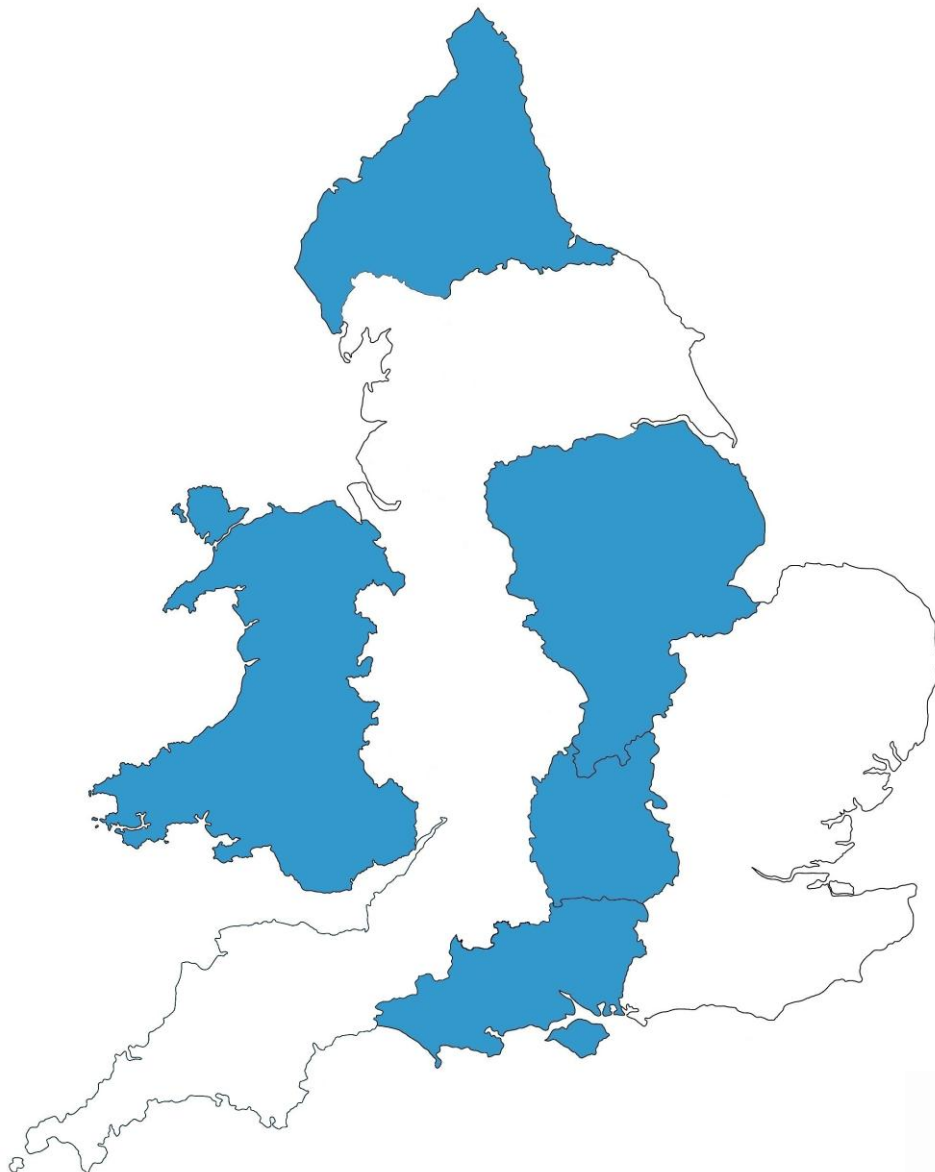




# British Isles Network of Congenital Anomaly Registers

## Congenital Anomaly Statistics 2009 England and Wales

December 2011



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## Foreword

All parents look forward to the birth of their child. However, from time to time, their excitement and joy is tempered by the realisation that their child has an anomaly of some kind. Serious anomalies are thankfully rare but as the data in this report show, are by no means exceptional. Many only come to light some months or years after birth.

Experience has shown that the key to understanding the root causes of these important events is careful documentation of patterns of occurrence of defined groups of anomalies. This information has been shown to be of great value for such research and to guide subsequent prevention programmes. It is also essential to ensure that the right services are available to treat affected children, and to support their families in other ways. It is increasingly being recognised that good quality registers are needed to generate reliable information on outcomes for prospective parents faced with difficult decisions in early pregnancy.

The process of anomaly registration and analysis is methodologically challenging. Collection of data from multiple sources allows a complete picture to emerge but requires great care and effort to eliminate double counting. BINOCAR members are to be congratulated on achieving consistently high standards over the years and making commensurate contribution to knowledge in this area. In particular, the new approach to collating data from regional registers at national level, in order to support larger scale analysis at national and international level, is very welcome.

Despite these achievements, there is no doubt that more could be done to improve coverage and use of the valuable data reported here. In future, responsibility for surveillance of congenital anomalies in England is likely to be given to the new organisation, Public Health England. This should allow the work of the English BINOCAR registers to be placed on a firmer footing, allow further development of this excellent work, and bring additional benefits to families and health services as a result.

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# Executive Summary

## Introduction

- This report collates data from five regional congenital anomaly registers, which together cover 28% of the population of England and Wales, to provide an estimate of the prevalence of congenital anomalies. More detailed information is available from the website ([www.binocar.org](http://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.
- Trends in congenital anomalies cannot be obtained by comparing earlier data from the Office for National Statistics National Congenital Anomaly System (ONS NCAS) with the 2009 data in this report; the prevalence reported by the British Isles Network of Congenital Anomaly Registers (BINOCAR) is consistently higher than that reported by NCAS due to active and multiple sources of ascertainment.

## Chapter 1: Congenital anomaly notifications

- In 2009, there were 4,181 notifications of congenital anomalies to the five BINOCAR registers (East Midlands & South Yorkshire, Northern England, Thames Valley, Wessex and Wales). The prevalence was 206 per 10,000 total births (1 in 49 births).
- The prevalence of births with congenital anomalies increased between 2005 and 2006 and then showed a 17% decrease from 248 per 10,000 total births in 2006 to 206 per 10,000 total births in 2009, probably partly due to a lag in registration in later years.

## Chapter 2: Timing of diagnosis and outcome

- Fifty-three percent of diagnoses occurred prenatally in 2009. The prevalence of prenatal diagnosis has decreased from 114 per 10,000 total births in 2005 to 109 per 10,000 total births in 2009.
- Of the pregnancies in which an anomaly was suspected prenatally, 43% resulted in a termination.
- Of those born alive, where the time of diagnosis was known, 68% were diagnosed at birth, 9% were diagnosed in the first week, 7% between the 2<sup>nd</sup> and 4<sup>th</sup> weeks and 17% after the 1<sup>st</sup> month.

## Chapter 3: Key public health indicators

- The perinatal mortality rate in the United Kingdom in 2009 was 76 per 10,000 total births of whom an estimated 11% had a congenital anomaly and 3% had congenital heart disease.
- Ninety-five percent of live births notified with congenital anomalies survived to one year of age. Live births with genital anomalies had the highest survival to one year of age (99%) and live births with respiratory anomalies had the lowest (79%).
- The overall rate of termination of pregnancy for fetal anomaly was 47 per 10,000 total births (1 in 213 births). Chromosomal anomalies accounted for 21 terminations of pregnancy per 10,000 total births (1 in 476 births).

## Chapter 4: Variation by register

- There is a significant difference in the prevalence of births with congenital anomalies in the five registers. CARIS (Wales) had the highest prevalence of 275 per 10,000 total births (1 in 36 births) and EMSYCAR (East Midlands & South Yorkshire) had the lowest prevalence of 172 per 10,000 total births (1 in 58 births).

- WANDA (Wessex) had the highest rate of termination of pregnancy (65 per 10,000 total births, 1 in 154 births) and EMSYCAR (East Midlands & South Yorkshire) had the lowest rate (38 per 10,000 total births, 1 in 263 births).
- Regional variations in prevalence and rates of termination of pregnancy for fetal anomaly may be due to both differences in ascertainment as well as differences in risk factors such as maternal age.

#### Chapter 5: Disease specific registers – National Down Syndrome Cytogenetic Register (NDSCR)

- In 2009, there were 1,887 notifications of Down syndrome (27 per 10,000 total births), 163 of Patau syndrome (2 per 10,000 total births) and 506 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-two percent of notifications of Down syndrome were diagnosed prenatally, 87% of Patau syndrome and 91% of Edwards syndrome.

#### Chapter 6: Disease specific registers – Cleft lip and palate (CRANE) database

- In 2009, the CRANE database registered 878 children born with a cleft lip and/or palate in England, Wales and Northern Ireland.
- Using Hospital Episode Statistics data for England and the CRANE data for Wales and Northern Ireland the estimated incidence of cleft lip and/or palate was 15.2 per 10,000 live births (1 in 658 live births).

# Glossary

Term	Definition
18 <sup>+0</sup> to 20 <sup>+6</sup> weeks fetal anomaly scan	A detailed ultrasound scan offered to all pregnant women and undertaken between 18 weeks + 0 days and 20 weeks + 6 days, for the purpose of assessing the fetus for structural anomalies.
95% confidence interval	This provides a range of values in which the observed prevalence will fall 95% of the time.
Antenatal/prenatal	The period from conception to birth.
Births/total births	Live births and stillbirths.
Case ascertainment	Proportion of notifications of congenital anomalies reported to the registers out of all cases of congenital anomaly in the population.
Congenital anomaly	A physiological or structural abnormality that develops at or before birth and is present at the time of birth, especially as a result of faulty development, infection, heredity, or injury.
Cytogenetics	The study of chromosomes. Clinical cytogenetics is the study of the relationship between chromosome aberrations and disease.
Live birth	Delivery of an infant, which, after complete separation from its mother, shows signs of life.
Miscarriage	Late fetal deaths from 20-23 weeks of gestation.
Neonatal death	Death of a live born baby occurring before 28 completed days after birth. Early = 0-6 completed days; Late = 7-27 completed day
Notifications	Live births, stillbirths, late miscarriages and terminations of pregnancy. A case with multiple anomalies will only count as one notification.
Perinatal mortality	The number of stillbirths and early neonatal deaths per 10,000 total births.
Prevalence	The total number of cases of congenital anomaly in a given population at a specific time.
Severe CHD	This includes the following congenital heart diseases: <ul style="list-style-type: none"> <li>• Common arterial truncus</li> <li>• Transposition of great vessels</li> <li>• Single ventricle</li> <li>• Atrioventricular septal defect</li> <li>• Tetralogy of Fallot</li> <li>• Tricuspid atresia and stenosis</li> <li>• Ebstein's anomaly</li> <li>• Pulmonary valve atresia</li> <li>• Aortic valve atresia/stenosis</li> <li>• Hypoplastic left heart</li> <li>• Hypoplastic right heart</li> <li>• Coarctation of aorta</li> <li>• Total anomalous pulmonary venous return</li> </ul>
Stillbirths	Late fetal deaths from 24 weeks of gestation.
Termination of pregnancy for fetal anomaly	Term used to describe the deliberate ending of a pregnancy with the intention that the fetus will not survive and which is carried out when the fetus is diagnosed prenatally as having a major congenital anomaly.
Ultrasound scan	A medical, non-invasive investigative screening examination which uses ultrasound to create real-time images on a monitor.

# Abbreviations

Abbreviation	Meaning
BINOCAR	British Isles Network of Congenital Anomaly Registers
CARIS	Congenital Anomaly Register and Information Service (for Wales)
CAROBB	Congenital Anomaly Register for Oxfordshire, Berkshire & Buckinghamshire
EMSYCAR	East Midlands and South Yorkshire Congenital Anomalies Register
EUROCAT	European Surveillance of Congenital Anomalies
LB	Live birth
Misc	Miscarriage
NCAS	National Congenital Anomaly System
NDSCR	National Down Syndrome Cytogenetic Register
NorCAS	Northern Congenital Abnormality Survey
ONS	Office for National Statistics
SB	Stillbirth
TOPFA	Termination of pregnancy for fetal anomaly
WANDA	Wessex Antenatally Detected Anomalies Register

# Introduction

The British Isles Network of Congenital Anomaly Registers (BINOCAR) is a group of regional and disease-specific registers collecting information about congenital anomalies occurring in England, Wales and Ireland.

This report includes data from the following regional registers: Congenital Anomaly Register and Information Service for Wales (CARIS, established in 1998); Congenital Anomaly Register for Oxfordshire, Berkshire and Buckinghamshire (CAROBB, 2005 but has data for Oxford from 1991); East Midlands and South Yorkshire Congenital Anomalies Register (EMSYCAR, 1997); Northern Congenital Abnormality Survey (NorCAS, 1985); Wessex Antenatally Detected Anomalies Register (WANDA, 1994). These five registers cover 28% of the births in England and Wales.

This report also includes data from two national disease-specific registers, the National Down Syndrome Cytogenetic Register (NDSCR) which collects data on Down (Trisomy 21), Edwards (Trisomy 18) and Patau (Trisomy 13) syndromes for the whole of England and Wales (1989) and the CRANE database which collects data on cleft lip and palate anomalies for the whole of England, Wales and Northern Ireland (1990). The data from these registers are presented in separate chapters.

Between 1964 and 2009 a National Congenital Anomaly System (NCAS) was run by the Office for National Statistics (ONS) to monitor changes in the numbers of births with congenital anomalies reported by health authorities in England and Wales. NCAS was acknowledged to have a lower ascertainment rate than BINOCAR and published its last set of national surveillance data in 2008. In January 2011 the Department of Health initiated funding of BINOCAR to provide national surveillance. The European Surveillance of Congenital Anomalies (EUROCAT) collates the anonymised data from the BINOCAR registers who are members of EUROCAT together with other European registers to provide European surveillance.

Anomaly notifications are classified using the EUROCAT exclusion list (Appendix A) and the BINOCAR Coding Framework (Appendix B).

## Aim of BINOCAR

The aim of BINOCAR is to provide continuous epidemiological monitoring of the frequency, nature and outcomes of congenital anomalies for the population of the British Isles by means of national, regional and disease-specific registers of congenital anomalies.

## Objectives of BINOCAR

The objectives of BINOCAR are:

- Surveillance and analysis of congenital anomalies
- Monitoring and audit of health provision, detection and outcomes for congenital anomalies
- Provision of information to support planning and administration of the provision made for health and social care for pregnancies and infants affected by congenital anomalies
- Medical research, approved by research ethics committees, into the causes and consequences of congenital anomalies
- Provision of information to clinicians to support their clinical practice

## Data collection, processing and validation

Congenital anomaly data are collected from a number of different sources, these include:

- Maternity units
- Diagnostic departments (paediatric, neonatal, clinical genetics, antenatal ultrasound, fetal medicine, pathology)
- Cytogenetic laboratories
- NHS Trust IT departments
- Neighbouring regional registers
- Disease-specific registers
- Child health systems
- Local audit schemes

This multiple source reporting is performed in order for the BINOCAR registers to achieve the highest possible ascertainment of congenital anomalies in their population.

Data are collected on all suspected and confirmed congenital anomalies identified in utero, at birth or in childhood. In addition to live births and stillbirths affected by anomalies, terminations of pregnancy for fetal anomaly following prenatal diagnosis (TOPFA) and late miscarriages (20-23 weeks gestation) where an anomaly is present are also collected.

BINOCAR collects information on the mother and child, including postcode of residence, mother's age, length of pregnancy, pregnancy outcome, when and how the anomalies were discovered and the details of the anomalies. Some identifiable information is collected on the mother and baby but only enough information to enable regional registers to avoid duplicate registrations and for the validation of cases, ensuring accurate matching between antenatally diagnosed anomalies and postnatal notifications.

BINOCAR is authorised under Section 251 of the NHS Act 2006 to collect personal information without individual consent. This exemption was granted by the National Information Governance Board (NIGB).

All regional databases have their own validation checks carried out on the data identifying missing data, values lying outside the expected range and data items that are not consistent.

Some of the BINOCAR registers are also members of EUROCAT. With European Union Joint Action funding, EUROCAT collates the anonymised data from these registers together with other European registers and carries out further data cleaning. Prevalence, key public health indicators and prenatal diagnosis data for some of the BINOCAR registers as well as other European registers can be found on the EUROCAT website [www.eurocat-network.eu](http://www.eurocat-network.eu).

## Data confidentiality

In line with the Code of Practice for Official Statistics<sup>1</sup>, all statistics in this report have been disclosure-controlled to protect confidentiality. Thus in tables containing individual register data, any cells containing less than five cases will be suppressed. Any rates based on such small numbers will also be suppressed. Secondary suppressions will be applied as necessary to avoid the possibility of disclosure through differencing/subtraction. Data at the combined register and national level is considered to be low risk so no suppression is required.

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<sup>1</sup> Code of Practice for Official Statistics, UK Statistics Authority, January 2009.  
<http://www.statisticsauthority.gov.uk/assessment/code-of-practice/index.html> [Accessed: 02/11/2011]

## Comparison of BINOCAR with National Congenital Anomaly Statistics (NCAS)

In 1998, BINOCAR registers began providing data to NCAS. This resulted in areas covered by BINOCAR registers having a higher prevalence of births with congenital anomalies than areas that were not covered. In 2005, NCAS began to present the data for the areas of England and Wales covered by registers separately to those areas that were not.

Table 1 shows the number of cases published by NCAS for the areas covered by registers and the number of cases published by BINOCAR for approximately the same areas in 2007 (the most recent year for which accurate data are available). At the time of the NCAS publication, the data on terminations was subject to greater suppression so actual numbers cannot be provided in Table 1.

One of the reasons for the greater ascertainment in the BINOCAR data is in the timing of the extract for analysis. The extract of the ONS data usually took place 9-10 months after the data year ended and was not updated. The BINOCAR data is continually updated. Thus, the data presented in Table 1 was extracted just over three years after the data year end. Extra cases will have been notified, anomalies confirmed or cases completed in this time period.

NCAS and BINOCAR calculated their anomaly subgroups separately so there may be some variation in the cases coded within each group. There are also some differences in the exclusion criteria for the NCAS and BINOCAR data, so some cases would be removed from the NCAS data that would not be removed from the BINOCAR data.

### NCAS exclusions:

- Date of birth not known
- LMP or gestation at delivery not known
- Gestation at delivery  $\leq 18$  wks or  $\geq 50$  wks
- Miscarriage
- Postcode not known
- Not complete

### BINOCAR exclusions:

- Miscarriage <20 wks
- Type of birth (e.g. live/still birth) not known
- Minor anomalies
- No confirmation of suspected anomalies

Table 2 shows the NCAS estimates of prevalence for England and Wales, NCAS estimates of prevalence for the areas covered by BINOCAR registers and the BINOCAR estimates of prevalence of all major congenital anomaly subgroups. The prevalence reported by NCAS for the areas not covered by BINOCAR registers (not shown) were considerably lower.

The prevalence reported by BINOCAR was generally 50% higher than that reported by NCAS for the areas covered by registers. The largest differences were in digestive system anomalies where the BINOCAR prevalence was 90% higher and congenital heart disease which was 80% higher. Therefore, trends in the prevalence of any congenital anomaly cannot be estimated by comparing NCAS data before 2009 with BINOCAR data from 2009.

**Table 1: Comparison of the number of notifications published by NCAS and BINOCAR: 2007**

Congenital anomaly <sup>1</sup>	Number						
	NCAS data for areas covered by registers <sup>2</sup>			BINOCAR <sup>2</sup>			
	LB <sup>3</sup>	SB <sup>3</sup>	TOPFA <sup>4</sup>	LB <sup>5</sup>	SB <sup>5</sup>	Misc <sup>5</sup>	TOPFA <sup>5</sup>
<b>All notifications</b>	<b>3,709</b>	<b>154</b>	<b>863</b>	<b>4,751</b>	<b>107</b>	<b>57</b>	<b>1,263</b>
<b>Nervous system</b>	<b>179</b>	<b>34</b>	<b>217</b>	<b>245</b>	<b>12</b>	<b>3</b>	<b>384</b>
Neural tube defects	39	12	134	40	6	1	233
Anencephalus and similar	7	5	69	8	2	1	93
Encephalocele	5	2	15	6	1	-	28
Spina bifida	28	5	50	26	3	-	112
Hydrocephaly	50	12	12	76	3	2	85
Microcephaly	19	-	*	41	-	-	5
Arhinencephaly/holoprosencephaly	7	2	21	6	-	-	33
<b>Congenital heart disease</b>	<b>896</b>	<b>40</b>	<b>49</b>	<b>1,344</b>	<b>30</b>	<b>11</b>	<b>191</b>
Common arterial truncus	5	1	*	4	-	-	6
Transposition of great vessels	56	1	*	74	1	-	6
Single ventricle	5	-	*	9	-	-	4
Ventricular septal defect	417	8	*	585	5	3	47
Atrial septal defect	128	4	*	192	5	1	10
Atrioventricular septal defect	52	5	*	79	5	1	30
Tetralogy of Fallot	43	1	*	84	1	1	13
Tricuspid atresia and stenosis	9	-	*	13	1	-	1
Ebstein's anomaly	5	4	*	7	2	1	3
Pulmonary valve stenosis	65	-	*	122	1	-	-
Pulmonary valve atresia	12	3	*	23	1	-	5
Aortic valve atresia/stenosis	17	-	*	34	-	-	1
Hypoplastic left heart	32	4	15	42	2	-	42
Hypoplastic right heart	7	2	*	7	1	2	2
Coarctation of aorta	78	1	*	104	1	-	2
Total anomalous pulmonary venous return	14	-	*	20	-	-	1
<b>Respiratory</b>	<b>88</b>	<b>12</b>	<b>*</b>	<b>130</b>	<b>6</b>	<b>6</b>	<b>36</b>
Choanal atresia	12	-	*	20	-	-	-
Cystic adenomatoid malformation of lung	23	1	*	33	-	1	4
<b>Oro-facial clefts</b>	<b>324</b>	<b>7</b>	<b>*</b>	<b>414</b>	<b>4</b>	<b>1</b>	<b>57</b>
Cleft lip with or without cleft palate	217	5	*	256	3	-	42
Cleft palate	107	2	*	158	1	1	15
<b>Digestive system</b>	<b>227</b>	<b>11</b>	<b>*</b>	<b>371</b>	<b>7</b>	<b>3</b>	<b>67</b>
Oesophageal atresia with or without tracheo-oesophageal fistula	39	2	*	45	1	-	6
Duodenal atresia or stenosis	31	-	*	39	1	-	2
Atresia or stenosis of other parts of the small intestine	19	-	*	29	-	-	-
Ano-rectal atresia and stenosis	38	4	*	48	2	-	23
Hirschsprung's disease	22	-	*	38	-	-	-
Diaphragmatic hernia	51	4	*	59	-	1	17
<b>Abdominal wall defects</b>	<b>125</b>	<b>6</b>	<b>12</b>	<b>148</b>	<b>3</b>	<b>3</b>	<b>74</b>
Gastroschisis	91	1	*	103	1	1	12
Omphalocele	32	5	*	43	2	2	52
<b>Urinary</b>	<b>619</b>	<b>15</b>	<b>39</b>	<b>617</b>	<b>11</b>	<b>11</b>	<b>127</b>
Bilateral renal agenesis including Potter syndrome	3	2	*	3	1	1	28

1 Some of the babies shown in this table will have multiple anomalies and appear in more than one row of the table.

2 NCAS and BINOCAR covered East Midlands and South Yorkshire, Northern England, Oxfordshire, Berkshire and Buckinghamshire, South West England, Wales and Wessex. Merseyside and Cheshire are only included in the NCAS data.

3 NCAS, 25 September 2008

4 Department of Health, Abortion Statistics 2007, England and Wales, June 2008

5 BINOCAR, February 2011

- = 0

\* NCAS suppression of termination of pregnancy counts less than 10 to protect the confidentiality of individuals

LB = Live birth, SB = Stillbirth (≥24 weeks), Misc = Miscarriage (20-23 weeks), TOPFA = Termination of pregnancy for fetal anomaly

**Table 1 cont'd: Comparison of the number of notifications published by NCAS and BINOCAR: 2007**

Congenital anomaly <sup>1</sup>	Number						
	NCAS data for areas covered by registers <sup>2</sup>			BINOCAR <sup>3</sup>			
	LB <sup>3</sup>	SB <sup>3</sup>	TOPFA <sup>4</sup>	LB <sup>5</sup>	SB <sup>5</sup>	Misc <sup>5</sup>	TOPFA <sup>5</sup>
Renal dysplasia	103	2	12	103	3	3	28
Congenital hydronephrosis	334	2	*	269	1	-	13
Bladder exstrophy and/or epispadias	11	1	*	10	-	1	1
Posterior urethral valve and/or prune belly	14	-	*	17	-	-	7
<b>Genital</b>	<b>408</b>	<b>2</b>	<b>*</b>	<b>511</b>	<b>4</b>	<b>1</b>	<b>24</b>
Hypospadias	338	-	*	415	-	1	4
Indeterminate sex	21	-	*	20	-	-	5
<b>Limbs</b>	<b>695</b>	<b>38</b>	<b>14</b>	<b>872</b>	<b>19</b>	<b>14</b>	<b>121</b>
Limb reductions	87	12	*	98	8	4	51
Club foot - talipes equinovarus	202	12	*	240	6	4	35
Hip dislocation and/or dysplasia	106	-	*	166	-	-	1
Polydactyly	172	9	*	208	3	1	11
Syndactyly	89	2	*	106	2	3	8
<b>Musculo-skeletal</b>	<b>101</b>	<b>16</b>	<b>23</b>	<b>158</b>	<b>8</b>	<b>4</b>	<b>87</b>
<b>Other malformations</b>	<b>61</b>	<b>5</b>	<b>27</b>	<b>121</b>	<b>7</b>	<b>5</b>	<b>44</b>
<b>Genetic syndromes + microdeletions</b>	<b>89</b>	<b>4</b>	<b>*</b>	<b>136</b>	<b>3</b>	<b>1</b>	<b>40</b>
<b>Chromosomal</b>	<b>320</b>	<b>45</b>	<b>319</b>	<b>415</b>	<b>38</b>	<b>21</b>	<b>555</b>
Down syndrome	217	17	182	241	18	7	281
Patau syndrome/trisomy 13	7	5	24	11	3	3	53
Edwards syndrome/trisomy 18	18	9	59	19	5	2	113
Turner's syndrome	18	3	14	22	3	2	35
Klinefelters syndrome	12	-	*	16	-	-	5

1 Some of the babies shown in this table will have multiple anomalies and appear in more than one row of the table.

2 NCAS and BINOCAR covered East Midlands and South Yorkshire, Northern England, Oxfordshire, Berkshire and Buckinghamshire, South West England, Wales and Wessex. Merseyside and Cheshire are only included in the NCAS data.

3 NCAS, 25 September 2008

4 Department of Health, Abortion Statistics 2007, England and Wales, June 2008

5 BINOCAR, February 2011

- = 0

\* NCAS suppression of termination of pregnancy counts less than 10 to protect the confidentiality of individuals

LB = Live birth, SB = Stillbirth (≥24 weeks), Misc = Miscarriage (20-23 weeks), TOPFA = Termination of pregnancy for fetal anomaly

**Table 2: Comparison of the prevalence published by NCAS and BINOCAR: 2007**

Congenital anomaly <sup>1</sup>	Prevalence per 10,000 total births [95% CI]			Ratio of BINOCAR to NCAS (areas covered by registers)
	NCAS data for all England and Wales	NCAS data for areas covered by registers	BINOCAR	
<b>All notifications</b>	<b>73.2 [71.2, 75.2]</b>	<b>127.5 [123.5, 131.6]</b>	<b>196.7 [191.2, 202.3]</b>	<b>1.5</b>
Nervous system	4.1 [3.6, 4.6]	7.0 [6.1, 8.0]	10.4 [9.2, 11.8]	1.5
Congenital heart disease	15.4 [14.5, 16.3]	30.9 [28.9, 32.9]	55.6 [52.7, 58.6]	1.8
Respiratory	1.7 [1.4, 2.0]	3.3 [2.7, 4.0]	5.5 [4.6, 6.5]	1.7
Oro-facial clefts	7.0 [6.4, 7.7]	10.9 [9.8, 12.2]	16.9 [15.3, 18.6]	1.5
Digestive system	3.7 [3.3, 4.2]	7.9 [6.9, 8.9]	15.3 [13.8, 16.9]	1.9
Abdominal wall defects	2.4 [2.1, 2.8]	4.3 [3.6, 5.1]	6.1 [5.2, 7.2]	1.4
Urinary	11.4 [10.6, 12.2]	20.9 [19.3, 22.6]	25.4 [23.5, 27.5]	1.2
Genital	7.8 [7.2, 8.5]	13.5 [12.3, 14.9]	20.8 [19.1, 22.7]	1.5
Limb	15.8 [14.9, 16.8]	24.2 [22.5, 26.0]	36.1 [33.7, 38.5]	1.5
Musculo-skeletal	2.3 [2.0, 2.7]	3.9 [3.2, 4.6]	6.7 [5.7, 7.8]	1.7
Chromosomal	7.2 [6.5, 7.8]	12.0 [10.8, 13.3]	18.3 [16.7, 20.1]	1.5

1 Some of the babies shown in this table will have multiple anomalies and appear in more than one row of the table.

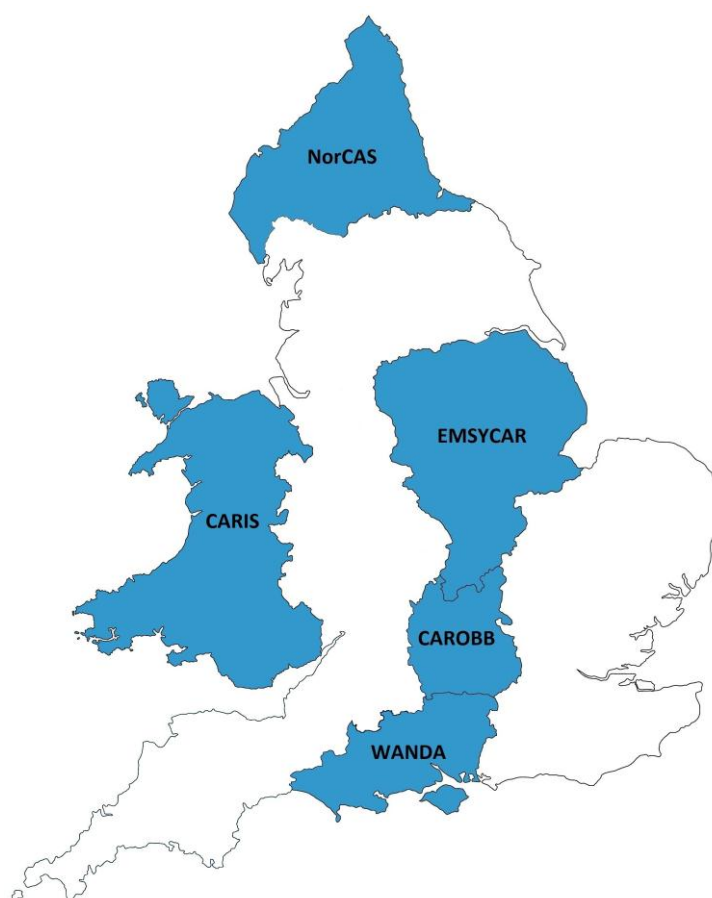
Note: Live births and stillbirths only

# Chapter 1: Congenital anomaly notifications

## 1.1 Notifications according to congenital anomaly subgroups

In 2009, there were 4,181 notifications of congenital anomalies to the following BINOCAR registers: Congenital Anomaly Register and Information Service for Wales (CARIS); Congenital Anomaly Register for Oxfordshire, Berkshire and Buckinghamshire (CAROBB); East Midlands and South Yorkshire Congenital Anomalies Register (EMSYCAR); Northern Congenital Abnormality Survey (NorCAS); Wessex Antenatally Detected Anomalies Register (WANDA) (see Figure 1.1 for geographical coverage and see Appendix C for the areas covered by each register).

**Figure 1.1: Map of registers included in this report**



Of the 4,181 notifications, 3,098 were live births, 87 were stillbirths ( $\geq 24$  weeks), 37 were miscarriages (20-23 weeks) and 959 were TOPFAs, out of a total of 203,264 births in the areas covered by the registers. This gives an overall prevalence of 206 per 10,000 total births (95% CI: 200, 212). After excluding chromosomal anomalies, there were 3,372 notifications giving a prevalence of 166 per 10,000 total births (95% CI: 160, 172).

Table 1.1 shows the number of notifications and the prevalence for all congenital anomaly subgroups. The most common congenital anomaly group is congenital heart disease with 1,100 notifications in 2009 (55 per 10,000 total births, 95% CI: 51, 58).

**Table 1.1:** Number of notifications and prevalence (per 10,000 total births) according to congenital anomaly subgroup; five BINOCAR registers (coverage: 28% of births in England and Wales): 2009

Congenital anomaly <sup>1</sup>	Including chromosomal					Excluding chromosomal		
	Number					Prevalence per 10,000 total births [95% CI]	Number	Prevalence per 10,000 total births [95% CI]
	LB	SB	Misc	TOPFA	Total		Total	
<b>Total births</b>	<b>201,902</b>	<b>1,043</b>	<b>..</b>	<b>..</b>	<b>202,945</b>	<b>..</b>	<b>..</b>	<b>..</b>
<b>All notifications</b>	<b>3,098</b>	<b>87</b>	<b>37</b>	<b>959</b>	<b>4,181</b>	<b>205.7 [199.5, 212.0]</b>	<b>3,372</b>	<b>165.9 [160.3, 171.6]</b>
<b>Nervous system</b>	<b>148</b>	<b>20</b>	<b>5</b>	<b>310</b>	<b>483</b>	<b>23.8 [21.7, 26.0]</b>	<b>440</b>	<b>21.6 [19.7, 23.8]</b>
Neural tube defects	41	6	1	207	255	12.5 [11.1, 14.2]	240	11.8 [10.4, 13.4]
Anencephalus and similar	4	3	-	95	102	5.0 [4.1, 6.1]	97	4.8 [3.9, 5.8]
Encephalocele	6	2	-	19	27	1.3 [0.9, 1.9]	26	1.3 [0.8, 1.9]
Spina Bifida	31	1	1	93	126	6.2 [5.2, 7.4]	117	5.8 [4.8, 6.9]
Hydrocephaly	48	10	1	58	117	5.8 [4.8, 6.9]	106	5.2 [4.3, 6.3]
Microcephaly	22	2	2	4	30	1.5 [1.0, 2.1]	26	1.3 [0.8, 1.9]
Arhinencephaly/holoprosencephaly	5	2	1	18	26	1.3 [0.8, 1.9]	17	0.8 [0.5, 1.3]
<b>Congenital heart disease (CHD)</b>	<b>925</b>	<b>27</b>	<b>7</b>	<b>141</b>	<b>1,110</b>	<b>54.6 [51.4, 57.9]</b>	<b>933</b>	<b>45.9 [43.0, 48.9]</b>
Severe CHD	313	13	2	84	412	20.3 [18.4, 22.3]	344	16.9 [15.2, 18.8]
Common arterial truncus	10	-	-	2	12	0.6 [0.3, 1.0]	11	0.5 [0.3, 1.0]
Transposition of great vessels	69	2	-	5	76	3.7 [2.9, 4.7]	75	3.7 [2.9, 4.6]
Single ventricle	7	1	-	1	9	0.4 [0.2, 0.8]	9	0.4 [0.2, 0.8]
Ventricular septal defect	407	6	2	23	438	21.5 [19.6, 23.7]	373	18.4 [16.5, 20.3]
Atrial septal defect	148	1	1	11	161	7.9 [6.7, 9.2]	126	6.2 [5.2, 7.4]
Atrioventricular septal defect	53	4	-	19	76	3.7 [2.9, 4.7]	35	1.7 [1.2, 2.4]
Tetralogy of Fallot	64	2	-	9	75	3.7 [2.9, 4.6]	69	3.4 [2.6, 4.3]
Tricuspid atresia and stenosis	12	-	-	5	17	0.8 [0.5, 1.3]	14	0.7 [0.4, 1.2]
Ebstein's anomaly	8	2	-	2	12	0.6 [0.3, 1.0]	11	0.5 [0.3, 1.0]
Pulmonary valve stenosis	86	-	-	4	90	4.4 [3.6, 5.4]	86	4.2 [3.4, 5.2]
Pulmonary valve atresia	17	1	-	5	23	1.1 [0.7, 1.7]	21	1.0 [0.6, 1.6]
Aortic valve atresia/stenosis	20	-	-	1	21	1.0 [0.6, 1.6]	21	1.0 [0.6, 1.6]
Hypoplastic left heart	24	3	1	33	61	3.0 [2.3, 3.9]	54	2.7 [2.0, 3.5]
Hypoplastic right heart	8	-	-	6	14	0.7 [0.4, 1.2]	13	0.6 [0.3, 1.1]
Coarctation of aorta	50	-	1	4	55	2.7 [2.0, 3.5]	49	2.4 [1.8, 3.2]
Total anomalous pulmonary venous return	16	-	-	2	18	0.9 [0.5, 1.4]	16	0.8 [0.4, 1.3]

<sup>1</sup> Some of the babies shown in this table will have multiple anomalies and appear in more than one row of the table.

- = 0

.. = Not applicable

LB = Live birth, SB = Stillbirth (≥24 weeks), Misc = Miscarriage (20-23 weeks), TOPFA = Termination of pregnancy for fetal anomaly

**Table 1.1 cont'd: Number of notifications and prevalence (per 10,000 total births) according to congenital anomaly subgroup; five BINOCAR registers (coverage: 28% of births in England and Wales): 2009**

Congenital anomaly <sup>1</sup>	Including chromosomal					Excluding chromosomal		
	Number					Prevalence per 10,000 total births [95% CI]	Number	Prevalence per 10,000 total births [95% CI]
	LB	SB	Misc	TOPFA	Total		Total	
<b>Respiratory</b>	<b>79</b>	<b>7</b>	<b>2</b>	<b>29</b>	<b>117</b>	<b>5.8 [4.8, 6.9]</b>	<b>109</b>	<b>5.4 [4.4, 6.5]</b>
Choanal atresia	12	-	-	-	12	0.6 [0.3, 1.0]	12	0.6 [0.3, 1.0]
Cystic adenomatous malformation of lung	28	-	-	3	31	1.5 [1.0, 2.2]	31	1.5 [1.0, 2.2]
<b>Oro-facial clefts<sup>2</sup></b>	<b>264</b>	<b>3</b>	<b>3</b>	<b>34</b>	<b>304</b>	<b>15.0 [13.3, 16.7]</b>	<b>281</b>	<b>13.8 [12.3, 15.5]</b>
Cleft lip with or without cleft palate	156	2	2	26	186	9.2 [7.9, 10.6]	171	8.4 [7.2, 9.8]
Cleft palate	108	1	1	8	118	5.8 [4.8, 7.0]	110	5.4 [4.4, 6.5]
<b>Digestive system</b>	<b>279</b>	<b>10</b>	<b>1</b>	<b>47</b>	<b>337</b>	<b>16.6 [14.9, 18.4]</b>	<b>302</b>	<b>14.9 [13.2, 16.6]</b>
Oesophageal atresia with or without trachea-oesophageal fistula	48	3	1	3	55	2.7 [2.0, 3.5]	51	2.5 [1.9, 3.3]
Duodenal atresia or stenosis	36	-	-	1	37	1.8 [1.3, 2.5]	29	1.4 [1.0, 2.0]
Atresia or stenosis of other parts of small intestine	11	-	-	3	14	0.7 [0.4, 1.2]	14	0.7 [0.4, 1.2]
Ano-rectal atresia and stenosis	49	1	1	14	65	3.2 [2.5, 4.1]	60	3.0 [2.3, 3.8]
Hirschsprung's disease	27	-	-	-	27	1.3 [0.9, 1.9]	25	1.2 [0.8, 1.8]
Diaphragmatic hernia	48	2	-	15	65	3.2 [2.5, 4.1]	53	2.6 [2.0, 3.4]
<b>Abdominal wall defects</b>	<b>114</b>	<b>5</b>	<b>3</b>	<b>68</b>	<b>190</b>	<b>9.3 [8.1, 10.8]</b>	<b>162</b>	<b>8.0 [6.8, 9.3]</b>
Gastroschisis	85	2	2	7	96	4.7 [3.8, 5.8]	96	4.7 [3.8, 5.8]
Omphalocele	28	3	1	48	80	3.9 [3.1, 4.9]	53	2.6 [2.0, 3.4]
<b>Urinary</b>	<b>421</b>	<b>6</b>	<b>2</b>	<b>87</b>	<b>516</b>	<b>25.4 [23.2, 27.7]</b>	<b>492</b>	<b>24.2 [22.1, 26.4]</b>
Bilateral renal agenesis including Potter syndrome	3	-	1	26	30	1.5 [1.0, 2.1]	30	1.5 [1.0, 2.1]
Renal dysplasia	83	2	-	21	106	5.2 [4.3, 6.3]	103	5.1 [4.1, 6.1]
Congenital hydronephrosis	203	1	1	8	213	10.5 [9.1, 12.0]	204	10.0 [8.7, 11.5]
Bladder exstrophy and/or epispadias	14	-	-	3	17	0.8 [0.5, 1.3]	16	0.8 [0.4, 1.3]
Posterior urethral valve and/or prune belly	14	-	-	6	20	1.0 [0.6, 1.5]	20	1.0 [0.6, 1.5]
<b>Genital</b>	<b>287</b>	<b>2</b>	<b>-</b>	<b>15</b>	<b>304</b>	<b>15.0 [13.3, 16.7]</b>	<b>300</b>	<b>14.8 [13.1, 16.5]</b>
Hypospadias	251	1	-	-	252	12.4 [10.9, 14.0]	251	12.3 [10.9, 14.0]
Indeterminate sex	8	1	-	5	14	0.7 [0.4, 1.2]	13	0.6 [0.3, 1.1]
<b>Limb</b>	<b>503</b>	<b>14</b>	<b>7</b>	<b>92</b>	<b>616</b>	<b>30.3 [28.0, 32.8]</b>	<b>579</b>	<b>28.5 [26.2, 30.9]</b>
Limb reduction	71	3	2	39	115	5.7 [4.7, 6.8]	104	5.1 [4.2, 6.2]
Upper limb reduction	53	2	2	29	86	4.2 [3.4, 5.2]	77	3.8 [3.0, 4.7]

1 Some of the babies shown in this table will have multiple anomalies and appear in more than one row of the table.

2 Information on cleft lip and/or palate for the whole of England, Wales and Northern Ireland can be found in Chapter 6.

- = 0

LB = Live birth, SB = Stillbirth (≥24 weeks), Misc = Miscarriage (20-23 weeks), TOPFA = Termination of pregnancy for fetal anomaly

**Table 1.1 cont'd: Number of notifications and prevalence (per 10,000 total births) according to congenital anomaly subgroup; five BINOCAR registers (coverage: 28% of births in England and Wales): 2009**

Congenital anomaly <sup>1</sup>	Including chromosomal					Excluding chromosomal		
	Number					Prevalence per 10,000 total births [95% CI]	Number	Prevalence per 10,000 total births [95% CI]
	LB	SB	Misc	TOPFA	Total		Total	
Lower limb reduction	22	1	1	16	40	2.0 [1.4, 2.7]	35	1.7 [1.2, 2.4]
Club foot – talipes equinovarus	175	4	1	31	211	10.4 [9.0, 11.9]	202	9.9 [8.6, 11.4]
Hip dislocation and/or dysplasia	45	-	-	-	45	2.2 [1.6, 3.0]	45	2.2 [1.6, 3.0]
Polydactyly	140	2	1	15	158	7.8 [6.6, 9.1]	150	7.4 [6.2, 8.7]
Syndactyly	62	3	2	4	71	3.5 [2.7, 4.4]	67	3.3 [2.6, 4.2]
<b>Musculo-skeletal</b>	<b>73</b>	<b>9</b>	<b>4</b>	<b>51</b>	<b>137</b>	<b>6.7 [5.7, 8.0]</b>	<b>135</b>	<b>6.6 [5.6, 7.9]</b>
<b>Other malformations</b>	<b>51</b>	<b>3</b>	<b>1</b>	<b>25</b>	<b>80</b>	<b>3.9 [3.1, 4.9]</b>	<b>73</b>	<b>3.6 [2.8, 4.5]</b>
<b>Genetic syndromes + microdeletions</b>	<b>74</b>	<b>2</b>	<b>2</b>	<b>22</b>	<b>100</b>	<b>4.9 [4.0, 6.0]</b>	<b>96</b>	<b>4.7 [3.8, 5.8]</b>
<b>Chromosomal</b>	<b>339</b>	<b>29</b>	<b>16</b>	<b>425</b>	<b>809</b>	<b>39.8 [37.1, 42.6]</b>	-	-
Down syndrome <sup>3</sup>	215	10	6	210	441	21.7 [19.7, 23.8]	-	-
Patau syndrome/trisomy 13 <sup>3</sup>	4	1	2	33	40	2.0 [1.4, 2.7]	-	-
Edwards syndrome/trisomy 18 <sup>3</sup>	16	9	2	97	124	6.1 [5.1, 7.3]	-	-
Turner's syndrome	14	2	4	33	53	2.6 [2.0, 3.4]	-	-
Klinefelters syndrome	9	-	-	2	11	0.5 [0.3, 1.0]	-	-

<sup>1</sup> Some of the babies shown in this table will have multiple anomalies and appear in more than one row of the table.

<sup>3</sup> Information on Down syndrome, Patau syndrome and Edwards syndrome for the whole of England and Wales can be found in Chapter 5.

- = 0

LB = Live birth, SB = Stillbirth (≥24 weeks), Misc = Miscarriage (20-23 weeks), TOPFA = Termination of pregnancy for fetal anomaly

## 1.2 Estimated numbers of notifications in England and Wales

The number of congenital anomalies in the whole of England and Wales in 2009 can be estimated by multiplying the prevalence estimates from the BINOCAR registers for 2009 by the total number of births in England and Wales in 2009 (709,936). Table 1.2 shows that in 2009 there were an estimated 14,603 cases of congenital anomalies in England and Wales. This assumes that the prevalence is consistent over the whole of England and Wales. In some areas without registers (particularly London) there are larger proportions of older mothers and so the prevalence of most chromosomal anomalies will be higher in these areas and the estimated number of cases for chromosomal anomalies is likely to be an under-estimate. NDSCR provides information on Down, Patau and Edwards syndromes for all of England and Wales in chapter 5.

**Table 1.2: Estimated numbers of notifications of congenital anomalies in England and Wales: 2009**

Congenital anomaly <sup>1</sup>	Prevalence per 10,000 total births [95% CI]	Estimated number of notifications in England and Wales
<b>All notifications</b>	<b>205.7 [199.5, 212.0]</b>	<b>14,603</b>
Nervous system	23.8 [21.7, 26.0]	1,687
Congenital heart disease	54.6 [51.4, 57.9]	3,877
Respiratory	5.8 [4.8, 6.9]	409
Oro-facial clefts <sup>2</sup>	15.0 [13.3, 16.7]	1,062
Digestive system	16.6 [14.9, 18.4]	1,178
Abdominal wall defects	9.3 [8.1, 10.8]	664
Urinary	25.4 [23.2, 27.7]	1,803
Genital	15.0 [13.3, 16.7]	1,062
Limb	30.3 [28.0, 32.8]	2,152
Musculo-skeletal	6.7 [5.7, 8.0]	479
Chromosomal <sup>3</sup>	39.8 [37.1, 42.6]	2,826

1 Some of the babies shown in this table will have multiple anomalies and appear in more than one row of the table.

2 Information on cleft lip and/or palate for the whole of England, Wales and Northern Ireland can be found in Chapter 6.

3 Information on Down syndrome, Patau syndrome and Edwards syndrome for the whole of England and Wales can be found in Chapter 5.

## 1.3 Trends over time

To determine trends over time the BINOCAR data alone must be used, not data from NCAS. We present here data for 5 years. Earlier data are available for some registers from the BINOCAR and EUROCAT websites ([www.binocar.org/Data](http://www.binocar.org/Data) or [www.eurocat-network.eu/ACCESSPREVALENCEDATA/PrevalenceTables](http://www.eurocat-network.eu/ACCESSPREVALENCEDATA/PrevalenceTables)).

There has been a non-linear change in the prevalence of births with congenital anomalies over the last 5 years. The prevalence increased between 2005 and 2006 and then significantly decreased from 248 per 10,000 total births in 2006 to 206 per 10,000 total births in 2009 (Table 1.3).

Caution should be taken when interpreting trends as the data in the later years are likely to be incomplete as it takes a while for notifications to be sent to the registers and some anomalies are sometimes not diagnosed until later in childhood.

**Table 1.3: Trends in prevalence (per 10,000 total births) according to congenital anomaly subgroup; five BINOCAR registers (coverage: 28% of births in England and Wales): 2005-2009**

Congenital anomaly <sup>1</sup>	Prevalence per 10,000 total births [95% CI]					Rate of change per year (%)
	2005	2006	2007 <sup>2</sup>	2008	2009	
<b>All notifications</b>	<b>240.6 [233.6, 247.7]</b>	<b>247.9 [240.9, 255.0]</b>	<b>235.9 [229.2, 242.7]</b>	<b>222.5 [216.1, 229.0]</b>	<b>205.7 [199.5, 212.0]</b>	◇
<b>Nervous system</b>	<b>26.1 [23.8, 28.5]</b>	<b>26.1 [23.8, 28.4]</b>	<b>26.0 [23.8, 28.3]</b>	<b>24.0 [22.0, 26.3]</b>	<b>23.8 [21.7, 26.0]</b>	<b>-2.6 [-5.3, 0.1]</b>
Neural tube defects	12.6 [11.1, 14.4]	12.9 [11.3, 14.6]	11.4 [10.0, 13.0]	11.3 [9.9, 12.8]	12.5 [11.1, 14.2]	-1.5 [-5.3, 2.6]
Anencephalus and similar	6.1 [5.0, 7.3]	5.5 [4.5, 6.7]	4.3 [3.5, 5.3]	4.9 [4.0, 6.0]	5.0 [4.1, 6.1]	-5.0 [-10.7, 1.1]
Encephalocele	1.5 [1.0, 2.1]	1.6 [1.0, 2.2]	1.5 [1.0, 2.2]	1.2 [0.8, 1.8]	1.3 [0.9, 1.9]	-4.1 [-14.8, 7.9]
Spina Bifida	5.1 [4.1, 6.2]	5.8 [4.8, 7.0]	5.6 [4.6, 6.7]	5.1 [4.2, 6.2]	6.2 [5.2, 7.4]	2.7 [-3.3, 9.0]
Hydrocephaly	6.0 [4.9, 7.2]	5.6 [4.6, 6.8]	7.4 [6.3, 8.7]	6.4 [5.4, 7.6]	5.8 [4.8, 6.9]	0.4 [-5.0, 6.2]
Microcephaly	1.6 [1.0, 2.2]	1.7 [1.1, 2.3]	1.7 [1.2, 2.4]	1.4 [1.0, 2.0]	1.5 [1.0, 2.1]	-2.6 [-13.0, 8.9]
Arhinencephaly/holoprosencephaly	1.2 [0.8, 1.9]	1.4 [0.9, 2.0]	1.6 [1.1, 2.2]	1.5 [1.0, 2.2]	1.3 [0.8, 1.9]	1.3 [-10.0, 14.1]
<b>Congenital heart disease (CHD)</b>	<b>66.5 [62.8, 70.3]</b>	<b>70.0 [66.4, 73.9]</b>	<b>63.5 [60.0, 67.1]</b>	<b>58.0 [54.7, 61.4]</b>	<b>54.1 [51.0, 57.4]</b>	<b>-5.8 [-7.5, -4.1]</b>
Severe CHD	24.4 [22.2, 26.7]	23.2 [21.1, 25.4]	23.8 [21.7, 26.1]	21.6 [19.6, 23.7]	20.3 [18.4, 22.3]	-4.3 [-7.0, -1.4]
Common arterial truncus	1.1 [0.7, 1.7]	1.3 [0.9, 2.0]	0.5 [0.2, 0.9]	1.0 [0.6, 1.6]	0.6 [0.3, 1.0]	-13.5 [-25.5, 0.5]
Transposition of great vessels	3.2 [2.5, 4.2]	3.5 [2.7, 4.5]	3.3 [2.5, 4.2]	3.1 [2.4, 4.0]	3.7 [2.9, 4.7]	1.9 [-5.6, 10.0]
Single ventricle	0.4 [0.2, 0.8]	0.6 [0.3, 1.1]	0.5 [0.2, 0.9]	0.4 [0.2, 0.8]	0.4 [0.2, 0.8]	-3.4 [-21.1, 18.4]
Ventricular septal defect	26.6 [24.3, 29.1]	29.6 [27.3, 32.2]	25.3 [23.1, 27.6]	23.9 [21.9, 26.2]	21.5 [19.6, 23.7]	-6.2 [-8.7, -3.5]
Atrial septal defect	9.8 [8.4, 11.3]	10.6 [9.2, 12.1]	8.9 [7.6, 10.3]	7.3 [6.2, 8.6]	7.9 [6.7, 9.2]	-7.6 [-11.9, -3.1]
Atrioventricular septal defect	4.9 [3.9, 6.0]	4.2 [3.3, 5.2]	5.0 [4.0, 6.1]	4.5 [3.6, 5.5]	3.7 [2.9, 4.7]	-4.4 [-10.6, 2.1]
Tetralogy of Fallot	3.8 [3.0, 4.8]	3.5 [2.7, 4.4]	4.1 [3.2, 5.1]	4.5 [3.6, 5.5]	3.7 [2.9, 4.6]	1.9 [-5.1, 9.4]
Tricuspid atresia and stenosis	0.8 [0.5, 1.3]	0.8 [0.4, 1.3]	0.6 [0.3, 1.1]	0.8 [0.4, 1.3]	0.8 [0.5, 1.3]	1.0 [-14.0, 18.7]
Ebstein's anomaly	0.5 [0.3, 1.0]	0.5 [0.2, 0.9]	0.6 [0.3, 1.1]	0.4 [0.2, 0.8]	0.6 [0.3, 1.0]	0.6 [-17.2, 22.3]
Pulmonary valve stenosis	6.1 [5.0, 7.3]	5.1 [4.1, 6.2]	5.2 [4.3, 6.3]	4.0 [3.1, 4.9]	4.4 [3.6, 5.4]	-8.5 [-14.1, -2.5]
Pulmonary valve atresia	0.9 [0.5, 1.4]	0.9 [0.6, 1.5]	1.2 [0.8, 1.8]	1.2 [0.8, 1.7]	1.1 [0.7, 1.7]	7.6 [-6.2, 23.3]
Aortic valve atresia/stenosis	1.8 [1.2, 2.5]	1.2 [0.8, 1.9]	1.5 [1.0, 2.1]	1.3 [0.9, 1.9]	1.0 [0.6, 1.6]	-9.8 [-20.0, 1.8]
Hypoplastic left heart	3.4 [2.6, 4.3]	3.8 [3.0, 4.8]	3.8 [3.0, 4.8]	2.4 [1.8, 3.2]	3.0 [2.3, 3.9]	-6.5 [-13.5, 1.0]
Hypoplastic right heart	1.1 [0.7, 1.7]	0.5 [0.2, 0.9]	0.6 [0.3, 1.0]	0.2 [0.1, 0.5]	0.7 [0.4, 1.2]	-15.9 [-30.1, 1.3]
Coarctation of aorta	4.2 [3.3, 5.2]	3.8 [3.0, 4.8]	4.3 [3.4, 5.3]	3.9 [3.1, 4.9]	2.7 [2.0, 3.5]	-7.4 [-13.8, -0.4]
Total anomalous pulmonary venous return	0.5 [0.3, 1.0]	0.9 [0.6, 1.5]	0.9 [0.5, 1.4]	0.6 [0.3, 1.1]	0.9 [0.5, 1.4]	5.1 [-10.5, 23.4]
<b>Respiratory</b>	<b>6.6 [5.4, 7.8]</b>	<b>6.9 [5.8, 8.2]</b>	<b>6.3 [5.3, 7.5]</b>	<b>6.2 [5.2, 7.4]</b>	<b>5.8 [4.8, 6.9]</b>	<b>-3.7 [-8.9, 1.9]</b>
Choanal atresia	0.9 [0.5, 1.4]	0.6 [0.3, 1.1]	0.8 [0.4, 1.2]	0.7 [0.4, 1.2]	0.6 [0.3, 1.0]	-5.8 [-20.2, 11.3]

1 Some of the babies shown in this table will have multiple anomalies and appear in more than one row of the table.

2 The data presented in the BINOCAR column of Table 2 are different to the data presented here. The data in Table 2 includes the South West register and excludes terminations of pregnancy, whereas the data here 3 excludes the South West register and includes terminations of pregnancy.

◇ = Non-linear change

**Table 1.3 cont'd: Trends in prevalence (per 10,000 total births) according to congenital anomaly subgroup; five BINOCAR registers (coverage: 28% of births in England and Wales): 2005-2009**

Congenital anomaly <sup>1</sup>	Prevalence per 10,000 total births [95% CI]					Rate of change per year (%)
	2005	2006	2007 <sup>2</sup>	2008	2009	
Cystic adenomatous malformation of lung	1.4 [0.9, 2.0]	1.7 [1.2, 2.4]	1.7 [1.2, 2.4]	2.0 [1.4, 2.7]	1.5 [1.0, 2.2]	3.2 [-7.4, 15.1]
<b>Oro-facial clefts<sup>3</sup></b>	<b>17.9 [16.0, 19.9]</b>	<b>16.8 [15.1, 18.8]</b>	<b>19.5 [17.6, 21.5]</b>	<b>17.0 [15.2, 18.9]</b>	<b>15.0 [13.3, 16.7]</b>	<b>-3.3 [-6.5, 0.0]</b>
Cleft lip with or without cleft palate	10.5 [9.1, 12.1]	10.1 [8.7, 11.6]	12.3 [10.8, 13.9]	10.4 [9.0, 11.9]	9.2 [7.9, 10.6]	-2.3 [-6.5, 2.0]
Cleft palate	7.4 [6.2, 8.7]	6.8 [5.7, 8.1]	7.2 [6.1, 8.5]	6.6 [5.5, 7.8]	5.8 [4.8, 7.0]	-4.8 [-9.8, 0.5]
<b>Digestive system</b>	<b>18.1 [16.2, 20.1]</b>	<b>18.2 [16.3, 20.2]</b>	<b>16.5 [14.8, 18.4]</b>	<b>15.7 [14.0, 17.5]</b>	<b>16.6 [14.9, 18.4]</b>	<b>-3.1 [-6.4, 0.2]</b>
Oesophageal atresia with or without trachea-oesophageal fistula	2.3 [1.6, 3.1]	1.8 [1.2, 2.5]	2.1 [1.5, 2.8]	2.3 [1.6, 3.0]	2.7 [2.0, 3.5]	6.7 [-3.0, 17.3]
Duodenal atresia or stenosis	1.4 [0.9, 2.0]	2.0 [1.4, 2.7]	1.5 [1.0, 2.2]	2.0 [1.4, 2.7]	1.8 [1.3, 2.5]	4.9 [-5.8, 16.7]
Atresia or stenosis of other parts of small intestine	0.7 [0.4, 1.2]	1.4 [0.9, 2.0]	1.1 [0.7, 1.6]	0.6 [0.3, 1.0]	0.7 [0.4, 1.2]	-9.4 [-22.0, 5.2]
Ano-rectal atresia and stenosis	2.3 [1.6, 3.1]	3.1 [2.4, 4.0]	3.0 [2.3, 3.8]	2.7 [2.0, 3.5]	3.2 [2.5, 4.1]	5.2 [-3.3, 14.3]
Hirschsprung's disease	1.9 [1.3, 2.6]	1.7 [1.1, 2.3]	1.3 [0.9, 1.9]	1.2 [0.8, 1.7]	1.3 [0.9, 1.9]	-10.2 [-20.1, 0.9]
Diaphragmatic hernia	3.7 [2.9, 4.7]	3.3 [2.5, 4.2]	3.1 [2.4, 4.0]	2.8 [2.2, 3.7]	3.2 [2.5, 4.1]	-4.3 [-11.5, 3.5]
<b>Abdominal wall defects</b>	<b>8.9 [7.6, 10.3]</b>	<b>10.3 [8.9, 11.8]</b>	<b>9.5 [8.2, 10.9]</b>	<b>10.4 [9.1, 11.9]</b>	<b>9.3 [8.1, 10.8]</b>	<b>1.0 [-3.4, 5.7]</b>
Gastroschisis	4.4 [3.5, 5.5]	5.9 [4.9, 7.1]	4.8 [3.9, 5.9]	5.7 [4.7, 6.9]	4.7 [3.8, 5.8]	0.8 [-5.3, 7.2]
Omphalocele	3.9 [3.1, 4.9]	3.6 [2.8, 4.6]	4.2 [3.4, 5.2]	3.6 [2.8, 4.5]	3.9 [3.1, 4.9]	0.0 [-6.9, 7.4]
<b>Urinary</b>	<b>29.6 [27.2, 32.2]</b>	<b>30.6 [28.2, 33.1]</b>	<b>29.3 [27.0, 31.8]</b>	<b>30.5 [28.1, 33.0]</b>	<b>25.4 [23.2, 27.7]</b>	<b>-2.9 [-5.4, -0.4]</b>
Bilateral renal agenesis including Potter syndrome	0.9 [0.5, 1.4]	1.5 [1.0, 2.1]	1.5 [1.0, 2.1]	1.6 [1.1, 2.2]	1.5 [1.0, 2.1]	10.2 [-2.4, 24.3]
Renal dysplasia	5.0 [4.0, 6.1]	5.3 [4.4, 6.5]	5.9 [4.9, 7.0]	5.7 [4.7, 6.9]	5.2 [4.3, 6.3]	1.4 [-4.5, 7.7]
Congenital hydronephrosis	13.0 [11.4, 14.8]	12.7 [11.2, 14.4]	11.4 [10.0, 13.0]	11.3 [9.9, 12.8]	10.5 [9.1, 12.0]	-5.4 [-9.2, -1.5]
Bladder exstrophy and/or epispadias	1.0 [0.6, 1.5]	0.9 [0.5, 1.4]	0.5 [0.2, 0.9]	0.8 [0.5, 1.3]	0.8 [0.5, 1.3]	-3.6 [-17.6, 12.9]
Posterior urethral valve and/or prune belly	1.2 [0.7, 1.8]	1.7 [1.2, 2.4]	0.9 [0.5, 1.4]	1.1 [0.7, 1.7]	1.0 [0.6, 1.5]	-8.1 [-19.4, 4.6]
<b>Genital</b>	<b>16.8 [15.0, 18.7]</b>	<b>19.5 [17.6, 21.6]</b>	<b>17.5 [15.7, 19.4]</b>	<b>15.6 [13.9, 17.4]</b>	<b>15.0 [13.3, 16.7]</b>	<b>-4.5 [-7.7, -1.2]</b>
Hypospadias	13.6 [12.0, 15.4]	15.7 [14.0, 17.6]	14.2 [12.6, 16.0]	13.0 [11.5, 14.7]	12.4 [10.9, 14.0]	-3.7 [-7.3, 0.0]
Indeterminate sex	0.9 [0.5, 1.4]	1.6 [1.1, 2.3]	1.2 [0.7, 1.7]	0.8 [0.5, 1.3]	0.7 [0.4, 1.2]	-10.7 [-22.3, 2.6]
<b>Limb</b>	<b>36.9 [34.2, 39.8]</b>	<b>35.7 [33.1, 38.5]</b>	<b>37.5 [34.8, 40.2]</b>	<b>35.1 [32.6, 37.8]</b>	<b>30.3 [28.0, 32.8]</b>	<b>-3.9 [-6.2, -1.6]</b>
Limb reduction	6.7 [5.6, 8.0]	7.0 [5.9, 8.3]	6.5 [5.5, 7.8]	5.9 [4.9, 7.0]	5.7 [4.7, 6.8]	-5.1 [-10.3, 0.3]
Upper limb reduction	4.4 [3.5, 5.5]	4.4 [3.5, 5.4]	4.4 [3.5, 5.5]	4.7 [3.8, 5.7]	4.2 [3.4, 5.2]	-0.3 [-6.7, 6.6]
Lower limb reduction	3.1 [2.3, 4.0]	2.5 [1.9, 3.4]	2.0 [1.4, 2.7]	2.0 [1.4, 2.7]	2.0 [1.4, 2.7]	-11.3 [-19.2, -2.6]

1 Some of the babies shown in this table will have multiple anomalies and appear in more than one row of the table.

2 The data presented in the BINOCAR column of Table 2 are different to the data presented here. The data in Table 2 includes the South West register and excludes terminations of pregnancy, whereas the data here 3 excludes the South West register and includes terminations of pregnancy.

3 Information on cleft lip and/or palate for the whole of England, Wales and Northern Ireland can be found in Chapter 6.

**Table 1.3 cont'd: Trends in prevalence (per 10,000 total births) according to congenital anomaly subgroup; five BINOCAR registers (coverage: 28% of births in England and Wales): 2005-2009**

Congenital anomaly <sup>1</sup>	Prevalence per 10,000 total births [95% CI]					Rate of change per year (%)
	2005	2006	2007 <sup>2</sup>	2008	2009	
Club foot – talipes equinovarus	13.0 [11.4, 14.7]	13.1 [11.5, 14.8]	13.2 [11.6, 14.9]	13.4 [11.9, 15.1]	10.4 [9.0, 11.9]	-3.8 [-7.6, 0.1]
Hip dislocation and/or dysplasia	3.3 [2.5, 4.2]	3.3 [2.5, 4.2]	4.0 [3.2, 5.0]	3.6 [2.8, 4.5]	2.2 [1.6, 3.0]	-5.6 [-12.7, 2.0]
Polydactyly	7.1 [5.9, 8.4]	6.8 [5.7, 8.1]	8.1 [6.9, 9.4]	7.5 [6.4, 8.8]	7.8 [6.6, 9.1]	2.8 [-2.3, 8.3]
Syndactyly	5.3 [4.3, 6.4]	5.0 [4.1, 6.1]	4.4 [3.5, 5.5]	3.9 [3.1, 4.8]	3.5 [2.7, 4.4]	-10.2 [-16.0, -4.0]
<b>Musculo-skeletal</b>	<b>9.7 [8.4, 11.3]</b>	<b>8.8 [7.5, 10.2]</b>	<b>9.3 [8.0, 10.7]</b>	<b>10.5 [9.1, 12.0]</b>	<b>6.7 [5.7, 8.0]</b>	◇
<b>Other malformations</b>	<b>5.4 [4.4, 6.5]</b>	<b>6.0 [4.9, 7.2]</b>	<b>5.9 [4.9, 7.1]</b>	<b>4.6 [3.7, 5.6]</b>	<b>3.9 [3.1, 4.9]</b>	<b>-8.1 [-13.6, -2.2]</b>
<b>Genetic syndromes + microdeletions</b>	<b>9.0 [7.7, 10.4]</b>	<b>8.4 [7.2, 9.8]</b>	<b>7.6 [6.5, 9.0]</b>	<b>5.0 [4.1, 6.1]</b>	<b>4.9 [4.0, 6.0]</b>	<b>-15.5 [-20.0, -10.8]</b>
<b>Chromosomal</b>	<b>46.4 [43.4, 49.6]</b>	<b>45.1 [42.1, 48.2]</b>	<b>40.0 [37.3, 42.9]</b>	<b>39.9 [37.2, 42.7]</b>	<b>39.8 [37.1, 42.6]</b>	<b>-4.3 [-6.3, -2.2]</b>
Down syndrome <sup>4</sup>	24.3 [22.1, 26.7]	22.5 [20.4, 24.7]	21.2 [19.2, 23.3]	21.7 [19.8, 23.9]	21.7 [19.7, 23.8]	-2.6 [-5.5, 0.3]
Patau syndrome/trisomy 13 <sup>4</sup>	2.6 [1.9, 3.4]	2.6 [2.0, 3.5]	2.4 [1.8, 3.2]	2.3 [1.7, 3.1]	2.0 [1.4, 2.7]	-6.4 [-14.6, 2.5]
Edwards syndrome/trisomy 18 <sup>4</sup>	6.2 [5.2, 7.5]	5.8 [4.7, 6.9]	5.6 [4.6, 6.7]	5.9 [4.9, 7.1]	6.1 [5.1, 7.3]	-0.1 [-5.7, 5.8]
Turner's syndrome	2.9 [2.2, 3.8]	3.8 [3.0, 4.8]	2.3 [1.7, 3.1]	3.1 [2.4, 3.9]	2.6 [2.0, 3.4]	-4.5 [-12.0, 3.6]
Klinefelters syndrome	0.5 [0.2, 0.9]	1.1 [0.7, 1.7]	0.9 [0.5, 1.4]	0.4 [0.2, 0.8]	0.5 [0.3, 1.0]	-8.0 [-22.5, 9.0]

<sup>1</sup> Some of the babies shown in this table will have multiple anomalies and appear in more than one row of the table.

<sup>2</sup> The data presented in the BINOCAR column of Table 2 are different to the data presented here. The data in Table 2 includes the South West register and excludes terminations of pregnancy, whereas the data here <sup>3</sup> excludes the South West register and includes terminations of pregnancy.

<sup>4</sup> Information on Down syndrome, Patau syndrome and Edwards syndrome for the whole of England and Wales can be found in Chapter 5.

◇ = Non-linear change

## Chapter 2: Timing of diagnosis and outcome

Of the 4,181 notifications of congenital anomalies in 2009, 53% were diagnosed prenatally, 42% were diagnosed postnatally and for 5% the timing of diagnosis was not known. Of the 2,207 notifications diagnosed prenatally, 950 (43%) pregnancies resulted in a termination of pregnancy for fetal anomaly. Seventy-two percent (421/545) of chromosomal anomalies resulted in a termination of pregnancy for fetal anomaly and 32% (529/1662) of non-chromosomal anomalies.

Of the 1,714 live births diagnosed postnatally, the time of diagnosis was known for 1,311 (76%); of these 68% were diagnosed at birth, 9% in the first week, 7% in the 2<sup>nd</sup> to 4<sup>th</sup> week and 17% after the 1<sup>st</sup> month.

**Table 2.1: Timing of first diagnosis for all anomalies; five BINOCAR registers (coverage: 28% of births in England and Wales): 2009**

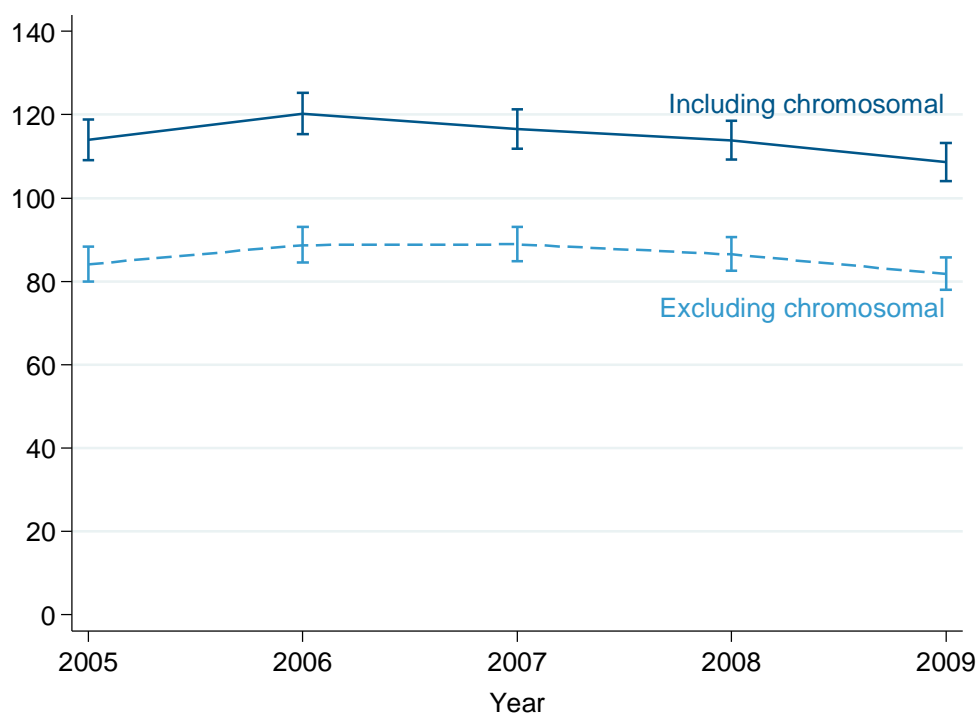
Timing of diagnosis	Number	Percentage [95% CI]
<b>All notifications</b>	<b>4,181</b>	<b>100.0</b>
<b>Prenatal</b>	<b>2,207</b>	<b>52.8 [51.3, 54.3]</b>
Termination of pregnancy	950	22.7 [21.5, 24.0]
Miscarriage (20-23 weeks)	19	0.5 [0.3, 0.7]
Stillbirth (≥24 weeks)	66	1.6 [1.2, 2.0]
Live birth	1,172	28.0 [26.7, 29.4]
<b>Postnatal</b>	<b>1,751</b>	<b>41.9 [40.4, 43.4]</b>
Miscarriage (20-23 weeks)	17	0.4 [0.3, 0.7]
Stillbirth (≥24 weeks)	20	0.5 [0.3, 0.7]
Live birth	1,714	41.0 [39.5, 42.5]
At birth	886	21.2 [20.0, 22.5]
Less than 1 week	114	2.7 [2.3, 3.3]
1-4 weeks	86	2.1 [1.7, 2.5]
Over 1 month	225	5.4 [4.7, 6.1]
Age not known	403	9.6 [8.8, 10.6]
<b>Timing of diagnosis not known</b>	<b>223</b>	<b>5.3 [4.7, 6.1]</b>
Termination of pregnancy	9	0.2 [0.1, 0.4]
Miscarriage (20-23 weeks)	1	0.0 [0.0, 0.1]
Stillbirth (≥24 weeks)	1	0.0 [0.0, 0.1]
Live birth	212	5.1 [4.4, 5.8]

Timing of diagnosis is affected by many factors, including mother's engagement with antenatal services, her choices regarding screening and diagnostic testing, her body mass index, detection rates for screening procedures and difficulty in diagnosing certain anomalies on ultrasound.

### 2.1 Trends over time in prenatal diagnosis

The prevalence of prenatal diagnosis has decreased significantly over the last 5 years when including chromosomal anomalies but is not significantly different when excluding chromosomal anomalies (Figure 2.1 and Table 2.2). When chromosomal anomalies are included, the prevalence decreased from 114 per 10,000 total births in 2005 to 109 per 10,000 total births in 2009.

**Figure 2.1:** Prevalence (per 10,000 total births) of prenatal diagnosis; five BINO CAR registers (coverage: 28% of births in England and Wales): 2005-2009



**Table 2.2:** Number and prevalence (per 10,000 total births) of prenatal diagnosis; five BINO CAR registers (coverage: 28% of births in England and Wales): 2005-2009

Year	Total births	Including chromosomal anomalies		Excluding chromosomal anomalies	
		Number prenatally diagnosed	Prevalence of prenatal diagnosis per 10,000 total births [95% CI]	Number prenatally diagnosed	Prevalence of prenatal diagnosis per 10,000 total births [95% CI]
2005	185,958	2,118	113.9 [109.1, 118.9]	1,564	84.1 [80.0, 88.4]
2006	193,025	2,320	120.2 [115.4, 125.2]	1,712	88.7 [84.5, 93.0]
2007	198,900	2,317	116.5 [111.8, 121.3]	1,768	88.9 [84.8, 93.1]
2008	204,292	2,326	113.9 [109.3, 118.6]	1,766	86.4 [82.5, 90.6]
2009	203,264	2,207	108.6 [104.1, 113.2]	1,662	81.8 [77.9, 85.8]

# Chapter 3: Key public health indicators

## 3.1 Perinatal mortality

In 2009, the perinatal mortality rate in the United Kingdom was 76 per 10,000 total births,<sup>2</sup> of which an estimated 11% had a congenital anomaly (Table 3.1; 8.4/76).

Table 3.1 shows that there were 87 stillbirths and 83 early neonatal deaths notified with congenital anomalies among 203,264 total births giving a perinatal mortality rate of 8 per 10,000 total births. The main congenital anomaly subgroups contributing to perinatal mortality are congenital heart disease (27% of anomaly related perinatal mortality rate), chromosomal (24%), nervous system (20%) and limb (16%). All four of these subgroups contribute more to stillbirths than to early neonatal deaths. Caution should be taken when interpreting this data as some of the births will have more than one anomaly and so may have died from one anomaly but will still appear in the data of the other anomalies.

**Table 3.1: Numbers and mortality rates (per 10,000 total births) according to congenital anomaly subgroups; five BINOCAR registers (coverage: 28% of births in England and Wales): 2009**

Congenital anomaly <sup>1</sup>	Number of stillbirths (SB)	Number of early neonatal deaths (END)	Perinatal mortality rate (per 10,000 total births) [95% CI]
<b>All notifications</b>	<b>87</b>	<b>83</b>	<b>8.4 [7.2, 9.7]</b>
<b>All notifications excluding chromosomal anomalies</b>	<b>58</b>	<b>71</b>	<b>6.3 [5.3, 7.5]</b>
Nervous system	20	14	1.7 [1.2, 2.3]
Congenital heart disease	27	19	2.3 [1.7, 3.0]
Respiratory	7	13	1.0 [0.6, 1.5]
Digestive system	10	14	1.2 [0.8, 1.8]
Abdominal wall defects	5	3	0.4 [0.2, 0.8]
Urinary	6	17	1.1 [0.7, 1.7]
Limb	14	13	1.3 [0.9, 1.9]
Musculo-skeletal	9	3	0.6 [0.3, 1.0]
Chromosomal	29	12	2.0 [1.4, 2.7]

<sup>1</sup> Some of the babies shown in this table will have multiple anomalies and appear in more than one row of the table.

## 3.2 Survival to one year of age

In 2009, there were 3,098 live births notified with a congenital anomaly. Of these 95% survived to one year of age (Table 3.2). Live births diagnosed with genital anomalies had the highest survival rate of 99% to one year of age and live births diagnosed with respiratory anomalies had the lowest survival rate of 79% to one year of age. Caution should be taken when looking at this data as some of the births will have more than one anomaly and so may have died from one anomaly but will still appear in the data of the other anomalies.

<sup>2</sup> Centre for Maternal and Child Enquiries (CMACE) Perinatal Mortality 2009: United Kingdom. CMACE: London, 2011.

**Table 3.2: Number and percentage of notification that survived to one year of age according to major congenital anomaly subgroups; five BINOCAR registers (coverage: 28% of births in England and Wales): 2009**

Congenital anomaly <sup>1</sup>	Live births	Number survived to one year of age	Percentage survived to one year of age [95% CI]
<b>All notifications</b>	<b>3,098</b>	<b>2,943</b>	<b>95.0 [94.2, 95.7]</b>
<b>All notifications excluding chromosomal anomalies</b>	<b>2,759</b>	<b>2,626</b>	<b>95.2 [94.3, 95.9]</b>
Nervous system	148	125	84.5 [77.8, 89.4]
Congenital heart disease	925	867	93.7 [92.0, 95.1]
Respiratory	79	62	78.5 [68.2, 86.1]
Digestive system	279	254	91.0 [87.1, 93.9]
Abdominal wall defects	114	108	94.7 [89.0, 97.6]
Urinary	421	392	93.1 [90.3, 95.2]
Genital	287	284	99.0 [97.0, 99.6]
Limb	503	482	95.8 [93.7, 97.3]
Musculo-skeletal	73	65	89.0 [79.8, 94.3]
Chromosomal	339	317	93.5 [90.4, 95.7]

<sup>1</sup> Some of the babies shown in this table will have multiple anomalies and appear in more than one row of the table.

### 3.3 Rates of termination of pregnancy for fetal anomaly

Table 3.3 shows the rates of TOPFA by timing. The overall rate of TOPFA for the five BINOCAR registers was 47 per 10,000 total births; 26 per 10,000 total births before 20 weeks' gestation and 20 per 10,000 total births from 20 weeks' gestation onwards.

The highest rate of TOPFA was associated with chromosomal anomalies (21 per 10,000 total births), followed by nervous system anomalies (15 per 10,000 total births) and congenital heart diseases (7 per 10,000 total births) (Table 3.3). The majority of notifications with chromosomal anomalies are terminated before 20 weeks' gestation due to screening for Down syndrome<sup>3</sup> whereas notifications with nervous system anomalies and congenital heart disease are terminated from 20 weeks' gestation as they are structural anomalies which will be primarily detected at the 18<sup>+0</sup> to 20<sup>+6</sup> weeks fetal anomaly scan.

<sup>3</sup> <http://fetalanomaly.screening.nhs.uk/> [Accessed: 02/11/2011]

**Table 3.3: Rates of termination of pregnancy (per 10,000 total births) according to major congenital anomaly subgroups; five BINOCAR registers (coverage: 28% of births in England and Wales): 2009**

Congenital anomaly <sup>1</sup>	TOPFA <20 weeks rate per 10,000 total births [95% CI]	TOPFA 20+ weeks rate per 10,000 total births [95% CI]	Total TOPFA rate per 10,000 total births [95% CI]
<b>All notifications</b>	<b>26.4 [24.2, 28.7]</b>	<b>20.4 [18.5, 22.5]</b>	<b>47.2 [44.2, 50.3]</b>
Nervous system	6.8 [5.7, 8.1]	8.4 [7.2, 9.7]	15.3 [13.6, 17]
Congenital heart disease	1.8 [1.2, 2.5]	5.1 [4.2, 6.2]	6.9 [5.8, 8.2]
Respiratory	0.4 [0.2, 0.8]	1.0 [0.6, 1.6]	1.4 [1.0, 2.0]
Digestive system	0.7 [0.4, 1.2]	1.6 [1.1, 2.2]	2.3 [1.7, 3.1]
Abdominal wall defects	2.8 [2.1, 3.6]	0.5 [0.3, 1.0]	3.3 [2.6, 4.2]
Urinary	1.4 [0.9, 2.0]	2.9 [2.2, 3.7]	4.3 [3.4, 5.3]
Genital	0.1 [0.0, 0.4]	0.6 [0.3, 1.0]	0.7 [0.4, 1.2]
Limb	1.4 [1.0, 2.0]	3.1 [2.4, 4.0]	4.5 [3.6, 5.6]
Musculo-skeletal	1.0 [0.6, 1.5]	1.5 [1.0, 2.2]	2.5 [1.9, 3.3]
Chromosomal	15.7 [14.0, 17.5]	4.9 [4.0, 6.0]	20.9 [19.0, 23.0]

<sup>1</sup> Some of the babies shown in this table will have multiple anomalies and appear in more than one row of the table, i.e. The pregnancy may not have been terminated because of that anomaly.

## Chapter 4: Variation by register

Table 4.1 shows the number of notifications of congenital anomalies to each register and Table 4.2 shows the prevalence. There were regional differences in the prevalence estimates for the five registers ( $p < 0.001$ ). The prevalence for CARIS (Wales) was significantly higher and the prevalence for EMSYCAR (East Midlands & South Yorkshire) was significantly lower than the prevalence for all five registers combined. The prevalence will be influenced by both the ascertainment of the register plus the underlying true prevalence which may vary regionally.

**Table 4.1: Number of notifications (LB + FD + TOPFA) by register according to congenital anomaly subgroup; five BINOCAR registers (coverage: 28% of births in England and Wales): 2009**

Congenital anomaly <sup>1</sup>	Number				
	CARIS (Wales)	CAROBB (Thames Valley)	EMSYCAR (East Midlands & South Yorkshire)	NorCAS (Northern England)	WANDA (Wessex)
<b>Total births</b>	<b>35,117</b>	<b>30,392</b>	<b>74,101</b>	<b>33,365</b>	<b>30,289</b>
<b>All notifications</b>	<b>964</b>	<b>608</b>	<b>1,275</b>	<b>717</b>	<b>617</b>
Nervous system	106	70	152	88	67
Congenital heart disease	313	142	234	303	108
Respiratory	45	13	29	12	18
Oro-facial clefts	73	48	82	50	51
Digestive system	96	42	90	55	54
Abdominal wall defects	39	26	65	31	29
Urinary	101	86	153	102	74
Genital	69	48	114	25	48
Limb	145	89	279	25	78
Musculo-skeletal	28	29	35	23	22
Chromosomal	146	121	215	148	179

<sup>1</sup> Some of the babies shown in this table will have multiple anomalies and appear in more than one row of the table.

- = 0

**Table 4.2: Prevalence (per 10,000 total births) by register according to congenital anomaly subgroup; five BINOCAR registers (coverage: 28% of births in England and Wales): 2009**

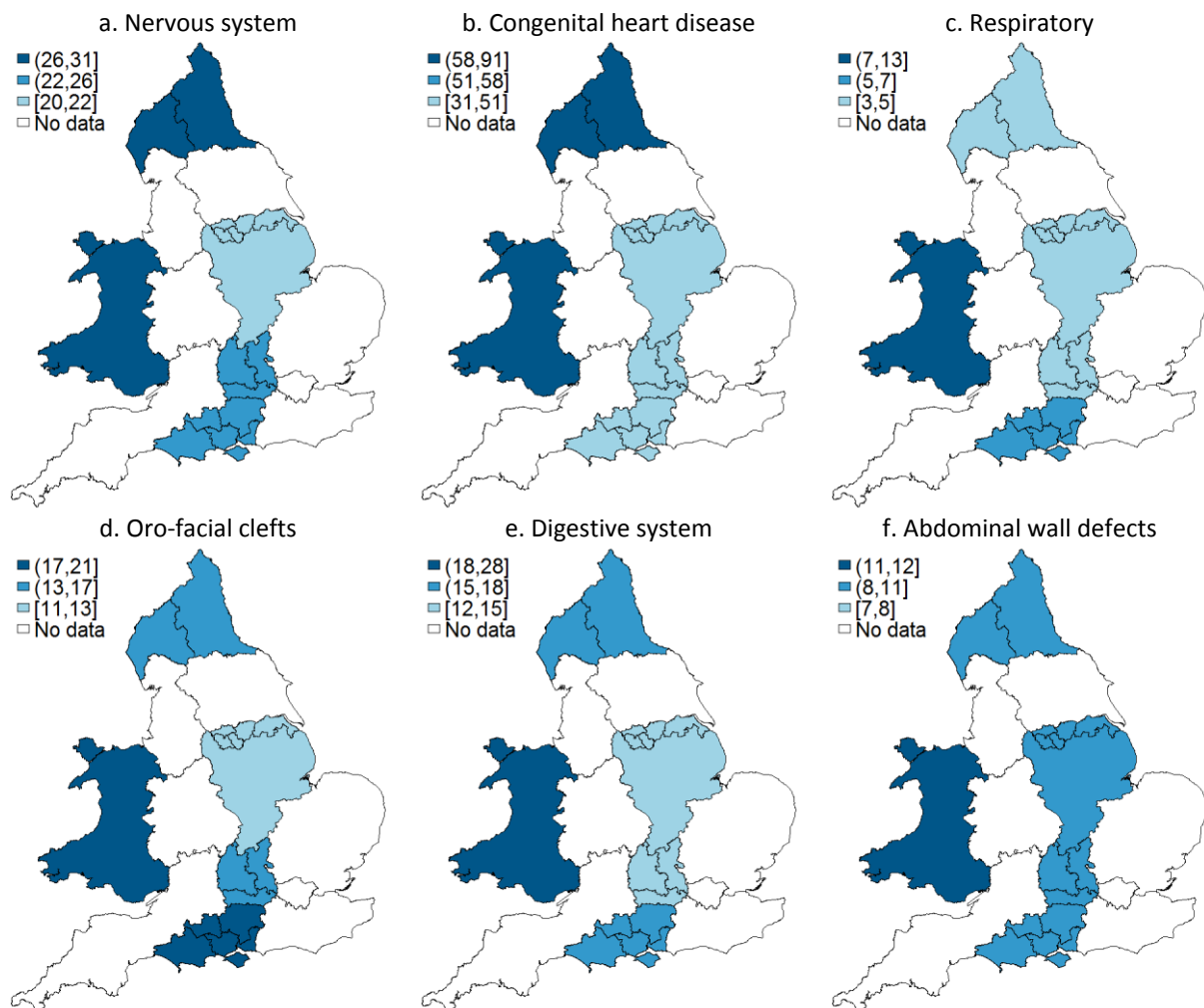
Congenital anomaly <sup>1</sup>	Prevalence per 10,000 total births [95% CI]				
	CARIS (Wales)	CAROB (Thames Valley)	EMSYCAR (East Midlands & South Yorkshire)	NorCAS (Northern England)	WANDA (Wessex)
<b>All notifications</b>	<b>274.5 [257.5, 292.4]</b>	<b>200.1 [184.5, 216.6]</b>	<b>172.1 [162.8, 181.8]</b>	<b>214.9 [199.5, 231.2]</b>	<b>203.7 [188.0, 220.4]</b>
Nervous system	30.2 [24.7, 36.5]	23.0 [18.0, 29.1]	20.5 [17.4, 24.0]	26.4 [21.2, 32.5]	22.1 [17.1, 28.1]
Congenital heart disease	89.1 [79.5, 99.6]	46.7 [39.4, 55.1]	31.6 [27.7, 35.9]	90.8 [80.9, 101.6]	35.7 [29.3, 43.0]
Respiratory	12.8 [9.3, 17.1]	4.3 [2.3, 7.3]	3.9 [2.6, 5.6]	3.6 [1.9, 6.3]	5.9 [3.5, 9.4]
Oro-facial clefts	20.8 [16.3, 26.1]	15.8 [11.6, 20.9]	11.1 [8.8, 13.7]	15.0 [11.1, 19.8]	16.8 [12.5, 22.1]
Digestive system	27.3 [22.1, 33.4]	13.8 [10.0, 18.7]	12.1 [9.8, 14.9]	16.5 [12.4, 21.5]	17.8 [13.4, 23.3]
Abdominal wall defects	11.1 [7.9, 15.2]	8.6 [5.6, 12.5]	8.8 [6.8, 11.2]	9.3 [6.3, 13.2]	9.6 [6.4, 13.8]
Urinary	28.8 [23.4, 34.9]	28.3 [22.6, 34.9]	20.6 [17.5, 24.2]	30.6 [24.9, 37.1]	24.4 [19.2, 30.7]
Genital	19.6 [15.3, 24.9]	15.8 [11.6, 20.9]	15.4 [12.7, 18.5]	7.5 [4.8, 11.1]	15.8 [11.7, 21.0]
Limb	41.3 [34.8, 48.6]	29.3 [23.5, 36.0]	37.7 [33.4, 42.3]	7.5 [4.8, 11.1]	25.8 [20.4, 32.1]
Musculo-skeletal	8.0 [5.3, 11.5]	9.5 [6.4, 13.7]	4.7 [3.3, 6.6]	6.9 [4.4, 10.3]	7.3 [4.6, 11.0]
Chromosomal	41.6 [35.1, 48.9]	39.8 [33.0, 47.6]	29.0 [25.3, 33.2]	44.4 [37.5, 52.1]	59.1 [50.8, 68.4]

<sup>1</sup> Some of the babies shown in this table will have multiple anomalies and appear in more than one row of the table.

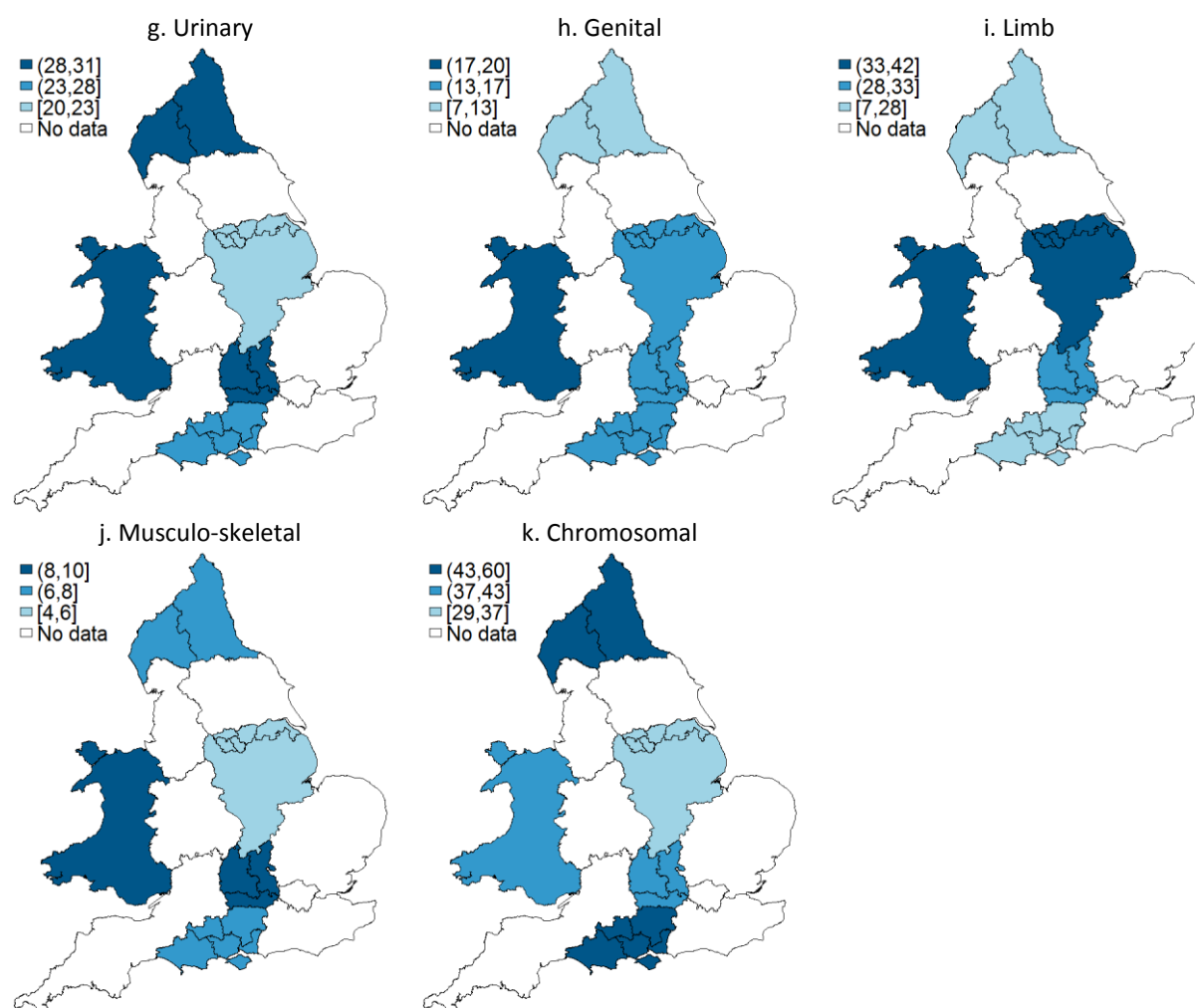
- = 0

Figures 4.1 a-k show the regional variation in the prevalence for major anomaly subgroups. All but abdominal wall defects and musculo-skeletal anomalies showed a significant difference between the registers. The lightest blue represents the registers with prevalence significantly lower than the prevalence for all registers combined, the mid blue represents the registers with prevalence comparable to the prevalence for all registers combined, and the darkest blue represent the registers with prevalence significantly higher than the prevalence for all registers combined. The prevalence will be influenced by both the ascertainment of the register plus the underlying true prevalence which may vary regionally.

**Figures 4.1 a-f: Regional variation in prevalence according to major anomaly subgroups; five BINOCAR registers (coverage: 28% of births in England and Wales): 2009**



**Figures 4.1 g-k: Regional variation in prevalence according to major anomaly subgroups; five BINOCA registers (coverage: 28% of births in England and Wales): 2009**



#### 4.1 Rates of termination of pregnancy for fetal anomaly

WANDA (Wessex) had the highest rate of TOPFA (65 per 10,000 total births) and EMSYCAR (East Midlands & South Yorkshire) had the lowest rate of TOPFA (38 per 10,000 total births) (Table 4.3). The rates will be influenced by many factors including the timing of diagnosis, the mother's choices about screening and diagnostic testing, the detection rates for different anomalies as well as the underlying prevalence of the anomalies.

**Table 4.3: Rates of termination of pregnancy (per 10,000 total births) according to register for all anomalies; five BINOCA registers (coverage: 28% of births in England and Wales): 2009**

Register	TOPFA <20 weeks per 10,000 total births [95% CI]	TOPFA 20+ weeks per 10,000 total births [95% CI]	Total TOPFA per 10,000 total births [95% CI]
CARIS (Wales)	23.6 [18.8, 29.3]	27.1 [21.9, 33.1]	52.1 [44.8, 60.2]
CAROBB (Thames Valley)	25.7 [20.3, 32.0]	21.7 [16.8, 27.6]	48.0 [40.6, 56.6]
EMSYCAR (East Midlands & South Yorkshire)	21.5 [18.3, 25.1]	16.6 [13.8, 19.8]	38.1 [33.7, 42.8]
NorCAS (Northern England)	27.6 [22.2, 33.8]	18.0 [13.7, 23.1]	45.6 [38.6, 53.4]
WANDA (Wessex)	41.3 [34.4, 49.2]	23.4 [18.3, 29.6]	64.7 [56.0, 74.4]

## Chapter 5: Disease specific registers – National Down Syndrome Cytogenetic Register (NDSCR)

The NDSCR was started in 1989 and collects all cytogenetically diagnosed notifications of Down syndrome (trisomy 21), Patau syndrome (trisomy 13) and Edwards syndrome (trisomy 18) occurring in England and Wales. Notifications to the other congenital anomaly registers are included in this data. More information on the NDSCR data is available in the annual report (<http://www.wolfson.qmul.ac.uk/ndscr/reports/NDCSRreport09.pdf>).

### 5.1 Diagnoses of Down, Patau and Edwards syndromes

In 2009, 1,887 Down syndrome, 163 Patau syndrome and 506 Edwards syndrome diagnoses were made.

Of the 1,887 Down syndrome diagnoses, 62% were diagnosed prenatally and 38% were diagnosed postnatally (Table 5.1). The prevalence of Down syndrome diagnoses in England and Wales in 2009 was 27 per 10,000 total births (Table 5.2). Assuming that the proportion of pregnancies resulting in a termination remains as before 2009, the live birth prevalence for Down syndrome diagnoses was 11 per 10,000 live births occurring in England and Wales in 2009.

Of the 163 Patau syndrome diagnoses, 87% were diagnosed prenatally and 13% were diagnosed postnatally (Table 5.1). The prevalence of Patau syndrome diagnoses in England and Wales in 2009 was 2 per 10,000 total births (Table 5.2).

Of the 506 Edwards syndrome diagnoses, 91% were diagnosed prenatally and 9% were diagnosed postnatally (Table 5.1). The prevalence of Edwards syndrome diagnoses in England and Wales in 2009 was 7 per 10,000 total births (Table 5.2).

**Table 5.1: NDSCR notifications diagnosed according to time of diagnosis and outcome; England and Wales: 2009**

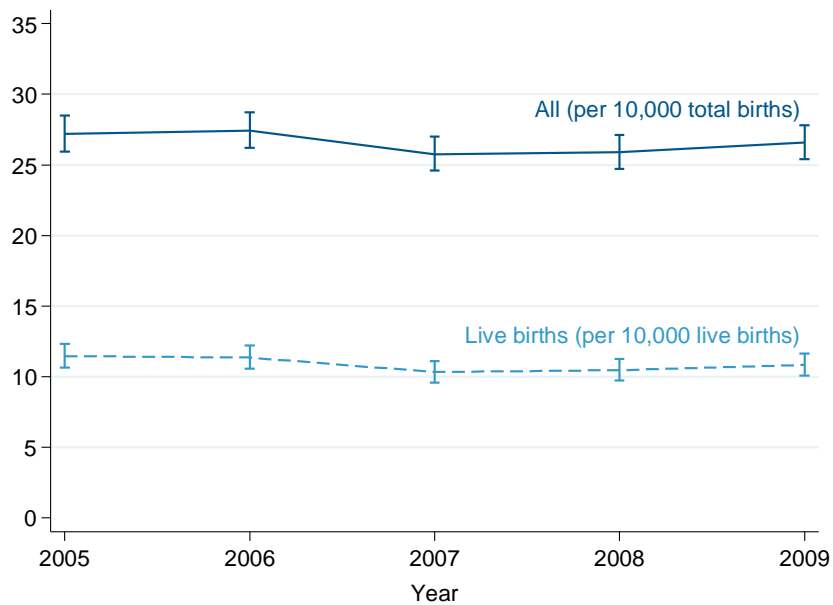
Time of diagnosis	Outcome	Number (Percentage)		
		Down syndrome	Patau syndrome	Edwards syndrome
Prenatal	Miscarriage (20-23 weeks)	24 (1.3)	4 (2.5)	13 (2.6)
	Termination of pregnancy	876 (46.4)	107 (65.6)	340 (67.2)
	Stillbirth (≥24 weeks)	13 (0.7)	2 (1.2)	16 (3.2)
	Live birth	63 (3.3)	4 (2.5)	9 (1.8)
	Unknown outcome	195 (10.3)	25 (15.3)	82 (16.2)
	<b>Total</b>	<b>1,171 (62.1)</b>	<b>142 (87.1)</b>	<b>460 (90.9)</b>
Postnatal	Miscarriage (20-23 weeks)	10 (0.5)	5 (3.1)	1 (0.2)
	Stillbirth (≥24 weeks)	16 (0.8)	1 (0.6)	15 (3.0)
	Live birth	690 (36.6)	15 (9.2)	30 (5.9)
	<b>Total</b>	<b>716 (37.9)</b>	<b>21 (12.9)</b>	<b>46 (9.1)</b>
<b>Total</b>		<b>1,887 (100.0)</b>	<b>163 (100.0)</b>	<b>506 (100.0)</b>

The prevalence of Down syndrome, Patau syndrome and Edwards syndrome have remained constant over the last 5 years (Table 5.2 and Figures 5.1 and 5.2).

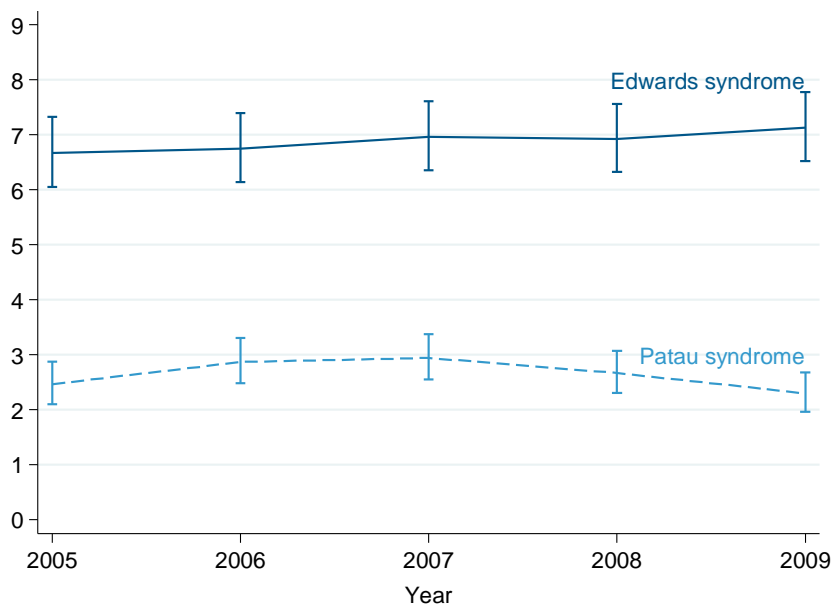
**Table 5.2: Trends over time (per 10,000 total births) in Down syndrome, Patau syndrome and Edwards syndrome; England and Wales: 2005-2009**

Year	Down syndrome		Patau syndrome		Edwards syndrome	
	Number	Prevalence per 10,000 total births [95% CI]	Number	Prevalence per 10,000 total births [95% CI]	Number	Prevalence per 10,000 total births [95% CI]
2005	1,766	27.2 [25.9, 28.5]	160	2.5 [2.1, 2.9]	433	6.7 [6.1, 7.3]
2006	1,846	27.4 [26.2, 28.7]	193	2.9 [2.5, 3.3]	454	6.7 [6.1, 7.4]
2007	1,787	25.8 [24.6, 27.0]	204	2.9 [2.6, 3.4]	483	7.0 [6.4, 7.6]
2008	1,845	25.9 [24.7, 27.1]	190	2.7 [2.3, 3.1]	493	6.9 [6.3, 7.6]
2009	1,887	26.6 [25.4, 27.8]	163	2.3 [2.0, 2.7]	506	7.1 [6.5, 7.8]

**Figure 5.1: Prevalence (per 10,000 births) of Down syndrome; England and Wales: 2005-2009**



**Figure 5.2: Prevalence (per 10,000 total births) of Patau syndrome and Edwards syndrome; England and Wales: 2005-2009**



## 5.2: Regional differences

Table 5.3 shows the number and prevalence of diagnoses of Down syndrome, Patau syndrome and Edwards syndrome across England and Wales according to the maternal region of residence.

London had the highest prevalence of Down syndrome (34 per 10,000 total births) and North West had the lowest (21 per 10,000 total births). The differences in Patau and Edwards syndromes may be due to small numbers.

There is no reason to expect different ascertainment rates and therefore the differences in the prevalence probably reflect the different maternal age distributions in the regions.

**Table 5.3: Number and prevalence (per 10,000 total births) of Down syndrome, Patau syndrome and Edwards syndrome diagnoses according to region of maternal residence; England and Wales: 2009**

Region	Down syndrome		Patau syndrome		Edwards syndrome	
	Number	Prevalence per 10,000 total births [95% CI]	Number	Prevalence per 10,000 total births [95% CI]	Number	Prevalence per 10,000 total births [95% CI]
North East	72	24.1 [18.8, 30.3]	7	2.3 [0.9, 4.8]	18	6.0 [3.6, 9.5]
North West	186	21.1 [18.2, 24.4]	14	1.6 [0.9, 2.7]	46	5.2 [3.8, 7.0]
Yorkshire and Humberside	163	24.4 [20.8, 28.5]	11	1.6 [0.8, 3.0]	36	5.4 [3.8, 7.5]
East Midlands	121	22.4 [18.6, 26.7]	12	2.2 [1.1, 3.9]	33	6.1 [4.2, 8.6]
West Midlands	154	21.6 [18.3, 25.2]	10	1.4 [0.7, 2.6]	43	6.0 [4.4, 8.1]
East England	183	25.5 [22.0, 29.5]	19	2.7 [1.6, 4.1]	58	8.1 [6.1, 10.5]
London	444	34.2 [31.1, 37.5]	39	3.0 [2.1, 4.1]	119	9.2 [7.6, 11.0]
South East	300	28.8 [25.6, 32.3]	25	2.4 [1.6, 3.5]	90	8.6 [7.0, 10.6]
South West	169	28.8 [24.7, 33.5]	19	3.2 [2.0, 5.1]	47	8.0 [5.9, 10.7]
Wales	89	25.3 [20.4, 31.2]	6	1.7 [0.6, 3.7]	15	4.3 [2.4, 7.0]
<b>Total</b>	<b>1,881*</b>	<b>26.5 [25.3, 27.7]</b>	<b>163**</b>	<b>2.3 [2.0, 2.7]</b>	<b>506***</b>	<b>7.1 [6.5, 7.8]</b>

\*6 have unknown region

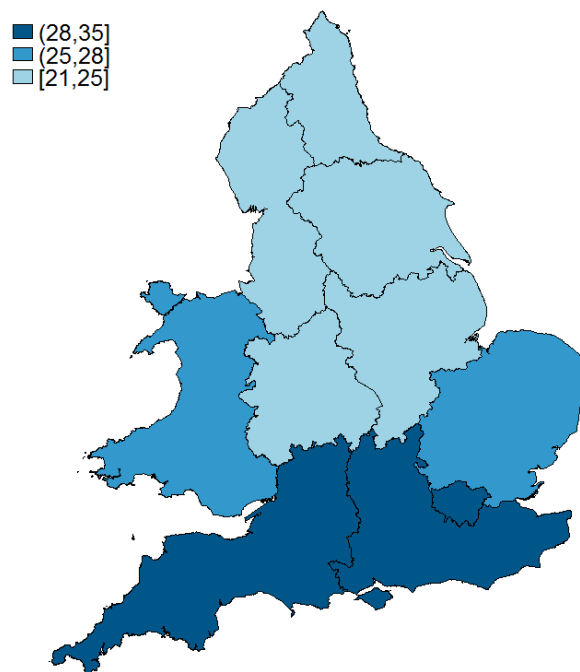
\*\*1 has unknown region

\*\*\*1 has unknown region

Figure 5.3 shows that the south of England had higher prevalence of Down syndrome than the midlands and the north.

The lightest blue represents the regions with prevalence significantly lower than the prevalence for England and Wales, the mid blue represents the regions with prevalence comparable to the prevalence for England and Wales, and the darkest blue represent the regions with prevalence significantly higher than the prevalence for England and Wales.

**Figure 5.3:** Map showing the difference between the regional and national prevalence of Down syndrome; England and Wales: 2009



### 5.3: Prenatal diagnosis

The proportion of notifications prenatally diagnosed with Edwards syndrome has significantly increased; Down syndrome and Patau syndrome has remained constant over the last 5 years (Table 5.4).

**Table 5.4:** Percentage of notifications of Down syndrome, Patau syndrome and Edwards syndrome that were prenatally diagnosed; England and Wales: 2009

Year	Down syndrome		Patau syndrome		Edwards syndrome	
	Number prenatally diagnosed	Percentage prenatally diagnosed [95% CI]	Number prenatally diagnosed	Percentage prenatally diagnosed [95% CI]	Number prenatally diagnosed	Percentage prenatally diagnosed [95% CI]
2005	1,055	59.7 [57.4, 62.0]	138	86.3 [80.1, 90.7]	388	89.6 [86.4, 92.1]
2006	1,118	60.6 [58.3, 62.8]	175	90.7 [85.7, 94.0]	393	86.6 [83.1, 89.4]
2007	1,110	62.1 [59.8, 64.3]	183	89.7 [84.8, 93.2]	442	91.5 [88.7, 93.7]
2008	1,139	61.7 [59.5, 63.9]	171	90.0 [84.9, 93.5]	458	92.9 [90.3, 94.9]
2009	1,171	62.1 [59.8, 64.2]	142	87.1 [81.1, 91.4]	460	90.9 [88.1, 93.1]

#### 5.3.1: Regional differences

Table 5.5 shows the number and percentage of notifications of Down syndrome, Patau syndrome and Edwards syndrome that were prenatally diagnosed. For Down syndrome, South East had the highest percentage prenatally diagnosed (70%) and North East had the lowest percentage (47%). For Patau and Edwards syndromes the regional differences between the proportions of prenatal diagnosis may be due to the small number of cases.

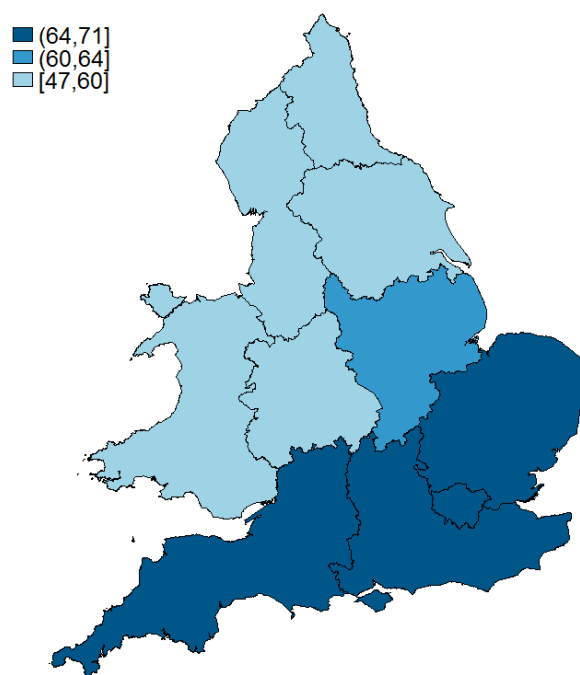
**Table 5.5: Percentage of notifications of Down syndrome, Patau syndrome and Edwards syndrome that were prenatally diagnosed by mother's region of residence; England and Wales: 2009**

Region	Down syndrome		Patau syndrome		Edwards syndrome	
	Number prenatally diagnosed	Percentage prenatally diagnosed [95% CI]	Number prenatally diagnosed	Percentage prenatally diagnosed [95% CI]	Number prenatally diagnosed	Percentage prenatally diagnosed [95% CI]
North East	34	47 [36, 59]	6	86 [49, 97]	16	89 [67, 97]
North West	97	52 [45, 59]	10	71 [45, 88]	38	83 [69, 91]
Yorkshire and Humberside	83	51 [43, 58]	9	82 [52, 95]	35	97 [86, 100]
East Midlands	75	62 [53, 70]	12	100 [76, 100]	33	100 [90, 100]
West Midlands	84	55 [47, 62]	9	90 [60, 98]	35	81 [67, 90]
East England	127	69 [62, 76]	17	89 [69, 97]	55	95 [86, 98]
London	298	67 [63, 71]	35	90 [76, 96]	110	92 [86, 96]
South East	210	70 [65, 75]	25	100 [87, 100]	81	90 [82, 95]
South West	109	64 [57, 71]	14	74 [51, 88]	42	89 [77, 95]
Wales	51	57 [47, 67]	4	67 [30, 90]	14	93 [70, 99]
<b>Total</b>	<b>1,168</b>	<b>62 [60, 64]</b>	<b>142*</b>	<b>87 [81, 91]</b>	<b>460**</b>	<b>91 [88, 93]</b>

\*1 has unknown region  
 \*\*1 has unknown region

The lightest blue represents the regions with significantly lower proportions of prenatal diagnosis than England and Wales, the mid blue represents the regions with comparable proportions to England and Wales, and the darkest blue represent the regions with significantly higher proportions of prenatal diagnosis than England and Wales.

**Figure 5.4: Map showing the difference between the regional and national percentage of notifications of Down syndrome that were prenatally diagnosed; England and Wales: 2009**



For more information about NDSCR or to see their publications go to the following website: <http://www.wolfson.qmul.ac.uk/ndscr/>.

## Chapter 6: Disease specific registers – Cleft lip and palate (CRANE) database

The CRANE database was established in 2000 and collects birth, demographic and diagnosis data on all children born in England, Wales and Northern Ireland with the congenital anomaly of cleft lip and/or palate. The database also records information about cleft-related treatment and outcomes. Hospital Episode Statistics data, linked at the patient level, are used to further examine treatment for cleft lip and/or palate in England.

### 6.1: Births with cleft lip and/or palate

In 2009, the CRANE database registered 878 children born with a cleft lip and/or palate. Of these, 178 (20%) were cleft lip only, 360 (41%) were cleft palate only and 255 (29%) were cleft lip and palate (Table 6.1).

Contributing hospitals report only those patients who have consented to be included on the CRANE database. While CRANE can be used to estimate incidence rates in Wales and Northern Ireland, Hospital Episode Statistics data better reflect incidence rates in England. Using both data sources, the incidence of cleft lip and/or palate in England, Wales and Northern Ireland in 2009 was estimated to be 15 per 10,000 live births (Table 6.2).

**Table 6.1: Number and percentage of children born with a cleft lip and/or palate according to cleft type; England, Wales and Northern Ireland; 2009**

Cleft type	Number	Percentage [95% CI]
Cleft lip	178	20.3 [17.7, 23.1]
Cleft palate	360	41.0 [37.8, 44.3]
Unilateral cleft lip & palate	169	19.2 [16.8, 22.0]
Bilateral cleft lip & palate	86	9.8 [8.0, 11.9]
Not specified	85	9.7 [7.9, 11.8]
<b>Total</b>	<b>878</b>	<b>100.0</b>

Note: Data based on cleft type reported to CRANE

### 6.2: Trends over time

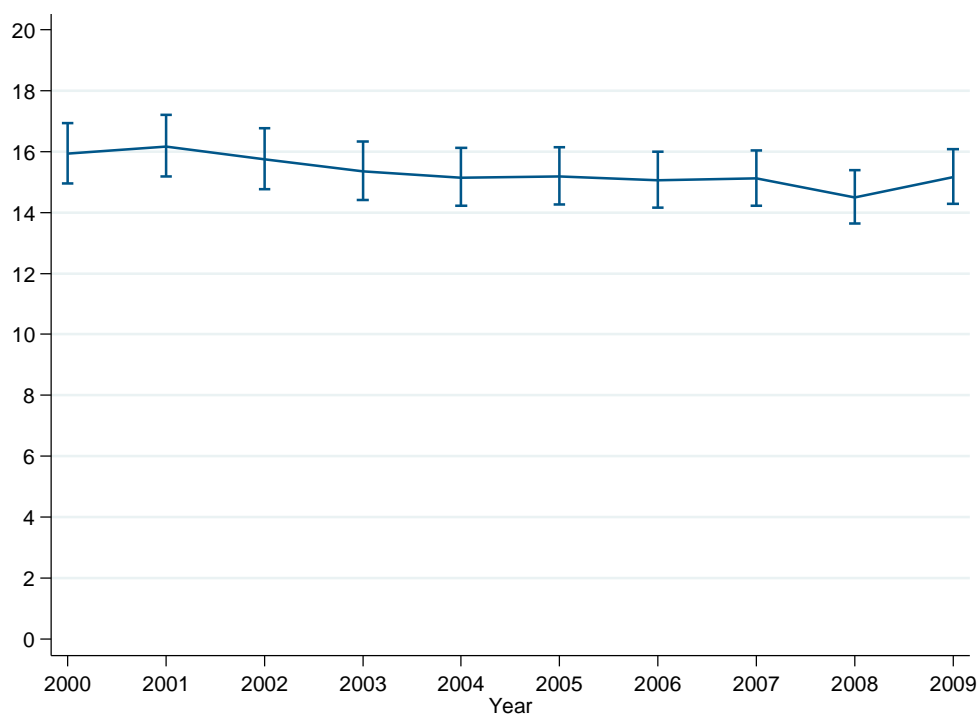
There has been a significant decrease ( $p=0.01$ ) in the incidence of cleft lip and/or palate over the last 10 years in England, Wales and Northern Ireland. The incidence was highest in 2001, when 16.2 per 10,000 live births were affected with cleft lip and/or palate. The lowest rate was in 2008, when 14.5 per 10,000 live births were affected (Table 6.2 and Figure 6.1).

**Table 6.2: Trend in cleft lip and/or palate; England, Wales and Northern Ireland: 2000-2009**

Year	Number	Incidence per 10,000 live births [95% CI]
2000	997	15.9 [15.0, 16.9]
2001	997	16.2 [15.2, 17.2]
2002	972	15.7 [14.8, 16.8]
2003	987	15.3 [14.4, 16.3]
2004	1,003	15.2 [14.2, 16.1]
2005	1,015	15.2 [14.3, 16.2]
2006	1,044	15.1 [14.2, 16.0]
2007	1,080	15.1 [14.2, 16.0]
2008	1,064	14.5 [13.6, 15.4]
2009	1,109	15.2 [14.3, 16.1]

Note: Cleft trends based on Hospital Episode Statistics data for England and CRANE data for Wales and Northern Ireland. Numbers reflect children who were both diagnosed and treated for cleft lip and/or palate.

**Figure 6.1: Incidence (per 10,000 live births) of cleft lip and/or palate; England, Wales and Northern Ireland: 2009**



Note: Cleft trends based on Hospital Episode Statistics data for England and CRANE data for Wales and Northern Ireland. Numbers reflect children who were both diagnosed and treated for cleft lip and/or palate.

Approximately 22% of all facial clefts are associated with additional congenital anomalies or syndromes. Table 6.3 shows that over the last 10 years, the proportion of children born in England with a cleft lip and/or palate who were diagnosed with associated syndromes has ranged from 20.7% to 24.7%. Although there have been some small annual fluctuations, the proportion of children with syndromic clefts has remained fairly consistent.

**Table 6.3: Trend in non-syndromic and syndromic cleft lip and palate; England: 2000-2009**

Year	Non-syndromic clefts		Syndromic clefts	
	Number	Percentage [95% CI]	Number	Percentage [95% CI]
2000	738	78.9 [76.2, 81.4]	197	21.1 [18.6, 23.8]
2001	699	75.3 [72.4, 78.0]	229	24.7 [22.0, 27.6]
2002	727	79.3 [76.5, 81.8]	190	20.7 [18.2, 23.5]
2003	710	76.8 [74.0, 79.4]	214	23.2 [20.6, 26.0]
2004	733	78.0 [75.2, 80.5]	207	22.0 [19.5, 24.8]
2005	732	78.3 [75.5, 80.8]	203	21.7 [19.2, 24.5]
2006	735	76.9 [74.1, 79.4]	221	23.1 [20.6, 25.9]
2007	785	78.9 [76.3, 81.3]	210	21.1 [18.7, 23.7]
2008	768	77.3 [74.6, 79.8]	225	22.7 [20.2, 25.4]
2009	816	79.2 [76.6, 81.6]	214	20.8 [18.4, 23.4]

Note: Syndromic-cleft trends based on Hospital Episode Statistics data. Numbers reflect children who were both diagnosed and treated for cleft lip and/or palate.

### 6.3: Regional differences

Since 1998, cleft services in the United Kingdom have undergone a process of centralisation. There are now 15 specialist units providing cleft treatment to children in England, Wales and Northern Ireland. Table 6.4 shows that Great Ormond Street hospital reported treating the greatest proportion of children born with cleft lip and/or palate in 2009 (11%), while Salisbury reported treating the smallest proportion (3%). The number of treated children reported by individual units is determined by the size of the population in its catchment area as well as case ascertainment (i.e. proportion of children born with a cleft reported to CRANE out of all children born with cleft), and does not reflect regional variations in cleft lip and/or palate incidence rates.

**Table 6.4: Number and percentage of children born with a cleft lip and/or palate reported to CRANE by the unit providing the cleft treatment; England, Wales and Northern Ireland: 2009**

Regional centre / Managed Clinical Network	Specialists Unit	Number	Percentage [95% CI]
Northern & Yorkshire	Newcastle	57	6.5 [5.0, 8.3]
	Leeds	64	7.3 [5.7, 9.2]
North West	Liverpool	74	8.4 [6.8, 10.5]
	Manchester	65	7.4 [5.9, 9.3]
Trent	Nottingham	83	9.5 [7.7, 11.6]
West Midlands	Birmingham	75	8.5 [6.9, 10.6]
East	Cambridge	60	6.8 [5.3, 8.7]
North Thames	Great Ormond Street	93	10.6 [8.7, 12.8]
	Chelmsford	35	4.0 [2.9, 5.5]
The Spires	Oxford	46	5.2 [4.0, 6.9]
	Salisbury	30	3.4 [2.4, 4.8]
South Wales/South West	Swansea	45	5.1 [3.9, 6.8]
	Bristol	45	5.1 [3.9, 6.8]
South Thames	Guy's	75	8.5 [6.9, 10.6]
Northern Ireland	Belfast	31	3.5 [2.5, 5.0]
<b>Total</b>		<b>878</b>	<b>100.0</b>

Note: Data based on registrations in the CRANE database.

For more information about the CRANE database or to see their publications go to the following website: <https://www.crane-database.org.uk/>.

# Appendix A – List of conditions for exclusion

## Head

- Aberrant scalp hair patterning
- Flat occiput
- Dolichocephaly
- Plagiocephaly – head asymmetry
- Bony occipital spur
- Third fontanel
- Macrocephalus
- Facial asymmetry
- Compression facies
- Other cong deformities of skull, face and jaw

## Eyes

- Epicanthic folds
- Epicanthus inversus
- Upward slanting palpebral fissures
- Downward slanting palpebral fissures
- Short palpebral fissures
- Congenital ectropion
- Congenital entropion
- Other congenital malformations of eyelid
- Dystopia canthorum
- Hypertelorism
- Hypotelorism
- Stenosis or stricture of lacrimal duct
- Synophrys
- Blue sclera
- Crocodile tears

## Ears

- Primitive shape
- Lack of helical fold
- Asymmetric size
- Posterior angulation
- Microtia
- Macrotia
- Protuberant ears
- Absent tragus
- Double lobule
- Accessory auricle, preauricular appendage, tag or lobule
- Auricular pit
- Preauricular sinus or cyst
- Narrow external auditory meatus
- Low set ears
- Bat ear, prominent ear
- Unspecified and minor malformation of ear
- Congenital malformation of face and neck unspecified

## Nose

- Small nares
- Notched alas

## Oral regions

- Borderline small mandible
- Aberrant frenula
- Enamel hypoplasia
- Malformed teeth
- High arched palate
- Tongue tie
- Macroglossia
- Macrostomia
- Microstomia
- Macrocheilia
- Microcheilia
- Ranula

## Neck

- Mild webbed neck
- Sinus, fistula or cyst of branchial cleft
- Preauricular sinus or cyst
- Other branchial cleft malformations
- Congenital malformation of face and neck, unspecified
- Torticollis

## Hands

- Duplication of thumbnail
- Enlarged or hypertrophic nails
- Single/abnormal palmar crease
- Unusual dermatoglyphics
- Clinodactyly (5th finger)
- Short fingers (4th and 5th fingers)
- Accessory carpal bones

## Feet – Limb

- Syndactyly (2nd and 3rd toes)
- Gap between toes (1st and 2nd)
- Short great toe
- Recessed toes (4th and 5th)
- Enlarged or hypertrophic nails
- Prominent calcaneus
- Clicking hip, subluxation or unstable hip
- Metatarsus varus or metatarsus adductus
- Hallux varus – other cong varus deformities of feet
- Talipes or pes calcaneovalgus
- Congenital pes planus
- Metatarsus varus – other cong valgus deformities of feet
- Pes cavus
- Clubfoot of postural origin - other cong deformities of feet
- Congenital deformity of feet, unspecified

## Skin

- Haemangioma (other than face or neck)

- Pigmented naevus – cong non-neoplastic naevus
- Nevus flammeus
- Strawberry nevus
- Lymphangioma
- Angioma
- Persistent lanugo
- Mongoloid spot (whites)
- Depigmented spot
- Unusual placement of nipples
- Accessory nipples
- Cafe-au-lait spot

#### **Skeletal**

- Cubitus valgus
- Prominent sternum
- Depressed sternum
- Sternum bifidum
- Shieldlike chest, other cong deformities of chest
- Congenital deformity of spine
- Genua valgum
- Genua varum
- Genu recurvatum
- Congenital bowing of femur
- Congenital bowing of fibula and tibia
- Congenital bowing of long bones of leg, unspecified
- Spina bifida occulta
- Sacral dimple
- Cervical rib
- Fused ribs
- Accessory rib
- Congenital lordosis, postural

#### **Brain**

- Arachnoid cyst
- Choroid plexus cyst
- Anomalies of septum pellucidum

#### **Cardiovascular**

- Absence or hypoplasia of umbilical artery, single umbilical artery
- Functional or unspecified cardiac murmur
- Patent ductus arteriosus if gestational age < 37 weeks
- Peripheral pulmonary artery stenosis
- Patent or persistent foramen ovale

#### **Pulmonary**

- Accessory lobe of lung
- Congenital laryngeal stridor
- Laryngomalacia
- Tracheomalacia
- Azygos lobe of lung

#### **Gastro-intestinal**

- Hiatus hernia
- Pyloric stenosis
- Diastasis recti
- Umbilical hernia
- Inguinal hernia
- Meckel's diverticulum
- Functional gastro-intestinal disorders
- Anterior anus

#### **Renal**

- Vesico-urethral-renal reflux
- Hydronephrosis with a pelvis dilatation less than 10 mm
- Hyperplastic and giant kidney

#### **External genitals**

- Deficient or hooded foreskin
- Undescended testicle
- Unspecified ectopic testis
- Retractable testis
- Hydrocele of testis
- Phimosi
- Bifid scrotum
- Curvature of penis lateral
- Hypoplasia of penis
- Hymen imperforatum
- Fusion of labia

#### **Other**

- Congenital malformation, unspecified

#### **Chromosomal**

- Balanced translocations or inversions in normal individuals

#### **“Non-congenital” anomalies**

- Hydrocephaly where a result of preterm birth rather than congenital: all cases among preterm births

#### **Poorly specified anomalies**

- Functional or unspecified cardiac murmur
- Laryngomalacia and tracheomalacia
- Functional gastro-intestinal disorders
- Undescended testicle. Registries may choose to record this locally if they can follow up all to ascertain whether the testis descends normally.
- Unspecified ectopic testis
- Vesico-ureteral reflux. Registries should record and transmit to EUROCAT the underlying anomaly, if present.
- Clicking hip
- Clubfoot where there is no further specification of whether malformation or postural origin.

## Appendix B – BINOCAR Coding Framework

Subgroups	ICD-10 Codes	ICD-10 Condition
<b>Nervous system</b>	<b>Q00–Q07</b>	
Neural tube defects	Q00, Q01, Q05	Anencephaly and similar malformations; Encephalocele; Spina bifida
Anencephalus and similar	Q00	Anencephaly and similar malformations
Encephalocele	Q01	Encephalocele
Spina Bifida	Q05	Spina bifida
Hydrocephaly	Q03	Hydrocephalus
Microcephaly	Q02	Microcephaly
Arhinencephaly/holoprosencephaly	Q04.1–Q04.2	Arhinencephaly/holoprosencephaly
<b>Eye</b>	<b>Q10.0, Q10.4, Q10.6–Q10.7, Q11–Q15 Exclude: Q13.5</b>	
Anophthalmos/microphthalmos	Q11.0–Q11.2	Anophthalmos, microphthalmos
Anophthalmos	Q11.0–Q11.1	
Congenital cataract	Q12.0	Congenital cataract
Congenital glaucoma	Q15.0	Congenital glaucoma Buphthalmos Glaucoma of newborn Hydrophthalmos Keratoglobus, congenital Macrophthalmos in congenital glaucoma Megalocornea
<b>Ear, face and neck</b>	<b>Q16, Q17.8, Q18.3, Q18.7–Q18.8</b>	
Anotia	Q16.0	Congenital absence of (ear) auricle
<b>Congenital heart disease</b>	<b>Q20–Q26 Exclude: Q25.0 with gestation &lt;37wks</b>	
Common arterial truncus	Q20.0	Common arterial trunk Persistent truncus arteriosus
Transposition of great vessels	Q20.3	Discordant ventriculoarterial connection Dextrotransposition of aorta Transposition of great vessels (complete)
Single ventricle	Q20.4	Double inlet ventricle Common ventricle Cor triloculare biatriatum Single ventricle
Ventricular septal defect	Q21.0	Ventricular septal defect
Atrial septal defect	Q21.1	Atrial septal defect Coronary sinus defect Patent or persistent: · foramen ovale · ostium secundum defect (type II) Sinus venosus defect
Atrioventricular septal defect	Q21.2	Atrioventricular septal defect Common atrioventricular canal Endocardial cushion defect Ostium primum atrial septal defect (type I)
Tetralogy of Fallot	Q21.3	Tetralogy of Fallot Ventricular septal defect with pulmonary stenosis or atresia, dextroposition of aorta and hypertrophy of right ventricle.
Tricuspid atresia and stenosis	Q22.4	Congenital tricuspid stenosis Tricuspid atresia
Ebstein's anomaly	Q22.5	Ebstein's anomaly
Pulmonary valve stenosis	Q22.1	Congenital pulmonary valve stenosis
Pulmonary valve atresia	Q22.0	Pulmonary valve atresia
Aortic valve atresia/stenosis	Q23.0	Congenital stenosis of aortic valve Congenital aortic: · atresia · stenosis Excludes: congenital subaortic stenosis (Q24.4); that in hypoplastic left heart syndrome (Q23.4)
Hypoplastic left heart	Q23.4	Hypoplastic left heart syndrome Atresia, or marked hypoplasia of aortic orifice or valve, with hypoplasia of ascending aorta and defective development

		of left ventricle (with mitral valve stenosis or atresia)
Hypoplastic right heart	Q22.6	Hypoplastic right heart syndrome
Coarctation of aorta	Q25.1	Coarctation of aorta Coarctation of aorta (preductal) (postductal)
Total anomalous pulmonary venous	Q26.2	Total anomalous pulmonary venous connection
<b>Respiratory</b>	<b>Q30–Q34</b> <b>Excludes: Q31.4, Q32.0</b>	
Choanal atresia	Q30.0	Choanal atresia Atresia, Congenital stenosis; (of nares [anterior][posterior])
Cystic adenomatous malformation of lung	Q33.80	Cystic adenomatoid malformation of lung
<b>Oro-facial clefts</b>	<b>Q35–Q37</b>	
Cleft lip with or without palate	Q36-Q37	Cleft lip - cleft palate with cleft lip
Cleft palate	Q35	Cleft palate Includes: fissure of palate palatoschisis Excludes: cleft palate with cleft lip (Q37)
<b>Digestive system</b>	<b>Q38–Q39, Q40.2–Q40.9, Q41–Q45, Q79.0</b> <b>Excludes: Q38.1, Q38.2, Q38.50, Q40.21, Q43.0, Q43.20, Q43.81, Q43.82</b>	
Oesophageal atresia with or without tracheo-oesophageal fistula	Q39.0–Q39.1	Atresia of oesophagus without fistula Atresia of oesophagus NOS - Oesophageal web
Duodenal atresia or stenosis	Q41.0	Congenital absence, atresia and stenosis of duodenum
Atresia or stenosis of other parts of the small intestine	Q41.1–Q41.8	Atresia or stenosis of other parts of the small intestine
Ano-rectal atresia and stenosis	Q42.0–Q42.3	Ano-rectal atresia and stenosis
Hirschsprung's disease	Q43.1	Hirschsprung's disease Aganglionosis Congenital (aganglionic) megacolon
Atresia of bile ducts	Q44.2	Atresia of bile ducts
Annular pancreas	Q45.1	Annular pancreas
Diaphragmatic hernia	Q79.0	Congenital diaphragmatic hernia Excludes: congenital hiatus hernia (Q40.1)
<b>Abdominal wall defects</b>	<b>Q79.2, Q79.3, Q79.5</b>	
Gastroschisis	Q79.3	Gastroschisis
Omphalocele	Q79.2	Exomphalos Omphalocele Excludes: umbilical hernia (K42)
<b>Urinary</b>	<b>Q60–Q64, Q79.4</b> <b>Excludes: Q62.7, Q63.3</b>	
Bilateral renal agenesis including Potter syndrome	Q60.1, Q60.6	
Renal dysplasia	Q61.4	Renal Dysplasia
Congenital hydronephrosis	Q62.0	Congenital hydronephrosis
Bladder exstrophy and or epispadias	Q64.1, Q64.0	Exstrophy of urinary bladder Ectopia vesicae Extroversion of bladder Epispadias Excludes: hypospadias (Q54)
Posterior urethral valve and/or prune belly	Q64.20, Q79.4	
<b>Genital</b>	<b>Q50–Q52, Q54–Q56</b> <b>Excludes: Q52.3, Q52.5</b>	
Hypospadias	Q54	
Indeterminate sex	Q56	Indeterminate sex and pseudohermaphroditism Excludes: pseudohermaphroditism: · female, with adrenocortical disorder (E25) · male, with androgen resistance (E34.5) · with specified chromosomal anomaly (Q96-Q99)
<b>Limbs</b>	<b>Q65.0-Q65.2, Q65.8-Q65.9, Q66.0, Q68.1-Q68.2, Q68.8, Q69-Q74</b> <b>Excludes: Q68.21</b>	
Limb reduction	Q71–Q73	Reduction defects of upper limb - Reduction defects of unspecified limb

Upper limb reduction	Q71	
Lower limb reduction	Q72	
Complete absence of a limb	Q71.0, Q72.0, Q73.0	
Club foot – talipes equinovarus	Q66.0	Talipes equinovarus
Hip dislocation and/or dysplasia	Q65.0–Q65.2, Q65.80, Q65.81	
Polydactyly	Q69	Polydactyly
Syndactyly	Q70	Syndactyly
Arthrogryposis multiplex congenita	Q74.3	Arthrogryposis multiplex congenita
<b>Musculo-skeletal</b>	<b>Q75.0–Q75.1, Q75.4–Q75.9, Q76.1–Q76.4, Q76.6–Q76.9, Q77, Q78, Q79.6–Q79.9</b>	
Thanatophoric dwarfism	Q77.1	Thanatophoric short stature
Jeune syndrome	Q77.2	Short rib syndrome Asphyxiating thoracic dysplasia [Jeune]
Achondroplasia	Q77.4	Achondroplasia/Hypochondroplasia
Craniosynostosis	Q75.0	Craniosynostosis Acrocephaly Imperfect fusion of skull Oxycephaly Trigonocephaly
Congenital constriction bands/amniotic bands	Q79.8	
<b>Other malformations</b>	<b>Q27, Q28, Q80–Q85, Q89</b> <b>Excludes: Q27.0, Q82.5, Q82.80, Q83.3, Q84.5, Q89.9</b>	
Asplenia	Q89.0	
Situs inversus	Q89.3	Situs inversus Dextrocardia with situs inversus Mirror-image atrial arrangement with situs inversus Situs inversus or transversus: · abdominalis · thoracis Transposition of viscera: · abdominal · thoracic Excludes: dextrocardia NOS (Q24.0)
Conjoined twins	Q89.4	Conjoined twins Craniopagus Dicephaly Double monster Pygopagus Thoracopagus
Disorders of skin	Q80–Q82	Disorders of skin
<b>Teratogenic syndromes with malformations</b>	<b>Q86, P35.0, P35.1, P37.1</b>	
Fetal alcohol syndrome	Q86.0	Fetal alcohol syndrome (dysmorphic)
Valproate syndrome	Q86.8	
Warfarin syndrome	Q86.2	Dysmorphism due to warfarin
Maternal infections resulting in malformations	P35.0, P35.1, P37.1	Maternal infections resulting in malformations
<b>Genetic syndromes + microdeletions</b>	<b>Q87, Q93.6, D82.1</b>	
<b>Chromosomal</b>	<b>Q90–Q93, Q96–Q99</b> <b>Excludes: Q93.6, Q95</b>	
Down syndrome	Q90	Down's syndrome
Patau syndrome/trisomy 13	Q91.4–Q91.7	
Edwards syndrome/trisomy 18	Q91.0–Q91.3	
Turner's syndrome	Q96	Turner's syndrome Excludes: Noonan's syndrome (Q87.1)
Klinefelters syndrome	Q98.0–Q98.4	
Cri-du-chat syndrome	Q93.4	Deletion of short arm of chromosome 5 Cri-du-chat syndrome
Wolf-Hirschhorn syndrome	Q93.3	Deletion of short arm of chromosome 4 Wolff-Hirschorn syndrome

## Appendix C – Area coverage of each register

CARIS (Wales)	All of Wales
CAROB (Thames Valley)	<b>Counties:</b> Oxfordshire Buckinghamshire <b>Unitary Authorities:</b> Bracknell West Berkshire Reading Slough Windsor & Maidenhead Wokingham Milton Keynes
EMSYCAR (East Midlands & South Yorkshire)	Derbyshire Leicestershire Lincolnshire Northamptonshire Nottinghamshire South Yorkshire North Lincolnshire PCT North East Lincolnshire PCT  Areas excluded in 2006: South Derbyshire, Lincolnshire and Nottinghamshire.
NorCAS (Northern England)	<b>Strategic Health Authority:</b> North East <b>Districts:</b> Allerdale Carlisle Copeland Eden
WANDA (Wessex)	<b>Counties:</b> Dorset Hampshire (excluding parts of Hart and East Hampshire and the whole of Rushmore) <b>Unitary Authorities:</b> Poole Isle of Wight Bournemouth Portsmouth Southampton <b>Districts:</b> Salisbury Parts of Arun and Chichester



British Isles Network of Congenital Anomaly Registers  
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