ILLINOIS DEPARTMENT OF PUBLIC HEALTH
IDPH
PROTECTING HEALTH, IMPROVING LIVES

Trends in the Prevalence of Birth Defects in Illinois 2002-2018

Epidemiologic Report Series 21:02

April 2021

TRENDS IN THE PREVALENCE OF BIRTH DEFECTS IN ILLINOIS 2002-2018

Illinois Department of Public Health
Division of Epidemiologic Studies

April 2021

ACKNOWLEDGMENTS

This report was possible because of the special efforts of many individuals and organizations. Particular thanks are due to the following Adverse Pregnancy Outcomes Reporting System (APORS) staff:

Jodi Snow, APORS Abstraction Liaison
Julie Rowden, APORS Coder
Latina Iverson-Simmons, APORS Abstractor
Lisa Lingleo, APORS Abstractor
Angela Butler, APORS Abstractor

Suggested Citation

Sandidge T, Fornoff JE, Shen T. Trends in the Prevalence of Birth Defects in Illinois 2002-2018. Epidemiologic Report Series 21:02, Springfield, IL. Illinois Department of Public Health, April 2021.

TRENDS IN THE PREVALENCE OF BIRTH DEFECTS IN ILLINOIS 2002-2018

TABLE OF CONTENTS

INDEX OF TABLES AND FIGURES iii

INTRODUCTION 1

METHODS 4

Calculation and Interpretation of Rates and Confidence Intervals 4
Analysis of Trends 5
Multiple Comparisons 5

FINDINGS 5

Rates of Birth Defects 5
Trend Analysis 5
Discussion of Illinois Results 7

REFERENCES 277

Description and ICD-9-CM Codes for Selected Birth Defects 29

INDEX OF TABLES AND FIGURES

Table 1: Projects to Identify Cases and Birth Defects 2

Table 2: Criteria that Determine Which Charts are Reviewed 3

Table 3. Birth Defects Showing a Significant Trend in Incidence Rate 7

Table 4. Number and Rate of Selected Birth Defects for 2018 (Illinois) 9

Table 5. Number and Rate of Selected Birth Defects for 2018 (Chicago) 131

Figure 1. Trends in the Reported Prevalence Rates of Neural Tube Defects 16

Figure 2. Trends in the Reported Prevalence Rates of Eye and Ear Defects 177

Figure 3A. Trends in the Reported Prevalence Rates of Cardiac Defects 188

Figure 3B. Trends in the Reported Prevalence Rates of Cardiac Defects 199

Figure 4. Trends in the Reported Prevalence Rates of Circulatory Defects 20

Figure 5. Trends in the Reported Prevalence Rates of Respiratory and Oral Defects 21

Figure 6. Trends in the Reported Prevalence Rates of Gastrointestinal Defects 222

Figure 7. Trends in the Reported Prevalence Rates of Genitourinary Defects 233

Figure 8. Trends in the Reported Prevalence Rates of Musculoskeletal Defects 244

Figure 9. Trends in the Reported Prevalence Rates of Chromosomal Defects 255

Figure 10. Trends in the Reported Prevalence Rates of Trisomy 21 (Down Syndrome), By Maternal Age at Delivery 26

INTRODUCTION

Since 1989, the Illinois Department of Public Health (IDPH) Adverse Pregnancy Outcomes Reporting System (APORS) has collected statewide data about congenital anomalies (birth defects) and other serious neonatal conditions identified in newborn infants. Birth defects are the leading cause of infant mortality in the United States (Ely & Driscoll, 2020) and the second leading cause of infant mortality in Illinois (Illinois Department of Public Health, 2021) and they contribute substantially to childhood morbidity and long-term disability. By collecting information about infants with birth defects, APORS is able to both study disease patterns and refer infants to local health departments and other providers for follow-up services.

Although they can occur at any stage of pregnancy, most birth defects occur during the first trimester when a baby's organs are developing. While the causes of some birth defects are known, there is still a lot to learn. Most defects are likely caused by a complex combination of genetic, human behavioral, and environmental factors (Centers for Disease Control and Prevention, 2021, March 2) and research is ongoing to assess how these factors interact to cause birth defects. Research to date indicates that some factors (alcohol, tobacco, or drug use) or maternal conditions (obesity, diabetes, older maternal age, genetic patterns) present during pregnancy may increase the chance of a birth defect. However, it is also important to note that birth defects can occur when none of these conditions or exposures are present. Consulting with a physician and preparing to be as healthy as possible, both before and during pregnancy, may increase the likelihood of having a healthy baby.

APORS is the most complete source of data on birth defects in Illinois. At its inception, APORS relied primarily on reports sent from hospitals to identify cases, but the program has evolved over time and currently uses multiple sources of data and active surveillance methods to identify and verify cases. All Illinois hospitals are mandated to report infants with adverse pregnancy outcomes born to women who are Illinois residents. (Perinatal centers in St. Louis also participate.) Birth, death, and fetal death certificates (maintained by the IDPH's Division of Vital Records) are an additional data source, allowing APORS to identify infants with certain birth defects or other conditions unreported by the hospitals. The IDPH Division of Patient Safety and Quality, which collects patient level discharge data from Illinois acute care hospitals, provides

information about children under the age of 2 with a documented birth defect. This allows APORS to identify children whose birth defect diagnosis was made after their newborn stay, or who were unidentified for other reasons.

Importantly, APORS undertakes systematic active case verification of cases reported to APORS and those identified through other sources. APORS staff members review charts for infants reported with selected serious birth defects. As the charts are reviewed, APORS staff correct and add to information reported by hospitals. An abstractor liaison oversees these activities to ensure the most accurate and complete information is recorded.

Table 1 below describes the activities APORS has undertaken over the years to expand case and birth defect finding since 2002.

Table 1: Projects to Identify Cases and Birth Defects

Birth years	Activity Implemented	Purpose
2002 onwards	Active Case Verification	Identify unreported or misreported diagnoses through review of infant charts using criteria listed in Table 2.
2012 onwards	Abstractor Liaison Oversight	Oversee case abstraction accuracy and completeness.
2013 onwards	Electronic Case Reporting	Systematic prompting of hospitals to report birth defects when documented on the birth certificate.
2013 onwards	Hospital Discharge Case Finding	Identify birth defects noted in review of hospital discharge data, but not reported to APORS.
2015, 2016	Rapid Case Identification	Identify conditions potentially related to the Zika virus, including brain and eye anomalies, neural tube defects, and arthrogryposis.

Since systematic active case verification began in 2002, there have been modifications to the criteria that establish which charts are reviewed. Details are given in Table 2.

Table 2: Criteria that Determine Which Charts are Reviewed

Birth years	Chart Review Criteria
2002 -2007	• one or more birth defects, • very low birth weight (< 1500 g), • exposure to alcohol, • a diabetic mother, • a disturbance in neonatal tooth eruption, or • death before discharge.
2008-2012	• selected birth defects including those covered in this report, • exposure to alcohol. • a diabetic mother. • a disturbance in neonatal tooth eruption, or • death before discharge.
2013 onwards	• selected birth defects including those covered in this report, • exposure to alcohol, • a disturbance in neonatal tooth eruption.

Each of these conditions has a high likelihood of being associated with one or more birth defects. As a result, the use of active case verification allows APORS to identify and verify more birth defects each year when compared to past years when this methodology was not used.

This increase in the number of verified diagnoses is the combined result of a number of factors:

i. The APORS chart review takes place several months after discharge, and additional diagnoses have been made since the children were reported to APORS.
ii. The diagnostic test results are placed in the chart after discharge and are not seen by the reporting hospital staff.
iii. Hospital reporting staffs are likely to report one or two major birth defects for each child and may not include associated, but less significant birth defects.

However, at times defects may be reported that do not meet APORS' criteria because:

i. a clinical diagnosis was suggested and reported, which was later ruled out by a diagnostic test;
ii. some defects are only collected in special circumstances that were not met upon chart review;
iii. a diagnosis was reported in general terms (e.g., heart anomaly) when a more specific diagnosis was available; or
iv. the hospital report was in error, and there was no evidence in the chart for the reported diagnoses.

APORS case finding is an ongoing process; children with birth defects identified during the newborn stay are added for previous years whenever they are found. This report presents birth

defect rates among newborns and infants up to 2 years of age, born between 2002 and 2018. The rates are presented for two distinct time periods reflecting the evolution of data collection methodologies over time. The first time period spans 2002 to 2012, during which active case verification was first introduced and utilized. The second time period spans 2013 to 2018 during which several changes took place. In 2013, APORS fully implemented an electronic reporting system for hospitals which aides in reporting by systematically flagging certain reportable conditions, including birth defects, when noted on the birth certificate. In addition to ongoing active case verification, hospital discharge data were used to ascertain new cases and the International Classification of Diseases, Tenth Revision Clinical Modification (ICD-10-CM) coding was implemented to categorize birth defects. In cooperation with CDC, rapid case identification techniques were employed (2015 and 2016 births) to quickly ascertain certain birth defects potentially related to the Zika virus. Because of the new data collection activities implemented during the 2013 to 2018 time period, rates are not comparable to the 2002 to 2012 period when active case verification alone was used.

In addition to trends for the two time periods, this report as also provides and compares current birth defects prevalence for the geographies of Illinois and Chicago alone for the year 2018.

METHODS

Calculation and Interpretation of Rates and Confidence Intervals

Thirty-five categories of birth defects are included in this study. A listing of the International Classification of Diseases – Ninth and Tenth Revisions Clinical Modification (ICD-9-CM and ICD-10-CM) codes for the selected birth defects is provided in Appendix A, together with a brief description of each birth defect.

Annual incidence rates (per 10,000 live births) for selected congenital anomalies identified during the newborn hospital stay up to 2 years of age or associated with a fetal death were calculated as:

$$\frac{\text{Number of infants and fetuses with selected congenital anomaly}}{\text{number of live births}} \times 10{,}000$$

The numbers of live births were obtained from the IDPH's master birth files. Occurrence of a specific birth defect is assumed to be a rare event, therefore following a Poisson distribution. Exact confidence intervals were calculated for each rate (Hardeo & Khurshid, 1993). Where there are a large number of birth defect cases, the confidence interval is narrow, indicating that

the rate is stable. Where there are few birth defect cases, the confidence interval becomes very wide, indicating that the rate is not very stable and a small change in the number of infants born with the specific birth defect could result in a large change in the rate.

To compare two rates, it is important to look not just at their value, but also their confidence intervals. As a conservative approximation, if two confidence intervals overlap, then there is no evidence that the two rates are really different. If two confidence intervals do not overlap, then the rates are said to be statistically different. In this report, 95% confidence intervals are used; where the confidence intervals do not overlap, the rates are statistically different at the 5% level ($p < 0.05$).

Analysis of Trends

Trends in Illinois birth defect rates for 2002-2018 were modeled using a log-linear regression model (which is appropriate for data following a Poisson distribution). Analyses were performed using the Joinpoint Regression Program (Version 4.8.0.1, April 2020, Statistical Research and Applications Branch, National Cancer Institute). This software compares a linear model with a single slope to linear models with different slopes joined by one or more join-points. The model tests whether the slope(s) are significantly different from 0 (whether there is a change over time) and whether any change in slope between two segments is statistically significant.

Multiple Comparisons

Because this report examines a large number of birth defects, the corresponding statistical tests are subject to the "multiple comparison problem." In this report, no explicit corrections were made for multiple comparisons because the focus was to detect trends, not compare trends; instead, exact probabilities are reported when discussing trends. The smaller the reported probability, the more likely it is that the difference is not simply the result of chance.

FINDINGS

Rates of Birth Defects for Illinois and Chicago, 2018

Birth defect rates for selected categories among Illinois and Chicago newborns in 2018 are presented in tables 4 and 5. Rates for Chicago are similar to those for Illinois as a whole. Statistically significant differences were not seen overall or for individual defect categories.

Trend Analysis

Table 3 shows statistically significant trends were found for 10 birth defects (See Table 3). Six defects showed significance from 2002 to 2012, and three defects displayed significance during the 2013 to 2018 period. Since the latter time period is shorter and reflects a time of changes in data collection methodologies, the results may be less reliable. One defect (hypospadias) showed a significant trend during both time periods. Table 3 includes columns with average annual percentage change that gives an estimate of how quickly the rate changed over time for each time period. For example, the rate of anotia/microtia was increasing by an average of 8.3% each year during 2002 and 2012 and 3.3% each year between 2013 and 2018. The change over the second time period was not statistically significant, however.

Figures 1 to 9 show the rates over time for birth defects during the two time periods of 2002-2012 and 2013-2018. Rates for Down syndrome are also plotted by maternal age group in Figure 10. Regression lines are plotted for each birth defect and time period. The regression lines are usually log-linear but may be made up of several straight line segments with different slopes. Although examination of the graphs may show some birth defects with a marked slope, the small number of cases means that the slope is not statistically significantly different from horizontal (no change with time).

Several defects may show trending in differing directions for the two time periods. This may be caused by several factors including instability of the estimates of trend for the latter period due to the small number of time points between 2013 and 2018, or changes in data collection methods during that period. Some estimates may have been affected by the change from the ICD-9 to ICD-10 coding scheme used by hospitals and officially enacted in October 2015 (Salemi *et al.*, 2019). Finally, it is also possible the trends are sound and will remain as additional years are examined in future trend studies.

Illinois data do not include birth defects that are diagnosed prenatally where the fetuses are subsequently terminated. This means that the Illinois observed incidence rates for conditions where terminations occur are lower than they should be. When examining trends, this will not affect the trend, provided the termination rate does not vary over time.

Table 3. Birth Defects Showing a Significant Trend in Incidence Rate Between 2002 and 2012 or Between 2013 and 2018

Selected Birth Defect	Significance of trend (P-value)	Average annual % change between 2002 and 2012	Significance of trend (P-value)	Average annual % change between 2013 and 2018
Anotia/Microtia	0.00	8.3	not significant	3.3
Ventricular septal defect	0.02	2.8	not significant	5.2
Endocardial cushion defect	0.00	3.7	not significant	-0.3
Tricuspid valve atresia, stenosis and other anomalies	0.00	5.8	not significant	-23.3
Coarctation of aorta	not significant	1.9	0.00	9.3
Aortic valve stenosis	not significant	2.2	0.00	22.2
Hypoplastic left heart syndrome	not significant	-0.1	0.00	17.3
Rectal and large intestinal atresia/stenosis	0.00	-1.8	not significant	2.2
Renal agenesis/hypoplasia	0.00	7.0	not significant	8.2
Hypospadias	0.00	2.7	0.00	6.9

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2021

Discussion of Illinois Results

The rate of anotia/microtia increased significantly from 2002 to 2012. Between 2013 and 2018 the rate increased as well but was not significant. The CDC reports that for most babies, the causes of anotia/microtia are not known. However, use of the medication isotretinoin (Accutane®) during pregnancy has been identified as one cause, and genetic mutations or complex genetic environmental factors may also elicit these defects. Women with (pre-existing) diabetes and women who consume a low carbohydrate diet during pregnancy may also be at an increased risk of having a baby with anotia/microtia (CDC, 2021, March 25).

A recent study covering 30 birth defects surveillance systems in the United States, including Illinois, found the highest prevalence rates of anotia/microtia among infants born to Hispanic

women when compared to other racial/ethnic groups. Prevalence rates were also found to be higher among infants born to non-Hispanic Asian Indian/Alaska Native and non-Hispanic Asian/Pacific Islander women when compared to non- Hispanic Black and White women (Stallings *et al.*, 2018). The same study also noted a higher prevalence of these defects among infants of women ages 40 and older when compared with other age groups. In Illinois, while the proportion of births to Hispanic women has decreased slightly during the study period (2002-2018), the proportion of births to non-Hispanic Asian/Pacific Islander women increased from 4.6% in 2002 to 6.7% in 2018. Also, the proportion of births to women 40 or more years of age has increased from 2.4% in 2002 to 3.5% in 2018. The increases in these populations may partly explain the rising rates of anotia/microtia in Illinois. The change from ICD-9 to ICD-10 coding may also explain rising rates after 2015 (Salemi *et al.*, 2019).

Significant increases were seen in three cardiovascular defects from 2002 to 2012, including ventricular septal defect, endocardial cushion defect, tricuspid atresia/stenosis, and other anomalies. During the period of 2013 to 2018, significant increases were noted for an additional three defects, including coarctation of aorta, aortic valve stenosis, and hypoplastic left heart syndrome. Heart defects are the most common type of birth defects, and the prevalence of some types of heart defects are increasing while the rates of others remain steady (CDC, 2021, April 6). In Illinois, APORS hospital discharge case-finding and universal pulse oximetry screening for critical congenital heart defects may explain some of the increase seen during 2013-2018. Of note is a marked (nonsignificant) decrease in prevalence of tricuspid atresia/stenosis and other anomalies during the 2013 to 2018 time period which is in line with findings from Salemi *et al.* who reported an immediate decrease in the prevalence for this defect as a result of the conversion from ICD-9 to ICD-10 coding.

Rectal and large intestinal atresia/stenosis rates decreased significantly between 2002 and 2012. However, between 2013 and 2018 the rates showed a nonsignificant increase. Gastrointestinal defects may be associated with chromosomal anomalies including Down syndrome, which also showed a slight nonsignificant decrease during the first time period. During the latter time period it is possible that hospital discharge case finding contributed to the increased rates.

Renal agenesis/hypoplasia rates increased significantly from 2002 to 2012. Between 2013 and 2018 the rate also increased, but not significantly. These conditions are largely genetic in nature. The increase in rates is in part due to improved chart abstraction, with the identification of additional cases of hypoplasia. Additionally, hospital discharge case-finding and the conversion from ICD-9 to ICD-10 during the latter period may also explain an increase in rates.

Hypospadias rates increased during both periods of study (2002 to 2012 and 2013 to 2018). The specific causes of most cases of hypospadias remain unknown, although certain factors have been identified that may increase the risk of having a child with this condition. These include older maternal age, obesity, use of assisted reproductive technology (ART), and use of hormones (CDC, 2021, April 8). In Illinois, the proportion of births in Illinois to women ages 35 and older has increased from 14.5% in 2002 to 20.1% in 2018. Use of ART has increased steadily in the United States since it was introduced in the early 1980s, and Illinois is one of a number of states consistently reporting the highest use of ART nationally (Sunderam *et al.*, 2017; Wright *et al.*, 2005). Multistate studies using the Pregnancy Risk Assessment Monitoring System and U.S. National Vital Statistics System natality data have revealed an increase in the prevalence of pre-pregnancy obesity nationwide during the years 2003 to 2015 (Deputy *et al.*, 2018).

While overall rates of Down syndrome are higher as maternal age increases (see Figure 10), no significant changes were seen in the rates over either study periods for this report.

It is noteworthy to mention that the (nonsignificant) increase in prevalence rates of microcephalus starting in 2015 (see Figure 1) correlates directly with APORS' participation in CDC's enhanced rapid data collection efforts (active case finding and standard case definition) used to gather information nationwide about birth defects possibly related to the Zika virus. A regression line is not shown for this defect for the time period of 2013 to 2018.

Birth Defect Rates for Selected Categories Among Illinois and Chicago Newborns

2018

Table 4. Number and Rate of Selected Birth Defects for 2018, Illinois

Selected Birth Defects Groups	N	Rate[1]	95% CI[2]	
A. Central Nervous System				
Anencephalus	17	1.2	(0.7,	1.9)
Spina bifida	50	3.5	(2.6,	4.6)
Encephalocele	14	1.0	(0.5,	1.6)
Microcephalus	356	24.6	(22.1,	27.3)
Total Selected CNS Defects	*437*	*30.2*	*(27.4,*	*33.1)*
B. Eye				
Anophthalmia/Microphthalmia	29	2.0	(1.3,	2.9)
Congenital cataract	14	1.0	(0.5	1.6)
Total Selected Eye Defects	*43*	*3.0*	*(2.1,*	*4.0)*
C. Ear				
Anotia/Microtia	32	2.2	(1.5,	3.1)
D. Cardiovascular				
Common truncus	9	0.6	(0.3,	1.2)
Transposition of great vessels	62	4.3	(3.3,	5.5)
Tetralogy of Fallot	82	5.7	(4.5,	7.0)
Ventricular septal defect	770	53.2	(49.5,	57.1)
Double outlet right ventricle	31	2.1	(1.5,	3.0)
Endocardial cushion defect	92	6.4	(5.1,	7.8)
Pulmonary valve atresia and stenosis	126	8.7	(7.2,	10.4)
Tricuspid valve atresia, stenosis and other anomalies	22	1.5	(1.0,	2.3)
Ebstein anomaly	10	0.7	(0.3,	1.3)
Aortic valve stenosis	53	3.7	(2.7,	4.8)
Hypoplastic left heart syndrome	52	3.6	(2.7,	4.7)
Coarctation of aorta	98	6.8	(5.5,	8.2)
Total Selected Cardiovascular Defects	*1,407*	*97.1*	*(92.1,*	*102.4)*
F. Orofacial				
Cleft palate without cleft lip	94	6.5	(5.2,	7.9)
Cleft lip with and without cleft palate	137	9.5	(7.9,	11.2)
Choanal atresia	17	1.2	(0.7,	1.9)
Total Selected Orofacial Defects	*248*	*17.1*	*(15.1,*	*19.4)*
G. Gastrointestinal				
Esophageal atresia/Tracheoesophageal fistula	37	2.6	(1.8,	3.5)
Rectal and large intestinal atresia/stenosis	69	4.8	(3.7,	6.0)
Biliary atresia	8	0.6	(0.2,	1.1)
Total Selected Gastrointestinal Defects	*114*	*7.9*	*(6.5,*	*9.5)*

Selected Birth Defects Groups	N	Rate[1]	95% CI[2]	
H. Genitourinary				
Renal agenesis/hypoplasia	128	8.8	(7.4,	10.5)
Bladder exstrophy	5	0.3	(0.1,	0.8)
Hypospadias	501	34.6	(31.6,	37.8)
Total Selected Genitourinary Defects	*634*	*43.8*	*(40.4,*	*47.3)*
I. Musculoskeletal				
Reduction deformity, upper and lower limbs	65	4.5	(3.5,	5.7)
Gastroschisis	49	3.4	(2.5,	4.5)
Omphalocele	30	2.1	(1.4,	3.0)
Diaphragmatic hernia	37	2.6	(1.8,	3.5)
Total Selected Musculoskeletal Defects	*181*	*12.5*	*(10.7,*	*14.5)*
J. Chromosomal				
Trisomy 13 (Patau syndrome)	17	1.2	(0.7,	1.9)
Trisomy 21 (Down syndrome)	204	14.1	(12.2,	16.2)
Trisomy 18 (Edward syndrome)	40	2.8	(2.0,	3.8)
Total Selected Chromosomal Defects	*261*	*18.0*	*(15.9*	*20.3)*
Total All Selected Defects	***3,357***	***231.8***	***(224.0,***	***239.8)***

[1] Rate per 10,000 live births
[2] 95 percent confidence interval for rate

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2021

Table 5. Number and Rate of Selected Birth Defects for 2018, Chicago

Selected Birth Defects Groups	N	Rate[1]	95% CI[2]
A. Central Nervous System			
Anencephalus	5	1.4	(0.5, 3.3)
Spina bifida	9	2.6	(1.2, 4.9)
Encephalocele	4	1.1	(0.3, 2.9)
Microcephalus	110	31.6	(25.9, 38.0)
Total Selected CNS Defects	*128*	*36.7*	*(30.6 43.7)*
B. Eye			
Anophthalmia/Microphthalmia	4	1.1	(0.3, 2.9)
Congenital cataract	3	0.9	(0.2, 2.5)
Total Selected Eye Defects	*7*	*2.0*	*(0.8, 4.1)*
C. Ear			
Anotia/Microtia	11	3.2	(1.6, 5.6)
D. Cardiovascular			
Common truncus	3	0.9	(0.2, 2.5)
Transposition of great vessels	18	5.2	(3.1, 8.2)
Tetralogy of Fallot	18	5.2	(3.1, 8.2)
Ventricular septal defect	183	52.5	(45.2, 60.7)
Double outlet right ventricle	7	2.0	(0.8, 4.1)
Endocardial cushion defect	17	4.9	(2.8, 7.8)
Pulmonary valve atresia and stenosis	31	7.7	(5.2, 11.0)
Tricuspid valve atresia, stenosis and other anomalies	9	2.6	(1.2, 4.9)
Ebstein anomaly	4	1.1	(0.3, 2.9)
Aortic valve stenosis	10	2.9	(1.4, 5.3)
Hypoplastic left heart syndrome	13	3.7	(2.0, 6.4)
Coarctation of aorta	21	6.0	(3.7, 9.2)
Total Selected Cardiovascular Defects	*334*	*95.8*	*(85.8, 106.7)*
F. Orofacial			
Cleft palate without cleft lip	21	6.0	(3.7, 9.2)
Cleft lip with and without cleft palate	33	9.5	(6.5, 13.3)
Choanal atresia	4	1.1	(0.3, 2.9)
Total Selected Orofacial Defects	*58*	*16.6*	*(12.6, 21.5)*
G. Gastrointestinal			
Esophageal atresia/Tracheosophageal fistula	11	3.2	(1.6, 5.6)
Rectal and large intestinal atresia/stenosis	21	6.0	(3.7, 9.2)
Biliary atresia	2	0.6	(0.1, 2.2)
Total Selected Gastrointestinal Defects	*34*	*9.8*	*(6.8 13.6)*

Selected Birth Defects Groups	N	Rate[1]	95% CI[2]
H. Genitourinary			
Renal agenesis/hypoplasia	39	11.2	(8.0, 15.3)
Bladder exstrophy	2	0.6	(0.1, 2.1)
Hypospadias	119	34.1	(28.3, 40.9)
Total Selected Genitourinary Defects	*160*	*45.9*	*(39.1, 53.6)*
I. Musculoskeletal			
Reduction deformity, upper and lower limbs	19	5.5	(3.3, 8.5)
Gastroschisis	14	4.0	(2.2, 6.7)
Omphalocele	7	2.0	(0.8, 4.1)
Diaphragmatic hernia	14	4.0	(2.2, 6.7)
Total Selected Musculoskeletal Defects	*54*	*15.5*	*(11.6, 20.2)*
J. Chromosomal			
Trisomy 13 (Patau syndrome)	4	1.1	(0.3, 2.9)
Trisomy 21 (Down syndrome)	41	11.8	(8.4, 16.0)
Trisomy 18 (Edward syndrome)	8	2.3	(1.0, 4.5)
Total Selected Chromosomal Defects	*53*	*15.2*	*(11.4, 19.9)*
Total All Selected Defects	***839***	***240.7***	***(224.7, 257.6)***

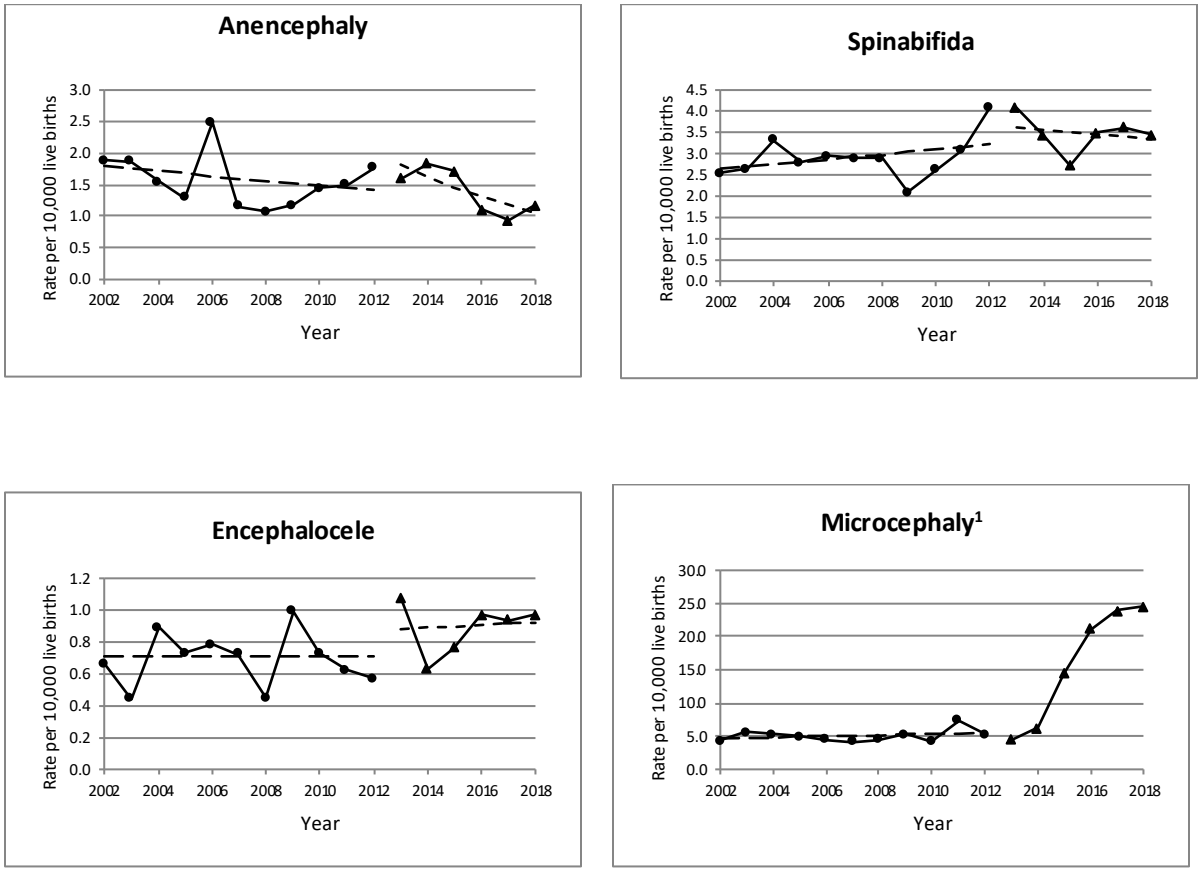
[1] Rate per 10,000 live births
[2] 95 percent confidence interval for rate

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2021

Trends in Birth Defect Rates for Selected Categories Among Illinois Newborns

2002-2018

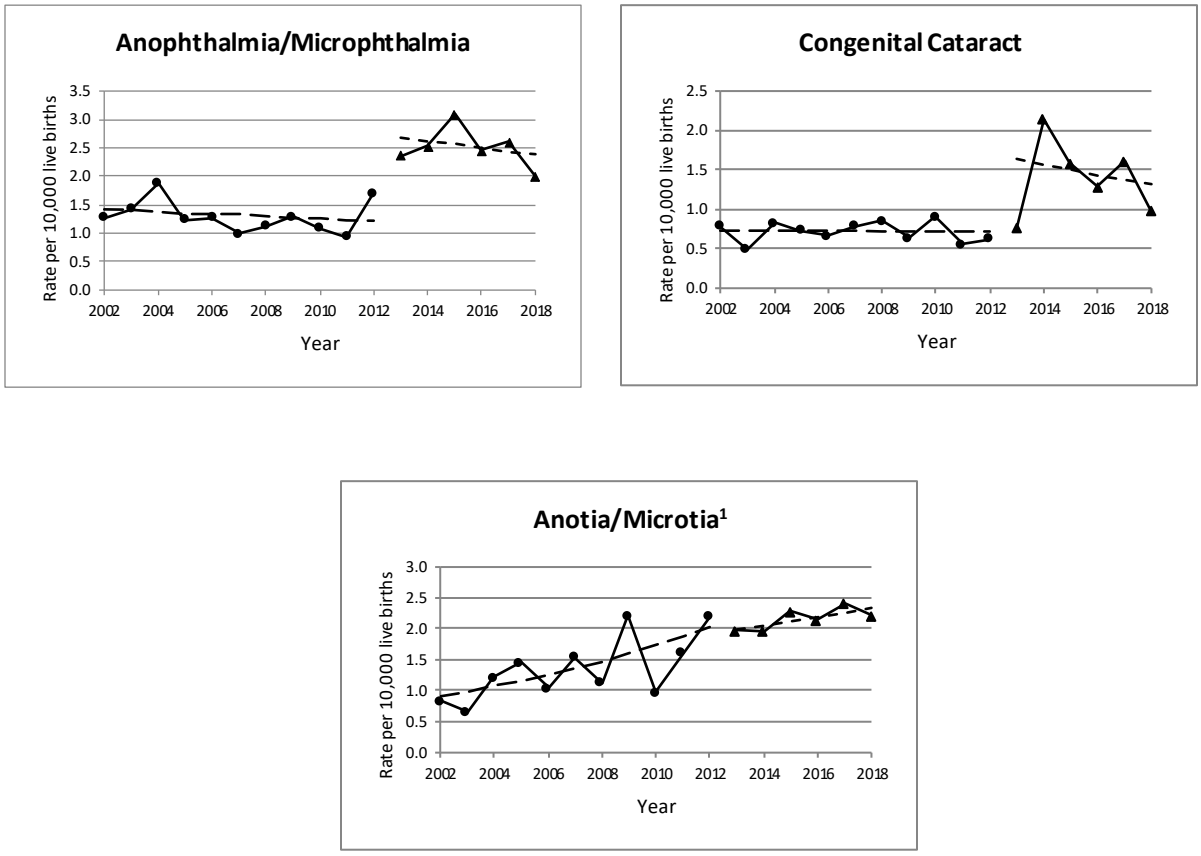
Figure 1. Trends in the Reported Prevalence Rates of Neural Tube Defects per 10,000 Live Births 2002-2018



[1] A trend line is not presented for microcephaly, because the observed increase is likely due to extra efforts to obtain microcephaly during 2016 and 2017 due to its suspected association with Zika virus exposure. See page nine for details.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2021

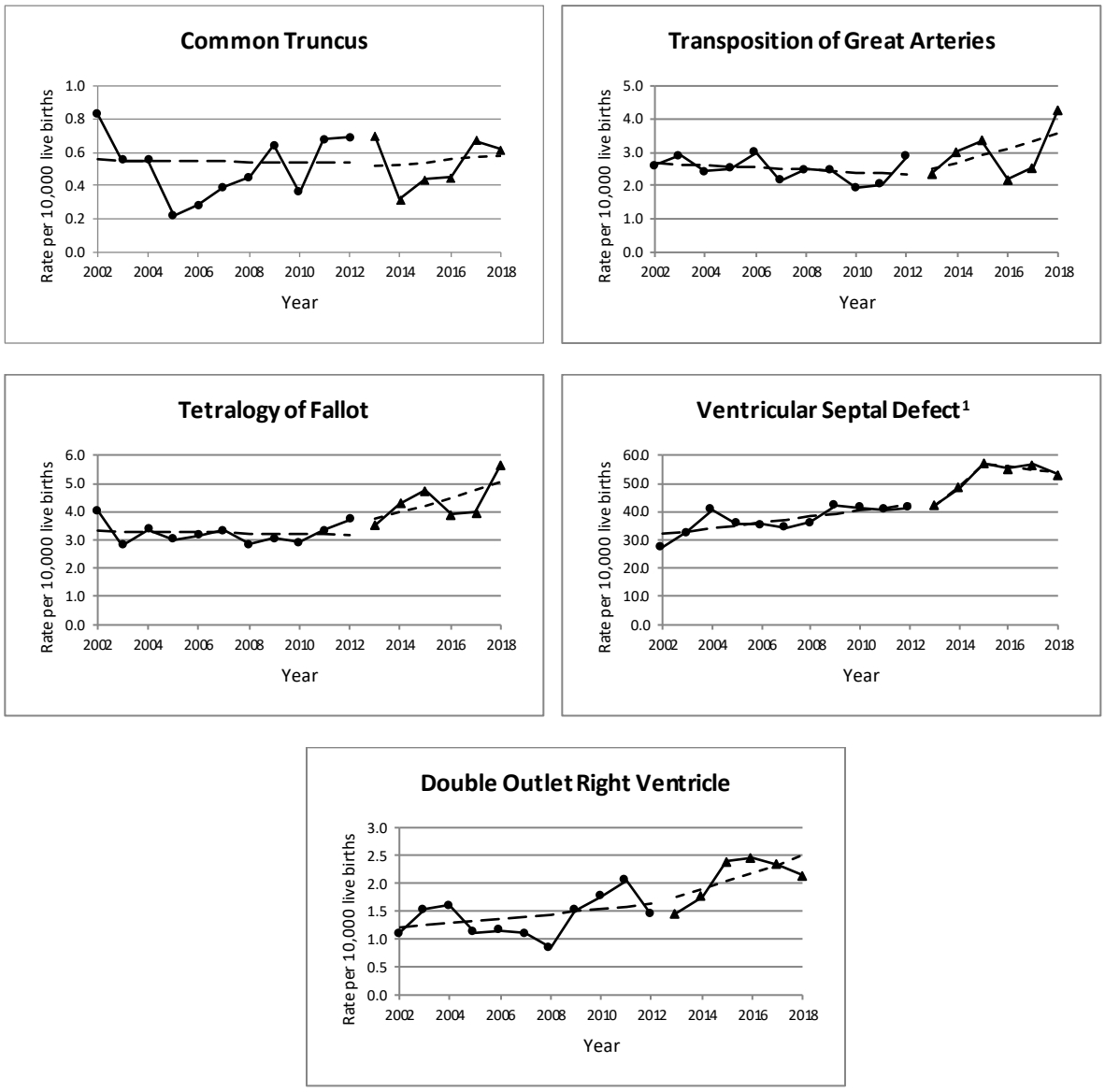
Figure 2. Trends in the Reported Prevalence Rates of Eye and Ear Defects per 10,000 Live Births 2002-2018



[1]Trend is significant for 2002-2012 but not for 2013-2018; See Table 3 for details.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2021

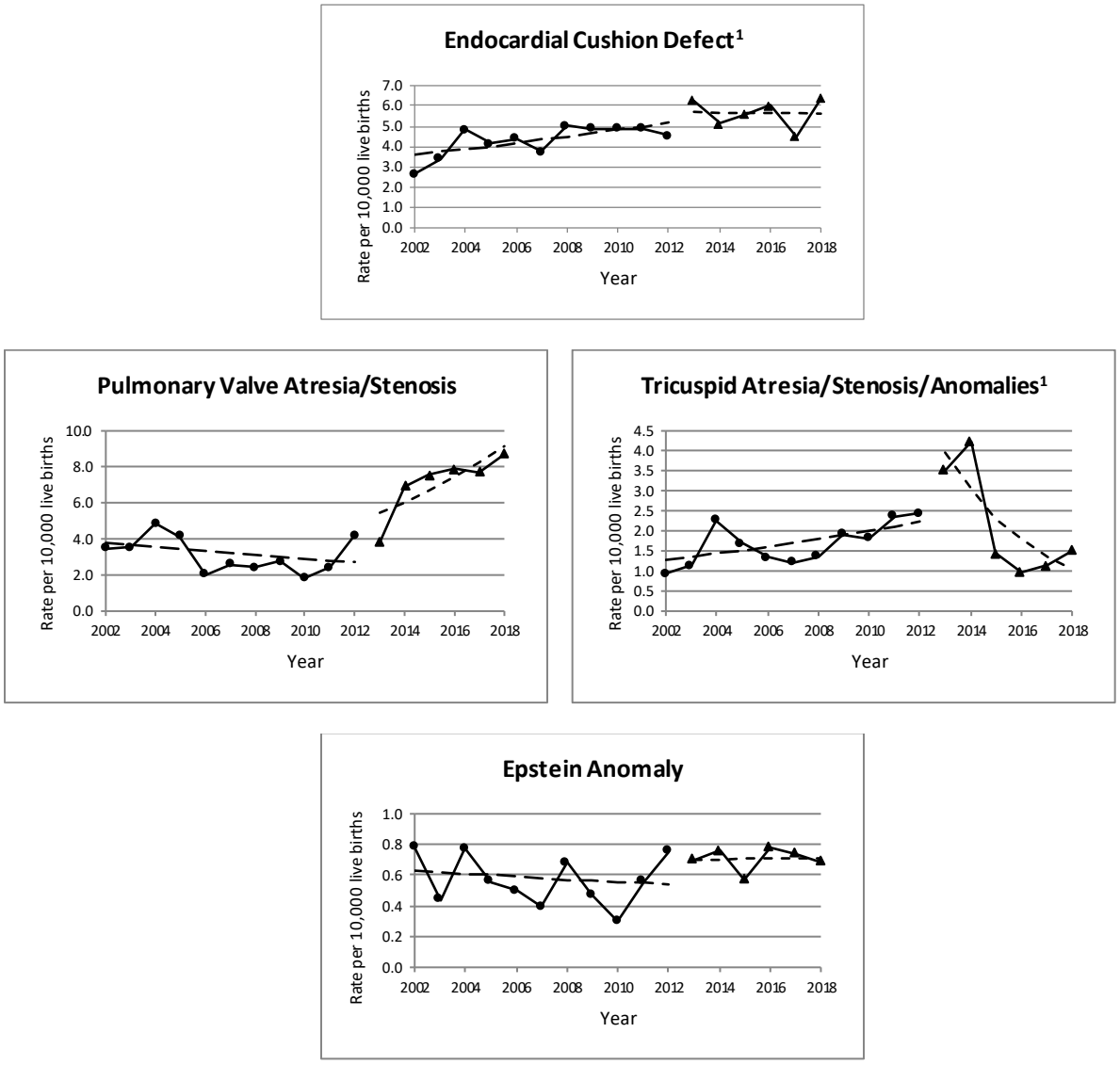
Figure 3A. Trends in the Reported Prevalence Rates of Cardiac Defects per 10,000 Live Births 2002-2018



[1]Trend is significant for 2002-2012 but not for 2013-2018; See Table 3 for details.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2021

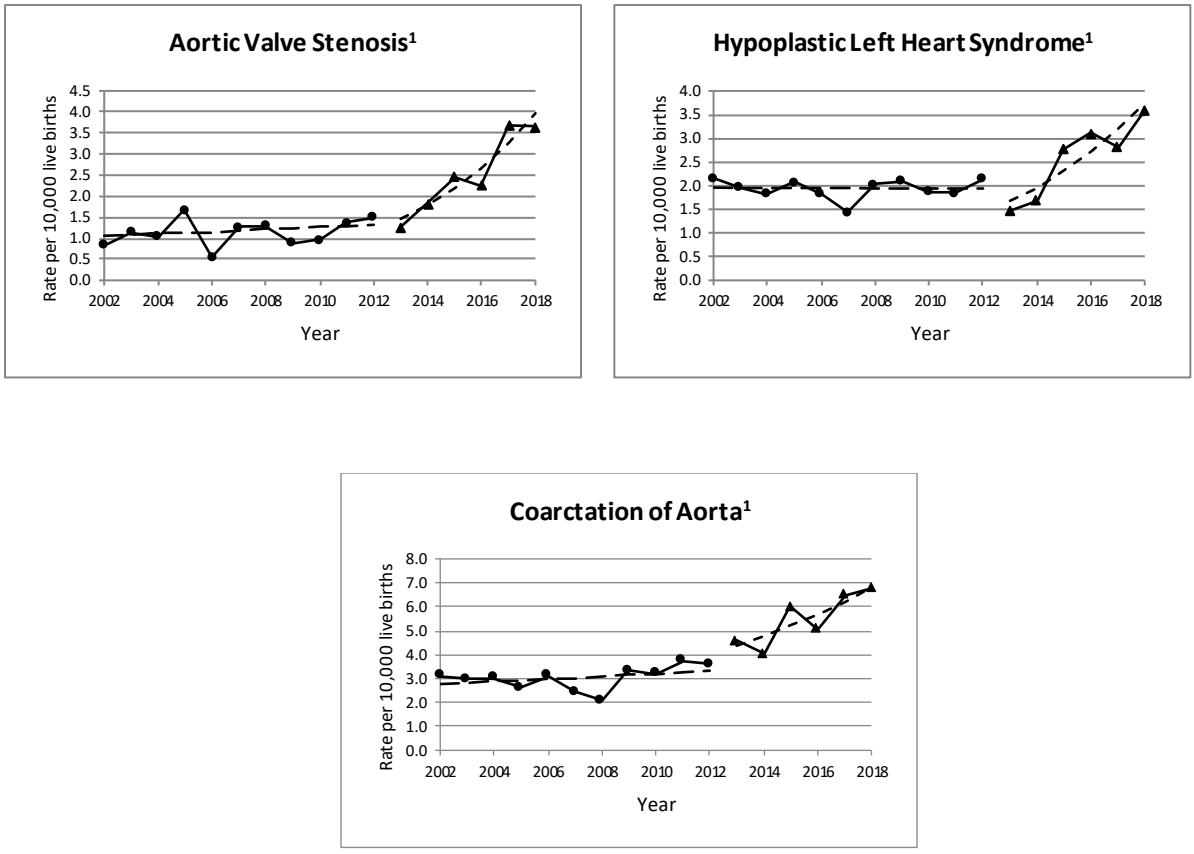
Figure 3B. Trends in the Reported Prevalence Rates of Cardiac Defects per 10,000 Live Births 2002-2018



[1]Trend is significant for 2002-2012, but not for 2013-2018; See Table 3 for details.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2021

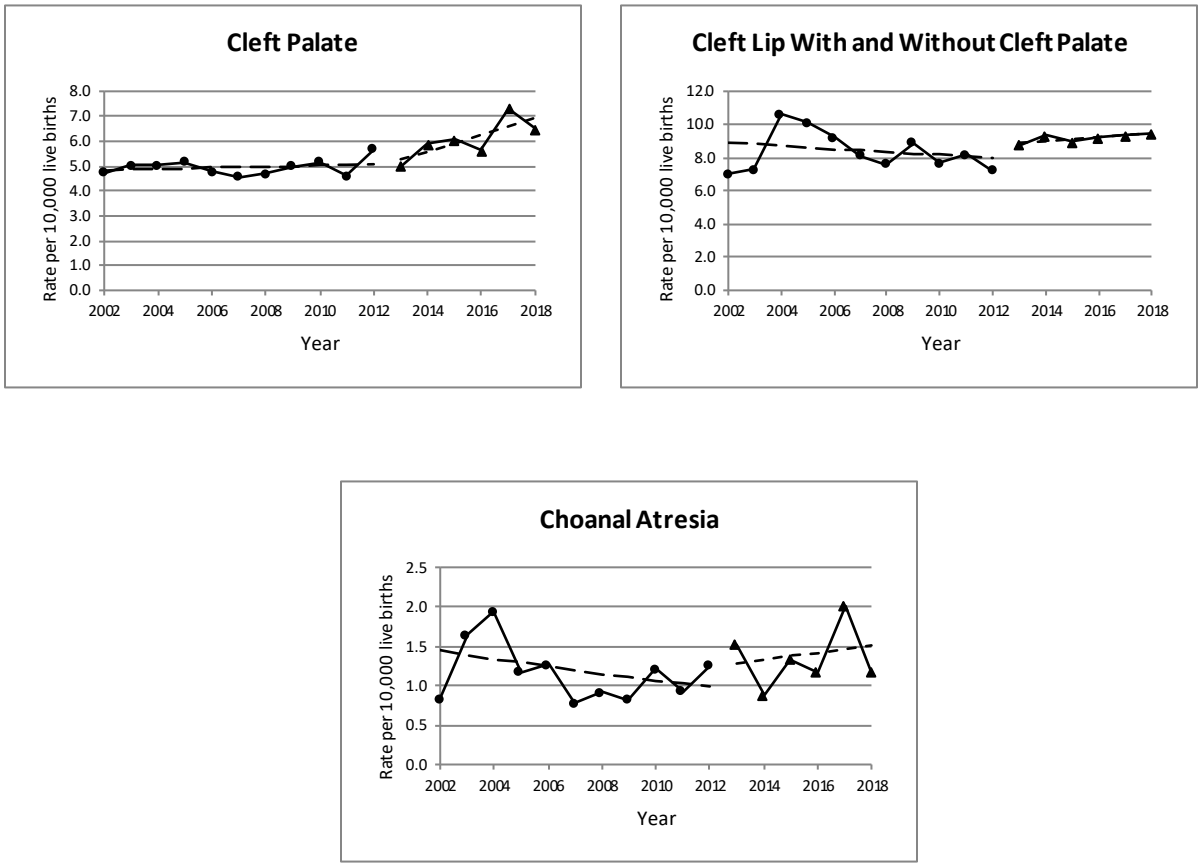
Figure 4. Trends in the Reported Prevalence Rates of Circulatory Defects per 10,000 Live Births 2002-2018



[1]Trend is significant for 2013-2018 only; See Table 3 for details.

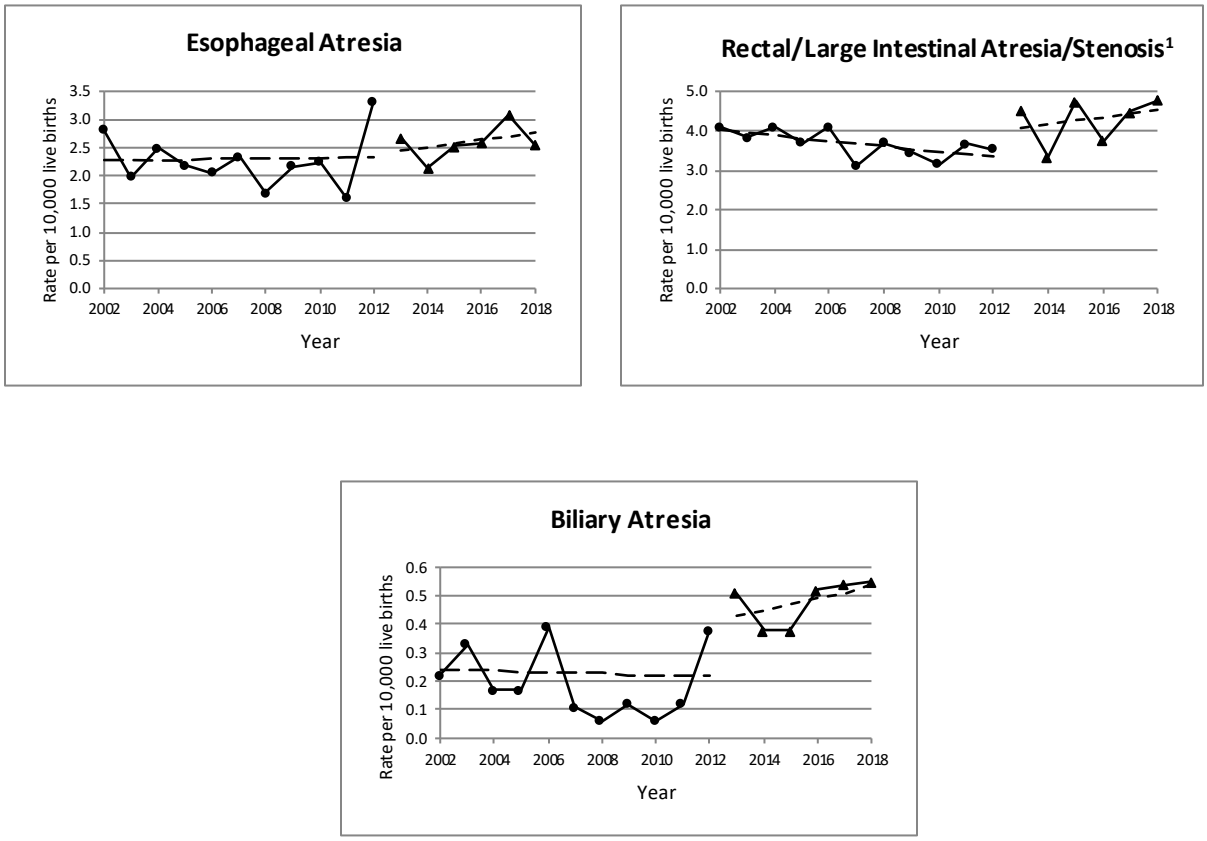
Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2021

Figure 5. Trends in the Reported Prevalence Rates of Respiratory and Oral Defects per 10,000 Live Births 2002-2018



Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2021

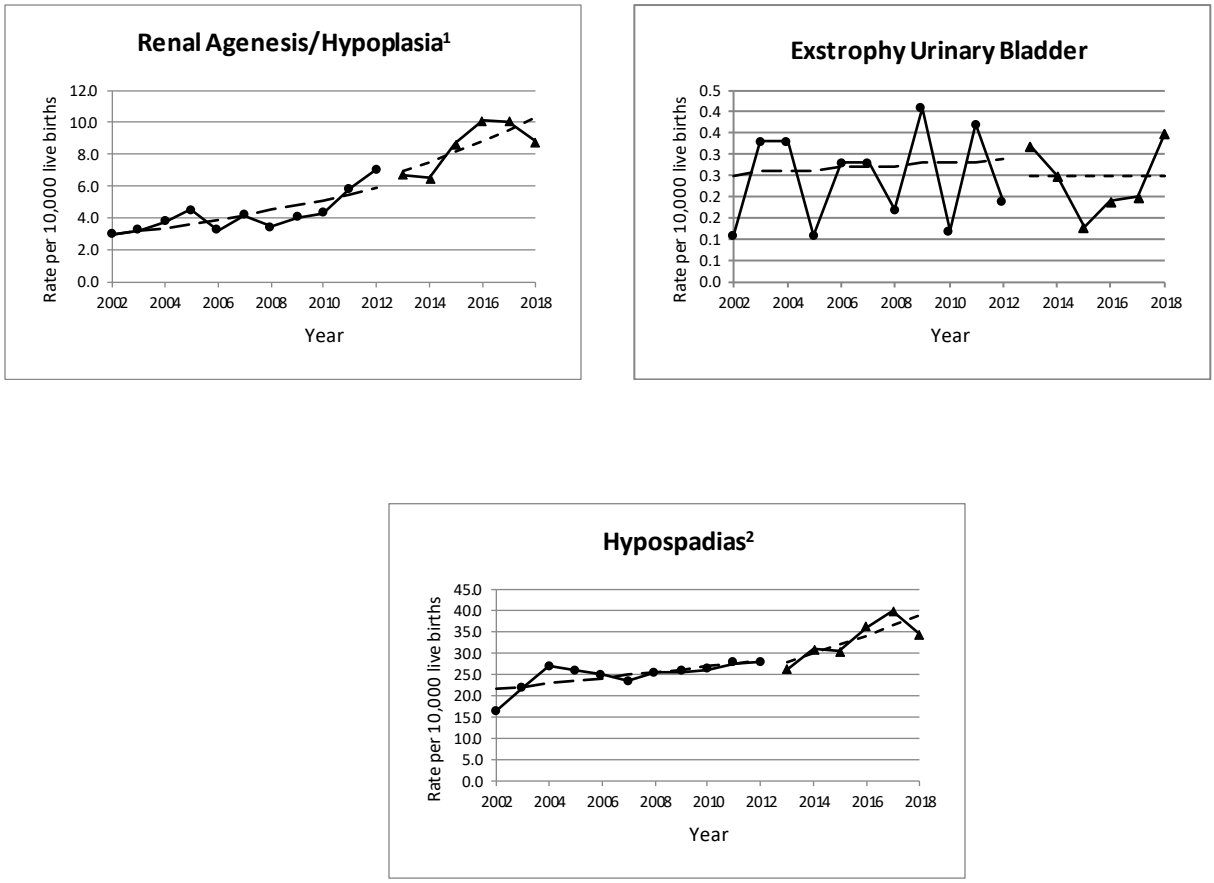
Figure 6. Trends in the Reported Prevalence Rates of Gastrointestinal Defects per 10,000 Live Births 2002-2018



[1]Trend is significant for 2002-2012 but not for 2013-2018; See Table 3 for details.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2021

Figure 7. Trends in the Reported Prevalence Rates of Genitourinary Defects per 10,000 Live Births 2002-2018

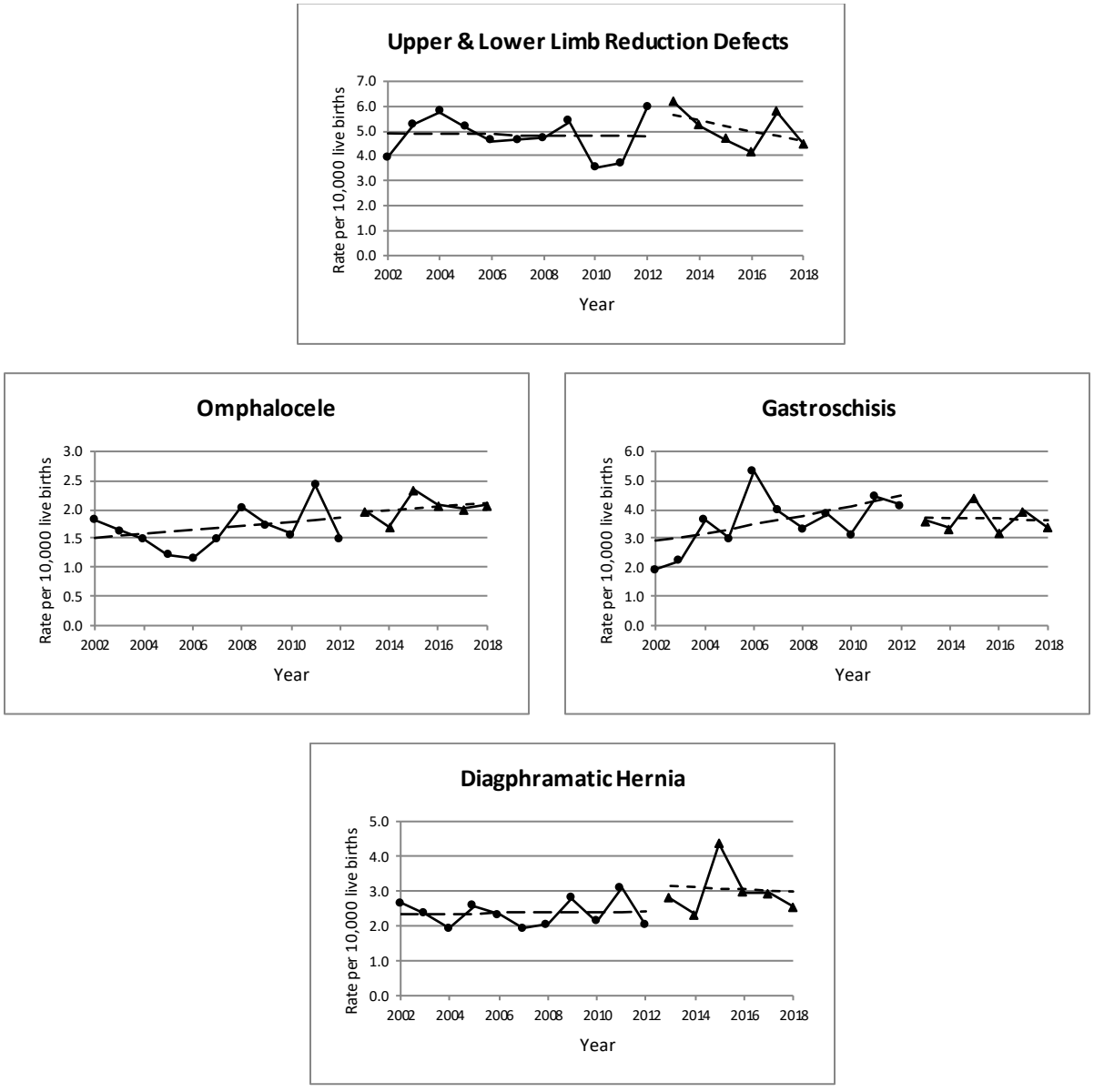


[1]Trend is significant for 2002-2012 but not for 2013-2018; See Table 3 for details.

[2]Trend is significant for 2002-2012 and 2013-2018; See Table 3 for details.

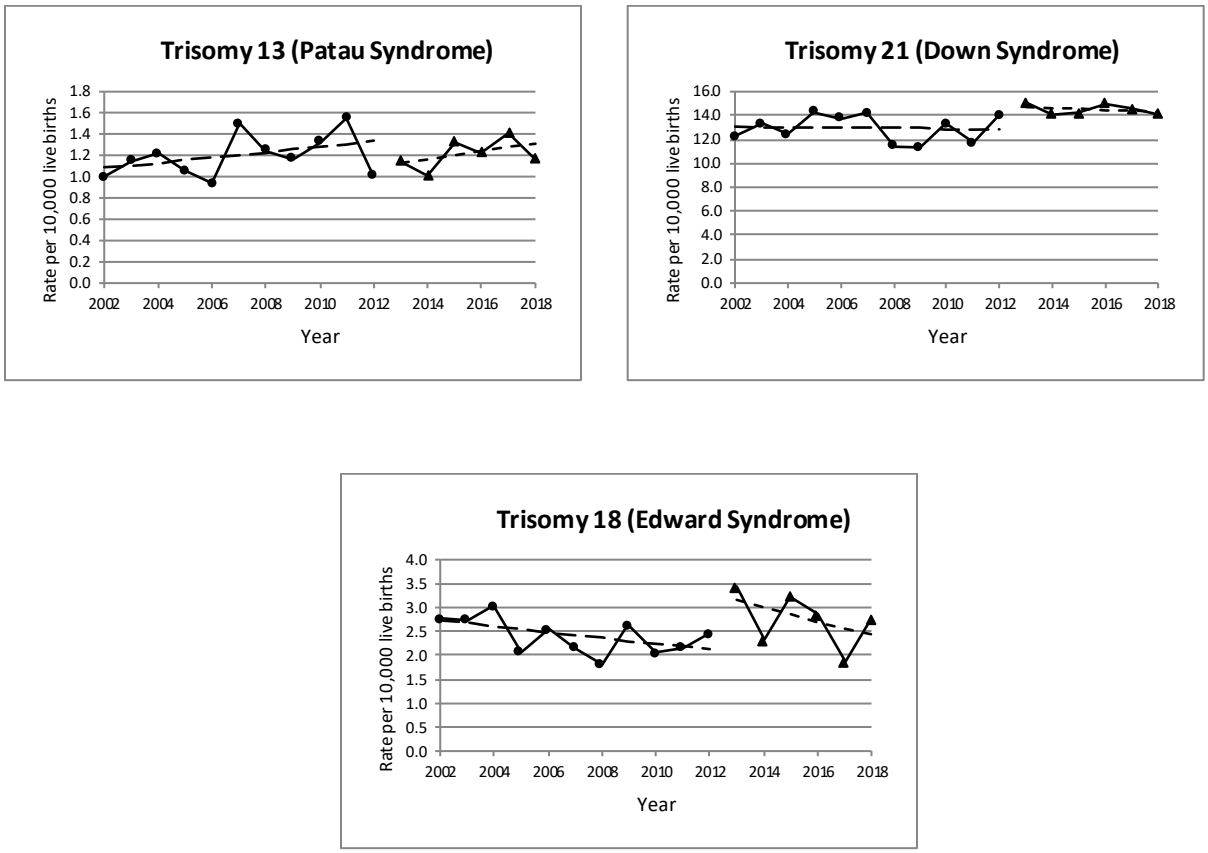
Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2021

Figure 8. Trends in the Reported Prevalence Rates of Musculoskeletal Defects per 10,000 Live Births 2002-2018



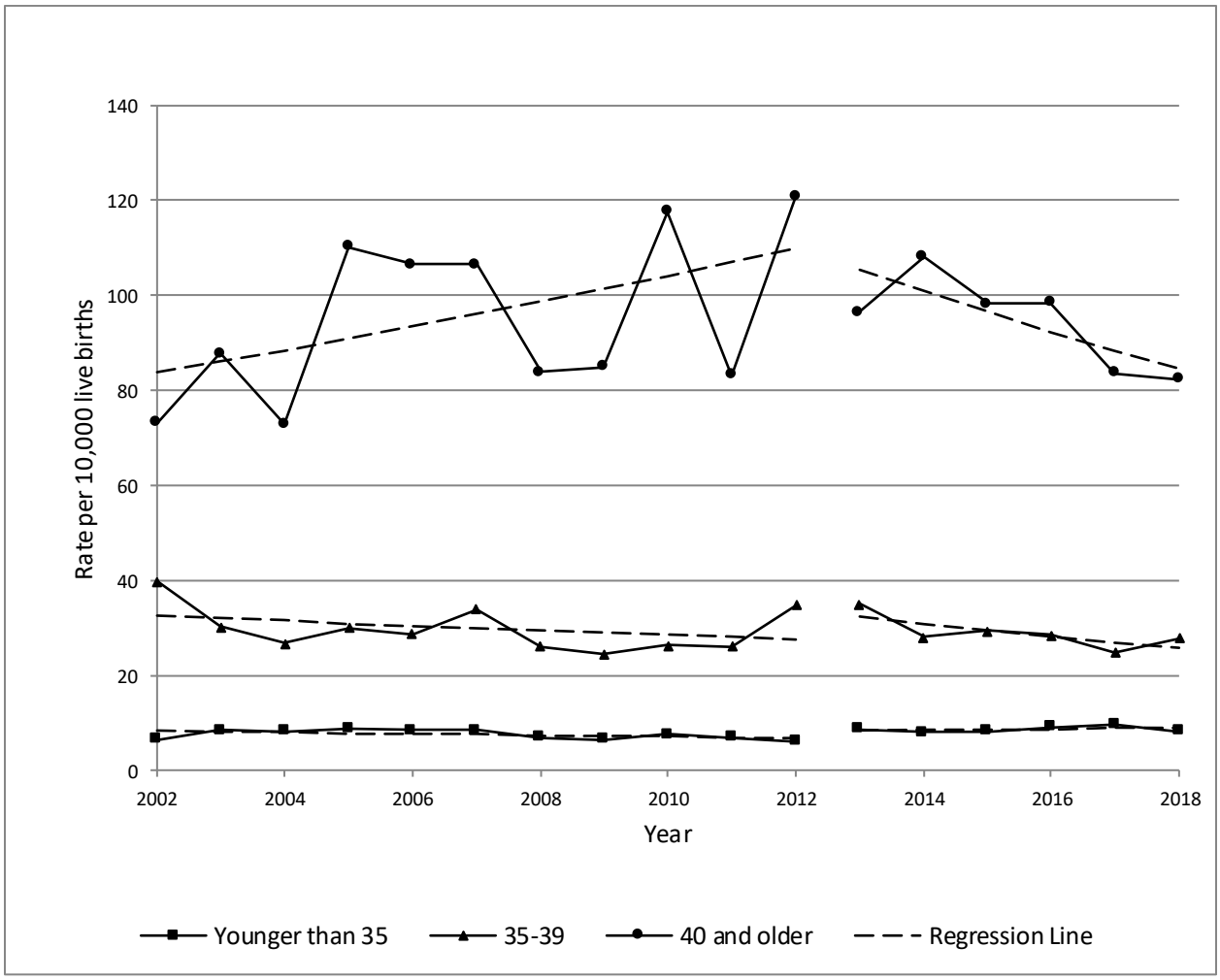
Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2021

Figure 9. Trends in the Reported Prevalence Rates of Chromosomal Defects per 10,000 Live Births 2002-2018



Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2021

Figure 10. Trends in Reported Prevalence of Trisomy 21 (Down Syndrome) by Maternal Age at Delivery, 2002-2018



Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2021

REFERENCES

Centers for Disease Control and Prevention (CDC). https://www.cdc.gov/ncbddd/birthdefects/facts.html Accessed March 2, 2021.

CDC.https://www.cdc.gov/ncbddd/birthdefects/anotia-microtia.html Accessed March 25, 2021.

CDC. https://www.cdc.gov/ncbddd/heartdefects/data.html Accessed April 6, 2021.

CDC. https://www.cdc.gov/ncbddd/birthdefects/hypospadias.html Accessed April 8, 2021.

Deputy NP, Dub B, Sharma AJ. Prevalence and Trends in Prepregnancy Normal Weight — 48 States, New York City, and District of Columbia, 2011–2015. *MMWR Morbidity and Mortality Weekly Report,* 2018;66:1402–1407. DOI: http://dx.doi.org/10.15585/mmwr.mm665152a3

Ely M, Driscoll AK. Infant Mortality in the United States, 2018: Data from the Period Linked Birth/Infant Death File. National Vital Statistics Reports; vol 69 no 7; Hyattsville, MD: National Center for Health Statistics. 2020. https://www.cdc.gov/nchs/data/nvsr/nvsr69/NVSR-69-7-508.pdf

Hardeo S, Khurshid A. Confidence Intervals for the Mean of a Poisson Distribution: A Review. *Biometrical Journal;35(1993)7, 857-867.* http://citeseerx.ist.psu.edu/viewdoc/download?doi=10.1.1.900.3700&rep=rep1&type=pdf Accessed August 7, 2019.

Illinois Department of Public Health. Leading Causes of Infant Death, Illinois Residents, 2018. http://www.dph.illinois.gov/sites/default/files/Infant%20causes%202019.pdf Accessed March 26, 2021.

Salemi JL, Tanner JP, Kirby RS, Cragan JD. The impact of the ICD-9-CM to ICD-10-CM transition on the prevalence of birth defects among infant hospitalizations in the United States. *Birth Defects Research*. 2019;111:1365-1379. https://doi.org/10.1002/bdr2.1578.

Stallings EB, Isenburg JL , Mai CT, *et al*. Population-based birth defects data in the United States, 2011-2015: A focus on eye and ear defects. Birth Defects Research. 2018;110:1478-1486. https://doi.org/10.1002/bdr2.1413

Sunderam S, Kissin DM, Zhang Y, *et al*. Assisted Reproductive Technology Surveillance — United States, 2017. *MMWR Surveillance Summaries*, 2020;69(No. SS-9):1–20. DOI: http://dx.doi.org/10.15585/mmwr.ss6909a1external icon.

Wright VC, Schieve LA, Reynolds MA, Jeng GJ. Assisted Reproductive Technology Surveillance --- United States, 2002. *MMWR Surveillance Summaries***,** 2005; 54(SS02);1-24.

APPENDIX 1

Description and ICD-9-CM Codes for Selected Birth Defects

Birth Defect	ICD-9-CM Codes	ICD-10-CM Codes	Description
Anencephalus	740.0-740.1	Q000.0-Q000.1	A neural tube defect that occurs when the head end of the neural tube fails to close, resulting in the absence of a major portion of the brain, skull, and scalp. Includes craniorachischisis in which there is incomplete closure of the skull and spinal column.
Spina bifida without anencephalus	741.xx	Q05, Q070.01, Q07.3	A birth defect in which there is a bony defect in the vertebral column so that part of the spinal cord, which is normally protected within the vertebral column, is exposed. May be associated with hydrocephalus.
Encephalocele	742.0	Q01	A neural tube defect affecting the skull, resulting in the protrusion of the meninges and portions of the brain through a bony midline defect in the skull.
Microcephalus	742.1	Q02	An abnormally small head due to failure of brain growth. In precise terms, microcephaly is a head circumference that is more than two standard deviations below the normal mean for age, sex, race, and gestation.
Anophthalmia	743.0x	Q11.0	Absence of the eye, as a result of a congenital malformation of the globe.
Microphthalmia	743.1x	Q11.2	An abnormally small eye, a congenital malformation of the globe.
Congenital cataract	743.30-743.34	Q12.0	Opacity of the lens that occurs in the fetus at some time during the pregnancy and is present at birth.
Anotia	744.01	Q16.0	Congenital absence of the external ear (the auricle).
Microtia	744.23	Q17.2	Smallness of the auricle of the ear with a blind or absent external auditory meatus.
Common truncus	745.0	Q20.0	Failure of the fetal truncus arteriosus to divide into the aorta and pulmonary artery.
Transposition of great vessels	745.1x	Q20.3,Q20.5	A congenital heart defect in which the position of the two major vessels that carry blood away from the heart, the aorta and the pulmonary artery, is transposed.

Birth Defect	ICD-9-CM Codes	ICD-10-CM Codes	Description
Tetralogy of Fallot	745.2	Q21.3	A congenital defect of the heart consisting of four abnormalities (a ventricular septal defect, an overriding aorta, right ventricular hypertrophy, and pulmonary valve or artery stenosis or atresia) that results in insufficiently oxygenated blood pumped to the body.
Ventricular septal defect	745.4	Q21.0	A hole in the wall between the lower chambers of the heart.
Double outlet right ventricle	745.11	Q20.1	Pulmonary artery and aorta both connect to the right ventricle.
Endocardial cushion defect	745.6x	Q21.2	A spectrum of septal defects associated with persistence of the embryonic atrioventricular canal due to incomplete growth and fusion of the endocardial cushion.
Pulmonary valve stenosis and atresia	746.01/746.02	Q22.0, Q22.1	Absence or narrowing of the valve between the right ventricle and the pulmonary artery.
Tricuspid valve stenosis, atresia and other anomalies	746.1	Q22.4	Tricuspid atresia is the absence or pathological narrowing of the valve between the right atrium and ventricle, with the presence of an atrial defect through which all the systemic venous return reaches the left heart.
Ebstein anomaly	746.2	Q22.5	Deformation or displacement of the tricuspid valve with the septal and posterior leaflets being attached to the wall of the right ventricle.
Aortic valve stenosis	746.3	Q23.0	A narrowing or obstruction of the aortic heart valve, causing it to not open properly and to obstruct the flow of blood from the left ventricle to the aorta.
Hypoplastic left heart syndrome	746.7	Q23.4	A form of congenital heart disease in which the whole left half of the heart (including the aorta, aortic valve, left ventricle, and mitral valve) is underdeveloped.
Coarctation of aorta	747.10	Q25.1	A birth defect in which the major artery from the heart (aorta) is narrowed somewhere along its length; most commonly the narrowing is just past the point where the aorta and the subclavian artery come together.
Choanal atresia	748.0	Q30.0	A congenital narrowing or blockage of the nasal airway by membranous or bony tissue.
Cleft palate without cleft lip	749.0x	Q35.1-Q35.9	An opening in the roof of the mouth (the palate) due to a failure of the palatal shelves to come fully together from either side of the mouth and fuse during embryonic development.

Birth Defect	ICD-9-CM Codes	ICD-10-CM Codes	Description
Cleft lip with and without cleft palate	749.1x/749.20-749.25	Q36.0-Q36.9, Q37.0-Q37.9	The presence of one or two vertical fissures in the upper lip resulting from failure of the normal process of fusion of the lip to come to completion during embryonic life.
Esophageal atresia/ Tracheoesophageal fistula	750.3	Q39.0-Q39.4	A narrowing or obstruction of the esophagus sometimes with a connection or hole between the lower esophagus and the trachea.
Rectal and large intestinal atresia and stenosis	751.2	Q42.0-Q42.9	Absence, abnormal localization, or blockage of the large intestine or rectum.
Biliary atresia	751.61	Q44.2-Q44.3	Congenital absence or closure of the major bile ducts that drain bile from the liver.
Hypospadias	752.61	Q54.0-Q54.3, Q54.5-Q54.9	A relatively common abnormality of the penis that appears as an abnormal opening of the penis on the underside of the penis rather than at the end. (In females, the opening to the urinary tract is below the normal opening.)
Renal agenesis/hypoplasia	753.0	Q60.0-Q60.6	The absence or underdevelopment of the kidneys; may be bilateral or unilateral.
Bladder exstrophy	753.5	Q64.10,Q64.19	An exstrophic bladder is one that is turned inside out like a rubber glove. Part of the abdominal wall and bladder wall are missing.
Reduction deformity, upper and lower limbs	755.2-755.4	Q71.0-Q73.8	A shortening or absence of one or both limbs; it may be of upper or lower limbs.
Diaphragmatic hernia	756.6	Q79.0, Q79.1	A failure of the diaphragm to form completely, leaving a hole. Abdominal organs can protrude through the hole into the chest cavity and interfere with development of the heart and lungs.
Omphalocele	756.72	Q79.2	The intestine or other abdominal organs protrude from the base of the belly button. The intestines are covered by a thin layer of tissue.
Gastroschisis	756.73	Q79.3	A herniation of the abdominal contents through a defect in the abdominal wall.

Birth Defect	ICD-9-CM Codes	ICD-10-CM Codes	Description
Down syndrome	758.0	Q90.0-Q90.9	A syndrome arising from the presence of an extra number 21 chromosome resulting in mild- to-moderate intellectual disabilities, distinctive malformations of the head and face, and other abnormalities.
Patau syndrome	758.1	Q91.4-Q91.7	A syndrome arising from the presence of an extra number 13 chromosome. Newborns have numerous internal and external abnormalities, including profound developmental disabilities.
Edward syndrome	758.2	Q91.0-Q91.3	A syndrome arising from the presence of an extra number 18 chromosome. It causes major physical abnormalities and severe developmental disabilities.