic glaucoma. The boy's intraocular pressure (IOP) was significantly elevated in the right eye (25 mmHg) and the left eye (21 mmHg). The normal IOP is below 21 mmHg. He was found to have glaucomatous optic neuropathy of the right eye without obvious corneal opacity. His tear ducts were probed. Topical medical therapy was commenced with one drop daily of timolol 0.25% (Tenopt, Timoptol, Timoptol-XE) in each eye and the patient was maintained on this regimen. His general health precluded surgical management of his glaucoma. At his last visit, when the patient was 3 years of age, his vision was good at 6/15 (right) and 6/19 (left), tested with letter matching. The patient's IOP was also controlled and his epiphora (tear overflow) had improved but a component of NLDO was still present.

## Comment

### A difficult presentation

Congenital NLDO is common and affects up to 20% of children in the first year of life. The condition is particularly prevalent in children with craniofacial disorders and has been shown to resolve spontaneously by end of the first year in 96% of cases.[1] A significant component of the child's case of persistently watering eyes was attributable to raised IOP, which can cause oedema of the corneal epithelium and the stroma in young children. Children usually present with epiphora and photophobia, both of which would have contributed to the difficulties associated with this patient's eye examination.

Figure 1. Chronic watery right eye without ocular injection or purulent discharge.

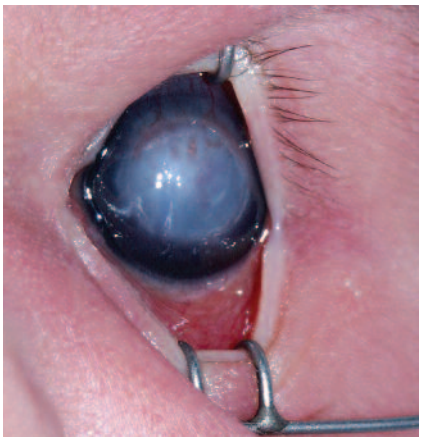

Figure 2. Severe cloudy cornea from congenital glaucoma with anterior segment dysgenesis.

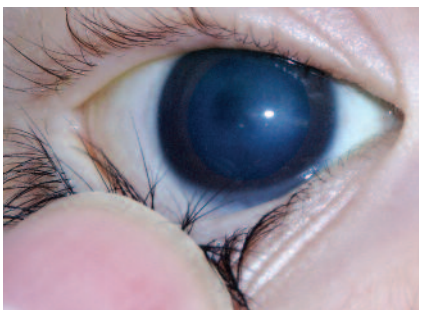

Figure 3. Less severe patchy corneal clouding.

### Congenital glaucoma

Congenital glaucoma is a rare sight-threatening condition that usually presents with a cloudy cornea in young infants. The cloudy cornea is usually apparent to the observer (Figures 2 and 3). Urgent treatment to lower the IOP and clear the optical media is required to prevent irreversible vision loss from optic neuropathy or amblyopia.

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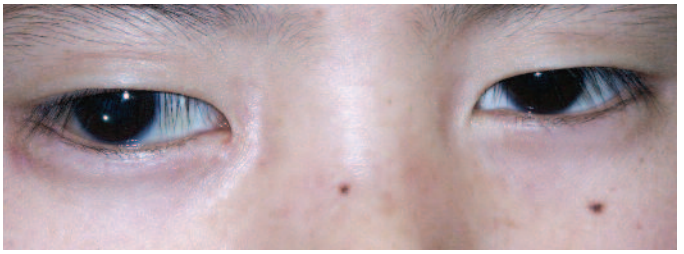

Figure 4. Right buphthalmos: glaucoma has enlarged the whole globe in all dimensions.

It is important to note that the cornea may not always have macroscopic opacity and particularly in older children the condition may present with the classical triad of:

- epiphora (excessive watering of the eyes)
- photophobia
- blepharospasm (excess squeezing of the eyelids).

## Causes of congenital glaucoma

Elevated IOP causes characteristic optic nerve damage with corresponding visual field loss. It may be associated with, or due to, congenital abnormalities of the eye or body. Primary congenital (or infantile) glaucoma is not associated with any other developmental abnormality. Secondary glaucoma occurs in a large number of ocular and systemic conditions, including:

- anterior segment dysgenesis
- phakomatoses – e.g. neurofibromatosis and Sturge–Weber syndrome
- congenital infections – e.g. rubella and herpes viruses
- Lowe's oculocerebrorenal syndrome.

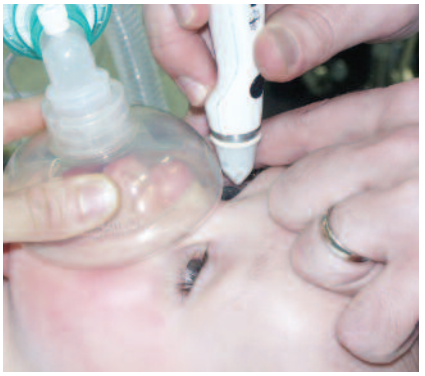

Figure 5. Accurate intraocular pressure measurement is often best obtained under general anaesthesia.

### Primary glaucoma

Primary congenital glaucoma is a rare condition with an incidence of around one in 10,000 births.[2] It usually occurs sporadically with a recessive inheritance pattern.[2] Mostly diagnosed in the first year of life, the condition usually appears bilaterally but can be asymmetric. It is more common in males than females and some genetic loci have been identified.

### Congenital infections

Congenital rubella was a well recognised cause of congenital cataract, corneal opacity, glaucoma and deafness, but is now very uncommon due to the widespread availability of rubella vaccination. Other congenital infections are only rarely associated with glaucoma, except as a complication of cataract or uveitis.

### Anterior segment dysgenesis

Anterior segment dysgenesis (ASD) refers to a broad group of congenital developmental anomalies that involve the front half of the eye including the cornea, iris, angle and lens. The molecular genetics for the most common ASD syndromes associated with glaucoma, such as Axenfeld–Rieger anomaly (ASD with iris strands crossing the angle), Peter's anomaly (ASD with corneal opacity) and aniridia (hypoplastic or absent iris), are reasonably well identified. Clinical geneticists are able to provide genetic counselling, carrier detection and prenatal diagnosis for affected families.

Figure 6. Gonioscopy using goniolens and portable slit lamp. Glaucoma surgery (goniotomy) can be performed using a similar lens.

### Phakomatoses

The phakomatoses are a group of systemic disorders that affect nerves, brain, the eye and skin. Neurofibromatosis (type 1) is an autosomal dominant disorder that causes skin neurofibromas and café-au-lait spots with tumours throughout the nervous system. Glaucoma is prominent when a neurofibroma affects the upper eyelid. Sturge–Weber syndrome is a neurocutaneous disorder characterised by meningeal angiomas, 'port-wine stain' haemangioma of the face, and glaucoma.

### Lowe's oculocerebrorenal syndrome

Lowe's oculocerebrorenal syndrome is an X-linked disorder causing developmental delay, dysmorphic facial features with frontal bossing, aminoaciduria, cataracts and glaucoma.

## Paediatric presentations

Children with congenital glaucoma classically present with a variety of signs and symptoms: an opaque cornea or prominent enlargement of the globe is usually readily apparent. A red eye is less common but symptoms of excessive tearing, photophobia and blepharospasm should raise suspicion about glaucoma in an infant.

Because the connective tissue of the cornea and sclera are much softer and more elastic in children than adults, children with glaucoma can present quite differently from adults with this condition. Elevated IOP often causes enlargement of the globe because of the sustained elevated pressure in the first few years of life. This is apparent at the corneal–scleral

junction because it causes measurable enlargement of the corneal diameter and axial (anterior–posterior) elongation of the whole globe, resulting in secondary myopia. The cornea may opacify from oedema or scarring, with focal breaks in one of the innermost corneal layers known as Descemet's membrane.

Like in adults, the primary concern in children is the characteristic optic nerve cupping with secondary visual field losses arising from the loss of nerve fibre at the optic nerve head. Unlike adults, cupping can occur rapidly in children and reverse with normalisation of IOP, due to the elasticity of the sclera. The treatment goal in such cases is the avoidance of significant and permanent optic nerve damage and the minimisation of pathological globe enlargement buphthalmos (Figure 4). Additionally, unlike adults, it is uncommon for the presenting sign to be optic nerve cupping in children.

### Optic nerve examination under general anaesthesia

Given children with congenital glaucoma have corneal irritation and opacity with photophobia, adequate examination of the optic nerve head often requires general anaesthesia (Figures 5 and 6). General anaesthesia allows for a full assessment of the eye, including:

- measurement of the child's IOP and corneal diameter
- gonioscopy
- refraction test
- optic disc photography.

These ophthalmological measures are also used to monitor the child's response to treatment and progression.

### Treatment

#### Medical therapy

Medical therapy is generally only used in the short term as the response is often poor and short-lived. A significant majority of the topical therapies used in adults either do not work at all in infants or the side effects preclude their use. For example, the systemic cardiorespiratory side effects that can occur with topical beta–blocker use occur more commonly in children and topical brimonidine (Alphagan Eye Drops) is dangerously sedating in infants.

Figure 7. Fluorescein 2% in drops instilled in the conjunctival fornix of each eye are still present five minutes later in the left tear lake, confirming nasolacrimal duct obstruction.

#### Surgery

When glaucoma is suspected clinically an examination under anaesthesia is usually booked with a view to proceeding to surgery if the diagnosis is confirmed. For patients with congenital glaucoma, the operations of choice are goniotomy or trabeculotomy (if the cornea is too opaque). Both surgical procedures are techniques for opening the drainage angle of the eye to decrease the IOP.

Given the rarity of congenital glaucoma, these procedures are typically only performed at tertiary referral children's hospitals by surgeons familiar and comfortable with these subspecialist techniques. These procedures have a success rate of around 80 to 90% in patient's with primary congenital glaucoma.[3]

## Conclusion

GPs are most likely to be involved in the co-management of children with a known ocular or systemic condition that predisposes them to glaucoma. The occasional infant will present, however, with clinical signs secondary to raised IOP. Occasionally congenital NLDO can present a diagnostic challenge, so any cause of asymmetric globe enlargement or corneal opacity warrants a complete ophthalmological assessment. The difficulty for the primary care physician arises in the differential diagnosis of the watering and sometimes red eye. It should be remembered that conjunctivitis is acute with prominent ocular injection and discharge, features that are not commonly apparent with congenital glaucoma.

Congenital glaucoma may present with or exacerbate a watering eye. The overwhelming majority of infantile epiphora is due to congenital NLDO and, fortunately, simple clinical tests can be performed to support the diagnosis (Figure 7). Although conservative therapy resolves greater than 90% of congenital NLDO, the spectre of congenital glaucoma is one of the primary reasons an infant with nonresolving epiphora warrants specialist referral. MT

### Congential glaucoma: key points

- Congenital glaucoma is a rare sight-threatening condition.
- Most epiphora in infants is due to congenital nasolacrimal duct obstruction.
- Congenital glaucoma may present with epiphora, photophobia and blepharospasm.
- An infant with epiphora that does not resolve should be referred to exclude glaucoma.
- Any infant with a cloudy cornea or asymmetric globe enlargement requires urgent ophthalmic assessment.
- Surgical treatment of congenital glaucoma is usually required to prevent irreversible globe enlargement and optic nerve damage.

## References


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DECLARATION OF INTERESTS: None.