Measuring and Interpreting the Incidence of Congenital Ocular Anomalies: Lessons from a National Study of Congenital Cataract in the UK

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Purpose. Prevention of visual impairment due to congenital cataract is an international priority. Estimates of incidence are required for implementation and assessment of preventive strategies, but are not widely available, despite routine monitoring of birth defects at a national level in many industrialized countries. The purpose of this study was to determine the incidence of new diagnosis of congenital and infantile cataract in the United Kingdom.

Methods. All children with newly diagnosed congenital and infantile cataract in the United Kingdom in 1 year from October 1995 through September 1996 were identified using independent ophthalmic and pediatric national active surveillance schemes. Capture–recapture analysis was used to estimate completeness of ascertainment. Annual age-specific and cumulative incidence were estimated and adjusted for ascertainment.

Results. Two hundred forty-eight children with newly diagnosed congenital or infantile cataract were identified—an estimated 92% of eligible cases. The adjusted annual age-specific incidence of new diagnosis of congenital and infantile cataract was highest in the first year of life, being 2.49 per 10,000 children (95% confidence interval [CI], 2.10–2.87). Adjusted cumulative incidence at 5 years was 3.18 per 10,000 (95% CI, 2.76–3.59), increasing to 3.46 per 10,000 by 15 years (95% CI, 3.02–3.90). Incidence of bilateral cataract was higher than that of unilateral, but incidence did not vary by sex or country of residence.

Conclusions. These estimates of congenital and infantile cataract incidence were higher than reported previously from routine sources relying on passive notification around the time of birth. Studies of congenital ocular anomalies that are not always readily diagnosed at birth should consider the potential influence on disease frequency of diagnostic practices as well as of underlying disease risk. (*Invest Ophthalmol Vis Sci.* 2001; 42:1444–1448)

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Birth defects are an important cause of ill health and death among children throughout the world.[1] Although many anomalies are individually uncommon, their collective impact is considerable, and thus much epidemiologic research in child health seeks to elucidate their causes.[2] In industrialized countries, there are national monitoring schemes to routinely scrutinize trends in the frequency of such anomalies over time and between countries.[3] Congenital ocular anomalies are major contributors to childhood visual morbidity. Congenital cataract is one of the few of these visually handicapping disorders that is amenable to primary prevention—for example, through a rubella immunization program—or more commonly, to secondary prevention, which depends on early diagnosis, mainly through screening, to ensure prompt treatment.[4–6]

Nevertheless, congenital cataract remains a major cause of visual impairment and blindness in childhood throughout the world,[5–7] the prevention of which is now an international priority.[7] Estimates of the incidence of congenital and infantile cataract are required for implementation and assessment of preventive strategies and provision of appropriate services, as well as for identification and investigation of secular trends in etiology and diagnostic practices within and between countries. Such incidence data are currently not available in many settings.[5] We report the incidence of congenital and infantile cataract in the United Kingdom, the influence of diagnostic practices on disease frequency, and associations with demographic factors.

Methods

Case Ascertainment by Population-Based Active Surveillance

Children with newly diagnosed congenital cataract were ascertained simultaneously, but independently, through ophthalmologists and pediatricians. This reflects the delivery of screening and treatment services for congenital cataract in the United Kingdom, to which there is universal, cost-free access through the National Health Service. Ophthalmologists undertake treatment, and pediatricians are responsible for universal routine ocular examinations of newborn infants undertaken to detect cataract as well as for the management of any underlying or associated systemic disorders.[8] Subsequently, at specified ages throughout childhood, routine assessments of all children are undertaken, by a variety of health professionals, to monitor normal visual development and to detect less severe disorders, such as strabismus.[8]

All children with congenital or infantile cataract in the United Kingdom, newly diagnosed during the 12-month period from October 1995 to September 1996 inclusive, were identified prospectively through two independent national active surveillance schemes. The ophthalmic surveillance scheme was established for this study, through the British Congenital Cataract Interest Group, after a national survey of practice.[9] The long-established pediatric scheme, run by the British Paediatric Surveillance Unit of the Royal College of Paediatrics and Child Health, has successfully facilitated incidence studies of a number of uncommon childhood conditions.[10]

For the duration of the study, pediatricians were sent reporting cards monthly and ophthalmologists every 2 months, with which to notify of new cases (as defined in the next section) or to confirm that no new cases had been identified. Independence between the two schemes was maintained throughout in all aspects, including the notification procedure, data collection, and other communication with reporting clinicians.

Case Definition

Because the clinical management of both congenital and infantile cataract is the same, these terms are usually used interchangeably in clinical practice,[4] although standard disease classification systems[11] distinguish between these and further subcategories of cataract occurring in the first year of life. Thus, a clinical case definition was adopted for this study. Clinicians were asked to notify any child, aged 15 years or less and born in the United Kingdom, with newly diagnosed congenital or infantile cataract, irrespective of treatment undertaken. Children who died after diagnosis were eligible. Notified children in whom cataract was diagnosed after the age of 1 year were eligible for inclusion only if, on review, the cataracts were confirmed to be due to a congenital cause or had specific ophthalmic features indicative of early onset, such as cataract morphology (e.g., polar cataract), associated congenital ocular anomaly (e.g., persistent hyperplastic primary vitreous), or presence of nystagmus. Only children with visually significant cataract were eligible. Those with minor lens opacities or in whom cataract was acquired—for example as result of trauma, uveitis, irradiation, or drugs—were excluded.

Data Collection

After notification of a new case, the reporting clinician was sent a form requesting detailed information about the patient. This included unique identifiers, such as initials, date of birth, gender, eye(s) affected, and country of residence, to allow matching of cases notified by both surveillance schemes, together with age at diagnosis and underlying or associated cause(s) of cataract. Up to two reminders were sent to nonresponding clinicians at eight-week intervals.

Analysis

Cumulative incidence (risk) and the annual age-specific incidence (rate) of new diagnosis of congenital and infantile cataract were estimated, together with their 95% confidence intervals (CIs), for all cases combined and according to laterality, sex, and country of residence at diagnosis, using the method of Breslow and Day.[12] Incidence estimates in the first year of life were calculated from the ratio of new cases to total annual live births, and for other ages from the ratio of new cases to the midyear population of children in each age group. During the study period, there were 735,000 live births in the United Kingdom: 615,000 in England, 61,000 in Scotland, 35,000 in Wales, and 24,000 in Northern Ireland.[13] Of the10.63 million children between 1 and 15 years living in the United Kingdom during this time, 8.82 million were resident in England, 0.91 million in Scotland, 0.53 million in Wales, and 0.37 million in Northern Ireland.[13]

It is important to measure undercounting in studies of rare conditions to ensure accurate estimation of disease frequency. Capture–recapture analysis is a recognized method for quantifying underascertainment and provides a means of adjusting estimates of frequency appropriately.[14] Two-source capture–recapture analysis[14–16] was used to assess completeness of ascertainment by active surveillance, as reported in detail elsewhere.[9] An estimate was made of the number of eligible cases not identified by either surveillance scheme, based on the closed population, effectively a birth cohort, of children born in 1995 or 1996 and in whom cataract was diagnosed by 12 months of age.

Method of calculation of ascertainment-corrected number of cases using two-source capture–recapture analysis[15,17]

$$N = [(a + b + 1)(a + c + 1)/(a + 1)] - 1 \qquad (1)$$

where N is total (ascertainment-corrected) number of cases in the population, a is cases reported by both schemes, b is cases reported by the pediatric scheme only, and c is cases reported by the ophthalmic scheme only as follows

		Reported by Ophthalmic Scheme	
		Yes	No
Reported by the pediatric scheme	Yes	a	b
	No	c	d (true unidentified cases)

Method for calculating confidence intervals for ascertainment-adjusted estimate of total number of cases[15,16]

$$\mathrm{Var}(N) = \frac{[(a + b + 1)(a + c + 1)(b)(c)]}{(a + 1)^2(a + 2)} \qquad (2)$$

and 95% confidence intervals $= N \pm 1.96\ [\sqrt{\mathrm{Var}(N)}]$.

This was used to derive rates adjusted for ascertainment for this age group. In the absence of a formal estimate of ascertainment of older children,[9] this adjustment factor was also applied to incidence rates for children older than 1 year. Analyses were undertaken by computer (SAS ver. 6.11; SAS Institute, Cary NC).

The study protocol was approved by the Research Ethics Committee of the Institute of Child Health/Great Ormond Street Hospital and conformed to the Declaration of Helsinki for research involving human subjects.

Results

In 1 year from October 1995, 248 children with newly diagnosed congenital or infantile cataract were notified. Of these, 161 (65%) had bilateral disease, 118 (48%) were girls, and 136 (55%) had isolated cataract—that is, not associated with another ipsilateral ocular anomaly or a systemic disorder. Age at diagnosis was missing in 10 cases, thus complete data for incidence estimation were available for 238 (96%) children. Of these, five children, all with bilateral cataract associated with a systemic disorder, died shortly after diagnosis. Although the median age at diagnosis was 10 weeks, cataract was diagnosed in 70 (30%) children after their first birthday.

The risk (cumulative incidence) of congenital and infantile cataract increased from 2.29 per 10,000 (95% CI, 1.94–2.64) by 1 year of age to 2.93 per 10,000 (95% CI, 2.54–3.31) by 5 years and was 3.19 per 10,000 (95% CI, 2.79–3.59) by 15 years (Table 1). At all ages, the risk of bilateral disease was higher than that of unilateral. There were no sex differences in risk. The highest risk was among children living in England; however, due to the small number of children resident outside England, the 95% CIs for risk estimates by country overlapped.

The annual rate (age-specific incidence) of new diagnosis of congenital and infantile cataract was highest in the first year of life, being 2.29 per 10,000 children (95% CI, 1.94–2.64; Table 2). The rate decreased with increasing age in a similar fashion for bilateral and unilateral disease, among boys and girls, and by country of residence. All estimated rates for children in the oldest age group were based on small numbers, resulting in overlapping 95% CIs.

Capture–recapture analysis indicated that 92% (95% CI, 86–99%) of all children with cataract diagnosed in the first year of life had been ascertained.[9] Based on this, the ascertainment-adjusted incidence of cataract in the first year of life in the United Kingdom was estimated to be 2.49 per 10,000 children (95% CI, 2.10–2.87; Table 3). By the age of 5 years, the adjusted risk was 3.18 per 10,000 (95% CI, 2.76–3.59), increasing to 3.46 per 10,000 (95% CI, 3.02–3.90) by the age of 15

TABLE 1. Risk (Cumulative Incidence) of Congenital and Infantile Cataract in the United Kingdom

	By 1 Year		By 5 Years		By 15 Years	
	Risk* (*n*)†	95% CI	Risk (*n*)	95% CI	Risk (*n*)	95% CI
All (unadjusted)	2.29 (168)	1.94–2.64	2.93 (218)	2.54–3.31	3.19 (238)§	2.79–3.59
By laterality						
Bilateral	1.47 (108)	1.19–1.74	1.90 (141)	1.59–2.21	2.10 (156)	1.77–2.43
Unilateral	0.82 (60)	0.60–1.04	1.04 (77)	0.80–1.28	1.10 (82)	0.86–1.34
By sex						
Female	2.31 (83)	1.81–2.81	2.95 (107)	2.38–3.52	3.12 (113)	2.55–3.69
Male	2.26 (85)	1.79–2.74	2.92 (111)	2.37–3.47	3.28 (125)	2.71–3.85
By country						
England	2.47 (152)	2.08–2.86	3.14 (195)	2.71–3.57	3.39 (211)	2.94–3.84
Scotland	1.64 (10)	0.62–2.66	2.10 (13)	0.97–3.23	2.41 (15)	1.19–3.53
Wales	0.86 (3)	0.0–1.82	1.39 (5)	0.18–2.62	1.39 (5)	0.18–2.62
Northern Ireland	1.25 (3)	0.0–2.66	2.03 (5)	0.25–3.81	2.79 (7)	0.73–4.85

* Per 10,000 children in each age group.
† Number of cases.
§ If 10 cases with missing age at diagnosis are included, then cumulative incidence by age 15 years is 3.33 per 10,000 children.

years, or 3.62 per 10,000 (95% CI, 3.17–4.06) if the 10 children with missing age at diagnosis are included.

DISCUSSION

The findings of this study indicate that currently almost 3 children of every 10,000 born in the United Kingdom each year will have congenital or infantile cataract diagnosed by their first birthday. Two of every three of these children will have bilateral cataract, which has greater visual impact. Although new diagnosis after this age is less common, 1 further child of every 10,000 will have cataract diagnosed by the age of 15 years. Thus, assuming that all children are diagnosed by 15 years, a given child with congenital or infantile cataract in the United Kingdom has a 72% probability of cataract being diagnosed by his or her first birthday, increasing to 92% by the age of 5 years.

In this study, the use of two active surveillance systems to identify incident cases, together with capture–recapture analysis to assess completeness of ascertainment, has allowed reliable estimates of both risk and rate of new diagnosis of congenital and infantile cataract to be determined. These apply to the population of children born in the United Kingdom in the past 15 years, who were subject to both the prevailing etiologic factors and established screening and diagnostic practices[8] for this disorder during that time, which we have assumed to be stable, to estimate cumulative incidence from a cross-sectional study. Given that the method of identification or reporting of congenital disorders can bias estimates of frequency,[18–23] we suggest this approach may be effective in future studies of congenital anomalies in other settings.

Routine notification systems for monitoring congenital anomalies in large populations are well established in Europe and the United States. From these sources, the current annual birth prevalence of congenital or infantile cataract has been estimated to be approximately 1 per 10,000 total births.[1,3] These systems differ from those used in the present study to identify cases, in that they rely on passive reporting procedures, often around the time of birth and from a single source, which are prone to being incomplete, even for major anomalies that are obvious in early life.[18,23] Thus, the disparity between the findings of the present study and these routine data sources is likely to reflect differences in surveillance methods, completeness of ascertainment, and age range included. In contrast, the frequency of congenital cataract reported in previous special studies involving systematic and specific clinical examination of infants to identify congenital ocular anomalies[24] is similar to that found in the present study.

TABLE 2. Rate (Annual Age-Specific Incidence) of New Diagnosis of Congenital and Infantile Cataract in the United Kingdom

	0–1 Years		>1–5 Years		>5–15 Years	
	Rate* (*n*)†	95% CI	Rate (*n*)	95% CI	Rate (*n*)	95% CI
All (unadjusted)	2.29 (168)	1.94–2.64	0.16 (50)	0.10–0.22	0.03 (20)	0.02–0.04
By laterality						
Bilateral	1.47 (108)	1.19–1.74	0.11 (33)	0.07–0.15	0.02 (15)	0.01–0.03
Unilateral	0.82 (60)	0.60–1.04	0.05 (17)	0.03–0.07	0.01 (5)	0.00–0.02
By sex						
Female	2.31 (83)	1.81–2.81	0.16 (24)	0.10–0.28	0.02 (6)	0.01–0.03
Male	2.26 (85)	1.79–2.74	0.16 (26)	0.11–0.22	0.04 (14)	0.02–0.06
By country						
England	2.47 (152)	2.08–2.86	0.17 (43)	0.12–0.22	0.03 (16)	0.01–0.04
Scotland	1.64 (10)	0.62–2.66	0.11 (3)	0.0–0.21	0.03 (2)	0.0–0.06
Wales	0.86 (3)	0.0–1.82	0.13 (2)	0.0–0.31	0	0
Northern Ireland	1.25 (3)	0.0–2.66	0.20 (2)	0.0–0.47	0.08 (2)	0.0–0.18

* Per 10,000 children in each age group per year.
† Number of cases.

TABLE 3. Ascertainment-Adjustment Cumulative and Annual Age-Specific Indicence of New Diagnosis of Congenital and Infantile Cataract in the United Kingdom

	By 1 Year	By 5 Years	By 15 Years
Cumulative incidence	2.49 (2.10–2.87)	3.18 (2.76–3.59)	3.46 (3.02–3.90)*
	0–1 Years	**>1–5 Years**	**>5–15 Years**
Annual age-specific incidence	2.49 (2.10–2.87)	0.17 (0.11–0.24)	0.03 (0.02–0.04)

Data are expressed as incidence per 10,000 children with 95% CI in parentheses.

* If 10 cases with missing age at diagnosis are included, then annual adjusted cumulative incidence by age 15 years is 3.62 per 10,000 children (95% CI 3.17–4.06).

Birth prevalence is the measure of disease frequency most commonly used in monitoring congenital ocular anomalies.[1,3] However, only half of the children in the present study were diagnosed by the age of 10 weeks, with 30% being diagnosed after the first year of life, suggesting that reliance on birth prevalence alone may underestimate total burden of disease in the population. The measures of disease frequency used in the present study, cumulative and annual age-specific incidence, reflect both underlying risk of disease and diagnostic practices and notification procedures.

Cumulative incidence to age 15 years of congenital cataract is an appropriate measure for comparing risk between different populations and, in the longer term, for monitoring secular trends within a population, with a view to identifying emerging causative factors or the impact of primary preventive strategies, such as an immunization program to reduce the risk of prenatally acquired rubella infection. For example, the higher estimates of cumulative incidence reported in longitudinal studies in North America in the 1950s and 1960s[25,26] are likely to reflect the high frequency, at that time, of relevant etiologic factors, including prenatal rubella infection.[27] This has since become a rare cause of congenital and infantile cataract in countries with effective immunization programs.[28] Such detailed longitudinal studies are difficult to perform, and there are no similar recent studies for comparison. Nevertheless there is scope for further research, using methods and case definition comparable to those of the present study, to allow comparison of cumulative incidence of congenital and infantile cataract between and within other countries.

We suggest that annual age-specific incidence of congenital cataract, reflecting patterns of diagnosis, may be useful in assessing secular trends in the performance of the screening program for congenital and infantile cataract within a country, assuming a relatively constant etiologic pattern and ascertainment-adjusted estimates. Similarly, it may be of use in comparing the outcomes of screening strategies between countries with similar health care systems and risk factors for congenital cataract. Furthermore, this measure may be useful for assessing diagnostic practices regarding other congenital conditions that are not always readily recognized at birth, such as congenital sensorineural hearing loss and congenital heart disease.

In the present study, bilateral disease was almost twice as common as unilateral disease at all ages. It is possible that unilateral cataract is less readily diagnosed than bilateral disease. However, investigation of the timing, mode, and context of detection did not reveal any differences between bilateral and unilateral cases in the present study.[29] This suggests that observed differences in incidence by laterality are unlikely to be due to diagnostic patterns. Although there are few sources with which to compare these findings directly, it is probable that bilateral disease is more common than unilateral in the United Kingdom, reflecting differences in relevant etiologic factors. The similar incidence of cataract among boys and girls concurs with current knowledge about hereditary cataract[4,6,30] and with investigations of other congenital ocular anomalies.[24,31,32] Although no clear geographical variations emerged in the present study, small area geographical variations in congenital ocular anomalies have been postulated in relation to potential environmental risk factors.[6,30–33]

Cataract in infancy confers life-long morbidity.[6,30] The estimated 200,000 children blind in the world from congenital cataract represent a burden of 12 million person-years of blindness,[5] and this disorder accounts for approximately 4% of blindness in adults in industrialized countries.[34] Thus prevention of childhood visual impairment due to congenital cataract is a priority of the World Health Organization's new international initiative for the elimination of avoidable blindness by 2020.[7]

The estimates of risk and rate of congenital and infantile cataract in the present study are relevant to implementation and evaluation of preventive strategies. Epidemiologic studies of congenital anomalies, which may be individually uncommon, are challenging but are an important first step toward reducing their considerable combined impact on the health of children.[2] We suggest that sole reliance on birth prevalence should be avoided, whenever possible, in future research on congenital anomalies that are not always readily diagnosed at birth. Consideration should be given to the relative influences on apparent disease frequency of both underlying risk of disease and patterns of diagnosis.

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APPENDIX

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