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

- In 2010, there were 1,868 notifications of Down syndrome (26 per 10,000 total births), 213 of Patau syndrome (3 per 10,000 total births) and 514 of Edwards syndrome (7 per 10,000 total births) in England and Wales.
- There is regional variation in the prevalence of Down, Patau and Edwards syndromes with the south having a higher prevalence than the north probably reflecting the different maternal age distributions in the regions.
- Sixty-four percent of cases of Down syndrome notified were diagnosed prenatally, as were 90% of Patau syndrome and 91% of Edwards syndrome.

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

- In 2010, the CRANE database registered 935 children born with a cleft lip and/or palate 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 2010

England and Wales

July 2012

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

HQIP
Healthcare Quality Improvement Partnership

Introduction

- This report collates data from six regional congenital anomaly registers, which together cover 35% of the births in England and Wales, to provide an estimate of the 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, which includes data from six regional registers, with data from the 2009 report, which included data from five regional registers. Prevalence rather than case numbers must be compared.
- This report also provides a comparison between the prevalence of congenital anomalies in the BINOCAR registers and the prevalence in the other European registers.

Congenital anomaly notifications

- In 2010, there were 5,818 notifications of congenital anomalies to six BINOCAR registers (East Midlands & South Yorkshire, Northern England, Oxfordshire, Berkshire & Buckinghamshire, South West England, Wessex and Wales). The prevalence was 224 per 10,000 total births (1 in 45 births).
- The prevalence of congenital anomalies decreased from 268 per 10,000 total births in 2006 to 224 per 10,000 total births in 2010. However, data in the later years are likely to be incomplete as it takes time for notifications to be sent to the registers and some anomalies, for example some cardiac anomalies, are not diagnosed until later in infancy and childhood.

Timing of diagnosis and outcome

- Sixty percent of cases, where the time of diagnosis was known, were prenatally diagnosed in 2010.
- Of the pregnancies in which an anomaly was suspected prenatally, 44% resulted in a termination of pregnancy for fetal anomaly. Seventy-nine percent of fetuses with chromosomal anomalies and 31% 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, 65% were diagnosed at birth, 11% were diagnosed in the first week, 5% between the 2nd and 4th weeks and 20% after the 1st month.
- The overall rate of termination of pregnancy for fetal anomaly was 50 per 10,000 total births (1 in 202 births). Chromosomal anomalies accounted for 24 terminations of pregnancy per 10,000 total births (1 in 426 births).

The NHS Fetal Anomaly Screening Programme in England

- Four out of the five English registers were able to provide data for this chapter.
- These registers exceeded the target detection rates for six out of the 11 FASP anomalies and in two others there was no evidence that they were significantly below the target. However, the proportions of prenatal diagnosis are likely to be an underestimate of the true rates as accurate adjustment cannot be made for women that declined or booked too late for screening and babies terminated for another severe anomaly found earlier.
- The majority of cases with FASP anomalies were prenatally diagnosed before 21 weeks' gestation. The proportion ranges from 54% of cases with bilateral renal agenesis to 97% of cases with anencephaly.

Key public health indicators

- The perinatal mortality rate in England and Wales in 2010 was 74 per 10,000 total births of which an estimated 16% had a congenital anomaly. This proportion is likely to be an underestimate of the true rate as some pregnancies with severe congenital anomalies are terminated before 24 weeks whereas without intervention the majority of these pregnancies would have been likely to end in a perinatal death.
- Ninety-five percent of live births notified with congenital anomalies survived to one year of age. Infants diagnosed with genital anomalies had the highest survival to one year of age (98%).
- Mothers who were between 25 and 29 years of age had the lowest 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

- There were regional differences in the reported prevalence of congenital anomalies in the six registers. The prevalence for SWCAR (South West England) was significantly higher and the prevalence for EMSYCAR (East Midlands & South Yorkshire) was significantly lower than the prevalence for all six registers combined.
- The prevalence of the majority of congenital anomalies for the six BINOCAR registers combined was consistent with that from the other European registers.
- The prevalence of abdominal wall defects was significantly higher in the BINOCAR registers and the prevalence of congenital heart defects and urinary anomalies were significantly lower in the BINOCAR registers when compared to the other European registers.
- The regional rates of termination of pregnancy for fetal anomaly ranged from 38 per 10,000 total births (1 in 265 births) to 70 per 10,000 total births (1 in 143 births).
- Geographical variations in prevalence and rates of termination of pregnancy for fetal anomaly may be due to both differences in ascertainment, differences in risk factors such as maternal age, differences in the screening tests that are offered and the uptake of the test, which are influenced by social factors.