Paediatric photophobia

The pressure of a timely diagnosis

Gizem Ashraf, Rahul Chakrabarti

CASE

A girl aged eight months was brought to see her family's general practitioner (GP) with a four-month history of persistent bilateral epiphora (overflow of tears onto the face) and light sensitivity. There were no other symptoms such as decreased appetite or vomiting. Medical history included an uncomplicated vaginal delivery at 41 weeks' gestation, with no significant history other than mild pre-eclampsia. Clinically, the infant was afebrile and hemodynamically stable. On examination, the infant appeared to have poor ability to fixate and follow a near target. The eyes appeared prominent, and the corneas appeared mildly cloudy. However, the eyes were not obviously red, and there were no periorbital rashes or inflammation of the periocular skin. There was no obvious head and neck lymphadenopathy. There were no rashes or focal neurological signs. Systemic examination was otherwise unremarkable. The GP referred the infant immediately to an ophthalmologist for review.

QUESTION 1

What features suggest light sensitivity in an infant?

QUESTION 2

What differential diagnoses should be considered in paediatric patients presenting with photophobia?

QUESTION 3

What differential diagnoses should be considered in infants presenting with a cloudy cornea?

QUESTION 4

What are the distinguishing factors for differential diagnosis for infants presenting with photophobia and epiphora?

QUESTION 5

How do you assess visual acuity in a child?

ANSWER 1

Infants with photosensitivity are likely to have an aversion to standard environment lighting, with features such as involuntarily closing eyes and rubbing the affected eyes.

ANSWER 2

In paediatric patients presenting with photophobia, differential diagnoses that should be considered include:

- ocular
  - congenital glaucoma
  - subtarsal foreign body
  - corneal foreign body/corneal abrasion
  - inflammation in various anatomical locations, including uveitis, conjunctivitis, keratoconjunctivitis, iritis, scleritis, blepharitis and optic neuritis
  - retinal conditions such as Leber's congenital amaurosis, aniridia, albinism and achromatopsia
- non-ocular
  - meningitis
  - encephalitis
  - migraine
  - subarachnoid haemorrhage
  - epilepsy
  - pituitary tumours
  - developmental and sensory disorders.

ANSWER 3

In infants presenting with cloudy cornea, differential diagnoses that should be considered include:

- sclerocornea
- tear in Descemet's membrane in the cornea
- metabolic (eg mucopolysaccharidoses, mucolipidosis)
- endothelial dystrophy of the cornea
- dermoid
- anterior segment dysgenesis
- ulcers (eg herpes simplex virus, rubella).

ANSWER 4

The distinguishing factors for the differential diagnosis for infants presenting with photophobia and epiphora are outlined in Table 1.

ANSWER 5

Visual acuity assessment of a child needs to be age appropriate.[1] There are different tools that can be used on the basis of the child's age.

0–3 months

- Blinks to light

3–6 months

- Fixation and following
- Teller acuity testing
- Forced preferential looking

6 months – 2 years
- Allen pictures
- Preferential looking tests
- Hundreds and thousands
- Teller acuity cards
- Cardiff acuity cards

2–5 years
- Kay pictures
- Sheridan Gardner test

School-aged children
- Snellen chart
- Logarithm of the minimum angle of resolution (LogMAR) chart

CASE CONTINUED

At the patient's ophthalmology assessment, general examination was unremarkable, and vital signs were normal. On examination of the eyes, the corneas were mildly cloudy, and fundi were difficult to visualise. Intraocular pressures of both eyes were very high at 37 mmHg and 45 mmHg (reference range <12 mmHg for children aged 6–9 months). The patient was diagnosed with bilateral primary infantile glaucoma, commonly referred to as congenital glaucoma. She was started on dorzolamide eyedrops to reduce intraocular pressures and immediately referred to hospital for urgent surgery.

QUESTION 6

What examination findings indicate primary infantile glaucoma?

QUESTION 7

How can congenital glaucoma be distinguished from nasolacrimal duct obstruction?

ANSWER 6

Primary infantile glaucoma is a rare disease occurring in one in 10,000 live births, with evidence suggesting 10–27% of cases are inherited.[2]

In more than two-thirds of cases, the disease affects both eyes but may do so asymmetrically.[3] Onset may occur at birth or within the first few years of life. The classic triad of symptoms consists of epiphora, photophobia and cloudy cornea.[4] Examination findings may include:
- corneal enlargement and Haab striae – in young children, increased intraocular pressure causes stretching of the cornea, leading to increased corneal diameter (megalocornea). A corneal diameter of >12 mm in children aged <1 year requires urgent ophthalmology referral.[5] Additionally, corneal stretching causes permanent scars called Haab striae.
- astigmatism – irregular enlargement may lead to astigmatism[6]
- corneal oedema – appears as corneal clouding[2]
- optic nerve cupping – elevated intraocular pressure damages the optic nerve, leading to cupping[2]
- ocular enlargement (buphthalmos) – in infants, increased intraocular pressure causes stretching of the sclera, leading to increase in the size of the globe[2]
- myopia – may be an early sign due to an enlargement of the globe
- amblyopia – may develop secondary to refractive error.

ANSWER 7

Occurring more commonly than primary infantile glaucoma, nasolacrimal duct obstruction affects 6% of newborns and is therefore a potential cause for delay in the diagnosis of primary infantile glaucoma.[7] Distinction between the two diseases can often be made clinically. Although both cause epiphora, children with nasolacrimal duct obstruction do not have blepharospasm or photophobia.

Key points

- In infants presenting with epiphora and photophobia, congenital glaucoma diagnosis should be considered.
- Visual acuity assessment methods of paediatric patients should be age appropriate.
- For patients presenting with red flag ocular symptoms, urgent ophthalmology review should be arranged.

Authors

Gizem Ashraf BMedSc, MD, Doctor, Monash Health, Vic

Rahul Chakrabarti MBBS (Hons), BMedSc (Hons), DPhil, MSurgEd, FRANZCO, Director of Hospital Medical Officer Training, The Royal Victorian Eye and Ear Hospital, Vic; Lecturer, Department of Ophthalmology, University of Melbourne, Vic

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Table 1. Differential diagnosis for infants presenting with photophobia and epiphora

Differential diagnosis	Distinguishing factors
Corneal abrasion	Examined under anaesthesia with fluorescein light. The dye will get trapped in any areas of corneal defect.
Subtarsal or corneal foreign body	Identification of foreign body under slit lamp examination with inversion of the eyelid.
Conjunctivitis	Diagnosis is usually history based but may require swab and microbiology. It is important to differentiate between unilateral and bilateral conjunctivitis as this may indicate different aetiologies. Pre-auricular lymph nodes may be palpable. Viral conjunctivitis is unlikely to last for four months.
Uveitis	Systemic features such as rashes or swollen and tender joints.
Herpes simplex keratoconjunctivitis	Observation of a corneal dendritic ulcer with fluorescein staining.
Primary infantile glaucoma	Triad of epiphora, photophobia and cloudy cornea. Examination findings may also include corneal enlargement, corneal oedema, optic nerve cupping, ocular enlargement, astigmatism, myopia and amblyopia.
Posterior fossa tumour	Triad of photophobia, epiphora and torticollis. Diagnosis confirmed with magnetic resonance imaging.

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Correspondence to:
rahul.chakrabarti@eyeandear.org.au

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