Alberta Health

Alberta Congenital Anomalies Surveillance System: Eleventh Report

1997 – 2014

Analytics and Performance Reporting Branch

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Alberta Congenital Anomalies Surveillance System

Eleventh Report

1997 – 2014

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ACRONYMS FOR ORGANIZATIONS MENTIONED IN THE REPORT

ACASS Alberta Congenital Anomalies Surveillance System

AHS Alberta Health Services
www.albertahealthservices.ca

AH Alberta Health
www.health.alberta.ca

CCASN Canadian Congenital Anomalies Surveillance Network
http://www.phac-aspc.gc.ca/ccasn-rcsac/index-eng.php

CCASS Canadian Congenital Anomalies Surveillance System

CPSS Canadian Perinatal Surveillance System
http://www.phac-aspc.gc.ca/rhs-ssg/

ICBDSR International Clearinghouse for Birth Defects Surveillance and Research
www.icbdsr.org

NBDPN National Birth Defects Prevention Network
www.nbdpn.org

PHAC Public Health Agency of Canada
www.phac-aspc.gc.ca/index-eng.php

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1. ACASS Activities and Report Summary

1. This is the eleventh in a series of reports detailing the birth prevalence of congenital anomalies in Alberta particularly the years 1997–2014 inclusive.

2. The International Classification of Diseases – 10th Edition (ICD-10-CA) has been adopted by Alberta hospital reporting data systems and ACASS 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 Guidelines for Conducting Birth Defects Surveillance (http://www.nbdpn.org/docs/SGSC_-_Ch3_Case_Definition_-_final_draft_3-24-15.pdf) 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 disorders of metabolism are monitored by ACASS. Data on such disorders can be provided to interested parties upon request.

3. The numerator data includes not only live births and stillbirths, but also fetal losses <20 weeks gestation with congenital anomalies. Denominator data include live births and stillbirths only. By including fetal losses in the numerator, the reported rates should be more representative of true congenital anomaly rates. Fetal losses have been ascertained since 1997. Data provided in this report include the years 2000-2014 however data from 1980 onward can be accessed at http://www.health.alberta.ca/newsroom/pub-pregnancy-birth.html and by request. Fetal losses will not be included in the numerators before 1997.

4. Microcephaly rates have been stable in Alberta. This report documents rates that precede the current concerns about Zika virus and its effect on neurological development of the fetus and infant, specifically microcephaly. However it is of value to have long term baseline data from which to investigate potential rate changes over time **(p. 14-15)**.

5. 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. Gastroschisis continues to show an increase and is particularly prevalent in young mothers (especially those <20 years of age), which is consistent with worldwide observations from other jurisdictions **(p. 20)**.

    5.2. Omphalocele rates are also increasing but these rates are driven by a higher frequency found in higher maternal ages (i.e. 40 years of age and older). Because omphalocele is more often associated with chromosome abnormalities, it is not unexpected that the rates would be higher in older mothers **(p. 21)**. In fact, 52% of cases of omphalocele in mothers over 40 years of age had a chromosome anomaly.

    5.3. The continued increase in Down syndrome is likely attributable to the increased number of women giving birth aged 35 years or older however the rates seem to have stabilized over the past two years **(p. 23)**.

    5.4. Trisomy 13 and Trisomy 18 are increasing, again likely attributable to advanced maternal age at birth **(p. 23)**.

    5.5. Rates of anotia/microtia **(p. 16)** appear to be increasing somewhat although the increase is not statistically significant (p=0.0714). Earlier reports demonstrated a more marked upward trend. ACASS will continue to monitor the rates.

5.6. Obstructive genitourinary defects are also increasing, perhaps related to better reporting and follow-up **(p. 19)**.

5.7. Anorectal malformation rates have continued to decline significantly since 1997 (<0.0001) **(p. 25-26)**.

6. Hypospadias rates are rising in Alberta. Rates vary world-wide with conflicting data whether trends are increasing, decreasing or remaining unchanged. Methodological issues such as ascertainment methods, definitions etc. can influence the results **(p. 29-30)**.

7. The percentage of births to women 35 years of age and over continues to increase with almost nineteen per cent of women in this age category giving birth in 2014 compared to four per cent in 1980 (over 280% increase in 37 years) **(p. 22)**.

8. The total number of Alberta births (live births and stillbirths) to Alberta residents has increased from 36,797 in 1997 to 55,490 in 2014, a 51 percent increase over 15 years.

9. Although the formal Canadian Congenital Anomalies Surveillance Network (CCASN) (http://www.phac-aspc.gc.ca/ccasn-rcsac/index-eng.php) has been disbanded (a Public Health Agency of Canada (PHAC) initiative), members of ACASS (T. Bedard, B. Sibbald) continue to be involved on an informal basis with the Canadian Congenital Anomalies Surveillance System (CCASS), administered by the Maternal and Infant Health Section of PHAC. An ACASS member (B. Sibbald) also belongs to the External Advisory Committee for the Canadian Perinatal Surveillance System (CPSS) whose mandate is to provide expert advice on national perinatal health surveillance which includes issues involving congenital anomalies surveillance.

10. An ACASS member (T. Bedard) also participates in the Stakeholders Partnering for Arthrogryposis Research Client-Centred Care (SPARC) Network. The Network is funded by the Canadian Institutes of Health Research and Shriners Hospitals for Sick Children. The goals are to establish an international arthrogryposis registry, 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.

11. ACASS continues its affiliation with the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR) (http://www.icbdsr.org/page.asp?p=9895&l=1) and has participated in group studies in a number of congenital anomalies including craniofacial defects, very rare defects, gastroschisis, holoprosencephaly and Down syndrome ascertainment (**see Surveillance and Research Projects, p. 32**).

12. An ACASS member (B. Sibbald) also belongs to the Alberta Perinatal Health Programme (APHP) Data Process Working Group whose mandate is to review data requests that come to the APHP to ensure the request is fully understood, the request is appropriate, the data are available and the request complies with data privacy and confidentiality policies of the APHP.

2. Introduction

This report provides updated data on congenital anomalies ascertained in Alberta from the years 1997–2014 inclusive. For the current release, the anomalies outlined in the National Birth Defects Prevention Network's (NBDPN) Guidelines for Conducting Birth Defects Surveillance (2004) are reported along with some others that might be of interest, however, data on other anomalies can be provided upon request.

The numerator data includes all fetal losses <20 weeks gestation with congenital anomalies. This differs from reports prior to 1997 where live births and stillbirths only were used. The reported rates should be 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 http://www.health.alberta.ca/newsroom/pub-pregnancy-birth.html; 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. Since 1996, funding has been provided by Alberta Health, Analytics and Performance Reporting Branch. ACASS continues to work closely with Alberta Vital Statistics and relies on them for the provision of notifications of births, deaths and stillbirths (**see Case Ascertainment, p.6**).

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 or antenatal screening);
5) assist with health related program planning and development through the provision of data;
6) participate in research into the etiology and natural history of birth defects;
7) assist with research through provision of congenital anomalies data; and
8) 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 to determine whether they belong to an existing or new syndrome complex can be explored.

A principle feature of a surveillance system is timeliness; however data collection and analysis 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 received. There can also be a lengthy delay in obtaining published data from Vital Statistics which is used for the denominators in our calculations.

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 an 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, after 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 ascertainment of the same infant can occur and is encouraged, as this frequently improves the quality and reliability of the data.

As several malformations may occur in the same infant, it is advantageous to allow 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** (**p. 38**) report anomaly rates.

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. 36**) shows the 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. 37**) 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 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 in 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 Research Assistant and Manager. If the information is unclear, the Manager, on behalf of the Medical Consultant, writes to the physician responsible for the case seeking clarification. A stamped, addressed envelope is included with the letter and the physician is asked to respond at the bottom of the letter thus making the mechanics of replying easy. The response from physicians has been very satisfactory and usually this is sufficient to make a decision whether to accept or reject an anomaly or case. Any questionable diagnosis that is not confirmed is not entered into the database. Some cases also excluded contain diagnoses that do not belong in a congenital anomaly system 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 is comprised of a paediatric cardiologist, neonatologist/epidemiologist, paediatric pathologist, medical geneticist (medical consultant) with occasional input from a paediatric neurologist, paediatric nephrologist, paediatric orthopaedic surgeon, paediatric general surgeon and a perinatologist/obstetrician.

3.4 Anomaly Coding

Coding is done at the Calgary office 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 (Medical Geneticist). In the past, we were able to code only six anomalies per case but since 1997 we have been coding all eligible anomalies 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 source, thus the unique identifier reduces the risk of duplicate entries for a case.

Data linkage has been achieved with the Alberta Perinatal Health Programme (APHP) by way of the personal health number to ascertain maternal risk factor data, such as maternal smoking, drinking and use of street drugs 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 Manager, Research Assistant, Secretary, Clerk 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.

Should record level data be required for research purposes, a request should be made to ACASS, however such data are ultimately released through Alberta Health, Analytics and Performance Reporting Branch. In this situation, it would be appropriate to first contact ACASS with an outline of the proposal to determine the feasibility of the study and whether or not ACASS has the necessary data. An e-mail should then be sent to health.resdata@gov.ab.ca complete with the proposal and appropriate ethics approvals.

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 per cent) 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 per cent 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 to us does not allow studies to determine etiology. If increasing trends indicate there is a potentially serious problem, then separate investigative studies need to be done. However, 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. Patterns of Selected Congenital Anomalies in Alberta

4.1 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 2014. Sentinel anomalies are those which the International Clearinghouse of 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. 53)** for other anomalies listed in the report**.**

Table 4.1.1 Chi Squared Linear Trend Analysis and p-values for Selected Anomalies 1997–2014 Inclusive (Live Births, Stillbirths & ToPs)

Anomaly	Trend Direction	Chi Squared Analysis (χ^2LT)	p-value
Neural Tube Defects	No significant change	1.07	0.3009
Anencephaly	No significant change	1.20	0.2733
Spina Bifida	No significant change	0.00	1.0000
Hydrocephalus	No significant change	0.20	0.6547
Cleft Lip +/- Cleft Palate	No significant change	0.62	0.4310
Cleft Palate	**Decreasing**	3.97	0.0463
Oesophageal Atresia/Stenosis	No significant change	0.23	0.6315
Anorectal & Large Intestine Atresia/Stenosis	**Decreasing**	23.28	<0.0001
Hypospadias*	**Increasing**	51.10	<0.0001
Epispadias*	No significant change	0.61	0.4348
Renal Agenesis/Hypoplasia	No significant change	2.33	0.1269
Limb Reductions - upper	No significant change	0.28	0.5967
Limb Reductions - lower	No significant change	0.40	0.5271
Gastroschisis	**Increasing**	5.96	0.0146
Omphalocele	**Increasing**	7.92	0.0049
Down Syndrome	**Increasing**	23.02	0.0001
Hypoplastic Left Heart Syndrome	No significant change	0.68	0.4096

*Hypospadias and Epispadias calculated for male births only

Table 4.1.2 presents a comparison of birth prevalence rates between ACASS and a selection of other countries or regions as reported in the most recently published Annual Report of the International Clearinghouse for Birth Defects Surveillance and Research, 2014 (with data through 2012).

Table 4.1.2 Selected Anomaly Rates for Alberta and Other Jurisdictions Reporting to the ICBDSR, 2007-2011* Rates per 1000 Total Births, ToPs included.

Anomaly	Alberta	Western Australia	Atlanta	Utah	Wales	Texas	Hungary
Neural Tube Defects	0.80	1.33	0.94	0.70	1.43	0.76	0.63
Anencephaly	0.25	0.55	0.33	0.27	0.51	0.27	0.16
Spina Bifida	0.42	0.57	0.46	0.35	0.72	0.39	0.39
Hydrocephalus	0.62	0.77	0.92	0.22	0.82	0.72	0.53
Cleft Lip +/- Cleft Palate	1.39	1.08	0.91	1.36	1.14	1.04	0.73
Cleft Palate	0.78	1.02	0.48	0.55	0.84	0.59	0.51
Oesophageal Atresia/Stenosis	0.26	0.41	0.24	0.28	0.29	0.21	0.25
Anorectal Atresia/Stenosis	0.39	0.46	0.43	0.32	0.42	0.49	0.27
Hypospadias†	2.33	3.46	1.76	0.95	3.13	1.66	2.69
Limb Reductions	1.24§ (**anomalies**)	0.70	0.49	0.64	0.77	0.60	0.33
Gastroschisis	0.48	0.38	0.44	0.46	0.51	0.61	0.11
Omphalocele	0.41	0.43	0.33	0.29	0.41	0.20	0.16
Down Syndrome	2.25	2.92	1.93	1.45	2.27	1.40	1.91
Renal Agenesis	0.15	0.56	0.41	0.33	0.16	0.20	0.17
Hypoplastic Left Heart Syndrome	0.31	0.22	0.24	0.34	0.28	0.22	0.25

* http://www.icbdsr.org/filebank/documents/ar2005/Report2014.pdf

† Alberta, Western Australia, Wales and Hungary report all hypospadias; Atlanta, Texas and Utah exclude glanular or 1st degree hypospadias

§1997-2012 **case** rate 0.59/1000 total births AJMG 2015, 167A:2599-2609

4.2 Selected Anomalies

4.2.1 Selected Anomaly Definitions

(Adapted from NBDPN guidelines: http://www.nbdpn.org/)

Abdominal Wall Defects

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

Anorectal Atresia Stenosis

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

Anotia/Microtia

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

Chromosome Anomalies

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

Cleft Lip and Palate

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

Congenital Heart Disease

- ***Aortic valve stenosis –*** obstruction or narrowing of the aortic valve impairing blood flow from the left ventricle to the aorta.
- ***Atrial Septal Defect (ASD) –*** opening in the septum that divides the right and left atria of the heart.
- ***Coarctation of the aorta –*** narrowing of the descending aorta obstructing blood flow from the heart to the rest of the body.
- ***Hypoplastic Left Heart Syndrome*** – a condition in which the structures on the left side of the heart and the aorta are extremely small. Classically, this condition includes hypoplasia of the left ventricle, atresia or severe hypoplasia of the mitral and aortic valves, 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.

Epispadias
Displacement of the opening of the urethra dorsally and proximally (on the top and closer to the body) in relation to the tip of the glans of the penis.

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) herniated.
- ***Encephalocele*** – herniation of brain tissue and/or meninges through a defect in the skull.

Oesophageal Atresia/Stenosis
A condition in which the oesophagus ends in a blind pouch and fails to connect with the stomach.

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.

4.2.2 Neural Tube Defects

There is no significant trend with neural tube defects as a whole or within the sub-groups anencephaly, spina bifida and encephalocele after the initial drop in rates after 1997. This decrease was likely due to the implementation of folic acid fortification (FAF) in flour and cereals. It appeared, as mentioned in previous reports, that the rates of spina bifida might have been inching upward from a low in 2002 but after the unexplained increase in spina bifida in 2006 and a slight increase in NTDs in 2009, the rates appear to be returning to lower levels. The increase in 2006, where the prevalence rate returned to pre fortification levels, was explained by the fact that 42% of the cases (11/26) were likely not responsive to FAF (6 chromosomal, 3 lipomeningocele, 2 syndromes). Overall the rates have been stable over the past 10 years (2005-2014 rate 0.76 per 1000 total births; 95% CI 0.69-0.84). It is normal to see fluctuations in rates from year to year particularly with small numbers of births with NTDs and degree of ascertainment. ACASS is dependent upon cases being reported to us so missing even a small number will affect rates considerably. Of note, ACASS codes NTDs hierarchically. For example, an infant with both anencephaly and spina bifida will be counted once as a case of anencephaly or if born with 2 spina bifida lesions, the higher of the two will be counted.

Figure 4.2.1 All Neural Tube Defects 1997– 2014 (Rate per 1000 total births)

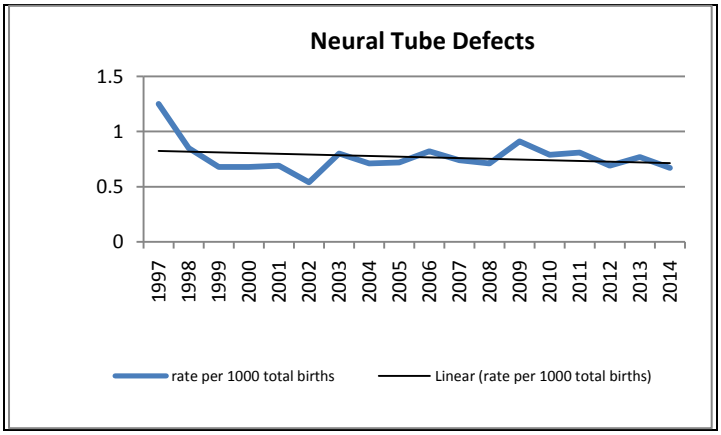
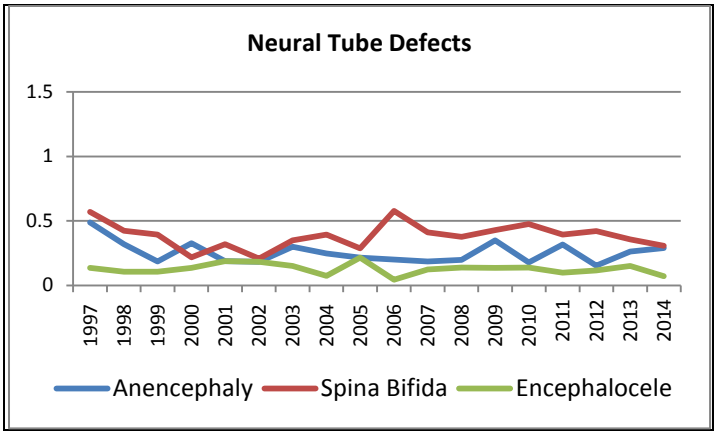


Figure 4.2.2 Neural Tube Defects: Spina Bifida, Anencephaly and Encephalocele 1997–2014 (Rate per 1000 total births)



4.2.3 Microcephaly

There are many known causes of microcephaly such as single gene disorders; chromosome abnormalities; teratogens such as alcohol, rubella, cytomegalovirus (CMV); or other events such as anoxia or haemorrhage, that disrupt the developing brain resulting in a smaller head circumference. There are also some cases for which we do not yet know or understand the cause for the unusually small head circumference. Although there are standard definitions for microcephaly, ACASS does not always have the head circumference measurement provided to us. We do accept a diagnosis of microcephaly when indicated. Nevertheless, despite our not receiving actual measurements in all cases, we can provide a useful guide to what is occurring in Alberta.

Although the data presented in this report precedes the current concerns about Zika virus and its association with microcephaly, good long term baseline data is important to help determine whether or not there has been a significant increase in rates or whether or not there should be cause for concern.

The following graphs indicate that microcephaly rates have been stable in the province over the 15 years between 2000 and 2014. Denominator data from Alberta Vital Statistics and from ACASS are not yet available for 2015. ACASS ascertains congenital anomalies up to one year of age thus the 2015 congenital anomalies data will not be complete until the end of 2016.

As the graphs demonstrate, whether or not we include known potential causes of the microcephaly, there have been no significant changes in the rates.

Figure 4.2.3 Microcephaly – all cases 2000-2014 (rate per 1000 total births)

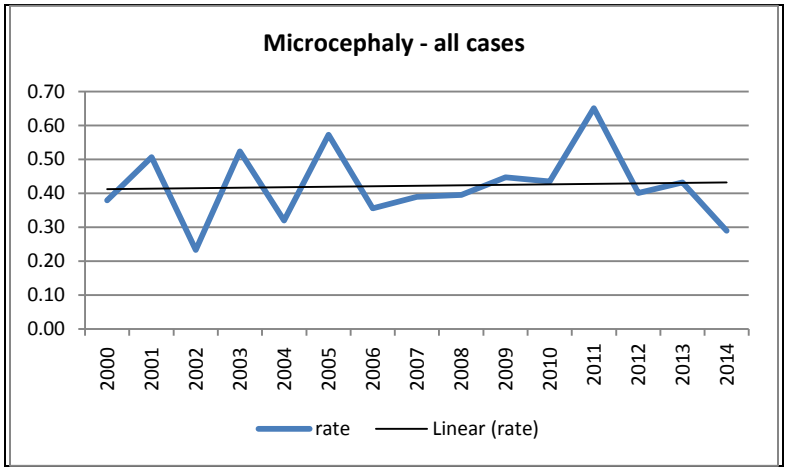
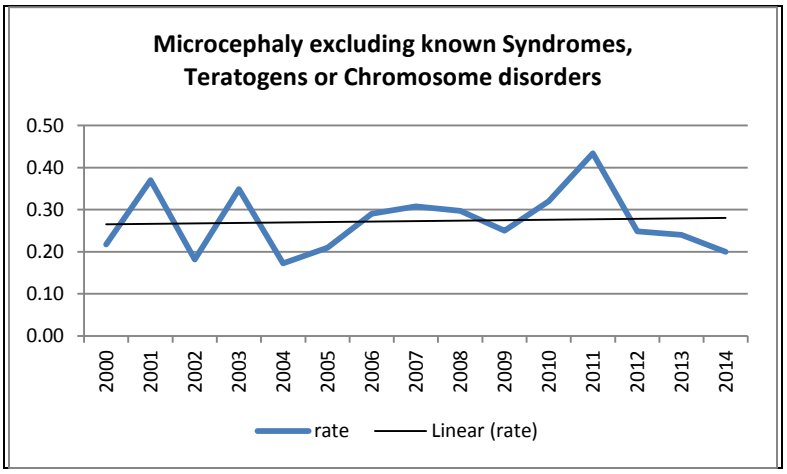


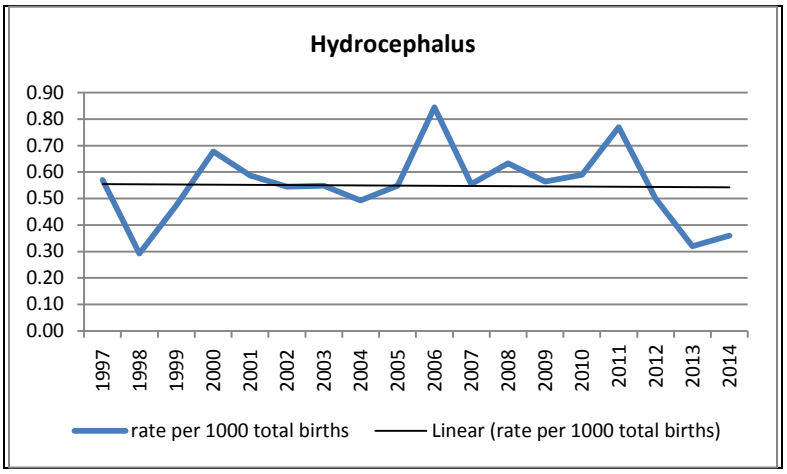
Figure 4.2.4 Microcephaly – excluding known syndromes, teratogens or chromosome disorders 2000-2014 (rate per 1000 total births)



4.2.4 Hydrocephalus

Rates of hydrocephalus do fluctuate from year to year but there has been no significant trend, either up or down, since 1997 (p=0.6547). There appears to have been a drop in rates since 2012 likely due to ascertainment however ACASS will continue to monitor the rates to determine whether this decrease is sustained over time.

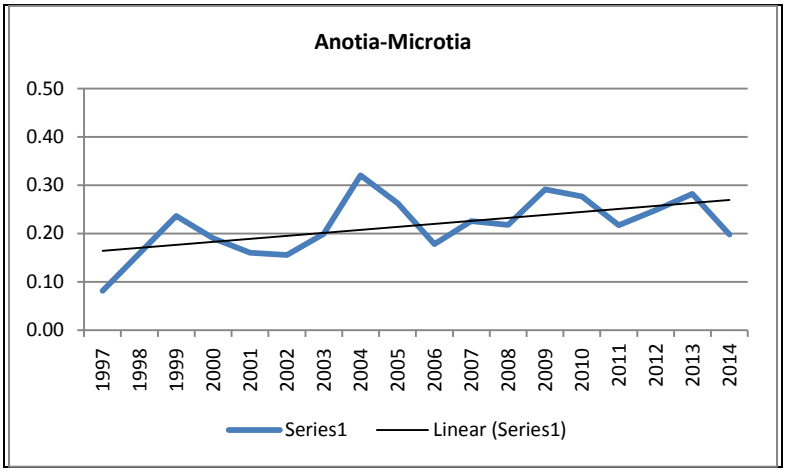
Figure 4.2.5 Hydrocephalus 1997–2014 (Rate per 1000 total births)



4.2.5 Anotia/Microtia

Anotia/Microtia rates appear to be increasing slightly although the trend is not significant (p=0.0714). The trend upward in the previous report was more marked, however an investigation was done and no particular pattern emerged. ACASS will continue to monitor the rates. As noted in previous years, the numbers are small so the addition of even 1 case can affect rates dramatically.

Figure 4.2.6 Anotia/Microtia 1997–2014 (Rate per 1000 total births)



4.2.6 Cleft Lip and Palate

The rate for cleft lip with or without cleft palate (CL ± CP) has remained stable (**Figure 4.2.7**). A review of studies with data from 10 years or more showed fluctuations similar to Alberta but there are no consistent trends (Lowry et al. 2014). The decline in the cleft palate alone (CP) rate is unexplained (**Figure 4.2.8)**.

Studies of many varieties – observational, case-control and randomised control trials - have indicated a lowering of the prevalence of CL ± CP and/or CP following appropriate timing and intake of folic acid (FA) ± multivitamins and/or a high diet quality. Folic acid fortification (FAF) alone may not be sufficient as was seen in Alberta and elsewhere (Bell et al. 2013). High daily doses of FA (3-6 mg) were cited by Czeizel (1999) and an even higher dose (10 mg daily) reduced the recurrence of orofacial clefts (Tolarova and Harris 1995).

Wehby and Murray (2010) summarised a large number of studies of the role of FA and concluded that the evidence remained inconclusive. Studies from two countries, at differing periods of time in each, revealed not only inconsistent but also contradictory results with both positive and negative effects (Norway – Wilcox et al. 2007 and Gildestad et al. 2015; Netherlands – van Rooij et al. 2004 and Rozendaal et al. 2013). Indeed in the latter study there was an increase in CP prevalence however Czeizel (2013) pointed out some of the problems with observational studies such as Rozendaal's.

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Figure 4.2.7 Cleft Lip +/- Cleft Palate (CL+/-CP) 1997–2014 (Rate per 1000 total births)

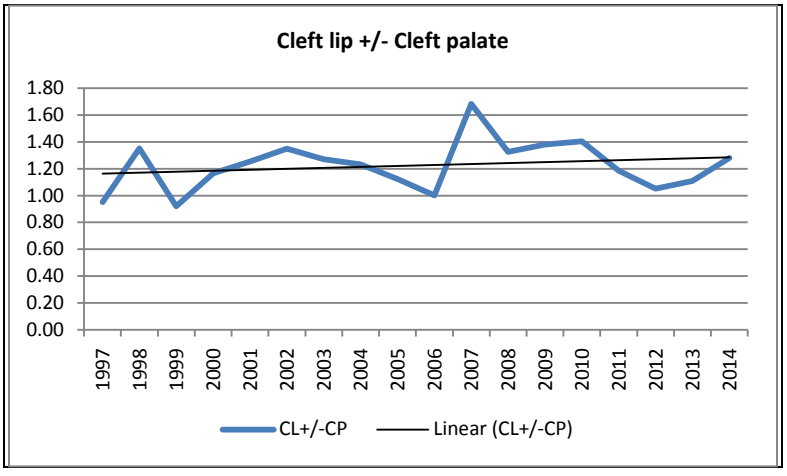
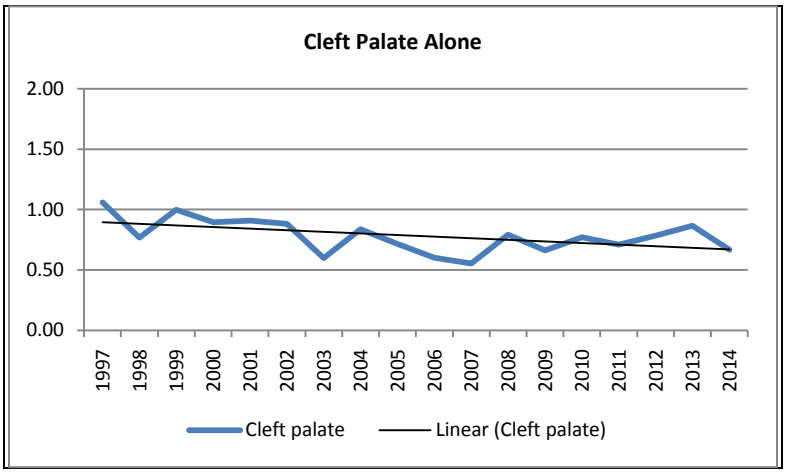


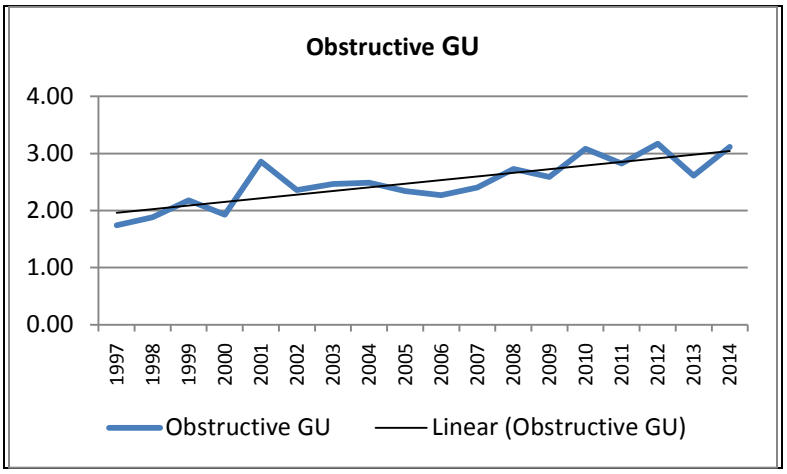
Figure 4.2.8 Cleft Palate Alone 1997–2014 (Rate per 1000 total births)



4.2.7 Obstructive Genitourinary

Obstructive GU anomalies are increasing (p<0.0001). Vesicoureteric junction (VUJ) obstruction and hydronephrosis seem to be the main drivers of this increase although there are very few cases of VUJ per year (n= 0-4). Pelviureteric junction obstruction (UPJ) on the other hand does not demonstrate a significant upward trend (p=0.3802). Hydronephrosis is increasing significantly (p<0.0001) perhaps related to better prenatal detection with subsequent ultrasound follow-up after birth.

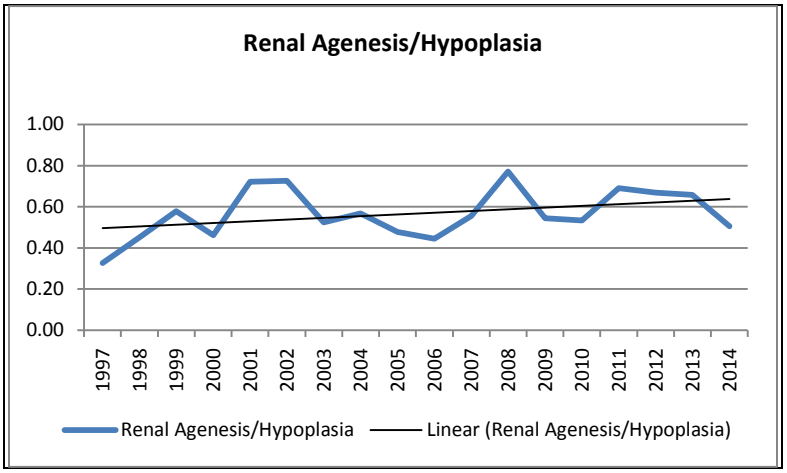
Figure 4.2.9 Obstructive Genitourinary tract anomalies 1997–2014 (Rate per 1000 total births)



4.2.8 Renal Agenesis/Hypoplasia

The rates of renal agenesis have been stable since 1997 with no significant trend either up or down (p=0.1269).

Figure 4.2.10 Renal Agenesis/Hypoplasia 1997–2014 (Rate per 1000 total births)



4.2.9 Abdominal Wall Defects

Abdominal wall defects include mainly gastroschisis and omphalocele (**Figure 4.2.11**). Although the total rates of gastroschisis have declined over the past 3 years, the overall trend is in keeping with many other jurisdictions both provincially and internationally (Moore et al. 2013). Maternal risk factors are well described and include for example, younger maternal age (**Figure 4.2.12**), smoking and pre-gestational or gestational diabetes (Skarsgard et al. 2015). Survival of an infant with gastroschisis exceeds 90% but in addition to surgery require prolonged hospitalisation in high intensity nurseries making them among the most expensive of congenital anomalies to treat (Skarsgard et al. 2008).

Figure 4.2.11 Abdominal Wall Defects – Gastroschisis and Omphalocele in Total Births Alberta, 1997–2014 (Rate per 1000 total births)

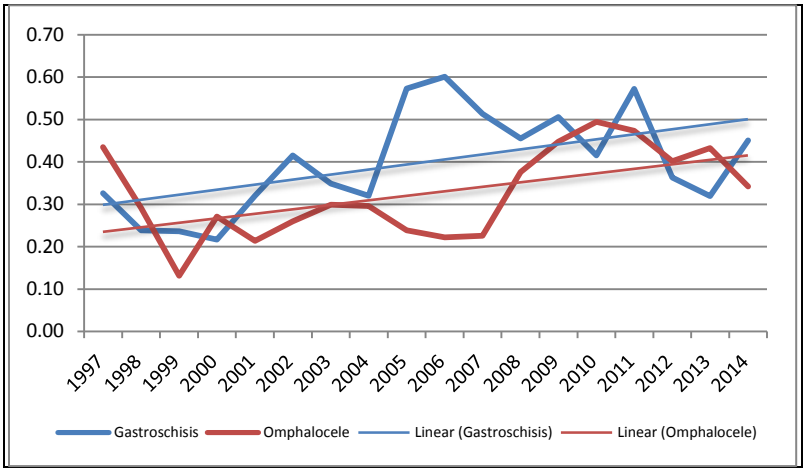
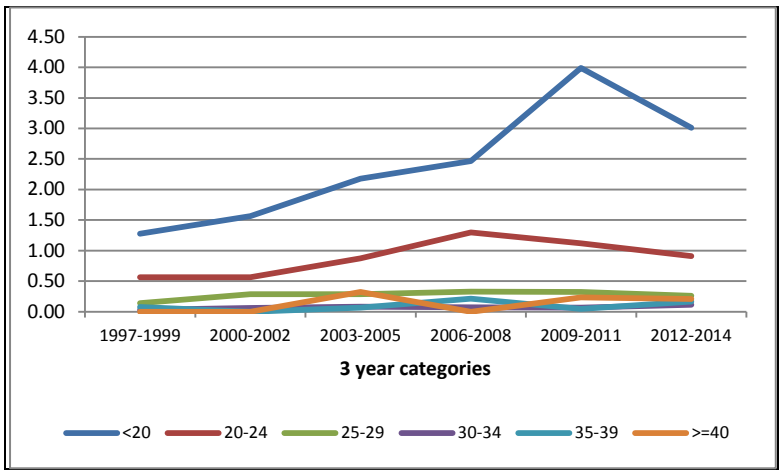
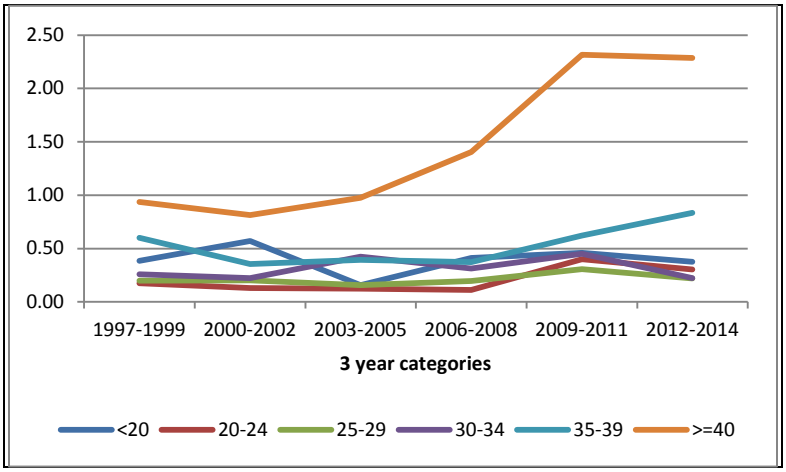


Figure 4.2.12 Gastroschisis by Maternal Age Groups - 3 Year Increments 1997–2014 (Rate per 1000 total births)



Omphalocele rates are also rising significantly but this increase is driven mainly by births to older mothers. Due to the fact that omphalocele is more associated with chromosome abnormalities than is gastroschisis, the prevalence in the older mothers likely explains the increased rate particularly in mothers 40 years of age and older (**Figure 4.2.13)**. In fact, 52% of cases of omphalocele in mothers over 40 at birth had a chromosome anomaly.

Figure 4.2.13 Omphalocele by Maternal Age Groups in 3 Year Increments 1997–2014 (Rate per 1000 total births)



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4.2.10 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=0.0090; T18 p=0.0015) (**Table 4.1.1 p.9; Figure 4.2.16**) but are strongly correlated with increasing maternal age **(Table 4.2.1 p. 23).**

In 1983, approximately four per cent of mothers were 35 years of age or over at the birth of their infant whereas in 2014, there were approximately19 per cent in the same age category (**Figure 4.2.15**). Births to women over 40 years of age have increased almost 6 times since 1983 while births to women 20-29 have steadily declined over the same time period.

Figure 4.2.14 Maternal Age at birth as a percent of total births, Alberta, 1983–2014

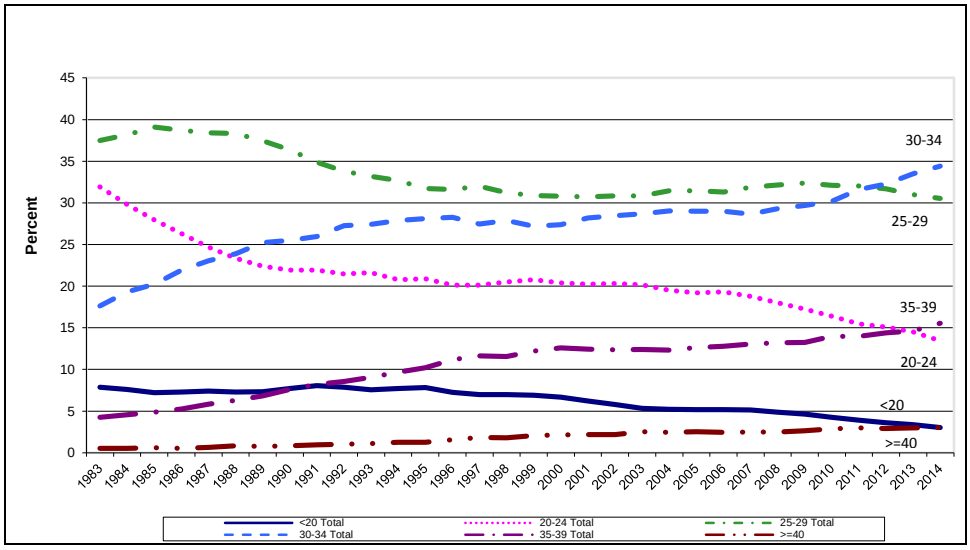


Figure 4.2.15 Maternal Age (>=35 years) at birth as a percent of total births, Alberta, 1983–2014

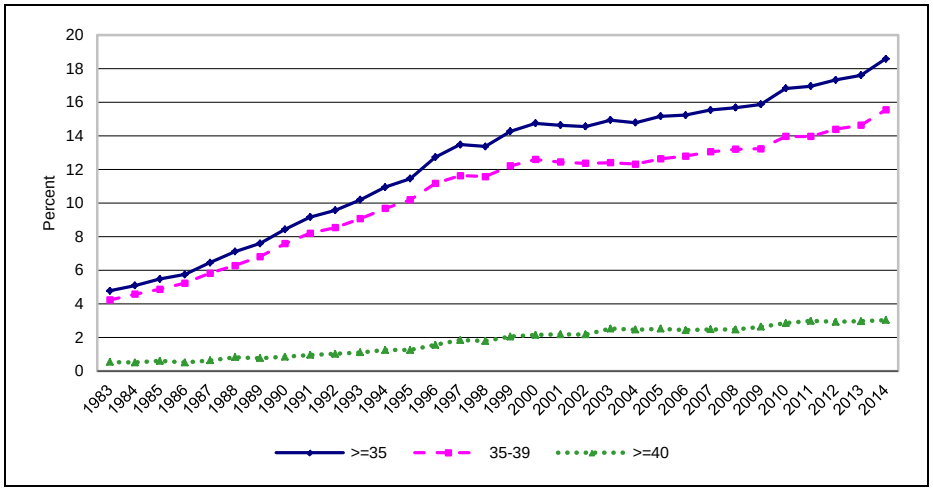


Figure 4.2.16 Chromosome Anomalies: Trisomy 13, Trisomy 18, Trisomy 21, 1997–2014 (Rate per 1000 total births)

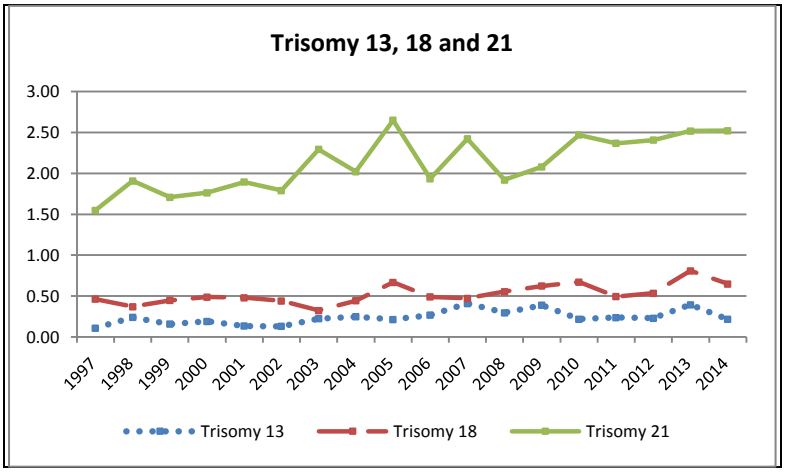


Table 4.2.1 Down Syndrome by Maternal Age, 2010-2014
Rates per 1000 Total Births (Live + Still + ToP)

Maternal Age	Year				
	2010	2011	2012	2013	2014
<20	1.39	0	0.53	1.11	0.62
20–24	0.84	1.02	1.14	1.30	0.67
25–29	1.17	0.99	0.84	0.85	1.24
30–34	2.29	1.62	2.07	1.68	2.25
35–39	4.67	5.66	6.11	6.81	4.99
≥40	19.38	19.13	13.05	15.18	15.88
All ages	2.47	2.37	2.41	2.52	2.52

Infants with Down Syndrome often have associated anomalies. As previously noted in earlier reports, ACASS does not code minor anomalies associated with Down syndrome such as single palmar crease, upslanting palpebral fissures, and increased space between the first and second toes. On the other hand, major malformations are entered routinely into the database as most live born infants with Trisomy 21 survive and require ongoing health services. Major malformations are entered into the database for Trisomies 13 and 18 as well. Although mortality is high among infants born with Trisomies 13 and 18, some infants survive to require medical care and treatment thus counting the anomalies associated with these diagnoses can help with future health care planning.

4.2.11 Limb Reductions

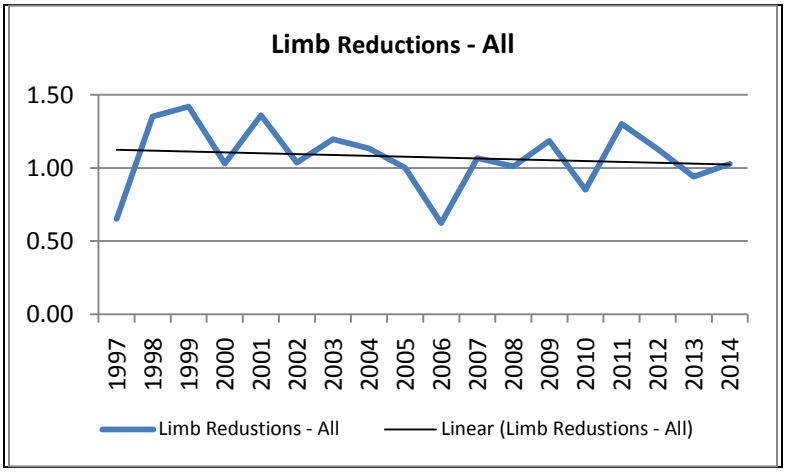
Birth prevalence rates for limb reductions have remained stable from 1997–2014 (**Figure 4.2.17**) with no significant trend evident (p=0.4237). The same holds true when upper and lower limb reduction defects are examined separately (upper – p=0.5967; lower – p=0.5271).

ACASS continues to participate in an international study, co-ordinated through the ICBDSR, on the epidemiology of very rare defects. Some of the more uncommon limb reduction defects such as true phocomelia (absence of all limb bones proximal to the hand or foot - the hand or foot attaching directly to the trunk) and amelia (complete absence of one or more limbs) are included in that study.

It appears our rates are somewhat higher than those of several countries that report to the ICBDSR **(Table 4.1.2, p. 10)**. A review of all cases in the database for 1980-2012 was undertaken and found that in fact when we look at cases as opposed to anomalies (ACASS reports anomalies), our rates are comparable with other jurisdictions (0.59/1000 total births) (Bedard et al. 2015). Problems with classification systems continue to be a factor, making comparisons with different jurisdictions difficult (Lowry et al. 2016).

Despite many studies of risk factors, the strongest evidence points to cigarette smoking (Caspers et al. 2013) but not alcohol exposure (Caspers et al. 2014).

Figure 4.2.17 Limb Reduction Defects 1997–2014 (Rate per 1000 total births)



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Caspers Conway KM, Romitti PA, Holmes L, et al. National Birth Defects Prevention Study. 2014. Maternal periconceptional alcohol consumption and congenital limb deficiencies. Birth Defects Res A Clin Mol Teratol 100:863-876.

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4.2.12 Anorectal Atresia/Stenosis

The malformations included in this category are accessed using the ICD-10 RCPCH codes Q42.0, Q42.1, Q42.2, and Q42.3 but data for fistulae or the defect level are not always provided. The current rates (1997-2014) show a significant decline (p<0.0001) (**Figure 4.2.18**) which is not only unexplained but is in complete contrast to a significant increase reported for 1980-1996 (p<0.0097) (**Figure 4.2.19**). The early period does not include terminations of pregnancy or foetal losses, whereas the later period does, and hence ascertainment should be more complete for this period.

We had investigated the increase in rates previously, which resulted in a publication (Lowry et al 2007) and showed no overall trend at that time. Our rates are very comparable to other population studies, which were summarized by Jenetzky (2007) for Texas, Hawaii, British Columbia, Bavaria, and EUROCAT, but none of these rates covers the last ten years. A ten year report (1999-2008) from EUROCAT (Loane et al 2011) showed a significantly decreasing trend in four registries, an increase in two, no change in one, and no data or too little to analyze in fourteen.

We found 24% of our cases were an isolated anomaly and the 76% associated group contained chromosomal, syndromic, or recognized conditions. These numbers are very comparable to de Blaauw et al (2013) from the European consortium on anorectal malformations. They found a 1:1 sex ratio, whereas ours shows a definite male excess 1.5:1. The latter is comparable to differential sex ratios found in other multifactorial disorders (e.g. cleft lip and palate), but some of our cases could be due to X-linked genes.

Risk factors that have been found by more than one investigator include cigarette smoking (Miller et al 2008), maternal obesity (BMI>30kg/m²) (Wijers et al 2010), family history of anorectal malformation and maternal occupational exposures such as organic solvents (van Rooij et al 2010).

A two year study in China compared a group of women taking 400 mg of folic acid daily from the time of premarital examination until the end of their first trimester with another group that did not take folic acid and showed that the rate among the supplemented group was 1.6/10,000 compared to 3.1/10,000 in the unsupplemented group(Myers et al 2001).

Figure 4.2.18 Anorectal Atresia/Stenosis
1997–2014 (Rate per 1000 total births)

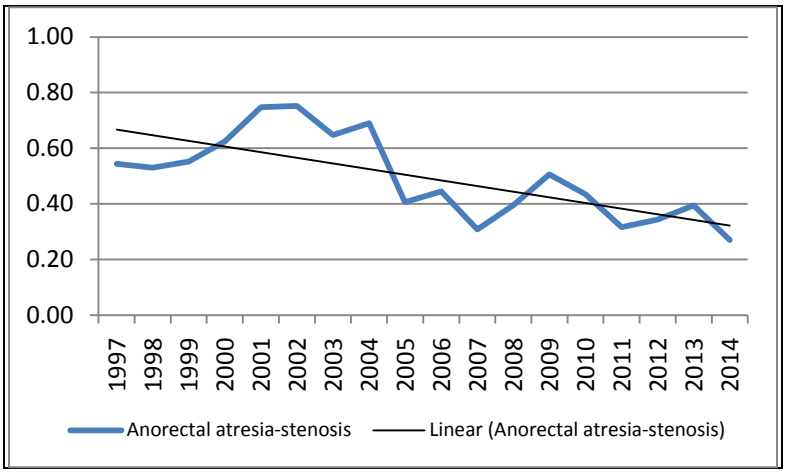
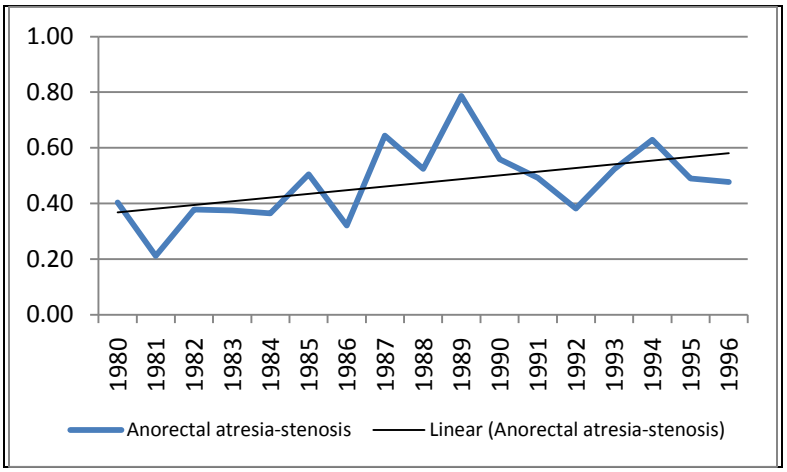


Figure 4.2.19 Anorectal Atresia/Stenosis 1980–1996 (Rate per 1000 total births)



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4.2.13 Congenital Heart Disease (CHD)

Tetralogy of Fallot (**Figure 4.2.20**) appears to be showing an increase over time (p=0.0381) although there has been a drop in rates over the last two years reported here. On the other hand, Hypoplastic Left Heart Syndrome (**Figure 4.2.21**) has remained stable between the years 1997–2014 (p=0.4096). There may be a slight upward trend for ventricular septal defects however it is barely significant (p=0.0528). In our previous report it appeared that ASD rates were declining however with the addition of data from 2012-2014, there is now no significant trend in either direction (p=0.5967). **(Figure 4.2.22)**.

Figure 4.2.20 Tetralogy of Fallot 1997–2014 (Rate per 1000 total births)

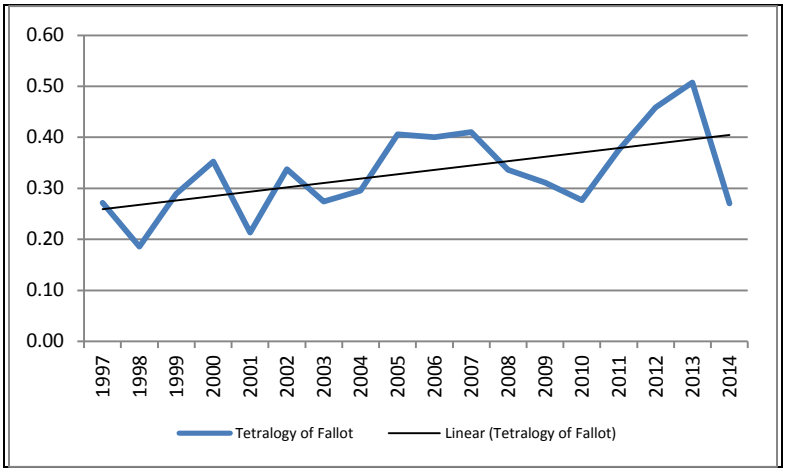


Figure 4.2.21 Hypoplastic Left Heart Syndrome 1997– 2014 (Rate per 1000 total births)

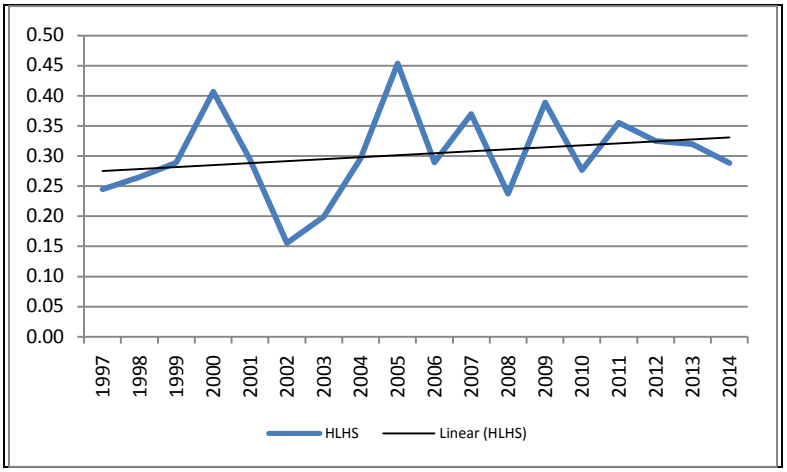
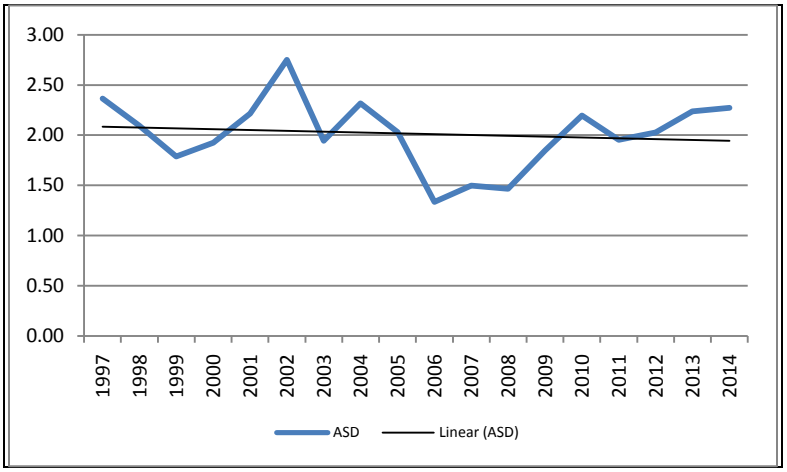


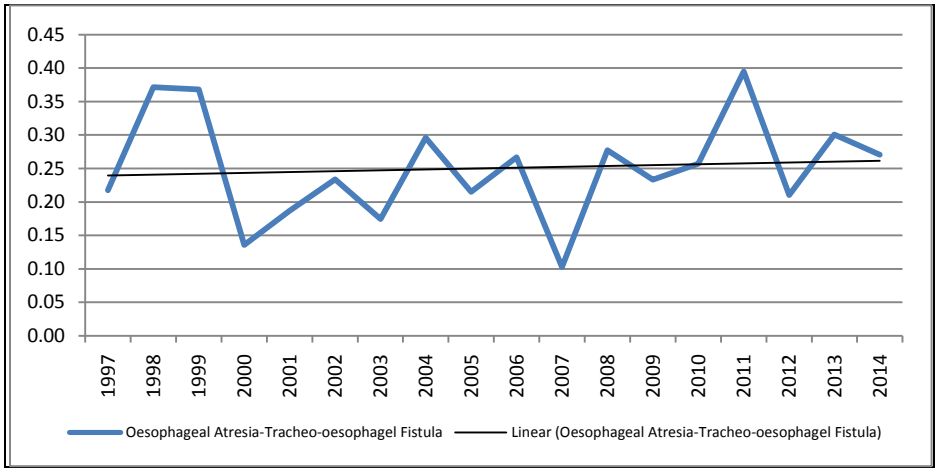
Figure 4.2.22 Atrial Septal Defect 1997– 2014 (Rate per 1000 total births)



4.2.14 Oesophageal Atresia/Stenosis

There has been no significant change in the rates of oesophageal atresia and stenosis with or without a tracheo-oesophageal fistula since 1997 (p=0.6315).

Figure 4.2.23 Oesophageal Atresia/Stenosis 1997–2014 (Rate per 1000 total births)



4.2.15 Hypospadias and Epispadias

Ascertainment of hypospadias includes all degrees of severity but excludes congenital chordee without hypospadias. Epispadias continues to be separated from hypospadias for this report. It should be noted that our rates are expressed as total male births in contrast to many rates in the literature which are cited as live births and do not differentiate the male proportion.

Hypospadias rates are increasing significantly ($p<0.0001$) and clearly 2013 and 2014 show a significant rise. The literature has recorded an increasing trend in the earlier decades (1970s, 1980s) followed by decreasing trends. A recent review (Springer et al. 2016) summarized the worldwide prevalence of hypospadias and showed a wide variation as well as conflicting data as to whether there was an increasing, decreasing or no trend. Many of the differences can be attributed to methodological issues such as ascertainment methods, definitions etc. For example, Bergman et al.'s (2015) study from 23 EUROCAT registries showed different trends with prevalence rates ranging from 3.68/1000 births in Germany-Mainz, to 0.51/1000 births in south Portugal.

A major concern is whether environmental chemical contaminants are causative factors for congenital anomalies, especially endocrine disruptive compounds and hypospadias. Two recent reviews summarized a number of the studies and pointed out methodological problems with many. For example, Foster et al. (2016) suggested that the literature did not support either a conclusion that environmental contaminants are or are not associated with increased risk for congenital anomalies in the general population. They paid particular attention to phthalates and developmental abnormalities of the male reproductive tract. Bonde et al. (2016) came to much the same conclusion. They stated that the current evidence is compatible with a small increased risk of male reproductive disorders, but that the evidence was limited.

Risk factors for hypospadias include advanced maternal age, increased BMI, in vitro fertilization and low birth weight. A novel protective factor for hypospadias is that of altitude. Fernandez et al. (2016) studied six different South American countries (Columbia, Bolivia, Brazil, Argentina, Chile, Uruguay) and showed that centres below 2000 metres had an increasing trend whereas in two centres in the Andes mountains where the altitude was 2000 metres or more, lower prevalence rates were observed. They also suggested there may be ethnic reasons for the difference, not just altitude.

In a case-control study in an area in the south of France, 300 boys with hypospadias with a negative family history and who were tested and negative for three mutations associated with hypospadias (AR, SRD5A2 and MAMLD2) were compared with 302 control boys. Fetal exposure to paint/solvents, detergents and pesticides during the time of genital differentiation was more frequent in the cases of hypospadias.

Although it appears that the trend for epispadias is increasing there has been no significant change since 1997 ($p=0.4348$) however the numbers are so few (range = 1-5 per year) that ascertaining one or two cases per year can alter the rates substantially.

Figure 4.2.24 Hypospadias 1997–2014 (Rate per 1000 total births)

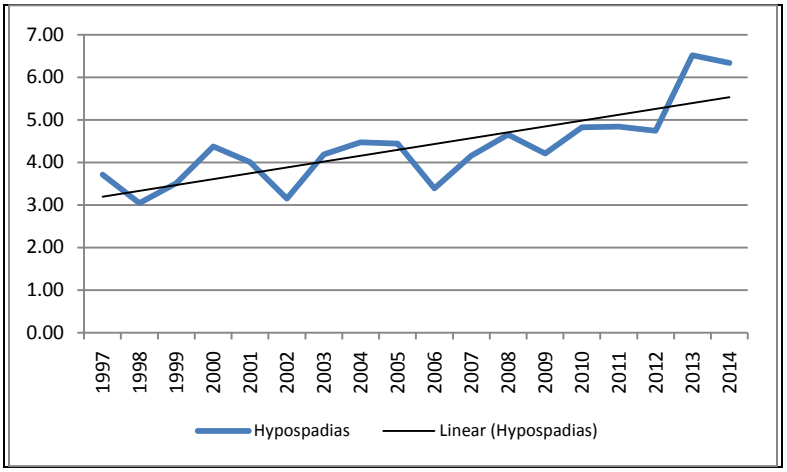


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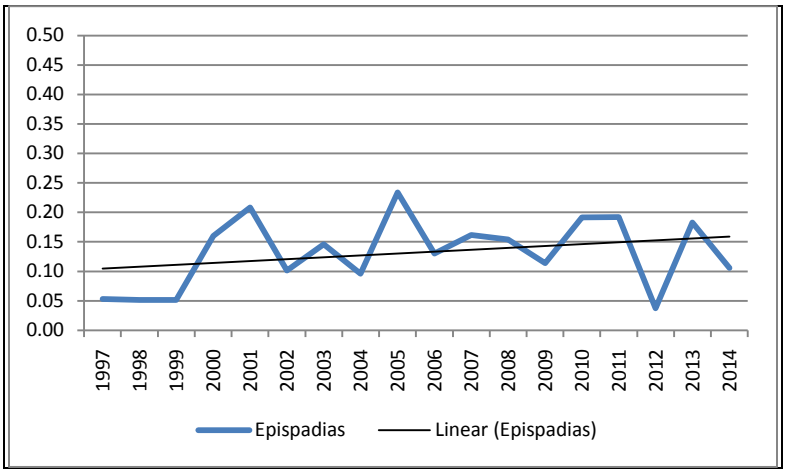
Figure 4.2.25 Epispadias 1997–2014 (Rate per 1000 total births)



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4.3 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.

5. SURVEILLANCE AND RESEARCH PROJECTS

5.1 Surveillance and Research Projects/Collaborations and Consultations/Papers

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Articles for Canadian Congenital Anomalies Surveillance System Current Contents (http://www.phac-aspc.gc.ca/ccasn-rcsac/index.html):

i. Sibbald B and Lowry RB Orofacial clefts in Alberta 1980–2004 inclusive (winter 2005) http://www.phac-aspc.gc.ca/ccasn-rcsac/ct2005/or-cl-alberta_e.html
ii. Sibbald B and Lowry RB Abdominal wall defects- Alberta 1980–2002 (winter 2004) http://www.phac-aspc.gc.ca/ccasn-rcsac/ct2004/awd-alb.html
iii. Sibbald B and Lowry RB Down Syndrome in Alberta: Alberta Congenital Anomalies Surveillance System (fall 2003) http://www.phac-aspc.gc.ca/ccasn-rcsac/ct2003/abds_e.html

Alberta Congenital Anomalies Surveillance System

6. Appendices

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

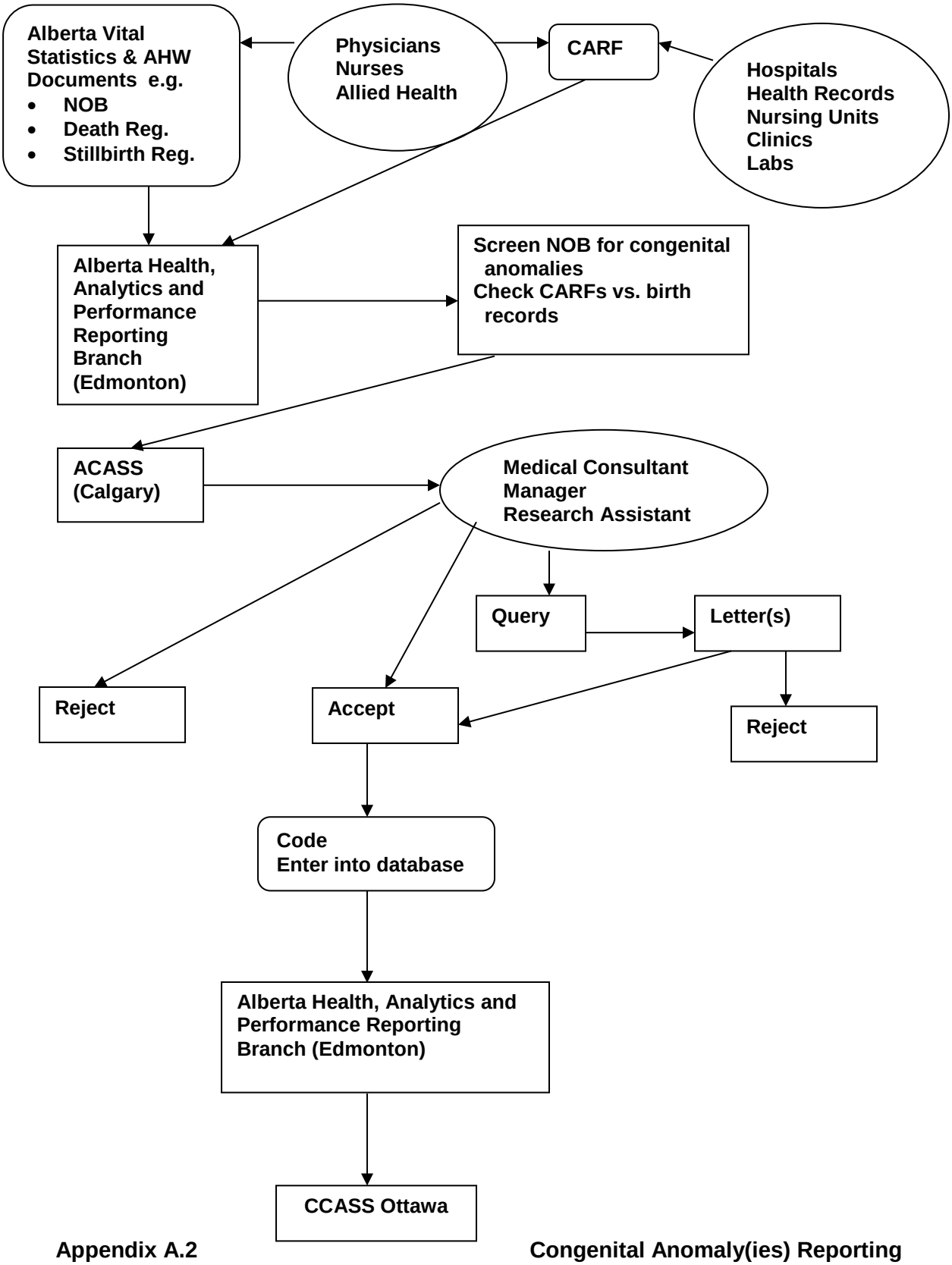
Appendix A.2 Congenital Anomaly(ies) Reporting Form (CARF)

Appendix A.3 Single and Aggregate Year Anomaly rates

Appendix A.4 Numbers of cases, anomalies and anomalies per case 1980–2011

Appendix A.5 Chi Trend table for reported anomalies 1997-2011

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

PLEASE PRINT CLEARLY

Fetus / Infant

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)* *Month by Name* Day Year	Infant's Discharge *Month by Name* Day Year
Location *(City/Town)*		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 Year Anomaly Rates per 1,000 Total Births
Total Births (Live, still and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		2004	2005	2006	2007	2008	2009	2010	2011	2012	2013	2014
Anencephaly	NUMBER	10	9	9	9	10	18	9	16	8	14	16
	RATE	**0.25**	**0.21**	**0.20**	**0.18**	**0.20**	**0.35**	**0.18**	**0.32**	**0.15**	**0.26**	**0.29**
	Lower CI	0.12	0.10	0.09	0.08	0.10	0.21	0.08	0.18	0.07	0.14	0.17
ICD-10 Q00.00, Q00.01, Q00.1	Upper CI	0.45	0.41	0.38	0.35	0.36	0.55	0.34	0.51	0.30	0.44	0.47
Spina Bifida without	NUMBER	16	12	26	20	19	22	24	20	22	19	17
Anencephaly	**RATE**	**0.39**	**0.29**	**0.58**	**0.41**	**0.38**	**0.43**	**0.47**	**0.39**	**0.42**	**0.36**	**0.31**
	Lower CI	0.23	0.15	0.38	0.25	0.23	0.27	0.30	0.24	0.26	0.22	0.18
ICD-10 Q05..	Upper CI	0.64	0.50	0.85	0.63	0.59	0.65	0.71	0.61	0.64	0.56	0.49
Encephalocele	NUMBER	3	9	2	6	7	7	7	5	6	8	4
	RATE	**0.07**	**0.21**	**0.04**	**0.12**	**0.14**	**0.14**	**0.14**	**0.10**	**0.11**	**0.15**	**0.07**
	Lower CI	0.01	0.10	0.01	0.05	0.06	0.05	0.06	0.03	0.04	0.06	0.02
ICD-10 Q01..	Upper CI	0.21	0.41	0.15	0.26	0.28	0.28	0.28	0.23	0.25	0.29	0.18
Neural Tube Defects (all)	NUMBER	29	30	37	36	36	47	40	41	36	41	37
	RATE	**0.71**	**0.72**	**0.82**	**0.74**	**0.71**	**0.91**	**0.79**	**0.81**	**0.69**	**0.77**	**0.67**
	Lower CI	0.48	0.48	0.58	0.52	0.50	0.67	0.57	0.58	0.48	0.55	0.47
ICD-10 Q00.., Q01.., Q05..	Upper CI	1.03	1.02	1.13	1.02	0.99	1.22	1.08	1.10	0.95	1.05	0.92
Hydrocephalus without Spina	NUMBER	20	23	38	27	32	29	30	39	26	17	20
Bifida	**RATE**	**0.49**	**0.55**	**0.85**	**0.55**	**0.63**	**0.56**	**0.59**	**0.77**	**0.50**	**0.32**	**0.36**
(Excludes hydranencephaly)	Lower CI	0.30	0.35	0.60	0.37	0.43	0.38	0.40	0.55	0.33	0.19	0.22
ICD-10 Q03	Upper CI	0.76	0.82	1.16	0.81	0.89	0.81	0.85	1.05	0.73	0.51	0.56
Arrhinencephaly/	NUMBER	7	7	7	13	16	16	9	11	12	18	8
Holoprosencephaly	**RATE**	**0.17**	**0.17**	**0.16**	**0.27**	**0.32**	**0.31**	**0.18**	**0.22**	**0.23**	**0.34**	**0.14**
	Lower CI	0.07	0.07	0.06	0.14	0.18	0.18	0.08	0.11	0.12	0.20	0.06
ICD-10 Q04.1, Q04.2, Q87.03	Upper CI	0.35	0.34	0.32	0.46	0.51	0.50	0.34	0.39	0.40	0.53	0.28
Microcephaly	NUMBER	13	22	16	19	20	23	22	33	20	22	16
	RATE	**0.32**	**0.53**	**0.36**	**0.39**	**0.40**	**0.45**	**0.43**	**0.65**	**0.38**	**0.41**	**0.29**
	Lower CI	0.17	0.33	0.20	0.24	0.24	0.28	0.27	0.45	0.23	0.26	0.17
ICD-10 Q02	Upper CI	0.55	0.79	0.58	0.61	0.61	0.67	0.66	0.92	0.59	0.63	0.47
Anophthalmia/microphthalmia	NUMBER	8	13	5	7	10	4	6	8	7	9	6
	RATE	**0.20**	**0.31**	**0.11**	**0.14**	**0.20**	**0.08**	**0.12**	**0.16**	**0.13**	**0.17**	**0.11**
	Lower CI	0.09	0.17	0.04	0.06	0.10	0.02	0.04	0.07	0.05	0.08	0.04
ICD-10 Q11.0, Q11.1, Q11.2	Upper CI	0.39	0.53	0.26	0.29	0.36	0.19	0.26	0.31	0.27	0.32	0.23
Congenital cataract	NUMBER	4	5	4	9	4	11	13	7	6	9	10
	RATE	**0.10**	**0.12**	**0.09**	**0.18**	**0.08**	**0.21**	**0.26**	**0.14**	**0.11**	**0.17**	**0.18**
	Lower CI	0.03	0.04	0.02	0.08	0.02	0.11	0.14	0.06	0.04	0.08	0.09
ICD-10 Q12.0	Upper CI	0.25	0.27	0.22	0.35	0.20	0.38	0.44	0.28	0.25	0.32	0.33

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
Total Births (Live, still and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		00-04 (5 years)	05-09 (5 years)	10-14 (5 years)	00-14 (15 years)	(years)	(years)
Anencephaly	NUMBER	48	55	63	166		
	RATE	**0.25**	**0.23**	**0.24**	**0.24**		
	Lower CI	0.18	0.17	0.18	0.20		
ICD-10 Q00.00, Q00.01, Q00.1	Upper CI	0.33	0.30	0.31	0.28		
Spina Bifida without Anencephaly	NUMBER	57	99	102	258		
	RATE	**0.29**	**0.42**	**0.39**	**0.37**		
	Lower CI	0.22	0.34	0.32	0.33		
ICD-10 Q05..	Upper CI	0.38	0.51	0.47	0.42		
Encephalocele	NUMBER	28	31	30	89		
	RATE	**0.14**	**0.13**	**0.11**	**0.13**		
	Lower CI	0.10	0.09	0.08	0.10		
ICD-10 Q01..	Upper CI	0.21	0.19	0.16	0.16		
Neural Tube Defects (all)	NUMBER	134	186	195	515		
	RATE	**0.69**	**0.78**	**0.74**	**0.74**		
	Lower CI	0.58	0.67	0.64	0.68		
ICD-10 Q00.., Q01.., Q05..	Upper CI	0.82	0.90	0.86	0.81		
Hydrocephalus without Spina Bifida	NUMBER	110	149	132	391		
	RATE	**0.57**	**0.63**	**0.50**	**0.56**		
(Excludes hydranencephaly)	Lower CI	0.47	0.53	0.42	0.51		
ICD-10 Q03	Upper CI	0.68	0.74	0.60	0.62		
Arrhinencephaly/ Holoprosencephaly	NUMBER	37	59	74	154		
	RATE	**0.19**	**0.25**	**0.28**	**0.22**		
	Lower CI	0.13	0.19	0.22	0.19		
ICD-10 Q04.1, Q04.2, Q87.03	Upper CI	0.26	0.32	0.35	0.26		
Microcephaly	NUMBER	76	100	113	289		
	RATE	**0.39**	**0.42**	**0.43**	**0.42**		
	Lower CI	0.31	0.34	0.36	0.37		
ICD-10 Q02	Upper CI	0.49	0.51	0.52	0.47		
Anophthalmia/microphthalmia	NUMBER	28	39	36	103		
	RATE	**0.14**	**0.16**	**0.14**	**0.15**		
	Lower CI	0.10	0.12	0.10	0.12		
ICD-10 Q11.0, Q11.1, Q11.2	Upper CI	0.21	0.22	0.19	0.18		
Congenital cataract	NUMBER	16	33	45	94		
	RATE	**0.08**	**0.14**	**0.17**	**0.14**		
	Lower CI	0.05	0.10	0.13	0.11		
ICD-10 Q12.0	Upper CI	0.13	0.20	0.23	0.17		

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
Total Births (Live, still and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		2004	2005	2006	2007	2008	2009	2010	2011	2012	2013	2014
Anotia/microtia	NUMBER	13	11	8	11	11	15	14	11	13	15	11
	RATE	**0.32**	**0.26**	**0.18**	**0.23**	**0.22**	**0.29**	**0.28**	**0.22**	**0.25**	**0.28**	**0.20**
	Lower CI	0.17	0.13	0.08	0.11	0.11	0.16	0.15	0.11	0.13	0.16	0.10
ICD-10 Q16.0, Q17.2	Upper CI	0.54	0.47	0.35	0.40	0.39	0.48	0.46	0.38	0.42	0.46	0.35
Congenital Heart Defects (all)	NUMBER	528	508	420	539	528	586	661	642	686	749	671
	RATE	**13.01**	**12.13**	**9.34**	**11.07**	**10.45**	**11.40**	**13.07**	**12.67**	**13.11**	**14.08**	**12.09**
	Lower CI	11.93	11.10	8.47	10.15	9.58	10.49	12.09	11.71	12.15	13.09	11.20
ICD-10 Q20.. – Q26..	Upper CI	14.17	13.23	10.28	12.04	11.38	12.36	14.10	13.69	14.13	15.13	13.04
Common Truncus	NUMBER	3	2	2	5	3	2	4	2	5	8	3
Excludes AP window	**RATE**	**0.07**	**0.05**	**0.04**	**0.10**	**0.06**	**0.04**	**0.08**	**0.04**	**0.10**	**0.15**	**0.05**
	Lower CI	0.01	0.01	0.01	0.03	0.01	0.00	0.02	0.00	0.03	0.06	0.01
ICD-10 Q20.0	Upper CI	0.21	0.16	0.15	0.24	0.17	0.13	0.20	0.14	0.22	0.29	0.15
Transposition of Great Arteries	NUMBER	18	19	15	13	14	13	13	20	20	23	29
	RATE	**0.44**	**0.45**	**0.33**	**0.27**	**0.28**	**0.25**	**0.26**	**0.39**	**0.38**	**0.43**	**0.52**
	Lower CI	0.26	0.27	0.19	0.14	0.15	0.13	0.14	0.24	0.23	0.27	0.35
ICD-10 Q20.11, Q20.3, Q20.5	Upper CI	0.70	0.71	0.55	0.46	0.46	0.43	0.44	0.61	0.59	0.65	0.75
Tetralogy of Fallot	NUMBER	12	17	18	20	17	16	14	19	24	27	15
(Includes Tetralogy with ASD	**RATE**	**0.30**	**0.41**	**0.40**	**0.41**	**0.34**	**0.31**	**0.28**	**0.38**	**0.46**	**0.51**	**0.27**
aka Pentalogy of Fallot)	Lower CI	0.15	0.24	0.24	0.25	0.20	0.18	0.15	0.23	0.29	0.34	0.15
ICD-10 Q21.3.., Q21.82	Upper CI	0.51	0.65	0.63	0.63	0.54	0.50	0.46	0.59	0.68	0.74	0.44
Ventricular Septal Defect	NUMBER	157	148	121	140	143	158	169	167	167	175	161
	RATE	**3.87**	**3.53**	**2.69**	**2.87**	**2.83**	**3.07**	**3.34**	**3.30**	**3.19**	**3.29**	**2.90**
	Lower CI	3.29	2.99	2.23	2.42	2.39	2.61	2.86	2.82	2.73	2.82	2.47
ICD-10 Q21.0	Upper CI	4.52	4.15	3.22	3.39	3.33	3.59	3.88	3.84	3.71	3.82	3.39
Atrial Septal Defect	NUMBER	94	85	60	73	74	95	111	99	106	119	126
	RATE	**2.32**	**2.03**	**1.33**	**1.50**	**1.46**	**1.85**	**2.19**	**1.95**	**2.03**	**2.24**	**2.27**
	Lower CI	1.87	1.62	1.02	1.18	1.15	1.50	1.81	1.59	1.66	1.85	1.89
ICD-10 Q21.1..	Upper CI	2.83	2.51	1.72	1.88	1.84	2.26	2.64	2.38	2.45	2.68	2.70
Endocardial Cushion Defect	NUMBER	27	15	13	26	25	20	32	26	29	33	33
	RATE	**0.67**	**0.36**	**0.29**	**0.53**	**0.49**	**0.39**	**0.63**	**0.51**	**0.55**	**0.62**	**0.59**
	Lower CI	0.44	0.20	0.15	0.35	0.32	0.24	0.43	0.34	0.37	0.43	0.41
ICD-10 Q21.2..	Upper CI	0.97	0.59	0.49	0.78	0.73	0.60	0.89	0.75	0.80	0.87	0.84
Pulmonary Valve Atresia And Stenosis	NUMBER	19	27	20	33	24	29	36	28	33	31	41
	RATE	**0.47**	**0.64**	**0.44**	**0.68**	**0.48**	**0.56**	**0.71**	**0.55**	**0.63**	**0.58**	**0.74**
	Lower CI	0.28	0.43	0.27	0.47	0.30	0.38	0.50	0.37	0.43	0.40	0.53
ICD-10 Q22.0, Q22.1	Upper CI	0.73	0.94	0.69	0.95	0.71	0.81	0.99	0.80	0.89	0.83	1.00

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
Total Births (Live, still and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		00-04 (5 years)	05-09 (5 years)	10-14 (5 years)	00-14 (15 years)
Anotia/microtia	NUMBER	40	56	64	160
	RATE	**0.21**	**0.24**	**0.24**	**0.23**
	Lower CI	0.15	0.18	0.19	0.20
ICD-10 Q16.0, Q17.2	Upper CI	0.28	0.31	0.31	0.27
Congenital Heart Defects (all)	NUMBER	2489	2581	3995	8479
	RATE	**12.86**	**10.87**	**15.23**	**12.23**
	Lower CI	12.36	10.45	14.77	11.97
ICD-10 Q20.., – Q26..	Upper CI	13.38	11.30	15.71	12.49
Common Truncus	NUMBER	14	14	22	50
Excludes AP window	**RATE**	**0.07**	**0.06**	**0.08**	**0.07**
	Lower CI	0.04	0.03	0.05	0.05
ICD-10 Q20.0	Upper CI	0.12	0.10	0.13	0.10
Transposition of Great Arteries	NUMBER	78	74	105	257
	RATE	**0.40**	**0.31**	**0.40**	**0.37**
	Lower CI	0.32	0.24	0.33	0.33
ICD-10 Q20.11, Q20.3, Q20.5	Upper CI	0.50	0.39	0.48	0.42
Tetralogy of Fallot	NUMBER	57	88	99	244
(Includes Tetralogy with ASD aka	**RATE**	**0.29**	**0.37**	**0.38**	**0.35**
Pentalogy of Fallot)	Lower CI	0.22	0.30	0.31	0.31
ICD-10 Q21.3.., Q21.82	Upper CI	0.38	0.46	0.46	0.40
Ventricular Septal Defect	NUMBER	635	710	839	2184
	RATE	**3.28**	**2.99**	**3.20**	**3.15**
	Lower CI	3.03	2.77	2.99	3.02
ICD-10 Q21.0	Upper CI	3.55	3.22	3.42	3.29
Atrial Septal Defect	NUMBER	432	387	561	1380
	RATE	**2.23**	**1.63**	**2.14**	**1.99**
	Lower CI	2.03	1.47	1.97	1.89
ICD-10 Q21.1..	Upper CI	2.45	1.80	2.32	2.10
Endocardial Cushion Defect	NUMBER	100	99	153	352
	RATE	**0.52**	**0.42**	**0.58**	**0.51**
	Lower CI	0.42	0.34	0.49	0.46
ICD-10 Q21.2..	Upper CI	0.63	0.51	0.68	0.56
Pulmonary Valve Atresia and Stenosis	NUMBER	124	133	169	426
	RATE	**0.64**	**0.56**	**0.64**	**0.61**
	Lower CI	0.53	0.47	0.55	0.56
ICD-10 Q22.0, Q22.1	Upper CI	0.76	0.66	0.75	0.68

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
Total Births (Live, still and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		2004	2005	2006	2007	2008	2009	2010	2011	2012	2013	2014
Tricuspid Valve Atresia and Stenosis	NUMBER	3	1	4	7	4	5	8	2	3	3	8
	RATE	**0.07**	**0.02**	**0.09**	**0.14**	**0.08**	**0.10**	**0.16**	**0.04**	**0.06**	**0.06**	**0.11**
	Lower CI	0.01	0.00	0.02	0.06	0.02	0.03	0.07	0.00	0.01	0.01	0.04
ICD-10 Q22.4	Upper CI	0.21	0.12	0.22	0.29	0.20	0.22	0.31	0.14	0.16	0.16	0.23
Ebstein's Anomaly	NUMBER	1	1	4	2	3	3	4	3	3	8	8
	RATE	**0.02**	**0.02**	**0.09**	**0.04**	**0.06**	**0.06**	**0.08**	**0.06**	**0.06**	**0.15**	**0.14**
	Lower CI	0.00	0.00	0.02	0.00	0.01	0.01	0.02	0.01	0.01	0.06	0.06
ICD-10 Q22.5	Upper CI	0.12	0.12	0.22	0.14	0.17	0.17	0.20	0.17	0.16	0.29	0.28
Aortic Valve Atresia/Stenosis	NUMBER	11	4	4	11	7	6	11	5	9	10	12
(excludes sub& supra aortic stenosis and	**RATE**	**0.27**	**0.10**	**0.09**	**0.23**	**0.14**	**0.12**	**0.22**	**0.10**	**0.17**	**0.19**	**0.22**
Aortic stenosis found with HLHS)	Lower CI	0.14	0.03	0.02	0.11	0.06	0.04	0.11	0.03	0.08	0.09	0.11
ICD-10 Q23.0	Upper CI	0.48	0.24	0.22	0.40	0.28	0.25	0.39	0.23	0.32	0.34	0.38
Hypoplastic Left Heart Syndrome (HLHS)	NUMBER	12	19	13	18	12	20	14	18	17	17	16
	RATE	**0.30**	**0.45**	**0.29**	**0.37**	**0.24**	**0.39**	**0.28**	**0.36**	**0.32**	**0.32**	**0.29**
	Lower CI	0.15	0.27	0.15	0.22	0.12	0.24	0.15	0.21	0.19	0.19	0.17
ICD-10 Q23.4	Upper CI	0.51	0.71	0.49	0.58	0.41	0.60	0.46	0.56	0.52	0.51	0.47
Coarctation of the Aorta	NUMBER	13	16	13	19	30	21	26	23	27	25	21
	RATE	**0.32**	**0.38**	**0.29**	**0.39**	**0.59**	**0.41**	**0.51**	**0.45**	**0.52**	**0.47**	**0.38**
	Lower CI	0.17	0.22	0.15	0.24	0.40	0.25	0.34	0.29	0.34	0.30	0.23
ICD-10 Q25.1..	Upper CI	0.55	0.62	0.49	0.61	0.85	0.62	0.75	0.68	0.75	0.69	0.58
Cleft Palate without Cleft Lip	NUMBER	34	30	27	27	40	34	39	36	41	46	37
(i.e. cleft palate alone)	**RATE**	**0.84**	**0.72**	**0.60**	**0.55**	**0.79**	**0.66**	**0.77**	**0.71**	**0.78**	**0.86**	**0.67**
	Lower CI	0.58	0.48	0.40	0.37	0.57	0.46	0.55	0.50	0.56	0.63	0.47
ICD-10 Q35..	Upper CI	1.17	1.02	0.87	0.80	1.08	0.92	1.05	0.98	1.06	1.15	0.92
Cleft Lip without Cleft Palate	NUMBER	14	15	22	27	32	26	17	21	24	24	27
(i.e. cleft lip alone)	**RATE**	**0.35**	**0.36**	**0.49**	**0.55**	**0.63**	**0.51**	**0.34**	**0.41**	**0.46**	**0.45**	**0.49**
	Lower CI	0.19	0.20	0.31	0.37	0.43	0.33	0.20	0.26	0.29	0.29	0.32
ICD-10 Q36..	Upper CI	0.58	0.59	0.74	0.81	0.89	0.74	0.54	0.63	0.68	0.67	0.71
Cleft Lip and Cleft Palate	NUMBER	36	32	23	55	35	45	54	39	31	35	44
	RATE	**0.89**	**0.76**	**0.51**	**1.13**	**0.69**	**0.88**	**1.07**	**0.77**	**0.59**	**0.66**	**0.79**
	Lower CI	0.62	0.52	0.32	0.85	0.48	0.64	0.80	0.55	0.40	0.46	0.58
ICD-10 Q37..	Upper CI	1.23	1.08	0.77	1.47	0.96	1.17	1.39	1.05	0.84	0.92	1.07
Cleft Lip with and without Cleft Palate	NUMBER	50	47	45	82	67	71	71	60	55	59	71
	RATE	**1.23**	**1.12**	**1.00**	**1.68**	**1.33**	**1.38**	**1.40**	**1.18**	**1.05**	**1.11**	**1.28**
	Lower CI	0.92	0.83	0.73	1.34	1.03	1.08	1.10	0.90	0.79	0.85	1.00
ICD-10 Q36.., Q37..	Upper CI	1.62	1.49	1.34	2.09	1.68	1.74	1.77	1.52	1.37	1.43	1.61

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
Total Births (Live, still and fetal losses*)

Diagnostic Category and ICD-10 RCPCH Code		00-04 (5 years)	05-09 (5 years)	10-14 (5 years)	00-14 (15 years)
Tricuspid Valve Atresia and Stenosis	NUMBER	13	21	22	56
	RATE	**0.07**	**0.09**	**0.08**	**0.08**
	Lower CI	0.04	0.05	0.05	0.05
ICD-10 Q22.4	Upper CI	0.11	0.14	0.13	0.13
Ebstein's Anomaly	NUMBER	14	13	26	53
	RATE	**0.07**	**0.05**	**0.10**	**0.08**
	Lower CI	0.04	0.03	0.06	0.06
ICD-10 Q22.5	Upper CI	0.12	0.09	0.15	0.10
Aortic Valve Atresia/Stenosis	NUMBER	50	32	47	129
(excludes sub& supra aortic stenosis and	**RATE**	**0.26**	**0.13**	**0.18**	**0.19**
Aortic stenosis found with HLHS)	Lower CI	0.19	0.09	0.13	0.16
ICD-10 Q23.0	Upper CI	0.34	0.19	0.24	0.22
Hypoplastic Left Heart Syndrome	NUMBER	52	82	82	216
	RATE	**0.27**	**0.35**	**0.31**	**0.31**
	Lower CI	0.20	0.27	0.25	0.27
ICD-10 Q23.4	Upper CI	0.35	0.43	0.39	0.36
Coarctation of the Aorta	NUMBER	63	99	122	284
	RATE	**0.33**	**0.42**	**0.47**	**0.41**
	Lower CI	0.25	0.34	0.39	0.36
ICD-10 Q25.1..	Upper CI	0.42	0.51	0.56	0.46
Cleft Palate without Cleft Lip	NUMBER	159	158	199	516
(i.e. cleft palate alone)	**RATE**	**0.82**	**0.67**	**0.76**	**0.74**
	Lower CI	0.70	0.57	0.66	0.68
ICD-10 Q35..	Upper CI	0.96	0.78	0.87	0.81
Cleft Lip without Cleft Palate	NUMBER	82	122	113	317
(i.e. cleft lip alone)	**RATE**	**0.42**	**0.51**	**0.43**	**0.46**
	Lower CI	0.34	0.43	0.36	0.41
ICD-10 Q35..	Upper CI	0.53	0.61	0.52	0.51
Cleft Lip and Cleft Palate	NUMBER	161	190	203	554
	RATE	**0.83**	**0.80**	**0.77**	**0.80**
	Lower CI	0.71	0.69	0.67	0.73
ICD-10 Q37..	Upper CI	0.97	0.92	0.89	0.87
Cleft Lip with and without Cleft Palate	NUMBER	243	312	316	871
	RATE	**1.26**	**1.31**	**1.20**	**1.26**
	Lower CI	1.10	1.17	1.08	1.17
ICD-10 Q36.., Q37..	Upper CI	1.42	1.47	1.35	1.34

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
Total Births (Live, still and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		2004	2005	2006	2007	2008	2009	2010	2011	2012	2013	2014
Choanal Atresia/Stenosis	NUMBER	9	7	8	5	6	9	8	1	10	8	12
	RATE	**0.22**	**0.17**	**0.18**	**0.10**	**0.12**	**0.18**	**0.16**	**0.02**	**0.19**	**0.15**	**0.22**
	Lower CI	0.10	0.07	0.08	0.03	0.04	0.08	0.07	0.00	0.09	0.06	0.11
ICD-10 Q30.0..	Upper CI	0.42	0.34	0.35	0.24	0.26	0.33	0.31	0.10	0.35	0.29	0.38
Oesophageal Atresia/	NUMBER	12	9	12	5	14	12	13	20	11	16	15
Tracheo-oesphageal Fistula	**RATE**	**0.30**	**0.21**	**0.27**	**0.10**	**0.28**	**0.23**	**0.26**	**0.39**	**0.21**	**0.30**	**0.27**
	Lower CI	0.15	0.10	0.14	0.03	0.15	0.12	0.14	0.24	0.11	0.17	0.15
ICD-10 Q39.0 – Q39.4	Upper CI	0.51	0.41	0.46	0.24	0.46	0.41	0.44	0.61	0.37	0.49	0.44
Pyloric Stenosis	NUMBER	33	42	43	51	57	53	44	44	51	33	49
	RATE	**0.81**	**1.00**	**0.96**	**1.05**	**1.13**	**1.03**	**0.87**	**0.87**	**0.97**	**0.62**	**0.88**
	Lower CI	0.56	0.72	0.69	0.78	0.86	0.77	0.63	0.63	0.73	0.43	0.65
ICD-10 Q40.0	Upper CI	1.14	1.36	1.29	1.38	1.46	1.35	1.17	1.17	1.28	0.87	1.17
Small Intestinal	NUMBER	13	8	15	11	24	14	18	22	24	18	22
Atresia/Stenosis (all)	**RATE**	**0.32**	**0.19**	**0.33**	**0.23**	**0.48**	**0.27**	**0.36**	**0.43**	**0.46**	**0.34**	**0.40**
	Lower CI	0.17	0.08	0.19	0.11	0.30	0.15	0.21	0.27	0.29	0.20	0.25
ICD-10 Q41…	Upper CI	0.55	0.37	0.55	0.40	0.71	0.46	0.56	0.66	0.68	0.53	0.60
Duodenal Atresia/Stenosis	NUMBER	6	5	6	6	16	3	11	14	15	9	14
	RATE	**0.15**	**0.12**	**0.13**	**0.12**	**0.32**	**0.06**	**0.22**	**0.28**	**0.29**	**0.17**	**0.25**
	Lower CI	0.05	0.04	0.05	0.05	0.18	0.01	0.11	0.15	0.16	0.08	0.14
ICD-10 Q41.0…	Upper CI	0.32	0.27	0.29	0.26	0.51	0.17	0.39	0.46	0.47	0.32	0.42
Rectal and Large Intestinal	NUMBER	34	21	24	15	22	31	24	18	20	22	18
Atresia/Stenosis (all)	**RATE**	**0.84**	**0.50**	**0.53**	**0.31**	**0.44**	**0.60**	**0.47**	**0.36**	**0.38**	**0.41**	**0.32**
	Lower CI	0.58	0.31	0.34	0.17	0.27	0.41	0.30	0.21	0.23	0.26	0.19
ICD-10 Q42..	Upper CI	1.17	0.77	0.79	0.51	0.66	0.86	0.71	0.56	0.59	0.63	0.51
Rectal Atresia/Stenosis	NUMBER	4	0	4	1	1	1	0	1	2	2	1
	RATE	**0.10**	**0**	**0.09**	**0.02**	**0.02**	**0.02**	**0**	**0.02**	**0.04**	**0.04**	**0.02**
	Lower CI	0.03		0.02	0.00	0.00	0.00		0.00	0.00	0.00	0.00
ICD-10 Q42.0…, Q42.1…	Upper CI	0.25		0.22	0.10	0.10	0.10		0.10	0.13	0.13	0.09
Anal Atresia/Stenosis	NUMBER	24	17	16	14	19	25	22	15	16	19	14
	RATE	**0.59**	**0.41**	**0.36**	**0.29**	**0.38**	**0.49**	**0.43**	**0.30**	**0.31**	**0.36**	**0.25**
	Lower CI	0.38	0.24	0.20	0.16	0.23	0.32	0.27	0.17	0.18	0.22	0.14
ICD-10 Q42.2…, Q42.3…	Upper CI	0.88	0.65	0.58	0.48	0.59	0.72	0.66	0.49	0.50	0.56	0.42
Other Large Intestinal	NUMBER	6	4	4	0	2	5	2	2	2	1	3
Atresia/Stenosis	**RATE**	**0.15**	**0.10**	**0.09**	**0**	**0.04**	**0.10**	**0.04**	**0.04**	**0.04**	**0.02**	**0.05**
	Lower CI	0.05	0.03	0.02		0.00	0.03	0.00	0.00	0.00	0.00	0.01
ICD-10 Q42.8…, Q42.9…	Upper CI	0.32	0.24	0.22		0.14	0.22	0.14	0.14	0.13	0.10	0.15

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
Total Births (Live, still and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		00-04 (5 years)	05-09 (5 years)	10-14 (5 years)	00-14 (15 years)
Choanal Atresia/Stenosis	NUMBER	41	35	39	115
	RATE	**0.21**	**0.15**	**0.15**	**0.17**
	Lower CI	0.15	0.10	0.11	0.12
ICD-10 Q30.0..	Upper CI	0.29	0.21	0.20	0.19
Oesophageal Atresia/	NUMBER	40	52	75	167
Tracheo-oesphageal Fistula	**RATE**	**0.21**	**0.22**	**0.29**	**0.24**
	Lower CI	0.15	0.16	0.23	0.21
ICD-10 Q39.0 – Q39.4	Upper CI	0.28	0.29	0.36	0.28
Pyloric Stenosis	NUMBER	179	246	221	646
	RATE	**0.92**	**1.04**	**0.84**	**0.93**
	Lower CI	0.79	0.91	0.74	0.86
ICD-10 Q40.0	Upper CI	1.07	1.17	0.96	1.01
Small Intestinal Atresia/	NUMBER	66	72	104	242
Stenosis (all)	**RATE**	**0.34**	**0.30**	**0.40**	**0.35**
	Lower CI	0.26	0.24	0.32	0.31
ICD-10 Q41…	Upper CI	0.43	0.38	0.48	0.40
Duodenal Atresia/Stenosis	NUMBER	34	36	63	133
	RATE	**0.18**	**0.15**	**0.24**	**0.19**
	Lower CI	0.12	0.11	0.18	0.16
ICD-10 Q41.0…	Upper CI	0.25	0.21	0.31	0.23
Rectal and Large Intestinal	NUMBER	146	113	102	361
Atresia/Stenosis (all)	**RATE**	**0.75**	**0.48**	**0.39**	**0.52**
	Lower CI	0.64	0.39	0.32	0.47
ICD-10 Q42..	Upper CI	0.89	0.57	0.47	0.58
Rectal Atresia/Stenosis	NUMBER	14	7	6	27
	RATE	**0.07**	**0.03**	**0.02**	**0.04**
	Lower CI	0.04	0.01	0.01	0.03
ICD-10 Q42.0…., Q42.1….	Upper CI	0.12	0.06	0.05	0.06
Anal Atresia/Stenosis	NUMBER	120	91	86	297
	RATE	**0.62**	**0.38**	**0.33**	**0.43**
	Lower CI	0.51	0.31	0.26	0.38
ICD-10 Q42.2…., Q42.3….	Upper CI	0.74	0.47	0.40	0.48
Other Large Intestinal	NUMBER	12	15	10	37
Atresia/Stenosis	**RATE**	**0.06**	**0.06**	**0.04**	**0.05**
	Lower CI	0.03	0.04	0.02	0.04
ICD-10 Q42.8…., Q42.9….	Upper CI	0.11	0.10	0.07	0.07

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Alberta Congenital Anomalies Surveillance System
RCPCH version ICD-10 Q Chapter (Q00-Q99)
Single Year Anomaly Rates per 1,000 Total Births
Total Births (Live, still and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		2004	2005	2006	2007	2008	2009	2010	2011	2012	2013	2014
Hirschsprung Disease	NUMBER	5	5	12	6	6	8	10	7	9	13	6
	RATE	**0.12**	**0.12**	**0.27**	**0.12**	**0.12**	**0.16**	**0.20**	**0.14**	**0.17**	**0.24**	**0.11**
	Lower CI	0.04	0.04	0.14	0.05	0.04	0.07	0.09	0.06	0.08	0.13	0.04
ICD-10 Q43.1..	Upper CI	0.28	0.27	0.46	0.26	0.26	0.30	0.36	0.28	0.32	0.42	0.23
Biliary Atresia	NUMBER	5	5	2	3	2	5	1	5	3	4	4
	RATE	**0.12**	**0.12**	**0.04**	**0.06**	**0.04**	**0.10**	**0.02**	**0.10**	**0.06**	**0.08**	**0.07**
	Lower CI	0.04	0.04	0.01	0.01	0.00	0.03	0.00	0.03	0.01	0.02	0.02
ICD-10 Q44.2	Upper CI	0.28	0.27	0.15	0.17	0.14	0.22	0.10	0.23	0.16	0.19	0.18
Undescended Testes	NUMBER	112	95	114	131	123	165	133	136	158	145	146
(denominator MALE births only)	**RATE**	**5.38**	**4.44**	**4.95**	**5.29**	**4.73**	**6.25**	**5.09**	**5.22**	**5.86**	**5.31**	**5.14**
(>36 weeks gestation)	Lower CI	4.43	3.59	4.09	4.42	3.93	5.34	4.27	4.39	4.98	4.48	4.34
ICD-10 Q53…	Upper CI	6.47	5.42	5.95	6.28	5.64	7.28	6.03	6.18	6.84	6.24	6.04
Hypospadias	NUMBER	93	95	78	103	121	111	126	126	128	178	180
(denominator MALE births only)	**RATE**	**4.47**	**4.44**	**3.39**	**4.16**	**4.65**	**4.21**	**4.82**	**4.84**	**4.74**	**6.51**	**6.34**
	Lower CI	3.61	3.59	2.68	3.40	3.86	3.46	4.02	4.03	3.96	5.59	5.45
ICD-10 Q54 (excl. Q54.4)	Upper CI	5.47	5.42	4.23	5.04	5.56	5.07	5.74	5.76	5.64	7.54	7.33
Epispadias	NUMBER	2	5	3	4	4	3	5	5	1	5	3
(denominator MALE births only)	**RATE**	**0.10**	**0.23**	**0.13**	**0.16**	**0.15**	**0.11**	**0.19**	**0.19**	**0.04**	**0.18**	**0.11**
	Lower CI	0.01	0.08	0.03	0.04	0.04	0.02	0.06	0.06	0.00	0.06	0.02
ICD-10 Q64.0	Upper CI	0.33	0.54	0.37	0.40	0.39	0.32	0.44	0.44	0.19	0.42	0.30
Renal Agenesis/Hypoplasia	NUMBER	23	20	20	27	39	28	27	35	35	35	28
	RATE	**0.57**	**0.48**	**0.44**	**0.55**	**0.77**	**0.54**	**0.53**	**0.69**	**0.67**	**0.66**	**0.50**
	Lower CI	0.36	0.29	0.27	0.37	0.55	0.36	0.35	0.48	0.47	0.46	0.34
ICD-10 Q60..	Upper CI	0.85	0.74	0.69	0.81	1.06	0.79	0.78	0.96	0.93	0.92	0.73
Cystic Kidney	NUMBER	32	39	27	36	30	37	44	35	44	37	35
(exclude single renal cyst	**RATE**	**0.79**	**0.93**	**0.60**	**0.74**	**0.59**	**0.72**	**0.87**	**0.69**	**0.84**	**0.70**	**0.63**
Q61.0)	Lower CI	0.54	0.66	0.40	0.52	0.40	0.51	0.63	0.48	0.61	0.49	0.44
Q61..	Upper CI	1.11	1.27	0.87	1.02	0.85	0.99	1.17	0.96	1.13	0.96	0.88
Bladder Exstrophy	NUMBER	0	1	1	2	1	1	3	0	1	1	2
	RATE	**0.00**	**0.02**	**0.02**	**0.04**	**0.02**	**0.02**	**0.06**	**0.00**	**0.02**	**0.02**	**0.04**
	Lower CI		0.00	0.00	0.00	0.00	0.00	0.01		0.00	0.00	0.00
ICD-10 Q64.1 (excl. Q64.10)	Upper CI		0.12	0.11	0.14	0.10	0.10	0.17		0.10	0.10	0.12
Obstructive Genitourinary	NUMBER	101	98	102	117	138	133	156	143	166	139	173
Defects (All)	**RATE**	**2.49**	**2.34**	**2.27**	**2.40**	**2.73**	**2.59**	**3.08**	**2.82**	**3.17**	**2.61**	**3.12**
ICD-10 Q62.0 – Q62.3, Q64.2,	Lower CI	2.03	1.90	1.85	1.99	2.30	2.17	2.62	2.38	2.71	2.20	2.67
Q64.3	Upper CI	3.02	2.85	2.75	2.88	3.23	3.07	3.61	3.32	3.69	3.09	3.62

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Alberta Congenital Anomalies Surveillance System
RCPCH version ICD-10 Q Chapter (Q00-Q99)
Aggregate Year Anomaly Rates per 1,000 Total Births
Total Births (Live, still and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		00-04 (5 years)	05-09 (5 years)	10-14 (5 years)	00-14 (15 years)
Hirschsprung Disease	NUMBER	23	37	45	105
	RATE	**0.12**	**0.16**	**0.17**	**0.15**
	Lower CI	0.08	0.11	0.13	0.12
ICD-10 Q43.1..	Upper CI	0.18	0.21	0.23	0.18
Biliary Atresia	NUMBER	12	17	17	46
	RATE	**0.06**	**0.07**	**0.06**	**0.07**
	Lower CI	0.03	0.04	0.04	0.05
ICD-10 Q44.2	Upper CI	0.11	0.11	0.10	0.09
Undescended Testes	NUMBER	498	628	718	1844
(denominator MALE births only)	**RATE**	**5.03**	**5.17**	**5.32**	**5.19**
(>36 weeks gestation)	Lower CI	4.60	4.77	4.94	4.95
ICD-10 Q53….	Upper CI	5.49	5.59	5.73	5.43
Hypospadias	NUMBER	400	508	738	1646
(denominator MALE births only)	**RATE**	**4.04**	**4.18**	**5.47**	**4.63**
	Lower CI	3.65	3.82	5.08	4.41
ICD-10 Q54 (excl. Q54.4)	Upper CI	4.45	4.56	5.88	4.86
Epispadias	NUMBER	14	19	19	52
(denominator MALE births only)	**RATE**	**0.14**	**0.16**	**0.14**	**0.15**
	Lower CI	0.08	0.09	0.08	0.11
ICD-10 Q64.0	Upper CI	0.24	0.24	0.22	0.19
Renal Agenesis/Hypoplasia	NUMBER	116	134	160	410
	RATE	**0.60**	**0.56**	**0.61**	**0.59**
	Lower CI	0.50	0.47	0.52	0.54
ICD-10 Q60..	Upper CI	0.72	0.67	0.71	0.65
Cystic Kidney	NUMBER	156	169	195	520
(excludes single renal cyst Q61.0)	**RATE**	**0.81**	**0.71**	**0.74**	**0.75**
	Lower CI	0.68	0.61	0.64	0.69
ICD-10 Q61..	Upper CI	0.94	0.83	0.86	0.82
Bladder Exstrophy	NUMBER	9	6	7	22
	RATE	**0.05**	**0.03**	**0.03**	**0.03**
	Lower CI	0.02	0.01	0.01	0.02
ICD-10 Q64.1 (excl. Q64.10)	Upper CI	0.09	0.05	0.05	0.05
Obstructive Genitourinary	NUMBER	469	588	777	1834
Defects (All)	**RATE**	**2.42**	**2.48**	**2.96**	**2.65**
	Lower CI	2.21	2.28	2.76	2.53
ICD-10 Q62.0 – Q62.3, Q64.2, Q64.3	Upper CI	2.65	2.68	3.18	2.77

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Alberta Congenital Anomalies Surveillance System
RCPCH version ICD-10 Q Chapter (Q00-Q99)
Single Year Anomaly Rates per 1,000 Total Births
Total Births (Live, still and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		2004	2005	2006	2007	2008	2009	2010	2011	2012	2013	2014
Hydronephrosis	NUMBER	63	62	62	79	90	95	104	95	111	98	99
	RATE	**1.55**	**1.48**	**1.38**	**1.62**	**1.78**	**1.85**	**2.06**	**1.88**	**2.12**	**1.84**	**1.78**
	Lower CI	1.19	1.14	1.06	1.28	1.43	1.50	1.68	1.52	1.75	1.50	1.45
ICD-10 Q62.0..	Upper CI	1.99	1.90	1.77	2.02	2.19	2.26	2.49	2.29	2.56	2.25	2.17
Pelviureteric Junction Obstruction	NUMBER	12	8	7	9	12	9	7	11	10	9	12
	RATE	**0.30**	**0.19**	**0.16**	**0.18**	**0.24**	**0.18**	**0.14**	**0.22**	**0.19**	**0.17**	**0.22**
	Lower CI	0.15	0.08	0.06	0.08	0.12	0.08	0.06	0.11	0.09	0.08	0.11
ICD-10 Q62.10 & Q62.11	Upper CI	0.51	0.37	0.32	0.35	0.41	0.33	0.28	0.39	0.35	0.32	0.38
Vesicoureteric Junction Obstruction	NUMBER	3	1	2	2	4	4	0	4	2	2	0
	RATE	**0.07**	**0.02**	**0.04**	**0.04**	**0.08**	**0.08**	**0**	**0.08**	**0.04**	**0.04**	**0**
	Lower CI	0.01	0.00	0.01	0.00	0.02	0.02		0.02	0.00	0.00	
ICD-10 Q62.12 & Q62.13	Upper CI	0.21	0.12	0.15	0.14	0.20	0.19		0.20	0.13	0.13	
Posterior Urethral Valves	NUMBER	3	4	3	6	7	2	5	3	3	4	9
	RATE	**0.07**	**0.10**	**0.07**	**0.12**	**0.14**	**0.04**	**0.10**	**0.06**	**0.06**	**0.08**	**0.16**
	Lower CI	0.01	0.03	0.01	0.05	0.06	0.00	0.03	0.01	0.01	0.02	0.07
ICD-10 Q64.20	Upper CI	0.21	0.24	0.19	0.26	0.28	0.13	0.23	0.17	0.16	0.19	0.31
Congenital Deformities Hip (All)	NUMBER	90	63	80	96	122	108	119	103	125	95	75
	RATE	**2.22**	**1.50**	**1.78**	**1.97**	**2.42**	**2.10**	**2.35**	**2.03**	**2.39**	**1.79**	**1.35**
	Lower CI	1.78	1.16	1.41	1.60	2.01	1.72	1.95	1.66	1.99	1.45	1.06
ICD-10 Q65	Upper CI	2.73	1.92	2.21	2.41	2.88	2.54	2.81	2.47	2.85	2.18	1.69
Congenital Hip Dislocation Subluxation and Dysplasia	NUMBER	55	49	51	72	84	74	84	72	85	65	59
	RATE	**1.36**	**1.17**	**1.13**	**1.48**	**1.66**	**1.44**	**1.66**	**1.42**	**1.62**	**1.22**	**1.06**
ICD-10 Q65.0-Q65.5 & Q65.80-Q65.81	Lower CI	1.02	0.87	0.85	1.16	1.33	1.13	1.33	1.11	1.30	0.94	0.81
	Upper CI	1.76	1.55	1.49	1.86	2.06	1.81	2.06	1.79	2.01	1.56	1.37
Reduction Deformity, Upper Limbs	NUMBER	32	27	18	33	33	39	29	44	40	32	40
	RATE	**0.79**	**0.64**	**0.40**	**0.68**	**0.65**	**0.76**	**0.57**	**0.87**	**0.76**	**0.60**	**0.72**
	Lower CI	0.54	0.43	0.24	0.47	0.45	0.54	0.38	0.63	0.55	0.41	0.52
ICD-10 Q71..	Upper CI	1.11	0.94	0.63	0.95	0.92	1.04	0.82	1.17	1.04	0.85	0.98
Reduction Deformity, Lower Limbs	NUMBER	14	15	10	19	18	22	14	22	19	18	17
	RATE	**0.35**	**0.36**	**0.22**	**0.39**	**0.36**	**0.43**	**0.28**	**0.43**	**0.36**	**0.34**	**0.31**
	Lower CI	0.19	0.20	0.11	0.24	0.21	0.27	0.15	0.27	0.22	0.20	0.18
ICD-10 Q72..	Upper CI	0.58	0.59	0.41	0.61	0.56	0.65	0.46	0.66	0.56	0.53	0.49
Diaphragmatic Hernia	NUMBER	8	17	12	18	18	20	17	18	23	12	15
	RATE	**0.20**	**0.41**	**0.27**	**0.37**	**0.36**	**0.39**	**0.34**	**0.36**	**0.44**	**0.23**	**0.27**
	Lower CI	0.09	0.24	0.14	0.22	0.21	0.24	0.20	0.21	0.28	0.12	0.15
ICD-10 Q79.0, Q79.11, Q79.12	Upper CI	0.39	0.65	0.46	0.58	0.56	0.60	0.54	0.56	0.66	0.39	0.44

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Alberta Congenital Anomalies Surveillance System
RCPCH version ICD-10 Q Chapter (Q00-Q99)
Aggregate Year Anomaly Rates per 1,000 Total Births
Total Births (Live, still and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		00-04 (5 years)	05-09 (5 years)	10-14 (5 years)	00-14 (15 years)
Hydronephrosis	NUMBER	292	388	507	1187
	RATE	**1.51**	**1.63**	**1.93**	**1.71**
	Lower CI	1.34	1.48	1.77	1.62
ICD-10 Q62.0..	Upper CI	1.69	1.80	2.11	1.81
Pelviureteric Junction Obstruction	NUMBER	41	45	49	135
	RATE	**0.21**	**0.19**	**0.19**	**0.19**
	Lower CI	0.15	0.14	0.14	0.16
ICD-10 Q62.10 & Q62.11	Upper CI	0.29	0.25	0.25	0.23
Vesicoureteric Junction Obstruction	NUMBER	4	13	8	25
	RATE	**0.02**	**0.05**	**0.03**	**0.04**
	Lower CI	0.01	0.03	0.01	0.02
ICD-10 Q62.12 & Q62.13	Upper CI	0.05	0.09	0.06	0.05
Posterior Urethral Valves	NUMBER	15	22	24	61
	RATE	**0.08**	**0.09**	**0.09**	**0.09**
	Lower CI	0.04	0.06	0.06	0.07
ICD-10 Q64.20	Upper CI	0.13	0.14	0.14	0.11
Congenital Deformities Hip (All)	NUMBER	418	469	517	1404
	RATE	**2.16**	**1.97**	**1.97**	**2.03**
	Lower CI	1.96	1.80	1.81	1.92
ICD-10 Q65	Upper CI	2.38	2.16	2.15	2.13
Congenital Hip Dislocation, Subluxation and Dysplasia	NUMBER	285	330	365	980
	RATE	**1.47**	**1.39**	**1.39**	**1.41**
ICD-10 Q65.0-Q65.5 & Q65.80-Q65.81	Lower CI	1.31	1.24	1.25	1.33
	Upper CI	1.65	1.55	1.54	1.51
Reduction Deformity, Upper Limbs	NUMBER	142	150	185	477
	RATE	**0.73**	**0.63**	**0.71**	**0.69**
	Lower CI	0.62	0.53	0.61	0.63
ICD-10 Q71..	Upper CI	0.86	0.74	0.81	0.75
Reduction Deformity, Lower Limbs	NUMBER	81	84	90	255
	RATE	**0.42**	**0.35**	**0.34**	**0.37**
	Lower CI	0.33	0.28	0.28	0.32
ICD-10 Q72..	Upper CI	0.52	0.44	0.42	0.42
Diaphragmatic Hernia	NUMBER	80	85	85	250
	RATE	**0.41**	**0.36**	**0.32**	**0.36**
	Lower CI	0.33	0.29	0.26	0.32
ICD-10 Q79.0.., Q79.11, Q79.12	Upper CI	0.51	0.44	0.40	0.41

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Alberta Congenital Anomalies Surveillance System
RCPCH version ICD-10 Q Chapter (Q00-Q99)
Single Year Anomaly Rates per 1,000 Total Births
Total Births (Live, still and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		2004	2005	2006	2007	2008	2009	2010	2011	2012	2013	2014
Abdominal Wall Defects (all)	NUMBER	33	41	40	39	48	52	52	57	49	47	48
	RATE	**0.81**	**0.98**	**0.89**	**0.80**	**0.95**	**1.01**	**1.03**	**1.13**	**0.94**	**0.88**	**0.87**
	Lower CI	0.56	0.70	0.64	0.57	0.70	0.76	0.77	0.85	0.69	0.65	0.64
ICD-10 Q79.2 to Q79.5	Upper CI	1.14	1.33	1.21	1.10	1.26	1.33	1.35	1.46	1.24	1.18	1.15
Omphalocele	NUMBER	12	10	10	11	19	23	25	24	21	23	19
	RATE	**0.30**	**0.24**	**0.22**	**0.23**	**0.38**	**0.45**	**0.49**	**0.47**	**0.40**	**0.43**	**0.34**
	Lower CI	0.15	0.11	0.11	0.11	0.23	0.28	0.32	0.30	0.25	0.27	0.21
ICD-10 Q79.2	Upper CI	0.51	0.44	0.41	0.40	0.59	0.67	0.73	0.70	0.61	0.65	0.53
Gastroschisis	NUMBER	13	24	27	25	23	26	21	29	19	17	25
	RATE	**0.32**	**0.57**	**0.60**	**0.51**	**0.46**	**0.51**	**0.42**	**0.57**	**0.36**	**0.32**	**0.45**
	Lower CI	0.17	0.37	0.40	0.33	0.29	0.33	0.26	0.38	0.22	0.19	0.29
ICD-10 Q79.3	Upper CI	0.55	0.85	0.87	0.76	0.68	0.74	0.63	0.82	0.57	0.51	0.66
All Chromosome Anomalies	NUMBER	170	219	198	230	223	251	244	228	237	284	285
	RATE	**4.19**	**5.23**	**4.40**	**4.72**	**4.41**	**4.88**	**4.82**	**4.50**	**4.53**	**5.34**	**5.14**
	Lower CI	3.59	4.56	3.81	4.13	3.86	4.30	4.24	3.94	3.97	4.74	4.56
ICD-10 Q90-Q99	Upper CI	4.87	5.97	5.06	5.37	5.03	5.53	5.47	5.12	5.14	6.00	5.77
Trisomy 13	NUMBER	10	9	12	20	15	20	11	12	12	21	12
	RATE	**0.25**	**0.21**	**0.27**	**0.41**	**0.30**	**0.39**	**0.22**	**0.24**	**0.23**	**0.39**	**0.22**
	Lower CI	0.12	0.10	0.14	0.25	0.17	0.24	0.11	0.12	0.12	0.24	0.11
ICD-10 Q91.4-Q91.7	Upper CI	0.45	0.41	0.46	0.63	0.49	0.60	0.39	0.41	0.40	0.60	0.38
Trisomy 18	NUMBER	18	28	22	23	28	32	34	25	28	43	36
	RATE	**0.44**	**0.67**	**0.49**	**0.47**	**0.55**	**0.62**	**0.67**	**0.49**	**0.54**	**0.81**	**0.65**
	Lower CI	0.26	0.44	0.31	0.30	0.37	0.43	0.47	0.32	0.36	0.59	0.45
ICD-10 Q91.0-Q91.3	Upper CI	0.70	0.97	0.74	0.71	0.80	0.88	0.94	0.73	0.77	1.09	0.90
Down Syndrome (Trisomy 21)	NUMBER	82	111	87	118	97	107	125	120	126	134	140
	RATE	**2.02**	**2.65**	**1.94**	**2.42**	**1.92**	**2.08**	**2.47**	**2.37**	**2.41**	**2.52**	**2.52**
	Lower CI	1.61	2.18	1.55	2.01	1.56	1.71	2.06	1.96	2.01	2.11	2.12
ICD-10 Q90..	Upper CI	2.51	3.19	2.39	2.90	2.34	2.51	2.94	2.83	2.87	2.98	2.98

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
Total Births (Live, still and fetal losses)

Diagnostic Category and ICD-10 RCPCH Code		00-04 (5 years)	05-09 (5 years)	10-14 (5 years)	00-14 (15 years)
Abdominal Wall Defects (all)	NUMBER	143	220	253	616
	RATE	**0.74**	**0.93**	**0.96**	**0.89**
	Lower CI	0.62	0.81	0.85	0.82
ICD-10 Q79.2-Q79.5	Upper CI	0.87	1.06	1.09	0.96
Omphalocele	NUMBER	52	73	112	237
	RATE	**0.27**	**0.31**	**0.43**	**0.34**
	Lower CI	0.20	0.24	0.35	0.30
ICD-10 Q79.2	Upper CI	0.35	0.39	0.51	0.39
Gastroschisis	NUMBER	63	125	111	299
	RATE	**0.33**	**0.53**	**0.42**	**0.43**
	Lower CI	0.25	0.42	0.35	0.38
ICD-10 Q79.3	Upper CI	0.42	0.63	0.51	0.48
All Chromosome Anomalies	NUMBER	769	1121	1278	3168
	RATE	**3.97**	**4.72**	**4.87**	**4.57**
	Lower CI	3.70	4.45	4.61	4.41
ICD-10 Q90-Q99	Upper CI	4.26	5.01	5.15	4.73
Trisomy 13	NUMBER	36	76	68	180
	RATE	**0.19**	**0.32**	**0.26**	**0.26**
	Lower CI	0.13	0.25	0.20	0.22
ICD-10 Q91.4-Q91.7	Upper CI	0.26	0.40	0.33	0.30
Trisomy 18	NUMBER	84	133	166	383
	RATE	**0.43**	**0.56**	**0.63**	**0.55**
	Lower CI	0.35	0.47	0.54	0.50
ICD-10 Q91.0-Q91.3	Upper CI	0.54	0.66	0.74	0.61
Down Syndrome (Trisomy 21)	NUMBER	379	520	645	1544
	RATE	**1.96**	**2.19**	**2.46**	**2.23**
	Lower CI	1.77	2.01	2.27	2.12
ICD-10 Q90..	Upper CI	2.17	2.39	2.66	2.34

Appendix A.4 Numbers of Cases, Anomalies and Anomalies per Case 1997–2014 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	1981	53.84	1.76
1998	37715	1193	31.63	2186	57.96	1.83
1999	38044	1223	32.15	2422	63.66	1.98
2000	36860	1288	34.94	2358	63.97	1.83
2001	37454	1384	36.95	2592	69.20	1.87
2002	38540	1374	35.65	2539	65.88	1.85
2003	40120	1518	37.84	2602	64.86	1.71
2004	40570	1550	38.21	2887	71.16	1.86
2005	41890	1609	38.41	2887	68.92	1.79
2006	44954	1621	36.06	2716	60.42	1.68
2007	48708	1873	38.45	3132	64.30	1.67
2008	50512	2007	39.73	3458	68.46	1.72
2009	51420	2091	40.67	3626	70.52	1.73
2010	50590	2191	43.31	3695	73.04	1.69
2011	50665	2089	41.23	3632	71.69	1.74
2012	52318	2123	40.58	3705	70.82	1.75
2013	53180	2136	40.17	3763	70.76	1.76
2014	55490	2162	38.96	3795	68.39	1.76
1997–2014	**805827**	**30558**	**37.92**	**53976**	**66.98**	**1.77**

Alberta Total Births from: Alberta Vital Statistics Annual Reviews for 1980-2013
Alberta Perinatal Health Programme for 2014

Appendix A.5 Chi Trend Table for Reported Anomalies 1997–2014

Anomaly	X^2	p Value	Direction*
Anencephaly	1.20	0.2733	?↓
Spina bifida without anencephaly	0.00	1.00	↔
Encephalocele	0.49	0.4839	?↓
Neural tube defects (all)	1.07	0.3009	?↓
Hydrocephalus without spina bifida	0.20	0.6547	↔
Arhinencephaly/ Holoprosencephaly	2.05	0.1522	?↑
Microcephaly	1.35	0.2453	?↑
Anophthalmia/Microphthalmia	3.13	0.0769	?↓
Congenital cataract	0.97	0.3247	?↑
Anotia/Microtia	3.25	0.0714	?↑
Congenital heart defects (all)	1.28	0.2579	↔
Common truncus	0.93	0.3349	?↑
Transposition of great arteries	0.35	0.5541	↔
Tetralogy of Fallot	4.30	0.0381	↑
Ventricular septal defect	3.75	0.0528	?↑
Atrial septal defect	0.28	0.5967	↔
Endocardial cushion defect	2.29	0.1302	?↑
Pulmonary valve atresia/stenosis	0.19	0.6629	↔
Tricuspid valve atresia/stenosis	0.11	0.7401	↔
Ebstein's anomaly	2.12	0.1454	?↑
Aortic valve atresia/stenosis	4.99	0.0255	↓
Hypoplastic left heart syndrome	0.68	0.4096	↔
Coarctation of the aorta	5.65	0.0175	↑
Cleft palate without cleft lip (CPO)	3.97	0.0463	↓
Cleft lip without cleft palate (CLO)	1.12	0.2899	↔
Cleft lip and cleft palate (CL+CP)	0.03	0.8625	↔
Cleft lip with and without cleft palate (CL+/-CP)	0.62	0.4310	↔
Choanal atresia/stenosis	0.04	0.8415	↔
Oesophageal atresia/trachea-oesophageal fistula	0.23	0.6315	↔
Pyloric stenosis	0.76	0.3833	↔
Small intestinal atresia/stenosis (all)	0.18	0.6714	↔

Anomaly	X^2	p Value	Direction*
Duodenal atresia/stenosis	1.61	0.2045	?↑
Rectal and large intestinal atresia/stenosis (all)	23.28	<0.0001	↓
Rectal atresia/stenosis	8.11	0.0044	↓
Anal atresia/stenosis	13.67	0.0002	↓
Other large intestinal atresia/stenosis	4.27	0.0388	↓
Hirschsprung's disease	2.21	0.1371	?↑
Biliary atresia	0.16	0.6892	↔
Undescended testes (male denominator)	6.37	0.0116	↑
Hypospadias (male denominator)	51.10	<0.0001	↑
Epispadias (male denominator)	0.61	0.4348	?↑
Renal agenesis/hypoplasia	2.33	0.1269	?↑
Cystic kidney	0.72	0.3961	↔
Bladder exstrophy	0.92	0.3375	?↓
Obstructive genitourinary defects (all)	34.14	<0.0001	↑
Hydronephrosis	34.19	<0.0001	↑
UPJ obstruction	0.77	0.3802	?↑
VUJ obstruction (based on very few cases per yr - range 0-4)	4.10	0.0429	↑
Posterior urethral valves	0.26	0.6101	↔
Congenital deformities of hip (all)	0.13	0.7184	↔
Congenital hip dislocation, subluxation, dysplasia	0.30	0.5839	↔
Reduction deformity, upper	0.28	0.5967	↔
Reduction deformity, lower	0.40	0.5271	↔
Diaphragmatic hernia	0.38	0.5376	↔
Abdominal wall defects (all)	9.58	0.0020	↑
Omphalocele	7.92	0.0049	↑
Gastroschisis	5.96	0.0146	↑
All chromosome anomalies	52.57	<0.0001	↑
Trisomy 13	6.83	0.0090	↑
Trisomy 18	10.13	0.0015	↑
Trisomy 21	23.02	0.0001	↑

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

