Early detection of visual defects in infancy

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Abstract

To determine the part played by screening in detecting visual defects questionnaires were sent to 240 families with blind or partially sighted children identified from the Family Fund's database. Questions were asked on social and family background, the visual disorder and its severity, any other disability, and how and when the disabilities were discovered and subsequently managed. Data from 189 families were analysed, constituting all those with children with major visual defects from the 219 families who replied. The visual defect was first discovered in 111 children by parents, friends, and neighbours, and in 36 by a doctor at the neonatal examination. In only three children who did not have a family history of visual impairment was the defect discovered during a formal screening examination at a child health clinic. Dissatisfaction about medical services was expressed by about a third of the parents, particularly a lack of provision of information and consideration of their worries and a failure to refer the child promptly to educational and treatment services.

Visual defects in children under 5 are generally detected by family and friends, not by screening, but detection by the medical profession could be improved by increased awareness and observation and quick referral.

Introduction

Detection of visual defects is one of the goals of developmental screening. Early diagnosis is thought to be desirable for many reasons: treatment may be more effective, developmental guidance is helpful for infants with severely reduced visual acuity, and genetic counselling may help to avoid the birth of another affected child. Perhaps the most compelling reason is that parents themselves value early diagnosis.[1]

In a previous study of vision screening in the under 5s we observed that screening played little part in detecting serious visual defects, which were usually detected by the parents or at the neonatal examination.[2] This observation was based only on clinical experience. We know of no study of how serious visual defects are first recognised. The purpose of this study was to determine whether screening does indeed play only a small part in detecting visual defects and whether early diagnosis is followed by prompt referral for developmental guidance and genetic counselling.

Patients and methods

Children with severe visual defects were identified through the Family Fund's database, which provides a unique means of locating children with severe disabilities.[3] The fund provides financial help to buy items such as laundry equipment, which may help to relieve the stress of caring for a severely disabled child. Grants are related to the family's economic circumstances, and most of the recipients are in the lower socioeconomic groups. Families were each sent a preliminary letter and if they agreed to participate were asked to complete a questionnaire (copy available on request). Only the staff of the Family Fund knew the identity of the families.

Each respondent was asked, "Who was the *first* person to *suspect* the defect?" Information was collected on social and family factors; the visual disorder and severity of visual impairment; other disabilities; and how and when these were detected and subsequently managed. A space was left for comment at the end of the questionnaire. The questionnaires were coded for analysis by computer and appropriate tabulations and significance tests performed.

Results

Altogether 240 questionnaires were dispatched. A total of 219 were returned, but 30 were rejected because of insufficient data or because the child was reported to have only a squint, unilateral defect, or minor refractive error. The analysis is based on the remaining 189 questionnaires. For 88 children the visual defect was the only problem. The remaining 101 included 62 children with multiple disabilities, 20 with mental handicap alone, and 19 with miscellaneous other defects.

The severity of visual impairment was as follows: totally blind, 39 children; able to perceive light, 33; able to detect movement or shapes, 27; able to recognise people or objects, 55; able to read print, 14; and uncertain (because of severe mental handicap), 18. Forty five different disorders were represented; the commonest were defects of the visual cortex associated with other handicaps, 30 children; retinopathy of prematurity, 20; and hypoplasia or atrophy of the optic nerve, 18.

The table shows who first suspected the visual defect. Family and friends between them detected nearly 60% of the cases. Examination of the neonate before discharge was the next most productive means of finding serious visual defects, with a yield of 37; for 10 of these children a high index of suspicion already existed because a family member had an inheritable eye disorder. In seven children a visual defect was discovered when their vision was assessed after they were found to have another major disability, such as mental handicap or cerebral palsy.

Person who first suspected visual defect

	Children with visual defect alone (n=88)	Children with visual defect and other disability (n=101)*	All children (n=189)*
Parent	50	50	100
Grandparent	4	2	6
Other relative	1	2	3
Friend or neighbour	2		2
Doctor (at neonatal examination)	16	20†	36†
Midwife	1		1
Health visitor	6	3	9
Child health doctor	1	2	3
General practitioner	3	1	4
Paediatrician (at follow up clinic)	4	9	13
Staff of child development centre		9	9
School doctor		1	1
Community orthoptist		1	1

*Data missing for one child.
†Includes three infants from special care found to have retinopathy of prematurity before discharge.

For only three children who did not have a family history of visual disorders and were apparently normal could the detection of a visual defect be attributed to a formal developmental screening procedure. One of these children was a baby found subsequently to have Norrie's disease, whose poor vision was noticed by a doctor at a clinic during the examination at 6

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weeks. For the two others the visual defect was detected by health visitors at 9 months of age, one in association with poor visual fixation and general developmental delay and the other in association with abnormal responses to a hearing test. In the six other children for whom the discovery of a visual defect was attributed to a health visitor the defects were recognised by informed observation rather than from the results of a screening test.

In 53 children the visual defect was suspected in the first week of life, in 79 by 6 weeks of age, and in 163 by 6 months of age. In children with more than one disability the visual defect was suspected first in 42, the other defect first in 40, and both simultaneously in 17.

The feature that first drew attention to the visual defect was the abnormal appearance of the eyes in 70 children, lack of fixation or following in 51, and abnormal movements of the eyes in 38. Squint, photophobia, epiphora, or reluctance to open the eyes was the main feature in 11, though these were present in many more. One baby was noted to "startle when touched."

Developmental advice was provided for 155 children; this was given by a peripatetic teacher of the visually handicapped for just under half of them and by various other staff for the remainder. Although 70 families received this advice within three months after diagnosis, 38 waited over a year. Genetic counselling was offered to and accepted by 72 families, although over a quarter of them experienced at least a year's delay between the diagnosis and their attending a clinic for genetic counselling. For 17 families referral was discussed but no consultation occurred. For a further 38 the history suggested strongly that genetic advice should have been offered.

About a third of the parents expressed dissatisfaction about medical services. The main complaints were the failure of doctors to listen to parents' worries; the lack of information about the child's disorder (almost one third of respondents were unable to give the precise name of the child's condition); inconsiderate care from medical and nursing staff in hospital; negative attitudes about intervention and management; and failure to refer the child promptly to educational and therapeutic services (many parents discovered these services for themselves). There were sometimes long delays between referral to an ophthalmologist and the actual consultation, although surprisingly few parents made any specific complaint about this.

Discussion

These results show that, although serious visual defects were detected in various ways, the formal vision testing procedures commonly used in clinics for developmental screening made little or no contribution. Most serious defects were discovered before 6 months of age. As we suspected, parents were efficient at detecting visual defects. The Family Fund's database contained an excess of families of low socioeconomic state, and it might be suggested that such families would be less capable than others of recognising defects in their infants. There were not enough families of social class I or II in our survey to test this hypothesis, but our finding that nearly 60% of the defects were detected by family or friends suggests that families of low socioeconomic state are perfectly capable of making relevant observations about defective vision.

Our previous study indicated that formal screening tests made little contribution to the detection of refractive error or squints in the under 5s,[2] and we can now confirm that they are not needed to detect serious defects. This does not mean that a knowledge of visual disorders is unnecessary for those practising developmental surveillance. On the contrary, a high level of awareness, sensitivity, and knowledge is necessary so that parents can rely on an appropriate response to their worries and opportunities for early detection are exploited efficiently.

Our results have some important implications for the training of junior hospital doctors who examine neonates and of health visitors and the other primary care professionals who are responsible for developmental surveillance in the community. We suggest that eight points should be emphasised:

(1) A careful inspection of the newborn infant's eyes in a good light will detect many defects. The examination is not complete until this is done. The mother should also be asked if she has noted any abnormality in the eyes. We suspect that mothers commonly detect a problem very early but, because of uncertainty about its importance, fail to mention it immediately.

(2) Any infant with a family history of a serious visual disorder, particularly one that is or could be inherited, should be examined with extra care, preferably by an ophthalmologist.

(3) The high prevalence of retinopathy of prematurity emphasised again the need to examine the eyes of all infants at risk in neonatal intensive care units.

(4) Parents are the most effective detectors of visual defects, but they need access to an efficient and responsive professional network to obtain appropriate referrals and advice.

(5) The main factors that alert parents are the appearance of the eyes, lack of fixation or following, and wandering eye movements. Photophobia, epiphora, squint, reluctance to open the eyes, and starting when touched are much less common but are nevertheless important. The importance of persistent uniocular squint in a young baby must continue to be emphasised. Primary care staff should be taught to ask about and look for these symptoms and signs.

(6) The symptoms and signs listed are uncommon and can be used as indications for prompt and urgent referral without fear of overwhelming clinics. Although most of our respondents seemed remarkably tolerant of long delays, they must have experienced considerable worry. Referring doctors should request priority consultations for infants with suspected visual defects, and ophthalmic clinics should ensure that they respond quickly.

(7) Among children with poor vision and other disabilities about half come to attention because of the visual defect and half because of the other problems. This finding emphasises again that infants with a visual defect need paediatric assessment and that other major defects are an indication for ophthalmic consultation.[4]

(8) Abnormal visual behaviour may be noted during hearing tests or developmental screening examinations, particularly between 6 months and 1 year of age, and is often associated with severe mental handicap or cerebral palsy.

Three additional points emerged from our questionnaire that are of particular relevance to ophthalmic clinics. Firstly, many parents resented the lack of information provided, and as many of the conditions were rare and had obscure names they found it difficult to make inquiries on their own. For the same reason they were unlikely to know another family with a similar problem.

Secondly, although children were usually referred to the peripatetic teacher, long delays were often experienced before this or any other help was suggested. Both of these problems were sometimes avoided by early referral to child development or assessment centres, which can provide appropriate treatment and support and are familiar with educational networks and organisations for parents. Although the services of the teacher are invaluable, additional advice is often needed, particularly for children with multiple disabilities.

Thirdly, genetic counselling services seem to be underused.[5] Many families were not referred at all despite a diagnosis of an inheritable disorder. In several children an eye defect was attributed to events of dubious relevance, such as asphyxia or pneumonia. More often, genetic counselling was suggested, but only after a long delay, which could have allowed the birth of another disabled child. Furthermore, learning that the child's defect may be inherited causes further distress when parents are coming to terms with the disability: parents seem to prefer to receive *all* the bad news at once.

These results and those from our previous survey suggest that knowledge, awareness, astute inquiry and observation, and an efficient referral system are the ingredients for a successful early detection system for visual defects. The available screening tests contribute little to detecting defects in the under 5s.

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