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Original quantitative research

The Alberta Congenital Anomalies Surveillance System: a 40-year review with prevalence and trends for selected congenital anomalies, 1997–2019

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Abstract

Introduction: Current published long-term provincial or territorial congenital anomaly data are lacking for Canada. We report on prevalence (per 1000 total births) and trends in 1997–2019, in Alberta, Canada, for selected congenital anomalies. Associated risk factors are also discussed.

Methods: We used data from the Alberta Congenital Anomalies Surveillance System (ACASS) to calculate the prevalence and perform chi-square linear trend analyses.

Results: From 1997 to 2019, the overall prevalence of neural tube defects was stable, at 0.74 per 1000 total births. The same was true for spina bifida (0.38), orofacial clefts (1.99), more severe CHDs (transposition of the great arteries, 0.38; tetralogy of Fallot, 0.33; and hypoplastic left heart syndrome, 0.32); and gastroschisis (0.38). Anencephaly, cleft palate and anorectal malformation significantly decreased with a prevalence of 0.23, 0.75 and 0.54 per 1000 total births, respectively. Significantly increasing trends were reported for anotia/microtia (0.24), limb reduction anomalies (0.73), omphalocele (0.36) and Down syndrome (2.21) and for hypospadias and undescended testes (4.68 and 5.29, respectively, per 1000 male births).

Conclusion: Congenital anomalies are an important public health concern with significant social and societal costs. Surveillance data gathered by ACASS for over 40 years can be used for planning and policy decisions and the evaluation of prevention strategies. Contributing genetic and environmental factors are discussed as is the need for continued surveillance and research.

Keywords: *congenital anomalies, surveillance, prevalence, trends, Alberta*

Highlights

- The Alberta Congenital Anomalies Surveillance System reports prevalence of anomalies and trends from 1997 to 2019 among live births, stillbirths and terminations of pregnancy at less than 20 weeks gestation.
- Overall prevalence of each of the following was stable, showing no significant trends: neural tube defects, spina bifida, orofacial clefts, cleft lip with or without cleft palate, severe congenital heart defects and gastroschisis.
- Anencephaly, cleft palate and anorectal malformations show significantly decreasing trends.
- Anotia/microtia, ventricular septal defects, hypospadias, undescended testes, limb reductions, omphalocele and Down syndrome show significantly increasing trends.
- Precise risk factors are challenging to address, supporting the need for continued congenital anomalies surveillance and research to be integral to public health.

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Introduction

Congenital anomalies surveillance in Alberta started in 1963 in response to malformations caused by thalidomide in the late 1950s. In addition to collecting data on structural congenital anomalies, this surveillance system included all physical and neurodevelopmental disabilities of children and included adults. In 1979, the Alberta government restricted surveillance to only congenital anomalies. In 1982, the government proposed to discontinue congenital anomaly surveillance completely, but agreed to transfer the surveillance system to the Alberta Children's Hospital, Department of Medical Genetics, without transfer of funds. Funds were secured by grant applications until 1994, when the government resumed funding.

The Alberta Congenital Anomalies Surveillance System (ACASS), a provincial population-based program, has data from 1980. Current, published long-term national, provincial or territorial congenital anomaly data are lacking for Canada.

The objectives of this paper are to report prevalence rates and trends for neural tube defects (NTDs), anotia/microtia, orofacial clefts, anorectal malformations, specific congenital heart defects (CHDs), hypospadias, undescended testes, limb reduction anomalies, gastroschisis, omphalocele and Down syndrome for 1997 to 2019, using data from ACASS. These data allow for other Canadian provinces and territories and the Canadian Congenital Anomalies Surveillance System (CCASS) to compare rates and trends.

ACASS is one of the only surveillance systems in Canada with data on termination of pregnancies at less than 20 weeks gestation with more complete ascertainment to contribute to better prevalence estimates. Although inclusion of ascertainment of termination of pregnancies was recommended for CCASS in 1997,[1] it has not been sufficiently achieved.

ACASS also provides context to the reported rates and trends, which is necessary for valid interpretation. Currently, CCASS reports numbers without context via their Public Health Infobase,[2] with their last comprehensive report published in 2013 with data to 2009.[3]

Methods

ACASS is primarily a passive system that relies on health care professionals and administrative data for case ascertainment as opposed to an active system where trained surveillance staff abstract case data. Still, it is best described as a hybrid system because we have both aspects, with legal permission to access patient medical records including supporting documentation (e.g. reports from consultations, operations, cytogenetics, diagnostic imaging and pathology). Thus, we can verify or clarify diagnoses including those that occur after termination of pregnancies at less than 20 weeks gestation. Eligible cases are born in Alberta to mothers who reside in Alberta at the time of delivery. Cases that have structural, syndromic, chromosomal, neoplasm, endocrine and/or metabolic abnormalities are ascertained for up to 1 year after delivery. Anomalies are coded using the Royal College of Paediatrics and Child Health (RCPCH) adaptation of the International Classification of Diseases, 10th Revision (ICD-10). Only selected congenital anomalies are included in this paper; however, data for additional anomalies are available.[4]

Multiple ascertainment sources are used (see Table 1) to include live births and stillbirths (>20 weeks gestation and/or >500 g) born since 1 January 1980. Data since 1 January 1997 also include early fetal deaths and termination of pregnancies (<20 weeks gestation and/or <500 g), which is why this paper focusses on the period 1 January 1997 to 31 December 2019.

The ACASS methodology is described in greater detail by Lowry et al.[4]

Alberta Vital Statistics provided denominators. We calculated prevalence as number of cases divided by total number of live births and stillbirths and, for hypospadias and undescended testes, as number of cases divided by total number of male live births and stillbirths, with 95% confidence intervals. Chi-square linear trend analysis was performed.

As this Registry is a part of public health surveillance in Alberta, which is covered by provincial legislation, no ethics approval is required from Alberta Health or the University of Calgary.

Results

Table 2 shows the case prevalence for selected congenital anomalies per 1000 total births. The overall rate of NTDs is 0.74 and of orofacial clefts is 1.99. The most frequent CHDs are septal defects (ventricular septal defects [VSDs] at 3.10 and atrial septal defects [ASDs] at 2.01). The rates of the more severe CHDs, including hypoplastic left heart syndrome (HLHS; 0.32), transposition of the great arteries (0.38) and tetralogy of Fallot (0.33), are comparable. Gastroschisis and omphalocele rates are similar (0.38 and 0.36, respectively). The rate of Down syndrome is 2.21. The prevalence, per 1000 total male births, of hypospadias is 4.68 and of undescended testes is 5.29.

TABLE 1
Sources of data for the Alberta Congenital Anomalies Surveillance System

Alberta Vital Statistics	Physicians' Notice of Birth
	Medical Certificate of Death
	Medical Certificate of Stillbirth
All Alberta hospitals	Case notifications from Alberta Hospital Health Records Department via the Congenital Anomalies Reporting Form (CARF)
	Alberta Children's Hospital (Calgary) and Stollery Children's Hospital (Edmonton)
Specialty data sources	Outpatient Clinics (e.g. genetics, prenatal, metabolics)
	Alberta Precision Laboratories (e.g. Cytogenetics, Newborn Metabolic Screening)
	Calgary and Edmonton Pathology

TABLE 2
Case prevalence of selected congenital anomalies in Alberta, 1997–2019

Congenital anomaly	ICD-10 RCPCH code	Prevalence per 1000 total births[a] (95% CI)
NTDs (all)	Q00…, Q01…, Q05…	0.74 (0.69–0.79)
Anencephaly	Q00.00, Q00.01, Q00.1	0.23 (0.20–0.26)
Spina bifida	Q05…	0.38 (0.35–0.42)
Anotia/microtia	Q16.0, Q17.2	0.24 (0.21–0.27)
Orofacial clefts (all)	Q35…, Q36…, Q37…	1.99 (1.91–2.08)
CLP	Q36…, Q37…	1.23 (1.17–1.30)
Cleft palate only	Q35…	0.75 (0.70–0.81)
Anorectal malformations	Q42…	0.54 (0.50–0.58)
CHDs		
Transposition of the great arteries	Q20.11, Q20.3, Q20.5	0.38 (0.34–0.42)
Tetralogy of Fallot	Q21.3…, Q21.82	0.33 (0.30–0.36)
VSD	Q21.0	3.10 (3.00–3.21)
ASD[b]	Q21.1…	2.01 (1.93–2.10)
Hypoplastic left heart syndrome	Q23.4	0.32 (0.29–0.35)
Hypospadias[c]	Q54… (exclude Q54.4)	4.68 (4.51–4.87)
Undescended testes[b,c]	Q53…	5.29 (5.10–5.48)
Limb reduction	Q71…, Q72…	0.73 (0.68–0.78)
Gastroschisis	Q79.3	0.38 (0.35–0.42)
Omphalocele	Q79.2	0.36 (0.32–0.39)
Down syndrome	Q90…	2.21 (2.12–2.30)

Source: Alberta Congenital Anomalies Surveillance System

Abbreviations: ASD, atrial septal defect; CHD, congenital heart defect; CI, confidence interval; CLP, cleft lip with or without cleft palate; ICD-10, International Classification of Diseases, 10th Revision; NTD, neural tube defect; RCPCH, Royal College of Paediatrics and Child Health; VSD, ventricular septal defect.

[a] Total number of births (1997–2019) = 1 074 927.

[b] >36 weeks gestation.

[c] Per male births only; total number of male births (1997–2019) = 550 712.

Results from the chi-square linear trend analyses for 1997–2019 are shown in Table 3. While there are no significant trends for NTDs overall or for spina bifida, anencephaly is significantly decreasing. Rates for cleft palate are also decreasing, while cleft lip with or without cleft palate (CLP) and overall orofacial clefts rates show no significant change. Anorectal malformation rates are significantly decreasing. Although the majority of selected CHD rates show no change, VSD rates are significantly increasing. Rates of both hypospadias and undescended testes show significant increases, as do limb reductions. Gastroschisis has stabilized, while omphalocele is significantly increasing, as is Down syndrome.

Discussion

Neural tube defects

NTDs show no significant change (p = 0.0585). Anencephaly prevalence rates started to decline in 2016 and continued to 2019. In contrast, spina bifida rates have remained stable.

Anencephaly rates are influenced by very early termination of pregnancies and perhaps by the terminology used to describe prenatal findings, for example, "absent calvarium," which is coded in ICD-10 RCPCH under the musculoskeletal system (Q75.8) and not with anencephaly/exencephaly. As a result, such cases are not classified as NTDs.

Acrania can progress to exencephaly and anencephaly.[5] The method of termination often precludes an accurate postmortem diagnosis.

The most recent statistics from the Public Health Agency of Canada (PHAC) use data to 2014 and show no trend for NTDs.[2]

Folic acid fortification was introduced in Canada in 1998 and has had a significant impact on the prevalence of NTDs. Nevertheless, a substantial number of such defects remain,[6] which may be due to red blood cell folate levels being below 906 nmol/L and/or the need to supplement with vitamin B12[7] or inositol.[8] Additional risk factors include maternal obesity, diabetes mellitus and the use of anticonvulsants and folic acid antagonists.[9]

Anotia/microtia

Most clinicians and surveillance programs classify anotia/microtia into four categories, with type 4 anotia the most severe and type 1 being a smaller ear with normal structure. Some studies record only types 2 to 4,[10] which include most ACASS cases. While the rate sharply dropped in 2019, ACASS closely monitors the overall significantly increasing trend, which remains unexplained.

Risk factors include male sex, maternal diabetes and obesity, Hispanic ethnicity, advanced maternal age, high parity, multifetal gestation, cold symptoms and viral infection.[10] Luquetti et al. summarized the epidemiology and genetics of microtia, including higher risks associated with Asian, Pacific Islander, Native/Alaskan and Indigenous ethnicities.[11] Living at an altitude greater than 2000 m is a risk factor, but this risk factor does not apply in Alberta (Calgary is at 1048 m and Edmonton at 645 m). Maternal smoking and alcohol are reported as risk factors in nonisolated cases[10] and alcohol exposure in isolated cases.[11] Known teratogens include thalidomide, isotretinoin and mycophenolate mofetil.[11]

Orofacial clefts

The overall rates for CLP have remained stable in Alberta for over 40 years and in other jurisdictions for over 30–50 years.[12] In contrast, cleft palate has shown a significantly declining trend (see Table 3). A decline in California has been reported for CLP, but not for cleft palate (1987–2010), suggesting a possible contribution of folic acid fortification to this decline.[13] Lowry et al.[14] compared the period prior to the introduction of folic acid fortification (1993–1997) with two periods after (2000–2004 and 2012–2016), in Alberta, and reported no decline for total CLP cases or for isolated cases over the three timeframes.

TABLE 3
Chi-square linear trend analyses and p values for selected anomalies in Alberta, 1997–2019

Congenital anomaly	Trend direction	Chi-square analysis (X^2LT)	*p* value
NTDs (all)	No significant change	3.58	0.0585
Anencephaly	Decreasing	7.00	0.0082
Spina bifida	No significant change	0.01	0.9203
Anotia/microtia	Increasing	5.67	0.0173
Orofacial clefts (all)	No significant change	0.88	0.3482
CLP	No significant change	0.32	0.5716
Cleft palate only	Decreasing	5.05	0.0246
Anorectal malformations	Decreasing	10.39	0.0013
CHDs			
Transposition of the great arteries	No significant change	1.14	0.2857
Tetralogy of Fallot	No significant change	0.90	0.3428
VSD	Increasing	4.79	0.0286
ASD[a]	No significant change	0.08	0.7773
Hypoplastic left heart syndrome	No significant change	2.26	0.1328
Hypospadias	Increasing	55.83	< 0.0001
Undescended testes[a]	Increasing	14.22	0.0002
Limb reduction	Increasing	4.49	0.0341
Gastroschisis	No significant change	0.07	0.7913
Omphalocele	Increasing	12.07	0.0005
Down syndrome	Increasing	23.54	< 0.0001

Abbreviations: ASD, atrial septal defect; CHD, congenital heart defects; CLP, cleft lip with or without cleft palate; LT, linear trend; NTD, neural tube defect; VSD, ventricular septal defect.

[a] >36 weeks gestation.

A decline was reported in prevalence of orofacial clefts for 1994–2017 in Ontario, especially for cleft palate; however, data from stillbirths and terminations of pregnancy were lacking.[15] The only national Canadian reported data covers 2005–2014 and show no change in trend for CLP but a possible downward trend for cleft palate.[2]

Risk factors include active and passive smoking; alcohol consumption, particularly binge drinking; and maternal obesity. Gene polymorphisms also play a role.[16,17] Meta-analysis of maternal supplementation suggests that periconception intake of folic acid plus multivitamins can reduce occurrence as well as recurrence.[18] The Hutterite Brethren, whose smoking and alcohol consumption is limited and nutrition probably adequate, had zero cases of cleft lip with cleft palate in 1980–2016.[19]

Anorectal malformations

The overall trend is significantly decreasing (p = 0.0013). A 2007 ACASS study for the years 1990–2004 showed stable rates[20] that compared favourably with the results of other studies of that time. The current decline is for both isolated and associated anomaly cases. Khanna et al.[21] reviewed genetic factors contributing to the etiopathogenesis of isolated cases and concluded that a number of copy number variants and/or single nucleotide variants contributed to the defect. Families with autosomal dominant inheritance are reported to exist.[21]

Risk factors include maternal smoking, maternal body mass index (BMI) greater than 30, assisted reproductive technology, maternal chronic respiratory disease, maternal use of anti-asthmatic medications, hypnotics and benzodiazepine.[22] Zwink and Jenetzky[22] report inconsistent results for the protective effects of folic acid supplements.

Zwink and Jenetzky[22] found that in the majority of studies in their systematic review approximately 60% of cases have an associated anomaly; this compares with 82% of ACASS cases. This difference should be interpreted with caution, as inclusion criteria and case classification differed. Other studies may only include live-born and surgically treated cases.

Congenital heart defects

The more severe anomalies show no significant trends with similar case prevalence rates (per 1000 total births: HLHS, 0.32; tetralogy of Fallot, 0.33; and transposition of the great arteries, 0.38). Öhman et al.[23] reported a decrease of live births with HLHS in Sweden, and suggest that this decrease was due to increased prenatal detection and termination of pregnancies. This highlights the importance of ascertaining termination of pregnancies to determine more accurate prevalence.

While the prevalence of ASDs remained stable between 1997 and 2019 (p = 0.7773), the prevalence of VSDs has statistically significantly increased (p = 0.0286), likely because small septal defects are better diagnosed as a result of advances in echocardiography and heart ultrasound. However, ACASS does not accept patent foramen ovales, ASDs in premature infants or ASDs that are smaller than 3 mm and spontaneously close; conversely, ACASS does accept VSDs, regardless of their size, the need for intervention or their spontaneous closing.

Although most CHDs are multifactorial, genetic diagnoses have been reported in 15.7% of cases that have a severe CHD requiring surgery or therapeutic intervention in the first year of life.[24] Cases with a known aneuploidy were excluded.[24] There is emerging evidence that complex single-gene disorders often present as isolated CHDs prenatally, as complete phenotyping may not be possible.[25] In the past two decades, genetic variants have been associated with nonsyndromic or isolated CHDs, particularly for highly conserved transcription factors essential for cardiac development (e.g. *GATA4* variants associated with tetralogy of Fallot, ASDs, VSDs, atrioventricular septal defects and pulmonary stenosis).[26]

Reported risk factors for CHDs include teratogens (e.g. thalidomide, isotretinoin, anticonvulsants, potassium channel blockers, lithium, alcohol), nutritional deficiencies (e.g. vitamin A, vitamin B3) and maternal conditions (diabetes, obesity, phenylketonuria, viral infections and hyperthermia).[27] Dolk et al.[28] reported significant associations with low maternal

education, vaginal infections, maternal clotting disorders and prescriptions for the anticlotting medication enoxaparin. With limited evidence to support such an association, more research is needed to confirm this reported increased risk with enoxaparin. Although the data did not support a protective effect of folic acid supplementation, risk was significantly increased for mothers with diets particularly low in fruits and vegetables, emphasizing the need to consider the entire dietary context.[28]

Placental abnormalities (e.g. low placental weight, altered gene expression in placental tissue) have also been reported to be associated with CHDs.[29] A more comprehensive framework has been proposed to include the environmental complement to the genome, an emerging field of the exposome.[30] Instead of a siloed approach, the interplay between internal and external prenatal environmental exposures that influence placental vascularization and subsequent fetal growth and development needs to be advanced.[30]

Hypospadias

The prevalence of hypospadias for both isolated and nonisolated cases peaked in 2015 and shows an overall significant increase ($p < 0.0001$) for 1997–2019. It is difficult to compare prevalence rates because of methodological differences, such as differences in the degree of severity, the inclusion of surgical cases only and whether rates are for total births versus male births. The EUROCAT report showed wide variability per 1000, with Portugal at 0.51 and Mainz (Germany) at 3.68.[31]

George et al.[32] have described the challenges with associations to determine the etiology of hypospadias and summarized the genetic and environmental factors. Consistent associated risk factors include a positive family history, low birth weight and/or small gestational age, maternal hypertension, preeclampsia, multiple gestations, placental insufficiency, diabetes mellitus and exposures to certain drugs such as progesterone derivatives or valproic acid. Evidence is inconsistent for risk factors such as maternal age and weight, paternal or maternal occupations and agriculture practices.

Genetic variants, such as the diacylglycerol kinase kappa (DGKK) variants, have been shown to be significant risk factors.[33] In California, cases with the DGKK variants and residential proximity to pesticide application had the highest odds ratios for hypospadias.[34] In Nova Scotia, the highest prevalence rates of hypospadias were in two counties that were associated with intense farming.[35] The prevalence of isolated hypospadias in the Hutterite Brethren is approximately double that of the general Alberta population, which may be associated with farming and agricultural practices.[19]

Undescended testes

While there was a sharp drop in rates of undescended testes in 2019, the trend from 1997 to 2019 shows a significant increase ($p = 0.0002$). These results have to be interpreted with caution, as this condition may resolve spontaneously or may in fact be retractile testes. A more accurate prevalence would be determined by knowing which full-term and normal birthweight cases came to orchidopexy. Surgical numbers could include preterm and low birth-weight babies. Hence, the difficulty in obtaining a true prevalence rate.

ACASS does not accept cases born before 37 weeks gestation or with a birth weight of less than 2500 g, but considers these to be physiological and caused by immaturity.

Although the etiology is likely multifactorial, there are some familial cases as well as multiple susceptibility genes.[36] Consistent risk factors are maternal smoking and diabetes, while maternal obesity, alcohol use, use of analgesics and exposure to endocrine-disrupting chemicals, such as agricultural pesticides, are inconsistently reported as risk factors.[37] No differences were reported in the prevalence of undescended testes in the Hutterite population and the general Alberta population.[19]

Limb reductions

Since 1980, rates have fluctuated[38] and we report a significant increase ($p = 0.0341$) for 1997–2019. As one case may have multiple limb reduction anomalies, we report both anomaly and case rates. Our rate of 0.73/1000 total births is comparable to studies from, for example, northern Netherlands (0.64/1000 for 1981–2017), which did not report a trend.[39]

Results of studies of folic acid, with or without supplements, reducing the risk of limb reductions are equivocal,[40] but it is clear that folic acid fortification has had no effect in Alberta. In most cases, the precise cause is unknown. Bergman et al.[39] recently found that an etiological cause was more likely to be identified in a case when more than one limb is affected or in a multiple congenital anomalies case with one affected limb, compared to cases with one limb affected and no other congenital anomalies. Risk factors include maternal smoking, pregestational diabetes, gestational hypertension, maternal age less than 25 years, upper respiratory tract infection in the first trimester, antiepileptic medications and lower educational level of parents.[41]

Gastroschisis

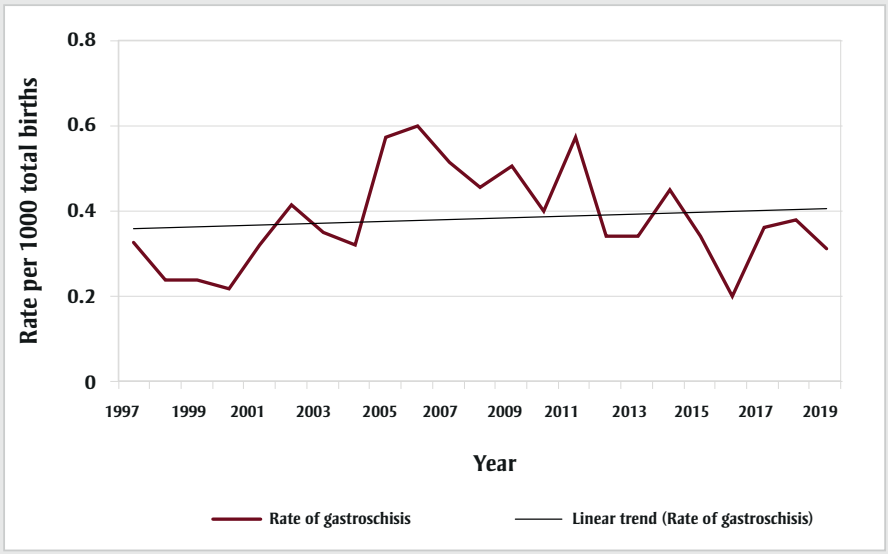
An increase in the prevalence of gastroschisis was noted in the early 1970s in many jurisdictions. In Alberta, the rate rose from 0.15 to 0.57/1000 total births between 1980 and 2011. Rates subsequently declined every year and have now stabilized (see Figure 1), which coincides with a decline in the number of teenage pregnancies (mothers < 20 years old) (see Figure 2). Young maternal age is a known risk factor, and the percentage of mothers younger than 20 years in Alberta fell from 7.3% in 2000 to 1.8% in 2019.[4]

A recent Canadian study using 2006–2017 data found similar results to those of ACASS for trend and a decrease in mothers younger than 20 years.[42] However, the North–South classification methodology the authors used and their interpretation of a geographical variation is problematic.[42] An Ontario study (2012–2018) reported no trend.[43] Neither study included early fetal deaths or terminations.[42,43]

Additional social risk factors include maternal smoking, use of marijuana, illicit drugs and alcohol, low BMI, poor nutrition and socioeconomic disadvantage.[43] There are fewer exposures to many of these risk factors in the Hutterite population, where there were no cases of gastroschisis between 1980 and 2016.[19] A recently recognized risk factor is exposure to wildfires during pre-pregnancy and the first trimester.[44]

While gastroschisis is usually an isolated anomaly, 28% of ACASS cases (data not shown) had a co-occurring anomaly; this is similar to findings in a study from

FIGURE 1
Trend for gastroschisis in Alberta, 1997–2019



* $p = 0.7913$.

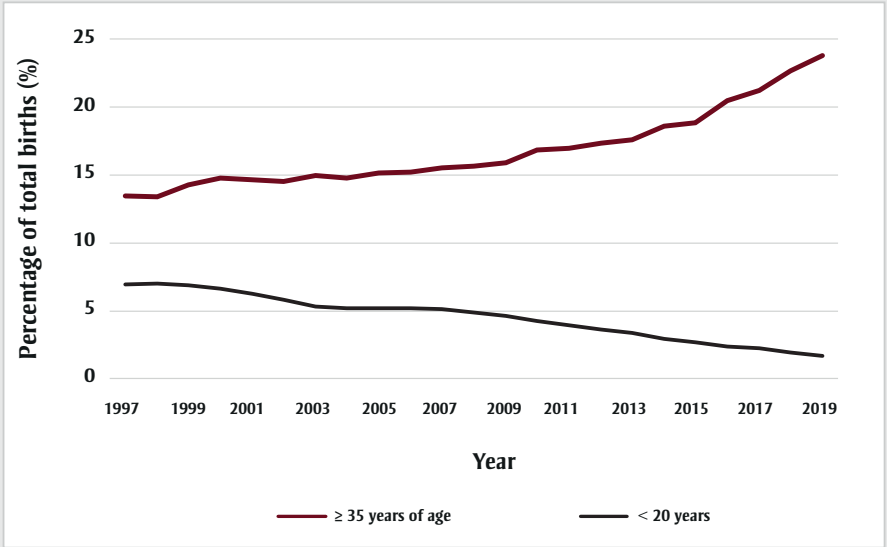
Sweden,[45] but was not mentioned by either of the recent Canadian studies.[42,43] Although gastroschisis is usually sporadic, there are familial reports of inheritance, including parent to child, full siblings, half siblings and distant relatives.[46] Geospatial studies have reported some provincial differences and clusters, with urban/rural differences in Ontario.[43]

Omphalocele

Comparable prevalence rates of omphalocele per 1000 total births have been recorded for several jurisdictions despite differing study years: 0.31 for 1997–2016;[47] 0.47 for 1993–2014;[48] and 0.38 for 2005–2011.[49] Neither trends for live births[47,48] nor for total births[49] were reported, but ACASS has a significantly increased trend for 1997–2019 ($p = 0.005$) (see Figure 3).

Associated anomalies, which include a malformation in another organ system, chromosomal abnormalities and syndromes, are present in 78% of cases recorded by ACASS. Trisomy 18 is very common, but a wide variety of abnormal karyotypes have been reported.

FIGURE 2
Proportion of births to women ≥35 years compared with women <20 years, as a percentage of total births in Alberta, 1997–2019



Risk factors include maternal age greater than 35 years or less than 20 years, maternal obesity and diabetes mellitus. Risks for exposures to smoking and alcohol are inconclusive.[50] A recent study has linked first trimester broad spectrum penicillin treatment with a reduced risk.[51]

Down syndrome

Down syndrome is significantly increasing ($p < 0.0001$) and is strongly correlated with increasing maternal age. In 1983, approximately 4% of mothers were 35 years or older; in 2019, 24% were in that age group.[4]

Frequently associated major malformations include CHDs and duodenal atresia. As most live-born infants with trisomy 21 require ongoing health services, ascertaining associated anomalies can help with future health care planning.

Strengths and limitations

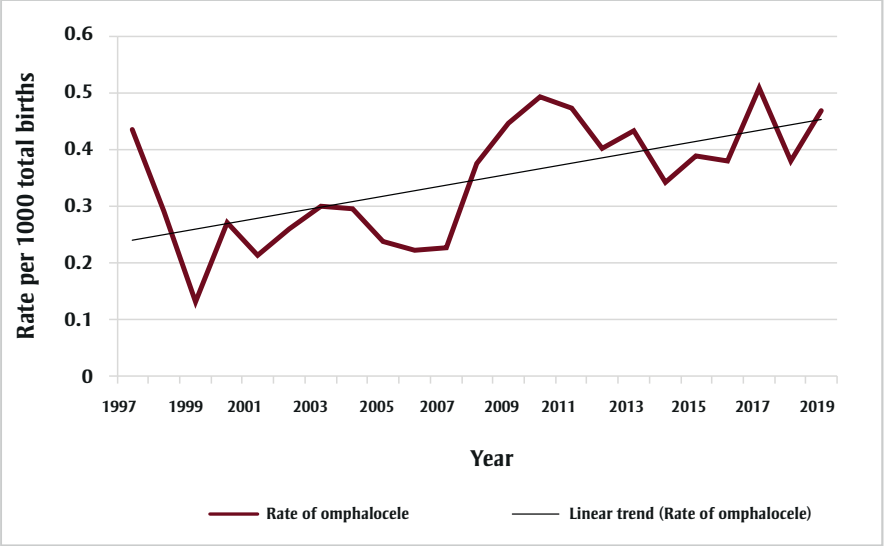
The strengths of this study are supported by the principal features of ACASS and include long-term baseline data, which are fundamental for valid descriptive and analytic studies. Additional features include provincial population-based coverage, multiple sources of ascertainment, ability to critically assess notifications and verify diagnoses that are reported to the system, and the expertise of ACASS personnel.

A limitation is that ACASS is technically a "passive" system, although it is augmented by active components, such as access to hospital records and correspondence with attending physicians for verification. ACASS primarily depends on others for case notifications and thus may not have complete ascertainment. The best systems, practised in many US States (e.g. Texas, Utah) and European and South American countries, have "active" ascertainment.

Conclusion

Congenital anomalies occur in approximately 3–5% of live births and 15% of stillbirths. They are an important public health concern and have significant social and societal costs. The majority of congenital anomalies are multifactorial, with

FIGURE 3
Trend for omphalocele in Alberta, 1997–2019



* $p = 0.0005$.

established risk factors often requiring a change in behaviour, which can be challenging (e.g. smoking and alcohol cessation, better control of maternal obesity and diabetes, folic acid/multivitamin supplementation and better nutrition).

Congenital anomalies surveillance data can be used for planning and policy decisions and the evaluation of prevention strategies, as exemplified by the success of folic acid fortification in the prevention of NTDs. These data are also required to respond to real and potential emerging threats such as Zika virus and the identification of congenital Zika syndrome. Many congenital anomalies surveillance programs now track outcomes of COVID-19 infection in pregnancy.

While funding is often challenging to obtain and maintain in Canada, PHAC is working with the provinces and territories to enhance CCASS data with more local datasets, which will provide more accurate prevalence rates of congenital anomalies across Canada. The last comprehensive congenital anomaly report published by PHAC used Canadian Institute for Health Information data from 1998–2009.[3] The British Columbia Health Status Registry was a world-class congenital anomalies surveillance system and after 70 years, the data were archived in 2021. Their last report was in 2005 using data to 2002.

With over 40 years in operation, ACASS has the most published prevalence data in Canada and provides context for more prevention. Congenital anomalies surveillance constitutes an essential data source for further research and to guide public health actions.[52]

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Conflicts of interest

The authors declare no conflicts of interest.

Authors' contributions and statement

RBL: Writing – Original draft. RBL, TB: Conceptualization of the work. RBL, TB, XG, SC, MAT: Data curation and analysis.

All the authors revised the manuscript for relevant and important intellectual content, edited the working manuscript and approved the final version for submission.

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