Congenital Anomaly Register for
Oxfordshire, Berkshire & Buckinghamshire

Fourth report of the

Congenital Anomaly Register for Oxfordshire, Berkshire and Buckinghamshire

(CAROBB)

Births in 2005-2012

Births within Oxfordshire 1991-2012

2014

National Perinatal Epidemiology Unit

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Confidentiality and policy on non-disclosure of small numbers

As a member of BINOCAR (British Isles Network of Congenital Anomaly Registers), CAROBB has the approval of the Trent MREC and the Confidentiality Advisory Group (CAG) of the Health Research Authority, (and previously the National Information Governance Board (NIGB) and Patient Information Advisory Group (PIAG)), to collect identifiable information without explicit consent from individuals registered. See documentation in Appendix 5.

We have followed the BINOCAR policy concerning the disclosure of small numbers (*www.binocar.org/methods/dataconfidentiality*).

Foreword

The Oxford Congenital Anomaly Register (OXCAR) was established in 1991 at the John Radcliffe Hospital by Dr Patricia Boyd, Clinical Geneticist for Prenatal Diagnosis. The aim of OXCAR was to collect information about the unselected local population of fetuses and babies affected by congenital anomalies for women resident in the area with an OX postcode. One of the main goals of the register was to monitor and evaluate the impact of the then newly emerging prenatal diagnosis technologies and in particular to assess the accuracy of antenatal ultrasound scanning used in the screening for structural congenital anomalies.

In 2002 the Department of Health put out a call for competitive bids to fund research active disease registers. With colleagues from the National Perinatal Epidemiology Unit (NPEU) Tricia successfully bid to expand OXCAR to become a fully established population-based based congenital anomaly register covering Berkshire and Buckinghamshire as well as Oxfordshire and aptly named the Congenital Anomaly Register for Oxfordshire, Berkshire and Buckinghamshire (CAROBB). With Cath Rounding appointed as Co-ordinator, Tricia as Clinical Director and the move to NPEU CAROBB has gone from strength to strength.

Funding for disease registers is typically difficult to secure since the registers themselves are a platform for research rather than a research activity *per se*. However, given the rarity of the very many conditions which fall under the category of congenital anomalies, the cost of establishing a data collection system and mounting a new study each time a new condition needs to be investigated would be prohibitively expensive. Furthermore, congenital anomalies registers fulfil other vital functions which go beyond research into the causes and consequences of anomalies which include: contributing to the evaluation of the national fetal anomaly screening programme; providing data to support health care policy development and service planning; monitoring prevalence rates to identify and investigate potential clusters of anomalies and putative teratogens; and monitoring the impact of other aspects of population trends on congenital anomalies including the effects of maternal age, ethnicity and social inequalities.

Data from CAROBB together with data from five of the other regional registers, which form part of the British Isles Network of Congenital Anomaly Registers (BINOCAR), covering 36% of births in England and Wales, have been combined to provide national monitoring of congenital anomalies taking over this function from the Office for National Statistics run National Congenital Anomaly System (NCAS) in 2010. This was necessitated by the evidence that NCAS under-reported congenital anomalies by at least 50% when directly compared to data collected by the regional congenital anomaly registers. Thus the transfer to BINOCAR led to a vast improvement in ascertainment and accuracy of congenital anomaly data through the use of a combination of routine and bespoke data sources. Public Health England (PHE) which has taken over the funding of the registers has plans to develop a truly national English system by expanding to cover all areas of England starting in 2015. BINOCAR are actively working with PHE to try and ensure that the model of data collection used in the regional registers is used as a basis for the development of the national system to maintain high levels of ascertainment, accuracy and direct local clinical involvement

In this, the fourth report from CAROBB, we present the congenital anomalies data for births in 2005 to 2012 and for OXCAR data for Oxfordshire births in 1991 to 2012. This will be the last CAROBB report from the register as it currently functions. PHE plans for national expansion involve moving the existing congenital anomaly registers, including CAROBB, into PHE and expanding the use of notifications from routine sources of data provided electronically following the model of the national cancer registry system. The active engagement between PHE and BINOCAR over the past 18 months will hopefully ensure that this move does not compromise future case ascertainment or data quality.

On the cusp of this major change to the functioning of CAROBB and the other registers it seems timely to reflect on the achievements of CAROBB. Led by Tricia Boyd and supported by Cath Rounding, Kay Randall, Jane Forrester-Barker and in the past Yvonne Kenworthy and Charlotte White, data from CAROBB and OXCAR have: led to 78 peer reviewed publications including three papers in The Lancet and five in the British Medical Journal; contributed to 148 separate projects ranging from local audits to national research programmes and the European collection of congenital anomaly data led by EUROCAT; and countless local presentations supporting local service delivery, most importantly in the form of data to support counselling of parents and prospective parents.

Providing information for counselling of parents and prospective parents has been a central focus of the work of CAROBB and BINOCAR and it will be incumbent upon those responsible for the new national congenital anomaly system to ensure that parents and prospective parents remain a central focus of the new system which they are establishing.

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Part 1 - Introduction and Summary

Introduction

This report provides data on prenatally suspected and postnatally confirmed congenital anomalies notified to the Congenital Anomaly Register for Oxfordshire, Berkshire and Buckinghamshire (CAROBB) for births occurring in the eight years from 2005 – 2012. It also provides 24 years of data (1991 - 2012) from within Oxfordshire (OXCAR) (Appendix 1).

In April 2003 the Department of Health awarded funding for the expansion and development of the Oxford Congenital Anomaly Register (OXCAR), for research purposes. A new population-based register, covering the three counties which made up the Thames Valley was formed, called the Congenital Anomaly Register for Oxfordshire, Berkshire and Buckinghamshire (CAROBB). CAROBB is based at the National Perinatal Epidemiology Unit (NPEU), University of Oxford. This is the fourth full report from CAROBB and provides population based information on congenital anomalies affecting births between 2005 and 2012 to mothers resident in the three counties at the time of delivery.

The National Congenital Anomaly System (NCAS), set up in 1964 in response to the Thalidomide disaster, was responsible for surveillance of congenital anomalies in England and Wales. This ceased to function in 2010 and the British Isles Network of Congenital Anomaly Registers (BINOCAR *www.binocar.org*), now funded by Public Health England, provides surveillance in England and Wales in the areas covered by BINOCAR registries. CAROBB is now the only source of data for surveillance of congenital anomalies in the three counties of Oxfordshire, Berkshire and Buckinghamshire. Anonymised data for surveillance purposes and research are also sent to the European Congenital Anomaly Surveillance System (EUROCAT, *www.eurocat.ulster.ac.uk*).

Since the last report we are pleased to have appointed Kay Randall as clinical co-ordinator to CAROBB. Kay is also a specialist midwife with the Oxford Fetal Medicine Unit and works with CAROBB and Fetal Medicine to the mutual benefit of both organisations. Kay is in regular contact with the relevant clinical areas across the region and is exploring with the clinical staff ways of improving our case ascertainment. Please contact us if you have any suggestions regarding improvement of ascertainment or if you are interested in using the data for audit or research purposes. For details of projects that CAROBB is involved with, publications to which CAROBB has contributed, ethical approval for CAROBB and how to request data, please see Appendices 3 - 5.

The principal objectives of CAROBB

- Provide data for research on the aetiology and natural history of congenital anomalies to enable better advice based on accurate information to be given to parents and prospective parents.

- Enable the evaluation and monitoring of new invasive and non-invasive prenatal diagnostic tests and screening programmes.

- Provide data to support health care policies and planning.

- Provide data to investigate clusters of abnormalities and putative teratogens by the monitoring of rates over time and of population trends such as maternal age, ethnicity, and health inequalities.

- Improve ascertainment to BINOCAR and to EUROCAT.

The population studied for this report

- This report has information on congenital anomalies suspected and/or confirmed in fetuses / babies born to mothers resident in Oxfordshire, Berkshire and Buckinghamshire at the time of delivery.

- Data are provided on cases notified to CAROBB by February 2014 and with a date of birth/ delivery 2005-2012 inclusive. For this report a 'case' is a birth with a suspected and / or confirmed congenital anomaly notified to CAROBB. The term 'birth' (unless otherwise stated) is used to cover all pregnancies (from 10 weeks' gestation) ending in live birth, stillbirth, and termination of pregnancy for fetal anomaly (TOPFA). Late miscarriages (20^{+0} - 23^{+6} weeks' gestation) are also included.

These inclusion criteria are the same as those used by BINOCAR and EUROCAT and have been adopted for the first time in this report so CAROBB data may be directly compared with UK and European data. The difference between the inclusion criteria for this report and previous CAROBB reports is that miscarriages with a gestation of less than 20 weeks' were included in previous reports and they are excluded from this report. The other inclusion criteria of live and stillbirths and TOPFAs (from 10 weeks' gestation) remain the same as in previous reports because they mirror the inclusion criteria of UK and European data.

- Denominator data are provided by the Office for National Statistics and include only live births and stillbirths of 24 weeks' gestation or more. There were 240,687 total births in the CAROBB region between 2005 and 2012.

- The proportions of births with congenital anomalies are given as a percentage of total births or as a rate per 10,000 total births. This is a change from previous CAROBB reports, where rates per 1,000 total births were reported, and has been implemented to allow for direct comparison with UK and European data.

The report gives data on anomalies, their rate and, where appropriate, their prenatal detection, in Oxfordshire, Berkshire and Buckinghamshire. Information on cases for the hospital at which the mother booked for delivery can be provided and presented for individual hospitals.

Definition and coding of congenital anomalies

The definition of congenital anomaly, used by CAROBB is 'a structural or functional anomaly, presumed to be of prenatal origin'. All anomalies present at birth or diagnosed after birth are recorded. Prenatally suspected anomalies including ultrasound 'soft markers' (normal variants) are also recorded including those occurring in cases subsequently confirmed to be structurally normal babies. In line with other

UK and European registers each anomaly is coded using the ICD10 classification with the British Paediatric Association (BPA) extensions where appropriate. Certain minor congenital anomalies are excluded by CAROBB, also in line with other UK and European registers. A list of these exclusions is presented in Appendix 7.

Summary of findings

- From January 2005 to December 2012 there were 4985 births with a confirmed congenital anomaly (2.1% of all births), to mothers resident in Oxfordshire, Berkshire and Buckinghamshire, notified to CAROBB.

- More male than female births were affected by a congenital anomaly; male:female 1.3:1

- In 60% of births there was prenatal suspicion of a congenital anomaly.

- Overall 1,430 births (29% of all births notified with a congenital anomaly) resulted in terminations of pregnancy for fetal anomaly (TOPFA).

- There were 655 births with Down's syndrome; 404 (62%) were prenatally diagnosed. A high risk first trimester screening test result was the most common means of prenatal diagnosis. Not all mothers, with a positive Down's syndrome screening test or suspicion on ultrasound scan, underwent a diagnostic test. If all women with a prenatal suspicion had undergone karyotyping, the prenatal detection rate would have been 72%.

- There is known to be some under ascertainment of postnatally diagnosed anomalies to CAROBB, particularly anomalies diagnosed after the mother has been discharged from the maternity hospital and those not requiring surgery under the age of one year. However, ascertainment has gradually improved over time and for 2012, is similar to other UK congenital anomaly registers for postnatally diagnosed anomalies. Births to mothers resident in the CAROBB area but delivering outside the CAROBB area (e.g. in London) may not at present be notified to CAROBB.

Table 1 summarises the prenatal detection rates and prevalence per 10,000 births for selected congenital anomalies which form part of the fetal anomaly screening programme (FASP) overseen by the National Screening Committee (*www.fetalanomaly.screening.nhs.uk*). The prenatal detection figures cannot be directly compared with the FASP targets for prenatal detection because these CAROBB figures are for isolated anomalies whereas the FASP figures include all instances of the anomaly, for example where a syndrome or chromosome anomaly is also present. Individual hospitals monitor their performance against these targets and a national overview from BINOCAR registries may be found in the BINOCAR Annual Report (*www.binocar.org/Publications/Reports*).

Table 1: Prenatal detection of specific congenital anomalies in Oxfordshire, Berkshire and Buckinghamshire, 2005 – 2012

Anomaly	Test performed	Number of pregnancies notified with prenatal suspicion of anomaly[1]	Number of cases notified with anomaly confirmed at birth	Prevalence per 10,000 total births	Prenatal detection rate
Isolated neural tube defects	Ultrasound Scanning +/- MS AFP[2]	221	232	9.6	95%
Isolated cardiac anomaly	Ultrasound scanning	279	851	35.4[3]	33%
Isolated cleft lip +/- palate	Ultrasound scanning	114	160	6.7	70%
Down's Syndrome	Screening tests, ultrasound scanning, karyotyping	404[4]	655	27.2	62%[2]
Isolated diaphragmatic hernia	Ultrasound scanning	37	51	2.1	73%
Isolated exomphalos	Ultrasound scanning +/- MS AFP	38	41	1.7	93%
Isolated gastroschisis	Ultrasound scanning +/- MS AFP	66	66	2.7	100%

[1] Not including false positive diagnoses

[2] Maternal Serum Alpha Feto Protein (MS AFP) screening is not part of combined screening so has not been routinely performed since combined screening was implemented in 2009

[3] Low prevalence most likely due to low ascertainment of cases diagnosed after birth

[4] Only includes those karyotyped prenatally – it excludes those with high risk screening result or suspicion on scan who were not karyotyped

Part 2 - Routine statistics, area covered by CAROBB and outcome of pregnancies

Population and area covered

There were over two million people resident in Oxfordshire, Berkshire and Buckinghamshire between 2005 and 2012, with Berkshire having the largest and Oxfordshire the smallest population. Both the population and total births have increased in the eight year period from 2005 to 2012. The figures in Tables 2 and 3 are supplied by the Office for National Statistics.

Table 2: Total population covered – mid-year estimates by county and year of birth

	Oxfordshire	Berkshire	Buckinghamshire	Total
2005	627,500	808,800	704,700	**2,141,000**
2006	629,600	817,000	710,100	**2,156,700**
2007	632,300	828,800	717,600	**2,178.700**
2008	635,500	841,800	724,400	**2,201,700**
2009	640,300	854,000	731,400	**2,225,700**
2010	648,700	865,100	739,600	**2,253,400**
2011	654,800	863,900	756,500	**2,275,200**
2012	660,800	871,000	763,800	**2,295,600**

Table 3: Total births (live and stillbirths), by county and year of birth

	Oxfordshire	Berkshire	Buckinghamshire	Total
2005	7616	10920	8762	**27298**
2006	8028	11391	9276	**28695**
2007	8184	12130	9402	**29716**
2008	8347	12490	9893	**30730**
2009	8175	12443	9774	**30392**
2010	8485	12770	10066	**31321**
2011	8537	12694	10125	**31356**
2012	8248	12798	10133	**31179**
Total	65620	97636	77431	**240687**

Figure 1: **Map of the CAROBB area, Oxfordshire, Berkshire and Buckinghamshire**

Total births with congenital anomalies, pre and postnatal diagnosis

There appears to be a lower rate of congenital anomalies in Berkshire (Table 4). This almost certainly does not reflect a lower prevalence but is probably due to lower ascertainment, partly because more babies with congenital anomalies born to mothers resident in Berkshire are delivered outside the CAROBB area (e.g. London) and although are eligible to be notified to CAROBB it is likely that this does not occur for all cases. The congenital anomaly rate in Oxfordshire appears higher and this is probably due to the fact that there are much longer established practices in place for ascertaining cases because the Oxford congenital anomaly register (OXCAR) was established in 1991.

Table 4: Number of cases (% of all births) with congenital anomaly[1], by year of birth

	Oxfordshire n (%)	Berkshire n (%)	Buckinghamshire n (%)	Total n (%)
2005	158 (2.1%)	158 (1.4%)	153 (1.7%)	469 (1.7%)
2006	196 (2.4%)	170 (1.5%)	177 (1.9%)	543 (1.9%)
2007	225 (2.7%)	169 (1.4%)	181 (1.9%)	575 (1.9%)
2008	225 (2.7%)	188 (1.5%)	190 (1.9%)	603 (2.0%)
2009	274 (3.4%)	222 (1.8%)	195 (2.0%)	691 (2.3%)
2010	270 (3.2%)	223 (1.7%)	217 (2.2%)	710 (2.3%)
2011	271 (3.2%)	208 (1.6%)	227 (2.2%)	706 (2.3%)
2012	258 (3.1%)	203 (1.6%)	227 (2.2%)	688 (2.2%)
Total	1877 (2.9%)	1541 (1.6%)	1567 (2.0%)	4985 (2.1%)

[1]including termination of pregnancy for fetal anomaly

Table 5 shows the number and percentage of cases prenatally and postnatally diagnosed. The percentage of cases with a prenatal suspicion of anomaly which were apparently normal at birth has decreased over time. Most of these cases in the early years were associated with ultrasound 'soft markers' (normal variants) such as choroid plexus cysts and the decrease probably represents changes in practice, following local protocols and recommendations from the Fetal Anomaly Screening Programme (*http://fetalanomaly.screening.nhs.uk/programmestatements*) for the reporting of these normal variants. See Appendix 1, Table 2A for related data from Oxfordshire, 1991-2012.

The apparent early increase in the rate of anomalies overall is almost certainly due to improved ascertainment over time as the wider CAROBB register became established.

Table 5: Total births and case notifications; number prenatally suspected with and without congenital anomaly at birth and total births with anomalies, by year of birth

Year	2005	2006	2007	2008	2009	2010	2011	2012	Total
Total births	27298	28695	29716	30730	30392	31321	31356	31179	240687
Total cases notified to CAROBB[1]	656	790	790	821	943	962	921	888	6771
Number of cases notified prenatally including normal variants (ultrasound soft markers) (% of total notified)	472 (72%)	588 (74%)	514 (65%)	514 (63%)	552 (59%)	535 (56%)	476 (52%)	473 (53%)	4124 (61%)
Number of cases notified prenatally with anomaly confirmed at birth (% of total cases with anomaly)	303 (65%)	374 (69%)	334 (58%)	354 (59%)	409 (60%)	407 (58%)	378 (54%)	386 (56%)	2945 (60%)
Number of cases notified prenatally & considered normal at birth (% of total notified prenatally)	126 (27%)	171 (29%)	142 (28%)	116 (23%)	105 (19%)	87 (16%)	73 (15%)	61 (13%)	881 (21%)
Total cases with anomaly at birth, miscarriage or TOPFA; excludes those notified prenatally and lost to follow up (% of total births)	469 (1.7%)	543 (1.9%)	575 (1.9%)	603 (2.0%)	691 (2.3%)	710 (2.3%)	706 (2.3%)	688 (2.2%)	4985 (2.1%)

[1]Including prenatally suspected cases without an anomaly present at birth.

Outcome of pregnancy

The proportion of notified anomalies resulting in termination of pregnancy for fetal anomaly (TOPFA) is lower in Oxfordshire than Berkshire and Buckinghamshire (Table 6; Figure 2)

While the TOPFA rate (based on percentages) appears to be lower in Oxfordshire than Berkshire and Buckinghamshire, this however, is not the case. The rate of TOPFA per 10,000 births shows Berkshire has the lowest TOPFA rate at 54.1 per 10,000 with Oxfordshire and Berkshire having rates of 62.8 and 63.3 respectively (Figure 3). The apparent disparity in outcomes between the three counties is most likely to be explained by ascertainment. Oxfordshire has very good ascertainment of congenital anomalies for all outcomes including those diagnosed after birth because CAROBB has access to detailed information from hospital systems from the local health care trust. This is not the case in the other two counties. The lower TOPFA rate per 10,000 births in Berkshire might also be influenced by the location of terminations. We are aware that women resident in Berkshire are referred, or choose to deliver in other areas, including London. This might also be the case for TOPFAs. As there is no currently functioning congenital anomaly register in the London area, and we tend to have a lower rate of case ascertainment when such referrals occur.

Table 6: Outcome of pregnancy of cases notified with congenital anomaly confirmed at birth from 2005 to 2012, by county (n = 4985)

	Oxfordshire n (%)[1]	Berkshire n (%) [1]	Buckinghamshire n (%) [1]	Total n (%) [1]
Live birth	1376 (73%)	925 (59%)	950 (62%)	**3251 (65%)**
Neonatal death	45 (2%)	54 (3%)	42 (3%)	**141 (3%)**
Stillbirth	34 (2%)	42 (3%)	47 (3%)	**123 (2%)**
Miscarriage (20-23^{+6} weeks')	10 (1%)	18 (1%)	12 (1%)	**40 (1%)**
Termination for fetal anomaly	412 (22%)	528 (34%)	490 (32%)	**1430 (29%)**
Total notified	1877 (100%)	1567 (100%)	1541 (101%)*	**4985 (100%)**

*Percentages may not add up to 100% because of rounding errors

Figure 2: **Outcome of pregnancy (percentage of live births, stillbirths, neonatal deaths, miscarriages or terminations of pregnancy) with congenital anomaly, 2005-2012, by county, n = 4985**

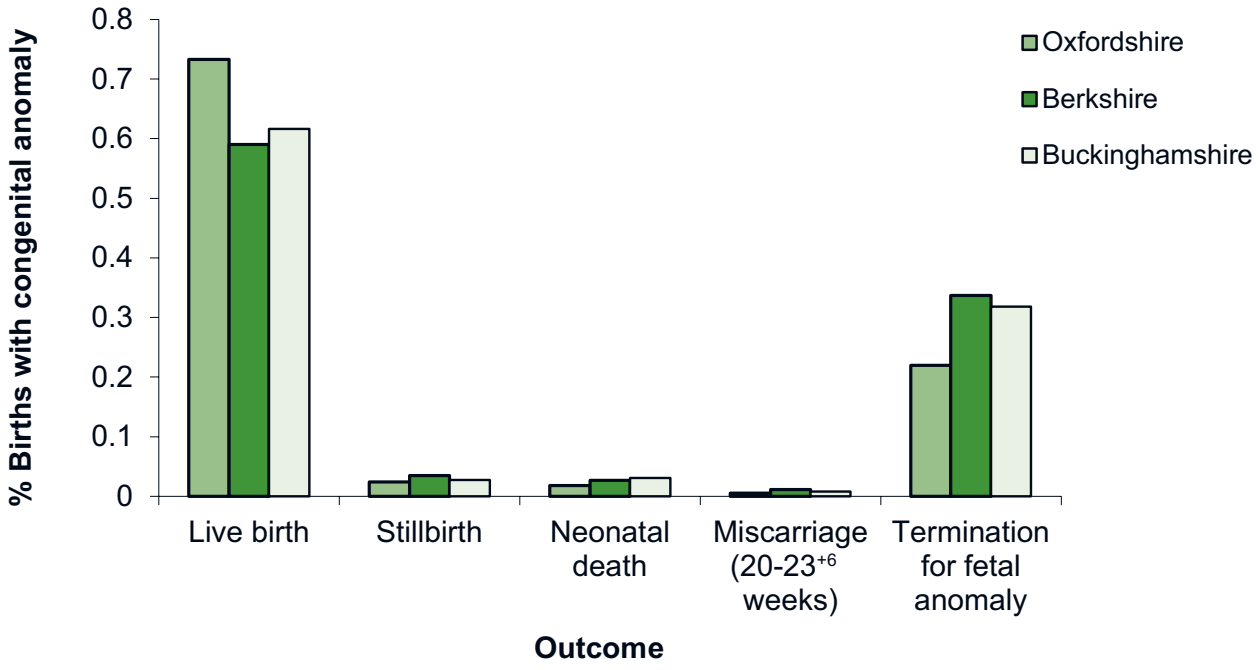
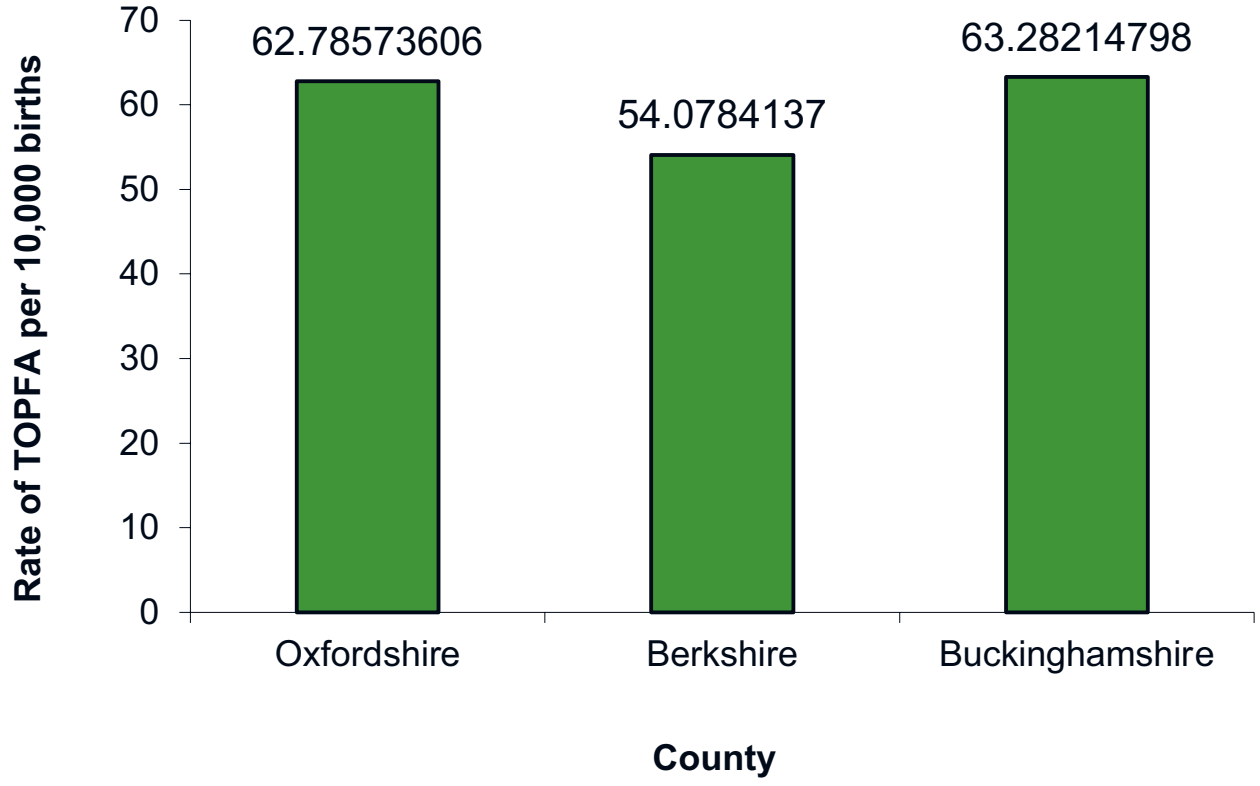


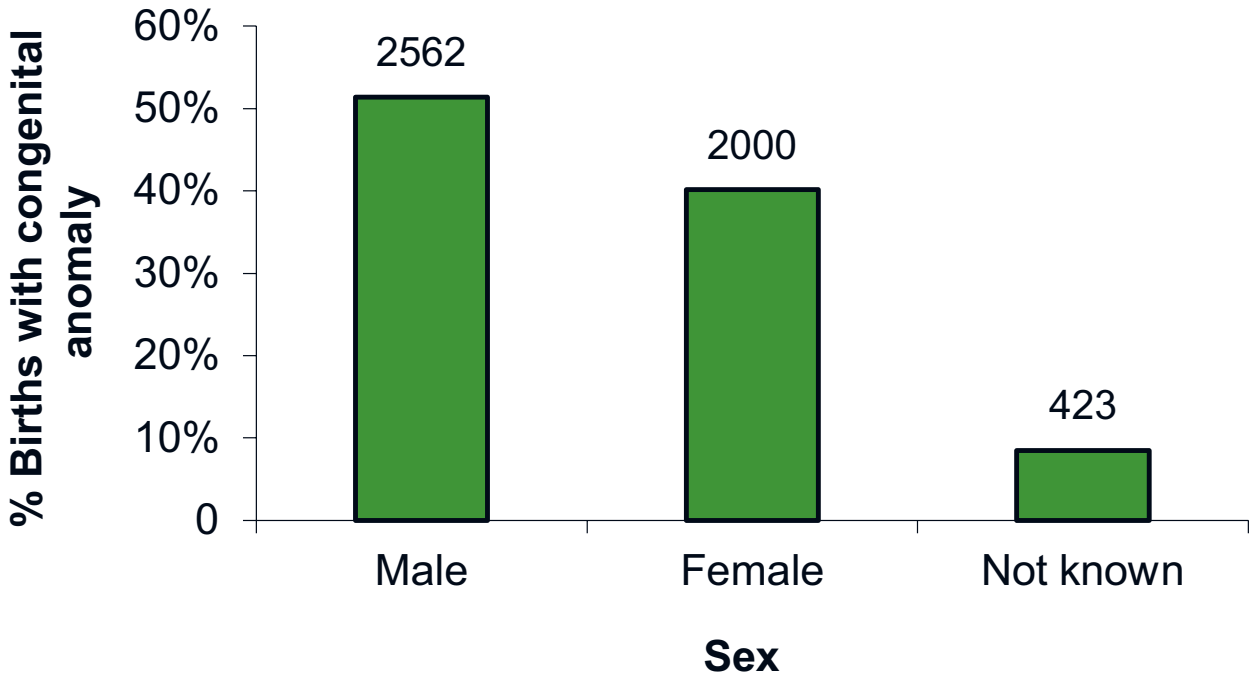
Figure 3: **Rates of termination of pregnancy for fetal anomaly (TOPFA) per 10,000 births, by county, 2005-2012**



Sex ratio of births with congenital anomalies

The sex ratio for births with a congenital anomaly in the CAROBB area, in 2005-2012 is male:female 1.3:1 (Figure 4). This is in contrast to the background sex ratio for all births in the UK which is 1.1:1 male to female births (data source: Department of Health).

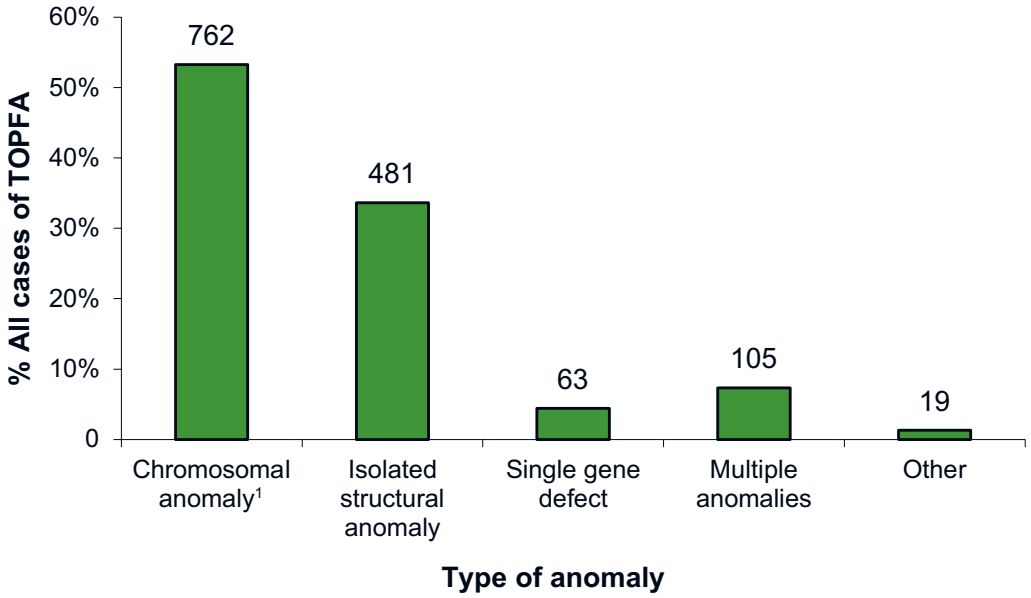
Figure 4: Percentage and number of male and female births with congenital anomaly



Termination of pregnancy for fetal anomaly (TOPFA), 2005 - 2012

Figure 5a shows the percentage and number of cases resulting in TOPFA by type of anomaly. Chromosome anomalies accounted for 53% of cases terminated, isolated structural anomalies for 34%, single gene defects for 4%, 7% were for non-chromosomal multiple structural anomalies and 1% were for other anomalies, including undiagnosed syndromes, and infections, such as cytomegalovirus (CMV). Of the chromosome anomalies 50% had Down's syndrome (trisomy 21) (Figure 5b). Neural tube defects (including anencephaly) were the most common isolated structural defect resulting in TOPFA (Figure 5c). Overall, 94% of TOPFAs were performed before 24 weeks' gestation (Figure 5d).

Figure 5a: Percentage and number of cases resulting in TOPFA by type of anomaly, n = 1430



[1]Please note that the chromosomal anomaly total includes 22Q deletion, whereas the EUROCAT chromosomal anomaly figures list this separately (Table 7).

Figure 5b: TOPFA, chromosome anomalies by type, n = 762

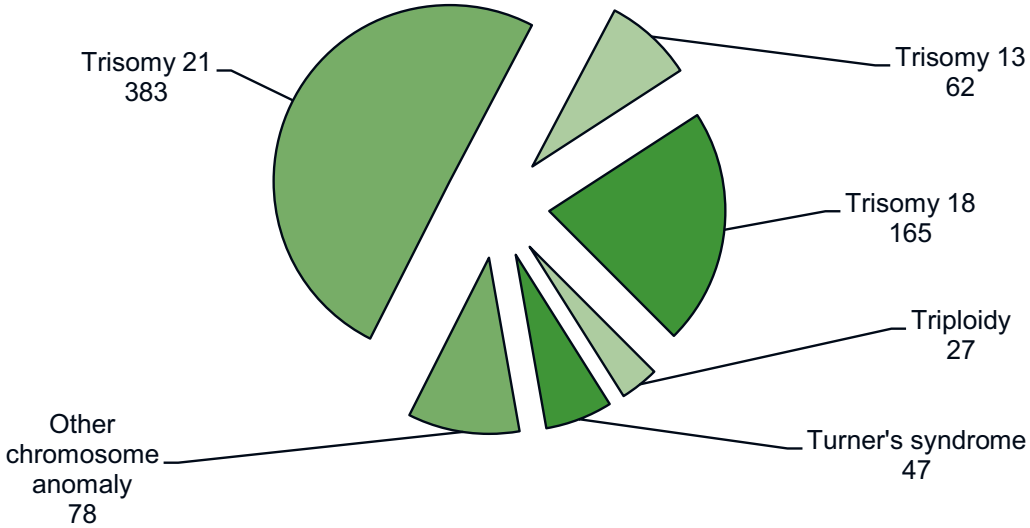


Figure 5c: **TOPFA, isolated anomalies by type, n = 481**

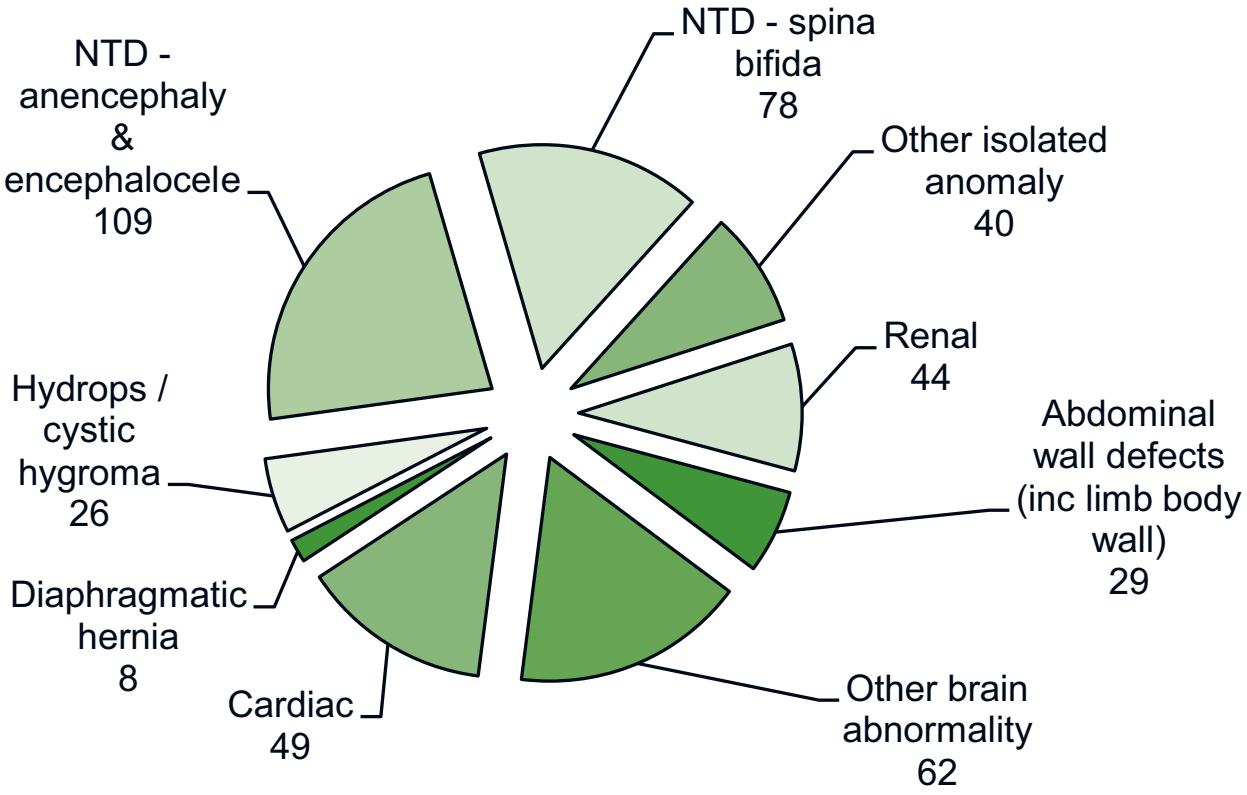
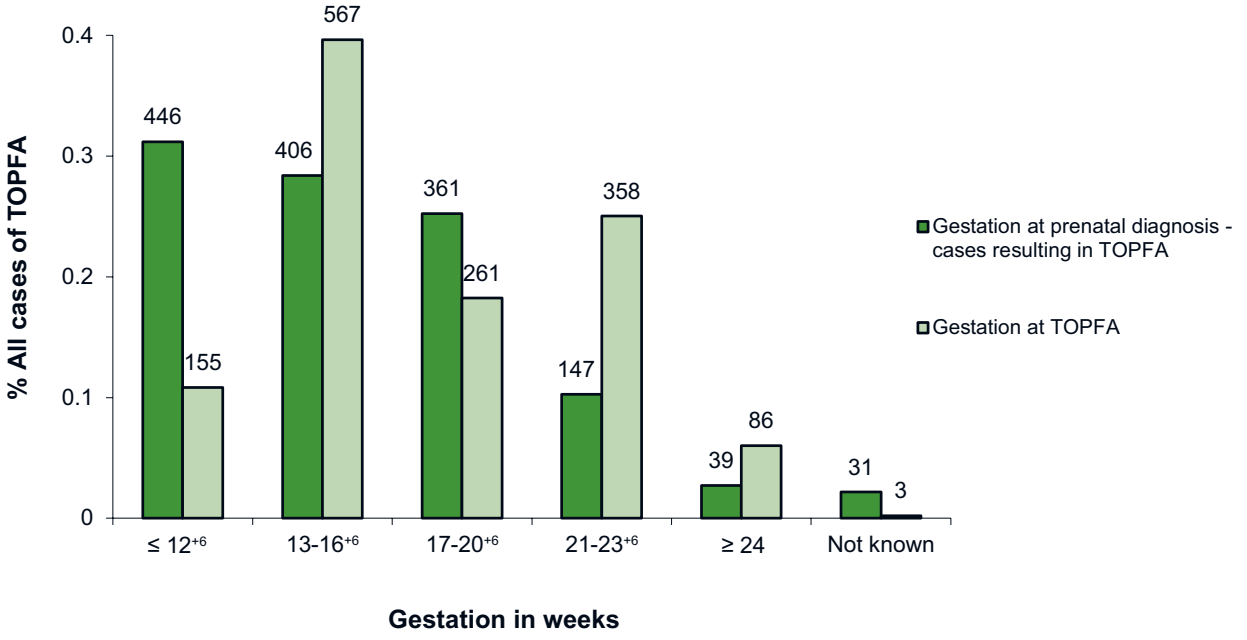


Figure 5d: **Percentage and number of cases resulting in termination of pregnancy for fetal anomaly (TOPFA), by gestation period at prenatal diagnosis and at termination, n = 1430**



Part 3 - Rates of congenital anomalies

Table 7 shows the overall numbers and rates of congenital anomalies for births within the CAROBB area using subgroups of anomalies defined by EUROCAT (*www.eurocat-network.eu/content/EUROCAT-Guide-1.4-Section-3.3.pdf*). The table shows information about individual anomalies, meaning that a case with multiple anomalies will appear more than once in the table, with an inclusion for each anomaly present. This means that the outcome of pregnancy may not be directly attributed to the anomaly listed because other anomalies may also have been present.

Table 7: Number and rates of anomalies per 10,000 births delivered in CAROBB area, year of birth 2005 - 2012 (Total births: 240,687)

Diagnostic Category	ICD 10 code	Live births, stillbirths (n)	Fetal deaths >=20 weeks' gestation (n)	Termination of pregnancy (n)	Including chromosomal anomalies rate per 10,000 births		Excluding chromosomal anomalies rate per 10,000 births	
					Live & still births, fetal deaths and termination of pregnancy^ (n)	Live & still births, fetal deaths and termination of pregnancy (rate (95% CI))	Live & still births, fetal deaths and termination of pregnancy (n)	Live & still births, fetal deaths and termination of pregnancy (rate (95% CI))
All births with congenital anomalies		**3392**	**163**	**1430**	**4985**	**207.12 (201.41 - 212.95)**	**3762**	**156.30 (151.35 - 161.38)**
The list below is a list of all anomalies, not individual births. Some births will have more than one anomaly present. An anomaly listed as resulting in termination of pregnancy may be part of a multiple anomaly case. * = Data suppressed because of small numbers, to protect the confidentiality of individuals								
Nervous system anomalies	Q00 – Q07	184	28	384	**596**	**24.76 (22.82 - 26.83)**	528	21.94 (20.11 - 23.89)
Neural Tube Defects		41	7	229	**277**	**11.51 (10.19 - 12.95)**	259	10.76 (9.49 - 12.15)
Anencephalus, and similar	Q00 – Q01	6	5	112	**123**	**5.11 (4.25 - 6.10)**	117	4.86 (4.02 - 5.83)
Encephalocoele	Q00 – Q01	*	*	22	**26**	**1.08 (0.71 - 1.58)**	24	1.00 (0.64 - 1.48)
Spina Bifida	Q05	*	*	95	**128**	**5.32 (4.44 - 6.32)**	118	4.90 (4.06 - 5.87)
Hydrocephaly	Q03	52	13	83	**148**	**6.15 (5.20 - 7.22)**	129	5.36 (4.48 - 6.37)
Congenital heart anomalies	Q20 - Q26	1019	34	175	**1228**	**51.02 (48.21 - 53.96)**	1046	43.46 (40.87 - 46.17)
Severe CHD†	Q200, Q203-4, Q212-3, Q220, Q230, Q234. Q224-6, Q251, Q262	337	13	95	**445**	**18.49 (16.81 - 20.29)**	379	15.75 (14.20 - 17.41)
Oro-facial clefts	Q35 - Q37	357	5	34	**396**	**16.45 (14.87 - 18.16)**	363	15.08 (13.57 - 16.72)

Diagnostic Category	ICD 10 code	Live births, stillbirths (n)	Fetal deaths >=20 weeks' gestation (n)	Termination of pregnancy (n)	Including chromosomal anomalies rate per 10,000 births		Excluding chromosomal anomalies rate per 10,000 births	
					Live & still births, fetal deaths and termination of pregnancy^ (n)	Live & still births, fetal deaths and termination of pregnancy (rate (95% CI))	Live & still births, fetal deaths and termination of pregnancy (n)	Live & still births, fetal deaths and termination of pregnancy (rate (95% CI))
Digestive system anomalies	Q38 – Q39, Q402, Q408-09, Q41 –Q45	282	11	60	**353**	**14.67 (13.18 - 16.28)**	308	12.80 (11.41 - 14.31)
Oesophageal atresia with or without tracheo-oesophagal fistula	Q390 - Q3914	48	*	*	**61**	**2.53 (1.94 - 3.26)**	54	2.24 (1.69 - 2.93)
Duodenal atresia or stenosis	Q410	**25**	*	*	**30**	**1.25 (0.84 - 1.78)**	20	0.83 (0.51 - 1.28)
Hirschspung's disease	Q431	37	*	*	**37**	**1.54 (1.08 - 2.12)**	32	1.33 (0.91 - 1.88)
Genital anomalies	Q50 – Q52, Q54 – Q56	280	*	*	**307**	**12.76 (11.37 - 14.26)**	295	12.26 (10.90 - 13.74)
Urinary anomalies	Q60 - Q64, Q794	449	15	105	**569**	**23.64 (21.74 - 25.66)**	545	22.64 (20.78 - 24.63)
Limb anomalies		495	17	95	**607**	**25.22 (23.25 - 27.31)**	564	23.43 (21.54 - 25.45)
Reduction defects	Q71 – Q73	58	*	*	**92**	**3.82 (3.08 - 4.69)**	88	3.66 (2.93 - 4.50)
Club foot – talipes equinovarus	Q660	164	6	42	**212**	**8.81 (7.66 - 10.08)**	196	8.14 (7.04 - 9.37)
Skeletal dysplasias	Q77 – Q78	*	*	39	65	2.70 (2.08 - 3.44)	62	2.58 (1.98 - 3.30)
Craniosynostosis	Q750-1, Q754-9	91	*	*	94	3.91 (3.16 - 4.78)	90	3.74 (3.01 - 4.60)

Diagnostic Category	ICD 10 code	Live births, stillbirths (n)	Fetal deaths >=20 weeks' gestation (n)	Termination of pregnancy (n)	Including chromosomal anomalies rate per 10,000 births		Excluding chromosomal anomalies rate per 10,000 births	
					Live & still births, fetal deaths and termination of pregnancy^ (n)	Live & still births, fetal deaths and termination of pregnancy (rate (95% CI))	Live & still births, fetal deaths and termination of pregnancy (n)	Live & still births, fetal deaths and termination of pregnancy (rate (95% CI))
Abdominal wall defects		100	9	98	**207**	**8.60 (7.47 - 9.85)**	154	6.40 (5.43 - 7.49)
Diaphragmatic Hernia	Q790	51	*	*	**71**	**2.95 (2.30 - 3.72)**	60	2.49 (1.90 - 3.21)
Gastroschisis	Q793	64	*	*	**68**	**2.83 (2.19 - 3.58)**	68	2.83 (2.19 - 3.58)
Omphalocele	Q792	35	8	83	**126**	**5.24 (4.36 - 6.23)**	76	3.16 (2.49 - 3.95)
Genetic syndromes & microdeletions	Q87, Q936, D821	116	6	39	**161**	**6.69 (5.70 - 7.81)**	155	6.44 (5.47 - 7.54)
Chromosomal anomalies	Q90 – Q93, Q96 – Q99	408	67	748	**1223**	**50.81 (48.01 - 53.74)**	N/A	N/A
Down's Syndrome (Trisomy 21)	Q90	249	23	383	**655**	**27.21 (25.17 - 29.38)**	N/A	N/A
Patau syndrome (Trisomy 13)	Q914 – Q917	13	7	62	**82**	**3.41 (2.71 - 4.23)**	N/A	N/A
Edward syndrome (Trisomy 18)	Q910 – Q913	23	14	165	**202**	**8.39 (7.28 - 9.63)**	N/A	N/A
Turner's syndrome	Q96	19	10	47	**76**	**3.16 (2.49 - 3.95)**	N/A	N/A

†Severe cardiac anomalies subgroup, as defined by EUROCAT (Boyd PA, Haeusler M, Barisic I, Loane M, Garne E, Dolk H. Paper 1: The EUROCAT network--organization and processes. Birth Defects Res A Clin Mol Teratol. 2011;91 Suppl 1:S2-15).

Part 4 - Information about specific anomalies

1. Open Neural Tube Defects (NTD), year of birth 2005 -2012

Anencephaly: **Definition:** Total or partial absence of the cranial vault, covering skin and brain tissue.

Encephalocoele: **Definition:** Herniation of the brain and/or meninges through a defect in the skull.

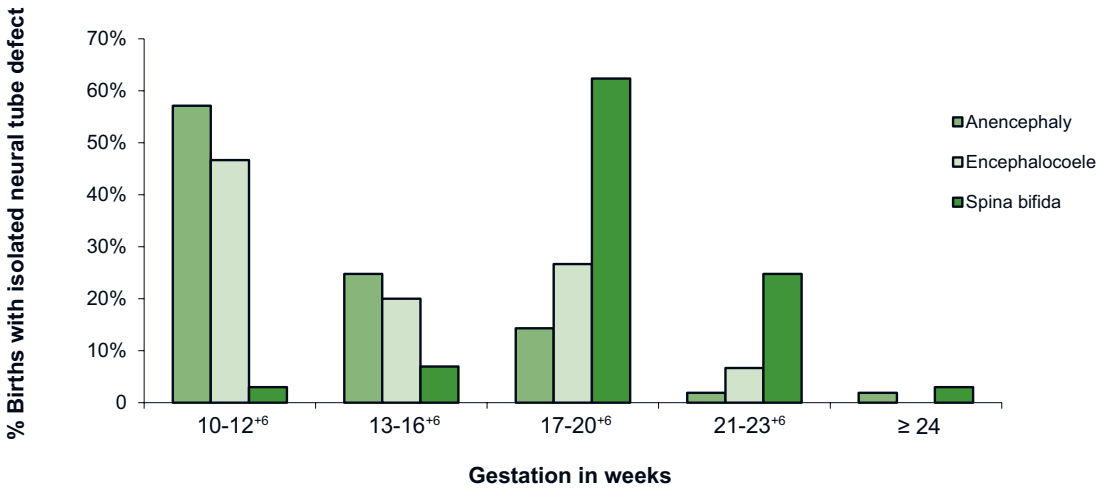
Spina bifida: **Definition:** Non-closure of the spine resulting in herniation or exposure of the spinal cord and /or meninges. Hydrocephaly may or may not be present.

Summary Information

Prenatal investigation:	Ultrasound scan +/- maternal serum alpha feto protein screening*
Rate: **Isolated and non-isolated neural tube defects** **Isolated neural tube defects**	 11.5 per 10,000 births n=277 9.6 per 10,000 births n = 232
Prenatal detection rate for isolated cases:	221/232 (95%)
ICD 10 codes:	Q00.0 (anencephaly); Q01 – Q01.9 (encephalocoele); Q05 – Q05.9 (spina bifida)

*Maternal Serum Alpha Feto Protein (MS AFP) screening is not part of combined screening so has not been routinely performed since combined screening was implemented in 2009

Figure 6: **Gestation at prenatal diagnosis of isolated neural tube defects - percentage of each type (anencephaly, encephalocele, spina bifida) diagnosed by gestational periods**



2. Cardiac Anomalies, year of birth 2005 -2012

Definition: Group of anomalies with abnormal structure of the heart.

Summary information

All Cardiac anomalies

Prenatal investigation:	Ultrasound scan
Rate: **Isolated and non-isolated structural cardiac anomalies** **Isolated structural cardiac anomalies**	 51.0* per 10,000 n = 1228 35.4 per 10,000 n = 851
Prenatal detection rate of isolated cardiac cases:	279/851 (33%)
ICD 10 Codes Q20 - Q26.9	Q20 - Q26.9

*Expected rate 70-80 per 10,000 (Knowles R et al. Newborn screening for congenital heart defects: a systematic review and cost-effectiveness analysis. *Health Technol Assess* 2005;9(44),1-152)

It has always been recognised that there is under ascertainment by CAROBB of cardiac abnormalities, particularly those diagnosed after the mother has left the maternity unit. In the last two years there has been some improvement due to new outpatient sources of cases. Figure 7 illustrates the prenatal diagnosis rate for selected isolated cardiac anomalies and Figure 8 the prenatal diagnosis rate for all isolated cardiac anomalies in the eight year period, 2005 to 2012. The lower percentages from 2009 to 2012 most likely reflect the improvement in postnatal ascertainment rather than a reduction in the actual prenatal detection rate.

Figure 7: Selected isolated anomalies, number of cases and percentage prenatally diagnosed

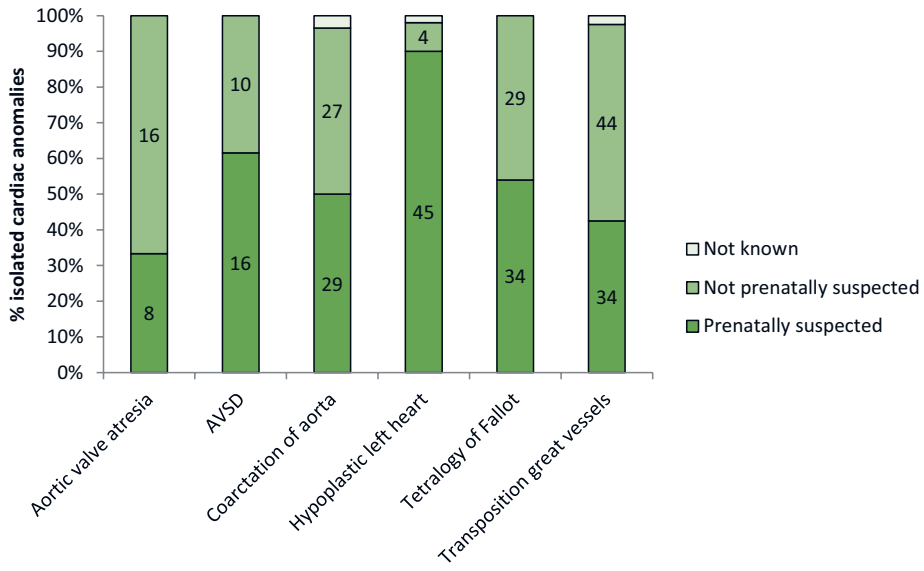


Figure 8: Isolated cardiac anomalies, percentage and number prenatally diagnosed, by year

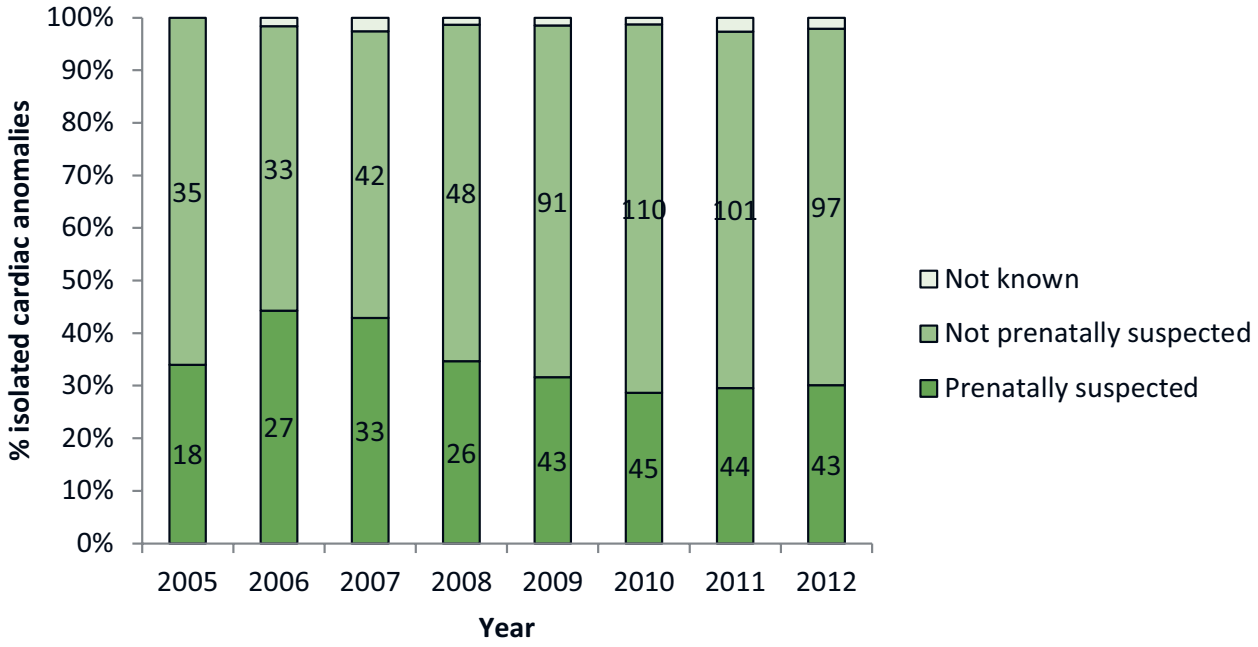
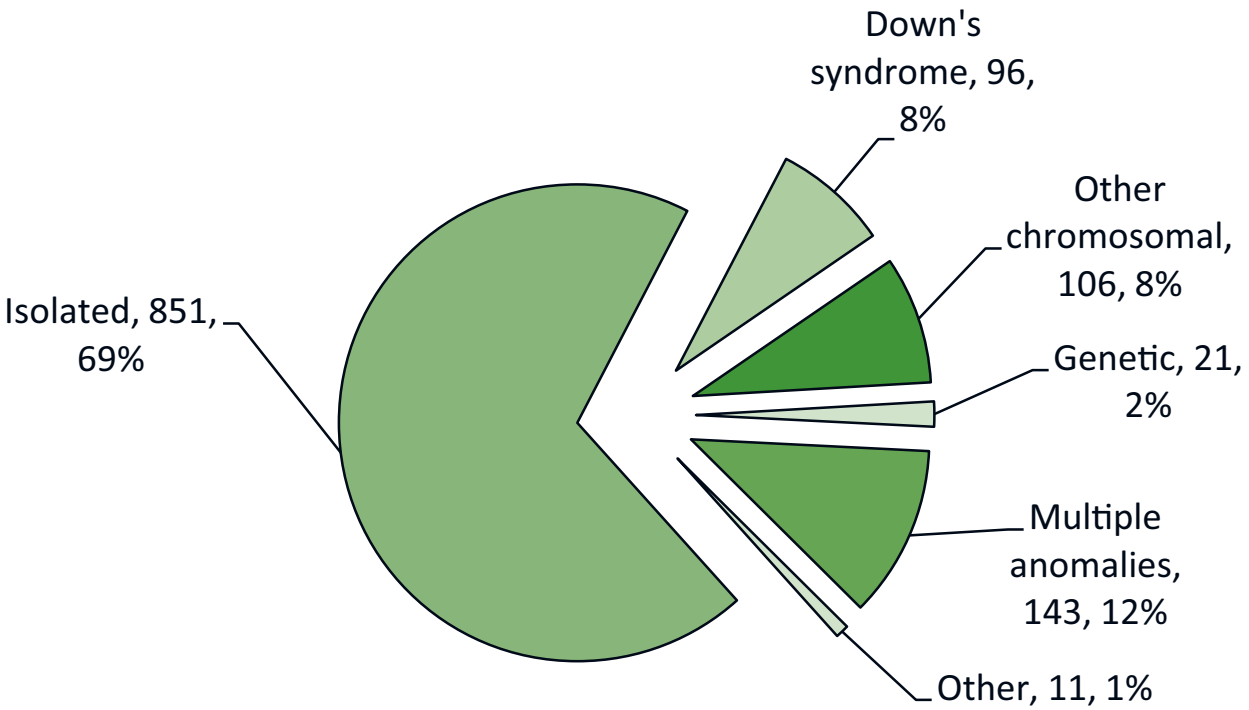


Figure 9 shows the proportion of cases with a cardiac anomaly which are isolated and the proportion which are associated with other conditions including chromosomes, genetic and multiple anomalies.

Figure 9: Number of births and percentage with a cardiac anomaly categorised by type, n=1228



3. Cleft Lip with or without Cleft Palate (Cleft lip +/- Palate), year of birth 2005 -2012

Cleft lip: **Definition:** Clefting of the upper lip without clefting of the alveolar ridge and palate.

Cleft lip and palate: **Definition:** Clefting of the upper lip with clefting of the alveolar ridge and palate.

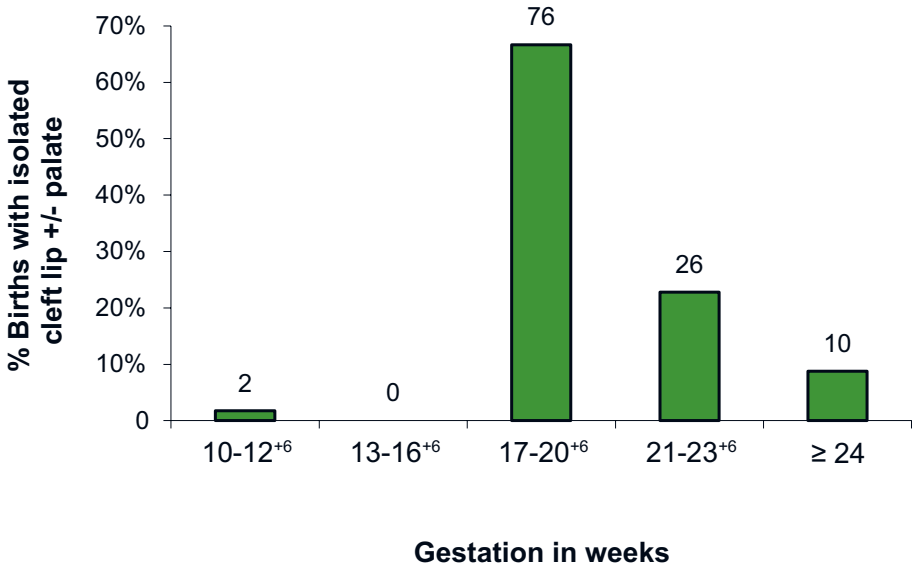
Summary Information

Prenatal investigation:	Ultrasound scan
Rate: **Isolated and non-isolated cleft lip +/- palate** **Isolated cleft lip +/- palate**	 8.6 / 10,000 n = 208 6.7 / 10,000 n = 160
Prenatal detection rate	114 / 160 (70%)
ICD 10 Codes Q36 – 37.9	Q36 – 37.9

We report the prenatal detection of cleft lip with or without cleft palate. It is not usually possible to visualise isolated cleft palate by ultrasound prenatally. Very minor clefts (forme fruste) have been excluded from this analysis.

There were 160 cases of isolated cleft lip +/- palate of which 114 (70%) were prenatally diagnosed. Of the 48 cases of non-isolated cleft lip +/- cleft palate, 20 (42%) were associated with chromosome anomalies.

Figure 10: **Percentage and number of births with prenatally diagnosed isolated cleft lip +/- palate diagnosed at different gestational age periods, n = 114**



4. Diaphragmatic Hernia, Exomphalos and Gastroschisis, year of birth 2005 -2012

a. Diaphragmatic hernia: **Definition:** Defect in the diaphragm resulting in herniation of the abdominal organs into the thorax.

b. Exomphalos: **Definition:** Herniation of the abdominal contents through the umbilical insertion and covered by a membrane which may or may not remain intact.

c. Gastroschisis: **Definition:** Visceral herniation through an abdominal wall defect lateral to an intact umbilical cord.

Summary information

	Diaphragmatic Hernia	Exomphalos	Gastroschisis
Prenatal Investigation	Ultrasound scan	Ultrasound scan +/- maternal serum AFP screening†	Ultrasound scan +/- maternal serum AFP screening†
Number of isolated cases	51*	41	66
Number of non-isolated and isolated cases	71 (eg chromosomal, cardiac and renal anomalies)	126 (eg Trisomy 18, Beckwith-Wiedemann syndrome)	68 (multiple anomalies)
Rate:			
Isolated cases	2.1 / 10,000	1.7 / 10,000	2.7 / 10,000
Isolated and non-isolated cases	2.9 /10,000	5.2 / 10,000	2.8 / 10,000
Prenatal detection rate for isolated cases	37/51* (73%)	38/41 (93%)	66/66 (100%)
ICD 10 Codes	Q79.0	Q79.2	Q79.3

* There were 2 additional cases where there was a suspicion of an anomaly but diaphragmatic hernia was not diagnosed

† Maternal Serum Alpha Feto Protein (MS AFP) screening is not part of combined screening so has not been routinely performed since combined screening was implemented in 2009

There was a high prenatal diagnosis rate for cases with isolated gastroschisis (100%) and for isolated exomphalos (93%). Overall for 73% of isolated diaphragmatic hernia cases there was a suspicion on scan prenatally.

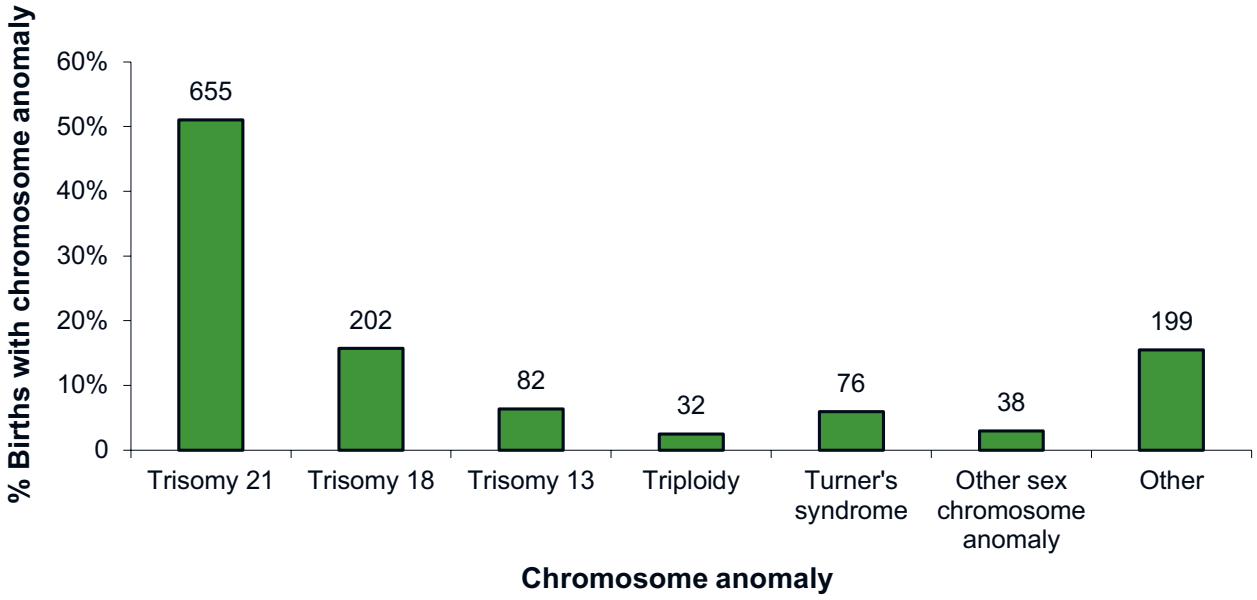
It is well recognised that gastroschisis is more common in babies born to younger mothers and that it is more likely to be an isolated lesion compared to both diaphragmatic hernia and exomphalos. All but two of the gastroschisis cases, 72% of diaphragmatic herniae and 33% of exomphalos had isolated lesions. The mean age (range) of mothers with babies with gastroschisis was 24 years (17-41 years) compared to 32 years (18-46 years) for isolated exomphalos and 31 years (19-38 years) for isolated diaphragmatic hernia.

5. Chromosome Anomalies, year of birth 2005 -2012

Figure 11 shows that Trisomy 21 makes up the largest proportion (51%) of chromosome anomaly cases, followed by Trisomy 18 (16%) and 'other' (15%).

There has been a slight increase in the proportion of 'other' chromosome anomalies (15% in this report compared to 11% in the previous CAROBB report). As Array Comparative Genomic Hybridisation (ARRAY CGH) is becoming established the proportion of 'other' chromosome anomalies is likely to increase further. The main benefit of ARRAY CGH is that it is able to detect much smaller genetic changes than was previously possible. It is currently used selectively for cases of prenatally suspected abnormality where the karyotype has been normal and postnatally for cases of developmental delay and congenital abnormality of uncertain cause. Over the past two years use of ARRAY CGH has increased both prenatally and postnatally.

Figure 11: All Chromosome anomalies, percentage of cases and number by chromosome type, n = 1284



6. Down's Syndrome (Trisomy 21)

Definition: Additional chromosome 21.

Summary information

Prenatal Investigation:	First and second trimester screening tests. Karyotyping performed because higher risk for Down's syndrome for one of the following reasons: positive family history, (previously, increased maternal age alone), translocation carrier, higher risk screening test or suspicion on ultrasound scan.
Rate: From 12 weeks' gestation	27.2 / 10,000 n = 655
Prenatal detection rate 2005-2012:	468/655 (72%) – some suspicion 404/655 (62%) – prenatally diagnosed
Prenatal detection rate 2009 – 2012*:	241/318 (76%) – some suspicion 206/318 (68%) – prenatally diagnosed
ICD 10 Codes	Q90 – Q90.9

* all hospitals offering first trimester combined screening for Down's syndrome

Over the past fifteen years there has been a move from offering pregnant women at higher risk of having a baby with Down's syndrome a prenatal diagnostic test, to a national programme for prenatal screening tests to be offered to all pregnant women.

In the CAROBB area there was a variety of screening tests for Down's syndrome in place in 2005 but by 2009 all NHS hospitals were offering first trimester combined screening, as recommended by the National Screening Committee Fetal Anomaly Screening Programme *www.fetalanomaly.screening.nhs.uk*. This is reflected by the increase in suspicion and detection of trisomy 21 since this new screening recommendation was implemented.

There were 655 cases with Down's syndrome between 2005 and 2012 inclusive. Three hundred and eighty nine (60%) of the 655 cases were karyotyped prenatally before 24 weeks' gestation, and 15 (2%) cases were karyotyped from 24 weeks' gestation; the remainder were not karyotyped. In 468/655 (72%) of cases there was some prenatal suspicion of abnormality either due to a higher risk screening test result or scan appearance although karyotyping was not performed in all cases. Figures 12a shows the percentage of Down's syndrome cases prenatally diagnosed, those with some prenatal suspicion and those with no suspicion prenatally, by year. Figure 12b shows the percentage of cases prenatally diagnosed at different gestational ages, by year. These show a tendency towards a higher prenatal diagnosis rate and earlier gestation at diagnosis over time.

Fig 12a: **Percentage of Down's Syndrome cases prenatally diagnosed, percentage with some prenatal suspicion, and percentage with no prenatal suspicion, by year (n=655)**

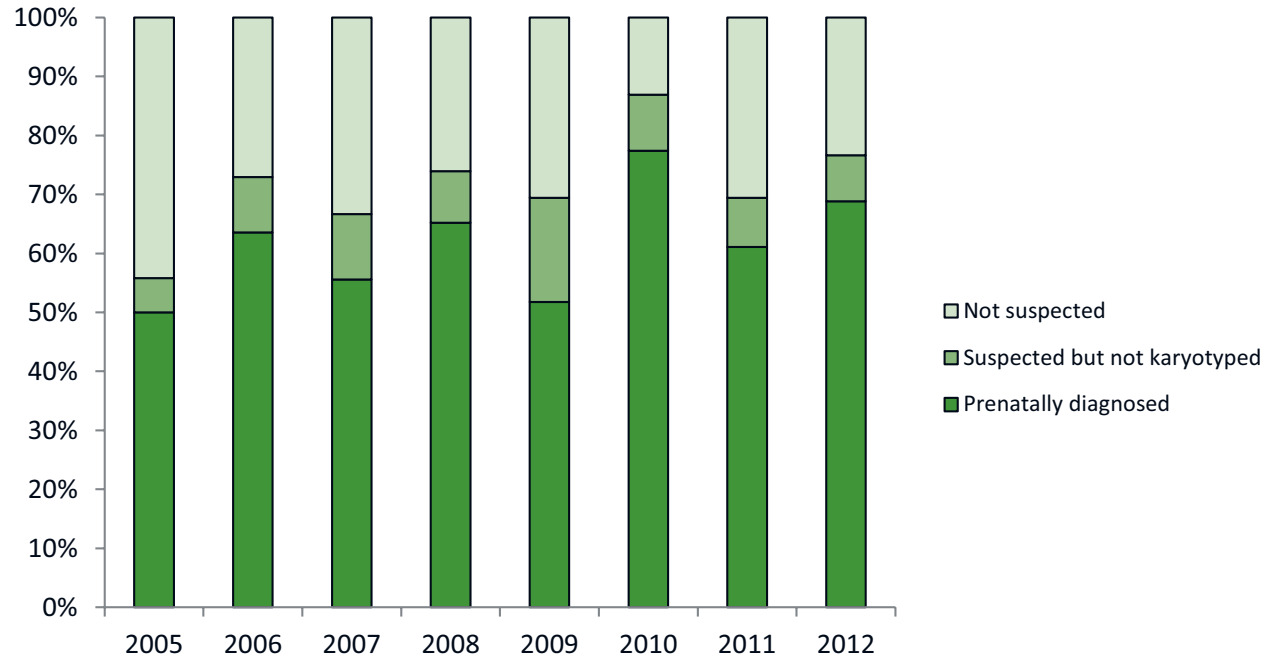
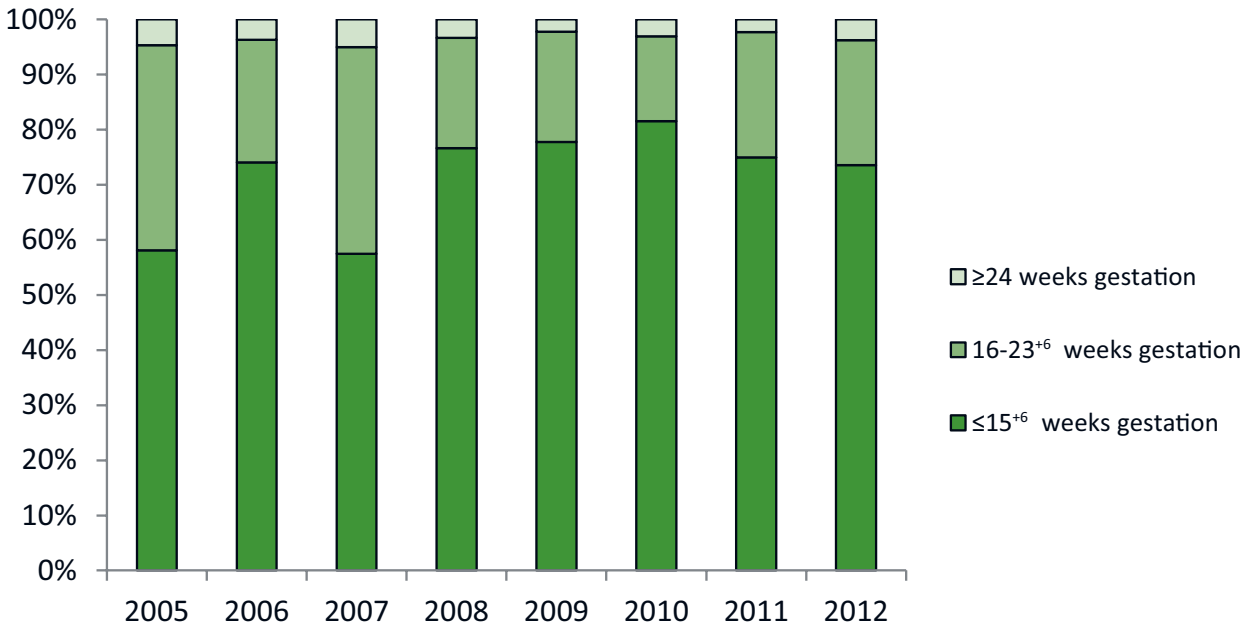


Fig 12b: **Percentage of prenatally diagnosed Down's syndrome cases diagnosed at different gestational ages, by year (n=404)**



Appendices

Appendix 1:

Congenital Anomalies from an unselected population within Oxfordshire, 1991-2012 using data from OXCAR and CAROBB

Background

The Oxford Congenital Anomaly Register (OXCAR) was established 24 years ago, in 1991, after consultation with local experts (obstetricians, midwives, paediatricians, neonatologists, paediatric cardiologists, paediatric pathologists, geneticists, biochemists and public health physicians) who gave full support to the register. One of the main aims of the register at that time was to monitor the newly developing techniques used in prenatal diagnosis and particularly the accuracy of antenatal ultrasound scanning. The first six years of data were summarised in a paper published in the Lancet(70) in 1998. This paper was followed up in 2012 with a publication in BJOG(10), summarizing 18 years of data (Appendix 4).

Other aims were to improve ascertainment to the then National Congenital Anomaly System for surveillance (now carried out by BINOCAR), to provide data for health care policy and planning and for research into the aetiology and natural history of congenital anomalies to enable better advice to be given to parents and prospective parents. In 2003 funding from the Department of Health enabled the expansion of OXCAR to Berkshire and Buckinghamshire (i.e. to cover Thames Valley) and the name was changed to CAROBB. Because there is now 22 years of data for the Oxford area, we are, in this Appendix to the main CAROBB report, summarising these data. More detailed information is available about individual anomalies, prenatal detection rates and outcome of pregnancy. Please contact us by email at carobb@npeu.ox.ac.uk if you would like further information.

The population studied

Anomalies suspected and or confirmed in fetuses / babies booked for delivery at the Oxford Women's Centre, John Radcliffe Hospital, community hospital or home delivery within the catchment area of the Women's Centre and with an OX postcode during 1991 - 2012 inclusive. Denominator data for this population were provided by the Oxford Radcliffe Hospitals NHS Trust Performance & Information Department. There were 143,592 total births in this category in the 22 year study period. Please note this population does not equate with the data from the whole of Oxfordshire used in the CAROBB report. The population used here gives the best approximation available to the unselected local Oxford population.

Table 1 summarises the prenatal detection rates and prevalence per 10,000 births for selected congenital anomalies which form part of the fetal anomaly screening programme (FASP) overseen by the National Screening Committee *(www.fetalanomaly.screening.nhs.uk)*. The prenatal detection figures cannot be directly compared with the FASP targets for prenatal detection because these local figures are for isolated anomalies whereas the FASP figures include all instances of the anomaly, for

example where a syndrome or chromosome anomaly is also present. Individual hospitals monitor their performance against these targets and a national overview from BINOCAR registries may be found in the BINOCAR Annual Report *(www.binocar.org/Publications/Reports)*.

Summary table

Table 1A: Prenatal detection of selected congenital anomalies from an unselected population within Oxfordshire, 1991 – 2012

Defect	Prenatal investigation	Number of pregnancies notified with prenatal suspicion of anomaly (not including false positive diagnoses)	Number of cases notified with anomaly confirmed at birth	Prevalence per 10,000 total births	Prenatal detection rate
Isolated open neural tube defects (anencephaly & spina bifida)	Ultrasound Scanning +/- MS AFP[2]	152	163	11.4	93%
Isolated cardiac anomaly	Ultrasound scanning	161	545	38.0	30%
Isolated cleft lip +/- palate	Ultrasound scanning	69	102	7.1	68%
Down's syndrome	Karyotyping Prenatal detection because MA>35 or 1st or 2nd trimester screening test or ultrasound scanning	278 (226 karyotyped)	392	27.3	71% (58%)
Isolated diaphragmatic hernia	Ultrasound scanning	26	42	2.9	62%
Isolated exomphalos (excludes exomphalos minor)	Ultrasound scanning +/- MS AFP[2]	30	34	2.4	88%
Isolated gastroschisis	Ultrasound scanning +/- MS AFP[2]	33	33	2.3	100%

[1] There is under reporting of cardiac anomalies diagnosed after discharge from the maternity unit particularly for years 1991-2007

[2] Maternal Serum Alpha Feto Protein (MS AFP) screening is not part of combined screening so has not been routinely performed since combined screening was implemented in 2009

Table 2A gives the number of notifications to the OXCAR population from 1991 – 2012, in five periods, four of five years and the most recent period of two years,. The most recent period of only two years has been used to demonstrate the effect of the 2009 the Fetal Anomaly Screening Programme's (FASP) national guidelines on how to manage the reporting of ultrasound normal variants. *www.fetalanomaly.screening.nhs.uk/standardsandpolicies*.

During these time periods the number of cases where there was a prenatal suspicion but the baby was apparently normal at birth rose from 32% of prenatal notifications (19% of total notifications) in 1991–1995 to 49% (42% of total notifications) in 1996-2000 but dropped back to 32% (18% of total notifications) and then 14% (6% of total notifications) for the years 2006-2010 and 2011-2012.

This demonstrates the evolution of reporting ultrasound normal variants (soft markers) such as echogenic bowel and nuchal thickening. These started to be reported regularly in the 1990s. By the late-1990s it was realised that most babies with these markers were usually normal. Local protocols were drawn up to guide professionals on the management of such markers (referred to by this time as 'normal variants'), when to report specific markers and what further tests might be indicated. In 2009 the Fetal Anomaly Screening Programme (FASP) produced national guidelines on how to manage the reporting of ultrasound normal variants. *www.fetalanomaly.screening.nhs.uk/standardsandpolicies*. The years 2011-2012 demonstrate the effect of the FASP normal variants policy, when fully implemented across the region. In this period only 1 in 416 babies had a prenatal suspicion of anomalies and were normal at birth compared to the time period 1996-2000 when 1 in 58 babies had a prenatal suspicion and were subsequently normal at birth.

This trend is illustrated in Figure 2A which, using 3 year running averages shows the percentage of notification made prenatally and those considered to be normal at birth.

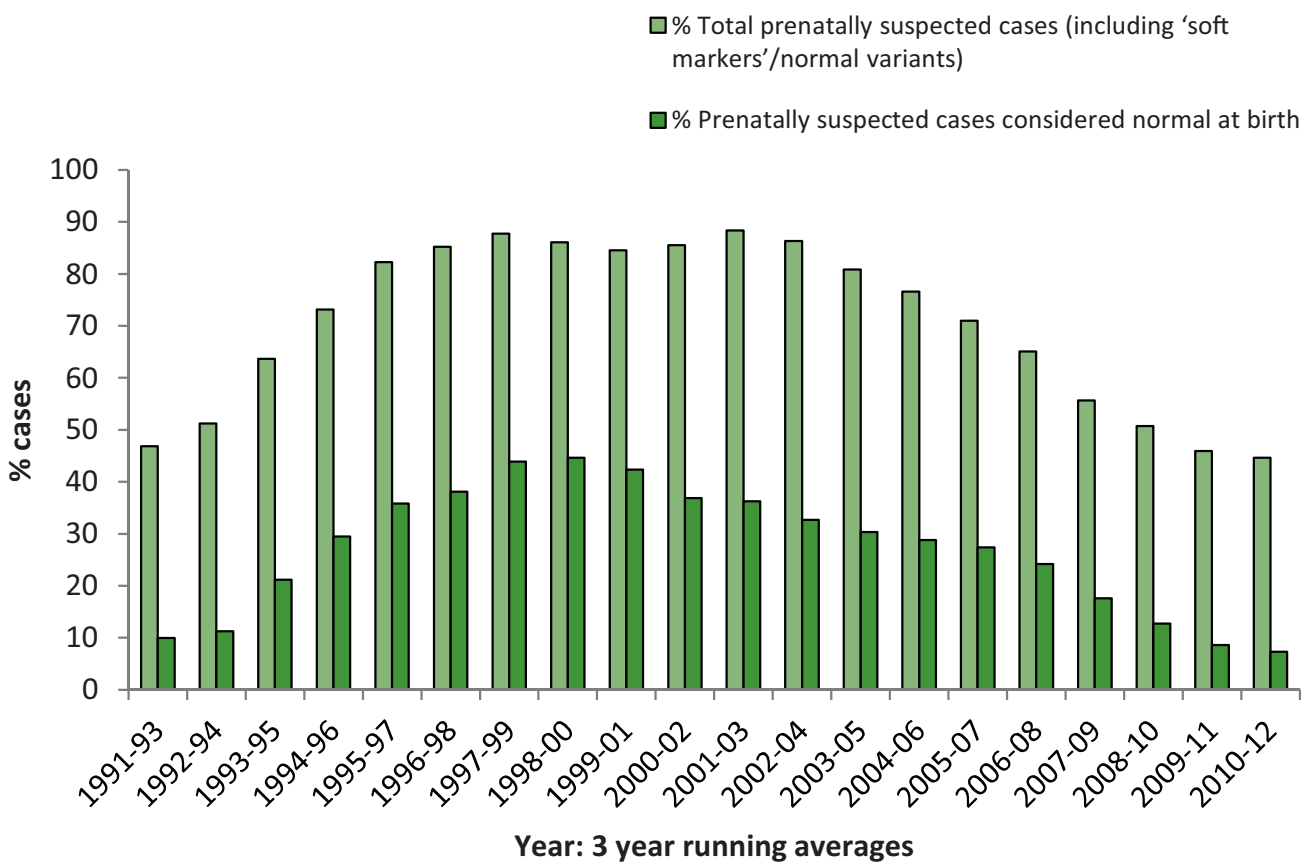
In the same time periods the percentage of cases notified prenatally changed from 58% in the first five years (1991 – 1995), to 86% / 83% in the next two time periods (1996-2005) and then dropped to 58% and 44% in the periods 2006-2010 and 2011-2012 respectively. The apparent fall in the percentage of anomalies detected prenatally (from 51% in 2001-2005 to 39% in 2006-2012) is due to improvement in the ascertainment of postnatally diagnosed anomalies from new sources of ascertainment – particularly for cardiac anomalies.

Table 2A: **Total births and notifications from an unselected population within Oxfordshire, (John Radcliffe Women's Centre booking, with OX postcodes), 1991-2012 inclusive; number prenatally suspected with and without congenital anomaly at birth, number resulting in termination of pregnancy for fetal anomaly (TOPFA), in five time periods**

Year	1991-1995	1996-2000	2001-2005	2006-2010	2011-2012	1991-2012
Total births	28833	28960	33231	37593	14975	143592
Total notifications	751	1205	992	1413	566	4927
Total notifications made prenatally (including 'soft markers'/normal variants)	435	1032	826	818	250	3361
(% of total notified)	(58%)	(86%)	(83%)	(58%)	(44%)	(68%)
Notifications made prenatally with anomaly present at birth	296	530	502	557	214	2099
(% of total)	(39%)	(44%)	(51%)	(39%)	(38%)	(43%)
Notifications made prenatally & considered normal at birth	139	502	324	261	36	1262
(% of total notified prenatally)	(32%)	(49%)	(39%)	(32%)	(14%)	(38%)
Notifications made prenatally and resulting in TOPFA	132	211	209	254	84	890
(% of prenatally diagnosed cases with anomaly confirmed)	(45%)	(40%)	(42%)	(46%)	(39%)	(42%)
Total with anomaly at delivery.	612	703	668	1151	513	3647
(% of total births)	(2.1%)	(2.4%)	(2%)	(3.1%)	(3.4%)	(2.5%)
Proportion of total births with prenatal suspicion & baby normal at birth*	1 in 207	1 in 58	1 in 103	1 in 144	1 in 416	1 in 114

< 1% lost to follow up

Figure 2A **Percentage of cases notified prenatally and percentage of those considered normal at birth using 3 year running averages**



Appendix 2:

CAROBB Notification form

The standard notification form is shown overleaf but we are happy to accept information in other ways eg copies of discharge letters or clinic lists.

Please contact us if you would like to discuss how best to notify cases to the register.

We will provide copies of forms on request or forms can be printed from our website: www.npeu.ox.ac.uk/carobb

CAROBB NOTIFICATION FORM

Congenital Anomaly Register for Oxfordshire, Berkshire and Buckinghamshire

Please register any actual OR prenatally suspected anomaly - structural, chromosomal or biochemical in fetus/baby. (See reverse of form for more information about the register and exclusion list)

Office use only - Case no		
Dup	Com	From

MOTHER DETAILS

(Sticky label, if available)

Surname……

Forename……..**Hosp No**…….

NHS Number……

Postcode *(essential field)*

Mother's DoB *(essential field)*

Booking hosp……

To deliver at…….....
(if different from booking hospital)

EDD *(essential field)*

Mother BMI..... **Smoker in 1st trim?** Y / N
(At booking. Or height & weight)

Assisted conception / IVF? ……
If yes, (please state method, if known)

No of previous pregnancies/births

…..Livebirth …..Miscarriage/TOP *(<24 weeks)* …..Stillbirth/TOP *(>24 weeks)*

Ethnic origin of mother *(pls circle or state)*……..

White	Pakistani	Black Caribbean	Chinese
Mixed	Bangladeshi	Black African	Other
Indian	Other Asian	Other Black	Not known

BABY DETAILS

(Sticky label, if available)

Surname……

Forename……..**Hosp No**……...

NHS Number……

Sex Male / Female / Ambiguous / Not known
(please circle)

Date of delivery / TOP
(and date of feticide if performed)

Place of delivery……....

Gest at delivery……weeks

Weight **g** ☐ Not weighed

Multiple pregnancy?……**Zygosity:** MCMA / MCDA / DCDA

Outcome *(when possible, please report date of delivery, gest, sex, weight and details of any anomalies, whatever the outcome)*

- ☐ Liveborn, no anomaly identified, no follow up requested
- ☐ Liveborn, anomaly present or req' further tests *(please give details)*
- ☐ Miscarriage/IUD *(<24 weeks)*
- ☐ Stillbirth/IUD *(>24 weeks)*
- ☐ Termination
- ☐ Neonatal death

Date of neonatal death

Post mortem? Yes / No / Not known

PRENATAL INVESTIGATIONS

Screening and Diagnostic tests

Gest	Test *(please circle)*	Result
…… ……. ….....	Nuchal / Combined Triple / Quad Other ….....	NT measurement ……mm Down's risk 1 in ……. Tri 13 / 18 risk 1 in ……. Not offered / Declined / Too late
…… …….	CVS / Amnio FISH / PCR	Normal / Abnormal *(state karyotype if known)* …….. Offered / Not offered / Declined
…….	Other*(please state)* ……	

Gest	Ultrasound scan findings *(& any other relevant details)*
…… ……. …… …… …… ……	**Anomaly scan undertaken?** *(please circle)* Yes / Declined / Not offered / Late booker….. …… …… …… …… ……

POSTNATAL DETAILS OF ANOMALY

Prenatally suspected? ☐ **Yes** ☐ **No**

……..
……..
……
……..
……..
……..

Surgery? Performed / Expected in 1st year / Expected after 1st year

Additional details *(eg previous congenital anomalies, illness in mother, exposure to potentially harmful substances)*

……..
……
……..
……..

Referred to:……..

Consanguinity? 1st cousins / other relation……
(if applicable, please circle / state)

Notified by:……..Date:……Hospital:…..Dept:……Tel:……

Confidential: Please send in a sealed envelope to: CAROBB, NPEU, RDB, Old Road Campus, Oxford OX3 7LF or use confidential fax: 01865 289720. Any queries contact Kay Randall: Tel: 01865 289723, E-mail: CAROBB@npeu.ox.ac.uk.

Congenital Anomaly Register for Oxfordshire, Berkshire and Buckinghamshire (CAROBB)

Please complete the form overleaf as fully as possible, registering any anomalies found at whatever stage you become aware of them in the pregnancy/postnatal period.

Uses of the register:

- Audit for prenatal diagnosis
- Evaluation and monitoring of new invasive and non invasive prenatal tests
- Evaluation of new screening programmes
- Provision of data for health care policies and planning
- Provision of data for the investigation of cluster of abnormalities
- Investigation of putative teratogens
- Research on aetiology and natural history of particular malformations
- Improving ascertainment for surveillance by the British Isles Network of Congenital Anomaly Registers (BINOCAR).

Congenital anomalies exclusion list
It is not necessary to report any of the following conditions to us POSTNATALLY, unless there was a prenatal suspicion of an anomaly.

- Spina bifida occulta uncomplicated
- Phymosis
- Stenosis or stricture of lacrimal duct
- Minor skin anomalies less than 4cm^2: skin tag, naevus, angioma, haemangioma, glomus tumor, lymphangioma, birth mark
- Minor anomaly of auricle
- Clicking hip
- Minor anomaly of face or nose
- Minor anomaly of nipple, accessory or ectopic nipple
- Postural clubfoot
- Minor anomalies of the foot: hallux valgus/varus, “orteil en marteau”, metatarus valgus/adductus
- Postural talipes calcaneovalgus or pes calcaneovalgus
- Congenital umbilical hernia, inguinal or para umbilical
- Functional or unspecified cardiac murmur
- Absence or hypoplasia of umbilical artery
- Congenital hydrocele or hydrocele of testis

If in doubt, report to us, we will feed back any inappropriate reporting

Confidentiality and data protection

All information held on the register is strictly confidential. Data are stored in a secure environment at the National Perinatal Epidemiology Unit, University of Oxford (data protection registration number: Z575783X). Any research undertaken is subject to ethical approval. The register holds Confidentiality Advisory Group (CAG) approval and NHS IG Toolkit approval (submission number: 8J017)

Confidential: Please fax or send in a sealed envelope to:

Confidential fax: 01865 289720

CAROBB Co-ordinator
National Perinatal Epidemiology Unit
University of Oxford,
Old Road Campus
Headington
Oxford OX3 7LF
Website: www.npeu.ox.ac.uk/carobb/

Please do not hesitate to contact us with any queries, or requests for more forms.

Tel: 01865 289723
E-mail: carobb@npeu.ox.ac.uk
kay.randall@nhs.net

PLEASE DO NOT SEND ANY NOTIFICATIONS BY E-MAIL, UNLESS USING NHS.NET

Appendix 3:

Research Projects using data from CAROBB

Ongoing projects

1.	**Project title:**	The prevalence of additional anomalies in babies with trisomy 13 or trisomy 18
	Investigators:	Joan Morris, Anna Springett
	Collaboration:	EUROCAT
	Status of study:	Ongoing
2.	**Project title:**	Epidemiology of Transposition of Great Arteries
	Investigators:	Judith Rankin (student MSc project)
	Collaboration:	BINOCAR
	Status of study:	Ongoing
3.	**Project title:**	The epidemiology of tetralogy of fallot and Ebstein's with special emphasis on medication exposure
	Investigators:	Breidge Boyle, Helen Dolk, Maria Loane, Ester Garne
	Collaboration:	EUROCAT
	Status of study:	Ongoing
4.	**Project title:**	The changing epidemiology of gastroschisis in Europe: a register-based study
	Investigators:	Elizabeth Draper, Judith LS Budd, Laura Berry, Lucy K Smith
	Collaboration:	EUROCAT
	Status of study:	Ongoing
5.	**Project title:**	Prevalence of neural tube defects (NTDs) within ethnic communities in the UK
	Investigators:	Jordana Peake
	Collaboration:	BINOCAR
	Status of study:	Ongoing
6.	**Project title:**	Availability/usefulness/value of fetal magnetic resonance imaging for prenatal diagnosis
	Investigators:	Yoshiko Yamamoto
	Collaboration:	Local
	Status of study:	Ongoing
7.	**Project title:**	Survival and predictors of survival of children born with congenital heart disease (CHD)
	Investigators:	Kate Best; Judith Rankin
	Collaboration:	BINOCAR
	Status of study:	Ongoing
8.	**Project title:**	Epidemiology of congenital heart disease in the UK
	Investigators:	Kate Best; Judith Rankin
	Collaboration:	BINOCAR
	Status of study:	Ongoing

9.	**Project title:**	Schmallenburg virus- enhanced surveillance of arthyrogryposis for the Health Protection Agency
	Investigators:	Judith Rankin
	Collaboration:	BINOCAR
	Status of study:	Ongoing
10.	**Project title:**	Trends in hypospadias in Europe in the period 2001-2010
	Investigators:	JEH Bergman, MK Bakker
	Collaboration:	EUROCAT
	Status of study:	Ongoing
11.	**Project title:**	Trends in uptake of post mortem (PM) examination following termination of pregnancy for fetal abnormality (TOPFA)
	Investigators:	Patricia A Boyd, Yoshiko Yamamoto
	Collaboration:	Local
	Status of study:	Ongoing
12.	**Project title:**	Epidemiology of rare syndromes in Europe
	Investigators:	Helen Dolk, Ingeborg Barisic
	Collaboration:	EUROCAT
	Status of study:	Ongoing
13.	**Project title:**	Epidemiology of congenital diaphragmatic hernia in Europe: a register-based study
	Investigators:	Judith Rankin, Mark McGivern, Kate Best
	Collaboration:	EUROCAT
	Status of study:	Ongoing
14.	**Project title:**	The impact of prenatal screening and susbsequent terminations on the prevalence of congential heart disease anomalies in live born babies with Down's syndrome
	Investigators:	Prof Joan Morris, Ester Garne, Diana Wellesley, Anna Springett
	Collaboration:	EUROCAT
	Status of study:	Ongoing
15.	**Project title:**	Epidemiology of Hirschsprung's disease in Europe: a register-based study
	Investigators:	Judith Rankin, Kate Best
	Collaboration:	EUROCAT
	Status of study:	Ongoing
16.	**Project title:**	Investigating the association between congenital anomalies and childhood cancer: a population-based data- linkage study
	Investigators:	Judith Rankin, Peter Tennant
	Collaboration:	BINOCAR
	Status of study:	Ongoing
17.	**Project title:**	Investigation into the genetic basis of renal tract anomalies - feasibility study
	Investigators:	Deirdre Cilliers
	Collaboration:	Local
	Status of study:	Ongoing
18.	**Project title:**	Antenatal diagnosis of lissencephaly
	Investigators:	Paul Griffiths, Mike Reeves

	Collaboration:	BINOCAR
	Status of study:	Ongoing

19.	**Project title:**	Termination of pregnancy for non lethal fetal anomaly: professional perspectives.
	Investigators:	Lisa Crowe, Ruth Graham, Judith Rankin, Steve Robson
	Collaboration:	BINOCAR
	Status of study:	Ongoing

20.	**Project title:**	Prevalence of neural tube defects (NTD) in younger mothers in Europe 2000-2008: analysis of the European surveillance system of congenital anomalies (EUROCAT) database
	Investigators:	M Loane, H Dolk, J Morris, H de Walle, L Abramsky & EUROCAT Working Group
	Collaboration:	EUROCAT
	Status of study:	Ongoing

21.	**Project title:**	The risk of congenital anomalies in multiple births: European registry based study
	Investigators:	Breidge Boyle
	Collaboration:	EUROCAT
	Status of study:	Ongoing

22.	**Project title:**	Gastroschisis study
	Investigators:	Elizabeth Draper
	Collaboration:	BINOCAR
	Status of study:	Ongoing

Completed projects and one-off data requests

23.	**Project title:**	Prevalence, prenatal diagnosis and clinical features of oculoauriculovertebral spectrum: a registry-based study in Europe
	Investigators:	Ljubica Odak, Ingeborg Barisic, Maria Loane, Ester Garne, Diana Wellesley et al
	Collaboration:	EUROCAT
	Status of study:	Complete
24.	**Project title:**	Termination of pregnancy for fetal abnormality report for Department of Health
	Investigators:	Ann Tonks
	Collaboration:	BINOCAR
	Status of study:	Complete
25.	**Project title:**	Epidemiology of multiple congenital anomalies in Europe: A European surveillance system of congenital anomalies (EUROCAT) population-based registry study
	Investigators:	Elisa Calzolari, Ingeborg Barisic , Ester Garne et al
	Collaboration:	EUROCAT
	Status of study:	Complete
26.	**Project title:**	Birth Outcomes in Buckinghamshire - An unidentified problem?
	Investigators:	Lynn Hryhorskyj, Ruchi Baxi
	Collaboration:	Local
	Status of study:	Complete
27.	**Project title:**	Congenital heart defects in Europe: prevalence and perinatal mortality
	Investigators:	Helen Dolk, Maria Loane, Ester Garne.
	Collaboration:	EUROCAT
	Status of study:	Complete
28.	**Project title:**	Epidemiology of orofacial clefts and associated malformations in a geographically defined region.
	Investigators:	Jenaleen Law
	Collaboration:	Local
	Status of study:	Complete
29.	**Project title:**	The evolution of prenatal screening and diagnosis and its impact on an unselected population over an 18 year period
	Investigators:	Patricia Boyd
	Collaboration:	Local
	Status of study:	Complete
30.	**Project title:**	Investigating the epidemiology of partial urorectal septum malformation sequence: a population-based study using data from the British Isles Network of Congenital Anomaly Registers (BINOCAR)
	Investigators:	Judith Rankin, Peter Tennant, Svetlana Glinianaia, Diana Wellesley
	Collaboration:	BINOCAR
	Status of study:	Complete

31.	**Project title:**	Children with language, reading and communication problems
	Investigators:	Dorothy Bishop, Debbie Shears, Patricia Boyd
	Collaboration:	Local
	Status of study:	Complete
32.	**Project title:**	Consanguinity and child health - A brief health needs assessment (Buckinghamshire PCT)
	Investigators:	Lucy Jessop
	Collaboration:	Local
	Status of study:	Complete
33.	**Project title:**	Total & livebirth prevalence of Down's syndrome and other trisomies in Europe 1990-2007: impact of increasing maternal age, prenatal screening and termination of pregnancy
	Investigators:	Maria Loane, Helen Dolk, Joan K Morris, Marie-Claude Addor, Larraitz Arriola,Berenice Doray, Patricia Boyd, Elizabeth Draper or Judith BuddEster Garne, Miriam Gatt, Martin Haeusler, Babak Khoshnood, et al
	Collaboration:	EUROCAT
	Status of study:	Complete
34.	**Project title:**	Consanguinity and child health - A brief health needs assessment (Oxfordshire PCT)
	Investigators:	Rosamund Southgate
	Collaboration:	Local
	Status of study:	Complete
35.	**Project title:**	UK Obstetric Surveillance System (UKOSS) /British Association of Paediatric Surgeons-Congenital Anomalies Surveillance system(BAPS-CASS) study on congential diaphragmatic hernia (CDH)
	Investigators:	Marian Knight
	Collaboration:	Other
	Status of study:	Complete
36.	**Project title:**	Understanding the basis of abnormal haematopoeisis in babies with Down's syndrome
	Investigators:	Mark Anthony
	Collaboration:	Local
	Status of study:	Complete
37.	**Project title:**	Audit of outcome of antenatally diagnosed pulmonary lesions, ie congenital cystic adenomatoid malformation of lung (CCAM), pulmonary sequestration.
	Investigators:	Peter Yeh
	Collaboration:	Local
	Status of study:	Complete
38.	**Project title:**	Data exchange with Down's register
	Investigators:	Cath Rounding, Joan Morris
	Collaboration:	Inter-register
	Status of study:	Complete

39.	**Project title:**	European surveillance system of congenital anomalies (EUROCAT) Website data on prenatal detection rates of congenital anomalies.
	Investigators:	Ester Garne, Helen Dolk, Maria Loane, Patricia Boyd on behalf of EUROCAT
	Collaboration:	EUROCAT
	Status of study:	Complete
40.	**Project title:**	Second report of the Congenital Anomaly Register for Oxfordshire, Berkshire and Buckinghamshire (CAROBB) births 2005-2008 and Oxford births 1991-2008
	Investigators:	Patricia Boyd, Catherine Rounding, Jennifer Kurinczuk
	Collaboration:	Local
	Status of study:	Complete
41.	**Project title:**	Survey of congenital diaphragmatic hernia
	Investigators:	Mary Anthony, Spr Vikranth Venugopalan
	Collaboration:	Local
	Status of study:	Complete
42.	**Project title:**	Ambient air pollution and risk of congenital anomalies in England, 1991-99
	Investigators:	Dolk H, Armstrong B, Lachowycz K, Vrijheid M, Rankin J, Abramsky L, Boyd PA, Wellesley D
	Collaboration:	EUROCAT
	Status of study:	Complete
43.	**Project title:**	Pulse oximetry trial (PulseOX trial) searching for cross border cardiac cases
	Investigators:	Ann Tonks
	Collaboration:	Inter-register
	Status of study:	Complete
44.	**Project title:**	Audit of known fetal abnormalities communicated to neonatologists for CNST Standard 5
	Investigators:	Mary Anthony
	Collaboration:	Local
	Status of study:	Complete
45.	**Project title:**	Report on the data collected on congenital anomalies in South East Region for surveillance and for monitoring the national antenatal Down's's syndrome and fetal anomaly screening programmes.
	Investigators:	Val Armstrong, Patricia A Boyd, Diana Wellesley and Catherine Rounding
	Collaboration:	Inter-register
	Status of study:	Complete
46.	**Project title:**	The outcomes of antenatally diagnosed isolated heart anomalies
	Investigators:	Moira Blyth and Diana Wellesley
	Collaboration:	Inter-register
	Status of study:	Complete
47.	**Project title:**	Schizencephaly prevalence, prenatal diagnosis and clues to etiology: a register-based study
	Investigators:	David Howe, Judith Rankin, Elizabeth Draper
	Collaboration:	BINOCAR
	Status of study:	Complete

48.	**Project title:**	Audit of soft markers in a population already screened for aneuploidy in the first trimester.
	Investigators:	Lawrence Impey
	Collaboration:	Local
	Status of study:	Complete
49.	**Project title:**	Analysing the rare unbalanced chromosome abnormalities reported to European surveillance system of congenital anomalies (EUROCAT)
	Investigators:	Diana Wellesley, Ingeborg Barisic, Patricia A Boyd, Helen Dolk, Ruth Greenlees
	Collaboration:	EUROCAT
	Status of study:	Complete
50.	**Project title:**	British Isles Network of Congenital Anomaly Registers (BINOCAR) Down's syndrome prenatal screening audit
	Investigators:	
	Collaboration:	BINOCAR
	Status of study:	Complete
51.	**Project title:**	A descriptive epidemiological study of small intestinal atresia in Europe
	Investigators:	K E Best, Judith Rankin et al
	Collaboration:	EUROCAT
	Status of study:	Complete
52.	**Project title:**	Evaluation of prenatal diagnosis rates for major structural congenital anomalies across areas covered by the British Isles network of congenital anomaly registers: 2005 to 2006
	Investigators:	Patricia A Boyd, Ann M Tonks, Judith Rankin, Catherine Rounding, Diana Wellesley, Elizabeth S Draper, and the BINOCAR working group
	Collaboration:	BINOCAR
	Status of study:	Complete
53.	**Project title:**	Oesophageal atresia: Population based study of epidemiology and outcome in european regions.
	Investigators:	Rikke Neess Pedersen, Ester Garne, Steffen Husby
	Collaboration:	EUROCAT
	Status of study:	Complete
54.	**Project title:**	Prevalence of congenital cystic adenomatoid malformation (CCAM) and other thoracic anomalies
	Investigators:	Steve Gould
	Collaboration:	Local
	Status of study:	Complete
55.	**Project title:**	To define the outcome of prenatally diagnosed gastroschisis with intra abdominal bowel dilatation vs those with no dilatation in the Thames valley region
	Investigators:	Kokila Lakhoo
	Collaboration:	Local
	Status of study:	Complete
56.	**Project title:**	Fraser Syndrome
	Investigators:	Helen Dolk, Ingeborg Barisic
	Collaboration:	EUROCAT
	Status of study:	Complete

57.	**Project title:**	Cornelia de Lange syndrome
	Investigators:	Helen Dolk, Ingeborg Barisic
	Collaboration:	EUROCAT
	Status of study:	Complete
58.	**Project title:**	Cognitive and behavioural outcomes of children with an extra sex chromosome
	Investigators:	Pat Jacob, Dorothy Bishop, Gaia Scerif
	Collaboration:	Dept of Experimental Psychology, Oxford University; Wessex Regional Genetics Laboratory
	Status of study:	Complete
59.	**Project title:**	Outcome of prenatally diagnosed exomphalos
	Investigators:	Kokila Lakhoo, N Shenker, J Sadiq
	Collaboration:	Local
	Status of study:	Complete
60.	**Project title:**	Audit of craniofacial anomalies
	Investigators:	Paul Chamberlain
	Collaboration:	Local
	Status of study:	Complete
61.	**Project title:**	Sex chromosome trisomies in europe: prevalence, prenatal detection and outcome of pregnancy
	Investigators:	Patricia A Boyd, M Loane, E Garne, B Khoshnood, H Dolk, and a EUROCAT working group
	Collaboration:	EUROCAT
	Status of study:	complete
62.	**Project title:**	The epidemiology of hospital admission rates for cleft lip and palate in the United Kingdom
	Investigators:	Aadil A Khan, Tim Goodacre
	Collaboration:	Local
	Status of study:	Complete
63.	**Project title:** of	Terminations of pregnancy ≥ 24 weeks of gestation after prenatal diagnosis fetal abnormality in Europe
	Investigators: Babak Khoshnood	Ester Garne, Helen Dolk, Patricia A Boyd, Maria Loane, Catherine de Vigan,
	Collaboration:	EUROCAT
	Status of study:	Complete
64.	**Project title:**	Audit cystic hygroma and neonatal outcome
	Investigators:	Kokila Lakhoo
	Collaboration:	Local
	Status of study:	Complete
65.	**Project title:**	Maternal age-specific risk of non-chromosomal anomalies
	Investigators:	M Loane, H Dolk, JK Morris, EUROCAT Working Group
	Collaboration:	EUROCAT
	Status of study:	Complete
66.	**Project title:**	Sacrococcygeal teratoma audit
	Investigators:	Kokila Lakhoo
	Collaboration:	Local
	Status of study:	Complete

67.	**Project title:**	Antenatal diagnosis of duodenal atresia and postnatal outcome
	Investigators:	Ms PG Roy, Kokila Lakhoo, Patricia A Boyd
	Collaboration:	Local
	Status of study:	Complete
68.	**Project title:**	Oro-facial Clefts. World-wide recent total prevalence data.
	Investigators:	Pierpaolo Mastroiacovo
	Collaboration:	Other
	Status of study:	Complete
69.	**Project title:**	Prenatal screening in Europe
	Investigators:	Patricia A Boyd, Ester Garne
	Collaboration:	EUROCAT
	Status of study:	Complete
70.	**Project title:**	Isolated cleft lip and palate audit
	Investigators:	Dorothy Halliday, Patricia Boyd
	Collaboration:	Local
	Status of study:	Complete
71.	**Project title:**	Audit of prenatal lung lesions versus pathological diagnosis
	Investigators:	P Teong, K Lakhoo, L Impey
	Collaboration:	Local
	Status of study:	Complete
72.	**Project title:**	Audit of gastroschisis 1995-2005
	Investigators:	Gail Whitehead
	Collaboration:	Local
	Status of study:	Complete
73.	**Project title:**	Arthrogryposis multiplex congenital (AMC) causes and risk factors
	Investigators:	Jana Midelfart Hoff
	Collaboration:	EUROCAT
	Status of study:	Complete
74.	**Project title:**	Assessment of ultrasound markers and their value
	Investigators:	National Screening Committee
	Collaboration:	Local
	Status of study:	Complete
75.	**Project title:**	Survey of congenital lung anomalies
	Investigators:	Mary Anthony
	Collaboration:	Local
	Status of study:	Complete
76.	**Project title:**	Audit of screening offered to parents of those babies born with Down syndrome
	Investigators:	Gail Whitehead
	Collaboration:	Local
	Status of study:	Complete
77.	**Project title:**	Audit of screening of fetuses with echogenic bowel
	Investigators:	Gail Whitehead
	Collaboration:	Local
	Status of study:	Complete

78.	**Project title:**	Understanding congenital anomaly hotspots within Oxon (postcode mapping)
	Investigators:	Angela Baker
	Collaboration:	Local
	Status of study:	Complete
79.	**Project title:**	Absent stomach bubble/Tracheo-oesophageal fistula/oesophageal atresia
	Investigators:	Paul Chamberlain, Kokila Lakhoo, Patricia A Boyd
	Collaboration:	Local
	Status of study:	Complete
80.	**Project title:**	Clinical genetics audit of late termination of pregnancy
	Investigators:	Dorothy Halliday, Patricia A Boyd
	Collaboration:	Local
	Status of study:	Complete
81.	**Project title:**	How have babies born with spina bifida in the 1990's fared?
	Investigators:	Jenny Kurinczuk, Jenny Calvert, Patricia A Boyd, Paul Chamberlain, Mary Anthony
	Collaboration:	Local
	Status of study:	Complete
82.	**Project title:**	Follow-up of children with congenital anomalies long-term (FOCAL) Pilot study of diaphragmatic hernia
	Investigators:	FOCAL
	Collaboration:	BINOCAR & BDF Newlife
	Status of study:	Complete
83.	**Project title:**	Geographical variation in overall rates of congenital abnomalities and the rates for specific abnormalities
	Investigators:	Helen Dolk
	Collaboration:	EUROCAT
	Status of study:	Complete
84.	**Project title:**	Myotonic dystrophy audit
	Investigators:	Paul Chamberlain
	Collaboration:	Local
	Status of study:	Complete
85.	**Project title:**	Concern from member of public re rise in no of anomalies since 1995
	Investigators:	Don Sinclair
	Collaboration:	Local
	Status of study:	Complete
86.	**Project title:**	National congenital anomalies system (NCAS) alert re cardiac & urogenital anomalies
	Investigators:	Monica Dent
	Collaboration:	Other
	Status of study:	Complete
87.	**Project title:**	Investigation of neural tube defects (NTDs) near landfill site
	Investigators:	Nick Hicks
	Collaboration:	Local
	Status of study:	Complete

88.	**Project title:**	Local investigation of potential cluster
	Investigators:	G Dean
	Collaboration:	Local
	Status of study:	Complete
89.	**Project title:**	Chlorination of water supplies and birth defects
	Investigators:	Paul Elliott
	Collaboration:	SASHU
	Status of study:	Complete
90.	**Project title:**	Congenital hydrocephalus: a population based study on prevalence and outcome
	Investigators:	Ester Garne
	Collaboration:	EUROCAT
	Status of study:	Complete
91.	**Project title:**	John Radcliffe fetal medicine termination of pregnancy for fetal abnormality audit
	Investigators:	Lawrence Impey, Kay Randall
	Collaboration:	Local
	Status of study:	One off data request**Project title:** Fetal anomalies screening programme (FASP) Downs livebirth case matching
	Investigators:	Anne Roberts
	Collaboration:	Local
	Status of study:	One off data request
92.	**Project title:**	Berkshire perinatal morbidity and ethnicity
	Investigators:	Request via Jenny Kurinczuk from Berkshire Public Health
	Collaboration:	Local
	Status of study:	One off data request
93.	**Project title:**	East Midlands and South Yorkshire Congenital Anomalies Register (EMSYCAR) records data exchange
	Investigators:	C Rounding, J Budd
	Collaboration:	Inter-register
	Status of study:	One off data request
94.	**Project title:**	Outcome of prenatally detected fetal brain abnormalities
	Investigators:	Usha Kini
	Collaboration:	Local
	Status of study:	One off data request
95.	**Project title:**	CAROBB / NDSCR Data exchange
	Investigators:	Anna Springett
	Collaboration:	Inter-register
	Status of study:	One off data request
96.	**Project title:**	The increasing reported incidence of echogenic lung lesions
	Investigators:	DT Howe; Diana Wellesley
	Collaboration:	EUROCAT
	Status of study:	One off data request

97.	**Project title:**	Fetal anomaly screening programme (FASP) audit data supply
	Investigators:	Annie Roberts
	Collaboration:	Local
	Status of study:	One off data request
98.	**Project title:**	Fetal anomaly screening programme (FASP) audit data supply
	Investigators:	Powatti Ramchand
	Collaboration:	Local
	Status of study:	One off data request
99.	**Project title:**	Improving care for infants and their families before, during and after surgery.
	Investigators:	Jenny Kurinczuk
	Collaboration:	Local
	Status of study:	One off data request
100.	**Project title:**	Down babies case matching exercise for annual report
	Investigators:	Catryn Dixon, Alison Wainwright
	Collaboration:	Local
	Status of study:	One off data request
101.	**Project title:**	Congenital cystic adenomatoid malformation of lung (CCAM) incidence to compare with Wessex region
	Investigators:	Diana Wellesley
	Collaboration:	Inter-register
	Status of study:	One off data request
102.	Project title:	Audit of 11 conditions for the 20 week ultrasound scan
	Investigators:	Jeanne Harris
	Collaboration:	Local
	Status of study:	One off data request
103.	Project title:	Fetal anomaly screening programme (FASP) gastroschisis audit
	Investigators:	Anne Roberts
	Collaboration:	Local
	Status of study:	One off data request
104.	Project title:	Spina bifida rates for statement in response to increase in Scotland
	Investigators:	Liz Draper
	Collaboration:	BINOCAR
	Status of study:	One off data request
105.	Project title:	Neural tube defect (NTDs) and cardiac anomaly numbers
	Investigators:	Judith Rankin
	Collaboration:	Inter-register
	Status of study:	One off data request
106.	Project title:	Parity information for selected anomalies for Berkshire
	Investigators:	Jenny Kurinczuk and Liz Ollerhead
	Collaboration:	Local
	Status of study:	One off data request

107.	Project title:	Tracheo-oesophageal fistula (TOF) cases data exchange with UKOSS
	Investigators:	Marian Knight
	Collaboration:	Other
	Status of study:	One off data request
108.	Project title:	Comparative incidence and prevalence of abdominal wall defects
	Investigators:	Kokila Lakhoo
	Collaboration:	Local
	Status of study:	One off data request
109.	Project title:	Gastroschisis numbers 2002-08
	Investigators:	Kokila Lakhoo
	Collaboration:	Local
	Status of study:	One off data request
110.	Project title:	Bladder exstrophy cases numbers prenatally detected.
	Investigators:	Diana Wellesley
	Collaboration:	Inter-register
	Status of study:	One off data request
111.	Project title:	Exomphalos audit
	Investigators:	Elizabeth Draper
	Collaboration:	BINOCAR
	Status of study:	One off data request
112.	Project title:	Gastroschisis audit
	Investigators:	Elizabeth Draper
	Collaboration:	BINOCAR
	Status of study:	One off data request
113.	Project title:	Gastroschisis case matching exercise with UK Obstetric Surveillance System (UKOSS)
	Investigators:	Marian Knight
	Collaboration:	Other
	Status of study:	One off data request
114.	Project title:	Echogenic bowel audit - cross referencing of cases
	Investigators:	Jackie Lovstrom
	Collaboration:	Local
	Status of study:	One off data request
115.	Project title:	Abdominal cyst audit
	Investigators:	Kokila Lakhoo
	Collaboration:	Local
	Status of study:	One off data request
116.	Project title:	Gastroschisis case matching exercise for UKOSS
	Investigators:	Marian Knight
	Collaboration:	Other
	Status of study:	One off data request

117.	Project title:	Supply of data for national screening committee - Down's cases
	Investigators:	Anne Roberts
	Collaboration:	Local
	Status of study:	One off data request
118.	Project title:	Supply of data for national screening committee - cases of anencephaly and gastroschisis
	Investigators:	Anne Roberts
	Collaboration:	Local
	Status of study:	One off data request
119.	Project title:	Gastroschisis rates for 2002-2006 for TVSHA to compare with South West congenital anomaly register (SWCAR)
	Investigators:	Aileen McLoughlin
	Collaboration:	Local
	Status of study:	One off data request
120.	Project title:	Risk management review of cleft lips/palates
	Investigators:	Michelle Errington
	Collaboration:	Local
	Status of study:	One off data request
121.	Project title:	Annual report of anomalies to feedback to antenatal department
	Investigators:	Ann Folkes
	Collaboration:	Local
	Status of study:	One off data request
122.	Project title:	Prenatally suspected heart defects in Down syndrome
	Investigators:	Nick Archer
	Collaboration:	Local
	Status of study:	One off data request
123.	Project title:	Atrioventricular septal defect audit
	Investigators:	Paul Chamberlain
	Collaboration:	Local
	Status of study:	One off data request
124.	Project title:	Incidence of brain anomalies
	Investigators:	Marion Knight
	Collaboration:	Other
	Status of study:	One off data request
125.	Project title:	Neural tube defects figures for England and Wales
	Investigators:	Elizabeth Draper
	Collaboration:	BINOCAR
	Status of study:	One off data request

Appendix 4: Publications to which CAROBB / OXCAR have contributed information

1. Calzolari E, Barisic I, Loane M, Morris J, Wellesley D, Dolk H, et al. Epidemiology of multiple congenital anomalies in Europe: A EUROCAT population-based registry study. Birth Defects Res A Clin Mol Teratol. 2014;100(4):270-6.

2. Barisic I, Odak L, Loane M, Garne E, Wellesley D, Calzolari E, et al. Prevalence, prenatal diagnosis and clinical features of oculo-auriculo-vertebral spectrum: a registry-based study in Europe. Eur J Hum Genet. 2014.

3. Loane M, Morris JK, Addor MC, Arriola L, Budd J, Doray B, et al. Twenty-year trends in the prevalence of Down syndrome and other trisomies in Europe: impact of maternal age and prenatal screening. Eur J Hum Genet. 2013;21(1):27-33.

4. Khoshnood B, Loane M, Garne E, Addor MC, Arriola L, Bakker M, et al. Recent decrease in the prevalence of congenital heart defects in Europe. The Journal of pediatrics. 2013;162(1):108-13 e2.

5. Boyle B, McConkey R, Garne E, Loane M, Addor MC, Bakker MK, et al. Trends in the prevalence, risk and pregnancy outcome of multiple births with congenital anomaly: a registry-based study in 14 European countries 1984-2007. BJOG. 2013;120(6):707-16.

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13. Loane M, Dolk H, Garne E, Greenlees R. Paper 3: EUROCAT data quality indicators for population-based registries of congenital anomalies. Birth Defects Res A Clin Mol Teratol. 2011;91 Suppl 1:S23-30.

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15. IPDTOC Working Group. Prevalence at Birth of Cleft Lip With or Without Cleft Palate: Data From the International Perinatal Database of Typical Oral Clefts (IPDTOC). The Cleft Palate-Craniofacial Journal. 2011;48(1):66-81.

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Appendix 5: Data Protection and handling requests for data

5a Confidentiality Advisory Group (CAG) approval documentation

5b National Research Ethics Committee approval documentation

5c Application form and guidelines for use of CAROBB data

Confidentiality Advisory Group (CAG) approval for CAROBB (as part of BINOCAR) to collect identifiable information without explicit consent from individuals registered.

Application Number	0009	
Reference	PIAG 2-08(e)/2002	
Other Refs		
Application Title	Congenital Anomalies Register (BINOCAR)	
Application Summary	To provide continuous epidemiological monitoring of the frequency, nature, cause and outcomes of congenital anomalies by means of national, regional and disease specific registers of congenital anomalies.**Dec 08 Application extended to contain address info at conception**. Amended 07/08/2012 to disclose baby's date of birth/date of death data to EUROCAT and link local registry data at the BINOCAR hub. Amended 16/07/2013 allowing a member of Department of Health (DH) staff access to National Down Syndrome Cytogenetic Register (NDSCR) data in order to compare with DH notification data. The aim of the activity would be to try and match each termination recorded in the NDSCR with that recorded by DH and determine whether the case has been correctly notified to DH.	
Applicant Organisation Name	British Isles Network of Congenital Anomalies Register (BINOCAR)	
Contact Name	Elizabeth S Draper,Chair of BINOCAR	
Address	Department of Health Sciences, University of Leicester	
	22-28 Princess Road West	
	Leicester	
Postcode	LE1 6TP	
Telephone	0116 252 3210	
Email	*msn@leicester.ac.uk*	
Medical Purposes	Y	preventative medicine
	□	medical diagnosis
	□	medical research, approved by a research ethics committee
	□	the provision of care and treatment
	□	the management of health and social care services
	□	informing individuals about their physical or mental health or condition, the diagnosis of their condition or their care and treatment
Cohort/Population	UK-wide: patients with congenital anomalies	
Description of confidential patient information used	Mother's name, address, postcode, hospital number, NHS number, date of birth. Baby's name, address, postcode, hospital number, NHS number, date of birth, date of death. Address at conception.	
S251 Class(es)	□	Specific Support
	Y	Class I - making the person less readily identifiable
	Y	Class II - present or past geographical locations of patients
	Y	Class III - to identify and contact patients to obtain consent
	Y	Class IV - linking multiple sources;validating quality and completeness; avoiding error
	Y	Class V - audit, monitoring, & analysis of healthcare provision
	Y	Class VI - granting of access to data for purposes I-V
Sponsor		
Status	Approved	
Outcome Date	20/06/2002	
Next Review Date	04/07/2014	
Notes	NorCAS and WMCAR activities have been novated into the PHE applications process.	

NHS
National Research Ethics Service
Trent Research Ethics Committee
Research Ethics Office
Derwent Shared Services
Laurie House
Colyear Street
Derby
DE1 1LJ

Telephone: 01332 868765
Facsimile: 01332 868930

11 October 2009

Professor Elizabeth Draper
Dept of Health Sciences
22-28 Princess Road West
Leicester
LE1 6TP

Dear Professor Draper

Title of the Database: **British Isles Network of Congenital Anomaly Registers (BINOCAR)**

REC reference: **09/H0405/48**

The Research Ethics Committee reviewed the above application at the meeting held on 1 October 2009. Thank you for attending to discuss the application.

Ethical opinion

The members of the Committee present gave a favourable ethical opinion of the above research database on the basis described in the application form and supporting documentation.

Duration of ethical opinion

The favourable opinion is given for a period of five years from the date of this letter and provided that you comply with the conditions set out in the attached document. You are advised to study the conditions carefully. The opinion may be renewed for a further period of up to five years on receipt of a fresh application. It is suggested that the fresh application is made 3-6 months before the 5 years expires, to ensure continuous approval for the research database.

Approved documents

The documents reviewed and approved at the meeting were:

Document	*Version*	*Date*
Covering Letter		14 August 2009
REC application	IRAS Research Database Form V 2.3 (lock code 25660/56406/9/606)	19 August 2009
Participant Information Sheet	V 1.1	12 August 2009
Protocol	V 2.0	12 August 2009

Research governance

Under the Research Governance Framework (RGF), there is no requirement for NHS research permission for the establishment of research databases in the NHS. Applications to NHS R&D offices through IRAS are not required as all NHS organisations are expected to have included management review in the process of establishing the database.

Research permission is also not required by collaborators at data collection centres (DCCs) who provide data under the terms of a supply agreement between the organisation and the database. DCCs are not research sites for the purposes of the RGF.

Database managers are advised to provide R&D offices at all DCCs with a copy of the REC application for information, together with a copy of the favourable opinion letter when available. All DCCs should be listed in Part C of the REC application.

NHS researchers undertaking specific research projects using data supplied by a database must apply for permission to R&D offices at all organisations where the research is conducted, whether or not the database has ethical approval.

Site-specific assessment (SSA) is not a requirement for ethical review of research databases. There is no need to inform Local Research Ethics Committees.

Membership of the Committee

The members of the Ethics Committee who were present at the meeting are listed on the attached sheet.

Statement of compliance

The Committee is constituted in accordance with the Governance Arrangements for Research Ethics Committees (July 2001) and complies fully with the Standard Operating Procedures for Research Ethics Committees in the UK.

After ethical review

Now that you have completed the application process please visit the National Research Ethics Service website > After Review

Here you will find links to the following:

a) Providing feedback. You are invited to give your view of the service that you have received from the National Research Ethics Service and the application procedure. If you wish to make your views known please use the feedback form available on the website.
b) Annual Reports. Please refer to the attached conditions of approval.
c) Amendments. Please refer to the attached conditions of approval.

Continued/

We would also like to inform you that we consult regularly with stakeholders to improve our service. If you would like to join our Reference Group please email referencegroup@nres.npsa.nhs.uk

09/H0405/48	**Please quote this number on all correspondence**

Yours sincerely

Dr Ian Gaywood
Chair

E-mail: jenny.hancock@derwentsharedservices.nhs.uk

Enclosures: *List of names and professions of members who were present at the meeting and those who submitted written comments*

Approval conditions

DATA REQUEST FORM

- This form is intended for requests for data for research purposes.
- Please read the CAROBB Guidelines and the notes on page 2 of this form before you sign.
- All requests will be approved by CAROBB Management Committee.
- Please complete, then **email and post a hard copy** (with signature and supporting documentation eg protocol) to Cath Rounding (CAROBB Co-ordinator) at the address at the bottom of this sheet.
- Please include any details of ethical approvals sought / granted.

Requester details	
Name:	
Job Title/Position:	
Organisation:	
Address:	
Contact phone number:	
Email address:	
Lead Clinician/Supervisor:	

Requester agreement	
Details of funding and source for project	
Name of person responsible for data security	
Request details	
Name of Project	
What question do you wish to answer?	
Intended use of information (e.g. Background, intended presentation/meeting/report)	

CAROBB, NPEU, University of Oxford, Old Road Campus, Headington, Oxford OX3 7LF
Direct: 01865 289721, Confidential fax: 01865 289720, E-mail: catherine.rounding@npeu.ox.ac.uk

DATA REQUEST FORM

Main outcome measures		
Case definition: (e.g. pre / postnatal diagnosis, live / stillbirths / TOPs.)		
Population: (e.g. CAROBB, Oxfordshire only)		
Time period (births):	By EDD or by Date of Birth? from to:	
Justification for identifiable data		
Do you plan to seek ethical approval / R&D approval for this study? (Please give details if yes)		
Signature:		Date:
Date required by:		

Please tick to confirm that you agree to the following:

□ To supply CAROBB with a 6 monthly update report.

□ On completion of the project all individual records will be destroyed and a CAROBB data destruction form completed and returned (requests for individual records only).

□ Any publications/reports arising from the use of data supplied must include a standard acknowledgement paragraph (CAROBB will supply content).

□ Any publications arising from the use of data supplied must be sent to Register Leads for approval while at draft stage. The register is also obliged to send the draft to the register funding body for approval.

□ I have read and agree to the CAROBB Guidelines

We are keen for the CAROBB information to be used for research purposes and will do our best to help with any requests for data. Please do not hesitate to contact us with any queries.

CAROBB, NPEU, University of Oxford, Old Road Campus, Headington, Oxford OX3 7LF
Direct: 01865 289721, Confidential fax: 01865 289720, E-mail: catherine.rounding@npeu.ox.ac.uk

GUIDELINES for users CAROBB

Congenital Anomaly Register for Oxfordshire, Berkshire and Buckinghamshire

CAROBB (Congenital Anomaly Register for Oxfordshire, Berkshire and Buckinghamshire) was awarded funding by the Department of Health in 2003 to establish a database of information on babies born with suspected or confirmed congenital anomalies for the three counties. Prior to 2003, the register was known as OXCAR and included cases seen at the John Radcliffe Hospital since 1991.

The principal objectives of CAROBB are:

- Provide data for research on the aetiology and natural history of particular malformations to enable better advice based on accurate information to be given to parents

- Enable the evaluation and monitoring of new invasive and non invasive prenatal tests.

- Evaluate new prenatal screening programmes and to provide data for health care policies and planning

- Provide data to investigate clusters of abnormalities and putative teratogens by the monitoring of incidence over time and in population trends such as maternal age, ethnicity, and health inequalities.

CAROBB can be used as a basis for other studies and there are increasing numbers of requests for access to the data for research purposes. The Management Group wishes to encourage the use of the register in this way and the following guidelines have been drawn up to help potential register users. CAROBB

conforms to the Data Protection Act 1998 and the Health and Social Care Act 2001.

Please feel free to contact the Register Co-ordinator for a discussion of your proposal at an early stage. It is important to be clear about what information you wish to collect and what information you will be able to obtain through the register.

1. All requests for access to CAROBB data should be made through the research co-ordinator using the accompanying form.

2. The request should be accompanied by a study protocol. The protocol must be approved by CAROBB. Approval by an ethics committee will not guarantee approval by CAROBB. Any amendments required by an ethics committee must be approved by CAROBB before data will be released.

3. If appropriate, the researcher will be responsible for obtaining approval from Ethics Committees in the areas in which the cases live. A copy of the approval must be supplied to the register co-ordinator before data will be released for the study.

4. Researchers are expected to seek peer review of the proposed study.

5. Researchers will need to seek the permission of the parent/child's general practitioner prior to contacting parents and children. If necessary, permission must also be sought from the appropriate consultant for access to hospital notes.

6. If the researcher has little or no previous experience of research the Management Group will require a written assurance from a supervisor that the work will be carried out and completed satisfactorily.

7. It is the responsibility of the researcher to apply for funds to carry out the proposed study. A small administrative charge may be made to cover the cost of accessing cases from CAROBB.

8. Data supplied by CAROBB must not be passed to a third party, nor should it be re-used for later study without applying to CAROBB for permission. Personal data must not be uploaded to a researchers home computer. Researchers are expected to deposit datasets which have been derived from the original data, with suitable documentation, in the CAROBB database.

9. In compliance with the Data Protection Act, 1998, to keep the database as accurate as possible, researchers will be expected to inform CAROBB of changes to subjects details during the course of the study.

10. The Management Group will request a short progress report at intervals during the course of the study and evidence of the final results in the form of a report or paper. Any change in contact addresses or personnel working on the project should be notified to the Management Group.

11. The Management Group would like to see an advanced draft of any publication, or abstract submitted for a meeting, in which CAROBB data have been used. Congenital Anomaly Register for Oxfordshire, Berkshire and Buckinghamshire should be acknowledged in any publication or presentation, arising from CAROBB data, using the sentence "The Management Group of Congenital Anomaly Register for Oxfordshire, Berkshire and Buckinghamshire approved the release of register data for this study. CAROBB is funded by the Department of Health."

12. On completion of the analysis and after copy datasets have been supplied to CAROBB, ALL PERSONAL IDENTIFIABLE INFORMATION MUST BE DESTROYED, in accordance with any requirements of the ethics approval for the study. If you are unsure on this point, contact CAROBB for clarification.

Please complete the application form enclosed

and return to the CAROBB office.

Appendix 6: Publicity

6a Poster for clinic waiting rooms

6b Leaflet for clinic waiting rooms

Congenital Anomaly Register for Oxfordshire, Berkshire & Buckinghamshire

Congenital Anomaly Register for Oxfordshire, Berkshire and Buckinghamshire

Most babies are born healthy,

but

if a baby is born with a birth defect (congenital anomaly)

or

a problem is suspected on scan before birth

information about the defect and the pregnancy is recorded on a local register and on a national one at the Office of National Statistics which was set up in the 1960s following the birth of babies affected by Thalidomide.

Why is this information collected?

- To improve our understanding of congenital anomalies and help research into causes, treatment and prevention
- To help identify possible clusters of birth defects
- To check how good antenatal scans and screening tests are at picking up problems
- To help plan and develop NHS services

The information collected is held securely and is strictly confidential. If you have any questions or concerns about the information that might be held about you or your baby, please contact:

CAROBB, National Perinatal Epidemiology Unit, University of Oxford, Old Road Campus, Headington, Oxford OX3 7LF
E-mail: CAROBB@npeu.ox.ac.uk.
Website: www.npeu.ox.ac.uk/carobb

npeu National Perinatal Epidemiology Unit

How is information collected?

A member of staff from the hospital who treats you or your baby, completes a notification to the register when the anomaly is identified. Any information reported in the early stages can be improved or confirmed later by sending another notification.

Names and postcodes are included so that information can be updated on the correct case and the same baby is not counted several times.

Information is collected on paper and stored electronically on a computer. This information is held securely by CAROBB.

Does my name or my baby's name have to go on the Register?

We hope everyone will want to be included on the Register, to help us plan and improve services for future mothers and babies. However, your details can be removed at any time.

Will the database be secure and confidential?

The information recorded on the Register about you or your baby is confidential. It is held in a responsible way which respects the rights and privacy of individuals.

The Register follows a strict policy on security and confidentiality. This policy is available to the public. The register conforms to the requirements of legislation on data protection.

How can I find out more about CAROBB?

If you have any questions or concerns regarding the information that could be held on you or your baby, please contact the registry:

CAROBB
National Perinatal Epidemiology Unit
University of Oxford
Old Road Campus
Headington
Oxford OX3 7LF

Tel: 01865 289721
Fax: 01865 289720
E-mail: carobb@npeu.ox.ac.uk
Website: www.npeu.ox.ac.uk/carobb/

CAROBB is funded by Public Health England.

Congenital Anomaly Register for Oxfordshire, Berkshire and Buckinghamshire

Information for parents

Every parent hopes that their baby will be healthy and most babies are.

However, a few babies do have problems (abnormalities) such as cleft palate, spina bifida, or Down's syndrome. These are sometimes called congenital anomalies or congenital malformations.

Some congenital anomalies are detected during pregnancy, some are found at birth, while others become apparent only as a baby grows older.

Why is information collected about babies with congenital anomalies?

CAROBB collects information:

- To increase our understanding of congenital anomalies and help research into their causes, treatment and prevention.
- To monitor how good antenatal screening tests (serum screening and ultrasound scans) are at picking-up problems.
- To look at trends - for example changes in the number of babies born with congenital anomalies, or changes in the pattern of where they are born.
- To give health professionals information to help them advise families about their chances of having a baby with a congenital anomaly.
- To help plan and develop NHS services.

What is CAROBB?

***CAROBB* is a database of information on babies born with suspected or confirmed congenital anomalies. It is part of, and contributes to, the British Isles Network of Congenital Anomalies (BINOCAR)**
www.binocar.org

What information is collected?

Information held by the register includes:

- Descriptions of each anomaly.
- Details and results of any investigations carried out during pregnancy (for example, the results of any ultrasound scans).
- Possible risk factors in the pregnancy including consanguinity.
- Details about mother and baby including names and dates of birth.

Who sees the information?

There are very strict regulations controlling access to personal information - that is names and addresses. This information will only be available to members of hospital staff treating you or your baby, and to those who work on CAROBB.

Information is sent to BINOCAR and also the European Surveillance of Congenital Anomalies, which collects information for many countries in Europe. When this happens no identifiable data are sent.

Information that is used by researchers or published in reports does not contain anything to identify either mother or baby, such as names and addresses.

Can I see the records on the Register?

Yes - you have the right to request a copy of the information held on you or your baby.

To do this, please make your wishes known to a member of your healthcare team or contact CAROBB by telephone or e-mail.

Appendix 7: List of Congenital Anomalies for Exclusion

	Specified ICD10-BPA – if present
Head	
Aberrant scalp hair patterning	
Flat occiput	
Dolichocephaly	Q67.2
Plagiocephaly – head asymmetry	Q67.3
Bony occipital spur	
Third fontanel	
Macrocephalus	Q75.3
Facial asymmetry	Q67.0
Compression facies	Q67.1
Other cong deformities of skull, face and jaw	Q67.4
Eyes	
Epicanthic folds	
Epicenthus inversus	
Upward slanting palpebral fissures	
Downward slanting palpebral fissures	
Short palpebral fissures	
Congenital ectropion	Q10.1
Congenital entropion	Q10.2
Other congenital malformation of eyelid	Q10.3
Dystopia canthorum	
Hypertelorism	Q75.2
Hypotelorism	
Stenosis of stricture of lacrimal duct	Q10.5
Synophrys	Q18.80
Blue sclera	Q13.5
Crocodile tears	Q07.82
Ears	
Primitive shape	Q17.3
Lack of helical fold	Q17.3
Asymmetric size	Q17.3
Posterior angulation	Q17.3
Microtia	Q17.2
Macrotia	Q17.1
Protuberant ears	Q17.3
Absent tragus	
Double lobule	Q17.0
Accesorry auricle, preauricular appendage, tag or lobule	Q17.0
Auricular pit	
Preauricular sinus or cyst	Q18.1
Narrow external auditory meatus	
Low set ears	Q17.4
Bat ear, prominent ear	Q17.4
Unspecified and minor malformation of ear	Q17.9

Nose	
Small nares	
Notched alas	
Oral regions	
Borderline small mandible/ minor micrognathia	
Aberrant frenula	
Enamel hypoplasia	
Malformed teeth	
High arched palate	Q38.50
Tongue tie or cyst of tongue	Q38.1
Macroglossia	Q38.2
Macrostomia	Q18.4
Microstomia	Q18.5
Macrocheilia	Q18.6
Microcheilia	Q18.7
Ranula	
Neck	
Mild webbed neck	
Sinus, fistula or cyst of branchial cleft	Q18.0
Preauricular sinus or cyst	Q18.1
Other branchial cleft malformation	Q18.2
Congenital malformation of face and neck, unspecified	Q18.9
Torticollis	Q68.0
Hands	
Duplication of thumbnail	
Enlarged or hypertrophic nails	Q84.5
Single/abnormal palmar crease	Q82.80
Unusual dermatoglyphics	
Clinodactyly (5th finger)	
Short fingers (4. 5. th finger)	
Accessorry carpal bones	Q74.00
Feet -Limb	
Syndactyly (2nd-3rd toes)	
Gap between toes (1st-2nd)	
Short great toe	
Recessed toes (4th, 5th)	
Enlarged or hypertrophic nails	Q84.5
Prominent calcaneus	
Clicking hip, subluxation of unstable hip	Q65.3-Q65.6
Metatarsus varus or metatarsus adductus	Q66.2
Hallux varus – other cong varus deformities of feet	Q66.3
Talipes or pes calcaneovalgus	Q66.4
Congenital pes planus	Q66.5
Metatarsus varus – other cong valgus deformities of feet	Q66.6
Pes cavus	Q66.7

Clubfoot of postural origin – other cong deformities of feet	Q66.8
Congenital deformity of feet, unspecified	Q66.9
Skin	
Hemangioma (other than face or neck)	
Pigmented naevus – cong non-neoplastic naevus	Q82.5
Neavus flammeus	Q82.50
Strawberry naevus	Q82.51
Lymphangioma	
Angioma	
Persistent lanugo	
Mongoloid spot (whites)	Q82.52
Depigmented spot	
Unusual placement of nipples	
Accessory nipples	Q83.3
Cafe-au-lait spot	
Skeletal	
Cubitus valgus	
Prominent sternum	Q67.7
Depressed sternum	Q67.6
Sternum bifidum	Q76.71
Shieldlike chest, other cong deformities of chest	Q67.8
Congenital deformity of spine	Q67.5
Genua valgum	
Genus varum	
Genu recurvatum	Q68.21
Congenital bowing of femur	Q68.3
Congenital bowing of fibula and tibia	Q68.4
Congenital bowing of long bones of leg, unspecified	Q68.5
Spina bifida occulta	Q76.0
Sacral dimple	
Cervical rib	Q76.5
Absence of rib	Q76.60
Accessory rib	Q76.62
Congenital lordosis, postural	Q76.43
Brain	
Arachnoid cyst	
Choroid plexus cyst	
Anomalies of septum pellucidum	
Cardiovascular	
Absence or hypoplasia of umbilical artery, single umbilical artery	Q27.0
Functional or unspecified cardiac murmur	
Patent ductus arteriosus if GA < 37 weeks	Q25.0 if GA <37 weeks
Peripheral pulmonary artery stenosis	
Patent or persistent foramen ovale	Q21.11

Pulmonary	
Accessory lobe of lung	Q33.1
Congenital laryngeal stridor	Q31.4
Laryngomalacia	Q31.4, Q31.5
Tracheomalacia	Q32.0
Azygos lobe of lung	Q33.10
Gastro-intestinal	
Hiatus hernia	Q40.1
Pyloric stenosis	Q40.0
Diastasis recti	
Umbilical hernia	
Inguinal hernia	
Meckel's diverticulum	Q43.0
Functional gastro-intestinal disorders	Q40.21, Q43.20, Q43.81, Q43.82
Transient choledochal cyst	
Anterior anus	
Renal	
Vesico-ureteral-renal reflux	Q62.7
Hydronephrosis with a pelvis dilatation less than 10 mm	
Hyperplastic and giant kidney	Q63.3
Single renal cyst	Q61.0
External genitals	
Deficient or hooded foreskin	
Undescended testicle	Q53
Unspecified ectopic testis	
Retractile testis	Q55.20
Hydrocele of testis	
Phymosis	
Bifid scrotum	Q55.21
Curvature of penis lateral	
Hypoplasia of penis	
Hymen imperforatum	Q52.3
Fusion of labia	Q52.5
Prominent labia minora	
Enlarged clitoris	
Vaginal skin tag	
Cysts of vulva	
Transient ovarian cyst	
Other	
Congenital malformation, unspecified	Q89.9
Chromosomal	
Balanced translocations or inversions in normal individuals	Q95.0, Q95.1

“Non-congenital” anomalies

Pyloric stenosis – there is controversy about the congenital nature of the majority of cases.

Patent ductus arteriosus in babies <37 weeks

Hydrocephaly where a result of preterm birth rather than congenital: all cases among preterm births should be thoroughly checked before registration.

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 babies 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 this is no further specification of whether malformation or postural origin

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