Prevalence of congenital anomalies: time trends 2010-2019
Update: March 2021

One of the goals of the registration is monitoring the prevalence of congenital anomalies over time. In the graphs presented below the prevalence rates per year are shown for a period of nine years. We present the graphs from 2010 onwards because since that year we included the ‘non responders’ in the prevalence rates (see for definition: Introduction and Methods). The prevalence of total births are shown, (with 95% confidence interval) except for those anomalies where a major impact of prenatal diagnosis and selective termination of pregnancy can be expected. In those cases, total births and terminated pregnancies are presented. The Y-axis represents the observed prevalence of the selected congenital anomaly per 10,000 births, with adjustment of scale according to the magnitude of the counts.

First, the groups of all congenital anomalies together were analyzed. Subsequently, we analyzed chromosomal and monogenic anomalies. Finally, we studied specific malformations excluding chromosomal and monogenic disorders and deletions. This resulted in a group of so-called ‘non genetic’ anomalies, where environmental factors could play a role.

In addition to graphical presentation of prevalences, 95% confidence intervals and chi-square testing for trend was performed. Results of chi-square test are presented under each figure and the period tested is 2010-2019.

All congenital anomalies
There is gradual (significant) decrease in overall prevalence of all congenital anomalies together as a group. The decline in numbers in the most recent years is largely caused by incomplete registration of all cases for these years.

\[ X^2 \text{ for trend } = 92.2, \ p = .001 \]
**Chromosomal anomalies**
The prevalence of the group of Chromosomal anomalies shows a fluctuating pattern but no trend. This is true for the total prevalence as well as for the proportion of terminated pregnancies.

\[ X^2 \text{ for trend} = 0.6, \ p = 0.454 \]

**Trisomy 21**
Trisomy 21 (Down syndrome) is the most prevalent chromosomal anomaly. There is no statistically significant difference between in time. The proportion of terminated pregnancies in this period shows a heterogeneous pattern but not statistically significant.

\[ X^2 \text{ for trend} = 0.05, \ p = 0.81 \]
**Trisomy 13**

Trisomy 13 (Patau syndrome) is a rare chromosomal anomaly. The total number of cases is very low (40 in nine years), which causes some fluctuations in prevalence. The recent years show an increasing trend but non-significant. In 2019 there were no cases registered at Eurocat with trisomy 13.

![Trisomy 13 (Patau) prevalence and 95% CI per 10,000 births, per birthyear (N=40)](image)

\[ X^2 \text{ for trend} = 0.03, \ p = 0.859 \]

**Trisomy 18**

Trisomy 13 (Edwards syndrome) does not show a significant trend over the last nine years, although the most recent years show an (non-significant) increasing trend.

![Trisomy 18 (Edwards) prevalence and 95% CI per birthyear per 10,000 births (N=128)](image)

\[ X^2 \text{ for trend} = 0.12, \ p = 0.726 \]
Neural tube defects
The prevalence of neural tube defects seems to fluctuate over the last nine years, but not in a statistically significant way.

Spina bifida
Spina bifida, the most common neural tube defect, does not show a significant difference per year.
Heart anomalies
Heart anomalies are among the most common congenital anomalies. The prevalence of all congenital heart anomalies combined, not associated with a genetic or syndromal condition, fluctuates between 76 per 10,000 births in 2012 and 33 in 2019. There is also a significant decrease over time. This might be due to a delay in collection of heart anomalies. Pregnancy terminations do not increase substantially.

\[ \chi^2 \text{ for trend} = 25.66, \ p = <.001 \]

ASD
Atrium septum defects do not show a significant trend over the last nine years.

\[ \chi^2 \text{ for trend} = 0.74, \ p = 0.39 \]
**VSD**
Ventricular septum defects (VSD) are one of the most common heart defects. They show a significant decrease over time, especially the last two years. This might be due to a delay in collection of heart anomalies.

![Ventricular septum defect (non-genetic)
prevalence and 95% CI per 10,000 births, per birthyear (N=506)](image1)

$X^2$ for trend = 14.88, $p = <.001$

**Clefts**
Clefts are relatively common group of anomalies with a prevalence varying between 11 and 18.6 per 10,000 births. The prevalence is relatively stable over time.

![Oro-facial clefts (non-genetic)
prevalence and 95% CI per 10,000 births, per birthyear (N=235)](image2)

$X^2$ for trend = 0.01, $p = 0.92$
**Hypospadia**  
The prevalence of hypospadia seems to decrease over time, however this is not statistically significant.  

![Graph showing the prevalence of hypospadia from 2010 to 2019](image)

\[X^2 \text{ for trend } = 3.49, p = 0.062\]

**Renal dysplasia**  
The prevalence of renal dysplasia also shows fluctuation over the years, between 3.0 and 8.8 per 10,000 births, but not in a statistically significant trend.

![Graph showing the prevalence of renal dysplasia from 2010 to 2019](image)

\[X^2 \text{ for trend } = 0.001, p = 0.977\]
Reduction defects Limb
The prevalence looks heterogeneous over the birth years but not statistically significant.

\[ X^2 \text{ for trend } = 0.52, \ p = 0.47 \]