Congenital Anomaly Register & Information Service for Wales

# caris review 2018

data from 1998 to 2017

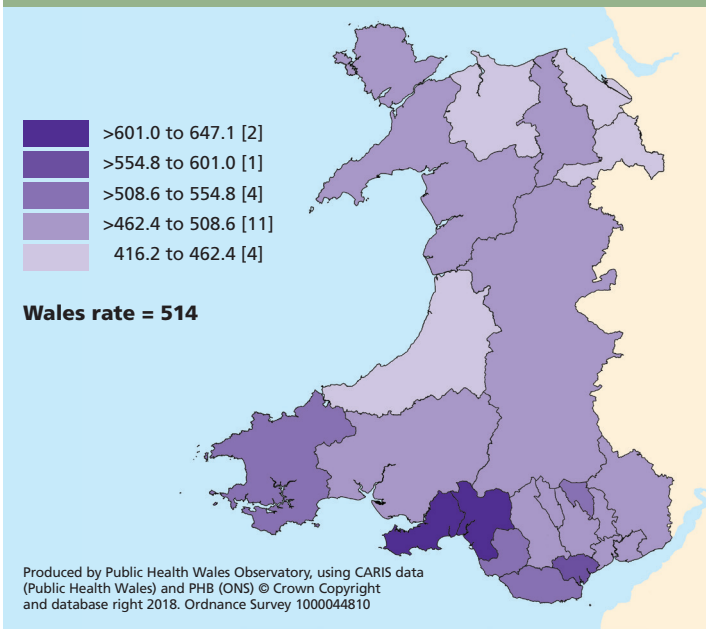
This annual update includes prevalence rates for some key congenital anomalies and rare diseases, and is released as Official Statistics. This short report summarises the updated information published in full on www.caris.wales.nhs.uk

By the end of 2017 there were 34,353 congenital anomaly cases recorded on the CARIS database. The rate of congenital anomalies has been stable for several years with 5.1% of all pregnancies affected. However as can be seen from the map, rates vary across Wales. It appears again that there is greater prevalence of anomalies in the Swansea area, this may reflect better recording because the team are located in the Singleton Hospital, but further investigation is planned in 2019.

Most cases (almost 60%) are registered with a single anomaly. 85.8% of cases are live-born, giving a live-born rate of 4.4% of all births affected, and 97% of babies survive to the age of one year. Where sex is known, 57.6% were male and 39.8% were female. The remainder did not have a sex assigned except 14 who were intersex. This year we have combined the count of limb conditions and musculoskeletal conditions so this now accounts for the largest group of conditions recorded. Circulatory conditions are the second largest group.

Each year this short report focuses on two areas of congenital anomalies, and this year we focus on trisomies and on miscarriages.

## Gross Rate of Congenital Anomaly Cases per 10,000 births, 1998-2017

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## Trisomies

Most people have 23 pairs of chromosomes, so 46 in each cell. A trisomy is a chromosomal disorder characterised by an additional chromosome in all or some cells, so instead of a pair of chromosomes, there are three. A person with a trisomy disorder has 47 chromosomes in a cell. A trisomy occurs at the cell division stage if one of the normal pairs of chromosomes fail to separate so that the egg or sperm carry an extra copy.

While trisomies are rare and many result in miscarriage, there are three more commonly occurring, which may be compatible with life. These are Trisomy 21 (Down syndrome); 18 (Edwards syndrome) and 13 (Patau syndrome). Antenatal screening for these three conditions changed in Wales in April this year with the introduction of the non-invasive prenatal test (NIPT). As part of the antenatal screening pathway, women are offered the option of a simple blood test (NIPT) rather than more invasive amniocentesis or chorionic villus sampling, if their earlier antenatal tests suggest they have a higher chance of having an affected pregnancy.

More information on the screening pathway can be found here: http://www.antenatalscreening.wales.nhs.uk

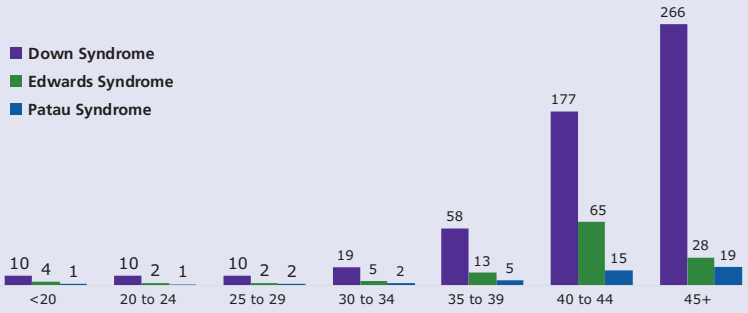
The number of cases recorded on the CARIS database for each of the three main trisomies is as above. It does appear prevalence is rising although the annual number of cases is small and detecting trend in rare events can be difficult. This increase in prevalence is thought to be associated with increases with maternal age.

### Cases with chromosomal anomalies, rates per 10,000 total births and percentage of cases liveborn, Wales, 1998-2017

| Anomaly | Total cases | Average cases per year | Rate | % of cases liveborn | Trend (3 year rolling rate) |
|---|---|---|---|---|---|
| T21 Down Syndrome | 1,556 | 78 | 23.3 | 46.0 | 21.0 … 25.5 |
| T18 Edwards Syndrome | 410 | 21 | 6.1 | 15.6 | 4.8 … 6.7 |
| T13 Patau Syndrome | 169 | 8 | 2.5 | 14.2 | 2.6 … 2.8 |

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

### Down Syndrome, Edwards Syndrome and Patau Syndrome rate per 10,000 total births by maternal age group, Wales, 1998-2017

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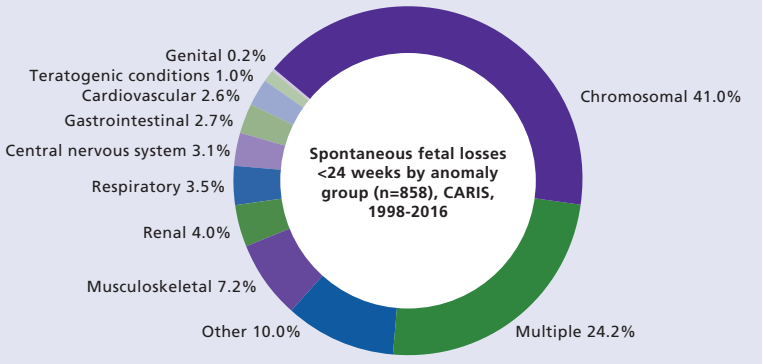
### Change in prevalence (per 10,000 births) CARIS 1998-2016

| Chromosomal anomaly | Prevalence excluding spontaneous fetal loss | Prevalence including spontaneous fetal loss | % increase in cases |
|---|---|---|---|
| Trisomy 21 (n–74) | 22.2 | 23.36 | 5.5 |
| Trisomy 18 (n–48) | 5.38 | 6.13 | 14.0 |
| Trisomy 13 (n–28) | 2.09 | 2.53 | 21.1 |
| Triploidy (n–72) | 3.35 | 4.34 | 29.6 |
| Turners (n–63) | 1.27 | 2.41 | 88.9 |

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## Miscarriages with Congenital Anomalies

Evidence suggests that over 50% of miscarriages (spontaneous fetal losses before 24 weeks gestation) result from chromosomal abnormalities and that 96% of these anomalies are aneuploidies – that is that there are an abnormal number of chromosomes present (too many or too few). Trisomies 22, 15 and 16 are thought to be the most common[1]. 3% of anomaly cases reported to CARIS are fetal losses and 41% of these were confirmed chromosomal anomalies.

CARIS records all spontaneous fetal losses and includes these in the numerator when rates are analysed. Many other congenital anomaly registers exclude earlier miscarriages when calculating prevalence rates of conditions. For comparability, when EUROCAT (the European network of registers) reports data, including data from CARIS, the report excludes cases where gestation is under 20 weeks.

The prevalence rates in certain chromosomal conditions were compared this year using and excluding miscarriage data. This showed an increase in rates particularly with Trisomy 13, triploidy and Turners syndrome and supports the use of miscarriage data in providing true prevalence rates and a more accurate description of outcomes.

1 Detection of aneuploidies in spontaneous abortions by quantitative fluorescent PCR with short tandem repeat markers: a retrospective study, Coelho et al, 2016, Genetics and Molecular Research 15 (3): gmr.15038617