

**TRENDS IN THE PREVALENCE OF  
BIRTH DEFECTS IN  
ILLINOIS AND CHICAGO  
1989-2001**

Illinois Department of Public Health  
Division of Epidemiologic Studies

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**TABLE OF CONTENTS**

INDEX OF TABLES AND FIGURES .....	ii
INTRODUCTION .....	1
METHODS .....	2
Calculation and Interpretation of Rates and Confidence Intervals .....	2
Analysis of Trends .....	3
Multiple Comparisons .....	3
FINDINGS .....	3
Rates of Birth Defects .....	3
Trend Analysis .....	4
Defects Associated with Inadequate Folic Acid Intake .....	4
Other Defects Showing Significant Trends .....	5
BIRTH DEFECT RATES FOR SELECTED CATEGORIES AMONG ILLINOIS AND CHICAGO NEWBORNS, 1996-1998 .....	7
TRENDS IN BIRTH DEFECT RATES FOR SELECTED CATEGORIES AMONG ILLINOIS NEWBORNS, 1989-2001 .....	20
REFERENCES .....	32
APPENDIX .....	33

## INDEX OF TABLES AND FIGURES

Table 1. Projects to identify cases and birth defects.....	2
Table 2. Birth Defects Associated with Low Intake of Folic Acid Showing a Significant Trend in Incidence Rate Between 1989 and 2001 .....	5
Table 3. Birth Defects Not Believed to Be Associated with Folic Acid Intake Showing a Significant Trend Between 1989 and 2001 .....	6
Table 4. Number and Rate of Selected Birth Defects for 2001; Illinois .....	8
Table 5. Number and Rate of Selected Birth Defects for 2000; Illinois .....	10
Table 6. Number and Rate of Selected Birth Defects for 1999; Illinois .....	12
Table 7. Number and Rate of Selected Birth Defects for 2001; Chicago .....	14
Table 8. Number and Rate of Selected Birth Defects for 2000; Chicago .....	16
Table 9. Number and Rate of Selected Birth Defects for 1999; Chicago .....	18
Figure 1. Trends in The Reported Prevalence Rates of Neural Tube Defects Identified During The Newborn Stay, Per 10,000 Live Births 1989-2001 .....	21
Figure 2. Trends in The Reported Prevalence Rates of Eye And Ear Defects Identified During The Newborn Stay, Per 10,000 Live Births 1989-2001 .....	22
Figure 3a. Trends in The Reported Prevalence Rates of Cardiac Defects Identified During The Newborn Stay, Per 10,000 Live Births 1989-2001 .....	23
Figure 3b. Trends in The Reported Prevalence Rates of Cardiac Defects Identified During The Newborn Stay, Per 10,000 Live Births 1989-2001 .....	24
Figure 4. Trends in The Reported Prevalence Rates of Circulatory Defects Identified During The Newborn Stay, Per 10,000 Live Births 1989-2001 .....	25
Figure 5. Trends in The Reported Prevalence Rates of Respiratory And Oral Defects Identified During The Newborn Stay, Per 10,000 Live Births 1989-2001 .....	26
Figure 6. Trends in The Reported Prevalence Rates of Gastrointestinal Defects Identified During The Newborn Stay, Per 10,000 Live Births 1989-2001 .....	27
Figure 7. Trends in The Reported Prevalence Rates of Genitourinary Defects Identified During The Newborn Stay, Per 10,000 Live Births 1989-2001 .....	28
Figure 8. Trends in The Reported Prevalence Rates of Musculoskeletal Defects Identified During The Newborn Stay, Per 10,000 Live Births 1989-2001 .....	29
Figure 9. Trends in The Reported Prevalence Rates of Chromosomal Defects Identified During The Newborn Stay, Per 10,000 Live Births 1989-2001 .....	30
Figure 10. Trends in The Reported Prevalence Rates of Down Syndrome, by Maternal Age at Delivery, Identified During The Newborn Stay, Per 10,000 Live Births 1989-2001 .....	31

## 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. Information about congenital anomalies identified in newborn infants was first collected statewide by APORS in 1989.

This information is collected for two major reasons. First, infants with a birth defect often need special services to help assure that they reach their full potential. These babies are referred to the Illinois Department of Human Services 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 and the fifth-leading cause of years of potential life lost in the United States, and they contribute substantially to childhood morbidity and long-term disability. There are three major categories of known causes:

- \$ chromosomal disorders (either hereditary or arising during conception)
- \$ exposure to an environmental chemical (for example, medications, alcohol, cigarettes, or solvents)
- \$ mother's illness during pregnancy, exposing her baby to viral or bacterial infection.

The stage of fetal development at the time of exposure to one of the latter two 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. 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 born to Illinois women. (Perinatal centers in St. Louis voluntarily participate.) APORS is a passive surveillance system, since reports are sent to IDPH rather than APORS staff going to hospitals to identify children with birth defects. 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 on the next page. More information about these studies is available from APORS.

**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
1999-2002	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.

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 1999, 2000 and 2001. Similar information is presented for Chicago alone.

## METHODS

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

Forty-eight 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 IDPH 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 is 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 2.7, September 2003, 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***

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, suggest 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 of the multiple comparison problem were made; instead, exact probabilities are reported when discussing trends. The smaller the reported probability, the more likely 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 2001 are presented in Table 4. Birth defect rates for 2000 and 1999 are presented in tables 5 and 6 respectively. These latter tables include rates for both Illinois newborns and Illinois infants up to 1 year of age. The corresponding information for Chicago is presented in tables 7, 8 and 9.

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 anencephalus, ventricular septal defect, atrial septal defect, pulmonary valve stenosis and atresia, patent ductus arteriosus, pulmonary artery anomalies, cleft palate, cleft lip, renal agenesis/hypoplasia, penile anomalies, hypospadias/epispadias, club foot and Down syndrome are significantly lower for Chicago than for the rest of Illinois. Rates of microcephalus are significantly higher 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/omphalocele cases also are contained in the category of abdominal wall anomalies and hypospadias/epispadias cases also are contained in the category of penile anomalies. In each case, the specific coding needed to identify the defects more precisely was not used by the APORS birth defects registry until 1998 and, 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 is also plotted for each birth defect, with the exception of aniridia for which there are too few cases to perform a regression analysis. The regression lines are usually log-linear, but may be made up of two straight lines with different slopes. Statistically significant trends were found for 13 birth defects (listed in tables 2 and 3). Although examination of the graphs may show some other birth defects with a marked trend, the small number of cases means that the slope is not statistically significantly different from horizontal (no change with time).

Tables 2 and 3 also include a column (average percentage change) that gives an estimate of how quickly the rate is changing over time. For example, the rate of spina bifida without anencephalus is significantly decreasing by an average of 2.2 percent each year.

### ***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). Since then, a number of other low folic acid levels have been potentially implicated with other birth defects: certain heart defects, cleft palate and lip, Down syndrome, limb reduction defects, urinary tract defects, brain defects and pylorus muscle defects (Werler *et al.*, James *et al.*).

Folic acid intake has increased in the general population, partly 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.

Several of the defects are showing a significant decrease in the incidence rate, while others are increasing. Low folic acid intake is only one reason that these defects might develop. For example, incidence rates of Down syndrome increase markedly with parental age (Jyothy *et al.*).

In addition, pyloric stenosis and obstructive genitourinary blockage may not be symptomatic until several weeks into a baby's life. Improved and more readily available diagnostic techniques may have contributed to the increase in diagnosis during the newborn stay.

**Table 2. Birth Defects Associated with Low Intake of Folic Acid Showing a Significant Trend in Incidence Rate Between 1989 and 2001**

<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>
Spina bifida without anencephalus	0.0347	-2.2%
Pyloric stenosis	0.0263	7.6%
Obstructive genitourinary blockage	0.0001	5.6%

<sup>1</sup> Birth defects listed in tables 4-9 but not in this table nor in Table 3 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. The time interval is 1989-2001.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2003.

The regression line for Down syndrome is very close to significance ( $p = 0.0535$ ), with an annual percentage change of 2.1 percent. 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.

***Other Defects Showing Significant Trends***

One defect unassociated with folic acid intake, club foot, showed statistical evidence of a decline between 1989 and 2001 (Table 3). Collection protocols have placed less emphasis on reporting club foot since it is readily corrected and this, in turn, may have contributed to the decline in observed incidence rates.

Hirschsprung disease is believed to be a result of genetic abnormalities. It is a condition in which improved and more readily available diagnostic techniques may have contributed to the increase in diagnosis during the newborn stay. The Hawaii Birth Defects Program (Merz and Forrester) reports declining rates of this birth defect. The Hawaii program uses active case management to identify cases through 1 year of age, so that the data collected would be less susceptible to variations resulting from changes in use of diagnostic techniques.

**Table 3: Birth Defects Not Believed to Be Associated with Folic Acid Intake  
Showing a Significant Trend Between 1989 and 2001**

Selected Birth Defect <sup>1</sup>	Significance of trend (P-value)	Average % change between 1989 and 2001 <sup>2</sup>
Atrial septal defect	0.0001	11.0%
Endocardial cushion defect	0.0032	6.3%
Ebstein Anomaly	0.0137	6.5%
Pulmonary valve stenosis and atresia	0.0041	7.2%
Coarctation of aorta	0.0151	3.6%
Pulmonary artery anomalies	0.0123	3.8%
Hirschsprung disease	0.0103	6.0%
Abdominal wall anomalies	0.0001	7.2%
Club foot	0.0006	-5.0%
Trisomy 18	0.0247	3.8%

<sup>1</sup> Birth defects listed in the tables 4-9 but not in this table nor in Table 2 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 2003

The rates of abdominal wall anomalies significantly increased between 1989 and 1998. 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, particularly with regard to gastroschisis (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. There are thought to be a number of causes of abdominal wall anomalies, including a genetic component.

The remaining defects are cardiovascular and are all significantly increasing. 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. However again, some of the increases may be a result of changes in the availability of diagnostic techniques.

APORS staff have also been working to improve awareness of birth defects and the need to report them to the Illinois birth defects registry. This activity may have contributed to the increases, but should have been effective over the whole range of birth defects.

**Birth Defect Rates for Selected Categories  
Among  
Illinois and Chicago Newborns  
1999-2001**

**Table 4. Number and Rate of Selected Birth Defects for 2001  
Illinois**

Selected Birth Defects Groups	Newborn Identification			Up to 1 year Identification		
	N	Rate <sup>1</sup>	95% CI <sup>2</sup>	N	Rate <sup>1</sup>	95% CI <sup>2</sup>
<b>A. CENTRAL NERVOUS SYSTEM</b>						
Anencephalus	30	1.63	(1.10, 2.33)	35	1.90	(1.32, 2.65)
Spina bifida without anencephalus	51	2.77	(2.06, 3.64)	51	2.77	(2.06, 3.64)
Hydrocephalus without spina bifida	92	5.00	(4.03, 6.13)	94	5.11	(4.13, 6.25)
Encephalocele	8	0.43	(0.19, 0.86)	8	0.43	(0.19, 0.86)
Microcephalus	62	3.37	(2.58, 4.32)	66	3.59	(2.77, 4.56)
<i>Total Selected CNS Defects</i>	<i>243</i>	<i>13.20</i>	<i>(11.60, 14.97)</i>	<i>254</i>	<i>13.80</i>	<i>(12.16, 15.61)</i>
<b>B. EYE</b>						
Coloboma of the eye	3	0.16	(0.03, 0.48)	3	0.16	(0.03, 0.48)
Anophthalmos/Microphthalmos	13	0.71	(0.38, 1.21)	13	0.71	(0.38, 1.21)
Congenital cataract	7	0.38	(0.15, 0.78)	7	0.38	(0.15, 0.78)
Aniridia	0	0.00	(0.00, 0.20)	0	0.00	(0.00, 0.20)
<i>Total Selected Eye Defects</i>	<i>23</i>	<i>1.25</i>	<i>(0.79, 1.88)</i>	<i>23</i>	<i>1.25</i>	<i>(0.79, 1.88)</i>
<b>C. EAR</b>						
Anotia/Microtia	5	0.27	(0.09, 0.63)	5	0.27	(0.09, 0.63)
<b>D. CARDIOVASCULAR</b>						
Common truncus	7	0.38	(0.15, 0.78)	7	0.38	(0.15, 0.78)
Transposition of great vessels	55	2.99	(2.25, 3.89)	55	2.99	(2.25, 3.89)
Tetralogy of fallot	40	2.17	(1.55, 2.96)	40	2.17	(1.55, 2.96)
Ventricular septal defect	339	18.42	(16.51, 20.49)	345	18.75	(16.82, 20.83)
Atrial septal defect	414	22.50	(20.38, 24.77)	422	22.93	(20.80, 25.23)
Endocardial cushion defect	36	1.96	(1.37, 2.71)	36	1.96	(1.37, 2.71)
Pulmonary valve stenosis and atresia	45	2.45	(1.78, 3.27)	47	2.55	(1.88, 3.40)
Tricuspid valve stenosis and atresia	5	0.27	(0.09, 0.63)	5	0.27	(0.09, 0.63)
Ebstein anomaly	10	0.54	(0.26, 1.00)	10	0.54	(0.26, 1.00)
Aortic valve stenosis	11	0.60	(0.30, 1.07)	11	0.60	(0.30, 1.07)
Hypoplastic left heart syndrome	28	1.52	(1.01, 2.20)	29	1.58	(1.06, 2.26)
Patent ductus arteriosus	790	42.93	(39.99, 46.03)	804	43.69	(40.72, 46.82)
Coarctation of aorta	34	1.85	(1.28, 2.58)	34	1.85	(1.28, 2.58)
Pulmonary artery anomalies	212	11.52	(10.02, 13.18)	218	11.85	(10.33, 13.53)
<i>Total Selected Cardiovascular Defects</i>	<i>2,026</i>	<i>110.10</i>	<i>(105.35, 115.00)</i>	<i>2,063</i>	<i>112.11</i>	<i>(107.32, 117.05)</i>
<b>E. RESPIRATORY</b>						
Lung agenesis/hypoplasia	59	3.21	(2.44, 4.14)	62	3.37	(2.58, 4.32)
<b>F. OROFACIAL</b>						
Cleft palate without cleft lip	70	3.80	(2.97, 4.81)	70	3.80	(2.97, 4.81)
Cleft lip with and without cleft palate	100	5.43	(4.42, 6.61)	102	5.54	(4.52, 6.73)
Choanal atresia	18	0.98	(0.58, 1.55)	18	0.98	(0.58, 1.55)
<i>Total Selected Orofacial Defects</i>	<i>188</i>	<i>10.22</i>	<i>(8.81, 11.79)</i>	<i>190</i>	<i>10.32</i>	<i>(8.91, 11.90)</i>
<b>G. GASTROINTESTINAL</b>						
Esophageal atresia/Tracheoesophageal fistula	35	1.90	(1.32, 2.65)	35	1.90	(1.32, 2.65)
Rectal and large intestinal atresia/stenosis	43	2.34	(1.69, 3.15)	43	2.34	(1.69, 3.15)
Pyloric stenosis	9	0.49	(0.22, 0.93)	11	0.60	(0.30, 1.07)
Hirschsprung disease (congenital megacolon)	32	1.74	(1.19, 2.45)	33	1.79	(1.23, 2.52)
Biliary atresia	3	0.16	(0.03, 0.48)	3	0.16	(0.03, 0.48)
<i>Total Selected Gastrointestinal Defects</i>	<i>122</i>	<i>6.63</i>	<i>(5.51, 7.92)</i>	<i>125</i>	<i>6.79</i>	<i>(5.65, 8.09)</i>

Selected Birth Defects Groups	Newborn Identification			Up to 1 year Identification		
	N	Rate <sup>1</sup>	95% CI <sup>2</sup>	N	Rate <sup>1</sup>	95% CI <sup>2</sup>
<b>H. GENITOURINARY</b>						
Renal agenesis/hypoplasia	36	1.96	(1.37, 2.71)	42	2.28	(1.64, 3.09)
Bladder exstrophy	4	0.22	(0.06, 0.56)	4	0.22	(0.06, 0.56)
Obstructive genitourinary defect	196	10.65	(9.21, 12.25)	211	11.47	(9.97, 13.12)
Penile anomalies	259	14.07	(12.41, 15.90)	259	14.07	(12.41, 15.90)
Hypospadias/Epispadias <sup>3</sup>	201	10.92	(9.46, 12.54)	214	11.63	(10.12, 13.30)
<i>Total Selected Genitourinary Defects</i>	<i>495</i>	<i>26.90</i>	<i>(24.58, 29.38)</i>	<i>516</i>	<i>28.04</i>	<i>(25.67, 30.57)</i>
<b>I. MUSCULOSKELETAL</b>						
Club foot	157	8.53	(7.25, 9.98)	164	8.91	(7.60, 10.39)
Reduction deformity, upper limbs	33	1.79	(1.23, 2.52)	33	1.79	(1.23, 2.52)
Reduction deformity, lower limbs	6	0.33	(0.12, 0.71)	6	0.33	(0.12, 0.71)
Abdominal wall defects	107	5.81	(4.77, 7.03)	107	5.81	(4.77, 7.03)
Gastroschisis/Omphalocele <sup>3</sup>	105	5.71	(4.67, 6.91)	105	5.71	(4.67, 6.91)
Congenital hip dislocation	19	1.03	(0.62, 1.61)	24	1.30	(0.84, 1.94)
Diaphragmatic hernia	48	2.61	(1.92, 3.46)	49	2.66	(1.97, 3.52)
<i>Total Selected Musculoskeletal Defects</i>	<i>370</i>	<i>20.11</i>	<i>(18.11, 22.26)</i>	<i>383</i>	<i>20.81</i>	<i>(18.78, 23.01)</i>
<b>J. CHROMOSOMAL</b>						
Trisomy 13 (Patau syndrome)	12	0.65	(0.34, 1.14)	13	0.71	(0.38, 1.21)
Down syndrome	194	10.54	(9.11, 12.13)	198	10.76	(9.31, 12.37)
Trisomy 18 (Edward syndrome)	44	2.39	(1.74, 3.21)	47	2.55	(1.88, 3.40)
<i>Total Selected Chromosomal Defects</i>	<i>250</i>	<i>13.59</i>	<i>(11.95, 15.38)</i>	<i>258</i>	<i>14.02</i>	<i>(12.36, 15.84)</i>
<b><i>Total All Selected Defects</i></b>	<b><i>3,781</i></b>	<b><i>205.46</i></b>	<b><i>(198.97, 212.12)</i></b>	<b><i>3,879</i></b>	<b><i>210.79</i></b>	<b><i>(204.21, 217.53)</i></b>

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

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

<sup>3</sup> Subset of previous birth defect group

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2003

**Table 5. Number and Rate of Selected Birth Defects for 2000  
Illinois**

Selected Birth Defects Groups	Newborn Identification			Up to 1 year Identification		
	N	Rate <sup>1</sup>	95% CI <sup>2</sup>	N	Rate <sup>1</sup>	95% CI <sup>2</sup>
<b>A. CENTRAL NERVOUS SYSTEM</b>						
Anencephalus	28	1.51	(1.01, 2.19)	29	1.57	(1.05, 2.25)
Spina bifida without anencephalus	44	2.38	(1.73, 3.19)	52	2.81	(2.10, 3.69)
Hydrocephalus without spina bifida	126	6.81	(5.67, 8.11)	135	7.30	(6.12, 8.64)
Encephalocele	11	0.59	(0.30, 1.06)	14	0.76	(0.41, 1.27)
Microcephalus	48	2.59	(1.91, 3.44)	76	4.11	(3.24, 5.14)
<i>Total Selected CNS Defects</i>	<i>257</i>	<i>13.89</i>	<i>(12.25, 15.70)</i>	<i>306</i>	<i>16.54</i>	<i>(14.74, 18.50)</i>
<b>B. EYE</b>						
Coloboma of the eye	4	0.22	(0.06, 0.55)	4	0.22	(0.06, 0.55)
Anophthalmos/Microphthalmos	21	1.14	(0.70, 1.74)	21	1.14	(0.70, 1.74)
Congenital cataract	9	0.49	(0.22, 0.92)	12	0.65	(0.34, 1.13)
Aniridia	0	0.00	(0.00, 0.20)	0	0.00	(0.00, 0.20)
<i>Total Selected Eye Defects</i>	<i>34</i>	<i>1.84</i>	<i>(1.27, 2.57)</i>	<i>37</i>	<i>2.00</i>	<i>(1.41, 2.76)</i>
<b>C. EAR</b>						
Anotia/Microtia	9	0.49	(0.22, 0.92)	10	0.54	(0.26, 0.99)
<b>D. CARDIOVASCULAR</b>						
Common truncus	5	0.27	(0.09, 0.63)	8	0.43	(0.19, 0.85)
Transposition of great vessels	31	1.68	(1.14, 2.38)	40	2.16	(1.54, 2.94)
Tetralogy of fallot	38	2.05	(1.45, 2.82)	57	3.08	(2.33, 3.99)
Ventricular septal defect	332	17.95	(16.07, 19.98)	469	25.35	(23.11, 27.75)
Atrial septal defect	400	21.62	(19.55, 23.85)	529	28.59	(26.21, 31.14)
Endocardial cushion defect	30	1.62	(1.09, 2.31)	39	2.11	(1.50, 2.88)
Pulmonary valve stenosis and atresia	39	2.11	(1.50, 2.88)	69	3.73	(2.90, 4.72)
Tricuspid valve stenosis and atresia	3	0.16	(0.03, 0.47)	5	0.27	(0.09, 0.63)
Ebstein anomaly	7	0.38	(0.15, 0.78)	7	0.38	(0.15, 0.78)
Aortic valve stenosis	17	0.92	(0.54, 1.47)	23	1.24	(0.79, 1.87)
Hypoplastic left heart syndrome	32	1.73	(1.18, 2.44)	38	2.05	(1.45, 2.82)
Patent ductus arteriosus	801	43.30	(40.35, 46.40)	920	49.73	(46.57, 53.05)
Coarctation of aorta	36	1.95	(1.36, 2.69)	42	2.27	(1.64, 3.07)
Pulmonary artery anomalies	247	13.35	(11.74, 15.12)	262	14.16	(12.50, 15.98)
<i>Total Selected Cardiovascular Defects</i>	<i>2,018</i>	<i>109.08</i>	<i>(104.37, 113.94)</i>	<i>2,508</i>	<i>135.57</i>	<i>(130.31, 140.98)</i>
<b>E. RESPIRATORY</b>						
Lung agenesis/hypoplasia	45	2.43	(1.77, 3.25)	47	2.54	(1.87, 3.38)
<b>F. OROFACIAL</b>						
Cleft palate without cleft lip	73	3.95	(3.09, 4.96)	81	4.38	(3.48, 5.44)
Cleft lip with and without cleft palate	118	6.38	(5.28, 7.64)	129	6.97	(5.82, 8.29)
Choanal atresia	22	1.19	(0.75, 1.80)	24	1.30	(0.83, 1.93)
<i>Total Selected Orofacial Defects</i>	<i>213</i>	<i>11.51</i>	<i>(10.02, 13.17)</i>	<i>234</i>	<i>12.65</i>	<i>(11.08, 14.38)</i>
<b>G. GASTROINTESTINAL</b>						
Esophageal atresia/Tracheoesophageal fistula	39	2.11	(1.50, 2.88)	39	2.11	(1.50, 2.88)
Rectal and large intestinal atresia/stenosis	54	2.92	(2.19, 3.81)	58	3.14	(2.38, 4.05)
Pyloric stenosis	11	0.59	(0.30, 1.06)	63	3.41	(2.62, 4.36)
Hirschsprung disease (congenital megacolon)	30	1.62	(1.09, 2.31)	34	1.84	(1.27, 2.57)
Biliary atresia	2	0.11	(0.01, 0.39)	7	0.38	(0.15, 0.78)
<i>Total Selected Gastrointestinal Defects</i>	<i>136</i>	<i>7.35</i>	<i>(6.17, 8.70)</i>	<i>201</i>	<i>10.86</i>	<i>(9.41, 12.47)</i>

Selected Birth Defects Groups	Newborn Identification			Up to 1 year Identification		
	N	Rate <sup>1</sup>	95% CI <sup>2</sup>	N	Rate <sup>1</sup>	95% CI <sup>2</sup>
<b>H. GENITOURINARY</b>						
Renal agenesis/hypoplasia	35	1.89	(1.32, 2.63)	35	1.89	(1.32, 2.63)
Bladder exstrophy	4	0.22	(0.06, 0.55)	4	0.22	(0.06, 0.55)
Obstructive genitourinary defect	170	9.19	(7.86, 10.68)	233	12.59	(11.03, 14.32)
Penile anomalies	354	19.13	(17.19, 21.24)	354	19.13	(17.19, 21.24)
Hypospadias/Epispadias <sup>3</sup>	184	9.95	(8.56, 11.49)	272	14.70	(13.01, 16.56)
<i>Total Selected Genitourinary Defects</i>	<i>563</i>	<i>30.43</i>	<i>(27.97, 33.05)</i>	<i>626</i>	<i>33.84</i>	<i>(31.24, 36.59)</i>
<b>I. MUSCULOSKELETAL</b>						
Club foot	157	8.49	(7.21, 9.92)	212	11.46	(9.97, 13.11)
Reduction deformity, upper limbs	29	1.57	(1.05, 2.25)	31	1.68	(1.14, 2.38)
Reduction deformity, lower limbs	15	0.81	(0.45, 1.34)	17	0.92	(0.54, 1.47)
Abdominal wall defects	106	5.73	(4.69, 6.93)	106	5.73	(4.69, 6.93)
Gastroschisis/Omphalocele <sup>3</sup>	95	5.14	(4.15, 6.28)	96	5.19	(4.20, 6.34)
Congenital hip dislocation	30	1.62	(1.09, 2.31)	67	3.62	(2.81, 4.60)
Diaphragmatic hernia	49	2.65	(1.96, 3.50)	50	2.70	(2.01, 3.56)
<i>Total Selected Musculoskeletal Defects</i>	<i>386</i>	<i>20.86</i>	<i>(18.83, 23.05)</i>	<i>483</i>	<i>26.11</i>	<i>(23.83, 28.54)</i>
<b>J. CHROMOSOMAL</b>						
Trisomy 13 (Patau syndrome)	15	0.81	(0.45, 1.34)	15	0.81	(0.45, 1.34)
Down syndrome	226	12.22	(10.68, 13.92)	263	14.22	(12.55, 16.04)
Trisomy 18 (Edward syndrome)	29	1.57	(1.05, 2.25)	32	1.73	(1.18, 2.44)
<i>Total Selected Chromosomal Defects</i>	<i>270</i>	<i>14.59</i>	<i>(12.91, 16.44)</i>	<i>310</i>	<i>16.76</i>	<i>(14.94, 18.73)</i>
<b><i>Total All Selected Defects</i></b>	<b><i>3,931</i></b>	<b><i>212.48</i></b>	<b><i>(205.89, 219.23)</i></b>	<b><i>4,762</i></b>	<b><i>257.40</i></b>	<b><i>(250.14, 264.82)</i></b>

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

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

<sup>3</sup> Subset of previous birth defect group

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2003

**Table 6. Number and Rate of Selected Birth Defects for 1999  
Illinois**

Selected Birth Defects Groups	Newborn Identification			Up to 1 year Identification		
	N	Rate <sup>1</sup>	95% CI <sup>2</sup>	N	Rate <sup>1</sup>	95% CI <sup>2</sup>
<b>A. CENTRAL NERVOUS SYSTEM</b>						
Anencephalus	32	1.76	(1.20, 2.48)	32	1.76	(1.20, 2.48)
Spina bifida without anencephalus	58	3.19	(2.42, 4.12)	74	4.07	(3.19, 5.10)
Hydrocephalus without spina bifida	107	5.88	(4.82, 7.10)	131	7.20	(6.02, 8.54)
Encephalocele	11	0.60	(0.30, 1.08)	15	0.82	(0.46, 1.36)
Microcephalus	35	1.92	(1.34, 2.67)	69	3.79	(2.95, 4.80)
<i>Total Selected CNS Defects</i>	243	13.35	(11.72, 15.14)	321	17.63	(15.76, 19.67)
<b>B. EYE</b>						
Coloboma of the eye	4	0.22	(0.06, 0.56)	4	0.22	(0.06, 0.56)
Anophthalmos/Microphthalmos	13	0.71	(0.38, 1.22)	13	0.71	(0.38, 1.22)
Congenital cataract	9	0.49	(0.23, 0.94)	9	0.49	(0.23, 0.94)
Aniridia	1	0.05	(0.00, 0.31)	1	0.05	(0.00, 0.31)
<i>Total Selected Eye Defects</i>	27	1.48	(0.98, 2.16)	27	1.48	(0.98, 2.16)
<b>C. EAR</b>						
Anotia/Microtia	9	0.49	(0.23, 0.94)	9	0.49	(0.23, 0.94)
<b>D. CARDIOVASCULAR</b>						
Common truncus	3	0.16	(0.03, 0.48)	4	0.22	(0.06, 0.56)
Transposition of great vessels	32	1.76	(1.20, 2.48)	43	2.36	(1.71, 3.18)
Tetralogy of fallot	37	2.03	(1.43, 2.80)	49	2.69	(1.99, 3.56)
Ventricular septal defect	298	16.37	(14.57, 18.34)	408	22.41	(20.29, 24.70)
Atrial septal defect	293	16.10	(14.31, 18.05)	374	20.55	(18.52, 22.74)
Endocardial cushion defect	34	1.87	(1.29, 2.61)	48	2.64	(1.94, 3.50)
Pulmonary valve stenosis and atresia	34	1.87	(1.29, 2.61)	71	3.90	(3.05, 4.92)
Tricuspid valve stenosis and atresia	2	0.11	(0.01, 0.40)	3	0.16	(0.03, 0.48)
Ebstein anomaly	7	0.38	(0.15, 0.79)	7	0.38	(0.15, 0.79)
Aortic valve stenosis	13	0.71	(0.38, 1.22)	15	0.82	(0.46, 1.36)
Hypoplastic left heart syndrome	22	1.21	(0.76, 1.83)	25	1.37	(0.89, 2.03)
Patent ductus arteriosus	675	37.08	(34.34, 39.99)	693	38.07	(35.29, 41.01)
Coarctation of aorta	29	1.59	(1.07, 2.29)	34	1.87	(1.29, 2.61)
Pulmonary artery anomalies	150	8.24	(6.97, 9.67)	158	8.68	(7.38, 10.14)
<i>Total Selected Cardiovascular Defects</i>	1,629	89.49	(85.20, 93.95)	1,932	106.14	(101.46, 110.98)
<b>E. RESPIRATORY</b>						
Lung agenesis/hypoplasia	65	3.57	(2.76, 4.55)	65	3.57	(2.76, 4.55)
<b>F. OROFACIAL</b>						
Cleft palate without cleft lip	51	2.80	(2.09, 3.68)	51	2.80	(2.09, 3.68)
Cleft lip with and without cleft palate	114	6.26	(5.17, 7.52)	114	6.26	(5.17, 7.52)
Choanal atresia	16	0.88	(0.50, 1.43)	16	0.88	(0.50, 1.43)
<i>Total Selected Orofacial Defects</i>	181	9.94	(8.55, 11.50)	181	9.94	(8.55, 11.50)
<b>G. GASTROINTESTINAL</b>						
Esophageal atresia/Tracheoesophageal fistula	36	1.98	(1.39, 2.74)	40	2.20	(1.57, 2.99)
Rectal and large intestinal atresia/stenosis	45	2.47	(1.80, 3.31)	58	3.19	(2.42, 4.12)
Pyloric stenosis	13	0.71	(0.38, 1.22)	85	4.67	(3.73, 5.77)
Hirschsprung disease (congenital megacolon)	32	1.76	(1.20, 2.48)	45	2.47	(1.80, 3.31)
Biliary atresia	2	0.11	(0.01, 0.40)	12	0.66	(0.34, 1.15)
<i>Total Selected Gastrointestinal Defects</i>	128	7.03	(5.87, 8.36)	240	13.18	(11.57, 14.96)

Selected Birth Defects Groups	Newborn Identification			Up to 1 year Identification		
	N	Rate <sup>1</sup>	95% CI <sup>2</sup>	N	Rate <sup>1</sup>	95% CI <sup>2</sup>
<b>H. GENITOURINARY</b>						
Renal agenesis/hypoplasia	26	1.43	(0.93, 2.09)	26	1.43	(0.93, 2.09)
Bladder exstrophy	8	0.44	(0.19, 0.87)	8	0.44	(0.19, 0.87)
Obstructive genitourinary defect	154	8.46	(7.18, 9.91)	154	8.46	(7.18, 9.91)
Penile anomalies	246	13.51	(11.88, 15.31)	246	13.51	(11.88, 15.31)
Hypospadias/Epispadias <sup>3</sup>	193	10.60	(9.16, 12.21)	193	10.60	(9.16, 12.21)
<i>Total Selected Genitourinary Defects</i>	<i>434</i>	<i>23.84</i>	<i>(21.65, 26.19)</i>	<i>434</i>	<i>23.84</i>	<i>(21.65, 26.19)</i>
<b>I. MUSCULOSKELETAL</b>						
Club foot	153	8.41	(7.13, 9.85)	153	8.41	(7.13, 9.85)
Reduction deformity, upper limbs	25	1.37	(0.89, 2.03)	25	1.37	(0.89, 2.03)
Reduction deformity, lower limbs	20	1.10	(0.67, 1.70)	20	1.10	(0.67, 1.70)
Abdominal wall defects	78	4.29	(3.39, 5.35)	78	4.29	(3.39, 5.35)
Gastroschisis/Omphalocele <sup>3</sup>	42	2.31	(1.66, 3.12)	42	2.31	(1.66, 3.12)
Congenital hip dislocation	26	1.43	(0.93, 2.09)	26	1.43	(0.93, 2.09)
Diaphragmatic hernia	55	3.02	(2.28, 3.93)	55	3.02	(2.28, 3.93)
<i>Total Selected Musculoskeletal Defects</i>	<i>357</i>	<i>19.61</i>	<i>(17.63, 21.76)</i>	<i>357</i>	<i>19.61</i>	<i>(17.63, 21.76)</i>
<b>J. CHROMOSOMAL</b>						
Trisomy 13 (Patau syndrome)	19	1.04	(0.63, 1.63)	19	1.04	(0.63, 1.63)
Down syndrome	164	9.01	(7.68, 10.50)	209	11.48	(9.98, 13.15)
Trisomy 18 (Edward syndrome)	47	2.58	(1.90, 3.43)	50	2.75	(2.04, 3.62)
<i>Total Selected Chromosomal Defects</i>	<i>230</i>	<i>12.64</i>	<i>(11.06, 14.38)</i>	<i>278</i>	<i>15.27</i>	<i>(13.53, 17.18)</i>
<b><i>Total All Selected Defects</i></b>	<b><i>3,303</i></b>	<b><i>181.46</i></b>	<b><i>(175.32, 187.75)</i></b>	<b><i>3,844</i></b>	<b><i>211.18</i></b>	<b><i>(204.55, 217.96)</i></b>

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

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

<sup>3</sup> Subset of previous birth defect group

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2003

**Table 7. Number and Rate of Selected Birth Defects for 2001  
Chicago**

Selected Birth Defects Groups	Newborn Identification			Up to 1 year Identification		
	N	Rate <sup>1</sup>	95% CI <sup>2</sup>	N	Rate <sup>1</sup>	95% CI <sup>2</sup>
<b>A. CENTRAL NERVOUS SYSTEM</b>						
Anencephalus	9	1.81	(0.83, 3.44)	10	2.02	(0.97, 3.71)
Spina bifida without anencephalus	13	2.62	(1.40, 4.48)	13	2.62	(1.40, 4.48)
Hydrocephalus without spina bifida	21	4.23	(2.62, 6.47)	22	4.44	(2.78, 6.72)
Encephalocele	3	0.60	(0.12, 1.77)	3	0.60	(0.12, 1.77)
Microcephalus	35	7.06	(4.92, 9.81)	37	7.46	(5.25, 10.28)
<i>Total Selected CNS Defects</i>	81	16.33	(12.97, 20.30)	85	17.14	(13.69, 21.19)
<b>B. EYE</b>						
Coloboma of the eye	1	0.20	(0.01, 1.12)	1	0.20	(0.01, 1.12)
Anophthalmos/Microphthalmos	2	0.40	(0.05, 1.46)	2	0.40	(0.05, 1.46)
Congenital cataract	0	0.00	(0.00, 0.74)	1	0.20	(0.01, 1.12)
Aniridia	0	0.00	(0.00, 0.74)	0	0.00	(0.00, 0.74)
<i>Total Selected Eye Defects</i>	3	0.60	(0.12, 1.77)	4	0.81	(0.22, 2.07)
<b>C. EAR</b>						
Anotia/Microtia	3	0.60	(0.12, 1.77)	3	0.60	(0.12, 1.77)
<b>D. CARDIOVASCULAR</b>						
Common truncus	1	0.20	(0.01, 1.12)	1	0.20	(0.01, 1.12)
Transposition of great vessels	6	1.21	(0.44, 2.63)	6	1.21	(0.44, 2.63)
Tetralogy of fallot	9	1.81	(0.83, 3.44)	9	1.81	(0.83, 3.44)
Ventricular septal defect	54	10.89	(8.18, 14.21)	55	11.09	(8.35, 14.43)
Atrial septal defect	58	11.69	(8.88, 15.12)	60	12.10	(9.23, 15.57)
Endocardial cushion defect	5	1.01	(0.33, 2.35)	5	1.01	(0.33, 2.35)
Pulmonary valve stenosis and atresia	7	1.41	(0.57, 2.91)	8	1.61	(0.70, 3.18)
Tricuspid valve stenosis and atresia	1	0.20	(0.01, 1.12)	1	0.20	(0.01, 1.12)
Ebstein anomaly	3	0.60	(0.12, 1.77)	3	0.60	(0.12, 1.77)
Aortic valve stenosis	2	0.40	(0.05, 1.46)	2	0.40	(0.05, 1.46)
Hypoplastic left heart syndrome	7	1.41	(0.57, 2.91)	8	1.61	(0.70, 3.18)
Patent ductus arteriosus	111	22.38	(18.41, 26.95)	116	23.39	(19.33, 28.05)
Coarctation of aorta	5	1.01	(0.33, 2.35)	5	1.01	(0.33, 2.35)
Pulmonary artery anomalies	27	5.44	(3.59, 7.92)	28	5.65	(3.75, 8.16)
<i>Total Selected Cardiovascular Defects</i>	296	59.68	(53.08, 66.88)	307	61.90	(55.17, 69.23)
<b>E. RESPIRATORY</b>						
Lung agenesis/hypoplasia	8	1.61	(0.70, 3.18)	8	1.61	(0.70, 3.18)
<b>F. OROFACIAL</b>						
Cleft palate without cleft lip	15	3.02	(1.69, 4.99)	15	3.02	(1.69, 4.99)
Cleft lip with and without cleft palate	15	3.02	(1.69, 4.99)	15	3.02	(1.69, 4.99)
Choanal atresia	4	0.81	(0.22, 2.07)	4	0.81	(0.22, 2.07)
<i>Total Selected Orofacial Defects</i>	34	6.86	(4.75, 9.58)	34	6.86	(4.75, 9.58)
<b>G. GASTROINTESTINAL</b>						
Esophageal atresia/Tracheoesophageal fistula	7	1.41	(0.57, 2.91)	7	1.41	(0.57, 2.91)
Rectal and large intestinal atresia/stenosis	9	1.81	(0.83, 3.44)	9	1.81	(0.83, 3.44)
Pyloric stenosis	2	0.40	(0.05, 1.46)	3	0.60	(0.12, 1.77)
Hirschsprung disease (congenital megacolon)	9	1.81	(0.83, 3.44)	10	2.02	(0.97, 3.71)
Biliary atresia	0	0.00	(0.00, 0.74)	0	0.00	(0.00, 0.74)
<i>Total Selected Gastrointestinal Defects</i>	27	5.44	(3.59, 7.92)	29	5.85	(3.92, 8.40)

Selected Birth Defects Groups	Newborn Identification			Up to 1 year Identification		
	N	Rate <sup>1</sup>	95% CI <sup>2</sup>	N	Rate <sup>1</sup>	95% CI <sup>2</sup>
<b>H. GENITOURINARY</b>						
Renal agenesis/hypoplasia	7	1.41	(0.57, 2.91)	8	1.61	(0.70, 3.18)
Bladder exstrophy	1	0.20	(0.00, 1.12)	1	0.20	(0.01, 1.12)
Obstructive genitourinary defect	23	4.64	(2.94, 6.96)	26	5.24	(3.42, 7.68)
Penile anomalies	28	5.65	(3.75, 8.16)	31	6.25	(4.25, 8.87)
Hypospadias/Epispadias <sup>3</sup>	25	5.04	(3.26, 7.44)	27	5.44	(3.59, 7.92)
<i>Total Selected Genitourinary Defects</i>	<i>59</i>	<i>11.90</i>	<i>(9.06, 15.35)</i>	<i>66</i>	<i>13.31</i>	<i>(10.29, 16.93)</i>
<b>I. MUSCULOSKELETAL</b>						
Club foot	24	4.84	(3.10, 7.20)	24	4.84	(3.10, 7.20)
Reduction deformity, upper limbs	6	1.21	(0.44, 2.63)	6	1.21	(0.44, 2.63)
Reduction deformity, lower limbs	1	0.20	(0.01, 1.12)	1	0.20	(0.01, 1.12)
Abdominal wall defects	29	5.85	(3.92, 8.40)	29	5.85	(3.92, 8.40)
Gastroschisis/Omphalocele <sup>3</sup>	29	5.85	(3.92, 8.40)	29	5.85	(3.92, 8.40)
Congenital hip dislocation	1	0.20	(0.01, 1.12)	1	0.20	(0.01, 1.12)
Diaphragmatic hernia	7	1.41	(0.57, 2.91)	7	1.41	(0.57, 2.91)
<i>Total Selected Musculoskeletal Defects</i>	<i>68</i>	<i>13.71</i>	<i>(10.65, 17.38)</i>	<i>68</i>	<i>13.71</i>	<i>(10.65, 17.38)</i>
<b>J. CHROMOSOMAL</b>						
Trisomy 13 (Patau syndrome)	3	0.60	(0.12, 1.77)	3	0.60	(0.12, 1.77)
Down syndrome	39	7.86	(5.59, 10.75)	41	8.27	(5.93, 11.21)
Trisomy 18 (Edward syndrome)	6	1.21	(0.44, 2.63)	7	1.41	(0.57, 2.91)
<i>Total Selected Chromosomal Defects</i>	<i>48</i>	<i>9.68</i>	<i>(7.14, 12.83)</i>	<i>51</i>	<i>10.28</i>	<i>(7.66, 13.52)</i>
<b><i>Total All Selected Defects</i></b>	<b><i>627</i></b>	<b><i>126.42</i></b>	<b><i>(116.72, 136.72)</i></b>	<b><i>655</i></b>	<b><i>132.07</i></b>	<b><i>(122.15, 142.58)</i></b>

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

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

<sup>3</sup> Subset of previous birth defect group

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2003

**Table 8. Number and Rate of Selected Birth Defects for 2000  
Chicago**

Selected Birth Defects Groups	Newborn Identification			Up to 1 year Identification		
	N	Rate <sup>1</sup>	95% CI <sup>2</sup>	N	Rate <sup>1</sup>	95% CI <sup>2</sup>
<b>A. CENTRAL NERVOUS SYSTEM</b>						
Anencephalus	10	1.97	(0.94, 3.61)	10	1.97	(0.94, 3.61)
Spina bifida without anencephalus	11	2.16	(1.08, 3.87)	14	2.75	(1.50, 4.62)
Hydrocephalus without spina bifida	42	8.25	(5.95, 11.16)	47	9.24	(6.79, 12.28)
Encephalocele	2	0.39	(0.05, 1.42)	2	0.39	(0.05, 1.42)
Microcephalus	19	3.73	(2.25, 5.83)	38	7.47	(5.28, 10.25)
<i>Total Selected CNS Defects</i>	84	16.51	(13.17, 20.44)	111	21.81	(17.95, 26.27)
<b>B. EYE</b>						
Coloboma of the eye	1	0.20	(0.00, 1.09)	1	0.20	(0.00, 1.09)
Anophthalmos/Microphthalmos	1	0.20	(0.00, 1.09)	1	0.20	(0.00, 1.09)
Congenital cataract	0	0.00	(0.00, 0.72)	2	0.39	(0.05, 1.42)
Aniridia	0	0.00	(0.00, 0.72)	0	0.00	(0.00, 0.72)
<i>Total Selected Eye Defects</i>	2	0.39	(0.05, 1.42)	4	0.79	(0.21, 2.01)
<b>C. EAR</b>						
Anotia/Microtia	2	0.39	(0.05, 1.42)	2	0.39	(0.05, 1.42)
<b>D. CARDIOVASCULAR</b>						
Common truncus	2	0.39	(0.05, 1.42)	3	0.59	(0.12, 1.72)
Transposition of great vessels	6	1.18	(0.43, 2.57)	12	2.36	(1.22, 4.12)
Tetralogy of fallot	10	1.97	(0.94, 3.61)	15	2.95	(1.65, 4.86)
Ventricular septal defect	48	9.43	(6.96, 12.51)	99	19.46	(15.81, 23.69)
Atrial septal defect	47	9.24	(6.79, 12.28)	84	16.51	(13.17, 20.44)
Endocardial cushion defect	3	0.59	(0.12, 1.72)	8	1.57	(0.68, 3.10)
Pulmonary valve stenosis and atresia	2	0.39	(0.05, 1.42)	13	2.55	(1.36, 4.37)
Tricuspid valve stenosis and atresia	1	0.20	(0.00, 1.09)	2	0.39	(0.05, 1.42)
Ebstein anomaly	1	0.20	(0.00, 1.09)	1	0.20	(0.00, 1.09)
Aortic valve stenosis	2	0.39	(0.05, 1.42)	3	0.59	(0.12, 1.72)
Hypoplastic left heart syndrome	5	0.98	(0.32, 2.29)	6	1.18	(0.43, 2.57)
Patent ductus arteriosus	134	26.33	(22.06, 31.19)	167	32.82	(28.03, 38.19)
Coarctation of aorta	5	0.98	(0.32, 2.29)	6	1.18	(0.43, 2.57)
Pulmonary artery anomalies	26	5.11	(3.34, 7.49)	29	5.70	(3.82, 8.18)
<i>Total Selected Cardiovascular Defects</i>	292	57.38	(50.99, 96.58)	448	88.04	(80.08, 96.58)
<b>E. RESPIRATORY</b>						
Lung agenesis/hypoplasia	12	2.36	(1.22, 4.12)	14	2.75	(1.50, 4.62)
<b>F. OROFACIAL</b>						
Cleft palate without cleft lip	11	2.16	(1.08, 3.87)	16	3.14	(1.80, 5.11)
Cleft lip with and without cleft palate	18	3.54	(2.10, 5.59)	20	3.93	(2.40, 6.07)
Choanal atresia	3	0.59	(0.12, 1.72)	3	0.59	(0.12, 1.72)
<i>Total Selected Orofacial Defects</i>	32	6.29	(4.30, 8.88)	39	7.66	(5.45, 10.48)
<b>G. GASTROINTESTINAL</b>						
Esophageal atresia/Tracheoesophageal fistula	7	1.38	(0.55, 2.83)	7	1.38	(0.55, 2.83)
Rectal and large intestinal atresia/stenosis	10	1.97	(0.94, 3.61)	12	2.36	(1.22, 4.12)
Pyloric stenosis	2	0.39	(0.05, 1.42)	41	8.06	(5.78, 10.93)
Hirschsprung disease (congenital megacolon)	8	1.57	(0.68, 3.10)	10	1.97	(0.94, 3.61)
Biliary atresia	1	0.20	(0.00, 1.09)	1	0.20	(0.00, 1.09)
<i>Total Selected Gastrointestinal Defects</i>	28	5.50	(3.66, 7.95)	71	13.95	(10.90, 17.60)

Selected Birth Defects Groups	Newborn Identification			Up to 1 year Identification		
	N	Rate <sup>1</sup>	95% CI <sup>2</sup>	N	Rate <sup>1</sup>	95% CI <sup>2</sup>
<b>H. GENITOURINARY</b>						
Renal agenesis/hypoplasia	4	0.79	(0.21, 2.01)	4	0.79	(0.21, 2.01)
Bladder exstrophy	0	0.00	(0.00, 0.72)	0	0.00	(0.00, 0.72)
Obstructive genitourinary defect	25	4.91	(3.18, 7.25)	55	10.81	(8.14, 14.07)
Penile anomalies	31	6.09	(4.14, 8.65)	66	12.97	(10.03, 16.50)
Hypospadias/Epispadias <sup>3</sup>	27	5.31	(3.50, 7.72)	51	10.02	(7.46, 13.18)
<i>Total Selected Genitourinary Defects</i>	<i>60</i>	<i>11.79</i>	<i>(9.00, 15.18)</i>	<i>125</i>	<i>24.57</i>	<i>(20.45, 29.27)</i>
<b>I. MUSCULOSKELETAL</b>						
Club foot	19	3.73	(2.25, 5.83)	29	5.70	(3.82, 8.18)
Reduction deformity, upper limbs	1	0.20	(0.00, 1.09)	1	0.20	(0.00, 1.09)
Reduction deformity, lower limbs	3	0.59	(0.12, 1.72)	4	0.79	(0.21, 2.01)
Abdominal wall defects	39	7.66	(5.45, 10.48)	40	7.86	(5.62, 10.70)
Gastroschisis/Omphalocele <sup>3</sup>	35	6.88	(4.79, 9.57)	36	7.07	(4.96, 9.79)
Congenital hip dislocation	5	0.98	(0.32, 2.29)	17	3.34	(1.95, 5.35)
Diaphragmatic hernia	12	2.36	(1.22, 4.12)	12	2.36	(1.22, 4.12)
<i>Total Selected Musculoskeletal Defects</i>	<i>79</i>	<i>15.53</i>	<i>(12.29, 19.35)</i>	<i>103</i>	<i>20.24</i>	<i>(16.52, 24.55)</i>
<b>J. CHROMOSOMAL</b>						
Trisomy 13 (Patau syndrome)	5	0.98	(0.32, 2.29)	5	0.98	(0.32, 2.29)
Down syndrome	44	8.65	(6.28, 11.61)	57	11.20	(8.48, 14.51)
Trisomy 18 (Edward syndrome)	9	1.77	(0.81, 3.36)	10	1.97	(0.94, 3.61)
<i>Total Selected Chromosomal Defects</i>	<i>58</i>	<i>11.40</i>	<i>(8.66, 14.73)</i>	<i>72</i>	<i>14.15</i>	<i>(11.07, 17.82)</i>
<b><i>Total All Selected Defects</i></b>	<b><i>649</i></b>	<b><i>127.54</i></b>	<b><i>(117.92, 137.74)</i></b>	<b><i>989</i></b>	<b><i>194.36</i></b>	<b><i>(182.43, 206.86)</i></b>

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

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

<sup>3</sup> Subset of previous birth defect group

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2003

**Table 9. Number and Rate of Selected Birth Defects for 1999  
Chicago**

Selected Birth Defects Groups	Newborn Identification				Up to 1 year Identification			
	N	Rate <sup>1</sup>	95% CI <sup>2</sup>		N	Rate <sup>1</sup>	95% CI <sup>2</sup>	
<b>A. CENTRAL NERVOUS SYSTEM</b>								
Anencephalus	6	1.19	(0.44,	2.58)	6	1.19	(0.44,	2.58)
Spina bifida without anencephalus	15	2.97	(1.66,	4.89)	23	4.55	(2.88,	6.83)
Hydrocephalus without spina bifida	31	6.13	(4.17,	8.71)	38	7.52	(5.32,	10.32)
Encephalocele	4	0.79	(0.22,	2.03)	6	1.19	(0.44,	2.58)
Microcephalus	14	2.77	(1.51,	4.65)	38	7.52	(5.32,	10.32)
<i>Total Selected CNS Defects</i>	70	13.85	(10.80,	17.50)	111	21.96	(18.07,	26.45)
<b>B. EYE</b>								
Coloboma of the eye	1	0.20	(0.01,	1.10)	1	0.20	(0.01,	1.10)
Anophthalmos/Microphthalmos	1	0.20	(0.01,	1.10)	1	0.20	(0.01,	1.10)
Congenital cataract	0	0.00	(0.00,	0.73)	1	0.20	(0.01,	1.10)
Aniridia	0	0.00	(0.00,	0.73)	0	0.00	(0.00,	0.73)
<i>Total Selected Eye Defects</i>	2	0.40	(0.05,	1.43)	3	0.59	(0.12,	1.73)
<b>C. EAR</b>								
Anotia/Microtia	1	0.20	(0.01,	1.10)	1	0.20	(0.01,	1.10)
<b>D. CARDIOVASCULAR</b>								
Common truncus	1	0.20	(0.01,	1.10)	1	0.20	(0.01,	1.10)
Transposition of great vessels	4	0.79	(0.22,	2.03)	12	2.37	(1.23,	4.15)
Tetralogy of fallot	7	1.38	(0.56,	2.85)	14	2.77	(1.51,	4.65)
Ventricular septal defect	48	9.50	(7.00,	12.59)	111	21.96	(18.07,	26.45)
Atrial septal defect	44	8.71	(6.33,	11.69)	90	17.81	(14.32,	21.89)
Endocardial cushion defect	5	0.99	(0.32,	2.31)	13	2.57	(1.37,	4.40)
Pulmonary valve stenosis and atresia	1	0.20	(0.01,	1.10)	20	3.96	(2.42,	6.11)
Tricuspid valve stenosis and atresia	1	0.20	(0.01,	1.10)	2	0.40	(0.05,	1.43)
Ebstein anomaly	1	0.20	(0.01,	1.10)	1	0.20	(0.01,	1.10)
Aortic valve stenosis	1	0.20	(0.01,	1.10)	3	0.59	(0.12,	1.73)
Hypoplastic left heart syndrome	2	0.40	(0.05,	1.43)	3	0.59	(0.12,	1.73)
Patent ductus arteriosus	98	19.39	(15.74,	23.63)	109	21.57	(17.71,	26.02)
Coarctation of aorta	7	1.38	(0.56,	2.85)	11	2.18	(1.09,	3.89)
Pulmonary artery anomalies	12	2.37	(1.23,	4.15)	19	3.76	(2.26,	5.87)
<i>Total Selected Cardiovascular Defects</i>	232	45.90	(40.19,	89.16)	409	80.92	(73.27,	89.16)
<b>E. RESPIRATORY</b>								
Lung agenesis/hypoplasia	16	3.17	(1.81,	5.14)	16	3.17	(1.81,	5.14)
<b>F. OROFACIAL</b>								
Cleft palate without cleft lip	12	2.37	(1.23,	4.15)	12	2.37	(1.23,	4.15)
Cleft lip with and without cleft palate	18	3.56	(2.11,	5.63)	18	3.56	(2.11,	5.63)
Choanal atresia	3	0.59	(0.12,	1.73)	3	0.59	(0.12,	1.73)
<i>Total Selected Orofacial Defects</i>	33	6.53	(4.49,	9.17)	33	6.53	(4.49,	9.17)
<b>G. GASTROINTESTINAL</b>								
Esophageal atresia/Tracheoesophageal fistula	8	1.58	(0.68,	3.12)	10	1.98	(0.95,	3.64)
Rectal and large intestinal atresia/stenosis	11	2.18	(1.09,	3.89)	16	3.17	(1.81,	5.14)
Pyloric stenosis	1	0.20	(0.01,	1.10)	43	8.51	(6.16,	11.46)
Hirschsprung disease (congenital megacolon)	18	3.56	(2.11,	5.63)	24	4.75	(3.04,	7.07)
Biliary atresia	1	0.20	(0.01,	1.10)	5	0.99	(0.32,	2.31)
<i>Total Selected Gastrointestinal Defects</i>	39	7.72	(5.49,	10.55)	98	19.39	(15.74,	23.63)

Selected Birth Defects Groups	Newborn Identification			Up to 1 year Identification		
	N	Rate <sup>1</sup>	95% CI <sup>2</sup>	N	Rate <sup>1</sup>	95% CI <sup>2</sup>
<b>H. GENITOURINARY</b>						
Renal agenesis/hypoplasia	6	1.19	(0.44, 2.58)	6	1.19	(0.44, 2.58)
Bladder exstrophy	0	0.00	(0.00, 0.73)	0	0.00	(0.00, 0.73)
Obstructive genitourinary defect	34	6.73	(4.66, 9.40)	34	6.73	(4.66, 9.40)
Penile anomalies	36	7.12	(4.99, 9.86)	36	7.12	(4.99, 9.86)
Hypospadias/Epispadias <sup>3</sup>	32	6.33	(4.33, 8.94)	32	6.33	(4.33, 8.94)
<i>Total Selected Genitourinary Defects</i>	<i>76</i>	<i>15.04</i>	<i>(11.85, 18.82)</i>	<i>76</i>	<i>15.04</i>	<i>(11.85, 18.82)</i>
<b>I. MUSCULOSKELETAL</b>						
Club foot	13	2.57	(1.37, 4.40)	13	2.57	(1.37, 4.40)
Reduction deformity, upper limbs	2	0.40	(0.05, 1.43)	2	0.40	(0.05, 1.43)
Reduction deformity, lower limbs	3	0.59	(0.12, 1.73)	3	0.59	(0.12, 1.73)
Abdominal wall defects	26	5.14	(3.36, 7.54)	26	5.14	(3.36, 7.54)
Gastroschisis/Omphalocele <sup>3</sup>	10	1.98	(0.95, 3.64)	10	1.98	(0.95, 3.64)
Congenital hip dislocation	3	0.59	(0.12, 1.73)	3	0.59	(0.12, 1.73)
Diaphragmatic hernia	13	2.57	(1.37, 4.40)	13	2.57	(1.37, 4.40)
<i>Total Selected Musculoskeletal Defects</i>	<i>60</i>	<i>11.87</i>	<i>(9.06, 15.28)</i>	<i>60</i>	<i>11.87</i>	<i>(9.06, 15.28)</i>
<b>J. CHROMOSOMAL</b>						
Trisomy 13 (Patau syndrome)	3	0.59	(0.12, 1.73)	3	0.59	(0.12, 1.73)
Down syndrome	32	6.33	(4.33, 8.94)	57	11.28	(8.54, 14.61)
Trisomy 18 (Edward syndrome)	7	1.38	(0.56, 2.85)	7	1.38	(0.56, 2.85)
<i>Total Selected Chromosomal Defects</i>	<i>42</i>	<i>8.31</i>	<i>(5.99, 11.23)</i>	<i>67</i>	<i>13.26</i>	<i>(10.27, 16.84)</i>
<b><i>Total All Selected Defects</i></b>	<b><i>571</i></b>	<b><i>112.98</i></b>	<b><i>(103.90 122.60)</i></b>	<b><i>874</i></b>	<b><i>172.93</i></b>	<b><i>(161.65, 184.78)</i></b>

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

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

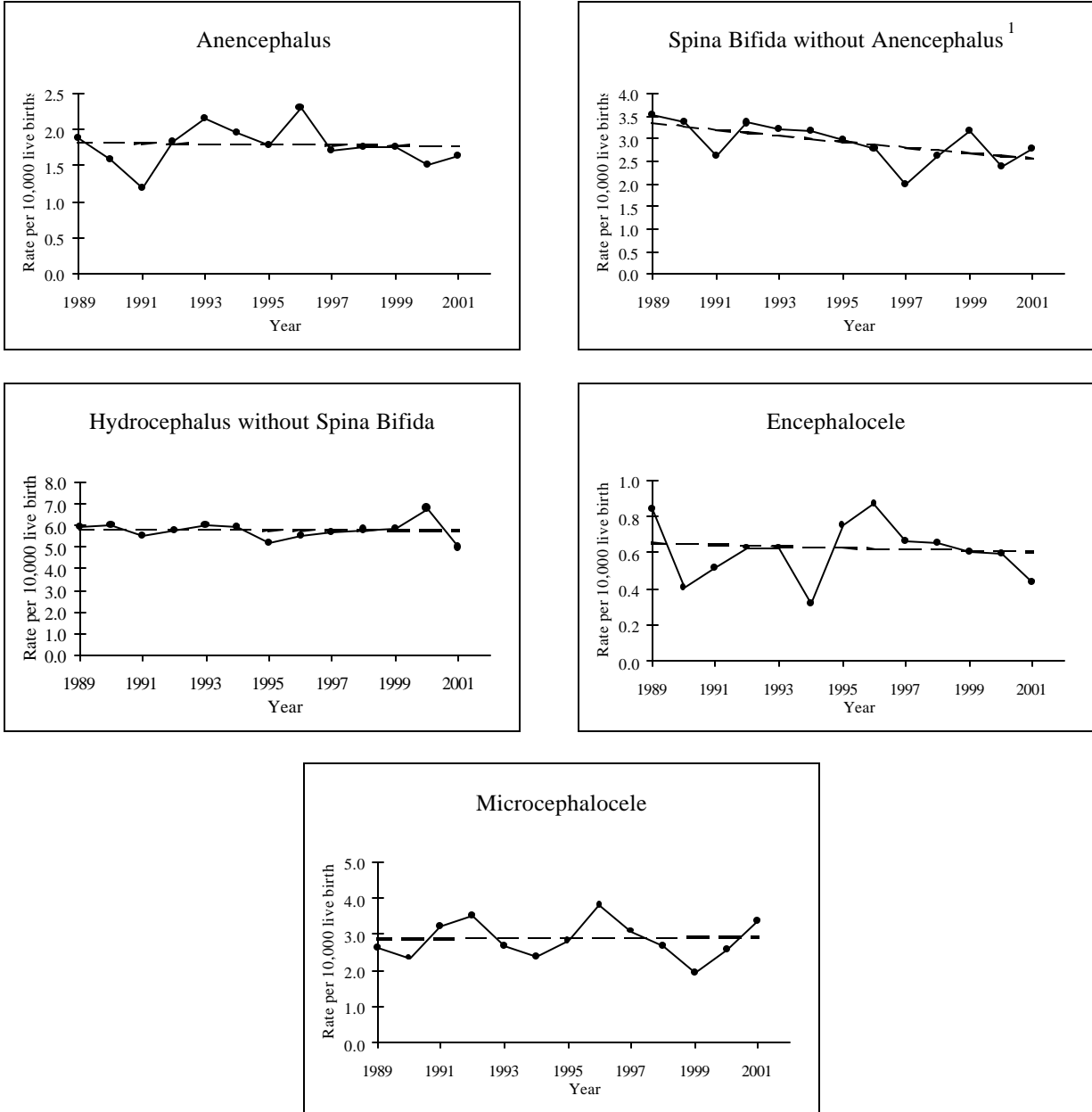
<sup>3</sup> Subset of previous birth defect group

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2003

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

**1989-2001**

**FIGURE 1. TRENDS IN THE REPORTED PREVALENCE RATES OF NEURAL TUBE DEFECTS IDENTIFIED DURING THE NEWBORN STAY, PER 10,000 LIVE BIRTHS 1989-2001**

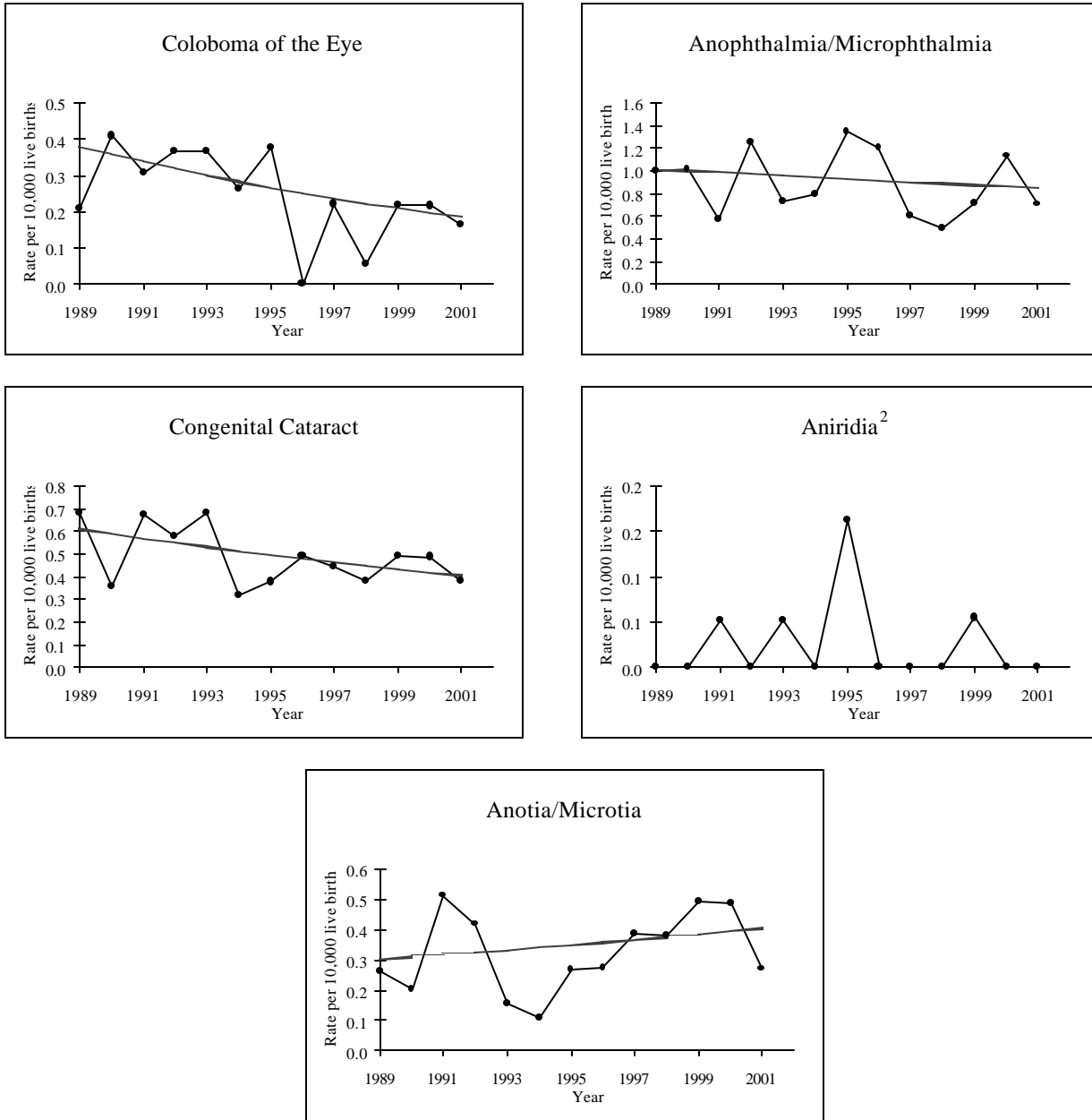


●—● Observed Rates      — Regression Line

<sup>1</sup>Trend is significant; details are given in Table 2.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2003

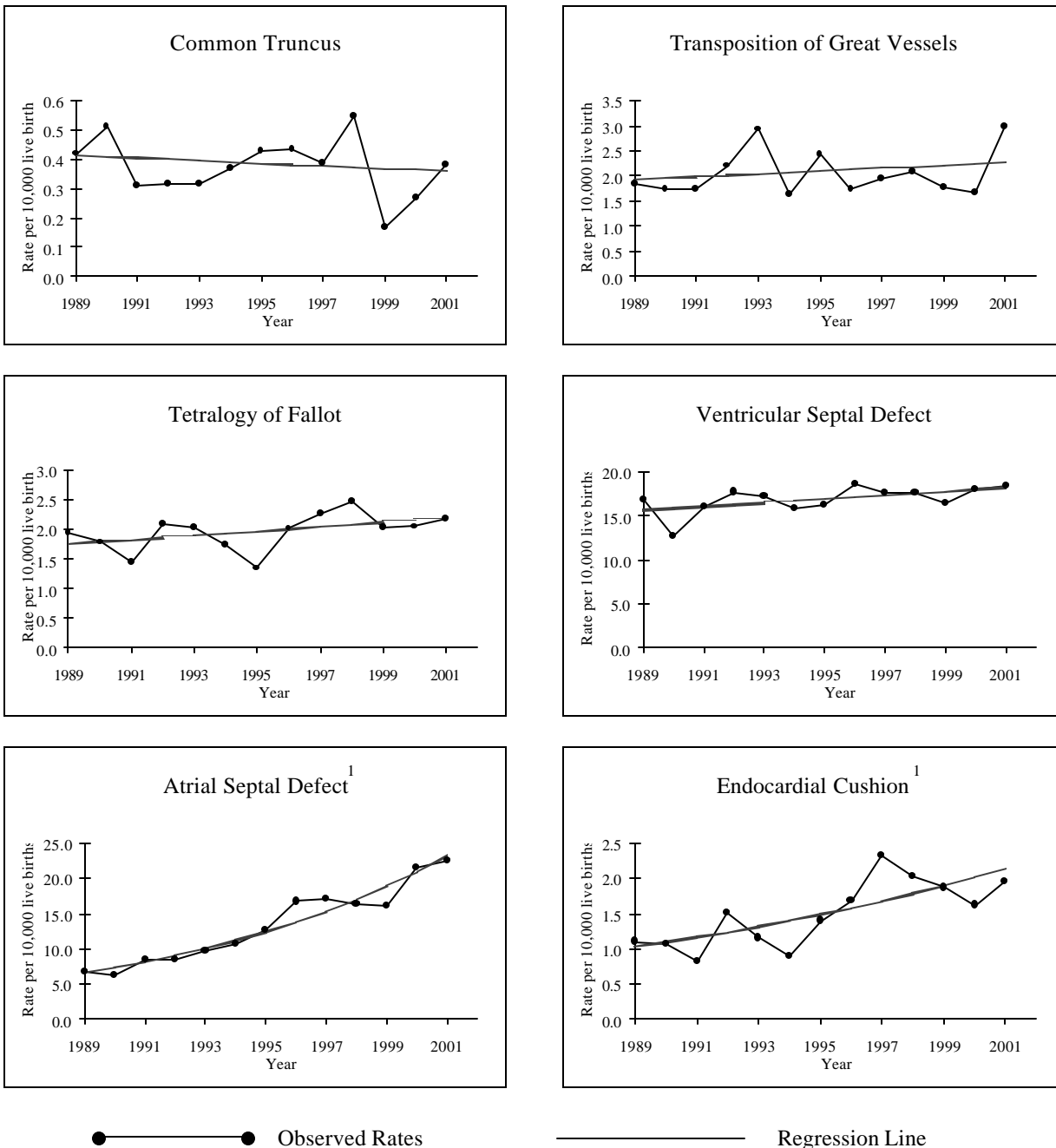
**FIGURE 2. TRENDS IN THE REPORTED PREVALENCE RATES OF EYE AND EAR DEFECTS IDENTIFIED DURING THE NEWBORN STAY, PER 10,000 LIVE BIRTHS 1989-2001**



●—● Observed Rates      — Regression Line

<sup>2</sup>There is no regression line for aniridia because there are too few cases to perform such an analysis.  
 Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2003

**FIGURE 3a. TRENDS IN THE REPORTED PREVALENCE RATES OF CARDIAC DEFECTS IDENTIFIED DURING THE NEWBORN STAY, PER 10,000 LIVE BIRTHS 1989-2001**



<sup>1</sup>Trend is significant; details are given in Table 3.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2003

**FIGURE 3b. TRENDS IN THE REPORTED PREVALENCE RATES OF CARDIAC DEFECTS IDENTIFIED DURING THE NEWBORN STAY, PER 10,000 LIVE BIRTHS 1989-2001**

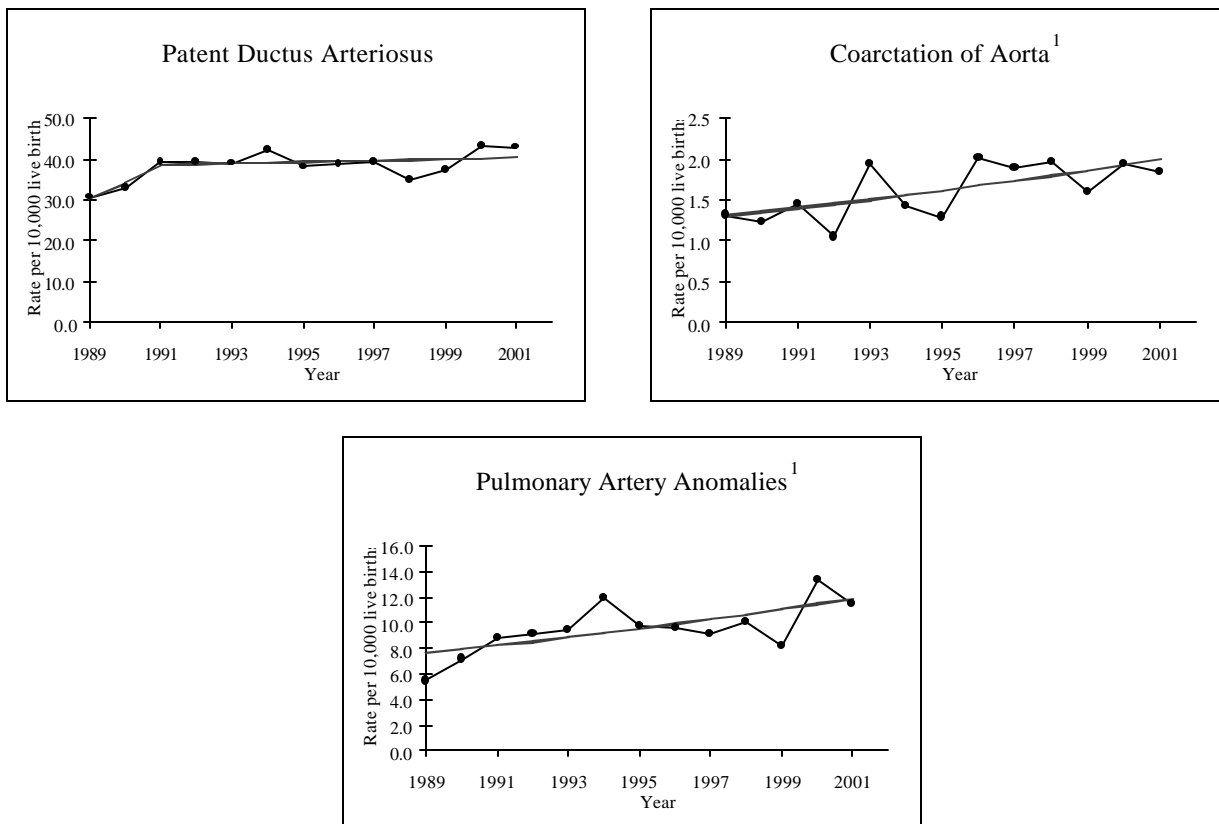


●—● Observed Rates      — Regression Line

<sup>1</sup>Trend is significant; details are given in Table 3.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2003

**FIGURE 4. TRENDS IN THE REPORTED PREVALENCE RATES OF CIRCULATORY DEFECTS IDENTIFIED DURING THE NEWBORN STAY, PER 10,000 LIVE BIRTHS 1989-2001**

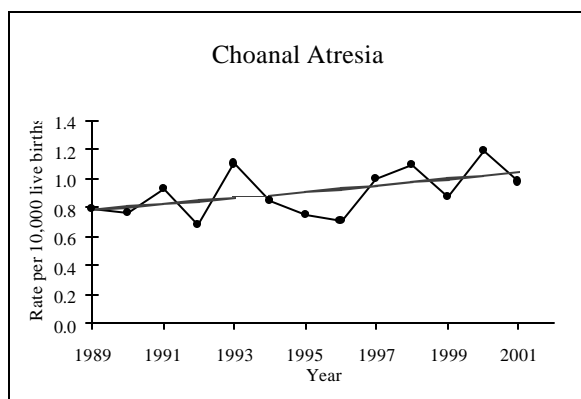
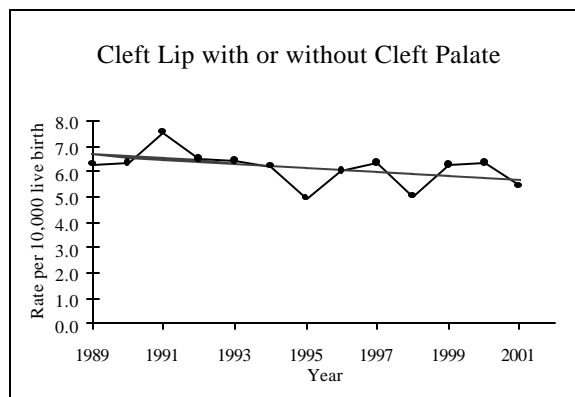
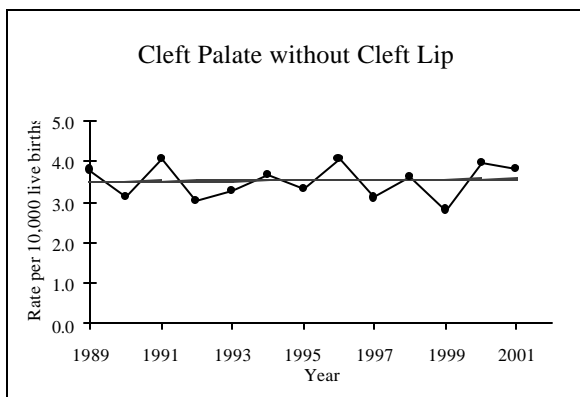
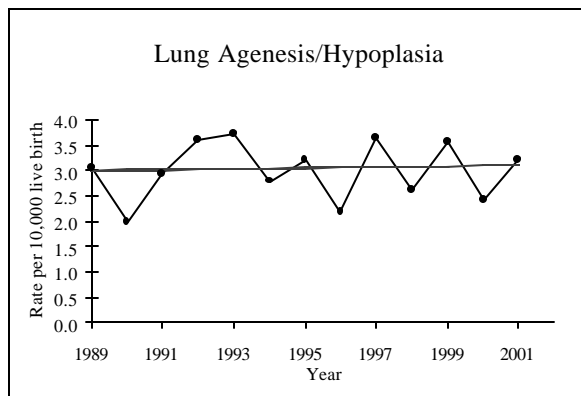


●—● Observed Rates      — Regression Line

<sup>1</sup>Trend is significant; details are given in Table 3.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2003

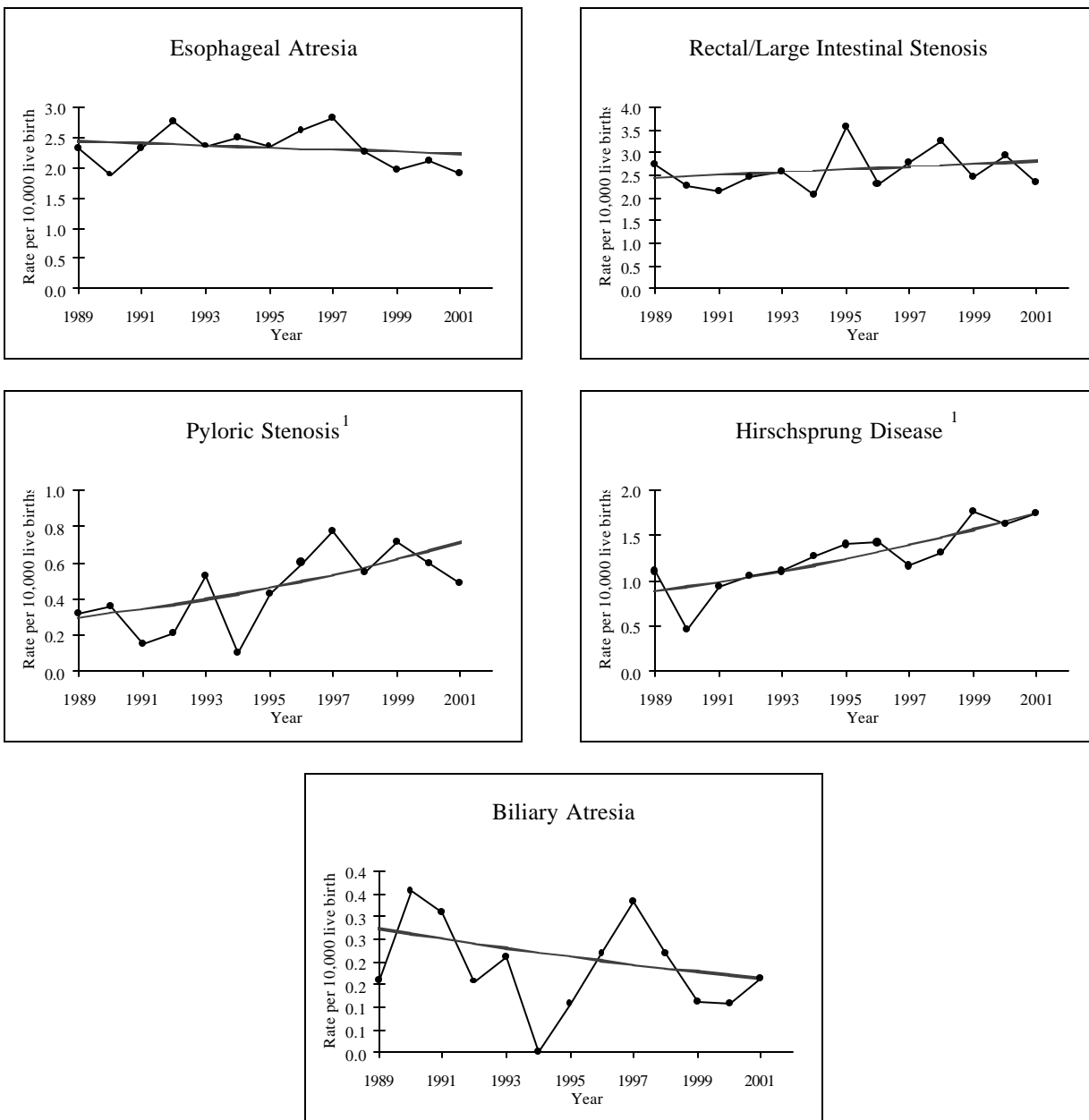
**FIGURE 5. TRENDS IN THE REPORTED PREVALENCE RATES OF RESPIRATORY AND ORAL DEFECTS IDENTIFIED DURING THE NEWBORN STAY, PER 10,000 LIVE BIRTHS 1989-2001**



●—● Observed Rates      — Regression Line

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2003

**FIGURE 6. TRENDS IN THE REPORTED PREVALENCE RATES OF GASTROINTESTINAL DEFECTS IDENTIFIED DURING THE NEWBORN STAY, PER 10,000 LIVE BIRTHS 1989-2001**

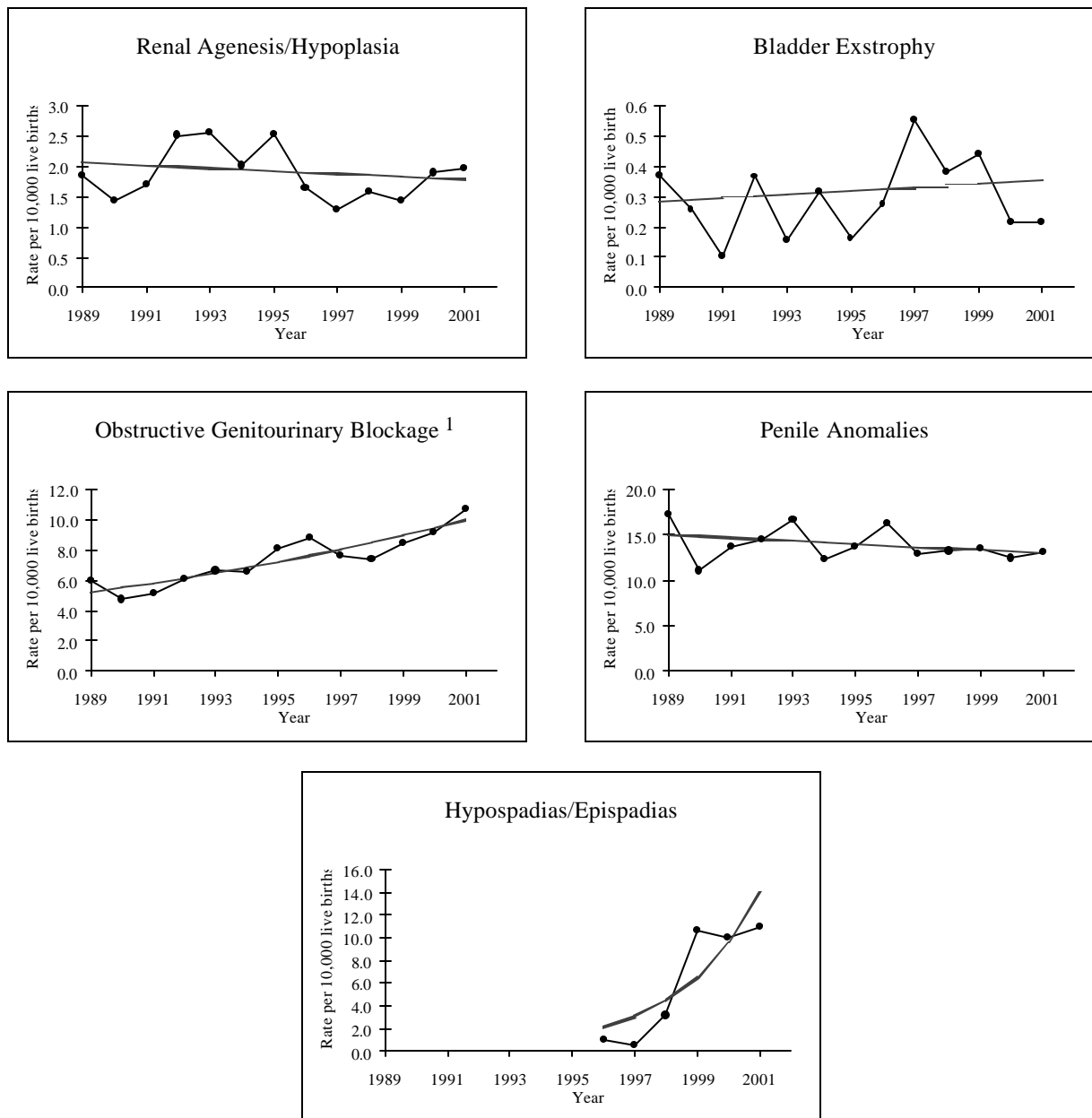


●—● Observed Rates      — Regression Line

<sup>1</sup>Trend is significant; details are given in tables 2 and 3.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2003

**FIGURE 7. TRENDS IN THE REPORTED PREVALENCE RATES OF GENTOURINARY DEFECTS IDENTIFIED DURING THE NEWBORN STAY, PER 10,000 LIVE BIRTHS 1989-2001**



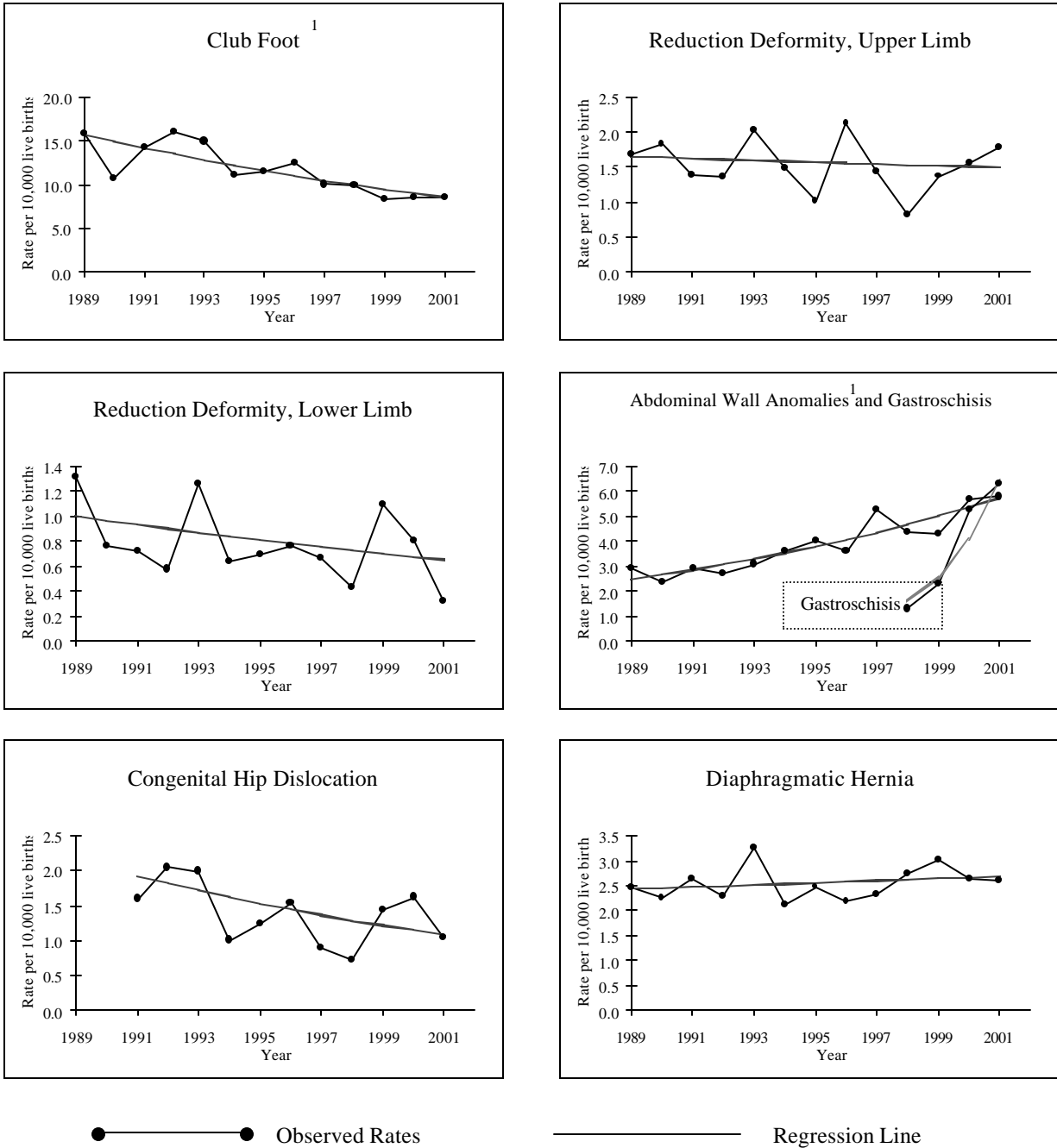
●—● Observed Rates      — Regression Line

<sup>1</sup>Trend is significant; details are given in Table 2.

<sup>2</sup> Includes hypospadias/epispadias; data collection changes do not allow specific analysis prior to 1996.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2003

**FIGURE 8. TRENDS IN THE REPORTED PREVALENCE RATES OF MUSCULOSKELETAL DEFECTS IDENTIFIED DURING THE NEWBORN STAY, PER 10,000 LIVE BIRTHS 1989-2001**

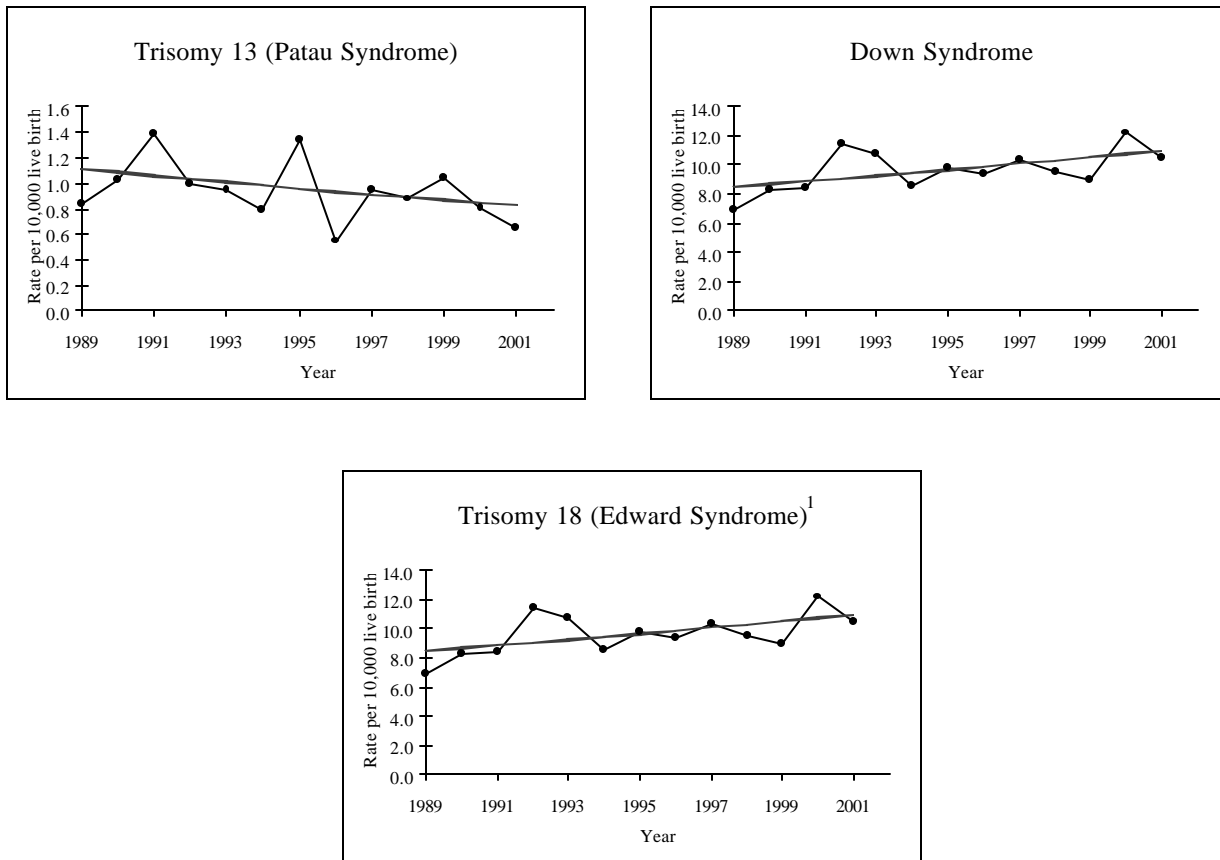


<sup>1</sup>Trend is significant; details are given in tables 2 and 3.

<sup>2</sup> Includes gastrochisis/omphalocele; data collection changes do not allow specific analysis before 1998.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2003

**FIGURE 9. TRENDS IN THE REPORTED PREVALENCE RATES OF CHROMOSOMAL DEFECTS IDENTIFIED DURING THE NEWBORN STAY, PER 10,000 LIVE BIRTHS 1989-2001**

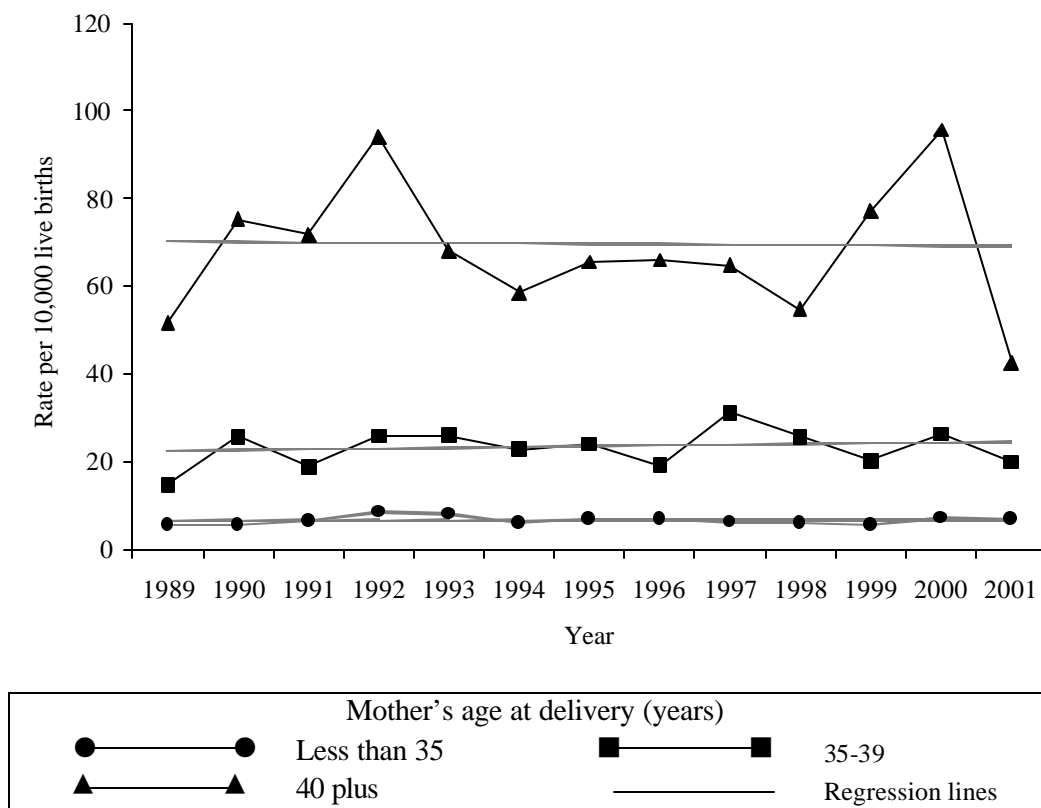


●—● Observed Rates      — Regression Line

<sup>1</sup>Trend is significant; details are given in Table 2.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2003

**FIGURE 10. TRENDS IN THE REPORTED PREVALENCE RATES OF DOWN SYNDROME, BY MATERNAL AGE AT DELIVERY, IDENTIFIED DURING THE NEWBORN STAY, PER 10,000 LIVE BIRTHS 1989-2001**



Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, July 2003

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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. (Hernia of the brain)
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.

**APPENDIX 1**

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

<b>Birth Defect</b>	<b>ICD-9-CM Codes</b>	<b>Description</b>
Congenital cataract	743.30-743.34	An opacity of the lens that occurs in the fetus at some time during the pregnancy and is present at birth.
Coloboma of the eye	743.41-743.44	A deformity where a portion of the structure of the eye is lacking. This gap can occur in the eyelid, iris, lens, choroid or optic disc, and be large or small. The most common form of gap is caused by an imperfect closure of a cleft, present in the womb but usually closed by birth date.
Aniridia	743.45	A 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.
Tetralogy of Fallot	745.2	A congenital defect of the heart consisting of four abnormalities that result in insufficiently oxygenated blood pumped to the body. This condition results in a blue baby at birth due to inadequate oxygenation.
Ventricular septal defect	745.4	A ventricular septal defect is a hole in the wall between the lower chambers of the heart.

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Birth Defect	ICD-9-CM Codes	Description
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 atresia	746.01	Obstruction of the outflow of blood from the right ventricle of the heart at the pulmonary heart valve.
Pulmonary valve stenosis	746.02	Narrowing of the pulmonary heart valve.
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 (hypoplastic).
Patent ductus arteriosus	747.0	A condition when the channel between the pulmonary artery and the aorta fails to close at birth.

**APPENDIX 1**

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

<b>Birth Defect</b>	<b>ICD-9-CM Codes</b>	<b>Description</b>
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.
Pulmonary artery anomalies	747.3	An abnormality in the formation of the pulmonary artery such as stenosis or atresia.
Choanal atresia	748.0	A congenital narrowing or blockage of the nasal airway by membranous or bony tissue.
Lung agenesis/hypoplasia	748.5	The absence or underdevelopment of the lungs that may be bilateral or unilateral.
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 (clefts) in the upper lip--cleft lip can be on one side only or on both sides --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/ 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).

**APPENDIX 1**

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

<b>Birth Defect</b>	<b>ICD-9-CM Codes</b>	<b>Description</b>
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 (meatus) on the under side 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.

## APPENDIX 1

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

Birth Defect	ICD-9-CM Codes	Description
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.
Club foot	754.5x, 754.6x, 754.70, 754.71	A deformity of the foot that results from a malformation of the muscle during the child's fetal development.
Reduction deformity	755.2x, 755.3x	May be of upper or lower limbs. A shortening or absence of one or both 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.
Omphalocele	756.79	A congenital malformation in which part of the intestine protrudes through a physical opening in the abdominal wall into the base of the umbilical cord.
Down syndrome	758.0	A chromosome abnormality resulting in mental retardation, distinctive malformations of the head and face and other abnormalities.
Patau syndrome	758.1	A syndrome associated with the presence of a third (extra) number 13 chromosome. Newborns have numerous internal and external abnormalities, including profound retardation.

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

<b>Birth Defect</b>	<b>ICD-9-CM Codes</b>	<b>Description</b>
Edward syndrome	758.2	A syndrome associated with the presence of a third (extra) number 18 chromosome. It causes major physical abnormalities and severe mental retardation.