Have we stopped looking for a red reflex in newborn screening?

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Abstract

Best medical evidence indicates that surgical treatment of significant congenital cataracts is required within the first 3 months of life for optimal visual outcome. The aim of the present study was to review when the diagnosis of congenital cataracts was made in our region, by whom it was made, and the visual outcome at 2 years of age or more. There were 455 cases of cataracts on the Cork and Kerry Paediatric Eye Database. Children with a cataract(s) present from infancy had a poor visual outcome. A recent national study in the United Kingdom indicated that almost half of congenital cataracts (47%) in total were diagnosed soon after birth and at 6-8 weeks of age. The American Academy Of Paediatrician has also recommended that all infants should have an examination of the red reflex of the eyes performed during the first 2 months of life by a Paediatrician or other Primary Care Clinician trained in this examination technique. The aim of the present study is to review when the diagnosis of congenital cataract was made, by whom it was made and visual outcome at 2 years of age or later.

Introduction

The Royal College Of Paediatrics and Child Health and the Royal College Of Ophthalmologists have recommended that ophthalmic examination of all babies is required in the first few weeks of life. Partly as a result of delayed diagnosis, only 2 out of 19 babies diagnosed with congenital cataracts before three months of age had a good visual outcome after surgery in contrast to only 2 out of 19 who were diagnosed after 3 months of age (p <0.005). Early detection of this ocular disorder is not only important for a good outcome, but also allows genetic counselling, advice and support to be offered at the earliest opportunity.

Methods

This was a retrospective study in the counties of Cork and Kerry, which have a population of 546,000 and approximately 8500 births per annum. This region was served by a single regional ophthalmology centre based at Cork University Hospital. All children under 15 years, diagnosed with Congenital Cataract (CC) over a 10-year period (1991-2002), were included in this study. Information was retrieved from the Hospital In-Patient Enquiry (RIPE) database. Those children with a cataract(s) present from infancy had a poor visual outcome. The medical notes were manually reviewed for demographic data, mode of detection, age at diagnosis, first person to detect the disease, unilateral/bilateral diseases and visual outcome at 2 years of age and more.

Results

27 cases of congenital and infantile cataract (15 males, 12 females) were identified. Of the 27 cases with congenital cataracts, 17 (63%) were diagnosed with bilateral disease and 10 (37%) were unilateral. Over half, 17/27 (63%) with congenital cataract were diagnosed following presentation with parental/carer concerns about visual function (usually a squint). The remaining cases however, only 17 of the 17 cases detected by carers/parents presented before 3 months of age. The remaining cases of congenital cataracts were diagnosed by general practitioners 8 (24%), paediatricians 4 (12%), optometrists 3 (9%) and School Medical Officer 1 (3%).

In total, 8 cases presented before 3 months of age (range 2 days to 10 weeks), while the remaining presented after then, though 2 cases presented at 4 months of age, others presented between 7 months and 11 years. Good visual outcome was defined as a visual acuity better than 6/24 at 2 years of age. Six out of eight children diagnosed with congenital cataracts before three months of age had a good visual outcome after surgery in contrast to only 2 out of 19 who were diagnosed after 3 months of age (p <0.005).

Discussion

This study has re-affirmed the need for early diagnosis and treatment of congenital cataracts. The role of the general practitioner in early diagnosis and referral to specialist care is highlighted. However, we cannot wait until parental concerns bring the child to medical attention as this is often too late for a satisfactory visual outcome for the child.

Much to our dismay, no baby with congenital cataracts was diagnosed by the neonatal screening examination for defects during the 10 years of the study. This is in marked contrast to the UK national study where one-third of cases were picked up by newborn screening. Partly as a result of delayed diagnosis, only 2 out of 19 babies diagnosed with congenital cataracts before three months of age had a good visual outcome after surgery in contrast to only 2 out of 19 who were diagnosed after 3 months of age (p <0.005). Early detection of this ocular disorder is not only important for a good outcome, but also allows genetic counselling, advice and support to be offered at the earliest opportunity.

Though other factors also contributed to poor visual outcome, these include association of other syndromes/ocular disorders. For example, a case of Hallerman-Streiff syndrome, and another case of Alstrom syndrome, both of these had a poor visual outcome. Despite being diagnosed with unilateral cataract at 7 days and 5 months respectively, other risk factors include hyperplastic vitreous and hyphaema in a 9 month and 7 days old respectively, both with unilateral cataract and ending up with a poor visual outcome after surgery.

The results of this study are a poor reflection (no pun intended) on the neonatal screening practices in our region. Though a number of factors could be involved. The poor detection could be because of lack of interest, lack of training or lack of resources (working ophthalmoscopes easily available). Lack of interest may have been related to the unavailability of the necessity for early diagnosis and treatment. Doctors and other health professionals must be kept informed about the need for a high index of suspicion and the need for early diagnosis of congenital cataract.

Conclusion

The ocular examination of young infants is quite simple, though it requires specific knowledge and skills. However, with appropriate training it can be performed by clinical staff with limited previous experience. The procedure of red reflex examination is well described in the policy statement released by the American Academy Of Paediatrics in May 2002 – an ophthalmoscope is held close to the examiners eye and focussed on the pupil is used to detect a red reflex. Dark spots in the red reflex or the presence of a white reflex (retina reflection) are all warning signs of potential anomalies or defects. Early detection of this disorder is not only important for a good outcome, but also allows genetic counselling, advice and support to be offered at the earliest opportunity.

Newborn screening should also become familiar with the light reflex in dark-skinned infants, who tend to have a white rather than a red reflex. This should not be confused with the appearance of a congenital retinoblastoma, which also appears as a white reflex and also warrants early diagnosis and treatment.

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Some institutions have trained nurses to perform newborn screening examinations for congenital anomalies. There is no evidence that health examinations carried out by nurses are of lower quality in detecting health problems than those carried out by physicians. Indeed, in one study overall maternal satisfaction was high and higher when a midwife rather than an SHO examined.

There are existing recommendations about teaching paediatric ophthalmic disorder and visual assessment of children during the postgraduate training of paediatricians. The reasons why the red light reflex examination is not consistently part of newborn screening needs to be explored further. Education as to the importance of early diagnosis, instruction to the performance of red light reflex and easy access to ophthalmoscopes on the postnatal wards need to be addressed.

General Practitioners form an important link in the detection of cataract by the red light reflex, since they conduct a routine 6 weeks check and see babies at 2, 4 and 6 months for immunisations. While not denying the responsibilities of doctors who perform neonatal screening examinations, performing the red light reflex at 6 weeks of age is usually easier than examination in the newborn period, in that babies are more visually alert and the eyelids are not oedematous as it is commonly seen in the newborn immediately after birth. We have found that performing the examination while standing at the baby's head, using the index and thumb to separate the eyelids increases the successful visualisation of the red light reflex. It is important that the examination is done while the baby is not crying, since babies usually shut their eyes tightly while crying.

Parents are also important in the chain of diagnosis. However, most cases presenting with parental concern were diagnosed after 3 months of age, which would not have altered visual outcome to a significant degree. Finally, all babies with possible syndromes should be screened and referred to specialist attention urgently. With these small numbers, we have confirmed the very poor prognosis of congenital cataract diagnosed after 3 months of age. This same conclusion was reached in other related studies. We must not stop looking at the red light reflex as an essential part of our routine examination of newborn and older infants.

References


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