Childhood blindness in the Republic of Ireland: a national survey

Michael Goggin, Michael O'Keefe

Abstract

We completed a national study of blindness in children under 16. Approximately 80% of the blind children (that is, with vision of 3/60 or less) in the Republic of Ireland (172 children) were seen, 93 males and 79 females. The survey was carried out between July 1989 and June 1990. It is the first such study to be carried out. Ninety seven (56%) children had lesions due to factors acting before the perinatal period. Of these, 28 (16% of the total number surveyed) had lesions due to genetic causes; 69 (40%) had lesions due to factors operating in the prenatal period other than genetic factors. Forty six (27%) had lesions due to factors acting in the perinatal period. Twenty two (13%) had lesions due to factors acting in childhood. (4% could not be categorised in this way). The commonest single primary diagnoses were birth asphyxia in 19 (11%) cases and retinopathy of prematurity in 19 (11%) cases.

This report outlines the major findings of a national survey of blind children in the Republic of Ireland. A survey carried out in Northern Ireland in 1976[1] found 142 patients under 21 years blind by World Health Organisation (WHO) standards – that is, visual acuity of 3/60 or less.[2] This represents about 0·02% of the under 21 population at that time. The current population of the Republic aged under 16 is 1 094 932 (at the last census in 1986). These figures would suggest that there are about 220 blind children under 16 in the Republic at present. Information on childhood blindness was not available in the Republic before this survey.

We set out to identify and record all blind children in the Republic of Ireland, and to institute a national live register of blind children.

Table 1 Ophthalmic diagnoses by category

Category: chromosomal/genetic: 28 cases			
Leber's amaurosis	3	Anophthalmos	2
Cortical blindness	3	Optic atrophy	2
Optic nerve hypoplasia	3	Rothmund-Thompson	2
Albinism	3	Microphthalmos/coloboma	2
Cataract	3	Buphthalmos	1
Leber's optic atrophy	2	Usher's syndrome	1
Retinitis pigmentosa	2	Stargardt's syndrome	1
Metabolic retinopathy	2		
Category: prenatal: 69 cases			
Optic nerve hypoplasia	25	Congenital toxoplasmosis	2
Optic atrophy	21	Peters's anomaly	2
Cortical blindness	10	Buphthalmos	2
Pigmentary retinopathy	6	Corneal dystrophy	1
Microphthalmos/coloboma	5	Motor nystagmus	1
Congenital rubella	5	Bilateral amblyopia	1
Cararact	5	Uveitis (? cause)	1
Anophthalmos	4		
Category: perinatal: 46 cases			
Retinopathy of prematurity	19	Cortical blindness	11
Optic atrophy	14	Optic nerve hypoplasia	9
Category: childhood: 22 cases			
Optic atrophy	9	Uveitis (? cause)	1
Retinoblastoma	8	Retinal detachment	1
Cortical blindness	3		
Category: unassigned: 7 cases			
Tractional retinal detachment			2
Optic atrophy ?cause			2
Optic nerve hypoplasia with hormone deficiencies			1
? Viral retinal pigment epitheliitis			1
Isolated microphthalmos			1

Materials and methods

SOURCES OF REFERRAL

A national survey of blind children under 16 years old on 1 July 1989 (that is, born on or after 2 July 1973) whose best corrected vision was 3/60 or worse in their better eye was carried out. Patients were sought by several methods. All consultant ophthalmologists were circularised. The schools for blind and physically handicapped, the mental handicap institutions, the social service and educational bodies responsible for care of the blind were contacted nationwide to refer suitable patients and also asked for any records of patients known to them who fitted the criteria. They provided comprehensive lists of such cases.

EXAMINATION OF PATIENTS

We examined all the patients. A history was taken when possible from a parent or a responsible adult. This included a family history, a history of the pregnancy, birth, and early postnatal period, and any relevant previous medical and ophthalmic history. Confirmatory medical data were sought when necessary from the relevant medical institutions.

The best corrected visual acuity was measured by the appropriate method for age, ability to co-operate, and level of intelligence. When possible this was by Snellen chart for distant vision. When the child was too young for the use of subjective tests of visual acuity, an assessment of the ability to fixate was made. Patients with central unsteady, unmaintained fixation or worse were deemed eligible for registration. When possible with the more co-operative patients and when the patients were able to attend the Children's Hospital more accurate measurements were made by Forced Choice Preferential Looking techniques. Ability to distinguish a pattern of 1·5 cycles/degree or less at 38 cm (that is, an approximate Snellen equivalent of 3/60 or less) was taken as the criterion for inclusion in the study. Electrophysiological examinations were carried out when necessary.

A full ophthalmic examination was under-

Department of Ophthalmology, Children's Hospital, Temple Street, Dublin 1, and UCD Department of Ophthalmology, Mater Misericordiae Hospital, Eccles Street, Dublin 7
M Goggin
M O'Keeffe

Correspondence to:
Michael O'Keeffe, FRCS, Department of Paediatric Ophthalmology, Children's Hospital, Temple Street, Dublin 1.

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taken in every case. A systemic examination was carried out by a paediatrician at the Children's Hospital if it was required to establish a diagnosis. Laboratory investigations were also carried out if necessary for a diagnosis. X rays, CT scanning, nuclear magnetic resonance (NMR) imaging, and ultrasound examination were undertaken for the same purpose.

All the relevant data were collected on data sheets and put in a computer database for future retrieval.

Results

A total of 172 blind children were seen. There were 93 males and 79 females, a ratio of 1·18:1. Seventy nine (46%) of the children registered were aged under 5, 39 (23%) were aged 5 to 10 years inclusive, and 54 (31%) were 11 or over but under 16 on 1 July 1989.

An ophthalmic diagnosis was recorded for all the children seen. A systemic diagnosis was also recorded in 112 cases (65%). The diagnosis was assigned to a category, genetic or chromosomal, prenatal, perinatal, or childhood depending on the aetiology. A complete list of diagnoses by category is given in Table 1. Many children had several related diagnoses.

CHROMOSOMAL OR GENETIC

Twenty eight children (16%) had diagnoses that fell into this group. Leber's amaurosis, albinism, and cortical blindness associated with genetic or chromosomal disorders were the most frequent diagnoses, with three cases each. The lesions causing cortical blindness were an extra chromosome 15, Pelizaeus-Merzbacher disease, and a peroxisomal disorder. Two cases of Rothmund-Thompson syndrome were discovered. Two cases of retinopathy associated with metabolic disorders were seen, one with Tay-Sachs disease and the other with a peroxisomal disorder. One case of Patau's syndrome with bilateral anophthalmos was seen. A deletion of the short arm of chromosome 11 was associated with aniridia, corneal opacity, and cataract in one child. Dicarboxylic aciduria was associated with optic atrophy in another.

In three families a history of parental consanguinity was obtained (1·74%). In two of these the parents were first cousins and in the third family second cousins. The diagnoses were bilateral anophthalmos and optic nerve hypoplasia in the first two cases, and optic atrophy in a background of hydrocephalus from aqueduct stenosis in the third.

PRENATAL

These were cases with lesions occurring before the perinatal period that could not be classified as chromosomal or genetic: 69 cases (40%) fell into this category. The most frequent findings here were optic nerve hypoplasia in 25 cases and optic atrophy in 21. Cortical blindness was diagnosed in 10 cases. Many of the latter two diseases were secondary to systemic abnormalities such as hydrocephalus or metabolic disorders (many of which were not fully elucidated at the time of the study) and were associated in the same patient. Optic nerve hypoplasia was associated with absent corpus callosum, fetal alcohol syndrome, twin-to-twin transfusion, spina bifida, and septo-optic dysplasia. Rubella syndrome was diagnosed in five, the youngest being 11 years old. A further five each had colobomata or non-hereditary cataract. One child with colobomata was thought to have Goltz syndrome, but because the evidence was inconclusive she was categorised as having a coloboma of prenatal cause rather than hereditary (Goltz syndrome is thought to have an X-linked pattern of inheritance).

PERINATAL

A total of 46 (27%) cases were in this group. Nineteen cases of retinopathy of prematurity were seen, 14 of optic atrophy, and 11 of cortical blindness. The latter two diseases were frequently seen together in the same patient and caused by hypoxic ischaemia from birth trauma. Nineteen cases of hypoxic ischaemic damage causing blindness were seen. In nine further cases there was documented birth trauma, but examination revealed optic nerve hypoplasia associated with the primary cause of blindness (namely, cortical damage or optic atrophy usually), implying some underlying prenatal abnormality.

CHILDHOOD

Twenty two patients (13%) had lesions occurring after the perinatal period. Eight of these had bilateral retinoblastoma, for which seven were treated by bilateral enucleation. The eighth received radiotherapy to the second eye, which became blind subsequently. It is likely that some of these bilateral cases have a genetic aetiology. However, none had affected parents, siblings, or more distant relatives. None were shown to have an abnormal karyotype. In the absence of firm evidence to suggest a genetic background they have been put in this category. Nine showed optic atrophy, six following infective encephalopathies (two of whom also had cortical vision defects) and three secondary to intracranial tumours. One other child was cortically blind secondarily to Reye's syndrome. One case of blindness due to self-injurious behaviour associated with mental handicap was seen. Finally, one child with Down's syndrome was seen with bilateral phthisis following bilateral retinal detachment.

CATEGORY NOT ASSIGNED

Seven cases (4%) could not be assigned to the above categories owing to difficulties in arriving at specific diagnoses. Two had tractional detachments of unknown aetiology. One had a pigmentary retinopathy that may have been secondary to a viral retinal pigment epitheliopathy.

MENTAL HANDICAP

Seventy seven of the 172 cases seen had mental handicap (44·5%). Of those, six had profound handicap, 23 severe, nine moderate, and four

mild. The other 35 had not had formal measurement of the intelligence quotient at the time of the survey since they were too young. Most of the latter cases would have been functioning in the 'severe' range as assessed by those caring for them. Seventy five children had developmental delay, usually associated with either mental or physical handicap or both. Twenty six of those with developmental delay, however, were too young to have had a full assessment of their intelligence. Twenty three of this 26 had systemic diagnoses (such as profound cerebral palsy) that made the confirmation of mental handicap at a later date likely. This would then give a total of about 59% with possible mental handicap.

VISUAL FUNCTION

For the purposes of inclusion in the survey and the register of blind children visual function was assessed in one of several ways by means of the most suitable for the child being examined. In 105 cases this was based on the fixation pattern. Snellen acuity, or perception of light, hand movement, or finger counting were used in 64, and three were assessed by forced choice preferential looking techniques. Visual function was the same in both eyes in 150 children. In 72 cases this consisted of 'uncentral', unsteady, and unmaintained fixation (ucusum) (probably representing a Snellen vision of less than 6/60). A further 28 had central, unsteady, and unmaintained fixation (cusum) (roughly equivalent to a Snellen acuity of 6/60.[3] Twenty three had no perception of light (NPL), 14 had definite light perception (PL), and three had possible PL. Two patients had a Snellen acuity of 1/60 in both eyes and two had 3/60. One child could count fingers and two had hand movement vision bilaterally. Three children had their vision assessed by preferential looking techniques, and their resolving power was estimated at 1·5, 1·3, and 0·32 cycles/degree testing at 38 cm respectively. Visual function was asymmetrical in 22 children. Ten had PL in the better eye, three had HM, three had cusum, two had ucusum, two had 1/60, and two had 3/60, all in the better eye (Table 2).

SURGERY

Eleven patients underwent cataract extraction or clear lens extraction as part of another procedure. Nine had enucleations, seven bilateral, six for retinoblastoma, and one for buphthalmos. Five had had corneal transplantations. Three had vitrectomies and two conventional retinal detachment surgery. Thirty one patients had had surgery of some description (Table 3).

Discussion

It has been pointed out in the past that several African and other third world countries have more accurate statistics on the prevalence of blinding disease than the developed nations.[4] Last year the results of the national survey of blindness and low vision in The Gambia were published.[5] A large random stratified sample of this country's population was examined (8174 people). The main causes of blindness found were cataract and uncorrected aphakia, non-trachomatous corneal opacity and phthisis bulbi, and trachoma. Similar studies have been published from Malawi in the past[6] and more recently from Kenya.[7] Two articles in 1989 documented trends in blind registration in two health authorities in the UK.[8,9] Though these latter studies contain accurate and interesting information, it cannot be assumed that they reflect accurately the make-up of the blind population of either the relevant health authorities or the UK as a whole, because the statistics are not derived from a random sample of the population but a highly selected one, namely, those registered as visually impaired in specific areas. Our survey found 108 blind children not previously registered on the Blind Register maintained by the National Association for the Blind in Ireland out of a total of 172 blind children seen (that is, 62·79% were not previously registered). Reliance on registered data is therefore suspect.

For the statistics from any register to be of use the register must actively seek out those visually impaired people in the population, document them accurately (this involves ontinued review of those registered), and delete those who die or whose acuity improves to a level greater than that at which they would qualify for registration – that is, a 'live' register.

Our study is an attempt to document

Table 2 Visual function by method of assessment

Fixation pattern assessment: 105 cases		
	Symmetrical vision	*Asymmetrical vision (vision in better eye)*
ucusum	72	2
cusum	28	3
Snellen visual acuity (including NPL, PL, HM, CF): 64 cases		
	Symmetrical vision	*Asymmetrical vision (vision in better eye)*
NPL	23+	0
PL	14 (3 ?PL)	10
HM	2	3
CF	1	0
1/60	2	2
3/60	2	2
Forced choice preferential looking: 3 cases		
	Symmetrical vision	*Asymmetrical vision (vision in better eye)*
1·5 cycles/degree	1	0
1·3 cycles/degree	1	0
0·32 cycle/degree	1	0

ucusum=uncentral unsteady unmaintained.
cusum=central unsteady unmaintained.
NPL=no perception of light.
PL=perception of light.
HM=hand movements.
CF=counting fingers.
Where vision was asymmetrical, the vision in the better eye is recorded in this table.

Table 3 Surgery carried out on children registered (number of procedures)

Cataract extraction/clear lensectomy	11
Enucleation	9 (7 bilateral)
Corneal transplant	5
Vitrectomy	3
Conventional retinal detachment surgery	2
Squint surgery	2
Trabeculectomy	2
Radiotherapy	2
Probing of nasolacrimal duct	1
Peripheral iridectomy	1
Correction of entropion	1
YAG laser capsulotomy	1
Removal of band keratopathy	1
Total patients	31

accurately the population of blind children (with vision less than or equal to 3/60 in the better eye) in the Republic of Ireland by examining them all ourselves. This information is now incorporated in the Blind Register as the start of a live register. Since such measures of visual function as fixation pattern or even preferential looking do not always correspond with eventual Snellen acuity, some children are registered who will turn out not to be 'blind' by the Snellen criteria set out above. However, this is a live register, and when these children come for review they can be deleted. Because of this we recommend patients should be registered as soon as it is established that their level of visual function, as measured by the most appropriate test available in the individual case, meets the criteria set out. Reliance on Snellen acuity testing as a criterion for blind registration is inappropriate for the preverbal child or for the mentally handicapped.

It is interesting to note that a similar study was carried out in Northern Ireland in the mid 1970s. The ascertainment rate was estimated to be similar to ours, namely 80%.

In Britain several monographs have been published by the Department of Health and Social Security on blindness and partial sight. In recent years these have been confined to the adult population; the most recent containing information on children was published in 1979[10] and referred to the years 1969–76 in England. The information contained in this report is derived from blind registers and not active ascertainment of all cases in the population. The statistics therefore must not be taken as an accurate reflection of the actual incidence and prevalence of the conditions described, but merely as the best available. Our survey, however, has attempted to assess the whole population so that the statistics produced are as accurate as is possible.

The criteria in this study for 'blindness' are stringent by most standards. Legal blindness in the Republic of Ireland is usually taken to be 6/60 or worse in the better eye, or 20° field or worse in the better eye. However, the WHO definition is 3/60 or less in the better eye.[2] For many children a vision of 6/60 does not preclude education by visual methods. Snellen acuity cannot be measured in preverbal infants or in the more severely mentally handicapped. Yet many blind children fall into these categories. For this reason we have set out different, but roughly equivalent, criteria for registration in these groups. An exact correlation between Snellen acuity and fixation pattern or even preferential looking measurements cannot be made, but, as long as those registered are subject to continual review, those patients who with maturation can subsequently be measured by Snellen methods will have this carried out and inappropriate registration altered.

The major causes of blindness in children have been estimated in several publications. The most recent DHSS monograph[10] shows that during 1969–75 in England the commonest causes of new registration in children under 16 were optic atrophy and cataract (no comment is made on the primary systemic diagnoses in these groups, which can vary considerably) followed by congenital abnormalities. In European studies in the mid 1970s in Holland, Belgium, Norway, and Denmark optic atrophy, cataract, congenital abnormalities, and retinopathy of prematurity figured largely also, but many cases of tapetoretinal dystrophy were seen.[11] Foster's review of worldwide statistics concluded that in the developed world childhood blindness was largely due to hereditary causes and factors operating at the time of birth (such as hypoxia).[12] Of note is his comment that better care of preterm infants may have led to an increase in visual handicap.

In countries where there are medical services of intermediate quality intrauterine infections, in particular rubella, are an important preventable cause of blindness. In areas where the medical services are poor, such as many rural and urban slum areas in Asia, Africa, and parts of South America, vitamin A deficiency, measles, and the lack of ophthalmic services are responsible for 50–75% of childhood blindness. Bryars's study in Northern Ireland of visually handicapped children[1] showed that 95% of his cases had pre- or perinatal causes, and 51% were genetic in origin. Of note is the fact that birth injury caused only 2% of his cases. However, 18% were attributed to optic atrophy, with no comment on the primary diagnosis, and presumably some of these are as a result of birth asphyxia. He does note a high incidence of optic atrophy among the mentally handicapped.

In this survey 28 out of 172 (16·28%) had genetically determined disease. This figure is considerably less than those quoted by Baraitser[13] and Jay.[14] Sixty nine cases, however, had causes of their disease that were categorised as prenatal. Many of these had conditions which, though not definitely genetic in origin, may have some genetic background in a multifactorial aetiology, such as optic nerve hypoplasia. It is possible that our strict criteria for inclusion in the genetic category, namely an accurate diagnosis with proved genetic aetiology, may have decreased our total. An associated systemic diagnosis was made in 112 cases (65%). Little reference is made in the DHSS reports (or any of the other large surveys) to this important aspect of the subject. Without this information the significance of a morphological diagnosis such as optic atrophy cannot be assessed, since the aetiology is so diverse.

The commonest morphological diagnoses (which often coexisted in the same patient) were optic atrophy (48 cases), optic nerve hypoplasia (39 cases), and cortical blindness (27 cases). The commonest single causative factor in the first and last of these was birth asphyxia (19 cases). The prominence of optic nerve hypoplasia in this list probably indicates increased recognition of this sometimes subtle condition. A small number have only this isolated abnormality (3 cases) but a great many more are associated with other congenital abnormalities, such as albinism, hydrocephalus, agenesis of the corpus callosum, and septo-optic dysplasia. Retinopathy of prematurity is still a major cause of childhood blindness, seen here in 19 children. However, none of the children so affected were born after September 1987, so perhaps the management of the condition is improving. Recent experience in the

field of retinal cryotherapy in this condition gives reason for hope, suggesting a reduction by a half in the numbers of eyes with an unfavourable outcome.[15]

Eleven children were found to be blind owing to tumours. Eight had blinding bilateral retinoblastoma, and three more had intracranial tumours causing damage to the anterior visual pathways (a craniopharyngioma, a hypothalamic astrocytoma, and an optic chiasm glioma).

The two commonest single primary causative agents are prematurity and birth asphyxia, both with 19 cases (11% of the total each). It is important to mention that we did not come across any cases of blindness from bilateral untreated or poorly treated amblyopia. Only three cases of blindness from childhood glaucoma were seen. Rubella eye disease seems to have been almost eradicated, the youngest patient (of only five) was 11 years old. The pattern of diagnoses fits roughly into that seen in other western nations.[12] 44% of the children seen were mentally handicapped. This is similar to other European studies. But if those with developmental delay and diseases likely to lead to mental handicap are included the total is nearer 60%. This can be explained by our confining our study to the severely visually impaired. These children tend to have a higher prevalence of mental handicap.[11]

A large proportion of our cases were under 5 years (46%). This may be attributable to our easy access to the three largest maternity units in the country, all located in Dublin and all referring cases directly to our own paediatric ophthalmology service.

This study shows that almost 30% of the causes of blindness in this country are potentially remediable – genetic causes (16%) through genetic counselling, retinopathy of prematurity (11%), and birth asphyxia (11%) – and 40% cannot be tackled with present knowledge, comprising those prenatal causes that have not yet become amenable to treatment.

One of the chief reasons for undertaking this survey was to upgrade the quality of the ophthalmic and background medical data on the current blind register and to register as many new blind children as possible. The data collected will now constitute the start of a live register to be maintained by the National Council for the Blind in Ireland. A live register would provide information on incidence and prevalence and demographic risk factors, and give invaluable aid to researchers and to those who provide services to the target population.

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