Information in practice

Congenital anomaly surveillance in England—ascertainment deficiencies in the national system

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Abstract

Objective Firstly, to assess the completeness of ascertainment in the National Congenital Anomaly System (NCAS), the basis for congenital anomaly surveillance in England and Wales, and its variation by defect, geographical area, and socioeconomic deprivation. Secondly, to assess the impact of the lack of data on pregnancies terminated because of fetal anomaly.

Design Comparison of the NCAS with four local congenital anomaly registers in England.

Setting Four regions in England covering some 109 000 annual births.

Participants Cases of congenital anomalies registered in the NCAS (live births and stillbirths) and independently registered in the four local registers (live births, stillbirths, fetal losses from 20 weeks' gestation, and pregnancies terminated after prenatal diagnosis of fetal anomaly).

Main outcome measure The ratio of cases identified by the national register to those in local registry files, calculated for different specified anomalies, for whole registry areas, and for hospital catchment areas within registry boundaries.

Results Ascertainment by the NCAS (compared with data from local registers, from which terminations of pregnancy were removed) was 40% (34% for chromosomal anomalies and 42% for non-chromosomal anomalies) and varied markedly by defect, by local register, and by hospital catchment area, but not by area deprivation. When terminations of pregnancy were included in the register data, ascertainment by NCAS was 27% (19% for chromosomal anomalies and 31% for non-chromosomal anomalies), and the geographical variation was of a similar magnitude.

Conclusion The surveillance of congenital anomalies in England is currently inadequate because ascertainment to the national register is low and non-uniform and because no data exist on termination of pregnancy resulting from prenatal diagnosis of fetal anomaly.

Introduction

A major congenital anomaly affects 2-3% of newborn babies. Congenital anomalies are an important cause of fetal, neonatal, and child mortality and morbidity, accounting for 21% of perinatal and infant deaths in the United Kingdom in 2001.[1] Monitoring of anomalies is vital to identify possible clusters and trends and to address concerns about putative environmental teratogens. The importance of registering the type and number of congenital anomalies has been recognised for many years; in Birmingham, information on congenital anomalies has been collected since 1949. A national register for England and Wales, now called the National Congenital Anomaly System (NCAS), was proposed by the minister of health in 1963 after the thalidomide "epidemic," and this is run by the Office for National Statistics (ONS; www.statistics.gov.uk).[2]

Notification of anomalies in live and stillbirths to NCAS is voluntary and usually done through a standard paper form (SD56) filled in by midwives, health visitors, and other health professionals. Local congenital anomaly registers have been set up alongside the NCAS, partly to deal with the known under-ascertainment[3–6] and partly to meet local needs and research needs, such as the audit of prenatal diagnosis and research into putative teratogens. Some 50% of births in England are covered by local congenital anomaly registers. These registers are all members of the British Isles Network of Congenital, Anomaly Registers (BINOCAR, www.statistics.gov.uk/binocar/) and belong to EUROCAT (the European Network of Congenital Anomaly Registers, www.eurocat.ulster.ac.uk). In contrast to the NCAS, these local registers record fetuses terminated for fetal anomaly. Ascertainment of cases to the local registers is actively sought and provided from multiple sources, such as cytogenetic and postmortem reports; prenatal diagnosis; and paediatric, neonatal, orthopaedic, and surgical units.

As part of a study of the geographical variation in the prevalence of birth defects[7] we measured the extent to which the under-ascertainment in the NCAS data compared with four local registers, varied by defect, geographical area, and socioeconomic deprivation, during the period 1991-9. We also assessed the impact of the absence of data on pregnancies terminated because of fetal anomaly from the national data set.

Methods

We used data from four local English congenital anomaly registers for comparison with the NCAS. North Thames (West) Congenital Malformation Register (NTW) covers 45 000 births per year; Northern Congenital Abnormality Survey (NorCAS), 33 000 births per year; Wessex Antenatally Diagnosed Congenital Anomalies Register (WANDA), 25 000 births per year; and Oxford Congenital Anomaly Register (OXCAR), 6000 births per year.[8–11] The study period was nine years (1991-9) for all registers except WANDA, which started in 1994 and contributed cases from 1994 to 1999 inclusive. The four local registers use similar methods, with active case finding and multiple sources of ascertainment. Each register collects information on all congenital anomalies occurring in miscarriages after 20 weeks' gestation, in live births and stillbirths, and in fetuses terminated after prenatal diagnosis of anomaly.

Table 1 Congenital anomalies studied in the National Congenital Anomaly System (NCAS) for England and Wales and four local congenital anomaly registers in England, 1991-9. Values are numbers of cases unless otherwise indicated

Anomaly group and subgroups	NCAS	Local registries (terminations excluded)*	NCAS cases as % of local registry cases (terminations excluded)*	All cases in registries (terminations included)*	NCAS cases as % of all registry cases*
All cases	**2483**	**6240**	**40**	**9245**	**27**
Type of anomaly					
All chromosomal anomalies†	555	1641	34	2927	19
Down's syndrome	428	834	51	1496	29
All non-chromosomal anomalies‡	1928	4599	42	6273	31
Some specific non-chromosomal anomalies:					
All neural tube defects	119	176	68	1041	11
Spina bifida	84	112	75	457	18
Cardiac anomalies (excluding ventricular septal defects)	241	1800	13	2050	12
Hypoplastic left heart	19	98	19	181	11
Fallot's tetralogy	26	140	19	151	17
Cleft lip	452	547	83	601	75
Cleft palate	208	292	71	307	68
Digestive system (fistulas and atresias)	188	415	45	471	40
Gastroschisis§	58	132	44	146	40
Exomphalos§	28	58	48	106	26
Diaphragmatic hernia§	29	85	34	123	24
Cystic kidneys	82	299	27	393	21
Limb reduction	217	246	88	296	73

* One of the four local registers provided data for 1994-9 only.
† Includes anomalies coded with the following ICD10 codes: Q90-94, Q96-99.
‡Includes anomalies coded with the following ICD10 codes: Q00-03, Q041-042, Q05, Q110-112, Q160, Q172, Q20, Q211-219, Q22-23, Q25-26, Q300-348, Q36-37, Q35, Q390-394, Q41, Q42, Q600-605, Q61, Q641-643, Q645, Q71-73, Q77, Q78, Q790-793,
§Analysis limited to 1995-9 to achieve coding comparability between NCAS and local register data.

As three of the local registers were not entirely population based, we reduced their populations to those census wards where at least 80% of mothers delivered in hospitals reporting to the register, as calculated from ONS birth data.[7] Cases could be allocated to wards on the basis of their postcode at birth. Average birth coverage in wards was 97%, and only one hospital catchment area (the lowest geographical area considered) had coverage below 90%. We extracted cases reported to NCAS for the same wards and occurring in the same time period covered by the four local registers. Cases were not matched directly because of confidentiality constraints in the use of NCAS data. We therefore compared total numbers of notified cases from the two sources (NCAS and local registers) by condition. We defined hospital catchment areas as the collection of census wards in which most resident mothers delivered in a particular hospital, as calculated by using ONS data on births. We calculated the Carstairs deprivation index[12] for each enumeration district on the basis of the 1991 census.

We selected for study major defects for which the degree of ascertainment is high, agreement on case definition by all registries is good, and ICD-10 lists specific codes (table).

We calculated the ratio of the number of cases in the NCAS data to the number in the local register data, overall and by anomaly type, region, hospital catchment area, and deprivation group dividing at quintiles. We also used a logistic model to adjust the results for deprivation group (dividing at quintiles) by hospital catchment area and region.[7] For these models, the number of NCAS cases was the numerator and the number of local register cases the denominator. We carried out all analyses twice; the first analysis excluded terminations of pregnancy present in local registers and the second included them.

Results

Ascertainment by NCAS was 40% (42% for non-chromosomal anomalies and 34% for chromosomal anomalies) when terminations of pregnancy were excluded from register data. This varied markedly by register, hospital catchment area (not shown), and congenital anomaly subgroup (table, fig 1); all variations were significant ($P<0.001$).

When terminations of pregnancy were included in register data, ascertainment of cases by NCAS (compared with the registers) was 27% (31% for non-chromosomal anomalies and 19% for chromosomal anomalies; table) and again varied markedly by register (fig 1), hospital catchment areas within register areas (fig 2), and congenital anomaly subgroup; all variations were significant ($P<0.001$).

The lowest ascertainment was for neural tube defects (11% when terminations are in the local register data, 68% when

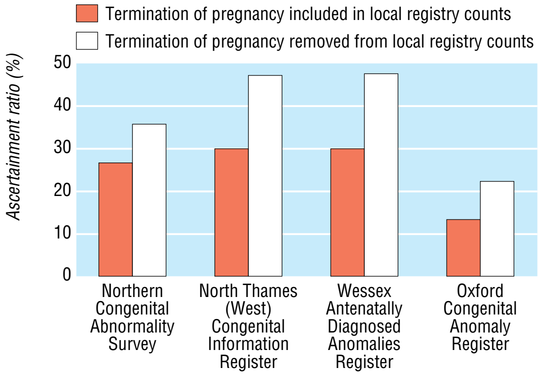

Fig 1 Percentage of all defects in the NCAS compared with local registers, by register

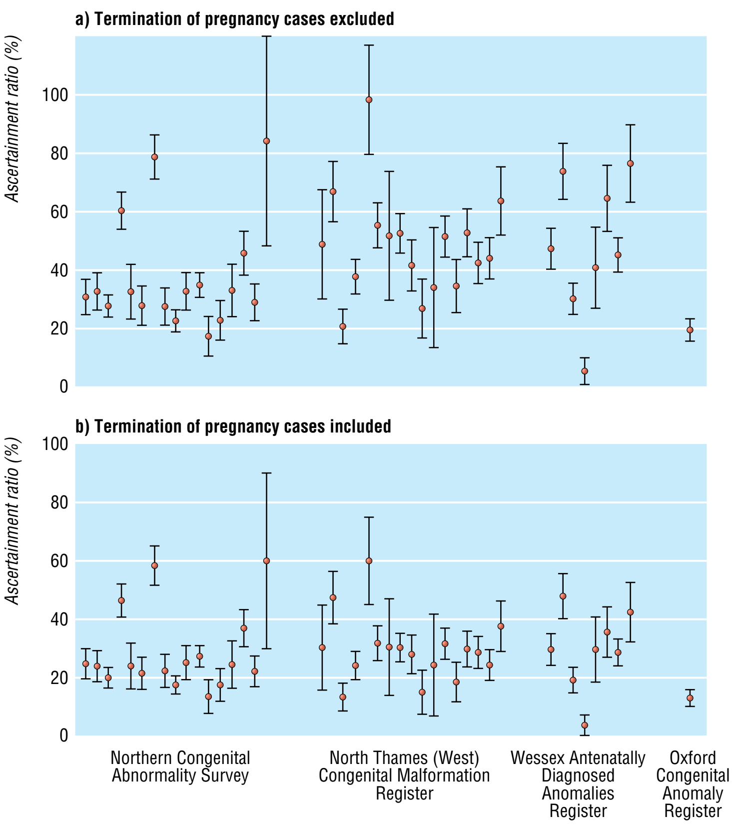

Fig 2 Ascertainment ratios to NCAS by hospital catchment area for all cases. (a) Termination of pregnancy excluded. (b) Termination of pregnancy cases included. Ascertainment ratios are shown for each hospital catchment, with 95% confidence intervals. Catchments with fewer than 10 anomalies identified by either source were omitted, to avoid distracting imprecise ratios

terminations are removed) and cardiac defects (12% with termination included and 13% with termination excluded from local data). The highest ascertainment to NCAS was for cleft lip (75% and 83% with termination included and excluded from local data, respectively) and for limb reduction defects (73% and 88% with terminations included and excluded from local data, respectively).

Figure 1 shows the variation in ascertainment in NCAS across local register areas. The highest ascertainment to NCAS was from the regions covered by NTW and WANDA and the lowest from the OXCAR area. However, ascertainment from the different locations was not consistent when individual defects were compared. For example, ascertainment for cleft lip (overall ascertainment 75%) was highest (91%) from NorCAS area and lowest (44%) from OXCAR. Ascertainment of atresias and fistulas of the digestive system (overall ascertainment 40%) was highest from WANDA and OXCAR areas (57% and 69%) and lowest from NTW and NorCAS (33% and 37%).

The proportion of cases ascertained by NCAS varied little by area deprivation (fig 3), certainly less than could be explained by chance ($P > 0.1$). This pattern did not change on adjustment for differences in ascertainment by registry and hospital catchment area.

Discussion

Surveillance of congenital anomalies in England is currently inadequate. NCAS identified only 40% of the live and stillborn cases it was set up to survey. Moreover, NCAS identified little more than a quarter of all cases including terminations, which are now numerically important in England. Whether terminations were included or excluded, case ascertainment, while always low, varied by anomaly, register, and hospital catchment area. Some hospitals or trusts were clearly giving a higher priority to notification of congenital anomaly than others. Some of the variation between anomalies is explained by the fact that those obvious at birth (such as cleft lip, limb defects) are more likely to be ascertained than "hidden" defects (such as renal anomalies, cardiac defects), which may be diagnosed after mother and child have left the maternity unit. Under-ascertainment by NCAS has long been known to be a problem.[3–6]

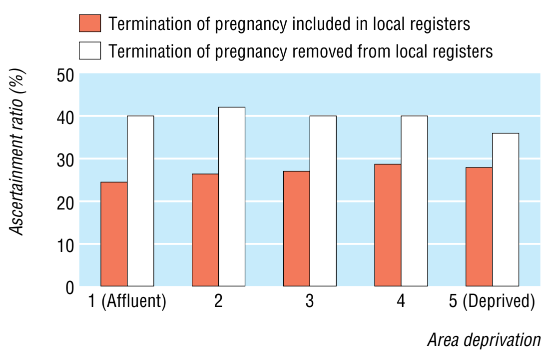

Fig 3 Ascertainment by National Congenital Anomaly System compared with local registers, by area deprivation

Purpose of surveillance and deficiencies of the current system

The original and main purpose of the NCAS is surveillance over time. If levels of under-ascertainment remain constant it is still possible to monitor substantial increases in notifications. However, there is no way of knowing whether an increase in notification is due to improved ascertainment or to a true increase in incidence. Further, for other uses of the data,[13 14] constant ascertainment over time does not ensure against bias due to under-ascertainment.

In an attempt to redress the deficiencies, electronic transmission of data on live births and stillbirths from some registers (Wales and Trent) to the national register was instituted in 1998 and 1999[15] and from others, including those participating in this study, more recently. This will presumably bring the standard of national registration of live births and stillbirths to that of local registries where these exist. However, at present only 50% of births in England are covered by local registers. Moreover, NCAS data before 1999 have been used in epidemiological studies.[13 14 16]

Impact of terminations

Prenatal diagnosis of congenital anomalies by ultrasound examination, cytogenetic testing, or molecular genetic testing has become increasingly available during the past 30 years—that is, since the NCAS was set up. Given that for some anomalies (Down's syndrome, neural tube defects) most pregnancies with affected fetuses in England result in termination of pregnancy,[17 18] the lack of data on termination of pregnancy in NCAS is an important omission. The number of terminations carried out varies geographically, probably because of differences in prenatal screening practice. Data from statutory notifications of terminations are collected by the Department of Health, but these data are relatively inaccessible and have never been validated in terms of their completeness or of the accuracy of malformation coding. A change in the NCAS system to enable data on terminations for fetal anomaly to be recorded on the national register would result in a much more valuable data set.

Impact of poor national data

The poor quality of NCAS data has implications for the interpretation of epidemiological studies seeking to establish risks of congenital anomaly related to residence in relation to environmental pollution sources.[16] Such studies need to be retrospective in order to collect large enough case numbers for analysis.[7] Low ascertainment levels leave a potential for substantial bias if ascertainment is higher or lower near pollution sources. It is reassuring that we could find no ascertainment bias in relation to socioeconomic deprivation. However, given the high level of variation in ascertainment between hospital catchment areas, we recommend that a minimum requirement in using these data is to take this into account in statistical analyses. Communicating results to the public may be difficult when families are aware that their affected child was more likely not to be included in the data than to be included.

Impact of local data

Ascertainment by local registers is not 100%, but, given the active ascertainment of cases from multiple sources, it is not surprising that they have more complete and accurate data than those on the national register. The variation in NCAS ascertainment ratio between registers has a different pattern and is greater than what is known of variation in local register ascertainment.[7] For example, we know that NorCAS has more complete ascertainment of some postnatally diagnosed anomalies than the other three registers. To communicate effectively locally and to ensure high quality of local data and their valid interpretation, local registries cooperate closely with medical specialties such as medical genetics, paediatrics, obstetrics, and pathology, as well as using epidemiological expertise. Therefore a hierarchical system of local data collection, which feeds into a national register (as is the case for cancer registration), should be the most effective model of national surveillance. However, for this system to work it would be necessary for the whole population to be covered by local registers. This does not necessarily mean that all local registers should follow the same model—some may be more research oriented than others, particularly with regard to aetiological factors—but we recommend a basic surveillance dataset.

What is already known on this topic

The National Congenital Anomaly System (NCAS) is the basis for surveillance of congenital anomalies in England and Wales

Ascertainment of cases by NCAS is incomplete

What this study adds

The surveillance of congenital anomalies in England is currently inadequate because ascertainment of affected live and stillbirths by the national register is very low (40%), varying by defect, region, and hospital and because NCAS currently does not include data on terminations of pregnancy after prenatal diagnosis of fetal anomaly

Outlook

If it is important to conduct surveillance of congenital anomalies to look for associations with potential environmental teratogens, to support health service planning, and to monitor prenatal diagnosis and screening programmes then ascertainment of defects at national level must be improved. We support moves to obtain data from local registers, to extend coverage of local registers to the whole country, and to institute an effective national data collection system for terminations of pregnancy.

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