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# **Trends in the Prevalence of Birth Defects in Illinois and Chicago 1989-2007**

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**TRENDS IN THE PREVALENCE OF  
BIRTH DEFECTS IN  
ILLINOIS AND CHICAGO  
1989-2007**



Illinois Department of Public Health  
Division of Epidemiologic Studies

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1989-2007**

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## INTRODUCTION

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

- chromosomal 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.

The stage of fetal development at the time of exposure to one of the latter three causes is critical. 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 about 70 percent of all birth defects remain unknown (CDC Foundation). 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 hospitals are mandated to report infants with adverse pregnancy outcomes born to Illinois women. (Perinatal centers in St. Louis voluntarily participate.) Until 2002, APORS was a passive surveillance system, relying primarily on reports sent to the Department. Such passive systems are likely to underestimate birth defect rates. The Trust for America's Health gave

APORS a rating of B because of this lack of active surveillance activities. (Only eight states received an A rating).

Since 1998, a number of projects have been carried out to identify cases and birth defects that have not been reported to APORS using the passive surveillance mechanism described above. These do not make a systematic active surveillance system, but are important elements of such a system. These projects are described in Table 1.

**Table 1: Projects to Identify Cases and Birth Defects**

<b>Birth years</b>	<b>Study Title</b>	<b>Purpose</b>
1996-1997	Out to One Year Study	Identify birth defects diagnosed in the first year of life, after the newborn hospital stay.
1999-2002	Active Case Finding	Identify major birth defects diagnosed in the first year of life at large Chicago hospitals, after the newborn hospital stay.
2000	Hospital Discharge Study	Identify birth defects noted in the hospital newborn stay discharge record, but not reported to APORS
1999-2005	Very Low Birth Weight Study	Identify children not reported to APORS, but with low birth weights or low APGAR scores recorded on their birth certificate.
2002 -2007	Active Case Verification	Identify unreported or misreported diagnoses through review of infant charts using criteria listed in Table 2

In 2002, APORS began systematic active case verification. This has continued with some modifications to the criteria that establish which charts are reviewed. Details are given in Table 2. Each of these conditions has a high likelihood of being associated with one or more birth defects.

**Table 2: Criteria that Determine Which Charts are Reviewed**

<b>Birth years</b>	<b>Chart Review Criteria</b>
2002 -2007	<ul style="list-style-type: none"> <li>• one or more birth defects;</li> <li>• very low birth weight (&lt; 1500 g);</li> <li>• exposure to alcohol;</li> <li>• a diabetic mother;</li> <li>• a disturbance in neonatal tooth eruption; or</li> <li>• death before discharge.</li> </ul>

2008 onwards	<ul style="list-style-type: none"> <li>• one or more of the birth defects included in this report;</li> <li>• a birth defect associated with birth defects included in this report;</li> <li>• exposure to alcohol;</li> <li>• a diabetic mother;</li> <li>• a disturbance in neonatal tooth eruption; or</li> <li>• death before discharge.</li> </ul>
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As the charts are reviewed, APORS staff correct and add to the information reported by the hospitals. Using active case verification, the APORS program has identified an additional 40 percent of the birth defects included in this report.

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. The 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 1 year of age, born in 2006 and 2007. Similar information is presented for Chicago alone.

As a result of the active case verification begun in 2002, the estimates of birth defect rates from 1989-2001 are not comparable to those for 2002 onwards. Therefore, for this report, the figures illustrating trends in birth defect rates show the data for all years, but indicate the change in collection methods by a break in the plotted lines.

## METHODS

### *Calculation and Interpretation of Rates and Confidence Intervals*

Forty-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 or associated with a fetal death were calculated as

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

Similar rates were calculated for selected congenital anomalies identified in children up to 1 year of age. The numbers of live births were obtained from the Department's master birth files, provided by the Department's Center for Health Statistics.

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 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 3.3.1, April 2008, 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.

In this report, the trends have been examined for 1989-2001, and 2002-2007 separately since data for these two time periods are not comparable because of the different data collection methods used during each.

### ***Multiple Comparisons***

Since this report examines a large number of birth defects, the corresponding statistical tests are subject to the “multiple comparison problem.” For a given birth defect, the observed rate is an estimate of the true birth defect rate in the population. When two rates from different times or groups are compared, statisticians will assert that the observed rates are evidence of the groups having differing birth defect rates, if the observed rates are so different that the chance of them coming from the same underlying population is less than 5 percent. The 5 percent type I error rate, however, suggests that when 100 comparisons are made, on average, five will provide statistical evidence that there are two true differing rates, when in fact there is no difference between the two groups. Therefore, the more comparisons that are made, the more may be statistically significant, just by chance. In this report, no explicit corrections were made for multiple comparisons; 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***

Birth defect rates for selected categories among Illinois newborns in 2006 and 2007 are presented in tables 4 and 5. The tables include rates based on the defects identified through the active case verification described above. The corresponding information for Chicago is presented in tables 6 and 7.

In general, rates for Chicago are lower than those for Illinois as a whole. Differences in hospital reporting are likely to account for at least part of this difference. Rates of heart defects – endocardial cushion defects (2007) and patent ductus arteriosus (2006 and 2007) – are significantly lower for Chicago than the rest of Illinois. In 2006, rates of cleft palate without cleft lip were lower in Chicago, although much more similar in 2007. For both years, rates of hypospadias/epispadias are also significantly lower for Chicago than for the rest of Illinois. Caution should be used in interpreting these results because of the large number of comparisons made (see the discussion of multiple comparisons above).

### ***Trend Analysis***

Two birth defect categories can only be analyzed over a short time period. Gastroschisis is an abdominal wall anomaly; the specific coding needed to identify the defects precisely has been used by the APORS birth defects registry since 2002. Hypospadias and epispadias are penile anomalies; the specific coding needed to identify these defects precisely has been used by the APORS birth defects registry since 1998. For each of these categories, therefore, there are only limited data available to examine trends.

For the remainder of the categories of birth defects, graphs of each birth defect rate over time are plotted in Figures 1 through 9. A regression line also is plotted for each birth defect, with the exception of aniridia for which there are too few cases to perform a regression analysis prior to 2002. For the 1989-2001 data, the regression lines are usually log-linear, but may be made up of several straight line segments with different slopes. Statistically significant trends were found for 10 birth defects during this time period (listed in 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). For the data collected between 2002 and 2007 three defects were found to have significant trends: hypoplastic left heart syndrome, Hirschsprung disease and gastroschisis. Since this is a short period of time, and reflects a time of changes in data collection methods, the results from this time period are less reliable. Only log-linear regression lines were fitted.

For several defects, the direction of the trends between 1989 and 2001 and between 2002 and 2007 are in different directions. These apparent direction changes may be explained by the instability of the estimates of trend because of the small number of time points available since 2002. They also may reflect changes in data collection as APORS staff became more familiar with the new methods, and as diagnoses were more clearly defined. Alternatively, the trends may be real; the situation should become clearer as further years of data are collected using the new approach.

Table 3 also includes two columns (average percentage change) that give an estimate of how quickly the rate is changing over time (1989-2001 and 2002-2007). For example, the rate of endocardial cushion defect was significantly increasing by an average of 6.2 percent each year between 1989 and 2001, while from 2002 to 2007 the rate for this defect increased 5.5 percent each year. However, the change for the second time period is not statistically significant.

**Table 3. Birth Defects Showing a Significant Trend in Incidence Rate  
Between 1989 and 2001 or Between 2002 and 2007**

<b>Selected Birth Defect<sup>1</sup></b>	<b>Significance of trend (P-value)</b>	<b>Average annual % change between 1989 and 2001<sup>2</sup></b>	<b>Significance of trend (P-value)</b>	<b>Average annual % change between 2002 and 2007<sup>2</sup></b>
Atrial septal defect	0.00	9.6	not significant	-2.6
Endocardial cushion defect	0.00	6.2	not significant	5.5
Ebstein anomaly	0.02	6.2	not significant	-9.9
Pulmonary valve stenosis and atresia	0.00	7.1	not significant	-7.4
Hypoplastic left heart syndrome	not significant	5.6	0.05	-6.1
Patent ductus arteriosus	0.00	3.8	not significant	-4.5
Coarctation of aorta	0.02	3.5	not significant	-2.8
Pyloric stenosis	0.01	8.0	not significant	-5.2
Hirschsprung disease	0.00	5.9	0.04	-9.9
Obstructive genitourinary blockage	0.00	5.3	not significant	1.6
Gastroschisis	--	--	0.04	18.5
Trisomy 18 (Edward Syndrome)	0.01	4.5	not significant	-5.4

<sup>1</sup> Birth defects listed in tables 4 and 5 but not in this table did not show any significant changes in incidence rate over time.

<sup>2</sup> This is a measure of how quickly the rate is changing over time.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2010.

***Defects Associated With Inadequate Folic Acid Intake***

In 1992, the U.S. Centers for Disease Control and Prevention recommended that all women of childbearing age consume 0.4 mg of folic acid daily. The recommendation originated in research indicating that inadequate levels of folic acid in the first weeks of pregnancy increased the risk of having a baby with a neural tube defect (MRC Vitamin Study Research Group). In 1996, the U.S. Food and Drug Administration agreed that enriched cereal grain products must be fortified with folic acid, as a means to reduce the rate of neural tube defects in the United States (with implementation mandatory by January 1, 1998). Since then, a number of other low folic acid levels have been potentially implicated with other birth defects: Canfield *et al.* examined 16 congenital anomalies, previously examined by others, and found 11 defects with significant changes in prevalence pre- and post-fortification. These included neural tube defects, certain heart defects, cleft palate, omphalocele, Down syndrome, renal agenesis, limb reduction defects, urinary tract defects, brain defects and pylorus muscle defects.

Folic acid intake has increased in the general population since fortification in 1996, in part because many foods (cereals and flours) have been fortified with folic acid. It might, therefore, be expected that declines in the defects believed to be associated with low folic acid levels would emerge. While folate levels in women of child-bearing age have more than doubled since fortification, there has been a modest decline since 1999 (McDowell *et al.*)

### ***Discussion of Illinois Results***

Between 1989 and 2001, spina bifida (without anencephalus) showed a marked, close to significant, decrease in the incidence rate of 2.0 percent each year ( $P=0.07$ ). However, since 2002 the slope of the regression line appears to have changed direction, although it is not significantly different from a horizontal line indicating no change of slope. It is possible that the reported decrease in women's folate levels may be giving rise to very modestly increasing rates of spina bifida. Alternatively, the increase is likely due to chance.

Anotia and microtia occur more often in multiple births; in addition, Hispanic and Asian women are more likely to have a baby with anotia or microtia than non-hispanic white women (Shaw *et al.*, Forrester & Hertz, Husain *et al.*) In Illinois between 1989 and 2007, 45.3 percent of anotia/microtia cases were born to Hispanic or Asian women; while they made up only 23.3 percent of births. The proportion of births to this group of women has been steadily increasing, and has more than doubled between 1989 and 2007. It is, therefore, not surprising that the rate of anotia/microtia is increasing in Illinois.

Between 1989 and 2001, six of the 13 cardiovascular defects increased significantly. This is in line with the findings reported by Botto *et al.* who reported increasing rates of ventricular septal defects, tetralogy of Fallot, atrioventricular defects and pulmonary stenosis. Much of these increases may be a result of the greater availability and use of diagnostic techniques. Between 2002 and 2007, most of these had decreased (although not significantly). Because there are only six years being analyzed, the trends for the later period are still unstable; this may explain the direction change. It is also possible that the use of diagnostic techniques is reaching a plateau, causing the trend in these defects to level.

The rates of pyloric stenosis observed in Illinois appeared to be increasing between 1989 and 2001. Between 2002 and 2007, there is a non-significant decrease. Other authors (Canfield *et al.*, Sommerfield *et al.*) have reported decreasing rates of pyloric stenosis over similar time frames. Since pyloric stenosis is often first diagnosed between 3 and 8 weeks of age, usually

well after discharge from hospital, the increasing rates in Illinois may be a result of improving case ascertainment. Hirschsprung's disease is another gastrointestinal condition usually diagnosed after discharge from hospital. The rates of this birth defect show rather different behaviors before and after 2001: increasing significantly between 1989 and 2001 but decreasing significantly between 2002 and 2007. Again, the significant increase may be a result of improving case ascertainment as the birth defect registry became better established.

Obstructive genitourinary blockages may not be symptomatic until several weeks into a baby's life. Substantially more cases are being identified through active case verification – tripling the number of cases found. Increases in the number of cases are found for both time periods, although only the 1989-2001 increase is statistically significant.

The rate of gastroschisis significantly increased between 2002 and 2007. These conditions are unlikely to be missed during a newborn stay, and so the observed increases cannot be explained by changing case identification. Incidence rate increases have been reported in other studies, (Merz and Forrester, Rankin et al., Tan et al., Martinez-Frias et al., Roeper et al.). The same studies indicate that younger women are more likely to have babies with gastroschisis. A recent study (Feldkamp et al.) demonstrated a significant association between gastroschisis and the presence of urinary tract infection plus sexually transmitted infection around the time of conception or during the first trimester.

Since incidence rates of Down syndrome (Trisomy 21) increase markedly with parental age (Jyothy *et al.*) Figure 10 examines the rates of Down syndrome by maternal age at time of delivery. There is no statistical evidence that rates of Down syndrome are changing in any of the three age groups (< 35 years old, 35-39 years old and 40 years and older). This suggests that the observed increase probably reflects the increasing number of pregnancies among older women in Illinois, rather than a change in the pattern of Down syndrome itself. The rate of Trisomy 18 increased between 1989 and 2001; this incidence of this condition also increases with maternal age (Baty et al.) so again the observed increase probably reflects the increasing number of pregnancies among older women in Illinois.

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

**2006-2007**

**Table 4. Number and Rate of Selected Birth Defects for 2006  
Illinois**

<b>Selected Birth Defects Groups</b>	<b>N</b>	<b>Rate<sup>1</sup></b>	<b>95% CI<sup>2</sup></b>
<b>A. Central Nervous System</b>			
Anencephalus	44	2.4	(1.8, 3.3)
Spina bifida without anencephalus	53	2.9	(2.2, 3.8)
Hydrocephalus without spina bifida	146	8.1	(6.8, 9.5)
Encephalocele	14	0.8	(0.4, 1.3)
Microcephalus	80	4.4	(3.5, 5.5)
<i>Total Selected CNS Defects</i>	337	18.7	(16.7, 20.8)
<b>B. Eye</b>			
Anophthalmos/Microphthalmos	22	1.2	(0.8, 1.8)
Congenital cataract	12	0.7	(0.3, 1.2)
Aniridia	0	0.0	(0.0, 0.2)
<i>Total Selected Eye Defects</i>	34	1.9	(1.3, 2.6)
<b>C. Ear</b>			
Anotia/Microtia	19	1.1	(0.6, 1.6)
<b>D. Cardiovascular</b>			
Common truncus	12	0.7	(0.3, 1.2)
Transposition of great vessels	54	3.0	(2.2, 3.9)
Tetralogy of Fallot	56	3.1	(2.3, 4.0)
Ventricular septal defect	637	35.3	(32.6, 38.1)
Atrial septal defect <sup>3</sup>	445	24.7	(22.4, 27.1)
Endocardial cushion defect	72	4.0	(3.1, 5.0)
Pulmonary valve stenosis and atresia	36	2.0	(1.4, 2.8)
Tricuspid valve stenosis and atresia	24	1.3	(0.9, 2.0)
Ebstein anomaly	9	0.5	(0.2, 0.9)
Aortic valve stenosis	10	0.6	(0.3, 1.0)
Hypoplastic left heart syndrome	34	1.9	(1.3, 2.6)
Patent ductus arteriosus <sup>3</sup>	316	17.5	(15.6, 19.5)
Coarctation of aorta	57	3.2	(2.4, 4.1)
<i>Total Selected Cardiovascular Defects</i>	1,762	97.6	(93.1, 102.3)
<b>F. Orofacial</b>			
Cleft palate without cleft lip	83	4.6	(3.7, 5.7)
Cleft lip with and without cleft palate	164	9.1	(7.7, 10.6)
Choanal atresia	23	1.3	(0.8, 1.9)
<i>Total Selected Orofacial Defects</i>	270	15.0	(13.2, 16.9)
<b>G. Gastrointestinal</b>			
Esophageal atresia/Tracheoesophageal fistula	38	2.1	(1.5, 2.9)
Rectal and large intestinal atresia/stenosis	68	3.8	(2.9, 4.8)
Pyloric stenosis	7	0.4	(0.2, 0.8)
Hirschsprung disease (congenital megacolon)	18	1.0	(0.6, 1.6)
Biliary atresia	7	0.4	(0.2, 0.8)
<i>Total Selected Gastrointestinal Defects</i>	138	7.6	(6.4, 9.0)

<b>Selected Birth Defects Groups</b>	<b>N</b>	<b>Rate<sup>1</sup></b>	<b>95% CI<sup>2</sup></b>
<b>H. GENITOURINARY</b>			
Renal agenesis/hypoplasia	64	3.5	(2.7, 4.5)
Bladder exstrophy	5	0.3	(0.1, 0.6)
Obstructive genitourinary defect	432	23.9	(21.7, 26.3)
Hypospadias/Epispadias	465	25.8	(23.5, 28.2)
<i>Total Selected Genitourinary Defects</i>	<i>966</i>	<i>53.5</i>	<i>(50.2, 57.0)</i>
<b>I. MUSCULOSKELETAL</b>			
Reduction deformity, upper limbs	53	2.9	(2.2, 3.8)
Reduction deformity, lower limbs	21	1.2	(0.7, 1.8)
Gastroschisis	99	5.5	(4.5, 6.7)
Congenital hip dislocation	60	3.3	(2.5, 4.3)
Diaphragmatic hernia	49	2.7	(2.0, 3.6)
<i>Total Selected Musculoskeletal Defects</i>	<i>282</i>	<i>15.6</i>	<i>(13.9, 17.6)</i>
<b>J. CHROMOSOMAL</b>			
Trisomy 13 (Patau syndrome)	17	0.9	(0.5, 1.5)
Trisomy 21 (Down syndrome)	249	13.8	(12.1, 15.6)
Trisomy 18 (Edward syndrome)	46	2.5	(1.9, 3.4)
<i>Total Selected Chromosomal Defects</i>	<i>312</i>	<i>17.3</i>	<i>(15.4, 19.3)</i>
<b><i>Total All Selected Defects</i></b>	<b><i>3,982</i></b>	<b><i>220.6</i></b>	<b><i>(213.8, 227.6)</i></b>

<sup>1</sup> Rate per 10,000 live births

<sup>2</sup> 95 percent confidence interval for rate

<sup>3</sup> Only children with gestational ages of 36 weeks or greater and who have another heart defect are included among cases with patent ductus arteriosus diagnoses and some atrial septal defect (ASD) cases (those where the chart describes a defect as either an ASD or a patent foramen ovale). These conditions are necessary for normal fetal circulation.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2010

**Table 5. Number and Rate of Selected Birth Defects for 2007  
Illinois**

<b>Selected Birth Defects Groups</b>	<b>N</b>	<b>Rate<sup>1</sup></b>	<b>95% CI<sup>2</sup></b>
<b>A. Central Nervous System</b>			
Anencephalus	21	1.2	(0.7, 1.8)
Spina bifida without anencephalus	51	2.8	(2.1, 3.7)
Hydrocephalus without spina bifida	129	7.1	(6.0, 8.5)
Encephalocele	13	0.7	(0.4, 1.2)
Microcephalus	76	4.2	(3.3, 5.3)
<i>Total Selected CNS Defects</i>	<i>290</i>	<i>16.1</i>	<i>(14.3, 18.0)</i>
<b>B. Eye</b>			
Anophthalmos/Microphthalmos	17	0.9	(0.5, 1.5)
Congenital cataract	14	0.8	(0.4, 1.3)
Aniridia	0	0.0	(0.0, 0.2)
<i>Total Selected Eye Defects</i>	<i>31</i>	<i>1.7</i>	<i>(1.2, 2.4)</i>
<b>C. Ear</b>			
Anotia/Microtia	27	1.5	(1.0, 2.2)
<b>D. Cardiovascular</b>			
Common truncus	8	0.4	(0.2, 0.9)
Transposition of great vessels	38	2.1	(1.5, 2.9)
Tetralogy of Fallot	63	3.5	(2.7, 4.5)
Ventricular septal defect	616	34.1	(31.5, 36.9)
Atrial septal defect <sup>3</sup>	402	22.3	(20.1, 24.6)
Endocardial cushion defect	67	3.7	(2.9, 4.7)
Pulmonary valve stenosis and atresia	47	2.6	(1.9, 3.5)
Tricuspid valve stenosis and atresia	21	1.2	(0.7, 1.8)
Ebstein anomaly	7	0.4	(0.2, 0.8)
Aortic valve stenosis	23	1.3	(0.8, 1.9)
Hypoplastic left heart syndrome	26	1.4	(0.9, 2.1)
Patent ductus arteriosus <sup>3</sup>	334	18.5	(16.6, 20.6)
Coarctation of aorta	45	2.5	(1.8, 3.3)
<i>Total Selected Cardiovascular Defects</i>	<i>1,697</i>	<i>94.0</i>	<i>(89.6, 89.6)</i>
<b>F. Orofacial</b>			
Cleft palate without cleft lip	78	4.3	(3.4, 5.4)
Cleft lip with and without cleft palate	144	8.0	(6.7, 9.4)
Choanal atresia	15	0.8	(0.5, 1.4)
<i>Total Selected Orofacial Defects</i>	<i>237</i>	<i>13.1</i>	<i>(11.5, 14.9)</i>
<b>G. Gastrointestinal</b>			
Esophageal atresia/Tracheoesophageal fistula	40	2.2	(1.6, 3.0)
Rectal and large intestinal atresia/stenosis	55	3.0	(2.3, 4.0)
Pyloric stenosis	8	0.4	(0.2, 0.9)
Hirschsprung disease (congenital megacolon)	19	1.1	(0.6, 1.6)
Biliary atresia	2	0.1	(0.0, 0.4)
<i>Total Selected Gastrointestinal Defects</i>	<i>124</i>	<i>6.9</i>	<i>(5.7, 8.2)</i>

<b>Selected Birth Defects Groups</b>	<b>N</b>	<b>Rate<sup>1</sup></b>	<b>95% CI<sup>2</sup></b>
<b>H. GENITOURINARY</b>			
Renal agenesis/hypoplasia	81	4.5	(3.6, 5.6)
Bladder exstrophy	5	0.3	(0.1, 0.6)
Obstructive genitourinary defect	400	22.2	(20.0, 24.4)
Hypospadias/Epispadias	441	24.4	(22.2, 26.8)
<i>Total Selected Genitourinary Defects</i>	<i>927</i>	<i>51.3</i>	<i>(48.1, 54.8)</i>
<b>I. MUSCULOSKELETAL</b>			
Reduction deformity, upper limbs	43	2.4	(1.7, 3.2)
Reduction deformity, lower limbs	28	1.6	(1.0, 2.2)
Gastroschisis	75	4.2	(3.3, 5.2)
Congenital hip dislocation	67	3.7	(2.9, 4.7)
Diaphragmatic hernia	38	2.1	(1.5, 2.9)
<i>Total Selected Musculoskeletal Defects</i>	<i>251</i>	<i>13.9</i>	<i>(12.2, 15.7)</i>
<b>J. CHROMOSOMAL</b>			
Trisomy 13 (Patau syndrome)	27	1.5	(1.0, 2.2)
Trisomy 21 (Down syndrome)	259	14.3	(12.7, 16.2)
Trisomy 18 (Edward syndrome)	39	2.2	(1.5, 3.0)
<i>Total Selected Chromosomal Defects</i>	<i>325</i>	<i>18.0</i>	<i>(16.1, 20.1)</i>
<b><i>Total All Selected Defects</i></b>	<b><i>3,909</i></b>	<b><i>216.5</i></b>	<b><i>(209.8, 223.4)</i></b>

<sup>1</sup> Rate per 10,000 live births

<sup>2</sup> 95 percent confidence interval for rate

<sup>3</sup> Only children with gestational ages of 36 weeks or greater and who have another heart defect are included among cases with patent ductus arteriosus diagnoses and some atrial septal defect (ASD) cases (those where the chart describes a defect as either an ASD or a patent foramen ovale). These conditions are necessary for normal fetal circulation.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2010

**Table 6. Number and Rate of Selected Birth Defects for 2006  
Chicago**

<b>Selected Birth Defects Groups</b>	<b>N</b>	<b>Rate<sup>1</sup></b>	<b>95% CI<sup>2</sup></b>
<b>A. Central Nervous System</b>			
Anencephalus	9	2.0	(0.9, 3.7)
Spina bifida without anencephalus	9	2.0	(0.9, 3.7)
Hydrocephalus without spina bifida	36	7.9	(5.5, 10.9)
Encephalocele	5	1.1	(0.4, 2.5)
Microcephalus	26	5.7	(3.7, 8.3)
<i>Total Selected CNS Defects</i>	85	18.5	(14.8, 22.9)
<b>B. Eye</b>			
Anophthalmos/Microphthalmos	3	0.7	(0.1, 1.9)
Congenital cataract	0	0.0	(0.0, 0.8)
Aniridia	0	0.0	(0.0, 0.8)
<i>Total Selected Eye Defects</i>	3	0.7	(0.1, 1.9)
<b>C. Ear</b>			
Anotia/Microtia	3	0.7	(0.1, 1.9)
<b>D. Cardiovascular</b>			
Common truncus	1	0.2	(0.0, 1.2)
Transposition of great vessels	15	3.3	(1.8, 5.4)
Tetralogy of Fallot	14	3.1	(1.7, 5.1)
Ventricular septal defect	142	31.0	(26.1, 36.5)
Atrial septal defect <sup>3</sup>	89	19.4	(15.6, 23.9)
Endocardial cushion defect	11	2.4	(1.2, 4.3)
Pulmonary valve stenosis and atresia	7	1.5	(0.6, 3.1)
Tricuspid valve stenosis and atresia	4	0.9	(0.2, 2.2)
Ebstein anomaly	1	0.2	(0.0, 1.2)
Aortic valve stenosis	0	0.0	(0.0, 0.8)
Hypoplastic left heart syndrome	6	1.3	(0.5, 2.8)
Patent ductus arteriosus <sup>3</sup>	53	11.6	(8.7, 15.1)
Coarctation of aorta	9	2.0	(0.9, 3.7)
<i>Total Selected Cardiovascular Defects</i>	352	76.8	(69., 85.2)
<b>F. Orofacial</b>			
Cleft palate without cleft lip	7	1.5	(0.6, 3.1)
Cleft lip with and without cleft palate	33	7.2	(5.0, 10.1)
Choanal atresia	5	1.1	(0.4, 2.5)
<i>Total Selected Orofacial Defects</i>	45	9.8	(7.2, 13.1)
<b>G. Gastrointestinal</b>			
Esophageal atresia/Tracheoesophageal fistula	10	2.2	(1.0, 4.0)
Rectal and large intestinal atresia/stenosis	17	3.7	(2.2, 5.9)
Pyloric stenosis	2	0.4	(0.1, 1.6)
Hirschsprung disease (congenital megacolon)	4	0.9	(0.2, 2.2)
Biliary atresia	2	0.4	(0.1, 1.6)
<i>Total Selected Gastrointestinal Defects</i>	35	7.6	(5.3, 10.6)

<b>Selected Birth Defects Groups</b>	<b>N</b>	<b>Rate<sup>1</sup></b>	<b>95% CI<sup>2</sup></b>
<b>H. GENITOURINARY</b>			
Renal agenesis/hypoplasia	12	2.6	(1.4, 4.6)
Bladder exstrophy	1	0.2	(0.0, 1.2)
Obstructive genitourinary defect	82	17.9	(14.2, 22.2)
Hypospadias/Epispadias	63	13.7	(10.6, 17.6)
<i>Total Selected Genitourinary Defects</i>	<i>158</i>	<i>34.5</i>	<i>(29.3, 40.3)</i>
<b>I. MUSCULOSKELETAL</b>			
Reduction deformity, upper limbs	5	1.1	(0.4, 2.5)
Reduction deformity, lower limbs	6	1.3	(0.5, 2.8)
Gastroschisis	22	4.8	(3.0, 7.3)
Congenital hip dislocation	8	1.7	(0.8, 3.4)
Diaphragmatic hernia	10	2.2	(1.0, 4.0)
<i>Total Selected Musculoskeletal Defects</i>	<i>51</i>	<i>11.1</i>	<i>(8.3, 14.6)</i>
<b>J. CHROMOSOMAL</b>			
Trisomy 13 (Patau syndrome)	1	0.2	(0.0, 1.2)
Trisomy 21 (Down syndrome)	60	13.1	(10.0, 16.8)
Trisomy 18 (Edward syndrome)	8	1.7	(0.8, 3.4)
<i>Total Selected Chromosomal Defects</i>	<i>69</i>	<i>15.1</i>	<i>(11.7, 19.0)</i>
<b><i>Total All Selected Defects</i></b>			
	<b>801</b>	<b>174.7</b>	<b>(162.8, 187.3)</b>

<sup>1</sup> Rate per 10,000 live births

<sup>2</sup> 95 percent confidence interval for rate

<sup>3</sup> Only children with gestational ages of 36 weeks or greater and who have another heart defect are included among cases with patent ductus arteriosus diagnoses and some atrial septal defect (ASD) cases (those where the chart describes a defect as either an ASD or a patent foramen ovale). These conditions are necessary for normal fetal circulation.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2010

**Table 7. Number and Rate of Selected Birth Defects for 2007  
Chicago**

<b>Selected Birth Defects Groups</b>	<b>N</b>	<b>Rate<sup>1</sup></b>	<b>95% CI<sup>2</sup></b>
<b>A. Central Nervous System</b>			
Anencephalus	6	1.3	(0.5, 2.8)
Spina bifida without anencephalus	11	2.4	(1.2, 4.3)
Hydrocephalus without spina bifida	32	6.9	(4.7, 9.8)
Encephalocele	5	1.1	(0.4, 2.5)
Microcephalus	23	5.0	(3.2, 7.5)
<i>Total Selected CNS Defects</i>	77	16.6	(13.1, 20.8)
<b>B. Eye</b>			
Anophthalmos/Microphthalmos	5	1.1	(0.4, 2.5)
Congenital cataract	6	1.3	(0.5, 2.8)
Aniridia	0	0.0	(0.0, 0.8)
<i>Total Selected Eye Defects</i>	11	2.4	(1.2, 4.3)
<b>C. Ear</b>			
Anotia/Microtia	12	2.6	(1.3, 4.5)
<b>D. Cardiovascular</b>			
Common truncus	4	0.9	(0.2, 2.2)
Transposition of great vessels	17	3.7	(2.1, 5.9)
Tetralogy of Fallot	16	3.5	(2.0, 5.6)
Ventricular septal defect	142	30.7	(25.9, 36.2)
Atrial septal defect <sup>3</sup>	95	20.5	(16.6, 25.1)
Endocardial cushion defect	2	0.4	(0.1, 1.6)
Pulmonary valve stenosis and atresia	8	1.7	(0.7, 3.4)
Tricuspid valve stenosis and atresia	5	1.1	(0.4, 2.5)
Ebstein anomaly	3	0.6	(0.1, 1.9)
Aortic valve stenosis	1	0.2	(0.0, 1.2)
Hypoplastic left heart syndrome	4	0.9	(0.2, 2.2)
Patent ductus arteriosus <sup>3</sup>	59	12.8	(9.7, 16.5)
Coarctation of aorta	8	1.7	(0.7, 3.4)
<i>Total Selected Cardiovascular Defects</i>	364	78.7	(70.8, 87.2)
<b>F. Orofacial</b>			
Cleft palate without cleft lip	15	3.2	(1.8, 5.3)
Cleft lip with and without cleft palate	27	5.8	(3.8, 8.5)
Choanal atresia	6	1.3	(0.5, 2.8)
<i>Total Selected Orofacial Defects</i>	48	10.4	(7.7, 13.8)
<b>G. Gastrointestinal</b>			
Esophageal atresia/Tracheoesophageal fistula	9	1.9	(0.9, 3.7)
Rectal and large intestinal atresia/stenosis	19	4.1	(2.5, 6.4)
Pyloric stenosis	3	0.6	(0.1, 1.9)
Hirschsprung disease (congenital megacolon)	5	1.1	(0.4, 2.5)
Biliary atresia	2	0.4	(0.1, 1.6)
<i>Total Selected Gastrointestinal Defects</i>	38	8.2	(5.8, 11.3)

<b>Selected Birth Defects Groups</b>	<b>N</b>	<b>Rate<sup>1</sup></b>	<b>95% CI<sup>2</sup></b>
<b>H. GENITOURINARY</b>			
Renal agenesis/hypoplasia	16	3.5	(2.0, 5.6)
Bladder exstrophy	2	0.4	(0.1, 1.6)
Obstructive genitourinary defect	94	20.3	(16.4, 24.9)
Hypospadias/Epispadias	67	14.5	(11.2, 18.4)
<i>Total Selected Genitourinary Defects</i>	<i>179</i>	<i>38.7</i>	<i>(33.2, 44.8)</i>
<b>I. MUSCULOSKELETAL</b>			
Reduction deformity, upper limbs	12	2.6	(1.3, 4.5)
Reduction deformity, lower limbs	3	0.6	(0.1, 1.9)
Gastroschisis	18	3.9	(2.3, 6.2)
Congenital hip dislocation	19	4.1	(2.5, 6.4)
Diaphragmatic hernia	11	2.4	(1.2, 4.3)
<i>Total Selected Musculoskeletal Defects</i>	<i>63</i>	<i>13.6</i>	<i>(10.5, 17.4)</i>
<b>J. CHROMOSOMAL</b>			
Trisomy 13 (Patau syndrome)	9	1.9	(0.9, 3.7)
Trisomy 21 (Down syndrome)	57	12.3	(9.3, 16.0)
Trisomy 18 (Edward syndrome)	7	1.5	(0.6, 3.1)
<i>Total Selected Chromosomal Defects</i>	<i>73</i>	<i>15.8</i>	<i>(12.4, 19.8)</i>
<b><i>Total All Selected Defects</i></b>			
	<b>865</b>	<b>187.0</b>	<b>(174.8, 199.9)</b>

<sup>1</sup> Rate per 10,000 live births

<sup>2</sup> 95 percent confidence interval for rate

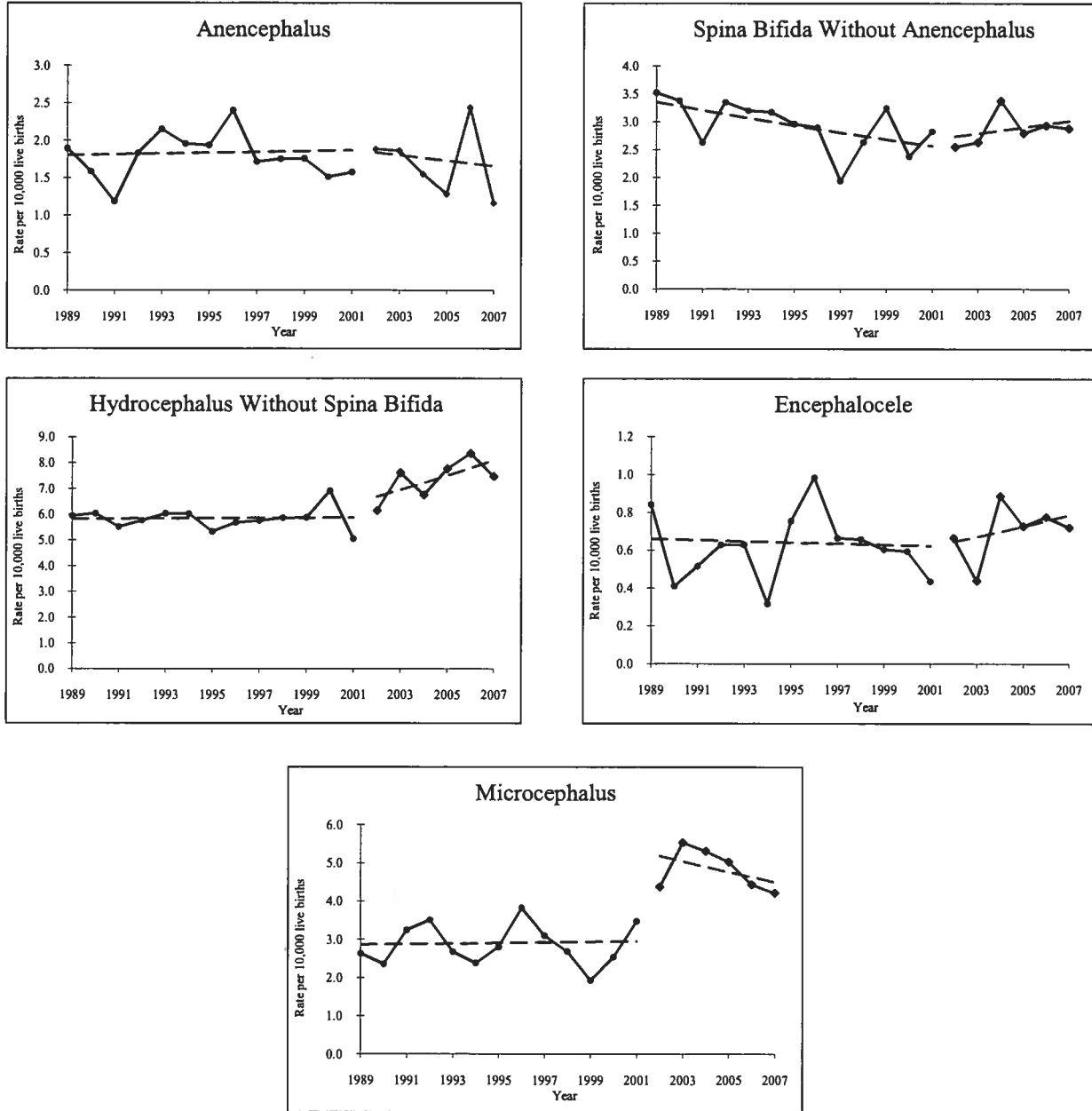
<sup>3</sup> Only children with gestational ages of 36 weeks or greater and who have another heart defect are included among cases with patent ductus arteriosus diagnoses and some atrial septal defect (ASD) cases (those where the chart describes a defect as either an ASD or a patent foramen ovale). These conditions are necessary for normal fetal circulation.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2010

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

**1989-2007**

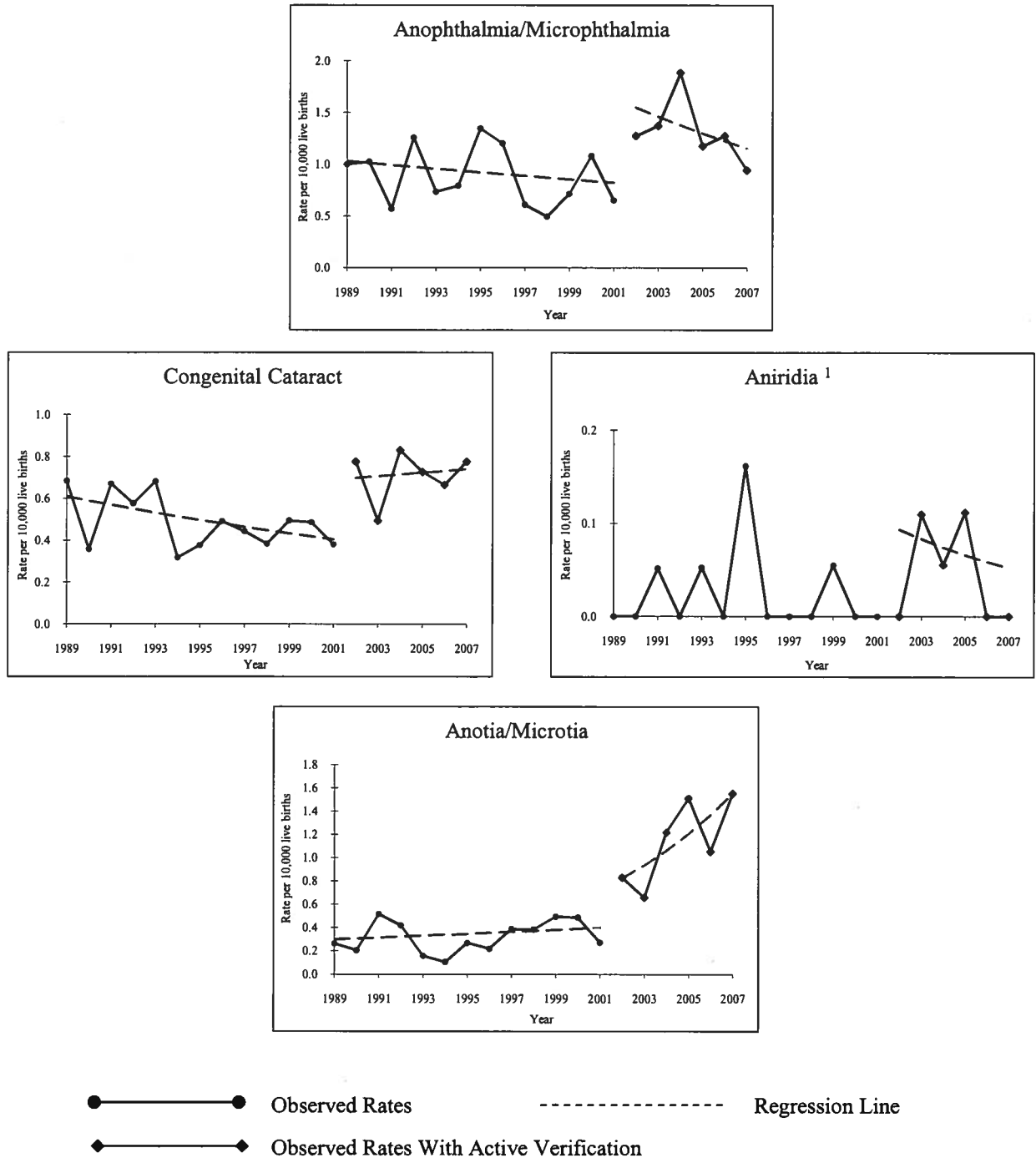
**Figure 1. Trends in the Reported Prevalence Rates of Neural Tube Defects per 10,000 Live Births 1989-2007**



● Observed Rates     
  Regression Line  
 ◆ Observed Rates With Active Verification

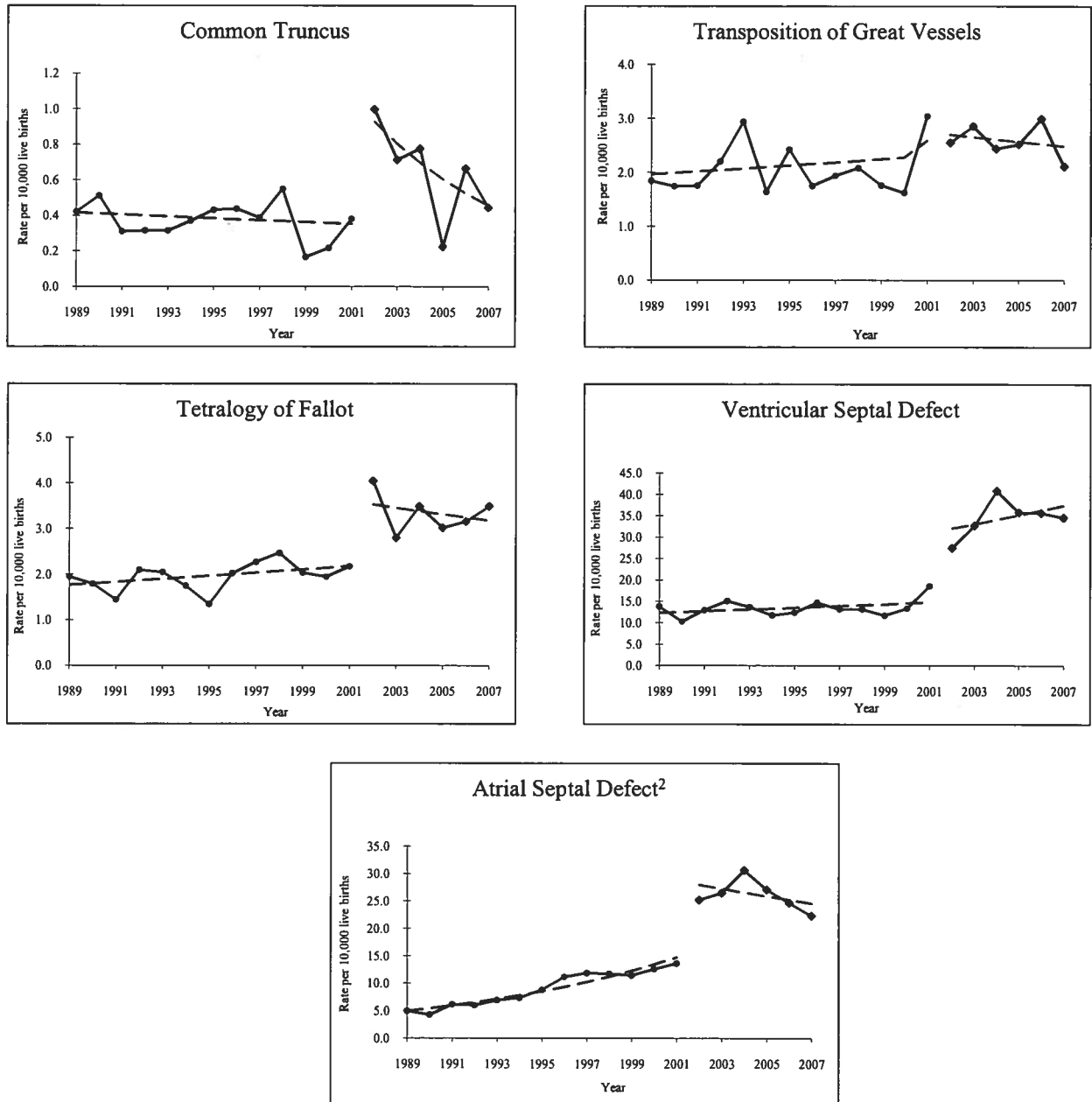
Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2010

**Figure 2. Trends in the Reported Prevalence Rates of Eye and Ear Defects per 10,000 Live Births 1989-2007**



<sup>1</sup>There is no regression line for aniridia (1989-2001) because there are too few cases to perform such an analysis.

**Figure 3A. Trends in the Reported Prevalence Rates of Cardiac Defects per 10,000 Live Births 1989-2007**

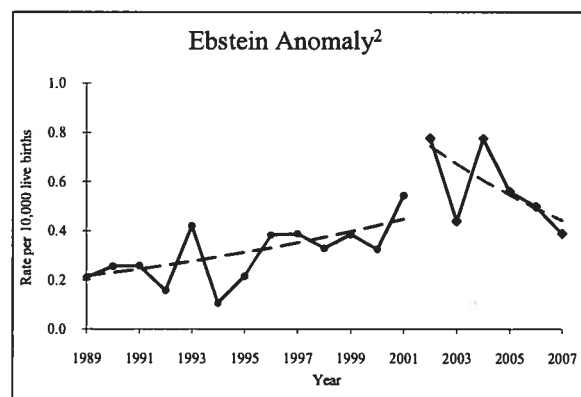
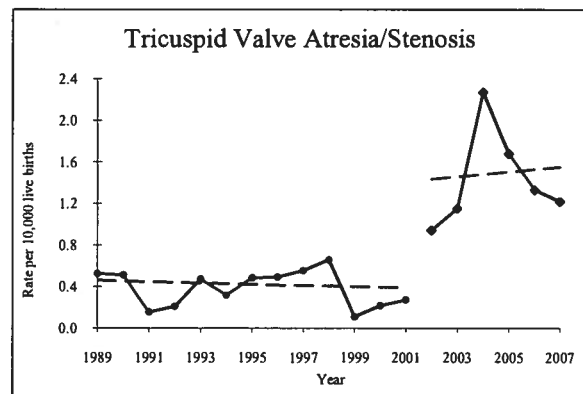
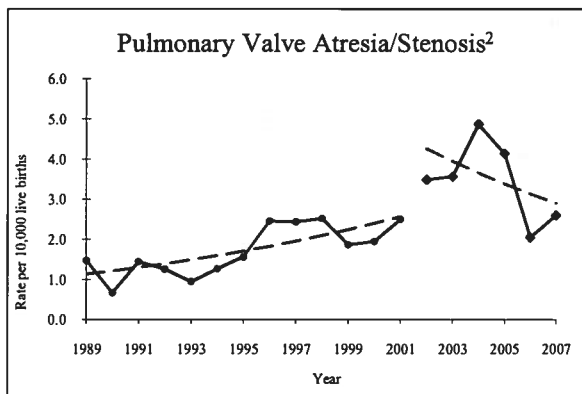
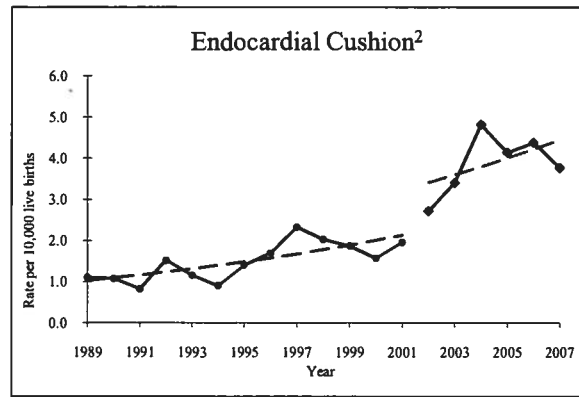


●—● Observed Rates      - - - - - Regression Line  
 ◆—◆ Observed Rates With Active Verification

<sup>2</sup>Trend is significant for 1989-2001, but not for 2002-2007; details are given in Table 2.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2010

**Figure 3B. Trends in the Reported Prevalence Rates of Cardiac Defects per 10,000 Live Births 1989-2007**

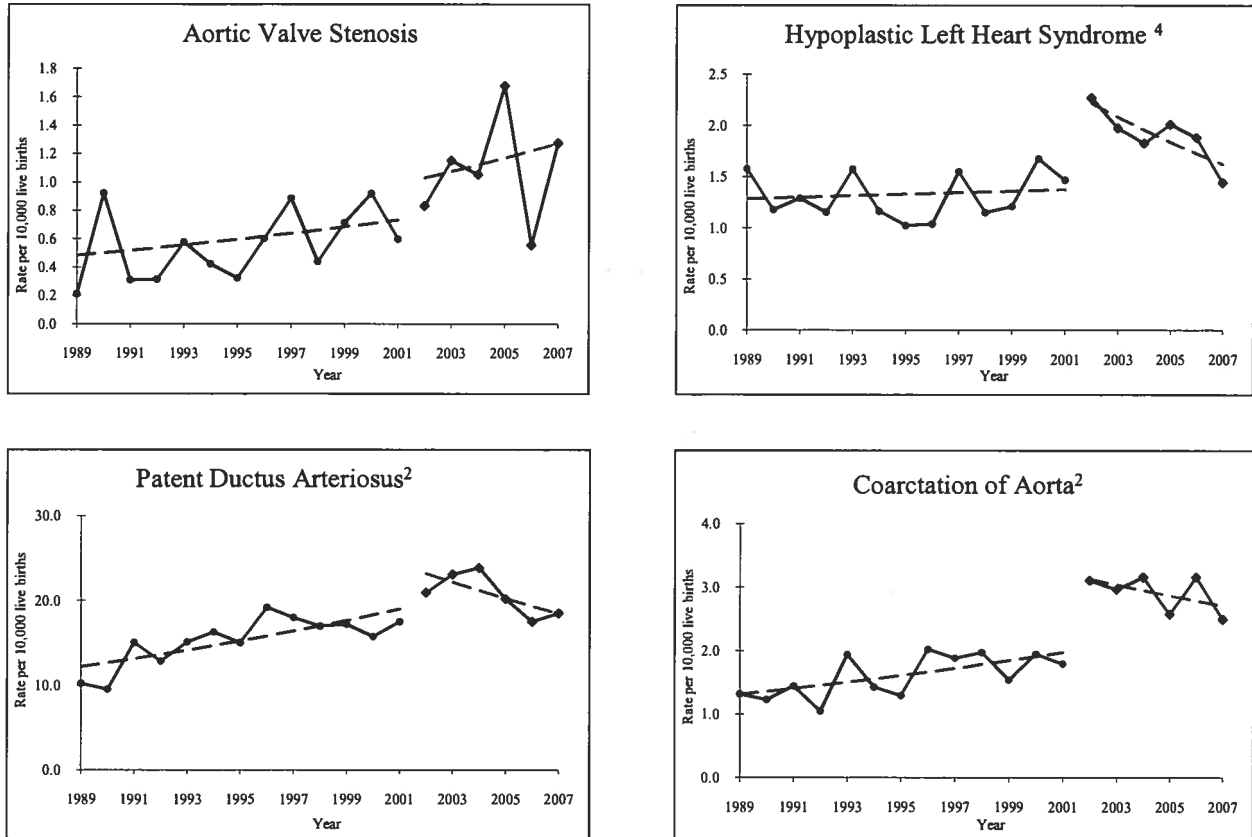


●● Observed Rates
 ----- Regression Line  
◆◆ Observed Rates With Active Verification

<sup>2</sup>Trend is significant for 1989-2001, but not for 2002-2007; details are given in Table 2.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2010

**Figure 4. Trends in the Reported Prevalence Rates of Circulatory Defects per 10,000 Live Births 1989-2007**



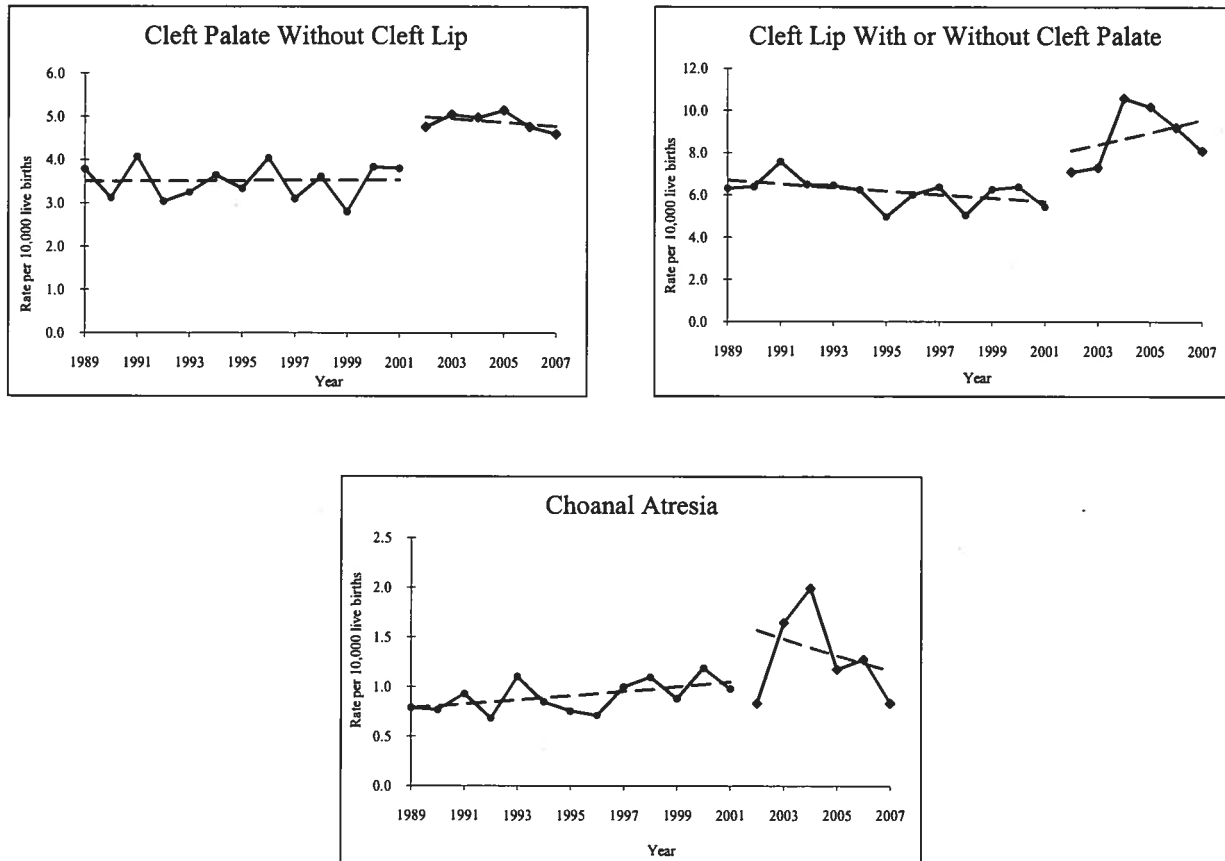
●—● Observed Rates      - - - - - Regression Line  
 ◆—◆ Observed Rates With Active Verification

<sup>2</sup>Trend is significant for 1989-2001, but not for 2002-2007; details are given in Table 2.

<sup>4</sup>Trend is significant for 2002-2007; details are given in Table 2.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2010

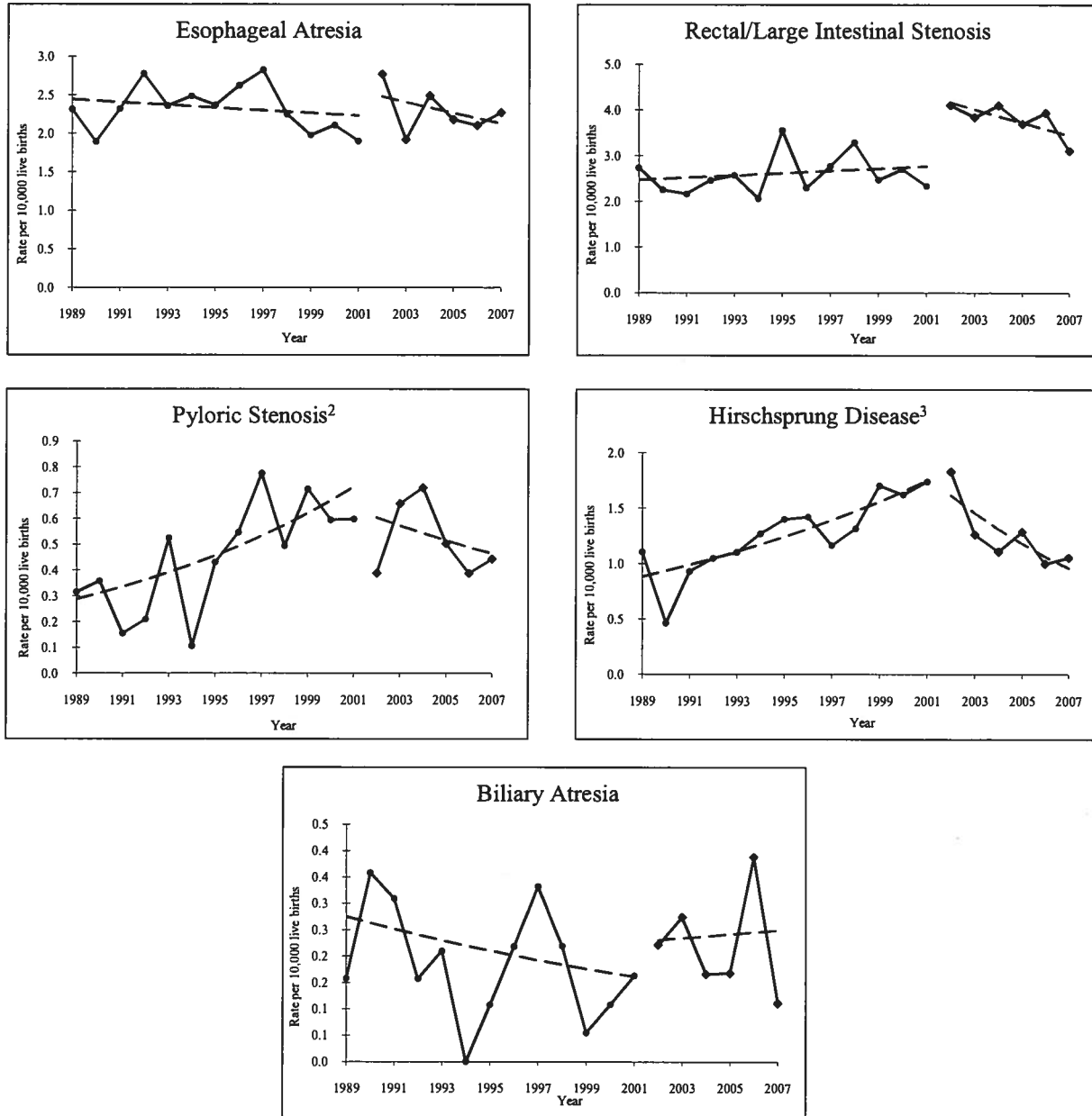
**Figure 5. Trends in the Reported Prevalence Rates of Respiratory and Oral Defects per 10,000 Live Births 1989-2007**



●—● Observed Rates      - - - - - Regression Line  
 ◆—◆ Observed Rates With Active Verification

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2010

**Figure 6. Trends in the Reported Prevalence Rates of Gastrointestinal Defects per 10,000 Live Births 1989-2007**



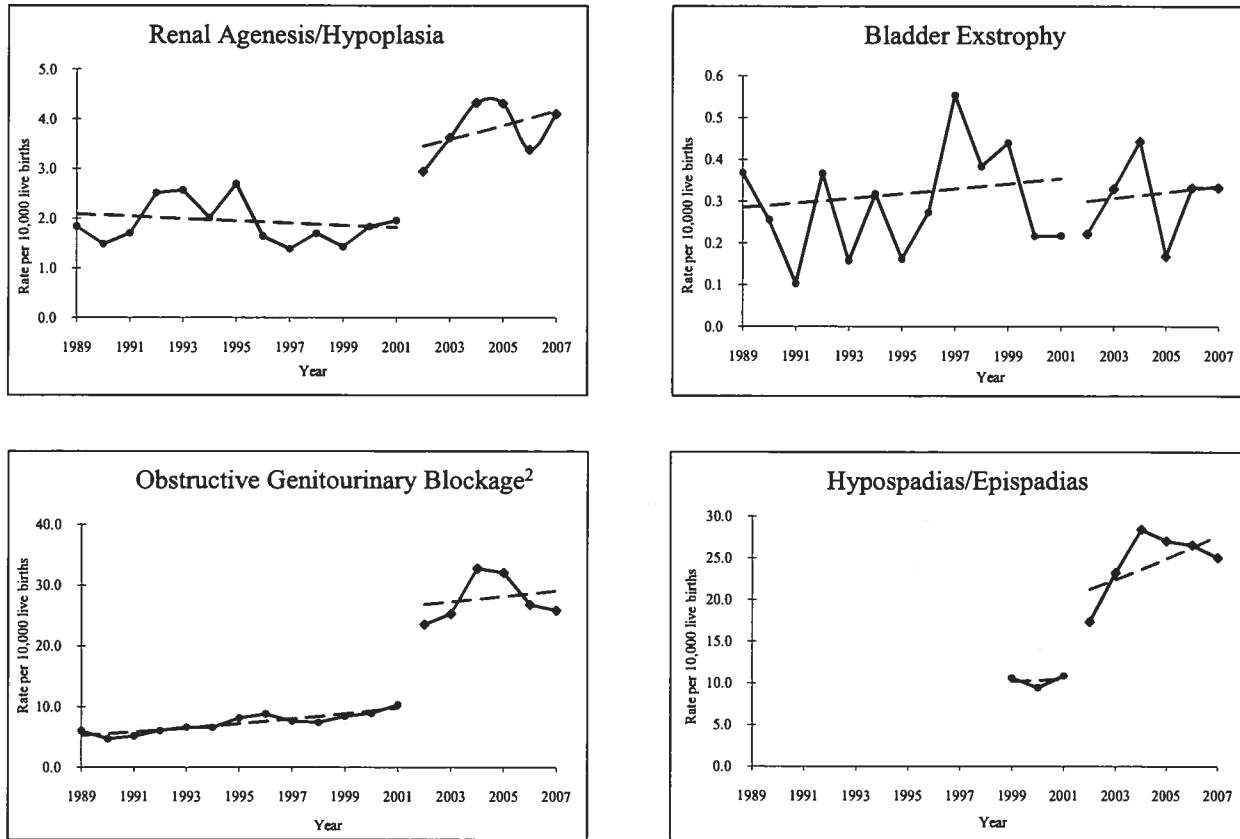
● Observed Rates     
  Regression Line  
◆ Observed Rates With Active Verification

<sup>2</sup>Trend is significant for 1989-2001, but not for 2002-2007; details are given in Table 2.

<sup>3</sup>Trend is significant for both 1989-2001 and 2002-2007; details are given in Table 2.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2010

**Figure 7. Trends in the Reported Prevalence Rates of Genitourinary Defects per 10,000 Live Births 1989-2007**

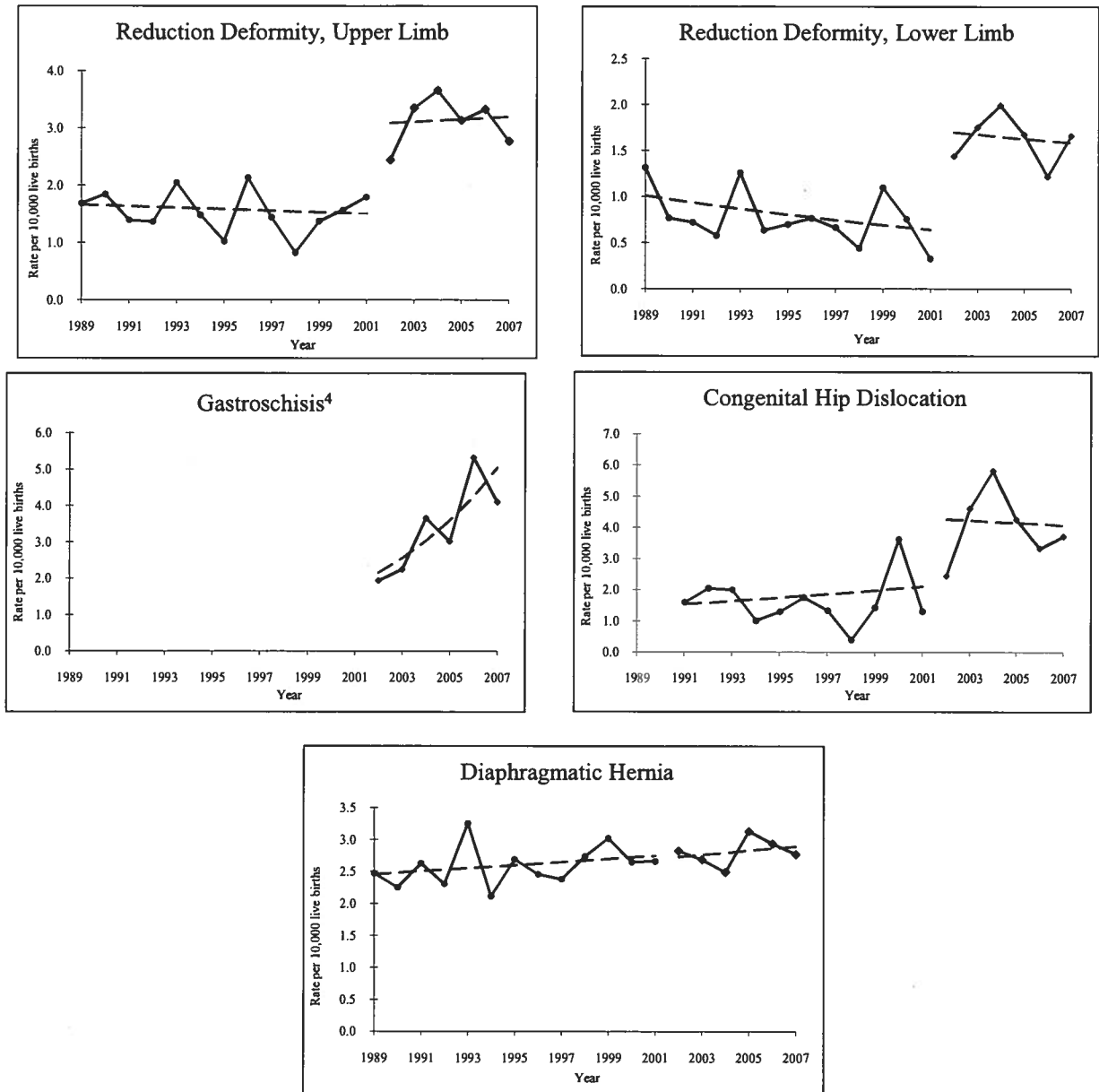


●—● Observed Rates      - - - - - Regression Line  
 ◆—◆ Observed Rates With Active Verification

<sup>2</sup>Trend is significant for 1989-2001, but not for 2002-2007; details are given in Table 2.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2010

**Figure 8. Trends in the Reported Prevalence Rates of Musculoskeletal Defects per 10,000 Live Births 1989-2007**

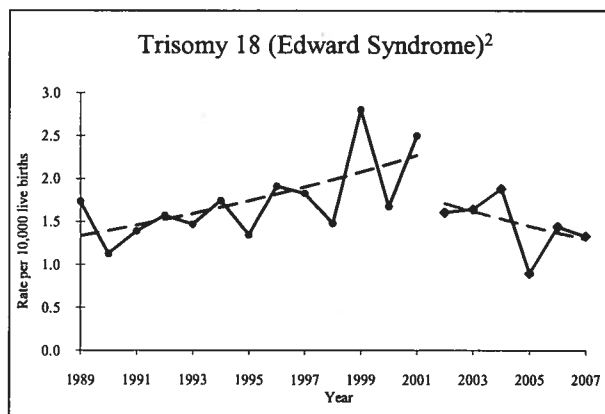
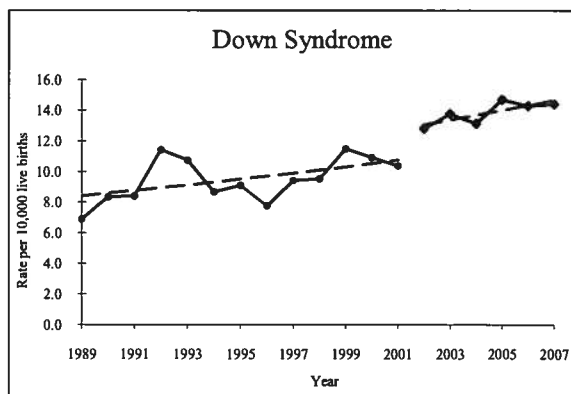
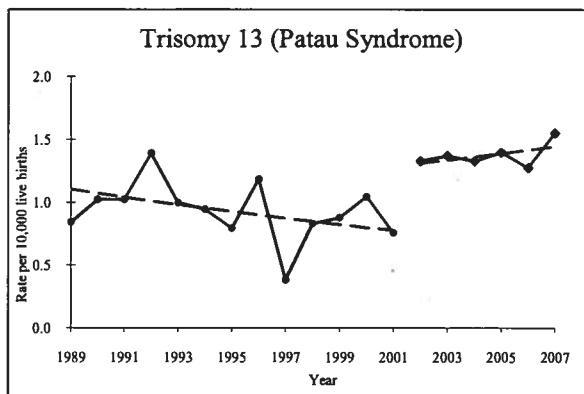


●—● Observed Rates      - - - - - Regression Line  
 ◆—◆ Observed Rates With Active Verification

<sup>4</sup>Trend is significant for 2002-2007; details are given in Table 2.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2010

**Figure 9. Trends in the Reported Prevalence Rates of Chromosomal Defects per 10,000 Live Births 1989-2007**

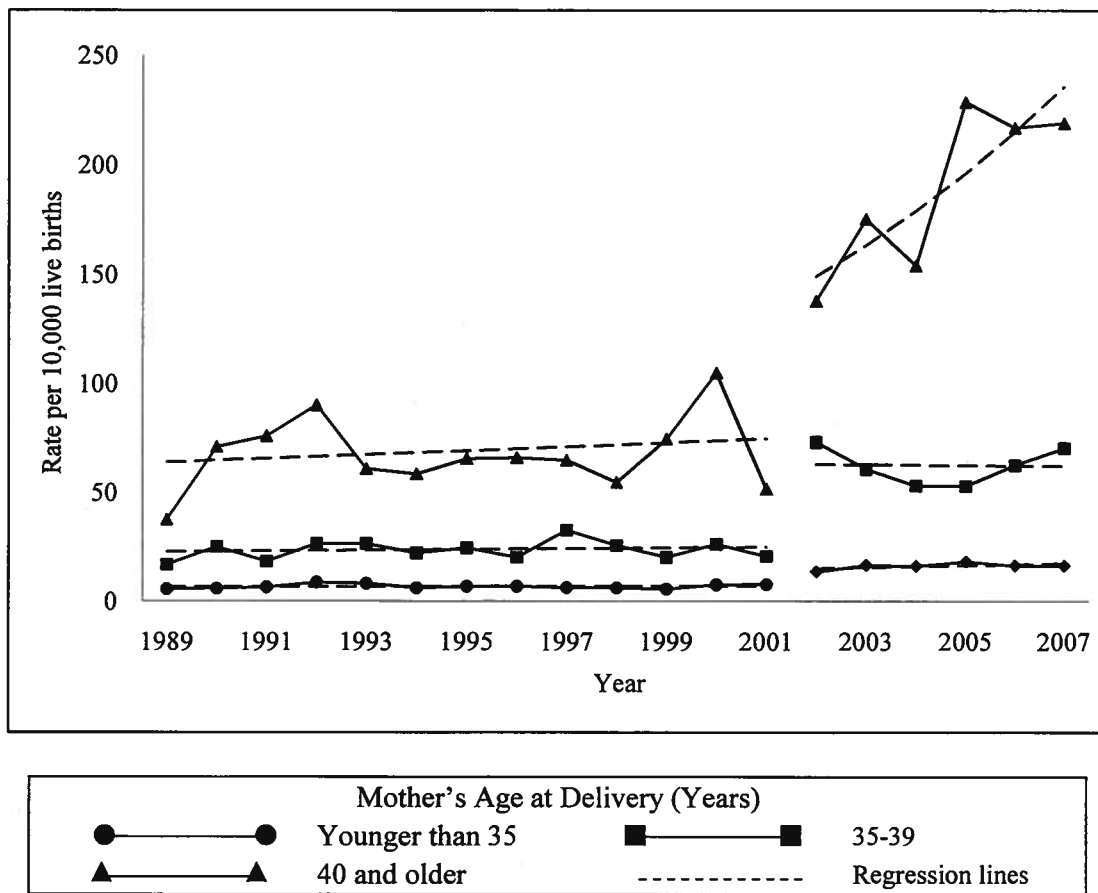


●● Observed Rates      - - - - - Regression Line  
◆◆ Observed Rates With Active Verification

<sup>2</sup>Trend is significant for 1989-2001, but not for 2002-2007; details are given in Table 2.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2010

**Figure 10. Trends in the Reported Prevalence Rates of Trisomy 21 (Down Syndrome),  
By Maternal Age at Delivery per 10,000 Live Births  
1989-2007**



Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2010

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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.
Anophthalmos	743.0x	Absence of the eye, as a result of a congenital malformation of the globe.
Microphthalmos	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.
Aniridia	743.45	Congenital absence of the iris in the eye.
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.

<b>Birth Defect</b>	<b>ICD-9-CM Codes</b>	<b>Description</b>
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.
Ventricular septal defect	745.4	A hole in the wall between the lower chambers of the heart.
Atrial septal defect	745.5	A hole in the wall between the upper chambers of the heart.
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 and atresia	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.
Patent ductus arteriosus	747.0	A condition when the channel between the pulmonary artery and the aorta fails to close at birth.
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	749.1x	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.

<b>Birth Defect</b>	<b>ICD-9-CM Codes</b>	<b>Description</b>
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.
Pyloric stenosis	750.5	A narrowing of the outlet from the stomach to the small intestine (the pylorus).
Rectal and large intestinal atresia and stenosis	751.2	Absence, abnormal localization or blockage of the large intestine or rectum.
Hirschsprung disease	751.3	A congenital abnormality of the bowel in which there is absence of the ganglia (nerves) in the wall of the bowel.
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.)
Epispadias	752.62	A rare congenital defect, most common in males, in which the urethra opens on the top (dorsal) surface of the penis. (In females, the opening to the urinary tract is above 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.
Congenital hip dislocation	754.30, 754.31, 354.35	A congenital defect in which the head of the femur does not articulate with the acetabulum of the pelvis because of an abnormal shallowness of the acetabulum.
Reduction deformity	755.2x, 755.3x	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.
Gastroschisis	756.79	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.

Birth Defect	ICD-9-CM Codes	Description
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.