

PRACTICE

EASILY MISSED?

Congenital cataract

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This is one of a series of occasional articles highlighting conditions that may be more common than many doctors realise or may be missed at first presentation. The series advisers are Anthony Harnden, university lecturer in general practice, Department of Primary Health Care, University of Oxford, and Richard Lehman, general practitioner, Banbury. To suggest a topic for this series, please email us at easilymissed@bmj.com.

Congenital cataract is an important preventable cause of visual impairment and blindness in childhood. Advances in surgical management and visual rehabilitation mean that early diagnosis is vital to optimise visual outcome and prevent irreversible visual impairment.

Why is congenital cataract missed?

In ideal conditions, examination for the red reflex by an experienced practitioner readily identifies congenital cataract; however, its effectiveness as a screening tool has yet to be formally evaluated. A national UK study assessing all diagnoses of congenital cataract during one year found that less than half were detected at either the newborn or 6-8 week examinations (35% at the newborn examination and a further 12% at the 6-8 week examination).² A more recent regional study from the Republic of Ireland found that over a 10 year period, none of the 27 cases of congenital cataract was detected at the neonatal check and only 24% were detected by the general practitioner on subsequent examination.³ Although recommendations for red reflex detection as part of the newborn and 6-8 week examinations were in place at the time of these studies, no data were available on the percentage of infants who had such testing, so whether delays in diagnosis were caused by problems performing the test or by failure to test (assuming that most cataracts were present from birth) is unclear.^{4,5}

Red reflex examination can be difficult to perform. Eyelid swelling at birth can make eye opening difficult, especially if the infant is distressed. Examination conditions can be suboptimal, with brightly lit rooms, background noise, and interruptions. Examination of infants aged 6-8 weeks is usually

easier when eyelid swelling has resolved and they are more visually alert and maintain gaze.

Why does this matter?

Visually significant congenital cataracts lead to irreversible changes in the developing visual system owing to form-deprivation amblyopia, and they can also cause nystagmus. These disorders result in severe and lifelong visual impairment. Some evidence suggests that these changes start to develop after only 6 weeks of life for unilateral cataract and 10 weeks of life for bilateral cataracts.^{6,7} Other evidence suggests that irreversible changes occur earlier, with long term visual outcomes showing an average loss of one Snellen visual acuity line for every three weeks of surgical delay during the first 14 weeks of life.⁸ Cataract surgery is essential before these irreversible changes take place. Early detection is therefore vital.

How is congenital cataract diagnosed?

Screening for congenital cataract, other ocular media opacities, and ocular malformations requires the red reflex to be sought. Although recommendations for performance of this test shortly after birth and again at 6-8 weeks have been in place for many years,^{4,5} red reflex examination has only recently achieved "screening programme" status as part of the National Screening Committee's NHS Newborn and Infant Physical Examination Programme (<http://newbornphysical.screening.nhs.uk/>).

During the test the room must be calm, quiet, and very dark. The infant is positioned comfortably on the mother's lap, with the head against her stomach. Calmly singing to the infant holds attention, often with spontaneous eye opening, and a bottle feed or soother can have the same effect. Alternatively, the infant can be positioned either over the mother's shoulder, or held head up at a 45° angle from the horizontal with one hand supporting the chest and the other hand used to jiggle the child's bottom (fig 1).⁹ Both positions result in spontaneous eyelid opening. The largest white-light circle on the direct

Case scenario

A mother brings her 8 week old baby to her general practitioner for her 6-8 week child health surveillance check. At the baby's initial neonatal hospital check the doctor had difficulty performing the red reflex examination owing to neonatal eyelid swelling, but took no further action. The general practitioner cannot detect the red reflex in the right eye so makes a direct referral to the ophthalmologist that day by telephone. The baby is seen the following day and a cataract in the right eye is diagnosed. Cataract surgery is performed four days later.

How common is congenital cataract?

- In the United Kingdom the incidence of detected cataract of congenital origin affecting vision has been estimated to be 2.49 per 10 000 population by age 1 year
- Owing to some delayed diagnoses, the incidence increases to 3.46 per 10 000 population by age 15 years. This equates with 200-300 children being born with congenital cataract each year in the UK¹

ophthalmoscope is used unless the pupils are very small, when the narrower light source is used. The lens is set at 0 or to the examiner's prescription if his or her glasses are removed. The examiner sits or stands at arm's length from the child and looks at the child's face through the aperture. If the infant is asleep, the eyelids can be gently opened with clean fingers. If the eyes are turned up (owing to Bell's phenomenon), the head can be turned gently from side to side to evoke the doll's eye phenomenon, which usually centres the eyes long enough to gain a view of the red reflex. Illumination of both pupils simultaneously is preferable to allow comparison. If a clear red reflex is not seen in one or both eyes, they are examined individually and compared.

Same day telephone referral to a paediatric ophthalmologist is warranted if the examination shows:

- The presence of opacities in the reflex (fig 2)
- The absence of any reflex
- A white pupillary reflex (leukocoria)

Urgent written referral to the ophthalmologist is recommended if the examination shows:

- Inequality in colour, intensity, or clarity of the reflection
- No detectable abnormality but a parent or observer describes a history suspicious of leukocoria on observation or in a photograph (recognising, however, that the commonest cause of a white pupil in flash photography is reflection from the optic disc of the in-turning eye when fixation is off-axis to the camera).¹⁰

In addition to the detection of media opacities, giving attention to eyes and vision at the time of the 6-8 week neonatal check has the potential to identify other conditions affecting sight, such as delayed visual maturation and nystagmus.

Screeners should be aware that the normal red reflex seen in dark skinned infants tends to be more yellow than red. This should not be confused with the white reflex of leukocoria, which may indicate underlying retinoblastoma.

Low specificity is expected to be the consequence of increasing the sensitivity of the red reflex test, leading to more false positive referrals. However, this is arguably acceptable in view of the serious and irreversible consequences of missed diagnoses.¹¹

How is congenital cataract managed?

Visually significant congenital cataracts are managed by prompt cataract surgery, and the resulting aphakia is corrected with prolonged wear contact lenses, primary intraocular lens implantation, or aphakic spectacles. Long term follow-up with the ophthalmologist is needed.

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Key points

- Congenital cataract is uncommon but is an important preventable cause of visual impairment and blindness
- For optimal visual outcome, surgical correction is needed within the first three months of birth as visual impairment may develop after only 6 weeks of life for unilateral cataract and after 10 weeks for bilateral cataracts
- In many countries, including the UK, ocular examination is recommended shortly after birth and again at 6-8 weeks
- Red reflex examination is used to detect opacities such as cataract, retinoblastoma, and malformations
- Red reflex examination needs optimal conditions, experience, and patience
- Detection of any abnormality warrants urgent ophthalmological referral

Figures

Fig 1 Positioning of an infant to induce spontaneous eye opening. Left: The child is held leaning forward at 45° to the horizontal, with one hand supporting the chest and the other supporting and jiggling the infant's bottom. Right: The child is positioned over the mother's shoulder

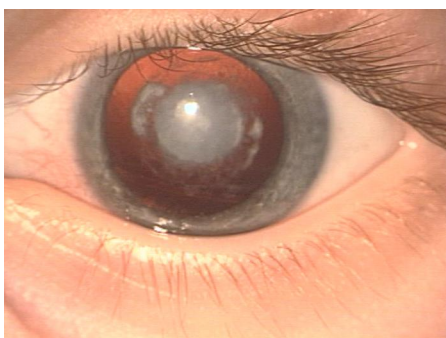


Fig 2 Red reflex showing a nuclear cataract