Congenital Anomaly Register & Information Service for Wales

caris review 2019

data from 1998 to 2018

This annual report includes the prevalence rates of key congenital anomalies and rare diseases in Wales, with a focus on hips. The updated prevalence rates includes the Official Statistics release of 2018 data.

Hip Dysplasia

Failure of the 'ball and socket' joint of the hip to form properly is termed Developmental Dysplasia of the Hip (DDH). This term describes a spectrum of structural anomaly ranging from instability, to subluxation or dislocation in more severe cases. It is widely accepted that early diagnosis and treatment is associated with better outcomes. There were 1,560 cases reported in Wales between 1998 to 2018 equating to approximately 74 cases per annum or a rate of 22.3 per 10,000 total births. Most cases (99.6%) were live born and 81% were female. The numbers of cases with dislocation or subluxation were 537 (26 per annum) and 78 (4 per annum) respectively. Talipes equinovarus was also present in 6.3% of DDH cases.

Detection of dislocation did not occur until after 1 year of age for 20% of the reported cases. However, early DDH detection rates have improved since around 2014, see Figure 1. Please note that the reduction in detections at birth observed between 1998 and 2005 is likely to have occurred as a result of protocol changes to the timing of diagnostic scans.

FIGURE 1 All hip dysplasias, date when anomaly first suspected, 3 - year rolling rate per10,000 total births, Wales, 1998 - 2018

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

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Perthes Disease

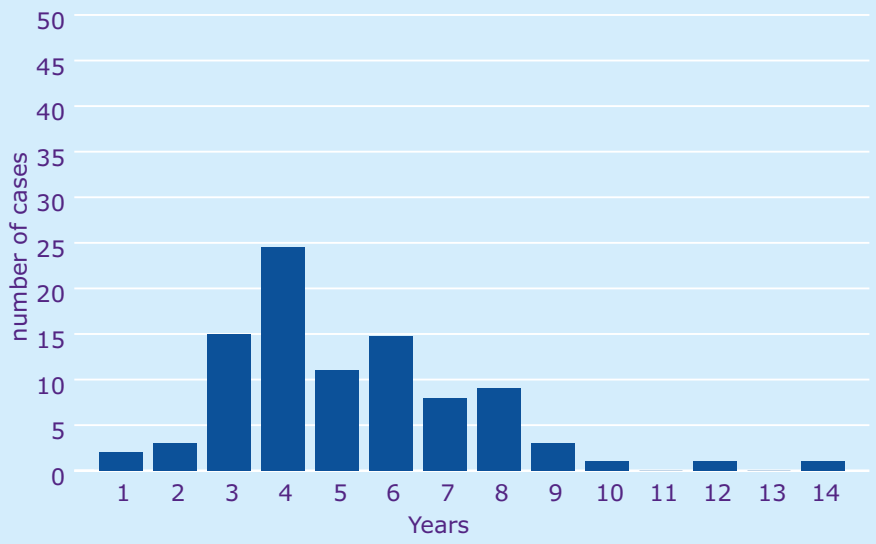
Perthes Disease in Wales

In its evolution to become a Rare Disease Register CARIS has reviewed Perthes (Legg-Calve-Perthes) disease this year. This is a condition of the hip joint that affects children and can cause premature osteoarthritis. It is a rare disease[1] (classified as an prevalence of less than 1 in 2000 births) and it is unclear why it occurs.

The blood supply to the head of the femur is reduced so the bone becomes soft and collapses.

Typically a boy of about 5 years will complain of knee pain (referred from the hip) and a left sided limp. Imaging confirms the diagnosis and treatment varies between simple monitoring, bed rest and traction. Surgery, usually osteotomy, may be necessary. Healing takes about 2 years.

FIGURE 2 Age at diagnosis



Data from the Community Childhealth Database and the Patient Episode Database in Wales (PEDW) were analysed. This yielded 303 cases between the years of 1998 and 2014. Prevalence rates have varied widely with lower rates latterly varying between 4.9 (2007) and 0.6 (2013) per 10,000 births.

Calculated from population figures the incidence rate per 100,000 children aged from 0-14 varied from 3 in 2006 to 0.6 in 2014. This means that in 2006, the highest year, there was one case diagnosed in 33,000 of children.

Of the 303 cases there were 113 records for analysis. In these there was a male to female ratio of 3:1 (86 males and 27 females).

Time of diagnosis was available in 111 cases and ranged from just under 2 years of age to over 14 years (see figure 2). The mean time of diagnosis was 5 years and 8 months.

Laterality details were available in 100 cases. 57 (57%) cases were left sided, 36 (36%) cases were right sided and 7 (7%) cases were bilateral. In unilateral cases, the left to right ratio was 1.6:1.

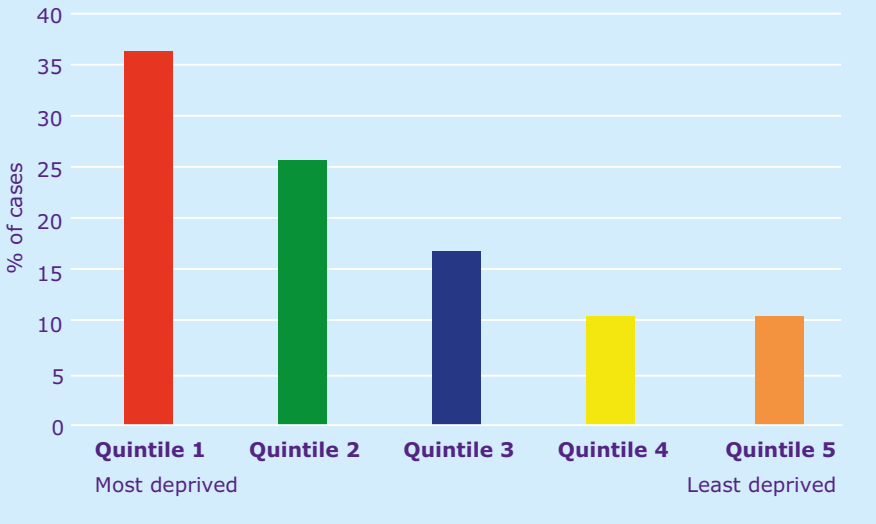
There is a record of surgery in 35 (31%) cases with the most frequent procedure being osteotomy and joint fixation.

Of the 113 cases, 10 (8.8%) had congenital anomalies including 3 children with cleft lip/palate anomalies.

There was a linear relationship with deprivation (see figure 3) which has been previously described in the literature[2].

One of a set of twins had the condition. No siblings were recorded.

FIGURE 3 Perthes disease in Wales by deprivation quintiles



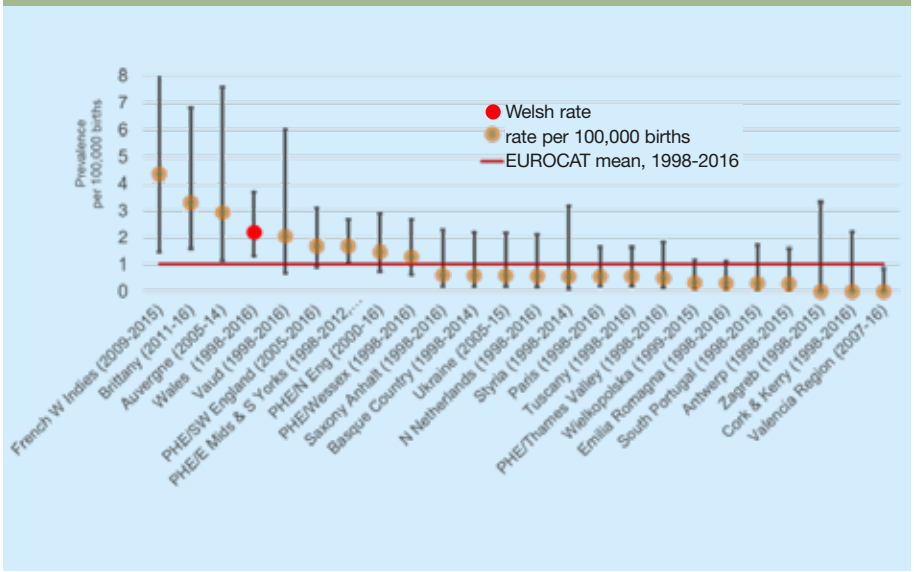
1 www.orphanet.net

2 Perry DC, Bruce CE, Pope D, et al
Perthes' disease of the hip: socioeconomic inequalities and the urban environment
Archives of Disease in Childhood 2012;**97**:1053-1057.

Sirenomelia

Sirenomelia is a rare congenital anomaly. There is no definitive cause, although evidence has been published that suggests links to environmental pollution. There is no treatment and live births are very rare. CARIS recorded 17 cases between 1998 and 2016, suggesting the prevalence in Wales may be higher than in other regions. CARIS reviewed the evidence to consider whether there is a higher prevalence of the condition in Wales compared with other regions and whether there have been any clusters of cases. Data were requested from The European Surveillance of Congenital Anomalies (EUROCAT) for the period and analysis suggested that Wales (with 2.2 cases per 100,000 births) had a higher rate than Europe as a whole (1 case per 100,000, see Figure 4). Almost half (47%) of cases were recorded in the South Wales valleys, a post-industrial area which has experienced past environmental pollution. However, statistical analyses yielded no evidence of epidemiological links between Welsh cases. For further rare disease rates see www.caris.wales.nhs.uk/rare-diseases

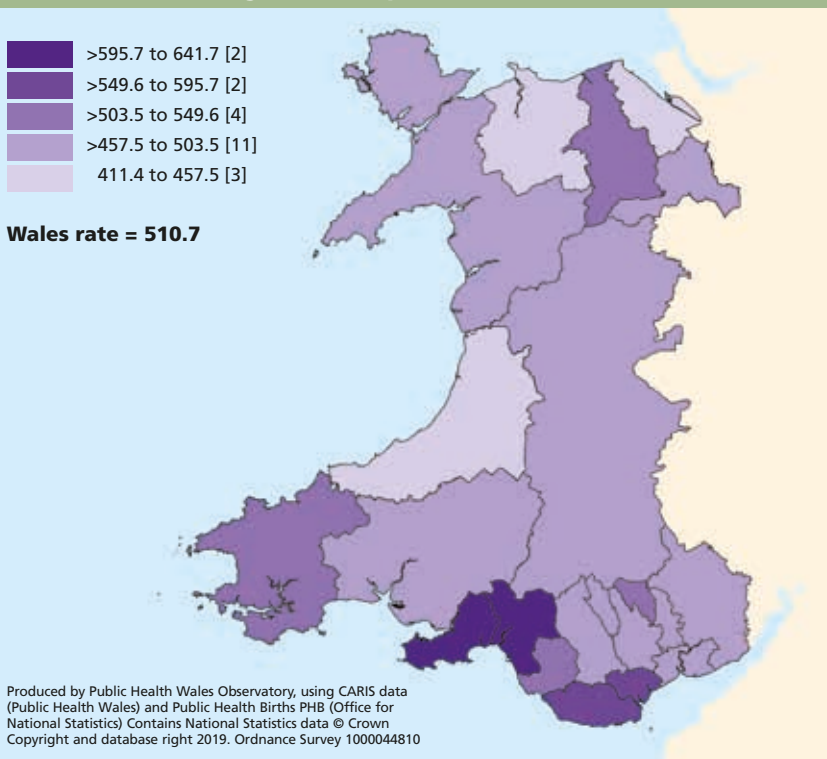
FIGURE 4 Sirenomelia Prevalence per 100,000 births by Country



Update on Congenital Anomalies 2018

An additional 1,386 congenital anomaly cases were reported to CARIS in 2018 taking the total number of cases in the database since its inception in 1998 to 35,739. The proportion of all births (live and still) with a congenital anomaly remained stable at 5.1%. Since 1998 the proportion of babies with a congenital anomaly that were live born was 85.7%, equating to 4.38% of all live born babies. Most live born babies with a congenital anomaly (97.0%) survive to 1 year of age. The proportion of cases with a single anomaly has remained stable at nearly 60%. Of those with a recorded gender, 59.1% were male. The prevalence rates by local authority region is summarised in Figure 5, and remains largely unchanged from previous years reports.

FIGURE 5 Rate of CARIS cases per 10,000 births, 1998-2018 Local authorities, gross rate per 10,000 births



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Circulatory conditions account for the largest anomalies group. The number of cases and prevalence rates for the main groups are summarised in Table 1. Please note that limb and musculoskeletal conditions were combined last year. They are separated once again this year.

TABLE 1 Main anomaly groups for cases reported to CARIS1998-2018, rate per 10,000 total births

	Count	Rate per10,000 total births
Circulatory	8,665	123.8
Genetic / multi-site	5,569	79.6
Limbs	5,447	77.8
Musculoskeletal	5,127	73.3
Digestive	4,626	66.1
Genital	4,267	61.0
Urinary	4,111	58.7
Neurological	3,257	46.5
Eye / ear	3,244	46.4
Skin	1,845	26.4
Respiratory	1,598	22.8
Endocrine, metabolic	1,575	22.5
Blood, immune, lymphatic	654	9.3
Neoplastic	374	5.3

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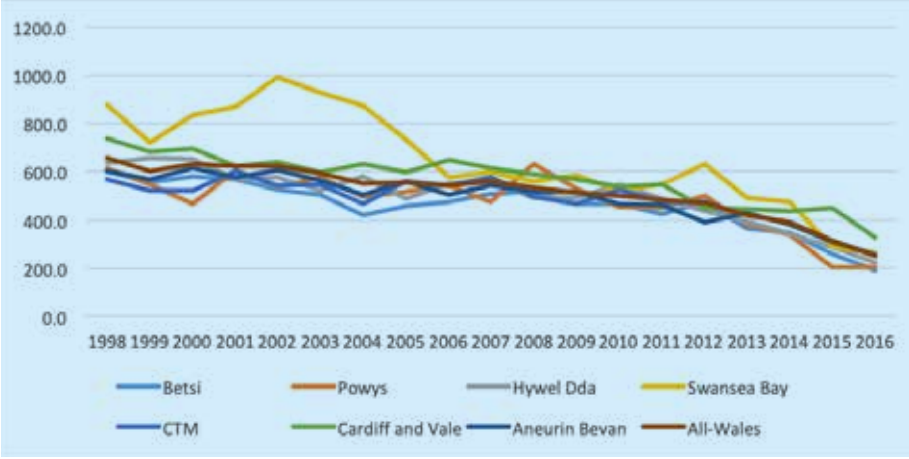
Geographical Variation in Congenital Anomaly Prevalence Rates in Wales

Geographical variation in congenital anomaly prevalence rates across Wales exists with higher prevalence in Swansea and Neath Port Talbot local authority areas. A retrospective analysis of CARIS cases from 1998 to 2016 was undertaken to identify if reporting bias due to the location of the CARIS office in Swansea was implicated.

FIGURE 6 Gross rate of congenital anomaly cases per 10,000 births by health board from 1998-2016



Significantly higher prevalence rates for Swansea Bay University Health Board (UHB) region in the early years of the register were observed, before returning to all-Wales levels from 2006 (see Figure 6). Specific conditions were identified with high prevalence in Swansea Bay UHB region, however, all were on a spectrum of disease severity and clinical significance that may influence their reporting to CARIS in different settings. The implementation of standardised antenatal screening and utilisation of electronic reporting systems to ensure comprehensive data collection across Wales may explain the Swansea Bay UHB rates being similar to the rest of Wales from 2006 onwards.

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