



Public Health
England

Protecting and improving the nation's health

National Congenital Anomaly and Rare Disease Registration Service

Congenital anomaly statistics 2016

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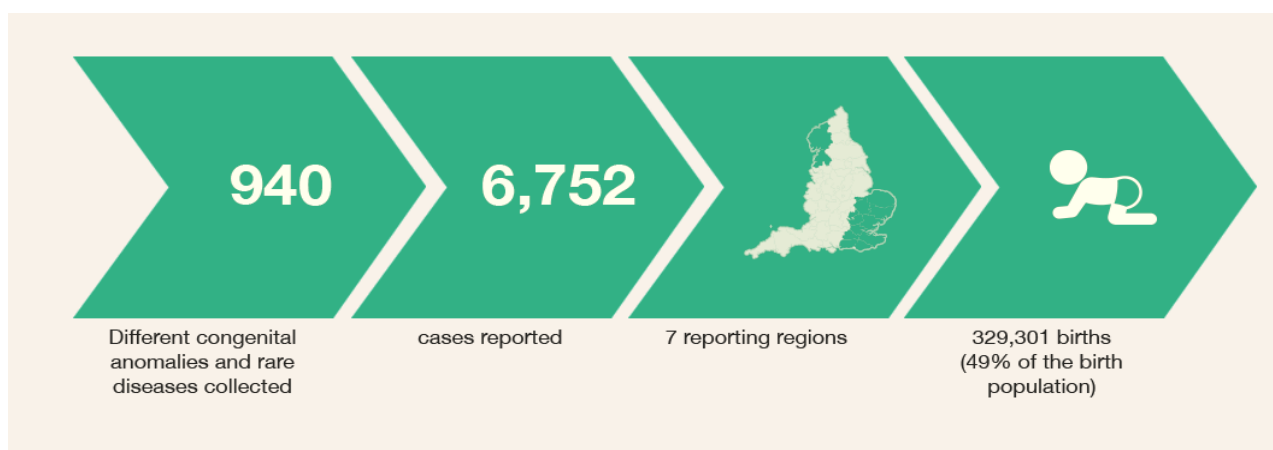
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Executive summary

This is the second National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) annual report. Public Health England launched NCARDRS on 1 April 2015. Prior to this, registries existed in only some regions of England. In response to the UK Rare Disease Strategy and the Chief Medical Officer's recommendation to ensure national coverage, three new regions covering the East of England, London and the South East, and the North West were established. Data collection in the newly established regions started from 1 April 2017.

The first annual report, published in 2015, reported on 21% of births in England. In this 2016 report, data from seven NCARDRS reporting regions, representing 49% coverage of births is presented; an increase that marks an important step towards our ambition to report national coverage.



NCARDRS currently collects data on 940 different congenital anomalies and rare diseases. In 2016, there were a total of 6,752 cases with one or more congenital anomalies notified to the 7 NCARDRS reporting regions covering 329,301 total births (live births and stillbirths). This gives an overall birth prevalence for these regions of 205 per 10,000 total births – or 1 in 49.

Some congenital anomalies are detectable during pregnancy and others are not. In 2016, the timing of first diagnosis was known for 6,408 (94.9%) cases and of these, 64.9% were diagnosed antenatally. Where a congenital anomaly was detected antenatally 57.6% of cases resulted in a live birth and where a congenital anomaly was diagnosed postnatally 95.0% were diagnosed following a live birth.

Of the 6,752 congenital anomaly cases reported to NCARDRS, 336 resulted in an infant death, giving an infant mortality rate of 10.2 per 10,000 live births. Congenital

heart anomalies accounted for 47% of infant deaths, followed by chromosomal anomalies (25%) and digestive system anomalies (18%).

Data recorded in 2016 shows that the prevalence of genetic congenital anomalies increased with maternal age; the prevalence of these anomalies was 7 times higher in older mothers (women aged 40+ years) compared with younger mothers (women aged less than 20 years).

The data collected for the period 2015 to 2016, representing 33.8% coverage of NHS trusts providing antenatal services in England, indicates that detection targets were met for 6 of the 11 structural anomalies auditable under the NHS Fetal Anomaly Screening Programme. Where the target was not met, this was statistically significant only in the case of Edwards' syndrome. This was based on a small number of cases. Robust reported detection rates will be available for 2017 to 2018 data in 2019.

This second annual report has demonstrated improvements over the past year towards achieving national data collection to enable robust information about the prevalence of congenital anomalies. NCARDRS looks forward to being able to present, for the first time in England, national coverage of congenital anomalies from 1 April 2017 and will be able to report on this data from 2019.

Chapter 1: Introduction

This report is the National Congenital Anomaly and Rare Disease Registration Service's (NCARDRS) second annual report. It primarily describes the number of cases with 1 or more congenital anomalies delivered between 1 January and 31 December 2016.

Readers who are interested in cases prior to 2016 are directed to our [first report](#), published in July 2017, which reports on data from 2015 and cites relevant sources of information for historical data collected before the inception of NCARDRS.

Public Health England (PHE) launched NCARDRS on 1 April 2015. Prior to this, registries existed in some regions of England, and they reported data for the benefit of clinicians, epidemiologists, researchers and patients. In response to the [UK Rare Disease Strategy](#) and the Chief Medical Officer's recommendation to ensure national coverage, 3 new regions covering the East of England, London and the South East, and the North West were established. Data collection in new regions started from 1 April 2017. This means, for the first time, national coverage of congenital anomaly reporting will be possible in 2019. Information about the current level of coverage presented in this report is explained below.

This report is intended primarily as a useful resource for commissioners and providers of healthcare needed for the diagnosis and management of congenital anomalies. It also provides high quality data for the use of researchers and those seeking detailed information about congenital anomaly prevalence in England. It is hoped that this annual public report will be of interest to healthcare workers involved in the direct care of patients.

This report describes the congenital anomaly data collected by NCARDRS and provides context for the detailed reports provided to each notifying trust about case ascertainment of the 11 structural anomalies (which include Edwards' syndrome and Patau's syndrome) and Down's syndrome screened for, under the NHS Fetal Anomaly Screening Programme (FASP). A short summary document, which draws on the key messages from this report, is also available.

The ambition to provide a comprehensive national register relies on the commitment of healthcare professionals across the country to report cases to NCARDRS. The multiple source approach to data collection in NCARDRS relies on the dedication of healthcare staff in a range of settings including maternity units, neonatal units, diagnostic departments, genetic laboratories and many more. This multiple source approach enables high ascertainment and completeness of cases and ensures consistency and standardisation across the country.

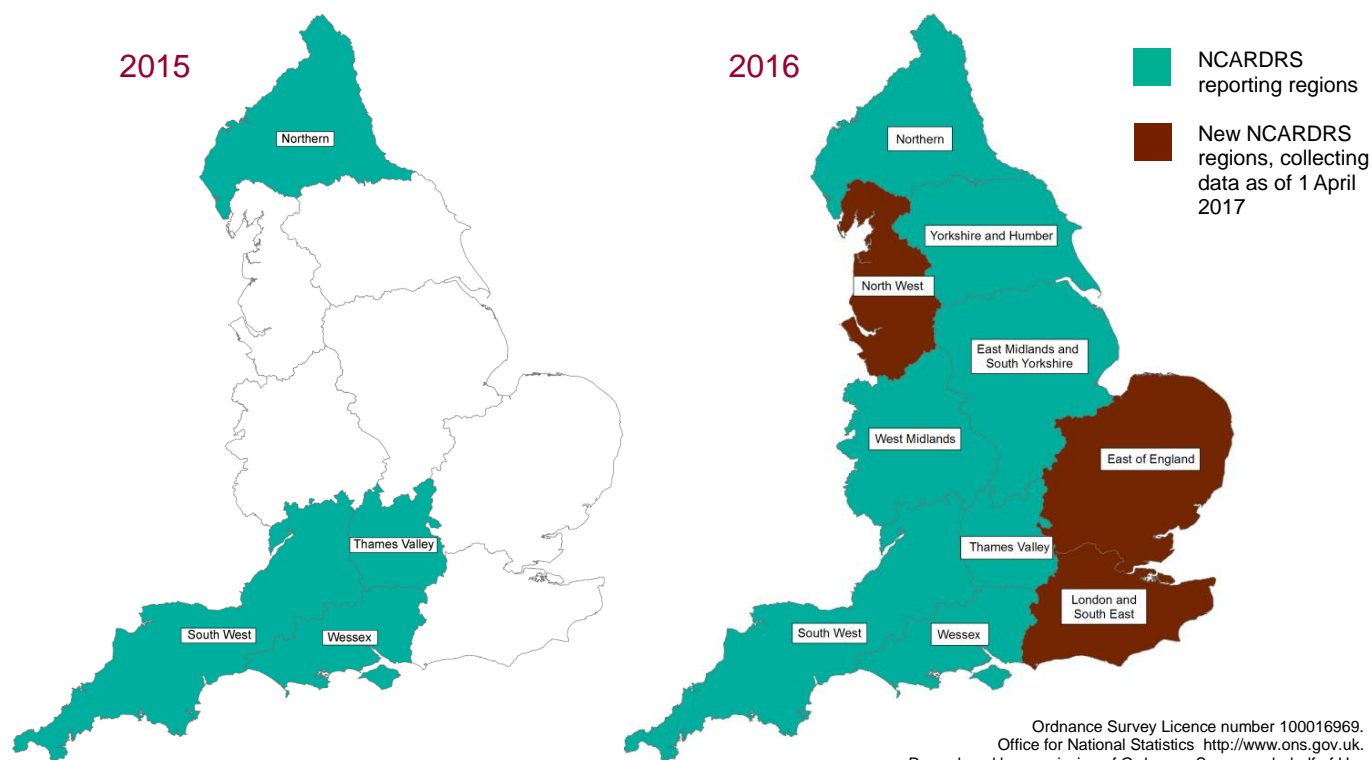
Thanks to the dedication of notifying healthcare staff, important and reliable information is available for clinicians, researchers, patients and their families. It is hoped this report encourages notifying trusts to continue to work with NCARDRS to realise their ambition. More information about the data collection process can be found in the accompanying technical details document.

1.1 NCARDRS reporting regions in 2016

NCARDRS is made up of 10 reporting regions and the information presented in this document reports on data for 7 of those regions (Figure 1). This is a significant improvement compared to last year's report where 2015 data was sourced from 4 regions (Northern, South West, Thames Valley and Wessex). The newly included 3 regions for 2016 data are East Midlands and South Yorkshire, West Midlands, and Yorkshire and Humber. See the technical details document for more information about geographical coverage of NCARDRS regions.

In 2015, 21% coverage of births was reported; the addition of three more regions for 2016 takes coverage to 49% of births. This increase in coverage marks an important step towards the ambition for NCARDRS to report national coverage with data collected starting as of 1 April 2017. This means, for the first time, national coverage of congenital anomaly reporting will be possible from 2019.

Figure 1: Map of NCARDRS reporting regions England, 2015 and 2016



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1.2 Data in this report

Congenital anomalies are defined as being present at delivery, probably originating before birth, and include structural, chromosomal, genetic and biochemical malformations.¹ Data is collected according to definitions and guidelines of European Surveillance of Congenital Anomalies (EUROCAT), the World Health Organization (WHO) Collaborating Centre for the Surveillance of Congenital Anomalies. Congenital anomalies are coded using the International Classification of Disease version 10 (ICD-10) with British Paediatric Association (BPA) extension, which gives supplementary 1 digit extensions to ICD-10 codes to allow greater specificity of coding. For more information about data collection, definitions and coding see the technical details document which accompanies this report.

The 2015 report included a focus on Down's syndrome, Edwards' syndrome and Patau's syndrome for cases reported in England up to 2014. NCARDRS plans to release a separate publication to include data for these syndromes up to the 2016 reporting year.

In this report, comparisons have been intentionally not made between 2015 and 2016 data. This is because:

- data are not directly comparable as regional coverage varies between years
- comparing year on year data in any case could lead to unreliable conclusions
- as a minimum, 3 years' worth of data are required to consider trend analysis

This 2016 report is organised in 4 further chapters covering information about:

- prevalence of congenital anomalies (chapter 2)
- timing of diagnosis and outcome (chapter 3)
- key public health indicators (chapter 4)
- the Fetal Anomaly Screening Programme (FASP) (chapter 5)

Information about the prevalence of congenital anomalies in chapter 2 outlines the types of anomaly most frequently reported to NCARDRS.

Chapter 3 describes the timing of diagnosis and the outcome of pregnancy. This relays important information about the number of babies born with congenital anomalies, some of which may need ongoing health and social care service provision.

¹ EUROCAT SYNDROME GUIDE: Definition and Coding of Syndromes (Revised 2017), available from: www.eurocat-network.eu/aboutus/datacollection/guidelinesforregistration/malformationcodingguides

Currently available public health information is the focus of chapter 4. This includes estimates about the contribution congenital anomalies make to perinatal and infant mortality rates as well as information about how prevalence varies by maternal age.

Chapter 5 provides an indication of national detection rates² for auditable conditions under the NHS Fetal Anomaly Screening Programme. Information in this chapter relates to cases reported by 43 NHS trusts, representing 33.8% coverage of NHS trusts providing antenatal services in England.

Information presented in chapters 2 to 4 relates to cases where one or more congenital anomalies were detected for deliveries between 1 January and 31 December 2016. Chapter 5, described above, reports on a separate cohort of cases, with expected dates of delivery between 1 April 2015 and 31 March 2016.

The final chapter of this report celebrates some of the success of the reporting year and outlines plans to move towards national coverage of congenital anomalies and expand our work to report on the prevalence of rare diseases. The aim of NCARDRS is to improve data collection across the service to be able to report on a range of indicators to improve knowledge about possible risk factors, or protective factors, affecting the prevalence of congenital anomalies and rare diseases.

A separate summary document, highlighting key messages about the prevalence of congenital anomalies, accompanies this report. A glossary and technical detail document, and detailed data tables also accompany this report.

² Detection rate: the proportion of affected individuals with a positive screening result.

Chapter 2: Prevalence of congenital anomalies

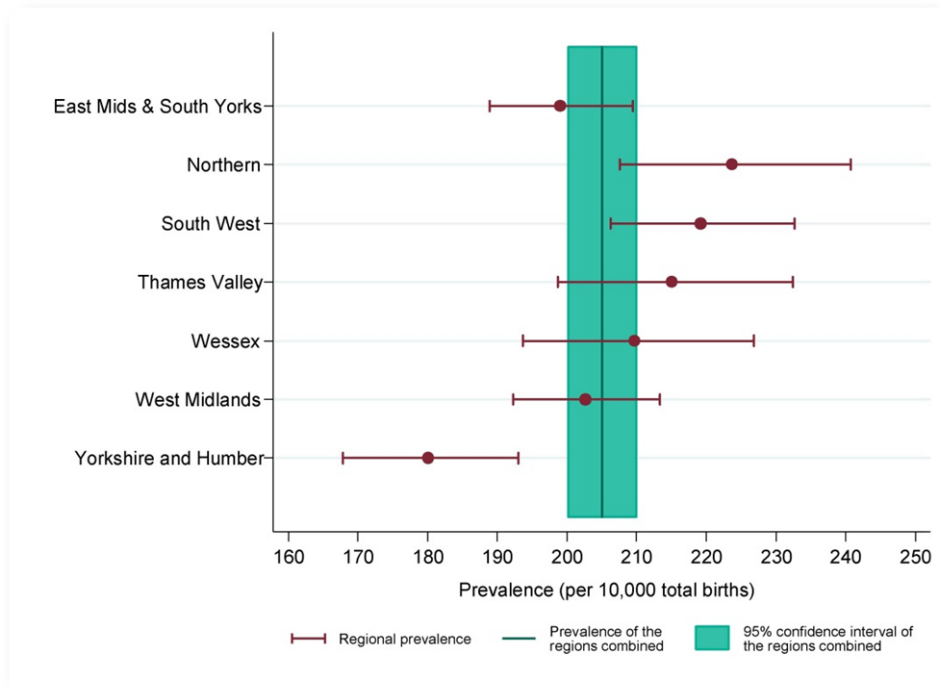
In 2016, there were a total of 6,752 cases with one or more congenital anomalies notified to the 7 NCARDRS reporting regions covering 329,301 total births (live births and stillbirths). This gives an overall birth prevalence for these regions of 205 per 10,000 total births (95% CI: 200.2-210.0) or 1 in 49 births (Table 1).

The birth prevalence of congenital anomalies by reporting region (Figure 2, Table 2) was not statistically significantly different from the combined result for the majority of reporting regions. The one exception was the Yorkshire and Humber region (180.1, 95% CI 167.9 - 193.0) where the prevalence was statistically significantly lower than the combined result for all reporting regions. This is the first year that the Yorkshire and Humber data has been included in national reporting, and there are still some improvements in case ascertainment needed in the region.

The data for Yorkshire and Humber was reviewed by the EUROCAT committee and accepted as being of high enough quality to be reportable, although the committee identified weaknesses in the reporting of less severe anomalies that are affecting overall prevalence in the region. NCARDRS is working with clinicians to improve notification, in particular, of less severe cardiac anomalies not requiring surgery and urinary anomalies such as hydronephrosis and hypospadias. There was no significant under reporting of the more severe anomalies identified. The overall birth prevalence for the NCARDRS reporting regions excluding Yorkshire and Humber data is 208.9 (95% CI 203.7 – 214.3) (Table 2).

Possible reasons for geographical variations could include disease clustering, exposure to teratogens, demographic variation including age and deprivation profiles between regions and also the genetic composition of the local population. As NCARDRS moves to national coverage and the collection of longitudinal data, greater insights will be gained into underlying population characteristics contributing to regional variation, as well as the ability to analyse associations with environmental factors which may potentially be modifiable.

Figure 2: Prevalence (per 10,000 total births) and 95% confidence intervals for NCARDRS reporting regions compared to regions combined, 2016



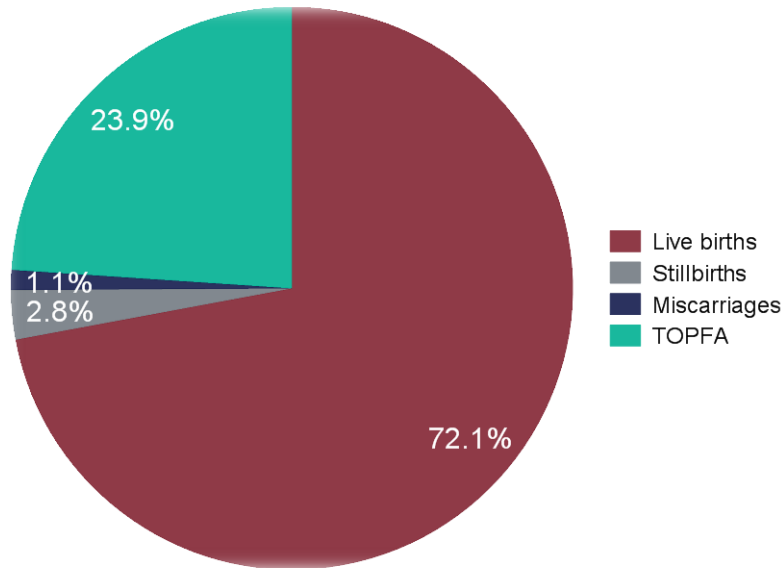
In the 2016 reporting year, congenital anomaly prevalence in England was statistically significantly lower than that reported by EUROCAT for European registries; 205 (95% CI 200.2 – 210.0) compared to 271.2 (95% CI 265.3 – 272.2) (Table 2). The breakdown by anomaly subgroup shows that prevalence in England was statistically significantly lower compared to European registries in 4 of these groups: congenital heart defects (CHD), urinary, genital and limb. However, more specific data within these subgroups shows that prevalence was not statistically significantly different for severe anomalies. For example, severe CHD, bilateral renal agenesis and limb reduction defects were not statistically significantly different. This suggests that the lower prevalence of congenital anomalies in England compared to European registries is likely due to lower ascertainment in England of less severe anomalies. Less severe anomalies are often not detectable at screening, not obvious at birth or do not require surgery or treatment shortly after birth. This means that these anomalies are less likely to be recorded and notified to NCARDRS – an area which the service is seeking to improve upon.

Figure 3 shows that of the 6,752 cases with one or more congenital anomalies, 4,867 (72.1%) were live births, 191 (2.8%) were stillbirths (24+ weeks’ gestation), 77 (1.1%) were late miscarriages (20-23 weeks’ gestation) and 1,617 (23.9%) were terminations of pregnancy with fetal anomaly (TOPFA).³ This includes terminations of pregnancy with

³ Where a pregnancy ends in a TOPFA, the baby may be born dead, or if parents have not opted for prior feticide the baby may be born alive but die shortly after. If the gestation at which a TOPFA takes place is 24 weeks or later, the civil birth registration must be recorded as a stillbirth. For the purposes of this report we record these as TOPFAs.

fetal anomaly as well as terminations of pregnancy for other reasons where a fetal anomaly was present. The data presented relate to both antenatal and postnatal diagnoses. The timing of diagnosis is explored in more detail in chapter 3.

Figure 3: Percentage of cases with one or more congenital anomalies by outcome of pregnancy in 7 NCARDRS reporting regions, 2016



TOPFA = Terminations of pregnancy with fetal anomaly

Figure 4a and 4b show the prevalence of the 12 congenital anomaly subgroups for all cases and for those cases resulting in a live birth. In 2016, the prevalence for total births (Figure 4a) was highest in the congenital heart anomalies subgroup (65.9 per 10,000, 95% CI 63.1 – 68.7), followed by those that are chromosomal in origin (49.9 per 10,000, 95% CI 47.5 – 52.4) (Table 1). The prevalence for live births (Figure 4b) was also highest in congenital heart anomalies (55.3 per 100,000, 95% CI 52.7 – 57.9), follow by those that are chromosomal in origin (22.4 per 10,000, 95% CI 20.8 – 24.1) (Table 3).

The pattern for all cases and live births is similar for the majority of subgroups apart from chromosomal and nervous system anomalies where prevalence is lower for cases resulting in a live birth than other outcomes. A more detailed breakdown by specific congenital anomaly, including the number of cases reported is available in Tables 1 and 3.

Figure 4a: Total birth prevalence (per 10,000 total births) by congenital anomaly subgroup in 7 NCARDRS reporting regions, 2016

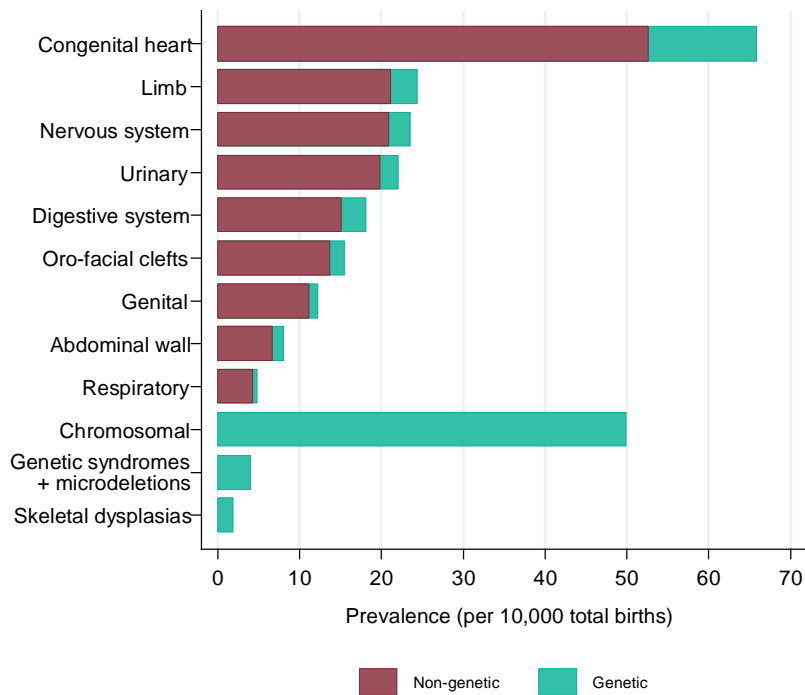
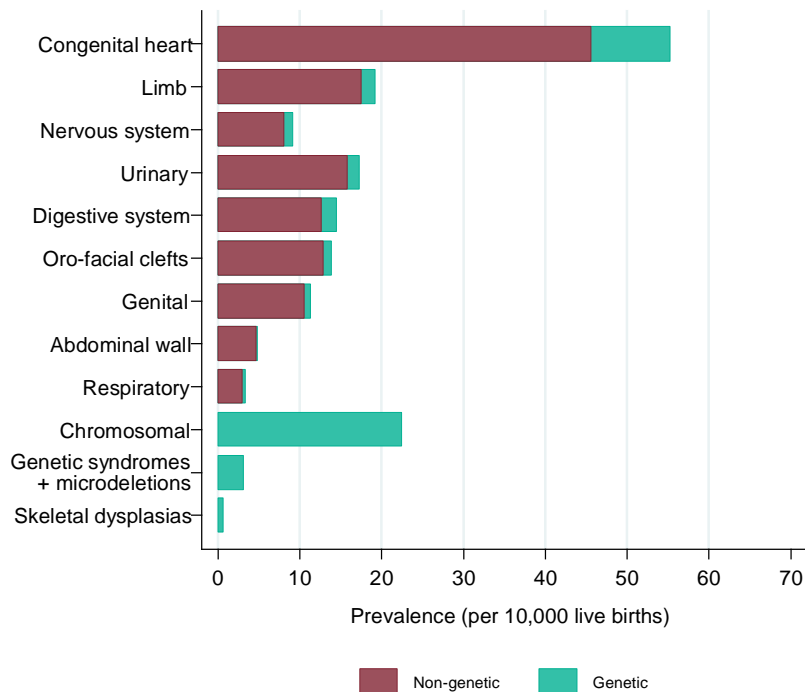


Figure 4b: Live birth prevalence (per 10,000 live births) by congenital anomaly subgroup in 7 NCARDRS reporting regions, 2016



Some of the cases shown in these figures will have more than one anomaly and appear in more than one bar. Genetic conditions include those cases with an identified chromosomal anomaly, skeletal dysplasia, genetic syndrome and/or microdeletion. Non-genetic conditions include those cases with one or more congenital anomaly with no identified anomalies that are chromosomal, skeletal dysplasias, genetic syndromes or microdeletions (not all cases undergo genetic testing and it is likely some of these are of genetic origin).

The approximate number of congenital anomaly cases in the whole of England in 2016 can be estimated by applying the birth prevalence estimate for the 7 NCARDRS reporting regions in 2016 to the total number of births in England in 2016 (666,050).

This extrapolation is an indication of prevalence and assumes that the birth prevalence was consistent across England. Currently, this is the best available method available to estimate national prevalence, though it should be noted that birth prevalence is likely to vary across England due to demographic differences. Using this method it is estimated that there were 13,657 registerable congenital anomalies in England in 2016 (Table 4). A breakdown by congenital anomaly subgroup is available in table 4.

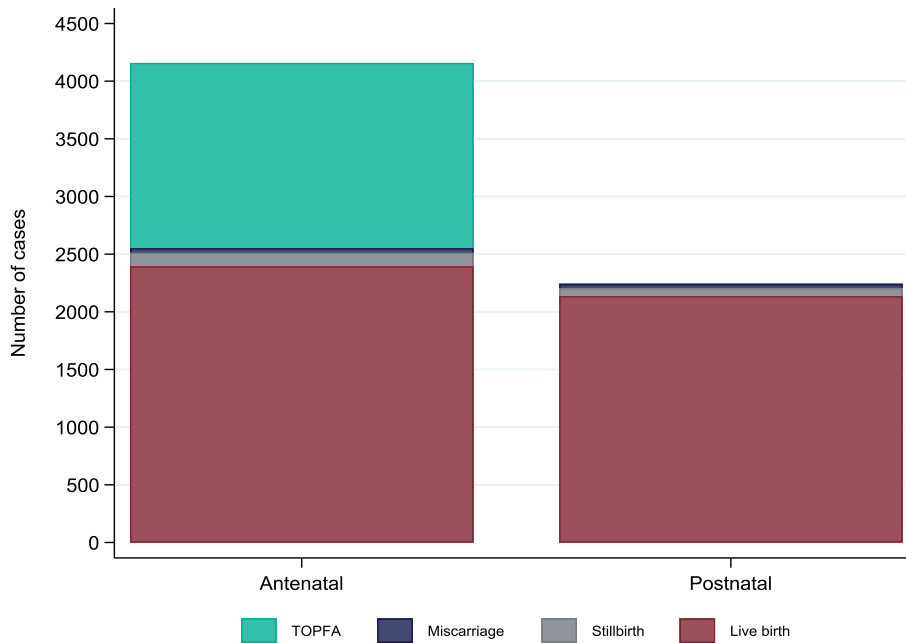
Chapter 3: Timing of diagnosis and outcome

Some congenital anomalies are detectable during pregnancy and others are not. Screening programmes are offered by NHS maternity services to maximise antenatal detection of specified conditions where women choose, and present in time to have screening. NCARDRS provides a separate audit of the Fetal Anomaly Screening Programme (FASP) to PHE and to individual NHS trusts to monitor the performance of this screening. Early diagnosis (as early as possible in the pregnancy) is preferable as it gives women and their partners greater choice about their pregnancy and enables better planning for the delivery of babies where specialist intervention may be required soon after birth. More detailed FASP data and detection rates can be found in chapter 5.

The timing of first diagnosis of a congenital anomaly was known for 6,408 (94.9%) cases. Of these 64.9% were diagnosed antenatally in 2016.

Figure 5 shows that of the 4,159 cases where a congenital anomaly was diagnosed antenatally, 1,608 (38.7%) resulted in termination of pregnancy with fetal anomaly (TOPFA) (Table 5). Where a congenital anomaly was detected antenatally 57.6% of cases resulted in a live birth and where a congenital anomaly was diagnosed postnatally 95.0% were diagnosed following a live birth (Table 5).

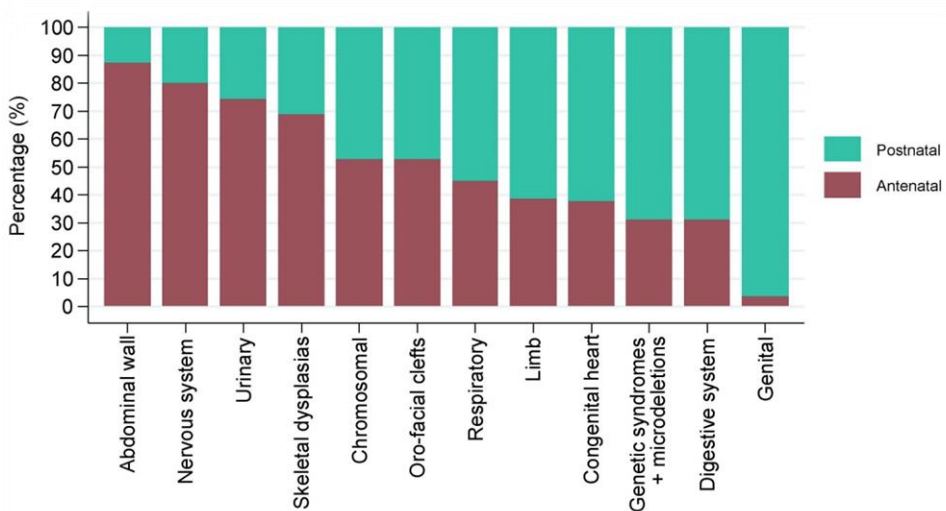
Figure 5: Timing of first diagnosis and pregnancy outcome in 7 NCARDRS reporting regions, 2016



TOPFA = Termination of pregnancy with fetal anomaly

Figure 6 shows that abdominal wall, nervous system and urinary anomalies are the conditions most frequently diagnosed antenatally. Genital and digestive system anomalies are the least frequently diagnosed antenatally. It should be noted that individual anomalies within these subgroups may not follow this pattern. A more detailed breakdown by specific congenital anomaly, including the number of cases reported is available in Table 6.

Figure 6: Timing of diagnosis by congenital anomaly subgroup (percentage) in 7 NCARDRS reporting regions, 2016

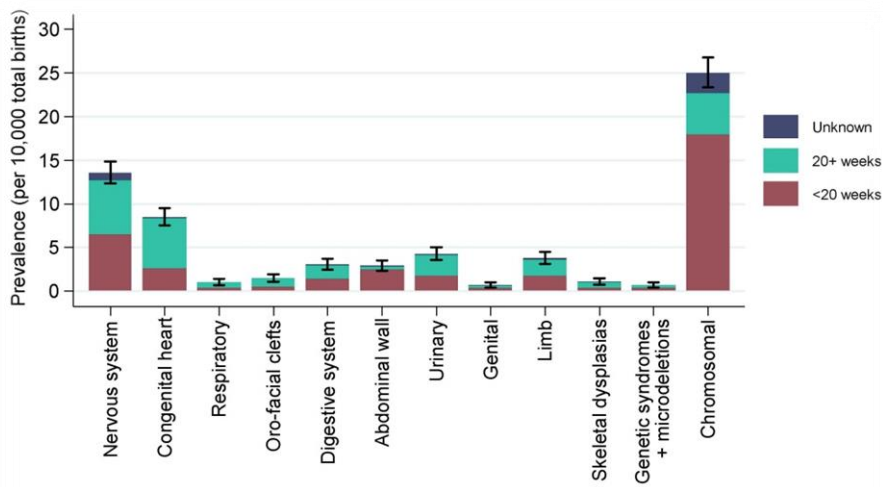


Some of the cases shown in this figure will have more than one anomaly and appear in more than one bar

The overall rate of TOPFA for the 7 NCARDRS reporting regions was 49.1 per 10,000 total births. The rate of TOPFA at over 20 weeks’ gestation was 17.4 per 10,000 total births (Table 7).

The data in Figure 7 (and Table 7) show the highest rate of TOPFA was associated with chromosomal anomalies (25.0 per 10,000 births). The majority of these cases resulted in TOPFA before 20 weeks’ gestation. This was also the case for abdominal wall anomalies, but not for any other major congenital anomaly subgroups. In the case of congenital heart anomalies the TOPFA rate is higher after 20 weeks gestation than before 20 weeks’ gestation. Where congenital heart anomalies are diagnosed antenatally they are often first suspected at the routine fetal anomaly scan, which takes place between 18 weeks and zero days’ and 20 weeks and 6 days’ gestation and require a referral to a tertiary service provider for confirmation.

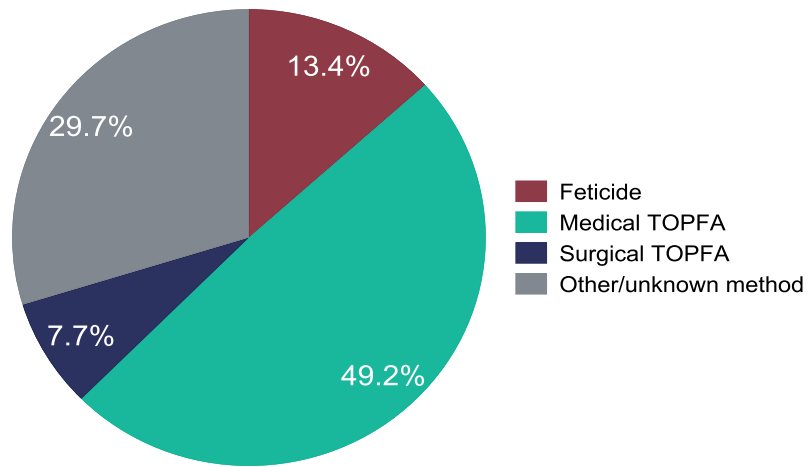
Figure 7: Prevalence (per 10,000 total births) and 95% confidence intervals of termination of pregnancy with fetal anomaly (TOPFA) in 7 NCARDRS reporting regions, 2016



Some of the cases shown in this figure will have more than one anomaly and appear in more than one bar, so may not have had the TOPFA for the anomaly shown. More information about the anomalies included in these sub-groups is available in the technical detail document.

The type of TOPFA was of other or unknown method in 481 (29.7%) cases (Table 8). Figure 8 shows that where type of TOPFA was known, most were medical TOPFAs (49.2%) or feticide (13.4%). NCARDRS is working to improve the quality of the type of TOPFA data for 2017.

Figure 8: Type of termination of pregnancy (percentage) in 7 NCARDRS reporting regions, 2016



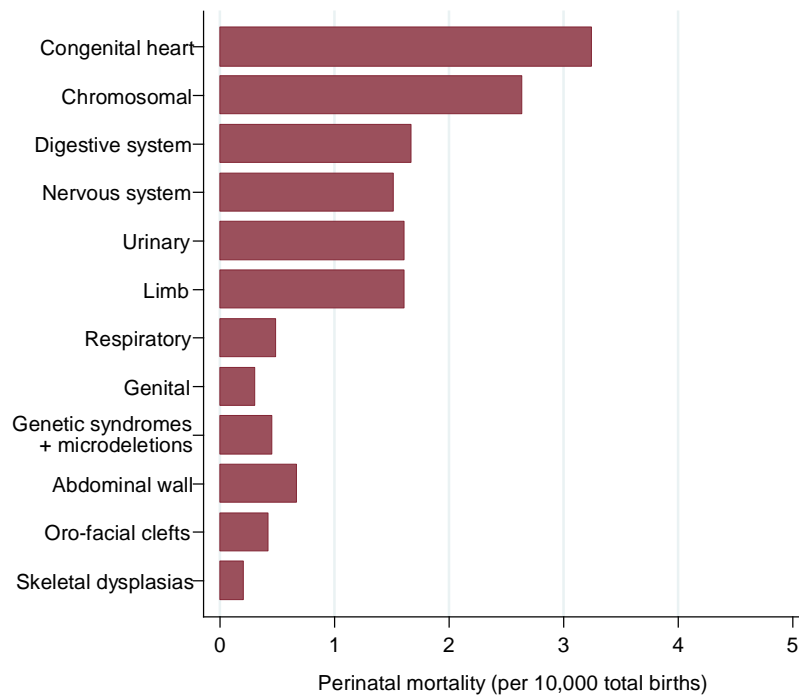
TOPFA = Termination of pregnancy with fetal anomaly

Chapter 4: Key public health indicators

4.1 Perinatal and infant mortality

There were 336 infant deaths with 1 or more congenital anomaly in the 327,900 live births in 2016, giving an infant mortality rate of 10.2 per 10,000 live births. Congenital anomalies accounted for approximately 25.2% of all infant deaths.⁴ The rate of perinatal mortality was similar, at 10.8 per 10,000 live births (Table 9). Figures 9a and 9b show that congenital heart anomalies (4.8, 3.2 per 10,000 births respectively) had the highest infant and perinatal mortality rates, followed by chromosomal anomalies (2.5, 2.7 per 10,000 births respectively) and digestive anomalies (1.8, 1.7 per 10,000 births respectively).

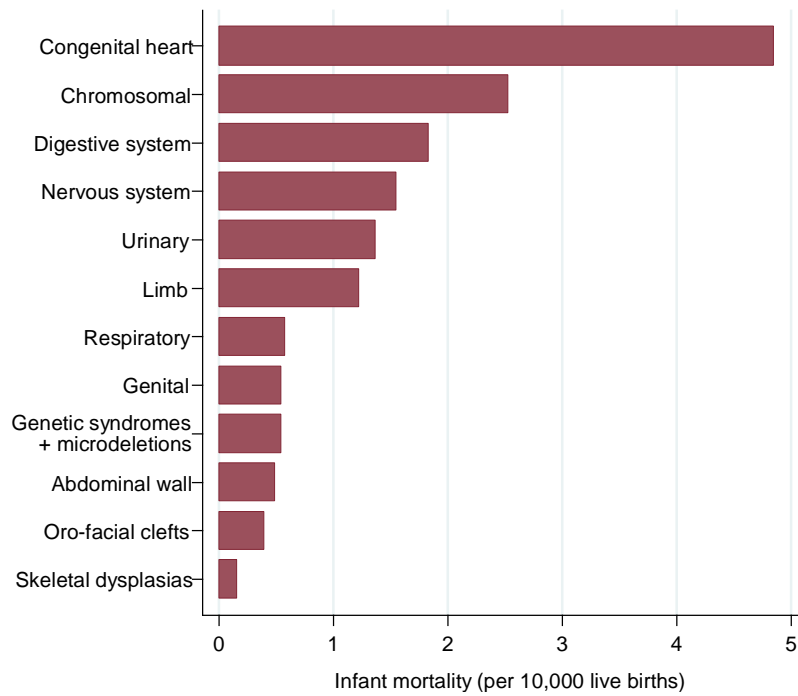
Figure 9a: Perinatal mortality by congenital anomaly subgroup in 7 NCARDS reporting regions, 2016



Some of the cases shown in this figure will have more than 1 anomaly and appear in more than one bar. Caution should be taken when interpreting this data as the cause of death for cases may not have been due to the congenital anomaly.

⁴ This is the percentage of infant deaths with a congenital anomaly out of all infant deaths in the 7 NCARDS reporting regions in 2016. All infant deaths = ONS - Source: Office for National Statistics licensed under the Open Government Licence.

Figure 9b: Infant mortality by congenital anomaly subgroup in seven NCARDRS reporting regions, 2016



Some of the cases shown in this figure will have more than one anomaly and appear in more than one bar. Caution should be taken when interpreting these data as the cause of death for cases may not have been due to the congenital anomaly.

4.2 Maternal age

All anomalies

Mothers aged between 25 and 29 years at delivery had the lowest birth prevalence of all anomalies (176.9 per 10,000 total births, Table 10). Compared to that group, the birth prevalence was significantly higher in the less than 20 years age group (220.7 per 10,000 total births), 20-24 years (197.6 per 10,000) and in the 35-39 years and 40 years and over age groups (247.8 per 10,000 total births and 431.7 per 10,000 total births respectively). Mothers in the 30-34 years age group (183.1 per 10,000 births) had a broadly similar rate to the 25-29 years age group.

Non-genetic anomalies

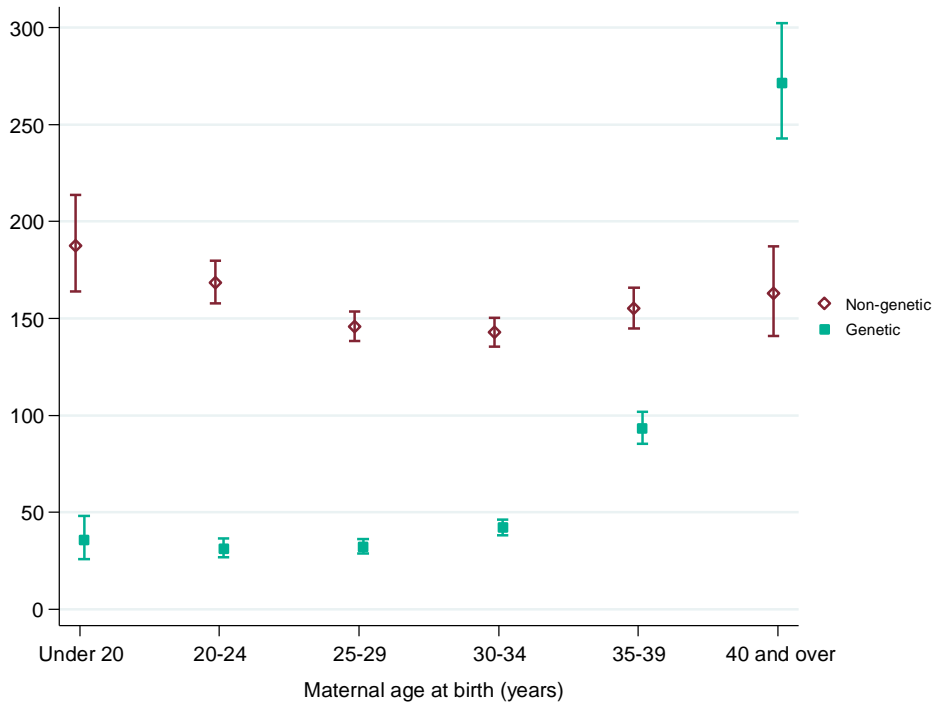
The birth prevalence of non-genetic⁵ anomalies (anomalies without evidence of a genetic cause) also varied by maternal age (Figure 10, Table 10). Younger mothers (less than 20 years) had the highest prevalence of non-genetic anomalies, and those in the 30-34 years age group had the lowest prevalence. The higher rate of non-genetic anomalies among women in the less than 20 years age group appears to be driven by abdominal wall anomalies. In particular gastroschisis, which is known to be associated with lower maternal age and accounts for 70% of the abdominal wall defects. Abdominal wall anomalies were significantly more common than the average for the less than 20 years and 20-24 years age groups (Figure 11). Anomalies of the nervous system, urinary system and limbs tended to be higher in the less than 20 years age group but not significantly so.

Genetic congenital anomalies

It is known that Down's syndrome is more common in older mothers and this accounted for 45% of genetic congenital anomalies and therefore, the birth prevalence of genetic congenital anomalies was higher in older mothers (Figure 10). The birth prevalence was 7 [7.4; 95% CI: 5.4-10.2] times higher for the oldest (40+ years) mothers (n=330) compared with the youngest (less than 20 years) age group (N=43) (Table 10). The birth prevalence of genetic congenital anomalies in age categories under 30 years old was broadly similar to each other.

⁵ Non-genetic conditions include those cases with one or more congenital anomaly with no identified anomalies that are chromosomal, skeletal dysplasias, genetic syndromes or microdeletions (not all cases undergo genetic testing and it is likely some of these are of genetic origin).

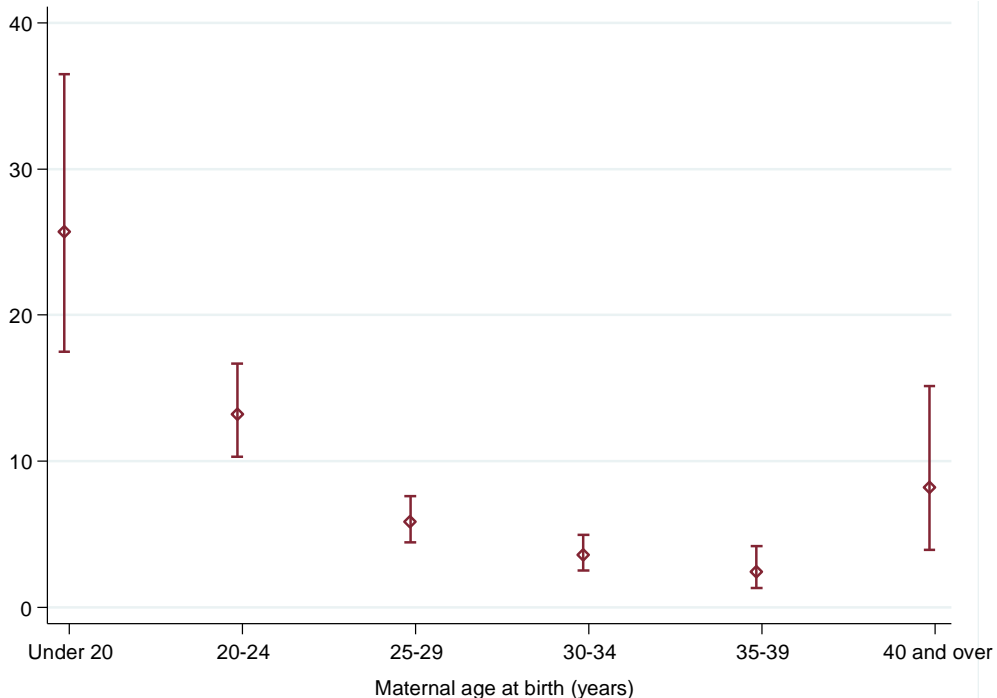
Figure 10: Prevalence (per 10,000 total births) and 95% confidence intervals of genetic and non-genetic congenital anomalies by maternal age in 7 NCARDRS reporting regions, 2016



Genetic conditions include those cases with an identified chromosomal anomaly, skeletal dysplasia, genetic syndrome and/or microdeletion.

Non-genetic conditions include those cases with one or more congenital anomaly with no identified anomalies that are chromosomal, skeletal dysplasias, genetic syndromes or microdeletions (not all cases undergo genetic testing and it is likely some of these are of genetic origin).

Figure 11: Prevalence (per 10,000 total births) and 95% confidence intervals of abdominal wall anomalies by maternal age in 7 NCARDRS reporting regions, 2016



Chapter 5: Fetal Anomaly Screening Programme

Data presented in this chapter provides detection rates for the 11 structural anomalies and Down's syndrome auditable under the NHS Fetal Anomaly Screening Programme (FASP). It is recognised that whilst antenatal screening should be offered to women and their partners where appropriate, whether tests are accepted or not is a personal choice.

Readers are advised to note the following data limitations:

- full national data coverage has not yet been achieved, so data presented here is purely indicative; no conclusions should therefore be drawn about national detection rate target performance (full coverage is expected for the 2017 to 2018 reporting year)
- work is underway to improve postnatal detection notifications; whilst this is mitigated by only including data assessed to be of high quality in terms of completeness, detection bias cannot be ruled out (more information about plans to achieve national coverage and improve postnatal notifications can be found in the final chapter of this report)
- data presented is based on small numbers and a single year of data; results are therefore subject to natural variation – the intention is to present long-term trend analysis data when available in the future
- data presented here relates to cases reported with an estimated date of delivery between 1 April 2015 and 31 March 2016; this is a different cohort to data presented in previous chapters (which reports on cases reported to 7 NCARDRS regions with 1 or more anomaly delivered between 1 January and 31 December 2016) – the inclusion criteria for both cohorts can be found in the accompanying technical detail document
- combined screening for Patau's syndrome and Edwards' syndrome had not yet been introduced in this reporting year
- the detection criteria for bilateral renal agenesis is currently being reviewed and the detection rates described here may be increased following that review

For the purposes of estimating detection rates, cases are included where there was no prior suspicion or diagnosis of the anomaly before the fetal anomaly scan, or women were booked on or before 19 weeks and 6 days' gestation and there was no documented evidence of fetal anomaly screening being declined. Cases are considered as 'detected' only where the scan has occurred within the gestational window specified by FASP and findings indicating the anomaly were reported.

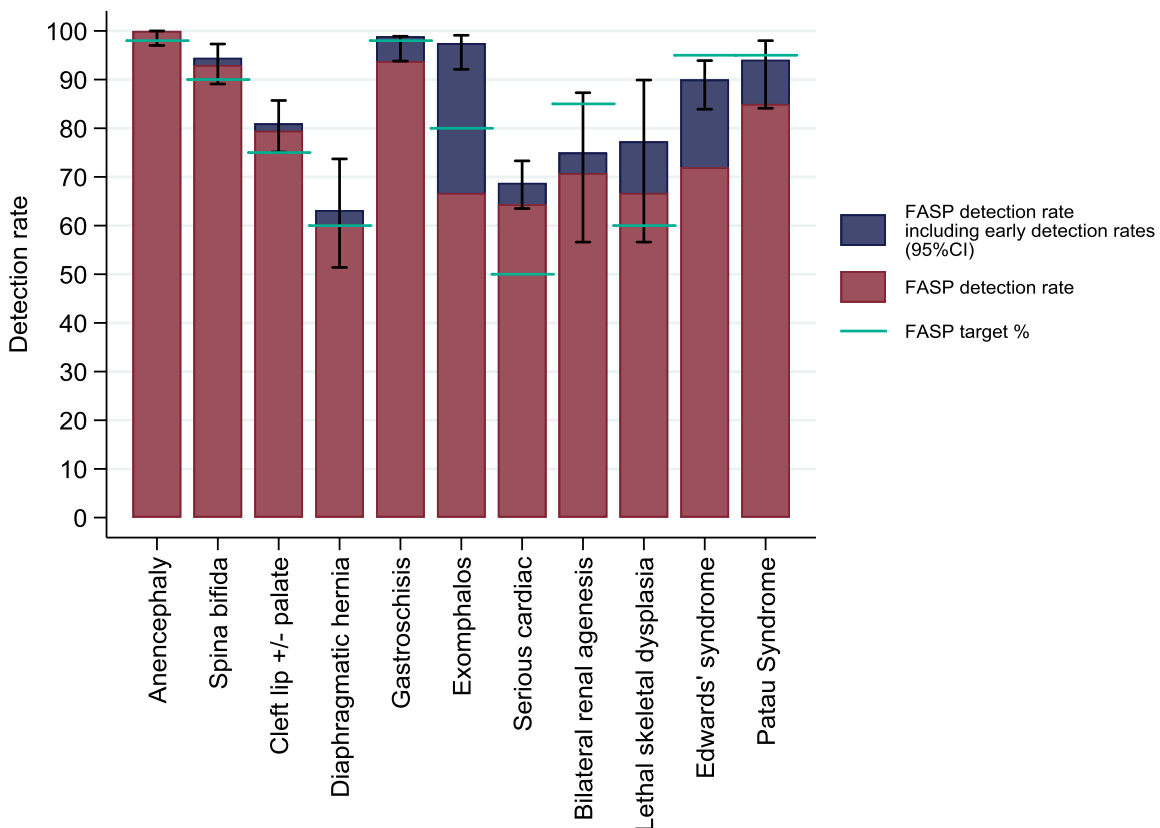
The national FASP detection rates presented here are sourced from data submitted to NCARDRS by 46 NHS trusts. This represents 33.8% coverage of NHS trusts providing antenatal services in England.

Figure 12 shows that of 46 reporting trusts in England, detection targets were met for 6 of the 11 structural anomalies. Where the target was not met, this was statistically significant only in the case of Edwards’ syndrome. This was based on a small number of cases (Table 11).

Detection rates were also calculated including cases where there was suspicion or diagnosis before the fetal anomaly scan. Early diagnosis enables earlier detailed investigations and can provide women with greater choice about their pregnancy.

For 5 of the 11 structural anomalies – anencephaly, gastroschisis, exomphalos, Patau’s syndrome, Edwards’ syndrome – the majority of cases that were detected were suspected/diagnosed at the first trimester scan. Figure 12 shows that, where early detections are included, targets were reached for 8 of the 11 conditions. Where the target was not met, this was statistically significant only in the case of Edwards’ syndrome

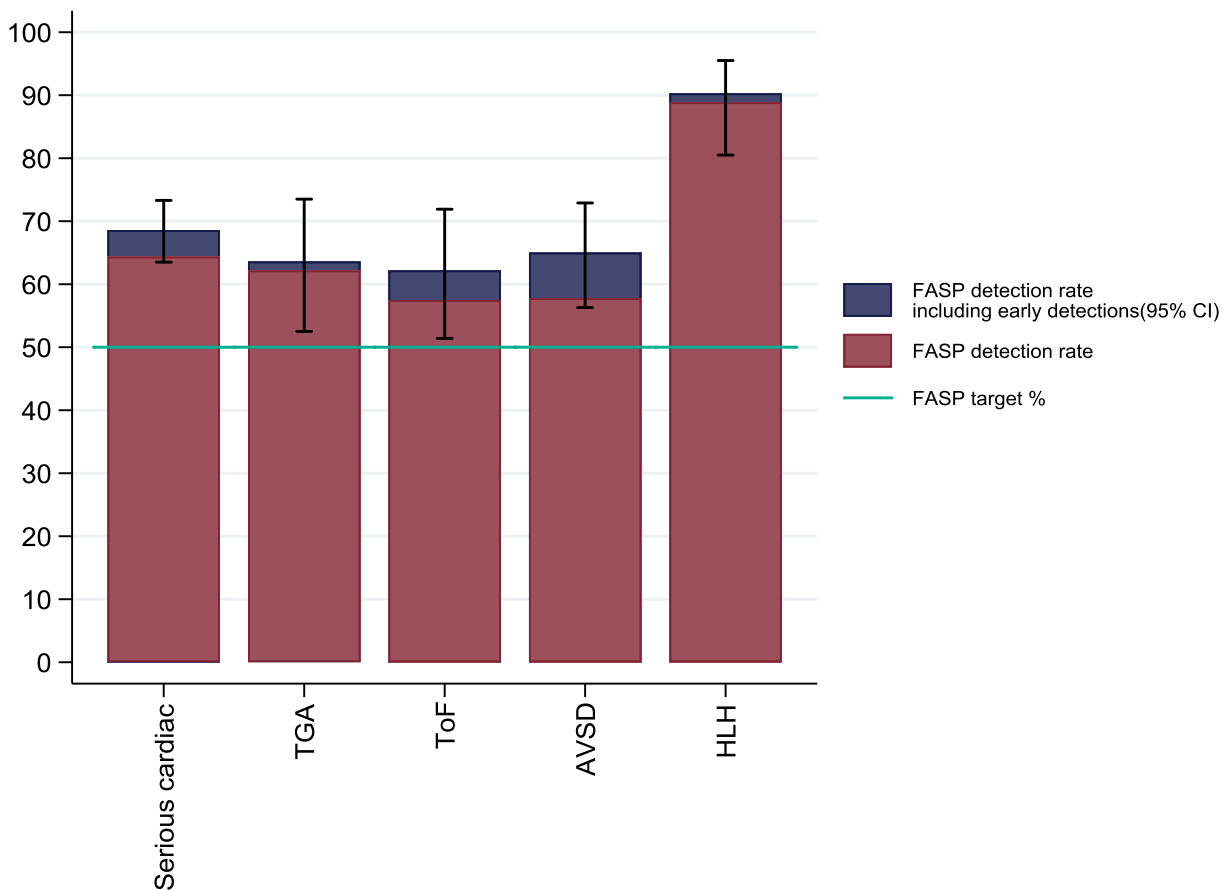
Figure 12: Detection rates, 95% confidence intervals and FASP targets by anomaly including early detections in 46 English reporting trusts, 2015 to 2016



Serious cardiac anomalies are defined here as cases with a confirmed diagnosis of 1 or more of the following: transposition of the great arteries (TGA), tetralogy of Fallot (ToF), atrioventricular septal defect (AVSD), and hypoplastic left heart (HLH). For these anomalies, the detection rate target is 50% for each condition and the group of conditions taken together (serious cardiac anomalies).

The data presented in Figure 13 shows that the target for serious cardiac anomalies combined, as well as for each of the 4 individual cardiac anomalies, was met when early detections are both excluded and included.

Figure 13: Detection rates, 95% confidence intervals and FASP targets for serious cardiac anomalies including and excluding early detections in 46 English reporting trusts, 2015 to 2016



For the 2015 to 2016 reporting year, detailed individualised reports were shared with NHS trusts containing data at trust, regional and national level. These reports contained information about data quality and made recommendations for improvement in data notification to NCARDRS. Each trust was also provided with a full list of FASP cases to enable review of those that were undetected.

The collection and analysis of this information marks an important step towards robust monitoring of the NHS Fetal Anomaly Screening Programme detection rate targets. More information about NCARDRS' work with NHS trusts to improve on the collection of high quality data is discussed in the following chapter.

Chapter 6: Next steps for NCARDRS

6.1 Congenital anomalies – achieving national coverage

This second annual report has shown a marked improvement towards achieving national coverage to enable robust information about the prevalence of congenital anomalies. NCARDRS looks forward to being able to present, for the first time in England, national coverage of congenital anomalies from 1 April 2017; this data will be reported on from 2019.

NCARDRS has pride in the achievements made within the past year to improve on the quality of data collected and reported. For example, there is awareness that postnatal notification data collection is an area for improvement to ensure robust and unbiased reporting going forward. To improve this, staff have been working with our NHS partners to ensure steps are in place to receive and process postnatal notifications. An example of success this year includes work delivered to electronically process data extracted from the BadgerNet Neonatal data system.

NCARDRS has been working to process and analyse congenital anomaly diagnostic data from cytogenetic laboratories recorded in 2015 and 2016. This work is now nearing completion; we plan to publish a national trisomy report in early 2019. This will provide updates to 2013 and 2014 data reported on Down's syndrome, Edwards' syndrome and Patau's syndrome presented in the 2015 NCARDRS annual report.

There is still more work to be done to improve postnatal notifications and to improve ascertainment of the less severe congenital anomalies. NCARDRS will continue work with NHS partners to improve this and create an efficient and effective data service for notifiers.

6.2 Rare disease expansion

In the introduction to this report, the ambition to expand our national service to include rare disease data collection was discussed. There are between 6,000 and 8,000 rare diseases and people with rare disease can present at any age and to any medical discipline. Because of this, NCARDRS values collaborating with experts to help prioritise conditions, understand how to collect retrospective and prospective data, find cases, and interpret and share findings.

NCARDRS is working with our national and international colleagues, including the devolved nations, the International Clearinghouse for Birth Defects and the EU Joint Research Centre, to ensure lessons learned are shared in the emerging area of rare

disease data collection. The Orphanet⁶ UK office sits within NCARDRS, and we are together aiming to implement Orphanet codes in the national reporting of rare disease, including existing congenital anomaly data streams.

Rare diseases are defined as conditions that have an Orphanet code and a prevalence rate of <1:2000 in the population at risk. Because there are so many rare diseases, a pragmatic, project-based approach is being taken to rare disease data collection. Currently, conditions that are not collected elsewhere (like malignant cancers) and are not infectious in origin are being prioritised.

Current areas of focus include proof of concept work on rare rheumatology conditions and Wilson's Disease. These projects should allow for sound estimates of prevalence for these conditions, inform service delivery, report on key outcomes and identify ways in which NCARDRS can continue to collect data on new cases prospectively and give baseline data for future projects. NCARDRS will build on the techniques used in these projects for data collection on other diseases.

Where data is available, stakeholders are being supported to access and interpret information about rare diseases. For example, using routinely collected data, it is estimated that the prevalence of Rett syndrome is 1.6 (95% CI: 1.5-1.7) per 100,000 people in England. This equates to between 843 and 960 people in England.

In addition to the work described above, efforts are being focused on:

- reporting congenital anomalies as rare disease by ensuring that diagnostic information collected allows for granular rare disease coding
- collecting data that is collected nationally but has not been collected in an identifiable format (like the newborn blood spot inherited metabolic disease data)
- exploring ways in which data may be collected on conditions where disease-specific data is currently not routinely collected
- supporting the development of rare disease project work being undertaken by key stakeholders such as clinicians, academics and third sector organisations

6.3 The important work and support of our stakeholders

Engagement with stakeholders has been key in enabling NCARDRS to realise achievements to date. Engagement with patient groups will continue to ensure that patients and their families lie at the heart of our work. NCARDRS will also continue to build on positive working relationships with data notifiers to ensure that together we can

⁶ Orphanet is a unique resource, gathering and improving knowledge on rare diseases so as to improve the diagnosis, care and treatment of patients with rare diseases.

understand and overcome the challenges of data sharing and work towards improved data quality and completeness.

Since the inception of NCARDRS on 1 April 2015, just over 150 data requests have been managed, the majority of which have been requested by existing notifying trusts, other clinicians, researchers, public health professionals, charities and patient groups. More information about data requests is available on the [NCARDRS website](#) and on the Public Health England [Office for Data Release website](#).

Finally, thanks must be given to the healthcare professionals dedicated to reporting cases to NCARDRS. NCARDRS also acknowledges the work and support of NCARDRS Expert Scientific and Clinical Advisory Panel.

Feedback on this report would be welcomed; if you would like to get in touch, please send your comments and suggestions to ncardrs@phe.gov.uk.