EASILY MISSED?

Congenital cataract

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- Giant cell arteritis (*BMJ* 2011;342:d3019)
- Metastatic spinal cord compression (*BMJ* 2011;342:d2402)
- Cholesteatoma (*BMJ* 2011;342:d1088)

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.

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 UK[1]

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 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.

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

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 inturning eye when fixation is off-axis to the camera).[10]

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

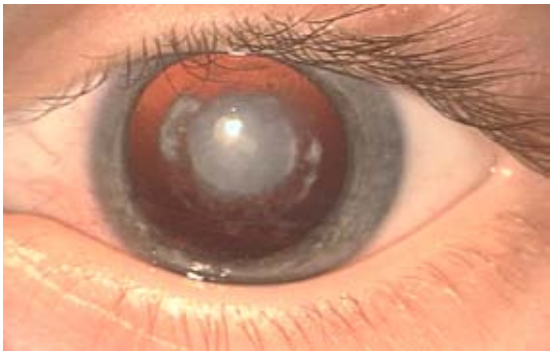

Fig 2 | Red reflex showing a nuclear cataract

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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Patient consent obtained for the photos but not required for the case scenario (patient anonymised, dead, or hypothetical).

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A PATIENT'S JOURNEY

Living with obstetric fistula

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This is one of a series of occasional articles by patients about their experiences that offer lessons to doctors. The *BMJ* welcomes contributions to the series. Please contact Peter Lapsley (plapsley@bmj.com) for guidance.

During the birth of her first child in 2000, Fatima Aliyu developed two fistulas: vesicovaginal fistula and a rectovaginal fistula. She is not yet fully cured and anticipates further surgery

When we arrived at the clinic I was just at the beginning of labour. I stayed there for a day and the pain kept increasing. One of the nurses asked me to push so as to increase the labour. I continued pushing with no sign of delivery. The next day a doctor came and told me that the labour was not progressing. He said I should be induced so that I could deliver.

There was no sign of delivery except the kicking of the baby in my womb. During my third day at the hospital the nurse on duty heard me screaming because of the terrible pain. She said "You are the only one giving us a problem and it is because you are so lazy. It is as if you are not a woman. Can't you see all the other mothers have delivered and gone home with their babies?" She told me to stand up and start going up and down. The nurse said that moving this way might help me to deliver. I started doing as she said and then something burst out of me. Black water started trickling down my legs. She said "Maybe now you are going to deliver." For over 30 minutes I was pushing on the bed but nothing progressed. On the fourth day when I felt like urinating nothing came out, only blood. By now I couldn't walk, my legs were very heavy as if they were frozen.

On the sixth day I started losing consciousness and my mother was terrified that I might not survive. My parents asked the management staff to refer me to a better hospital where I could have a caesarean section. By then we knew the baby was already dead because the heartbeat could not be felt. I went by bus to the next hospital. We were lucky to find a doctor there who was very sympathetic. She asked my relatives how they could leave me in this condition.

I was not aware that I had developed a fistula until after the caesarean. I was brought to the postoperative care unit with a catheter. After seven days, the catheter was removed and I found myself lying in a stream of water. I was shocked when I realised that I could not control my bladder as I had before. My mother saw me and asked why I had poured water on my bed. I told her it was not water and that it felt like urine was coming out uncontrolled. One of the nurses said it was vesicovaginal fistula. My mother screamed and I started crying. I knew it was something terrible because one of our neighbours has vesicovaginal fistula. My neighbour's husband rejected her and her relations left her; her baby was stillborn, and she never had another child because no other men would marry her. She became destitute and had to beg on the street, right into old age.

The diagnosis affected me seriously and I started thinking about the future. My hope was that I should recover and catch up with my colleagues who had started registering for their second year at university. I wondered how I could go back to university in this condition? How could I mix with my classmates with urine trickling down legs that don't work? I cried in silence in the dark, because I was left behind, my dignity had gone, and I didn't know when my problem could be solved. The healthcare staff referred me to the fistula unit in the Murtala Specialist Hospital at Kano.

At the fistula unit

The doctor said I had two different and difficult fistula complications, a vesicovaginal fistula and a rectovaginal fistula. I also had paralysed nerves in my legs (drop foot), which meant a lot of damage had been done during labour.

G M B AKASH/PANOS

Front cover image: patients at the fistula treatment centre in the Dhaka Medical College Hospital, Bangladesh

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Previous articles in this series

- Bilateral developmental dysplasia of the hips (*BMJ* 2011;342:d2152)
- Motor neurone disease (*BMJ* 2011;342:d1661)
- Chronic depression (*BMJ* 2011;342:d93)
- Adult atopic eczema (*BMJ* 2011;342:d644)

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- Sally Carter: Films, fistula, and an illiterate surgeon

A DOCTOR'S PERSPECTIVE

Annually many millions of women die or are injured and disabled in the process of giving birth. The most prominent among these injuries is obstetric fistula, which until the beginning of this century was common in many women around the world. The condition is caused by lack of prompt medical attention, which leads to prolonged, obstructed labour. The presenting fetal part, unable to pass through the birth canal presses against the soft tissues of the mother's pelvis for days. This impact can cut off the blood supply to large parts of the vagina, bladder, and rectum, resulting in the death of these tissues. If the patient survives labour, during the days and weeks after the birth the dead tissue falls away, leaving holes (fistulas), which allow a constant, uncontrollable loss of urine (vesicovaginal fistula) or stool (rectovaginal fistula) into the vagina. Unless they can get treatment, women with fistula have to live with this incontinence and are likely to face debilitating physical, psychological, social, and economic consequences because of the strong odour that accompanies them. The constant leaking can cause skin ulcerations in the genital area and the smell may drive away her family and community. The trauma is often compounded by the psychological trauma of delivering a stillborn baby.

Today most obstetric fistulas occur in developing countries of sub-Saharan Africa, south Asia, and the Middle East; more than two million women already have the problem and there are 50 000–100 000 new cases every year. Fistulas can be repaired surgically with a success rate of over 90% in experienced hands. However repairing fistula injuries is not the end of the challenge, because women will return to the same conditions that caused the injury in the first place. Subsequent pregnancies, if they are possible, can undo the success of treatment if women do not receive the care that they need. The capacity for surgical intervention for fistulas is limited; at best, 10 000 fistulas can be repaired a year. And too few surgeons have the expert knowledge and skill needed to cope with the large range of traumas present in obstetric fistula. In reality it is not simply a discrete entity of holes that need to be closed, but a widespread, complex trauma to multiple structures in the pelvis, such as the muscles, fascia, and nerves, which can result in a minute fistula with minimal tissue loss. In addition to such major tissue loss, damage can occur to other body systems, such as neurological damage and paralysis in the legs (drop foot) and a complete cloaca—where the rectum, vagina, bladder, and urethra conflux into a single common channel. Closing such a complicated fistula, which is estimated to occur in about 1% of patients, is a challenge and many women are rendered incurable, or need several attempts to close the fistula.

Because of these factors it is crucial that prevention efforts and new treatment strategies are strengthened and emphasised in tandem with surgical repair. One new way is supported by the National Nigerian fistula programme, which estimates that systematic early catheterisation of small fistulas within 75 days of leaking could cure over 25% of all new cases each year without the need for fistula surgery.

It is also important to not just simply focus on the development issues inextricably linked with fistula such as poverty, lack of education for girls, and early marriage and pregnancy. If marriage, childbirth or both were delayed by legislation or by promoting access to family planning services, this would only help women delay developing a fistula until later on if they could not obtain timely, high quality obstetric help. Nor is it any use to identify mothers who are at risk during the antenatal stage or to empower and educate women about their right to health care if, while in hospital, they are neglected as much as or more than they would be at home. As Fatima's case shows, timely skilled care during pregnancy and childbirth when women need it is crucial. Failure to address this aggressively will only ensure that the problem of maternal morbidity and mortality endures.

Gloria Esegbona

The surgery needed to be done by an experienced European surgeon who was coming in two weeks' time. The healthcare staff said that the specialist was always willing to do the work, and wanted to help solve our problems. I was happy to hear that the doctor was so dedicated and sympathetic.

After that the days were long and always the same. My mattress was always wet, with urine all over. In cold weather urine would penetrate down the mat and flow under my bed. My mother (I call her Hajiya) would make sure my bed was always dry by using large towels and a macintosh as if making a baby's bed. That would help me to sleep well. Without this, urine would disturb me at night and cause rashes and other infections on my skin.

I looked very thin and anaemic. I always passed everything I ate so I limited the amount of food I took.

Hajiya was always telling me to go out and stretch my legs. But I told her I didn't want to, because the pain in my legs became intense and I moved slowly because of my drop foot. In the middle of the night I could not sleep. My eyes were open, turning from one side to the other until morning. If Hajiya forced me to go out I would start crying.

Whenever I did go out I saw girls and women everywhere with wet, urine-soaked clothing who were overcome with sorrow and poverty, this touched my heart and made it beat fast. And every day new women with fistulas came to the medical centre, most of them from poor villages with little education. Some of them had had fistula for ten to 12 years and their ambition in life, their focus, was always to be cured and to be normal. Some had been cured but came again with one, two, three, or more fistulas after bearing more children because there was no one to deliver their babies properly. These women slept and lived under a tree in the fistula compound. They hung their property on the tree trunk, and put their bags under the tree. And in order to eat, to get clothes and a nappy to trap the urine, to survive and to maintain their pride, they went out begging, day and night. It was terrible, and I wondered why women were being injured like this every day.

When the surgeon arrived, he investigated me and told me there was a lot of damage to my body and that, before the repair could be made, I needed to eat more fish and chicken with green vegetables to make myself healthy.

I had several surgeries because the sphincters that controlled my urine and bowels were lost due to trauma so I was incontinent. So far I have had surgery four times and I am still not healed because of the extreme nature of my fistulas. I am still waiting for the next stage, which I hope

FURTHER READING

Useful resources for patients and health professionals

Campaign to end fistula (www.endfistula.org)—This campaign is an international effort that currently covers over 40 countries in sub-Saharan Africa, South Asia, and some Arab States. Launched by the United Nations Population Fund (UNFPA) in 2003, the campaign includes interventions to prevent fistula from occurring, to treat women who are affected, and to help women who have undergone treatment return to full and productive lives, with the ultimate goal of making fistula as rare in the developing world as it is in industrialised countries.

International society of obstetric fistula surgeons (www.isofs.org)—ISOFS is the leading international professional body for those engaged at the forefront of the treatment and rehabilitation of women with obstetric fistula. Its aim is to scale up the number of skilled professionals who can offer a quality repair.

The National Nigerian Fistula Project (http://www.isofs.org/wp-co ntent/uploads/2011/04/report_2010_000_full_report_LR.pdf)—Over the past 25 years this project has become the largest fistula repair and training centre in the world. Founded and led by Dr Kees Waaldjik, an experienced fistula surgeon. The project includes eight fistula repair centres in the north of Nigeria and one in the south, with additional support provided to three centres in Niger.

will be the last, but I am not sure when to go for that surgery, although my surgeon is always willing to assist. I feel good whenever I think of my surgeon. He always cares for my condition; he has always given me treatment whenever the need has arisen. But I am traumatised by having had a fistula at the age of 25, and by losing my status as a mother, wife, and university graduate for ten years. And it takes courage to face more painful surgery with no guarantee. Now I am searching for whatever might lead me to a better life and a better future. Because fistula is stressful you have to be strong or you could end up losing your whole life.

My family never rejected me. They continue to support me and encourage me to be strong. At first my husband loved and cared for me, but after I developed this problem he stopped everything he had been doing for me, seeing me as being of no use. I have been staying with my parents for the past 10 years. I am now divorced.

I have not given birth again and, in a society in which children are very important to every woman of child-bearing age that is very damaging to my pride. This is the most distressing aspect of my condition. I am always hoping for the day when I will heal and have a way of giving birth to a child again.

What has really made a difference is my level of education. If I was uneducated, like most women with fistula, the stigmatisation would be worse. Before my fistula I didn't even know what a non-governmental organisation was, but I have now learnt a lot about how to be an advocate for people with reproductive health problems and to mobilise them. I have learnt that fistula is a condition that is almost entirely preventable with timely medical intervention. It can be caused by bad service delivery in the medical world and thus negligence. I know that I am in the same position as any other woman with fistula, no matter what my educational background. I therefore have to join hands with the poorest women in the world, to prevent fistula and to help my sisters with fistula to fight for their dignity and rehabilitation. And we must do that, or the trauma of vesicovaginal fistula will continue to affect the lives of thousands of good mothers of childbearing age.

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ANSWERS TO ENDGAMES, p 1369.
For long answers go to the Education channel on bmj.com

ON EXAMINATION QUIZ

Heparin

Answer A is correct.

STATISTICAL QUESTION

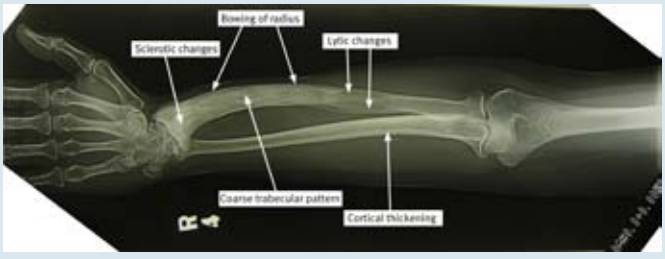
Patient preference trials

Answers *b*, *c*, and *d* are true, whereas *a* is false.

ANATOMY QUIZ

Computed tomography section through the aortic sinus of the heart

A Azygos vein
B Non-coronary sinus
C Pericardium
D Left internal thoracic artery
E Left atrial appendage



PICTURE QUIZ

A woman with raised alkaline phosphatase and forearm deformity

1 The diagnosis is Paget's disease of bone.
2 This radiograph shows cortical thickening of the radius and ulnar with a coarse trabecular pattern and bowing of the radius. Sclerosis of the distal radius is also seen, as well as lytic lesions in the shaft of the radius (figure).
3 Bisphosphonates are the gold standard treatment. The aim is to decrease the abnormal bone turnover caused by osteoclastic bone resorption.