CARIS

CONGENITAL ANOMALY REGISTER & INFORMATION SERVICE

COFRESTR ANOMALEDDAU CYNHENID

1999 Annual Report

NHS CYMRU WALES

Welcome to CARIS

Foreword

Welcome to the second CARIS annual report. Now in our third year of operation, this technical report outlines 1998 Welsh congenital anomaly data to the end of the first year of life, as well as initial data for 1999.

Once again we would like to express our sincere thanks to all the hard working health professionals who contribute to CARIS.

Already the data have been used to help with both research and service planning in Wales. We believe that this work will directly contribute to future developments and improvements in this important area of health.

We would like to thank the many people who have continued to offer support and assistance over the past year, including the West Wales Centre for Public Health, the Office for National Statistics, the National Assembly for Wales and especially Swansea NHS Trust.

MARGERY MORGAN

JUDITH GREENACRE

Swansea, July 2000

Contents

Introduction to CARIS

Welcome to the second Annual Report of the Congenital Anomaly Register and Information Service for Wales.

CARIS is based at Singleton Hospital, Swansea and is funded by the National Assembly for Wales. Professionals and staff involved with running CARIS are shown in Appendix A.
An overview of CARIS data flows can be found in Figure 1.

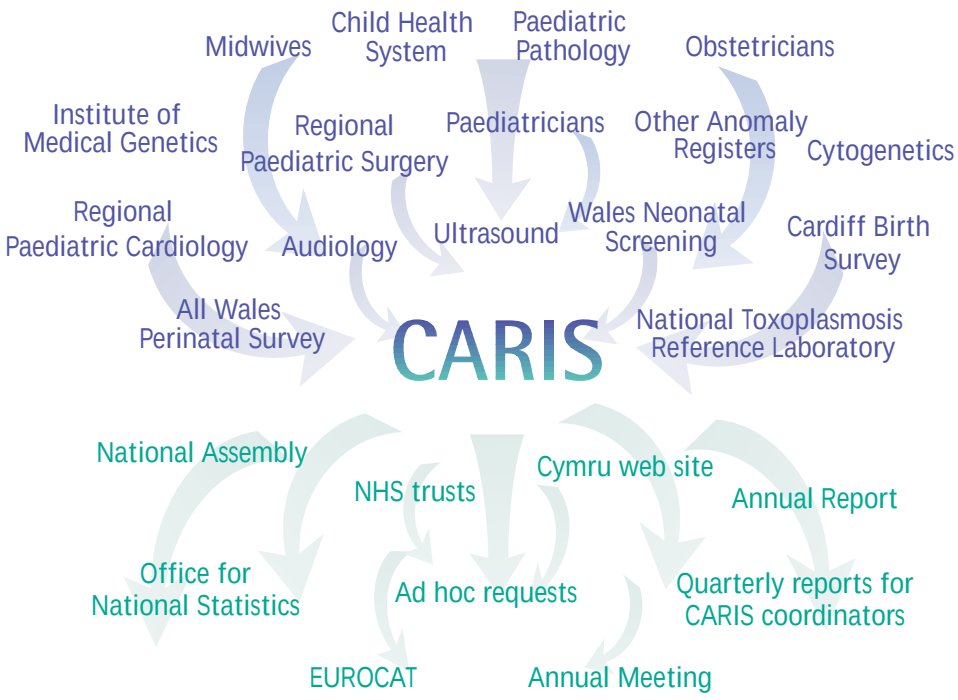
Aims of CARIS

CARIS aims to collect reliable data about congenital anomalies which can then be used to help:

- build up and monitor the picture of congenital anomalies in Wales.
- assess interventions intended to help prevent or detect congenital anomalies such as the folic acid campaign or antenatal screening.
- plan and co-ordinate provision of health services for affected babies and children.
- assess possible clusters of birth defects and their causes.

CARIS collects information about any fetus or baby who has, or is suspected of having, a congenital anomaly and whose mother is normally resident in Wales at the time of birth. It covers babies in whom anomalies are diagnosed at any time from conception to the end of the first year of life. Data collection commenced on 1st January 1998 and includes any baby where pregnancy ended after this date.

Figure 1
THE CARIS REPORTING SYSTEM



Congenital anomalies in Wales: agencies involved

Over 40,000 recorded pregnancies occur in Wales each year. Of these, about three quarters are registered as live or stillbirths, the rest ending in termination or spontaneous loss of the fetus before the 24th week of pregnancy.
About 2% of births take place at home. Wales has 17 consultant obstetric units and 11 general practitioner units. The majority of births take place in these units. However, a significant number of births to Welsh mothers occur in hospitals across the English border, most notably the Countess of Chester Hospital (500 births), the Royal Shrewsbury Hospital and the County Hospital, Hereford. Babies born in North Wales requiring specialist attention may be delivered or treated in Liverpool. Occasionally, babies that require very specialist help may be born further afield in other centres of expertise. It is very important therefore that CARIS has good links with these sites, and with the Mersey and the West Midlands congenital anomaly registers, that border Wales.

In each of these units, many different agencies may be involved in the diagnosis and management of a baby with a congenital anomaly. As well as the clinicians responsible for the mother and baby, diagnostic services such as radiology (especially ultrasound scanning) and pathology are often involved. Regional diagnostic services such as cytogenetics and paediatric pathology can have detailed information on the nature of anomalies, especially for those babies who die or where the pregnancy is terminated. Specialist clinical services may also be involved such as paediatric cardiology, paediatric surgery and plastic surgery.
Babies with congenital anomalies may be recorded on other databases such as the Cardiff Birth Survey, The All Wales Perinatal Survey or the Standard Child Health Computer System.
CARIS recognises that all these sources have the potential to give valuable information about congenital anomalies in Wales. It has worked hard to establish an effective multiple source reporting system with good links to as many of these agencies and units as possible.
At present 18 different types of source report to CARIS (Appendix B) and this has been a major contributory factor to the quality of CARIS data.
Despite this, direct clinical reporting remains the main source of information on many babies, especially those who

- die but do not have post mortems.
- survive and have anomalies that do not require immediate specialist help.

Data collected

Cases and anomalies

CASES

Babies and fetuses included in the CARIS data set (cases of congenital anomaly) are defined as follows:

Babies and fetuses born from 1st January 1998 to mothers normally resident in Wales at the time of birth and in whom an anomaly is detected during pregnancy, at birth or within the first year of life. The anomaly must be a defect which was present at the time of birth, regardless of when it was detected.

ANOMALIES

CARIS has defined an anomaly as involving a structural, metabolic, endocrine or genetic defect, present in the child/fetus at birth.

All anomalies reported to CARIS are coded using the Royal College of Paediatrics and Child Health expansion of ICD10[1].

For the 1998 annual report, CARIS adopted a slightly amended form of the Office for National Statistics groupings for congenital anomalies[2].

This involves a system of major group headings and sub-headings.

For example:

GROUP HEADING	GROUP CODE
Major group:	
Central Nervous System	A
Sub Groups:	
Anencephalus	0A
Spina Bifida	0B
Congenital Hydrocephalus	0C
Encephalocele	0E
Other CNS anomalies	0F

A full list of the CARIS modification of the ONS grouping system and the ICD10 codes included in them are given in Appendix C.

This grouping system comprehensively covers all congenital anomalies but it is difficult to pick out some groups of anomalies that may be of clinical or epidemiological significance. To help with this, CARIS is developing its own list of key congenital anomalies. (see data section and table 3).

The Department of Medical Genetics, University Hospital of Wales provide expertise to ensure accurate diagnosis of cases with multiple or complex anomalies.

In common with the ONS and EUROCAT reporting systems, some minor anomalies are excluded from reporting to CARIS, unless they are associated with other more significant defects. A list of such exclusions is given in Appendix D.

Coding of anomalies is undertaken by staff trained in clinical coding who adhere to the principles of ICD10.

This includes:

- Coding every anomaly identified.
- Using the minimum number of codes necessary to accurately describe the clinical picture.
- Coding to the greatest level of specificity available in the classification.

A coding audit programme is under way.

[1] Extension of the International Classification of Diseases and Related Health Conditions (ICD10). Royal College of Paediatrics and Child Health 1995

[2] Congenital Anomaly Statistics. Series MB3 no.12 ONS 1997

The CARIS reporting system

There are three main routes by which CARIS receives data:

- CARIS warning card.
- CARIS reporting form.
- Reports from specialist sources and databases.

Warning card

This is intended to flag up a potential case to CARIS before all the details are fully known, usually before the pregnancy has ended. A copy of the card is shown in Appendix E. About half the congenital anomalies reported to CARIS are suspected antenatally.
CARIS is keen to hear of any possible problems early on in pregnancy:

- to help improve the number of confirmed cases subsequently identified.
- to contribute to the future evaluation of antenatal screening.

Reporting form

This represents all the clinical data collected by CARIS on any baby or fetus. It would normally be filled in once the pregnancy has ended and there is reasonable evidence of at least one congenital anomaly. A copy of the form is shown in Appendix F.

Reports from specialist sources

Information from specialist sources usually involves detailed diagnostic data and is extremely useful to CARIS, both in improving the quality of information on known cases and in helping identify new ones. The data from such sources would not usually include the full CARIS data set and this is normally completed through the CARIS co-ordinators, if the register does not already know about the baby/fetus.

CARIS co-ordinators

All delivery units and most community trusts now have a nominated co-ordinator that is able to supply CARIS cards and forms. They can also help with filling in the form and retrieving notes. In many units the initial CARIS card is sent to the co-ordinator before being sent to the office so that the co-ordinator has a record of what anomalies are present in their unit.
CARIS also feeds back details of reported cases to all co-ordinators on a regular basis so that they are aware of any information CARIS may have received from other sources. This data is then available for local audit. A variety of people have taken up this role and CARIS is very grateful to them for helping make reporting so successful (Appendix G).

Over the first two years of operation, it has become clear that some units have periodic difficulties in getting data to CARIS. A reporting support system is being developed to help with this.

Data processing

Once data is received in the CARIS office:

- The case is checked against the Welsh NHS Administrative Register to validate the address, DOB and NHS numbers of mother and baby.
- The postcode is also checked on postcoding software from which the Unitary Authority, Ward and grid references are obtained.
- The CARIS database is searched to check if the case is already known and the case is either updated or added.
- The data is entered on the database. Data relating to diagnosis,[3] medication[4] and occupation[5] are coded. Diagnoses are checked against the CARIS exclusion list.
- The complete entry is validated independently and saved.
- Live and stillborn babies are flagged for reporting to the Office for National Statistics (ONS).

[3] Extension of the International Classification of Diseases and Related Health Conditions (ICD10). Royal College of Paediatrics and Child Health 1995

[4] Read Codes v3

[5] Standard Occupational Classification 2nd Edition. OPCS 1995

[6] Todd G. Cleft lip and palate services; Outline framework (Report of Director). SHSCW. Nov 1999

[7] Todd G. Report on future provision of a congenital heart disease service in Wales. SHSCW. Feb 2000

[8] Fielder HMP, Poon-King CM, Palmer SR, Moss N, Coleman G, Assessment of impact on health of residents living near the Nant-y-Gwyddon landfill site: retrospective analysis BMJ 2000; 320:19-22

[9] Greenacre J, Morgan M, Tucker D. Analyses require high quality data. (letter). BMJ 2000; 320:1542

[10] Abramsky L, Botting B, Chapple J et al.Has advice on peri conceptual folate supplementation reduced neural tube defects? Lancet 1999; 54:998-999

[11] Greenacre J, Morgan M. Diagnostic Features in Down Syndrome. BINOCAR conference 2000, Birmingham.

How the data is used

Feedback to clinicians

CARIS recognises the importance of regular feedback to those supplying the data. This is done in a number of ways:

- Quarterly feedback of data to CARIS co-ordinators.
- The CARIS newsletter.
- Visits and presentations around Wales.
- Responses to requests for information (– for example to assist with audit projects in Bangor and Wrexham).

Office for National Statistics

Once a case is reported to CARIS, it is transferred electronically to the ONS reporting system. This only includes data on live or stillborn babies. ONS operates a surveillance system to alert to possible rises in the numbers of notifications.

Office of the National Assembly for Wales

The numbers of recorded anomalies are fed back to the Office of the National Assembly for Wales (previously Welsh Office) every quarter.

Through the Office, CARIS can provide information to help answer questions from the National Assembly concerning congenital anomalies in Wales.

EUROCAT

At the end of each calendar year, anonymised congenital anomaly data recorded with CARIS is transferred to the EUROCAT register as part of the European surveillance programme.

Annual Report and meetings

Meetings were held in both North and South Wales during 1999. These reviewed the data for 1998 and provided an opportunity to discuss issues relating to neural tube defects and Down syndrome.

Further studies

CARIS data have been used for the following work:

- Specialist Health Services Commission for Wales have used data on both cleft lip/ palate[6] and congenital heart disease[7] to help assess the need for appropriate specialist services in Wales.
- Dept of Epidemiology and Public Health, University of Wales College of Medicine are using data for ongoing studies into the levels of congenital anomalies around landfill sites in South Wales[8].
- CARIS has also looked at levels of congenital anomaly around the Nant-y-Gwyddon landfill site in South Wales.[9]
- BINOCAR (British Isles Network of Congenital Anomaly Registers) has used data on neural tube defects to assess the impact of the folic acid campaign.[10]
- CARIS has used data relating to Down syndrome to assess aspects of antenatal screening. This was presented to the BINOCAR annual conference[11] (see page 20).

Figure 2
CARIS REGISTRATION 1998/99

Confidentiality

CARIS recognises that it has a duty to meet professional ethical standards and to maintain the confidentiality and security of the data it holds. Personal information held on a computer system is safeguarded by the Data Protection Act and CARIS is registered under this.

CARIS has adopted the following principles set out in the Caldicott Report,[12] which have been accepted by the NHS in Wales:

- Justify the purpose of transfer of patient identifiable information.
- Don't use patient identifiable information unless it is absolutely necessary.
- Use the minimum necessary patient identifiable information.
- Access to patient identifiable information should be on a strict need to know basis.
- Everyone with access to patient identifiable information should be aware of their responsibilities.
- Understand and comply with the law.

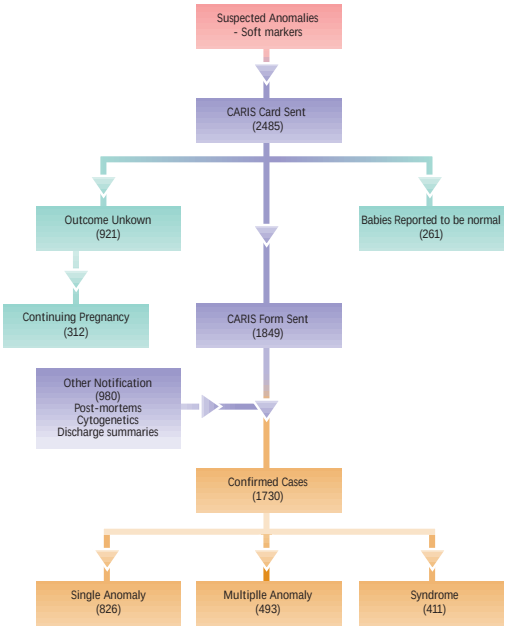
CARIS has drawn up its own confidentiality and security policy[13] to manage data requests and this is strictly followed.

Data quality

To ensure that the information it produces is as reliable as possible, CARIS has developed a data quality assurance programme.[14] A summary of some of the data quality indices for 1998 and 1999 is given in Appendix H. Receiving data from so many sources increases the risk of numbers being over-inflated from duplicate entries. The CARIS checking programme to exclude duplicates is listed in the Appendix.

Targets for data completeness are also given. In general, CARIS data shows high levels of completeness. However, some data items fall below the targets set. These items include date of LMP and birthweight. Such items highlight the need for continuing good quality clinical reporting as these are not easily available from specialist sources. CARIS has been investigating the possibility of developing real time access to the Standard Child Health Computer System, from which many of these data items could be retrieved. So far however, administrative logistics have prevented such a link being established.

[12] The Caldicott Committee. Report on the Review of Patient Identifiable Information. DoH London (1997)
[13] Confidentiality Security and Disclosure: Code of Practice. CARIS September 1998
[14] Data Quality Standards. CARIS March 1999



Data

Overall picture

The flow of data reported to CARIS is shown in Figure 2.

All Wales

1998 DATA

In the 1998 report, data on 849 confirmed cases (babies/fetuses) with congenital anomaly were presented. Since that time a further 95 cases with pregnancy ending in 1998 have been reported, giving a total of 944 cases for the year. 33,610 live and stillbirths (total births) occurred in Wales in 1998, giving an updated gross rate of 281 cases of congenital anomaly for every 10,000 total births. Of the 944 cases, 717 resulted in live or stillbirths. Thus the rate of cases of congenital anomaly among live and still births in Wales for 1998 was 213/10,000 total births (2.1%).
Among the 944 cases from 1998, 2334 anomalies or syndromes have been identified, giving a gross rate of 694 anomalies per 10,000 total births and a ratio of 2.5 anomalies per case.

1999 DATA

So far, 786 cases of congenital anomaly with pregnancy ending in 1999 have been reported to CARIS, less than that reported for 1998 at the same stage last year. The number of births fell in 1999 in Wales to 32,266 live and stillbirths (total births), giving a gross rate of 243 cases of congenital anomaly for every 10,000 total births. Of the 786 cases of congenital anomaly, 607 resulted in live or stillbirths. Thus the rate of cases of congenital anomaly reported so far among live and still births in Wales for 1999 was 188/10,000 total births (1.9%).
Among the 786 cases from 1999, 2016 anomalies or syndromes have been identified, giving a gross rate of 624/10,000 total births and a ratio of 2.6 anomalies per case.

COMMENT

The breakdown of the anomalies for both 1998 and 1999 to date, by ONS anomaly groups, is shown in Figure 3 for both 1998 and 1999. Details of the breakdown by cases is given in Table 1 and by anomalies in Table 2. The rates for 1999 are slightly lower than for 1998 this time last year,

This report is based on cases born during 1998 and 1999. All babies born in 1998 have now reached the end of their first year of life. However, some babies born during 1999 will complete their first year of life towards the end of 2000, so the 1999 data presented in this report must be regarded as provisional. The final data for 1999 will be presented in next year's report.

CARIS rates

CARIS receives reports on cases resulting in miscarriage or termination. It is not possible to describe accurately how many terminations or miscarriages occur that do not have congenital anomalies, making it difficult to estimate precise rates or to enable comparisons with other data sources.

In this report the term **gross rate** is used to describe the total number of cases or anomalies/10,000 live and stillbirths/year. Cases ending in live or stillbirth, termination of pregnancy or spontaneous fetal loss are included in this figure.

The term **rate** is used to describe the number of CARIS cases or anomalies that end in live or stillbirth, per 10,000 live/stillbirths/year.

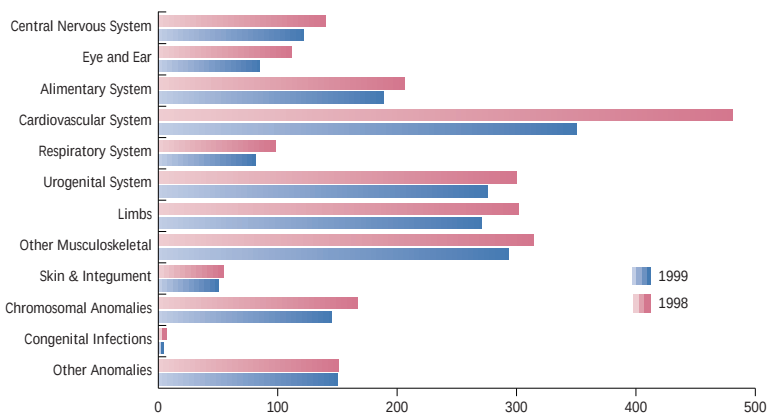


Figure 3
CONFIRMED ANOMALIES AND SYNDROMES BY ONS CONGENITAL ANOMALY GROUP REPORTED TO CARIS FOR 1998 AND 1999 (SO FAR)

but these differences are not statistically significant.

Further information on cases with malformations of particular clinical or epidemiological significance is given in Table 3. In general, it is still too early to pass comment on the significance of rates of particular anomalies. It is notable however that gross rates of gastroschisis in Wales have continued to rise during 1999. the gross rate for 1998 and 1999 combined is 5.3/10000 births (95% CIs 3.6 to 7.1). This figure is higher than that reported from many other sources for the early 1990s. Comparable data for 1998 and 1999 is not yet generally available.

Variations across Wales

In the 1998 annual report considerable variations in gross case prevalence rates were noted across Welsh Health and Unitary Authorities. In particular, Bro Taf and Morgannwg Health Authority areas had higher rates whilst Gwent had generally lower reporting rates.

The updated picture for both Health and Unitary Authorities is shown in Tables 4 and 5 and in Figures 4 and 5.

The high rates seen during 1998 for Bro Taf and Morgannwg are not reflected in the 1999 data so far. It is also interesting to note that these Health Authorities had the least increase in rate between the provisional and final 1998 figures, suggesting good initial reporting.

Over 1999, increased reporting of cases from the Gwent area has occurred as awareness of CARIS has increased.

At Unitary Authority level, numbers of cases of congenital anomaly remain relatively small, explaining some of the wide variations in rates. Figure 5 shows how combined 1998/1999 gross case rates for individual Unitary Authorities compare with the overall rate for Wales. Cardiff, Swansea and the Vale of Glamorgan have significantly higher rates. Isle of Anglesey, Bridgend, Rhondda Cynon Taff, Torfaen, Caerphilly and Monmouthshire have significantly lower rates. Once future years of data become available it should be clearer how much of the present picture is due to:

- small number variation.
- true differences in the prevalence of cases of congenital anomalies.
- differences in clinical reporting.

The numbers of anomalies and gross rates per 10,000 total births are given for each anomaly group, by Health Authority area, in Table 6.

Figure 4
PREVALENCE OF CARIS CASES OF CONGENITAL ANOMALY AT END OF PREGNANCY, WALES AND WELSH HEALTH AUTHORITIES, 1998 AND 1999 (SO FAR)
Gross case prevalence rates per 10,000 live and stillbirths (+95% confidence limits)

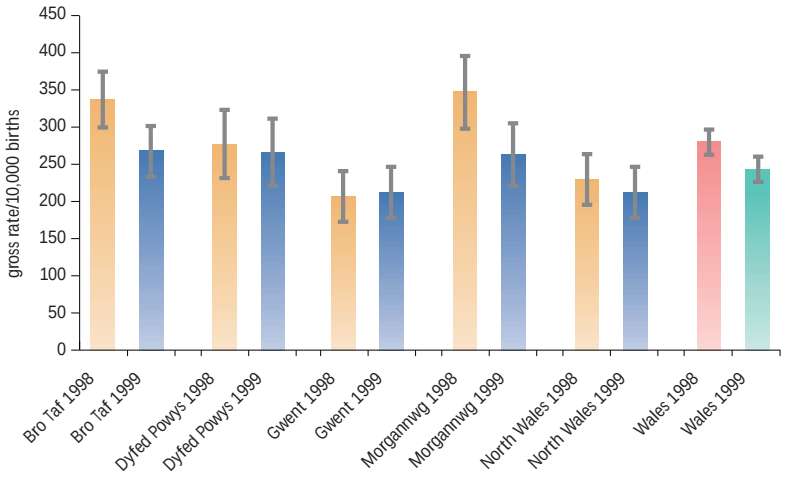
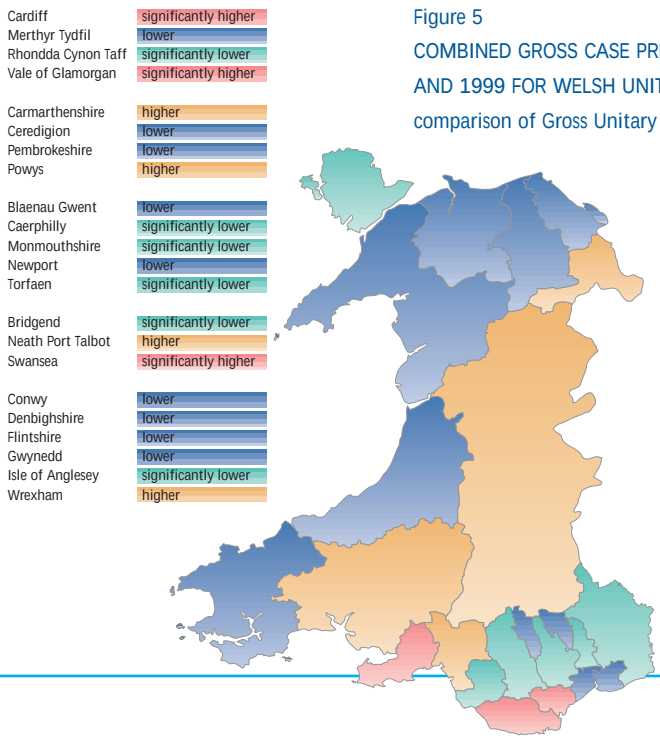


Figure 5
COMBINED GROSS CASE PREVALENCE RATES 1998 AND 1999 FOR WELSH UNITARY AUTHORITIES
comparison of Gross Unitary Authority and Welsh Rates



Relationship between cases and anomalies

A baby or case may have several anomalies, the average reported to CARIS being 2.6 anomalies per case.

Some of these anomalies can indicate the presence of an underlying syndrome. For example, a baby with Down syndrome may display some of the typical features of the condition. The baby could also have associated heart or gut anomalies. Alternatively, karyotyping may confirm the presence of the condition in utero, before any other features or anomalies are detected. Should the decision be made to terminate a pregnancy, the specific features and anomalies to be found in that particular case may never be known.

CARIS can now identify four broad patterns of anomalies among cases.

- single anomaly
  - involves a single defect that is not thought to be part of an underlying syndrome.
- multiple anomalies (single system)
  - involving a single organ or body system and not thought to be part of an underlying syndrome.
- multiple anomalies (different systems)
  - involving more than one organ or body system and not thought to be part of an underlying syndrome.
- syndrome
  – in which an underlying recognisable syndrome has been identified, regardless of the number of anomalies reported.

Table 3 shows the numbers and % of cases with specific malformations that fit into each of these groups. The table supports what is already known about some conditions. For example, in 29/35 (83%) of cases, gastroschisis occurs as the only defect. In cases of exomphalos however, only 3/22 (14%) occur as a single defect and over half are associated with an underlying syndrome.

Demographic features

GESTATIONAL AGE

Figure 6 shows the gestational age in weeks at the end of pregnancy for 1998 and 1999 CARIS cases together with the recorded outcome of the pregnancy.

As might be expected, there are 2 peaks in gestational age for babies with congenital anomaly. The first occurs between 16 - 22 weeks of pregnancy and is largely accounted for by terminations of pregnancy. The second and much larger peak occurs around term. As with the majority of births, most term babies are liveborn.

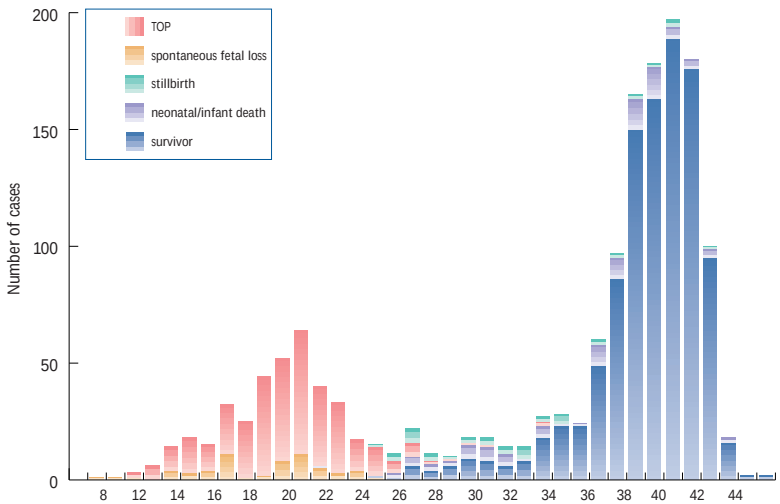


Figure 6
OUTCOME BY GESTATIONAL AGE AT END OF PREGNANCY
CARIS cases with pregnancy ending in 1998 and 1999 so far, where gestational age known

START OF PREGNANCY

CARIS uses the estimated date of last menstrual period (LMP) as a proxy for the time of conception. LMP is commonly calculated back from the estimated date of delivery (EDD), which is usually confirmed by antenatal ultrasound. For 1998 and 1999 together, CARIS has LMP dates for 91% of cases.

Seasonal variation in the development of congenital anomalies might be indicated by the month of the last maternal menstrual period. The month of LMP for cases is shown in Figure 7. It is not yet possible to identify any underlying trends.

MULTIPLE PREGNANCIES

Combined data for 1998 and 1999 shows that 70 out of 1730 cases (4%) were part of a multiple pregnancy (twins, triplets etc).

Using 1998 data, the percentages of pregnancies in which at least one fetus was affected by a congenital anomaly were as follows.

- 2.52% of all pregnancies (n=835/33159).
- 2.47% of singleton (single fetus) pregnancies (n=809/32709).
- 5.7% of multiple (twins triplets) pregnancies (n=26/450).

The relative risk of having a fetus affected by congenital anomaly in a multiple pregnancy is therefore 2.34 (95% CIs 1.60 TO 3.41).

SEX OF BABY / FETUS

Combined data for 1998 and 1999 shows that the reverse male to female ratio among CARIS cases suspected in the 1998 annual report was incorrect. For the 1707 cases in which the sex of the baby/fetus is known, 986 were male and 720 female, giving a female to male ratio of 1:1.37. (Gender was not determined in 23 cases).

MATERNAL AGE

Figure 8 shows the maternal age distributions at time of birth for babies reported to CARIS and for the general Welsh population of births for 1998 and 1999. The age distribution shows that the greatest numbers of mothers of babies with congenital anomalies were in the 25 - 29 year age group, similar to the pattern seen for all births in Wales. However this peak was not as high in the CARIS group.

CARIS reports a significantly greater proportion of mothers in the 35 year+ age group compared to the general population of expectant mothers. For mothers aged 35 and over, the relative risk of having a baby affected by a congenital anomaly is 1.88 (95% CIs 1.59 to 2.22) compared with mothers below this age. As the incidence of chromosomal anomalies is known to increase with maternal age, this is as expected.

Figure 7
MONTH OF MATERNAL LAST MENSTRUAL PERIOD
CARIS cases with pregnancy ending in 1998 and 1999 (so far).

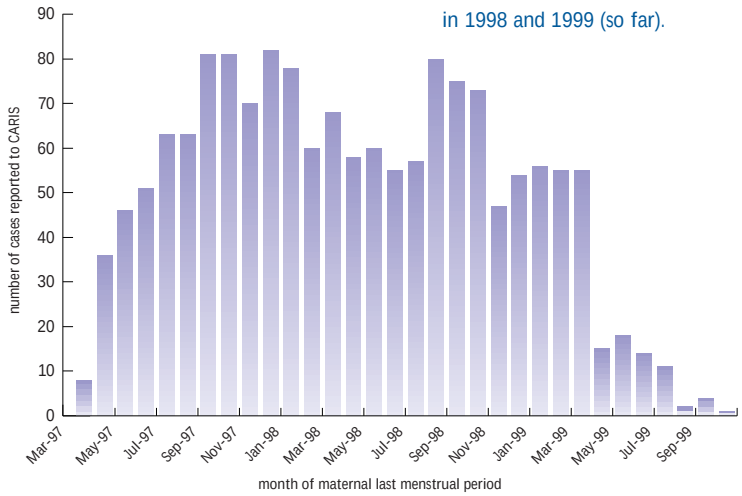
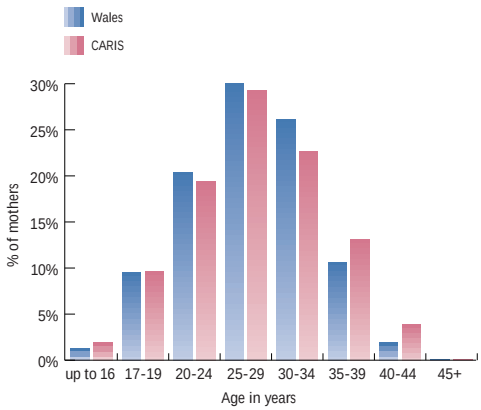


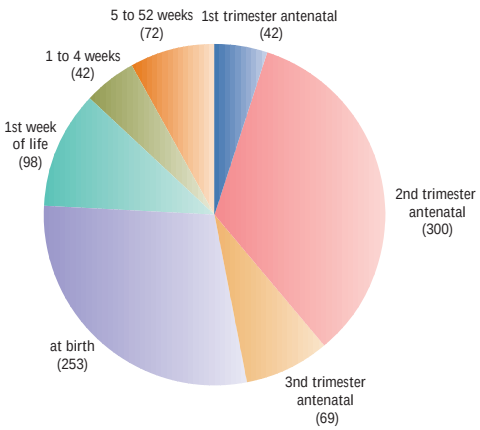
Figure 8
MATERNAL AGE DISTRIBUTION 1998/1999
Comparison of all mothers in Wales with mothers of confirmed CARIS cases



TIME OF FIRST DIAGNOSIS OF ANOMALY

Initial data from 1998 suggested that anomalies were indicated antenatally in about half of all cases. However, this could not be verified until data up until the end of the first year of life became available. Figure 9 shows the stage of development (where known) at which any anomaly was first suspected among cases with pregnancy ending during 1998. As expected, the figure shows that a slightly greater proportion of cases was detected at birth or beyond than was evident in last year's report. Antenatal indicators were reported in 47% of cases, the majority of which were during the 2nd trimester of pregnancy.

Figure 9
TIME OF FIRST DIAGNOSIS FOR CASES WITH PREGNANCY ENDING IN 1998
where time of first diagnosis known
(876 cases)



Outcomes for CARIS cases

ALL WALES PICTURE

The recorded outcomes for cases with pregnancy ending in 1998 are shown in Figure 10. This now represents a more complete picture for outcome than was available in the 1998 report, when not all babies had reached the end of their first year of life. Overall, about one fifth of cases resulted in termination of pregnancy and two thirds of cases continue to survive. The remainder were natural losses or postnatal deaths. The picture for 1999 so far is similar to 1998. The outcome of pregnancy by gestational age has already been shown in Figure 6. As might be expected, there is a difference in outcome between the patterns of anomalies. Survival is higher among babies recorded as having single anomalies and poorest for those recorded as having syndromes (Figure 11).

Figure 10
OUTCOME FOR CASES TO THE END OF THE FIRST YEAR OF LIFE
CARIS cases with pregnancy ending in 1998
(n=944)

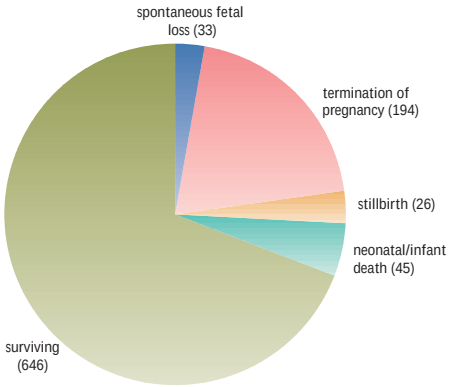
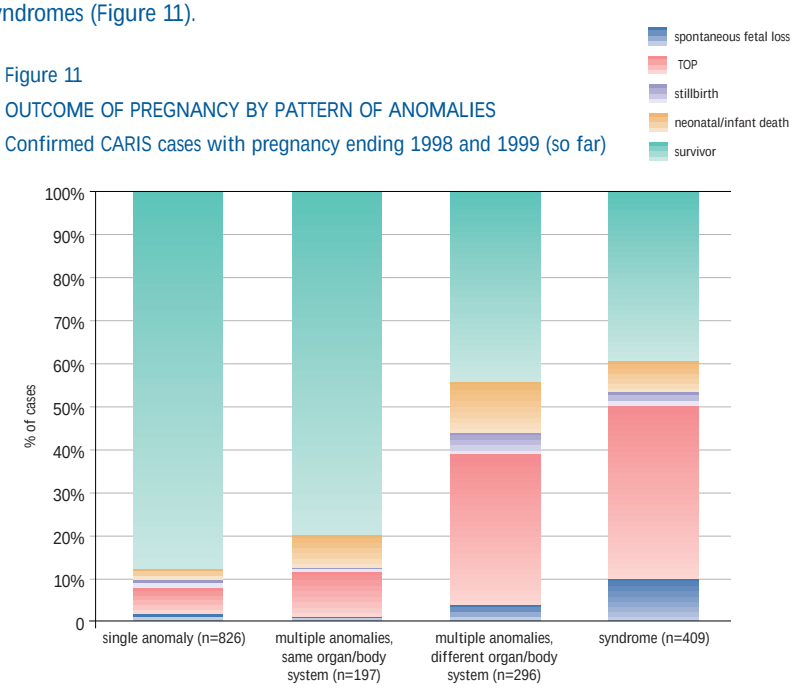


Figure 11
OUTCOME OF PREGNANCY BY PATTERN OF ANOMALIES
Confirmed CARIS cases with pregnancy ending 1998 and 1999 (so far)



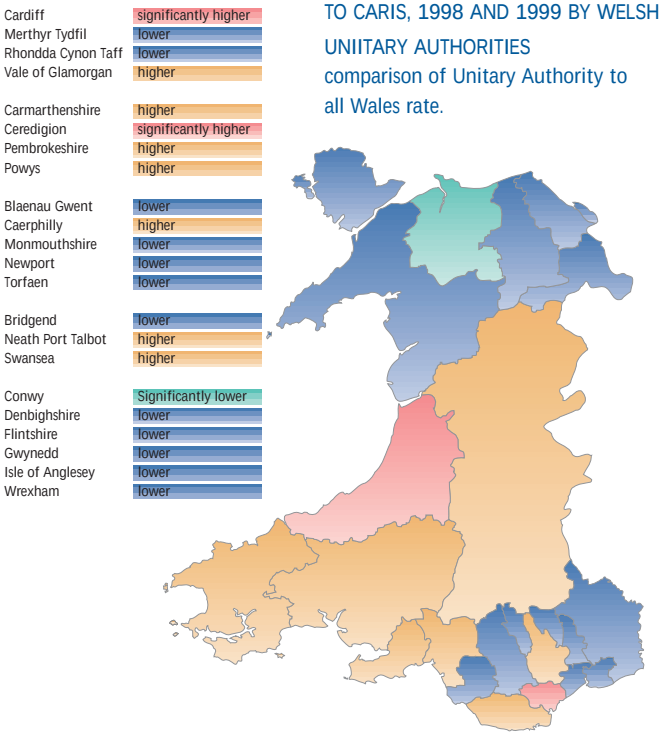
In general, much of the poorer rates of survival can be accounted for by higher rates of termination of pregnancy. The poorer rates may also be accounted for by postmortems in the babies who have died, revealing additional anomalies not apparent in survivors. Using combined data for 1998 and 1999 so far, Table 3 shows the recorded outcomes for cases with some specific conditions. At present, the numbers of cases involved are still relatively small and figures should be interpreted with caution. It should also be remembered that these figures are recorded retrospectively and cannot be taken as a predictor of outcome.

The stillbirth rate among cases reported to CARIS and reaching the 24th week of pregnancy was 3.6% for 1998 and 3.3% for 1999. These rates are significantly higher than that among the general population in Wales (0.54% in 1998 and 0.48% in 1999). This finding supports the suggestion of poorer outcomes for babies with congenital anomalies.

VARIATIONS ACROSS WALES

In the 1998 report CARIS noted differences across Wales in the outcomes of pregnancy for babies with congenital anomalies. The proportions of babies surviving are shown by Welsh Health and Unitary Authorities for 1998 in Table 4(a) and for 1999 in Table 4(b).

Figure 12
SURVIVAL RATES FOR CASES REPORTED TO CARIS, 1998 AND 1999 BY WELSH UNIITARY AUTHORITIES
comparison of Unitary Authority to all Wales rate.



Babies with congenital anomalies born to mothers resident in Gwent appear to have generally poorer overall survival rates than for other Welsh Health Authorities although these differences are not statistically significant. Figure 12 shows the degree of difference (as denoted by the 95% CIs) in case survival rates between Welsh Unitary Authorities and the all Wales rate for 1998 and 1999 combined.

Small number variation has a role in the differences noted to date. The poor survival rate seen in Torfaen in 1998 is not repeated in the 1999 data. These rates are based on very small numbers of cases. This should be less of a problem as further years of data are collated. Survival is also likely to seem poorer if good clinical reporting to CARIS is not achieved, as information about babies with anomalies who survive is not available from other CARIS data sources. Other reasons for these variations may be complex, and cannot be adequately investigated from the CARIS database alone. The outcome profile seen for babies reported to CARIS may simply be a reflection of similar trends for all births across Wales. Relevant factors relating to termination of babies with anomalies may include accessibility to services, availability of information, socio-economic indices and social or cultural norms. In addition, these figures give no indication of levels of disability, quality of life or the need for further medical treatment among survivors. Over the next year CARIS hopes to study relationships between case ascertainment and reported survival rates as part of an ongoing programme of data quality assurance.

Surveillance

Background

The Office for National Statistics operates a surveillance system to detect potential rises in the number of babies born with congenital anomalies[15]. Analysis of data is undertaken by ONS using statistical (CUSUM) techniques in which the number of anomalies reported for a particular geographical area and time period are compared with the expected number, based on the previous year of reporting. Analysis is usually undertaken monthly, by month of birth of baby and Health Authority area of residence. If the number of actual anomalies exceeds the number expected, a warning is sounded and Directors of Public Health are informed of a potential problem. Analyses are based on numbers of births and do not include termination data.

CARIS is not set up to provide detailed analyses of potential rises in reports on anomalies. However, it seeks to assist Directors of Public Health to assess potential problems when CUSUM warnings sound. In order to assist this process, the total number of anomalies by Health Authority and ONS malformation group are given in Table 6 for both 1998 and 1999. The proportions of the total that have resulted in live or stillbirth (and thus been included in current ONS surveillance systems) are also shown.

Factors likely to influence the sounding of CUSUM warnings

CUSUM warnings may be affected by the following:

IMPROVED REPORTING

It is known that considerable under-reporting of cases to ONS took place prior to CARIS being established. This led to an artificially low estimation of expected cases. Since 1998 CARIS has retrospectively reported to ONS any cases from earlier years that could be identified from other sources (EUROCAT, un-submitted SD56 forms etc), as well as new CARIS cases that met ONS reporting criteria. The numbers of cases reported from Wales (via health authorities) to ONS using the old system of form SD56 are shown in Figure 13, together with numbers of cases reported from CARIS. The figure clearly shows that, with CARIS becoming operational, under reporting has reduced considerably, leading to an apparent rise in cases, above expected levels. CARIS considers this to be a significant factor in causing 98 CUSUM warnings for Wales to be issued by ONS during 1998. In 1999, once the new reporting levels had stabilised, only 2 CUSUM warnings were issued.

At the end of 1999, ONS re-calibrated their CUSUM programme for Wales to take account of increased levels of reporting.

VARIATIONS IN TERMINATION RATES

ONS estimations are based on data that excludes terminations. As antenatal detection of congenital anomalies has improved, increasing numbers of parents are offered the option to terminate a

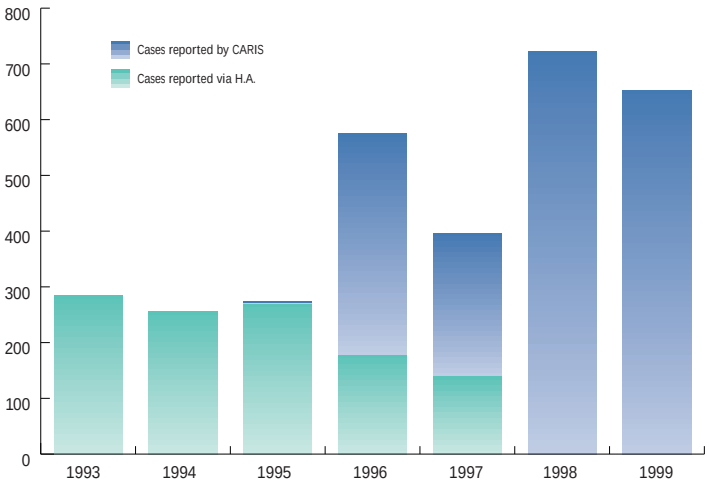


Figure 13
CASES REPORTED TO THE OFFICE FOR NATIONAL STATISTICS, 1993 TO 1999
comparison of number reported via Health Authorities and CARIS

[15] The OPCS Monitoring System for Congenital Malformation: Occasional Paper 43. OPCS 1995

pregnancy. This may lead to a reduction in reporting to ONS and thus a reduction in the estimated numbers of expected cases. Termination remains a difficult personal choice for parents and fluctuations in rates can be expected especially where small numbers of cases are involved. This may lead to variations in the number of cases reported to ONS and thus increase the likelihood of the CUSUM warning firing. Because the CUSUM system does not take account of termination data, it is less likely to be sensitive to picking up a true rise in cases of congenital anomaly that can be detected in the antenatal period. CARIS has asked ONS to look at ways of developing a more refined surveillance system in Wales that also includes data on cases that result in termination of pregnancy.

VARIATIONS DUE TO SMALL NUMBERS

At Health Authority level, numbers of specific anomalies are extremely small, and the presence of even one extra case may lead to an apparent increase in observed over expected numbers.

VARIATIONS IN THE LENGTH OF PREGNANCY

The CUSUM technique looks at the month of birth of the baby rather than the month of the start of pregnancy. Fluctuations in the number of babies reaching term may cause an apparent cluster over a short space of time. A refined surveillance system in Wales may be able to look at data in relation to month of LMP rather than birth. However, for this to succeed, good clinical reporting of maternal LMP is required.

VARIATIONS IN THE BIRTH RATE

The CUSUM technique measures expected numbers of cases, based on previous years reporting, rather than rates based on numbers of births. Should the birth rate increase, numbers of cases may go up even though the incidence rate of an anomaly remains the same. As the birthrate is currently falling in Wales, this is not likely to be a significant factor.

A TRUE RISE IN THE NUMBER OF ANOMALIES

The CUSUM warning may go off because of a true rise in the incidence of an anomaly, reflected in an increased prevalence at birth.

Special Topics

A genetic perspective

About 2-3% of children are born with congenital problems of significant severity. These can have a considerable impact on their lives and the lives of their families. Some congenital anomalies could have been prevented by (for example) the avoidance of specific teratogens, prophylactic folic acid or dietary measures in cases of maternal diabetes or phenylketonuria. Of particular importance are previously unrecognised teratogens, which are often only identifiable when surveying a group of affected individuals, rather than individual cases. This became painfully obvious in the delayed recognition of thalidomide as a devastating teratogen.

As a result, systems were set up in the UK (ONS) and elsewhere to monitor congenital anomalies. However the strength of such systems is very dependent on the level and accuracy of reporting. This has led to the setting up of more regional registers, like CARIS, which can have a much higher local impact.

Over the last year, three consultants from the Institute of Medical Genetics (UHW, Cardiff) with a particular interest in congenital anomalies were invited to review cases with multiple congenital anomalies reported to CARIS without a specific diagnosis or aetiology, such as a chromosome anomaly. We were asked whether additional information could provide more accurate classification. Although review outside a clinical consultation limited us to the information provided, we were able to provide more accurate classification for a number of cases. In some cases we had already seen the family in clinic and made a diagnosis. In others maternal information had not been correlated with the congenital anomalies at the time of reporting, but on review in one case it became clear that the anomalies were due to a valproate embryopathy. In a number of cases we reviewed the pathology slides and photographs, particularly the facial features, and provided a probable diagnosis, not obvious from the post mortem report. In all, 65 cases were reviewed.

As accurate classification improves understanding of the nature and aetiology of congenital anomalies, we feel that genetic input has been helpful in some cases. We have also modified our practice, and report children to CARIS we see in clinic born since January 1998, even if they are over a year old, as this will in some cases add the diagnosis to the previously reported information.

DANIELA PILZ
HELEN HUGHES
SALLY DAVIES

Genetics case scenario:

There had been some anxiety about Mrs X's baby from about 32 weeks gestation. The baby was felt to be small and serial ultrasound scans confirmed this.

The baby's abdominal and head circumference were growing but only on the 3rd centile. Reduced fetal movements and a poor cardiotocograph prompted an emergency Caesarean section at 36 weeks.

The baby was born in good condition but only weighed 1.9kg. The paediatric registrar examined the baby and found it had a small head, mild eyelid ptosis, an extra little finger, and syndactyly of the second and third toes. It was difficult to determine the baby's sex from the appearance of the genitalia. Chromosome analysis was arranged and showed a normal 46, XY karyotype. At this time the baby was reported to CARIS, without a specific diagnosis. The family was referred for genetic counselling and to discuss the likelihood of having another affected child.

When the geneticist saw the family in clinic, she felt that in view of the little boy's congenital problems, and his now apparent developmental delay, a condition known as Smith-Lemli-Opitz syndrome (SLO) should be considered. This rare condition is due to a change in the 7-dehydrocholesterol-reductase gene, which is part of cholesterol metabolism. SLO has an autosomal recessive inheritance pattern, with the parents having a 1 in 4 chance of having another affected child. The child was found to have high 7-dehydrocholesterol and low cholesterol levels, which confirmed the diagnosis. Antenatal testing if requested would be available during another pregnancy.

The geneticist reported the diagnosis to CARIS, which allowed updating of the record, and a more accurate assessment of the incidence of Smith-Lemli-Opitz syndrome.

Cleft lip and palate in Wales

The birth of a baby with a cleft lip can cause a mix of emotions in the labour ward with disappointment often taking the edge off the happy event. Despite this, a team based approach to the child can ensure excellent results for the future.

How & when clefts occur

The upper lip and front of the mouth is formed by fusion of the median nasal process and the lateral maxillary processes between the 5th and 12th week of development. Fusion of the palatal shelves forms the hard and soft palate between the 8th and 12th week.

- Cleft lip and palate are the most common malformations of the head and face.
- Cleft lip with or without cleft palate occurs in about 1 per 1000 live Caucasian births.
- The male to female ratio is 2:1.
- Cleft lip and palate is twice as common as isolated cleft lip.
- Most cleft lips (80%) occur unilaterally on the left.
- Isolated cleft palate appears to be a separate entity and more common than isolated cleft lip.

Cleft lip & palate in Wales

There were 137 confirmed cases reported to CARIS with pregnancy ending in the years 1998 and 1999. This gives a gross rate of 20.8 per 10,000 livebirths/stillbirths. Of the 137 cases, 95 were either live or stillborn, giving a rate of 14.5/10000 live or stillbirths. These rates may be slightly higher than elsewhere, as shown in the table above.

COMPARATVE RATES FOR CLEFT LIP AND PALATE FROM DIFFERENT CONGENITAL ANOMALY REGISTRIES.(RATES PER 10,000 LIVE/STILLBIRTHS)

CARIS 1998-1999		NW THAMES 1990-1998	TRENT 1997-1998	EUROCAT 1980-1994
All cases	Live/ stillborn cases	All Cases > 16 weeks gestation	Cases include live/stillbirths and terminations	Cases include live/stillbirths and terminations
20.8	14.5	14.8	13.9	15.3

The CARIS cases encompass all types of clefts and support isolated cleft palate being more common than isolated cleft lip. The outcome for all cases is shown below. Of the 137 cases reported, 90 (66%) were recorded as survivors.

Antenatal diagnosis

Although cleft lip and palate can be diagnosed early in the second trimester, most diagnoses are made at the anomaly scan at 18 to 20 weeks gestation.

Antenatal diagnosis gives parents the opportunity to come to terms with the situation and enables them to meet the team who will be involved with care and surgery after birth.

Reported detection rates for fetal facial anomalies by antenatal ultrasound range from 25% to 43%.[16] The classic ultrasound finding seen in a unilateral cleft lip is a vertical transonic area within the upper lip with extension into the nose if the palate is involved (see Figure 14). Isolated cleft palate is very difficult to demonstrate on ultrasound.

Polyhydramnios or difficulty visualising the fetal stomach may be suggestive of a facial cleft. This is probably due to leakage of amniotic fluid through the cleft into the nasopharynx and back into the amniotic cavity without being swallowed.

Following the diagnosis of a cleft, the ultrasonographer will review the fetus very closely for additional anomalies. If other anomalies are found, chromosome analysis is normally recommended.

The 137 cases reported to CARIS include 76 with cleft lip (with or without cleft palate). Of the 76, 26 were reported as being diagnosed antenatally. This equates to antenatal detection in 34% of cases with cleft lip and 19% of all cases of clefting. Other scan abnormalities were seen in 31/137 (22.6%) cases reported and include soft chromosomal markers and liquor volume problems.

NUMBERS OF CASES OF CLEFT LIP AND / OR CLEFT PALATE REPORTED TO CARIS 1998/99, SHOWING THE TYPE OF CLEFT AND OUTCOME.

	Total No. of cases	Spontaneous fetal loss	Termination of pregnancy	Stillbirth	Neonatal /infant death	Survivors
Cleft lip	25	0	2	0	1	22 (88%)
Cleft palate	61	8	14	1	3	35 (57%)
Cleft lip & palate	51	7	9	1	1	33 (65%)

[16] Twining, McHugo and Pilling. Textbook of Fetal Abnormalities. 2000 Churchill Livingstone

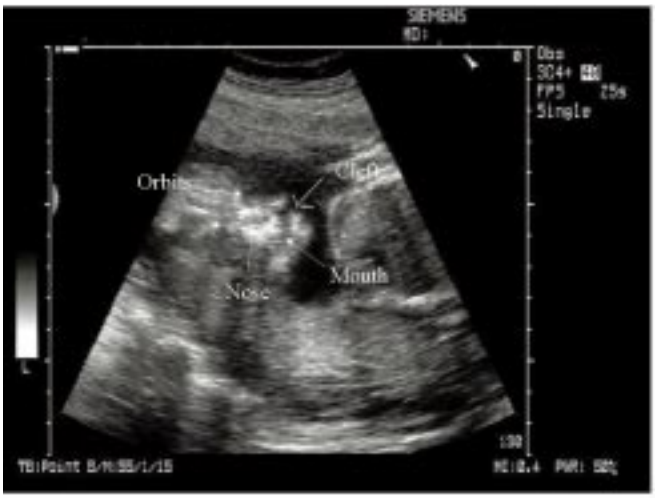


Figure 14
ULTRASOUND SCAN
SHOWING CLEFT LIP

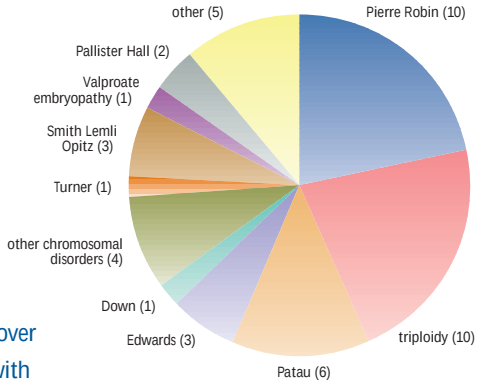


Figure 15
RECOGNISED SYNDROMES
ASSOCIATED WITH CLEFT LIP
AND/OR PALATE
Breakdown of 46 recognisable syndromes associated with 137 cases reported to CARIS with pregnancy ending in 1998-1999

Who is at risk?

The majority of cases have no obvious cause and may well be due to a number of factors. Some drugs taken in early pregnancy have been implicated including phenytoin, carbamazepine, steroids and diazepam. Four mothers with epilepsy were recorded in this group in Wales. There is an increased risk to both siblings and offspring of family members with facial clefting. Though not confirmed as yet, there may well be a genetic cause possibly involving transforming growth factor-α.

Other malformations

The presence of other malformations is much more common in cleft lip and palate and less frequent in isolated cleft lip. This is confirmed with CARIS data for 1998/1999. Recognisable syndromes are known to be associated with cleft lip and palate. Of the 137 cases, 46 were associated with syndromes. This accounted for just over two thirds of the cases associated with other anomalies. The types of syndrome involved are shown in Figure 15. Among the survivors, a variety of anomalies were reported, apart from the clefts themselves and any associated syndromes. The most frequent anomalies are shown below. Limb and musculoskeletal defects account for nearly half of the other anomalies associated with clefts.

ASSOCIATION BETWEEN TYPE OF CLEFT AND OTHER REPORTED ANOMALIES AMONG CASES OF CLEFT LIP AND/OR CLEFT PALATE REPORTED TO CARIS 1998/1999

	No other anomaly	Other anomalies	Total
Cleft lip	20	5	25
Cleft palate	17	44	61
Cleft lip with cleft palate	28	23	51

MAIN TYPES OF ANOMALY REPORTED AMONG CASES OF CLEFT LIP AND/OR PALATE (EXCLUDING THE CLEFTS THEMSELVES AND UNDERLYING SYNDROMES) 1998-1999

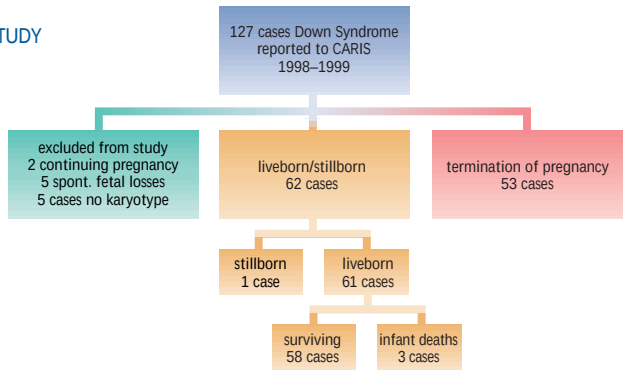
Other anomalies reported with cases of cleft lip/palate	Number	% of anomalies
All associated anomalies (not syndromes)	349	100%
Limb defects, of which	79	22.6%
Poly/syndactly	25	7.2%
Limb reductions	22	6.3%
Other musculoskeletal, of which	91	26.1%
Micrognathia	15	4.3%
Absence of rib	12	3.4%
Cardiovascular, of which	46	13.2%
Septal defects	15	4.3%
Anomaly of umbilical artery	9	2.6%
Respiratory, of which	31	8.9%
Agenesis/hypoplasia of lung	14	4.0%
Central Nervous System, of which	29	8.3%
Neural tube defect	7	2.0%
Hydrocephalus	6	1.7%

Down syndrome update

Down syndrome is the most common chromosomal anomaly - normally caused by an extra chromosome 21 (trisomy 21). Some initial information on Down syndrome in Wales was presented in the CARIS 1998 Report. With further data now available, CARIS has looked at the detection of Down syndrome in greater detail.

127 reported cases of Down syndrome[17] have been reviewed. Twelve cases were excluded from further study, as shown in Figure16, leaving 115 cases for analysis.

Figure 16
CASES IN STUDY



Features and anomalies associated with Down syndrome

Clinical reports and post-mortem findings often mention the characteristic features of Down syndrome as well as the anomalies that are associated with the syndrome. Postmortem reports were available in 42 of 57 cases (74%) in which the pregnancy was terminated, the baby was stillborn or died during infancy.

Features of Down syndrome commonly reported include:

- Facial dysmorphism, epicanthic folds, nose malformations, low set ears, protruding tongue.
- Sandal gap toes, single palmar creases.
- Hypotonia.

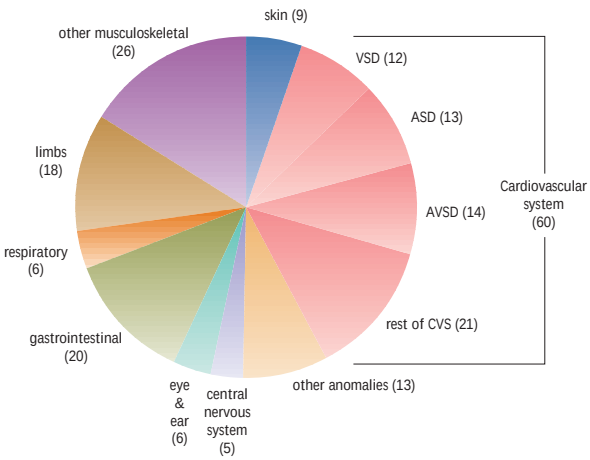
Once these common features of Down syndrome were excluded a total of 163 more significant anomalies were reported on the 115 cases. The breakdown of these is shown in Figure 17 . As can be seen in the diagram, cardiovascular anomalies were the most frequent type of anomaly, with septal defects accounting for over two thirds of this group. Malrotation of the colon accounted for the most common gut anomaly. Cystic hygroma (7) and hydrops (9) were also relatively common.

Antenatal detection

A review of the reporting forms of the 115 cases to CARIS suggest that just under half were first indicated antenatally.

- 7 (6%) were indicated in the 1st trimester of pregnancy
- 46 (40%) were indicated in the 2nd trimester of pregnancy
- 2 (2%) were indicated in the 3rd trimester of pregnancy
- 6 (5%) were indicated in investigations post termination of pregnancy
- 42 (37%) were indicated in the first week following live or stillbirth
- 4 (3%) were indicated later in infancy
- In 8 cases (7%), the time of first diagnosis of Down syndrome was not reported.

Figure 17
ANOMALIES REPORTED AMONG 115 CASES OF DOWN SYNDROME
Cases with pregnancy ending in 1998 and 1999. (Minor features of Down Syndrome excluded)



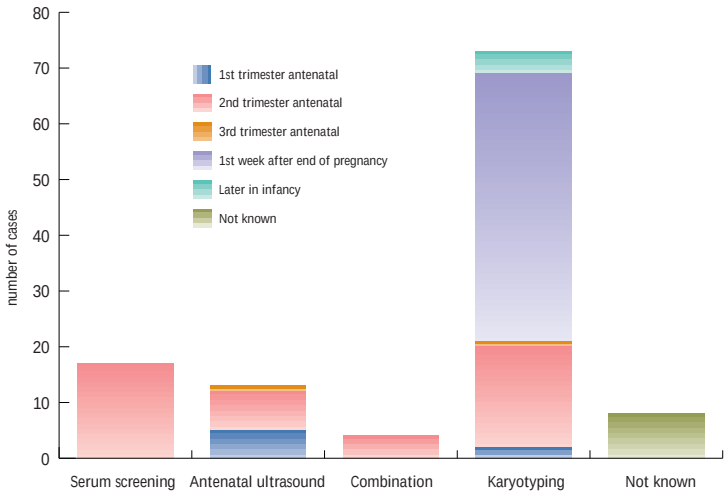
[17] 125 of these had a date of end of pregnancy in 1998 or 1999, in 2 cases, pregnancy continued into 2000

The diagnostic procedures that appear to have first suggested Down syndrome are summarised in Figure 18, together with the stage of pregnancy / infancy at which they were performed.

As expected, a large proportion of the terminated group (n=53) had a suspicion of Down syndrome recorded in early pregnancy, 7 in the first trimester and 38 in the second trimester. This included abnormal ultrasound, high-risk serum screening and amniocentesis done for age alone.

Of the liveborn/stillborn group, 8 cases were reported as having a suspicion of Down syndrome before the end of the second trimester. Three of these cases had second trimester confirmation of the diagnosis with karyotyping, suggesting parental choice to continue with the pregnancy.

Figure 18
PROCEDURE THAT APPEARS TO HAVE GIVEN FIRST INDICATION OF DOWN SYNDROME
Showing numbers of cases and stage of pregnancy/ infancy when performed (n=115)



Ultrasound findings in the antenatal period

Antenatal ultrasound scan findings were reported on 37 / 115 cases (32%). Of these, 19 (51%) included reports of nuchal translucency, increased nuchal fold thickness,[18] cystic hygroma or hydrops, suggesting the possibility of Down syndrome.

Other common antenatal scan anomalies included echogenic bowel (6 cases) and dilated renal pelves (5 cases).

Current evidence suggests that scanning for nuchal translucency between 10-14 weeks is an effective way of determining babies at risk of Down syndrome. The SURUSS (Serum, Urine, and Ultrasound Screening study) trial has yet to report but it is likely that a combination of serum testing and nuchal translucency measurement will produce the highest sensitivity for the lowest false positive rate[19] for antenatal detection of Down syndrome.

SERUM SCREENING

Of the 115 cases, 35 (30%) had a recorded result of a double serum screening test for Down syndrome (HCG/AFP). Of these, 23 were reported antenatally as being at increased risk (1:300 or greater) of having the condition, giving an antenatal detection rate of 66% in this small group. Conversely, 12/35 cases were reported antenatally as being at low risk for Down syndrome, giving a false negative rate of 34%.

- Of the 23 high-risk cases, 16 resulted in termination of pregnancy and 7 babies were liveborn.
- Of the 12 low risk results, 1 resulted in termination and 11 in livebirths.

Maternal age and outcome

The incidence of Down syndrome is well known to increase with maternal age. Of the cases reported to CARIS:

- 52/115 (45%) were associated with a maternal age of 35 years and above.
- 62/115 (54%) were associated with a maternal age of less than 35 years.
- in 1 case, maternal age was not recorded.

The table shows differences in antenatal karyotyping and outcome of pregnancy with maternal age. The differences between the 2 age groups are statistically significant on Chi Square testing.

[18] Greater than 3mm in 1st trimester; greater than 6mm in 2nd or 3rd trimesters
[19] Routine ultrasound screening in pregnancy - RCOG 2000

Comments

The study suggests that, at an all Wales level, there is relatively low overall use of antenatal screening to detect Down syndrome. Of the 115 cases:

- 80 (70%) had no reported evidence of serum screening.
- at least 78 (68%) had no reported evidence of antenatal ultrasound assessment of the nuchal fold.
- 38 (33%) had no record of antenatal serum screening, ultrasound anomalies or antenatal karyotyping.
- 12 (10%) had only antenatal karyotyping reported.

It is quite possible that CARIS has not received reports on the antenatal scan findings in all cases so that this part of the picture may be incomplete. Serum screening and karyotype results are reported directly to CARIS from the laboratories as well as via clinical reporting, so these data are thought to be a fairly accurate reflection of uptake of these procedures.

In 1999, a CARIS telephone survey of the main maternity units used by Welsh mothers indicated wide variation in the pattern of antenatal screening for Down syndrome across Wales. The survey suggested that about two thirds of mothers would routinely be offered serum screening for Down syndrome and just over half would be offered nuchal translucency assessment. (Where these services were not offered routinely, they may still be available to high-risk mothers). It is also known that where routine screening is offered, many mothers choose to opt out of testing. Both these factors may well explain the low overall levels of screening among cases of Down syndrome identified in this study.

Of the 53 cases detected before the end of the second trimester, 45 resulted in termination of pregnancy (85%). However, in the remaining 15% of cases, parents chose to continue with the pregnancy in the light of a diagnosis of Downs. This study has not evaluated the effect of an antenatal diagnosis of Down syndrome but this remains an interesting question.

SUMMARY

(1) Congenital anomalies with Down syndrome were as expected with cardiovascular anomalies being the most frequent.

(2) Low overall use of antenatal testing for Down syndrome has been identified across Wales.

(3) There appears to be wide variation in availability of antenatal screening techniques.

(4) In Down Syndrome pregnancies, women over 35 years of age are more likely to have an amniocentesis and a termination of pregnancy compared with those under 35 years of age.

(5) Antenatal detection of Down Syndrome appears to alter the course of events in the pregnancy in many but not all cases.

CASES UNDERGOING ANTENATAL KARYOTYPING AND TERMINATION OF PREGNANCY, BY MATERNAL AGE.

	Maternal age		
	Under 35 yrs at end of pregnancy	35 yrs + at end of end of pregnancy	significance on Chi square testing
Antenatal karyotyping undertaken	14/62 (23%)	27/52 (52%)	p<0.005
Pregnancy terminated	18/62 (29%)	34/52 (65%)	p<0.001

Data tables

TABLE 1
Confirmed CASES of congenital anomaly reported to CARIS, for babies/fetuses with pregnancy ending in 1998/1999 by ONS malformation groups. Numbers for Wales and gross rates per 10,000 live/still births.

TABLE 2
Confirmed congenital ANOMALIES reported to CARIS, for babies/fetuses with pregnancy ending in 1998/1999 by ONS malformation groups. Numbers for Wales and gross rates per 10,000 live/still births.

TABLE 3
Cases reported to CARIS with dates of end of pregnancy 1998/1999, by specific conditions. Rates, pattern of malformations and outcome for the baby/fetus.

TABLE 4(A)
Confirmed cases and anomalies reported to CARIS with pregnancy ending in 1998. Numbers and gross rates per 10,000 livebirths/stillbirths, anomalies per case and outcome for Wales, Welsh Health Authorities and Unitary Authorities.

TABLE 4(B)
Confirmed cases and anomalies reported to CARIS with pregnancy ending in 1999. Numbers and gross rates per 10,000 livebirths/stillbirths, anomalies per case and outcome for Wales, Welsh Health Authorities and Unitary Authorities.

TABLE 5
Variations in Congenital Anomalies by Welsh Health and Unitary Authorities (combined data for 1998 & 1999). Gross case and anomaly rates, anomalies per case, pattern of anomalies and outcome for baby/fetus.

TABLE 6
All confirmed congenital anomalies reported to CARIS, for babies/fetuses with pregnancy ending in 1998 and 1999 so far by ONS malformation groups used in CUSUM surveillance. Numbers for Wales and Welsh Health Authorities.

TABLE 1

Confirmed CASES of congenital anomaly reported to CARIS, for babies/fetuses with pregnancy ending in 1998/1999 by ONS malformation groups. Numbers for Wales and gross rate* per 10,000 live/still births.

	ONS Congenital Malformation Group	1998 - initial data reported in 1998 Annual Report		1998 - updated data to end of first year of life		1999 – data so far	
		number	*gross rate**	number	*gross rate**	number	*gross rate**
A	**Central Nervous System**	**102**	**30.3**	**111**	**33.0**	**95**	**29.4**
OA	Anencephalus	25	7.4	28	8.3	23	7.1
OB	Spina Bifida	25	7.4	26	7.7	21	6.5
OC	Congenital Hydrocephalus	28	8.3	28	8.3	27	8.4
OE	Encephalocele	7	2.1	7	2.1	11	3.4
OF	Other CNS	40	11.9	44	13.1	31	9.6
B	**Eye and Ear**	**76**	**22.6**	**88**	**26.2**	**64**	**19.8**
1B	Congenital Lens Anomalies	8	2.4	13	3.9	2	0.6
1C	Other & Unspecified Eye Anomalies	17	5.1	22	6.5	27	8.4
1D	Ear, All	57	17.0	61	18.1	45	13.9
1E	Other Anophthalmos	8	2.4	8	2.4	4	1.2
C	**Alimentary System**	**173**	**51.5**	**175**	**52.1**	**153**	**47.4**
2A	Cleft of Palate Only	29	8.6	32	9.5	29	9.0
2B	Cleft of Lip Only	14	4.2	15	4.5	10	3.1
2C	Cleft Palate with Cleft Lip	26	7.7	27	8.0	24	7.4
2D	Tracheo-oesophageal Fistula/Stenosis	9	2.7	9	2.7	12	3.7
2E	Atresia/Stenosis of Large Intestine, Rectum & Anal Canal	16	4.8	16	4.8	16	5.0
2F	Other or Unspecified Anomalies of Alimentary System	106	31.5	90	26.8	81	25.1
D	**Cardiovascular System**	**256**	**76.2**	**274**	**81.5**	**196**	**60.7**
3A	Tetralogy of Fallot	9	2.7	11	3.3	8	2.5
3B	Ventricular Septal Defect	94	28.0	103	30.6	74	22.9
3C	Other Septal Defects	92	27.4	97	28.9	70	21.7
3E	Patent Ductus Arteriosus	41	12.2	37	11.0	23	7.1
3F	Anomalies of the Umbilical Artery	27	8.0	28	8.3	20	6.2
3G	Other Congenital Cardiac or Great Vessel Anomalies	109	32.4	116	34.5	94	29.1
3H	Congenital Anomalies of Other Vessels	7	2.1	7	2.1	12	3.7
E	**Respiratory System**	**78**	**23.2**	**84**	**25.0**	**67**	**20.8**
4A	Congenital Anomalies of the Respiratory System	78	23.2	84	25.0	67	20.8
F	**Urogenital System**	**177**	**52.7**	**204**	**60.7**	**192**	**59.5**
5A	Hypospadias/Epispadias	25	7.4	30	8.9	29	9.0
5B	Other Anomalies of the Male Genitalia	13	3.9	15	4.5	15	4.6
5C	Anomalies of the Female Genitalia	10	3.0	9	2.7	9	2.8
5D	Bladder Extrophy	0	0.0	1	0.3	4	1.2
5E	Renal Agenesis/Hypoplasia	38	11.3	37	11.0	21	6.5
5F	Other or Unspecified Defects of Urogenital System	30	8.9	37	11.0	35	10.8
5G	Indeterminate Sex	2	0.6	2	0.6	0	0.0
5H	Cystic Kidney Disease	28	8.3	33	9.8	31	9.6
5J	Congenital Obstructive Defects of Renal Pelvis or Anomalies of Ureter	80	23.8	100	29.8	91	28.2
G	**Limbs**	**189**	**56.2**	**208**	**61.9**	**174**	**53.9**
6A	Polydactyly/Syndactyly	56	16.7	62	18.4	58	18.0
6B	Limb Reductions	34	10.1	36	10.7	35	10.8
6C	Deformity of Feet	83	24.7	93	27.7	68	21.1
6D	Dislocation of Hip(s)	13	3.9	15	4.5	22	6.8
6E	Other Limb or Limb Girdles	48	14.3	49	14.6	45	13.9
H	**Other Musculoskeletal**	**202**	**60.1**	**212**	**63.1**	**185**	**57.3**
7A	Other Anomalies of the Diaphragm	6	1.8	5	1.5	3	0.9
7B	Anomalies of the Face, Skull or Neck	47	14.0	51	15.2	36	11.2
7C	Other Musculoskeletal Anomalies of the Thorax or Neck	0	0.0	0	0.0	1	0.3
7D	Osteodystrophy or Chondrodystrophy	5	1.5	4	1.2	5	1.5
7E	Other or Unspecified Anomalies of the Musculoskeletal System	73	21.7	75	22.3	71	22.0
7F	Anomalies of the Abdominal Wall	7	2.1	10	3.0	14	4.3
7G	Exomphalos	14	4.2	15	4.5	7	2.2
7H	Anomalies of the Lips, Tongue & Pharynx	11	3.3	13	3.9	13	4.0
7K	Congenital Diaphragmatic Hernia	9	2.7	9	2.7	12	3.7
7L	Gastroschisis	16	4.8	16	4.8	19	5.9
7M	Prune Belly Syndrome	1	0.3	1	0.3	0	0.0
7P	Other Anomalies of Face and Neck	75	22.3	79	23.5	75	23.2
I	**Skin & Integument**	**51**	**15.2**	**54**	**16.1**	**49**	**15.2**
8C	Anomalies of the Skin or Integument	51	15.2	54	16.1	49	15.2
J	**Chromosomal Anomalies**	**151**	**44.9**	**163**	**48.5**	**143**	**44.3**
9C	Trisomy 21 - Down Syndrome	70	20.8	70	20.8	55	17.0
9D	Other Congenital Anomalies	83	24.7	95	28.3	89	27.6
K	**Other Anomalies**	**122**	**36.3**	**136**	**40.5**	**131**	**40.6**
9A	Congenital Neoplasms (other than benign skin)	5	1.5	5	1.5	2	0.6
9B	Endocrine and Metabolic Disorders	33	9.8	36	10.7	27	8.4
9E	Other & Unspecified Congenital Anomalies	84	25.0	93	27.7	100	31.0
9H	Congenital Infections	3	0.9	5	1.5	4	1.2
All defects		**849**	**252.6**	**944**	**280.9**	**786**	**243.6**

** Gross rate is calculated as total number of cases reported to CARIS, divided by number of total births (livebirths & stillbirths) for given year (derived from ONS birth data)*

	ONS Congenital Malformation Group	1998 - initial data reported in 1998 Annual Report		1998 - updated data to end of first year of life		1999 – data so far	
		number	*gross rate**	number	*gross rate**	number	*gross rate**
A	**Central Nervous System**	**131**	**39.0**	**141**	**42.0**	**122**	**37.8**
OA	Anencephalus	25	7.4	28	8.3	23	7.1
OB	Spina Bifida	27	8.0	28	8.3	21	6.5
OC	Congenital Hydrocephalus	29	8.6	30	8.9	27	8.4
OE	Encephalocele	7	2.1	7	2.1	11	3.4
OF	Other CNS	43	12.8	48	14.3	40	12.4
B	**Eye and Ear**	**94**	**28.0**	**112**	**33.3**	**85**	**26.3**
1B	Congenital Lens Anomalies	8	2.4	13	3.9	2	0.6
1C	Other & Unspecified Eye Anomalies	21	6.2	27	8.0	29	9.0
1D	Ear, All	57	17.0	64	19.0	50	15.5
1E	Other Anophthalmos	8	2.4	8	2.4	4	1.2
C	**Alimentary System**	**217**	**64.6**	**206**	**61.3**	**189**	**58.6**
2A	Cleft of Palate Only	29	8.6	32	9.5	29	9.0
2B	Cleft of Lip Only	14	4.2	15	4.5	10	3.1
2C	Cleft Palate with Cleft Lip	26	7.7	27	8.0	24	7.4
2D	Tracheo-oesophageal Fistula/Stenosis	9	2.7	9	2.7	12	3.7
2E	Atresia/Stenosis of Large Intestine, Rectum & Anal Canal	16	4.8	16	4.8	18	5.6
2F	Other or Unspecified Anomalies of Alimentary System	123	36.6	107	31.8	96	29.8
D	**Cardiovascular System**	**459**	**136.6**	**481**	**143.1**	**350**	**108.5**
3A	Tetralogy of Fallot	9	2.7	11	3.3	8	2.5
3B	Ventricular Septal Defect	94	28.0	103	30.6	74	22.9
3C	Other Septal Defects	95	28.3	101	30.1	71	22.0
3E	Patent Ductus Arteriosus	41	12.2	37	11.0	23	7.1
3F	Anomalies of the Umbilical Artery	27	8.0	28	8.3	20	6.2
3G	Other Congenital Cardiac or Great Vessel Anomalies	186	55.3	194	57.7	142	44.0
3H	Congenital Anomalies of Other Vessels	7	2.1	7	2.1	12	3.7
E	**Respiratory System**	**92**	**27.4**	**98**	**29.2**	**81**	**25.1**
4A	Congenital Anomalies of the Respiratory System	92	27.4	98	29.2	81	25.1
F	**Urogenital System**	**253**	**75.3**	**300**	**89.3**	**276**	**85.5**
5A	Hypospadias/Epispadias	26	7.7	32	9.5	34	10.5
5B	Other Anomalies of the Male Genitalia	13	3.9	16	4.8	17	5.3
5C	Anomalies of the Female Genitalia	10	3.0	9	2.7	11	3.4
5D	Bladder Exstrophy	0	0.0	1	0.3	5	1.5
5E	Renal Agenesis/Hypoplasia	43	12.8	38	11.3	21	6.5
5F	Other or Unspecified Defects of Urogenital System	34	10.1	43	12.8	39	12.1
5G	Indeterminate Sex	2	0.6	2	0.6	0	0.0
5H	Cystic Kidney Disease	29	8.6	34	10.1	32	9.9
5J	Congenital Obstructive Defects of Renal Pelvis or Anomalies of Ureter	96	28.6	125	37.2	117	36.3
G	**Limbs**	**278**	**82.7**	**302**	**89.9**	**271**	**84.0**
6A	Polydactyly/Syndactyly	65	19.3	72	21.4	72	22.3
6B	Limb Reductions	60	17.9	63	18.7	54	16.7
6C	Deformity of Feet	85	25.3	95	28.3	72	22.3
6D	Dislocation of Hip(s)	13	3.9	15	4.5	22	6.8
6E	Other Limb or Limb Girdles	55	16.4	57	17.0	51	15.8
H	**Other Musculoskeletal**	**291**	**86.6**	**314**	**93.4**	**293**	**90.8**
7A	Other Anomalies of the Diaphragm	6	1.8	5	1.5	3	0.9
7B	Anomalies of the Face, Skull or Neck	54	16.1	61	18.1	49	15.2
7C	Other Musculoskeletal Anomalies of the Thorax or Neck	0	0.0	0	0.0	1	0.3
7D	Osteodystrophy or Chondrodystrophy	5	1.5	4	1.2	5	1.5
7E	Other or Unspecified Anomalies of the Musculoskeletal System	87	25.9	95	28.3	87	27.0
7F	Anomalies of the Abdominal Wall	8	2.4	11	3.3	14	4.3
7G	Exomphalos	14	4.2	15	4.5	7	2.2
7H	Anomalies of the Lips, Tongue & Pharynx	11	3.3	13	3.9	14	4.3
7K	Congenital Diaphragmatic Hernia	9	2.7	9	2.7	12	3.7
7L	Gastroschisis	16	4.8	16	4.8	19	5.9
7M	Prune Belly Syndrome	1	0.3	1	0.3	0	0.0
7P	Other Anomalies of Face and Neck	80	23.8	84	25.0	82	25.4
I	**Skin & Integument**	**52**	**15.5**	**55**	**16.4**	**50**	**15.5**
8C	Anomalies of the Skin or Integument	52	15.5	55	16.4	50	15.5
J	**Chromosomal Anomalies**	**155**	**46.1**	**167**	**49.7**	**145**	**44.9**
9C	Trisomy 21 - Down Syndrome	70	20.8	70	20.8	55	17.0
9D	Other Congenital Anomalies	85	25.3	97	28.9	90	27.9
K	**Other Anomalies**	**137**	**40.8**	**158**	**47.0**	**154**	**47.7**
9A	Congenital Neoplasms (other than benign skin)	5	1.5	5	1.5	2	0.6
9B	Endocrine and Metabolic Disorders	34	10.1	37	11.0	27	8.4
9E	Other & Unspecified Congenital Anomalies	93	27.7	109	32.4	121	37.5
9H	Congenital Infections	5	1.5	7	2.1	4	1.2
All defects		**2159**	**642.4**	**2334**	**694.4**	**2016**	**624.8**

TABLE 2
Confirmed congenital anomalies reported to CARIS, for babies/fetuses with pregnancy ending in 1998/1999 by ONS malformation groups. Numbers for Wales and gross rates* per 10,000 live/still births.

** Gross rate is calculated as total number of cases reported to CARIS, divided by number of total births (livebirths & stillbirths) for given year (derived from ONS birth data)*

TABLE 3

Cases reported to CARIS with dates of end of pregnancy 1998/1999, by specific conditions. Rates, pattern of malformations and outcome for the baby/fetus.

ONS Congenital Malformation Group	Total Number of cases	Gross rate of all cases reported per 10,000 livebirths/ stillbirths			associated anomalies reported: single anomaly reported		other anomalies confined to to this body system		multiple anomalies involving other body systems		associated with syndrome	
		rate	L95%CI	U95%CI	no.	%	no.	%	no.	%	no.	%
A Central Nervous System												
All neural tube defects	115	17.5	14.3	20.6	39	34	11	10	51	44	14	12
Anencephalus	51	7.7	5.6	9.9	23	45	1	2	26	51	1	2
Spina Bifida	47	7.1	5.1	9.2	14	30	7	15	17	36	9	19
Encephalocele	18	2.7	1.5	4.0	2	11	4	22	8	44	4	22
Congenital Hydrocephalus	55	8.3	6.1	10.6	10	18	4	7	21	38	20	36
B Eye and Ear												
Anophthalmos/microphthalmos	12	1.8	0.8	2.9	2	17	2	17	2	17	6	50
Congenital cataracts	13	2.0	0.9	3.0	8	62	0	0	2	15	3	23
Sensori-neural hearing loss	9	1.4	0.5	2.3	3	33	0	0	4	44	2	22
C Alimentary System												
Cleft lip/palate	137	20.8	17.3	24.3	65	47	2	1	24	18	46	34
Cleft palate only	61	9.3	6.9	11.6	17	28	1	2	14	23	29	48
Cleft lip only	25	3.8	2.3	5.3	20	80	0	0	1	4	4	16
Cleft lip and palate	51	7.7	5.6	9.9	28	55	1	2	9	18	13	25
Pyloric stenosis	39	5.9	4.1	7.8	31	79	0	0	8	21	0	0
Large bowel atresia	32	4.9	3.2	6.5	8	25	1	3	10	31	13	41
Duodenal atresia	11	1.7	0.7	2.7	4	36	1	9	2	18	4	36
Other small bowel atresia	9	1.4	0.5	2.3	1	11	0	0	6	67	2	22
Tracheo-oesophageal fistula/stenosis	21	3.2	1.8	4.6	6	29	1	5	6	29	8	38
Hirschsprung's disease	5	0.8	0.1	1.4	4	80	0	0	0	0	1	20
D Cardiovascular System												
Patent ducus arteriosus (>37 weeks)	60	9.1	6.8	11.4	5	8	22	37	15	25	18	30
Atrial septal defects	130	19.7	16.3	23.1	12	9	44	34	37	28	37	28
Ventricular septal defects	177	26.9	22.9	30.8	50	28	46	26	30	17	51	29
Tetralogy of Fallot	19	2.9	1.6	4.2	10	53	1	5	6	32	2	11
Transposition of great arteries	25	3.8	2.3	5.3	2	8	13	52	6	24	4	16
Pulmonary artery stenosis	39	5.9	4.1	7.8	6	15	16	41	5	13	12	31
Aortic stenosis	7	1.1	0.3	1.8	3	43	2	29	0	0	2	29
Aortic valve stenosis	5	0.8	0.1	1.4	1	20	2	40	0	0	2	40
Co-arctation of aorta	17	2.6	1.4	3.8	4	24	5	29	2	12	6	35
Hypoplastic left heart syndrome	22	3.3	1.9	4.7	10	45	4	18	3	14	5	23
Atrioventricular septal defect	41	6.2	4.3	8.1	3	7	5	12	6	15	27	66
Mitral stenosis/atresia	5	0.8	0.1	1.4	0	0	1	20	1	20	3	60
Tricuspid stenosis/atresia	8	1.2	0.4	2.1	1	13	6	75	0	0	1	13
E Respiratory System												
Agenesis of lung	24	3.6	2.2	5.1	0	0	0	0	8	33	16	67
Hypoplasia of lung	83	12.6	9.9	15.3	3	4	0	0	39	47	41	49
F Urogenital System												
Pelvi-ureteric obstruction (all forms)	180	27.3	23.3	31.3	101	56	45	25	29	16	5	3
Renal agenesis - bilateral	11	1.7	0.7	2.7	0	0	1	9	5	45	5	45
Renal agenesis - unilateral	18	2.7	1.5	4.0	3	17	1	6	9	50	5	28
Hypospadias (male)	57	8.7	6.4	10.9	40	70	8	14	7	12	2	4
Epispadias	2	0.3	-0.1	0.7	0	0	1	50	1	50	0	0
Cystic kidney disease (all forms)	64	9.7	7.3	12.1	16	25	7	11	21	33	20	31
G Limbs												
Polydactyly, upper limb	47	7.1	5.1	9.2	21	45	7	15	5	11	14	30
Polydactyly, lower limb	26	3.9	2.4	5.5	9	35	7	27	4	15	6	23
Polydactyly, nos	9	1.4	0.5	2.3	5	56	0	0	1	11	3	33
Syndactyly, upper limb	16	2.4	1.2	3.6	0	0	4	25	2	13	10	63
Syndactyly, lower limb	35	5.3	3.6	7.1	12	34	6	17	5	14	12	34
Syndactyly, nos	11	1.7	0.7	2.7	2	18	0	0	1	9	8	73
Limb reductions, upper limb	58	8.8	6.5	11.1	10	17	4	7	15	26	29	50
Limb reductions, lower limb	21	3.2	1.8	4.6	0	0	2	10	8	38	11	52
Limb reductions, nos	5	0.8	0.1	1.4	0	0	1	20	3	60	1	20
Talipes (non postural)	125	19.0	15.7	22.3	52	42	3	2	34	27	36	29
Congenital dislocation of hip(s)	33	5.0	3.3	6.7	27	82	1	3	5	15	0	0
H Other Musculoskeletal												
Gastroschisis	35	5.3	3.6	7.1	29	83	1	3	4	11	1	3
Exomphalos	22	3.3	1.9	4.7	3	14	1	5	6	27	12	55
Congenital constriction bands	18	2.7	1.5	4.0	0	0	0	0	4	22	14	78
Congenital diaphragmatic hernia	21	3.2	1.8	4.6	10	48	0	0	9	43	2	10
I Skin & Integument												
Cystic hygroma	45	6.8	4.8	8.8	6	13	0	0	9	20	30	67
J Chromosomal Anomalies												
Down Syndrome – trisomy	125	19.0	15.7	22.3	0	0	0	0	0	0	125	100
Edward's syndrome	32	4.9	3.2	6.5	0	0	0	0	0	0	32	100
Patau syndrome	15	2.3	1.1	3.4	0	0	0	0	0	0	15	100
* Triploidy/polyploidy	23	3.5	2.1	4.9	0	0	0	0	0	0	23	100
* Turner's syndrome	23	3.5	2.1	4.9	0	0	0	0	0	0	23	100
Klinefelter's syndrome	8	1.2	0.4	2.1	0	0	0	0	0	0	8	100
Chomosomal deletions	20	3.0	1.7	4.4	0	0	0	0	0	0	20	100
K Other Anomalies												
Hypothyroidism	19	2.9	1.6	4.2	18	95	0	0	0	0	1	5
Congenital adrenal hyperplasia	5	0.8	0.1	1.4	4	80	0	0	0	0	1	20
Phenylketonuria	9	1.4	0.5	2.3	8	89	1	11	0	0	0	0
Congenital adrenal hypoplasia	30	4.6	2.9	6.2	1	3	0	0	18	60	11	37
Cystic fibrosis	18	2.7	1.5	4.0	18	100	0	0	0	0	0	0
Neoplasms (other than benign skin)	7	1.1	0.3	1.8	5	71	0	0	1	14	1	14
Congenital infections	8	1.2	0.4	2.1	4	50	3	38	1	13	0	0

** Outcome of pregnancy unknown for one case*

reported outcome for case									
spontaeneous fetal loss		termination of pregnancy		stillbirth		neonatal or infant death		survivors	
no.	%	no.	%	no.	%	no.	%	no.	%
3	3	98	85	2	2	4	3	7	6
1	2	46	90	1	2	3	6	0	0
1	2	40	85	0	0	0	0	6	13
1	6	14	78	1	6	1	6	1	6
1	2	30	55	4	7	10	18	10	18
1	8	2	17	1	8	3	25	5	42
0	0	0	0	0	0	1	8	12	92
0	0	0	0	0	0	1	11	8	89
15	11	25	18	2	1	5	4	90	66
8	13	14	23	1	2	3	5	35	57
0	0	2	8	0	0	1	4	22	88
7	14	9	18	1	2	1	2	33	65
0	0	0	0	0	0	0	0	39	100
3	9	9	28	1	3	6	19	13	41
0	0	2	18	0	0	2	18	7	64
0	0	3	33	0	0	2	22	4	44
0	0	6	29	1	5	1	5	13	62
0	0	0	0	0	0	0	0	5	100
0	0	0	0	1	2	11	18	48	80
0	0	15	12	1	1	14	11	100	77
4	2	30	17	2	1	13	7	128	72
0	0	0	0	0	0	2	11	17	89
1	4	9	36	0	0	1	4	14	56
1	3	8	21	0	0	1	3	29	74
0	0	0	0	0	0	1	14	6	86
0	0	1	20	0	0	1	20	3	60
0	0	0	0	0	0	1	6	16	94
0	0	15	68	1	5	4	18	2	9
1	2	20	49	2	5	6	15	12	29
0	0	2	40	0	0	1	20	2	40
0	0	0	0	0	0	2	25	6	75
4	17	15	63	4	17	1	4	0	0
8	10	48	58	12	14	13	16	2	2
1	1	9	5	2	1	4	2	164	91
1	9	8	73	2	18	0	0	0	0
0	0	8	44	1	6	1	6	8	44
0	0	1	2	0	0	1	2	55	96
0	0	0	0	0	0	0	0	2	100
2	3	29	45	0	0	5	8	28	44
2	4	8	17	1	2	5	11	31	66
0	0	6	23	0	0	1	4	19	73
0	0	3	33	0	0	0	0	6	67
4	25	2	13	2	13	0	0	8	50
2	6	5	14	3	9	3	9	22	63
2	18	4	36	0	0	2	18	3	27
6	10	19	33	5	9	3	5	25	43
2	10	9	43	2	10	4	19	4	19
0	0	0	0	0	0	1	20	4	80
6	5	40	32	3	2	8	6	68	54
0	0	0	0	0	0	1	3	32	97
1	3	4	11	3	9	0	0	27	77
3	14	10	45	0	0	4	18	5	23
6	33	7	39	0	0	0	0	5	28
0	0	8	38	0	0	4	19	9	43
10	22	31	69	0	0	0	0	4	9
4	3	52	42	2	2	6	5	61	49
0	0	22	69	2	6	6	19	2	6
3	20	9	60	0	0	2	13	1	7
11	48	9	39	0	0	1	4	1	4
6	26	11	48	0	0	1	4	4	17
0	0	4	50	0	0	0	0	4	50
1	5	6	30	0	0	1	5	12	60
0	0	0	0	0	0	0	0	19	100
0	0	0	0	0	0	1	20	4	80
0	0	0	0	0	0	0	0	9	100
3	10	21	70	4	13	2	7	0	0
0	0	0	0	0	0	0	0	18	100
0	0	2	29	0	0	0	0	5	71
2	25	2	25	0	0	2	25	3	38

TABLE 4(A)

Confirmed cases and anomalies reported to CARIS with pregnancy ending in 1998. Numbers and gross rates* per 10,000 livebirths/stillbirths, anomalies per case and outcome for Wales, Welsh Health Authorities and Unitary Authorities.

Authority Area	total births (ONS birth data)	Gross case rate*/10,000 total births			
		cases	gross rate	L95%CI	U95%CI
Wales	**33610**	**944**	**280.9**	**263.2**	**298.5**
Bro Taf	**9032**	**305**	**337.7**	**300.4**	**374.9**
Cardiff	4063	170	418.4	356.8	480.0
Merthyr Tydfil	709	14	197.5	95.1	299.9
Rhondda Cynon Taff	2824	62	219.5	165.5	273.6
Vale of Glamorgan	1436	59	410.9	308.2	513.5
Dyfed Powys	**4930**	**137**	**277.9**	**232.0**	**323.8**
Carmarthenshire	1745	54	309.5	228.2	390.7
Ceredigion	649	16	246.5	127.2	365.8
Pembrokeshire	1277	30	234.9	151.9	318.0
Powys	1259	37	293.9	200.6	387.2
Gwent	**6707**	**139**	**207.2**	**173.2**	**241.3**
Blaenau Gwent	830	21	253.0	146.2	359.8
Caerphilly	2154	42	195.0	136.6	253.4
Monmouthshire	868	13	149.8	69.0	230.6
Newport	1769	42	237.4	166.5	308.4
Torfaen	1086	21	193.4	111.5	275.3
Morgannwg	**5399**	**189**	**350.1**	**301.0**	**399.1**
Bridgend	1508	24	159.2	96.0	222.3
Neath Port Talbot	1455	49	336.8	244.1	429.5
Swansea	2436	116	476.2	391.6	560.8
North Wales	**7542**	**174**	**230.7**	**196.8**	**264.6**
Conwy	1178	31	263.2	171.7	354.6
Denbighshire	1037	23	221.8	132.2	311.4
Flintshire	1795	38	211.7	145.1	278.3
Gwynedd	1305	32	245.2	161.3	329.1
Isle of Anglesey	732	14	191.3	92.0	290.5
Wrexham	1495	36	240.8	163.1	318.5

*Gross rate is calculated as total number of cases reported to CARIS, divided by number of total births (livebirths & stillbirths) for given year (derived from ONS birth data)

TABLE 4(B)

Confirmed cases and anomalies reported to CARIS with pregnancy ending in 1999. Numbers and gross rates* per 10,000 livebirths/stillbirths, anomalies per case and outcome for Wales, Welsh Health Authorities and Unitary Authorities.

Authority Area	total births (ONS birth data)	Gross case rate*/10,000 total births			
		cases	gross rate	L95%CI	U95%CI
Wales	**32266**	**786**	**243.6**	**226.8**	**260.4**
Bro Taf	**8411**	**226**	**268.7**	**234.1**	**303.3**
Cardiff	3723	136	365.3	305.0	425.6
Merthyr Tydfil	664	14	210.8	101.6	320.1
Rhondda Cynon Taff	2655	38	143.1	97.9	188.3
Vale of Glamorgan	1369	38	277.6	190.6	364.6
Dyfed Powys	**4868**	**130**	**267.1**	**221.8**	**312.3**
Carmarthenshire	1730	55	317.9	235.2	400.6
Ceredigion	622	12	192.9	84.8	301.0
Pembrokeshire	1220	30	245.9	159.0	332.8
Powys	1296	33	254.6	168.9	340.4
Gwent	**6421**	**136**	**211.8**	**176.6**	**247.0**
Blaenau Gwent	818	17	207.8	110.1	305.6
Caerphilly	2066	44	213.0	150.7	275.2
Monmouthshire	837	16	191.2	98.4	283.9
Newport	1676	42	250.6	175.8	325.4
Torfaen	1024	17	166.0	87.8	244.3
Morgannwg	**5430**	**143**	**263.4**	**220.8**	**305.9**
Bridgend	1538	28	182.1	115.2	248.9
Neath Port Talbot	1374	42	305.7	214.7	396.7
Swansea	2518	73	289.9	224.4	355.4
North Wales	**7136**	**151**	**211.6**	**178.2**	**245.0**
Conwy	1090	14	128.4	61.6	195.3
Denbighshire	930	25	268.8	164.9	372.8
Flintshire	1714	39	227.5	156.9	298.1
Gwynedd	1277	24	187.9	113.5	262.4
Isle of Anglesey	729	9	123.5	43.3	203.6
Wrexham	1396	40	286.5	199.0	374.0

*Gross rate is calculated as total number of cases reported to CARIS, divided by number of total births (livebirths & stillbirths) for given year (derived from ONS birth data)

Gross Anomaly rate/10,000 total births					survival rates			
anomalies	gross rate	L95%CI	U95%CI	anomalies/ case	survivors	% of cases	L95%CI	U95%CI
2334	649.4	676.8	712.1	2.5	646	68	65	71
734	812.7	775.4	849.9	2.4	217	71	66	76
391	962.3	900.8	1023.9	2.3	125	74	67	80
38	536.0	433.6	638.4	2.7	7	50	24	76
172	609.0	555.0	663.1	2.8	37	60	47	72
133	926.2	823.5	1028.8	2.3	48	81	71	91
285	578.1	532.2	624.0	2.1	104	76	69	83
133	762.2	680.9	843.4	2.5	39	72	60	84
22	339.0	219.7	458.3	1.4	13	81	62	100
74	579.5	496.4	662.6	2.5	22	73	58	89
56	444.8	351.5	538.1	1.5	30	81	68	94
305	454.7	420.7	488.8	2.2	88	63	55	71
53	638.6	531.7	745.4	2.5	12	57	36	78
67	311.0	252.7	369.4	1.6	29	69	55	83
31	357.1	276.3	437.9	2.4	9	69	44	94
87	491.8	420.9	562.8	2.1	29	69	55	83
67	616.9	535.0	698.8	3.2	9	43	22	64
488	903.9	854.8	952.9	2.6	133	70	64	77
60	397.9	334.7	461.0	2.5	15	63	43	82
135	927.8	835.1	1020.5	2.8	34	69	56	82
293	1202.8	1118.2	1287.4	2.5	84	72	64	81
552	692.1	658.2	726.0	3.0	104	60	52	67
76	645.2	553.7	736.6	2.5	14	45	28	63
68	655.7	566.1	745.4	3.0	14	61	41	81
122	679.7	613.1	746.3	3.2	25	66	51	81
110	842.9	759.0	926.8	3.4	19	59	42	76
55	751.4	652.1	850.6	3.9	9	64	39	89
91	608.7	531.0	686.4	2.5	23	64	48	80

Gross Anomaly rate/10,000 total births					survival rates			
anomalies	gross rate	L95%CI	U95%CI	anomalies/ case	survivors	% of cases	L95%CI	U95%CI
2016	624.8	608.0	641.6	2.6	544	69	66	72
590	701.5	666.9	736.0	2.6	159	70	64	76
285	765.5	705.2	825.8	2.1	105	77	70	84
60	903.6	794.3	1012.9	4.3	9	64	39	89
148	557.4	512.3	602.6	3.9	20	53	37	69
97	708.5	621.5	795.6	2.6	25	66	51	81
300	616.3	571.0	661.6	2.3	94	72	65	80
147	849.7	767.0	932.4	2.7	38	69	57	81
28	450.2	342.1	558.3	2.3	11	92	76	107
56	459.0	372.1	545.9	1.9	21	70	54	86
69	532.4	446.6	618.2	2.1	24	73	58	88
372	579.3	544.1	614.6	2.7	80	59	51	67
47	574.6	476.8	672.3	2.8	9	53	29	77
116	561.5	499.2	623.7	2.6	30	68	54	82
55	657.1	564.3	749.9	3.4	5	31	9	54
109	650.4	575.5	725.5	2.6	26	62	47	77
45	439.5	361.2	517.7	2.6	10	59	35	82
330	607.7	565.1	650.3	2.3	103	72	65	79
88	572.2	505.4	639.0	3.1	19	68	51	85
88	640.5	549.4	731.5	2.1	34	81	69	93
154	611.6	546.1	677.1	2.1	50	68	58	79
424	594.2	560.8	627.6	2.8	108	72	64	79
54	495.4	428.6	562.3	3.9	6	43	17	69
91	978.5	874.5	1082.4	3.6	14	56	37	75
90	525.1	454.5	595.7	2.3	26	67	52	81
61	477.7	403.2	552.2	2.5	17	71	53	89
16	219.5	139.3	299.6	1.8	6	67	36	97
112	802.3	714.8	889.8	2.8	26	65	50	80

TABLE 5

Variations in congenital anomalies by Welsh Health and Unitary Authorities (combined data for 1998 & 1999). Gross case and anomaly rates, anomalies per case, pattern of anomalies

Authority Area	cases	gross case rate*/10,000 total births			gross anomaly rate*/10,000 total births			pattern of anomalies: case has single anomaly		pattern of anomalies: case has multiple anomalies, but no syndrome	
		rate	L95%CI	U95%CI	rate	L95%CI	U95%CI	no.	%	no.	%
Wales	**1730**	**262.5**	**250.3**	**274.7**	**660.2**	**648.0**	**672.4**	**826**	**47.7**	**492**	**28.4**
Bro Taf	**531**	**304.4**	**278.9**	**329.9**	**759.0**	**733.5**	**784.5**	**252**	**47.5**	**154**	**29.0**
Cardiff	306	393.0	349.9	436.2	868.2	825.1	911.4	154	50.3	80	26.1
Merthyr Tydfil	28	203.9	129.2	278.7	713.8	639.0	788.5	10	35.7	11	39.3
• Rhondda Cynon Taff	100	182.5	147.1	218.0	584.0	548.6	619.5	34	34.0	42	42.0
Vale of Glamorgan	97	345.8	278.2	413.4	820.0	752.3	887.6	54	55.7	21	21.6
Dyfed Powys	**267**	**272.5**	**240.3**	**304.7**	**597.1**	**564.8**	**629.3**	**137**	**51.3**	**70**	**26.2**
Carmarthenshire	109	313.7	255.7	371.6	805.8	747.8	863.7	49	45.0	31	28.4
Ceredigion	28	220.3	139.6	301.0	393.4	312.7	474.1	18	64.3	5	17.9
Pembrokeshire	60	240.3	180.2	300.4	520.6	460.6	580.7	32	53.3	17	28.3
Powys	70	274.0	210.7	337.3	489.2	425.9	552.5	38	54.3	17	24.3
Gwent	**275**	**209.5**	**185.0**	**234.0**	**515.7**	**491.2**	**540.2**	**134**	**48.7**	**74**	**26.9**
Blaenau Gwent	38	230.6	158.1	303.0	606.8	534.3	679.3	19	50.0	7	18.4
Caerphilly	86	203.8	161.2	246.4	433.6	391.0	476.3	46	53.5	23	26.7
Monmouthshire	29	170.1	108.7	231.5	504.0	443.0	565.8	8	27.6	13	44.8
Newport	84	243.8	192.3	295.3	568.9	517.4	620.4	47	56.0	19	22.6
• Torfaen	38	180.1	123.4	236.8	530.8	474.1	587.5	14	36.8	12	31.6
Morgannwg	**332**	**305.7**	**273.2**	**338.1**	**754.5**	**722.0**	**786.9**	**161**	**48.6**	**97**	**29.2**
Bridgend	52	170.7	124.7	216.7	485.9	439.9	531.9	20	38.5	22	42.3
Neath Port Talbot	91	318.1	253.5	382.8	784.7	720.1	849.4	51	56.0	24	25.3
Swansea	189	381.5	328.2	434.9	902.3	849.0	955.6	90	47.6	52	27.5
North Wales	**325**	**221.4**	**197.6**	**245.2**	**644.5**	**620.7**	**688.3**	**142**	**43.7**	**97**	**29.8**
Conwy	45	198.4	141.0	255.8	573.2	515.8	630.6	20	44.4	10	22.2
Denbighshire	48	244.0	175.8	312.2	808.3	740.1	876.5	21	43.8	17	35.4
Flintshire	77	219.4	171.0	267.9	604.2	555.7	652.6	42	54.5	17	22.1
Gwynedd	56	216.9	160.7	273.1	662.3	606.1	718.5	18	32.1	22	39.3
Isle of Anglesey	23	157.4	93.6	221.3	486.0	422.1	549.8	4	17.4	9	39.1
Wrexham	76	262.9	204.6	321.2	702.2	643.9	760.5	37	48.7	22	28.9

** gross rate is calculated as total number of cases reported to CARIS, divided by number of total births (livebirths & stillbirths) for given year (derived from ONS birth data)*

• outcome of pregnancy not known for one case

reported		reported outcome for all cases									
cases associated with syndromes		spontaneous fetal loss		termination of pregnancy		stillbirth		neonatal or infant death		survivors	
no.	%	no.	%	no.	%	no.	%	no.	%	no.	%
411	**23.8**	**66**	**3.8**	**337**	**19.5**	**46**	**2.7**	**101**	**5.8**	**1177**	**68.1**
125	**23.5**	**23**	**4.3**	**90**	**16.9**	**16**	**3.0**	**25**	**4.7**	**376**	**70.8**
72	23.5	12	3.9	46	15.0	8	2.6	10	3.3	230	75.2
7	25.0	0	0	8	28.6	1	3.6	3	10.7	16	57.1
24	24.0	5	5.0	23	23.0	4	4.0	10	10.0	57	57.0
22	22.7	6	6.2	13	13.4	3	3.1	2	2.1	73	75.3
60	**22.5**	**5**	**1.9**	**39**	**14.6**	**9**	**3.4**	**16**	**6.0**	**198**	**74.2**
29	26.6	1	0.9	20	18.3	5	4.6	6	5.5	77	70.6
5	17.9	1	3.6	2	7.1	1	3.6	0	0	24	85.7
11	18.3	3	5.0	7	11.7	1	1.7	6	10.0	43	71.7
15	21.4	0	0	10	14.3	2	2.9	4	5.7	54	77.1
67	**24.4**	**18**	**6.5**	**65**	**23.6**	**8**	**2.9**	**15**	**5.5**	**168**	**61.1**
12	31.6	4	10.5	10	26.3	0	0	3	7.9	21	55.3
17	19.8	3	3.5	15	17.4	5	5.8	4	4.7	59	68.6
8	27.6	4	13.8	8	27.6	1	3.4	2	6.9	14	48.3
18	21.4	5	6.0	17	20.2	2	2.4	5	6.0	55	65.5
12	31.6	2	5.3	15	39.5	0	0	1	2.6	19	50.0
73	**22.0**	**12**	**3.6**	**57**	**17.2**	**8**	**2.4**	**18**	**5.4**	**236**	**71.1**
10	19.2	0	0	11	21.2	3	5.8	4	7.7	34	65.4
16	17.6	3	3.3	16	17.6	1	1.1	2	2.2	68	74.7
47	24.9	9	4.8	30	25.9	4	2.1	12	6.3	134	70.9
86	**26.5**	**8**	**2.5**	**86**	**26.5**	**5**	**1.5**	**27**	**8.3**	**199**	**61.2**
15	33.3	0	0	17	37.8	0	0	8	17.8	20	44.4
10	20.8	0	0	13	27.1	0	0	7	14.6	28	58.3
18	23.4	3	3.9	19	24.7	0	0	4	5.2	51	66.2
16	28.6	1	1.8	16	28.6	1	1.8	2	3.6	36	64.3
10	43.5	0	0	6	26.1	0	0	2	8.7	15	65.2
17	22.4	4	5.3	15	19.7	4	5.3	4	5.3	49	64.5

TABLE 6

All confirmed congenital anomalies reported to CARIS for babies/fetuses with pregnancy ending in 1998 and 1999 so far by ONS malformation groups used in CUSUM surveillance. Numbers for Wales and Welsh Health Authorities.

ONS Congenital Malformation Group		Bro Taf		Dyfed Powys		Gwent	
		1998	*1999*	1998	*1999*	1998	*1999*
A	**Central Nervous System**	**41**	**39**	**17**	**7**	**20**	**22**
OA	Anencephalus	6	4	3	2	7	4
OB	Spina Bifida	7	8	2	0	3	4
OC	Congenital Hydrocephalus	11	6	6	2	4	8
OE	Encephalocele	2	4	1	1	0	2
OF	Other CNS	15	17	5	2	6	4
B	**Eye and Ear**	**33**	**26**	**8**	**15**	**12**	**13**
1B	Congenital Lens Anomalies	5	2	3	0	0	0
1C	Other & Unspecified Eye Anomalies	8	8	0	7	1	3
1D	Ear, All	19	14	4	7	11	9
1E	Other Anophthalmos	1	2	1	1	0	1
C	**Alimentary System**	**56**	**31**	**24**	**33**	**20**	**51**
2A	Cleft of Palate Only	5	6	1	6	4	5
2B	Cleft of Lip Only	5	3	4	2	2	1
2C	Cleft Palate with Cleft Lip	10	4	3	4	5	7
2D	Tracheo-oesophageal Fistula/Stenosis	4	2	2	1	1	4
2E	Atresia/Stenosis of Large Intestine, Rectum & Anal Canal	3	0	2	3	3	10
2F	Other or Unspecified Anomalies of Alimentary System	29	16	12	17	5	24
D	**Cardiovascular System**	**145**	**110**	**75**	**63**	**63**	**50**
3A	Tetralogy of Fallot	4	0	0	1	4	1
3B	Ventricular Septal Defect	28	25	17	13	11	10
3C	Other Septal Defects	29	22	15	11	8	8
3E	Patent Ductus Arteriosus	16	12	4	6	2	0
3F	Anomalies of the Umbilical Artery	10	6	4	1	4	5
3G	Other Congenital Cardiac or Great Vessel Anomalies	56	42	35	30	31	22
3H	Congenital Anomalies of Other Vessels	2	3	0	1	3	4
E	**Respiratory System**	**33**	**27**	**13**	**13**	**11**	**13**
4A	Congenital Anomalies of the Respiratory System	33	27	13	13	11	13
F	**Urogenital System**	**136**	**93**	**29**	**32**	**34**	**48**
5A	Hypospadias/Epispadias	16	16	6	3	2	5
5B	Other Anomalies of the Male Genitalia	7	4	2	1	1	3
5C	Anomalies of the Female Genitalia	4	1	0	2	1	0
5D	Bladder Extrophy	0	2	0	1	0	2
5E	Renal Agenesis/Hypoplasia	9	8	2	2	6	2
5F	Other or Unspecified Defects of Urogenital System	12	9	5	5	8	9
5G	Indeterminate Sex	1	0	0	0	0	0
5H	Cystic Kidney Disease	10	4	2	3	7	8
5J	Congenital Obstructive Defects of Renal Pelvis or Anomalies of Ureter	77	49	12	15	9	19
G	**Limbs**	**79**	**78**	**31**	**27**	**49**	**55**
6A	Polydactyly/Syndactyly	19	17	8	13	12	10
6B	Limb Reductions	12	17	5	1	10	17
6C	Deformity of Feet	29	19	6	8	20	20
6D	Dislocation of Hip(s)	4	8	4	1	0	0
6E	Other Limb or Limb Girdles	15	17	8	4	7	8
H	**Other Musculoskeletal**	**99**	**95**	**33**	**52**	**47**	**58**
7A	Other Anomalies of the Diaphragm	0	2	2	0	1	0
7B	Anomalies of the Face, Skull or Neck	19	12	7	14	4	11
7C	Other Musculoskeletal Anomalies of the Thorax or Neck	0	1	0	0	0	0
7D	Osteodystrophy or Chondrodystrophy	2	2	1	0	0	2
7E	Other or Unspecified Anomalies of the Musculoskeletal System	34	38	7	9	20	15
7F	Anomalies of the Abdominal Wall	3	2	2	4	1	2
7G	Exomphalos	4	1	1	0	4	3
7H	Anomalies of the Lips, Tongue & Pharynx	2	3	0	4	3	1
7K	Congenital Diaphragmatic Hernia	2	2	3	1	1	3
7L	Gastroschisis	8	4	1	6	3	4
7M	Prune Belly Syndrome	1	0	0	0	0	0
7P	Other Anomalies of Face and Neck	24	28	9	14	10	17
I	**Skin & Integument**	**16**	**12**	**6**	**13**	**6**	**7**
8C	Anomalies of the Skin or Integument	16	12	6	13	6	7
J	**Chromosomal Anomalies**	**53**	**43**	**27**	**23**	**21**	**26**
9C	Trisomy 21 - Down Syndrome	21	20	11	10	12	6
9D	Other Congenital Anomalies	32	23	16	13	9	20
K	**Other Anomalies**	**43**	**36**	**22**	**22**	**22**	**29**
9A	Congenital Neoplasms (other than benign skin)	0	1	0	0	1	0
9B	Endocrine and Metabolic Disorders	11	6	6	4	5	3
9E	Other & Unspecified Congenital Anomalies	30	29	12	17	16	26
9H	Congenital Infections	2	0	4	1	0	0
Total		**734**	**590**	**285**	**300**	**305**	**372**

Morgannwg		North Wales		Wales		% resulting in live or stillbirth (1998/99)
1998	*1999*	1998	*1999*	1998	*1999*	%
24	**13**	**39**	**41**	**141**	**122**	**32.1**
3	3	9	10	28	23	7.8
7	5	9	4	28	21	12.2
4	3	5	8	30	27	43.9
2	1	2	3	7	11	17.6
8	1	14	16	48	40	52.3
27	**16**	**32**	**15**	**112**	**85**	**72.1**
4	0	1	0	13	2	100.0
6	6	12	5	27	29	87.5
15	10	15	10	64	50	60.5
2	0	4	0	8	4	75.0
54	**30**	**52**	**44**	**206**	**189**	**58.5**
6	3	16	9	32	29	63.9
2	2	2	2	15	10	92.0
3	6	6	3	27	24	68.6
1	3	1	2	9	12	71.4
1	0	7	5	16	18	58.8
41	16	20	23	107	96	48.8
108	**72**	**90**	**55**	**481**	**350**	**71.5**
1	2	2	4	11	8	100.0
32	17	15	9	103	74	80.8
29	16	20	14	101	71	79.1
7	3	8	2	37	23	100.0
4	5	6	3	28	20	37.5
33	27	39	21	194	142	63.1
2	2	0	2	7	12	31.6
17	**14**	**24**	**14**	**98**	**81**	**36.9**
17	14	24	14	98	81	36.9
52	**50**	**49**	**53**	**300**	**276**	**74.1**
4	5	4	5	32	34	98.5
1	5	5	4	16	17	78.8
3	0	1	8	9	11	70.0
0	0	1	0	1	5	100.0
13	1	8	8	38	21	40.7
7	9	11	7	43	39	40.2
0	0	1	0	2	0	50.0
8	9	7	8	34	32	51.5
16	21	11	13	125	117	92.6
66	**48**	**77**	**63**	**302**	**271**	**62.0**
13	11	20	21	72	72	73.6
10	2	26	17	63	54	51.3
24	17	16	8	95	72	65.3
5	9	2	4	15	22	100.0
14	9	13	13	57	51	39.8
61	**34**	**74**	**54**	**314**	**293**	**47.0**
1	1	1	0	5	3	37.5
16	5	15	7	61	49	51.8
0	0	0	0	0	1	0.0
0	0	1	1	4	5	44.4
17	8	17	17	95	87	22.5
1	2	4	4	11	14	100.0
2	1	4	2	15	7	40.9
3	4	5	2	13	14	85.2
1	3	2	3	9	12	61.9
2	2	2	3	16	19	85.7
0	0	0	0	1	0	100.0
18	8	23	15	84	82	47.6
10	**4**	**17**	**14**	**55**	**50**	**49.5**
10	4	17	14	55	50	49.5
32	**22**	**34**	**31**	**167**	**145**	**47.8**
13	6	13	13	70	55	55.2
19	16	21	18	97	90	42.8
37	**27**	**34**	**40**	**158**	**154**	**65.8**
0	0	4	1	5	2	71.4
9	4	6	10	37	27	98.4
27	22	24	27	109	121	56.5
1	1	0	2	7	4	63.6
487	**330**	**522**	**424**	**2334**	**2016**	**59.6**

Glossary of terms and definitions

Amniocentesis
Taking a sample of amniotic fluid from the pregnant uterus

CUSUM
Cumulative sum statistical method used by ONS for surveillance

EDD
Estimated date of delivery

EUROCAT
European Concerted Action Of Congenital Anomalies & Twins (anonymised data sent annually from Wales)

Gross Rate
In this report the term is used to describe the number of all CARIS cases or anomalies (including spontaneous fetal losses and terminations of pregnancy) per 10,000 live and stillbirths in Wales. This is described as a gross rate rather than a rate as the all Wales birth figures do not include spontaneous fetal losses or terminations

H.A.(Health Authority)
There are currently five health authorities in Wales:

North Wales
covers North Wales

Dyfed Powys
covers Mid and South West Wales

Morgannwg
covers Swansea, Neath, Port Talbot and Bridgend

Bro Taf
covers Cardiff, Vale of Glamorgan, Merthyr Tydfil, Rhondda, Cynon & Taff valleys

Gwent
covers Newport, Monmouth, Caerphilly, Gwent Valleys

ICD10
International classification of disease, edition 10

Infant death
Death at age under 1 year

Karyotype
Chromosome make-up as seen with a microscopic preparation of dividing chromosomes

LMP
Last menstrual period

Neonatal death
Death occurring during the first 28 days of life

NHS Number
Unique 10-digit number assigned on registering with a GP

ONS
Office for National Statistics (all live and stillbirths with anomalies in Wales are reported by CARIS to ONS)

Rate
The number of events (eg cases, anomalies, deaths) that occur per unit of population (eg per 10,000 births) per unit of time (eg year)

Serum Screening
Testing maternal blood for substances that may suggest a congenital anomaly.

Spontaneous fetal loss
(Miscarriage) - loss of the fetus up to 24 weeks gestation

Stillbirth
Late fetal death after 24 weeks gestation

Syndrome
A combination of clinical features or anomalies forming a recognisable named condition

Total births
Livebirths + stillbirths

U.A. – Unitary Authority
The 8 counties of Wales were re-organised into 22 new unitary authorities in 1996

Appendices

Appendix A

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Appendix B

DATA SOURCES THAT REPORT TO CARIS

- Obstetricians
- Paediatricians
- Midwives
- Ultrasonography
- Special care baby units
- Welsh neonatal screening unit
- Welsh cytogenetics unit
- Paediatric pathology
- Regional paediatric surgery
- All Wales perinatal survey
- Cardiff birth survey
- Regional paediatric cardiology
- Medical Genetics
- Community services
- Maxillo-facial surgery
- Serum Screening
- Mersey congenital anomaly register
- West Midlands congenital anomaly register
- Other congenital anomaly registers
- National child health system

Appendix C

ONS MONITORING GROUPS AND ICD EQUIVALENT CODES

Central Nervous system	A	ICD10 codes
0A	Anencephaly	Q00.0-Q00.2
0B	Spina Bifida	Q05.0-Q05.9
0C	Congenital Hydrocephalus	Q03.0-Q03.9
0E	Encephalocele	Q01.0-Q01.9
0F	Other	G04.9, G12.0, G12.9, G40.9,G60.0,G62.9, G70.9, G71.1, G71.2,G80.9,G83.2, G93.0 ,G93.1, Q02, Q04, Q06, Q07
Eye and Ear	B	
1A	Cystic eyeball	Q11.0
1B	Congenital lens anomalies	Q12.0-Q12.9
1C	Other & Unspecified eye anomalies	Q10, Q11.3, Q13-Q15, H18.5, H50.0, H50.8, H54.0, H54.4, H55,
1D	Ear, all	Q16, Q17
1E	Other anophthalmos	Q11.1-Q11.2
Alimentary sytem	C	
2A	Cleft of palate only	Q35
2B	Cleft of lip only	Q36
2C	Cleft palate with cleft lip	Q37
2D	Tracheo-oesophageal fistula/stenosis	Q39.0-Q39.3
2E	Atresia/stenosis of large intestine. rectum & anal canal	Q42.0-Q42.9
2F	Other or unspecified anomalies of alimentary system	K74.0, K80.2, Q39.4-Q39.9, Q40.0 Q40.2-Q40.9, Q41, Q43-Q45, R14
Cardiovascular system	D	
3A	Tetralogy of Fallot	Q21.3
3B	Ventricular septal defect	Q21.0
3C	Other septal defects	Q21.1-Q21.2, Q21.4-Q21.9
3E	Patent Ductus Arteriosus	Q25.0
3F	Anomalies of the umbilical artery	Q27.0
3G	Other congenital cardiac, or great vessel anomalies	I45.6, I47.1, I49.1, M30.3, Q20, Q22-Q24, Q25.1-Q25.9, Q26
3H	Congenital anomalies of other vessels	Q27.1-Q27.9, Q28
Respiratory system	E	
4A	Congenital anomalies of the respiratory system	Q30-Q34
Urogenital system	F	
5A	Hyspospadias / epispadias	Q54, Q64.0
5B	Other anomalies of the male genitalia	N47, Q53, Q55
5C	Anomalies of the female genitalia	N89.8, Q50-Q52

5D	Bladder exstrophy	Q64.1
5E	Renal agenesis	Q60
5F	Other or unspecified defects of urogenital system	N25.8, Q63, Q64.2-Q64.9
5G	Indeterminate sex	Q56
5H	Cystic kidney disease	Q61
5J	Congenital obstructive defects of renal pelvis or anomalies of ureter	N13.9, Q62
Limbs	G	
6A	Polydactyly / syndactyly	Q69,Q70
6B	Limb reductions	Q71-Q73
6C	Deformities of feet	Q66
6D	Dislocation of hip(s)	Q65.0-Q65.6
6E	Other limb or limb girdle defects	M21.2, Q65.8, Q65.9, Q68.1-Q68.5, Q74
Other musculoskeletal	H	
7A	Other anomalies of diaphragm	Q79.1
7B	Anomalies of face, skull, or neck	Q67-Q68.0, Q75, R22.0,
7C	Other musculoskeletal anomalies of thorax and neck	Q76.8, Q76.9
7D	Osteodystrophy or chondrodystrophy	Q77, Q78
7E	Other or unspecifed anomalies of musculosketal system	M89.8, P94, Q68.8, Q76.0-Q76.7, Q79.5, Q79.9
7F	Anomalies of abdominal wall (hernias)	K40-K46
7G	Exomphalos	Q79.2
7H	Anomalies of the lips, tongue and pharynx	Q18.4-Q18.7, Q38
7J	Congenital hiatus hernia	Q40.1
7K	Congenital diaphragmatic hernia	Q79.0
7L	Gastroschisis	Q79.3
7M	Prune belly syndrome	Q79.4
7N	Branchial cleft, auricular sinus	Q18.0-Q18.2
7P	Other anomalies of face & neck	K07, Q18.3, Q18.8, Q18.9, R22
Skin and Integument	I	
8C	Anomalies of skin and integument	D18.0, D22.3, D22.6, D22.7, D22.9, D23.6, D23.7, L53.9, L81.3, Q80-Q84
Chromosomal Anomalies	J	
9C	Trisomy 21 – Down syndrome	Q90
9D	Other chromosomal anomalies	Q91-Q99
Other Anomalies	K	
9A	Congenital neoplasms (other than benign skin)	C49.2, Q69.2, C71.9, C74.9, D13.9, D14.3, D15.1, D16.6, D17.2, D17.9, D36.1, D37.0, D41.0, D43.2, D47.1, D48.0, D48.7, D48.9,
9B	Endocrine and metabolic disorders	D66, D67, D68.0, D68.2, D69.4, D81.9, D82.1, E03.0, E03.1, E07, E23-E25, E27-E30, E32, E34, E70-E80, E83-E85, E88, E90
9E	Other and unspecified congenital anomalies	B27.0, D18.1, D55.0, D56.3, D57.1, D57.3, D58.0, D68.0, F79, I42.4, K21.9, L05.9, P02.6, P10-P15, P20-P29 (excl P29.4), P36, P50, P61, P70-P78, P80-P83, P90-P93, P95, P96, Q85-Q89, R16.0, R16.2, R18, R19.0, R22.9
Congenital Infections		
9H		A50.0, P29.4, P35, P37, P52.5

Appendix D

ONS & EUROCAT EXCLUSION LIST

ONS and EUROCAT exclude the following minor defects from registration unless occurring in combination with other anomalies. These exclusion criteria also apply to CARIS (with the exception of hypospadias*)

ANOMALIES OF EYE

- Stenosis or stricture of lacrimal duct

ANOMALIES OF EAR

- Minor or unspecified anomaly of ear
- Preauricular appendage, tag or lobule
- Other appendage, tag or lobule

CARDIOVASCULAR SYSTEM

- Functional or unspecified cardiac murmur
- Absence or hypoplasia of umbilical artery, single umbilical artery
- Patent ductus arteriosus (in babies <37 wks or <2500gms)

DIGESTIVE SYSTEM

- Tongue-tie

EXTERNAL GENITALIA

- Undescended testicle and unspecified ectopic testis
- Congenital hydrocele or hydrocele of testis
- Phimosis
- Hypospadias when the meatus lies before the coronary sulcus, glandular or 1st degree hypospadias (Please report all cases to CARIS)

LIMBS

- Clicking hips
- Clubfoot of postural origin
- Postural or unspecified metatarsus varus or metatarsus adductus
- Postural or unspecified talipes calcaneovalgus or pes calcaneovalgus
- Minor or unspecified anomalies of toe such as hallux valgus, hallus varus, or “orteil en marteau”

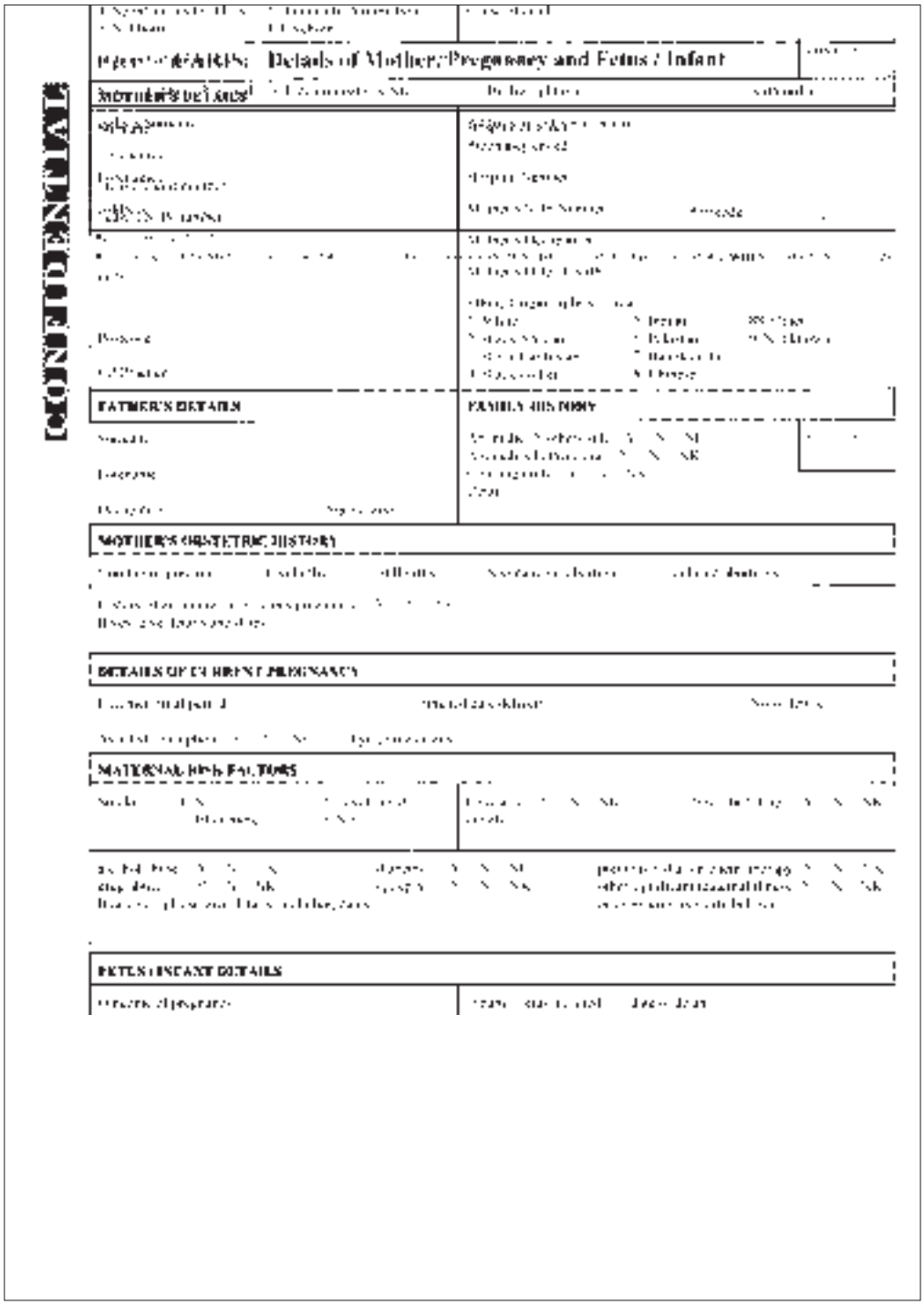
OTHER MUSCULOSKELETAL ANOMALIES AND ANOMALIES OF THE INTEGUMENT

- Spina Bifida occulta uncomplicated
- Pectus excavatum
- Minor or unspecified anomaly of nose
- Minor or unspecified deformity of face
- Minor anomaly of nipple
- Accessory or ectopic nipple
- Congenital umbilical hernia, inguinal hernia, para umbilical, ventral or incisional hernia, hiatus hernia
- Abnormal palmar crease
- Skin tag with surface less than $4cm^2$: skin tag, naevus, angioma, haemangioma, glomus tumour, lymphangioma, birthmark
- Sacral dimple

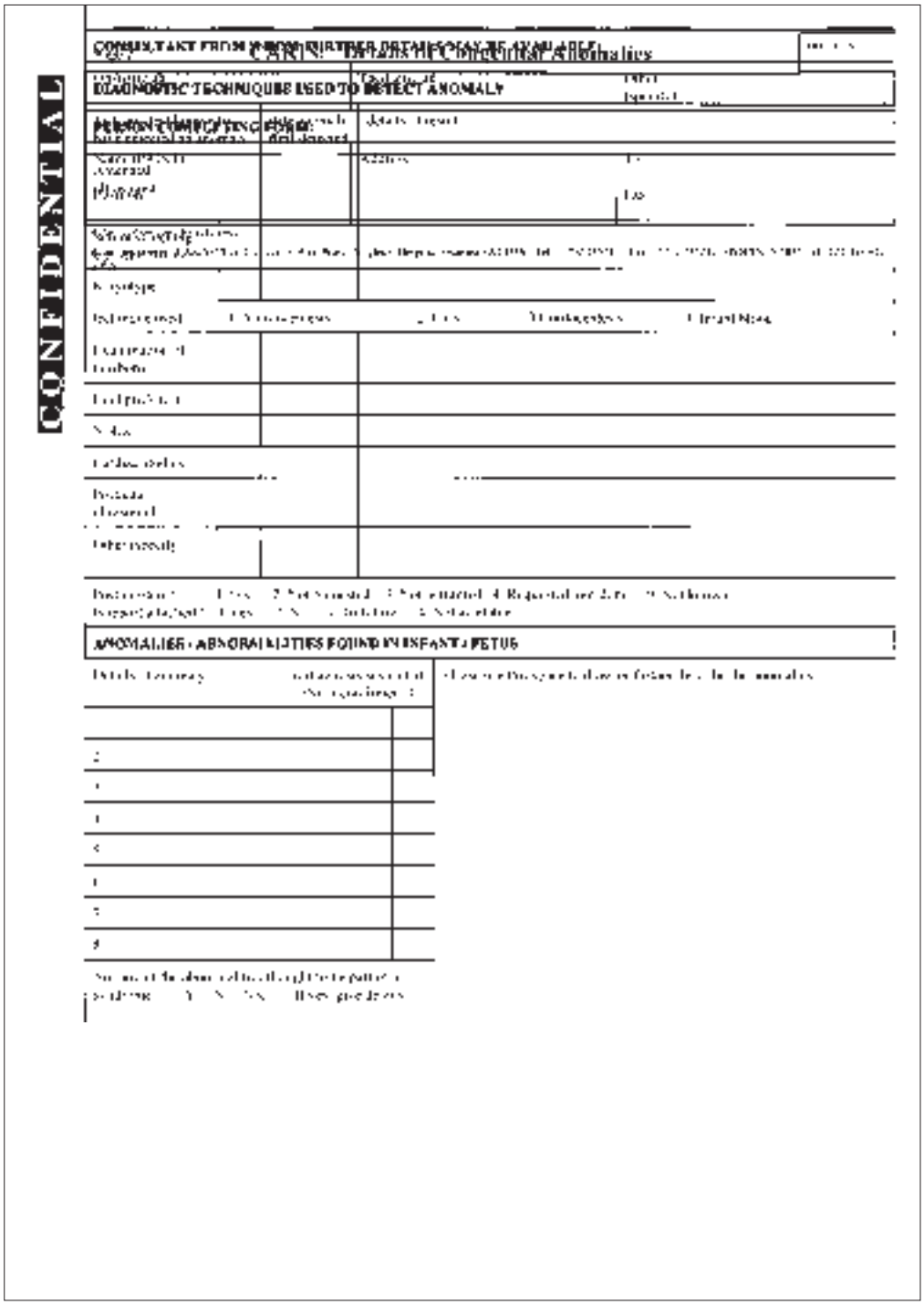
Appendix E

CARIS WARNING CARD

Appendix F


CONFIDENTIAL
Details of Mother/Pregnancy and Fetus / Infant
MOTHER'S DETAILS
FATHER'S DETAILS
FAMILY HISTORY
MOTHER'S OBSTETRIC HISTORY
DETAILS OF CURRENT PREGNANCY
MATERNAL RISK FACTORS
BIRTH / INFANT DETAILS

Appendix F


CONFIDENTIAL

DIAGNOSTIC TECHNIQUES USED TO DETECT ANOMALY

ANOMALIES / ABNORMALITIES FOUND IN INFANT / FETUS

Appendix G

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Appendix H

Data Quality Measures

DUPLICATE ENTRIES - ALL CASES WITH DATE OF END OF PREGNANCY IN 1998 AND 1999

(11 duplicates found on previous analysis of 1998 data)

Method of Identification (sequential searches)	What search identifies	Number of duplicate entries found	Duplicates deleted from databse prior to data analysis
Duplicates on Maternal NHS number	Duplicate cases	1	yes
Duplicates on Baby NHS number	Duplicate cases	4	yes
Postcode	Duplicate cases	1	yes
Maternal DOB	Duplicate cases	0	
Baby ID number and ICD10 anomaly code	Duplicate anomalies recorded on case	2	yes

CASES FOUND WITHOUT (ONS) CLASSIFIABLE CONGENITAL ANOMALY OR SYNDROME

-3 cases identified. Excluded from any data analysis

DATA COMPLETENESS - ALL CASES WITH DATE OF END OF PREGNANCY IN 1998 AND 1999

Data item	Number missing	% completeness
Postcode	0/1730	100%
Ward code (ie using AFD software)	3/1730	99.8%
Maternal date of birth	4/1730	99.8%
Maternal NHS number	39/1730	97.8%
Maternal date of last menstrual period	152/1730	91.2%
Outcome of pregnancy	2/1730	99.9%
Sex of baby/fetus	9/1730	99.5%
Birthweight of baby/fetus (all cases)	199/1730	88.5%
Birthweight of baby/fetus (livebirths and stillbirths)	116/1326	91.3%
Baby NHS number (livebirths only)	238/1279	81.5%

Conclusions

1 The CARIS reporting system has continued to grow during 1999, leading to improved reporting from various areas across Wales. With the many NHS staff changes that take place, continued vigilance is required to ensure good paths of communication and high quality reporting.

2 During 1999, a further 95 cases of congenital anomaly with dates of end of pregnancy in 1998 were identified, giving a gross rate of 281 cases of congenital anomaly per 10,000 livebirth/stillbirths for 1998. Gross rates for 1999 so far appear slightly lower than for 1998. This difference is not statistically significant.

3 Variations are evident around Wales in gross case rates and the percentage of cases that survive. The reasons for these variations are not yet clear, and may still be due to differences in reporting rather than true differences in the levels of anomalies that occur.

4 From the first two years of data collection only, Wales continues to experience rates of congenital anomaly that are at least as high as those reported from the rest of Britain or Europe. Comparisons should only be made with caution because of national and international differences in definitions of particular anomalies, reporting systems and methods of calculating rates. At present, little data has been published from elsewhere for the same years as CARIS.

5 Specific anomalies

a) Gastroschisis. The number of cases rose in 1999, despite a falling birthrate. The gross rate for 1998 and 1999 combined was 5.3/10,000 births (95% CIs 3.6 to 7.1). This figure is higher than reported from many other sources for the early 1990s. Comparable data from elsewhere for 1998 and 1999 is not yet generally available.

b) Further analysis of data on cases of Down syndrome from 1998 and 1999 suggest low overall use of serum screening in Wales. Where serum screening is undertaken it appears to detect about 2/3rds of cases.

CARIS

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