



Trends in the Prevalence of Birth Defects in Illinois 2002-2014

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**TRENDS IN THE PREVALENCE OF
BIRTH DEFECTS IN
ILLINOIS
2002-2014**



Illinois Department of Public Health
Division of Epidemiologic Studies

March 2018

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INTRODUCTION

Adverse pregnancy outcomes are recorded by the Illinois Department of Public Health (IDPH) for infants with congenital anomalies (birth defects) and other serious neonatal conditions. Each year in Illinois, IDPH's Adverse Pregnancy Outcomes Reporting System (APORS) obtains information on thousands of such births throughout the state. APORS first collected statewide data about congenital anomalies identified in newborn infants in 1989.

This information is collected for two important reasons. First, infants with a birth defect often need special services to help assure that they reach their full potential. These babies are, therefore, referred to their local health departments and other providers for follow-up services. Second, the data are collected for purposes of public health surveillance. These may include describing disease patterns, tracking trends, conducting cluster investigations, and developing education and intervention strategies.

Birth defects are the leading cause of infant mortality in the United States and the second in Illinois; they contribute substantially to childhood morbidity and long-term disability.

Known causes may be broadly divided into four categories:

- genetic disorders (either hereditary or arising during conception);
- exposures to environmental chemicals (for example, medications, alcohol, cigarettes, or solvents);
- mother's illness during pregnancy, exposing her baby to viral or bacterial infection; and
- diet

However, most birth defects are likely a result of a combination of these factors. Another critical determinant of whether a child will develop a birth defect is the stage of fetal development at the time of exposure to one of the latter three causes. Fetal development is particularly vulnerable to disruption in the first trimester of pregnancy. Despite the increasing understanding of factors that give rise to birth defects, the causes of many birth defects remain unknown (Centers for Disease Control and Prevention [CDC], 2018.) The same congenital anomaly may have completely different causes in different individuals.

APORS is the most complete source of data on birth defects that exists in Illinois. All Illinois birth hospitals are mandated to report infants with adverse pregnancy outcomes born to Illinois women. (Members of the Illinois Perinatal Network in St. Louis are also required to report.) Prior to 2002, APORS was a passive surveillance system, relying primarily on reports sent to IDPH. Because passive surveillance systems are likely to underestimate birth defect rates,

efforts have been made in recent years to expand case and birth defect identification by implementing elements more characteristic of an active surveillance system. Active systems utilize abstraction staff to review data from multiple sources on an ongoing basis to identify and verify birth defects (CDC, 2018). While resource intensive, this practice is generally recognized as improving case identification and quality. Table 1 below describes the activities APORS has undertaken 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	Hospital Discharge Case Finding	Identify birth defects noted in review of hospital discharge data, but not reported to APORS

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	<ul style="list-style-type: none"> • 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	<ul style="list-style-type: none"> • 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	<ul style="list-style-type: none"> • 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 the charts are reviewed, APORS staff correct and add to the information reported by the hospitals. 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.

At times however, 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 two years of age, born from 2002-2014. This study period was chosen as it represents a span of years in which active case verification methodology was used. Previous years' rates are not comparable as this approach was not utilized. Prevalence information is also presented for Illinois and Chicago alone for the year 2014.

METHODS

Calculation and Interpretation of Rates and Confidence Intervals

Thirty-six categories of birth defects are included in this study. A listing of the International Classification of Diseases – Ninth Revision Clinical Modification (ICD-9-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 two 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 (Armitage and Berry, page 134). 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 percent confidence intervals are used; where the confidence intervals do not overlap, the rates are statistically different at the 5 percent level ($p < 0.05$).

Analysis of Trends

Trends in Illinois birth defect rates for 2002-2014 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 4.5.0.1, June 2017, 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, 2014

Birth defect rates for 35 selected categories among Illinois and Chicago newborns in 2014 are presented in tables 4 and 5. In general, 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

Figures 1 to 9 show the rates over time for 36 birth defects in nine categories. A regression line is also plotted for each birth defect. The regression lines are usually log-linear, but may be made up of several straight line segments with different slopes. All defects were examined for the period of 2002-2014, with the exception of hydrocephalus. This condition was not actively collected during 2013 and 2014 so rates for those years are not shown. Rates for Down syndrome are also plotted by maternal age group in Figure 10.

Statistically significant trends were found for eight birth defects (See Table 3). Although examination of the graphs may show some other 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). Table 3 also includes a column (average annual percentage change) that gives an estimate of how quickly the rate is changing over time. For example, the rate of spina bifida was increasing by an average of 2.8 percent each year.

Table 3. Birth Defects Showing a Significant Trend in Incidence Rate 2002-2014

Selected Birth Defect	Average Annual Percent Change	Significance of trend (P-value)
Spina bifida	2.8	0.00
Anotia/Microtia	7.1	0.00
Ventricular Septal Defect	3.0	0.00
Endocardial Cushion Defect	3.9	0.00
Tricuspid valve atresia, stenosis and other anomalies	10.0	0.00
Coarctation of Aorta	3.4	0.00
Renal Agenesis/Hypoplasia	7.1	0.00
Hypospadias	2.6	0.00

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2018.

Discussion of Illinois Results

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.

A couple of process changes in APORS have affected rates during the study period as noted throughout the following discussion. APORS hired an abstractor liaison in 2012, whose job is to oversee case abstraction accuracy and completeness. With such oversight, previously unreported or misreported diagnoses may have been identified. Additionally, hospital discharge case finding began in 2013. This process identifies additional conditions diagnosed later than the newborn hospital stay.

Spina bifida exhibited an overall increasing trend in Illinois during the study period. In a recently published article, St. Louis et al. found no significant trend in the prevalence rate of spina bifida when examining birth defect surveillance data combined from 11 states, including Illinois, for the time period of 1999-2007. The study offers limited comparability to this trend report, however, as it does not include data for more recent years. In 2015, CDC reported that initial drops in rates of neural tube defects (NTDs) following mandatory folic acid fortification in the U. S. in 1998 had largely been maintained for more than a decade. However, opportunities still existed for improvement, as nearly a quarter of women of childbearing age still did not have adequate blood folate levels associated with a lower risk for NTDs. Compounding this challenge, recent research has suggested that carbohydrate-restricted diets during the year prior to conception may result in increased risks of NTDs, especially among women with unplanned pregnancies (*Desrosiers, et al*). The prevalence of NTDs among Hispanic births continues to be higher than that observed in other racial/ethnic groups, possibly due to a combination of genetic and dietary factors (*CDC,2015*).

The rate of anotia and microtia increased steadily from 2002-2014. These conditions occur more often in multiple births, and among Hispanic and Asian women (*Shaw et al., Forrester & Merz, Husain et al*). While the proportion of births to Hispanic women has been steady in Illinois during the study period, the proportion of births to women of other (Non-black Non-white) races, including Asian, has increased from 4.9 percent in 2002 to 8.1 percent in 2014. Also, the proportion of multiple births has risen slightly as well to 4.0 percent in 2014 from 3.7 percent in 2002. Other factors that may increase the risk of anotia or microtia include diabetes (pre-pregnancy), eating a low carbohydrate diet during pregnancy, and use of the medication

isotretinoin (Accutane®) (CDC, 2018).

Significant increases were seen in four cardiovascular defects including ventricular septal defect, endocardial cushion defect, tricuspid atresia, stenosis and other anomalies, and coarctation of the aorta. Heart defects are the most common type of birth defects, and CDC reports that the prevalence of some types of heart defects is increasing (CDC, 2018). Greater use of diagnostic techniques may also explain part of the increase in Illinois. Hospital discharge case-finding in Illinois has been a factor in identifying additional cases of valve anomalies since 2013. Although the overall trend was not significant for pulmonary valve atresia and stenosis, increases seen during the latter period of study were likely due in part to identification of cases via hospital discharge data.

Renal agenesis/hypoplasia rates increased during the study period. These conditions are largely genetic in nature. The increase in rates during the latter part of the study period is primarily due to improved chart abstraction review, with the identification of additional cases of hypoplasia.

Hypospadias rates increased during the period. CDC has recently identified several factors that may increase the risk of having a child with this condition including older maternal age, obesity, use of assisted reproductive technology (ART), and use of hormones (CDC, 2017). In Illinois, the proportion of births in Illinois to women ages 35 and over has increased from 14.5 percent in 2002 to 17.4 percent in 2014. Use of ART has steadily increased in the U.S. since its introduction in 1981, and in Illinois, its use exceeded the national rate in 2013 (CDC, 2015). Multistate studies using 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 2003-2015. (CDC, 2018).

While overall rates of Down syndrome are higher as maternal age increases, no significant changes were seen in the rates over time.

As for eye defects, an upward trend in the rate of anophthalmia/microphthalmia was detected from 2011-2014, which is largely reflective of improved chart review processes in Illinois. It is expected that this trend will level off in future years. As well, the number of cases of congenital cataract remained very steady throughout the entire study period until 2014, when there was a sizable spike in cases. Factors that might explain this outlier were examined, including hospital or geographical reporting variances, abstraction methods or coding misclassification, but no plausible explanation was identified. Current estimates of congenital cataract in 2015, although not complete, appear to be returning to rates seen during 2002-2013.

**Birth Defect Rates for Selected Categories Among
Illinois and Chicago Newborns**

2014

**Table 4. Number and Rate of Selected Birth Defects for 2014
Illinois**

Selected Birth Defects Groups	N	Rate ¹	95% CI ²
A. Central Nervous System			
Anencephalus	29	1.8	(1.2, 2.6)
Spina bifida	54	3.4	(2.6, 4.4)
Encephalocele	10	0.6	(0.3, 1.2)
Microcephalus	100	6.3	(5.1, 7.7)
<i>Total Selected CNS Defects</i>	<i>193</i>	<i>12.2</i>	<i>(10.5, 14.0)</i>
B. Eye			
Anophthalmia/Microphthalmia	40	2.5	(1.8, 3.4)
Congenital cataract	34	2.1	(1.5, 3.0)
<i>Total Selected Eye Defects</i>	<i>74</i>	<i>4.7</i>	<i>(3.7, 5.9)</i>
C. Ear			
Anotia/Microtia	31	2.0	(1.3, 2.8)
D. Cardiovascular			
Common truncus	5	0.3	(0.1, 0.7)
Transposition of great vessels	48	3.0	(2.2, 4.0)
Tetralogy of Fallot	68	4.3	(3.3, 5.4)
Ventricular septal defect	768	48.4	(45.1, 52.0)
Double outlet right ventricle	28	1.8	(1.2, 2.6)
Endocardial cushion defect	81	5.1	(4.1, 6.4)
Pulmonary valve atresia and stenosis	110	6.9	(5.7, 8.4)
Tricuspid valve atresia, stenosis and other anomalies	67	4.2	(3.3, 5.4)
Ebstein anomaly	12	0.8	(0.4, 1.3)
Aortic valve stenosis	29	1.8	(1.2, 2.6)
Hypoplastic left heart syndrome	27	1.7	(1.1, 2.5)
Coarctation of aorta	64	4.0	(3.1, 5.2)
<i>Total Selected Cardiovascular Defects</i>	<i>1,307</i>	<i>82.4</i>	<i>(78.0, 87.0)</i>
F. Orofacial			
Cleft palate without cleft lip	93	5.9	(4.7, 7.2)
Cleft lip with and without cleft palate	148	9.3	(7.9, 11.0)
Choanal atresia	14	0.9	(0.5, 1.5)
<i>Total Selected Orofacial Defects</i>	<i>255</i>	<i>16.1</i>	<i>(14.2, 18.2)</i>
G. Gastrointestinal			
Esophageal atresia/Tracheoesophageal fistula	34	1.6	(1.1, 2.4)
Rectal and large intestinal atresia/stenosis	53	3.6	(2.8, 4.6)
Biliary atresia	6	0.4	(0.8, 0.8)
<i>Total Selected Gastrointestinal Defects</i>	<i>93</i>	<i>5.9</i>	<i>(4.7, 7.2)</i>

Selected Birth Defects Groups	N	Rate¹	95% CI²	
H. Genitourinary				
Renal agenesis/hypoplasia	104	6.6	(5.4,	7.9)
Bladder exstrophy	4	0.3	(0.1,	0.6)
Hypospadias	491	31.0	(28.3,	33.8)
<i>Total Selected Genitourinary Defects</i>	<i>599</i>	<i>37.8</i>	<i>(34.8,</i>	<i>40.9)</i>
I. Musculoskeletal				
Reduction deformity, upper and lower limbs	83	5.2	(4.2,	6.5)
Gastroschisis	53	3.3	(2.5,	4.4)
Omphalocele	27	1.7	(1.1,	2.5)
Diaphragmatic hernia	37	2.3	(1.6,	3.2)
<i>Total Selected Musculoskeletal Defects</i>	<i>200</i>	<i>12.6</i>	<i>(10.9,</i>	<i>14.5)</i>
J. Chromosomal				
Trisomy 13 (Patau syndrome)	16	1.0	(0.6,	1.6)
Trisomy 21 (Down syndrome)	224	14.1	(12.3,	16.1)
Trisomy 18 (Edward syndrome)	37	2.3	(1.6,	3.2)
<i>Total Selected Chromosomal Defects</i>	<i>277</i>	<i>17.5</i>	<i>(15.5,</i>	<i>19.7)</i>
<i>Total All Selected Defects</i>	<i>3,029</i>	<i>191.1</i>	<i>(184.3,</i>	<i>198.0)</i>

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 2018

**Table 5. Number and Rate of Selected Birth Defects for 2014
Chicago**

Selected Birth Defects Groups	N	Rate ¹	95% CI ²
A. Central Nervous System			
Anencephalus	3	0.7	(0.2, 2.2)
Spina bifida	15	3.7	(2.1, 6.2)
Encephalocele	4	1.0	(0.3, 2.6)
Microcephalus	26	6.5	(4.2, 9.5)
<i>Total Selected CNS Defects</i>	48	12.0	(8.8, 15.9)
B. Eye			
Anophthalmia/Microphthalmia	10	2.5	(1.2, 4.6)
Congenital cataract	11	2.7	(1.4, 4.9)
<i>Total Selected Eye Defects</i>	21	5.2	(3.2, 8.0)
C. Ear			
Anotia/Microtia	15	3.7	(2.1, 6.2)
D. Cardiovascular			
Common truncus	3	0.7	(0.2, 2.2)
Transposition of great vessels	10	2.5	(1.2, 4.6)
Tetralogy of Fallot	23	5.7	(3.6, 8.6)
Ventricular septal defect	197	49.1	(42.5, 56.4)
Double outlet right ventricle	6	1.5	(0.5, 3.3)
Endocardial cushion defect	26	6.5	(4.2, 9.5)
Pulmonary valve atresia and stenosis	31	7.7	(5.2, 11.0)
Tricuspid valve atresia, stenosis and other anomalies	20	5.0	(3.0, 7.7)
Ebstein anomaly	3	0.7	(0.2, 2.2)
Aortic valve stenosis	7	1.7	(0.7, 3.6)
Hypoplastic left heart syndrome	9	2.2	(1.0, 4.3)
Coarctation of aorta	25	6.2	(4.0, 9.2)
<i>Total Selected Cardiovascular Defects</i>	360	89.7	(80.7, 99.5)
F. Orofacial			
Cleft palate without cleft lip	15	3.7	(2.1, 6.2)
Cleft lip with and without cleft palate	42	10.5	(7.5, 14.1)
Choanal atresia	5	1.2	(0.4, 2.9)
<i>Total Selected Orofacial Defects</i>	62	15.4	(11.8, 19.8)
G. Gastrointestinal			
Esophageal atresia/Tracheoesophageal fistula	9	2.2	(1.0, 4.3)
Rectal and large intestinal atresia/stenosis	12	3.0	(1.5, 5.2)
Biliary atresia	1	0.2	(0.0, 1.4)
<i>Total Selected Gastrointestinal Defects</i>	22	5.5	(3.4, 8.3)

Selected Birth Defects Groups	N	Rate¹	95% CI²
H. Genitourinary			
Renal agenesis/hypoplasia	25	6.2	(4.0, 9.2)
Bladder exstrophy	0	0.0	(0.0, 0.9)
Hypospadias	105	26.2	(21.4, 31.7)
<i>Total Selected Genitourinary Defects</i>	<i>130</i>	<i>32.4</i>	<i>(27.1, 38.5)</i>
I. Musculoskeletal			
Reduction deformity, upper and lower limbs	20	5.0	(3.0, 7.7)
Gastroschisis	13	3.2	(1.7, 5.5)
Omphalocele	5	1.2	(0.4, 2.9)
Diaphragmatic hernia	8	2.0	(0.9, 3.9)
<i>Total Selected Musculoskeletal Defects</i>	<i>46</i>	<i>11.5</i>	<i>(8.4, 15.3)</i>
J. Chromosomal			
Trisomy 13 (Patau syndrome)	5	1.2	(0.4, 2.9)
Trisomy 21 (Down syndrome)	48	12.0	(8.8, 15.9)
Trisomy 18 (Edward syndrome)	10	2.5	(1.2, 4.6)
<i>Total Selected Chromosomal Defects</i>	<i>63</i>	<i>15.7</i>	<i>(12.1, 20.1)</i>
<i>Total All Selected Defects</i>	<i>767</i>	<i>191.1</i>	<i>(177.8, 205.1)</i>

¹ Rate per 10,000 live births

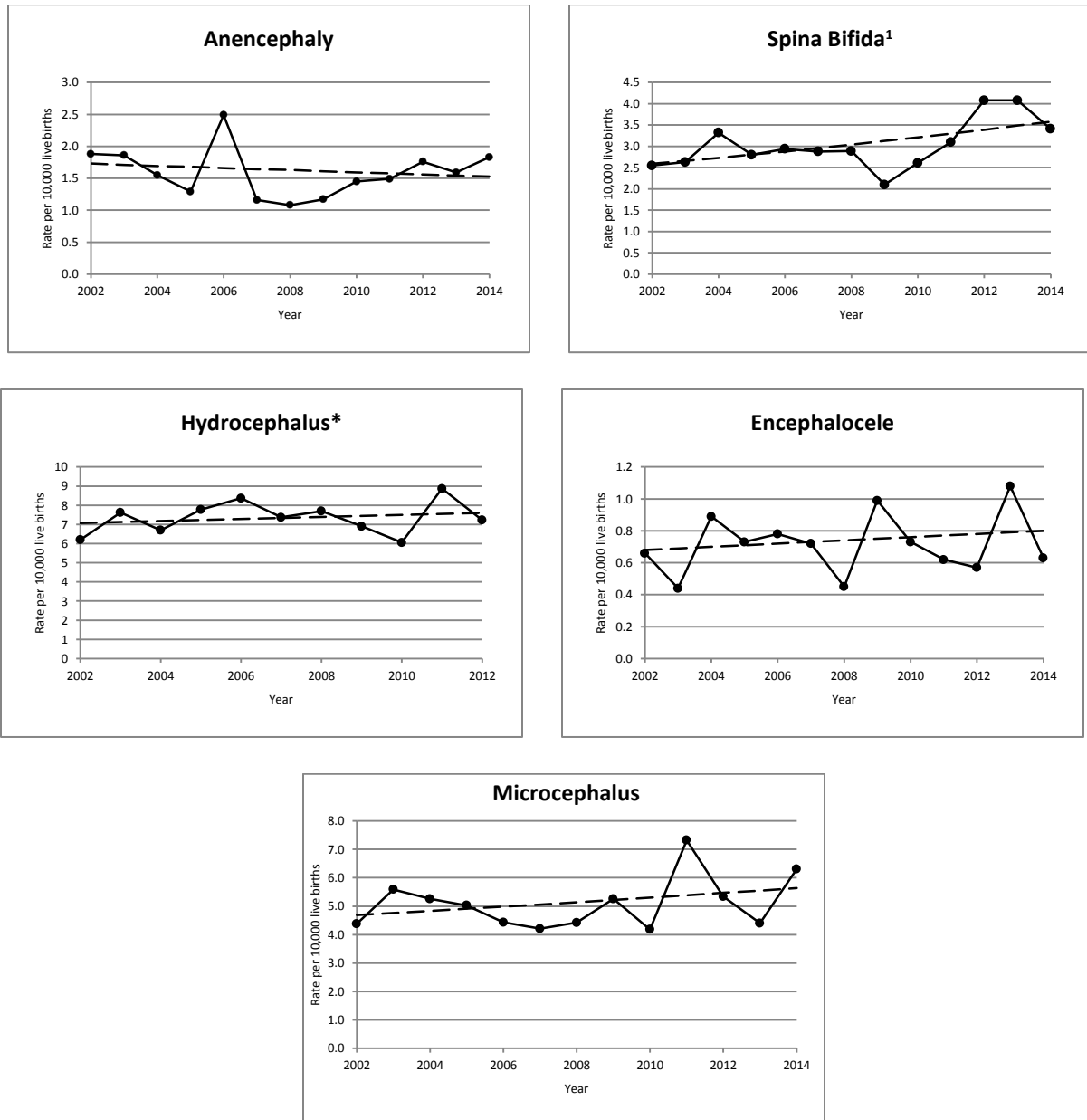
² 95 percent confidence interval for rate

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2018

**Trends in Birth Defect Rates for Selected Categories
Among Illinois Newborns**

2002-2014

Figure 1. Trends in the Reported Prevalence Rates of Neural Tube Defects per 10,000 Live Births 2002-2014



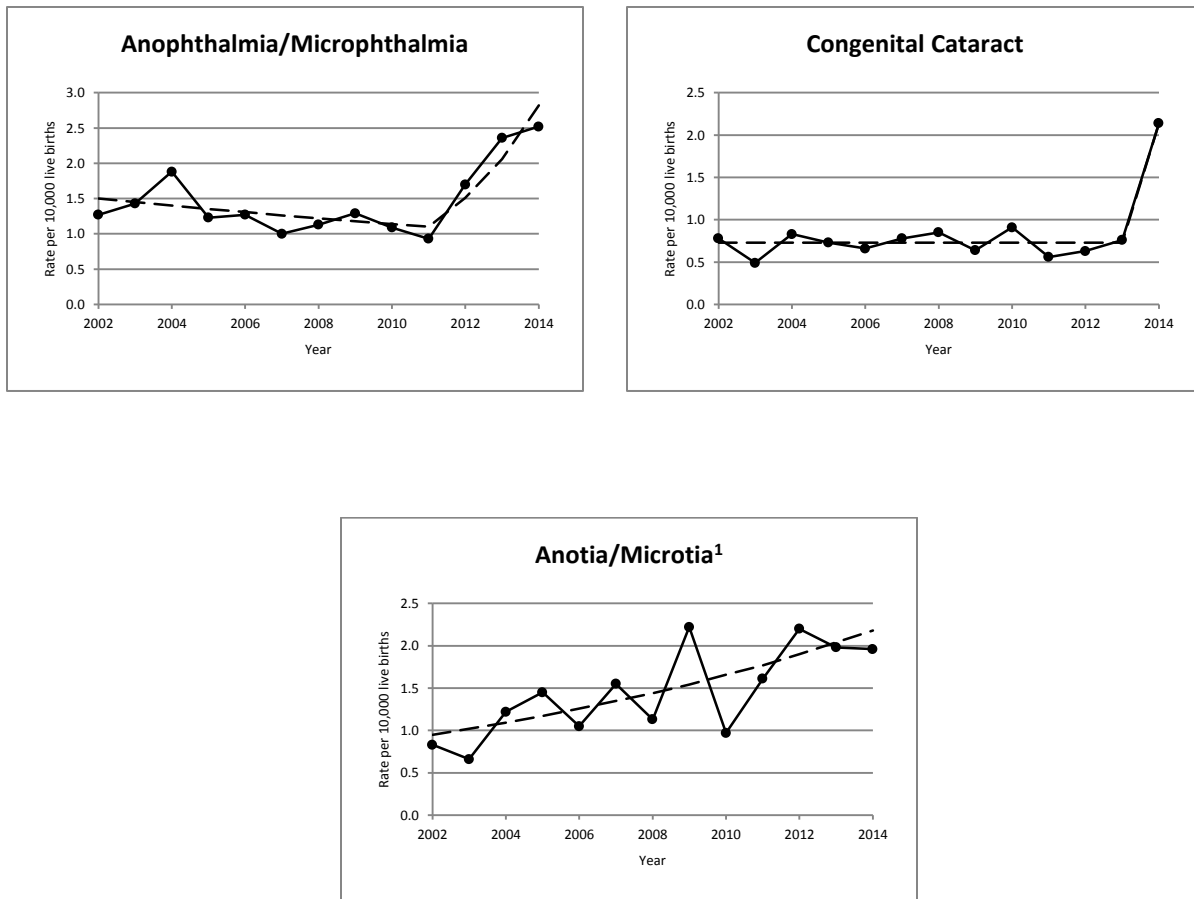
●—● Observed Rates - - - - - Regression Line

¹Trend is significant; See Table 3 for details.

*Hydrocephalus data no longer actively collected after 2012

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2018

Figure 2. Trends in the Reported Prevalence Rates of Eye and Ear Defects per 10,000 Live Births 2002-2014



●—● Observed Rates - - - - - Regression Line

¹Trend is significant; See Table 3 for details.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2018

Figure 3A. Trends in the Reported Prevalence Rates of Cardiac Defects per 10,000 Live Births 2002-2014

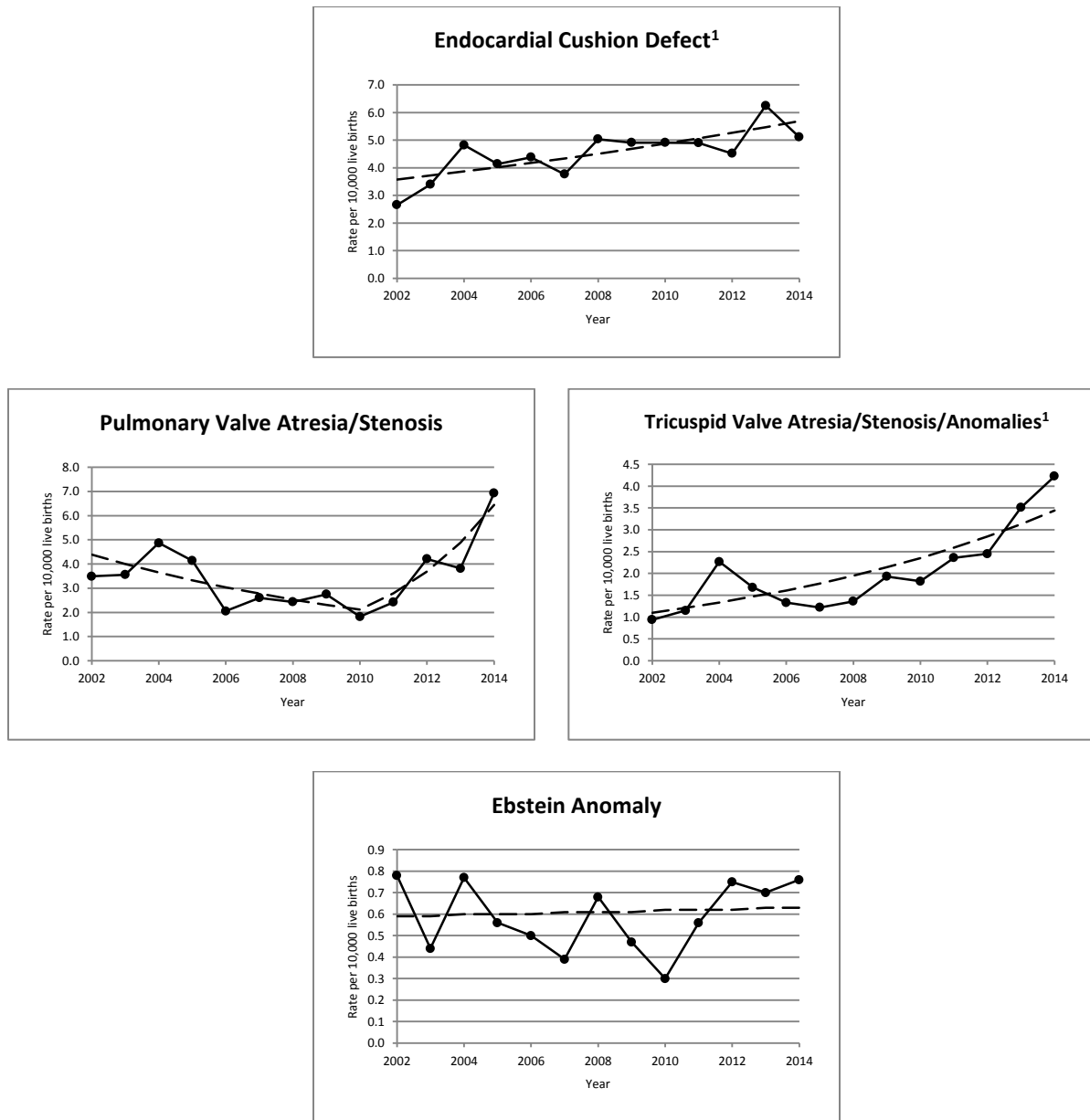


●—● Observed Rates - - - - - Regression Line

¹Trend is significant; See Table 3 for details.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2018

Figure 3B. Trends in the Reported Prevalence Rates of Cardiac Defects per 10,000 Live Births 2002-2014

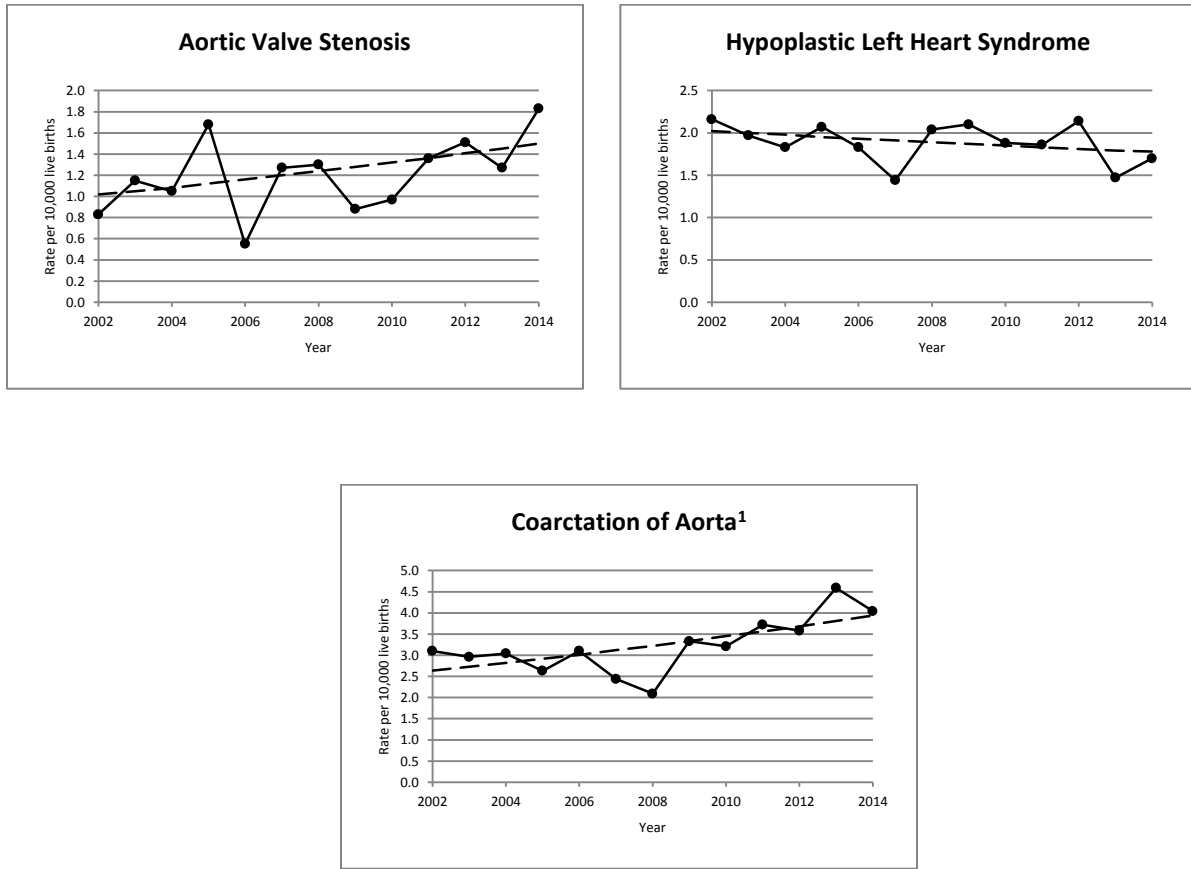


●—● Observed Rates - - - - - Regression Line

¹Trend is significant; See Table 3 for details.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2018

Figure 4. Trends in the Reported Prevalence Rates of Circulatory Defects per 10,000 Live Births 2002-2014

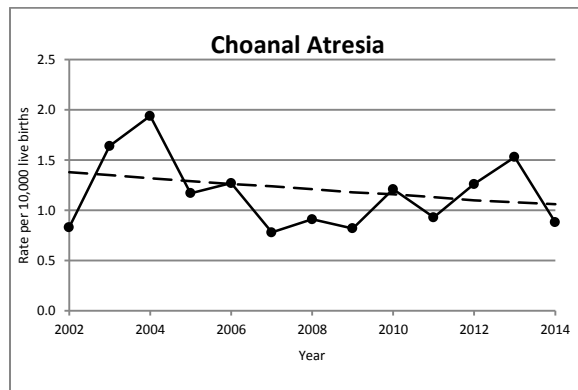
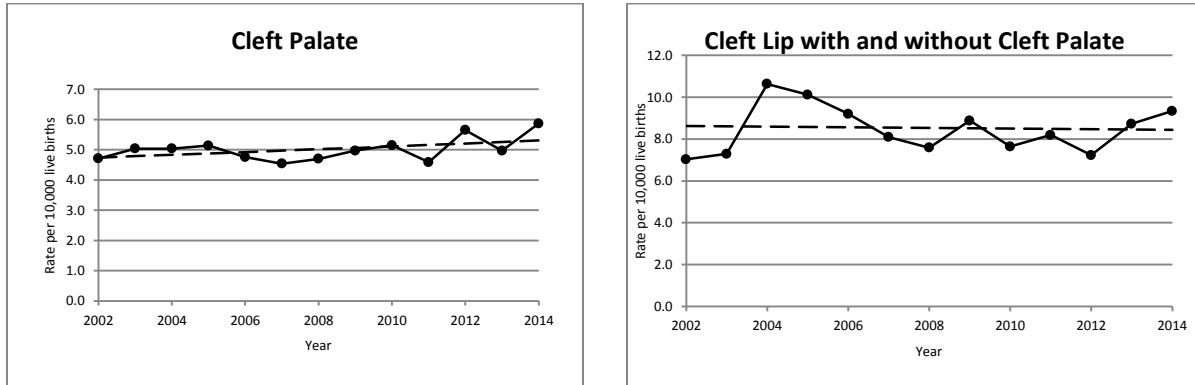


●—● Observed Rates - - - - - Regression Line

¹Trend is significant; See Table 3 for details.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2018

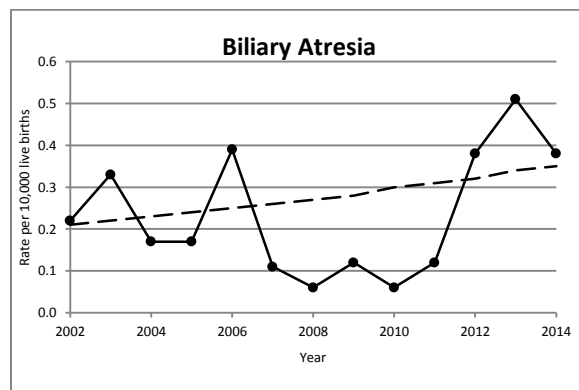
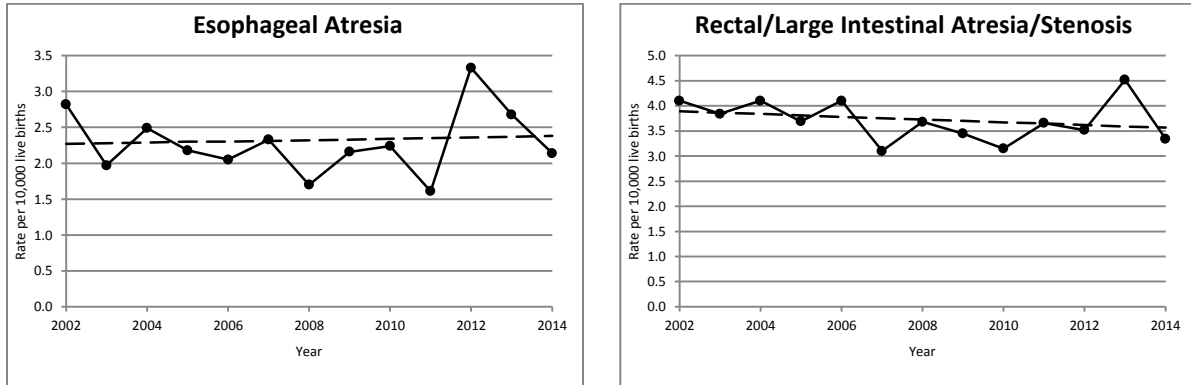
Figure 5. Trends in the Reported Prevalence Rates of Respiratory and Oral Defects per 10,000 Live Births 2002-2014



●—● Observed Rates - - - - - Regression Line

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2018

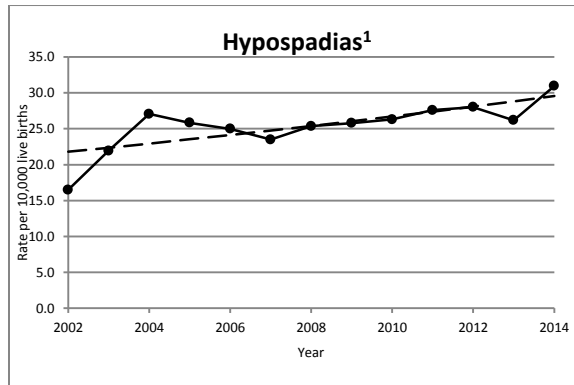
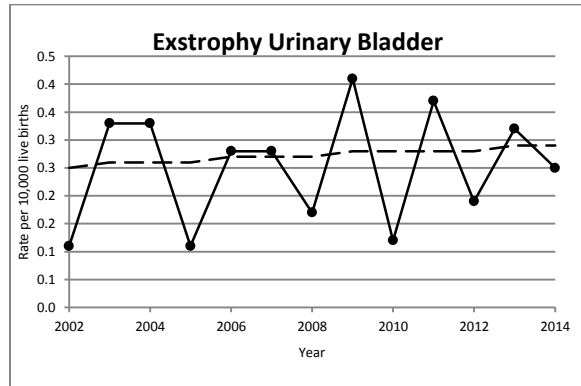
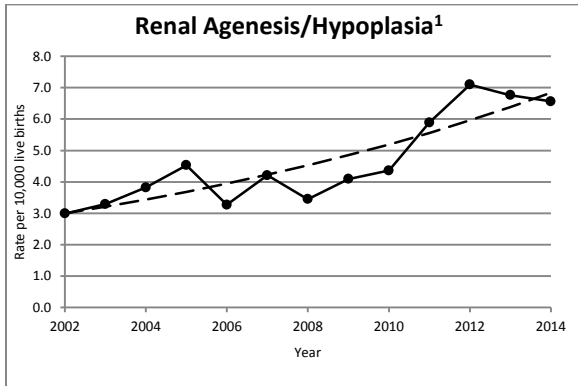
Figure 6. Trends in the Reported Prevalence Rates of Gastrointestinal Defects per 10,000 Live Births 2002-2014



●—● Observed Rates - - - - - Regression Line

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2018

Figure 7. Trends in the Reported Prevalence Rates of Genitourinary Defects per 10,000 Live Births 2002-2014

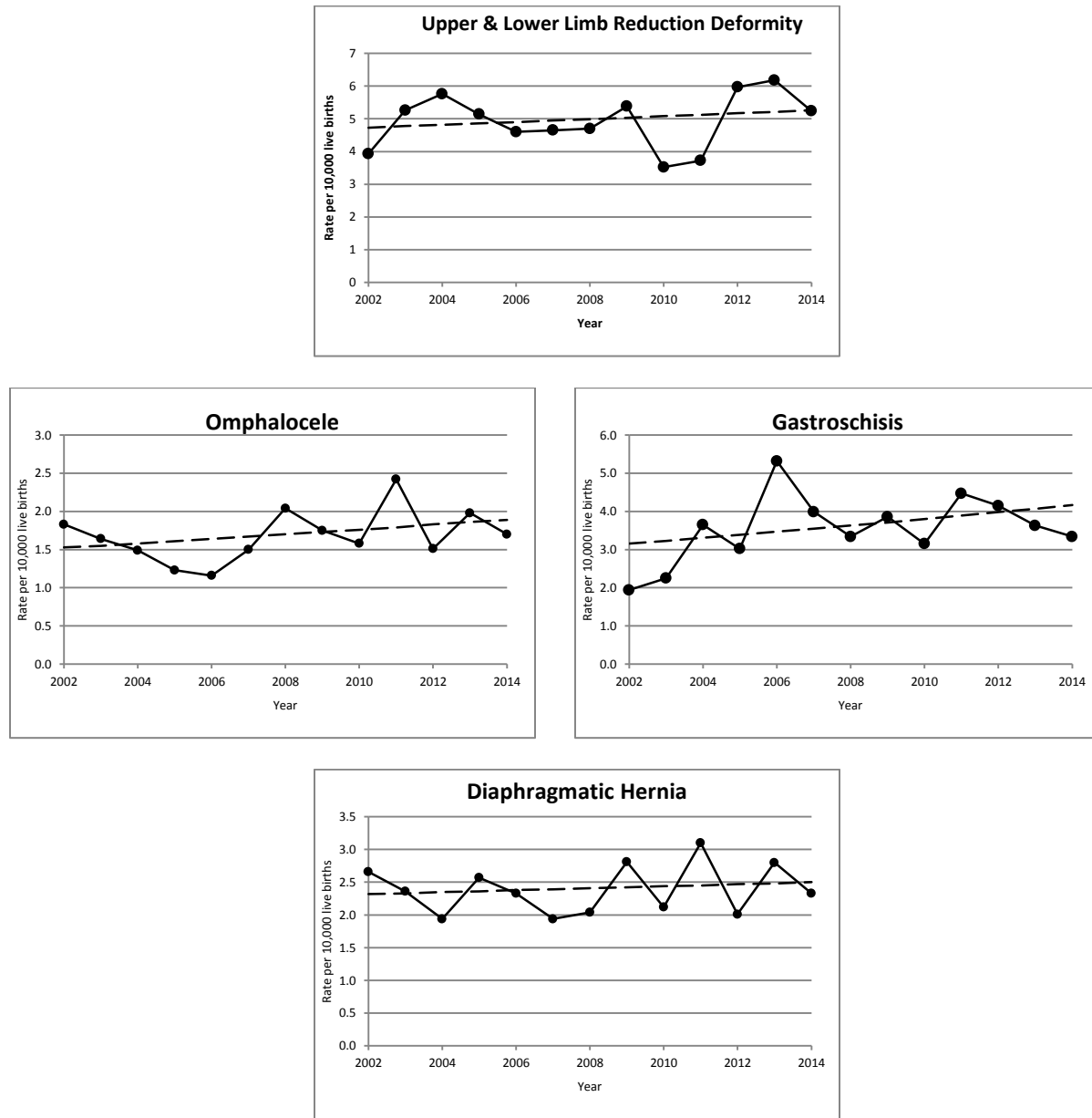


●—● Observed Rates - - - - - Regression Line

¹Trend is significant; See Table 3 for details.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2018

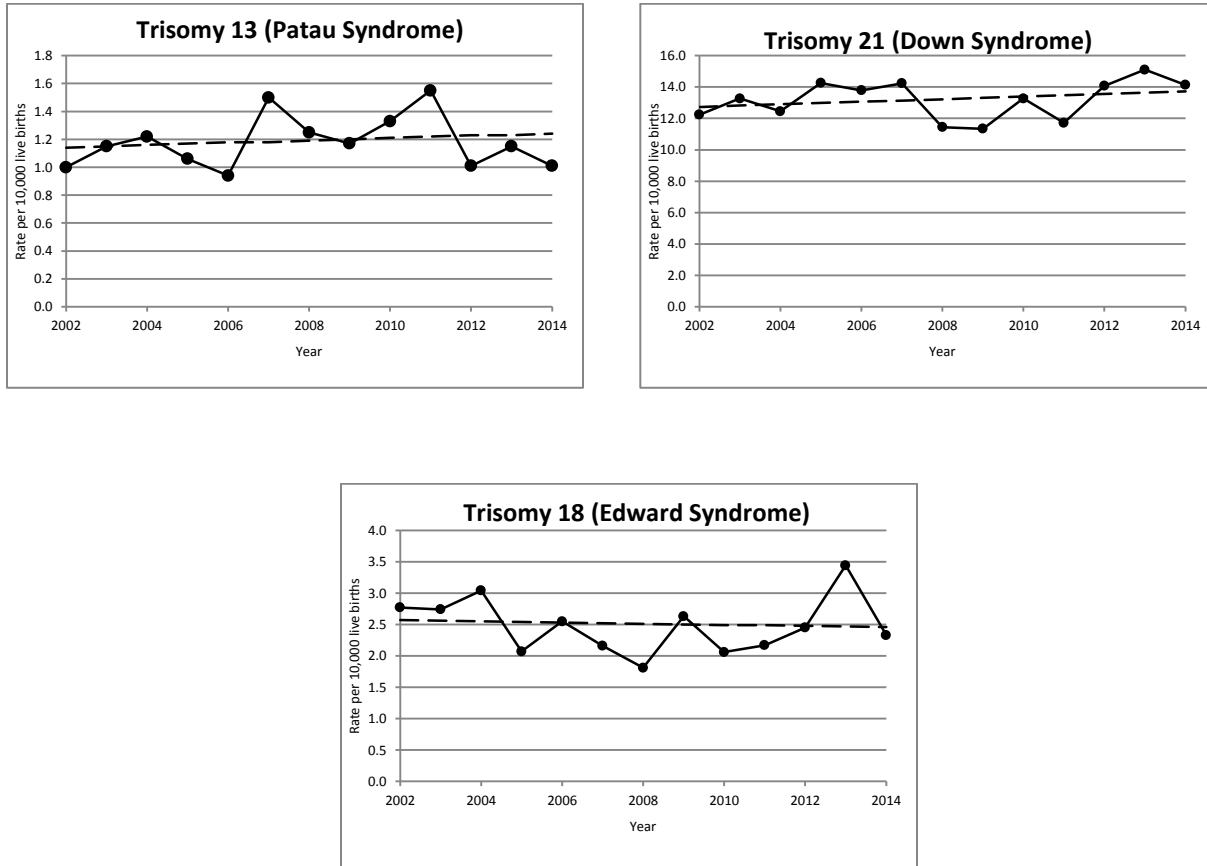
Figure 8. Trends in the Reported Prevalence Rates of Musculoskeletal Defects per 10,000 Live Births 2002-2014



●—● Observed Rates - - - - - Regression Line

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2018

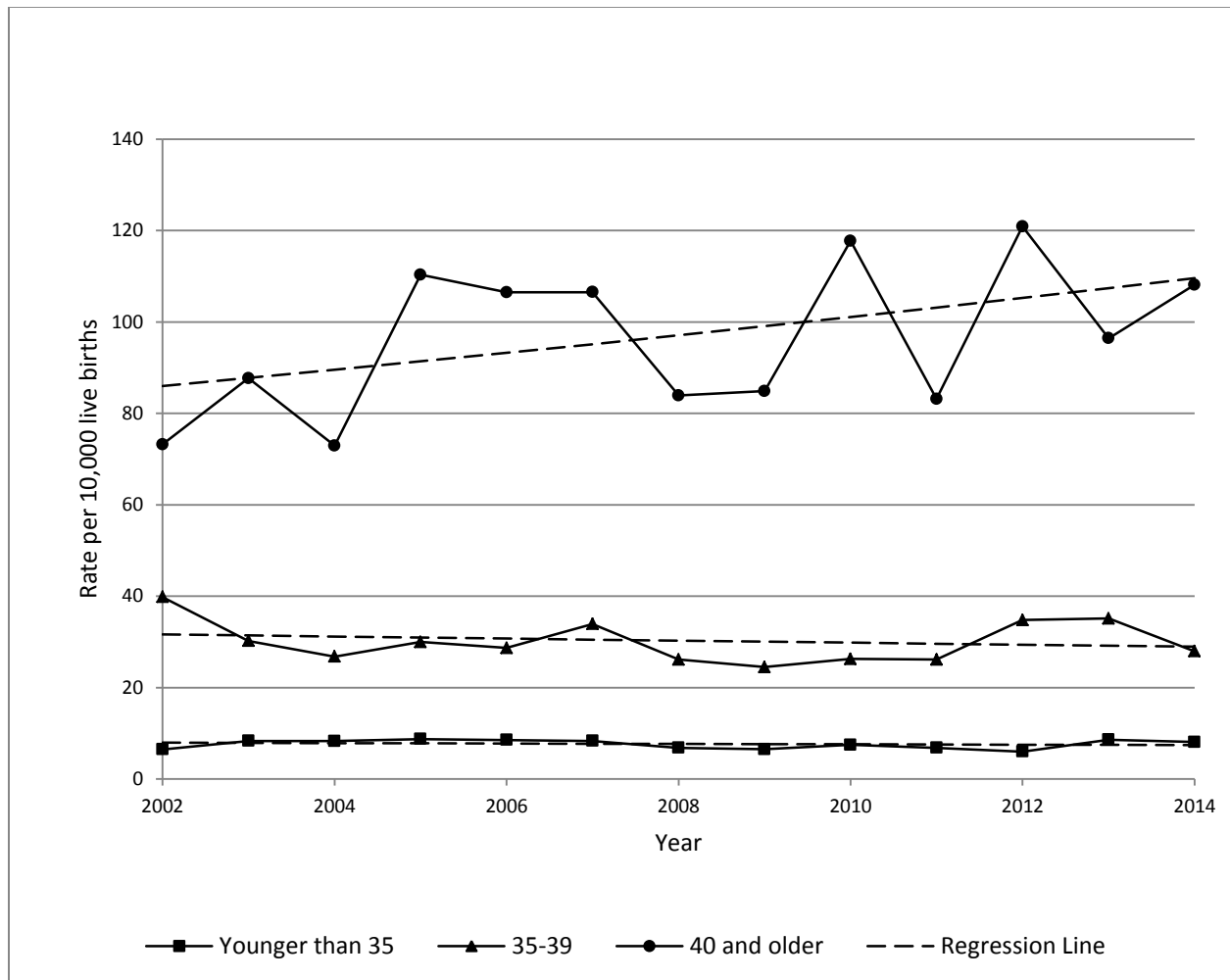
Figure 9. Trends in the Reported Prevalence Rates of Chromosomal Defects per 10,000 Live Births 2002-2014



● — ● Observed Rates - - - - - Regression Line

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2018

Figure 10. Trends in Reported Prevalence of Trisomy 21 (Down Syndrome) By Maternal Age at Delivery, 2002-2014



Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, March 2018

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APPENDIX 1

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

Birth Defect	ICD-9-CM Codes	Description
Anencephalus	740.0-740.1	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	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	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	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.
Hydrocephalus without spina bifida	742.3	An abnormal buildup of cerebrospinal fluid in the ventricles of the brain. The fluid is often under increased pressure and can compress and damage the brain.
Anophthalmia	743.0x	Absence of the eye, as a result of a congenital malformation of the globe.
Microphthalmia	743.1x	An abnormally small eye, a congenital malformation of the globe.
Congenital cataract	743.30-743.34	Opacity of the lens that occurs in the fetus at some time during the pregnancy and is present at birth.
Anotia	744.01	Congenital absence of the external ear (the auricle).
Microtia	744.23	Smallness of the auricle of the ear with a blind or absent external auditory meatus.
Common truncus	745.0	Failure of the fetal truncus arteriosus to divide into the aorta and pulmonary artery.
Transposition of great vessels	745.1x	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.
Tetralogy of Fallot	745.2	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.

Birth Defect	ICD-9-CM Codes	Description
Ventricular septal defect	745.4	A hole in the wall between the lower chambers of the heart.
Double outlet right ventricle	745.11	Pulmonary artery and aorta both connect to the right ventricle.
Endocardial cushion defect	745.6x	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	Absence or narrowing of the valve between the right ventricle and the pulmonary artery.
Tricuspid valve stenosis ,atresia and other anomolies	746.1	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	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	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	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	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	A congenital narrowing or blockage of the nasal airway by membranous or bony tissue.
Cleft palate without cleft lip	749.0x	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.
Cleft lip with and without cleft palate	749.1x/749.20-749.25	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	A narrowing or obstruction of the esophagus sometimes with a connection or hole between the lower esophagus and the trachea.

Birth Defect	ICD-9-CM Codes	Description
Rectal and large intestinal atresia and stenosis	751.2	Absence, abnormal localization or blockage of the large intestine or rectum.
Biliary atresia	751.61	Congenital absence or closure of the major bile ducts that drain bile from the liver.
Hypospadias	752.61	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	The absence or underdevelopment of the kidneys; may be bilateral or unilateral.
Obstructive genitourinary defect	753.2x, 753.6	Obstruction of ureter, renal pelvis, urethra or bladder neck.
Bladder exstrophy	753.5	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	A shortening or absence of one or both limbs, it may be of upper or lower limbs.
Diaphragmatic hernia	756.6	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	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	A herniation of the abdominal contents through a defect in the abdominal wall.
Down syndrome	758.0	A syndrome arising from the presence of an extra number 21 chromosome resulting in mental retardation, distinctive malformations of the head and face, and other abnormalities.
Patau syndrome	758.1	A syndrome arising from the presence of an extra number 13 chromosome. Newborns have numerous internal and external abnormalities, including profound retardation.
Edward syndrome	758.2	A syndrome arising from the presence of an extra number 18 chromosome. It causes major physical abnormalities and severe mental retardation.