

## The health of children and young people

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### Chapter 11

## Congenital anomalies

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### Table of Contents

	<i>page number</i>
Background and limitations of the National Congenital Anomaly System . . . . .	3
Completeness by condition . . . . .	4
Age restriction for notification . . . . .	4
Voluntary system . . . . .	5
Trends in congenital anomalies in live births and stillbirths 1991 to 2001 . . . . .	5
Abortions for congenital anomalies . . . . .	6
Specific conditions by maternal age . . . . .	7
Specific conditions by birthweight . . . . .	7
The impact of collecting data from local registers . . . . .	8
Ascertainment . . . . .	8
Data being provided by local registers on specific conditions 1991 to 2001 . . . . .	10
Specific conditions, comparison internationally . . . . .	11
New analyses using congenital anomaly data linked to birth records . . . . .	12
Congenital anomalies by country of birth using linked records 1997 to 2000 . . . . .	13
Congenital anomalies by multiplicity . . . . .	14
Survival of babies with congenital anomalies using linked data 1997 to 2000 . . . . .	14
Use of congenital anomaly data in environmental studies . . . . .	16
Conclusions . . . . .	17
References . . . . .	17



## List of Tables

	<i>page number</i>
Table 11.1	Ascertainment by NCAS for selected conditions . . . . . 20
Table 11.2	Notifications of congenital anomalies, 1964–2001. . . . . 21
Table 11.3	Notification rates of anencephalus, other central nervous system anomalies, and legal abortions due to central nervous system anomalies in fetus, 1979–2001 . . . . . 22
Table 11.4	Notification rates of congenital anomalies by condition and maternal age, 1991–2001 . . . . . 24
Table 11.5	Congenital anomalies notification rates from registers participating in electronic data transfer, 1991–2001. . . . . 25
Table 11.6	Observed notification rates for specific anomalies for local registers, 1991–2001 . . . . . 26
Table 11.7	Comparison of notification rates between four local registers in England and Wales, the remainder of England and the International Clearinghouse for Birth Defects Programmes in 1999, 2000 . . . . . 28
Table 11.8	Condition specific notification rates by mother's country of birth, 1997–2000 . . . . . 28
Table 11.9	Notification rates by multiplicity as recorded at birth registration, 1997–2000 . . . . . 29

## List of Figures

Figure 11.1	Congenital anomalies notification rates in live births, England and Wales, 1964–2001 . . . . . 5
Figure 11.2	Congenital anomalies notification rates in stillbirths, England and Wales, 1964–2001 . . . . . 6
Figure 11.3	Congenital anomalies notification rates by birthweight, England, and Wales, 1991–2001 . . . . . 7
Figure 11.4	Congenital anomalies notifications to NCAS from local registers and rest of England, 1991–2001 . . . . . 9
Figure 11.5	Survival to 48 months of babies born with selected conditions. . . . . 15



## Background and limitations of the National Congenital Anomaly System

Congenital anomalies are an important cause of infant and child mortality in England and Wales, and account for an increasing proportion of infant deaths.<sup>1</sup>

The England and Wales Congenital Malformation Notification Scheme (now known as the National Congenital Anomaly System or NCAS) began in 1964. NCAS has a number of limitations, described below. However, other sources of data on congenital anomalies in England and Wales are limited to local or condition-specific registers and surveys; NCAS is the only source of national data. Therefore, this Chapter is based primarily on data from NCAS. These are supplemented by statistical information resulting from the legal requirement to notify terminations of pregnancy.

Since its inception, NCAS has been co-ordinated by the Office for National Statistics (ONS), its predecessors the Office of Population Censuses and Surveys (OPCS) and the General Register Office. The principal source of data on congenital anomalies for Health Authorities (HAs) notifying NCAS was and is the birth notification form, as other sources are rarely used.

Since 1997 some local registers collaborated with NCAS in providing data for their locality. As a result, data for these areas are now more complete, and allow comparisons with other areas covered by a local register. The composite national data, however, should be used with care as they have been gathered by two different reporting systems – anomalies in some areas being notified by their local register, and in others by their local Health Areas.

NCAS was set up in the wake of the epidemic of limb reduction defects following maternal use of thalidomide during pregnancy in the late 1950s and early 1960s.<sup>2,3</sup> Although the epidemic led to a large number of victims world-wide, the increased numbers of babies with obvious limb-reduction defects were not spotted because no routine congenital-anomaly monitoring programmes existed anywhere in the world. Once the epidemic was recognised, less than six months elapsed before the etiological agent was identified and withdrawn from sale. Systems for the surveillance of congenital anomalies were then set up across the world. NCAS's principal purpose was rapid surveillance of localised and national increases in notifications of congenital anomalies.

Information collected by NCAS has been limited to live and stillbirths. Information on anomalies in fetuses aborted spontaneously earlier in pregnancy was not collected. Legal abortion was not introduced in England and Wales until April 1968, following the 1967 Abortion Act, so therapeutic abortions were not included in the original system. Before 1967, the Infant Life (Preservation) Act 1929 made it an offence to deliberately cause the death of a child before it had an existence independent of its mother except where the act which caused the death of the child was done in good faith for the purpose only of preserving the life of the mother. In the ensuing 35 years, ONS and its predecessors have included data about abortions in summary tabulations of congenital anomalies.

Grounds E of the 1967 Abortion Act allows a legal termination of pregnancy if 'there is a substantial risk that if the child were born it would suffer from such physical or mental abnormalities as to be seriously handicapped'. Data on abortions that took place under Grounds E are available in statistical form for complete analysis of congenital anomalies.



In the 1960s, when NCAS was set up, antenatal diagnosis techniques were largely unavailable. Some anomalies that would now be detected antenatally were not diagnosed until birth or later. For the last 25 years at least the limitations of not including abortions in analyses of congenital anomalies have been recognised. Since antenatal diagnosis has had a major impact on the birth prevalence of anomalies, a distorted picture is given if terminations are excluded from analyses of trends in notification rates for anomalies. It is now essential to consider abortion data alongside that for live and stillbirths.

### **Completeness by condition**

Research has shown that completeness of reporting to NCAS varied with the nature and severity of the anomaly, with some severe, life-threatening anomalies being better ascertained.<sup>4-8</sup> Other research has shown that ascertainment was better for children with multiple congenital anomalies than those with a single anomaly.<sup>9</sup> Some studies were conducted nationally<sup>10</sup> and others for a smaller geographical area.<sup>4, 5, 7</sup> Ascertainment also varied for different anomalies depending on whether it could be identified antenatally or postnatally, and whether there was a need for a specific test for diagnosis. If a condition was easily identified at birth then ascertainment would be expected to be high. If the condition was an internal anomaly, not easily visible at birth, more subjective and dependent on clinical judgement or dependent on investigations such as ultrasound, then true cases could have been missed.

Information on the prevalence of under-ascertainment is important for interpreting condition-specific notification rates. Table 11.1 shows the results of five studies that aimed to measure the prevalence of under-ascertainment in anencephaly and spina bifida,<sup>6</sup> anophthalmia and microphthalmia,<sup>11</sup> congenital cataracts,<sup>12</sup> cardiovascular anomalies<sup>13</sup> and Down's syndrome.<sup>14</sup> These conditions were selected as marker conditions, using examples of external major anomalies (anencephaly and spina bifida), external anomalies manifesting in a spectrum of severity (anophthalmia, microphthalmia), major anomalies requiring a specialised clinical diagnosis (congenital cataracts), internal anomalies (cardiac anomalies) and congenital anomalies requiring a test for confirmation (Down's syndrome). From known characteristics of the different conditions it was possible to generalise about ascertainment by NCAS of other similar conditions.

Table 11.1 presents levels of under-ascertainment for each of these conditions.

Easily visible conditions, such as anencephaly tend to be more completely notified to NCAS than those such as Down's syndrome and congenital cataracts, which require specialist confirmation or laboratory tests. Severe manifestations of the condition tend to be better notified than mild cases.

### **Age restriction for notification**

Notifications were at first requested within seven days of birth. It was accepted that some anomalies would not be identified during that period, and that only partial clinical information would be received for other notifications. As diagnostic tools were introduced to detect anomalies earlier in the postnatal period, the age cut-off period was extended to 10 days in January 1990. This allowed NCAS to be notified about cases identified in the immediate postnatal period. But in January 1995 the age cut-off limit was abolished to allow later-diagnosed cases to be notified and to meet the increased research and policy uses of NCAS data.



### Voluntary system

NCAS is a voluntary system whose design was driven by its main purpose of surveillance – to quickly detect any increases in notifications of external, visible anomalies. In the 1960s, it was believed<sup>15</sup> that since clinicians understood the importance of fast notification, statutory powers should not be necessary.

### Trends in congenital anomalies in live births and stillbirths 1991 to 2001

Apart from monitoring changes in the notification of anomalies, and despite the limitations described above, NCAS provides the most extensive data on birth prevalence of congenital anomalies in England and Wales. Table 11.2 shows the number and rate of notifications by live births and stillbirths since 1964, when the system first began. Between 1964 and 1975 the live-birth notification rate remained steady at around 160–180 per 10,000 live births. So, fewer than two per cent of live births were notified as having one or more anomalies. Notification rates increased between 1975 and 1983 reaching 214 per 10,000 live births, then declined to 114 per 10,000 live births in 2001.

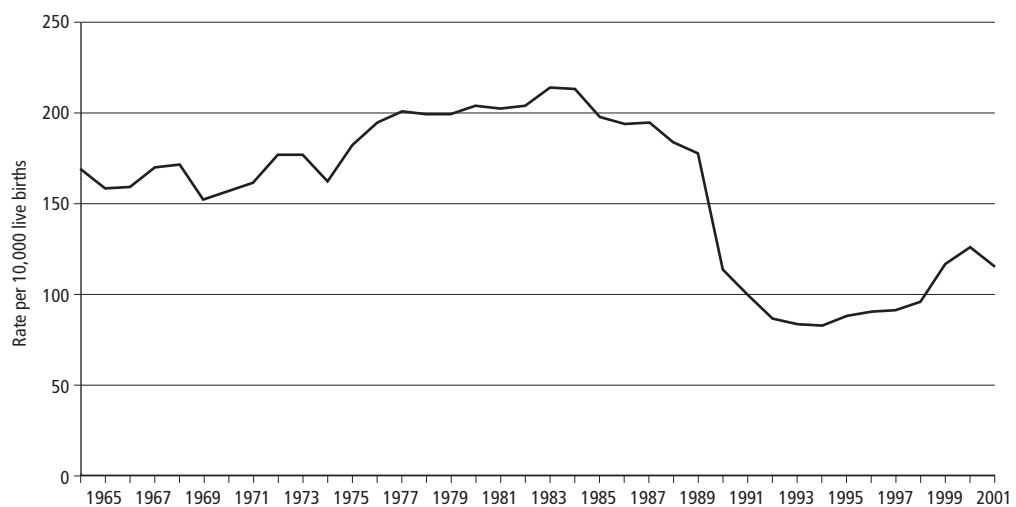
Before 1990 a large proportion of notifications sent to NCAS were for minor anomalies. An exclusion list was introduced in January 1990 to advise notifiers on which conditions should be notified. The aim was to reduce the burden on those notifying and to improve reporting compliance. This list, based on that used by EUROCAT (European Registration Of Congenital Abnormalities and Twins, a European collaboration), gives a number of minor conditions that should not be notified unless they occur in combination with other major anomalies.

As a result of introducing this list, the overall number of congenital anomalies notifications to NCAS fell sharply from 12,391 in 1989 to 8,154 in 1990. The number of notifications continued to fall as notifiers became familiar with the exclusion list, reaching a low of 5,574 in 1994. One reason for the fall in notifications for live births was the fall in notifications of

**Figure 11.1**

#### Congenital anomalies notification rates in live births, 1964–2001

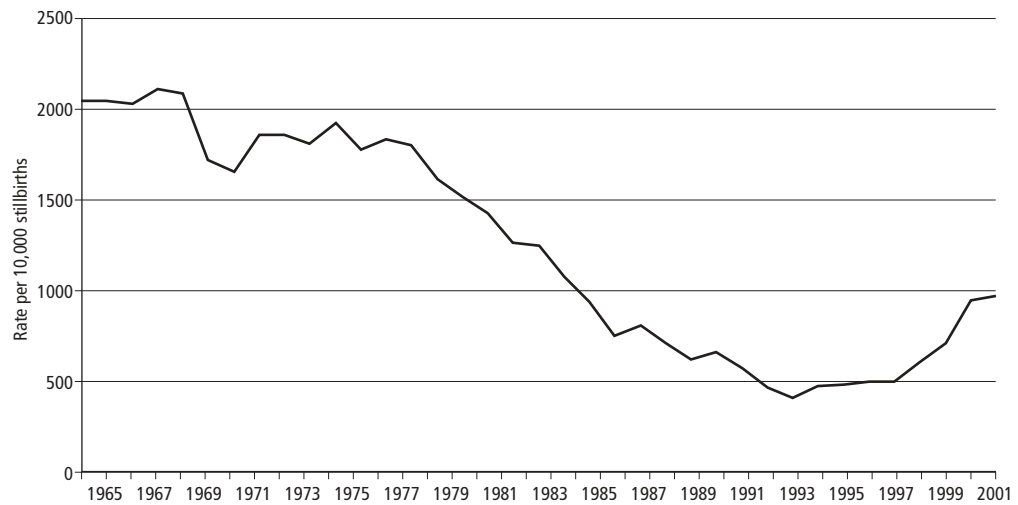
England and Wales



Source: National Congenital Anomaly System. Data at 27 November 2002

**Figure 11.2****Congenital anomalies notification rates in stillbirths, 1964–2001**

England and Wales



Source: National Congenital Anomaly System. Data at 27 November 2002

conditions on the exclusion list. Since the conditions on the list were largely minor and non-lethal, its introduction did not have such a noticeable impact on the number of notifications of stillbirths, which fell from 213 in 1990 to 145 in 1992.

Between 1994 and 2001, the number of notifications increased by 27 per cent to 7,081. This increase was largely due to a small number of local congenital anomaly registers sending their data to NCAs in place of those previously provided by the local health area.

These trends are shown in Figures 11.1 and 11.2.

### Abortions for congenital anomalies

As described above, one limitation of NCAS is the lack of therapeutic abortion data. Termination of pregnancy for fetal handicap was not legally possible when NCAS began in 1964, so abortions were not included. This could be another reason why notifications of congenital anomalies fell in the 1980s and 1990s. As more anomalies were detected antenatally, the parents might have chosen to terminate the pregnancy, resulting in fewer anomalies in live and stillbirths.

Table 11.3 shows the impact of including abortion data in an analysis of central nervous system (CNS) anomalies. The table shows the large fall in the number of CNS anomalies notified to NCAS. Changing patterns in recognising anomalies before birth lead to more antenatal diagnosis, more terminations, and fewer live and stillbirths with the specific congenital anomalies. However, only part of this decrease is the result of the anomalies being detected before birth and the parents choosing to terminate the pregnancy. Although notification of CNS anomalies to NCAS is known to be incomplete, these anomalies are among the better notified to NCAS,<sup>16</sup> and so some of the decrease seen in the table reflects a real decrease in prevalence. This trend has also been seen in other countries.<sup>17</sup>



## Specific conditions by maternal age

This section is based on pooled national data. Although these data have been collected through different notification procedures for different areas, there is no evidence that these different procedures introduce biases by age of mother.

Table 11.4 shows that all notification rates by maternal age group have followed overall trends in congenital anomaly notification rates. Between 1991 and 2001, the highest notification rates were seen among mothers aged 40 years and over, and the second highest rates were among mothers under 20-year-olds. Abdominal wall defects showed the familiar pattern of being highest for the youngest mothers. In contrast, notification rates for Down's syndrome followed the established pattern of increasing with increasing maternal age.

## Specific conditions by birthweight

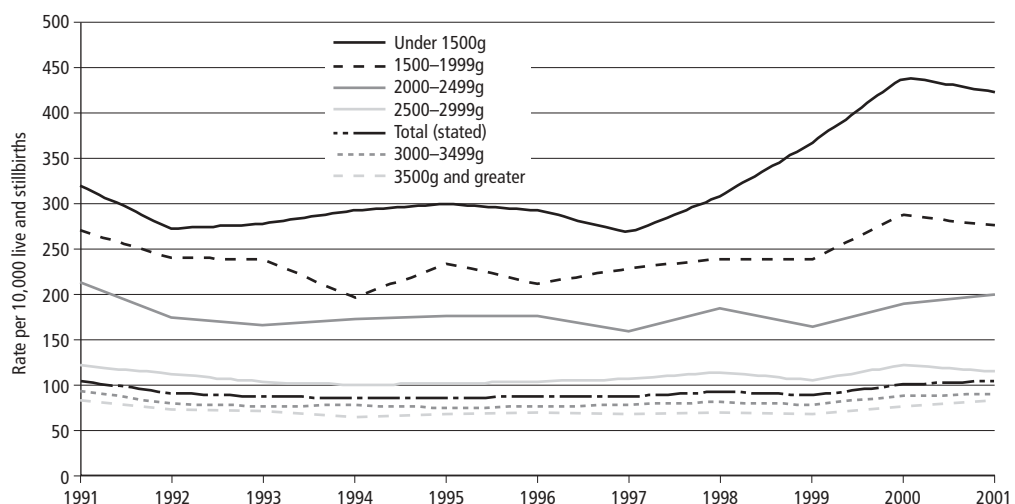
This section is based on pooled national data. These data have been collected through different notification procedures for different areas, which may introduce biases by birthweight.

Figure 11.3 shows that total notification rates by birthweight groups remained constant between 1991 and 1997, with rates being highest for babies with birthweights below 2,000g and lowest among those with birthweights of 3,500g and greater. However, between 1997 and 2001 there were slight increases in notification rates for all birthweight groups, but particularly for babies weighing under 1,500g at birth. The notification rate for these very low birthweight babies increased by 59 per cent from 266 per 10,000 live and stillbirths in 1997 to 423 per 10,000 in 2001. This is likely to be, at least in part, a result of receiving some data from local registers. These registers used more sources to identify babies with anomalies, and were better able to identify babies with other health problems at birth who were more likely to have a longer stay in hospital after birth, require further medical intervention, and in whom the specific anomalies were not notified through the usual birth notification process.

**Figure 11.3**

### Congenital anomalies notification rates by birthweight, 1991–2001

England and Wales



Source: National Congenital Anomaly System. Data at 27 November 2002



## The impact of collecting data from local registers

### Ascertainment

This section describes increases in ascertainment, and how the resulting condition-specific rates differ between different areas. These may be real, or a consequence of different reporting practices and the involvement of local clinical specialists.

Since 1997, some notifications to NCAS have been through ascertainment by local congenital anomaly registers with subsequent transfer of information to NCAS. Previously, all notifications to NCAS had been reported directly from Health Authorities (HAs).

In Britain, following the pioneering example of Birmingham,<sup>5</sup> local registers were created in several locations including Liverpool (now closed), Merseyside and Cheshire, South Wales (now covering all of Wales), Trent, Northern Region, West Midlands and Glasgow.<sup>18</sup>

NCAS was set up in 1964, independently of the local registers in existence at the time. Indeed, an analysis of 1966 NCAS data suggested that the competing interests of local surveys reduced notification to NCAS,<sup>19</sup> as cases notified to a local register were less likely to be notified to NCAS. This may in part have been due to the incorrect assumption by those notifying locally that the local register worked together with NCAS.

Local registers continued to operate in parallel to NCAS until the mid-1990s. One advantage of local registers was that they used multiple sources of information to identify children with congenital anomalies, which improved ascertainment.<sup>20</sup> They also followed-up affected children to verify diagnoses, which improved specificity.<sup>19</sup> Consequently, the local registers held data that were more complete and accurate. As a result, a review of NCAS in 1993<sup>21</sup> recommended that

‘where good congenital malformation registers exist outside OPCS (now ONS), information should be exchanged with these to improve the completeness and validity of both local and national data.’

Initially four registers volunteered to take part in electronic data transfer. In 2001 this combination covered 28 per cent of all births in England and Wales. All four local registers used multiple sources to ascertain cases. All included anomalies detected antenatally as well as postnatally, and collected more data items than were required by NCAS. Three of these registers were based on a geographically defined population, but the fourth register, North Thames (West) was based on the population of several hospitals.

In 1998, Wales began data transfer to NCAS of all congenital anomalies reported in live births or stillbirths, known to them from any source. The East Midlands and South Yorkshire (formerly known as Trent) register began transferring data to NCAS in 1998. Initially they only transferred the information they had received from HAs on the NCAS SD56 notification forms. In 1999, however, they began complete electronic data transfer, based on multiple source ascertainment. The analysis presented here is based on data from East Midlands and South Yorkshire CAR for births in 1999 onwards. Two other registers, North Thames (West) and Mersey, began data transfer of prospectively ascertained congenital anomalies in 2000.

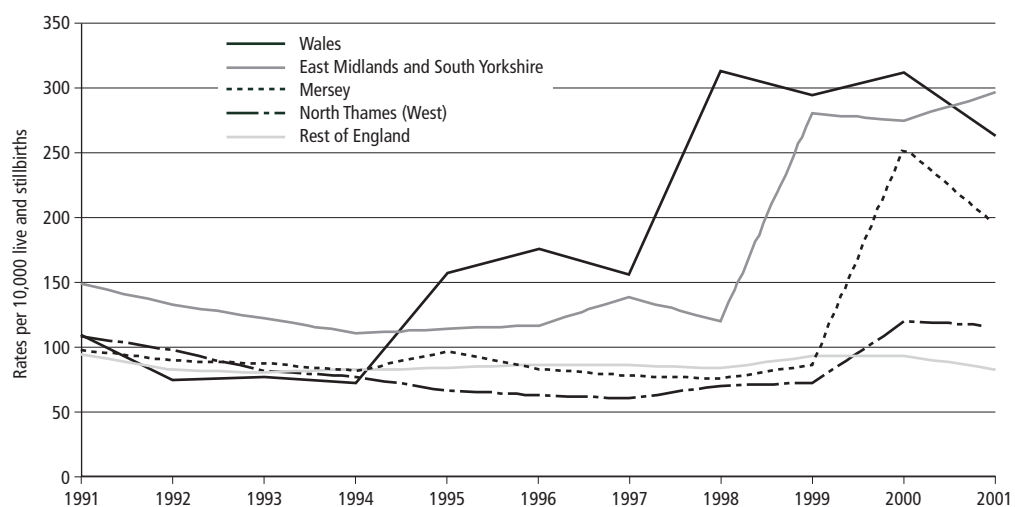


The North Thames (West) register specifically excludes some anomalies, such as polydactyly and hypospadias. Therefore, the notifications to NCAS from North Thames (West) are supplemented by notifications of the additional conditions received by the register from local health areas. The North Thames (West) register covers babies born in the hospitals within their area, rather than being based on residents.

Figure 11.4 and Table 11.5 show the notification rates between 1991 and 2001 for the four local registers, together with the total notification rate for the remainder of England excluding the three English local registers.

**Figure 11.4**

**Congenital anomalies notifications to NCAS from local registers and the rest of England, 1991–2001**



Source: National Congenital Anomaly System. Data at 27 November 2002

Three of the local registers have very similar rates for total anomalies and for some condition groups. This suggests that any differences in reported rates from the areas covered by these three registers before they began notification to NCAS were due to differences in under-notification to NCAS rather than any true difference in birth prevalence. It also shows that in some areas only a quarter of anomalies were being notified to NCAS in the year before the local register assumed responsibility for notification. North Thames (West) rates were lower, less similar and more variable from year to year.

Between 1991 and 1993, before the East Midlands and South Yorkshire and Mersey local registers were in existence, the notification rates for the areas covered by each of the local registers in England were higher than those for the rest of England. Despite East Midlands and South Yorkshire having falling notification rates in the years prior to data exchange, their notification rates were consistently higher than those for the rest of England. The rates for the rest of England were higher than those for North Thames (West) from 1994 to 1999, but substantially lower in 2000 (93 per 10,000 live and stillbirths in rest of England; 120 per 10,000 live and stillbirths in North Thames (West)) when the register began electronic data transfer.



The data in Figure 11.4 and Table 11.5 also show the steep increase in notification rates as data transfer begins. The increase in the NCAS total notification rate after data transfer was two-fold for Wales and East Midlands and South Yorkshire and three-fold for Mersey. Two to three per cent of all children born in these areas were notified as having at least one congenital anomaly following data transfer with NCAS. For Wales and East Midlands and South Yorkshire, there was a lower rate in the second year of data transfer compared with the first. This replicates an analysis by the Wales register, which showed this to be a temporary difference until late notifications had accumulated. The notification rate for North Thames (West) increased by 67 per cent after data transfer, but their notification rate after data transfer was considerably lower than the other local registers. The highest observed rate in the first year of data transfer was for East Midlands and South Yorkshire, which more than doubled between 1998 and 1999, from 119 to 280 per 10,000 live and stillbirths.

### **Data being provided by local registers on specific conditions 1991 to 2001**

Data from local congenital anomaly registers were compared with those for the remainder of England. Comparisons by geographical areas were complicated by the data being notified by two different procedures – some from local registers and some from local Health Areas. Comparisons by geographical area were therefore limited to those areas covered by a local congenital anomaly register where data was thought to be more complete. However, it should be noted that each local register operates a different system of data collection, therefore they are almost certainly not completely comparable. Any differences between areas should be interpreted with this in mind. Data are presented for the remainder of England to give a better understanding of the differences in notification rates due to different practices.

The specific anomalies included were neural tube defects, eye anomalies, cleft lip and palate, heart and circulatory system anomalies, respiratory anomalies, limb reduction defects, abdominal defects and Down's syndrome.

These conditions were analysed in groups according to:

- Whether they should be easily visible at birth (neural tube defects, cleft lip and palate, and limb reduction defects).
- Anomalies that tend to be less well detected at birth and are largely internal (heart and circulatory system anomalies, respiratory anomalies and abdominal defects).
- Those very poorly notified (eye anomalies).
- Those requiring a test to confirm clinical diagnosis (Down's syndrome).

These categories are fairly crude as specific anomalies from one group might be better placed in another group. However, the purpose of these groupings was to determine whether any changes in the notification rate on starting data transfer could be explained by previous levels of ascertainment by NCAS.

All analyses were based on notifications of cases born in the period 1991 to 2001 inclusive. At the time of the study, the most recent complete year of data available for analysis was 2001.



Table 11.6 shows the observed notification rates of specific congenital anomalies for the whole study period. In 2000 the notification rates for the rest of England were the lowest for each condition except eye anomalies. In 2001, the rest of England had the lowest rates for each condition except eye anomalies and limb reductions.

When the local registers began data transfer, the observed notification rates for specific conditions increased and most were significantly higher than those expected from a linear regression on their previous rates.

Neural tube defects, cleft lip and palate, and limb reduction defects are usually obvious at birth. All four local registers saw a significant increase ( $p < 0.01$ ) in notification rates for abdominal defects and for cleft lip and palate compared to the rates expected based on previous notifications for that area received from their local health area. All registers except East Midlands and South Yorkshire experienced a significant increase in notification rate for neural tube defects after data transfer. Mersey and North Thames (West) had the highest rate for neural tube defects. Wales and East Midlands and South Yorkshire had significant increases in notification rates for limb reduction defects following data transfer. In all registers except Mersey, the rates for abdominal wall defects were over five times higher than expected. Wales had the highest notification rates for abdominal anomalies.

Heart and circulatory and respiratory anomalies are largely internal anomalies and may not be diagnosed quickly unless detected antenatally. All four local registers saw a significant increase ( $p < 0.01$ ) in notification rates for heart and circulatory anomalies and abdominal defects, compared to expected rates.

Notification rates from Wales were the highest for both Down's syndrome and eye anomalies. Particularly noticeable were the notification rates for eye anomalies from Wales, which at 16 per 10,000 live and stillbirths in 1998 was four times higher than the next highest rate from the other registers. This is likely to be a result of diligent notification by local clinicians and may include more minor conditions than reported by other registers. However, all registers except Mersey experienced a significant increase in notification rates for eye anomalies on beginning data transfer. The notification rate was double that expected for East Midlands and South Yorkshire, five times that expected by North Thames (West) and over nine times that expected by Wales. Only Wales and Mersey had a significant increase in notification rate for Down's syndrome on starting data transfer, with the notification rate for Wales being four times that expected.

Down's syndrome had already been the subject of an earlier follow-up exercise for notifications in 1997 and 1998. This resulted in a 44 per cent increase in notifications.<sup>22</sup> So it was not surprising that data transfer had not lead to a significantly higher rate for Down's syndrome than expected for two of the three local registers that began data transfer in 1999 and 2000.

### **Specific conditions, comparison internationally**

Some congenital anomalies are so rare as to require large-scale global monitoring to have sufficient numbers to enable analyses. In 1974 NCAS became a founder member of the International Clearinghouse for Birth Defects Monitoring Systems (the 'Clearinghouse'). The Clearinghouse is a non-governmental organisation recognised by the World Health



Organisation. In 2000, the Clearinghouse had 36 programmes participating from 34 countries and collected information on almost three million births annually worldwide.<sup>23</sup>

Congenital anomaly monitoring systems vary; some are nationally based, some cover residents of smaller areas within a country, while others collect data from local hospitals. Some programmes include notifications of anomalies in terminations of pregnancy, whereas in other countries these are not included, or abortion is illegal. So any analyses of international data must be interpreted with care. NCAS contributes statistical summary data to the Clearinghouse quarterly and annually.

While the primary function of the Clearinghouse is to exchange data and detect changes in notification rates, these data are also used for other large scale epidemiological studies and monitoring the implementation of preventative policies.

In 2000, after the four local registers had begun data transfer, notification rates for the registers showed close agreement for cleft lip and palate, abdominal wall defects and Down's syndrome. To test whether a maximum detection level had been reached, notification rates for these conditions were compared with international notification rates, on the assumption that there are no real differences between different countries for the prevalence of these conditions.

Table 11.7 compares a selection of these notification rates with the range of rates for participating programmes in the International Clearinghouse for Birth Defects Monitoring Systems.<sup>23, 24</sup> The chosen conditions were those available from the annual published reports.

The range of rates for Clearinghouse programmes was wide, reflecting the diversity of programmes and their different prevalence and ascertainment rates. The notification rates from the local registers for these conditions fall within the range specified by the International Clearinghouse data. This indicates that either their rates were closer to true prevalence or that some maximum ascertainment had been reached. The rates for the rest of England also fall within the range of international data for these conditions but are all lower than the range for areas covered by a local registers.

## **New analyses using congenital anomaly data linked to birth records**

ONS carried out an evaluation to see if linkage of NCAS records with birth registration records would be feasible; to determine if linkage would improve the quality of NCAS data by reducing missing data; and if it would enable new analyses of information collected at birth registration but not at congenital anomaly notification, and of subsequent survival. Data items collected at birth registration but not by NCAS included mother's and father's country of birth, and the parent's marital status at the child's birth. Consequently, linkage can provide additional co-variables from the birth record for research into factors associated with congenital anomalies. The survival of babies with congenital anomalies was also analysed, through the existing linkage of death registration records to birth records.

The evaluation of linkage between NCAS and birth registration records was based on all NCAS notifications for babies born between 1997 and 2000 using data taken from the



NCAS database in January 2002. The final linkage rate was 97 to 98 per cent for each year.

This study did not include linkage the other way, linking stillbirths and infant deaths due to congenital anomalies to NCAS records. This additional work, however, would supplement these analyses by providing some measure of incomplete notification for lethal malformations.

### **Congenital anomalies by country of birth using linked records 1997 to 2000**

The country of birth of the parents had traditionally been used as a proxy for ethnic origin. However, an increasing proportion of mothers from some minority ethnic groups, particularly Afro-Caribbean, were born in the UK. These crude associations by mother's country of birth can be confounded. Possible confounders include mother's age, parity and consanguinity. This analysis was based on all linked records for England and Wales, pooling data from areas notifying from a local register with those notifying from their Health Areas. Although the data had been notified through two different processes, with different levels of under-notification, there was no evidence that either notification process introduced biases by mother's country of birth. Bias would be introduced if minority ethnic groups were concentrated in the areas covered by local registers, which achieve better ascertainment. Some minority ethnic groups are known to be concentrated in the area covered by the North Thames West register, but other groups are in areas where the local Health Areas notify NCAS. Some caution should therefore be used when interpreting these data.

Countries were included in the analysis if they had 20 or more notifications to NCAS from 1997 to 2000. As a result, it was not possible to divide the group of mothers born in Asia into the specific countries in which they were born.

Total notification rates were highest for mothers born in Asia and lowest for those born in Australia, New Zealand and Canada. The total notification rates for the rest of Europe, Australia, Canada, New Zealand and for the rest of Africa were significantly lower than the notification rates for mothers born in the UK. The rest of Europe also had significantly lower rates for cleft lip and palate, cardiovascular anomalies and urogenital anomalies. Australia, Canada and New Zealand had significant lower rates for CNS and urogenital anomalies. The rest of Africa had significantly lower rates for cleft lip and palate, cardiovascular anomalies, urogenital anomalies and limb reduction defects. Babies with mothers born in the Caribbean had significantly lower rates of urogenital anomalies.

The New Commonwealth and Asia had significantly higher than expected total notification rates. Both had significantly higher rates for CNS anomalies with notifications in Asia being more than double that for mothers born in the UK. Asia also had a significantly higher rate for eye anomalies. They had significantly lower than expected rates for abdominal wall defects, and the New Commonwealth had significantly lower rates for limb reduction defects. Notification rates for Down's syndrome for mothers born in Africa, excluding East Africa, were similar to those for mothers born in the UK. This is comparable with findings in African births.<sup>25</sup>

The rates of some anomalies were higher in babies of Asian-born women. One explanation has been the increased rates of consanguinity in Pakistani marriages resulting in higher rates of recessive inherited disorders.<sup>26</sup> Several studies have reported raised risks of



anomalies in children from consanguineous families. A study of armed-forces families in Pakistan found that the overall prevalence of congenital malformations in children of related parents was 4.0 per cent compared with 2.6 per cent for non-related parents.<sup>27</sup>

### **Congenital anomalies by multiplicity**

Analysing the linked data showed that there was a systematic bias in the NCAS data towards recording more babies as singletons. Fourteen per cent of linked records registered at birth as one of a multiple birth had not been recorded as such at congenital anomaly notification. This is likely to be due to those completing the NCAS notification being unaware that the child was one of a multiple birth, if this information was not present on the notes.

Ninety-four per cent of all NCAS records declared as one of a multiple birth were recorded as being part of a multiple birth at birth registration in the linked data. Other babies recorded as one of a multiple birth by NCAS were recorded as a singleton on birth notification. This could have been a result of linking the wrong record, or that the delivery was at less than 24 weeks' gestation and one or more babies were born dead. Babies born dead at less than 24 weeks' gestation are not legally able to be registered so the delivery would have been recorded at birth registration as a singleton.

In general, anomalies are more common in multiple births, than in singletons, particularly for monozygotic twinning.<sup>28,29</sup> Analyses of NCAS data have shown this excess, but not as great as shown in other studies.<sup>30,31</sup> This means that if the true multiplicity was that recorded at birth registration, then rates of anomalies among multiple births previously published by NCAS were likely to have been underestimated. More marked differences would be expected between the prevalence of congenital anomalies in singleton and multiple births if the multiplicity recorded at birth registration was used.

Using linked data from 1997 to 2000 and based on the multiplicity as given at birth registration, Table 11.9 shows that for all anomalies presented, twins had higher notification rates than singletons.

### **Survival of babies with congenital anomalies using linked data 1997 to 2000**

Linkage also allowed analyses of survival in babies with congenital anomalies. The survival analysis used the death registration information already linked to the birth record. The data used in this analysis included both data notified from local congenital anomaly registers and those notified by local Health Areas. The analysis uses censored data, following babies notified as having an anomaly to death or 48 months of age.

To summarise survival to 48 months of age, Kaplan-Meier survival curves were calculated using the linked NCAS-birth-death data. Survival of these children was followed up to 31 December 2001 or 48 months of age, whichever was sooner. Babies born between 1 January and 30 December 1997 had their data censored at 48 months, whereas for all babies born after 31 December 1997 their data were included up to 31 December 2001. Therefore, babies born in January 2000 would have contributed to the population at risk until age 1 year and 11 months. Similarly those born in January 1999 could have contributed to the population at risk of dying up to and including age 2 years 11 months, those born in January 1998 up to age 3 years 11 months.



Figure 11.5

## Survival to 48 months of babies born with selected condition

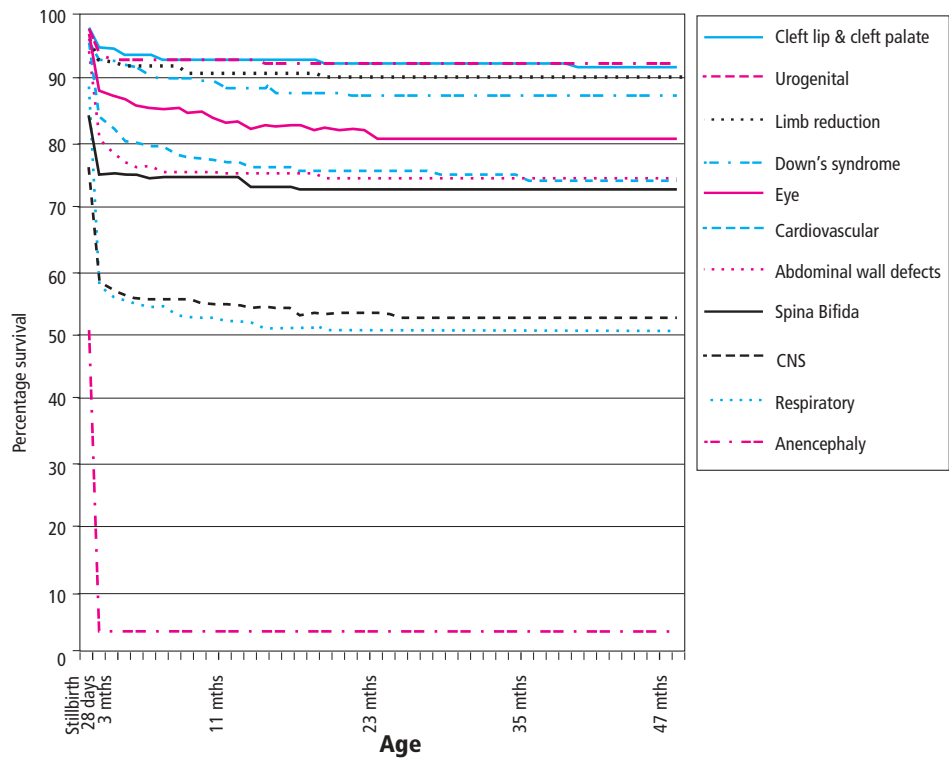


Figure 11.5 shows survival to 48 months according to type of congenital anomaly for the NCAS notifications that were linked to birth records.

The analysis included all babies with the notified anomalies, irrespective of whether they had other anomalies notified. Thus a baby could be included in more than one category.

Just over half of all children born with central nervous system (CNS) defects survived to their fourth birthday, compared with 91 per cent of children with limb defects.

In contrast, 27 per cent of babies with spina bifida died by four years of age, 15 per cent were stillborn, and five per cent died on the first day of life. Eighteen per cent of babies with eye anomalies died by 4 years of age compared with eight per cent of babies with cleft lip and/or cleft palate, 13 per cent of those with Down's syndrome, 25 per cent of those with cardiovascular anomalies and of those with abdominal wall defects, and 49 per cent of those with respiratory anomalies. These anomalies may have occurred with or without other notified anomalies, so the direct cause of death may not have been the anomaly analysed here.

Further analysis showed that at one month of age, 97 per cent of babies notified to NCAS with one anomaly were still alive, compared to 92 per cent of those with two or more notified anomalies. There were more deaths in the group with two or more notified anomalies than would be expected. Therefore, babies with less serious anomalies who die are more likely to have more than one anomaly.

Of the babies born with anencephaly (with or without other notified anomalies) almost 100 per cent were recorded as having died, half of these having been stillborn, 32 per cent



dying on the first day after live birth and the remainder dying during the first week of life. Since anencephaly is incompatible with life, the remaining cases of anencephaly, which the graph suggests were still alive at 48 months of age must be due to an incorrect linkage with a birth record or misclassification of the anomaly at notification. This probably means that the results here underestimate mortality since these problems are likely to affect classification and matching of other records as well.

## Use of congenital anomaly data in environmental studies

Surveillance through NCAS was intended to identify local increases, but because of poor ascertainment, these data could not be used to investigate specific clusters. Instead separate, expensive studies have been required to investigate local concerns resulting from apparent clusters of specific anomalies where the public concern is that toxins from a given location may have a teratogenic effect on the developing fetus. These studies have required good-quality local data to test whether there were higher rates of anomalies in a given area. Recent examples include the possible adverse effects of disinfectant by-products in drinking water, of which trihalomethanes (THMs) were the most common and routinely measured compounds. This has been largely fuelled by studies that have shown an increase risk of NTDs in areas where THMs are used to disinfect water.<sup>32, 33</sup> Another example is the EUROHAZCON study<sup>34</sup> which found that living within 3km of selected European landfill sites was associated with a significantly raised odds ratio for NTDs (odds ratio 1.86; 95 per cent confidence intervals 1.24–2.79).

Within England and Wales, there is a need to examine smaller geographical areas to investigate hypotheses of iatrogenic, environmental or occupational hazards. Testing these hypotheses or investigating identified clusters requires data at a small geographical area. Methodology for geographical epidemiology has become much more sophisticated in recent years. Postcode can be used to place cases within the boundaries of different local areas. Therefore, postcode has been collected by NCAS and held on the computer record since the early 1980s.

The Black Report<sup>35</sup> stated that

‘Encouragement should be given to an organisation such as the Office of Population Censuses and Surveys (now part of ONS) or the Medical Research Council, to co-ordinate centrally the monitoring of small area statistics around major installations producing discharges which might present a carcinogenic or mutagenic hazard to the public.’

Ideally the data required for these purposes would be as complete as possible, requiring good ascertainment not only in live and stillbirths, but also, for many anomalies, in abortions and spontaneous miscarriages. In the past the albeit limited NCAS data has been used to help allay media-driven fears, but better quality and more complete data could reduce the need for separate more intensive studies. To be more sensitive to changes the data needs to be more complete, requiring both more records and more complete data items within records.

For example, between February 1989 and May 1990 four children with transverse limb reduction defects (LRD) were born in the same town on the Isle of Wight. The only



common characteristic identified by the mothers was that they had swum in the sea during pregnancy. This raised the possibility that an environmental factor associated with living near to the coast and swimming in the sea might be implicated for these anomalies. Studies were undertaken in several countries to compare the prevalence of LRD in babies born in coastal areas with those born inland. The results from three different national congenital anomaly registers were published as letters in *The Lancet* on 23 April 1994.<sup>36–38</sup> These reports, from England and Wales, Latin America and Italy all found that there was no difference in the reported rates of LRD for babies born in coastal areas compared with those living inland.

Another example was the local fear that the pesticide Benomyl was associated with anophthalmia following a media report of a small cluster in Lincolnshire.<sup>39</sup> NCAS data showed no trend in the notification of anophthalmia and other related eye defects associated with the introduction of this pesticide, despite a one thousand-fold increase in the use of Benomyl. This strengthens the position of NCAS to investigate such alerts despite its under-ascertainment. A large-scale investigation by the Clearinghouse in other countries also found no association.<sup>40</sup>

## Conclusions

It is essential to have a national surveillance system for monitoring the occurrence of congenital anomalies in the population, and the NCAS serves that purpose. Although dependent on voluntary notifications, it is the largest database of congenital anomalies in England and Wales. Collaboration with local registers in recent years has greatly improved the quality and quantity of data, but more can still be done to encourage their use, and to incorporate data from other local and condition-specific registers where these exist.

The NCAS is currently being reviewed with the aim of enhancing its coverage and completeness to provide a surveillance programme that will generate more valuable information to its users.

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**Table 11.1** Ascertainment by NCAS for selected conditions

Condition	Year	Area		Estimated proportion notified to NCAS
Anophthalmia/ microphthalmia	1988–1994	National	Severe	24
	1988–1994	National	Mild	2
Anencephaly and spina bifida	1968–1990	National		66
	1970–1994	Oxfordshire & West Berkshire		60
Congenital cataracts	1995–1996	National	Diagnosed as neonates	16
	1995–1996	National	Later diagnosis	2
Cardiac anomalies	June 1993 to August 1994	SE Thames & Wessex		5–8
Down's syndrome	1974–1987	National		67
	1990–1993	National		48
	1995–1998	National		39–49

Sources: Hey et al (1994), Busby et al (1998), Rahi et al (1999), Smeeton et al (1999); Huang et al (1997)

**Table 11.2** Notifications of congenital anomalies, 1964–2001

England and Wales

Numbers and rates per 10,000

	Number of congenital anomalies notifications		Rates per 10,000 of total livebirths		Rates per 10,000 of total stillbirths	
	Live	Still	Live	Still	Live	Still
1964	14,729	2,971	168.1		2042.5	
1965	13,610	2,832	157.8		2046.1	
1966	13,479	2,681	158.6		2024.5	
1967	14,112	2,640	169.6		2107.3	
1968	14,011	2,474	171.0		2088.1	
1969	12,072	1,830	151.4		1717.7	
1970	12,246	1,712	156.1		1654.9	
1971	12,567	1,840	160.5		1858.8	
1972	12,782	1,629	176.3		1853.9	
1973	11,920	1,431	176.5		1804.8	
1974	10,315	..	161.3		..	
1975	10,968	1,209	181.9		1922.1	
1976	11,337	1,013	194.2		1777.5	
1977	11,380	990	200.1		1835.0	
1978	11,827	918	198.5		1798.6	
1979	12,667	825	198.7		1612.0	
1980	13,349	723	203.6		1516.4	
1981	12,774	600	201.5		1427.6	
1982	12,732	497	203.6		1262.7	
1983	13,444	452	213.8		1246.9	
1984	13,546	391	212.8		1075.9	
1985	12,949	342	197.4		939.6	
1986	12,758	265	193.1		747.3	
1987	13,237	275	194.3		804.1	
1988	12,723	239	183.5		708.4	
1989	12,191	200	177.4		618.8	
1990	7,941	213	112.5		655.8	
1991	6,889	186	98.6		572.5	
1992	5,909	145	85.7		461.0	
1993	5,565	156	82.7		406.7	
1994	5,394	180	81.2		474.7	
1995	5,657	171	87.3		477.5	
1996	5,784	175	89.1		497.6	
1997	5,756	171	89.7		499.1	
1998	6,035	204	95.0		599.8	
1999	7,179	232	115.5		706.7	
2000	7,575	301	125.4		948.3	
2001	6,776	305	114.0		969.5	

*Source: National Congenital Anomaly System Data as at 27 November 2002*

Note:

.. = Not available.

**Table 11.3 Notification rates of anencephalus, other central nervous system anomalies, and legal abortions due to central nervous system anomalies in fetus, 1979–2001**

England and Wales		Numbers and rates per 10,000 live births, stillbirths and abortions							
	1979	1980	1981	1982	1983	1984	1985	1986	
Anencephalus	455	342	247	162	114	89	59	52	
CNS notifications	1,637	1,476	1,229	1,016	917	806	728	637	
Abortions due to CNS Anomalies	285	418	441	486	511	536	517	556	
Total congenital anomaly and abortions	1,922	1,894	1,670	1,502	1,428	1,342	1,245	1,193	
Total live births and stillbirths	642,608	660,496	638,208	629,367	632,374	640,068	659,712	664,567	
Total Abortions Residents	120,611	128,927	128,581	128,553	127,375	136,388	141,101	147,619	
Total live births, stillbirths and abortions	763,219	789,423	766,789	757,920	759,749	776,456	800,813	812,186	
<b>Rates per 10,000 live births, stillbirths and abortions</b>									
Anencephalus	6.0	4.3	3.2	2.1	1.5	1.1	0.7	0.6	
All CNS notifications	21.4	18.7	16.0	13.4	12.1	10.4	9.1	7.8	
Abortions due to CNS Anomalies	3.7	5.3	5.8	6.4	6.7	6.9	6.5	6.8	
CA and Abortions due to CNS	25.2	24.0	21.8	19.8	18.8	17.3	15.5	14.7	
	1987	1988	1989	1990	1991	1992	1993	1994	
Anencephalus	31	41	34	26	22	32	14	26	
CNS notifications	511	509	426	360	324	316	273	264	
Abortions due to CNS Anomalies	529	464	475	452	492	519	521	465	
Total congenital anomaly and abortions	1,040	973	901	812	816	835	794	729	
Total live births and stillbirths	684,566	696,561	690,608	709,034	702,134	692,452	676,887	668,114	
Total Abortions Residents	156,191	168,298	170,463	173,900	167,376	160,501	157,846	156,539	
Total live births, stillbirths and abortions	840,757	864,859	861,071	882,934	869,510	852,953	834,733	824,653	
<b>Rates per 10,000 live births, stillbirths and abortions</b>									
Anencephalus	0.4	0.5	0.4	0.3	0.3	0.4	0.2	0.3	
All CNS notifications	6.1	5.9	4.9	4.1	3.7	3.7	3.3	3.2	
Abortions due to CNS Anomalies	6.3	5.4	5.5	5.1	5.7	6.1	6.2	5.6	
CA and Abortions due to CNS	12.4	11.3	10.5	9.2	9.4	9.8	9.5	8.8	

continued overleaf

**Table 11.3** Continued

England and Wales		Numbers and rates per 10,000 live births, stillbirths and abortions					
	1995	1996	1997	1998	1999	2000	2001
Anencephalus	35	27	30	22	25	27	27
CNS notifications	257	254	223	287	300	372	332
Abortions due to CNS Anomalies	494	520	416	448	434	460	411
Total congenital anomaly and abortions	751	774	639	735	734	832	743
Total live births, and stillbirths	651,315	652,595	646,148	638,950	624,862	607,304	597,506
Total Abortions Residents	154,315	167,916	170,145	177,871	173,701	175,542	176,364
Total live births, stillbirths and abortions	805,630	820,511	816,293	816,821	798,563	782,846	773,870
<b>Rates per 10,000 live births, stillbirths and abortions</b>							
Anencephalus	0.4	0.3	0.4	0.3	0.3	0.3	0.3
All CNS notifications	3.2	3.1	2.7	3.5	3.8	4.8	4.3
Abortions due to CNS Anomalies	6.1	6.3	5.1	5.5	5.4	5.9	5.3
CA and Abortions due to CNS	9.3	9.4	7.8	9.0	9.2	10.6	9.6

Source: NCAS, ONS Series AB: Abortion Statistics 1979–2001

**Table 11.4** Notification rates of congenital anomalies by condition and maternal age, 1991–2001

England and Wales		Rates per 10,000 live and stillbirths									
	1991	1992	1993	1994	1995	1996	1997	1998	1999	2000	2001
<b>All conditions</b>											
Under 20	112.6	88.4	83.9	95.7	93.6	113.9	104.6	110.0	131.6	148.1	125.1
20–24	103.7	91.3	88.6	85.2	91.4	92.4	93.3	100.8	122.9	134.2	127.9
25–29	94.5	81.3	79.6	79.9	84.9	88.8	85.1	92.3	111.8	116.3	107.7
30–34	93.2	85.5	82.9	78.2	81.9	82.9	82.6	85.1	104.7	118.2	107.7
35–39	118.7	99.0	88.2	88.6	95.7	88.4	98.8	108.7	109.3	122.6	115.0
40 and over	145.2	132.2	126.1	108.8	153.2	141.6	142.7	156.0	162.8	174.7	152.1
Total	101.5	88.0	84.9	84.0	89.8	91.8	92.1	97.9	119.0	130.0	118.8
<b>Central nervous system</b>											
Under 20	6.5	5.2	5.9	5.9	6.4	6.7	4.5	4.5	5.8	8.9	7.6
20–24	5.5	5.3	5.3	3.9	4.9	4.0	3.5	5.2	6.3	7.2	7.7
25–29	4.2	4.2	3.4	3.9	3.4	3.6	3.1	4.0	3.8	5.2	4.5
30–34	3.8	3.8	3.0	3.3	3.0	3.3	3.1	4.0	3.9	5.8	4.6
35–39	3.9	4.9	4.1	3.9	3.9	4.1	4.0	4.9	4.0	4.6	4.7
40 and over	7.1	5.8	7.5	1.8	2.6	4.1	4.6	4.4	4.9	7.3	4.9
Total	4.7	4.6	4.0	4.0	3.9	3.9	3.5	4.5	4.8	6.1	5.6
<b>Neural tube defects</b>											
Under 20	2.8	2.5	4.2	3.1	2.8	2.0	2.4	1.4	2.5	3.3	3.1
20–24	2.7	2.8	2.2	2.1	1.8	2.0	0.9	1.7	2.2	2.4	2.3
25–29	2.2	1.6	1.4	1.3	1.6	1.1	1.2	1.4	1.2	1.7	1.3
30–34	2.0	1.7	1.2	1.3	1.4	1.2	1.4	1.5	1.5	2.1	1.2
35–39	1.7	2.1	2.0	1.3	1.7	1.3	1.3	1.0	1.1	1.3	1.6
40 and over	3.0	1.9	0.9	1.8	0.9	1.6	0.0	1.5	1.4	2.6	1.2
Total	2.3	2.1	1.8	1.6	1.8	1.4	1.3	1.5	1.6	2.0	1.7
<b>Eye</b>											
Under 20	1.5	0.6	0.7	1.7	0.9	2.0	0.9	2.3	4.1	1.5	2.0
20–24	1.2	1.2	0.6	1.3	1.4	1.5	1.8	1.6	2.7	2.6	2.2
25–29	0.8	0.9	1.0	1.0	1.2	1.5	1.4	1.6	1.6	1.4	2.4
30–34	1.2	0.5	1.5	1.9	1.0	1.6	1.3	1.6	2.1	2.2	1.3
35–39	0.9	1.1	0.7	1.6	1.5	0.9	1.6	1.9	1.5	1.9	1.5
40 and over	2.0	1.0	1.9	0.0	2.6	2.5	4.6	3.7	3.5	2.6	1.2
Total	1.1	0.9	1.0	1.4	1.3	1.6	1.5	1.8	2.2	2.0	1.9
<b>Cleft lip and palate</b>											
Under 20	12.3	9.2	7.9	9.9	9.5	9.1	10.9	8.6	10.9	10.2	8.1
20–24	11.0	11.1	11.5	11.1	8.5	10.4	8.4	10.2	10.5	11.4	10.8
25–29	10.5	9.4	9.7	8.3	9.3	9.2	9.2	9.8	9.7	9.0	9.3
30–34	11.3	10.7	8.4	8.9	7.8	7.6	8.2	7.6	8.5	9.1	8.2
35–39	10.6	13.0	9.5	10.4	10.5	9.3	8.4	10.8	7.3	8.4	9.1
40 and over	14.1	17.5	12.2	6.5	14.0	9.8	15.3	8.8	8.3	14.5	9.8
Total	11.2	10.6	9.7	9.4	9.1	9.0	9.0	9.3	9.4	9.8	9.4
<b>Heart and circulatory</b>											
Under 20	6.6	5.8	5.3	7.8	8.3	11.1	10.9	13.8	15.8	22.6	18.4
20–24	6.3	6.6	6.4	7.2	8.9	10.8	9.6	14.5	14.6	20.5	20.4
25–29	6.2	6.1	5.7	6.0	8.9	9.3	9.3	13.4	13.4	16.7	15.7
30–34	7.2	6.7	5.9	5.9	9.1	10.5	7.9	10.3	14.6	18.8	16.6
35–39	11.1	6.1	10.1	7.4	9.4	10.0	12.6	15.3	15.1	24.2	16.2
40 and over	13.1	9.7	10.3	12.9	14.9	16.4	18.4	22.0	22.3	33.6	22.6
Total	7.0	6.4	6.4	6.7	9.2	10.4	10.1	13.5	15.6	22.1	18.4
<b>Respiratory</b>											
Under 20	1.1	0.6	0.7	0.5	0.9	1.1	2.1	1.0	2.5	2.4	3.1
20–24	0.9	0.8	0.5	0.7	1.4	1.4	1.7	1.8	1.3	3.3	2.1
25–29	0.5	0.6	0.5	0.7	1.1	1.4	1.4	1.4	1.6	1.7	1.2
30–34	0.4	0.7	0.6	0.7	0.8	0.6	0.6	0.7	1.1	2.3	2.2
35–39	0.6	0.5	1.2	1.1	1.1	0.7	1.7	1.4	2.2	2.0	2.0
40 and over	3.0	1.9	0.9	0.9	5.3	0.8	0.8	1.5	3.5	1.3	3.1
Total	0.7	0.7	0.6	0.8	1.1	1.1	1.3	1.3	1.8	2.4	2.0
<b>Limb reductions</b>											
Under 20	5.3	3.5	4.2	3.1	3.8	2.7	3.2	5.8	4.1	4.8	3.6
20–24	4.1	2.9	3.5	2.9	3.0	2.6	2.5	2.9	3.7	4.2	3.3
25–29	2.8	2.2	3.2	3.2	2.7	3.3	1.9	2.8	4.3	3.6	3.0
30–34	3.2	2.9	2.6	2.7	3.2	2.9	1.9	2.7	2.1	2.8	2.9
35–39	1.9	2.1	3.0	2.5	2.3	3.0	2.0	3.3	2.6	2.7	3.2
40 and over	4.0	5.8	2.8	3.7	2.6	5.7	2.3	2.9	3.5	3.3	3.1
Total	3.4	2.7	3.2	2.9	3.0	3.1	2.1	3.1	3.4	3.4	3.1
<b>Abdominal wall defects</b>											
Under 20	7.6	7.3	7.9	6.1	8.3	9.6	6.9	9.1	11.5	12.1	9.4
20–24	4.0	3.5	3.7	3.4	4.1	3.6	3.3	4.3	5.2	5.0	4.9
25–29	1.4	2.0	1.7	1.6	1.1	1.5	1.8	2.1	2.4	2.3	2.5
30–34	1.4	1.3	1.9	1.5	1.4	0.9	1.6	1.0	1.5	1.8	1.8
35–39	1.1	1.9	1.0	1.1	1.7	1.6	1.2	0.6	2.1	2.7	2.0
40 and over	5.0	1.0	2.8	0.0	5.3	3.3	1.5	3.7	2.8	2.6	3.7
Total	2.6	2.5	2.6	2.2	2.4	2.3	2.4	2.6	3.3	3.5	3.3
<b>Down's Syndrome</b>											
Under 20	2.7	2.9	2.4	3.5	2.8	4.7	2.8	3.5	4.3	4.8	3.6
20–24	4.6	3.4	3.4	2.8	2.8	4.0	3.9	3.5	3.5	3.5	3.4
25–29	4.0	4.0	3.4	3.5	2.8	3.6	3.4	3.9	3.9	4.3	2.4
30–34	7.2	6.0	4.9	5.1	5.1	4.9	6.1	6.7	6.3	5.7	5.9
35–39	16.7	15.6	7.9	9.9	10.6	9.7	14.1	15.9	12.7	11.5	12.8
40 and over	35.3	33.1	30.1	21.2	37.6	40.9	41.4	43.9	39.7	41.5	37.9
Total	6.3	5.7	4.6	4.7	4.9	5.5	6.5	7.2	6.8	6.7	6.3

Source: National Congenital Anomaly System. Data at 27 November 2002

**Table 11.5** Congenital anomalies notification rates from registers participating in electronic data transfer, 1991–2001

Rates per 10,000 live and stillbirths

Register	1991	1992	1993	1994	1995	1996	1997	1998	1999	2000	2001
Wales	109.5	74.3	77.2	71.7	157.3	175.7	155.3	<b>312.6</b>	<b>294.1</b>	<b>311.9</b>	<b>263.2</b>
East Midlands and South											
Yorkshire	148.5	132.6	122.6	111.0	114.0	116.5	138.2	119.3	<b>280.0</b>	<b>274.0</b>	<b>297.0</b>
Mersey	97.9	89.3	86.7	80.9	96.9	82.7	78.3	76.0	86.6	<b>253.1</b>	<b>194.0</b>
North Thames											
(West)	108.5	97.5	81.5	76.9	66.3	62.5	61.0	69.4	72.1	<b>120.3</b>	<b>115.7</b>
Rest of England	94.1	82.0	80.8	82.2	83.7	86.0	85.7	84.1	93.5	93.1	82.0

Source: National Congenital Anomaly System. Data at 27 November 2002

Note:

Rates in bold are those after electronic data transfer.

**Table 11.6** Observed notification rates for specific anomalies for local registers, 1991–2001  
Boundaries are those at 1 April 2001

Rates per 10,000 live and stillbirths

		1991	1992	1993	1994	1995	1996
<b>All Babies</b>	Wales	109.5	74.3	77.2	71.7	157.3	175.7
	East Midlands and South Yorkshire	148.5	132.6	122.6	111.0	114.0	116.5
	Mersey	97.9	89.3	86.7	80.9	96.9	82.7
	North Thames (West)	108.5	97.5	81.5	76.9	66.3	62.5
	Rest of England	94.1	82.0	80.8	82.2	83.7	86.0
<b>Central nervous system</b> ICD-9 740.0–742.9, 320.0–359.9 ICD-10 Q00.0–Q07.9, selection of G codes	Wales	5.8	3.7	2.7	3.1	4.9	4.8
	East Midlands and South Yorkshire	5.3	5.5	4.7	5.9	4.6	4.7
	Mersey	5.4	6.3	4.6	3.3	3.1	2.1
	North Thames (West)	5.8	5.7	4.8	4.1	3.6	3.8
	Rest of England	4.4	4.3	3.9	3.8	3.9	3.8
<b>Neural tube defects</b> ICD-9 740.0–742.0, 756.1 ICD-10 Q00.0–Q01.9, Q05.0–Q05.9, Q76.0	Wales	2.9	2.9	1.6	1.4	0.9	1.7
	East Midlands and South Yorkshire	2.2	1.8	1.7	2.2	1.9	1.5
	Mersey	3.0	2.5	2.0	0.7	1.0	0.7
	North Thames (West)	1.5	2.1	1.8	1.0	2.4	0.8
	Rest of England	2.3	2.0	1.8	1.6	1.8	1.5
<b>Eye</b> ICD-9 743.0–743.9, 360.0–379.9 ICD-10 Q10.0–Q15.9, selection of H codes	Wales	1.0	0.8	0.8	1.4	10.1	7.1
	East Midlands and South Yorkshire	1.9	1.3	1.1	2.2	1.4	2.3
	Mersey	0.9	0.3	0.7	0.3	0.7	1.0
	North Thames (West)	0.2	0.4	0.8	0.8	0.4	0.4
	Rest of England	1.1	0.9	1.1	1.4	0.7	1.2
<b>Cleft lip and palate</b> ICD9 749.0–749.2 ICD10 Q35.0–Q37.9	Wales	14.1	9.8	9.2	11.0	15.0	14.0
	East Midlands and South Yorkshire	12.6	13.2	12.9	9.6	11.2	9.1
	Mersey	9.9	11.3	11.1	7.4	8.7	5.9
	North Thames (West)	9.9	11.7	10.1	7.5	6.5	7.6
	Rest of England	11.0	10.1	9.2	9.6	8.7	9.0
<b>Heart and circulatory</b> ICD-9 745.0–747.9, 390.0–459.9 ICD-10 Q20.0–Q28.9, selection of I and M codes	Wales	9.1	9.0	8.7	10.4	48.5	58.7
	East Midlands and South Yorkshire	11.8	10.8	11.0	10.4	9.8	14.0
	Mersey	5.7	7.5	6.8	4.3	3.5	4.8
	North Thames (West)	6.2	5.2	6.0	7.7	3.0	5.6
	Rest of England	6.4	5.7	5.6	5.9	7.3	7.3
<b>Respiratory</b> ICD-9 748.0–748.9, 460.0–519.9 ICD-10 Q30.0–Q34.9	Wales	0.5	0.8	0.5	1.1	3.2	3.4
	East Midlands and South Yorkshire	0.6	1.2	1.4	0.3	1.4	3.1
	Mersey	0.9	0.6	0.3	0.7	0.3	1.0
	North Thames (West)	0.6	1.0	0.2	1.2	0.8	0.4
	Rest of England	0.7	0.6	0.5	0.8	1.0	0.7
<b>Limb reductions</b> ICD-9 755.2–755.4 ICD-10 Q71.0–Q73.8	Wales	2.6	3.2	1.1	2.0	3.2	2.9
	East Midlands and South Yorkshire	4.1	3.3	4.7	3.9	3.8	3.4
	Mersey	2.1	0.9	3.3	2.7	3.1	3.1
	North Thames (West)	2.1	2.7	3.4	2.2	2.8	2.0
	Rest of England	3.6	2.7	3.1	3.0	2.8	3.2
<b>Abdominal anomalies</b> ICD-9 756.7, 550.0–553.9 ICD-10 Q79.2–Q79.5, K40.0–K46.9	Wales	2.9	2.1	3.3	1.1	4.0	2.6
	East Midlands and South Yorkshire	3.7	3.7	4.4	2.3	2.9	1.1
	Mersey	3.9	3.1	3.3	2.3	2.1	3.1
	North Thames (West)	2.1	3.1	2.4	1.2	2.0	2.6
	Rest of England	2.4	2.3	2.3	2.4	2.3	2.4
<b>Down's Syndrome</b> ICD-9 758.0 ICD-10 Q90.0–Q90.9	Wales	5.8	6.1	7.3	8.2	8.4	8.0
	East Midlands and South Yorkshire	8.0	7.1	5.2	5.1	4.0	5.4
	Mersey	7.8	5.0	4.2	2.3	4.5	5.9
	North Thames (West)	8.5	7.7	4.4	3.9	6.5	4.6
	Rest of England	5.7	5.3	4.4	4.7	4.6	5.4

Source: National Congenital Anomaly System. Data at 27 November 2002.

Continued overleaf

Note:

Rates in bold are those after electronic data transfer.

Table 11.6 Continued

		Rates per 10,000 live and stillbirths				
		1997	1998	1999	2000	2001
<b>All Babies</b>	Wales	155.3	<b>312.6</b>	<b>294.1</b>	<b>311.9</b>	<b>263.2</b>
	East Midlands and South Yorkshire	138.2	119.3	<b>280.0</b>	<b>274.0</b>	<b>297.0</b>
	Mersey	78.3	76.0	86.6	<b>253.1</b>	<b>194.0</b>
	North Thames (West)	61.0	69.4	72.1	<b>120.3</b>	<b>115.7</b>
	Rest of England	85.7	84.1	93.5	93.1	82.0
<b>Central nervous system</b> ICD-9 740.0–742.9, 320.0–359.9 ICD10 Q00.0–Q07.9, selection of G codes	Wales	6.3	<b>13.1</b>	<b>14.6</b>	<b>9.5</b>	<b>10.4</b>
	East Midlands and South Yorkshire	5.3	5.5	<b>12.5</b>	<b>12.2</b>	<b>13.4</b>
	Mersey	3.9	4.0	1.9	<b>10.0</b>	<b>9.6</b>
	North Thames (West)	2.8	3.2	2.8	<b>11.2</b>	<b>8.9</b>
	Rest of England	3.0	3.9	3.5	4.3	3.6
<b>Neural tube defects</b> ICD-9 740.0–742.0, 756.1 ICD-10 Q00.0–Q01.9, Q05.0–Q05.9, Q76.0	Wales	0.9	<b>1.5</b>	<b>3.1</b>	<b>1.9</b>	<b>3.2</b>
	East Midlands and South Yorkshire	2.5	1.5	<b>2.4</b>	<b>2.3</b>	<b>2.2</b>
	Mersey	1.4	1.8	0.8	<b>3.9</b>	<b>2.8</b>
	North Thames (West)	0.8	1.4	0.8	<b>3.9</b>	<b>2.1</b>
	Rest of England	1.2	1.5	1.5	1.7	1.3
<b>Eye</b> ICD-9 743.0–743.9, 360.0–379.9 ICD-10 Q10.0–Q15.9, selection of H codes	Wales	9.5	<b>15.5</b>	<b>15.8</b>	<b>14.0</b>	<b>11.0</b>
	East Midlands and South Yorkshire	2.0	1.8	<b>4.4</b>	<b>4.1</b>	<b>2.9</b>
	Mersey	1.8	0.4	1.1	<b>0.4</b>	<b>3.2</b>
	North Thames (West)	0.4	0.8	0.4	<b>2.0</b>	<b>1.1</b>
	Rest of England	1.0	0.9	1.3	0.9	1.2
<b>Cleft lip and palate</b> ICD9 749.0–749.2 ICD10 Q35.0–Q37.9	Wales	8.1	<b>17.0</b>	<b>13.6</b>	<b>10.8</b>	<b>14.9</b>
	East Midlands and South Yorkshire	9.5	8.4	<b>15.5</b>	<b>14.2</b>	<b>16.4</b>
	Mersey	11.1	6.2	6.4	<b>10.4</b>	<b>16.3</b>
	North Thames (West)	5.8	6.8	8.5	<b>12.0</b>	<b>10.1</b>
	Rest of England	9.2	9.3	8.6	8.8	7.6
<b>Heart and circulatory</b> ICD-9 745.0–747.9, 390.0–459.9 ICD-10 Q20.0–Q28.9, selection of I and M codes	Wales	41.5	<b>103.8</b>	<b>85.2</b>	<b>98.6</b>	<b>90.7</b>
	East Midlands and South Yorkshire	18.0	17.8	<b>43.4</b>	<b>45.7</b>	<b>46.0</b>
	Mersey	2.5	4.4	7.5	<b>85.4</b>	<b>29.9</b>
	North Thames (West)	6.0	7.0	7.9	<b>26.1</b>	<b>20.6</b>
	Rest of England	7.7	7.7	8.5	9.7	8.7
<b>Respiratory</b> ICD-9 748.0–748.9, 460.0–519.9 ICD-10 Q30.0–Q34.9	Wales	2.9	<b>8.9</b>	<b>12.1</b>	<b>8.9</b>	<b>11.4</b>
	East Midlands and South Yorkshire	3.2	1.2	<b>4.4</b>	<b>6.1</b>	<b>3.9</b>
	Mersey	1.1	1.1	1.5	<b>2.3</b>	<b>2.8</b>
	North Thames (West)	1.0	0.4	0.4	<b>4.3</b>	<b>3.7</b>
	Rest of England	1.0	0.8	1.0	1.2	0.9
<b>Limb reductions</b> ICD-9 755.2–755.4 ICD-10 Q71.0–Q73.8	Wales	2.3	<b>5.4</b>	<b>7.1</b>	<b>6.7</b>	<b>4.5</b>
	East Midlands and South Yorkshire	2.8	2.7	<b>5.6</b>	<b>5.9</b>	<b>6.6</b>
	Mersey	2.5	2.9	4.1	<b>3.1</b>	<b>2.4</b>
	North Thames (West)	1.0	2.8	3.2	<b>3.5</b>	<b>4.8</b>
	Rest of England	2.1	3.1	2.8	2.9	2.4
<b>Abdominal anomalies</b> ICD-9 756.7, 550.0–553.9 ICD-10 Q79.2–Q79.5, K40.0–K46.9	Wales	2.6	<b>9.2</b>	<b>9.0</b>	<b>7.6</b>	<b>9.7</b>
	East Midlands and South Yorkshire	2.2	2.0	<b>7.8</b>	<b>5.4</b>	<b>7.6</b>
	Mersey	3.9	2.9	1.9	<b>7.3</b>	<b>5.6</b>
	North Thames (West)	1.8	1.6	1.4	<b>5.7</b>	<b>3.7</b>
	Rest of England	2.4	2.3	2.6	2.6	2.1
<b>Down's Syndrome</b> ICD-9 758.0 ICD-10 Q90.0–Q90.9	Wales	8.9	<b>11.0</b>	<b>11.8</b>	<b>9.5</b>	<b>9.4</b>
	East Midlands and South Yorkshire	3.0	7.4	<b>9.9</b>	<b>11.2</b>	<b>9.8</b>
	Mersey	4.3	5.5	5.6	<b>9.7</b>	<b>8.4</b>
	North Thames (West)	4.6	11.0	13.9	<b>9.8</b>	<b>12.2</b>
	Rest of England	7.0	6.6	5.3	5.4	4.7

Source: National Congenital Anomaly System. Data at 27 November 2002.

Note:

Rates in bold are those after electronic data transfer.

**Table 11.7** Comparison of notification rates between the four local registers in England and Wales, the remainder of England and the International Clearinghouse for Birth Defects Programmes in 1999, 2000

Rates per 10,000 live and stillbirths

	Condition (based on ICD codes as shown in Table 11.5)		Range of rates from the four local registers in England and Wales		Rate for the remainder of England Range of rates from the International Clearinghouse Programmes	
	1999	2000	1999	2000	1999	2000
All Babies	72.1–294.1	120.3–311.9	93.5	93.1	..	..
Cleft lip and palate	6.4–15.5	10.4–14.2	8.6	8.8	6.1–39.18	4.13–28.94
Abdominal defects	1.4–9.0	5.4–7.6	2.6	2.6	0.18–7.54	0.18–9.38
Down's Syndrome	5.6–13.9	9.5–11.2	5.3	5.4	4.34–25.35	2.99–28.13

Sources: National Congenital Anomaly System, International Clearinghouse for Birth Defects Annual Report 2001 (with data for 1999), International Clearinghouse for Birth Defects Annual Report 2002 (with data for 2000).

Note:

.. = Not available

**Table 11.8** Condition-specific notification rates by mothers country of birth, 1997–2000

Rates per 10,000 live and stillbirths

	Central nervous system	Eye	Cleft lip and palate	Cardio-vascular	Respiratory
United Kingdom	4.3	1.7	9.4	16.2	1.5
Rest of Europe	4.5	0.9	6.3**	10.1**	1.1
Australia, Canada and New Zealand	0.0**	0.7	5	14.4	0.7
New Commonwealth	8.0*	2	9.4	16.1	1.7
Asia	10.0*	2.7*	11.2	18.5	2.2
East Africa	6.9	0.6	7.4	12	1.1
Rest of Africa	5.4	1.6	6.0**	11.1**	0.9
Caribbean	2.9	1	6.7	11.4	1.9
Total	4.6	1.7	9.2	15.8	1.5

	Urogenital	Limb reduction defects	Abdominal wall	Down's Syndrome	Total
United Kingdom	22	3	3.9	6.4	131.1
Rest of Europe	16.9**	3	3.2	6.3	97.1**
Australia, Canada and New Zealand	13.6**	2.9	5	4.3	89.0**
New Commonwealth	20.2	2.0**	2.6**	6.6	139.2*
Asia	22.1	2.7	2.6**	6.4	157.0*
East Africa	24.6	0.6	1.7	7.4	125.4
Rest of Africa	13.9**	0.9**	2.8	6.6	101.9**
Caribbean	9.5**	1.9	6.7	9.5	135.3
Total	21.5	2.9	3.7	6.4	129.3

Sources: ONS unpublished linked data.

Table excludes countries with fewer than 20 notifications in 1997–2000.

Notes:

\* Significantly higher.

\*\* Significantly lower.

**Table 11.9** Notification rates by multiplicity as recorded at birth registration, 1997–2000

Rates per 10,000 live and stillbirths

	Singleton	Twin
All babies	101.7	111.0
Central Nervous System	4.0	12.5
Neural Tube Defects	1.3	6.8
Eye	1.5	1.5
Clefts	9.1	8.8
Heart	12.5	20.1
Respiratory	1.4	2.4
Limb reduction	2.9	2.7
Abdominal	2.7	3.6
Down's syndrome	6.4	5.6

Source: ONS unpublished linked data