Congenital anomalies in Scotland
2012 to 2017

Publication date
26 November 2019
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Introduction

This is the first publication from NHS National Services Scotland, Information Services Division (ISD) focused on congenital anomalies. The publication provides information on the project, started in 2018, to establish a national congenital anomaly register for Scotland. The national register will be known as the Congenital Anomalies and Rare Diseases Registration and Information Service for Scotland (CARDRISS).

The publication also provides current best estimates of the occurrence of congenital anomalies among pregnancies ending in Scotland in 2012 to 2017 inclusive. These estimates have been produced from an analysis of existing national records. We will update and publish these estimates every year until CARDRISS is up and running and can be used as the source of national statistics on the occurrence of anomalies.

Congenital anomalies

Congenital anomalies are abnormalities of body structure or function which are present from birth. They are the result of an abnormality in some aspect of the normal process of development of a baby in the womb.

There are many causes of anomalies\(^1\). Some are due to an underlying genetic defect such as an abnormality in the number or structure of a baby’s chromosomes, or an abnormality in a single gene. Others are the result of environmental factors that disrupt normal development. These include maternal exposure to certain infections; medications or other chemicals; and poor nutritional status, in particular inadequate intake of folic acid. In other cases, anomalies are the result of a baby being subject to abnormal mechanical forces in the womb, for example low levels of amniotic fluid leading to the baby being constrained in an unusual position. Many anomalies have no specific identified cause and are assumed to be the result of the interplay between many genes and environmental factors.

Structural anomalies are generally classified as ‘minor’ or ‘major’. Minor anomalies are those with limited or no functional impact, for example abnormal patterns of ‘creases’ on the palm of the hand, or skin ‘tags’. Minor anomalies are very common and generally of little or no significance. They are not usually included in anomaly statistics. Major anomalies by contrast are those causing significant functional impairment, such as a congenital heart defect or a limb reduction defect. Major anomalies have a substantial (often lifelong) impact on affected children and their families, and on health and other services. Although specific anomalies are generally rare, when all anomalies are considered together they are collectively common. Anomalies are a leading cause of fetal and child ill health and death.

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Major anomalies are present in at least 2-3% of live born children. They are more common in stillbirths and much more common (particularly chromosomal abnormalities) in early pregnancy losses (miscarriages).

Most children with a major anomaly have just one isolated anomaly (with or without other minor anomalies). A minority however have more than one major anomaly. This may occur by chance. Alternatively the specific combination of anomalies seen may result from a single, known underlying cause that simultaneously affects the development of multiple body structures. These cases are known as ‘syndromes’, for example Down syndrome or congenital rubella syndrome. In other cases a single underlying anomaly causes secondary anomalies through downstream/cascade effects. These cases are known as ‘sequences’, for example the Pierre Robin sequence where the underlying anomaly of a small lower jaw leads to an abnormal position of the tongue which in turn impedes normal development of the palate leading to cleft palate. Finally, some specific combinations of anomalies are seen more commonly than would be expected by chance alone, however no underlying cause is (yet) known. These are known as ‘associations’, for example the VATER/VACTERL association².

There are opportunities to prevent some major anomalies, for example through ensuring women who are planning a pregnancy, or in the early stages of a pregnancy, have an adequate intake of folic acid. Ensuring protection from infections through immunisation, and minimising exposure to harmful chemicals also contributes to preventing anomalies. Optimising the pre-pregnancy care of women with certain medical conditions which increase the risk of anomalies either directly (for example diabetes) or through the medications used in treatment (for example epilepsy) is also important.

Increasingly, major anomalies can be detected during pregnancy. All pregnant women in Scotland are currently offered antenatal screening tests to detect specific anomalies. Screening does not prevent anomalies, however it does offer parents and healthcare staff the opportunity to prepare for the birth of a child affected by an anomaly and hence improve their outcomes, for example by planning delivery in a hospital that can provide the level of medical and surgical care a baby may need after birth. In some cases, when a baby is affected by a very serious anomaly, parents may consider the option of terminating the pregnancy.

Establishing a national congenital anomaly register for Scotland

Many countries maintain high quality registers of all babies affected by congenital anomalies to monitor the occurrence of anomalies in the population. Registers can identify high or increasing rates of specific anomalies that may indicate exposure of women to harmful

environmental factors and hence help to inform the prevention of anomalies. Good quality data on numbers of babies affected by anomalies, and their outcomes (such as survival) provides important contextual information for parents and professionals that helps guide decision making when a baby is found to have an anomaly. It also informs the planning of relevant services and helps to ensure they meet the needs of affected children and their families. Data on how anomalies are identified (through antenatal screening or after a child is born) and the outcomes of affected pregnancies (live birth, spontaneous fetal loss/stillbirth, or termination of pregnancy) provides important information on the performance and impact of antenatal screening services. Finally, national registers can support research into anomalies to increase understanding of their occurrence, causes, treatment, and outcomes. Given that the numbers of babies affected by any specific anomaly in one country may be relatively small, national registers often collaborate in international networks to support larger scale monitoring of anomaly occurrence and research.

To date, Scotland has never had a national congenital anomaly register. This is a significant gap in Scotland’s population health data. As part of Scotland’s response to the UK wide strategy for rare diseases, in 2018 the Scottish Government (through the Rare Disease Implementation Oversight Group) therefore commissioned ISD to establish a register. The register will be known as the Congenital Anomalies and Rare Diseases Registration and Information Service for Scotland (CARDRISS).

ISD is currently in the process of setting up CARDRISS. Key initial decisions about how the register will work have been taken and are summarised here. In the first instance, CARDRISS will seek to register babies affected by anomalies in line with the minimum standards recommended by EUROCAT\(^3\). We will register babies affected by a major structural or chromosomal anomaly or recognised syndrome. Live born babies diagnosed within the first year of life; spontaneous stillbirths occurring at \(\geq 24\) weeks gestation; spontaneous late fetal losses occurring at 20-23 weeks gestation; and pregnancies terminated at any gestation due to an included anomaly will all be registered. In due course, when registration of major anomalies is securely established, we plan to widen the remit of CARDRISS to include registration of other rare diseases.

In addition to the central team based in ISD, CARDRISS will depend on a new national IT system and specialist registration staff based in hospitals across Scotland. Broadly speaking, the registration process will work as follows. Babies meeting EUROCAT registration inclusion criteria will be ascertained from existing national records already held by ISD, for example termination of pregnancy notifications; stillbirth records; and neonatal care

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3 EUROCAT is a network of European anomaly registers that was established in the 1970s. It provides standards and guidance on registration to support production of comparable, high quality data. Member registers submit data to a central database maintained by the European Commission's Joint Research Centre. EUROCAT uses this data to support international monitoring of trends in anomaly occurrence and large scale research studies.
discharge records. In addition, healthcare professionals will be able to notify CARDRISS directly of babies affected by an anomaly. Registration staff will then access local clinical records to confirm cases (ensure that they meet inclusion criteria), complete the agreed CARDRISS data items to be gathered on each case, and ensure accurate codes (including ICD10 and, where possible, ORPHA codes) for all anomalies present are appended to babies’ registration records. Information on confirmed cases will then be held by ISD as a new national dataset available for analysis alongside other existing datasets.

In addition to the processes set out above, we will work with colleagues across Scotland to bring relevant local data sources (i.e. data currently held by territorial Boards across Scotland) into the CARDRISS system. This local data will help to ensure we identify all relevant cases and/or efficiently provide key items of information required on cases.

We anticipate that setting up CARDRISS will be a three year project. Over the first year of the project we have agreed the key initial decisions as outlined above. We have successfully applied for Scotland to join EUROCAT as an affiliate member. We have agreed the specific data items to be captured on cases registered through CARDRISS and we have started the process of specifying and building the CARDRISS IT system. In addition we have started discussions with clinical geneticists and genetic scientists with the aim of bringing local data on genetic testing and associated results into CARDRISS.

For CARDRISS to deliver its intended benefits, it is essential that it captures as many affected babies that meet the inclusion criteria as possible. Delivering CARDRISS will be part of ISD’s public task as set out in the legislation that governs our work. Explicit consent will not be sought from affected families prior to babies being registered by CARDRISS. It is therefore particularly important that CARDRISS operates in a transparent way, actively seeking to inform affected families and the wider public about its work, and listening to and acting on any priorities and concerns expressed. To that end, the CARDRISS central team in ISD has already been involved in a range of engagement and communication activities, including an article in the Rare Disease UK newsletter and presentations at an education day hosted by the Office for Rare Conditions Glasgow and at the UK Rare Disease Policy Forum annual meeting.

Our key goals for the remaining second and third years of the project to establish CARDRISS include:

- Establishing a project advisory group
- Developing and disseminating information materials on CARDRISS for affected families, professionals, and the general public
- Securing all the governance approvals required to begin registration
- Completing development of the CARDRISS IT system
• Recruiting and training registration staff and developing the required standard operating procedures and coding guidance that they will require
• Planning the outputs that will be provided when CARDRISS is up and running, for example an annual publication and educational event
• Bringing local genetic testing data into CARDRISS and agreeing a prioritised programme of securing access to further local data resources
• Consolidating Scotland’s membership of EUROCAT to associate and, eventually, full membership status

Our current plan is that CARDRISS will prospectively register affected pregnancies ending in 2021 onwards.

Interim production of statistics on the numbers of babies affected by anomalies based on analysis of existing national records

There are currently no estimates of anomaly occurrence in Scotland for pregnancies ending in 2012 onwards. Whilst we work on establishing CARDRISS, we are therefore also producing the best possible estimates of anomaly occurrence in Scotland in recent years based on analysis of existing national records. In addition to addressing a key gap in national population health data, this analysis will also demonstrate the minimum level of case ascertainment that will be achieved by CARDRISS (i.e. the number of cases that can be ascertained from existing national records alone, without additional input from local data sources or direct notification of cases by clinicians).

This publication presents the results of this analysis. Results are provided for pregnancies ending in Scotland in 2012 to 2017 inclusive. The analysis has sought to include cases that will be registerable by CARDRISS, i.e. babies affected by a major structural or chromosomal anomaly or recognised syndrome. Affected live born babies diagnosed within the first year of life; spontaneous stillbirths occurring at ≥24 weeks gestation; spontaneous late fetal losses occurring at 20-23 weeks gestation; and pregnancies terminated at any gestation due to an included anomaly have all been included.

The following national records have been used in this analysis:
• National Records of Scotland (NRS) statutory live birth registration records
• NRS statutory stillbirth registration records
• Statutory termination of pregnancy notification records
• Hospital maternity care delivery and abortion discharge records (SMR02)
• Hospital neonatal care discharge records (Scottish Birth Record)
• General hospital discharge records for babies aged up to 1 year (SMR01)
• NRS statutory death registration records for babies aged up to 1 year
• Perinatal death enhanced surveillance records (Scottish Stillbirth and Infant Death (SSBID) records for 2012, Mothers and Babies: Reducing Risk through Audits and Confidential Enquiries across the UK (MBRRACE-UK) records for 2013 to 2016 as not yet available for 2017)

All records relating to individual babies have been linked together. EUROCAT recommended code lists have then been used to examine the diagnostic codes included in the records to (a) identify babies with an included anomaly, and then (b) to assign included babies to specific group(s) (for example nervous system anomalies) and type(s) (for example spina bifida) of anomalies as appropriate. If the records relating to a baby contain codes for more than one anomaly they have been counted against each relevant anomaly group and type, but only once in the overall count of affected babies.

The specific record types available for each baby, and the wider coding they contain, have then been used to determine the pregnancy outcome of affected babies: live birth; spontaneous stillbirth at ≥24 weeks gestation; spontaneous late fetal loss at 20-23 weeks gestation; or a termination of pregnancy for fetal anomaly (TOPFA) at any gestation. It is important to note that a baby with multiple anomalies recorded that is subject to TOPFA will be counted as a TOPFA against each of the specific anomaly types that they have (but, as above, only once in the overall count of babies subject to TOPFA). So, for example, a baby subject to TOPFA due to hypoplastic left heart (a severe congenital heart defect) who also has a cleft lip will be counted as a TOPFA against both the hypoplastic left heart and cleft lip anomaly types. It therefore cannot be assumed that babies counted as TOPFAs in any particular anomaly group/type were subject to TOPFA as a direct result of that particular anomaly alone.

Finally, each included baby has been categorised as having/not having a known genetic condition. Again, this follows standard EUROCAT practice. All babies recorded as having an anomaly in the chromosomal; skeletal dysplasias; or genetic syndromes and microdeletions anomaly groups have been assumed to have a known genetic condition. If a baby with one of these anomalies is also recorded as having an anomaly assigned to another group/type (for example a congenital heart defect), their other anomaly is assumed to also be caused by their underlying genetic condition and is classified as such. This distinction is made as anomalies due to an underlying genetic defect have different causes and risk factors (and hence potential for prevention) than anomalies without an underlying genetic defect.

Once the number of babies affected by anomalies was determined, this was used to calculate anomaly birth prevalence. Birth prevalence is the standard measure of anomaly frequency⁴. Total birth prevalence (reflecting the overall occurrence of anomalies in the

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population) is calculated as the total number of affected babies (live births, spontaneous stillbirths at \( \geq 24 \) weeks gestation; spontaneous late fetal loss at 20-23 weeks gestation; and TOPFAs at any gestation) divided by the total number of (live and still) births. Live birth prevalence (reflecting the occurrence of anomalies among live born babies that will require ongoing care and services) is calculated as the number of affected live born babies divided by the number of live births.

A technical report providing full details of methods used in the linked data analysis is provided alongside this publication.

Other sources of information on congenital anomalies

For babies born in Scotland up to 2011, ISD included high level estimates of the number of babies with certain types of anomalies in the annual Scottish Perinatal & Infant Mortality & Morbidity Report (SPIMMR). These previous estimates are not directly comparable to those provided in this publication as they were based on different underlying records and a different approach to linking together different records relating to the same baby. In addition the estimates provided in SPIMMR were only for singleton babies (babies from multiple pregnancies of twins or more were excluded), and bespoke ICD10 code lists (rather than EUROCAT standard lists) were used to define specific groups and types of anomalies.

Elsewhere in the UK, information on anomalies is provided for England by the National Congenital Anomaly and Rare Disease Registration Service (NCARDRS), and for Wales by the Congenital Anomaly Register and Information Service (CARIS). Northern Ireland does not currently have a national anomaly register. Information on anomalies at the European level is provided by EUROCAT, and at the global level by the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR).
Main Points

This is the first publication by ISD focused on congenital anomalies. The publication provides information on the project, started in 2018, to set up a national congenital anomaly register for Scotland. The publication also provides current best estimates of the number of babies affected by anomalies among pregnancies ending in Scotland in 2012 to 2017. These estimates have been produced from an analysis of existing national records. Information is provided on the number of babies affected by a serious congenital anomaly such as a major structural anomaly or a chromosomal anomaly. Live born babies diagnosed before their first birthday; miscarriages and stillbirths from 20 weeks of pregnancy onwards; and terminations of pregnancy at any stage of pregnancy are all counted.

- In 2017, 1,640 babies affected by a serious congenital anomaly were identified among pregnancies ending in Scotland. This is 308.4 per 10,000 total (live and still) births.
- 1,321 (81%) of affected babies were live born. This is 249.5 per 10,000 live births. This means that around 1 in 40 babies born alive in Scotland in 2017 had a serious congenital anomaly.
- Overall, the commonest group of anomalies seen was congenital heart defects (448 babies affected; 84.3 per 10,000 total births).
- Among the specific anomalies covered by the antenatal screening programme, the commonest anomaly seen was Down syndrome (84 babies affected; 15.8 per 10,000 total births).
- Anomalies due to an underlying genetic problem were more common in babies of older mothers. Other anomalies were more common in babies of younger mothers.
- Among live born babies, anomalies were more common in boys (796 boys affected; 292.3 per 10,000 live births) than girls (520 girls; 202.3/10,000); and in babies from a multiple pregnancy of twins or more (48 multiples; 307.5/10,000) than in singletons (1,273 singletons; 247.8/10,000), although the difference between multiples and singletons was not statistically significant.
Results and Commentary

Overall occurrence of included congenital anomalies

1,640 babies affected by an included anomaly (major structural or chromosomal anomaly or recognised syndrome) were identified among pregnancies ending in Scotland in 2017. 1,321 (81%) were live born babies (diagnosed at any time from during pregnancy up to their first birthday; 15 (<1%) were spontaneous stillbirths (delivered at ≥24 weeks gestation); 4 (<1%) were spontaneous late fetal losses (delivered at 20-23 weeks gestation); and 300 (18%) were terminations of pregnancy for fetal anomaly (TOPFA) at any gestation.

The total birth prevalence of included anomalies in 2017 was 308.4 (95% confidence interval 293.7 – 323.7) per 10,000 total (live and still) births. The live birth prevalence was 249.5 (236.2 – 263.3) per 10,000 live births. In other words, around 1 in every 40 babies born alive in Scotland in 2017 was affected by a major congenital anomaly.

Among the 1,640 total cases in 2017, 224 (14%) had a known genetic condition, that is they had an anomaly assigned to the chromosomal; skeletal dysplasias; or genetic syndromes and microdeletions groups, with or without other anomalies also recorded.

Detailed information on the occurrence of all included anomalies (and specific groups and types of anomalies), the outcome of affected pregnancies, and the number of cases with/without a known genetic condition is provided in supporting Table 1.

Occurrence of specific groups of anomalies

Considering the specific groups of anomalies as categorised by EUROCAT, in 2017 congenital heart defects accounted for the highest number of cases/total prevalence (448 cases, 84.3 (76.6 – 92.4) per 10,000 total births). Among the 448 babies affected by congenital heart defects, 54 (12%) had a known genetic condition (i.e. were also coded as having a condition such as Down syndrome).

The groups accounting for the next highest numbers of cases were limb; genital; chromosomal; urinary; and nervous system anomalies. Anomalies affecting the ear, face and neck; and skeletal dysplasias accounted for the fewest cases (Figure 1 and supporting Table 1).
Figure 1: Total birth prevalence of major structural and chromosomal anomalies in Scotland, 2017

For most of the anomaly groups considered, over 90% of affected babies were live born (Figure 2 and supporting Table 1). The exceptions were babies affected by chromosomal and nervous system anomalies. In 2017, among the 179 babies affected by chromosomal anomalies, 67 (37%) were live born. Among the 129 babies affected by a nervous system anomaly, 62 (48%) were live born. Termination of pregnancy for fetal anomaly accounted for almost all of the remaining cases, showing the impact of antenatal screening on the outcome of babies affected by these types of anomalies (see section on Occurrence of specific types of anomalies covered by antenatal screening below).
Occurrence of specific types of anomalies covered by antenatal screening

The **Scottish antenatal screening programme** currently offers women an early pregnancy ultrasound scan and blood test to assess the chance that their baby is affected by Down syndrome (trisomy 21); and a detailed mid pregnancy ultrasound scan to look for the following specific anomalies:

- **Anencephaly**
- **Open spina bifida** (see below)
- **Cleft lip** (with or without cleft palate)
- **Diaphragmatic hernia**
- **Gastroschisis**
- **Omphalocoele/exomphalos**
- **Severe congenital heart defects**, including
  - Transposition of the great vessels
  - Atrioventricular septal defect
  - Tetralogy of Fallot
  - Hypoplastic left heart
- Bilateral renal agenesis
- Lethal skeletal dysplasias
- Edwards syndrome (trisomy 18)
- Patau syndrome (trisomy 13)

Regarding spina bifida: the antenatal screening programme specifically seeks to detect open spina bifida, however ICD10 codes (and EUROCAT code lists) do not distinguish open and closed spina bifida hence this publication provides figures for all forms of spina bifida combined.

The mid pregnancy scan may detect other structural anomalies, but only those listed above have associated recommended antenatal detection rates specified by the screening programme. If, following antenatal screening, a baby is suspected of having a structural anomaly, further tests (for example a repeat specialist scan) would be offered to confirm the diagnosis. If a baby is suspected of having a chromosomal anomaly (trisomy 21, 18, or 13), a definitive diagnostic test would be offered. This involves detailed genetic testing of the fetus based on material obtained through chorionic villus sampling or amniocentesis.

Considering the specific conditions covered by the antenatal screening programme, in 2017 Down syndrome accounted for the highest number of cases/total prevalence (84 cases, 15.8 (12.6 – 19.6) per 10,000 total births). Lethal skeletal dysplasias accounted for the fewest cases (Figure 3 and supporting Table 1).

Note that the national records used to produce the estimates of anomaly birth prevalence provided in this publication do not include information on how or when specific anomalies were first suspected or definitively diagnosed. If a baby was subject to termination of pregnancy for fetal anomaly, it is reasonable to assume that their anomaly was detected during pregnancy. We cannot tell however whether affected live births, stillbirths, or late fetal losses had their anomaly detected during pregnancy or after birth/delivery. Once CARDRISS is up and running, detailed information on the timing and method of detection of affected babies’ anomalies will be captured. This will enable us to provide more detailed information on the proportion of babies affected by anomalies covered by the antenatal screening programme that were actually detected during pregnancy as a consequence of screening.

For most of the specific anomalies covered by antenatal screening, in 2017 over 70% of affected babies were live born. The exceptions were babies affected by anencephaly; spina bifida; hypoplastic left heart; lethal skeletal dysplasias; and each of Down, Edwards, and Patau syndromes. For all of the conditions covered by antenatal screening, TOPFA accounted for almost all of the remaining (non-live born) cases.
Figure 3: Total and live birth prevalence of specific anomalies covered by antenatal screening, Scotland, 2017

Trends over time in the occurrence of included congenital anomalies

The estimated total birth prevalence of all included anomalies increased somewhat between 2012 and 2015, then fell in 2016 and 2017 (Figure 4 and supporting Table 1). The decline seen in 2016 and, in particular, 2017 may in part reflect the following technical issues:

- Perinatal death enhanced surveillance (MBRRACE-UK) records are not currently available to ISD for babies born in 2017. We would not expect to identify many babies affected by anomalies by MBRRACE-UK records alone as these babies should also have other types of records available (such as a statutory stillbirth or infant death registration record), however the absence of MBRRACE-UK records may have marginally reduced ascertainment of affected babies in 2017.

- Hospital neonatal care discharge records (provided to ISD through the Scottish Birth Record, SBR) are an important source through which live born babies affected by anomalies have been ascertained for this analysis. NHS Borders and NHS Dumfries & Galloway have recently had difficulties submitting their SBR records to ISD. At the time that the data extracts used in this analysis were taken (October 2019), no ICD10 diagnostic codes were available through the SBR for babies discharged from the neonatal unit in NHS Borders from June 2017 onwards, and in NHS Dumfries & Galloway from April 2018 onwards. ISD is currently working to improve national neonatal care discharge records, but definite timescales for this work are not yet available.

- Finally, as we seek to include affected live born babies diagnosed at any time up to their first birthday, this means we rely on records for up to the end of 2018 to ascertain
cases born in 2017. Whilst records (for example general hospital discharge records) for late 2018 should have been complete by the time extracts were taken to support this analysis (October 2019), it is possible some relevant records have not yet been submitted to ISD.

Due to the reasons outlined above, results for 2017 provided in this publication should be considered provisional. In next year’s publication, in addition to providing new (provisional) results for babies born in 2018, we will provide updated figures for all previous years. At that point we will be better able to assess if the apparent decline in the prevalence of anomalies in pregnancies ending in 2017 is real, or simply a reflection of underlying data availability issues.

**Figure 4: Total and live birth prevalence of major structural and chromosomal abnormalities in Scotland, 2012 to 2017**

The live birth prevalence rate shows a similar pattern of increasing between 2012 and 2015, then falling in 2016 and 2017. The discrepancy between the live and total birth prevalence has widened slightly between 2012 and 2017. This reflects the fact that the proportion of all affected babies identified that were live born has declined somewhat over time (from 85% in 2012 to 81% in 2017), and the proportion that were TOPFA has conversely increased. This may reflect a genuine trend and/or may be the result of specific difficulties in ascertaining live born cases in 2017 due to problems with neonatal care discharge records as discussed above. Again, trends will become clearer over time as updated data for more years accumulates.
Variation across Scotland

The total birth prevalence of all included anomalies (and specific groups of anomalies) by the NHS Board area of residence of the mother at the time the affected pregnancy ended is provided in supporting Table 2.

The total birth prevalence of all included anomalies shows some variation across Scotland (Figure 5). In 2017, total anomaly birth prevalence was significantly lower than the Scottish average among babies born to women living in NHS Borders, NHS Dumfries & Galloway, NHS Fife, and NHS Tayside. Prevalence was significantly higher than the Scottish average among babies born to women living in NHS Grampian and NHS Highland. In general this pattern has been broadly consistent over the years examined (2012 to 2017). Prevalence rates for the island Boards are very variable year on year due to the small numbers of babies born in these areas.

Variation in anomaly prevalence rates between areas may reflect data availability/quality issues and/or genuine variation in population risk. As discussed above, NHS Borders and NHS Dumfries & Galloway are known to have missing neonatal care discharge (SBR) records from June 2017 and April 2018 respectively. This may have specifically constrained ascertainment of live born babies affected by anomalies in these areas since that time. Factors influencing population risk that may vary across Scotland include maternal age (see below) or maternal exposure to factors that disrupt normal development.

Figure 5: Total birth prevalence of major structural and chromosomal anomalies in Scotland, 2017, by NHS Board of maternal residence
Relationship with demographic characteristics

Maternal age

The total birth prevalence of all included anomalies (and specific groups of anomalies) by the age of the mother at the time the affected pregnancy ended is provided in supporting Table 3. Results are also provided separately for cases with and without a known genetic condition.

In 2017, the total birth prevalence of anomalies that were not due to a known genetic condition decreased with increasing maternal age. By contrast, the total birth prevalence of anomalies that were due to a known genetic condition increased with increasing maternal age, particularly for babies born to mothers aged 35-39 and ≥40 years (Figure 6). This reflects the well known specific association between increasing maternal age and increasing risk of having a baby affected by a chromosomal anomaly such as Down syndrome.

Figure 6: Total birth prevalence of major anomalies with and without a known genetic basis, Scotland, 2017, by maternal age

Maternal deprivation level

The total birth prevalence of all included anomalies (and specific groups of anomalies) by the deprivation level of the mother at the time the affected pregnancy ended is provided in supporting Table 4. Results are also provided separately for cases with and without a known genetic condition. The measure of deprivation used is the Scottish Index of Multiple Deprivation. This is based on the mother’s postcode of residence and reflects the average
level of maternal deprivation within the small geographical area (datazone) that includes her postcode. Results are shown by deprivation quintile: a fifth (20%) of the overall Scottish population lives in areas assigned to each of the 5 quintiles.

In 2017, the total birth prevalence of anomalies that were **not** due to a known genetic condition was highest amongst babies born to mothers living in the most deprived areas (SIMD quintile 1). This may reflect the fact that women living in more deprived areas are at higher risk of exposure to factors that disrupt normal development. By contrast, the total birth prevalence of anomalies that **were** due to a known genetic condition was lowest amongst babies born to mothers living in the most deprived areas (Figure 7). This is likely to reflect the fact that women living in the most deprived areas on average have their babies at a younger age than women living in less deprived areas, and will therefore be at relatively low risk of having a baby affected by a chromosomal anomaly.

**Figure 7: Total birth prevalence of major anomalies with and without a known genetic basis, Scotland, 2017, by maternal deprivation level**

[Graph showing total birth prevalence of major anomalies with and without a known genetic basis by maternal deprivation level.]

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**Without Known Genetic Condition**

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**With Known Genetic Condition**
Infant sex

The live birth prevalence of all included anomalies (and specific groups of anomalies) by the sex of the baby is provided in supporting Table 5. Total birth prevalence is not provided by infant sex as sex is often unknown (or unrecorded on the national records used in this analysis) for affected babies when the pregnancy ends in a TOPFA or late fetal loss.

In 2017, the live birth prevalence of all included anomalies was significantly higher in boys (292.3 (272.3 – 313.3) per 10,000 live births) than girls (202.3 (185.3 – 220.4) per 10,000 live births) (Figure 8). The higher rate in boys was mainly due to a higher rate of genital and urinary anomalies. Some genital anomalies by definition can only affect babies of one sex. For example, hypospadias is one of the most common genital anomalies. Hypospadias is abnormal placement of the urinary outlet on the penis and can only affect boys: girls are not at risk of this anomaly. This will account for some of the higher overall anomaly risk found for boys.

Infant singleton/multiple status

The live birth prevalence of all included anomalies (and specific groups of anomalies) by the baby’s singleton/multiple status (that is whether they were from a pregnancy involving a single baby, or a multiple pregnancy of twins or more) is provided in supporting Table 6. Total birth prevalence is not provided by infant singleton/multiple status as this is often unknown (or unrecorded on the national records used in this analysis) for affected babies when the pregnancy ends in a TOPFA or late fetal loss.

In 2017, the live birth prevalence of all included anomalies was higher among babies from a multiple pregnancy (307.5 (226.8 – 407.7) per 10,000 live births) than those from a singleton pregnancy (247.8 (234.3 – 261.7) per 10,000 live births), although the difference was not statistically significant due to the relatively small numbers of affected babies from multiple pregnancies (Figure 8). The live birth prevalence was higher for multiples than singletons for all other years examined (2012 to 2016). It is well recognised that babies from multiple pregnancies are at higher risk of anomalies than singletons. Some of the additional risk is due to anomalies that affect multiple pregnancies specifically (such as conjoined twinning and increased risk of ‘crowding’ in the womb leading to positional anomalies) however some of the additional risk is unexplained.
Comparison of Scotland’s figures on anomalies to those for the rest of the UK and Europe

The total birth prevalence of all included anomalies (and specific groups of anomalies) estimated for Scotland for each of the years 2012 to 2017 is compared to figures published by EUROCAT for (a) all other UK based full member registers and (b) all other non-UK full member registers with data available for the relevant year in supporting Table 7.

In 2017, the total birth prevalence of included anomalies estimated for Scotland was 308.4 (293.7 – 323.7) per 10,000 total births. This was significantly higher than the total birth prevalence published for other UK based registers (covering Wales and parts of England) (227.6 (221.6 - 233.7) per 10,000 total births) and for the other non-UK based European registers (254.0 (247.9 - 260.2) per 10,000 total births) (Figure 9). A similar pattern was seen for the previous years examined (2012 to 2016).

In the estimates produced for Scotland, the sum of the number of babies affected by each group of anomalies is similar to the total number of affected babies, indicating that most babies identified have just one type of anomaly coded within their national records available for analysis. By contrast, in the figures published by EUROCAT for UK and non-UK based registers, the sum of the number of babies affected by each group of anomalies is consistently around 20% higher than the total number of affected babies, indicating that a substantial minority of babies registered have more than one type of anomaly recorded. This suggests that in producing these estimated anomaly prevalence figures for Scotland, we
have identified a relatively high number of affected babies, but to a relatively low level of
detail.

There are a number of reasons why the anomaly birth prevalence estimates for Scotland
produced for this publication may differ from prevalence figures published by EUROCAT for
existing full member registers. EUROCAT full member registers vary in how they ascertain
babies affected by an included anomaly, for example using a combination of active searching
of local records, encouraging healthcare professionals to report cases, and automated
searching of routine records. As noted previously, the estimates produced for Scotland
presented in this publication are based on analysis of existing national records alone.
Secondly, all EUROCAT full member registers have processes in place to confirm that
identified cases definitely meet EUROCAT registration inclusion criteria, and to ensure that
accurate codes reflecting all anomalies present are then included in babies’ registration
records. Again, as noted previously, in time CARDRISS will adopt similar processes
however this current analysis has relied solely on the ICD10 codes contained within existing
national records.

Taking these differences into consideration, the estimates presented for Scotland in this
publication are likely to have some strengths and some limitations. Relying on high quality
existing national records to ascertain cases means that we are not dependent on healthcare
professionals remembering to notify cases. This is likely to lead to good ascertainment,
particularly of cases diagnosed some weeks or months after a baby is born. However, our
current inability to confirm that cases definitely meet EUROCAT inclusion criteria means that
we may have included some cases with relatively minor anomalies that (inappropriately) had
an included ICD10 code added to their records. In addition, EUROCAT recommends that
registers use special (5th digit) extensions to some ICD10 codes to provide a greater level of
detail when coding a baby’s anomaly/ies. These extension codes are then occasionally used
to define very specific conditions that should be included or, conversely, minor types of
anomalies that should be excluded from registration. As we only had access to the standard
ICD10 codes included on existing national records for this analysis, we could not implement
the small number of inclusion or exclusion criteria that rely on 5th digit extension codes.
Finally, it is likely that our current reliance on ICD10 codes contained within existing national
records has led to under-ascertainment of babies with multiple anomalies. For example, it is
possible that if a pregnancy is terminated following an antenatal diagnosis of Down syndrome
complicated by a severe congenital heart defect, whilst full details will be contained within
local clinical notes, only one of the anomalies may be recorded on the baby’s national
records.

On balance the estimates of anomaly birth prevalence presented in this publication are the
best currently available for Scotland. They are fit for purpose in terms of monitoring the
occurrence of anomalies within the Scottish population and for planning services for babies
and families affected by anomalies. When CARDRISS is up and running, we will be able to
further improve our national statistics on congenital anomalies, as our methods of
ascertaining babies affected by an included anomaly will widen; cases will be confirmed as definitely meeting EUROCAT inclusion criteria; and detailed coding of all anomalies present in included cases (including use of 5th digit ICD10 extension codes where appropriate) will be done. In addition, new items of information not currently available from existing national records, for example on the timing and method of diagnosis of a baby’s anomaly, will be available for analysis.

Figure 9: Total birth prevalence of major structural and chromosomal anomalies in 2017, Scotland compared to all other (full EUROCAT member) UK registries and all non-UK European registries
Supporting information accompanying this report

This publication is accompanied by

- A summary report
- A technical report giving background information on methods
- A list of inclusion and exclusion codes for congenital anomalies
- Summary data tables in Excel format (see below)

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Further Information

Further Information can be found on the ISD website.
For more information on Maternity and Births see the Maternity and Births section of our website.
The next release of this publication will October 2019.

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