

**Alberta Congenital Anomalies
Surveillance System
Fifteenth Report:
Data for 1997 – 2024**

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Data for 1997 – 2024

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Previous reports can be found at: <https://open.alberta.ca/dataset/1710-8594>

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ACRONYMS FOR JURISDICTIONS AND ORGANIZATIONS MENTIONED IN THE REPORT

ACASS	Alberta Congenital Anomalies Surveillance System
AHS	Alberta Health Services
CCASN	Canadian Congenital Anomalies Surveillance Network https://www.canada.ca/en/public-health/services/health-promotion/what-is-ccasn.html
ICBDSR	International Clearinghouse for Birth Defects Surveillance and Research http://www.icbdsr.org/
NBDPN	National Birth Defects Prevention Network https://www.nbdpn.org/
PHAC	Public Health Agency of Canada www.phac-aspc.gc.ca/index-eng.php
PPHS	Alberta Ministry of Primary and Preventative Health Services https://www.alberta.ca/primary-and-preventative-health-services
RCPCH	Royal College of Paediatrics and Child Health

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1. ACASS Activities and Report Summary

1. This is the fifteenth in a series of reports detailing the birth prevalence of congenital anomalies in Alberta, for the years 1997–2024 inclusive.
2. Currently the International Classification of Diseases – 10th Edition (ICD-10-CA) is used by Alberta hospital reporting data systems, and ACASS uses the Royal College of Paediatrics and Child Health (RCPCH) adaptation of ICD-10. Many of the anomalies reported by the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR) <https://www.icbdsr.org/> are reported in this document along with others that might be of interest. It should be noted that notwithstanding the reported anomalies, all items from the ICD-10 “Q” codes as well as other sections such as metabolic conditions are monitored by ACASS. Data on such conditions can be provided to interested parties upon request.
3. The numerator data include not only live births and stillbirths, but also fetal losses <20 weeks gestational age with congenital anomalies. Denominator data include live births and stillbirths only. By including fetal losses in the numerator, the reported rates are better estimates of true congenital anomaly rates. Fetal losses have been ascertained since 1997. Data provided in this report include the years 1997-2024 however data from 1980 onward can be accessed at <https://open.alberta.ca/dataset/1710-8594> and by request. Fetal losses will not be included in the numerators before 1997.
4. Most congenital anomaly rates have remained relatively stable over the years with fluctuations occurring on a year-to-year basis. There are, however, some exceptions:
 - 4.1. While the rates of spina bifida and encephalocele have remained relatively stable from 1997-2024, anencephaly rates continue to significantly decrease ($p=0.0045$) (**pp. 23-28**).
 - 4.2. There is a statistically significant increase of congenital heart defects as a group ($p<0.0001$). Notably, critical congenital heart defects such as tetralogy of Fallot ($p=0.0146$), pulmonary valve atresia/stenosis ($p=0.0019$), hypoplastic left heart syndrome ($p=0.0437$) and coarctation of the aorta ($p=0.0123$) are contributing to this increase (**pp. 29-35**).
 - 4.3. The prevalence of cleft lip with or without cleft palate has remained stable ($p=1.0000$), while cleft palate is trending downwards ($p=0.0979$) (**pp. 36-39**).
 - 4.4. Obstructive genitourinary anomalies continue to significantly increase ($p<0.0001$) (**pp. 40-42**).
 - 4.5. Both upper and lower limb reductions show significant increases ($p=0.0286$ and $p=0.0437$, respectively) (**pp. 43-48**).
5. The percentage of births to women 35 years of age and over continues to increase, at 27% in 2024, compared to 15% in 2000 and five per cent in 1980. Alternatively, the proportion of births to mothers less than 20 years of age has decreased from seven per cent in 2000 to 1.3% in 2024.
6. The total number of Alberta births (live births and stillbirths) to mothers who reside in Alberta has increased steadily from 36,797 in 1997 before peaking in 2015 at 56,524. Since 2015, the number

has decreased to a low of 46,967 in 2022 and has since increased to 51,207 total births in 2024 (p. 70).

7. Members of ACASS continue to participate and collaborate with the Canadian Congenital Anomalies Surveillance Network (CCASN) ([Provincial and Territorial Congenital Anomalies Surveillance: a summary of surveillance programs across Canada, HPCDP: Vol 44\(5\), May 2024 - Canada.ca](#)). T. Bedard is also a member of the British Columbia Congenital Anomaly Surveillance System Advisory Committee.
8. ACASS continues its affiliation with the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR) (<https://www.icbdsr.org>). M. Thomas is the Chair of the Executive Committee. ACASS has participated in many collaborative projects and publications and continues to submit annual data for the ICBDSR Report ([Welcome to the 2024 Report – ICBDSR 2024 Report](#)).
9. Since 2023, ACASS has been a World Affiliate member of The European Concerted Action on Congenital Anomalies and Twins (EUROCAT). This will provide additional collaborative opportunities for ACASS.
10. Since 2020, ACASS has co-authored 10 peer-reviewed publications in addition to presentations at national and international meetings (pp. 50-51).
11. Data from ACASS is routinely used to provide support to quality assurance reviews for the Alberta Newborn Screening Program and the Maternal Serum Prenatal Screen.

2. Introduction

This report provides updated data on congenital anomalies ascertained in Alberta for the years 1997–2024 inclusive. For the current release, many anomalies reported by the ICBDSR ([by congenital condition – ICBDSR 2024 Report](#)) are included with some others that might be of interest. Data on other anomalies can be provided upon request.

The numerator data additionally includes all fetal losses <20 weeks gestation with congenital anomalies. This differs from reports prior to 1997 where only live births and stillbirths were used. The reported rates are more representative of the true rates of congenital anomalies in Alberta. Fetal losses have been ascertained since 1997; thus aggregate data are reported from that year forward. Congenital anomalies data from 1980 onwards can be accessed from previous reports at <https://open.alberta.ca/dataset/1710-8594>; however fetal losses will not be included in the numerator. Denominator data includes live births and stillbirths only.

2.1 History

The history of the Alberta Congenital Anomalies Surveillance System (ACASS) has been described in previous reports. Between 1996 and 2017, funding was provided by Alberta Health. ACASS is now supported by Alberta Health Services and continues to work closely with the Ministry of Primary and Preventative Health Services and Alberta Vital Statistics relying on them for the provision of notifications of births, deaths, and stillbirths (**see Case Ascertainment, p. 12**).

2.2 Purpose of a Surveillance System

Public health surveillance, in general, has been defined by the Public Health Agency of Canada (PHAC) as the ongoing collection, analysis, interpretation, and dissemination of health data for the purpose of planning, implementing, and evaluating interventions to protect and improve the health of populations ([A vision for public health surveillance in Canada by 2030: Technical discussion guide - Canada.ca](#))

The purposes and objectives of surveillance for congenital anomalies (CAs) are to:

- 1) provide reliable and valid data on the birth prevalence of congenital anomalies in Alberta;
- 2) investigate any significant temporal or geographic changes in the frequency of congenital anomalies with a view to identifying environmental, and therefore, possible preventable causes;
- 3) measure trends;
- 4) assess the effectiveness of prevention (e.g., folic acid fortification);
- 5) evaluate the impact of new diagnostic assessments on prevalence trends (e.g. prenatal imaging, non-invasive prenatal screening);
- 6) assist with health-related program planning and development through the provision of data;
- 7) assist with quality assessments for Alberta provincial programs (e.g. Alberta Newborn Metabolic Screening Program, Edmonton Maternal Serum Screening Program);
- 8) participate in research for the etiology and natural history of birth defects;

- 9) assist with research through the provision of congenital anomalies data; and
- 10) provide advice to health care professionals about congenital anomalies, especially with respect to teaching and launching public health campaigns (e.g., folic acid campaign by Community Health in Calgary).

As well as the above, patterns or associations of malformations can be assessed to determine whether they belong to an existing or new syndrome complex.

A principle feature of a surveillance system is the timeliness of data collection, analysis, and reporting however, these should not be accomplished at the expense of an accurate diagnosis. Data are collected to the first birthday, and with the possibility of reporting delays, the data of a given calendar year may not be complete until at least December 31 of the subsequent year although the cases and anomalies are monitored as they are received.

3. Methodology

3.1 Case Definitions

A **congenital anomaly** is an abnormality that is present at birth, even if not diagnosed until months or years later. Most congenital anomalies are present long before the time of birth, some in the embryonic period (up to the end of the seventh week of gestation) and others in the fetal period (eighth week to term). The term “anomaly” covers all the major classes of abnormalities of development, of which there are four major categories as follows:

Malformation – a morphologic defect of an organ, part of an organ or a larger region of the body resulting from an intrinsically abnormal developmental process (e.g., spina bifida, cleft lip and palate).

Deformation – an abnormal form, shape or position of a part of the body caused by mechanical forces (e.g., extrinsic force such as intrauterine constraint causing some forms of clubfoot).

Disruption – a morphologic defect of an organ, part of an organ or a larger region of the body resulting from the extrinsic breakdown of, or interference with, an originally normal developmental process (e.g., an infection such as rubella or a teratogen such as thalidomide).

Dysplasia – the abnormal organization of cells into tissues and its morphologic result (e.g., Marfan Syndrome, osteogenesis imperfecta).

Other definitions related to pregnancy outcomes for the purposes of this report are as follows:

Live birth – a complete expulsion or extraction from the mother, *irrespective* of the duration of the pregnancy, of a fetus in which, after expulsion or extraction, there is breathing, beating of the heart, pulsation of the umbilical cord or definite movement of voluntary muscle (Alberta Vital Statistics Annual review, 2000).

Stillbirth – a complete expulsion or extraction from the mother, at 20 weeks of pregnancy or more **or** after attaining a weight of 500 grams or more, of a fetus in which, after the expulsion or extraction, there is no breathing, beating of the heart, pulsation of the umbilical cord or unmistakable movement of voluntary muscle (Alberta Vital Statistics Annual review, 2000).

Gestation – completed weeks of pregnancy at delivery.

Preterm birth (aka premature) – a birth before 37 weeks of gestation (<37 weeks).

Termination of Pregnancy (ToP) – for our purposes, includes any pregnancy loss before 20 weeks gestation (<20 weeks). Most cases are therapeutic terminations for congenital anomalies but spontaneous abortions or intrauterine fetal deaths with fetal anomalies could also be included.

Anomaly definitions are based, for the most part, on those provided by the ICBDSR and the National Birth Defects Prevention Network (NBDPN).

3.2 Case Ascertainment

An infant can be ascertained at any time up to the first birthday. Multiple ascertainment of the same infant can occur and is encouraged, as this frequently improves the quality and reliability of the data.

As several malformations may occur in the same infant, it is beneficial for each to be reported so that groups of associated malformations may be studied. This, however, leads to difficulties since the final tabulations may be reported as total congenital anomalies (anomaly rates) or as the total number of infants with a congenital anomaly (case rates). The tables in **Appendix A.3 (pp. 55-69)** report anomaly rates, which for most are similar to case rates (e.g. cleft palate, hypospadias, and microcephaly). Whereas with limb reduction anomalies, there can be multiple different limb reduction anomalies in the same infant.

ACASS obtains information about infants with congenital anomalies from a variety of independent sources. Acquisition of additional reporting agencies is always a priority since the use of multiple sources of information improves completeness of ascertainment as well as the verification of the diagnostic data.

ACASS screens many Ministry of Primary and Preventative Health Services (PPHS) and Alberta Vital Statistics documents for the presence of a congenital anomaly including:

- Notice of a Live Birth or a Stillbirth and Newborn Record often referred to as the Physician's Notice of Birth (NOB)
- Medical Certificate of Stillbirth
- Medical Certificate of Death

However, case ascertainment via the NOB has substantially decreased due to new workflows with the provincial implementation of Connect Care.

Also, ACASS screens a notification called the Congenital Anomalies Reporting Form (**CARF, Appendix A.2, p. 54**) that is completed by all acute care hospitals in the province on live births, stillbirths, admissions or hospital deaths of infants under one year of age as well as pregnancy losses involving one or more congenital anomalies. This form currently serves as the single most important source of case ascertainment.

Ascertainment at a continued high level requires each hospital health records department and each health care provider to co-operate with the system by notifying us as promptly as possible. We are fortunate and grateful for having such co-operative agencies and personnel.

Since many children with congenital anomalies are not admitted to hospital, it is very important to obtain out-patient information such as from the Calgary and Edmonton Departments of Medical Genetics.

The provincial implementation of Connect Care has provided the opportunity for ACASS to increase case and congenital anomaly ascertainment by using built reports within the Analytics

Catalog in Connect Care. Included in these reports are live birth records assigned an ICD-10 congenital anomalies code within the first year of life; individuals seen by genetics (prenatal and clinical) for identified eligible congenital anomalies and/or syndromic diagnosis; and those with abnormal cytogenetic or molecular genetic results.

ACASS and Health Information Management Professionals from the Provincial Data Collection group have also been advocating for an electronic case notification workflow within Connect Care. This would be more streamline, efficient, secure, and sustainable.

Appendix A.1 (p. 53) indicates the current ACASS flow of data.

3.3 Quality Control Measures

All case notifications are reviewed by the ACASS team and verified with electronic medical records via Connect Care and Netcare. Some cases will be excluded because they have a condition not considered to be a congenital anomaly i.e., a normal variant (e.g., patent foramen ovale) or are part of a normal developmental process such as a patent ductus arteriosus or undescended testes in a premature infant. Any reports requiring a medical decision are reviewed with the Medical Consultant. Policy decisions with respect to the acceptance or rejection of a case and its coding are referred to the ACASS Advisory Committee. This body includes medical geneticists, a paediatric general surgeon, paediatric pathologists, a paediatric cardiologist, a paediatric neurologist, a paediatric orthopaedic surgeon, a neonatologist, maternal fetal medicine specialists, epidemiologists, and a provincial health information management professional.

3.4 Anomaly Coding

Coding is done at the Calgary office mainly using the Royal College of Paediatrics and Child Health (RCPCH) adaptation of the International Classification of Diseases, tenth edition (ICD-10). Difficult cases are referred to the Medical Consultant. All eligible anomalies are coded that are reported to us. Of note, we have been updating our database as time permits, by going back to the original reports and reviewing all codes for consistency with current coding practices.

3.5 Data Linkage

Data from ACASS are linked to data from the Alberta Vital Statistics Birth Registry by the birth registration number ensuring a unique identifier for each case is entered into the ACASS Application (electronic database). This is important to ACASS because we ascertain cases from multiple sources, thus the unique identifier reduces the risk of duplicate entries for a case.

Data linkage has been achieved with legacy Alberta Perinatal Health Program (APHP) data by way of the personal health number to ascertain maternal risk factor data, such as maternal smoking, drinking and drug exposure(s) during pregnancy for babies with congenital anomalies.

On a quarterly basis, ACASS data are uploaded into Snowflake, which is a platform for data storage, management, linkage across multiple data sets, and analyses. The ACASS data in this

state can be used to support health system data quality and research projects with the required approvals, i.e. ethics and operational.

3.6 Confidentiality and Release of Data

Paper/hard copy notifications of Congenital Anomalies are sent to the Epidemiology and Surveillance Unit, Primary and Preventative Health Services, and from there to the ACASS office in Calgary where the ACASS Application is maintained. The notifications are handled by the ACASS-Lead, Research Assistant, Health Information Management Professional, Administrative Assistant, and Medical Consultant. The data are treated in a completely confidential manner, and these paper notifications are kept in locked files in a locked room. The ACASS Application is secured by limited access and is password protected. Direct contact is never made with the family. When data are requested from us, they are released in aggregate form with no personal identifiers.

3.7 Epidemiological and Statistical Measures

Unless otherwise stated, the birth defect rates presented in this report are calculated using the following formulae:

ANOMALY (DEFECT) RATE =

$$\frac{\text{Number of a specific congenital anomaly among live births + stillbirths + fetal losses}}{\text{Total number of live births and stillbirths}} \times 1000$$

CASE RATE =

$$\frac{\text{Number of individual infants (live or stillborn) or fetuses with } \geq 1 \text{ congenital anomaly}}{\text{Total number of live births and stillbirths}} \times 1000$$

Confidence intervals (95%) are also included because the rate obtained is actually only a point estimate of the unknown, true population rate. The confidence interval provides information about the precision of the estimate. Thus, the confidence intervals are an estimated range of values which there is a 95% probability that the true population rate will fall.

Chi Squared Linear Trend Analysis was performed and presented as appropriate.

3.8 Limitations of Data and Analysis

One of the major limitations of the surveillance system is that on its own, the information provided does not allow us to determine etiology. If increasing trends indicate there is a potentially serious problem, then separate investigative studies need to be done. However, with appropriate approvals in place, it would be possible to conduct linkage studies with other data sources to explore potential causes of specific birth defects.

The ACASS data are collected passively from Alberta Vital Statistics, hospitals, and other agencies. ACASS also uses Connect Care Analytics Catalog Reports to actively collect data. The completeness and accuracy of data are largely dependent on reporting and documentation.

4. Data Quality

As discussed in the last ACASS Report (Alberta Congenital Anomalies Surveillance System, 2024), the COVID-19 pandemic and the provincial implementation of Connect Care may have impacted the quality of ACASS data. A data quality assessment can provide more insight into the impact of these significant events on our data. Data quality indicators (DQIs) are measurements used to identify issues and evaluate interventions to improve the system. They provide opportunities to understand the data, support interpretation, and focus on areas that need improvement. Although there are different data quality assessment tools utilized by networks such as the National Birth Defects Prevention Network and EUROCAT, we applied the DQIs developed by Groisman et al., (2019) since ICBDSR program data were utilized for these DQIs and ACASS is an active member. This DQI tool is also widely applicable, readily available via ICBDSR [Data-Quality-Indicators-tool-project-v3-2022.xlsx](#), and easy to use.

While there are four specific surveillance processes assessed by the Groisman DQIs (ascertainment, description, coding, and classification), the results for the ascertainment and description processes are presented in Tables 4.1 and 4.2 for 2020-2024 data. Ascertainment and description would be the most affected processes. These are highly dependent on factors that are external to ACASS since we primarily rely on health professionals for case ascertainment, and documentation in the health records to describe the congenital anomaly.

The results presented are for selected congenital anomalies that have DQIs and are reported by ACASS.

The ascertainment DQIs in Table 4.1 compares the actual observed prevalence of the congenital anomaly with a 'threshold prevalence'. All DQIs were achieved for ascertainment except for the prevalence ratio for spina bifida and anencephaly and the prevalence by type for gastroschisis and omphalocele.

The spina bifida and anencephaly ratio should be between 1.00 and 1.33 however, the actual ratio is 2.16. This may reflect an under-ascertainment of anencephaly which would further be supported by the statistically significant declining trend reported for 1997-2024 ($p=0.0045$) and noted in the last two reports (Alberta Congenital Anomalies Surveillance System, 2024 and 2021). As most of these cases are early fetal losses due to earlier prenatal detection in recent times, this decline is likely attributed to the terminology used in first trimester prenatal ultrasound reports (e.g. "acrania" or "absent calvarium"). Congenital anomalies with either of those descriptions would be coded with an ICD-10 code outside of the anencephaly section and thus not counted as such. Further discussion can be found on pp. 23-26 of this report.

The prevalence of gastroschisis should be greater than omphalocele, however, the ACASS data shows the opposite. The prevalence of omphalocele is 5.12/10,000 total births (TB) and gastroschisis 2.25/10,000 TB. This is also supported by the statistically significant **increasing** trend of omphalocele for 1997-2024 ($p<0.0001$) and the **decreasing** trend for gastroschisis ($p=0.0249$). The increasing trend for omphalocele has also been reported in previous ACASS reports (Alberta Congenital Anomalies Surveillance System, 2024 and 2021).

An established risk factor for gastroschisis is young maternal age, less than 20 years with the highest prevalence observed in this maternal age group (Alberta Congenital Anomalies Surveillance System, 2024; Feldkamp et al., 2024; Raitio et al., 2020). Like gastroschisis, very young maternal age (less than 20 years) is associated with omphalocele. However, maternal age equal to or greater than 35 years is also an established risk factor (Alberta Congenital Anomalies Surveillance System, 2024; Stallings et al., 2019). The changing demographics of the Alberta population with respect to maternal age at the time of delivery, may be contributing to the observed trends for gastroschisis and omphalocele. The proportion of births to women less than 20 years of age has decreased from seven per cent in 2000 to 1.3% in 2024. Alternatively, births to women 35 years or older has increased from 15% in 2000 to 27% in 2024. These demographic changes would support the prevalence shift to omphalocele being more frequent than gastroschisis in the Alberta population. Thus, the application of the prevalence by type of abdominal wall defect DQI to the ACASS data is done so with caution.

The description DQIs, according to Groisman et al. (2019), indicates how accurate and complete the description and documentation of congenital anomalies are by program staff according to pre-established criteria. This provides the opportunity for congenital anomalies to be ascertained and coded with the most specific ICD-10 code as possible so that the severity (e.g. degree of hypospadias), type (e.g. location of limb reduction), and spectrum of associated anomalies (e.g. trisomy 21 associated with congenital heart defects) can be recorded and efficiently abstracted from the system for analyses and reporting.

The skin coverage of spina bifida, extension of cleft palate, and the degree of hypospadias DQIs were not achieved (Table 4.2). To improve these DQI metrics, perhaps additional training for program staff is required to identify this information in the health record, if it is routinely described (e.g. cleft lip and palate clinic consult, urology consult, operative reports), connecting with different groups responsible for caring for these babies to further describe these congenital anomalies, and/or assuming that although these anomalies are not explicitly described as such, assumptions can be made (e.g. if Chiari II is reported with spina bifida, an open defect can be assumed).

Although DQIs are useful in identifying areas of improvement, they need to be assessed within the local and historical context of the data. Data quality will be routinely assessed for subsequent ACASS reports to identify data issues, track improvements, and guide data interpretation.

Table 4.1: Data Quality Indicators for Ascertainment – 2020 to 2024 Data

Congenital Anomaly	Data Quality Indicator	Criteria Metric	Actual Metric	Criteria Met
Neural Tube Defects	Minimum Prevalence	<i>Prevalence > 5.7</i>	7.13	YES
	Prevalence by Type	<i>Prevalence of spina bifida > anencephaly > encephalocele</i>	3.81 > 1.76 > 1.56	YES
Spina Bifida and Anencephaly	Prevalence ratio Spina Bifida : anencephaly	<i>Prevalence Ratio between 1.00 and 1.33</i>	2.16	NO
Microtia/Anotia	Minimum Prevalence	<i>Prevalence > 0.6</i>	2.62	YES
Critical Congenital Heart Defects	Minimum Prevalence	<i>Prevalence > 21.5</i>	23.61	YES
Oral Clefts	Minimum Prevalence	<i>Prevalence > 11.1</i>	19.5	YES
	Prevalence by Type	<i>Prevalence of cleft lip and palate > cleft palate > cleft lip</i>	7.95 > 7.63 > 3.89	YES
Hypospadias	Minimum Prevalence	<i>Prevalence > 6.2</i>	25.38	YES
Limb Reductions	Minimum Prevalence	<i>Prevalence > 3.8</i>	10.86	YES
	Prevalence by Localization	<i>Prevalence of upper limb reduction cases > prevalence of lower limb reduction cases</i>	7.01 > 3.85	YES
	Prevalence by Axis	<i>Prevalence of transverse > preaxial > postaxial</i>	4.92 > 2.38 > 0.86	YES
Ventral Abdominal Wall Defects	Minimum Prevalence	<i>Prevalence > 3.0</i>	8.94	YES
	Prevalence by Type	<i>Prevalence of gastroschisis > omphalocele</i>	2.25 < 5.12	NO
Gastroschisis	Prevalence by Maternal Age	<i>Prevalence of gastroschisis to mothers under 20 years old > prevalence over 19 years old</i>	25.59 > 1.91	YES
Trisomy 21	Prevalence by Maternal Age	<i>Prevalence of Trisomy 21 to mothers over 34 years old > prevalence under 35 years old</i>	68.64 > 13.08	YES
	Observed : Expected Ratio	<i>Observed : Expected between 0.67 and 1.33</i>	0.76	YES

Table 4.2: Data Quality Indicators for Description – 2020 to 2024 Data

Congenital Anomaly	Data Quality Indicator	Criteria Metric	Actual Metric	Criteria Met
Spina Bifida	Level Specified	<i>Proportion of cases with specified level > 80%</i>	87.1%	YES
	Skin Coverage	<i>Proportion of cases with open or closed type specified > 80%</i>	51.6%	NO
Encephalocele	Localization	<i>Proportion of cases with specified localization > 80%</i>	86.8%	YES
Congenital Heart Defects	Specification	<i>Proportion of cases with specified congenital heart defects > 80%</i>	97.9%	YES
Cleft Lip	Laterality	<i>Proportion of cases with specified laterality of cleft lip > 80%</i>	96.8%	YES
Cleft Palate	Extension	<i>Proportion of cases with extension of cleft specified as hard and/or soft > 80%</i>	70.4%	NO
Hypospadias	Specification	<i>Proportion of cases with specified type > 80%</i>	63.6%	NO
Limb Deficiencies	Specification	<i>Proportion of cases with specified type and localization > 80%</i>	89.8%	YES
Ventral Abdominal Wall Defects	Specification	<i>Proportion of cases with specified type > 80%</i>	82.6%	YES
Trisomy 21	Cases with Congenital Heart Defects	<i>Proportion of cases with Trisomy 21 have a congenital heart defect $\geq 40\%$</i>	54.7%	YES

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5. Patterns of Selected Congenital Anomalies in Alberta

5.1 Birth Prevalence – Time Trends

The following table of selected sentinel anomalies indicate the trends in congenital anomaly rates in Alberta from 1997 through 2024. Sentinel anomalies are those which the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR), of which we are a member, watches worldwide with the rationale that they are quite easily identified hence more accurately reported. See **Appendix A.5 (p. 71)** for other anomalies listed in the report.

Table 5.1.1: Chi Squared Linear Trend Analysis - Selected Anomalies, 1997–2024

Anomaly	Trend Direction	Chi Squared Analysis (χ^2 LT)	p-value
Neural Tube Defects	No significant change	2.34	0.1261
Anencephaly	Decreasing	8.09	0.0045
Spina Bifida	No significant change	0.00	1.0000
Hydrocephalus	Decreasing	25.56	<0.0001
Cleft Lip +/- Cleft Palate	No significant change	0.00	1.0000
Cleft Palate	No significant change	2.74	0.0979
Hypoplastic Left Heart Syndrome	Increasing	4.07	0.0437
Oesophageal Atresia/Tracheo-oesophageal Fistula	Increasing	4.03	0.0447
Anorectal & Large Intestine Atresia/Stenosis	Decreasing	14.18	0.0002
Hypospadias (male births only)	Increasing	40.33	<0.0001
Undescended Testes (male births only)	No significant change	2.52	0.1124
Renal Agenesis/Hypoplasia	Increasing	29.08	<0.0001
Limb Reductions - upper	Increasing	4.79	0.0286
Limb Reductions - lower	Increasing	4.07	0.0437
Gastroschisis	Decreasing	5.03	0.0249
Omphalocele	Increasing	25.29	<0.0001
Down Syndrome	Increasing	49.75	<0.0001

5.2 Selected Anomalies

5.2.1 Selected Anomaly Definitions

(Adapted from the ICBDSR Reported Malformations Definitions: <http://www.icbdsr.org/> and the NBDPN guidelines: <http://www.nbdpn.org/>)

Abdominal Wall Defects

- **Gastroschisis** – a congenital opening or fissure in the anterior abdominal wall lateral to the umbilicus through which the small intestine, and occasionally the liver and spleen, may be herniated.
- **Omphalocele** – a defect in the anterior abdominal wall in which the umbilical ring is widened, allowing herniation of abdominal organs, including the small intestine, part of the large intestine, and occasionally the liver and spleen, into the umbilical cord. The herniating organs are covered by a nearly transparent sac.

Anorectal Atresia/Stenosis

Complete or partial occlusion of the lumen of one or more segments of the large intestine and/or rectum.

Anotia/Microtia

- **Anotia** – absence of external ear and canal
- **Microtia** – hypoplasia of external ear

Chromosome Anomalies

- **Trisomy 13** – *aka Patau syndrome* – the presence of three copies of all or a large part of chromosome 13.
- **Trisomy 18** – *aka Edwards syndrome* – the presence of three copies of all or a large part of chromosome 18.
- **Trisomy 21** – *aka Down syndrome* – the presence of three copies of all or a large part of chromosome 21.

Cleft Lip and Palate

- **Cleft Lip** – a defect in the upper lip resulting from incomplete fusion of the parts of the lip.
- **Cleft palate** – an opening in the roof of the mouth resulting from incomplete fusion of the shelves of the palate.

Congenital Heart Disease

- **Aortic valve stenosis** – obstruction or narrowing of the aortic valve impairing blood flow from the left ventricle to the aorta.
- **Atrial Septal Defect (ASD)** – opening in the septum that divides the right and left atria of the heart.
- **Coarctation of the aorta** – narrowing of the descending aorta obstructing blood flow from the heart to the rest of the body.

- **Hypoplastic Left Heart Syndrome** – a condition in which the structures on the left side of the heart and the aorta are extremely small. Classically, this condition includes hypoplasia of the left ventricle, atresia or severe hypoplasia of the mitral and aortic valves.
- **Tetralogy of Fallot** – the simultaneous presence of a ventricular septal defect (VSD), pulmonic stenosis, a malpositioned aorta that overrides the ventricular septum and right ventricular hypertrophy.
- **Ventricular Septal Defect (VSD)** – opening in the septum that divides the right and left ventricles of the heart.

Hydrocephalus

An increase in the amount of cerebrospinal fluid within the brain resulting in enlargement of the cerebral ventricles and increased intracranial pressure.

Hypospadias

Displacement of the opening of the urethra ventrally and proximally (underneath and closer to the body) in relation to the glans of the penis.

Limb Reductions

Complete or partial absence of upper and/or lower limbs.

Microcephaly

Commonly defined as a head circumference less than 2 standard deviations (SD) from the mean, or less than the 3rd percentile for age and sex (some jurisdictions use less than 3 SD).

Neural tube defects

- **Anencephaly** – partial or complete absence of the brain and skull.
- **Spina Bifida** – incomplete closure of the vertebral spine through which spinal cord tissue and/or the membranes covering the spine (meninges) are herniated.
- **Encephalocele** – herniation of brain tissue and/or meninges through a defect in the skull.

Obstructive genitourinary anomalies

Partial or complete obstruction of the flow of urine at any level of the genitourinary tract from the kidneys to the urethra.

Renal Agenesis/Hypoplasia

Complete absence or incomplete development of the kidney.

Undescended Testes

Bilateral or unilateral undescended testis in a term newborn.

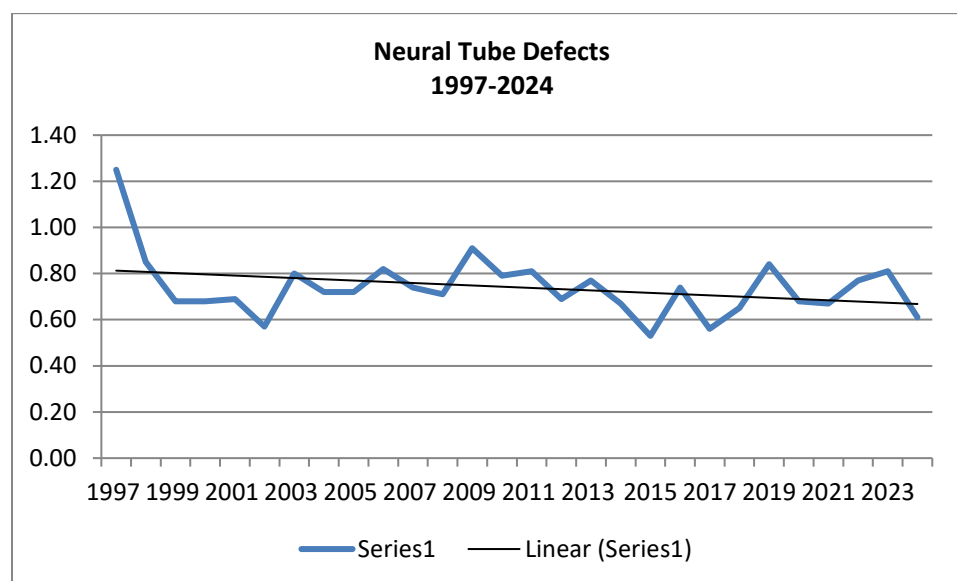
5.2.2 Neural Tube Defects

Between 1997 and 2024, the prevalence of neural tube defects (NTDs) has not significantly changed ($p=0.1261$), however it is a possible trend to watch (Figure 5.2.1). Although there continues to be a statistically significant decline in anencephaly ($p=0.0045$), the rates of spina bifida and encephalocele are stable.

The prevalence of NTDs as a group for the 25-year period of 2000-2024 is 0.72/1,000 total births (TB), which is much higher than the Canadian average of 0.48/1,000 TB for 2008-2023 (Public Health Agency of Canada, 2025). This comparison is done with caution, as the time periods differ slightly and the Public Health Agency of Canada exclusively uses data derived from the Canadian Institute for Health Information (CIHI) to perform national congenital anomaly surveillance. This CIHI data set does not include early fetal losses (less than 20 weeks gestation) which would contribute to an under ascertainment of congenital anomalies, including NTDs.

While the Alberta prevalence of NTDs is higher than the Canadian average, it is lower than that reported by Broughan et al. (2024) for England for 2000-2019 (1.25/1,000 TB), and 0.98/1,000 TB for Scotland for 2000-2021 (Kirolos et al., 2025). Mandatory folic acid was legislated in November 2024 with implementation by the end of 2026 in the United Kingdom, thus these rates will likely decrease.

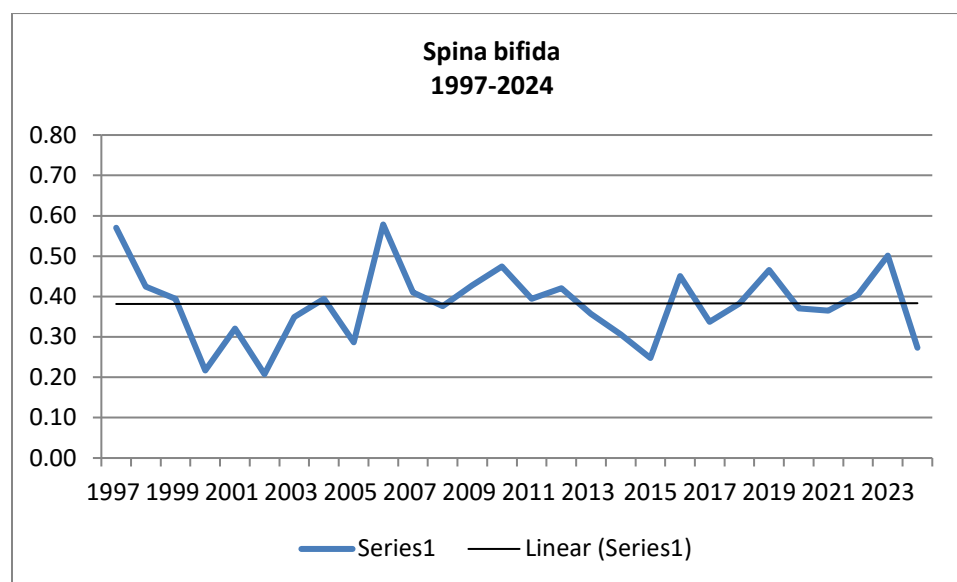
Figure 5.2.1: All Neural Tube Defects, 1997-2024 (Rate per 1,000 total births)



$p=0.1262$

Although the prevalence of spina bifida fluctuates yearly, the long-term prevalence has remarkably remained stable, with a range between 0.38 and 0.40/1,000 TB for 10-year and 25-year periods (p. 57), and no statistical change in the trend from 1997-2024 (p=1.0000) (Figure 5.2.2).

Figure 5.2.2: Spina Bifida, 1997-2024 (Rate per 1,000 Total Births)



p=1.0000

Mai et al. (2022), reported a 72% decrease of severe, upper lesion defects in the US after mandatory FAF. The authors defined “severe upper lesion” defects as cervical and thoracic level lesions since they are associated with increased disability and mortality risk when compared with sacral and lower lumbar level lesions (Sullivan & Herdt, 2022). The overall prevalence of lower-level lesions, that are considered less severe, remained relatively unchanged (Mai et al., 2022). In Alberta between 2000 and 2024, the majority of cases with spina bifida had either lower-level lesions or lipomeningocele, which is not as responsive to folic acid ($386/453 = 85.2\%$), compared with $36/453 = 7.9\%$ with lesions at the cervical and thoracic levels. The remaining 31 (6.8%) cases did not have the level of lesion recorded. A more comprehensive assessment of pre and post folic acid fortification prevalence by level of lesion for Alberta would provide more insight and contribute to a more thorough evaluation of the impact of folic acid and potentially other factors on spina bifida severity.

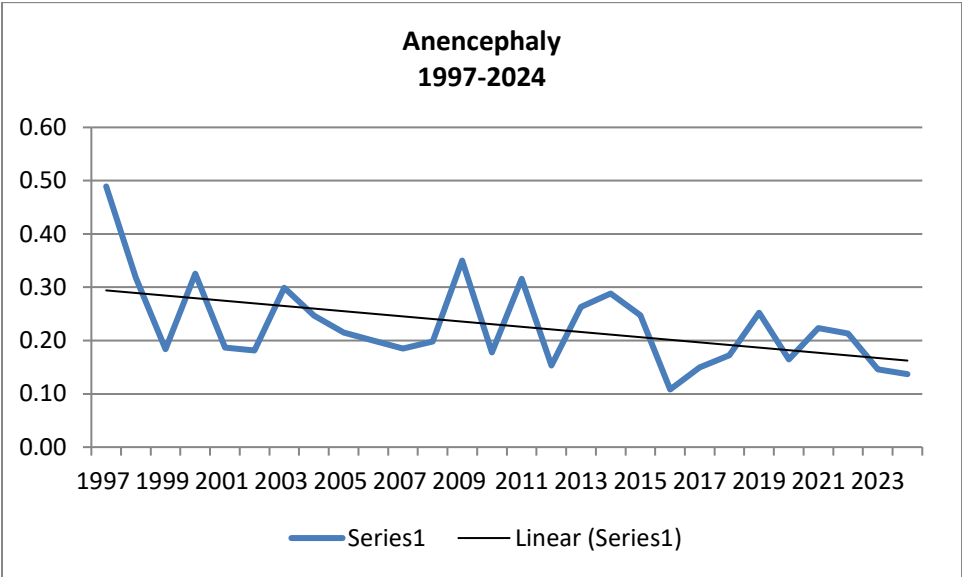
Inadequate red blood folate concentrations and other micronutrient deficiencies may be contributing to the observed frequency of this subset of spina bifida (Chandler et al., 2012; Colapinto et al., 2011). It has been suggested that the entirety of one-carbon metabolism, including other micronutrients that act as methyl donors or cofactors, are involved in the etiology

of NTDs. Petersen et al. (2023) reported a lower NTD risk, in the context of folic acid fortification, for women with higher periconceptional intake of methyl donors. This association was strongest (~ 75% lower NTD risk) with concurrent consumption of higher amounts of vitamins B6 and B12, choline, betaine, and methionine, compared with intake of only 1 or no methyl donors (Petersen et al., 2023).

The decline in anencephaly rates was first noted for 2016 (Alberta Congenital Anomalies Surveillance System, 2024 and 2021) and continues to statistically significantly decrease from 1997-2024 (Figure 5.2.3). Conversely, there have been relatively recent reported increases in the prevalence of anencephaly for England (2015-2019) and Scotland (2013-2021) (Broughan et al., 2024; Kirolos et al., 2025). This may be a true increase or it could be due to an increased detection of anencephaly in early pregnancy, improved data quality through increased ascertainment, and/or the impact of the established downward trend in RBC folate concentrations in women of childbearing age in the United Kingdom (Broughan et al., 2024; Kirolos et al., 2025).

The decline in Alberta may be due to the terminology used to report what is seen on first trimester prenatal ultrasounds (e.g. “acrania” or “absent calvarium”). When described as such, these anomalies are coded by ACASS with an ICD-10 code outside of the NTD section which is based on consultation and a subsequent 2010 ACASS coding policy amendment.

Figure 5.2.3: Anencephaly, 1997-2024 (Rate per 1,000 Total Births)



p=0.0045

Although the decline may be a true decline and there is heterogeneity regarding the etiology of subtypes of NTDs, the prevalence rates of anencephaly and spina bifida are generally expected

to show similar trend patterns over time (Kooshnood et al., 2015). Also notable is that for the years 2020-2024, the data quality indicator for anencephaly ascertainment was not achieved (pp. 15-18). The prevalence ratio for spina bifida and anencephaly should be between 1.00 and 1.33 however this ratio is 2.16. A comparison with other jurisdictions that include terminations of pregnancy and are population-based are shown in Table 5.2.1. These programs, along with ACASS, report to the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR) (ICBDSR, 2024). The United Kingdom was the only jurisdiction that achieved the ascertainment data quality indicator for anencephaly with a ratio of 1.09. While Texas and Alberta have mandatory folic acid fortification, the European countries listed in Table 5.2.1 all rely on voluntary fortification.

Table 5.2.1: Spina Bifida-Anencephaly Prevalence Ratio for Alberta and other Jurisdictions Reporting to the ICBDSR, 2015-2020, prevalence per 10,000 Total Births

Jurisdiction	Prevalence of Spina Bifida	Prevalence of Anencephaly	*Prevalence Ratio Spina Bifida : Anencephaly
Alberta	3.7	1.8	2.06
Texas	3.6	2.3	1.57
United Kingdom	5.9	5.4	1.09
Sweden	3.4	2.1	1.62
Germany	4.9	2.8	1.75

*Expected Ratio between 1.00 and 1.33

Since ICD-10 codes are assigned by what is described, the prevalence estimate of anencephaly may be higher. The statistically significant decline could be due to the terminology used for very early prenatal ultrasound findings with no or very limited postnatal examination to confirm the progression through the acrania-exencephaly-anencephaly sequence (Society for Maternal-Fetal-Medicine & Monteagudo, 2020; Santana et al., 2018). A review of ACASS coding policies, discussions with pediatric neurology and pathology, and maternal-fetal-medicine groups, with further analysis of data quality will support more accurate reporting of anencephaly.

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5.2.3 Congenital Heart Defects:

The Alberta Congenital Anomalies Surveillance System reports selected congenital heart defects (CHDs). This is a very heterogeneous group, comprising both less severe (e.g. small ventricular septal defects) and more severe (e.g. hypoplastic left heart syndrome) types of anomalies. Some CHDs such as small ventricular septal defects may close spontaneously and not require intervention, while some such as hypoplastic left heart syndrome may need multiple surgical procedures.

Table 5.2.2 presents a comparison of the ACASS birth prevalence of these anomalies with the Canadian Congenital Anomalies Surveillance System (CCASS) ([Congenital Anomalies in Canada — Canada.ca](#)), the Texas Birth Defects Registry ([Table 1. Overall Prevalence of Selected Birth Defects, Texas, 1999–2022](#)), and the European network of population-based registries for the epidemiological surveillance of congenital anomalies (EUROCAT) ([Prevalence charts and tables | European Platform on Rare Disease Registration](#)). Most rates are comparable, however there are notable differences, particularly for ventricular septal defects and atrial septal defects. The Texas rates are significantly higher than those reported by ACASS and EUROCAT, which may reflect a higher frequency of septal defects and/or an increased detection and inclusion of smaller defects.

While the prevalence presented for EUROCAT is the overall pooled rate from diverse congenital anomalies surveillance systems throughout Europe, this network has well-established guidelines, data quality assessments, and available tools to support uniform data transmission to the Central Registry for analysis. The ACASS CHD prevalence rates presented are most similar to those reported by EUROCAT.

Congenital heart defects (CHDs) as a group continue to statistically significantly increase from 1997-2024 ($p < 0.0001$). There are annual variations per 1,000 TB during this period, from a low of 9.37 in 2006 (data not shown) to a high of 15.76 in 2024. Atrial septal defects and ventricular septal defects are the most common types of CHDs and have statistically significantly increased ($p = 0.0495$ and $p = 0.0136$, respectively). Improved echocardiography which detects minor lesions such as small septal defects is perhaps contributing to this increase (Mamasoula et al., 2022). Although ACASS does not include patent foramen ovale as an eligible congenital anomaly, as they are relatively common and often spontaneously close, small secundum atrial septal defects and ventricular septal defects are included. Norway and the United Kingdom had decreasing trends for CHDs during 2008-2015 after adopting the EUROCAT recommendation to only include atrial septal defects if they remain open six months after birth (Mamasoula et al., 2022).

Table 5.2.2: Prevalence of Selected Congenital Heart Defects by Congenital Anomalies Surveillance System, per 1,000 Total Births

Congenital Heart Defect	ACASS (2000-2024)	CCASS (2008-2023)	Texas * (1999-2022)	EUROCAT (2000-2023)
Common Truncus	0.08 (0.06-0.09)	0.05 (0.04-0.06)	0.08 (0.07-0.08)	0.08 (0.07-0.08)
Transposition of Great Arteries	0.39 (0.36-0.43)	0.54 (0.52-0.56)	0.33 (0.32-0.35)	0.39 (0.38-0.41)
Tetralogy of Fallot	0.36 (0.32-0.39)	0.46 (0.44-0.48)	0.40 (0.39-0.41)	0.37 (0.36-0.38)
Ventricular Septal Defect	3.17 (3.07-3.27)	NR	6.25 (6.20-6.30)	3.78 (3.75-3.81)
Atrial Septal Defect	1.95 (1.87-2.03)	NR	7.38 (7.32-7.43)	1.77 (1.75-1.79)
Endocardial Cushion Defect/AVSD	0.55 (0.51-0.59)	0.46 (0.44-0.48)	0.44 (0.43-0.46)	0.49 (0.48-0.50)
Pulmonary Valve Atresia/Stenosis	0.69 (0.64-0.73)	NR	1.01 (0.99-1.03)	0.58 (0.56-0.60)
Tricuspid Valve Atresia/Stenosis	0.09 (0.07-0.11)	NR	0.19 (0.19-0.20)	0.08 (0.07-0.08)
Ebstein's Anomaly	0.07 (0.05-0.09)	NR	0.08 (0.07-0.08)	0.05 (0.05-0.06)
Aortic Valve Atresia/Stenosis	0.19 (0.17-0.22)	NR	0.24 (0.23-0.25)	0.15 (0.15-0.16)
Hypoplastic Left Heart Syndrome	0.33 (0.30-0.37)	0.28 (0.26-0.29)	0.22 (0.21-0.23)	0.29 (0.28-0.30)
Coarctation of Aorta	0.44 (0.40-0.48)	0.59 (0.57-0.61)	0.53 (0.52-0.55)	0.41 (0.40-0.42)

*per 1,000 live births only; NR, not reported

Endocardial cushion defects/atrioventricular septal defects (AVSDs) also show a statistically significant increase between 1997 and 2024 ($p=0.0003$) with a higher prevalence than other programs in Table 5.2.2 and non-overlapping confidence intervals for the period 2000-2024. Stallings et al. (2024) reported an increase for AVSD between 2016-2020, when compared with earlier time periods. The estimated United States national prevalence for this period was 0.58/1,000 TB, with non-overlapping confidence intervals when compared with 2010-2014, 2004-2006, and 1999-2001 (Stallings et al., 2024).

Atrioventricular septal defects are often associated with trisomy 21 with 45% of these CHD cases diagnosed with this aneuploidy (data not shown). Trisomy 21 also has a statistically significant increase ($p<0.0001$) (p. 71). Seventy-four per cent of cases with an AVSD have a chromosomal anomaly, including trisomy 21, a genetic syndrome (e.g. Noonan syndrome), or a recognized condition such as heterotaxy. Only 17% of cases with an AVSD were considered isolated and did not have a congenital anomaly outside of the cardiovascular system (Table 5.2.3).

It is notable that a few types of critical congenital heart defects are also statistically significantly increasing. Critical congenital heart defects are severe lesions requiring intervention early in life for an optimal health outcome (Wong et al., 2017). Those with a statistically significant increase for the period 1997-2024 include tetralogy of Fallot ($p=0.0146$), pulmonary valve atresia/stenosis ($p=0.0019$), hypoplastic left heart syndrome (0.0437), and coarctation of the aorta ($p=0.0123$). These were increasing in previous years, though not statistically significant (Alberta Congenital Anomalies Surveillance System, 2021; 2024).

An epidemiological description for these and other CHDs that ACASS routinely reports is in Table 5.2.3. The majority are live born with the exception of cases with common truncus (48%) or hypoplastic left heart syndrome (46%). Isolated cases, those without extra-cardiac congenital anomalies or syndrome diagnoses, ranged from 17% for AVSD to 63% for transposition of the great arteries. Notably, **non**-isolated cases may also include associated relatively minor anomalies (e.g. polydactyly). The average maternal age ranged from 29 years for Ebstein's anomaly to 33 years for AVSDs. On average, most live births with CHDs were delivered close to term, except for those with pulmonary valve atresia/stenosis which were delivered late preterm at 36 weeks.

Relatively recent meta-analyses assessed non-genetic and familial risk factors for CHDs (Khalilipalandi et al., 2024; Lemieux et al., 2024). Excluded from both analyses were cases with chromosomal anomalies, genetic mutations, multisystemic syndromes, consanguinity, exposures to pollution and/or contaminants, and socioeconomic status. Increased risks for CHDs were associated with maternal obesity, maternal diabetes with a larger effect-size for pre-gestational maternal diabetes compared with gestational diabetes, maternal smoking and increased maternal age greater than 35 years (Khalilipalandi et al., 2024). There were also increased risks associated with increased nuchal translucency, extracardiac anomalies, and a family history of CHD particularly first-degree (Lemieux et al., 2024). Antidepressants for all CHDs and assisted reproductive technology for 'severe' CHDs had statistically significant increased odds ratios (Lemieux et al., 2024).

Miao et al. (2023) reported a statistically significant association between higher maternal material deprivation (27% higher odds of having a child with a CHD), which represents poverty and the

(in)ability to purchase basic material needs. Infants with mothers living in more ethnically diverse neighbourhoods were associated with an 11% reduced risk of CHDs compared with infants to mothers in the least ethnically concentrated quintile in Ontario (Miao et al., 2023). The authors hypothesized that this protective association may be due to the healthy immigrant effect, whereby recent immigrant populations have a tendency to be healthier than those born in Canada (Miao et al., 2023). However, this needs further investigation since there is heterogeneity within CHDs and minority groups, where specific types of CHDs may be inherently lower in specific ethnic groups.

Table 5.2.3: Descriptive Epidemiology of Selected Critical Congenital Heart Defects, for the last 10 years of complete ACASS data, 2015 -2024

Congenital Heart Defect	Total number of Cases	Live Births N (%)	*Isolated CHD N (%)	Average Maternal Age (years)	Average Gestational Age at delivery, live births (weeks)
Common Truncus	42	20 (48)	15 (36)	32	38
Transposition of Great Arteries	212	157 (74)	133 (63)	30	38
Tetralogy of Fallot	187	143 (76)	100 (53)	32	37
Ventricular Septal Defect	1639	1442 (88)	940 (57)	31	37
Atrial Septal Defect	977	970 (99)	471 (48)	31	38
Endocardial Cushion Defect/AVSD	310	180 (58)	53 (17)	33	37
Pulmonary Valve Atresia/Stenosis	399	330 (83)	222 (56)	31	36
Tricuspid Valve Atresia/Stenosis	53	31 (58)	29 (55)	31	38
Ebstein's Anomaly	30	20 (67)	21 (70)	29	37
Aortic Valve Atresia/Stenosis	99	81 (82)	61 (62)	31	38
Hypoplastic Left Heart Syndrome	185	86 (46)	108 (58)	31	37
Coarctation of Aorta	241	199 (83)	144 (60)	30	37

*Isolated cases do **not** have extracardiac congenital anomalies or syndrome diagnoses.

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Miao Q, Dunn S, Wen SW, Loughheed J, Yang P, Davies M, Lavin Venegas C, Walker M. (2023). Association between maternal marginalization and infants born with congenital heart disease in Ontario Canada. *BMC Public Health*, 23:790. <https://doi.org/10.1186/s12889-023-15660-5>
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5.2.4 Orofacial Clefts

The prevalence of cleft lip with or without cleft palate (CL/P) (Figure 5.2.4) has remained remarkably stable in Alberta for 28 years ($p=1.0000$).

Cleft palate (CP) shows a slight downward trend that is not statistically significant ($p=0.0979$) (Figure 5.2.5), but no trend for isolated CP, i.e. excluding cases with associated anomalies, known syndromes, teratogens, or chromosome disorders ($p=0.6892$) (Figure 5.2.6). We have no explanation for the decline and have no reason to believe that there is any change with ascertainment.

Section 4 of this report shows the application of the data quality indicators (DQI) tool for congenital anomaly programs (<https://www.icbdsr.org/data-quality-indicators-tool/>) to ACASS data for the period 2020-2024. This tool was designed to assess congenital anomaly data quality and includes indicators for orofacial clefts (OFCs) (pp. 17 & 18). The minimum prevalence of OFCs should be greater than 11.1/10,000 total births (TB) and is 19.5/10,000 TB. The expected prevalence of OFCs per 10,000 TB should be $CL/P > CP > CL$ and the actual is $7.95 > 7.63 > 3.89$. This indicates that the ascertainment of OFC cases in Alberta is meeting expectations. While the DQI for the description of cleft lip was achieved with 97% of cases having the laterality of the cleft specified, the indicator for the extension of the cleft palate to hard and/or soft palate was not achieved. The expected metric was $>80\%$ of cases with this description and ACASS had this for 70.4% of cases. This may be due to a lack of description in the medical record such as in the surgical report which can be feedback to the surgeons or fetal cases with limited postnatal description.

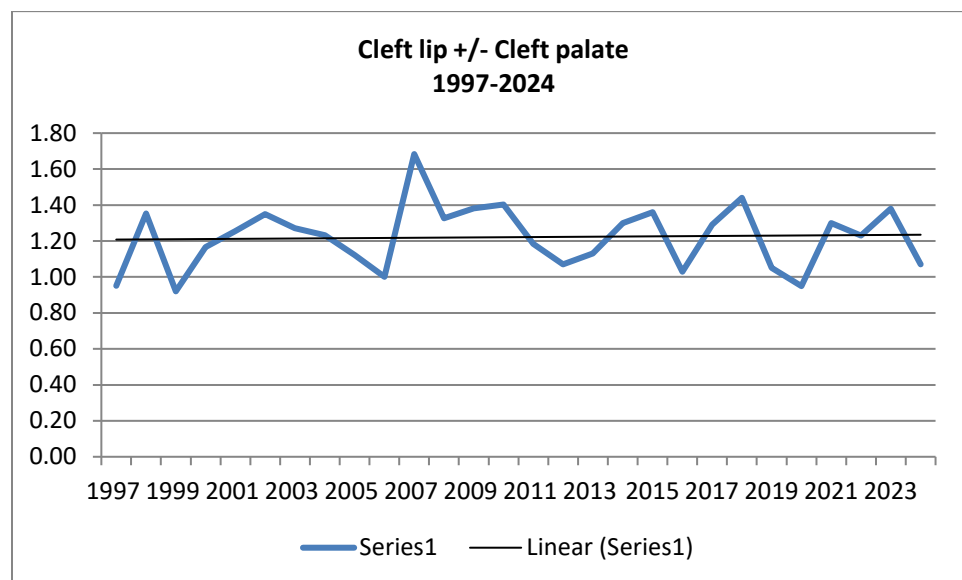
A comparison of the prevalence of CP and CL/P with other congenital anomalies networks and jurisdictions are reported in Table 5.2.4. The ACASS prevalence for both CP and CL/P are significantly more than those reported by CCASS, Texas, and EUROCAT. While the precise reason for this difference is unknown, potential causes could be more complete ascertainment of OFCs by ACASS, or this could be a true difference due to inherent population factors such as a genetic predisposition and/or increased exposures to associated environmental factors (Babai & Irving, 2023). Further study would be required to determine the factors contributing to the relatively high prevalence of OFCs in Alberta.

A population-based trend study using the EUROCAT Netherlands registry data (2002-2021) showed stable overall prevalence rates but showed a significant decrease in livebirths with CL/P that they attributed to increasing prenatal diagnosis and termination rather than a reduction in occurrence (Anania et al., 2025).

A recent cross-sectional population-based retrospective study using live birth data provided by the National Center for Health Statistics (NCHS) from the Centers for Disease Control and Prevention (CDC) showed stable trends from 2016-2021. It identified significant associations between CL/P and maternal age (20-24 years), smoking, obesity, pre-pregnancy hypertension, pre-pregnancy diabetes mellitus and the use of assisted reproductive technology (Heydari et al., 2024).

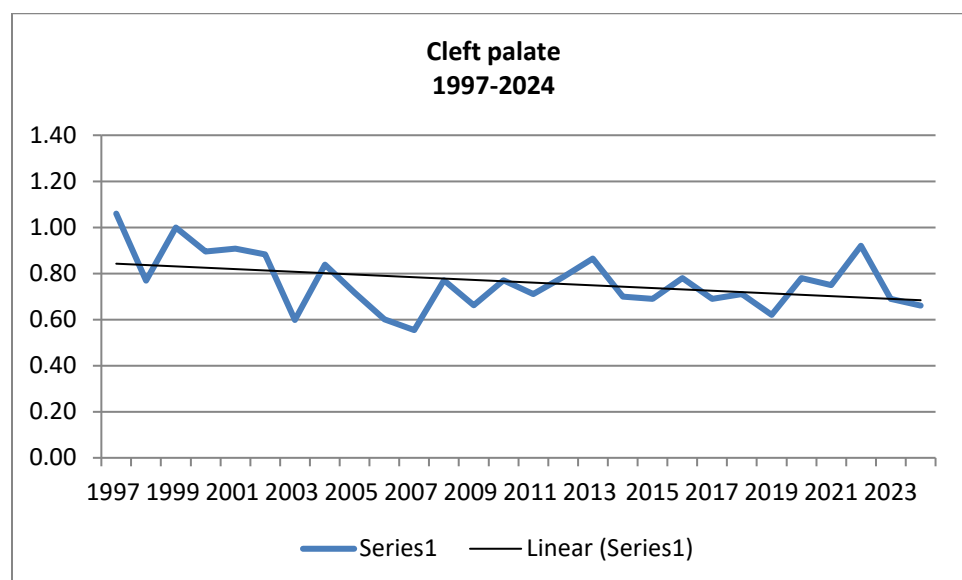
There have been no new publications regarding potential protective factors for OFC, and it is still considered a possible folic acid sensitive anomaly (Wilson et al., 2022).

Figure 5.2.4: Cleft Lip with and without Cleft Palate, 1997-2024 (Rate per 1,000 total births)



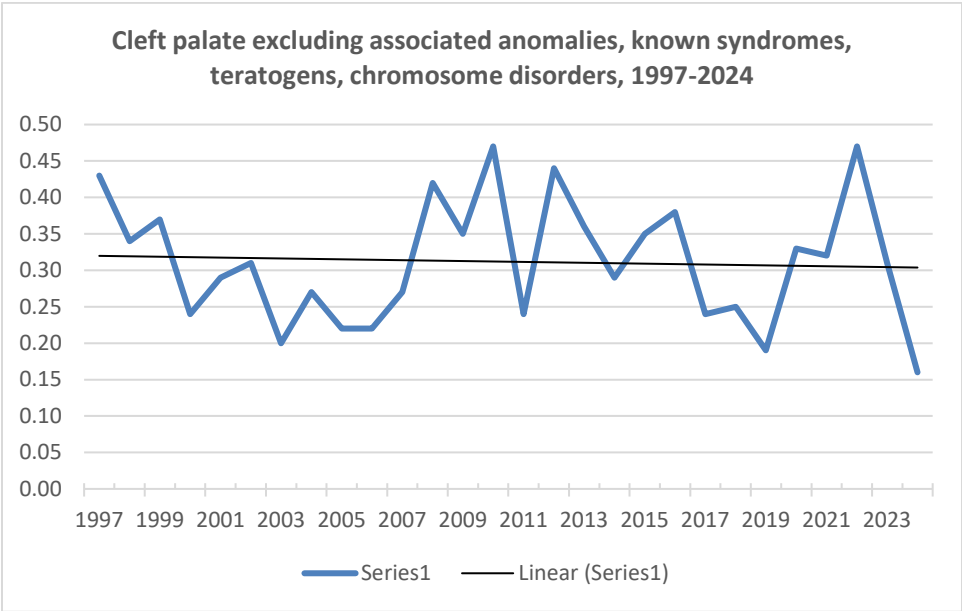
p=1.0000

Figure 5.2.5: Cleft Palate, 1997-2024 (Rate per 1,000 total births)



p=0.0979

Figure 5.2.6: Isolated Cleft Palate, 1997-2024 (Rate per 1,000 total births)



p=0.6892

Table 5.2.4: A Comparison of Orofacial Cleft Prevalence per 10,000 Total Births

Orofacial Cleft	ACASS (2000-2024)	CCASS (2008-2023)	Texas (1999-2022)	EUROCAT (2000-2023)
Cleft Palate Only	7.39 (6.92-7.90)	6.38 (6.17-6.58)	5.94 (5.78-6.10)	6.08 (5.96-6.20)
Cleft Lip with or Without Cleft Palate	12.39 (11.77-13.04)	9.93 (9.59-10.29)	10.72 (10.51-10.94)	9.14 (9.00-9.29)

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European Commission, European Platform on Rare Disease Registration, Prevalence Charts and Tables, [Prevalence charts and tables | European Platform on Rare Disease Registration](#) (accessed July 16, 2026).

Heydari MH, Sadeghian A, Khadivi G, Mustafa HJ, Javinani A, Nadjmi N, Khojasteh A. (2024). Prevalence, trend, and associated risk factors for cleft lip with/without cleft palate: a national study on live births from 2016 to 2021. *BMC Oral Health*, 24, 36. <https://doi.org/10.1186/s12903-023-03797-z>

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Texas Health and Human Services, Table 1, Overall Prevalence of Selected Birth Defects, Texas, 1999-2022 [Table 1. Overall Prevalence of Selected Birth Defects, Texas, 1999–2022](#) (accessed July 16, 2026).

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5.2.5 Obstructive Genitourinary Defects

Congenital anomalies of the kidney and urinary tract (CAKUT) represent a diverse group of anomalies. This section includes obstructive genitourinary (GU) anomalies with ICD-10 RCPCH codes Q62.0-62.3 (hydronephrosis, atresia or stenosis of the ureter, and other obstructive defects of the renal pelvis and ureter), Q64.2 (posterior urethral valves) and Q64.3 (other atresia and stenosis of urethra and bladder neck). Not included are other CAKUT anomalies such as renal agenesis or hypogenesis, or multicystic dysplastic kidney. This separation is felt to help study trends and risk factors for more homogeneous types of anomalies within the CAKUT group.

There is a statistically significant increase ($p < 0.0001$) of congenital obstructive GU tract anomalies from 1997-2024 (Figure 5.2.7). Hydronephrosis accounts for the majority of cases with obstructive GU anomalies (63%) and may be largely driving this upward trend ($p < 0.0001$). However, ureteropelvic junction (UPJ) and vesicoureteric junction (VUJ) obstructions also have increased trends that are statistically significant; $p < 0.0001$ and $p = 0.0025$ respectively. Of note, the annual case numbers with VUJ obstruction are very low, ranging from zero to six, thus this reported increase should be interpreted with caution.

The overall increase may be due to more frequent and accurate diagnostic imaging techniques. Multiple studies state that the upward trend in CAKUT, including obstructive GU anomalies, is reflecting improved prenatal detection on ultrasound rather than a true increase in occurrence (Li et al., 2024; van der Zanden et al., 2023). National estimates from 13 US population-based surveillance programs from 2016-2020, showed no statistically significant change in obstructive GU defect prevalence (Stallings et al., 2024).

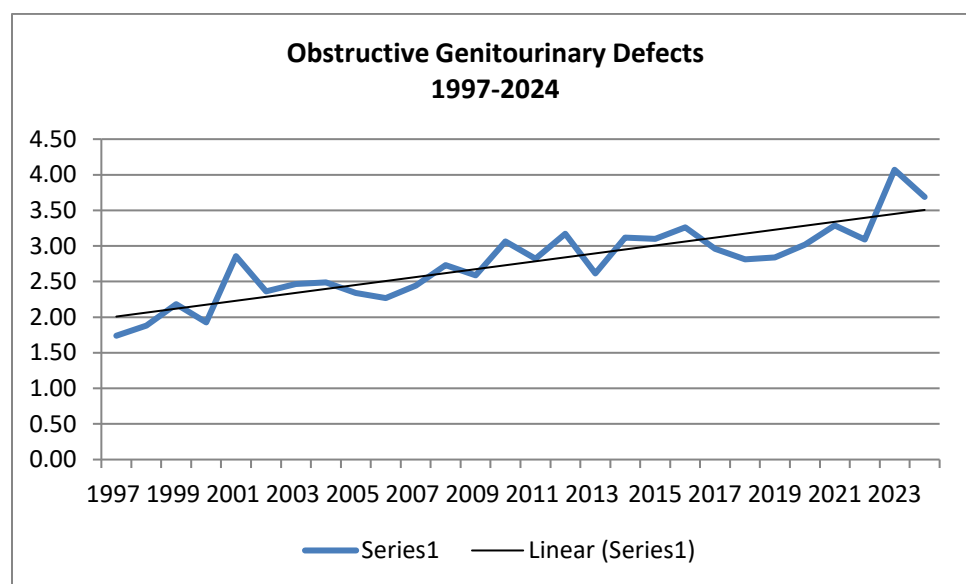
There is no routine follow up by ACASS of cases with obstructive GU anomalies, which may be particularly relevant for hydronephrosis. While only 2% of cases with hydronephrosis from 1997-2024 were detected by prenatal ultrasound without postnatal confirmation (e.g. early fetal loss with limited or no autopsy), even the ones confirmed postnatally may require further follow-up by ACASS as some may resolve spontaneously (Capolicchio et al., 2018; Gatto et al., 2024). Additionally, ACASS includes cases if the diagnostic imaging report states “hydronephrosis” regardless of measurement, if reported. Other congenital anomalies surveillance systems and networks such as EUROCAT have specific definitions for hydronephrosis which is based on a renal pelvis at or above 10mm after birth (EUROCAT, 2025). Thus, a change of ACASS practice for hydronephrosis may be needed to ensure that only cases with persistent hydronephrosis and a defined minimum measurement are included. The ICBDSR does not report obstructive GU anomalies.

A population-based case control study to assess maternal risk factors for posterior urethral valves (PUV) identifying all liveborn boys with PUV in the Netherlands from 1990-2019 failed to identify modifiable maternal risk factors and supports the hypothesis that PUV have strong genetic risk factors over environmental risk factors (van der Zanden, 2023).

A population-based data-linkage cohort study using nine EUROCAT congenital anomaly registries over a 20-year period assessed outcomes in children born with anomalies that included

congenital hydronephrosis and PUV, amongst others (Garne et al., 2025). For children with PUV, 18.3% had a diagnosis of kidney failure and 5% had a kidney transplantation by the age of 5 years. Children with congenital hydronephrosis had a low absolute risk of kidney failure (3% by age 5 years) but 13 times more likely to receive antihypertensive medication by age 10 years. Antibiotic prescriptions were high in early childhood, reflecting urinary tract infection burden associated with obstructive GU anomalies. (Garne et al., 2025).

Figure 5.2.7: Obstructive Genitourinary Defects, 1997-2024 (Rate per 1,000 total births)



p<0.0001

References

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5.2.6 Congenital Limb Reductions:

While ACASS routinely reports anomaly rates as in Appendix A.3, we can also report case rates. This is particularly relevant for congenital limb reductions since one case may have multiple types of limb reductions (e.g. one case can present with absent right 4th and 5th metacarpals and phalanges, hypoplastic right fibula, and absent right 3rd-5th metatarsals and phalanges).

The overall trends for upper and lower limb reduction cases and anomalies are showing statistically significant increases from 1997-2024 (Figures 5.2.8 – 5.2.11) with annual fluctuations. Cases, when compared with anomaly rates, have a greater increase suggesting that more cases are being identified with a congenital limb reduction rather than cases identified with more of these anomalies. Upper limb reduction anomalies, and cases numbers are showing a greater increase than lower limb reductions.

The assessment of data quality for the years 2020 through 2024 is reported in Section 4, including for limb reductions. All data quality indicators (DQIs) were achieved for this type of congenital anomaly as the minimum prevalence per 10,000 total births (TB) is greater than 3.8 (ACASS prevalence - 10.9) and the prevalence of upper limb reduction cases (ACASS prevalence - 7.01) is greater than the prevalence of lower limb reduction cases (ACASS prevalence - 3.85). Although the DQI for prevalence by axis was achieved by ACASS, with transverse (4.92) greater than preaxial (2.38) which is greater than postaxial (0.86), this comparison is done with caution because there is no consensus as to the classification of congenital limb reductions, particularly by axis (Lowry & Bedard, 2016). The description DQI was also achieved, as 90% of the cases on ACASS were specified by type and localization since we use X-ray reports, photographs, pathology reports (where applicable) and orthopedic consultations to code the specific bone(s) affected.

The overall limb reduction case prevalence is 7.8/10,000 TB for 2000-2024 and the anomaly rate, 11.7/10,000 TB. The anomaly rate is significantly higher than the overall Canadian rate of 4.0/10,000 TB for the years 2008 through 2023 (Public Health Agency of Canada, 2025).

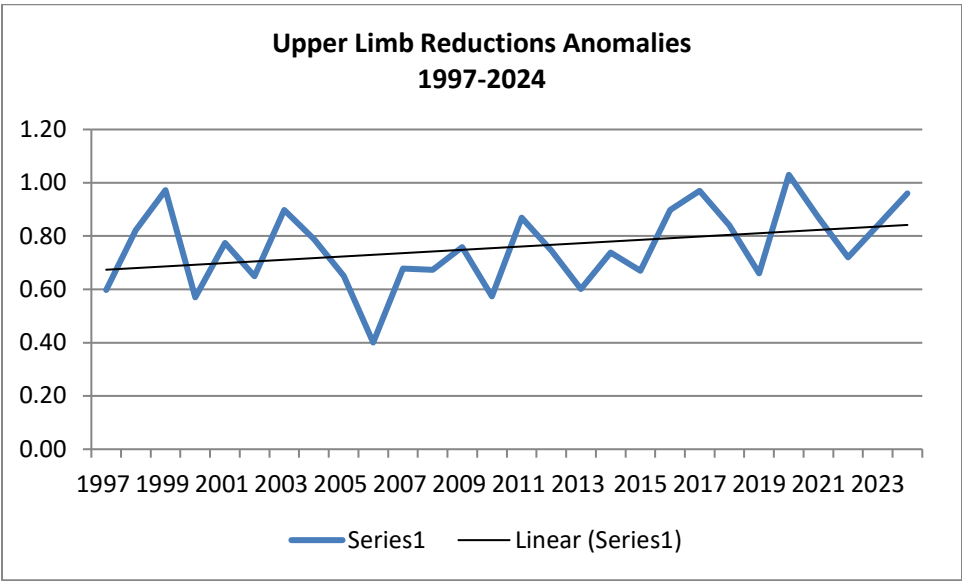
Although historically and in the most recent ICBDSR 2024 Report the prevalence of congenital limb reduction is based on an anomaly rate for ACASS, Table 5.2.5 includes the case rates and comparisons with other member programs. The prevalence of limb reductions varies widely, with Alberta having the highest prevalence of cases.

Bedard et al (2015) reviewed congenital limb reduction cases ascertained by ACASS, born between 1980 and 2012. The authors reported a case prevalence of 5.6/10,000 TB with no significant trend however, they only included cases with absence or severe hypoplasia that had a post-natal confirmation. The numbers presented in this report reflect all cases with a congenital limb reduction including those with mild hypoplasia, and/or with only a prenatal diagnosis. This increase may also reflect more complete case/anomaly ascertainment since ACASS is currently using Connect Care provincial reports for active case ascertainment. Active case ascertainment requires ACASS staff to go through clinic lists to identify eligible cases. This type of case ascertainment improves case detection and data quality (World Health Organization, 2020).

Nevertheless, further study may be warranted to confirm and/or establish specific factors contributing to the reported increased prevalence of congenital limb reductions in Alberta. The prevalence of isolated limb reduction cases (i.e. excluding cases with multiple congenital anomalies, known syndromes, chromosome abnormalities, and teratogens) is also increasing and is statistically significant ($p=0.0123$) Figure 5.2.12.

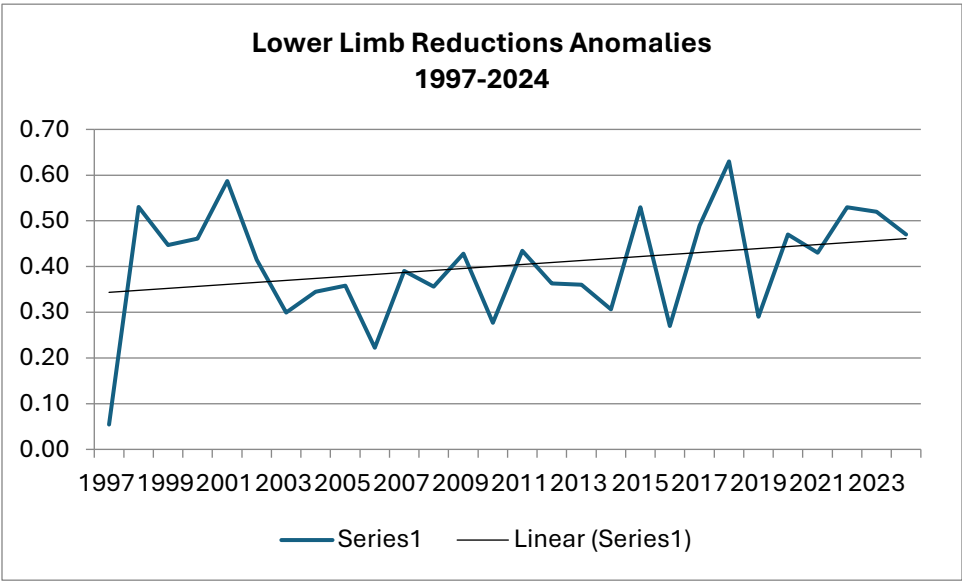
Although disruptive causes such as amniotic bands and the historical teratogen thalidomide are well-established, other contributing factors are largely unknown and are thought to be multifactorial. A relatively recent population-based case control study from Finland reported that pregestational diabetes, primiparity, young maternal age less than 25 years, and male sex were all associated with an increased risk of non-syndromic limb deficiencies (Syvänen et al., 2021). Maternal antiepileptic medication was associated with all limb deficiencies and with isolated cases (Syvänen et al., 2021).

Figure 5.2.8: Upper Limb Reduction Anomalies, 1997-2024 (Rate per 1,000 Total Births)



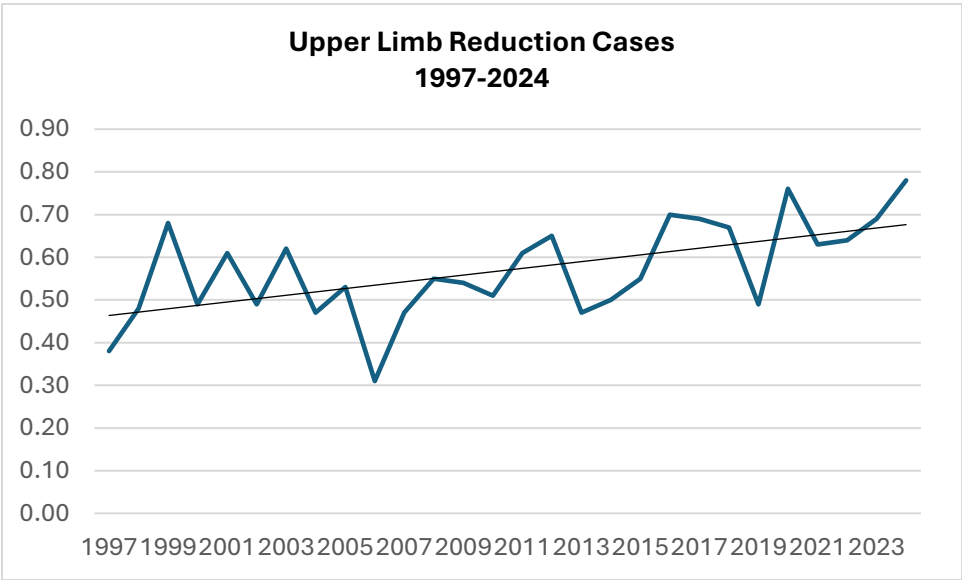
$p=0.0286$

Figure 5.2.9: Lower Limb Reduction Anomalies, 1997-2024 (Rate per 1,000 Total Births)



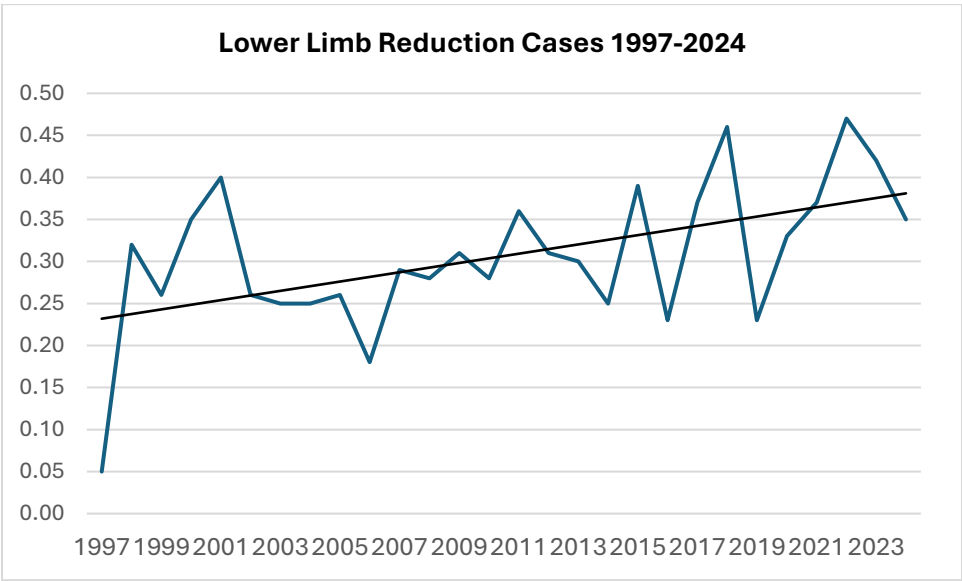
p=0.0437

Figure 5.2.10: Upper Limb Reduction Cases, 1997-2024 (Rate per 1,000 Total Births)



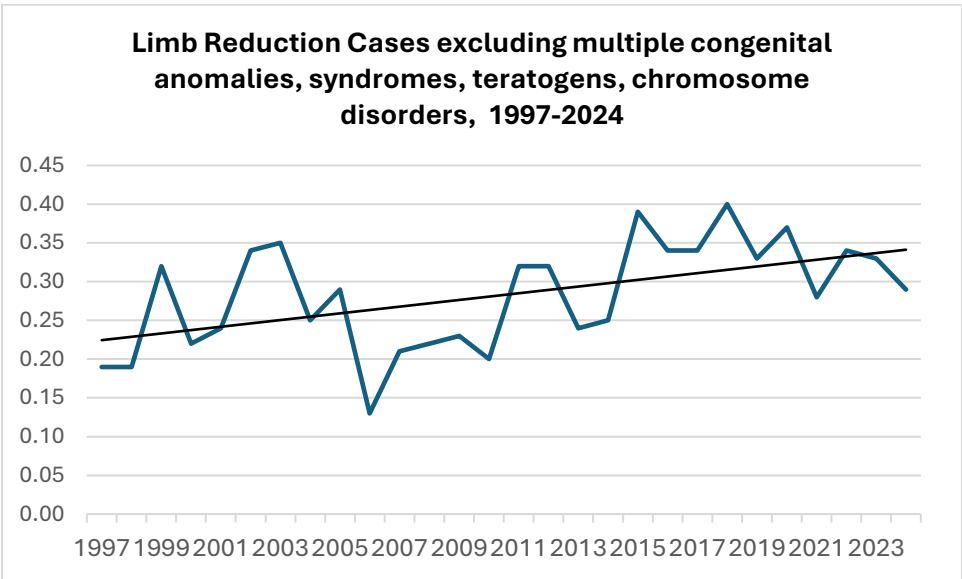
p=0.0023

Figure 5.2.11: Lower Limb Reduction Cases, 1997-2024 (Rate per 1,000 Total Births)



p=0.0049

Figure 5.2.12: Isolated Limb Reduction Cases, 1997-2024 (Rate per 1,000 Total Births)



p=0.0123

Table 5.2.5: Limb Reduction Case Prevalence for Alberta and other Jurisdictions Reporting to the ICBDSR, 2015-2020, prevalence per 10,000 Total Births

Jurisdiction	Prevalence	95% CI
Alberta	9.1	7.9-10.3
Texas	5.7	5.4-6.1
United Kingdom	2.5	2.3-2.7
Sweden	3.9	3.5-4.4
Germany	6.6	5.2-8.4

References:

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5.3 Summary

ACASS reviews anomalies that have been entered into the database on a regular basis. Detailed studies of some individual anomalies or anomaly groups aid in the assessment and maintenance of the data quality. With intensive review, some cases might be reassigned, recoded or discarded altogether from the database. This continuing review might explain some discrepancies in the data from earlier reports.

6. Surveillance and Research Projects since 2020

6.1 Peer Reviewed Surveillance and Research Publications

1. Lowry RB, Bedard T, Crawford S, Grevers X, Bernier FP, Thomas MA. **(2020)**. Prevalence rates study of selected isolated non-Mendelian congenital anomalies in the Hutterite population of Alberta, 1980-2016. *Am J Med Genet A*, 182(11):2594-2604. <https://doi.org/10.1002/ajmg.a.61834>
2. Machiraju P, Degtiarev V, Patel D, Hazari H, Lowry RB, Bedard T, . . . Khan A. **(2022)**. Phenotype and pathology of the dilated cardiomyopathy with ataxia syndrome in children. *J Inherit Metab Dis* 45(2):366-376. <https://doi.org/10.1002/jimd.12441>
3. Lowry RB, Bedard T, Grevers X, Crawford S, Greenway SC, Brindle ME, Sarnat HB, Harrop AR, Kiefer GN, Thomas MA. **(2023)**. The Alberta Congenital Anomalies Surveillance System: a 40-year review with prevalence and trends for selected congenital anomalies, 1997,2019. *Health Promot Chronic Dis Prev Can*, 43(1):40-48. <https://doi.org/10.24095/hpcdp.43.1.04>
4. Thomas MA, Bedard T, Crawford S, Grevers X, Lowry RB. **(2023)**. Craniofacial microsomia, associated congenital anomalies, and risk factors in 63 cases from the Alberta Congenital Anomalies Surveillance System. *J Pediatr*, 261:113528. <https://doi.org/10.1016/j.jpeds.2023.113528>
5. Bedard T, Lowry RB, Crawford S, Wang T, Bakal J, Metcalfe A, Harrop AR, Grevers X, Thomas MA. **(2024)**. Publicly funded healthcare costs associated with orofacial clefts for children born in Alberta, Canada between 2002 and 2018. *Birth Defects Res* <https://doi.org/10.1002/bdr2.2295>
6. Thomas MA, Bedard T, Crawford S, Lowry RB. **(2024)**. Prenatal findings in 11 cases with craniofacial microsomia using the Alberta Congenital Anomalies Surveillance System, 1997-2019. *Am J Med Genet A*, 194(8):e63594. <https://doi.org/10.1002/ajmg.a.63594>
7. Feldkamp ML, Canfield MA, Krikov S, Preto-Merino, Šipek A, LeLong N, . . . Thomas MA, . . . Botto LD. **(2024)**. Gastroschisis prevalence patterns in 27 surveillance programs from 24 countries, International Clearinghouse for Birth Defects Surveillance and Research, 1980-2017. *Birth Defects Res*, 116(2):e2306. <https://doi.org/10.1002/bdr2.2306>
8. Bedard T, Nava de Escalante Y, Cole C, Dang K, Jeyaraman M, Johnston K, Miao Q, Rickert L, Nelson C. **(2024)**. Provincial and territorial congenital anomalies surveillance: a summary of surveillance programs across Canada. *HPCDP Journal*, 44(5):229-235. <https://doi.org/10.24095/hpcdp.44.5.04>

9. Nava de Escalante Y, Bedard T, Cole C, Dang K, Jeyaraman M, Johnston K, Miao Q, Rickert L. **(2025)**. Innovation through collaboration: identifying opportunities to improve congenital anomalies surveillance in Canada. Can J Public Health, 116(3):456-463. <https://doi.org/10.17269/s41997-024-00949-8>
10. Thomas MA, Crawford S, Bedard T. **(2025)**. A population-based study of limb body wall complex with proposed features for prenatal diagnosis. Am J Med Genet A, 197(11):e64150. <https://doi.org/10.1002/ajmg.a.64140>

Alberta Congenital Anomalies Surveillance System

7. Appendices

Appendix A.1 ACASS Data Flow

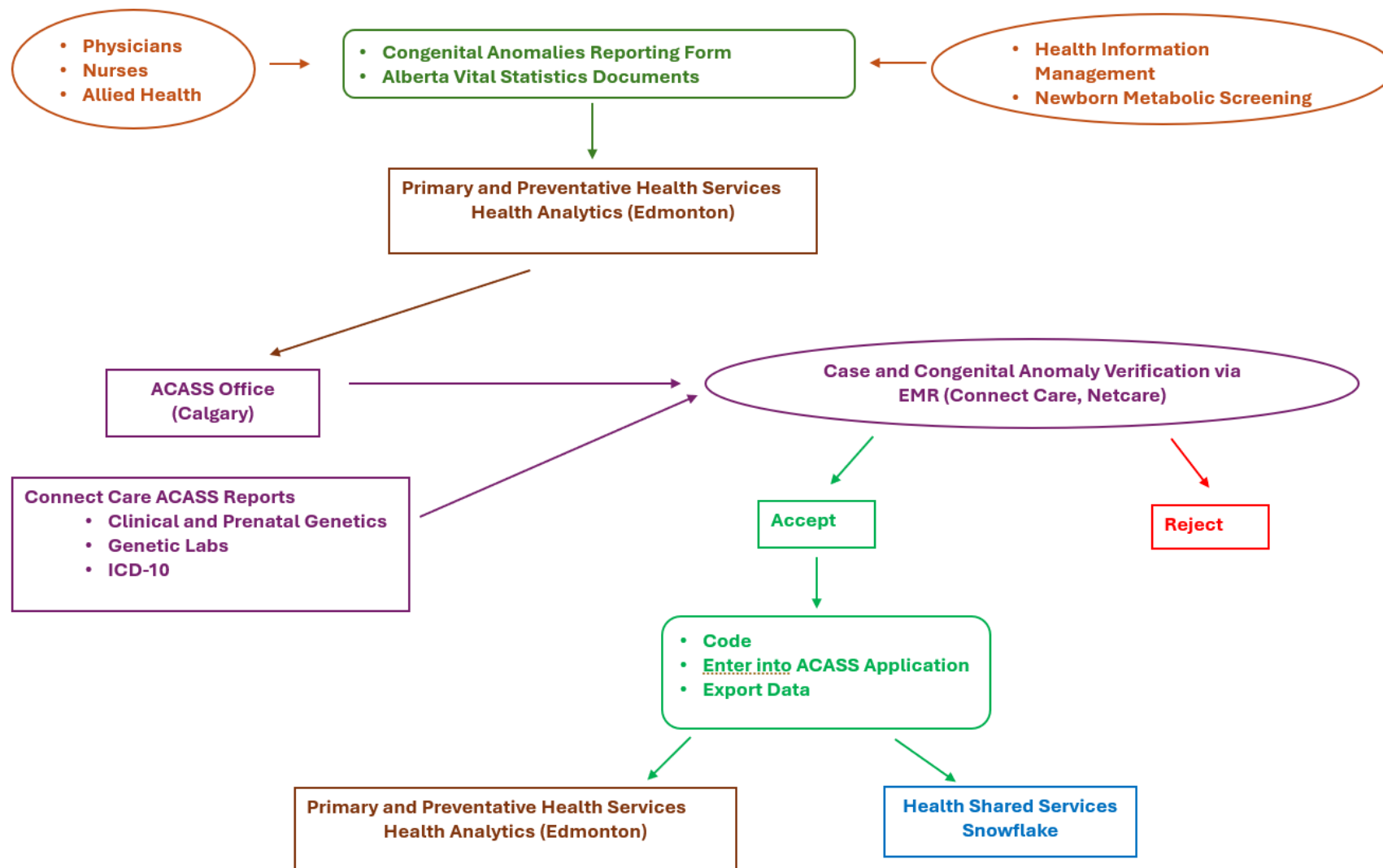
Appendix A.2 Congenital Anomaly(ies) Reporting Form (CARF)

Appendix A.3 Alberta Congenital Anomalies Surveillance System Anomaly Rates

Appendix A.4 Numbers of cases, anomalies and anomalies per case 1997–2024

Appendix A.5 Chi Trend table for reported anomalies 1997-2024

Appendix A.1 The Alberta Congenital Anomalies Surveillance System Routine Flow of Data



Appendix A.2 Congenital Anomaly(ies) Reporting Form (CARF)



Death Reg No	Birth Reg No
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Addressograph

Mail parts one and two to:
Alberta Health and Wellness
Surveillance and Environmental Health Branch
PO Box 1360 Stn Main
Edmonton AB T5J 2N3

Congenital Anomaly(ies) Reporting

Fetus / Infant		PLEASE PRINT CLEARLY	
Name (Last, First, Initial)		Date of Birth Month by Name Day Year	
Gender ☐ Male ☐ Female ☐ Unknown	Type of Birth ☐ Livebirth ☐ Stillbirth ☐ Fetus less than 20 weeks gestation	Name of Hospital of Birth	
Birthweight Grams	Gestation Age (Completed Weeks)	Location of Hospital of Birth (City/Town)	
Child's Personal Health Number		Attending Physician's Name	
Plurality of Birth ☐ Single ☐ Twin ☐ Triplets ☐ First ☐ Second ☐ Third		Physician Responsible for Ongoing Care (if different from above)	

Parents		Total Number of
Mother's Name (Last, First, Maiden)	Date of Birth or Age (if DOB unavailable) Month by Name Day Year	Livebirths
Permanent Address	Mother's Personal Health Number	Stillbirths
City/Town	Postal Code	Spontaneous Abortions
Father's Name (Last, First, Initial)	Date of Birth or Age (if DOB unavailable) Month by Name Day Year	Therapeutic Abortions

Reporting Hospital/Agency/Clinic		
Name	Infant's Admission (If different from birthdate) Month by Name Day Year	Infant's Discharge Month by Name Day Year
Location (City/Town)	Month by Name Day Year	Infant's Death (If Applicable) Month by Name Day Year

Full description of Congenital Anomaly(ies) and/or **SYNDROME DIAGNOSES** (If necessary, please attach supporting documents.)

Completed by	Position	Date
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HS0020-112 (2008/06)

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Appendix A.3

Alberta Congenital Anomalies Surveillance System Anomaly Rates

RCPCH version ICD-10 Q-Chapter (Q00-Q99)

**Single and Aggregate Year Anomaly Rates per
1,000 Total Births (live births + stillbirths)**

**Numerator
(live births, stillbirths and fetal losses)**

Appendix A.3

Single Year Anomaly Rates per 1,000 Total Births (live births + stillbirths)

Numerator (live births, stillbirths and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		2014	2015	2016	2017	2018	2019	2020	2021	2022	2023	2024
Anencephaly ICD-10 Q00.00, Q00.01, Q00.1	NUMBER	16	14	6	8	9	13	8	11	10	7	7
	RATE	0.29	0.25	0.11	0.15	0.17	0.25	0.16	0.22	0.21	0.15	0.14
	Lower CI	0.17	0.14	0.04	0.06	0.08	0.13	0.07	0.11	0.10	0.06	0.05
	Upper CI	0.47	0.41	0.23	0.29	0.33	0.43	0.32	0.40	0.39	0.30	0.28
Spina Bifida without Anencephaly ICD-10 Q05..	NUMBER	17	14	25	18	20	24	18	18	19	24	14
	RATE	0.31	0.25	0.45	0.34	0.38	0.47	0.37	0.37	0.40	0.50	0.27
	Lower CI	0.18	0.14	0.29	0.20	0.23	0.30	0.22	0.22	0.24	0.32	0.15
	Upper CI	0.49	0.41	0.67	0.53	0.59	0.69	0.58	0.58	0.63	0.75	0.46
Encephalocele ICD-10 Q01..	NUMBER	4	3	10	6	7	5	7	6	7	8	10
	RATE	0.07	0.05	0.18	0.11	0.13	0.10	0.14	0.12	0.15	0.17	0.20
	Lower CI	0.02	0.01	0.09	0.04	0.05	0.03	0.06	0.04	0.06	0.07	0.09
	Upper CI	0.18	0.15	0.33	0.24	0.27	0.22	0.29	0.26	0.30	0.33	0.36
Neural Tube Defects (all) ICD-10 Q00.., Q01.., Q05..	NUMBER	37	31	42	32	37	43	33	35	36	39	31
	RATE	0.67	0.55	0.76	0.60	0.71	0.84	0.68	0.71	0.77	0.81	0.61
	Lower CI	0.47	0.37	0.55	0.41	0.50	0.60	0.47	0.50	0.54	0.58	0.41
	Upper CI	0.92	0.78	1.02	0.85	0.98	1.13	0.95	0.99	1.06	1.11	0.86
Hydrocephalus without Spina Bifida (Excludes hydranencephaly) ICD-10 Q03	NUMBER	21	20	28	16	13	15	17	9	23	15	10
	RATE	0.38	0.35	0.50	0.30	0.25	0.29	0.35	0.18	0.49	0.31	0.20
	Lower CI	0.23	0.22	0.34	0.17	0.13	0.16	0.20	0.08	0.31	0.18	0.09
	Upper CI	0.58	0.55	0.73	0.49	0.42	0.48	0.56	0.34	0.73	0.52	0.36
Arrhinencephaly/ Holoprosencephaly ICD-10 Q04.1, Q04.2, Q87.03	NUMBER	8	16	17	22	11	15	6	6	12	12	13
	RATE	0.14	0.28	0.31	0.41	0.21	0.29	0.12	0.12	0.26	0.25	0.25
	Lower CI	0.06	0.16	0.18	0.26	0.11	0.16	0.05	0.04	0.13	0.13	0.14
	Upper CI	0.28	0.46	0.49	0.62	0.38	0.48	0.27	0.26	0.44	0.44	0.43
Microcephaly ICD-10 Q02	NUMBER	15	22	15	20	20	13	16	14	20	17	20
	RATE	0.27	0.39	0.27	0.37	0.38	0.25	0.33	0.28	0.43	0.36	0.39
	Lower CI	0.15	0.24	0.15	0.23	0.23	0.13	0.19	0.16	0.26	0.21	0.24
	Upper CI	0.44	0.59	0.45	0.58	0.59	0.43	0.53	0.48	0.66	0.57	0.60
Anophthalmia/Microphthalmia ICD-10 Q11.0, Q11.1, Q11.2	NUMBER	6	6	10	13	10	14	3	7	4	10	10
	RATE	0.11	0.11	0.18	0.24	0.19	0.27	0.06	0.14	0.09	0.21	0.20
	Lower CI	0.04	0.04	0.09	0.13	0.09	0.15	0.01	0.06	0.02	0.10	0.09
	Upper CI	0.23	0.23	0.33	0.42	0.35	0.46	0.17	0.29	0.21	0.38	0.36
Congenital cataract ICD-10 Q12.0	NUMBER	10	4	8	8	13	8	2	4	8	6	8
	RATE	0.18	0.07	0.14	0.15	0.25	0.16	0.04	0.08	0.17	0.13	0.16
	Lower CI	0.09	0.02	0.06	0.06	0.13	0.07	0.00	0.02	0.07	0.05	0.07
	Upper CI	0.33	0.18	0.28	0.29	0.42	0.30	0.14	0.20	0.33	0.27	0.31

Appendix A.3

Aggregate Year Anomaly Rates per 1,000 Total Births (live births + stillbirths)
Numerator (live births, stillbirths and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		2019- 2021 (3 years)	2022- 2024 (3 years)	2005- 2014 (10 years)	2015- 2024 (10 years)	2000- 2024 (25 years)
Anencephaly ICD-10 Q00.00, Q00.01, Q00.1	NUMBER	32	24	118	93	259
	RATE	0.21	0.16	0.24	0.18	0.21
	Lower CI	0.15	0.11	0.20	0.15	0.19
	Upper CI	0.30	0.24	0.28	0.22	0.24
Spina Bifida without Anencephaly ICD-10 Q05..	NUMBER	60	57	201	194	453
	RATE	0.40	0.39	0.40	0.38	0.38
	Lower CI	0.31	0.30	0.35	0.33	0.34
	Upper CI	0.52	0.51	0.46	0.44	0.41
Encephalocele ICD-10 Q01..	NUMBER	18	25	61	69	158
	RATE	0.12	0.17	0.12	0.13	0.13
	Lower CI	0.07	0.11	0.09	0.10	0.11
	Upper CI	0.19	0.25	0.16	0.17	0.15
Neural Tube Defects (all) ICD-10 Q00.., Q01.., Q05..	NUMBER	111	106	381	359	874
	RATE	0.74	0.73	0.76	0.70	0.72
	Lower CI	0.61	0.59	0.69	0.63	0.68
	Upper CI	0.90	0.88	0.84	0.78	0.77
Hydrocephalus without Spina Bifida (Excludes hydranencephaly) ICD-10 Q03	NUMBER	41	48	276	166	548
	RATE	0.27	0.33	0.55	0.32	0.45
	Lower CI	0.20	0.24	0.49	0.28	0.42
	Upper CI	0.37	0.44	0.62	0.38	0.49
Arrhinencephaly/ Holoprosencephaly ICD-10 Q04.1, Q04.2, Q87.03	NUMBER	27	37	117	130	284
	RATE	0.18	0.25	0.23	0.25	0.24
	Lower CI	0.12	0.18	0.19	0.21	0.21
	Upper CI	0.26	0.35	0.28	0.30	0.26
Microcephaly ICD-10 Q02	NUMBER	43	57	206	177	456
	RATE	0.29	0.39	0.41	0.34	0.38
	Lower CI	0.21	0.30	0.36	0.30	0.34
	Upper CI	0.39	0.51	0.47	0.40	0.41
Anophthalmia/Microphthalmia ICD-10 Q11.0, Q11.1, Q11.2	NUMBER	24	24	75	87	190
	RATE	0.16	0.16	0.15	0.17	0.16
	Lower CI	0.10	0.11	0.12	0.14	0.14
	Upper CI	0.24	0.24	0.19	0.21	0.18
Congenital cataract ICD-10 Q12.0	NUMBER	14	22	79	69	164
	RATE	0.09	0.15	0.16	0.13	0.14
	Lower CI	0.05	0.09	0.13	0.10	0.12
	Upper CI	0.16	0.13	0.20	0.17	0.16

Appendix A.3

Single Year Anomaly Rates per 1,000 Total Births (live births + stillbirths)
Numerator (live births, stillbirths and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		2014	2015	2016	2017	2018	2019	2020	2021	2022	2023	2024
Anotia/Microtia ICD-10 Q16.0, Q17.2	NUMBER	11	12	18	24	11	10	13	7	17	10	16
	RATE	0.20	0.21	0.32	0.45	0.21	0.19	0.27	0.14	0.36	0.21	0.31
	Lower CI	0.10	0.11	0.19	0.29	0.11	0.09	0.14	0.06	0.21	0.10	0.18
	Upper CI	0.35	0.37	0.51	0.67	0.38	0.36	0.46	0.29	0.58	0.38	0.51
Congenital Heart Defects (all) ICD-10 Q20.. to Q26..	NUMBER	678	752	729	676	758	611	595	564	610	738	807
	RATE	12.21	13.30	13.14	12.66	14.51	11.87	12.24	11.45	12.99	15.42	15.76
	Lower CI	11.31	12.37	12.20	11.72	13.49	10.94	11.28	10.53	11.98	14.32	14.69
	Upper CI	13.17	14.29	14.13	13.65	15.58	12.85	13.26	12.44	14.06	16.57	16.89
Common Truncus (Excludes AP window) ICD-10 Q20.0	NUMBER	3	3	8	7	3	4	3	4	5	3	2
	RATE	0.05	0.05	0.14	0.13	0.06	0.08	0.06	0.08	0.11	0.06	0.04
	Lower CI	0.01	0.01	0.06	0.05	0.01	0.02	0.01	0.02	0.03	0.01	0.00
	Upper CI	0.15	0.15	0.28	0.27	0.16	0.19	0.17	0.20	0.24	0.18	0.13
Transposition of Great Arteries ICD-10 Q20.11, Q20.3, Q20.5	NUMBER	29	25	22	19	26	19	23	10	17	22	29
	RATE	0.52	0.44	0.40	0.36	0.50	0.37	0.47	0.20	0.36	0.46	0.57
	Lower CI	0.35	0.29	0.25	0.21	0.33	0.22	0.30	0.10	0.21	0.29	0.38
	Upper CI	0.75	0.65	0.60	0.56	0.73	0.58	0.71	0.37	0.58	0.70	0.81
Tetralogy of Fallot (Includes Tetralogy with ASD aka Pentalogy of Fallot) ICD-10 Q21.3.., Q21.82	NUMBER	15	9	19	18	19	16	11	23	30	22	20
	RATE	0.27	0.16	0.34	0.34	0.36	0.31	0.23	0.47	0.64	0.46	0.39
	Lower CI	0.15	0.07	0.21	0.20	0.22	0.18	0.11	0.30	0.43	0.29	0.24
	Upper CI	0.44	0.30	0.53	0.53	0.57	0.50	0.40	0.70	0.91	0.70	0.60
Ventricular Septal Defect ICD-10 Q21.0	NUMBER	163	164	187	176	180	157	136	132	119	156	232
	RATE	2.94	2.90	3.37	3.30	3.45	3.05	2.80	2.68	2.53	3.26	4.53
	Lower CI	2.50	2.48	2.91	2.83	2.96	2.59	2.35	2.24	2.10	2.77	3.97
	Upper CI	3.42	3.38	3.89	3.82	3.99	3.56	3.31	3.18	3.03	3.81	5.15
Atrial Septal Defect ICD-10 Q21.1..	NUMBER	123	130	108	116	108	86	95	72	84	87	91
	RATE	2.22	2.30	1.95	2.17	2.07	1.67	1.95	1.46	1.79	1.82	1.78
	Lower CI	1.84	1.92	1.60	1.80	1.70	1.34	1.58	1.14	1.43	1.46	1.43
	Upper CI	2.64	2.73	2.35	2.61	2.50	2.06	2.39	1.84	2.21	2.24	2.18
Endocardial Cushion Defect ICD-10 Q21.2..	NUMBER	33	30	25	23	34	21	28	32	36	41	40
	RATE	0.59	0.53	0.45	0.43	0.65	0.41	0.58	0.65	0.77	0.86	0.78
	Lower CI	0.41	0.36	0.29	0.77	0.45	0.25	0.38	0.44	0.54	0.62	0.56
	Upper CI	0.84	0.76	0.67	0.65	0.91	0.62	0.83	0.92	1.06	1.16	1.06
Pulmonary Valve Atresia and Stenosis ICD-10 Q22.0, Q22.1	NUMBER	41	38	33	35	50	39	37	39	35	48	45
	RATE	0.74	0.67	0.59	0.66	0.96	0.76	0.76	0.79	0.75	1.00	0.88
	Lower CI	0.53	0.48	0.41	0.46	0.71	0.54	0.54	0.56	0.52	0.74	0.64
	Upper CI	1.00	0.92	0.84	0.91	1.26	1.05	1.05	1.08	1.04	1.33	1.18

Appendix A.3

Aggregate Year Anomaly Rates per 1,000 Total Births (live births + stillbirths)
Numerator (live births, stillbirths and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		2019- 2021 (3 years)	2022- 2024 (3 years)	2005- 2014 (10 years)	2015- 2024 (10 years)	2000-2024 (25 years)
Anotia/Microtia ICD-10 Q16.0, Q17.2	NUMBER	30	43	124	138	302
	RATE	0.20	0.29	0.25	0.27	0.25
	Lower CI	0.14	0.21	0.21	0.23	0.22
	Upper CI	0.29	0.40	0.30	0.32	0.28
Congenital Heart Defects (all) ICD-10 Q20.., Q26..	NUMBER	1770	2155	6001	6840	15337
	RATE	11.85	14.76	12.01	13.33	12.71
	Lower CI	11.31	14.14	11.71	13.02	12.51
	Upper CI	12.42	15.39	12.32	13.65	12.92
Common Truncus (Excludes AP window) ICD-10 Q20.0	NUMBER	11	10	36	42	92
	RATE	0.07	0.07	0.07	0.08	0.08
	Lower CI	0.04	0.03	0.05	0.06	0.06
	Upper CI	0.13	0.13	0.10	0.11	0.09
Transposition of Great Arteries ICD-10 Q20.11, Q20.3, Q20.5	NUMBER	52	68	180	212	470
	RATE	0.35	0.47	0.36	0.41	0.39
	Lower CI	0.26	0.36	0.31	0.36	0.36
	Upper CI	0.46	0.59	0.42	0.47	0.43
Tetralogy of Fallot (Includes Tetralogy with ASD aka Pentalogy of Fallot) ICD-10 Q21.3.., Q21.82	NUMBER	50	72	187	187	431
	RATE	0.33	0.49	0.37	0.36	0.36
	Lower CI	0.25	0.39	0.32	0.31	0.32
	Upper CI	0.44	0.62	0.43	0.42	0.39
Ventricular Septal Defect ICD-10 Q21.0	NUMBER	425	507	1551	1639	3827
	RATE	2.85	3.47	3.10	3.19	3.17
	Lower CI	2.58	3.18	2.95	3.04	3.07
	Upper CI	3.13	3.79	3.26	3.35	3.27
Atrial Septal Defect ICD-10 Q21.1..	NUMBER	253	262	941	977	2351
	RATE	1.69	1.79	1.88	1.90	1.95
	Lower CI	1.49	1.58	1.76	1.79	1.87
	Upper CI	1.92	2.02	2.01	2.03	2.03
Endocardial Cushion Defect ICD-10 Q21.2..	NUMBER	81	117	253	310	663
	RATE	0.54	0.80	0.51	0.60	0.55
	Lower CI	0.43	0.66	0.45	0.54	0.51
	Upper CI	0.67	0.96	0.57	0.68	0.59
Pulmonary Valve Atresia and Stenosis ICD-10 Q22.0, Q22.1	NUMBER	115	128	304	399	827
	RATE	0.77	0.88	0.61	0.78	0.69
	Lower CI	0.64	0.73	0.54	0.70	0.64
	Upper CI	0.92	1.04	0.68	0.86	0.73

Appendix A.3

Single Year Anomaly Rates per 1,000 Total Births (live births + stillbirths)

Numerator (live births, stillbirths and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		2014	2015	2016	2017	2018	2019	2020	2021	2022	2023	2024
Tricuspid Valve Atresia and Stenosis	NUMBER	6	5	6	6	4	7	5	4	6	7	3
	RATE	0.11	0.09	0.11	0.11	0.08	0.14	0.10	0.08	0.13	0.15	0.06
	Lower CI	0.04	0.03	0.04	0.04	0.02	0.05	0.03	0.02	0.05	0.06	0.01
	Upper CI	0.23	0.20	0.23	0.24	0.19	0.28	0.24	0.20	0.27	0.30	0.17
ICD-10 Q22.4												
Ebstein's Anomaly	NUMBER	8	3	1	0	5	5	2	1	4	6	3
	RATE	0.14	0.05	0.02	0.00	0.10	0.10	0.04	0.02	0.09	0.13	0.06
	Lower CI	0.06	0.01	0.00	0.00	0.03	0.03	0.00	0.00	0.02	0.05	0.01
	Upper CI	0.28	0.15	0.09	0.04	0.22	0.22	0.14	0.10	0.21	0.27	0.17
ICD-10 Q22.5												
Aortic Valve Atresia/Stenosis (excludes sub & supra aortic stenosis Aortic stenosis found with HLHS)	NUMBER	13	9	11	9	6	10	12	5	11	15	11
	RATE	0.23	0.16	0.20	0.17	0.11	0.19	0.25	0.10	0.23	0.31	0.21
	Lower CI	0.12	0.07	0.10	0.08	0.04	0.09	0.13	0.03	0.12	0.18	0.11
	Upper CI	0.40	0.30	0.35	0.32	0.25	0.36	0.43	0.23	0.42	0.52	0.38
ICD-10 Q23.0												
Hypoplastic Left Heart Syndrome (HLHS)	NUMBER	16	21	14	24	20	17	17	13	12	22	25
	RATE	0.29	0.37	0.25	0.45	0.38	0.33	0.35	0.26	0.26	0.46	0.49
	Lower CI	0.17	0.23	0.14	0.29	0.23	0.19	0.20	0.14	0.13	0.29	0.32
	Upper CI	0.47	0.57	0.42	0.67	0.59	0.53	0.56	0.45	0.44	0.70	0.72
ICD-10 Q23.4												
Coarctation of the Aorta	NUMBER	22	27	33	22	32	22	20	19	18	24	24
	RATE	0.40	0.48	0.59	0.41	0.61	0.43	0.41	0.39	0.38	0.50	0.47
	Lower CI	0.25	0.32	0.41	0.26	0.42	0.27	0.25	0.23	0.23	0.32	0.30
	Upper CI	0.60	0.70	0.84	0.62	0.86	0.65	0.63	0.60	0.61	0.75	0.70
ICD-10 Q25.1..												
Cleft Palate without Cleft Lip (i.e. cleft palate alone)	NUMBER	40	39	43	37	37	32	38	38	43	33	34
	RATE	0.72	0.69	0.78	0.69	0.71	0.62	0.78	0.77	0.92	0.69	0.66
	Lower CI	0.52	0.49	0.56	0.49	0.50	0.43	0.55	0.55	0.66	0.48	0.46
	Upper CI	0.98	0.94	1.04	0.96	0.98	0.88	1.07	1.06	1.23	0.97	0.93
ICD-10 Q35..												
Cleft Lip without Cleft Palate (i.e. cleft lip alone)	NUMBER	27	20	21	22	30	20	17	21	16	19	22
	RATE	0.49	0.35	0.38	0.41	0.57	0.39	0.35	0.43	0.34	0.40	0.43
	Lower CI	0.32	0.22	0.23	0.26	0.39	0.24	0.20	0.26	0.20	0.24	0.27
	Upper CI	0.71	0.55	0.58	0.62	0.82	0.60	0.56	0.65	0.55	0.62	0.65
ICD-10 Q36..												
Cleft Lip and Cleft Palate	NUMBER	45	57	36	47	45	34	29	43	42	47	33
	RATE	0.81	1.01	0.65	0.88	0.86	0.66	0.60	0.87	0.89	0.98	0.64
	Lower CI	0.59	0.76	0.45	0.65	0.63	0.46	0.40	0.63	0.65	0.72	0.44
	Upper CI	1.09	1.31	0.90	1.17	1.15	0.92	0.86	1.18	1.21	1.31	0.91
ICD-10 Q37..												
Cleft Lip with and without Cleft Palate	NUMBER	72	77	57	69	75	54	46	64	58	66	55
	RATE	1.30	1.36	1.03	1.29	1.44	1.05	0.95	1.30	1.23	1.38	1.07
	Lower CI	1.02	1.08	0.78	1.01	1.13	0.79	0.69	1.00	0.94	1.07	0.81
	Upper CI	1.63	1.70	1.33	1.63	1.80	1.37	1.26	1.66	1.60	1.75	1.40
ICD-10 Q36.., Q37..												

Appendix A.3

Aggregate Year Anomaly Rates per 1,000 Total Births (live births + stillbirths)
Numerator (live births, stillbirths and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		2019- 2021 (3 years)	2022- 2024 (3 years)	2005- 2014 (10 years)	2015- 2024 (10 years)	2000-2024 (25 years)
Tricuspid Valve Atresia and Stenosis ICD-10 Q22.4	NUMBER	16	16	43	53	109
	RATE	0.11	0.11	0.09	0.10	0.09
	Lower CI	0.06	0.06	0.06	0.08	0.07
	Upper CI	0.17	0.18	0.12	0.14	0.11
Ebstein's Anomaly ICD-10 Q22.5	NUMBER	8	13	39	30	83
	RATE	0.05	0.09	0.08	0.06	0.07
	Lower CI	0.02	0.05	0.06	0.04	0.05
	Upper CI	0.10	0.15	0.11	0.08	0.09
Aortic Valve Atresia/Stenosis (excludes sub & supra aortic stenosis & Aortic stenosis found with HLHS) ICD-10 Q23.0	NUMBER	27	37	80	99	230
	RATE	0.18	0.25	0.16	0.19	0.19
	Lower CI	0.12	0.18	0.13	0.16	0.17
	Upper CI	0.26	0.35	0.20	0.23	0.22
Hypoplastic Left Heart Syndrome ICD-10 Q23.4	NUMBER	47	59	164	185	402
	RATE	0.31	0.40	0.33	0.36	0.33
	Lower CI	0.23	0.31	0.28	0.31	0.30
	Upper CI	0.42	0.52	0.38	0.42	0.37
Coarctation of the Aorta ICD-10 Q25.1..	NUMBER	61	66	222	241	527
	RATE	0.41	0.45	0.44	0.47	0.44
	Lower CI	0.31	0.35	0.39	0.41	0.40
	Upper CI	0.52	0.57	0.51	0.53	0.48
Cleft Palate without Cleft Lip (i.e. cleft palate alone) ICD-10 Q35..	NUMBER	108	110	359	374	892
	RATE	0.72	0.75	0.72	0.73	0.74
	Lower CI	0.59	0.62	0.65	0.66	0.69
	Upper CI	0.87	0.91	0.80	0.81	0.79
Cleft Lip without Cleft Palate (i.e. cleft lip alone) ICD-10 Q36..	NUMBER	58	57	237	208	527
	RATE	0.39	0.39	0.47	0.41	0.44
	Lower CI	0.30	0.30	0.42	0.35	0.40
	Upper CI	0.50	0.51	0.54	0.46	0.48
Cleft Lip and Cleft Palate ICD-10 Q37..	NUMBER	106	122	394	413	968
	RATE	0.71	0.84	0.79	0.80	0.80
	Lower CI	0.58	0.69	0.71	0.73	0.75
	Upper CI	0.86	1.00	0.87	0.89	0.85
Cleft Lip with and without Cleft Palate ICD-10 Q36.., Q37..	NUMBER	164	179	631	621	1495
	RATE	1.10	1.23	1.26	1.21	1.24
	Lower CI	0.94	1.05	1.17	1.12	1.18
	Upper CI	1.28	1.42	1.37	1.31	1.30

Appendix A.3

Single Year Anomaly Rates per 1,000 Total Births (live births + stillbirths)
Numerator (live births, stillbirths and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		2014	2015	2016	2017	2018	2019	2020	2021	2022	2023	2024
Choanal Atresia/Stenosis	NUMBER	14	5	9	5	6	9	7	8	5	7	4
	RATE	0.25	0.09	0.16	0.09	0.11	0.17	0.14	0.16	0.11	0.15	0.08
	Lower CI	0.14	0.03	0.07	0.03	0.04	0.08	0.06	0.07	0.03	0.06	0.02
	Upper CI	0.42	0.20	0.31	0.22	0.25	0.33	0.29	0.32	0.24	0.30	0.20
	ICD-10 Q30.0..											
Oesophageal Atresia/ Tracheo-oesophageal Fistula	NUMBER	15	7	13	20	26	18	11	13	6	18	23
	RATE	0.27	0.12	0.23	0.37	0.50	0.35	0.23	0.26	0.13	0.38	0.45
	Lower CI	0.15	0.05	0.12	0.23	0.33	0.21	0.11	0.14	0.05	0.22	0.29
	Upper CI	0.44	0.25	0.40	0.58	0.73	0.55	0.40	0.45	0.27	0.59	0.67
	ICD-10 Q39.0 – Q39.4											
Pyloric Stenosis	NUMBER	49	35	29	29	25	20	21	20	7	22	15
	RATE	0.88	0.62	0.52	0.54	0.48	0.39	0.43	0.41	0.15	0.46	0.29
	Lower CI	0.65	0.43	0.35	0.36	0.31	0.24	0.27	0.25	0.06	0.29	0.16
	Upper CI	1.17	0.86	0.75	0.78	0.71	0.60	0.66	0.63	0.30	0.70	0.48
	ICD-10 Q40.0											
Small Intestinal Atresia/Stenosis (all)	NUMBER	22	20	18	28	16	20	15	14	12	15	12
	RATE	0.40	0.35	0.32	0.52	0.31	0.39	0.31	0.28	0.26	0.31	0.23
	Lower CI	0.25	0.22	0.19	0.35	0.18	0.24	0.17	0.16	0.13	0.18	0.12
	Upper CI	0.60	0.55	0.51	0.76	0.50	0.60	0.51	0.48	0.44	0.52	0.41
	ICD-10 Q41...											
Duodenal Atresia/Stenosis	NUMBER	14	12	9	14	9	11	8	10	5	10	8
	RATE	0.25	0.21	0.16	0.26	0.17	0.21	0.16	0.20	0.11	0.21	0.16
	Lower CI	0.14	0.11	0.07	0.14	0.08	0.11	0.07	0.10	0.03	0.10	0.07
	Upper CI	0.42	0.37	0.31	0.44	0.33	0.38	0.32	0.37	0.24	0.38	0.31
	ICD-10 Q41.0...											
Rectal and Large Intestinal Atresia/Stenosis (all)	NUMBER	18	21	27	38	34	25	20	17	21	30	18
	RATE	0.32	0.37	0.49	0.71	0.65	0.49	0.41	0.35	0.45	0.63	0.35
	Lower CI	0.19	0.23	0.32	0.50	0.45	0.31	0.25	0.20	0.28	0.42	0.21
	Upper CI	0.51	0.57	0.71	0.98	0.91	0.72	0.63	0.55	0.68	0.89	0.55
	ICD-10 Q42..											
Rectal Atresia/Stenosis	NUMBER	1	1	2	2	4	2	2	1	0	2	1
	RATE	0.02	0.02	0.04	0.04	0.08	0.04	0.04	0.02	0.00	0.04	0.02
	Lower CI	0.00	0.00	0.00	0.00	0.02	0.00	0.00	0.00	0.00	0.00	0.00
	Upper CI	0.09	0.09	0.12	0.13	0.19	0.13	0.14	0.10	0.04	0.14	0.10
	ICD-10 Q42.0..., Q42.1...											
Anal Atresia/Stenosis	NUMBER	14	19	21	32	27	21	17	14	18	26	16
	RATE	0.25	0.34	0.38	0.60	0.52	0.41	0.35	0.28	0.38	0.54	0.31
	Lower CI	0.14	0.20	0.23	0.41	0.34	0.25	0.20	0.16	0.23	0.36	0.18
	Upper CI	0.42	0.52	0.58	0.85	0.75	0.62	0.56	0.48	0.61	0.80	0.51
	ICD-10 Q42.2..., Q42.3...											
Other Large Intestinal Atresia/Stenosis	NUMBER	3	1	4	4	3	2	1	2	3	2	1
	RATE	0.05	0.02	0.07	0.07	0.06	0.04	0.02	0.04	0.06	0.04	0.02
	Lower CI	0.01	0.00	0.02	0.02	0.01	0.00	0.00	0.00	0.01	0.00	0.00
	Upper CI	0.15	0.09	0.18	0.19	0.16	0.13	0.10	0.14	0.18	0.14	0.10
	ICD-10 Q42.8..., Q42.9...											

Appendix A.3

Aggregate Year Anomaly Rates per 1,000 Total Births (live births + stillbirths)
Numerator (live births, stillbirths and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		2019- 2021 (3 years)	2022- 2024 (3 years)	2005- 2014 (10 years)	2015- 2024 (10 years)	2000-2024 (25 years)
Choanal Atresia/Stenosis ICD-10 Q30.0..	NUMBER	24	16	76	65	182
	RATE	0.16	0.11	0.15	0.13	0.15
	Lower CI	0.10	0.06	0.12	0.10	0.13
	Upper CI	0.24	0.18	0.19	0.16	0.17
Oesophageal Atresia/ Tracheo-oesophageal Fistula ICD-10 Q39.0 – Q39.4	NUMBER	42	47	127	155	322
	RATE	0.28	0.32	0.25	0.30	0.27
	Lower CI	0.20	0.24	0.21	0.26	0.24
	Upper CI	0.38	0.43	0.30	0.35	0.30
Pyloric Stenosis ICD-10 Q40.0	NUMBER	61	44	468	223	870
	RATE	0.41	0.30	0.94	0.43	0.72
	Lower CI	0.31	0.22	0.85	0.38	0.67
	Upper CI	0.52	0.40	1.03	0.50	0.77
Small Intestinal Atresia/ Stenosis (all) ICD-10 Q41...	NUMBER	49	39	176	170	412
	RATE	0.33	0.27	0.35	0.33	0.34
	Lower CI	0.24	0.19	0.30	0.28	0.31
	Upper CI	0.43	0.37	0.41	0.39	0.38
Duodenal Atresia/Stenosis ICD-10 Q41.0...	NUMBER	29	23	99	96	229
	RATE	0.19	0.16	0.20	0.19	0.19
	Lower CI	0.13	0.10	0.16	0.15	0.17
	Upper CI	0.28	0.24	0.24	0.23	0.22
Rectal and Large Intestinal Atresia/Stenosis (all) ICD-10 Q42..	NUMBER	62	69	214	251	611
	RATE	0.42	0.47	0.43	0.49	0.51
	Lower CI	0.32	0.37	0.37	0.43	0.47
	Upper CI	0.53	0.60	0.49	0.55	0.55
Rectal Atresia/Stenosis ICD-10 Q42.0....., Q42.1....	NUMBER	5	3	13	17	44
	RATE	0.03	0.02	0.03	0.03	0.04
	Lower CI	0.01	0.00	0.01	0.02	0.03
	Upper CI	0.08	0.06	0.04	0.05	0.05
Anal Atresia/Stenosis ICD-10 Q42.2....., Q42.3....	NUMBER	52	60	176	211	507
	RATE	0.35	0.41	0.35	0.41	0.42
	Lower CI	0.26	0.31	0.30	0.36	0.38
	Upper CI	0.46	0.53	0.41	0.47	0.46
Other Large Intestinal Atresia/Stenosis ICD-10 Q42.8....., Q42.9....	NUMBER	3	6	25	17	51
	RATE	0.02	0.04	0.05	0.03	0.04
	Lower CI	0.00	0.02	0.03	0.02	0.03
	Upper CI	0.06	0.09	0.07	0.05	0.06

Appendix A.3

Single Year Anomaly Rates per 1,000 Total Births (live births + stillbirths)
Numerator (live births, stillbirths and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		2014	2015	2016	2017	2018	2019	2020	2021	2022	2023	2024
Hirschsprung Disease	NUMBER	6	6	7	7	8	9	10	4	7	10	4
	RATE	0.11	0.11	0.13	0.13	0.15	0.17	0.21	0.08	0.15	0.21	0.08
	Lower CI	0.04	0.04	0.05	0.05	0.07	0.08	0.10	0.02	0.06	0.10	0.02
	Upper CI	0.23	0.23	0.26	0.27	0.30	0.33	0.38	0.20	0.30	0.38	0.20
ICD-10 Q43.1..												
Biliary Atresia	NUMBER	4	3	5	2	5	6	1	0	4	2	1
	RATE	0.07	0.05	0.09	0.04	0.10	0.12	0.02	0.00	0.09	0.04	0.02
	Lower CI	0.02	0.01	0.03	0.00	0.03	0.04	0.00	0.00	0.02	0.00	0.00
	Upper CI	0.18	0.15	0.21	0.13	0.22	0.25	0.10	0.04	0.21	0.14	0.10
ICD-10 Q44.2												
Undescended Testes (denominator MALE births only) (>36 weeks gestation)	NUMBER	146	191	151	170	169	123	133	125	139	108	108
	RATE	5.14	6.63	5.34	6.17	6.32	4.69	5.34	4.93	5.77	4.40	4.10
	Lower CI	4.34	5.72	4.52	5.28	5.41	3.90	4.48	4.11	4.85	3.61	3.37
	Upper CI	6.04	7.64	6.26	7.17	7.35	5.59	6.33	5.87	6.81	5.31	4.95
ICD-10 Q53...												
Hypospadias (denominator MALE births only)	NUMBER	189	169	149	154	133	126	144	120	144	115	126
	RATE	6.65	5.86	5.27	5.59	4.97	4.80	5.79	4.73	4.73	4.69	4.79
	Lower CI	5.74	5.01	4.46	4.74	4.17	4.00	4.88	3.93	3.90	3.87	3.99
	Upper CI	7.67	6.82	6.18	6.54	5.90	5.72	6.81	5.66	5.68	5.62	5.70
ICD-10 Q54 (excl. Q54.4)												
Epispadias (denominator MALE births only)	NUMBER	4	3	2	0	0	1	0	3	1	2	1
	RATE	0.14	0.10	0.07	0.00	0.00	0.04	0.00	0.12	0.04	0.08	0.04
	Lower CI	0.04	0.02	0.01	0.00	0.00	0.00	0.00	0.02	0.00	0.01	0.00
	Upper CI	0.35	0.29	0.24	0.08	0.08	0.19	0.08	0.34	0.21	0.28	0.19
ICD-10 Q64.0												
Renal Agenesis/Hypoplasia	NUMBER	30	27	43	47	40	42	38	42	49	48	36
	RATE	0.54	0.48	0.78	0.88	0.77	0.82	0.78	0.85	1.04	1.00	0.70
	Lower CI	0.37	0.32	0.56	0.65	0.55	0.59	0.55	0.62	0.77	0.74	0.49
	Upper CI	0.77	0.70	1.04	1.17	1.04	1.10	1.07	1.15	1.38	1.33	0.97
ICD-10 Q60..												
Cystic Kidney (exclude single renal cyst Q61.0) Q61..	NUMBER	36	51	41	39	47	39	45	42	36	52	39
	RATE	0.65	0.90	0.74	0.73	0.90	0.76	0.93	0.85	0.77	1.09	0.76
	Lower CI	0.45	0.67	0.53	0.52	0.66	0.54	0.68	0.62	0.54	0.81	0.54
	Upper CI	0.90	1.19	1.00	1.00	1.20	1.04	1.24	1.15	1.06	1.42	1.04
ICD-10 Q61..												
Bladder Exstrophy	NUMBER	2	2	1	1	1	1	1	2	1	0	1
	RATE	0.04	0.04	0.02	0.02	0.02	0.02	0.02	0.04	0.02	0.00	0.02
	Lower CI	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00
	Upper CI	0.12	0.12	0.09	0.09	0.09	0.10	0.10	0.14	0.11	0.04	0.10
ICD-10 Q64.1 (excl Q64.10)												
Obstructive Genitourinary Defects (All)	NUMBER	173	176	181	158	147	146	147	165	145	195	189
	RATE	3.12	3.11	3.26	2.96	2.81	2.84	3.02	3.35	3.09	4.07	3.69
	Lower CI	2.67	2.67	2.81	2.52	2.38	2.39	2.56	2.86	2.61	3.52	3.18
	Upper CI	3.62	3.61	3.77	3.46	3.31	3.33	3.55	3.90	3.63	4.69	4.26
ICD-10 Q62.0 – Q62.3, Q64.2, Q64.3												

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Aggregate Year Anomaly Rates per 1,000 Total Births (live births + stillbirths)
Numerator (live births, stillbirths and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		2019- 2021 (3 years)	2022- 2024 (3 years)	2005- 2014 (10 years)	2015- 2024 (10 years)	2000- 2024 (25 years)
Hirschsprung Disease ICD-10 Q43.1..	NUMBER	23	21	82	72	177
	RATE	0.15	0.14	0.16	0.14	0.15
	Lower CI	0.10	0.09	0.13	0.11	0.13
	Upper CI	0.23	0.22	0.20	0.18	0.17
Biliary Atresia ICD-10 Q44.2	NUMBER	7	7	34	29	75
	RATE	0.05	0.05	0.07	0.06	0.06
	Lower CI	0.02	0.02	0.05	0.04	0.05
	Upper CI	0.10	0.10	0.10	0.08	0.08
Undescended Testes <i>(denominator MALE births only)</i> <i>(>36 weeks gestation)</i> ICD-10 Q53....	NUMBER	381	355	1351	1417	3267
	RATE	4.98	4.73	5.27	5.39	5.28
	Lower CI	4.49	4.26	4.99	5.11	5.10
	Upper CI	5.51	5.25	5.56	5.68	5.47
Hypospadias <i>(denominator MALE births only)</i> ICD-10 Q54 (excl. Q54.4)	NUMBER	390	355	1260	1350	3009
	RATE	5.10	4.73	4.91	5.14	4.87
	Lower CI	4.61	4.26	4.65	4.87	4.69
	Upper CI	5.63	5.25	5.19	5.42	5.04
Epispadias <i>(denominator MALE births only)</i> ICD-10 Q64.0	NUMBER	4	4	39	13	66
	RATE	0.05	0.05	0.15	0.05	0.11
	Lower CI	0.01	0.01	0.11	0.03	0.08
	Upper CI	0.13	0.13	0.21	0.08	0.14
Renal Agenesis/Hypoplasia ICD-10 Q60..	NUMBER	122	133	299	412	827
	RATE	0.82	0.91	0.60	0.80	0.69
	Lower CI	0.68	0.76	0.53	0.73	0.64
	Upper CI	0.98	1.08	0.67	0.88	0.73
Cystic Kidney <i>(excludes single renal cyst Q61.0)</i> ICD-10 Q61..	NUMBER	126	127	363	431	950
	RATE	0.84	0.87	0.73	0.84	0.79
	Lower CI	0.70	0.73	0.65	0.76	0.74
	Upper CI	1.00	1.03	0.81	0.92	0.84
Bladder Exstrophy ICD-10 Q64.1 (excl Q64.10)	NUMBER	4	2	13	11	33
	RATE	0.03	0.01	0.03	0.02	0.03
	Lower CI	0.01	0.00	0.01	0.01	0.02
	Upper CI	0.07	0.05	0.04	0.04	0.04
Obstructive Genitourinary Defects (All) ICD-10 Q62.0 – Q62.3, Q64.2, Q64.3	NUMBER	458	529	1367	1649	3485
	RATE	3.07	3.62	2.74	3.21	2.89
	Lower CI	3.79	3.32	2.59	3.06	2.79
	Upper CI	3.36	3.94	2.88	3.37	2.99

Appendix A.3

Single Year Anomaly Rates per 1,000 Total Births (live births + stillbirths)
Numerator (live births, stillbirths and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		2014	2015	2016	2017	2018	2019	2020	2021	2022	2023	2024
Hydronephrosis ICD-10 Q62.0..	NUMBER	99	104	106	101	86	85	94	96	94	116	122
	RATE	1.78	1.84	1.91	1.89	1.65	1.65	1.93	1.95	2.00	2.42	2.38
	Lower CI	1.45	1.50	1.56	1.54	1.32	1.32	1.56	1.58	1.62	2.00	1.98
	Upper CI	2.17	2.23	2.31	2.30	2.03	2.04	2.37	2.38	2.45	2.91	2.84
Pelviureteric Junction Obstruction ICD-10 Q62.10 & Q62.11	NUMBER	12	18	19	17	16	20	15	18	19	20	23
	RATE	0.22	0.32	0.34	0.32	0.31	0.39	0.31	0.37	0.40	0.42	0.45
	Lower CI	0.11	0.19	0.21	0.19	0.18	0.24	0.17	0.22	0.24	0.26	0.29
	Upper CI	0.38	0.50	0.53	0.51	0.50	0.60	0.51	0.58	0.63	0.64	0.67
Vesicoureteric Junction Obstruction ICD-10 Q62.12 & Q62.13	NUMBER	0	3	3	1	1	3	4	4	0	6	4
	RATE	0.00	0.05	0.05	0.02	0.02	0.06	0.08	0.08	0.00	0.13	0.08
	Lower CI	0.00	0.01	0.01	0.00	0.00	0.01	0.02	0.02	0.00	0.05	0.02
	Upper CI	0.04	0.15	0.15	0.09	0.10	0.16	0.21	0.20	0.04	0.27	0.20
Posterior Urethral Valves (denominator MALE births only) ICD-10 Q64.20	NUMBER	9	8	2	1	5	6	6	5	2	5	1
	RATE	0.32	0.28	0.07	0.04	0.19	0.23	0.24	0.20	0.08	0.20	0.04
	Lower CI	0.15	0.12	0.01	0.00	0.06	0.08	0.09	0.06	0.01	0.07	0.00
	Upper CI	0.60	0.54	0.24	0.18	0.43	0.49	0.52	0.45	0.29	0.47	0.19
Congenital Deformities Hip (All) ICD-10 Q65	NUMBER	75	79	50	54	52	47	54	42	34	31	36
	RATE	1.35	1.40	0.90	1.01	1.00	0.91	1.11	0.85	0.72	0.65	0.70
	Lower CI	1.06	1.11	0.67	0.76	0.74	0.67	0.84	0.62	0.50	0.44	0.49
	Upper CI	1.69	1.74	1.19	1.32	1.30	1.21	1.45	1.14	1.01	0.92	0.97
Congenital Hip Dislocation Subluxation and Dysplasia ICD-10 Q65.0-Q65.5 & Q65.80-Q65.81	NUMBER	59	70	45	49	43	45	49	40	33	26	35
	RATE	1.06	1.24	0.81	0.92	0.82	0.87	1.01	0.81	0.70	0.54	0.80
	Lower CI	0.81	0.97	0.59	0.68	0.60	0.64	0.75	0.58	0.48	0.36	0.48
	Upper CI	1.37	1.56	1.09	1.21	1.11	1.17	1.33	1.11	0.99	0.80	0.95
Reduction Deformity, Upper Limbs ICD-10 Q71..	NUMBER	41	38	50	52	44	34	50	43	34	40	49
	RATE	0.74	0.67	0.90	0.97	0.84	0.66	1.03	0.87	0.72	0.84	0.96
	Lower CI	0.53	0.48	0.67	0.73	0.61	0.46	0.76	0.63	0.50	0.60	0.71
	Upper CI	1.00	0.92	1.19	1.28	1.13	0.92	1.36	1.18	1.01	1.14	1.27
Reduction Deformity, Lower Limbs ICD-10 Q72..	NUMBER	17	30	15	26	33	15	23	21	25	25	24
	RATE	0.31	0.53	0.27	0.49	0.63	0.29	0.47	0.43	0.53	0.52	0.47
	Lower CI	0.18	0.36	0.15	0.32	0.44	0.16	0.30	0.26	0.34	0.34	0.30
	Upper CI	0.49	0.76	0.45	0.71	0.89	0.48	0.71	0.65	0.79	0.77	0.70
Diaphragmatic Hernia ICD-10 Q79.0, Q79.11, Q79.12	NUMBER	15	18	17	20	13	19	20	17	17	22	23
	RATE	0.27	0.32	0.31	0.37	0.25	0.37	0.41	0.35	0.36	0.46	0.45
	Lower CI	0.15	0.19	0.18	0.23	0.13	0.22	0.25	0.20	0.21	0.29	0.29
	Upper CI	0.44	0.50	0.49	0.58	0.42	0.58	0.63	0.55	0.58	0.70	0.67

Appendix A.3

Aggregate Year Anomaly Rates per 1,000 Total Births (live births + stillbirths)
Numerator (live births, stillbirths and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		2019- 2021 (3 years)	2022- 2024 (3 years)	2005- 2014 (10 years)	2015- 2024 (10 years)	2000- 2024 (25 years)
Hydronephrosis ICD-10 Q62.0..	NUMBER	275	332	896	1004	2192
	RATE	1.84	2.27	1.79	1.96	1.82
	Lower CI	1.63	2.04	1.68	1.84	1.74
	Upper CI	2.07	2.53	1.91	2.08	1.89
Pelviureteric Junction Obstruction ICD-10 Q62.10 & Q62.11	NUMBER	53	62	95	185	321
	RATE	0.35	0.42	0.19	0.36	0.27
	Lower CI	0.27	0.33	0.15	0.31	0.24
	Upper CI	0.46	0.54	0.23	0.42	0.30
Vesicoureteric Junction Obstruction ICD-10 Q62.12 & Q62.13	NUMBER	11	10	22	29	55
	RATE	0.07	0.07	0.04	0.06	0.05
	Lower CI	0.04	0.03	0.03	0.04	0.03
	Upper CI	0.13	0.13	0.07	0.08	0.06
Posterior Urethral Valves <i>(denominator MALE births only)</i> ICD-10 Q64.20	NUMBER	17	8	46	41	102
	RATE	0.22	0.11	0.18	0.16	0.16
	Lower CI	0.13	0.05	0.13	0.11	0.13
	Upper CI	0.36	0.21	0.24	0.21	0.20
Congenital Deformities Hip (All) ICD-10 Q65	NUMBER	143	101	988	479	1885
	RATE	0.96	0.69	1.98	0.93	1.56
	Lower CI	0.81	0.56	1.86	0.85	1.49
	Upper CI	1.13	0.84	2.10	1.02	1.63
Congenital Hip Dislocation, Subluxation and Dysplasia ICD-10 Q65.0-Q65.5 & Q65.80- Q65.81	NUMBER	134	94	698	435	1418
	RATE	0.90	0.64	1.40	0.85	1.18
	Lower CI	0.75	0.52	1.30	0.77	1.12
	Upper CI	1.06	0.79	1.50	0.93	1.24
Reduction Deformity, Upper Limbs ICD-10 Q71..	NUMBER	127	123	336	434	914
	RATE	0.85	0.84	0.67	0.85	0.76
	Lower CI	0.71	0.70	0.60	0.77	0.71
	Upper CI	1.01	1.00	0.75	0.93	0.81
Reduction Deformity, Lower Limbs ICD-10 Q72..	NUMBER	59	74	175	237	494
	RATE	0.40	0.51	0.35	0.46	0.41
	Lower CI	0.30	0.40	0.30	0.41	0.37
	Upper CI	0.51	0.64	0.41	0.52	0.45
Diaphragmatic Hernia ICD-10 Q79.0.., Q79.11, Q79.12	NUMBER	56	62	170	186	436
	RATE	0.37	0.42	0.34	0.36	0.36
	Lower CI	0.28	0.33	0.29	0.31	0.33
	Upper CI	0.49	0.54	0.40	0.42	0.40

Appendix A.3

Single Year Anomaly Rates per 1,000 Total Births (live births + stillbirths)
Numerator (live births, stillbirths and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		2014	2015	2016	2017	2018	2019	2020	2021	2022	2023	2024
Abdominal Wall Defects (all)	NUMBER	48	48	45	52	45	47	29	43	41	49	57
	RATE	0.86	0.85	0.81	0.97	0.86	0.91	0.60	0.87	0.87	1.02	1.11
	Lower CI	0.64	0.63	0.59	0.73	0.63	0.67	0.40	0.63	0.63	0.76	0.84
	Upper CI	1.15	1.13	1.09	1.28	1.15	1.21	0.86	1.18	1.18	1.35	1.44
	ICD-10 Q79.2 to Q79.5											
Omphalocele	NUMBER	19	22	21	27	20	24	17	23	25	27	33
	RATE	0.34	0.39	0.38	0.51	0.38	0.47	0.35	0.47	0.53	0.56	0.64
	Lower CI	0.21	0.24	0.23	0.33	0.23	0.30	0.20	0.30	0.34	0.37	0.44
	Upper CI	0.53	0.59	0.58	0.74	0.59	0.69	0.56	0.70	0.79	0.82	0.91
	ICD-10 Q79.2											
Gastroschisis	NUMBER	25	19	11	19	20	16	8	12	14	13	8
	RATE	0.45	0.34	0.20	0.36	0.38	0.31	0.16	0.24	0.30	0.27	0.16
	Lower CI	0.29	0.20	0.10	0.21	0.23	0.18	0.07	0.13	0.16	0.14	0.07
	Upper CI	0.66	0.52	0.35	0.56	0.59	0.50	0.32	0.42	0.50	0.46	0.31
	ICD-10 Q79.3											
All Chromosome Anomalies	NUMBER	288	279	282	311	323	308	299	308	293	380	373
	RATE	5.19	4.94	5.08	5.82	6.18	5.98	6.15	6.25	6.24	7.94	7.28
	Lower CI	4.61	4.37	4.51	5.20	5.53	5.33	5.47	5.57	5.55	7.16	6.56
	Upper CI	5.82	5.55	5.71	6.51	6.90	6.69	6.89	6.99	7.00	8.78	8.06
	ICD-10 Q90-Q99											
Trisomy 13	NUMBER	12	19	13	17	13	17	8	18	16	24	19
	RATE	0.22	0.34	0.23	0.32	0.25	0.33	0.16	0.37	0.34	0.50	0.37
	Lower CI	0.11	0.20	0.12	0.19	0.13	0.19	0.07	0.22	0.20	0.32	0.22
	Upper CI	0.38	0.52	0.40	0.51	0.42	0.53	0.32	0.58	0.55	0.75	0.58
	ICD-10 Q91.4-Q91.7											
Trisomy 18	NUMBER	36	44	33	35	52	29	33	31	33	64	58
	RATE	0.65	0.78	0.59	0.66	1.00	0.56	0.68	0.63	0.70	1.34	1.13
	Lower CI	0.45	0.57	0.41	0.46	0.74	0.38	0.47	0.43	0.48	1.03	0.86
	Upper CI	0.90	1.05	0.84	0.91	1.30	0.81	0.95	0.89	0.99	1.71	1.46
	ICD-10 Q91.0-Q91.3											
Down Syndrome (Trisomy 21)	NUMBER	140	128	120	134	132	126	123	111	113	155	165
	RATE	2.52	2.26	2.16	2.51	2.53	2.45	2.53	2.25	2.41	3.24	3.22
	Lower CI	2.12	1.89	1.79	2.10	2.11	2.04	2.10	1.85	1.98	2.75	2.75
	Upper CI	2.98	2.69	2.59	2.97	3.00	2.92	3.02	2.71	2.89	3.79	3.75
	ICD-10 Q90..											

Appendix A.3

Aggregate Year Anomaly Rates per 1,000 Total Births (live births + stillbirths)

Numerator (live births, stillbirths and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		2019- 2021 (3 years)	2022- 2024 (3 years)	2005- 2014 (10 years)	2015- 2024 (10 years)	2000- 2024 (25 years)
Abdominal Wall Defects (all) ICD-10 Q79.2-Q79.5	NUMBER	119	147	472	456	1071
	RATE	0.80	1.01	0.94	0.89	0.89
	Lower CI	0.66	0.85	0.86	0.81	0.84
	Upper CI	0.95	1.18	1.03	0.97	0.94
Omphalocele ICD-10 Q79.2	NUMBER	64	85	185	239	476
	RATE	0.43	0.58	0.37	0.47	0.39
	Lower CI	0.33	0.47	0.32	0.41	0.36
	Upper CI	0.55	0.72	0.43	0.53	0.43
Gastroschisis ICD-10 Q79.3	NUMBER	36	35	235	140	438
	RATE	0.24	0.24	0.47	0.27	0.36
	Lower CI	0.17	0.17	0.41	0.23	0.33
	Upper CI	0.33	0.33	0.53	0.32	0.40
All Chromosome Anomalies ICD-10 Q90-Q99	NUMBER	915	1046	2436	3156	6374
	RATE	6.13	7.16	4.87	6.15	5.28
	Lower CI	5.74	6.73	4.68	5.94	5.16
	Upper CI	6.54	7.61	5.07	6.37	5.42
Trisomy 13 ICD-10 Q91.4-Q91.7	NUMBER	43	59	144	164	344
	RATE	0.29	0.40	0.29	0.32	0.29
	Lower CI	0.21	0.31	0.24	0.27	0.26
	Upper CI	0.39	0.52	0.34	0.37	0.32
Trisomy 18 ICD-10 Q91.0-Q91.3	NUMBER	93	155	299	412	795
	RATE	0.62	1.06	0.60	0.80	0.66
	Lower CI	0.50	0.90	0.53	0.73	0.61
	Upper CI	0.76	1.24	0.67	0.88	0.71
Down Syndrome (Trisomy 21) ICD-10 Q90..	NUMBER	360	433	1165	1307	2851
	RATE	2.41	2.96	2.33	2.55	2.36
	Lower CI	2.17	2.69	2.20	2.41	2.28
	Upper CI	2.67	3.26	2.47	2.69	2.45

Appendix A.4

Numbers of Cases, Anomalies and Anomalies per Case 1997–2024
Live Births (L), Stillbirths (S) and Fetal losses <20 weeks (T)

Year	Alberta Total Births (L & S)	# Cases (L, S & T)	Case Rate/1000 Total Births	# Anomalies (L, S & T)	Anomaly Rate/1000 Total Births	Average # Anomalies/Case
1997	36797	1126	30.60	1983	53.89	1.76
1998	37715	1193	31.63	2181	57.83	1.83
1999	38044	1222	32.12	2420	63.61	1.98
2000	36860	1288	34.94	2362	64.08	1.83
2001	37460	1384	36.95	2599	69.38	1.88
2002	38532	1373	35.63	2548	66.13	1.86
2003	40118	1516	37.79	2609	65.03	1.72
2004	40557	1550	38.22	2900	71.50	1.87
2005	41856	1609	38.44	2900	69.29	1.80
2006	44947	1620	36.04	2756	61.32	1.70
2007	48708	1872	38.43	3168	65.04	1.69
2008	50516	2005	39.69	3475	68.79	1.73
2009	51420	2090	40.65	3668	71.33	1.76
2010	50590	2187	43.23	3716	73.45	1.70
2011	50662	2086	41.17	3674	72.52	1.76
2012	52318	2133	40.77	3767	72.00	1.77
2013	53180	2154	40.50	3822	71.87	1.77
2014	55506	2192	39.49	3917	70.57	1.79
2015	56524	2264	40.05	4072	72.04	1.80
2016	55481	2209	39.82	4076	73.47	1.85
2017	53399	2282	42.73	4268	79.93	1.87
2018	52245	2090	40.00	4208	80.54	2.01
2019	51492	2133	41.42	3921	76.15	1.84
2020	48611	2002	41.18	3807	78.32	1.90
2021	49256	2052	41.66	4035	81.92	1.97
2022	46967	1899	40.43	3919	83.44	2.06
2023	47873	2122	44.33	4465	93.27	2.10
2024	51207	2300	44.92	4563	89.11	1.98
1997–2024	1318841	51953	39.39	95799	72.64	1.84

Alberta Total Births from: Alberta Vital Statistics Annual Reviews for 1997-2024

Appendix A.5 Chi Trend Table for Reported Anomalies, 1997–2024

Anomaly	X ²	p Value	Direction*
Anencephaly	8.09	0.0045	↓
Spina bifida without anencephaly	0.00	1.00	↔
Encephalocele	0.10	0.7518	↔
Neural tube defects (all)	2.34	0.1261	?↓
Hydrocephalus without spina bifida	25.56	<0.0001	↓
Arhinencephaly/Holoprosencephaly	2.11	0.1463	?↑
Microcephaly	0.20	0.6547	↔
Anophthalmia/Microphthalmia	0.29	0.5902	↔
Congenital cataract	0.04	0.8415	↔
Anotia/Microtia	5.03	0.0249	↑
Congenital heart defects (all)	35.81	<0.0001	↑
Common truncus	0.78	0.3771	↔
Transposition of great arteries	2.03	0.1542	?↑
Tetralogy of Fallot	5.97	0.0146	↑
Ventricular septal defect	6.09	0.0136	↑
Atrial septal defect	3.86	0.0495	↓
Endocardial cushion defect	13.11	0.0003	↑
Pulmonary valve atresia/stenosis	9.62	0.0019	↑
Tricuspid valve atresia/stenosis	1.47	0.2253	?↑
Ebstein's anomaly	0.05	0.8231	↔
Aortic valve atresia/stenosis	1.02	0.3125	?↓
Hypoplastic left heart syndrome	4.07	0.0437	↑
Coarctation of the aorta	6.26	0.0123	↑
Cleft palate without cleft lip (CPO)	2.74	0.0979	?↓
Cleft lip without cleft palate (CLO)	0.10	0.7518	↔
Cleft lip and cleft palate (CL+CP)	0.09	0.7642	↔
Cleft lip with and without cleft palate (CL+/-CP)	0.00	1.0000	↔
Choanal atresia/stenosis	1.63	0.2017	?↓
Oesophageal atresia/trachea-oesophageal fistula	4.03	0.0447	↑
Pyloric stenosis	65.02	<0.0001	↓
Small intestinal atresia/stenosis (all)	0.64	0.4237	↔
Duodenal atresia/stenosis	0.09	0.7642	↔
Rectal and large intestinal atresia/stenosis (all)	14.18	0.0002	↓
Rectal atresia/stenosis	5.92	0.0150	↓
Anal atresia/stenosis	6.67	0.0098	↓
Ano-rectal atresia/stenosis			↓
Other large intestinal atresia/stenosis	5.41	0.0200	↓
Hirschsprung's disease	0.31	0.5777	↔
Biliary atresia	0.40	0.5271	↔
Undescended testes (male denominator)	2.52	0.1124	?↑
Hypospadias (male denominator)	40.33	<0.0001	↑
Epispadias (male denominator)	5.01	0.0252	↓
Renal agenesis/hypoplasia	29.08	<0.0001	↑
Cystic kidney	6.64	0.0100	↑
Bladder exstrophy	2.19	0.1389	?↓
Obstructive genitourinary defects (all)	88.04	<0.0001	↑
Hydronephrosis	51.88	<0.0001	↑
UPJ obstruction	34.01	<0.0001	↑
VUJ obstruction (based on very few cases per yr. - range 0-6)	9.14	0.0025	↑
Posterior urethral valves (male denominator)	0.06	0.8065	↔

*Direction: ↑ (up); ↓ (down); ↔ (no change); ?↑ or ?↓ (not statistically significant but a possible trend to watch)

Appendix A.5 Chi Trend Table for Reported Anomalies, 1997–2024 cont'd.

Anomaly	X²	p Value	Direction*
Congenital deformities of hip (all)	175.38	<0.0001	↓
Congenital hip dislocation, subluxation, dysplasia	70.95	<0.0001	↓
Reduction deformity, upper	4.79	0.0286	↑
Reduction deformity, lower	4.07	0.0437	↑
Diaphragmatic hernia	0.17	0.6801	↔
Abdominal wall defects (all)	5.60	0.0180	↑
Omphalocele	25.29	<0.0001	↑
Gastroschisis	5.03	0.0249	↓
All chromosome anomalies	261.06	<0.0001	↑
Trisomy 13	13.85	0.0002	↑
Trisomy 18	52.70	<0.0001	↑
Trisomy 21	49.75	<0.0001	↑

*Direction: ↑ (up); ↓ (down); ↔ (no change); ?↑ or ?↓ (not statistically significant but a possible trend to watch)