GIG CYMRU NHS WALES
Iechyd Cyhoeddus Cymru
Public Health Wales

Congenital Anomaly Register & Information Service for Wales

CARIS review 2025

Data from 1998 to 2024

This annual report includes the prevalence rates of key congenital anomalies and rare diseases in Wales, with a focus on antenatal detection. The updated prevalence rates includes the Official Statistics release of 2024 data.

Contents

Introduction

This annual report:

1. Summarises the key congenital anomalies prevalence rates updates from the recent Official Statistics release that includes 2024 data.

2. Provides a detailed focus on antenatal detection rates.

This report is intended to be informative for health care professionals, patients, the public, and service planners.

Update on Congenital Anomalies 2024

Congenital anomalies data are reported at National level annually as an official statistic to monitor trends in prevalence and help inform public health or healthcare actions. To date the CARIS team have registered 745 cases for 2024 and further additional cases for previous years, taking the total number of registrations since establishment in 1998 to 41,530. Of these, 34,982 (84.2%) were liveborn.

Of all liveborn babies, 4% were affected by a congenital anomaly when considering all cases from 1998-2024. However, the proportion was lower at 3% for the last 10 years (2015-2024). Of all live and still births in Wales from 1998-2024, 4.8% were affected by a congenital anomaly. However, when considering the data for the last 10 years (2015-2024), the proportion was lower at 3.8%. Between 1998-2024 the rate of congenital anomalies was 477.3 per 10,000 births. The rate has reduced stepwise since 1998, see Table 1. We are unclear whether these changes are a true reduction in cases, or a function of under-reporting. Changes in practice with technology and due to the covid-19 pandemic means that the team undertake fewer site visits. This may have resulted in under-reporting.

Table 1: Congenital anomaly rate breakdown by 5-year bands

Years	Rate per 10,000 births
2005-2009	523.6
2010-2014	478.1
2015-2019	417.0
2020-2024	340.3*

** Expected to increase as further retrospective cases added for these most recent years*

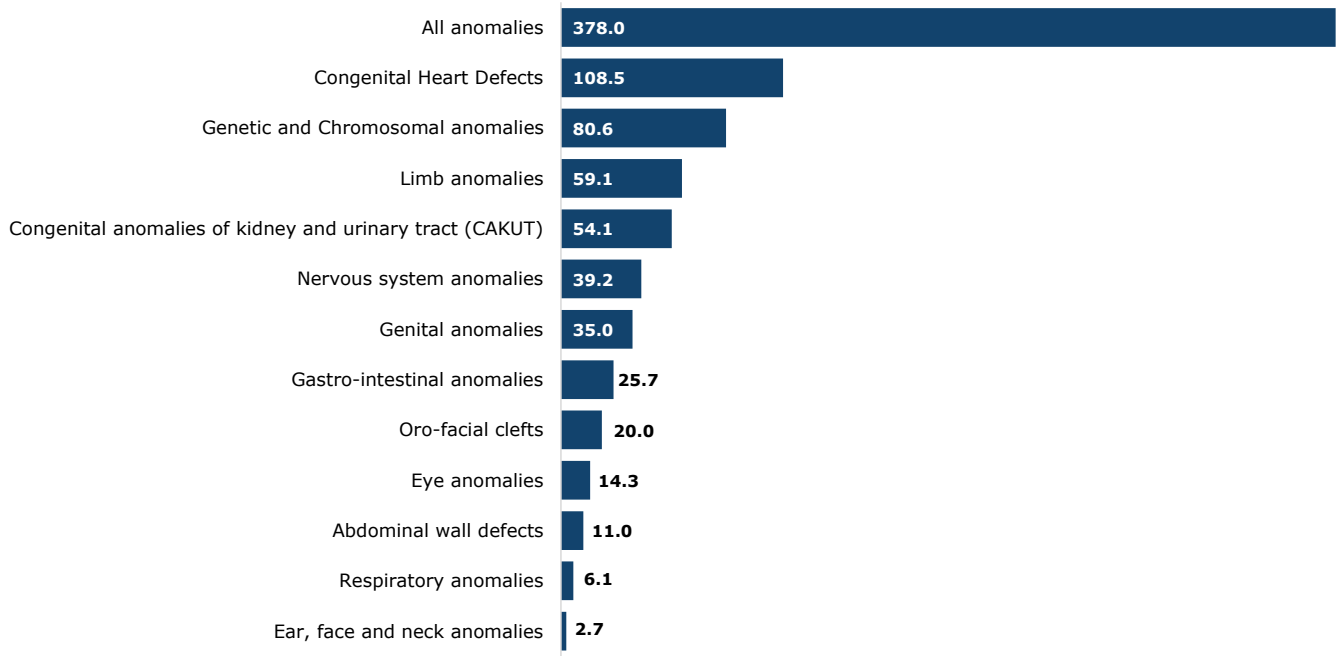
Of those with a recorded sex at birth between 1998-2024, 59.4% were male. This proportion remains similar to those reported previously and have not changed over time (58.7% for 2015-2024).

A singular anomaly was reported in 61.5% of cases between 1998-2024. When reviewing the past 10 years (2015-2024) data the proportion of singular anomaly was 56.9%. Singular anomaly cases are a proxy for ascertainment, because this will include less severe cases that are less likely to be reported. Our ascertainment of singular anomalies has historically hovered around 60%. However, the reduction over the past 10 years is suggestive that ascertainment may have reduced slightly during this period, possibly exacerbated by the covid-19 pandemic disruption to usual practice.

The main anomaly groups and corresponding prevalence rates when considering 1998-2024 data are given in Figure 1. When considering the data from the previous 10 years only (2015-2024) the top 5 groups of anomalies remain the same, although, 'Congenital anomalies of kidney and urinary tract (CAKUT)' have moved up to 3rd place, with a reciprocal drop down to 4th place for 'Limb anomalies'.

Figure 1

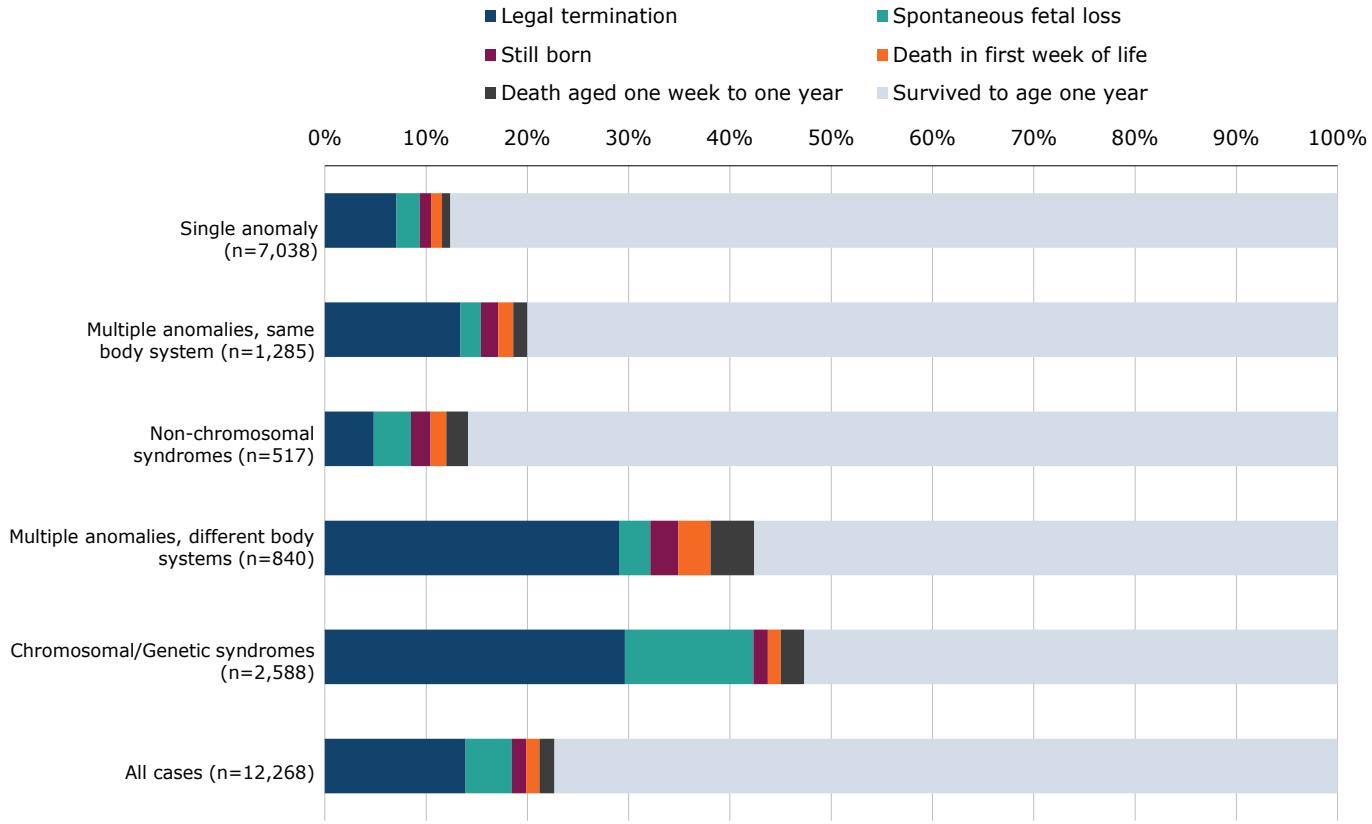
Main anomaly groups for cases reported to CARIS 1998-2024, rate per 10,000 total births
Produced by Public Health Wales Observatory, using CARIS (PHW) & PHB (ONS) & NCCHD (DHCW)



Of all cases reported to CARIS with a congenital anomaly between 1998-2024, 84.2% resulted in a live birth. During the past 10 years (2015-2024), 79.4% resulted in a live birth. This may be explained by decreasing ascertainment of less serious conditions. Survival to 1 year of age has remained constant with time at 96.8%. Survival summaries by high level groupings of anomaly types are shown in Figure 2. Survival is poorer for babies with multiple anomalies across different body systems, or babies with chromosomal syndromes.

Figure 2

Congenital anomalies, outcome of pregnancy by pattern of anomalies, pregnancies ending 2014-2023 (followed up to end 2024)
Produced by Public Health Wales Observatory, using CARIS (Public Health Wales)



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. The official statistics release also includes data on ultrasound scan antenatal screening detection rates. These data are reported to Antenatal Screening Wales for review as an annual audit.

Focus on antenatal detection rates

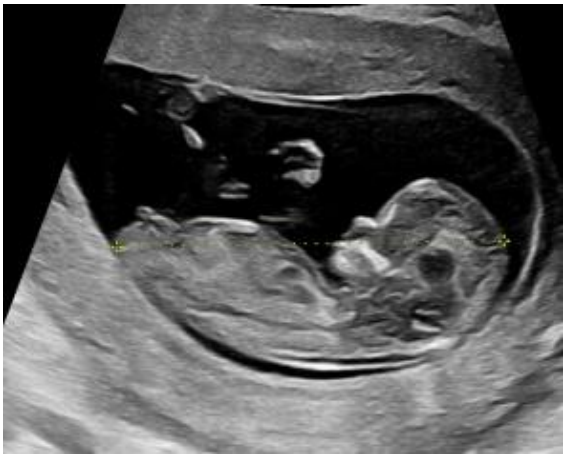
Antenatal screening in Wales includes a combination of maternal blood tests, an early ultrasound scan at booking (typically between 12–14 weeks), and a detailed anomaly scan at 20 weeks. The 20-week scan plays a crucial role in the detection of structural congenital anomalies.
Over recent years, the antenatal detection rate for congenital anomalies has continued to improve. This progress is largely attributed to:

- Advancements in ultrasound technology
- Enhanced training and expertise of sonographers
- Standardised protocols and pathways

For reporting purposes, we further analyse the conditions recommended by the Fetal Anomaly Screening Programme (FASP) to monitor the antenatal detection rate. To do so, we only include cases that are eligible for antenatal screening for consistency and accuracy.

Exclusions from the dataset include:

- Mothers who decline screening
- Early terminations or miscarriages
- Late bookers who miss the routine scans

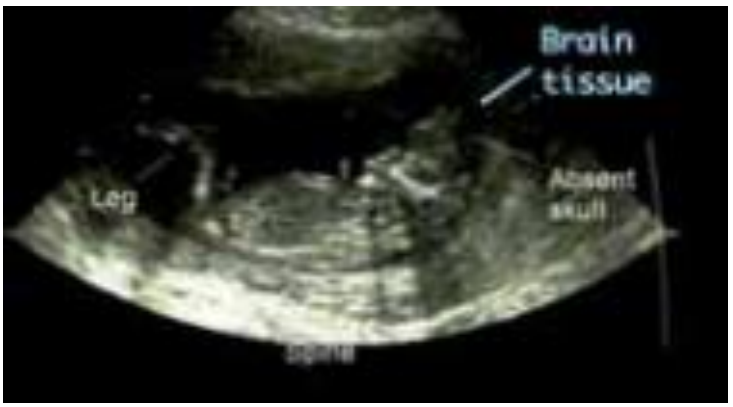


Picture 1: Booking scan picture showing Crown-Rump Length (CRL) measurement for gestational age.

Nervous system congenital anomalies

Neural Tube Defects (NTDs) are the commonest among this group, resulting from incomplete closure of the neural tube during early embryonic development. These defects typically occur within the first month of pregnancy and can lead to significant structural abnormalities of the brain and spinal cord. In Wales, the prevalence of neural tube defects is approximately 14.8 per 10,000 births (CARIS 2009-2024). These anomalies are highly detectable via prenatal ultrasound, often during the mid-trimester anomaly scan. The risk factors are multifactorial including genetic and non-genetic factors including exposure to a wide range of environmental hazards[1].

Anencephaly is one of the most severe forms, characterized by the absence of major portions of the brain and skull. It is invariably fatal. The detection remains consistently high at almost 100% for the period 2015-2024.



Picture 2: The ultrasound picture presents anencephalic fetus at booking scan.

Encephalocele involves herniation of brain tissue and meninges through a defect in the skull, forming a sac-like protrusion. The detection rate was around 90% from 2015 - 2019, then it dropped to the lowest point in 2020-2023 of ~73% and improved again rising to about 89% in 2022-2024.

Spina bifida is the most common NTD, where the spinal column fails to close completely, potentially leading to neurological impairment and physical disability. The detection rate for spina bifida was 90% in 2015-2017, improved over the years to 93% in 2020-2022, and was 92.9% in 2022-2024.

Hydrocephalus. Over a 10-year period 68 cases of hydrocephalus were reported to CARIS. Of these, 39 cases (57.7%) were detected during the routine anomaly scan, highlighting the moderate sensitivity of prenatal ultrasound in identifying hydrocephalus.

These data underscore the importance of anomaly scans in early detection, while also suggesting that a significant proportion of cases may be missed or diagnosed later in pregnancy or postnatally.

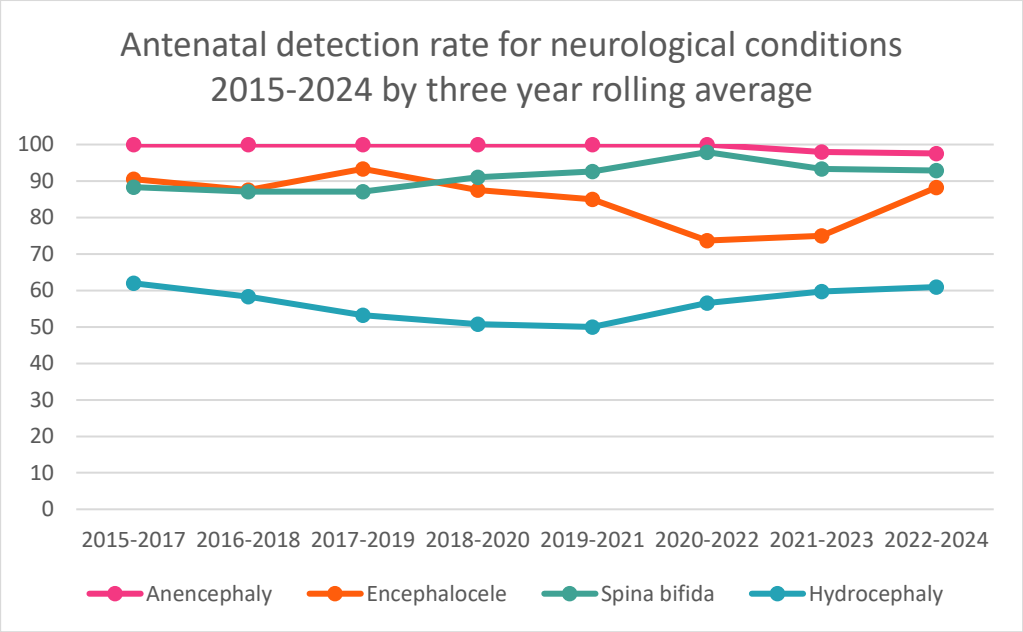


Figure:3: The graph presents 3 years rolling average number of cases for anencephaly, encephalocele, spina bifida and hydrocephalus reported to CARIS, 2015-2024.

Gastrointestinal tract congenital anomalies

Gastroschisis is a defect in the anterior abdominal wall, typically located to the right of the umbilical cord insertion, resulting in extrusion of abdominal contents such as intestines and occasionally the stomach. The exposed bowel is vulnerable to amniotic fluid damage, leading to inflammation and potential complications and exposure to air post-delivery. It requires surgical input postnatally.

- Prevalence: 4.1 per 10,000 births in Wales between 2009 and 2024. This decreased from 4.4 per 10,000 births in 2009-2017, to 3.7 per 10,000 births in 2018-2024. This decrease in prevalence is being observed across Europe[2].
- Risk Factors: Low maternal age, smoking, poor nutrition, low body mass index, illicit drug use and urinary tract infections[3,4].
- Prognosis: Excellent, with a 95% survival rate in live-born infants following immediate surgical repair post delivery[5].

Over a 10-year period (2015–2024), 106 cases were reported to CARIS, with 101 diagnosed before or during the anomaly scan, yielding a detection rate of ~95%.

Congenital Diaphragmatic Hernia (CDH) involves protrusion of abdominal organs into the thoracic cavity, impairing lung development. Early gestational onset and associated anomalies (e.g., cardiac defects, chromosomal abnormalities, hydrops, polyhydramnios, and fetal growth restriction) are linked to poor fetal prognosis.

The detection rate was ~68% in 2015–2017 and increased to ~77% during 2019–2021, then declined to ~60% in 2022–2024. In the last three years (2022–2024) 34 cases were registered, with 19 detected at anomaly scan. This decline in detection warrants further investigation into systemic or healthcare-related factors, potentially including resource limitations or service disruptions.

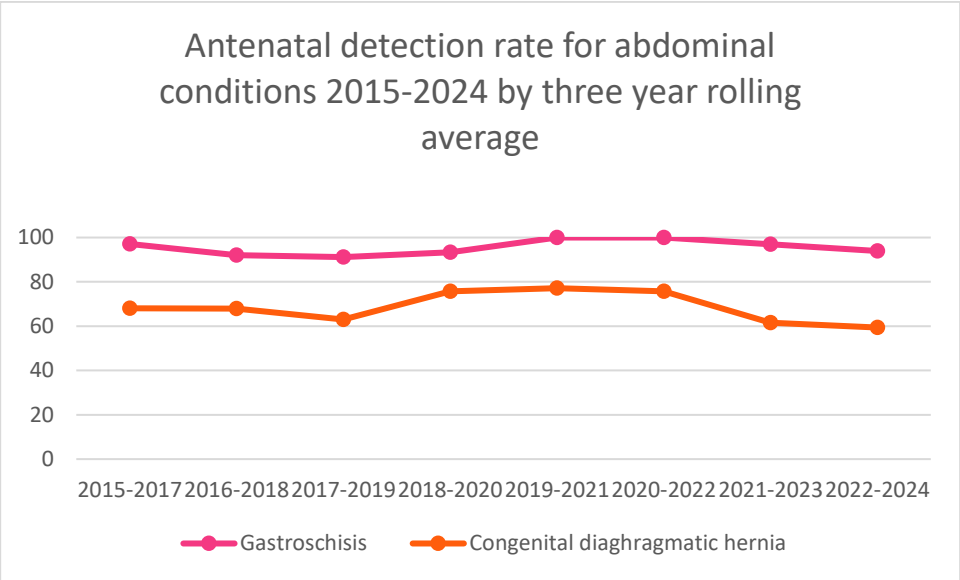


Figure 4. The graph presents 3 years rolling average number of cases for gastroschisis and congenital diaphragmatic hernia (CDH) reported to CARIS, 2015-2024.

Renal Congenital anomalies

Bilateral renal agenesis is a rare but severe congenital anomaly characterised by the complete absence of both kidneys. This condition is not compatible with life, so most mothers choose to terminate the pregnancy (~70%) with the remainder resulting in stillbirth or dying within a few hours of birth due to pulmonary hypoplasia (CARIS, 2015-2024).

The prevalence in Wales is 1.43 per 10,000 births (2015-2024).

The pathophysiology associated with oligohydramnios leads to Potter's syndrome or sequence, a constellation of fetal deformities caused by intrauterine compression that includes:

- Low-set ears
- Wide-set eyes
- Micrognathia
- Limb contractures
- Talipes
- Pulmonary hypoplasia

Antenatal detection trends vary slightly with a detection rate ~76% in 2015–2019, an increase to ~82% in 2017-2021, followed by a small decline to ~77% in 2020-2024. Bilateral renal agenesis detection rates are calculated over a 5-year rolling average due to small numbers.

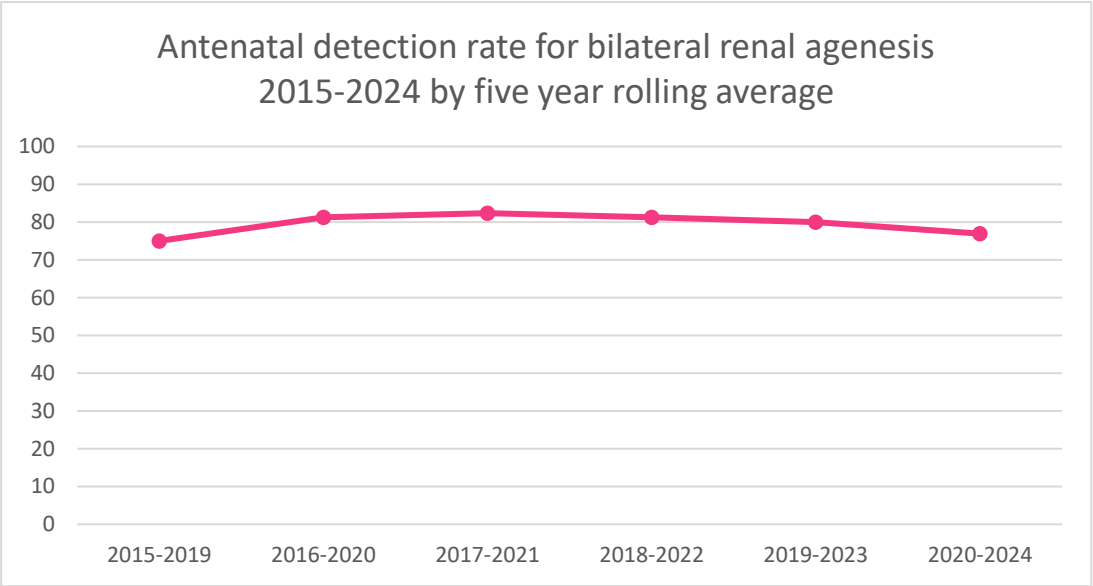


Figure 5: The graph presents 5 years rolling average number of cases for bilateral renal agenesis reported to CARIS, 2015-2024.

Cardiac Congenital anomalies

Congenital heart defects (CHDs) affect approximately 1% of live births, making them the most common birth defect and a leading cause of infant and childhood morbidity and mortality worldwide[6].

Antenatal detection of cardiac anomalies has improved globally, largely due to:

- Enhanced imaging techniques
- Systematic assessment of heart chambers and outflow tracts
- Increased awareness and training among sonographers

Early detection is crucial for planning the mode of delivery and ensuring birth occurs in a specialist-equipped unit where postnatal treatment for the fetus is immediately available. This significantly improves outcomes and survival rates.

Antenatal screening aims for optimal detection of major cardiac anomalies during the 20-week anomaly scan.

Hypoplastic Left Heart Syndrome (HLHS): Where the left side of the heart including left atrium, ventricle and aorta are underdeveloped. This results in serious problems for the baby after birth. Antenatal detection helps to improve outcomes by keeping the venous ductus arteriosus open after birth with medical treatment initially, followed by definitive surgical management[7].

The detection rate is 97–100% from 2015–2024.

Transposition of the Great Arteries (TGA): The aorta and pulmonary arteries are 'transposed' with the aorta arising from the right ventricle and the pulmonary artery from the left ventricle, causing two separate circulations of blood. A connection between the two circulations is needed for oxygenated blood to reach the body tissues.

The antenatal detection trends upwards from 72% in 2015-2017 to 93% in 2017-2019. It is followed by a steady decline to ~77% in 2022-2024. Overall, the prenatal detection of TGA has improved over the years[8].

Atrioventricular Septal Defects (AVSD): A congenital cardiac anomaly characterised by an incomplete formation of the atrial and ventricular septa along with malformation of the atrioventricular valves. This leads to left-to-right shunting and varying degrees of atrioventricular valve regurgitation. AVSD can be classed as partial, transitional or complete, with complete being the most severe[9].

The antenatal detection rates followed an upward trend from ~63% in 2015-2017 to ~77% in 2022-2024, with a dip noted in 2019-2021 to ~62%.

Tetralogy of Fallot (TOF): A congenital cardiac malformation that consists of four component parts which are ventricular septal defect, override of the ventricular septum by the aorta, pulmonary stenosis and right ventricular hypertrophy[10]. TOF occurs in 3 of every 10,000 live births and accounts for around 7% of all cardiac malformations[10,11].

The antenatal detection rate was ~59% in 2015-2017, peaked at ~77% during 2018–2020 then declined to ~68% in 2022–2024.

Coarctation of the Aorta (CoA): Coarctation of aorta is the narrowing of the aorta that can be at any location on the aortic arch or thoracic or abdominal aorta. The most common site is superior to the left subclavian artery at the insertion of the ductus arteriosus. Coarctation accounts for 5-8% of all congenital heart defects[11].

The antenatal detection is a persistent diagnostic challenge, the detection rate estimated at ~42% in 2015-2017, declined to ~16% by 2021–2023 with slight recovery to ~27% in 2022–2024. It remains the most difficult cardiac anomaly to detect antenatally, with concerns over the downward trend in recent years. Prenatal diagnosis of CoA is challenging and often produces both false-negative and false-positive results, with only the most severe cases showing earlier in pregnancy[12].

These trends highlight both the successes and ongoing challenges in antenatal cardiac screening. Whilst HLHS detection is nearly universally detected antenatally, CoA remains a diagnostic concern. Continued investment in training and technology is essential to improve detection rates across all cardiac anomalies.

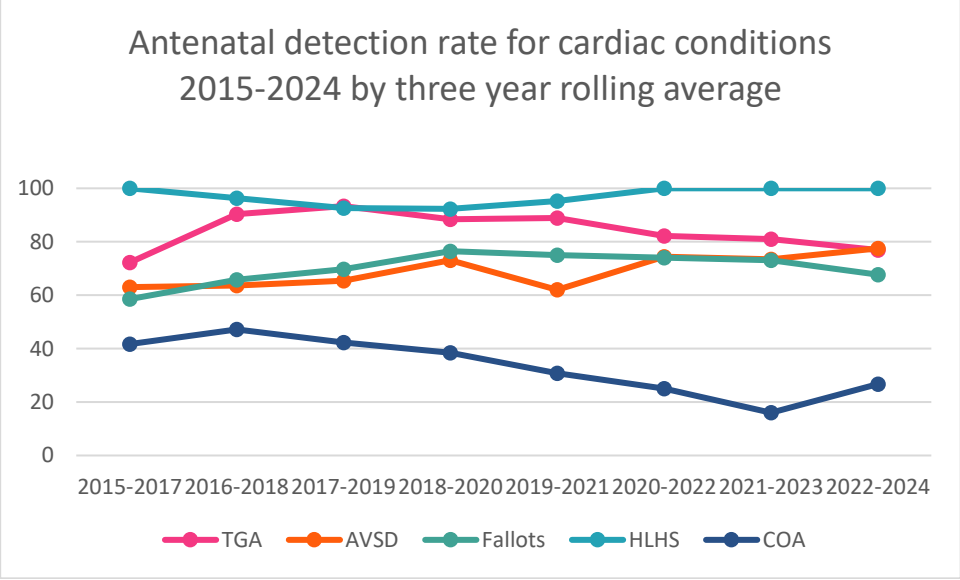


Figure 6: The graph presents 3 years rolling average number of cases for TGA, AVSD, Fallots, HLHS, and COA reported to CARIS, 2015-2024.

CARIS contribution to research in 2024/2025

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, et al.
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, et al.
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, et al.
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, et al.
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, et al.
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, et al.
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, et al.
European Journal of Human Genetics 32, 407-412

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, et al.
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, et al.
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, et al. 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, et al.
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, et al.
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.

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