online ar-y-we

CARIS

Congenital Anomaly Register & Information Service

caris review

including 1998-2003 data

inside...

- Key points on congenital anomalies in Wales
- Urinary tract anomalies
- Update on gastroschisis
- Reporting of anomalies in Wales

NHS WALES GIG CYMRU

Introduction

CARIS is the Congenital Anomaly Register and Information Service for Wales. Based at Singleton Hospital, Swansea, it is funded by the Welsh Assembly Government as part of NHS Wales.

The CARIS website contains detailed information about the register, an overview of congenital anomalies in Wales, special reports and data tables. This can be accessed at :

www.wales.nhs.uk/caris

The CARIS team at the 2003 South Wales Annual Meeting and website launch.

We are (left to right) David Tucker, Val Vye, Margery Morgan, Judith Greenacre, Debbie Rogers

online ar-y-we
CARIS

Write CARIS Office
Level 3 West Wing
Singleton Hospital
SWANSEA
SA2 8QA

Phone 01792 285241
(WHTN 0 1883 6122)

Fax 01792 285242
(WHTN 0 1883 6123)

e-mail dave.tucker@swansea-tr.wales.nhs.uk

web www.wales.nhs.uk/caris

Published by
CARIS ISBN 0-9537080-5-5

Designed by RIDLER WEBSTER LIMITED 01792 700070

* *also accessible through the HOWIS (NHS Wales) website at www.howis.wales.nhs.uk/caris*

What is CARIS?

The fundamental aim of CARIS is to provide reliable data on congenital anomalies in Wales. With this valuable data, studies can be done on the profile of congenital anomalies in

- **antenatal screening/interventions**
- **health service provision for affected babies and children**
- **possible clusters of birth defects and their causes.**

We collect data on any baby or fetus for whom pregnancy ended after 1st January 1998, where the mother was normally resident in Wales at the end of pregnancy.

CARIS uses a multiple source reporting system and, at present, over 100 individuals or agencies regularly send us information. Data from clinical and laboratory sources is reported via warning cards, reporting forms and data exchanges. CARIS Co-ordinators in each Trust are responsible for much of the clinical reporting. In the CARIS office, data is collated, the information is coded and the data quality carefully checked. The data is then available for feedback to clinicians – paediatricians, ultrasonographers, midwives, etc. as well as supplying information to the National Assembly for Wales, EUROCAT and the Office for National Statistics (for surveillance).

At the CARIS office we cannot over emphasise the importance of the confidential nature of our data and we operate a strict security and confidentiality policy. We have gained support under Section 60 of the Health and Social Care Act 2001 meaning that the register can continue collecting and analysing this valuable information.

Over 37,000 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 3% of births take place at home. Wales has 16 consultant obstetric units and 10 midwifery/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. Good links with congenital anomaly registers that border Wales (Mersey, West Midlands and the South West of England) remain very important.

Clinical reporting is the most important source of information for CARIS, especially for those babies who:

- **die but do not have a post mortem**
- **survive and have anomalies not requiring immediate specialist help.**

Diagnostic services, particularly ultrasound scanning and pathology, can alert us to a case or give valuable further information.

Regional services including cytogenetics and specialist clinical services can help by providing more details of the anomalies involved.

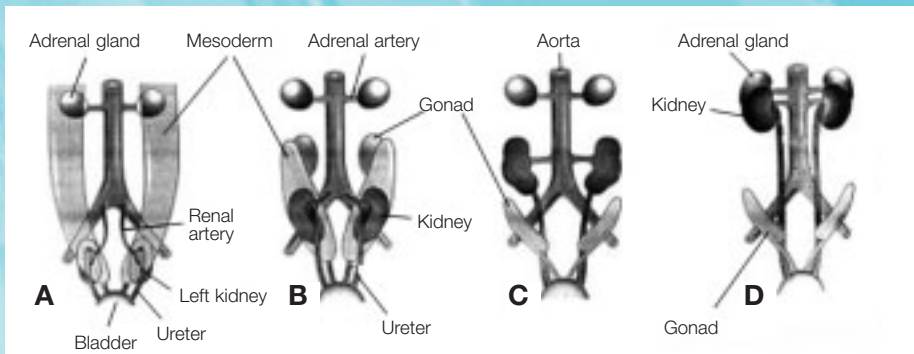
Babies with congenital anomalies may be recorded on other databases such as Protos (Cardiff), the All Wales Perinatal Survey or the Standard Child Health Computer System.

Key points on congenital anomalies in Wales 1998-2003

- 8,145 cases of congenital anomalies have so far been reported to CARIS with pregnancy ending between 1st January 1998 and 31st December 2003. These include live births, stillbirths, terminations of pregnancy for congenital anomalies and miscarriages (although reporting of miscarriages will inevitably be incomplete). This means that the "gross" rate of known pregnancies affected by congenital anomaly is 4.3%.
- 83% of cases were liveborn. The percentage of liveborn babies affected by congenital anomaly is 3.6%. Approximately two-thirds of live born cases survived to the end of their first year of life.
- In just over 50% of cases, only one anomaly or birth defect is recorded. In 11% of cases, an underlying chromosomal defect was identified that could account for many of the physical anomalies. The remainder of cases had multiple anomalies of varying levels of complexity.
- Defects of the heart and circulation are the largest single group of anomalies followed by defects of the limbs, urinary system and musculo-skeletal system.
- Even allowing for the fact that more male babies are born each year than females, there is a slight excess of congenital anomalies in male babies.
- In about 20% of cases, an indication that the fetus has a congenital anomaly is detected before the end of pregnancy. This figure varies with the type of anomaly. For example, 97% of cases of anencephaly (a form of neural tube defect) are suspected before the end of pregnancy.
- Variations in congenital anomaly rates occur around Wales, with apparently higher rates for Swansea and Neath Port Talbot compared to other Unitary Authorities. Apparently lower rates are seen in areas of the South Wales valleys, Mid Wales and parts of North Wales. Some of this variation is undoubtedly due to persistent differences in reporting practices across Wales. This is discussed in the section, *Reporting of Anomalies in Wales*.
  The areas with highest rates tend to have better survival rates for liveborn babies. This may, again, reflect better reporting of cases in infancy, allowing more survivors to be added to the numbers from better reporting areas.
- Rates for many anomalies in Wales appear relatively high when compared to other areas in Britain and Europe. This is considered in greater detail in the section, *Reporting of Anomalies in Wales.* We suspect that good reporting in Wales accounts for a large part of these differences but CARIS takes this finding seriously and we continue to keep the situation under review.

- gross rate of (all) congenital anomalies reported is 4.3%
- 83% of cases are liveborn
- 2/3rd liveborn cases survive to end of 1st year
- heart & circulatory defects are largest single group
- 20% of cases overall are suspected antenatally
- variations in rates exist around Wales

Urinary tract anomalies



Kidney development in the fetus from the 6th to the 9th week of gestation

Abnormalities of the urinary tract are among the most common birth defects and are said to account for as many as 20% of all fetal malformations. Some of these anomalies are minor problems causing no symptoms e.g. a double ureter leading from one kidney to the bladder. These are often undiagnosed until later investigations in childhood for an unrelated problem. Other anomalies can cause problems ranging from urinary tract infections to kidney damage or failure.

Why do they occur?

The cause of most urinary anomalies is unknown. Some disorders (e.g. polycystic kidneys) are inherited from parents with the defect. The urinary tract system undergoes several phases in its development and it is possible that environmental effects can influence this.

Urinary tract development

End of 4th week

Ureteric bud induces mesoderm in sacral region to form definitive kidneys. Ureteric bud forms the collecting tubules, calyces, renal pelvis and ureter.

4 to 6 weeks

Bladder forms from the anterior part of the cloaca. Ureters connect to the trigone of the bladder.

6 to 9 weeks

Kidneys ascend from sacral region to lumbar region

- failure results in pelvic kidney,
- fusion of the inferior poles results in horseshoe kidney

On transvaginal ultrasound the fetal kidneys and bladder are detectable

10th week

Nephrons of the developing kidney connect to collecting ducts.

- failure causes cystic renal disease

End of 10th week

URINE production starts.

12 weeks gestation

On ultrasound

- *Kidneys appear as hyperechoic structures in paravertebral regions.*
- *Bladder seen in the pelvis as an echo free area.*
- *Ureters are not normally visualised.*

Welsh picture

In Wales, the gross[1] rate of urinary anomalies (all cases) is 78.5 per 10,000 live and stillbirths. Around 250 babies a year in Wales are reported to have some form of urinary anomaly and these represent 18% of all cases reported to CARIS. About 85% of cases are liveborn, and 80% of these survive to the end of their first year.

Boys are more commonly affected than girls in a ratio of 3 to 1. In about one third of cases, additional defects involving other body systems have also been identified.

[1] *The gross rate includes fetal losses, terminations of pregnancy, live and stillborn babies.*

Common urinary defects

Renal Agenesis

Renal agenesis is the absence of one or both kidneys. If bilateral the condition is lethal. If unilateral the prognosis is generally good if the remaining kidney enlarges to compensate.
Renal agenesis is probably caused when the ureteric bud fails to start the development process in the mesoderm. Most cases are sporadic but genetic factors may be involved in view of the number of familial cases.

For the years 1998-2003, CARIS has reports of:

- 28 cases of bilateral renal agenesis. This gives a gross rate (all cases) of 1.5/10,000 live & stillbirths (95%CI 0.9-2.0). None of these cases were liveborn and 23/25 ended at termination of pregnancy following antenatal detection of this or other significant birth defects
- 74 cases of unilateral renal agenesis. 49 (66%) of these were live or stillborn – a rate of 2.5/10,000 births or about 1 in 2500 live and stillbirths

Data on bilateral renal agenesis is available at European level from EUROCAT for 1998-2002. The Wales rate in this analysis was 2.8 per 10,000 births[2] – slightly higher than the overall EUROCAT rate of 1.6 per 10,000 births.

[2] *Based on livebirths, stillbirths, fetal losses beyond 20 weeks gestation and termination of pregnancy for congenital anomaly*

Diagnosis

If no urine is being produced, very little liquor is present in the developing pregnancy, giving severe oligohydramnios. The assessment of the fetus can be extremely difficult as it is often curled up deep in the pelvis. Transvaginal scanning and intra-amniotic infusion may help with the diagnosis. The lack of liquor causes the typical Potter's syndrome with a fetal appearance of wide set eyes, low set ears, receding chin and limb defects. Lung development is severely restricted because of the absence of liquor.

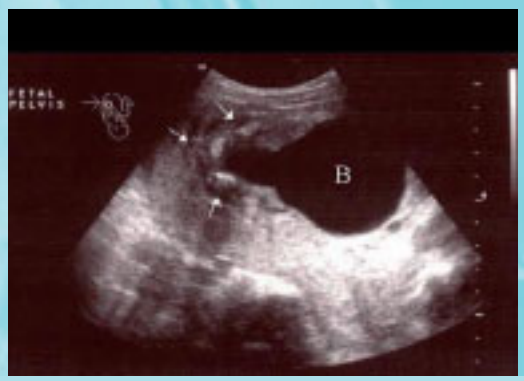
Outcome

Pulmonary hypoplasia is the result of severe oligohydramnios and this means that most infants with bilateral renal agenesis die shortly after birth. Babies born with unilateral renal agenesis can often lead normal lives but are at increased risk of kidney infections, kidney stones, high blood pressure and kidney failure. They are also more likely to have other congenital anomalies.

Hydronephrosis

This involves swelling of one or both kidneys with accumulation of urine. This cannot flow out of the kidneys because of a blockage somewhere in the urinary tract. This can show up on antenatal ultrasound in as many as 1 in 50 pregnancies, but significant renal pathology will exist in only 1 in 500. The two main causes are pelvi-ureteric junction obstruction and posterior urethral valves.

CARIS has reports on 632 cases of hydronephrosis from 1998 to 2003. This equates to about 1 in 300 affected pregnancies.



Posterior urethral valves. Sagittal section through the fetal abdomen showing a distended bladder (B). The arrows point to the dilated posterior urethra.

Pelvi-ureteric junction obstruction

This is the commonest cause of hydronephrosis. It is more common in boys and is more common unilaterally. Why it occurs is unclear as it is rare to see any true narrowing at the region where the ureter joins the renal pelvis.
Associated renal anomalies are said to be seen in 25% of cases and other anomalies in 12%. The risk of recurrence is low.

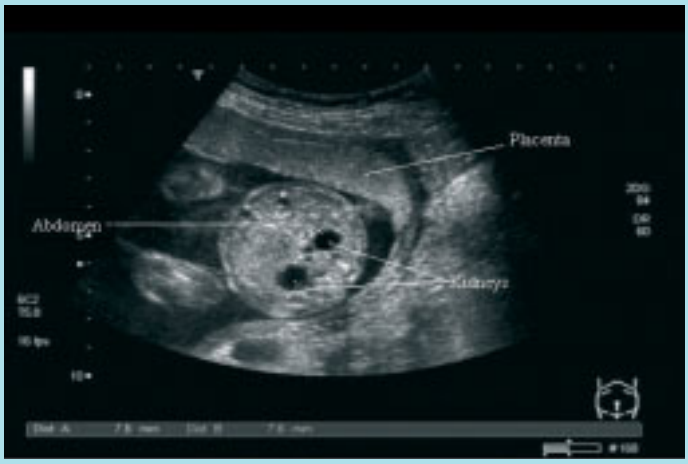
Diagnosis

Ultrasound easily demonstrates PUJ showing dilated renal pelvis and calyces with a normal ureter and bladder. The greater the dilatation the poorer the renal function in the neonate. In view of this, follow up antenatal and postnatal ultrasound is usually recommended for renal pelvic diameters at or greater than 5mm. Before the advent of ultrasound, hydronephrosis was poorly diagnosed and often missed. Antenatal diagnosis these days can improve management and outcome.

Outcome

This is usually good in both unilateral and bilateral disease. Surgery is reserved for cases with increasing hydronephrosis and worsening renal function.

Cross section of the fetal abdomen showing mildly dilated renal pelves



Posterior urethral valves

This is the most common form of severe urinary obstruction in children. It is caused by excess tissue present in the urethra draining the bladder. It is only seen in males and is the causative factor in as many as one third of boys under 4 years with end stage renal disease.
Other abnormalities include pulmonary hypoplasia if oligohydramnios has been severe, giving a Potter's syndrome appearance. Prune belly syndrome can also result from urethral obstruction.

Diagnosis

At antenatal ultrasound the bladder appears thick walled and is dilated, as are the posterior urethra and ureters. Hydronephrosis arises as the back pressure of urine reaches the kidneys. The obstruction may be so severe that the bladder ruptures into the peritoneal cavity thereby relieving the pressure on the kidneys. Liquor volume is not always reduced, but oligohydramnios is a sign of a poor outcome.

Outcome

Cases diagnosed early indicate that they have severe disease and usually a poor outcome. The overall mortality rate can be high with about a third of the survivors developing end-stage renal failure.
Fetal intervention designed to relieve the urinary obstruction has been proposed to prevent the development of renal dysplasia. This involves monitoring fetal urinary electrolytes and the appropriate insertion of a shunt to take the urine from the fetal bladder to the amniotic cavity. This is not without complications and management remains controversial.

Polycystic Kidney Disease

This covers a wide range of conditions, many of which have a genetic basis.
The resulting cysts can cause reduced renal function and often renal failure. Other effects are frequent urinary tract infections, pain and high blood pressure. Three of the commonest types of cystic kidneys are:

- Autosomal dominant polycystic renal disease
- Multicystic renal dysplasia
- Autosomal recessive (infantile) polycystic disease

Autosomal dominant polycystic renal disease

- 1 in 1000 live births
- traditionally presents with hypertension and renal failure in the 5th decade
- antenatal ultrasound appearance of enlarged echogenic kidneys
- poor outcome for antenatally diagnosed cases
- recurrence risk of 50%

CARIS is aware of only a few confirmed cases but this is not surprising for a condition that often presents in later life.

Multicystic renal dysplasia

- 1 in 3000 live births
- usually unilateral
- commoner in boys
- antenatal ultrasound appearance of kidney as paraspinal mass containing multiple cysts
- good outcome if unilateral
- no increased risk of recurrence

CARIS is aware of 93 cases reported in 1998-2003, 72 of which were unilateral. Overall, 61 cases (66%) were liveborn, although this rate was lower (26%) for bilaterally affected cases. This gives an occurrence in Wales of 1 in 3099 live births.

Autosomal recessive (infantile) polycystic disease

- 1 in 40-50,000 live births
- presents as symmetrically enlarged kidneys caused by cystic dilatation of the collecting tubules
- associated with liver fibrosis
- antenatal ultrasound appearance of enlarged echogenic kidneys with oligohydramnios
- outcome dependent on the severity of renal disease
- recurrence risk of 25%

14 cases have been reported to CARIS for 1998-2003, of which 11 were liveborn. This gives an occurrence in Wales of *1 in 17,189* livebirths.

Hypospadias

Hypospadias is a common birth defect that affects approximately 1 in 500 boys in Wales.
The urethra does not extend to the tip of the penis and its opening is located somewhere along the underside of the shaft.
Prevalence rates of hypospadias have been rising from the 1960s onwards.
It has been suggested that this is due to adverse environmental influences including the effect of female hormones and pesticides.

Diagnosis

Diagnosis is made at the newborn examination. The severity of the condition can vary from a minor abnormality near the usual position of the urethral opening to an opening at the scrotum or perineum. Often it is accompanied by a hooded prepuce, sometimes with a curvature of the penis called chordee.

Outcome

Mild hypospadias is usually insignificant and surgery is not required unless the opening is stenosed. If the opening is proximal to the glans (on the shaft of the penis or at the scrotum or perineum) surgical repair is necessary to move the urethral opening to the tip of the penis.
Circumcision, if required, should be delayed until corrective surgery is done as the prepuce may be needed for reconstruction of the urethra.
Without surgery most affected boys would have to urinate sitting down and as adults would suffer pain during intercourse.

Reports to CARIS have increased with the recent inclusion of child health data from several areas of Wales. For 1998-2003, 356 cases of hypospadias have been reported, although the degree of abnormality is recorded in only half of these. Over 99% (353 cases) were liveborn and over 97% of these survived to the end of their first year of life. This gives a liveborn rate of 18.7/10000 births. Rates to the end of 2002 were higher than the EUROCAT rate for the same period (see section, *Reporting of Anomalies in Wales*).

About two third of CARIS cases are reported as isolated anomalies. Of the 171 cases for which the type of hypospadias is given

- 135 (79%) are glanular
- 24 (14%) are penile
- 8 (5%) are peno-scrotal (proximal)
- 4(2%) have the meatus at other sites

As hypospadias is not evident on antenatal ultrasound, it is not surprising that none of the cases are reported as being detected antenatally.

Site of urethral meatus

Brock D, Rodeck C, Ferguson-Smith M, (1992) Prenatal Diagnosis and Screening, Churchill Livingstone

Thomas R and Harvey D (1997) Neonatology. 2nd edition, Churchill Livingstone

Moore K and Persaud T (1998) Before We Are Born. Essentials of embryology and birth defects, 5th edition W.B. Saunders

Twining P, McHugo J, Pilling D (2000) Textbook of Fetal Abnormalities, Churchill Livingstone

*March of Dimes – **www.modmes.org/professionals/681_1215.asp***

Update on Gastroschisis

Gastroschisis is a relatively rare condition that has attracted considerable recent attention. Rates of occurrence have risen, especially in the Western World and over the past 20 years. Very recently there has been a suggestion that rates may be increasing more rapidly, although this has yet to be confirmed.

Although reported rates of gastroschisis in Wales are not the highest in the world, they are certainly greater than for many other areas, and appear to have risen further during 2003 and early 2004. One reason for this may be good reporting, but it is unlikely that this accounts for the entire picture. CARIS is involved with others in a number of initiatives to look at gastroschisis in Wales in greater detail; to try and establish whether recent changes are a chance finding or part of a more significant event. Work is also underway to assess whether any rise is confined to parts of Wales or includes other areas of the British Isles.

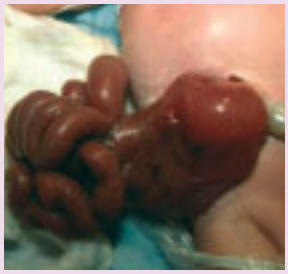
Here, we present CARIS data for Wales, based on cases with pregnancy ending up to the end of 2003.

What is gastroschisis

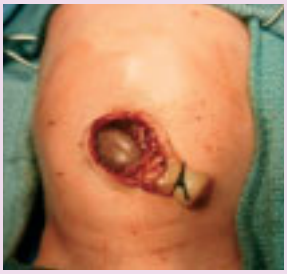
Gastroschisis is a condition in which there is a herniation of abdominal contents through a defect in the anterior abdominal wall. The abdominal wall defect often lies to the right of the insertion of the umbilical cord, but the cord is not involved.
In gastroschisis, the abdominal contents (most often loops of bowel) float freely in the amniotic cavity and are often visible by antenatal ultrasound. Following delivery in a specialist unit, the baby usually undergoes early surgery to repair the defect. There may be a prolonged stay in hospital. In the absence of complications or other congenital anomalies, the outlook for these babies is usually good.

The condition may be associated with small bowel atresia. Associations with other anomalies are not common.

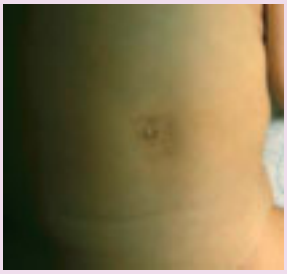
Occasional cases have been described where the condition is diagnosed antenatally but ‘disappears’ by birth. Such cases have been described with subsequent small bowel atresia / stenosis.



At birth
The defect is lying to the right of the umbilicus



Surgery
The gut has been put back inside the abdomen



At 6 months
The scar has healed well

The cause of gastroschisis is not understood. It is not usually associated with known genetic defects or other congenital anomalies. Possible links to various environmental pollutants have been suggested, including proximity to landfill sites.

It is thought that development of the anomaly may be a consequence of vasoconstrictive episodes in early pregnancy. Gastroschisis can affect any pregnancy but known risk factors include the following:

- younger mothers
- socially disadvantaged groups
- mothers who smoke
- maternal use of aspirin and anti cold remedies
- maternal substance misuse.

Gastroschisis in Wales 1998-2003

Table 1 shows the number of cases by year in which pregnancy ended and **Table 2** shows the breakdown of cases by year and area of residence of mother. Data show a rise in rates in 2003, although this difference is not statistically significant.

All but 2 (98%) of the cases reported to CARIS were detected antenatally, allowing the opportunity for planned delivery near a neonatal surgery unit. The top row of **Table 1** shows the number of liveborn cases of gastroschisis. This represents the cases likely to be treated in neonatal surgical units. The majority of cases undergo surgery in the University Hospital of Wales, Cardiff, although babies born to mothers living in mid or north Wales **(table 2 overleaf)** may be referred to Birmingham or Liverpool for treatment. Reporting of cases treated outside Wales is often delayed so that numbers of cases from mid and north Wales may increase further.

Table 1

Numbers of cases of gastroschisis among babies of mothers normally resident in Wales and reported to CARIS, showing year of end of pregnancy and survival outcome

outcome pregnancy	Total	1998	1999	2000	2001	2002	2003
live born	81	12	15	10	15	10	19
stillbirths, fetal losses and terminations	12	4	4	1	1	0	2
outcome not confirmed	1	0	0	1	0	0	0
total cases reported for year	94	16	19	12	16	10	21
gross rate (all cases) per 10000 live & still births (95%C.I.)	4.9 (3.9-5.9)	4.8 (2.4-7.1)	5.9 (3.2-8.5)	3.8 (1.7-6.0)	5.2 (2.7-7.7)	3.3 (1.3-5.4)	6.7 (3.8-9.5)
liveborn rate per 10000 livebirths (95%C.I.)	4.3 (3.4-5.2)	3.6 (1.6-5.6)	4.7 (2.3-7.0)	3.2 (1.2-5.2)	4.9 (2.4-7.4)	3.3 (1.3-5.4)	6.1 (3.3-8.8)

Table 2

All cases of gastroschisis born to mothers normally resident in Wales and reported to CARIS, showing numbers by year of end of pregnancy and maternal area of residence

Unitary Authority	Total	1998	1999	2000	2001	2002	2003
South East Wales	46	11	8	5	9	6	7
Mid & West Wales	31	3	8	3	5	3	9
North Wales	17	2	3	4	2	1	5
Total for Wales	94	16	19	12	16	10	21

Table 3

Comparison of features of cases of gastroschisis with all other cases of congenital anomaly reported to CARIS, with pregnancy ending 1998-2003

	Gastroschisis 1998-2003		All other cases of anomaly 1998-2003		Odds Ratio	*p* (X^2)
Factor	n	%	n	%		
maternal age <25yrs	69/94	73.4%	2375/8035	29.6%	6.6 (4.1-10.7)	<0.001*
+ve history maternal drug abuse	6/94	6.4%	69/8053	0.9%	7.9 (3.0-19.5)	<0.001*
+ve history maternal smoking	50/82	61.0%	1512/5373	28.1%	4.0 (2.5-6.4)	<0.001*
presence other anomalies	27/94	28.7%	3767/8052	46.8%	0.5 (0.3-0.7)	0.001*
+ve history consanguinity	0/30	0%	32/2796	1.1%	-	-

** significant at 0.05 level*

Table 3 shows features of cases of gastroschisis compared to all other reported cases, using information routinely collected on the CARIS database. This confirms the association with younger maternal age, maternal smoking and a history of maternal substance misuse, although it must be stressed that there is no evidence that all mothers of babies with gastroschisis undertake these activities. Other factors such as maternal diabetes and use of folic acid are not associated with this condition.

Among the 27 cases that had additional anomalies reported, two had chromosomal defects (one triploidy and one Turner syndrome). The remaining 25 cases had a variety of defects, most notably including:

- 6 cases of atrial septal defects (which may have been associated with early delivery)
- 4 cases of small bowel atresia and 1 of large bowel atresia. Small bowel atresia is a known associated defect for gastroschisis
- 2 cases of optic nerve hypoplasia. This rare defect is not often reported with gastroschisis although it too is known to occur more frequently in the babies of younger mothers.

Reporting of Anomalies in Wales

During 2003/2004, concern has been expressed in the media and by politicians concerning:

- the apparently high rates of congenital anomalies in Wales compared to other countries.
- differences in reported rates across Wales, particularly the higher reported rates for Swansea and Neath Port Talbot.

CARIS has always openly discussed these variations and their possible causes. We have now reviewed the situation again in the light of increased concerns.

Rates for Wales compared to elsewhere in Great Britain and Europe

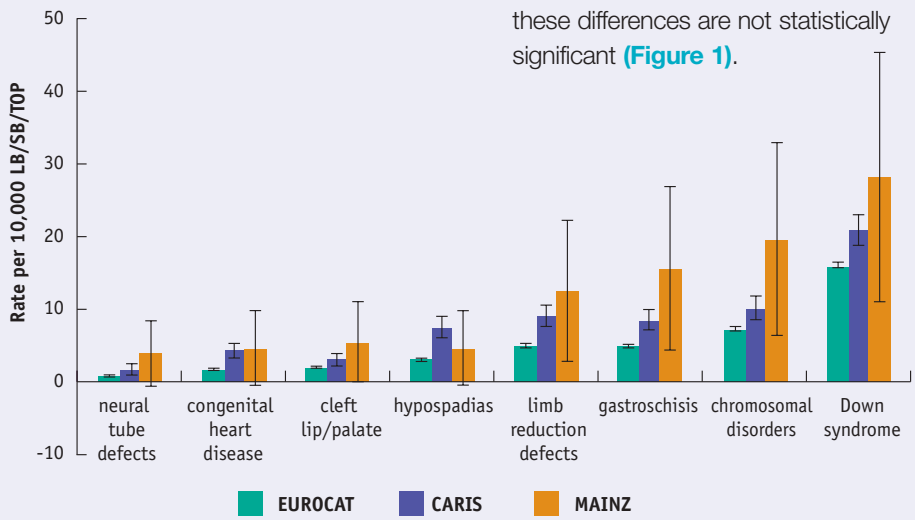
How do Welsh rates compare to Britain and Europe?

Before CARIS was established in 1998, there was very little evidence that Wales had higher rates of congenital anomalies than other parts of Great Britain or Europe. Anomaly data routinely submitted to the Office for National Statistics (ONS) and European Congenital Anomaly Monitoring Programme (EUROCAT) were broadly in keeping with data from other areas. Since 1998, reported congenital anomaly rates for Wales have risen sharply and now rates for Wales are often the highest of any regional area of Great Britain that reports to ONS. Welsh rates are also higher than the average European rates reported by EUROCAT. Having said this, one or two individual registries reporting to EUROCAT consistently have higher rates than CARIS. Rates from the register based at Mainz in Germany are almost always higher than those for CARIS, but the numbers from Mainz are small and these differences are not statistically significant **(Figure 1)**.

Figure 1

EUROCAT data: Comparison of reported rates of specific anomalies from EUROCAT, CARIS and Mainz (Germany) 1998-2002 (showing 95% confidence limits).

(source: EUROCAT)



Why could rates for Wales be higher than elsewhere?

There are three explanations suggested for the relatively high rates in Wales:

1 Better reporting to CARIS

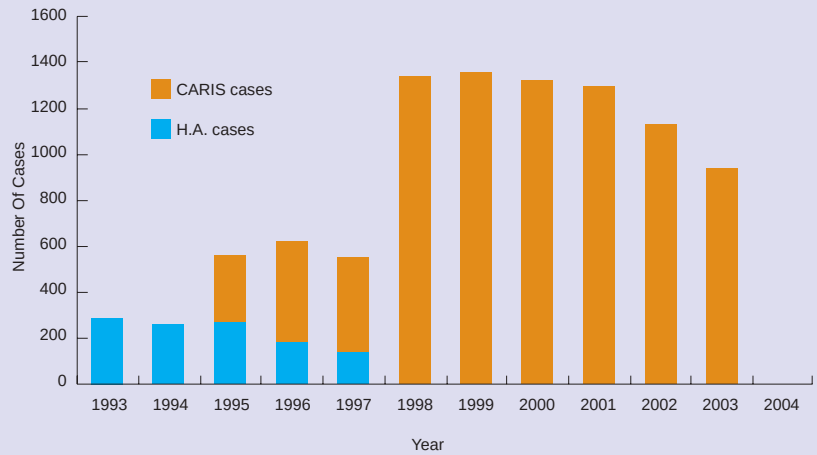
It could be possible that the true rates for Wales are broadly similar to elsewhere, but CARIS has a better reporting system than other areas and so misses fewer cases. This can be illustrated from analyses of reports made to ONS. Up to 31st December 1997, a Health Authority based reporting system to ONS operated in Wales (using Form SD56). This was replaced in 1998 by electronic reporting. As well as collecting data from 1998, CARIS also back-reported any cases for the previous three years (1995 – 1997) and this is shown in **Figure 2**. It illustrates some of the under-reporting that clearly occurred using the old Health Authority system and is evidence that the development of a modern system has led to improvements in data collection.

2 Double counting by CARIS

It could be possible that the true rates for Wales are broadly similar to elsewhere, but CARIS records cases incorrectly, duplicating cases and artificially inflating the figures. Because CARIS operates a multiple reporting system with bits of information coming from many sources, it would be quite easy to double count cases. To both assess the extent of any duplication and to remove any such cases from the system, CARIS undertakes a series of data quality checks every year before analysing data and calculating rates. Maternal and baby NHS numbers, postcode and maternal date of birth are used to identify duplicate cases. In the latest checks, 14 duplicate records and 1 triplicate record were found. Overall since CARIS began, a total of 56 duplicate cases have been identified out of 8201 potential cases – a duplication rate of less than 1%. Duplication of records is therefore unlikely to be a significant contributor to anomaly rates in Wales.

Figure 2

Cases of congenital anomaly reported to ONS using old Health Authority paper based system and CARIS



3 Rates of congenital anomaly in Wales are truly higher than many other areas in Britain and Europe

If congenital anomaly rates for Wales are truly higher than elsewhere, we would expect it to show up in other data sources apart from CARIS. However, CARIS was established because there was no mechanism for collecting accurate data on congenital anomalies in Wales.
This means that there is no parallel data source for congenital anomalies in Wales against which we can compare.
CARIS has looked at inpatient data and information from the child health system to see if these also reflect high rates. Unfortunately, coding issues and limited access to the child health computer system have so far prevented an in depth study. Inpatient statistics can only be useful to check anomalies where most cases are liveborn and are certain to be admitted to hospital. A recent small study into the usefulness of inpatient data suggests that it is not a good source against which to compare figures (although it is certainly helpful in identifying some cases).

Abortion statistics

Another source of routine data that may be helpful is abortion statistics. A legal abortion for congenital anomaly is justified under defined grounds:

Ground E – There is a substantial risk that if the child were born it would suffer from such physical or mental abnormalities as to be seriously handicapped.

Abortions overall in England and Wales have continued to rise in recent years with the vast majority of terminations being carried out for other grounds. Unfortunately, it is impossible to directly compare abortion statistics with CARIS data because of data confidentiality. However, if anomalies are truly higher and rising in Wales, similar trends might also be seen in official abortion statistics.

Between 1992 and 2002, the proportion of women in Wales who have had terminations under Ground E has varied from 0.7% to 1.1% with no particular trend over these years. Over the same period, the actual rate of terminations under Ground E for women aged 15 to 44 in Wales, has varied between 0.9 and 2.1 per 10,000, again with no discernable trend **(Figure 3 overleaf)**.
There is, therefore, no clear evidence from termination statistics to support the theory that congenital anomaly rates have risen in recent years.

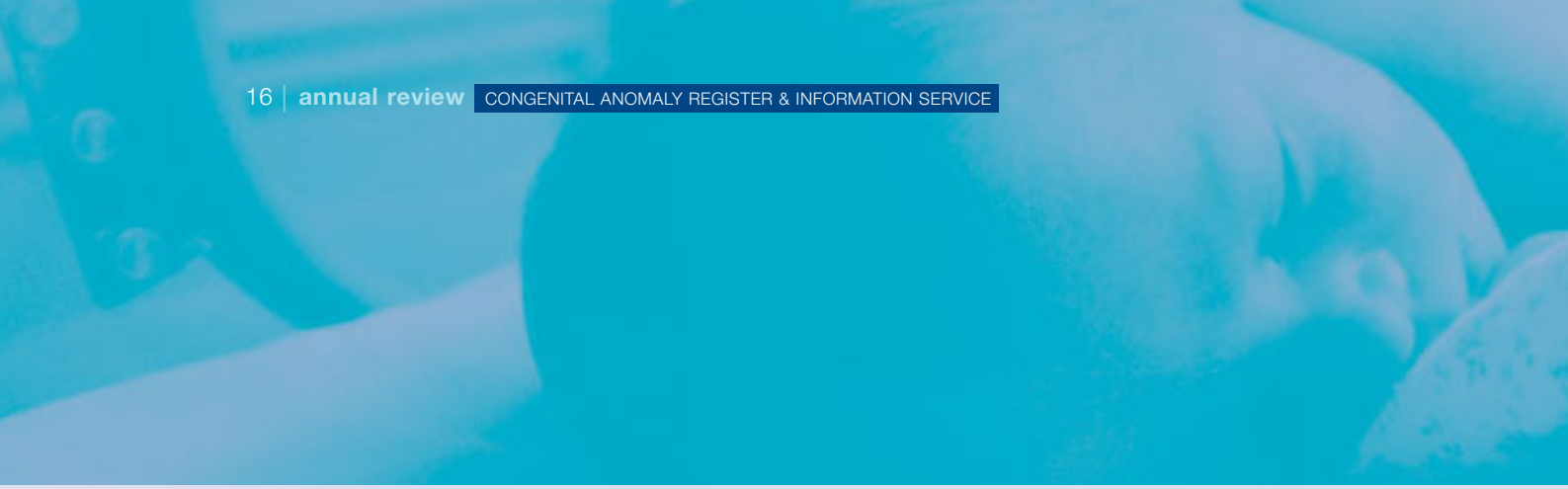
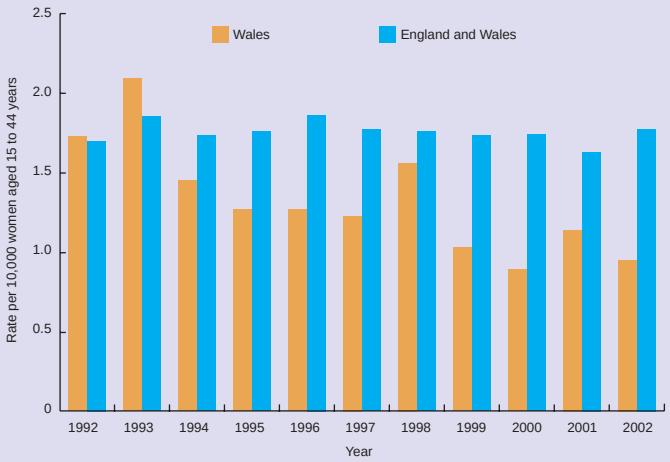


Figure 3

Abortions under ground E for England and Wales, 1992 to 2002

Source ONS



Differences across Wales

What is the situation?

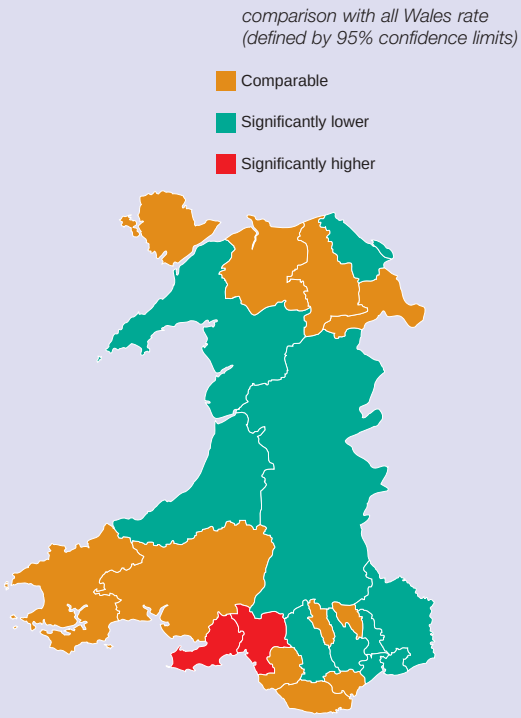
Differences in reported rates of anomaly are evident between Unitary Authorities in Wales, with Swansea and Neath Port Talbot particularly having higher rates than elsewhere **(Figure 4)**.

What are the explanations?

The three most likely explanations for the picture across Wales are differences in reporting, small number variation or a true difference in rates.

Figure 4

Comparison of gross rates of congenital anomaly for Welsh Unitary Authorities, 1998-2003



Differences in reporting across Wales

Table 4 summarises key features of reporting from different sources across Wales. Mid and West Wales have better reporting than elsewhere in Wales from various sources, particularly from Swansea NHS Trust, especially for cases identified after birth. This explains some of the apparently higher levels in Swansea and Neath Port Talbot. Chromosomal anomalies such as Down syndrome are reported via the national cytogenetics service and should not therefore be influenced by local reporting differences. In general, rates of Down syndrome for Swansea and Neath Port Talbot are not particularly higher than elsewhere, although for an accurate comparison, differences in maternal age between areas should also be taken into account (CARIS has not yet been in operation for long enough to allow this calculation).

Small number variation

Analysis of CARIS data at unitary authority level involves working with small numbers of cases, which can lead to wide variations in rates. As the database grows, figures will become more reliable as this effect reduces. Numbers for Swansea and Neath Port Talbot have been consistently high since CARIS began. It is unlikely that this would be the case if the rates were simply due to small number variation.

A true difference in rates

CARIS is aware that it would be dangerous to ascribe all differences in congenital anomaly rates simply to reporting practices and that some true differences will occur across Wales. Geographical variations in some anomalies have previously been reported – for example, the distribution of isolated cleft palate[3].

Reasons for differences need careful consideration, so that any underlying problems are correctly identified and acted upon. The public need to be kept informed of problems, but without causing inappropriate concern as a result of false alarms. Detailed investigations go beyond the service routinely provided by CARIS and may require complex studies and a multi-agency approach. An example of this is the current work on gastroschisis, which will be reported at a future date.

In order to provide useful information more quickly on the distribution of anomalies in Wales, we are now involved in a project in association with Health Solutions Wales and the University of Wales College of Medicine to assess the feasibility of developing a better surveillance system for congenital anomalies in Wales that would help us to detect potential problems more accurately and at an earlier stage.

The mainstay of good congenital anomaly data on which investigations can be based remains excellent clinical reporting of cases. CARIS continues to encourage clinicians to report as many cases as possible, to allow this work to continue.

[3] *CARIS Review 1998-2002. Published November 2003*

Table 4 Reporting sources for congenital anomalies across Wales

Reporting Source	South East Wales	Mid & West Wales	North Wales
Antenatal ultrasound	Variable between units	Excellent reporting	Excellent reporting
Other antenatal clinical reporting	Patchy	Patchy	Patchy
Postnatal clinical reporting	Remains generally weak	Excellent clinical reporting from Swansea NHS Trust, serving Swansea, Neath Port Talbot and East Carmarthenshire	Good reporting from paediatric audits in Wrexham, Conwy & Denbighshire in previous years
Karyotypes	Good national reporting via quarterly reports	Good national reporting via quarterly reports	Good national reporting via quarterly reports. Some cases not reported from Liverpool
Paediatric pathology	Good national reporting via quarterly reports	Good national reporting via quarterly reports	Fair national coverage via quarterly reporting. Some still done locally and some not reported from Liverpool
Patient Episode Database Wales (PEDW)	Good coverage for Monmouthshire, Blaenau Gwent and Merthyr. Partial coverage for Newport, Torfaen and Caerphilly. Cardiff data awaiting processing	Data processed for Bridgend only	Data processed from Conwy & Denbighshire. Wrexham data awaiting processing
Child Health System	Data processed from most trusts up to 2001	Data processed from most trusts up to 2002. No data from Carmarthenshire	Good reporting for the whole of North Wales
Paediatric cardiology	Good reporting via annual report from paediatric cardiologists	Good reporting via annual report from paediatric cardiologists	Good reporting via annual report from paediatric cardiologists at Alder Hey
Neonatal surgeons	Good reporting from 1998-2000. Declining reporting in 2001-2. No data yet for 2003	Served by UHW, Morriston, and Alder Hey. Good from Cardiff 1998-2000. Maxillo-facial and cleft repair good from Morriston. No link with Alder Hey	No direct link with surgeons at Alder Hey
Medical Genetics	Decline in reporting since 2002	Good reporting from Swansea	Decline in reporting since 2002
Postnatal ultrasound	Poor reporting	Fairly good, particularly for orthopaedics in Swansea	Poor reporting
Paediatric orthopaedics	None	Good reporting via Phillips Parade clinic in Swansea	Good reporting from Robert Jones & Agnes Hunt orthopaedic hospital Oswestry, covering North and Mid Wales
Ophthalmology	Occasional	Excellent clinical reporting from Swansea NHS Trust, serving Swansea, Neath Port Talbot and East Carmarthenshire	None