National cross sectional study of detection of congenital and infantile cataract in the United Kingdom: role of childhood screening and surveillance

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Abstract

Objectives To determine the mode of detection and timing of ophthalmic assessment of a nationally representative group of children with congenital and infantile cataract.
Design Cross sectional study.
Setting United Kingdom.
Subjects All children born in the United Kingdom and aged 15 years or under in whom congenital or infantile cataract was newly diagnosed between October 1995 and September 1996.
Main outcome measures Proportion of cases detected through routine ocular examination and proportion assessed by an ophthalmologist by 3 months and 1 year of age.
Results Data were complete for 235 (95%) of 248 children identified. Of these, 83 (35%) were detected at the routine newborn examination and 30 (12%) at the 6-8 week examination; 82 children presented symptomatically. 137 (57%) children had been assessed by an ophthalmologist by the age of 3 months but 78 (33%) were not examined until after 1 year of age. In 91 cases the child's carers suspected an eye defect before cataract was diagnosed.
Conclusions A substantial proportion of children with congenital and infantile cataract are not diagnosed by 3 months of age, although routine ocular examination of all newborn and young infants is recommended nationally. Strategies to achieve earlier detection through screening and surveillance are required.

Introduction

Cataract in infancy is an important avoidable cause of visual handicap worldwide.[1] Visual loss is mainly due to amblyopia.[2 3] This arises principally through stimulus deprivation as the cataract prevents normal retinal images forming and being transmitted to the visual cortex.[4] Experimental and clinical research suggests that surgical treatment of dense congenital cataract needs to take place within the first three months of life.[3 5–8] The management and outcome of congenital and infantile cataract have improved considerably in the past few decades with increased recognition of the importance of early detection and treatment.[2 6 9 10]

In industrialised countries the most common visually disabling disorders of children are present, or become manifest, in early childhood.[1] Routine ocular examination of young infants is widely recommended[11–14] to ensure that treatment, genetic counselling, and other advice and support are offered at the earliest opportunity. In the United Kingdom current guidance is based on the reports of two national joint working parties of the Royal College of Paediatrics and Child Health and the Royal College of Ophthalmologists.[13 14] These recommend inspection of the eyes and evaluation of the pupillary red reflex of all infants during the newborn period and again at 6 to 8 weeks, when assessment of visual behaviour and examination for the presence of squint are also advised.[13 14]

Although an ocular examination has been part of the routine newborn examination since the 1960s,[15] its effectiveness in detecting ophthalmic disorders is not known. We report the mode of detection and timing of ophthalmic assessment in children with newly diagnosed congenital or infantile cataract, ascertained through a national cross sectional study.

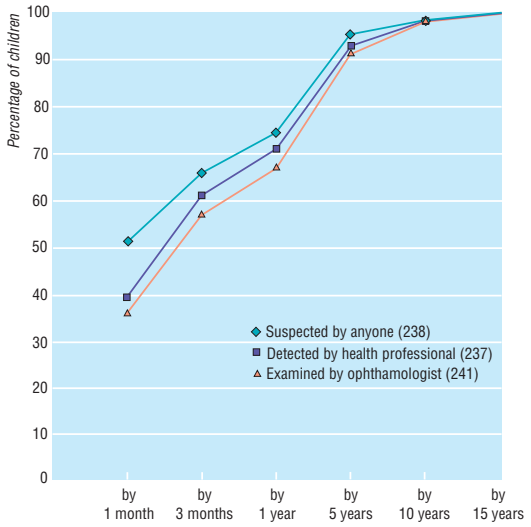
Subjects and methods

Although congenital and infantile cataract are separate categories in standard disease classification systems,[16] in clinical practice the terms are often used interchangeably.[9] We therefore used a clinical case definition encompassing both. All children aged 15 or under with newly diagnosed congenital cataract were eligible. This included all those with cataract present from infancy with a congenital cause and those first presenting outside infancy but with salient clinical features indicating early onset, such as cataract morphology, associated congenital ocular anomaly, or nystagmus.[6 9] All children requiring regular review to monitor visual development were included, irrespective of treatment given. We excluded children with minor lens opacities who did not require regular review or further assessment, children with possible or definite acquired cataract, and those born outside the United Kingdom.

All children with congenital or infantile cataract newly diagnosed during October 1995 to September 1996 were identified through active reporting to two independent national surveillance schemes. Cases were notified by ophthalmologists participating in a new scheme established for this study through the British Congenital Cataract Interest Group.[17] As paediatricians are responsible for routine ocular examination of young infants and for managing any associated systemic disease, cases were also notified by paediatricians reporting to the British Paediatric Surveillance Unit of the Royal College of Paediatrics and Child Health,[18] on which the ophthalmic scheme was modelled. Reporting cards were sent to paediatricians monthly, and to ophthalmologists every two months, for them to either notify new cases or confirm that no new cases had been seen. Notification of cases and communication with reporting clinicians in the two surveillance schemes were independent throughout.

Data collection and analysis

Reporting clinicians were asked by postal questionnaire to provide further details about each case. They were asked to report who first suspected the child had



Age at detection and ophthalmic assessment of all children with newly diagnosed congenital and infantile cataract, October 1995 to September 1996

an ocular defect and at what age; date and reason for contact with a health professional during which cataract was detected; and age at first referral to, and at first examination by, an ophthalmologist. Non-responding clinicians were sent up to three reminder requests at eight week intervals.

Unilateral and bilateral cases were analysed separately and together and within the following clinically relevant, but not mutually exclusive, age groups: neonates aged ≤1 month, infants aged ≤3 months, and infants aged ≤12 months. Observed differences between unilateral and bilateral cases were examined with the test of significance for the difference in two proportions.[19]

Results

During the 12 month study period 248 children (118 girls) with newly diagnosed congenital cataract were identified, of whom 236 (95%) were notified through the ophthalmic scheme and 90 (36%) through the paediatric scheme. Both eyes were affected in 161 (66%) cases. Complete data regarding detection and ophthalmic assessment were available in 235 cases. The denominators for each analysis are reported separately.

By 1 month of age an ocular defect had been suspected in 121 (51%) children, but only 87 (36%) had been assessed by an ophthalmologist, rising to 137 (57%) by 3 months. Median age (range) at first examination by an ophthalmologist was 10 weeks (birth to 15 years), but 78 (33%) children were not examined by an ophthalmologist until after 1 year (figure). In all, 178 (92%) cases were referred to an ophthalmologist within 1 month of detection by a non-ophthalmic health professional, and 195 (88%) were seen by an ophthalmologist within 1 month of referral. Among children aged ≤3 months at detection, nine had referral to an ophthalmologist delayed by more than 2 weeks. Similarly, assessment by an ophthalmologist was delayed by more than 2 weeks in 11 (8%) cases referred before age 3 months. Children with bilateral and unilateral cataracts did not differ significantly in age at detection or at ophthalmic assessment.

Of 69 cases detected after the age of 12 months, 25 (36%) were due to confirmed and nine to suspected prenatal factors. In the remaining 35 idiopathic cases, clinical findings consistent with infantile onset included morphology, associated congenital ocular anomalies, and nystagmus. Six of the 69 cases detected after 1 year had established amblyopia too severe for surgery, 38 had surgery, and five with bilateral asymmetric cataracts had occlusion therapy. The remaining 20 cases (29%) were initially managed conservatively as their level of visual function did not require immediate surgery.

Table 1 shows that 113 (47%) children were detected through child health surveillance or screening in early infancy: 83 at the neonatal examination and 30 at the 6-8 week examination. Twenty two children were detected during later routine examinations, including 15 during preschool or school entry vision screening. Similar proportions of children with unilateral and bilateral cataracts had them detected through these routine examinations (62% (52/83) unilateral, 53% (83/156) bilateral; 95% confidence interval for difference −4% to 22%, P=0.23) and had concerns before the examination about ophthalmic signs, symptoms, or known risk factors (17% (14) *v* 21% (33); −6% to 14%, P=0.56).

Eighty two (34%) children were detected as a result of established ophthalmic symptoms or signs such as reduced vision, strabismus, or nystagmus (table 1). This included one fifth (34) of all cases diagnosed in infancy.

Ten cases were detected through clinical examination of asymptomatic children with a family history of ocular or systemic disease and three through routine examination of preterm infants at risk of retinopathy of prematurity. Eight bilateral cases were detected while the child was being assessed because of a systemic disorder. In all cases with associated systemic diseases the non-ophthalmic features had been noted before detection of cataract.

Table 1 Context of detection of congenital or infantile cataract

	No of cases (n=239)
Child health screening or surveillance	135 (57)
Neonatal examination	83 (35)
6-8 week examination	30 (12)
8-9 month examination	3 (1)
13-18 month examination	4 (2)
Preschool vision screening	4 (2)
School entry vision screening	11 (5)
Presentation because of ophthalmic symptoms or signs	82 (34)
Targeted examination of high risk groups by ophthalmic professionals*	13 (5)
Sibling with ocular or systemic disease	5 (2)
Other ocular or systemic family history	5 (2)
Screening for retinopathy of prematurity	3 (1)
Detection during assessment or management of systemic disorder	8 (3)
Routine eye examination	1 (<1)

*Includes cases detected by a geneticist screening for an unrelated systemic disorder (1), a general practitioner (1), and a community paediatrician examining children with a family history of ocular disease (1).

Table 2 First health professional to detect congenital or infantile cataract

Health professional	No of cases (n=235)
Non-ophthalmic:	
Hospital paediatrician	96 (41)
General practitioner	63 (27)
Community paediatrician	13 (5)
Health visitor	10 (4)
School nurse or doctor	9 (4)
Other*	2 (1)
Ophthalmic:	
Optometrist	17 (7)
Ophthalmologist	14 (6)
Orthoptist	11 (5)

*1 midwife, 1 geneticist.

Most cases were detected by non-ophthalmic health professionals: 96 (41%) by a hospital paediatrician and 63 (27%) by a general practitioner (table 2). Two bilateral hereditary cases, suspected by an obstetrician during antenatal ultrasound examination, were subsequently confirmed by a paediatrician. Ophthalmic health professionals identified 42 (18%) cases.

In 91 (38%) cases parents or other family members were the first to suspect an ocular or vision defect. In 53 cases a problem was suspected within the first 3 months of life, but 13 (25%) of these were diagnosed after this age and 24 had established ocular symptoms or signs by the time of diagnosis. Four affected children were the first to note an ocular problem, and two cases were first suspected by a teacher.

Discussion

In this national cross sectional study, congenital and infantile cataracts were not detected by a health professional before the first birthday in 29% of cases, despite recommendations to examine all newborn and young infants routinely for cataract.[13 14] Capture-recapture analysis suggests that ascertainment in this study is 92% (95% confidence interval 86% to 98%).[17]

The clinical features of cases detected outside infancy were consistent with infantile onset.[6 9] Although the density of some forms of cataract may increase over time, making them clinically more important, others are relatively stable.[2 6 9] Thus, it is reasonable to suppose that an abnormality of the pupillary red reflex was present in infancy in those cases first diagnosed after this time. Most children in this study were born after the publication of current UK recommendations for routine ocular examinations of infants. We therefore believe our findings reflect current national practice.

Effectiveness of ocular examinations

The proportion of affected children having ophthalmic assessment by 3 months of age is a useful indicator of the performance of routine ocular examinations in infants[13 14] in the United Kingdom. By this age, 47% of children in our study had been detected through screening and 57% had been assessed by an ophthalmologist. Direct comparison with published case series[10 20] is difficult but suggests some improvement in the past decade. There is a lack of population based studies with which to assess secular trends in age and mode of detection. As neither process nor outcome of current practice is routinely monitored, little is known about the detection rate of children with congenital cataract. It has been assumed from previous studies that children with serious ophthalmic disorders, including cataract, are usually identified in early infancy and that later formal vision screening examinations contribute little to detection.[21 22] Our findings challenge these assumptions.

In a few cases delays occurred in referral to, and subsequent examination by, an ophthalmologist. This emphasises the importance of an effective chain of referral for children with suspected ophthalmic disease. However, as most cases were referred promptly the low proportion of children examined by an ophthalmologist by the age of 3 months is likely to be due to delays in detection. Although the proportion of babies having ocular examinations at birth and 6-8 weeks is thought to be high, we do not know whether all children in this study were examined. As congenital cataract is the disorder most often sought by paediatricians during these examinations,[23] the delays in ophthalmic assessment observed are likely to be due to problems with performing the test rather than failure to test.

Improving detection

Strategies are required to improve the effectiveness of existing routine examinations. Routine ocular examination of young infants requires specific knowledge and skills but with appropriate training can be performed by clinical staff with limited previous experience.[24] However, undergraduate and postgraduate training in ophthalmology varies, and its content and purpose have been questioned.[23 25] Existing recommendations about teaching paediatric ophthalmic disorders and visual assessment of children during the postgraduate training of paediatricians have not been widely implemented.[14]

A few cases in this study were identified through examination of asymptomatic children at high risk. Formal ophthalmic assessment is currently recommended for children at higher risk of ophthalmic disease,[13] including preterm infants and those with a family history or relevant systemic disorders. Effective liaison between paediatric, primary care, and ophthalmic health professionals and services is important to the success of this approach.

As lens opacity is difficult to identify by inspection alone, parents in this study who suspected a problem before diagnosis were probably alerted by their child's visual behaviour or features arising from reduced visual function such as strabismus or nystagmus. Parental concern did not always ensure prompt detection, suggesting parental uncertainty about the importance of the features noticed or a failure of health professionals to elicit or respond to these concerns.[21 22 26 27] Health professionals and parents should be made more aware of the importance of relevant clinical features and a family history of eye disease.[26 27]

Conclusions

We plan to follow the children identified in this study to assess their visual and educational outcomes. However, regular monitoring of the process and outcome of routine ocular examinations at national level is

Key messages

- Cataract in infancy is an important preventable cause of visual impairment and blindness in childhood
- Research suggests surgical treatment of dense cataracts is needed within the first 3 months of life
- In the United Kingdom routine ocular examination is recommended for all infants at birth and at 6-8 weeks
- In this national study, 47% of children newly diagnosed with congenital or infantile cataract were detected through these examinations
- 57% had been examined by an ophthalmologist by 3 months of age, but 33% were not assessed until after their first birthday
- Strategies are required to achieve earlier diagnosis and increase the proportion of cases detected through screening in the first 3 months of life

required to assess the performance and improve the quality of this component of health surveillance of young infants. The purpose and value of vision screening in later childhood are being reviewed in the United Kingdom,[28] and this review should include ophthalmic screening and surveillance in infants. Our findings suggest that measures are needed to improve the effectiveness of this practice.

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Endpiece

Mind over matter

Lady Talmash, that says she can do whatsover she will, cannot believe whatsover she pleases. 'Tis not unpleasant, methinks, to hear her talk how at such a time she was sick and the physicians told her she would have the smallpox, and showed her where they were coming out upon her; but she bethought herself that it was not all convenient for her to have them at that time; some business she had that required her going abroad; and so she resolved she would not be sick, nor was not.

The Letters of Dorothy Osborne, edited E A Parry, Everyman's Library, 1924

Submitted by A L Wyman, retired physician, London