GIG CYMRU NHS WALES
Iechyd Cyhoeddus Cymru
Public Health Wales

Congenital Anomaly Register & Information Service for Wales

CARIS Review 2024

Data from 1998 to 2023

This annual report includes the prevalence rates of key congenital anomalies and rare diseases in Wales, with a focus on cardiac anomalies. The updated prevalence rates includes the Official Statistics release of 2023 data.

Contents

Introduction

This annual report:

1. Describes the new childhood rare diseases Local Health Board level data output.

2. Summarises the key congenital anomalies prevalence rates updates from the recent Official Statistics release that includes 2023 data.

3. Provides a detailed focus on cardiac congenital anomalies.

This report is intended to be informative for patients, the public, healthcare professionals and service planners. Although, the more detailed information about cardiac anomalies is also intended to provide continuing professional development information targeted at healthcare professionals.

Update on Childhood Rare Diseases 2023

Rare diseases in children have been registered since 2014 with data going back to 1998. We now publish data on cases distributed over 380 diseases. The data tables and outputs that form our official statistics release are available here.

Following stakeholder discussions and feedback via the Rare Diseases Implementation Network (RDIN) Wales, we have provided data at Local Health Board (LHB) level for the first time. Data at LHB level was requested to help inform healthcare service needs. The LHB level data have focussed on 13 diseases in the first instance, selecting those with greater numbers of cases to allow meaningful reporting.

The key observation from the LHB level data is that patients with rare diseases are distributed across all LHB regions. Some data are suppressed for some diseases in the smaller LHBs, such as Powys, as the numbers are too low for reporting. Variation in disease prevalence rates are noted between LHBs, such as Kawasaki disease ranging from 3.26 per 10,000 total births in Cardiff and Vale University Health Board to 5.33 per 10,000 total births in Cwm Taf Morgannwg University Health Board. However, given the relatively low numbers of cases per LHB, such comparisons must be interpreted with caution as observed variations are expected.

We will share a survey with stakeholders to seek feedback about this new output.

Update on Congenital Anomalies 2023

Congenital anomalies data are reported at National level annually to monitor trends in prevalence and help inform public health or healthcare actions. Since 1998, the CARIS team have registered 40,261 congenital anomaly cases in Wales. Of all live and still births in Wales, 4.8% are affected by a congenital anomaly. Of those with a recorded sex at birth, 59.2% were male. These proportions remain similar to those reported previously.

A singular anomaly was reported in 57.2% of cases and the proportion of cases with an underlying chromosomal disorder remained at 14.3%. The main anomaly groups and corresponding prevalence rates are given in Table 1.

Table 1

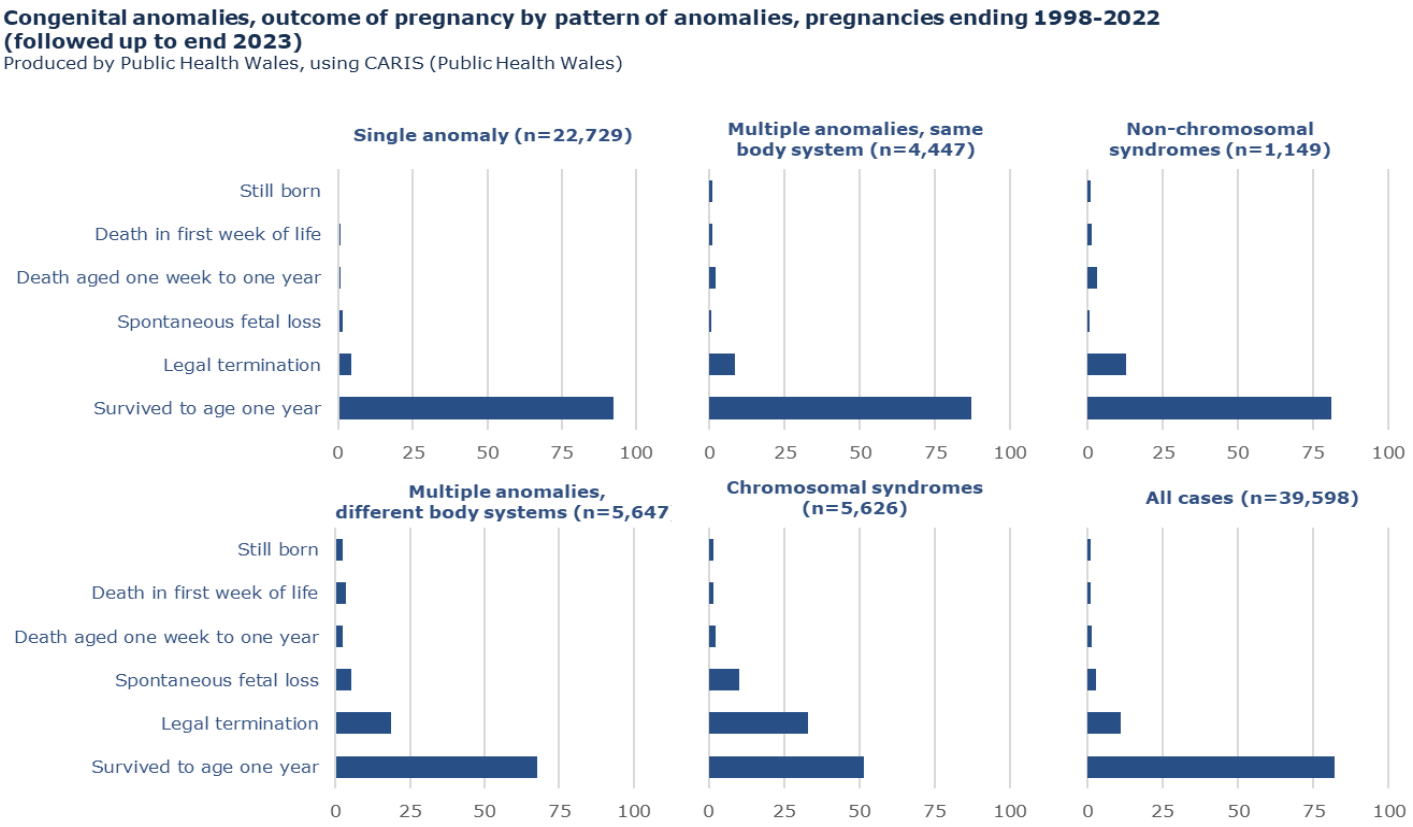
Main anomaly groups for cases reported to CARIS 1998-2023, rate per 10,000 total births

	Count	Rate per 10,000 total births
Circulatory	10,027	118.9
Genetic / multi-site	6,857	81.3
Limbs	6,455	76.6
Musculoskeletal	6,074	72.0
Digestive	5,338	63.3
Urinary	4,947	58.7
Genital	4,933	58.5
Neurological	3,742	44.4
Eye / ear	3,458	41.0
Skin	2,251	26.7
Respiratory	1,758	20.8
Blood, immune, lymph	346	4.1
Neoplastic	272	3.2

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

Of the cases reported to CARIS with a congenital anomaly, 84.5% resulted in a live birth, with 96.9% of these surviving to 1 year of age. Survival summaries by high level groupings of anomaly types are shown in Figure 1. Survival is poorer for babies with multiple anomalies across different body systems or babies with chromosomal syndromes.

Figure 1



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 screening ultrasound scan antenatal detection rates. These data provide an audit and are reported to Antenatal Screening Wales for review.

Cardiac focus

Figure 2

Cases with anomalies of the heart, rates per 10,000 total births and percentage of cases liveborn, Wales, 1998-2023

Anomaly	Total cases	Average cases per year	Rate	% of cases liveborn	Trend (3 year rolling rate)
Coarctation of aorta	448	17	5.3	92.6	6.6 … 2.7
Fallot's	313	12	3.7	91.4	3.2 … 3.5
Double outlet right ventricle	201	8	2.4	75.6	2.6 … 2.6
Hypoplastic left heart syndrome	263	10	3.1	45.2	3.2 … 3.2

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

Cases with anomalies of the heart, rates per 10,000 total births and percentage of cases liveborn, Wales, 2014-2023

Anomaly	Total cases	Average cases per year	Rate	% of cases liveborn	Trend (3 year rolling rate 2014-16 onwards)
Coarctation of aorta	134	13	4.4	92.5	5.3 … 2.7
Fallot's	119	12	3.9	86.6	4.0 … 3.5
Double outlet right ventricle	96	10	3.1	76.0	3.4 … 2.6
Hypoplastic left heart syndrome	85	9	2.8	42.4	2.3 … 3.2

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

Coarctation of aorta

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. Cases are usually sporadic and more common in males, although some familial links are reported[6].

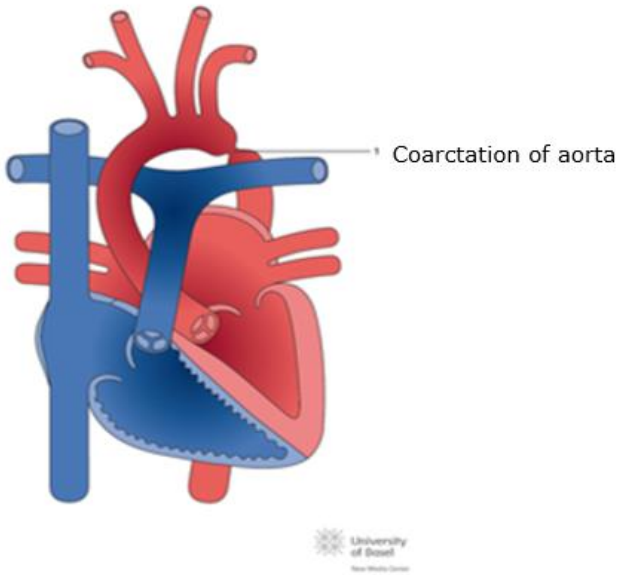


Illustration taken from http://www.chd-diagrams.com

The narrow lumen of the aorta results in high blood pressure in the upper body and, if untreated, may progress to ventricular dysfunction, aortic aneurysm, aortic dissection and cerebral artery disease in the third or fourth decade of life.

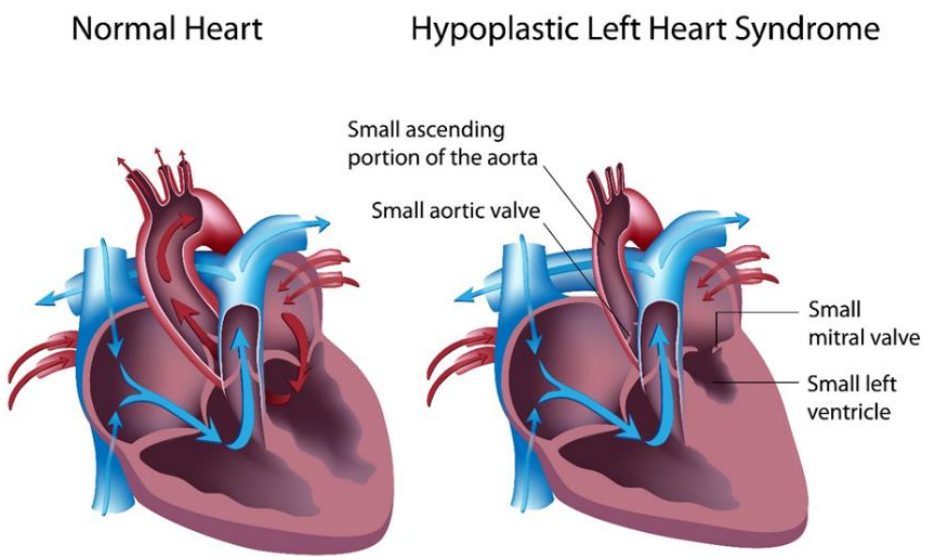
Patients with Turner syndrome (XO) have a higher risk of isolated coarctation of aorta[3]. Chromosome karyotype testing is recommended for female coarctation of aorta cases. Coarctation of aorta usually coexists with a bicuspid aortic valve and a ventricular septal defect.

In Wales from 1998-2023, 448 cases of coarctation of aorta were reported. This equates to 17 cases per year or 5.3 cases per 10,000 total births. The livebirth rate is almost 93% among these babies. The number of Coarctation of aorta cases are less than half in 2021- 2023 compared to 1998-2000, so the CARIS team will monitor and explore reasons. See Figure 2.

Hypoplastic left heart syndrome (HLHS)

Hypoplastic left heart syndrome (HLHS) is a congenital heart disease associated with under development of left sided heart structures such as the left atrium, left ventricle, mitral valve, aortic valve, aortic arch and ascending aorta. Mitral valve, aortic valve and foramen ovale atresia or stenosis results in HLHS. The most common abnormality is aortic valve stenosis. The defective blood flow results in hypoplasia of the left heart and affects the growth of the baby. The prevalence is 1 in 5,000 neonates[4] and accounts for 3% of congenital cardiac anomalies[4,5].

Previously, when no treatment options were available, HLHS was associated with an almost 100% mortality in the first weeks of life[4]. Now the continuous infusion of prostaglandin E1 can keep the ductus arteriosus (DA) patent until corrective surgery.



There were 263 cases reported in Wales from 1998-2023 for HLSH (see figure Y), equating to an average of approximately 10 cases per annum. Almost half of the cases were liveborn (45.2%). All cases since 2020 have been detected in the antenatal period. There were 3.1 cases per 10,000 total births.

Amongst liveborn babies over the past 10 years 26 (72.2%) were delivered in tertiary centres, whilst 10 (27.8%) were delivered in other centres and transferred to tertiary centres for surgery.

The prevalence rates have varied somewhat year on year. However, there has been a raise from 2.3 per 10,000 total births in 1998-2000 to 3.2 per 10,000 total births in 2021-2023.

Tetralogy of Fallot (TOF)

Tetralogy of Fallot (TOF) is 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[1]. TOF occurs in 3 of every 10,000 live births and accounts for around 7% of all cardiac malformations[1,2]. An aortic arch is present in 1 out of 4 cases.

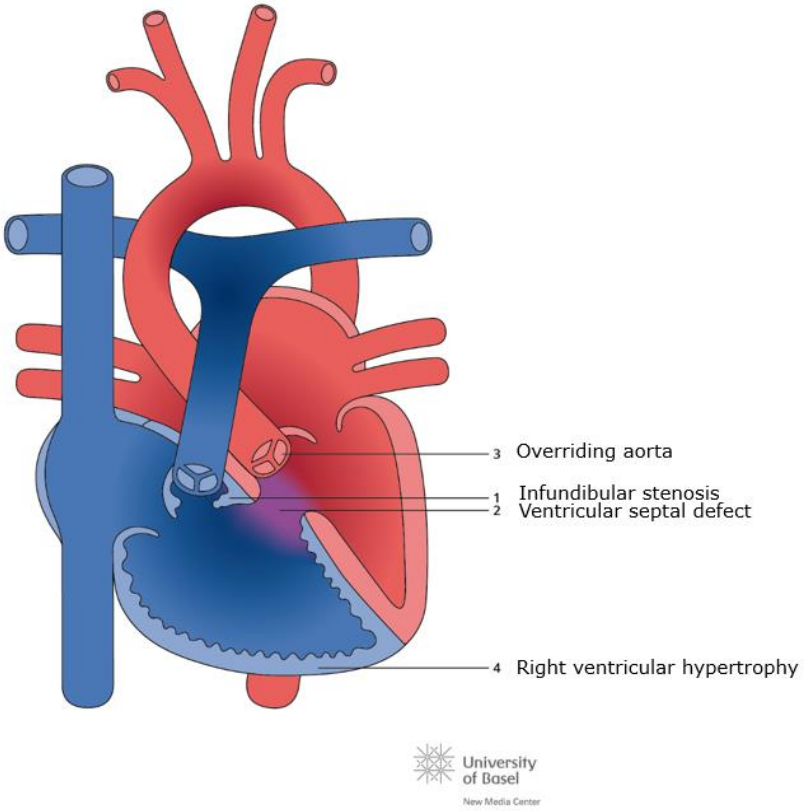


Illustration taken from http://www.chd-diagrams.com

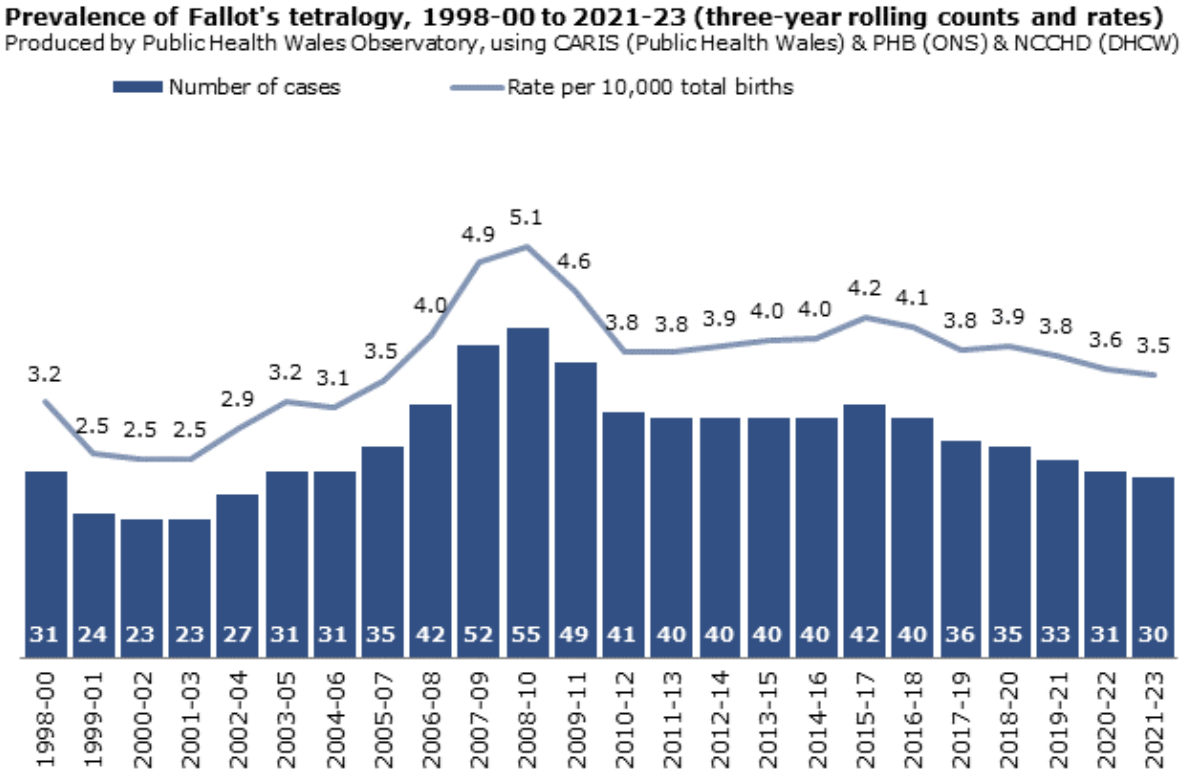
Most TOF cases (75%-80%) have isolated cardiac defects. The remaining cases are associated with syndromes or chromosomal anomalies. These include trisomy 21 (Down's syndrome) and 22q11.2 deletion syndrome (historically referred to as DiGeorge syndrome). Chromosome karyotype testing is recommended for all cases as outcomes are good in isolated cardiac cases. New advanced medical technology have helped in diagnosis and early treatment to improve the survival rates of TOF patients.

In Wales 313 cases have been registered between 1998 to 2023 (see Figure 2). This equates to approximately 12 cases per year or a prevalence rate of 3.7 per 10,000 total births. Nine out of 10 cases were liveborn. Modern ultrasound technology detected 72% of cases antenatally between

2021-2023. The remaining 28% were diagnosed in the postnatal period.

The 3 year rolling prevalence rates over time are shown in Figure 3. There was an increase in prevalence from 2005-2007 to a peak of 5.1 per 10,000 total births in 2008-2010. The prevalence dropped down to 3.8 per 10,000 total births in 2010-2012, and has been relatively stable over the past 10 years.

Figure 3



CARIS contribution to research in 2023/2024

Children with Hirschsprung's disease have high morbidity in the first 5 years of life
M Damkjær, J Tan, JK Morris, M Loane, J Given, C Cavero-Carbonell, ...
Birth Defects Research 116 (5), e2338

Hospital care in the first 10 years of life of children with congenital anomalies in six European countries: data from the EUROlinkCAT cohort linkage study
JK Morris, M Loane, C Wahlich, J Tan, S Baldacci, E Ballardini, ...
Archives of disease in childhood 109 (5), 402-408

Higher risk of cerebral palsy, seizures/epilepsy, visual-and hearing impairments, cancer, injury and child abuse in children with congenital anomalies: Data from the …

SK Urhoj, J Morris, M Loane, E Ballardini, L Barrachina-Bonet, ...
Acta paediatrica 113 (5), 1024-1031

Surveillance of multiple congenital anomalies; searching for new associations
JK Morris, JEH Bergman, I Barisic, D Wellesley, D Tucker, E Limb, ...
European Journal of Human Genetics 32 (4), 407-412

Gastroschisis prevalence patterns in 27 surveillance programs from 24 countries, International Clearinghouse for Birth Defects Surveillance and Research, 1980–2017
ML Feldkamp, MA Canfield, S Krikov, D Prieto-Merino, A Šípek Jr, ...
Birth defects research 116 (2), e2306

Updated EUROCAT guidelines for classification of cases with congenital anomalies
JEH Bergman, A Perraud, I Barišić, A Kinsner-Ovaskainen, JK Morris, ...
Birth Defects Research 116 (2), e2314

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 37 (8), 679-690

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

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