Alberta Congenital Anomalies Surveillance System

Fourteenth Report:

Data for 1997 – 2021

Alberta Health Services

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ACRONYMS FOR JURISDICTIONS AND ORGANIZATIONS MENTIONED IN THE REPORT

ACASS	Alberta Congenital Anomalies Surveillance System
AHS	Alberta Health Services
AH	Alberta Health
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
RCPCH	Royal College of Paediatrics and Child Health

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1. ACASS Activities and Report Summary

1. This is the fourteenth in a series of reports detailing the birth prevalence of congenital anomalies in Alberta, for the years 1997–2021 inclusive.

2. The International Classification of Diseases – 10th Edition (ICD-10-CA) has been adopted by Alberta hospital reporting data systems, and ACASS currently uses the Royal College of Paediatrics and Child Health adaptation of ICD-10. Many of the anomalies outlined in the National Birth Defects Prevention Network's (NBDPN) Guidelines for Conducting Birth Defects Surveillance (https://www.nbdpn.org/guidelines.php) 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 1997-2021 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. The prevalence of neural tube defects has remained relatively stable from 1997-2021, particularly for spina bifida and encephalocele. Anencephaly rates continue to significantly decrease **(pp. 20-23)**.

5. 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:

    5.1. Two of the most prevalent congenital genitalia anomalies, undescended testes ($p = 0.0015$) and hypospadias ($p < 0.0001$), continue to significantly increase **(pp. 23-25).**

    5.2. Renal agenesis/hypoplasia continues to significantly increase ($p < 0.0001$) **(pp. 25-27).**

    5.3. Although gastroschisis rates have stabilized ($p = 0.3032$), the prevalence of omphalocele continues to significantly increase ($p = 0.0005$). A special report on gastroschisis is provided **(pp. 27-36).**

    5.4. Chromosome anomalies as a group continues to significantly increase ($p < 0.0001$), particularly the common aneuploidies trisomy 13 ($p = 0.0160$), trisomy 18 ($p < 0.0001$), and trisomy 21 ($p < 0.0001$) **(pp. 36-37).**

6. The percentage of births to women 35 years of age and over has increased to one quarter in 2021, 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 1.5% in 2021.

7. The total number of Alberta births (live births and stillbirths) to Alberta mothers increased steadily from 36,797 in 1997 before peaking in 2015 at 56,524. Since 2015, the number has decreased to 49,256 births in 2021.

8. Members of ACASS continue to participate with the Canadian Congenital Anomalies Surveillance Network (CCASN) (https://www.canada.ca/en/public-health/services/health-promotion/what-is-ccasn.html). T. Bedard is also a member of the British Columbia Congenital Anomaly Surveillance System Advisory Committee and participates with the Stakeholders Partnering for Arthrogryposis Research Client-Centred Care (SPARC) Network, which is funded by the Canadian Institutes of Health Research and Shriners Hospitals for Sick Children. With this funding, an international arthrogryposis registry has been established to align research priorities and implement multi-site studies to promote evidence-based practice that will improve the overall health and well-being of individuals with arthrogryposis.

9. ACASS continues its affiliation with the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR) (http://www.icbdsr.org/). M. Thomas is a member of the Executive Committee. ACASS has participated in many group projects and publications. One of the most recent projects evaluated data quality indicators to promote and support a shared culture of quality assessment and improvement among ICBDSR programs. Data from ACASS was used to determine the prevalence and time trends of gastroschisis among programs in ICBDSR *(article in press)*.

10. The European Concerted Action on Congenital Anomalies and Twins (EUROCAT) has accepted the application from ACASS to become a World Affiliate member in 2023. This will provide additional collaborative opportunities for ACASS.

11. Over the past 10 years, ACASS has co-authored 23 peer-reviewed publications in addition to presentations at national and international meetings **(pp. 39-41)**.

12. 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 1997–2021 inclusive. For the current release, many anomalies outlined in the NBDPN Guidelines for Conducting Birth Defects Surveillance (https://www.nbdpn.org/guidelines.php) are reported along with some others that might be of interest. However, 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 Alberta Health 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 Centers for Disease Control and Prevention (CDC) in Atlanta, Georgia as the ongoing, systematic collection, analysis and interpretation of data (e.g., regarding agent/hazard, risk factor, exposure, health event) essential to the planning, implementation and evaluation of public health practice, closely integrated with the timely dissemination of these data to those responsible for prevention and control.

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, possibly 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 AHS provincial programs (e.g. Alberta Newborn Metabolic Screening Program, Edmonton Maternal Serum Screening Program);
8) participate in research into 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 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 NBDPN.

3.2 Case Ascertainment

An infant can be ascertained at any time up to the first birthday. Multiple ascertainments 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 malformations (anomaly rates) or as the total number of malformed infants (case rates). The tables in **Appendix A.3 (pp. 45-59)** report anomaly rates, which in most cases are similar to case rates (e.g. cleft palate, hypospadias, and microcephaly). Whereas with limb anomalies, there can be multiple different limb 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 not only the ease but also completeness of ascertainment as well as for verification of the diagnostic data. **Appendix A.1 (p. 43**) indicates the current process of data collection at ACASS.

ACASS screens many Alberta Health 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

Also, ACASS screens a notification called the Congenital Anomalies Reporting Form **(CARF, Appendix A.2, p. 44)** 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.

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.

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.

3.3 Quality Control Measures

When a copy of a reporting document reaches the ACASS office in Calgary, it is reviewed for content by the ACASS team. 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 orthopaedic surgeon, a paediatric nephrologist, a perinatologist/obstetrician, a professor of obstetrics and gynaecology, with occasional input from a paediatric neurologist.

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 entered into the 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 the Alberta Perinatal Health Program (APHP) 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.

3.6 Confidentiality and Release of Data

Notifications of Congenital Anomalies are sent to the Analytics and Performance Reporting Branch, Alberta Health, and from there to the ACASS office in Calgary where the database 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 the notifications are kept in locked files in a locked room. The database is secured by limited access and is password protected. Should further clarification about a case or anomaly become necessary, we communicate with the attending physician or the physician responsible for ongoing care. 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 =

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

CASE RATE =

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

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 within 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 Vital Statistics, hospitals, and other agencies but are augmented by active ascertainment from physicians and labs, etc. The completeness and accuracy of data are largely dependent on reporting.

4. Current Challenges and Future Opportunities

Compared to our last ACASS report, published in 2021 with data up to 2018, this current report includes data spanning the years impacted by the COVID-19 pandemic and the provincial implementation of Connect Care, a provincial electronic medical record. Since ACASS primarily relies on health professionals for case notifications, the reported prevalence from 2019 to 2021 should be interpreted with caution, as the health system was and still is significantly challenged. However, there are opportunities for ACASS to address these challenges.

4.1 COVID-19 Pandemic

As highlighted by Ludorf et al. (2020), the COVID-19 pandemic may have impacted the diagnoses and ascertainment of congenital anomalies due to hospitals attempting to discharge patients more quickly to increase capacity. Care may be delayed to mitigate risk of virus exposure. This could result in reduced opportunities to identify, thoroughly examine and test, document, and follow-up cases with congenital anomalies, particularly those not obvious at the time of birth or that are relatively mild. The

increased use of telemedicine and the downstream effects on congenital anomalies surveillance systems may also need to be considered.

While more severe and obvious congenital anomalies are still likely to be diagnosed and ascertained as before the pandemic, there are epidemiologic considerations. For example, there may be an over-representation of cases with severe congenital anomalies in surveillance data (Ludorf et al., 2020).

The case rates per 1,000 total births are comparable to pre-pandemic years. Although the anomaly rates are lower for the years 2019 and 2020 (Table 4.1.1) and (p. 60), there are annual fluctuations. A future data quality assessment will provide more insight as to the impact of the COVID-19 pandemic on ACASS data (Groisman et al., 2019).

Table 4.1.1 Case and Anomaly Rates per 1,000 Total Births, 2017-2021

Year	Case Rate/1,000 Total Births	Anomaly Rate/1,000 Total Births
2017	42.75	79.81
2018	40.02	80.47
2019	41.39	75.76
2020	41.02	77.45
2021	40.87	80.52

4.2 Provincial Implementation of Connect Care Electronic Medical Record

In addition to the COVID-19 pandemic, there have been phased launches of Connect Care that may have also impacted congenital anomalies surveillance in Alberta. The management of the pandemic and the implementation of Connect Care both continue to contribute to substantial differences in workload and workflows through the health system. There is a possibility of under reporting and under ascertainment during the different launches of Connect Care due to increased demands on learning a new system while maintaining health care services.

Currently, ACASS receives case notifications, primarily from Health Information Management Professionals throughout the province, in paper form (i.e., the Congenital Anomalies Reporting Form – CARF). With the provincial implementation of Connect Care, there are opportunities for ACASS to move from a paper to an electronic notification process which will increase ascertainment and thus data quality. Mai et al. (2015) report the benefits of using electronic health information systems for congenital anomalies surveillance include increasing the efficiency of record review and verification, and easing the reporting burden for the reporting source. Connect Care will also contribute to a more streamlined and sustainable data flow compared to the current flow (p. 43).

References:

Groisman B, Mastroiacovo P, Barbero P, Paz Bidondo M, Liascovich R, Botto LD. (2019). A proposal for the systematic assessment of data quality indicators in birth defects surveillance. Birth Defects Res, 111(6):324-332. https://doi.org/10.1002/bdr2.1474

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Mai CT, Correa A, Kirby RS, Rosenberg D, Petros M, Fagen MC. (2015). Assessing the practices of population-based birth defects surveillance programs using the CDC Strategic Framework, 2012. Public Health Rep, 130(6):722-730. https://doi.org/10.1177/003335491513000621

5. Patterns of Selected Congenital Anomalies in Alberta

5.1 Birth Prevalence – Time Trends

The following table and graphs of selected sentinel anomalies indicate the trends in congenital anomaly rates in Alberta from 1997 through 2021. 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. 61)** for other anomalies listed in the report**.**

Table 5.1.1 Chi Squared Linear Trend Analysis and p-values for Selected Anomalies 1997–2021 (Live Births, Stillbirths & ToPs)

Anomaly	Trend Direction	Chi Squared Analysis (χ^2LT)	p-value
Neural Tube Defects	No significant change	2.99	0.0838
Anencephaly	**Decreasing**	5.62	0.0178
Spina Bifida	No significant change	0.01	0.9203
Hydrocephalus	**Decreasing**	18.97	<0.0001
Cleft Lip +/- Cleft Palate	No significant change	0.01	0.9203
Cleft Palate	No significant change	3.68	0.0551
Hypoplastic Left Heart Syndrome	No significant change	1.35	0.2453
Oesophageal Atresia/Tracheo-oesophageal Fistula	No significant change	2.21	0.1371
Anorectal & Large Intestine Atresia/Stenosis	**Decreasing**	15.96	<0.0001
Hypospadias*	**Increasing**	55.23	<0.0001
Undescended Testes*	**Increasing**	10.12	0.0015
Renal Agenesis/Hypoplasia	**Increasing**	16.68	<0.0001
Limb Reductions - upper	No significant change	2.90	0.0886
Limb Reductions - lower	No significant change	0.99	0.3197
Gastroschisis	No significant change	1.06	0.3032
Omphalocele	**Increasing**	12.02	0.0005
Down Syndrome	**Increasing**	22.56	<0.0001

*Hypospadias and Undescended Testes calculated for male births only

5.2 Selected Anomalies

5.2.1 Selected Anomaly Definitions

(Adapted from NBDPN guidelines: http://www.nbdpn.org/ and ICBDSR Reported Malformations Definitions: http://www.icbdsr.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, and hypoplasia and coarctation of the aorta.

- ***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 2021, the prevalence of neural tube defects (NTDs) has not significantly changed (p=0.0838), however it is a possible trend to watch (Figure 5.2.1). Although there continues to be a significant decline in anencephaly (p=0.0178)**,** the rates of spina bifida and encephalocele have not changed significantly (Figure 5.2.2).

The decline in anencephaly rates were noted in the last ACASS report (Alberta Congenital Anomalies Surveillance System, 2021), potentially due to the terminology used to report what is seen on first trimester prenatal ultrasounds (e.g. “acrania” or “absent calvarium”), which would be coded by ACASS with an ICD-10 code outside of the NTD section. The decline may also be a true decline.

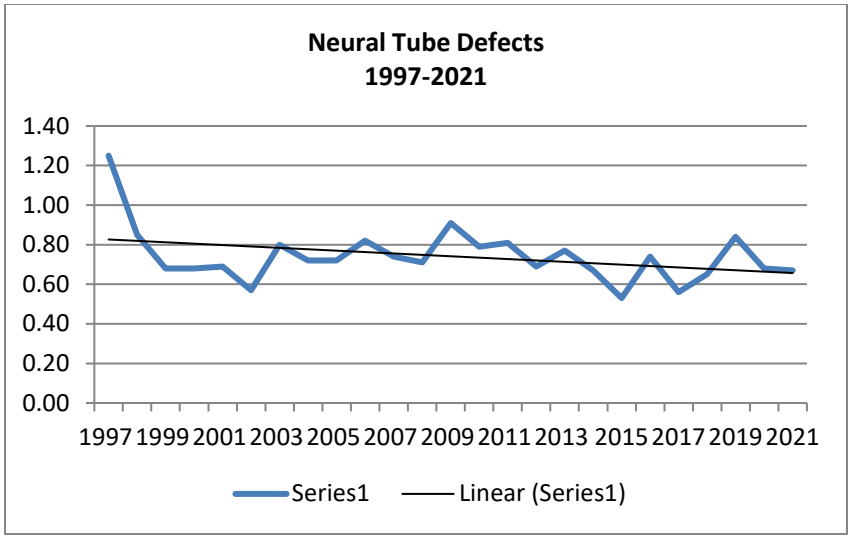
Spina bifida (SB) has remained relatively stable since the sharp decline following folic acid fortification (FAF) in 1998. More current rates range from 0.25/1,000 TB in 2015 to a high of 0.47/1,000 TB in 2019 (Appendix A.3 p.46). For the 5-year period, 2017-2021, we report a prevalence of SB of 0.38/1,000 TB. This is comparable per 1,000 live births to Utah (0.42) and Texas (0.36), where there is mandatory FAF, for the period 2016-2020 (National Birth Defects Prevention Network, 2024). However, this is much lower than the rates reported by Broughan et al. (2023) for England for 2015-2019 (0.61/1,000 TB) and for 2000-2019 (0.59/1,000 TB). During the reported period, no country in Europe had mandatory FAF. However, in September 2021, the UK government announced the mandatory FAF for non-wholemeal wheat flour to reduce the prevalence of NTDs. Broughan et al. (2023) aimed to assess the trend of NTDs to determine a baseline, which will be used for the future evaluation of the impact of FAF on NTDs. The authors included all cases with NTDs to determine prevalence, which may be challenging to compare to the post FAF period, since some types of SB are not as responsive to FAF due to differing etiologies (e.g., chromosomal, syndromic, lipomyelomeningocele).

Since 1997, the prevalence of spina bifida excluding lipomeningomyelocele, and those associated with known syndromes, teratogens, and chromosome abnormalities, is trending upwards in Alberta, although the increase is not significant (p=0.1936) (Figure 5.2.3). From 2016-2021, the proportion of these types of cases that perhaps should be more folate-responsive was 59%, which is very comparable to Lowry et al. (2019), who reported 58% from 2001-2015.

A more comprehensive study of cases with SB, that should be more folate-responsive, would provide more insight. Additional details regarding the level and severity of lesion(s), and associated risk factors may inform preventive efforts. 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).

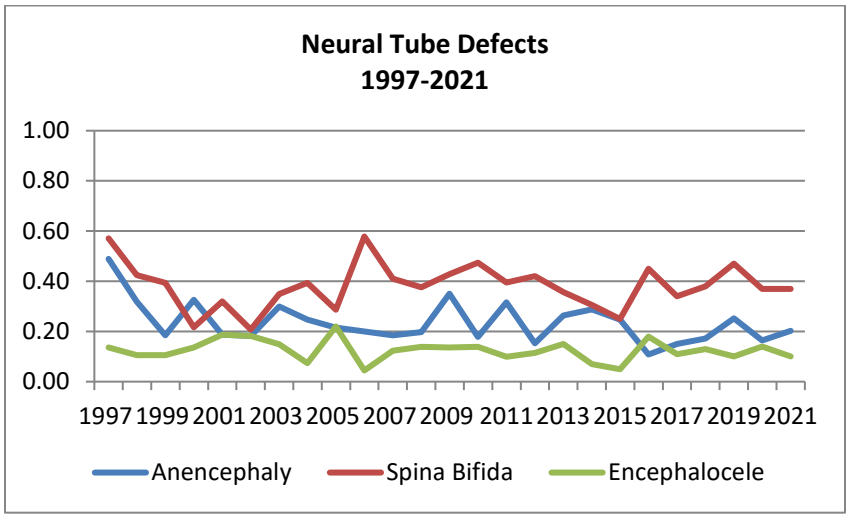
Inadequate red blood folate concentrations and other micronutrient deficiencies may be contributing to the observed frequency of this subset of SB (Chandler et al., 2012; Colapinto et al., 2011). It has been suggested that the entirety of one-carbon metabolism, including other micronutrients that act 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 FAF, 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).

Figure 5.2.1 All Neural Tube Defects, 1997-2021 (Rate per 1,000 total births)



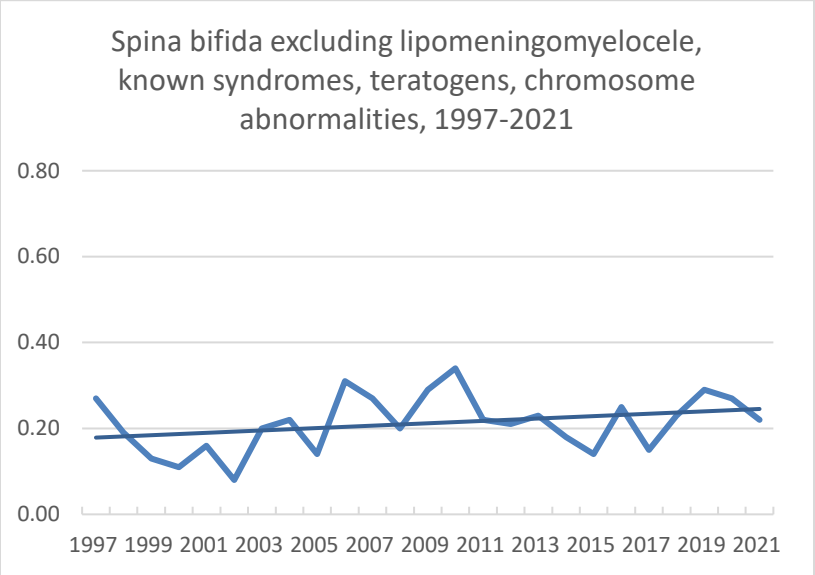
p=0.0838

Figure 5.2.2 Neural Tube Defects: Anencephaly, Spina Bifida, and Encephalocele, 1997-2021 (Rate per 1,000 total births)



Anencephaly p=0.0178; Spina Bifida p=0.9203; Encephalocele p=0.4708

Figure 5.2.3 Spina bifida excluding lipomeningomyelocele, known syndromes, teratogens, chromosome abnormalities, 1997-2021 (Rate per 1,000 total births)



p=0.1936

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5.2.3 Undescended Testes and Hypospadias

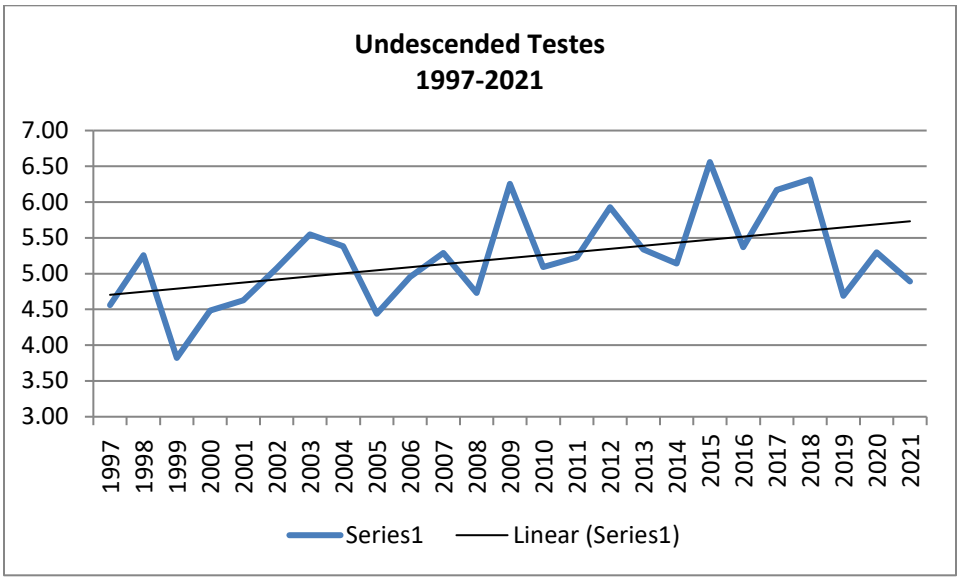
Two of the most common congenital anomalies of the external genitalia are undescended testes (UT) and hypospadias, which are hormone-mediated malformations that occur during male development. The rates reported are expressed as total male births in contrast to some reports which are cited as total live births and do not differentiate the male proportion. Regarding UT, ACASS does not accept cases born before 37 weeks gestation or a birth weight less than 2500 grams because these cases commonly have UT. The ascertainment of cases with hypospadias by ACASS includes all degrees of severity but excludes isolated chordee.

Both UT and hypospadias continue to statistically significantly increase in Alberta (Figures 5.2.4 and 5.2.5). The trend for UT needs to be interpreted with caution, since many may resolve spontaneously while others may be misdiagnosed and are actually retractile testes. We acknowledge that a more accurate prevalence would be estimated if cases were followed up for at least 6 months to determine if the testes spontaneously descend. For a birth cohort which was part of the Norwegian Human Milk Study (HUMIS 2002-2009) to assess levels of persistent organic pollutants in breast milk and health effects, Desalegn et al. (2021) reported that 56% of their cases with UT, descended spontaneously within 6 months of birth.

The prevalence for the 20 year-period (2002-2021) is 5.42 and 4.93 per 1,000 total male births for UT and hypospadias respectively. Lane et al. (2017) report a higher prevalence for Nova Scotia, between 1988 and 2013 of 7.5 for UT and 7.8 for hypospadias, per 1,000 total male births. While the prevalence of hypospadias was stable and slightly decreased for UT during the study period, the two anomalies significantly clustered in the same western region (county level) of Nova Scotia, indicating that their distribution is likely influenced by regional factors such as environmental exposures including those associated with intense agricultural activities (Lane at al., 2017). Mahboubi et al.

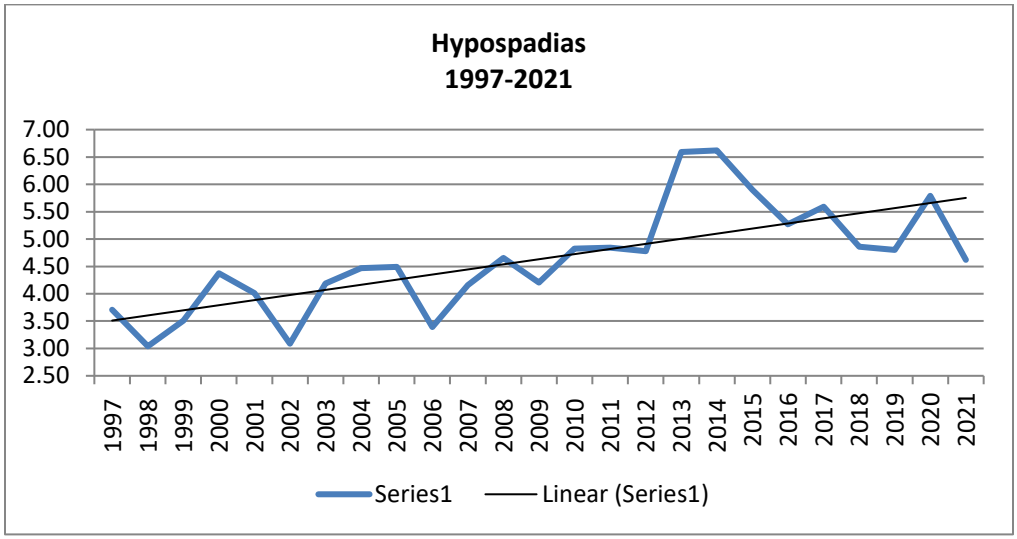
(2023) performed a more granular analysis using the same data and assessed clustering distributions at the postal code level. The authors report different clustering patterns than those reported by Lane et al. (2017), citing the unit of analysis as contributing to the discrepancies. Using more specific spatial units such as postal codes, helps reduce statistical bias associated with using larger units such as counties. Cryptorchidism was clustered in the Cape Breton region, most notably in areas surrounding Sydney, while hypospadias had a higher prevalence near the Halifax Regional Municipality and Cape Breton (Mahboubi et al., 2023). Further study is needed to assess routes for exposures of endocrine-disrupting chemicals, and the role of genetic factors. Lowry et al. (2020) reported that the Hutterite Brethren (HB) have a statistically significantly increased prevalence (p=0.0001) of hypospadias (7.70/1000 total male births) compared to the general Alberta population (3.79/1000 total male births) for the years 1997-2016. Since the HB are a farming and agriculture community, it does suggest that this increased prevalence may be related to agricultural practices. Thus, the findings reported by Lane et al. (2017) and Mahboubi et al. (2023) are pertinent to Alberta, where geospatial analyses may provide insight into the factors associated with the reported increasing trends of UT and hypospadias.

Figure 5.2.4 Undescended Testes - All, 1997-2021 (Rate per 1000 Male Births)



p=0.0015

Figure 5.2.5 Hypospadias – All, 1997-2021 (Rate per 1000 Male Births)



p<0.0001

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5.2.4 Renal Agenesis/Hypoplasia

ACASS reports the prevalence of both renal agenesis and hypoplasia, affecting one or both kidneys. The total case load is 744 for the period 1997-2021, with a prevalence of 0.63/1000 total births. The trend is significantly increasing (p<0.0001) (Figure 5.2.6).

Renal agenesis and hypoplasia are often considered to be part of the congenital anomalies of kidney and urinary tract (CAKUT) group. There is etiological heterogeneity including genetic causes (chromosome and single-gene), associations such as VACTERL and MURCS, developmental field defects, as well as multifactorial causes with genetic and environmental risk factors. There are genes that can result in both renal agenesis and other CAKUT anomalies, even within the same family (Kirschen et al., 2023).

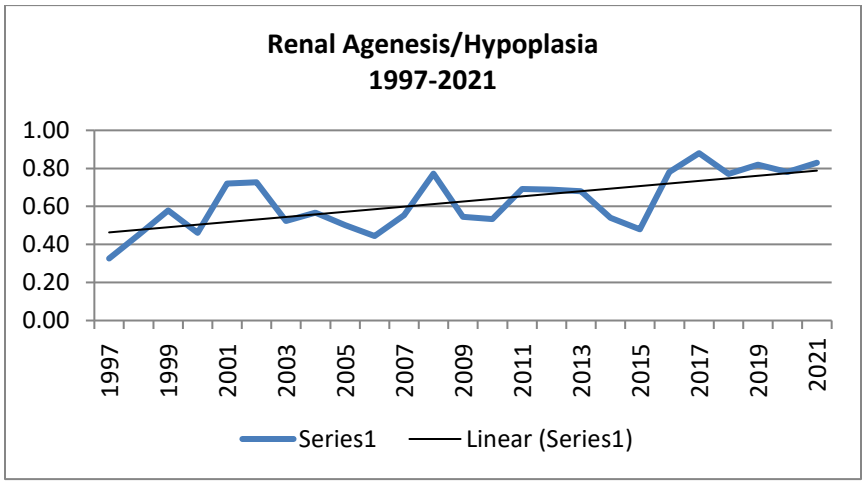
Renal agenesis can be seen in isolation or be one feature of a genetic syndrome with other congenital anomalies. The anhydramnios that is present in the second trimester when there is bilateral renal agenesis can limit the ultrasound detection of the other anomalies. (Kirschen et al., 2023).

A meta-analysis for renal agenesis based on 15,641,184 patients identified a pooled prevalence for renal agenesis being 0.3/1000 with no significant difference in frequency between males and females (Plutecki et al., 2023). Possible explanations for our higher reported prevalence are that renal hypoplasia is also included and all birth outcomes are included where one third of our cases were early fetal deaths (< 20 weeks gestational age), stillbirths, or infant deaths.

Table 5.2.1 shows that during 2017 and 2021, most cases with renal agenesis were unilateral (83.2%), and 16.3% were bilateral. The overall prenatal diagnosis rate was relatively high at 96.3% (Table 5.2.1).

Risk factors for renal agenesis include diabetes, both pre-gestation and gestational (Davis et al., 2010); BMI > 30Kg/m2; maternal smoking; and binge drinking (Slickers et al., 2008). Plutecki et al. (2023) reported a 2.72% rate of pre-gestational diabetes in 829 patients evaluated. An emerging risk factor may be maternal exposure to endocrine disrupting chemicals. In a population-based study by Spinder et al. (2022), this exposure was associated with certain CAKUT anomalies, particularly anomalies of the urinary collecting system, but not renal agenesis, so does not likely account for the increased trend in our data. Though, this risk factor will be worth following in the coming years.

Figure 5.2.6 Renal Agenesis-Hypoplasia, 1997-2021 (Rate per 1000 total births)



p<0.0001

Table 5.2.1 Numbers, Prevalence, and Prenatal Diagnosis of Renal Agenesis, 2017-2021

Renal Anomaly	Total Number of Cases (%)	Total Number of Cases prenatally diagnosed (%)	Total Prevalence per 1,000 total births
Bilateral Renal Agenesis	31 (16.3)	28 (90.3)	0.12 (0.08-0.17)
Unilateral Renal Agenesis	158 (83.2)	154 (97.5)	0.62 (0.53-0.72)
Unspecified Renal Agenesis	1 (0.5)	1 (100)	0.00
Total Renal Agenesis	190	183 (96.3)	0.75 (0.64-0.86)

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5.2.5 Abdominal Wall Defects

5.2.5a Special Report on Gastroschisis

The overall prevalence of gastroschisis (GS) in Alberta for 2002 to 2021, is 0.39/1000 TB, which is lower than that reported by ELHassan et al. (2020) for 1998-2015 (0.58/1000 live births (LB)); Stallings et al. (2019) for 2012-2016 (0.43/1000 LB); and Benjamin & Wilson (2014) for 1999-2008 (0.48/1000 TB). The reported lower Alberta prevalence may reflect the previously reported decreasing trend for GS after reaching a high of 0.57/1000 TB in 2011 (Alberta Congenital Anomalies Surveillance System, 2021). This has coincided with a decrease in pregnancies to mothers less than 20 years of age in Alberta, which is a known risk factor for GS (Lowry et al., 2023). The proportion of births to mothers

less than 20 years of age has decreased from 8% in 1983 and into the mid-1990s to 1.5% in 2021 (Figure 5.2.7). However, the proportion of cases in Alberta with GS born to this age group between 2002 and 2021 is still notable at 23%, with a prevalence of 2.53 per 1000 TB (Table 5.2.2 and Figure 5.2.8), compared to ELHassan et al. (2020) at 33%; Stallings et al. (2019) at 16%; although much lower than Egger at al. (2022) at 49%.

Other Canadian studies include Bourque et al. (2021) from Ontario, who found no change in prevalence between 2012 and 2018 however, they did not include termination of pregnancies for fetal anomalies (TOPFAs). Liu et al. (2021) used national data (excluding Quebec) from the Canadian Congenital Anomalies Surveillance System that included births form 2006-2017. The authors report annual variations with no significant increasing or decreasing trend for the study period. However, there was a high per 1000 TB of 0.50 in 2009 compared with 0.34 in 2017. Liu et al. (2021) primarily attributed the more current stabilization of GS prevalence to the decline of births to mothers less than 25 years.

While initially described as an isolated anomaly and non-familial, later work has established the occurrence of associated anomalies with varying proportions, from a low of 5.0 % (Tan et al., 1996; Rankin et al., 1999) to a high of 33.6 % (Stallings et al., 2019). This variability is in part due to the lack of consensus of what is part of GS and what is a separate entity.

A case review was recently completed to describe the congenital anomalies associated with GS, with ethics approval granted by the University of Calgary (REB22-0960). After excluding four cases (three with limb body wall complex and/or amniotic bands and one with Omphalocele-Exstrophy of the cloaca-Imperforate anus-Spinal defects complex), there were 417 eligible cases with GS born between 1997 and 2020. There were 347 (83.2%) cases that were classified as having an isolated anomaly, and 70 (16.8%) with an associated major anomaly that was unrelated to the GS. Congenital heart defects (CHDs), particularly septal defects, were the most frequently associated congenital anomalies occurring in 26 (37.1%) cases with non-isolated GS. This is followed by anomalies in the gastrointestinal system, 12 (17.1%) cases; musculoskeletal system 9 (12.9%) cases; and seven (10.0%) cases with anomalies affecting the head and neck. Syndromes/recognized conditions were relatively rare, being diagnosed in only 5.7% of cases with non-isolated GS.

Some studies (Benjamin & Wilson, 2014; Stallings et al., 2019) report a higher frequency of associated anomalies however, they include those considered to be GS sequence anomalies such as intestinal atresias, hydronephrosis, ureteral obstruction, and cryptorchidism, which we classified as isolated if no other major anomalies were reported (Hunter & Stevenson, 2008). If we moved our 34 cases with GS sequence anomalies from isolated to non-isolated, our frequency would increase to 24.9%, which would be more comparable to Benjamin & Wilson (2014), 32%; and Stallings et al. (2019), 33.6%. Feldkamp et al. (2016) had similar case classification criteria as our review and report a similar frequency of cases with non-isolated GS, 15.8%.

The concept, GS sequence, was used by Mastroiacovo et al. (2007) who reported an overall rate of 14.1% non-isolated cases from more than 30 registries, with variability between registries. The occurrence of cryptorchidism was first described by Kaplan et al. (1986). The descent of the testis requires intraabdominal pressure (IAP), which is lacking in patients with GS. Although, it is less clear how reduced IAP contributes to the presence of hydronephrosis or hydroureter in cases with GS. These anomalies, however, are often considered secondary to the dynamics of gastroschisis. In a small

case series, Reiss et al. (2000) describes how the herniation of abdominal contents through a gastroschisis can cause obstruction of a structurally normal urinary tract. Once the gastroschisis was repaired, the obstruction resolved spontaneously (Reiss et al., 2000).

Although these anomalies are considered GS sequence anomalies and cases are classified as 'isolated', particularly from a pathogenic perspective, cases with GS sequence anomalies require multiple neonatal surgeries and have an increased risk of morbidity and mortality when compared with cases with simple GS (Joyeux et al., 2021). Other GS sequence anomalies may also require additional surgical intervention (e.g. orchidopexy).

While GS often occurs as an 'isolated' anomaly, associated anomalies are reported, including those that are considered part of GS (GS sequence anomalies) and those that are primary anomalies in different organ systems. The main purpose of recording associated anomalies is to emphasize that every baby deserves a thorough evaluation to provide the best surgical and medical treatments for optimum health outcomes. The outstanding advances in pediatric surgery and neonatal care has resulted in 96-98% with a good outcome (Skarsgard et al., 2015; Räsänen et al., 2022).

It should also be emphasized that some cases with GS may be preventable as shown by the risk factors reported by Raitio et al. (2020), Brindle et al. (2012), Baldacci et al. (2020), and Bourque et al. (2021). This is supported by the fact that the Hutterite Brethren in Alberta has recorded zero cases of GS in the last 40 years which may be due to fewer exposures to many of these risk factors (Lowry et al. 2020). It is encouraging to see the reduction in teen-age mothers as shown in the Canadian studies by Bourque et al. (2021), Liu et al. (2021) and Lowry et al. (2023). However, in view of the increase in wildfires in many parts of Canada, which is an emerging risk factor (Park et al. 2021), it is essential that congenital anomalies surveillance continues.

Figure 5.2.7 Maternal Age Groups (all ages) Percent Total Births, 1983-2021

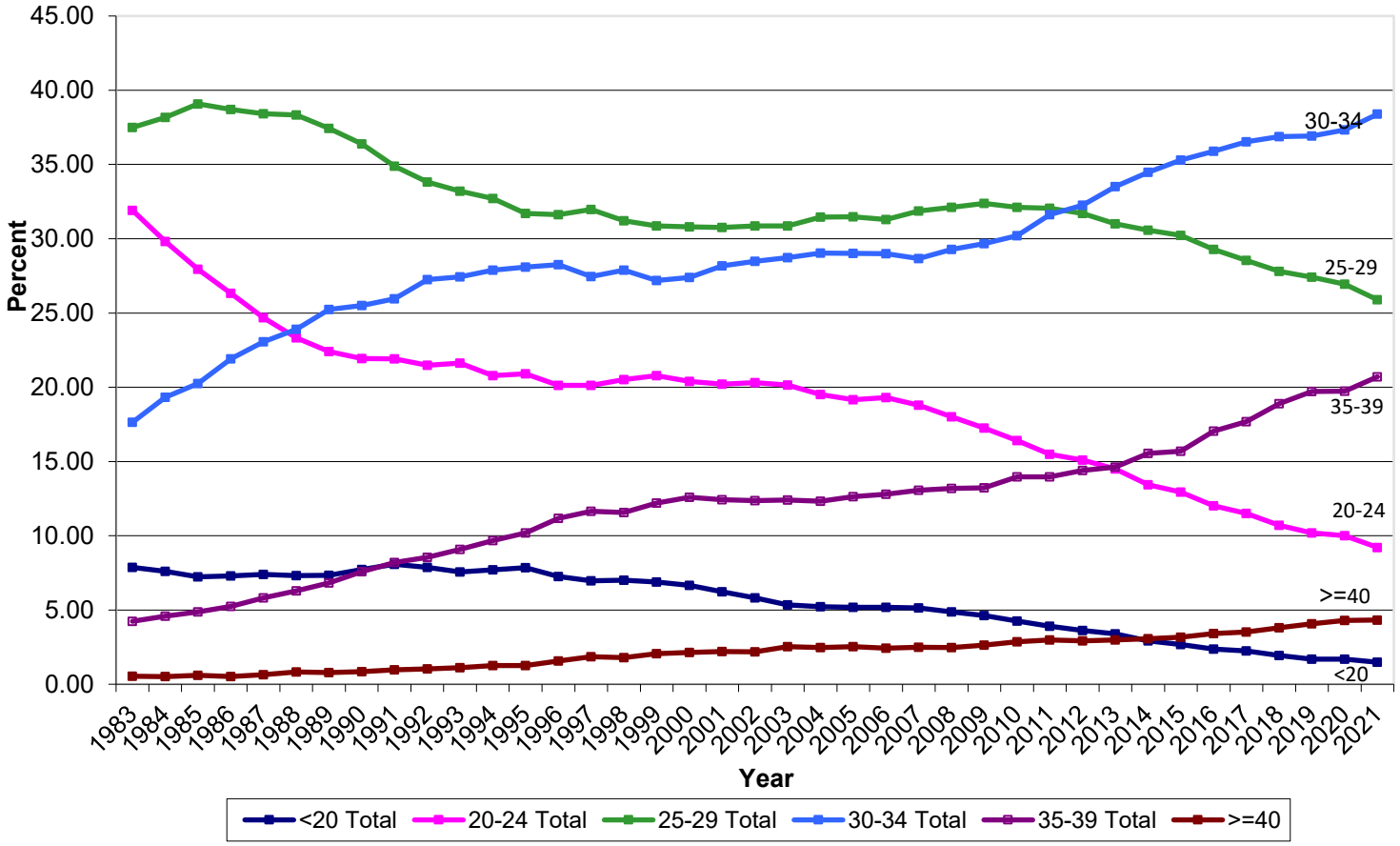
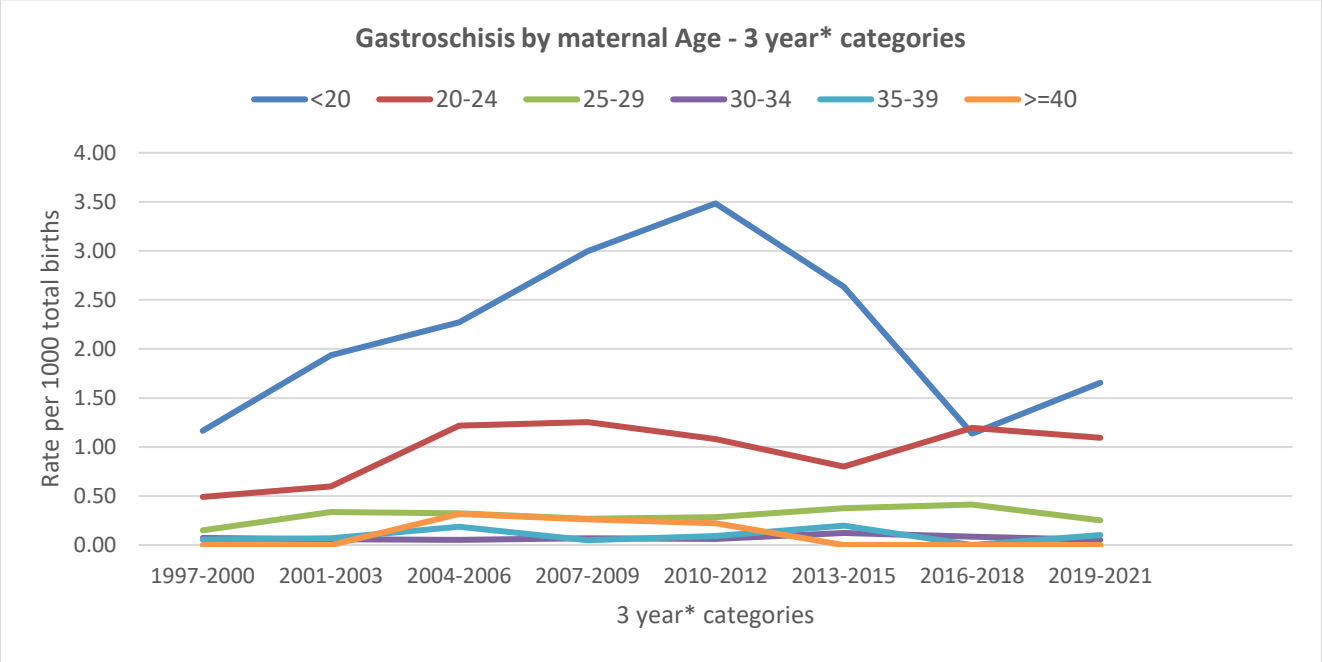


Table 5.2.2 Cases with gastroschisis by maternal age group, 2002-2021

Maternal Age Group (years)	Number of Cases with Gastroschisis	Proportion (%)	Prevalence per 1,000 Total Births (95% CI), for age group
<20	89	23.2	2.53 (2.03-3.11)
20-24	156	40.7	1.06 (0.90-1.24)
25-29	95	24.8	0.32 (0.26-0.39)
30-34	25	6.5	0.08 (0.05-0.11)
35-39	15	3.9	0.10 (0.06-0.16)
40-44	3	0.8	0.10 (0.02-0.30)
45+	0	0	0
Total	383	100	0.39 (0.35-0.43)

Figure 5.2.8 Prevalence of gastroschisis by maternal age group, 1997-2021



*except 4 years for 1997-2000

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5.2.5b Omphalocele

From 1997-2021, the linear trend for omphalocele is significantly increasing (p=0.0005). Like gastroschisis, very young maternal age (i.e., < 20 years) is associated with omphalocele. However, maternal age equal or greater than 35 years is also a risk factor. Table 5.2.3 shows that although the highest proportion of omphalocele cases is in the 30-34 years maternal age group (30%), the highest prevalence per 1,000 TB is in the > 40 years groups (1.67, 40-44 years and 1.80, 45+). While births to mothers < 20 years are decreasing, births to mothers in the 30+ years age groups are increasing (Figure 5.2.7) and is perhaps contributing to the significantly increasing trend of omphalocele in Alberta.

Additional risk factors include maternal obesity and diabetes with the co-occurrence increasing the risk further (Raitio et al., 2021). Feldkamp et al. (2014) in a self-reported maternal smoking study found no association but did find a possible association with second-hand smoke. There was no increased risk reported by Raitio et al. 2021 with maternal smoking. The authors did suggest that maternal use of oral extended spectrum penicillins, ascertained from the Register on Reimbursed Drug Purchases in Finland, significantly mitigated the risk (Raitio et al., 2021). They propose that microbiological factors may contribute to pathogenesis and explain the role of penicillins however, further study is required to confirm this association.

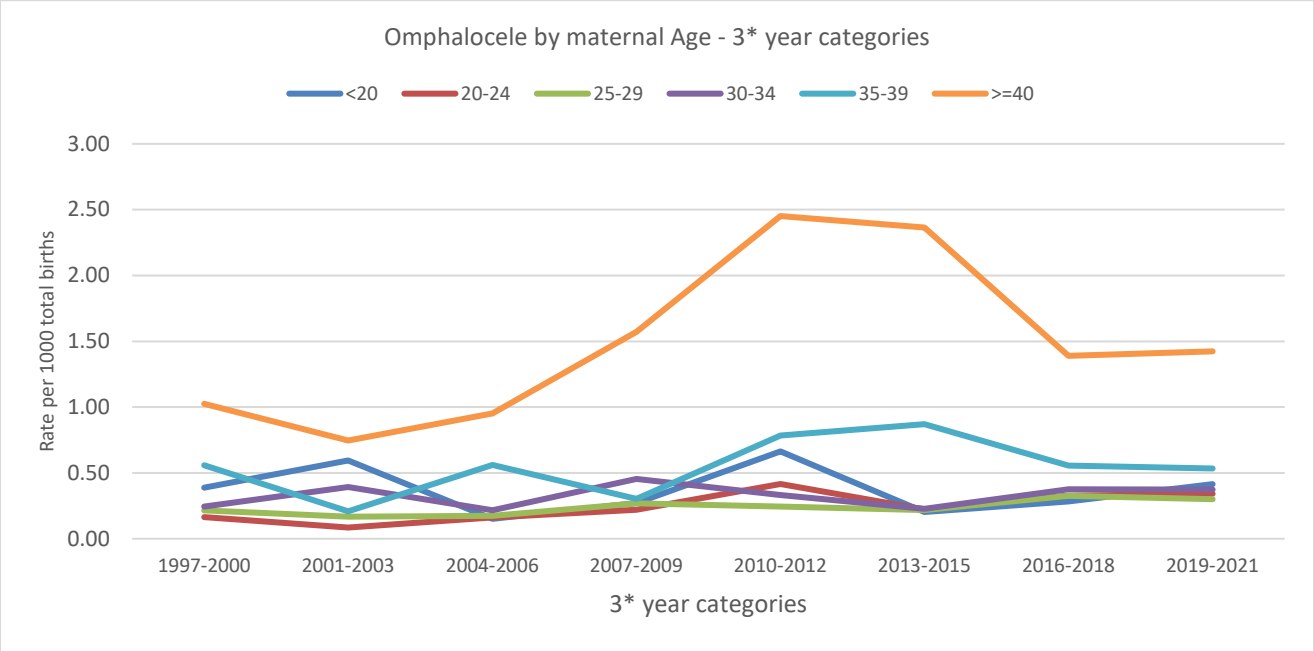
In contrast to gastroschisis, omphalocele often occurs with associated anomalies. The proportion of ACASS cases with omphalocele and co-occurring congenital anomalies is 76%, which is comparable with Stalling et al. (2019) (71.8%) and Stoll et al. (2021) (74.3%). These anomalies involve chromosome aneuploidies and other chromosome defects as well as malformations in many systems such as heart, gastrointestinal, genitourinary and neural tube defects. Many syndromes have omphalocele as one of their features, e.g. Beckwith-Wiedemann, Cantrell and OEIS (Adams et al., 2021; Frolov et al., 2010).

While isolated omphalocele is usually a sporadic event, nevertheless, there are reports of familial cases (Hershey et al., 1989) but of course, the recurrence risk to be cited depends on the diagnosis such as a syndrome.

Table 5.2.3 Cases with omphalocele by maternal age group, 2002-2021

Maternal Age Group (years)	Number of Cases with Omphalocele	Proportion (%)	Prevalence per 1,000 Total Births (95% CI), for age group
<20	12	3.2	0.34 (0.18-0.59)
20-24	38	10.2	0.26 (0.18-0.35)
25-29	74	19.8	0.25 (0.19-0.31)
30-34	111	29.8	0.34 (0.28-0.41)
35-39	87	23.3	0.58 (0.46-0.71)
40-44	48	12.9	1.67 (1.23-2.21)
45+	3	0.8	1.80 (0.36-5.10)
Total	373	100	0.38 (0.34-0.42)

Figure 5.2.9 Prevalence of omphalocele by maternal age group, 1997-2021



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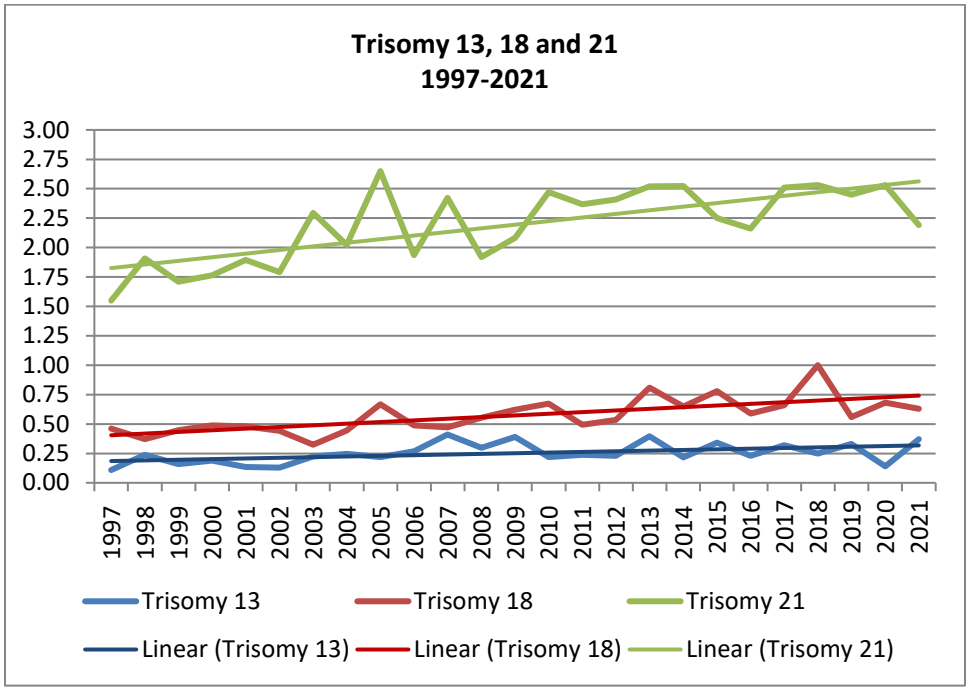
5.2.6 Chromosome Anomalies

Down syndrome (Trisomy 21) is the most commonly ascertained chromosome anomaly. As previously reported, rates of Down syndrome, Trisomy 13 and Trisomy 18 are increasing significantly (χ trend analyses: T21 $p<0.0001$; T13 $p=0.0160$; T18 $p<0.0001$) (Appendix A.5; Figure 5.2.10) and are strongly correlated with increasing maternal age. In 1980, approximately 5% of mothers were 35 years of age or over at the birth of their infant whereas, in 2021, one quarter of births were to women in this age category (Figure 5.2.7).

Although mortality is high among infants born with Trisomies 13 and 18, more infants are obtaining life saving medical and/or surgical intervention and will survive and require ongoing medical care and treatment, thus counting the anomalies associated with these diagnoses can help with future health care planning.

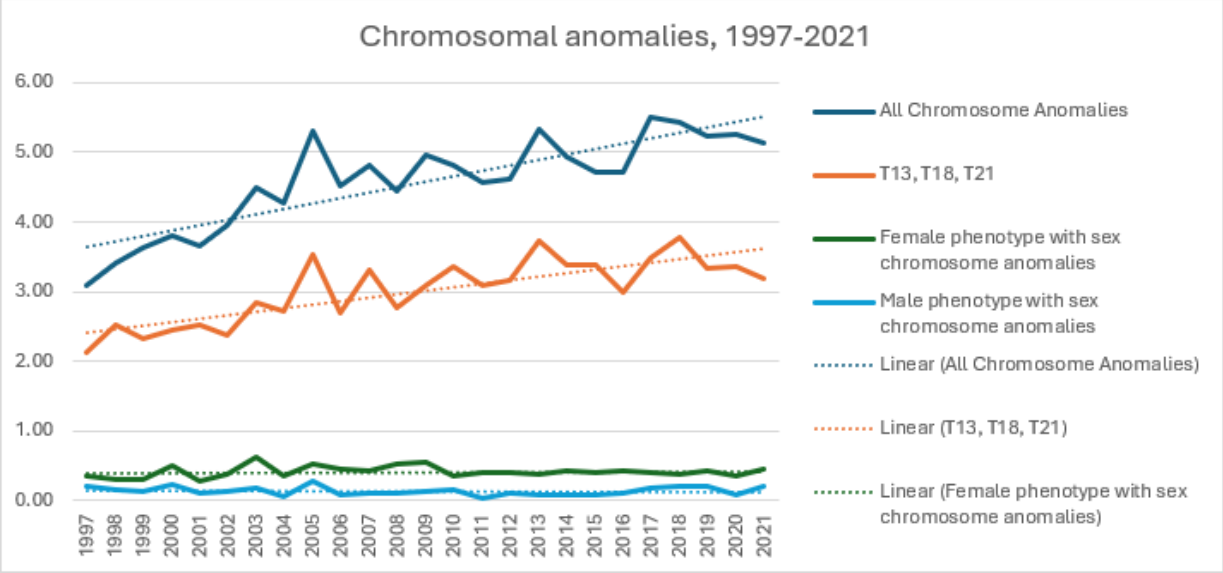
Figure 5.2.10 Prevalence of chromosome anomalies: Trisomy 13, Trisomy 18, Trisomy 21, 1997-2021



Trisomy 13, $p=0.0160$; Trisomy 18, $p<0.0001$; Trisomy 21, $p<0.0001$

Figure 5.2.11 compares different groups of chromosome anomalies. All chromosome anomalies include the common trisomies, rare trisomies, unbalanced chromosome anomalies including pathogenic/likely pathogenic chromosome deletions or duplications. Variants of uncertain significance were not included in this comparison. There has been a steady and significant increasing trend in all chromosome anomalies. Sex chromosome aneuploidies were stable. With increasing uptake of non-invasive prenatal screening (NIPS), and the option for sex chromosome aneuploidy (SCA) screening, there will likely be more patients prenatally diagnosed with SCAs, including 45,X and 47,XXY, as well as others. Prior to this, many were not diagnosed before 1 year of age and were often clinically diagnosed later in childhood. In an Australian population-based study, there was a noted significant increase in the rate of prenatal SCAs diagnosed on amniocentesis and CVS, from 5.8/10,000 births in 2005 to 8.7/10,000 births in 2020 (Loughry et al., 2023). This was predominantly due to 47, XXY cases, with 91% ascertained by a high-risk NIPS result (Loughry et al., 2023). We will continue to monitor local trends, particularly those that may be impacted by NIPS.

Figure 5.2.11 Comparison of chromosome groups, 1997 – 2021



All chromosome anomalies p<0.0001; T13, T18, T21 p<0.0001; female phenotype abnormal sex chromosomes p=0.8065; male phenotype abnormal sex chromosomes p=0.6468

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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 2010

6.1 Surveillance and Research Projects/Collaborations and Consultations/Papers

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Alberta Congenital Anomalies Surveillance System

7. Appendices

Appendix A.1 Flowchart of the Process of ACASS Data Collection

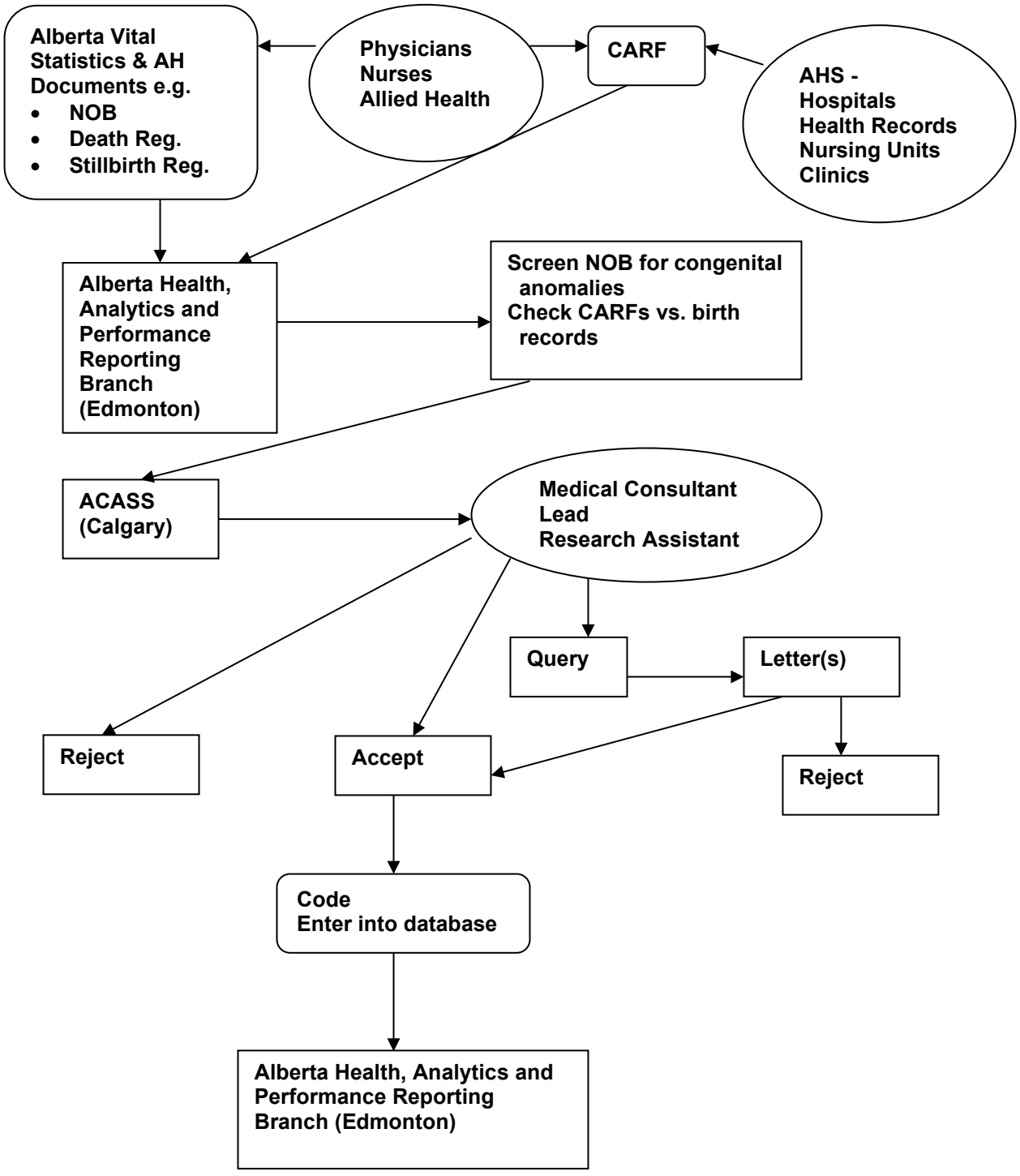
Appendix A.2 Congenital Anomaly(ies) Reporting Form (CARF)

Appendix A.3 Alberta Congenital Anomalies Surveillance System Anomaly Rates

Appendix A.4 Numbers of cases, anomalies and anomalies per case 1997–2021

Appendix A.5 Chi Trend table for reported anomalies 1997-2021

Appendix A.1 Flowchart of the Process of ACASS Data Collection



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

Alberta Health and Wellness

Death Reg No	Birth Reg No

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 ☐ First ☐ Second ☐ Triplets ☐ First ☐ Second ☐ Third		Physician Responsible for Ongoing Care *(if different from above)*

Parents

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

Reporting Hospital/Agency/Clinic

Name	Infant's Admission *(If different from birthdate)*	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.)*

OFFICE USE ONLY

Completed by	Position	Date

HS0020-112 (2008/06)

Send to Surveillance and Environmental Health

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 Alberta Congenital Anomalies Surveillance System Anomaly Rates
RCPCH version ICD-10 Q-Chapter (Q00-Q99)
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		2011	2012	2013	2014	2015	2016	2017	2018	2019	2020	2021
Anencephaly	NUMBER	16	8	14	16	14	6	8	9	13	8	10
	RATE	**0.32**	**0.15**	**0.26**	**0.29**	**0.25**	**0.11**	**0.15**	**0.17**	**0.25**	**0.16**	**0.20**
	Lower CI	0.18	0.07	0.14	0.17	0.14	0.04	0.06	0.08	0.13	0.07	0.10
ICD-10 Q00.00, Q00.01, Q00.1	Upper CI	0.51	0.30	0.44	0.47	0.41	0.23	0.29	0.33	0.43	0.32	0.37
Spina Bifida without Anencephaly	NUMBER	20	22	19	17	14	25	18	20	24	18	18
	RATE	**0.39**	**0.42**	**0.36**	**0.31**	**0.25**	**0.45**	**0.34**	**0.38**	**0.47**	**0.37**	**0.37**
	Lower CI	0.24	0.26	0.22	0.18	0.14	0.29	0.20	0.23	0.30	0.22	0.22
ICD-10 Q05..	Upper CI	0.61	0.64	0.56	0.49	0.41	0.67	0.53	0.59	0.69	0.58	0.58
Encephalocele	NUMBER	5	6	8	4	3	10	6	7	5	7	5
	RATE	**0.10**	**0.11**	**0.15**	**0.07**	**0.05**	**0.18**	**0.11**	**0.13**	**0.10**	**0.14**	**0.10**
	Lower CI	0.03	0.04	0.06	0.02	0.01	0.09	0.04	0.05	0.03	0.06	0.03
ICD-10 Q01..	Upper CI	0.23	0.25	0.29	0.18	0.15	0.33	0.24	0.27	0.22	0.29	0.23
Neural Tube Defects (all)	NUMBER	41	36	41	37	31	42	32	37	43	33	33
	RATE	**0.81**	**0.69**	**0.77**	**0.67**	**0.55**	**0.76**	**0.60**	**0.71**	**0.84**	**0.68**	**0.67**
	Lower CI	0.58	0.48	0.55	0.47	0.37	0.55	0.41	0.50	0.60	0.47	0.46
ICD-10 Q00.., Q01.., Q05..	Upper CI	1.10	0.95	1.05	0.92	0.78	1.02	0.85	0.98	1.13	0.95	0.94
Hydrocephalus without Spina Bifida	NUMBER	37	26	16	20	20	28	16	13	15	17	9
	RATE	**0.73**	**0.50**	**0.30**	**0.36**	**0.35**	**0.50**	**0.30**	**0.25**	**0.29**	**0.35**	**0.18**
(Excludes hydranencephaly)	Lower CI	0.51	0.33	0.17	0.22	0.22	0.34	0.17	0.13	0.16	0.20	0.08
ICD-10 Q03	Upper CI	1.01	0.73	0.49	0.56	0.55	0.73	0.49	0.42	0.48	0.56	0.34
Arrhinencephaly/ Holoprosencephaly	NUMBER	11	12	18	8	16	17	22	11	15	6	6
	RATE	**0.22**	**0.23**	**0.34**	**0.14**	**0.28**	**0.31**	**0.41**	**0.21**	**0.29**	**0.12**	**0.12**
	Lower CI	0.11	0.12	0.20	0.06	0.16	0.18	0.26	0.11	0.16	0.05	0.04
ICD-10 Q04.1, Q04.2, Q87.03	Upper CI	0.39	0.40	0.53	0.28	0.46	0.49	0.62	0.38	0.48	0.27	0.26
Microcephaly	NUMBER	33	20	21	15	22	15	20	20	13	16	14
	RATE	**0.65**	**0.38**	**0.39**	**0.27**	**0.39**	**0.27**	**0.37**	**0.38**	**0.25**	**0.33**	**0.28**
	Lower CI	0.45	0.23	0.24	0.15	0.24	0.15	0.23	0.23	0.13	0.19	0.16
ICD-10 Q02	Upper CI	0.92	0.59	0.60	0.44	0.59	0.45	0.58	0.59	0.43	0.53	0.48
Anophthalmia/Microphthalmia	NUMBER	8	7	9	6	6	10	13	10	14	3	6
	RATE	**0.16**	**0.13**	**0.17**	**0.11**	**0.11**	**0.18**	**0.24**	**0.19**	**0.27**	**0.06**	**0.12**
	Lower CI	0.07	0.05	0.08	0.04	0.04	0.09	0.13	0.09	0.15	0.01	0.04
ICD-10 Q11.0, Q11.1, Q11.2	Upper CI	0.31	0.27	0.32	0.23	0.23	0.33	0.42	0.35	0.46	0.17	0.26
Congenital cataract	NUMBER	7	6	9	10	4	8	8	13	8	2	4
	RATE	**0.14**	**0.11**	**0.17**	**0.18**	**0.07**	**0.14**	**0.15**	**0.25**	**0.16**	**0.04**	**0.08**
	Lower CI	0.06	0.04	0.08	0.09	0.02	0.06	0.06	0.13	0.07	0.00	0.02
ICD-10 Q12.0	Upper CI	0.28	0.25	0.32	0.33	0.18	0.28	0.29	0.42	0.30	0.14	0.20

Appendix A.3 Alberta Congenital Anomalies Surveillance System
RCPCH version ICD-10 Q-Chapter (Q00-Q99)
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		2007-2011 (5 years)	2012-2016 (5 years)	2017-2021 (5 years)	2012-2021 (10 years)	2002-2021 (20 years)
Anencephaly	NUMBER	62	58	48	106	215
	RATE	**0.25**	**0.21**	**0.19**	**0.20**	**0.22**
	Lower CI	0.19	0.16	0.14	0.16	0.19
ICD-10 Q00.00, Q00.01, Q00.1	Upper CI	0.32	0.27	0.25	0.24	0.25
Spina Bifida without Anencephaly	NUMBER	105	97	98	195	376
	RATE	**0.42**	**0.36**	**0.38**	**0.37**	**0.38**
	Lower CI	0.34	0.29	0.31	0.32	0.34
ICD-10 Q05..	Upper CI	0.50	0.43	0.47	0.43	0.42
Encephalocele	NUMBER	32	31	30	61	120
	RATE	**0.13**	**0.11**	**0.12**	**0.12**	**0.12**
	Lower CI	0.09	0.08	0.08	0.09	0.10
ICD-10 Q01..	Upper CI	0.18	0.16	0.17	0.15	0.15
Neural Tube Defects (all)	NUMBER	200	187	178	365	715
	RATE	**0.79**	**0.68**	**0.70**	**0.69**	**0.73**
	Lower CI	0.69	0.59	0.60	0.62	0.67
ICD-10 Q00.., Q01.., Q05..	Upper CI	0.91	0.79	0.81	0.77	0.78
Hydrocephalus without Spina Bifida	NUMBER	154	110	70	180	452
	RATE	**0.61**	**0.40**	**0.27**	**0.34**	**0.46**
(Excludes hydranencephaly)	Lower CI	0.52	0.33	0.21	0.29	0.42
ICD-10 Q03	Upper CI	0.72	0.49	0.35	0.39	0.50
Arrhinencephaly/ Holoprosencephaly	NUMBER	65	71	60	131	238
	RATE	**0.26**	**0.26**	**0.24**	**0.25**	**0.24**
	Lower CI	0.20	0.20	0.18	0.21	0.21
ICD-10 Q04.1, Q04.2, Q87.03	Upper CI	0.33	0.33	0.30	0.29	0.27
Microcephaly	NUMBER	112	93	83	176	368
	RATE	**0.44**	**0.34**	**0.33**	**0.33**	**0.37**
	Lower CI	0.37	0.28	0.26	0.29	0.34
ICD-10 Q02	Upper CI	0.54	0.42	0.40	0.39	0.41
Anophthalmia/Microphthalmia	NUMBER	35	38	46	84	148
	RATE	**0.14**	**0.14**	**0.18**	**0.16**	**0.15**
	Lower CI	0.10	0.10	0.13	0.13	0.13
ICD-10 Q11.0, Q11.1, Q11.2	Upper CI	0.19	0.19	0.24	0.20	0.18
Congenital cataract	NUMBER	45	37	35	72	136
	RATE	**0.18**	**0.14**	**0.14**	**0.14**	**0.14**
	Lower CI	0.13	0.10	0.10	0.11	0.12
ICD-10 Q12.0	Upper CI	0.24	0.19	0.19	0.17	0.16

Appendix A.3 **Alberta Congenital Anomalies Surveillance System**
RCPCH version ICD-10 Q Chapter (Q00-Q99)
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		2011	2012	2013	2014	2015	2016	2017	2018	2019	2020	2021
Anotia/Microtia	NUMBER	11	13	18	11	12	18	24	11	10	13	7
	RATE	**0.22**	**0.25**	**0.34**	**0.20**	**0.21**	**0.32**	**0.45**	**0.21**	**0.19**	**0.27**	**0.14**
	Lower CI	0.11	0.13	0.20	0.10	0.11	0.19	0.29	0.11	0.09	0.14	0.06
ICD-10 Q16.0, Q17.2	Upper CI	0.39	0.42	0.53	0.35	0.37	0.51	0.67	0.38	0.36	0.46	0.29
Congenital Heart Defects (all)	NUMBER	645	691	744	678	752	726	676	758	610	591	559
	RATE	**12.73**	**13.21**	**13.99**	**12.21**	**13.30**	**13.09**	**12.66**	**14.51**	**11.85**	**12.16**	**11.35**
	Lower CI	11.77	12.24	13.00	11.31	12.37	12.15	11.72	13.49	10.93	11.20	10.43
ICD-10 Q20.. to Q26..	Upper CI	13.76	14.23	15.03	13.17	14.29	14.08	13.65	15.58	12.83	13.18	12.33
Common Truncus	NUMBER	2	5	8	3	3	8	7	3	4	3	4
(Excludes AP window)	**RATE**	**0.04**	**0.10**	**0.15**	**0.05**	**0.05**	**0.14**	**0.13**	**0.06**	**0.08**	**0.06**	**0.08**
	Lower CI	0.00	0.03	0.06	0.01	0.01	0.06	0.05	0.01	0.02	0.01	0.02
ICD-10 Q20.0	Upper CI	0.14	0.22	0.29	0.15	0.15	0.28	0.27	0.16	0.19	0.17	0.20
Transposition of Great Arteries	NUMBER	20	21	23	29	25	22	19	26	19	23	10
	RATE	**0.39**	**0.40**	**0.43**	**0.52**	**0.44**	**0.40**	**0.36**	**0.50**	**0.37**	**0.47**	**0.20**
	Lower CI	0.24	0.25	0.27	0.35	0.29	0.25	0.21	0.33	0.22	0.30	0.10
ICD-10 Q20.11, Q20.3, Q20.5	Upper CI	0.61	0.61	0.65	0.75	0.65	0.60	0.56	0.73	0.58	0.71	0.37
Tetralogy of Fallot	NUMBER	19	24	27	15	9	19	18	19	16	11	24
(Includes Tetralogy with ASD	**RATE**	**0.38**	**0.46**	**0.51**	**0.27**	**0.16**	**0.34**	**0.34**	**0.36**	**0.31**	**0.23**	**0.49**
aka Pentalogy of Fallot)	Lower CI	0.23	0.29	0.34	0.15	0.07	0.21	0.20	0.22	0.18	0.11	0.31
ICD-10 Q21.3.., Q21.82	Upper CI	0.59	0.68	0.74	0.44	0.30	0.53	0.53	0.57	0.50	0.40	0.72
Ventricular Septal Defect	NUMBER	167	168	174	163	164	187	176	180	157	135	128
	RATE	**3.30**	**3.21**	**3.27**	**2.94**	**2.90**	**3.37**	**3.30**	**3.45**	**3.05**	**2.78**	**2.60**
	Lower CI	2.82	2.74	2.80	2.50	2.48	2.91	2.83	2.96	2.59	2.33	2.17
ICD-10 Q21.0	Upper CI	3.84	3.74	3.80	3.43	3.38	3.89	3.82	3.99	3.57	3.29	3.09
Atrial Septal Defect	NUMBER	99	107	114	123	130	108	116	108	86	95	71
	RATE	**1.95**	**2.05**	**2.14**	**2.22**	**2.30**	**1.95**	**2.17**	**2.07**	**1.67**	**1.95**	**1.44**
	Lower CI	1.59	1.68	1.77	1.84	1.92	1.60	1.80	1.70	1.34	1.58	1.13
ICD-10 Q21.1..	Upper CI	2.38	2.47	2.58	2.65	2.73	2.35	2.61	2.50	2.06	2.39	1.82
Endocardial Cushion Defect	NUMBER	26	30	33	33	30	25	23	34	21	28	32
	RATE	**0.51**	**0.57**	**0.62**	**0.59**	**0.53**	**0.45**	**0.43**	**0.65**	**0.41**	**0.58**	**0.65**
	Lower CI	0.34	0.39	0.43	0.41	0.36	0.29	0.27	0.45	0.25	0.38	0.44
ICD-10 Q21.2..	Upper CI	0.75	0.82	0.87	0.84	0.76	0.67	0.65	0.91	0.62	0.83	0.92
Pulmonary Valve Atresia and Stenosis	NUMBER	30	33	31	41	38	33	35	50	39	36	39
	RATE	**0.59**	**0.63**	**0.58**	**0.74**	**0.67**	**0.59**	**0.66**	**0.96**	**0.76**	**0.74**	**0.79**
	Lower CI	0.40	0.43	0.40	0.53	0.48	0.41	0.46	0.71	0.54	0.52	0.56
ICD-10 Q22.0, Q22.1	Upper CI	0.85	0.89	0.83	1.00	0.92	0.84	0.91	1.26	1.04	1.03	1.08

Appendix A.3 **Alberta Congenital Anomalies Surveillance System**
RCPCH version ICD-10 Q Chapter (Q00-Q99)
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		2007-2011 (5 years)	2012-2016 (5 years)	2017-2021 (5 years)	2012-2021 (10 years)	2002-2021 (20 years)
Anotia/Microtia	NUMBER	63	72	65	137	246
	RATE	**0.25**	**0.26**	**0.25**	**0.26**	**0.25**
	Lower CI	0.19	0.21	0.20	0.22	0.22
ICD-10 Q16.0, Q17.2	Upper CI	0.32	0.33	0.33	0.31	0.28
Congenital Heart Defects (all)	NUMBER	2960	3591	3194	6785	12235
	RATE	**11.75**	**13.15**	**12.53**	**12.85**	**12.41**
	Lower CI	11.33	12.73	12.09	12.55	12.19
ICD-10 Q20.., Q26..	Upper CI	12.18	13.59	12.97	13.16	12.63
Common Truncus	NUMBER	16	27	21	48	78
(Excludes AP window)	**RATE**	**0.06**	**0.10**	**0.08**	**0.09**	**0.08**
	Lower CI	0.04	0.07	0.05	0.07	0.06
ICD-10 Q20.0	Upper CI	0.10	0.14	0.13	0.12	0.10
Transposition of Great	NUMBER	73	120	97	217	377
Arteries	**RATE**	**0.29**	**0.44**	**0.38**	**0.41**	**0.38**
	Lower CI	0.23	0.36	0.31	0.36	0.34
ICD-10 Q20.11, Q20.3, Q20.5	Upper CI	0.36	0.53	0.46	0.47	0.42
Tetralogy of Fallot	NUMBER	86	94	88	182	339
(Includes Tetralogy with ASD aka	**RATE**	**0.34**	**0.34**	**0.35**	**0.34**	**0.34**
Pentalogy of Fallot)	Lower CI	0.27	0.28	0.28	0.30	0.31
ICD-10 Q21.3.., Q21.82	Upper CI	0.42	0.42	0.43	0.40	0.38
Ventricular Septal Defect	NUMBER	777	856	776	1632	3091
	RATE	**3.08**	**3.14**	**3.04**	**3.09**	**3.14**
	Lower CI	2.87	2.93	2.83	2.94	3.03
ICD-10 Q21.0	Upper CI	3.31	3.35	3.27	3.24	3.25
Atrial Septal Defect	NUMBER	452	582	476	1058	1934
	RATE	**1.79**	**2.13**	**1.87**	**2.00**	**1.96**
	Lower CI	1.63	1.96	1.70	1.88	1.88
ICD-10 Q21.1..	Upper CI	1.97	2.31	2.04	2.13	2.05
Endocardial Cushion Defect	NUMBER	129	151	138	289	504
	RATE	**0.51**	**0.55**	**0.54**	**0.55**	**0.51**
	Lower CI	0.43	0.47	0.45	0.49	0.47
ICD-10 Q21.2..	Upper CI	0.61	0.65	0.64	0.61	0.56
Pulmonary Valve Atresia and	NUMBER	152	176	199	375	641
Stenosis	**RATE**	**0.60**	**0.64**	**0.78**	**0.71**	**0.66**
	Lower CI	0.51	0.55	0.68	0.64	0.61
ICD-10 Q22.0, Q22.1	Upper CI	0.71	0.75	0.90	0.79	0.71

Appendix A.3 **Alberta Congenital Anomalies Surveillance System**
RCPCH version ICD-10 Q Chapter (Q00-Q99)
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		2011	2012	2013	2014	2015	2016	2017	2018	2019	2020	2021
Tricuspid Valve Atresia and Stenosis	NUMBER	2	3	3	6	5	6	6	4	7	5	4
	RATE	**0.04**	**0.06**	**0.06**	**0.11**	**0.09**	**0.11**	**0.11**	**0.08**	**0.14**	**0.10**	**0.08**
	Lower CI	0.00	0.01	0.01	0.04	0.03	0.04	0.04	0.02	0.05	0.03	0.02
ICD-10 Q22.4	Upper CI	0.14	0.16	0.16	0.23	0.20	0.23	0.24	0.19	0.28	0.24	0.20
Ebstein's Anomaly	NUMBER	3	3	8	8	3	1	0	5	5	2	1
	RATE	**0.06**	**0.06**	**0.15**	**0.14**	**0.05**	**0.02**	**0.00**	**0.10**	**0.10**	**0.04**	**0.02**
	Lower CI	0.01	0.01	0.06	0.06	0.01	0.00	0.00	0.03	0.03	0.00	0.00
ICD-10 Q22.5	Upper CI	0.17	0.16	0.29	0.28	0.15	0.09	0.04	0.22	0.22	0.14	0.10
Aortic Valve Atresia/Stenosis	NUMBER	5	9	10	13	9	11	9	6	10	12	5
(excludes sub & supra aortic stenosis &	**RATE**	**0.10**	**0.17**	**0.19**	**0.23**	**0.16**	**0.20**	**0.17**	**0.11**	**0.19**	**0.25**	**0.10**
Aortic stenosis found with HLHS)	Lower CI	0.03	0.08	0.09	0.12	0.07	0.10	0.08	0.04	0.09	0.13	0.03
ICD-10 Q23.0	Upper CI	0.23	0.32	0.34	0.40	0.30	0.35	0.32	0.25	0.36	0.43	0.23
Hypoplastic Left Heart Syndrome (HLHS)	NUMBER	18	17	17	16	21	14	24	20	17	16	13
	RATE	**0.36**	**0.32**	**0.32**	**0.29**	**0.37**	**0.25**	**0.45**	**0.38**	**0.33**	**0.33**	**0.26**
	Lower CI	0.21	0.19	0.19	0.17	0.23	0.14	0.29	0.23	0.19	0.19	0.14
ICD-10 Q23.4	Upper CI	0.56	0.52	0.51	0.47	0.57	0.42	0.67	0.59	0.53	0.53	0.45
Coarctation of the Aorta	NUMBER	23	27	25	22	27	33	22	32	22	20	19
	RATE	**0.45**	**0.52**	**0.47**	**0.40**	**0.48**	**0.59**	**0.41**	**0.61**	**0.43**	**0.41**	**0.39**
	Lower CI	0.29	0.34	0.30	0.25	0.32	0.41	0.26	0.42	0.27	0.25	0.23
ICD-10 Q25.1..	Upper CI	0.68	0.75	0.69	0.60	0.70	0.84	0.62	0.86	0.65	0.63	0.60
Cleft Palate without Cleft Lip	NUMBER	36	41	46	40	39	43	37	37	32	38	37
(i.e. cleft palate alone)	**RATE**	**0.71**	**0.78**	**0.86**	**0.72**	**0.69**	**0.78**	**0.69**	**0.71**	**0.62**	**0.78**	**0.75**
	Lower CI	0.50	0.56	0.63	0.52	0.49	0.56	0.49	0.50	0.43	0.55	0.53
ICD-10 Q35..	Upper CI	0.98	1.06	1.15	0.98	0.94	1.04	0.96	0.98	0.88	1.07	1.04
Cleft Lip without Cleft Palate	NUMBER	21	25	25	27	20	21	22	30	20	17	21
(i.e. cleft lip alone)	**RATE**	**0.41**	**0.48**	**0.47**	**0.49**	**0.35**	**0.38**	**0.41**	**0.57**	**0.39**	**0.35**	**0.43**
	Lower CI	0.26	0.31	0.30	0.32	0.22	0.23	0.26	0.39	0.24	0.20	0.26
ICD-10 Q36..	Upper CI	0.63	0.71	0.69	0.71	0.55	0.58	0.62	0.82	0.60	0.56	0.65
Cleft Lip and Cleft Palate	NUMBER	39	31	35	45	57	36	47	45	34	29	43
	RATE	**0.77**	**0.59**	**0.66**	**0.81**	**1.01**	**0.65**	**0.88**	**0.86**	**0.66**	**0.60**	**0.87**
	Lower CI	0.55	0.40	0.46	0.59	0.76	0.45	0.65	0.63	0.46	0.40	0.63
ICD-10 Q37..	Upper CI	1.05	0.84	0.92	1.09	1.31	0.90	1.17	1.15	0.92	0.86	1.18
Cleft Lip with and without Cleft Palate	NUMBER	60	56	60	72	77	57	69	75	54	46	64
	RATE	**1.18**	**1.07**	**1.13**	**1.30**	**1.36**	**1.03**	**1.29**	**1.44**	**1.05**	**0.95**	**1.30**
	Lower CI	0.90	0.81	0.86	1.02	1.08	0.78	1.01	1.13	0.79	0.69	1.00
ICD-10 Q36.., Q37..	Upper CI	1.53	1.39	1.45	1.63	1.70	1.33	1.64	1.80	1.37	1.26	1.66

Appendix A.3 **Alberta Congenital Anomalies Surveillance System**
RCPCH version ICD-10 Q Chapter (Q00-Q99)
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		2007-2011 (5 years)	2012-2016 (5 years)	2017-2021 (5 years)	2012-2021 (10 years)	2002-2021 (20 years)
Tricuspid Valve Atresia and Stenosis	NUMBER	26	23	26	49	88
	RATE	**0.10**	**0.08**	**0.10**	**0.09**	**0.09**
	Lower CI	0.07	0.05	0.07	0.07	0.07
ICD-10 Q22.4	Upper CI	0.15	0.13	0.15	0.12	0.11
Ebstein's Anomaly	NUMBER	15	23	13	36	65
	RATE	**0.06**	**0.08**	**0.05**	**0.07**	**0.07**
	Lower CI	0.03	0.05	0.03	0.05	0.05
ICD-10 Q22.5	Upper CI	0.10	0.13	0.09	0.09	0.08
Aortic Valve Atresia/Stenosis	NUMBER	40	52	42	94	175
(excludes sub & supra aortic stenosis &	**RATE**	**0.16**	**0.19**	**0.16**	**0.18**	**0.18**
Aortic stenosis found with HLHS)	Lower CI	0.11	0.14	0.12	0.14	0.15
ICD-10 Q23.0	Upper CI	0.22	0.25	0.22	0.22	0.21
Hypoplastic Left Heart Syndrome	NUMBER	82	85	90	175	315
	RATE	**0.33**	**0.31**	**0.35**	**0.33**	**0.32**
	Lower CI	0.26	0.25	0.28	0.28	0.29
ICD-10 Q23.4	Upper CI	0.40	0.39	0.43	0.38	0.36
Coarctation of the Aorta	NUMBER	119	134	115	249	439
	RATE	**0.47**	**0.49**	**0.45**	**0.47**	**0.45**
	Lower CI	0.39	0.41	0.37	0.41	0.40
ICD-10 Q25.1..	Upper CI	0.57	0.58	0.54	0.53	0.49
Cleft Palate without Cleft Lip	NUMBER	175	209	181	390	714
(i.e. cleft palate alone)	**RATE**	**0.69**	**0.77**	**0.71**	**0.74**	**0.72**
	Lower CI	0.60	0.67	0.61	0.67	0.67
ICD-10 Q35..	Upper CI	0.81	0.88	0.82	0.82	0.78
Cleft Lip without Cleft Palate	NUMBER	123	118	110	228	441
(i.e. cleft lip alone)	**RATE**	**0.49**	**0.43**	**0.43**	**0.43**	**0.45**
	Lower CI	0.41	0.35	0.38	0.38	0.41
ICD-10 Q36..	Upper CI	0.58	0.52	0.49	0.49	0.49
Cleft Lip and Cleft Palate	NUMBER	228	204	198	402	785
	RATE	**0.91**	**0.75**	**0.78**	**0.76**	**0.80**
	Lower CI	0.79	0.65	0.67	0.69	0.74
ICD-10 Q37..	Upper CI	1.03	0.86	0.89	0.84	0.85
Cleft Lip with and without Cleft Palate	NUMBER	351	322	308	630	1226
	RATE	**1.39**	**1.18**	**1.21**	**1.19**	**1.24**
	Lower CI	1.25	1.05	1.08	1.10	1.17
ICD-10 Q36.., Q37..	Upper CI	1.55	1.32	1.35	1.29	1.32

Appendix A.3 **Alberta Congenital Anomalies Surveillance System**
RCPCH version ICD-10 Q Chapter (Q00-Q99)
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		2011	2012	2013	2014	2015	2016	2017	2018	2019	2020	2021
Choanal Atresia/Stenosis	NUMBER	1	10	8	14	5	9	5	6	9	6	8
	RATE	**0.02**	**0.19**	**0.15**	**0.25**	**0.09**	**0.16**	**0.09**	**0.11**	**0.17**	**0.12**	**0.16**
	Lower CI	0.00	0.09	0.06	0.14	0.03	0.07	0.03	0.04	0.08	0.05	0.07
ICD-10 Q30.0..	Upper CI	0.10	0.35	0.29	0.42	0.20	0.31	0.22	0.25	0.33	0.27	0.32
Oesophageal Atresia/	NUMBER	20	11	16	15	7	13	20	26	18	11	13
Tracheo-oesphageal Fistula	**RATE**	**0.39**	**0.21**	**0.30**	**0.27**	**0.12**	**0.23**	**0.37**	**0.50**	**0.35**	**0.23**	**0.26**
	Lower CI	0.24	0.11	0.17	0.15	0.05	0.12	0.23	0.33	0.21	0.11	0.14
ICD-10 Q39.0 – Q39.4	Upper CI	0.61	0.37	0.49	0.44	0.25	0.40	0.58	0.73	0.55	0.40	0.45
Pyloric Stenosis	NUMBER	44	51	33	49	35	29	29	25	20	21	20
	RATE	**0.87**	**0.97**	**0.62**	**0.88**	**0.62**	**0.52**	**0.54**	**0.48**	**0.39**	**0.43**	**0.41**
	Lower CI	0.63	0.73	0.43	0.65	0.43	0.35	0.36	0.31	0.24	0.27	0.25
ICD-10 Q40.0	Upper CI	1.17	1.28	0.87	1.17	0.86	0.75	0.78	0.71	0.60	0.66	0.63
Small Intestinal	NUMBER	22	24	18	22	20	18	28	16	20	15	14
Atresia/Stenosis (all)	**RATE**	**0.43**	**0.46**	**0.34**	**0.40**	**0.35**	**0.32**	**0.52**	**0.31**	**0.39**	**0.31**	**0.28**
	Lower CI	0.27	0.29	0.20	0.25	0.22	0.19	0.35	0.18	0.24	0.17	0.16
ICD-10 Q41…	Upper CI	0.66	0.68	0.53	0.60	0.55	0.51	0.76	0.50	0.60	0.51	0.48
Duodenal Atresia/Stenosis	NUMBER	14	15	9	14	12	9	14	9	11	8	10
	RATE	**0.28**	**0.29**	**0.17**	**0.25**	**0.21**	**0.16**	**0.26**	**0.17**	**0.21**	**0.16**	**0.20**
	Lower CI	0.15	0.16	0.08	0.14	0.11	0.07	0.14	0.08	0.11	0.07	0.10
ICD-10 Q41.0…	Upper CI	0.46	0.47	0.32	0.42	0.37	0.31	0.44	0.33	0.38	0.32	0.37
Rectal and Large Intestinal	NUMBER	18	19	22	18	21	27	38	34	25	18	17
Atresia/Stenosis (all)	**RATE**	**0.36**	**0.36**	**0.41**	**0.32**	**0.37**	**0.49**	**0.71**	**0.65**	**0.49**	**0.37**	**0.35**
	Lower CI	0.21	0.22	0.26	0.19	0.23	0.32	0.50	0.45	0.31	0.22	0.20
ICD-10 Q42..	Upper CI	0.56	0.57	0.63	0.51	0.57	0.71	0.98	0.91	0.72	0.58	0.55
Rectal Atresia/Stenosis	NUMBER	1	2	2	1	1	2	2	4	2	2	1
	RATE	**0.02**	**0.04**	**0.04**	**0.02**	**0.02**	**0.04**	**0.04**	**0.08**	**0.04**	**0.04**	**0.02**
	Lower CI	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.02	0.00	0.00	0.00
ICD-10 Q42.0…, Q42.1…	Upper CI	0.10	0.13	0.13	0.09	0.09	0.12	0.13	0.19	0.13	0.14	0.10
Anal Atresia/Stenosis	NUMBER	15	15	19	14	19	21	32	27	21	15	14
	RATE	**0.30**	**0.29**	**0.36**	**0.25**	**0.34**	**0.38**	**0.60**	**0.52**	**0.41**	**0.31**	**0.28**
	Lower CI	0.17	0.16	0.22	0.14	0.20	0.23	0.41	0.34	0.25	0.17	0.16
ICD-10 Q42.2…, Q42.3…	Upper CI	0.49	0.47	0.56	0.42	0.52	0.58	0.85	0.75	0.62	0.51	0.48
Other Large Intestinal	NUMBER	2	2	1	3	1	4	4	3	2	1	2
Atresia/Stenosis	**RATE**	**0.04**	**0.04**	**0.02**	**0.05**	**0.02**	**0.07**	**0.07**	**0.06**	**0.04**	**0.02**	**0.04**
	Lower CI	0.00	0.00	0.00	0.01	0.00	0.02	0.02	0.01	0.00	0.00	0.00
ICD-10 Q42.8…, Q42.9…	Upper CI	0.14	0.13	0.10	0.15	0.09	0.18	0.19	0.16	0.13	0.10	0.14

Appendix A.3 **Alberta Congenital Anomalies Surveillance System**
RCPCH version ICD-10 Q Chapter (Q00-Q99)
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		2007-2011 (5 years)	2012-2016 (5 years)	2017-2021 (5 years)	2012-2021 (10 years)	2002-2021 (20 years)
Choanal Atresia/Stenosis	NUMBER	29	46	34	80	147
	RATE	**0.12**	**0.17**	**0.13**	**0.15**	**0.15**
	Lower CI	0.08	0.12	0.09	0.13	0.13
ICD-10 Q30.0..	Upper CI	0.17	0.22	0.19	0.18	0.18
Oesophageal Atresia/	NUMBER	64	62	88	150	263
Tracheo-oesphageal Fistula	**RATE**	**0.25**	**0.23**	**0.35**	**0.28**	**0.27**
	Lower CI	0.20	0.17	0.28	0.24	0.24
ICD-10 Q39.0 – Q39.4	Upper CI	0.32	0.29	0.43	0.33	0.30
Pyloric Stenosis	NUMBER	249	197	115	312	757
	RATE	**0.99**	**0.72**	**0.45**	**0.59**	**0.77**
	Lower CI	0.87	0.62	0.37	0.53	0.71
ICD-10 Q40.0	Upper CI	1.12	0.83	0.54	0.66	0.82
Small Intestinal Atresia/	NUMBER	89	102	93	195	345
Stenosis (all)	**RATE**	**0.35**	**0.37**	**0.36**	**0.37**	**0.35**
	Lower CI	0.28	0.30	0.29	0.32	0.31
ICD-10 Q41…	Upper CI	0.44	0.45	0.45	0.43	0.39
Duodenal Atresia/Stenosis	NUMBER	50	59	52	111	194
	RATE	**0.20**	**0.22**	**0.20**	**0.21**	**0.20**
	Lower CI	0.15	0.16	0.15	0.17	0.17
ICD-10 Q41.0…	Upper CI	0.26	0.28	0.27	0.25	0.23
Rectal and Large Intestinal	NUMBER	110	107	132	239	484
Atresia/Stenosis (all)	**RATE**	**0.44**	**0.39**	**0.52**	**0.45**	**0.49**
	Lower CI	0.36	0.32	0.43	0.40	0.45
ICD-10 Q42..	Upper CI	0.53	0.47	0.61	0.51	0.54
Rectal Atresia/Stenosis	NUMBER	4	8	11	19	33
	RATE	**0.02**	**0.03**	**0.04**	**0.04**	**0.03**
	Lower CI	0.00	0.01	0.02	0.02	0.02
ICD-10 Q42.0…., Q42.1….	Upper CI	0.04	0.06	0.08	0.06	0.05
Anal Atresia/Stenosis	NUMBER	95	88	109	197	402
	RATE	**0.38**	**0.32**	**0.43**	**0.37**	**0.41**
	Lower CI	0.31	0.26	0.35	0.32	0.37
ICD-10 Q42.2…., Q42.3….	Upper CI	0.46	0.40	0.52	0.43	0.45
Other Large Intestinal	NUMBER	11	11	12	23	49
Atresia/Stenosis	**RATE**	**0.04**	**0.04**	**0.05**	**0.04**	**0.05**
	Lower CI	0.02	0.02	0.02	0.03	0.04
ICD-10 Q42.8…., Q42.9….	Upper CI	0.08	0.07	0.08	0.07	0.07

Appendix A.3

Alberta Congenital Anomalies Surveillance System
RCPCH version ICD-10 Q Chapter (Q00-Q99)
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		2011	2012	2013	2014	2015	2016	2017	2018	2019	2020	2021
Hirschsprung Disease	NUMBER	7	9	13	6	6	7	7	8	9	10	4
	RATE	**0.14**	**0.17**	**0.24**	**0.11**	**0.11**	**0.13**	**0.13**	**0.15**	**0.17**	**0.21**	**0.08**
	Lower CI	0.06	0.08	0.13	0.04	0.04	0.05	0.05	0.07	0.08	0.10	0.02
ICD-10 Q43.1..	Upper CI	0.28	0.32	0.42	0.23	0.23	0.26	0.27	0.30	0.33	0.38	0.20
Biliary Atresia	NUMBER	5	3	4	4	3	5	2	5	6	1	0
	RATE	**0.10**	**0.06**	**0.08**	**0.07**	**0.05**	**0.09**	**0.04**	**0.10**	**0.12**	**0.02**	**0.00**
	Lower CI	0.03	0.01	0.02	0.02	0.01	0.03	0.00	0.03	0.04	0.00	0.00
ICD-10 Q44.2	Upper CI	0.23	0.16	0.19	0.18	0.15	0.21	0.13	0.22	0.25	0.10	0.04
Undescended Testes	NUMBER	136	160	147	146	192	152	170	170	123	132	124
(denominator MALE births only)	**RATE**	**5.22**	**5.93**	**5.38**	**5.14**	**6.66**	**5.37**	**6.17**	**6.36**	**4.69**	**5.30**	**4.89**
(>36 weeks gestation)	Lower CI	4.39	5.05	4.55	4.34	5.75	4.55	5.28	5.44	3.90	4.44	4.07
ICD-10 Q53…	Upper CI	6.18	6.93	6.33	6.05	7.68	6.30	7.17	7.39	5.60	6.29	5.84
Hypospadias	NUMBER	126	130	180	189	169	149	154	133	126	144	117
(denominator MALE births only)	**RATE**	**4.84**	**4.82**	**6.59**	**6.65**	**5.86**	**5.27**	**5.59**	**4.97**	**4.80**	**5.79**	**4.62**
	Lower CI	4.03	4.03	5.66	5.74	5.01	4.46	4.74	4.17	4.00	4.88	3.82
ICD-10 Q54 (excl. Q54.4)	Upper CI	5.77	5.73	7.63	7.68	6.82	6.19	6.55	5.90	5.72	6.82	5.53
Epispadias	NUMBER	5	1	5	4	3	2	0	0	1	0	3
(denominator MALE births only)	**RATE**	**0.19**	**0.04**	**0.18**	**0.14**	**0.10**	**0.07**	**0.00**	**0.00**	**0.04**	**0.00**	**0.12**
	Lower CI	0.06	0.00	0.06	0.04	0.02	0.01	0.00	0.00	0.00	0.00	0.02
ICD-10 Q64.0	Upper CI	0.44	0.19	0.42	0.35	0.29	0.24	0.08	0.08	0.19	0.08	0.34
Renal Agenesis/Hypoplasia	NUMBER	35	36	36	30	27	43	47	40	42	38	41
	RATE	**0.69**	**0.69**	**0.68**	**0.54**	**0.48**	**0.78**	**0.88**	**0.77**	**0.82**	**0.78**	**0.83**
	Lower CI	0.48	0.48	0.47	0.37	0.32	0.56	0.65	0.55	0.59	0.55	0.60
ICD-10 Q60..	Upper CI	0.96	0.95	0.94	0.77	0.70	1.04	1.17	1.04	1.10	1.07	1.13
Cystic Kidney	NUMBER	35	43	37	36	51	41	39	47	38	44	41
(exclude single renal cyst	**RATE**	**0.69**	**0.82**	**0.70**	**0.65**	**0.90**	**0.74**	**0.73**	**0.90**	**0.74**	**0.91**	**0.83**
Q61.0)	Lower CI	0.48	0.60	0.49	0.45	0.67	0.53	0.52	0.66	0.52	0.66	0.60
Q61..	Upper CI	0.96	1.11	0.96	0.90	1.19	1.00	1.00	1.20	1.01	1.22	1.13
Bladder Exstrophy	NUMBER	0	1	1	2	2	1	1	1	1	1	2
	RATE	**0.00**	**0.02**	**0.02**	**0.04**	**0.04**	**0.02**	**0.02**	**0.02**	**0.02**	**0.02**	**0.04**
	Lower CI	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00
ICD-10 Q64.1 (excl Q64.10)	Upper CI	0.04	0.10	0.10	0.12	0.12	0.09	0.09	0.10	0.10	0.10	0.14
Obstructive Genitourinary	NUMBER	143	167	139	173	176	181	158	147	146	147	162
Defects (All)	**RATE**	**2.82**	**3.19**	**2.61**	**3.12**	**3.11**	**3.26**	**2.96**	**2.81**	**2.84**	**3.02**	**3.29**
	Lower CI	2.38	2.73	2.20	2.67	2.67	2.81	2.52	2.38	2.39	2.56	2.80
ICD-10 Q62.0 – Q62.3, Q64.2, Q64.3	Upper CI	3.33	3.72	3.09	3.62	3.61	3.78	3.46	3.31	3.34	3.56	3.84

Appendix A.3 **Alberta Congenital Anomalies Surveillance System**
RCPCH version ICD-10 Q Chapter (Q00-Q99)
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		2007-2011 (5 years)	2012-2016 (5 years)	2017-2021 (5 years)	2012-2021 (10 years)	2002-2021 (20 years)
Hirschsprung Disease	NUMBER	37	41	38	79	149
	RATE	**0.15**	**0.15**	**0.15**	**0.15**	**0.15**
	Lower CI	0.10	0.11	0.11	0.12	0.13
ICD-10 Q43.1..	Upper CI	0.20	0.20	0.20	0.19	0.18
Biliary Atresia	NUMBER	16	19	14	33	65
	RATE	**0.06**	**0.07**	**0.05**	**0.06**	**0.07**
	Lower CI	0.04	0.04	0.03	0.04	0.05
ICD-10 Q44.2	Upper CI	0.10	0.11	0.09	0.09	0.08
Undescended Testes	NUMBER	688	797	719	1516	2740
(denominator MALE births only)	**RATE**	**5.32**	**5.70**	**5.50**	**5.60**	**5.42**
(>36 weeks gestation)	Lower CI	4.93	5.31	5.10	5.32	5.22
ICD-10 Q53….	Upper CI	5.73	6.11	5.92	5.89	5.63
Hypospadias	NUMBER	587	817	674	1491	2492
(denominator MALE births only)	**RATE**	**4.54**	**5.84**	**5.15**	**5.51**	**4.93**
	Lower CI	4.18	5.45	4.77	5.23	4.74
ICD-10 Q54 (excl. Q54.4)	Upper CI	4.92	6.26	5.56	5.80	5.13
Epispadias	NUMBER	21	15	4	19	55
(denominator MALE births only)	**RATE**	**0.16**	**0.11**	**0.03**	**0.07**	**0.11**
	Lower CI	0.10	0.06	0.01	0.04	0.08
ICD-10 Q64.0	Upper CI	0.25	0.18	0.08	0.11	0.14
Renal Agenesis/Hypoplasia	NUMBER	156	172	208	380	649
	RATE	**0.62**	**0.63**	**0.82**	**0.72**	**0.66**
	Lower CI	0.53	0.54	0.71	0.65	0.61
ICD-10 Q60..	Upper CI	0.72	0.73	0.93	0.80	0.71
Cystic Kidney	NUMBER	182	208	209	417	773
(excludes single renal cyst Q61.0)	**RATE**	**0.72**	**0.76**	**0.82**	**0.79**	**0.78**
	Lower CI	0.62	0.66	0.71	0.72	0.73
ICD-10 Q61..	Upper CI	0.84	0.87	0.94	0.87	0.84
Bladder Exstrophy	NUMBER	7	7	6	13	26
	RATE	**0.03**	**0.03**	**0.02**	**0.02**	**0.03**
	Lower CI	0.01	0.01	0.01	0.01	0.02
ICD-10 Q64.1 (excl Q64.10)	Upper CI	0.06	0.05	0.05	0.04	0.04
Obstructive Genitourinary	NUMBER	688	836	760	1596	2775
Defects (All)	**RATE**	**2.73**	**3.06**	**2.98**	**3.02**	**2.81**
	Lower CI	2.53	2.86	2.77	2.88	2.71
ICD-10 Q62.0 – Q62.3, Q64.2, Q64.3	Upper CI	2.94	3.28	3.20	3.17	2.92

Appendix A.3 **Alberta Congenital Anomalies Surveillance System**
RCPCH version ICD-10 Q Chapter (Q00-Q99)
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		2011	2012	2013	2014	2015	2016	2017	2018	2019	2020	2021
Hydronephrosis	NUMBER	96	111	98	99	104	106	101	86	85	94	93
	RATE	**1.89**	**2.12**	**1.84**	**1.78**	**1.84**	**1.91**	**1.89**	**1.65**	**1.65**	**1.93**	**1.89**
	Lower CI	1.54	1.75	1.50	1.45	1.50	1.56	1.54	1.32	1.32	1.56	1.52
ICD-10 Q62.0..	Upper CI	2.32	2.56	2.25	2.17	2.23	2.31	2.30	2.03	2.04	2.37	2.31
Pelviureteric Junction Obstruction	NUMBER	11	11	9	12	18	19	17	16	20	15	18
	RATE	**0.22**	**0.21**	**0.17**	**0.22**	**0.32**	**0.34**	**0.32**	**0.31**	**0.39**	**0.31**	**0.37**
	Lower CI	0.11	0.11	0.08	0.11	0.19	0.21	0.19	0.18	0.24	0.17	0.22
ICD-10 Q62.10 & Q62.11	Upper CI	0.39	0.37	0.32	0.38	0.50	0.53	0.51	0.50	0.60	0.51	0.58
Vesicoureteric Junction Obstruction	NUMBER	4	2	2	0	3	3	1	1	3	4	4
	RATE	**0.08**	**0.04**	**0.04**	**0.00**	**0.05**	**0.05**	**0.02**	**0.02**	**0.06**	**0.08**	**0.08**
	Lower CI	0.02	0.00	0.00	0.00	0.01	0.01	0.00	0.00	0.01	0.02	0.02
ICD-10 Q62.12 & Q62.13	Upper CI	0.20	0.13	0.13	0.04	0.15	0.15	0.09	0.10	0.16	0.21	0.20
Posterior Urethral Valves	NUMBER	3	3	4	9	8	2	1	5	6	6	6
(denominator MALE births only)	**RATE**	**0.12**	**0.11**	**0.15**	**0.32**	**0.28**	**0.07**	**0.04**	**0.19**	**0.23**	**0.24**	**0.24**
	Lower CI	0.02	0.02	0.04	0.15	0.12	0.01	0.00	0.06	0.08	0.09	0.09
ICD-10 Q64.20	Upper CI	0.33	0.31	0.37	0.60	0.54	0.24	0.18	0.43	0.49	0.52	0.51
Congenital Deformities Hip (All)	NUMBER	103	125	96	75	79	50	54	52	47	54	42
	RATE	**2.03**	**2.39**	**1.81**	**1.35**	**1.40**	**0.90**	**1.01**	**1.00**	**0.91**	**1.11**	**0.85**
	Lower CI	1.66	1.99	1.46	1.06	1.11	0.67	0.76	0.74	0.67	0.84	0.62
ICD-10 Q65	Upper CI	2.47	2.85	2.21	1.69	1.74	1.19	1.32	1.31	1.21	1.45	1.15
Congenital Hip Dislocation Subluxation and Dysplasia	NUMBER	72	85	66	59	70	45	49	43	45	49	40
	RATE	**1.42**	**1.62**	**1.24**	**1.06**	**1.24**	**0.81**	**0.92**	**0.82**	**0.87**	**1.01**	**0.81**
ICD-10 Q65.0-Q65.5 & Q65.80-Q65.81	Lower CI	1.11	1.30	0.96	0.81	0.97	0.59	0.68	0.60	0.64	0.75	0.58
	Upper CI	1.79	2.01	1.58	1.37	1.57	1.09	1.21	1.11	1.17	1.33	1.11
Reduction Deformity, Upper Limbs	NUMBER	44	39	32	41	38	50	52	44	34	48	43
	RATE	**0.87**	**0.75**	**0.60**	**0.74**	**0.67**	**0.90**	**0.97**	**0.84**	**0.66**	**0.99**	**0.87**
	Lower CI	0.63	0.53	0.41	0.53	0.48	0.67	0.73	0.61	0.46	0.73	0.63
ICD-10 Q71..	Upper CI	1.17	1.02	0.85	1.00	0.92	1.19	1.28	1.13	0.92	1.31	1.18
Reduction Deformity, Lower Limbs	NUMBER	22	19	19	17	28	15	26	33	15	22	21
	RATE	**0.43**	**0.36**	**0.36**	**0.31**	**0.50**	**0.27**	**0.49**	**0.63**	**0.29**	**0.45**	**0.43**
	Lower CI	0.27	0.22	0.22	0.18	0.33	0.15	0.32	0.44	0.16	0.28	0.26
ICD-10 Q72..	Upper CI	0.66	0.57	0.56	0.49	0.72	0.45	0.71	0.89	0.48	0.68	0.65
Diaphragmatic Hernia	NUMBER	18	23	12	15	18	17	20	13	19	20	17
	RATE	**0.36**	**0.44**	**0.23**	**0.27**	**0.32**	**0.31**	**0.37**	**0.25**	**0.37**	**0.41**	**0.35**
	Lower CI	0.21	0.28	0.12	0.15	0.19	0.18	0.23	0.13	0.22	0.25	0.20
ICD-10 Q79.0, Q79.11, Q79.12	Upper CI	0.56	0.66	0.39	0.44	0.50	0.49	0.58	0.42	0.58	0.63	0.55

Appendix A.3 **Alberta Congenital Anomalies Surveillance System**
RCPCH version ICD-10 Q Chapter (Q00-Q99)
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		2007-2011 (5 years)	2012-2016 (5 years)	2017-2021 (5 years)	2012-2021 (10 years)	2002-2021 (20 years)
Hydronephrosis	NUMBER	464	518	459	977	1750
	RATE	**1.84**	**1.90**	**1.80**	**1.85**	**1.77**
	Lower CI	1.68	1.74	1.64	1.74	1.69
ICD-10 Q62.0..	Upper CI	2.02	2.07	1.97	1.97	1.86
Pelviureteric Junction Obstruction	NUMBER	48	69	86	155	247
	RATE	**0.19**	**0.25**	**0.34**	**0.29**	**0.25**
	Lower CI	0.14	0.20	0.27	0.25	0.22
ICD-10 Q62.10 & Q62.11	Upper CI	0.25	0.32	0.42	0.34	0.28
Vesicoureteric Junction Obstruction	NUMBER	15	10	13	23	45
	RATE	**0.06**	**0.04**	**0.05**	**0.04**	**0.05**
	Lower CI	0.03	0.02	0.03	0.03	0.03
ICD-10 Q62.12 & Q62.13	Upper CI	0.10	0.07	0.09	0.07	0.06
Posterior Urethral Valves	NUMBER	23	26	24	50	88
	RATE	**0.18**	**0.19**	**0.18**	**0.18**	**0.17**
	Lower CI	0.11	0.12	0.12	0.14	0.14
ICD-10 Q64.20	Upper CI	0.27	0.27	0.27	0.24	0.21
Congenital Deformities Hip (All)	NUMBER	549	425	249	674	1599
	RATE	**2.18**	**1.56**	**0.98**	**1.28**	**1.62**
	Lower CI	2.00	1.41	0.86	1.18	1.54
ICD-10 Q65	Upper CI	2.37	1.71	1.11	1.38	1.70
Congenital Hip Dislocation, Subluxation and Dysplasia	NUMBER	388	325	226	551	1184
	RATE	**1.54**	**1.19**	**0.89**	**1.04**	**1.20**
ICD-10 Q65.0-Q65.5 & Q65.80-Q65.81	Lower CI	1.39	1.06	0.77	0.96	1.13
	Upper CI	1.70	1.33	1.01	1.13	1.27
Reduction Deformity, Upper Limbs	NUMBER	179	200	221	421	739
	RATE	**0.71**	**0.73**	**0.87**	**0.80**	**0.75**
	Lower CI	0.61	0.63	0.76	0.72	0.70
ICD-10 Q71..	Upper CI	0.82	0.84	0.99	0.88	0.81
Reduction Deformity, Lower Limbs	NUMBER	95	98	117	215	378
	RATE	**0.38**	**0.36**	**0.46**	**0.41**	**0.38**
	Lower CI	0.31	0.29	0.38	0.35	0.35
ICD-10 Q72..	Upper CI	0.46	0.44	0.55	0.47	0.42
Diaphragmatic Hernia	NUMBER	91	85	89	174	334
	RATE	**0.36**	**0.31**	**0.35**	**0.33**	**0.34**
	Lower CI	0.29	0.25	0.28	0.28	0.30
ICD-10 Q79.0.., Q79.11, Q79.12	Upper CI	0.44	0.39	0.43	0.38	0.38

Appendix A.3

Alberta Congenital Anomalies Surveillance System
RCPCH version ICD-10 Q Chapter (Q00-Q99)
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		2011	2012	2013	2014	2015	2016	2017	2018	2019	2020	2021
Abdominal Wall Defects (all)	NUMBER	57	47	48	48	48	45	52	45	47	29	43
	RATE	**1.13**	**0.90**	**0.90**	**0.86**	**0.85**	**0.81**	**0.97**	**0.86**	**0.91**	**0.60**	**0.87**
	Lower CI	0.85	0.66	0.67	0.64	0.63	0.59	0.73	0.63	0.67	0.40	0.63
ICD-10 Q79.2 to Q79.5	Upper CI	1.46	1.20	1.20	1.15	1.13	1.09	1.28	1.15	1.21	0.86	1.18
Omphalocele	NUMBER	24	21	23	19	22	21	27	20	24	17	23
	RATE	**0.47**	**0.40**	**0.43**	**0.34**	**0.39**	**0.38**	**0.51**	**0.38**	**0.47**	**0.35**	**0.47**
	Lower CI	0.30	0.25	0.27	0.21	0.24	0.23	0.33	0.23	0.30	0.20	0.30
ICD-10 Q79.2	Upper CI	0.70	0.61	0.65	0.53	0.59	0.58	0.74	0.59	0.69	0.56	0.70
Gastroschisis	NUMBER	29	18	18	25	19	11	19	20	16	8	12
	RATE	**0.57**	**0.34**	**0.34**	**0.45**	**0.34**	**0.20**	**0.36**	**0.38**	**0.31**	**0.16**	**0.24**
	Lower CI	0.38	0.20	0.20	0.29	0.20	0.10	0.21	0.23	0.18	0.07	0.13
ICD-10 Q79.3	Upper CI	0.82	0.54	0.53	0.66	0.52	0.35	0.56	0.59	0.50	0.32	0.42
All Chromosome Anomalies	NUMBER	232	244	284	287	278	281	311	321	308	295	302
	RATE	**4.58**	**4.66**	**5.34**	**5.17**	**4.92**	**5.06**	**5.82**	**6.14**	**5.98**	**6.07**	**6.13**
	Lower CI	4.01	4.10	4.74	4.59	4.36	4.49	5.20	5.49	5.33	5.40	5.46
ICD-10 Q90-Q99	Upper CI	5.21	5.29	6.00	5.81	5.53	5.69	6.51	6.86	6.69	6.80	6.87
Trisomy 13	NUMBER	12	12	21	12	19	13	17	13	17	7	18
	RATE	**0.24**	**0.23**	**0.39**	**0.22**	**0.34**	**0.23**	**0.32**	**0.25**	**0.33**	**0.14**	**0.37**
	Lower CI	0.12	0.12	0.24	0.11	0.20	0.12	0.19	0.13	0.19	0.06	0.22
ICD-10 Q91.4-Q91.7	Upper CI	0.41	0.40	0.60	0.38	0.52	0.40	0.51	0.42	0.53	0.29	0.58
Trisomy 18	NUMBER	25	28	43	36	44	33	35	52	29	33	31
	RATE	**0.49**	**0.54**	**0.81**	**0.65**	**0.78**	**0.59**	**0.66**	**1.00**	**0.56**	**0.68**	**0.63**
	Lower CI	0.32	0.36	0.59	0.45	0.57	0.41	0.46	0.74	0.38	0.47	0.43
ICD-10 Q91.0-Q91.3	Upper CI	0.73	0.77	1.09	0.90	1.05	0.84	0.91	1.31	0.81	0.95	0.89
Down Syndrome (Trisomy 21)	NUMBER	120	126	134	140	128	120	134	132	126	123	108
	RATE	**2.37**	**2.41**	**2.52**	**2.52**	**2.26**	**2.16**	**2.51**	**2.53**	**2.45**	**2.53**	**2.19**
	Lower CI	1.96	2.01	2.11	2.12	1.89	1.79	2.10	2.11	2.04	2.10	1.80
ICD-10 Q90..	Upper CI	2.83	2.87	2.99	2.98	2.69	2.59	2.97	3.00	2.92	3.02	2.65

Appendix A.3 **Alberta Congenital Anomalies Surveillance System**
RCPCH version ICD-10 Q Chapter (Q00-Q99)
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		2007-2011 (5 years)	2012-2016 (5 years)	2017-2021 (5 years)	2012-2021 (10 years)	2002-2021 (20 years)
Abdominal Wall Defects (all)	NUMBER	248	236	216	452	874
	RATE	**0.98**	**0.86**	**0.85**	**0.86**	**0.89**
	Lower CI	0.87	0.76	0.74	0.78	0.83
ICD-10 Q79.2-Q79.5	Upper CI	1.12	0.98	0.97	0.94	0.95
Omphalocele	NUMBER	102	106	111	217	373
	RATE	**0.40**	**0.39**	**0.44**	**0.41**	**0.38**
	Lower CI	0.33	0.32	0.36	0.36	0.34
ICD-10 Q79.2	Upper CI	0.49	0.47	0.52	0.47	0.42
Gastroschisis	NUMBER	123	91	75	166	383
	RATE	**0.49**	**0.33**	**0.29**	**0.31**	**0.39**
	Lower CI	0.41	0.27	0.23	0.27	0.35
ICD-10 Q79.3	Upper CI	0.58	0.41	0.37	0.37	0.43
All Chromosome Anomalies	NUMBER	1195	1374	1537	2911	5036
	RATE	**4.74**	**5.03**	**6.03**	**5.51**	**5.11**
	Lower CI	4.48	4.77	5.73	5.31	4.97
ICD-10 Q90-Q99	Upper CI	5.02	5.31	6.34	5.72	5.25
Trisomy 13	NUMBER	78	77	72	149	272
	RATE	**0.31**	**0.28**	**0.28**	**0.28**	**0.28**
	Lower CI	0.24	0.22	0.22	0.24	0.24
ICD-10 Q91.4-Q91.7	Upper CI	0.39	0.35	0.36	0.33	0.31
Trisomy 18	NUMBER	142	184	180	364	604
	RATE	**0.56**	**0.67**	**0.71**	**0.69**	**0.61**
	Lower CI	0.47	0.58	0.61	0.62	0.56
ICD-10 Q91.0-Q91.3	Upper CI	0.66	0.78	0.82	0.76	0.66
Down Syndrome (Trisomy 21)	NUMBER	567	648	623	1271	2279
	RATE	**2.25**	**2.37**	**2.44**	**2.41**	**2.31**
	Lower CI	2.07	2.19	2.26	2.28	2.22
ICD-10 Q90..	Upper CI	2.44	2.56	2.64	2.54	2.31

Appendix A.4 Numbers of Cases, Anomalies and Anomalies per Case 1997–2021
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	2183	57.88	1.83
1999	38044	1222	32.12	2421	63.64	1.98
2000	36860	1288	34.94	2362	64.08	1.83
2001	37460	1384	36.95	2602	69.46	1.88
2002	38532	1373	35.63	2551	66.20	1.86
2003	40118	1516	37.79	2612	65.11	1.72
2004	40557	1550	38.22	2901	71.53	1.87
2005	41856	1609	38.44	2900	69.29	1.80
2006	44947	1620	36.04	2734	60.83	1.69
2007	48708	1872	38.43	3165	64.98	1.69
2008	50516	2005	39.69	3472	68.73	1.73
2009	51420	2091	40.67	3663	71.24	1.75
2010	50590	2187	43.23	3707	73.28	1.70
2011	50662	2087	41.19	3671	72.46	1.76
2012	52318	2133	40.77	3757	71.81	1.76
2013	53180	2156	40.54	3825	71.93	1.77
2014	55506	2192	39.49	3909	70.42	1.78
2015	56524	2263	40.04	4071	72.02	1.80
2016	55481	2208	39.80	4069	73.34	1.84
2017	53399	2283	42.75	4262	79.81	1.87
2018	52245	2091	40.02	4204	80.47	2.01
2019	51492	2131	41.39	3901	75.76	1.83
2020	48611	1994	41.02	3765	77.45	1.89
2021	49256	2013	40.87	3966	80.52	1.97
1997–2021	**1172794**	**45587**	**38.87**	**82656**	**70.48**	**1.81**

Alberta Total Births from: Alberta Vital Statistics Annual Reviews for 1997-2021

Appendix A.5 Chi Trend Table for Reported Anomalies 1997–2021

Anomaly	X^2	p Value	Direction*
Anencephaly	5.62	0.0178	↓
Spina bifida without anencephaly	0.01	0.9203	↔
Encephalocele	0.52	0.4708	↔
Neural tube defects (all)	2.99	0.0838	?↓
Hydrocephalus without spina bifida	18.97	<0.0001	↓
Arhinencephaly/ Holoprosencephaly	1.83	0.1761	↔
Microcephaly	0.56	0.4543	↔
Anophthalmia/Microphthalmia	0.70	0.4028	?↓
Congenital cataract	0.00	1.00	↔
Anotia/Microtia	3.42	0.0644	?↑
Congenital heart defects (all)	4.13	0.0421	↑
Common truncus	1.58	0.2088	?↑
Transposition of great arteries	0.36	0.5485	↔
Tetralogy of Fallot	1.22	0.2694	?↑
Ventricular septal defect	0.90	0.3428	↔
Atrial septal defect	1.87	0.1715	?↓
Endocardial cushion defect	2.26	0.1328	?↑
Pulmonary valve atresia/stenosis	2.98	0.0843	?↑
Tricuspid valve atresia/stenosis	0.95	0.3297	?↑
Ebstein's anomaly	0.13	0.7184	↔
Aortic valve atresia/stenosis	5.00	0.0253	↓
Hypoplastic left heart syndrome	1.35	0.2453	?↑
Coarctation of the aorta	7.01	0.0081	↑
Cleft palate without cleft lip (CPO)	3.68	0.0551	?↓
Cleft lip without cleft palate (CLO)	0.01	0.9203	↔
Cleft lip and cleft palate (CL+CP)	0.00	1.00	↔
Cleft lip with and without cleft palate (CL+/-CP)	0.01	0.9203	↔
Choanal atresia/stenosis	0.63	0.4274	?↓
Oesophageal atresia/trachea-oesophageal fistula	2.21	0.1371	?↑
Pyloric stenosis	29.36	<0.0001	↓
Small intestinal atresia/stenosis (all)	0.02	0.8875	↔
Duodenal atresia/stenosis	0.85	0.3566	?↑
Rectal and large intestinal atresia/stenosis (all)	15.96	<0.0001	↓
Rectal atresia/stenosis	4.27	0.0388	↓
Anal atresia/stenosis	8.91	0.0028	↓
Ano-rectal atresia/stenosis	12.02	0.0005	↓
Other large intestinal atresia/stenosis	4.77	0.0290	↓
Hirschsprung's disease	0.51	0.4751	↔
Biliary atresia	0.05	0.8231	↔
Undescended testes (male denominator)	10.12	0.0015	↑
Hypospadias (male denominator)	55.23	<0.0001	↑
Epispadias (male denominator)	2.94	0.0864	?↓
Renal agenesis/hypoplasia	16.68	<0.0001	↑
Cystic kidney	3.91	0.0480	↑
Bladder exstrophy	1.11	0.2921	?↓
Obstructive genitourinary defects (all)	51.22	<0.0001	↑
Hydronephrosis	28.32	<0.0001	↑
UPJ obstruction	18.95	<0.0001	↑
VUJ obstruction (based on very few cases per yr. - range 0-4)	6.48	0.0109	↑
Posterior urethral valves (male denominator)	0.47	0.4930	?↑
Congenital deformities of hip (all)	92.37	<0.0001	↓
Congenital hip dislocation, subluxation, dysplasia	33.25	<0.0001	↓
Reduction deformity, upper	2.90	0.0886	?↑
Reduction deformity, lower	0.99	0.3197	?↑
Diaphragmatic hernia	0.25	0.6171	↔
Abdominal wall defects (all)	2.50	0.1138	?↑
Omphalocele	12.02	0.0005	↑
Gastroschisis	1.06	0.3032	↔
All chromosome anomalies	139.37	<0.0001	↑
Trisomy 13	5.80	0.0160	↑
Trisomy 18	19.47	<0.0001	↑
Trisomy 21	22.56	<0.0001	↑

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

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