GIG CYMRU NHS WALES
Iechyd Cyhoeddus Cymru
Public Health Wales

Congenital Anomaly Register & Information Service for Wales

CARIS review 2022

Data from 1998 to 2022

This annual report includes the prevalence rates of key congenital anomalies and rare diseases in Wales, with a focus on abdominal congenital anomalies that affect nutrition. The updated prevalence rates includes the Official Statistics release of 2022 data.

Contents

Update on Childhood Rare Diseases 2022

Rare diseases in children have been registered since 2014 with data going back to 1998. We now hold data on 24,042 cases distributed over 1,253 diseases. The data tables and outputs that form our official statistics release are available here.

Update on Congenital Anomalies 2022

Since 1998 the CARIS team have registered 39,160 congenital anomaly cases in Wales. Of all live and still births in Wales, 4.8% are affected by a congenital anomaly. This proportion remains slightly lower than the 5.1% reported at the 2018 update, however, it is acknowledged that the COVID pandemic may have impacted data collection.

The proportion of babies affected by a congenital anomaly that were liveborn was 84.7%, with 96.9% surviving to 1 year of age. Of those with a recorded gender, 59.2% were male. These proportions remain similar to those reported previously. The 10-year prevalence rates by local authority region remain largely unchanged from last year's report and are summarised in Figure one.

A singular anomaly was reported in 57.5% of cases, consistent with last year's report. However, the proportion of cases with an underlying chromosomal disorder was higher at 14.3%, compared with 13.9% last year. The five largest groups of anomalies remained the same as reported last year and were circulatory, limbs, musculoskeletal, genetic/multi-site disorder and urinary.

In addition to the headline summary provided in this report, the data tables and outputs that form our official statistics release are available here, including breakdowns at Local Health Board level. For the first time we have broken down the congenital anomalies data according to the European Concerted Action on Congenital Anomalies and Twins (EUROCAT) headings for relevant conditions. The official statistics release also includes data on childhood rare diseases and antenatal detection rates.

Introduction

About 10% of congenital anomalies are related to the gastrointestinal system. They include those resulting from defects in the abdominal wall and those with a defect of the bowel itself. Many of these are diagnosed antenatally and the majority will require surgery. As a result, the best outcomes are achieved by a multidisciplinary team of ultrasonographers, obstetricians, neonatal paediatricians, paediatric surgeons and specialist paediatric nursing staff.

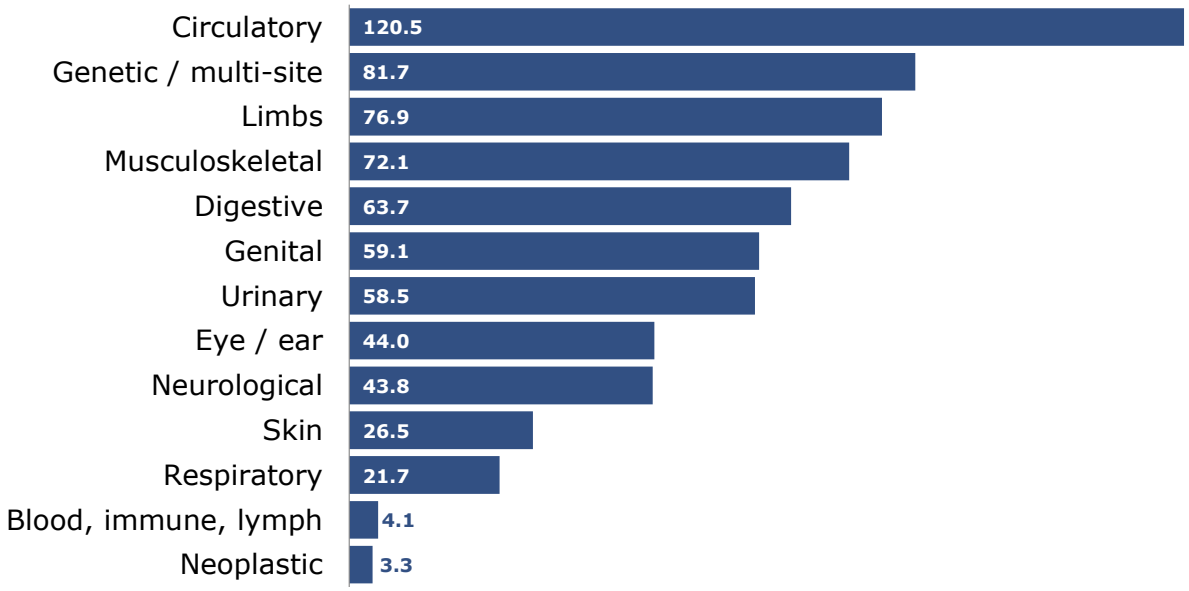
Anterior abdominal wall defects are the most common anomalies affecting nutrition though these are usually classified as anomalies of the musculoskeletal system.

Antenatal diagnosis for abdominal wall defects at the booking scan is very good with 98% detection in Wales . Fetal anomaly ultrasound standards (Antenatal Screening Wales) ensure that at the 20 week fetal anomaly scan the ultrasonographer can see:

- Normal sub diaphragmatic position of the stomach.
- Umbilical cord inserting into the anterior abdominal wall.
- No abdominal wall defect.

Other digestive anomalies are not as easy to detect with a rate of 36.9% quoted from England[1].

Main anomaly groups for cases reported to CARIS 1998-2022, rate per 10,000 total births



Produced by Public Health Wales Observatory, using CARIS (PHW) & PHB (ONS)

Exomphalos

This is also known as omphalocele and appears as a defect in the ventral abdominal wall through the umbilicus. The resulting hernia contains small bowel and liver covered in peritoneum. This herniation is a normal part of development from the sixth to the tenth week of development. An exomphalos is thought to be a failure of the bowel to return to the abdominal cavity[2].

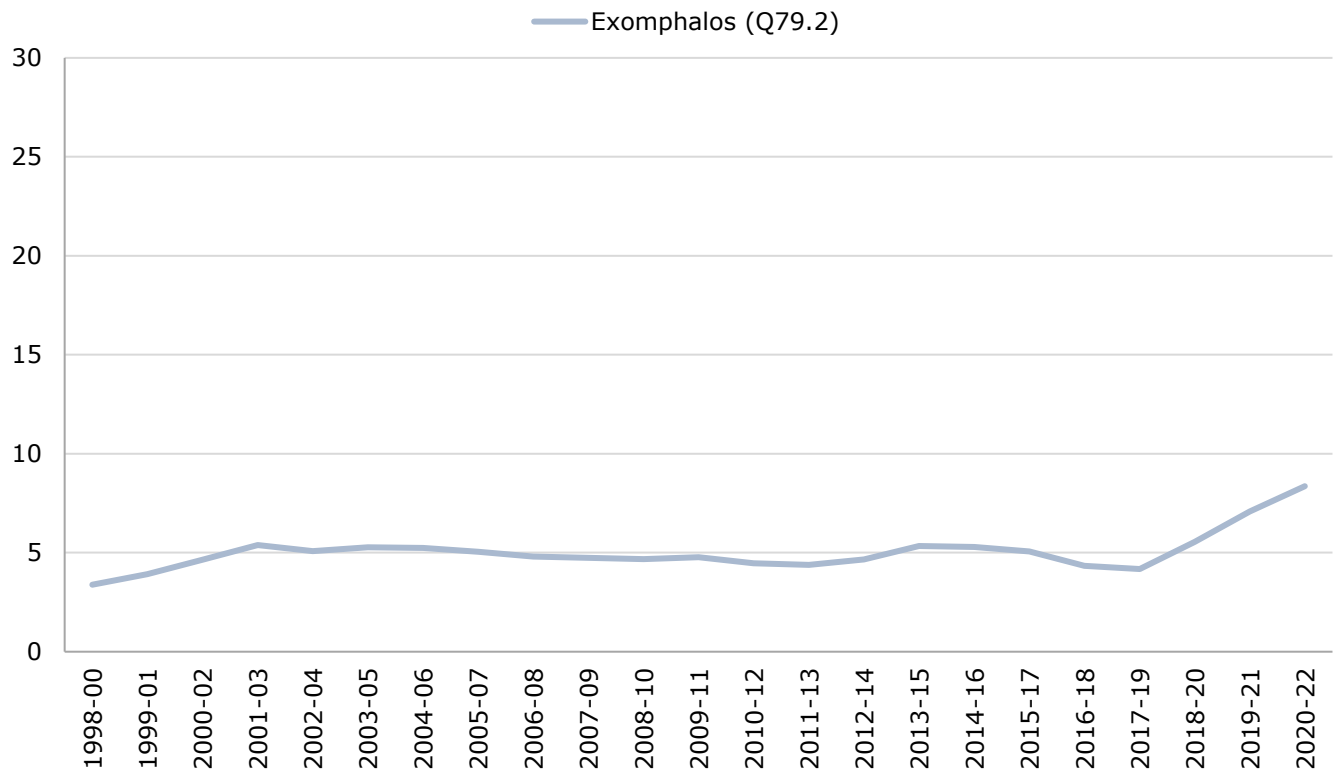
Risk Factors:

- Mother over 35years
- Mothers of Black ethnicity
- Multiple pregnancies
- More prevalent in the male fetus

Prevalence in Wales

The prevalence of exomphalos in Wales is 5 per 10,000 total births. From the graph there shows an increase in prevalence over the years. This may reflect the increase in older mothers with a concomitant increase in chromosomal anomalies.

Exomphalos cases, 3-year rolling rate per 10,000 births, Wales, 1998-00 to 2020-22
Produced by Public Health Wales Observatory, using CARIS (PHW) & PHB (ONS)



Associated anomalies

These are common occurring in as many as 75% of cases with cardiac anomalies reported as being the most common[3]. Chromosomal anomalies were present in 35.3% of cases in Wales.

Antenatal Diagnosis on ultrasound

A widened sac is seen at the umbilical cord insertion site containing bowel or liver. 95.3% were diagnosed antenatally in the 10 year period 2013 – 2022 in Wales.

Management

Vaginal delivery is possible. The baby is best delivered in a unit with access to early neonatal surgery. Management of any associated anomalies will need to be anticipated.

Outlook

This is dependent on the size of the exomphalos, the timing of diagnosis and the presence of other anomalies. The outlook is best when it is an isolated condition with no other anomalies.

Gastroschisis

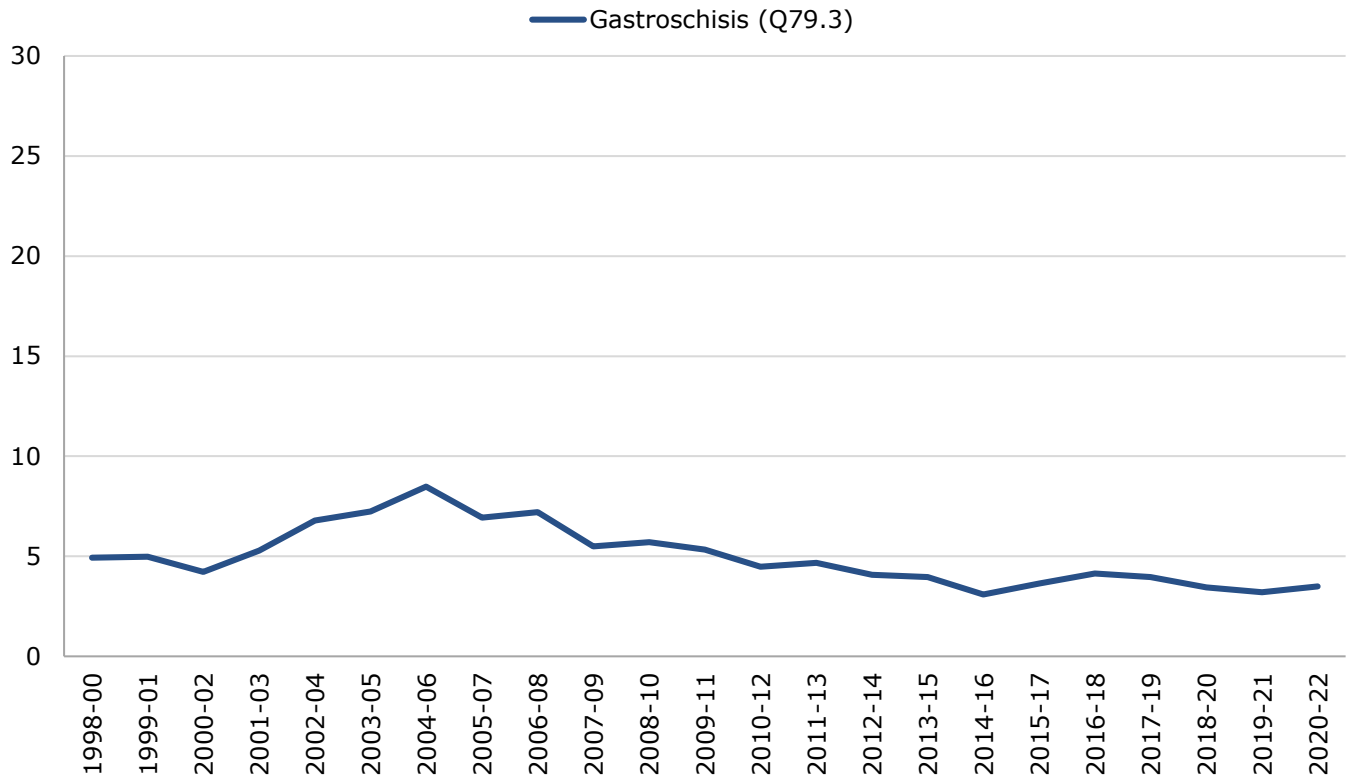
Gastroschisis is a defect in the anterior abdominal wall usually to the right of the insertion of the umbilical cord. This results in small bowel floating without a peritoneal covering in the amniotic cavity. It is classified as a musculoskeletal anomaly rather than a digestive one.

It is thought to occur in the fourth week of development as the lateral body wall folds form.

Prevalence

Gastroschisis is relatively common in Wales with a prevalence rate of 5 per 10.000 total births. This rate has fluctuated in the past but is latterly stable. The Welsh rate compares with an overall European figure of 2.75 per 10,000 total births[4].

Gastroschisis cases, 3-year rolling rate per 10,000 births, Wales, 1998-00 to 2020-22
Produced by Public Health Wales Observatory, using CARIS (PHW) & PHB (ONS)



Risk factors:

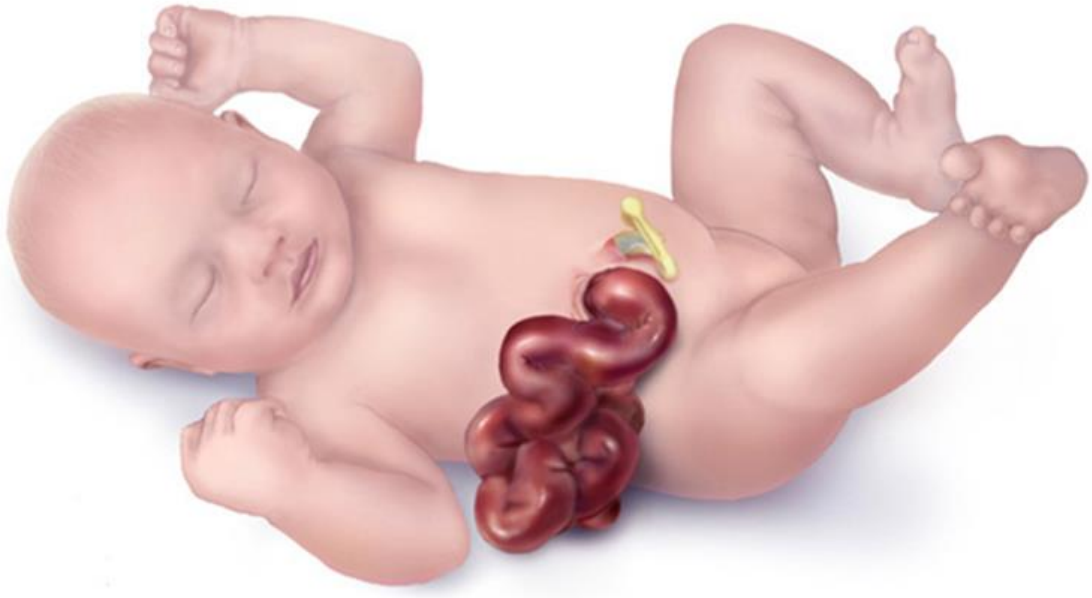
- Younger maternal age
- Maternal smoking
- Cannabis use
- Low maternal body mass index

Associated anomalies

These are uncommon looking at Welsh data and are mainly anomalies of the central nervous system. There are no known genetic associations but there may be a genetic susceptibility.

Antenatal Diagnosis on Ultrasound

In the last 3 years 100% were diagnosed on ultrasound in Wales with 75.8% detected at the dating scan at 12 weeks. The floating loops of bowel are readily seen in the amniotic cavity. There may be additional complications of oligohydramnios and fetal growth restriction.



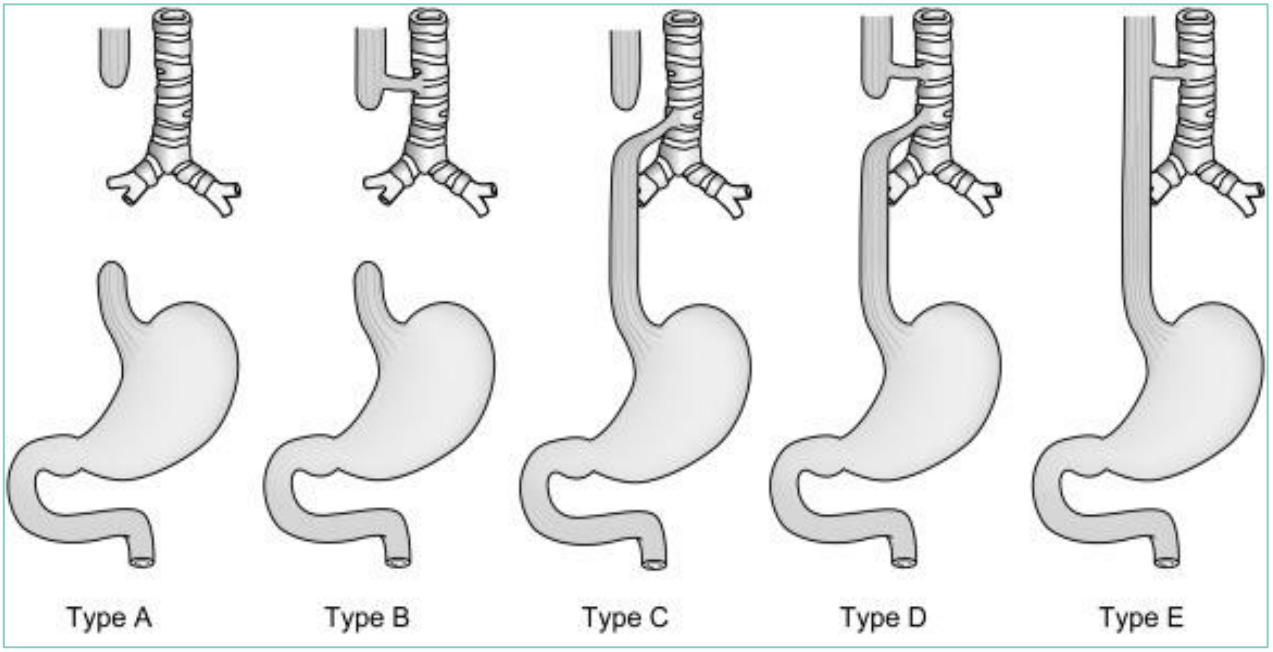
Outlook

From 1998 to 2022 6.4% were terminated with 2.7% stillborn. Overall neonatal survival is good with a 96% survival of live born babies at the age of 5 years. Postnatal complications can include sepsis, necrotising enterocolitis, short bowel syndrome, bowel obstruction and volvulus. These can contribute to difficulties with feeding.

Oesophageal atresia / Tracheo-oesophageal fistula (TOF)

What is oesophageal atresia/trachea oesophageal fistula?

This occurs when the upper part of the oesophagus is closed meaning that nothing can pass from the mouth to the stomach. In 89% of cases in Wales there is a connection with the trachea forming a tracheo-oesophageal fistula (TOF).



7-10%	**<1%**	**85-87%**	**<1%**	**4%**

When does it occur?

Early in pregnancy from the fourth week the foregut divides into oesophageal and respiratory parts and failure of this process can cause the problem.

How common is it?

It is the most common gastrointestinal congenital anomaly with 3.2 cases of oesophageal atresia/TOF for every 10,000 total births per year. This compares with 2.7 per 10,000 total births in 2021 in the European group of registers (EUROCAT).

Cases with Oesophageal atresia without fistula/Oesophageal atresia with fistula/Tracheo-oesophageal fistula without atresia, rate per 10,000 total births, Wales, 1998 to 2022

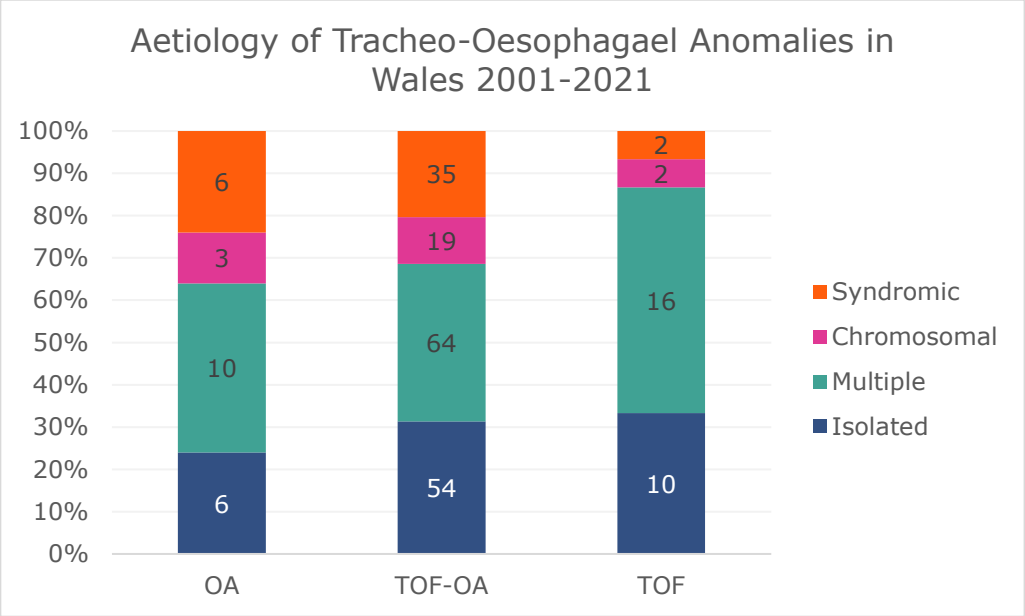
Anomaly	Total cases	Rate per 10,000 total birth
Oesophageal atresia without fistula	28	0.3
Oesophageal atresia with fistula	198	2.4
Tracheo-oesophageal fistula without atresia	39	0.5

Produced by Public Health Wales Observatory, using CARIS & PHB (ONS)

Causes and Associations

The cause is unclear and most cases occur sporadically with a recurrence risk of less than 1%. Additional anomalies can include trisomies 18 and 21, sirenomelia, Cri du Chat syndrome and VACTERL association (vertebral anomalies, anorectal anomalies, cardiac defects, trachea oesophageal fistula, limb defects).[5]

Results: Aetiology



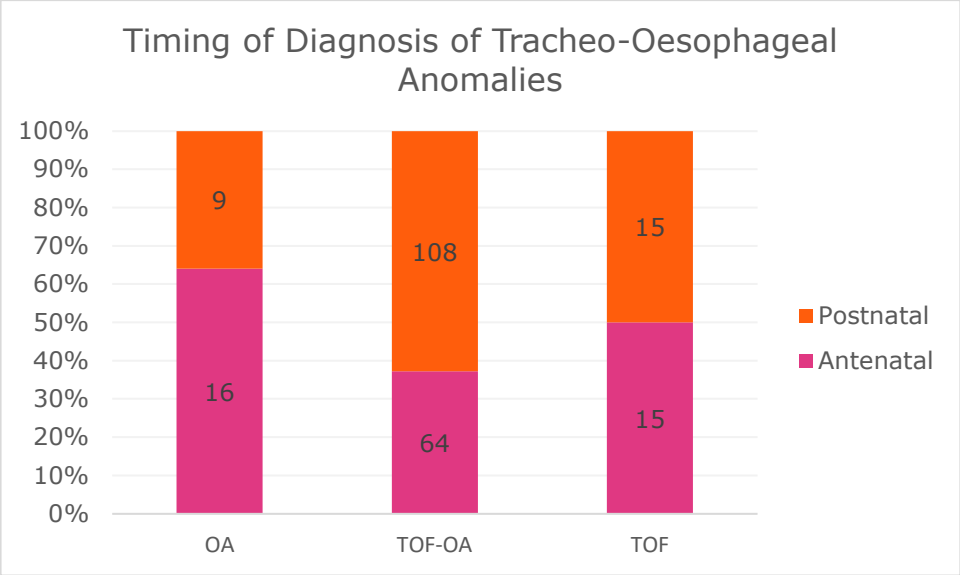
Goel N, Mendoza MK, Tucker, D, Morgan M, 2023

When is it diagnosed?

Often there are no clear signs at the 20 week scan except for a small or absent stomach. Polyhydramnios in the 3rd trimester due to an inability to swallow fluid can be an indication. 42% of cases in Wales were reported antenatally. The figure below shows that antenatal diagnosis is best in oesophageal atresia alone.

Diagnosis after birth is made when the baby produces frothy saliva, dribbling and breathing problems. The diagnosis is usually confirmed with an inability to pass a catheter down the oesophagus and a chest X ray showing gas in the bowel when there is a trachea-oesophageal fistula.

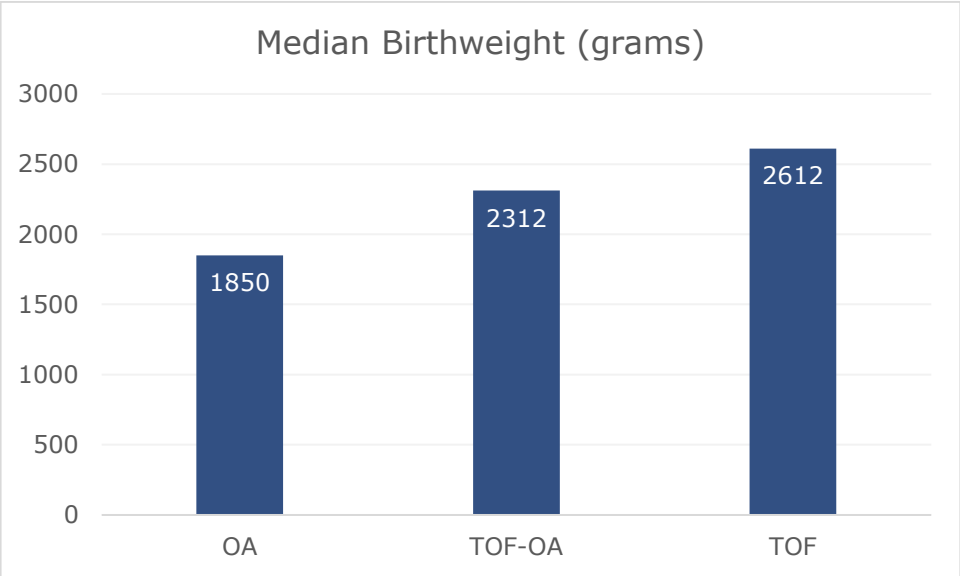
Results: Diagnosis



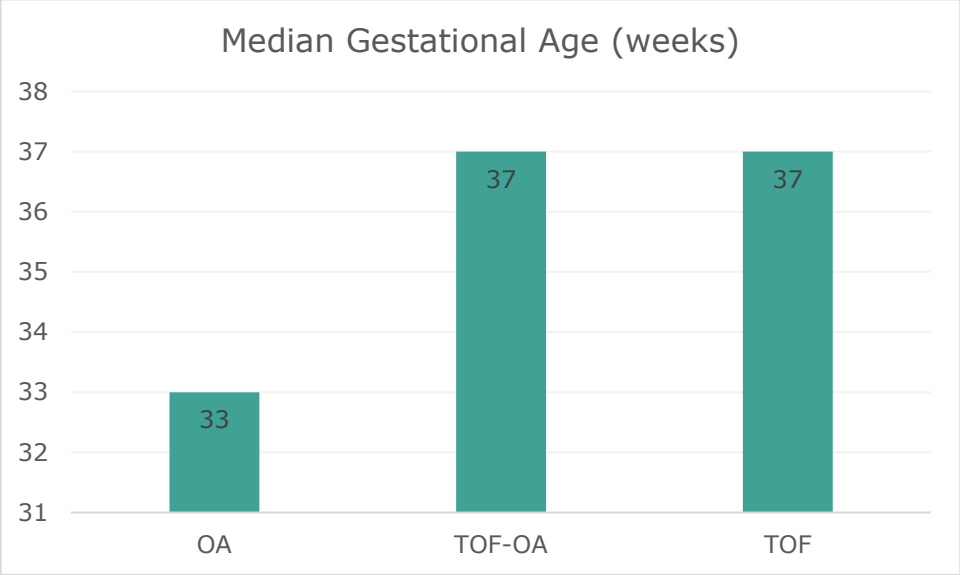
Goel N, Mendoza MK, Tucker, D, Morgan M, 2023

Outlook

Results: Demographics



Goel N, Mendoza MK, Tucker, D, Morgan M, 2023

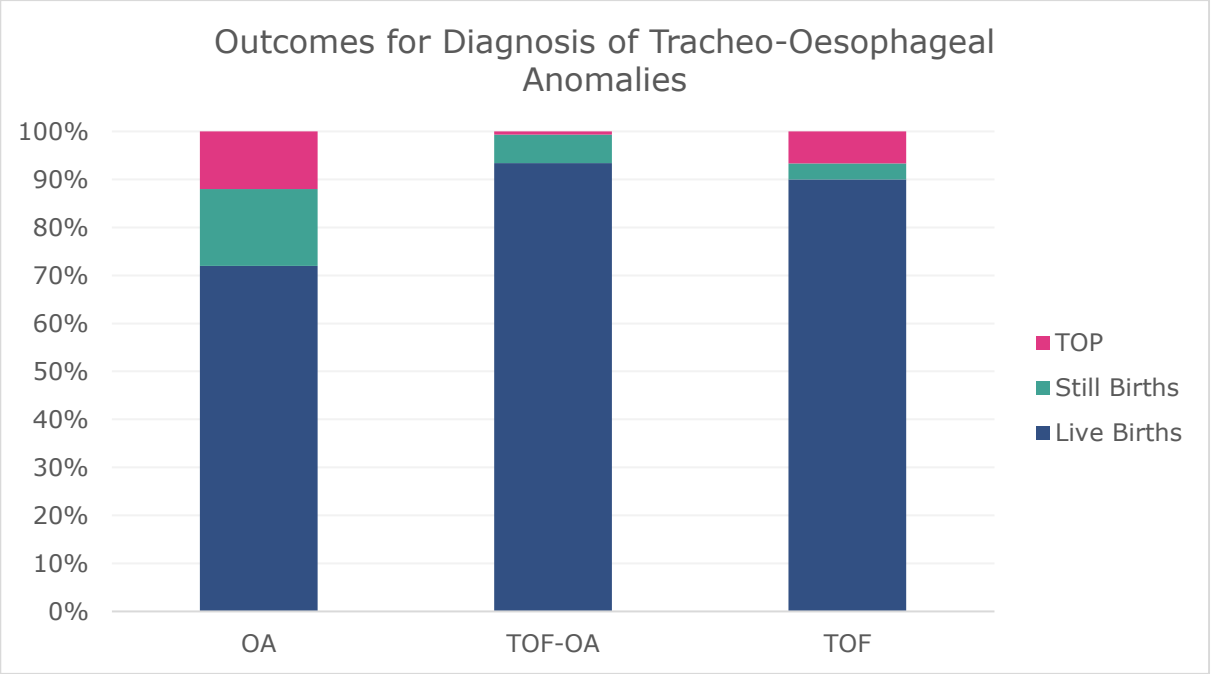


Goel N, Mendoza MK, Tucker, D, Morgan M, 2023

Polyhydramnios may precipitate premature delivery which can cause additional problems. This is more common in oesophageal atresia alone because of presumed difficulties with swallowing. Surgery for all the conditions soon after birth is required to repair the defects.

If there are no other anomalies the prognosis is good bearing in mind the challenge that feeding can pose in the postoperative period.

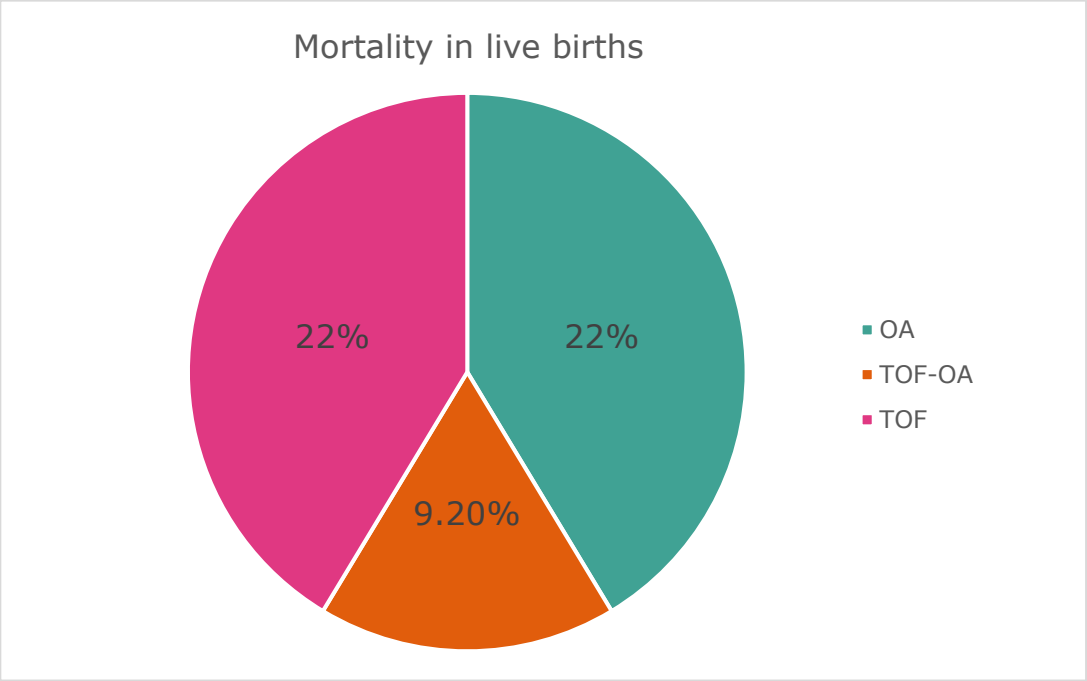
Results: Outcomes



Goel N, Mendoza MK, Tucker, D, Morgan M, 2023

Mortality in live births

Majority of babies who died were associated with multiple anomalies and had low birth weight.



Goel N, Mendoza MK, Tucker, D, Morgan M, 2023

Survival is the same (78%) for babies born with oesophageal atresia alone and those born with a trachea oesophageal fistula alone. Survival is better in those babies born with trachea-oesophageal fistula/oesophageal atresia combination (90.8%).

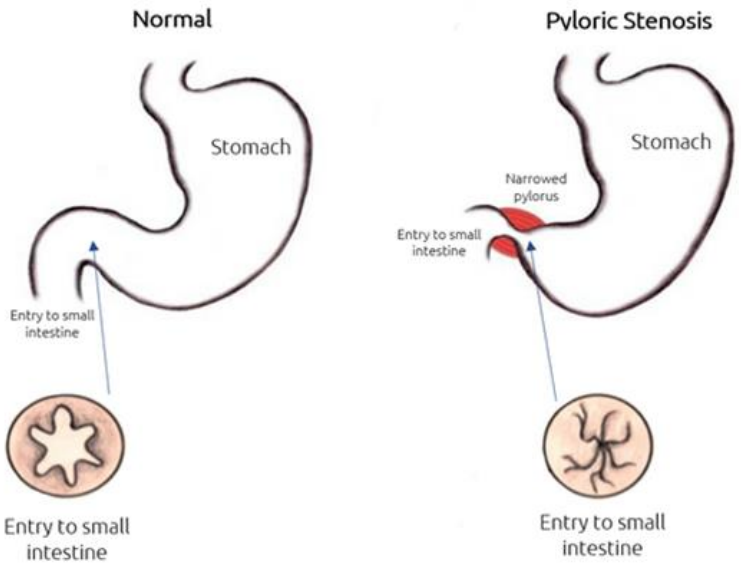
In a 6 year (2015-2021) post-surgery follow up, (Goel, personal communication) almost half of babies had an anastomotic stricture and a fifth required other surgical procedures.

Thanks to Dr Nitin Goel for his contribution to this update.

Pyloric Stenosis

This is the most frequent condition for a baby to need surgery in the first months of life.

Pyloric stenosis occurs when the pyloric sphincter or pylorus, the muscle ring between the stomach and duodenum, becomes thickened. This means that the stomach cannot empty. Projectile vomiting occurs as the stomach contents are expelled up through the oesophagus. The baby who is usually 3 to 6 weeks of age remains hungry and eager to feed.



Risk factors[6]:

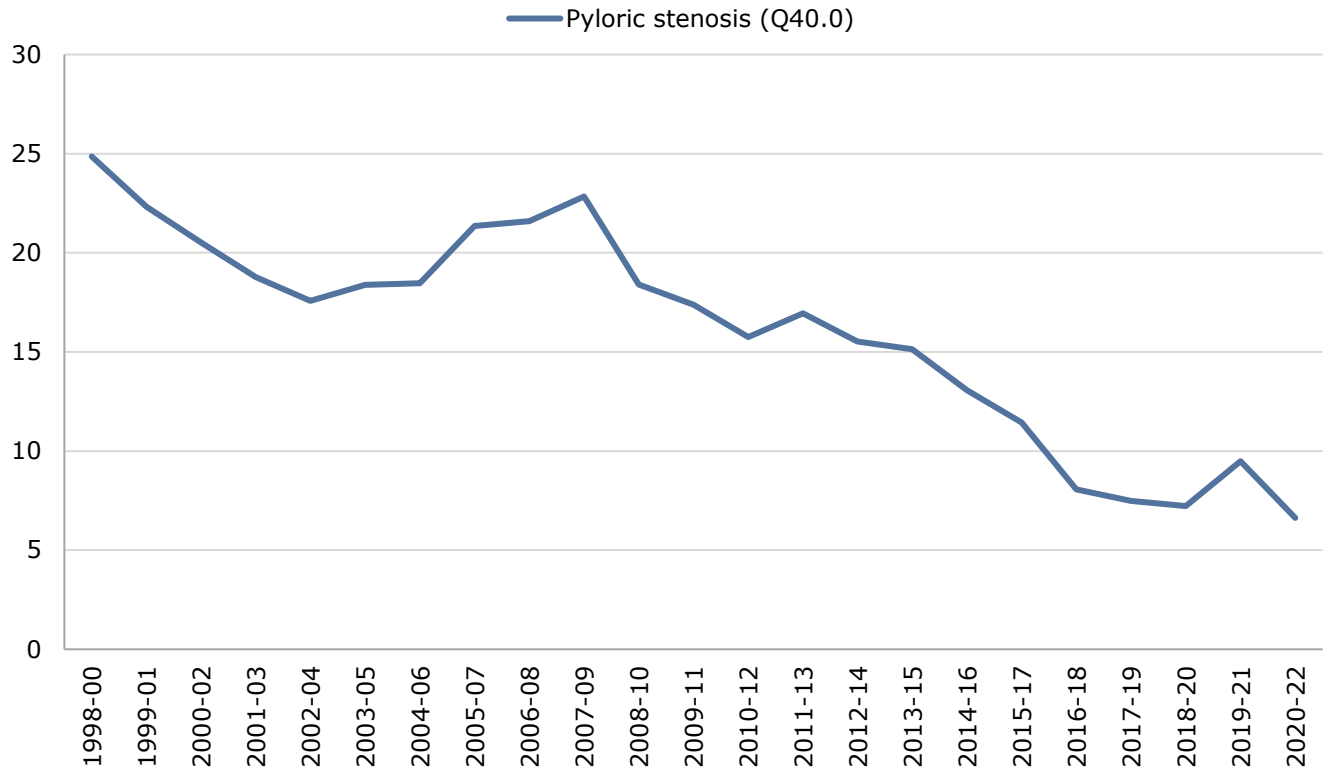
- Males
- Maternal smoking
- Caesarean section
- Primiparity
- Young maternal age
- Use of antibiotics (macrolides) in early neonatal life
- Possible genetic component

Prevalence

Overall, there were 16.3 babies born who developed pyloric stenosis per 10,000 total births (1998-2022). The graph shows a steady decline in prevalence from the 2010's. Other units have recorded a decline in prevalence[7] in the last decade also. The Wales data record a male to female ratio of 6:1. As pyloric stenosis is not classed as a congenital anomaly there are no comparable data from EUROCAT.

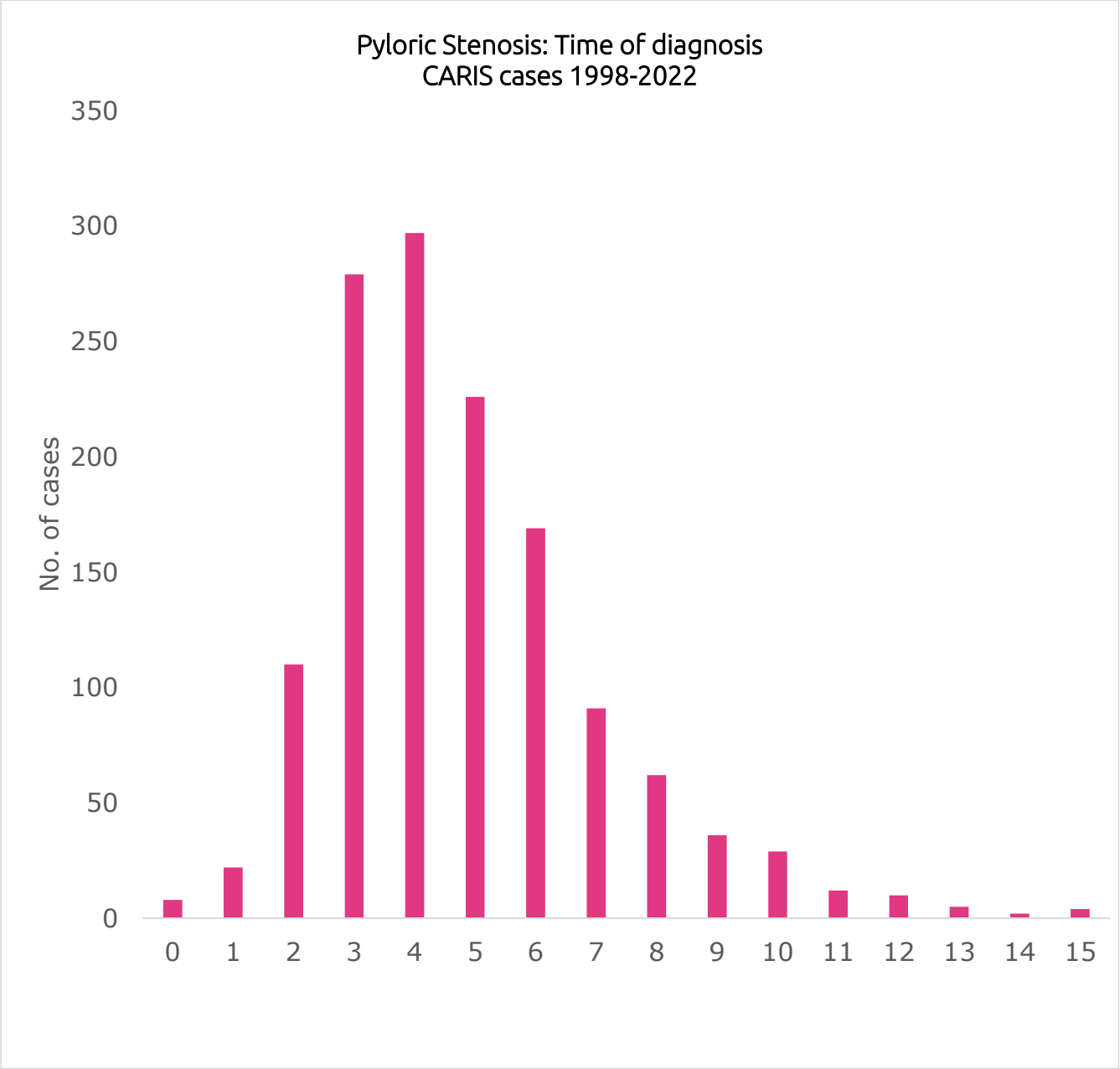
Of the 1427 CARIS cases 77 babies had a sibling with the same condition, including sets of twins. This supports the idea of a genetic origin.

Pyloric stenosis cases, 3-year rolling rate per 10,000 births, Wales, 1998-00 to 2020-22
Produced by Public Health Wales Observatory, using CARIS (PHW) & PHB (ONS)



Diagnosis

A history of non-bilious projectile vomiting usually prompts an examination of the baby. Visible gastric peristalsis may be seen and the thickened pylorus felt as similar to an olive. Ultrasound confirms the diagnosis with an increased thickness of the pylorus which is normally less than 3mm[8].



Management

Most babies begin to have symptoms between the third and the sixth week of life as can be seen in the graph.

After the baby has been rehydrated with normal electrolytes, surgery releasing the tight pylorus muscle (pyloromyotomy) is done. This can be done laparoscopically (keyhole surgery). Recovery is usually straightforward with a good outlook. From the CARIS data 98.5% of babies were operated within 7 days. The outcome of babies is excellent with a 99.5% survival rate at 1 year.

Thanks to Samantha Fisher (CARIS) for her contribution.

Multidisciplinary approach

Babies with abdominal anomalies affecting nutrition can pose challenges in their care that benefit from a multidisciplinary approach[9]. This begins with the initial antenatal ultrasound diagnosis and discussion with fetal medicine experts. Counselling can give an outline of the condition and its natural history to help parents navigate the rest of the pregnancy and the management afterwards. Involvement with paediatricians and paediatric surgeons is useful in the antenatal period to prepare parents for the birth, possible surgery and any future challenges. More recently paediatric palliative care has taken a valuable role when the outlook for the baby is unsure.

CARIS contribution to research in 2022/2023

Risk factors for mortality in infancy and childhood in children with major congenital anomalies: A European population-based cohort study.
J Tan, SV Glinianaia, J Rankin, A Pierini, M Santoro, A Coi, E Garne, ...
Paediatric and Perinatal Epidemiology

Ethics and legal requirements for data linkage in 14 European countries for children with congenital anomalies.
H Claridge, J Tan, M Loane, E Garne, I Barisic, C Cavero-Carbonell, ...
BMJ open 13 (7), e071687

Ten-year survival of children with trisomy 13 or trisomy 18: a multi-registry European cohort study.
SV Glinianaia, J Rankin, J Tan, M Loane, E Garne, C Cavero-Carbonell, ...
Archives of disease in childhood 108 (6), 461-467

Unravelling the clinical co-morbidity and risk factors associated with agenesis of the corpus callosum.
CJ Smith, ZG Smith, H Rasool, K Cullen, M Ghosh, TE Woolley, O Uzun, ...
Journal of Clinical Medicine 12 (11), 3623

Maternal age and the prevalence of congenital heart defects in Europe, 1995–2015: A register-based study.
C Mamasoula, T Bigirumurame, T Chadwick, MC Addor, ...
Birth Defects Research 115 (6), 583-594

Amniotic band syndrome and limb body wall complex in Europe 1980–2019.
JEH Bergman, I Barišić, MC Addor, P Braz, C Cavero-Carbonell, ...
American Journal of Medical Genetics Part A 191 (4), 995-1006

Surveillance of multiple congenital anomalies; searching for new associations.
J Morris, J Bergman, I Barisic, D Wellesley, D Tucker, E Limb, MC Addor, ...

Hospital Length of Stay and Surgery among European Children with Rare Structural Congenital Anomalies—A Population-Based Data Linkage Study.
E Garne, J Tan, M Damkjaer, E Ballardini, C Cavero-Carbonell, A Coi, ...
International Journal of Environmental Research and Public Health 20 (5), 4387

Epidemiology of aplasia cutis congenita: A population-based study in Europe.
A Coi, I Barisic, E Garne, A Pierini, M Addor, A Aizpurua Atxega, ...
Journal of the European Academy of Dermatology and Venereology 37 (3), 581-589

The burden of disease for children diagnosed with Klinefelter syndrome–a European cohort.
ALR Andersen, SK Urhoj, C Cavero-Carbonell, M Gatt, M Gissler, ... Researchsquare.com

Prevalence of vascular disruption anomalies and association with young maternal age: A EUROCAT study to compare the United Kingdom with other European countries.
JK Morris, D Wellesley, E Limb, JEH Bergman, A Kinsner-Ovaskainen, ...
Birth defects research 114 (20), 1417-1426

Prevalence of congenital heart defects in Europe, 2008–2015: A registry-based study.
C Mamasoula, MC Addor, CC Carbonell, CM Dias, ...
Birth defects research 114 (20), 1404-1416

A multicountry analysis of prevalence and mortality among neonates and children with bladder exstrophy.
Vijaya K, Sundar M, Lux A, Bakker M, Bergman J, Bermejo-Sánchez E, et al. American journal of perinatology (2022).

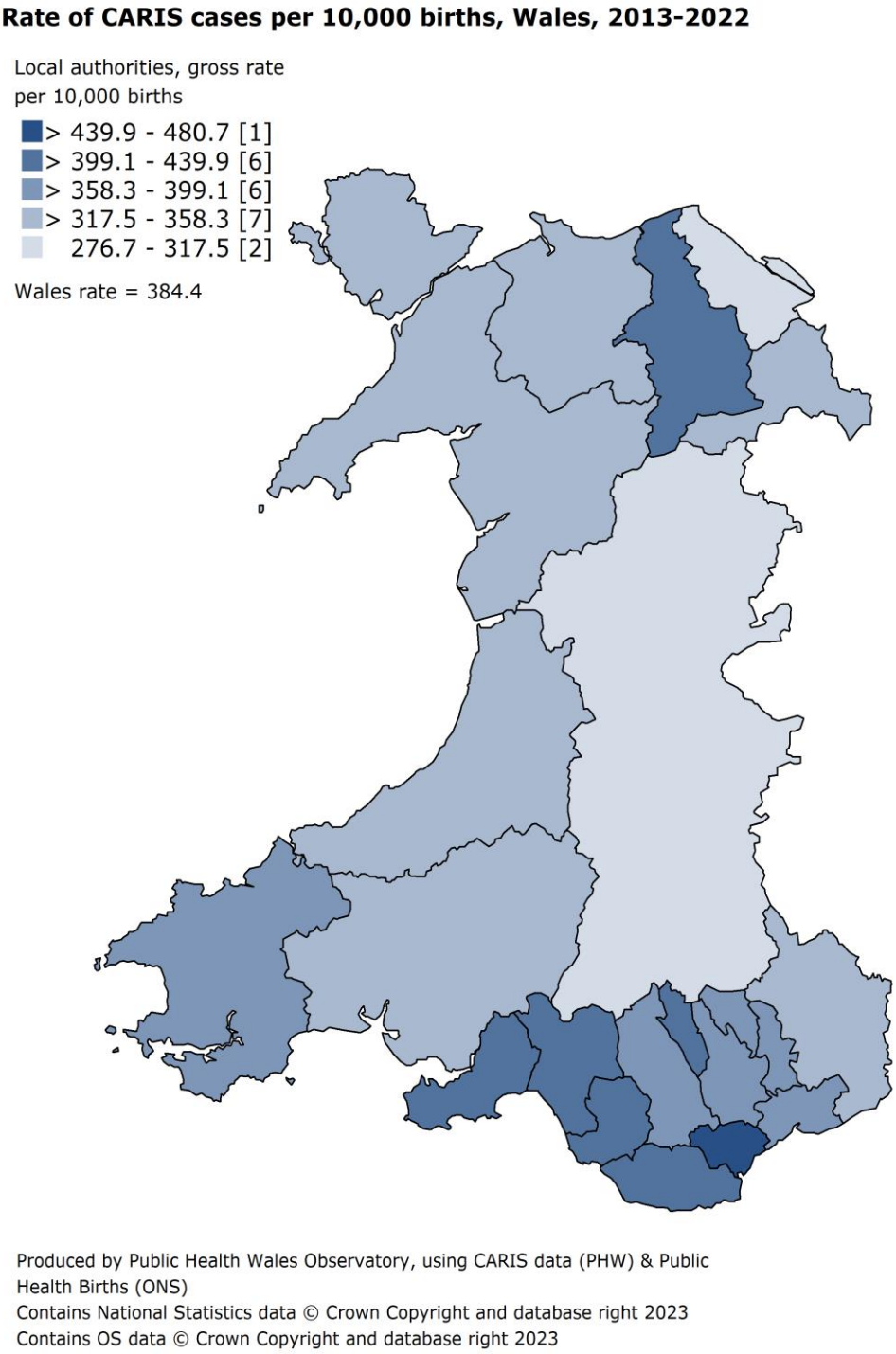
Prevalence and mortality among children with anorectal malformation: A multi-country analysis.
Vijaya K, Sundar M , Tandaki L, Lux A, Bakker M, Bergman J, Bermejo-Sánchez E, et al. Birth defects research 115, no. 3 (2023): 390-404.

A multi-program analysis of cleft lip with cleft palate prevalence and mortality using data from 22 International Clearinghouse for Birth Defects Surveillance and Research programs, 1974–2014.
Mc Goldrick, N., Revie, G., Groisman, B., Hurtado-Villa, P., Sipek, A., Khoshnood, B., Rissmann, A., Dastgiri, S., Landau, D., Tagliabue, G. and Pierini, A., et al. 2023. Birth Defects Research (2023)

A list of CARIS presentations and Journal Publications can be found here.

Figures

Figure one: Rate of CARIS cases per 10,000 births, Wales, 2013-2022



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Working together for a healthier Wales

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