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

- Sixty-five percent of cases of Down syndrome notified were diagnosed prenatally, as were 93% of Edwards syndrome and 90% of Patau syndrome.

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

- In 2011, the CRANE database registered 949 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 2010, when 13.7 per 10,000 live births were reported. However, the lower rates between 2008 and 2010 should be interpreted with caution. Primary surgical repairs usually take place between three months and two years after birth, with some repairs taking place even later, therefore the data for the latter years are likely to under-represent the true number of cleft-affected births.

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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 2011

England and Wales

September 2013

Public Health England

website: www.binocar.org

GIG CYMRU NHS WALES
Iechyd Cyhoeddus Cymru
Public Health Wales

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.
- Care must be taken in comparing data in this report and the 2010 report, which include data from six regional registers, with data from the 2009 report, which included data from five regional registers. Birth prevalence estimates rather than case numbers should be compared.
- This report also provides a comparison between the birth prevalence of congenital anomalies in the BINOCAR registers and the birth prevalence in the European Surveillance of Congenital Anomalies (EUROCAT) registers (excluding the BINOCAR registers).

Prevalence of congenital anomalies

- In 2011, there were 5,718 cases with one or more congenital anomalies notified to six BINOCAR registers (East Midlands & South Yorkshire, Northern England, Oxfordshire, Berkshire & Buckinghamshire, South West England, Wessex and Wales). The birth prevalence was 219 per 10,000 total births (1 in 46 total births).
- The birth prevalence of congenital anomalies decreased from 261 per 10,000 total births in 2007 to 219 per 10,000 total births in 2011. However, data in the later years are likely to be incomplete as it takes time for notifications to be sent to the registers, then checked and validated by the registers, and some anomalies, for example some congenital heart defects, are not diagnosed until later in infancy and childhood.

Timing of diagnosis and outcome

- Sixty-one percent of cases, where the time of diagnosis was known, were prenatally diagnosed in 2011.
- Of the pregnancies in which an anomaly was suspected prenatally, 44% resulted in a termination of pregnancy for fetal anomaly. Seventy-eight percent of cases with chromosomal anomalies and 32% of those with non-chromosomal anomalies resulted in a termination of pregnancy for fetal anomaly.
- Of the live born postnatally diagnosed cases, where the time of diagnosis was known, 68% were diagnosed at birth, 8% were diagnosed in the 1st week, 6% between the 2nd and 4th weeks and 18% after the 1st month.
- The overall rate of termination of pregnancy for fetal anomaly was 50 per 10,000 total births (1 in 200 births). Chromosomal anomalies accounted for 23 terminations of pregnancy per 10,000 total births (1 in 429 births).

The NHS Fetal Anomaly Screening Programme in England

- Data on prenatal detection were available from four English registers.
- The target detection rates were achieved or exceeded for four out of the 11 FASP anomalies. However, the proportions of prenatal diagnosis are likely to be underestimates of the true 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 2011 was 4.4 per 1,000 total births of which an estimated 15% had a congenital anomaly. The main congenital anomaly subgroups contributing to infant mortality are congenital heart defects (47%), chromosomal anomalies (19%) and digestive system anomalies (17%).
- Mothers who were between 25 and 29 years of age 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.

Geographical variations and comparison with EUROCAT registers

- There were regional differences in the reported birth prevalence of congenital anomalies in the six registers. The birth prevalence for CARIS (Wales) and SWCAR (South West England) were significantly higher and the birth prevalence for CAROBB (Oxfordshire, Berkshire and Buckinghamshire) and EMSYCAR (East Midlands & South Yorkshire) were significantly lower than the birth prevalence for all six registers combined. 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.
- The birth prevalence across the BINOCAR registers for all anomalies is significantly lower than the birth prevalence across all of the EUROCAT registers (excluding BINOCAR registers) (219 per 10,000 total births vs. 241 per 10,000 total births). The birth prevalence will be influenced by the ascertainment of cases by the registers within BINOCAR and EUROCAT as well as the underlying true birth prevalence which may vary regionally and nationally.
- The birth prevalence of abdominal wall defects was significantly higher in the BINOCAR registers (9 per 10,000 total births vs. 6 per 10,000 total births) and in severe congenital heart defects (24 vs. 20) when compared to the EUROCAT registers.
- The regional rates of termination of pregnancy for fetal anomaly in the BINOCAR registers ranged from 42 per 10,000 total births (1 in 240 births) to 67 per 10,000 total births (1 in 149 births).

Spotlight on congenital diaphragmatic hernia

- There was no significant change in the birth prevalence of non-chromosomal or chromosomal congenital diaphragmatic hernia (CDH) between 2002 and 2011.
- The 1-year survival in live born cases was 75% in isolated CDH compared to 64% in those with multiple anomalies and 57% with chromosomal CDH.
- 1-year survival is significantly higher in babies born at term. However, 1-year survival was much lower in prenatally diagnosed chromosomal CDH cases than in prenatally diagnosed isolated or multiple CDH cases, partially explained by 50% of these chromosomal cases having Edwards or Patau syndrome which have extremely low survival rates.

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

- In 2011, there were 1,873 cases of Down syndrome (26 per 10,000 total births), 504 of Edwards syndrome (7 per 10,000 total births) and 191 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 having a higher birth prevalence than the north. This reflects the different maternal age distributions between the regions, as once it has been adjusted for there was no difference in the birth prevalence of Down syndrome across England and Wales.