Ascertainment of congenital malformations: a comparative study of two systems

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Abstract

Study objective—The aim was to compare the Office of Population Censuses and Surveys (OPCS) notification system for congenital malformation surveillance and the Liverpool Congenital Malformations Registry (LCMR) with respect to efficiency and uniformity of ascertainment, diagnostic accuracy, and overreporting of minor malformations.

Design—Manual matching of computer listings was done, using date of birth, sex, birthweight, and health district of residence. Maternal age was used to confirm the match.

Setting—Data were collected in the English health districts of Liverpool, St Helens and Knowsley, Southport and Formby, South Sefton, and Wirral over the years 1980-1985.

Subjects—1959 malformed children notified to OPCS and 2649 notified to LCMR were assessed.

Main results—35.5% of malformed children ascertained by LCMR within seven days of birth were not notified to OPCS; 35.7% of cases reported to OPCS were exclusions from the LCMR protocol as being trivial malformations according to EUROCAT (European Registry of Congenital Anomalies) guidelines. Misclassification was infrequent but in 11.2% of cases the additional malformations present were not notified to OPCS. Conditions readily diagnosed at birth, such as neural tube defects, exomphalos, and facial clefts, were well ascertained by OPCS but others such as oesophageal atresia and Down's syndrome were not.

Conclusions—It is important that national surveillance of congenital malformations should continue. However, several modifications to the present OPCS monitoring system are necessary, including greater standardisation of data collection, the exclusion of trivial and clinically non-significant malformations, and the inclusion of data on therapeutic abortions performed for fetal abnormality. These issues are being addressed by OPCS.

prevalence of malformations detected at birth. Each health district of England and Wales notifies OPCS of any live births where an anomaly has been noted within the first seven days of life and of any stillbirths with anomalies. OPCS regularly informs health districts of any significant increase in prevalences of individual malformations in their district.

In 1984 Knox *et al*¹ published a study in which the quality of notification in the OPCS system was compared with that of the Birmingham Congenital Malformations Register, which is based on multisource ascertainment. Several serious deficiencies in the notifications to OPCS were identified. Some were due to structural errors in the notification system and others to defective implementation. He described four types of error: not all patients with major malformations were notified; some malformations were misclassified; additional malformations were not always ascertained; and there was overnotification of trivial malformations.

To confirm and clarify these deficiencies a similar comparative study was set up between the Liverpool Congenital Malformations Registry (LCMR) and OPCS for the years 1980 to 1985 inclusive. The aims of the study were to determine: (1) the efficiency of the OPCS system in the ascertainment of congenital malformations in the area of study; (2) the diagnostic accuracy of the OPCS system; (3) the extent of the overreporting by districts of trivial malformations to OPCS; and (4) any differences in accuracy and efficiency of notifications to OPCS in the five district health authorities under long term surveillance by the LCMR.

Methods

Ascertainment structures of OPCS and LCMR are shown in table I. LCMR collects data on malformations from numerous sources; its methods have been described elsewhere.² More recently data from Hospital Activities Analysis (HAA), based on hospital discharges and deaths collected by health authorities, have been included as an additional source. For the later years of the study HAA was replaced by the Korner system which is based on hospital admissions and consultant episodes. The basis of ascertainment for most health districts who take part in the OPCS system is the birth notification form completed by the midwife after delivery. Data are collected on babies noted with malformations up to seven days old. Although notification of a birth to the district medical officer by the birth attendant is statutory, this does not apply to the notification of congenital

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The Office of Population Censuses and Surveys (OPCS) notification system for monitoring congenital malformations was introduced in 1964 in the wake of the thalidomide tragedy. It was established in order to provide an early warning system for the detection of any increases in the

malformations. However, on the birth notification forms there is usually a box for information on congenital malformations which is nearly always filled in. The information required by the OPCS monitoring scheme is recorded on an SD56 form which is forwarded to OPCS by the district medical officer. This covers information such as date of birth, district of residence, district of birth, mother's and father's age and occupation, details of the birth, and any malformations present.

For the purposes of this study OPCS provided data on all children notified in the following district health authorities for the period 1980–85 inclusive: Liverpool, St Helens and Knowsley, Southport and Formby, South Sefton, and Wirral. The collection of data in order to complete the SD56 form varies between district health authorities (table II). All five districts use the birth notification forms as a basis for their malformation returns and all but one district rely on details from an additional questionnaire sent out to either the midwife or the health visitor. The remaining district relies solely on a description of the malformation from the birth notification form. Only two district medical officers or their deputies check the validity of the diagnosis on all cases. Copies of the SD56 forms, which are sent to OPCS with details of each malformed baby, have been forwarded to LCMR for the last few years.

Over the study period there were 122 703 live births and stillbirths in the five district health authorities. Information was obtained for each

child on health district of residence, maternal age, infant's date of birth, sex, birthweight, multiplicity of birth, malformation code (up to 8 codes from ICD9), and the OPCS identification number. The main variables used for matching cases were the infant's date of birth, sex, birthweight, and health district of residence. Maternal age was used to confirm the match, following which the data on malformations were compared.

Information regarding therapeutic abortions on malformed babies is collected by LCMR but at the time of the study period was not collected by the OPCS malformations monitoring scheme. Consequently therapeutic abortions have been excluded from the comparison. Complying with EUROCAT guidelines³ LCMR exclude the following non-significant malformations: abnormal palmar creases, congenital hydrocoele; undescended testes, glandular hypospadias; redundant prepuce, phimosis; clicky hips; positional talipes; minor skin anomalies; umbilical hernia; single umbilical artery; minor anomalies of toes, ear, and nose; uncomplicated spina bifida occulta; and all birth injuries. The latter are excluded since while they are sometimes worrying to parents, their definitions are vague and the majority do not require treatment. Conversely, at the time of the study period, OPCS was notified of data on all the above minor as well as the major malformations. In any comparison between LCMR and OPCS this must be taken into account.

Results

Details of the 1959 notifications of malformations to OPCS during the study period 1980–85 are summarised in table III. Only 1185 (60.5%) of notifications to OPCS were also registered by LCMR. However 699 (35.7%) would have been routinely excluded by LCMR according to EUROCAT guidelines. One hundred and forty five cases (7.4%) were considered to have a significant malformation which had not been registered by LCMR. This latter group was further investigated by obtaining addresses from OPCS and tracing names and case note numbers using hospital birth registers. Forty seven infants (32.4% of these "significant" malformations) could not be traced, due, for example, to birth registers or case notes being lost by the hospital of birth. These 47 cases represent 2.4% of the 1959 malformed infants notified to OPCS. The remaining 98 cases were traced and 13 were found to be resident outside the LCMR geographical area, 39 were either normal or had trivial malformations not registerable by LCMR, 16 had been registered by LCMR but not been correctly matched (usually due to incorrect date of birth or district health authority of residence), and 30 cases had validated malformations not previously ascertained by LCMR. The latter 30 cases represent 1.5% of the 1959 babies notified to OPCS and 1.1% of those ascertained by LCMR over the study period. The other 68 successfully traced cases have been redistributed as appropriate in table III.

In tables IV and V the 30 children with malformations notified to OPCS but not

Table I Ascertainment structure of Office of Population Censuses and Surveys (OPCS) and Liverpool Congenital Malformations Register (LCMR)

	OPCS	LCMR
Age at notification	7 days (mostly at birth)	Up to 16 years (mostly <1 year)
Area covered	England and Wales	5 DHAs
Ascertainment	Live and stillbirths	Livebirths, stillbirths, and abortions
Validation	Limited	Multiple sources, including case notes
Continuity	All DHAs different	Registry staff consistent
Exclusions	None	Minor/non-significant anomalies following EUROCAT guidelines

DHA = district health authority

Table II Differences in notification to Office of Population Censuses and Surveys

	District health authority				
	A	B	C	D	E
Birth notification Malformation questionnaires	Yes/no Midwives, Health visitors	Description No	Description Health visitors	Yes/no Health visitors	Yes/no Midwives
SD56 forms	M Q	Birth notification Occasionally SD56 forms	M Q	M Q	M Q
Validity check	No	No	Yes	Yes	No
Copies to LCMR	SD56 forms	SD56 forms	M Q	List	List

M Q = malformation questionnaires; LCMR = Liverpool Congenital Malformations Register

Table III Children notified to Office of Population Censuses and Surveys (OPCS) by district health authority 1980–1985

	District health authority					Total
	A	B	C	D	E	
Percentage notified to both OPCS and LCMR	73.3	37.7	70.7	58.0	63.6	60.5
Percentage notified to OPCS only, with malformations normally excluded by LCMR	22.9	55.6	27.7	37.6	32.8	35.7
Percentage notified to OPCS only, with true malformations	0.0	2.6	1.8	0.7	2.0	1.5
Percentage notified to OPCS only, not traced	3.8	4.0	0.2	3.8	1.6	2.4
Total number of children notified to OPCS	131	302	509	572	445	1959

LCMR = Liverpool Congenital Malformations Register

ascertained by LCMR are presented by year and by district health authority. The largest single group (10 cases) consists of infants with isolated syndactyly or polydactyly, malformations which are often very trivial in nature. A greater concern to LCMR are the seven cases of neural tube defect and four cases of multiple anomalies not originally ascertained. All of these were stillbirths or deaths

Table IV True malformations not ascertained by Liverpool Congenital Malformations Register, by year

Year	Total	Anencephaly/ spina bifida	Syndactyly/ polydactyly	Skull	Multiple	Other
1980	7	4	2			1
1981	7	1	3	1	1	1
1982	3	1	1	1		
1983	4		2			2
1984	2		1	1		
1985	7	1	1		3	2
Total	30	7	10	3	4	6

Table V True malformations not ascertained by Liverpool Congenital Malformations Register, by district health authority

DHA	Total	Anencephaly/ spina bifida	Syndactyly/ polydactyly	Skull	Multiple	Other
A	0					
B	8	1	6			1
C	9	2	2	2		3
D	4	1	1	1		1
E	9	3	1		4	1
Total	30	6	10	3	4	6

Table VI Children ascertained by Liverpool Congenital Malformations Register (LCMR), by district health authority and time of diagnosis, 1980–1985

	District health authority					Total
	A	B	C	D	E	
Percentage notified to both OPCS and LCMR	69.1	39.7	32.8	52.0	58.2	44.7
Percentage notified by LCMR but not notified to OPCS: within seven days of birth	15.1	34.8	47.9	29.4	21.6	35.5
more than seven days from birth	11.5	17.0	14.0	11.9	13.0	13.5
with unknown time of diagnosis	4.3	8.4	5.3	6.7	7.2	6.3
Number of children notified to LCMR (100%)	139	287	1098	639	486	2649

OPCS = Office of Population Censuses and Surveys

Table VII Classification of matched children

	District health authority					Total
	A	B	C	D	E	
Percentage children correctly matched ^a	95.8	83.9	82.2	90.5	82.8	86.0
Percentage misclassified	1.0	5.4	2.8	3.0	2.2	2.8
Percentage where additional malformations not notified to OPCS	3.1	10.7	15.0	6.5	15.0	11.2
Total number of children notified to both OPCS and LCMR	96	114	360	332	283	1185

^a This category includes syndromes and "multiple" ICD codes

OPCS = Office of Population Censuses and Surveys;
LCMR = Liverpool Congenital Malformations Register

Table VIII Specific malformations—All district health authorities, 1980–1985

	Percentage of malformations notified to:			Total (100%)
	Both MCMR and OPCS	OPCS only	LCMR only	
Malformations obvious at birth				
Anencephaly	82.0	12.0	6.0	50
Spina bifida	80.2	4.6 ^a	15.3	131
Facial clefts	72.7	1.8 ^a	25.5	161
Exomphalos	78.9	—	21.0	38
Structural talipes	34.4	46.7 ^b	18.9	488
Malformations not always obvious at birth				
Transposition of great vessels	28.9	—	71.1	38
Oesophageal atresia	46.7	0.3 ^a	50.0	30
Trisomy 21	55.5	0.8	43.7	128
Congenital heart disease	7.8	—	92.2	798
Congenital dislocation of the hip	11.1	27.5 ^b	61.4	153

^a Not all confirmed by perusal of case notes

^b Not traced for confirmation of diagnosis

LCMR = Liverpool Congenital Malformations Register; OPCS = Office of Population Censuses and Surveys

within a few hours of birth. District E had a number of very small maternity units in the earlier years leading to less efficient ascertainment by LCMR than the other district health authorities but this has been improved, partly due to centralisation of their maternity and paediatric services. In the latter part of the study period copies of OPCS returns were forwarded to LCMR by all district health authorities in the surveillance area. Thus the real shortfall of cases ascertained by LCMR was nine (0.3%) for the later years. However 47 (1.7%) could not be traced because of missing birth registers and case notes.

Of the 2649 live births and stillbirths with malformations registered by LCMR during the study period only 1185 (44.7%) were also notified to OPCS (table VI). However as the age limit for notification to OPCS is seven days of life the proportion of children registered by LCMR has been tabulated according to time of diagnosis. Of cases ascertained by LCMR 35.5% were not known to OPCS, but had been diagnosed within seven days of birth. Thus if we only take account of those cases diagnosed within seven days of birth, OPCS matched with 55.8% of the LCMR cases, and were not notified of 42.2% (1040). It can be seen from table VI that there was wide variation between district health authorities with 15.1% not being reported in district A and 47.9% in district C. Many of those ascertained in the latter district had congenital heart defects. There is much less interdistrict variation when the cases were ascertained by LCMR after the age of seven days and hence not within the OPCS notification period. These infants may have significant malformations and are referred to regional centres, in which case access to LCMR is facilitated.

In table VII the 1185 cases registered by both OPCS and LCMR are compared by health district for correctness of matching. The 86% who were correctly matched include infants with malformations coded by OPCS as "syndrome" or "multiple" but coded in detail by LCMR. A system that relies on information collected within seven days of birth will miss late diagnoses and it is therefore not surprising that in 11.2% of the OPCS cases data on additional malformations were lacking. On the other hand, only 2.8% of the OPCS notifications were misclassified.

Efficiency of ascertainment of specific malformations is compared in the two systems in table VIII. These malformations are divided into two groups: (1) those which are obvious at birth and which ought therefore to be ascertained by OPCS; and (2) those which may be difficult to diagnose at birth or which manifest themselves later. OPCS ascertained between 70 and 90% of cases in most of the former group of specific malformations. The notifications of anencephaly to OPCS exceeded the ascertained figures of LCMR. Comparison of rates for talipes (table VIII) is difficult but we suspect that OPCS includes many with positional lesions not recorded by LCMR. Follow up of the excess cases of talipes (46.7%) in order to validate the diagnosis was not possible. However, the 120 cases of talipes ascertained by LCMR over the study period suggest a birth prevalence of

approximately two per 1000 births. The study conducted by Wynne-Davies⁴ in Exeter found the birth prevalence of structural talipes equinovarus to be 1.24 per 1000 births.

The OPCS system failed to receive notifications of a large proportion of the selected malformations in table VIII which are not always obvious at birth. This was particularly true of congenital heart disease, where over 90% of cases were missed, although for transposition of the great vessels, which usually manifests itself in the early days of life, notification was slightly better. OPCS did not receive notifications of nearly half the cases of Down's syndrome. Neither system ascertained all cases of congenital dislocation of the hip, although LCMR was more efficient than OPCS. However, it is not known whether the 27.5% of cases notified to OPCS alone are true cases as it is often impossible to differentiate between congenital dislocation, instability, and clicky hip in the early neonatal period. Additionally the OPCS figures will almost certainly underestimate the prevalence of congenital dislocation as not all cases are identifiable at birth.⁵

Discussion

Our findings generally confirm those of Knox *et al.*,¹ that there is underascertainment of major malformations and overnotification of infants without significant malformations in the OPCS monitoring system. Of those malformations ascertained by LCMR within seven days of birth, 35.5% were not notified to OPCS (table VI), and 35.7% of cases recorded by OPCS were normally excluded by LCMR (table III). Misclassification, however, was uncommon (2.8%—table VII) but in 11.2%, additional malformations were not notified to OPCS (table VII).

When the ascertainment of specific malformations is analysed there are wide variations in the efficiency of the OPCS system. Knox *et al* found good notification rates for facial clefts, oesophageal atresia, exomphalos, and Down's syndrome, but poor rates for neural tube defects. While our study corroborates these findings for facial clefts and exomphalos, notification to OPCS was poor for oesophageal atresia (46.7%—table VIII), which is a condition not always immediately obvious at birth, and for Down's syndrome (55.5%). Conversely, the OPCS register missed few cases of neural tube defects and was more efficient than LCMR at the ascertainment of anencephaly (table VIII).

About one third of cases reported to OPCS were of a clinically insignificant nature and thus excluded by LCMR according to EUROCAT guidelines. These include minor orthopaedic problems such as positional talipes and clicky hip. Although OPCS recorded about twice as many cases of structural talipes as LCMR it is suspected for reasons stated above that this is due to misdiagnosis and overnotification of positional deformations. Other abnormalities found by Knox *et al* to be of doubtful significance included "systolic murmurs" and some malformations of the eye and ear.

An important feature noted during the study was the widely differing methods of data

collection in the five district health authorities (table II). This manifested itself in differences in efficiency of notification. For example, the notification of minor malformations by districts to OPCS varied from 22.9 to 55.6% (table III), and the percentage misclassified varied from 1.0 to 5.4% (table VII). The percentage ascertained by LCMR within seven days of birth but not notified to OPCS varied from 15.1 to 47.9% by district health authority (table VI). In each instance district A, the smallest in the study, was the most efficient.

A small proportion of validated cases was notified to OPCS but not ascertained by LCMR (1.5%—table III). These represent 30 patients, of whom 10 had syndactyly or polydactyly. Although it is of concern that seven cases of neural tube defect were not ascertained by LCMR, all but one of these missed cases were in the earlier years of the study and all were stillbirths or very early neonatal deaths. There is again wide interdistrict variation, with no missed cases in district A, and most underascertainment in district B.

In this study we did not include data on terminations performed after prenatal diagnosis, as the OPCS malformations notification scheme at the time of the study period excluded these. Although previous studies by LCMR^{6,7} showed that prior to 1979 the antenatal screening programme had little effect on the birth prevalence of neural tube defects and Down's syndrome this is no longer the case for the former. Since 1979 the screening programme for neural tube defects has become more effective so that in the period 1980–85 70.7% of anencephalic and 36.2% of spina bifida fetuses were aborted. Consequently previously published OPCS data for malformation prevalence at birth do not give a complete picture of the epidemiology of pregnancy complicated by neural tube defects. Other congenital malformations now diagnosed antenatally leading to a termination of pregnancy have a much smaller numerical effect on the at birth prevalence, but as screening of procedures improves this may change, particularly for a condition such as Down's syndrome.

Several comments are apposite regarding the OPCS monitoring programme. Firstly, there is no question but that monitoring on a national scale should continue, as there are geographical variations in the prevalence of certain anomalies. Also, some malformations are so rare that they require the large numbers collected by a National Register in order to study secular trends. Because of the way it has been set up, the OPCS system is the only birth defects register that is able to give an early warning of an increased frequency of any malformation in England and Wales. Ascertainment, including the validation process of each malformed infant by the LCMR, takes up to two years to be complete for a given annual cohort.

There are, however, several modifications that should be made. Firstly, the methods of data collection should be standardised for all health authorities. Secondly, OPCS should exclude from publication data on minor or insignificant malformations. Finally, methods should be sought of including in published OPCS data

information on terminations of pregnancy following antenatal screening. We note that since the completion of this study some of these issues are now being addressed.

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