Disease specific registers – National Down Syndrome Cytogenetic Register (NDSCR)

- In 2012, there were 1,982 cases of Down syndrome (27 per 10,000 total births), 526 of Edwards syndrome (7 per 10,000 total births) and 229 of Patau syndrome (3 per 10,000 total births) in England and Wales.
- There is regional variation in the birth prevalence of Down, Edwards and Patau syndromes with the south generally having higher birth prevalence than the north. This reflects the different maternal age distributions between the regions. Following adjustment maternal age there was no difference in the birth prevalence of Down syndrome across England and Wales.
- Sixty-four percent of cases of Down syndrome notified were diagnosed prenatally, compared to 88% of Edwards syndrome and 93% of Patau syndrome.

Disease specific registers – Cleft lip and palate (CRANE) database

- In 2012, the CRANE database registered 1,146 live born children with a cleft lip and/or palate in England, Wales and Northern Ireland.
- The birth prevalence was highest in 2001, when 16.2 per 10,000 live births were reported to be affected with cleft lip and/or palate. The lowest rate was in both 2010 and 2011, when 13.7 per 10,000 live births were reported.
- However, the lower rates between 2008 and 2010 should be interpreted with caution. As primary surgical repairs usually take place between three months and two years after birth, with some repairs taking place even later, the data for the latter years are likely to under-represent the true number of cleft-affected births in England, Wales and Northern Ireland.

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For more information about BINOCAR go to - www.binocar.org
or to see the full report go to - www.binocar.org/publications/reports

British Isles Network of Congenital Anomaly Registers

Executive Summary: Congenital Anomaly Statistics 2012

England and Wales

December 2014

website: www.binocar.org
link to full report: www.binocar.org/publications/reports

Introduction

- This report collates data from six BINOCAR regional congenital anomaly registers, which together cover 36% of the births in England and Wales, to provide an estimate of the birth prevalence of congenital anomalies nationally. More detailed information is available from the website (www.binocar.org).
- Information from the National Down Syndrome Cytogenetic Register (NDSCR) for England and Wales and the cleft lip and palate (CRANE) database for England, Wales and Northern Ireland are presented separately.

Prevalence of congenital anomalies

- In 2012, there were 5,911 cases (live births, fetal deaths and terminations) with one or more congenital anomaly notified to six BINOCAR registers (East Midlands & South Yorkshire; Northern England; Oxfordshire, Berkshire & Buckinghamshire; South West England; Wessex; and Wales).
- The provisional birth prevalence for 2012 was 227 per 10,000 total births (1 in 44 total births). However, in previous reports about 7% of cases are only confirmed by the registers over two years after their birth. Including these late notifications, by increasing the number of cases reported by 7% would result in a predicted total birth prevalence of 243 per 10,000 total births (1 in 41 total births) equivalent to 17,800 cases with one or more congenital anomaly in the whole of England and Wales in 2012.
- The birth prevalence of congenital anomalies decreased slightly from 250 per 10,000 total births in 2011 to 243 per 10,000 total births predicted for 2012.
- Significantly increasing trends can be seen in neural tube defects (5% per year) and respiratory anomalies (7%), and significantly decreasing trends in limb reduction anomalies (8%) and oro-facial clefts (4%).

Timing of diagnosis and outcome

- Sixty-one percent of cases, for whom the time of diagnosis was known, were prenatally diagnosed in 2012.
- Of the pregnancies in which an anomaly was suspected prenatally, 45% resulted in a termination of pregnancy. Seventy-eight percent of cases with chromosomal anomalies and 32% of those with non-chromosomal anomalies resulted in a termination of pregnancy.
- Of the live born postnatally diagnosed cases, where the time of diagnosis was known, 69% were diagnosed at birth, 8% were diagnosed in the 1st week, 6% between the 2nd and 4th weeks and 16% after the 1st month. Less than 2% of live births are diagnosed after 1 year of age.
- There were significantly increasing trends in the percentage of prenatally diagnosed cases with non-chromosomal anomalies (from 40% in 2007 to 45% in 2011) as well as in cases with chromosomal anomalies (from 66% in 2007 to 73% in 2011).
- The overall rate of termination of pregnancy with fetal anomaly was 53 per 10,000 total births (1 in 189 births). Chromosomal anomalies accounted for 25 terminations of pregnancy per 10,000 total births (1 in 400 births).

The NHS Fetal Anomaly Screening Programme in England

- The target detection rates were achieved or exceeded for five out of the 11 FASP anomalies. However, the percentage of prenatal diagnoses are likely to be underestimates of the true detection rates as accurate adjustment cannot be made for women that declined or booked too late for screening or for cases terminated for another severe anomaly found earlier.

Key public health indicators

- The infant mortality rate in England and Wales in 2012 was 40 per 10,000 total births, of which an estimated 22% had a congenital anomaly. The main congenital anomaly subgroups contributing to infant mortality are congenital heart defects (43%), digestive system anomalies (23%) and chromosomal anomalies (19%).
- Mothers who were between 25 and 29 years of age at delivery had the lowest birth prevalence for all anomalies. The prevalence was higher in the under 20 age group and considerably higher in the 40 and over age group, largely a consequence of the higher rate of chromosomal anomalies in women aged 40 years and older.

Geographical variations and comparison with EUROCAT registers

- There were regional differences in the reported birth prevalence of congenital anomalies between the six registers. The birth prevalence for CARIS (Wales) and SWCAR (South West England) were significantly higher and the birth prevalence for EMSYCAR (East Midlands & South Yorkshire) and WANDA (Wessex) were significantly lower than the overall birth prevalence. Although there are likely to be some regional variations in the true birth prevalence of specific anomalies, the regional variation is thought largely to be due to variations in case ascertainment at the time of data extraction for this report and variations in the severity of the anomalies.
- The birth prevalence across the BINOCAR registers for all anomalies is 15% (95% CI: 11, 18) lower than the average birth prevalence for all the other European congenital anomaly registers (EUROCAT).
- The birth prevalence of respiratory anomalies (10 compared with 6 per 10,000 total births), abdominal wall defects (9 compared with 5 per 10,000 total births) and chromosomal anomalies (43 compared with 36 per 10,000 total births) were significantly higher in the BINOCAR registers compared to the EUROCAT registers.
- The regional rates of termination of pregnancy with fetal anomaly in the BINOCAR registers ranged from 43 per 10,000 total births (1 in 233 births) to 67 per 10,000 total births (1 in 149 births).

Cluster analysis

- A total of six potential clusters were identified. However none of them were judged to be clusters due to the differing aetiology of the anomalies in the cluster and the regional disparity of the cases in one potential cluster. Clusters will continue to be monitored annually.

Spotlight on prenatal diagnosis

- The percentage of cases prenatally diagnosed varied according to the type of anomaly. Abdominal wall defects, urinary anomalies and nervous system anomalies were prenatally diagnosed in over 90% of isolated cases whereas only 10% of genital anomalies were prenatally diagnosed, due to the defects being very small.
- There were significant increases in the percentage of cases being prenatally diagnosed between 2008 and 2012 with isolated congenital heart defects and isolated limb anomalies.
- The decision to terminate was strongly associated with the severity of the anomaly. Following a prenatal diagnosis, 71% of pregnancies affected with an isolated nervous system anomaly were terminated whereas fewer than 3% of pregnancies affected by less severe anomalies were terminated.
- Prenatal diagnosis was not associated with improved survival, but this may be partly due to more severe anomalies being detected prenatally.