

**TRENDS IN THE PREVALENCE OF
BIRTH DEFECTS IN
ILLINOIS AND CHICAGO
1989-1998**

Illinois Department of Public Health
Division of Epidemiologic Studies

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INTRODUCTION

Adverse pregnancy outcomes are recorded by the Illinois Department of Public Health (IDPH) for infants with congenital anomalies (birth defects) and other serious neonatal conditions. Each year in Illinois, IDPH's Adverse Pregnancy Outcomes Reporting System (APORS) obtains information on thousands of such births throughout the state. 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 therefore 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 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, 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 Pew Environmental Health Commission gave APORS a rating of B because of this lack of active surveillance activities.

In 1998, a pilot project to look at the impact of active surveillance was undertaken in Illinois. APORS staff used hospital discharge records to identify unreported birth defects among children up to 1 year of age, born in 1996 or 1997.

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 an updated report of birth defect rates among newborns and infants up to 1 year of age, born in 1996 and 1997, and an initial report of birth defect rates among newborns in 1998. The same 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 Joinpoint Regression Software (Version 2.5, March 2000, 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 1998 are presented in Table 3. Birth defect rates for 1997 and 1996 are presented in tables 4 and 5 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 6, 7 and 8.

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 cannot be analyzed over time. Gastroschisis/omphalocele cases are also contained in the category of abdominal wall anomalies and hypospadias/epispadias cases are also 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 no 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 linear, but may be made up of two straight lines with different slopes. Statistically significant trends were found for 15 birth defects (listed in Tables 1 and 2). 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).

Table 1 also includes a column (average percentage change) that gives that 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 3.6 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 1. Birth Defects Associated with Low Intake of Folic Acid
Showing a Significant Trend in Incidence Rate Between 1989 and 1998**

Selected Birth Defect¹	Significance of trend (P-value)	Average annual % change between 1989 and 1998²
Spina bifida without anencephalus	0.0080	-3.6%
Cleft lip	0.0484	-2.32%
Pyloric stenosis	0.0176	10.8%
Obstructive genitourinary blockage	0.0001	5.4%
Reduction deformity, lower limb	0.0318	-7.0%
Down syndrome	0.0432	3.2%

¹ Birth defects listed in Tables 3-8 but not in this table or in Table 2 did not show any significant changes in incidence rate over time.

² This is a measure of how quickly the rate is changing over time. The time interval is 1989-1998. Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, 6/28/2001

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 increase noted in Table 1 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

Two defects unassociated with folic acid intake, club foot and congenital hip dislocation, showed statistical evidence of a decline between 1989 and 1998 (Table 2). However, the APORS program has clarified the definition of congenital hip dislocation to exclude hip click, which is likely to have contributed to the decline in observed incidence rates. 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 2: Birth Defects Not Believed to Be Associated with Folic Acid Intake
Showing a Significant Trend Between 1989 and 1998**

Selected Birth Defect ¹	Significance of trend (P-value)	Average % change between 1989 and 1998 ²
Atrial septal defect	0.0001	12.5%
Endocardial cushion defect	0.0002	9.3%
Pulmonary valve stenosis and atresia	0.0007	10.2%
Coarctation of aorta	0.0070	5.2%
Pulmonary artery anomalies	0.0304	4.1%
Hirschsprung disease	0.0301	5.2%
Abdominal wall anomalies	0.0001	7.69%
Club foot	0.0138	-4.1%
Congenital hip dislocation ³	0.0060	-12.6%

¹ Birth defects listed in the Tables 3-8 but not in this table or in Table 1 did not show any significant changes in incidence rate over time.

² This is a measure of how quickly the rate is changing over time. The time interval is 1989-1998 except as noted.

³ 1991-1998

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, 6/28/2001

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
1996-1998**

**Table 3. Number and Rate of Selected Birth Defects for 1998
Illinois**

Selected Birth Defects Groups	Newborn Identification		
	N	Rate ¹	95% CI ²
A. CENTRAL NERVOUS SYSTEM			
Anencephalus	32	1.75	(1.20, 2.48)
Spina bifida without anencephalus	49	2.68	(1.99, 3.55)
Hydrocephalus without spina bifida	106	5.81	(4.76, 7.02)
Encephalocele	12	0.66	(0.34, 1.15)
Microcephalus	49	2.68	(1.99, 3.55)
<i>Total Selected CNS Defects</i>	<i>248</i>	<i>13.59</i>	<i>(11.95, 15.39)</i>
B. EYE			
Coloboma of the eye	1	0.05	(0.00, 0.31)
Anophthalmos/Microphthalmos	9	0.49	(0.23, 0.94)
Congenital cataract	7	0.38	(0.15, 0.79)
Aniridia	0	0.00	(0.00, 0.20)
<i>Total Selected Eye Defects</i>	<i>17</i>	<i>0.93</i>	<i>(0.54, 1.49)</i>
C. EAR			
Anotia/Microtia	7	0.38	(0.15, 0.79)
D. CARDIOVASCULAR			
Common truncus	10	0.55	(0.26, 1.01)
Transposition of great vessels	38	2.08	(1.47, 2.86)
Tetralogy of fallot	45	2.47	(1.80, 3.30)
Ventricular septal defect	320	17.53	(15.67, 19.56)
Atrial septal defect	298	16.33	(14.53, 18.29)
Endocardial cushion defect	37	2.03	(1.43, 2.79)
Pulmonary valve stenosis and atresia	46	2.52	(1.85, 3.36)
Tricuspid valve stenosis and atresia	12	0.66	(0.34, 1.15)
Ebstein anomaly	6	0.33	(0.12, 0.72)
Aortic valve stenosis	8	0.44	(0.19, 0.86)
Hypoplastic left heart syndrome	20	1.10	(0.67, 1.69)
Patent ductus arteriosus	636	34.85	(32.19, 37.67)
Coarctation of aorta	36	1.97	(1.38, 2.73)
Pulmonary artery anomalies	184	10.08	(8.68, 11.65)
<i>Total Selected Cardiovascular Defects</i>	<i>1,696</i>	<i>92.93</i>	<i>(88.56, 97.46)</i>
E. RESPIRATORY			
Lung agenesis/hypoplasia	48	2.63	(1.94, 3.49)
F. OROFACIAL			
Cleft palate without cleft lip	66	3.62	(2.80, 4.60)
Cleft lip with and without cleft palate	92	5.04	(4.06, 6.18)
Choanal atresia	20	1.10	(0.67, 1.69)
<i>Total Selected Orofacial Defects</i>	<i>178</i>	<i>9.75</i>	<i>(8.37, 11.30)</i>
G. GASTROINTESTINAL			
Esophageal atresia/Tracheoesophageal fistula	41	2.25	(1.61, 3.05)
Rectal and large intestinal atresia/stenosis	60	3.29	(2.51, 4.23)
Pyloric stenosis	10	0.55	(0.26, 1.01)
Hirschsprung disease (congenital megacolon)	24	1.32	(0.84, 1.96)
Biliary atresia	4	0.22	(0.06, 0.56)
<i>Total Selected Gastrointestinal Defects</i>	<i>139</i>	<i>7.62</i>	<i>(6.40, 8.99)</i>

Selected Birth Defects Groups	Newborn Identification		
	N	Rate ¹	95% CI ²
H. GENITOURINARY			
Renal agenesis/hypoplasia	29	1.59	(1.06, 2.28)
Bladder exstrophy	8	0.44	(0.19, 0.86)
Obstructive genitourinary defect	135	7.40	(6.20, 8.76)
Penile anomalies	242	13.26	(11.64, 15.04)
Hypospadias/Epispadias ³	57	3.12	(2.37, 4.05)
<i>Total Selected Genitourinary Defects</i>	<i>414</i>	<i>22.68</i>	<i>(20.55, 24.98)</i>
I. MUSCULOSKELETAL			
Club foot	182	9.97	(8.58, 11.53)
Reduction deformity, upper limbs	15	0.82	(0.46, 1.36)
Reduction deformity, lower limbs	8	0.44	(0.19, 0.86)
Abdominal wall defects	82	4.49	(3.57, 5.58)
Gastroschisis/Omphalocele ³	25	1.37	(0.89, 2.02)
Congenital hip dislocation	13	0.71	(0.38, 1.22)
Diaphragmatic hernia	50	2.74	(2.03, 3.61)
<i>Total Selected Musculoskeletal Defects</i>	<i>350</i>	<i>19.18</i>	<i>(17.22, 21.30)</i>
J. CHROMOSOMAL			
Trisomy 13 (Patau syndrome)	16	0.88	(0.50, 1.42)
Down syndrome	173	9.48	(8.12, 11.00)
Trisomy 18 (Edward syndrome)	26	1.42	(0.93, 2.09)
<i>Total Selected Chromosomal Defects</i>	<i>215</i>	<i>11.78</i>	<i>(10.26, 13.47)</i>
<i>Total All Selected Defects</i>	<i>3,264</i>	<i>178.85</i>	<i>(172.76, 185.09)</i>

¹ Rate per 10,000 live births

² 95% confidence interval for rate

³ Subset of previous birth defect group

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, 6/28/2001

**Table 4. Number and Rate of Selected Birth Defects for 1997
Illinois**

Selected Birth Defects Groups	Newborn Identification			Up to 1 year Identification		
	N	Rate ¹	95% CI ²	N	Rate ¹	95% CI ²
A. CENTRAL NERVOUS SYSTEM						
Anencephalus	31	1.72	(1.17, 2.44)	31	1.72	(1.17, 2.44)
Spina bifida without anencephalus	35	1.94	(1.35, 2.69)	49	2.71	(2.01, 3.59)
Hydrocephalus without spina bifida	103	5.70	(4.65, 6.91)	117	6.48	(5.36, 7.76)
Encephalocele	12	0.66	(0.34, 1.16)	12	0.66	(0.34, 1.16)
Microcephalus	56	3.10	(2.34, 4.03)	69	3.82	(2.97, 4.83)
<i>Total Selected CNS Defects</i>	237	13.12	(11.50, 14.90)	278	15.39	(13.63, 17.31)
B. EYE						
Coloboma of the eye	4	0.22	(0.06, 0.57)	7	0.39	(0.16, 0.80)
Anophthalmos/Microphthalmos	11	0.61	(0.30, 1.09)	13	0.72	(0.38, 1.23)
Congenital cataract	8	0.44	(0.19, 0.87)	12	0.66	(0.34, 1.16)
Aniridia	0	0.00	(0.00, 0.20)	0	0.00	(0.00, 0.20)
<i>Total Selected Eye Defects</i>	23	1.27	(0.81, 1.91)	32	1.77	(1.21, 2.50)
C. EAR						
Anotia/Microtia	7	0.39	(0.16, 0.80)	7	0.39	(0.16, 0.80)
D. CARDIOVASCULAR						
Common truncus	7	0.39	(0.16, 0.80)	7	0.39	(0.16, 0.80)
Transposition of great vessels	34	1.88	(1.30, 2.63)	46	2.55	(1.86, 3.40)
Tetralogy of fallot	41	2.27	(1.63, 3.08)	57	3.16	(2.39, 4.09)
Ventricular septal defect	316	17.49	(15.62, 19.53)	438	24.25	(22.03, 26.63)
Atrial septal defect	304	16.83	(14.99, 18.83)	451	24.97	(22.71, 27.38)
Endocardial cushion defect	42	2.32	(1.68, 3.14)	52	2.88	(2.15, 3.77)
Pulmonary valve stenosis and atresia	43	2.38	(1.72, 3.21)	86	4.76	(3.81, 5.88)
Tricuspid valve stenosis and atresia	10	0.55	(0.27, 1.02)	13	0.72	(0.38, 1.23)
Ebstein anomaly	7	0.39	(0.16, 0.80)	11	0.61	(0.30, 1.09)
Aortic valve stenosis	16	0.89	(0.51, 1.44)	22	1.22	(0.76, 1.84)
Hypoplastic left heart syndrome	28	1.55	(1.03, 2.24)	32	1.77	(1.21, 2.50)
Patent ductus arteriosus	705	39.03	(36.20, 42.02)	825	45.67	(42.61, 48.89)
Coarctation of aorta	34	1.88	(1.30, 2.63)	53	2.93	(2.20, 3.84)
Pulmonary artery anomalies	162	8.97	(7.64, 10.46)	271	15.00	(13.27, 16.90)
<i>Total Selected Cardiovascular Defects</i>	1,749	96.82	(92.33, 101.46)	2,364	130.86	(125.64, 136.25)
E. RESPIRATORY						
Lung agenesis/hypoplasia	66	3.65	(2.83, 4.65)	75	4.15	(3.27, 5.20)
F. OROFACIAL						
Cleft palate without cleft lip	56	3.10	(2.34, 4.03)	60	3.32	(2.53, 4.28)
Cleft lip with and without cleft palate	115	6.37	(5.26, 7.64)	133	7.36	(6.16, 8.73)
Choanal atresia	18	1.00	(0.59, 1.57)	23	1.27	(0.81, 1.91)
<i>Total Selected Orofacial Defects</i>	189	10.46	(9.02, 12.06)	216	11.96	(10.42, 13.66)
G. GASTROINTESTINAL						
Esophageal atresia/Tracheoesophageal fistula	51	2.82	(2.10, 3.71)	52	2.88	(2.15, 3.77)
Rectal and large intestinal atresia/stenosis	50	2.77	(2.05, 3.65)	58	3.21	(2.44, 4.15)
Pyloric stenosis	14	0.77	(0.42, 1.30)	146	8.08	(6.82, 9.50)
Hirschsprung disease (congenital megacolon)	21	1.16	(0.72, 1.78)	32	1.77	(1.21, 2.50)
Biliary atresia	6	0.33	(0.12, 0.72)	10	0.55	(0.27, 1.02)
<i>Total Selected Gastrointestinal Defects</i>	142	7.86	(6.62, 9.26)	298	16.50	(14.68, 18.48)

Selected Birth Defects Groups	Newborn Identification			Up to 1 year Identification		
	N	Rate ¹	95% CI ²	N	Rate ¹	95% CI ²
H. GENITOURINARY						
Renal agenesis/hypoplasia	23	1.27	(0.81, 1.91)	29	1.61	(1.08, 2.31)
Bladder exstrophy	10	0.55	(0.27, 1.02)	11	0.61	(0.30, 1.09)
Obstructive genitourinary defect	138	7.64	(6.42, 9.03)	207	11.46	(9.95, 13.13)
Penile anomalies	233	12.90	(11.29, 14.66)	288	15.94	(14.15, 17.89)
Hypospadias/Epispadias ³	8	0.44	(0.19, 0.87)	46	2.55	(1.86, 3.40)
<i>Total Selected Genitourinary Defects</i>	<i>404</i>	<i>22.36</i>	<i>(20.24, 24.65)</i>	<i>535</i>	<i>29.62</i>	<i>(27.16, 32.23)</i>
I. MUSCULOSKELETAL						
Club foot	181	10.02	(8.61, 11.59)	216	11.9	(10.42, 13.66)
Reduction deformity, upper limbs	26	1.44	(0.94, 2.11)	34	1.88	(1.30, 2.63)
Reduction deformity, lower limbs	12	0.66	(0.34, 1.16)	18	1.00	(0.59, 1.57)
Abdominal wall defects	96	5.31	(4.30, 6.49)	99	5.48	(4.45, 6.67)
Gastroschisis/Omphalocele ³	0	0.00	(0.00, 0.20)	1	0.06	(0.00, 0.31)
Congenital hip dislocation	16	0.89	(0.51, 1.44)	24	1.33	(0.85, 1.98)
Diaphragmatic hernia	42	2.32	(1.68, 3.14)	44	2.44	(1.77, 3.27)
<i>Total Selected Musculoskeletal Defects</i>	<i>373</i>	<i>20.65</i>	<i>(18.61, 22.85)</i>	<i>435</i>	<i>24.08</i>	<i>(21.87, 26.45)</i>
J. CHROMOSOMAL						
Trisomy 13 (Patau syndrome)	16	0.89	(0.51, 1.44)	17	0.94	(0.55, 1.51)
Down syndrome	205	11.35	(9.85, 13.01)	205	11.35	(9.85, 13.01)
Trisomy 18 (Edward syndrome)	15	0.83	(0.46, 1.37)	32	1.77	(1.21, 2.50)
<i>Total Selected Chromosomal Defects</i>	<i>236</i>	<i>13.06</i>	<i>(11.45, 14.84)</i>	<i>254</i>	<i>14.06</i>	<i>(12.38, 15.90)</i>
<i>Total All Selected Defects</i>	<i>3,426</i>	<i>189.65</i>	<i>(183.35 196.11)</i>	<i>4,494</i>	<i>248.77</i>	<i>(241.55 256.15)</i>

¹ Rate per 10,000 live births

² 95% confidence interval for rate

³ Subset of previous birth defect group

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, 6/28/2001

**Table 5. Number and Rate of Selected Birth Defects for 1996
Illinois**

Selected Birth Defects Groups	Newborn Identification			Up to 1 year Identification		
	N	Rate ¹	95% CI ²	N	Rate ¹	95% CI ²
A. CENTRAL NERVOUS SYSTEM						
Anencephalus	42	2.29	(1.65, 3.10)	42	2.29	1.65, 3.10)
Spina bifida without anencephalus	50	2.73	(2.03, 3.60)	63	3.44	2.64, 4.40)
Hydrocephalus without spina bifida	101	5.52	(4.49, 6.70)	126	6.88	5.73, 8.19)
Encephalocele	15	0.82	(0.46, 1.35)	19	1.04	0.62, 1.62)
Microcephalus	70	3.82	(2.98, 4.83)	93	5.08	4.10, 6.22)
<i>Total Selected CNS Defects</i>	<i>278</i>	<i>15.18</i>	<i>(13.45, 17.08)</i>	<i>343</i>	<i>18.74</i>	<i>16.80, 20.83)</i>
B. EYE						
Coloboma of the eye	0	0.00	(0.00, 0.20)	2	0.11	0.01, 0.39)
Anophthalmos/Microphthalmos	22	1.20	(0.75, 1.82)	25	1.37	0.88, 2.02)
Congenital cataract	9	0.49	(0.22, 0.93)	21	1.15	0.71, 1.75)
Aniridia	0	0.00	(0.00, 0.20)	0	0.00	0.00, 0.20)
<i>Total Selected Eye Defects</i>	<i>31</i>	<i>1.69</i>	<i>(1.15, 2.40)</i>	<i>48</i>	<i>2.62</i>	<i>1.93, 3.48)</i>
C. EAR						
Anotia/Microtia	5	0.27	(0.09, 0.64)	6	0.33	0.12, 0.71)
D. CARDIOVASCULAR						
Common truncus	8	0.44	(0.19, 0.86)	9	0.49	0.22, 0.93)
Transposition of great vessels	31	1.69	(1.15, 2.40)	58	3.17	2.41, 4.10)
Tetralogy of fallot	37	2.02	(1.42, 2.79)	58	3.17	2.41, 4.10)
Ventricular septal defect	339	18.52	(16.60, 20.60)	502	27.42	25.07, 29.93)
Atrial septal defect	298	16.28	(14.48, 18.23)	474	25.89	23.61, 28.33)
Endocardial cushion defect	31	1.69	(1.15, 2.40)	53	2.89	2.17, 3.79)
Pulmonary valve stenosis and atresia	44	2.40	(1.75, 3.23)	103	5.63	4.59, 6.82)
Tricuspid valve stenosis and atresia	9	0.49	(0.22, 0.93)	17	0.93	0.54, 1.49)
Ebstein anomaly	7	0.38	(0.15, 0.79)	7	0.38	0.15, 0.79)
Aortic valve stenosis	10	0.55	(0.26, 1.00)	17	0.93	0.54, 1.49)
Hypoplastic left heart syndrome	19	1.04	(0.62, 1.62)	34	1.86	1.29, 2.60)
Patent ductus arteriosus	710	38.78	(35.98, 41.74)	859	46.92	43.83, 50.17)
Coarctation of aorta	35	1.91	(1.33, 2.66)	65	3.55	2.74, 4.53)
Pulmonary artery anomalies	174	9.50	(8.14, 11.03)	299	16.33	14.53, 18.29)
<i>Total Selected Cardiovascular Defects</i>	<i>1,75</i>	<i>95.70</i>	<i>(91.27, 100.28)</i>	<i>2,55</i>	<i>139.56</i>	<i>134.20, 145.08)</i>
E. RESPIRATORY						
Lung agenesis/hypoplasia	39	2.13	(1.51, 2.91)	53	2.89	2.17, 3.79)
F. OROFACIAL						
Cleft palate without cleft lip	74	4.04	(3.17, 5.07)	91	4.97	4.00, 6.10)
Cleft lip with and without cleft palate	110	6.01	(4.94, 7.24)	132	7.21	6.03, 8.55)
Choanal atresia	13	0.71	(0.38, 1.21)	17	0.93	0.54, 1.49)
<i>Total Selected Orofacial Defects</i>	<i>197</i>	<i>10.76</i>	<i>(9.31, 12.37)</i>	<i>240</i>	<i>13.11</i>	<i>11.50, 14.88)</i>
G. GASTROINTESTINAL						
Esophageal atresia/Tracheoesophageal fistula	48	2.62	(1.93, 3.48)	53	2.89	2.17, 3.79)
Rectal and large intestinal atresia/stenosis	42	2.29	(1.65, 3.10)	47	2.57	1.89, 3.41)
Pyloric stenosis	10	0.55	(0.26, 1.00)	202	11.03	9.56, 12.66)
Hirschsprung disease (congenital megacolon)	26	1.42	(0.93, 2.08)	37	2.02	1.42, 2.79)
Biliary atresia	4	0.22	(0.06, 0.56)	7	0.38	0.15, 0.79)
<i>Total Selected Gastrointestinal Defects</i>	<i>130</i>	<i>7.10</i>	<i>(5.93, 8.43)</i>	<i>346</i>	<i>18.90</i>	<i>16.96, 21.00)</i>

Selected Birth Defects Groups	Newborn Identification			Up to 1 year Identification		
	N	Rate ¹	95% CI ²	N	Rate ¹	95% CI ²
H. GENITOURINARY						
Renal agenesis/hypoplasia	28	1.53	(1.02, 2.21)	38	2.08	1.47, 2.85)
Bladder exstrophy	5	0.27	(0.09, 0.64)	5	0.27	0.09, 0.64)
Obstructive genitourinary defect	160	8.74	(7.44, 10.20)	211	11.53	10.02, 13.19)
Penile anomalies	298	16.28	(14.48, 18.23)	363	19.83	17.84, 21.98)
Hypospadias/Epispadias ³	14	0.76	(0.42, 1.28)	54	2.95	2.22, 3.85)
<i>Total Selected Genitourinary Defects</i>	<i>491</i>	<i>26.82</i>	<i>(24.50, 29.30)</i>	<i>617</i>	<i>33.70</i>	<i>31.09, 36.47)</i>
I. MUSCULOSKELETAL						
Club foot	229	12.51	(10.94, 14.24)	281	15.35	13.61, 17.25)
Reduction deformity, upper limbs	38	2.08	(1.47, 2.85)	41	2.24	1.61, 3.04)
Reduction deformity, lower limbs	13	0.71	(0.38, 1.21)	17	0.93	0.54, 1.49)
Abdominal wall defects	96	5.24	(4.25, 6.40)	99	5.41	4.39, 6.58)
Gastroschisis/Omphalocele ³	0	0.00	(0.00, 0.20)	0	0.00	0.00, 0.20)
Congenital hip dislocation	27	1.47	(0.97, 2.15)	31	1.69	1.15, 2.40)
Diaphragmatic hernia	40	2.18	(1.56, 2.98)	45	2.46	1.79, 3.29)
<i>Total Selected Musculoskeletal Defects</i>	<i>443</i>	<i>24.20</i>	<i>(22.00, 26.56)</i>	<i>514</i>	<i>28.08</i>	<i>25.70, 30.61)</i>
J. CHROMOSOMAL						
Trisomy 13 (Patau syndrome)	11	0.60	(0.30, 1.08)	11	0.60	0.30, 1.08)
Down syndrome	195	10.65	(9.21, 12.26)	196	10.71	9.26, 12.31)
Trisomy 18 (Edward syndrome)	11	0.60	(0.30, 1.08)	35	1.91	1.33, 2.66)
<i>Total Selected Chromosomal Defects</i>	<i>217</i>	<i>11.85</i>	<i>(10.33, 13.54)</i>	<i>242</i>	<i>13.22</i>	<i>11.61, 14.99)</i>
<i>Total All Selected Defects</i>	<i>3,583</i>	<i>195.71</i>	<i>(189.35, 202.22)</i>	<i>4,964</i>	<i>271.14</i>	<i>263.65, 278.79)</i>

¹ Rate per 10,000 live births

² 95% confidence interval for rate

³ Subset of previous birth defect group

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, 6/28/2001

**Table 6. Number and Rate of Selected Birth Defects for 1998
Chicago**

Selected Birth Defects Groups	Newborn Identification		
	N	Rate ¹	95% CI ²
A. CENTRAL NERVOUS SYSTEM			
Anencephalus	0	0.00	(0.00, 0.72)
Spina bifida without anencephalus	12	2.33	(1.20, 4.07)
Hydrocephalus without spina bifida	31	6.02	(4.09, 8.54)
Encephalocele	2	0.39	(0.05, 1.40)
Microcephalus	23	4.46	(2.83, 6.70)
<i>Total Selected CNS Defects</i>	68	13.20	(10.25, 16.73)
B. EYE			
Coloboma of the eye	0	0.00	(0.00, 0.72)
Anophthalmos/Microphthalmos	2	0.39	(0.05, 1.40)
Congenital cataract	2	0.39	(0.05, 1.40)
Aniridia	0	0.00	(0.00, 0.72)
<i>Total Selected Eye Defects</i>	4	0.78	(0.21, 1.99)
C. EAR			
Anotia/Microtia	0	0.00	(0.00, 0.72)
D. CARDIOVASCULAR			
Common truncus	3	0.58	(0.12, 1.70)
Transposition of great vessels	6	1.16	(0.43, 2.53)
Tetralogy of fallot	8	1.55	(0.67, 3.06)
Ventricular septal defect	43	8.35	(6.04, 11.24)
Atrial septal defect	28	5.44	(3.61, 7.86)
Endocardial cushion defect	9	1.75	(0.80, 3.32)
Pulmonary valve stenosis and atresia	4	0.78	(0.21, 1.99)
Tricuspid valve stenosis and atresia	5	0.97	(0.32, 2.26)
Ebstein anomaly	2	0.39	(0.05, 1.40)
Aortic valve stenosis	0	0.00	(0.00, 0.72)
Hypoplastic left heart syndrome	5	0.97	(0.32, 2.26)
Patent ductus arteriosus	102	19.80	(16.14, 24.03)
Coarctation of aorta	6	1.16	(0.43, 2.53)
Pulmonary artery anomalies	24	4.66	(2.98, 6.93)
<i>Total Selected Cardiovascular Defects</i>	245	47.56	(41.79, 53.90)
E. RESPIRATORY			
Lung agenesis/hypoplasia	9	1.75	(0.80, 3.32)
F. OROFACIAL			
Cleft palate without cleft lip	8	1.55	(0.67, 3.06)
Cleft lip with and without cleft palate	12	2.33	(1.20, 4.07)
Choanal atresia	2	0.39	(0.05, 1.40)
<i>Total Selected Orofacial Defects</i>	22	4.27	(2.68, 6.47)
G. GASTROINTESTINAL			
Esophageal atresia/Tracheoesophageal fistula	9	1.75	(0.80, 3.32)
Rectal and large intestinal atresia/stenosis	22	4.27	(2.68, 6.47)
Pyloric stenosis	1	0.19	(0.00, 1.08)
Hirschsprung disease (congenital megacolon)	7	1.36	(0.55, 2.80)
Biliary atresia	0	0.00	(0.00, 0.72)
<i>Total Selected Gastrointestinal Defects</i>	39	7.57	(5.38, 10.35)

Selected Birth Defects Groups	Newborn Identification		
	N	Rate ¹	95% CI ²
H. GENITOURINARY			
Renal agenesis/hypoplasia	3	0.58	(0.12, 1.70)
Bladder exstrophy	3	0.58	(0.12, 1.70)
Obstructive genitourinary defect	29	5.63	(3.77, 8.08)
Penile anomalies	32	6.21	(4.25, 8.77)
Hypospadias/Epispadias ³	8	1.55	(0.67, 3.06)
<i>Total Selected Genitourinary Defects</i>	<i>67</i>	<i>13.01</i>	<i>(10.08, 16.52)</i>
I. MUSCULOSKELETAL			
Club foot	32	6.21	(4.25, 8.77)
Reduction deformity, upper limbs	4	0.78	(0.21, 1.99)
Reduction deformity, lower limbs	2	0.39	(0.05, 1.40)
Abdominal wall defects	16	3.11	(1.78, 5.04)
Gastroschisis/Omphalocele ³	5	0.97	(0.32, 2.26)
Congenital hip dislocation	4	0.78	(0.21, 1.99)
Diaphragmatic hernia	15	2.91	(1.63, 4.80)
<i>Total Selected Musculoskeletal Defects</i>	<i>73</i>	<i>14.17</i>	<i>(11.11, 17.82)</i>
J. CHROMOSOMAL			
Trisomy 13 (Patau syndrome)	2	0.39	(0.05, 1.40)
Down syndrome	36	6.99	(4.89, 9.67)
Trisomy 18 (Edward syndrome)	4	0.78	(0.21, 1.99)
<i>Total Selected Chromosomal Defects</i>	<i>42</i>	<i>8.15</i>	<i>(5.88, 11.02)</i>
<i>Total All Selected Defects</i>	<i>569</i>	<i>110.45</i>	<i>(101.56, 119.91)</i>

¹ Rate per 10,000 live births

² 95% confidence interval for rate

³ Subset of previous birth defect group

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, 6/28/2001

**Table 7. Number and Rate of Selected Birth Defects for 1997
Chicago**

Selected Birth Defects Groups	Newborn Identification			Up to 1 year Identification		
	N	Rate ¹	95% CI ²	N	Rate ¹	95% CI ²
A. CENTRAL NERVOUS SYSTEM						
Anencephalus	0	0.00	(0.00, 0.72)	0	0.00	(0.00, 0.72)
Spina bifida without anencephalus	8	1.57	(0.68, 3.08)	8	1.57	(0.68, 3.08)
Hydrocephalus without spina bifida	29	5.67	(3.80, 8.15)	32	6.26	(4.28, 8.84)
Encephalocele	3	0.59	(0.12, 1.72)	3	0.59	(0.12, 1.72)
Microcephalus	28	5.48	(3.64, 7.92)	28	5.48	(3.64, 7.92)
<i>Total Selected CNS Defects</i>	68	13.30	(10.33, 16.86)	71	13.89	(10.85, 17.52)
B. EYE						
Coloboma of the eye	0	0.00	(0.00, 0.72)	0	0.00	(0.00, 0.72)
Anophthalmos/Microphthalmos	2	0.39	(0.05, 1.41)	2	0.39	(0.05, 1.41)
Congenital cataract	3	0.59	(0.12, 1.72)	3	0.59	(0.12, 1.72)
Aniridia	0	0.00	(0.00, 0.72)	0	0.00	(0.00, 0.72)
<i>Total Selected Eye Defects</i>	5	0.98	(0.32, 2.28)	5	0.98	(0.32, 2.28)
C. EAR						
Anotia/Microtia	2	0.39	(0.05, 1.41)	2	0.39	(0.05, 1.41)
D. CARDIOVASCULAR						
Common truncus	4	0.78	(0.21, 2.00)	4	0.78	(0.21, 2.00)
Transposition of great vessels	7	1.37	(0.55, 2.82)	9	1.76	(0.81, 3.34)
Tetralogy of fallot	7	1.37	(0.55, 2.82)	8	1.57	(0.68, 3.08)
Ventricular septal defect	46	9.00	(6.59, 12.00)	55	10.76	(8.11, 14.01)
Atrial septal defect	42	8.22	(5.92, 11.11)	50	9.78	(7.26, 12.90)
Endocardial cushion defect	11	2.15	(1.07, 3.85)	14	2.74	(1.50, 4.60)
Pulmonary valve stenosis and atresia	0	0.00	(0.00, 0.72)	3	0.59	(0.12, 1.72)
Tricuspid valve stenosis and atresia	4	0.78	(0.21, 2.00)	5	0.98	(0.32, 2.28)
Ebstein anomaly	2	0.39	(0.05, 1.41)	2	0.39	(0.05, 1.41)
Aortic valve stenosis	1	0.20	(0.00, 1.09)	1	0.20	(0.00, 1.09)
Hypoplastic left heart syndrome	1	0.20	(0.00, 1.09)	1	0.20	(0.00, 1.09)
Patent ductus arteriosus	145	28.37	(23.94, 33.38)	155	30.32	(25.74, 35.49)
Coarctation of aorta	8	1.57	(0.68, 3.08)	9	1.76	(0.81, 3.34)
Pulmonary artery anomalies	24	4.70	(3.01, 6.99)	30	5.87	(3.96, 8.38)
<i>Total Selected Cardiovascular Defects</i>	302	59.08	(52.60, 66.13)	346	67.69	(60.74, 75.21)
E. RESPIRATORY						
Lung agenesis/hypoplasia	13	2.54	(1.35, 4.35)	15	2.93	(1.64, 4.84)
F. OROFACIAL						
Cleft palate without cleft lip	13	2.54	(1.35, 4.35)	14	2.74	(1.50, 4.60)
Cleft lip with and without cleft palate	23	4.50	(2.85, 6.75)	24	4.70	(3.01, 6.99)
Choanal atresia	4	0.78	(0.21, 2.00)	5	0.98	(0.32, 2.28)
<i>Total Selected Orofacial Defects</i>	40	7.83	(5.59, 10.66)	43	8.41	(6.09, 11.33)
G. GASTROINTESTINAL						
Esophageal atresia/Tracheoesophageal fistula	15	2.93	(1.64, 4.84)	16	3.13	(1.79, 5.08)
Rectal and large intestinal atresia/stenosis	15	2.93	(1.64, 4.84)	15	2.93	(1.64, 4.84)
Pyloric stenosis	0	0.00	(0.00, 0.72)	3	0.59	(0.12, 1.72)
Hirschsprung disease (congenital megacolon)	7	1.37	(0.55, 2.82)	9	1.76	(0.81, 3.34)
Biliary atresia	1	0.20	(0.00, 1.09)	2	0.39	(0.05, 1.41)
<i>Total Selected Gastrointestinal Defects</i>	38	7.43	(5.26, 10.20)	45	8.80	(6.42, 11.78)

Selected Birth Defects Groups	Newborn Identification			Up to 1 year Identification		
	N	Rate ¹	95% CI ²	N	Rate ¹	95% CI ²
H. GENITOURINARY						
Renal agenesis/hypoplasia	4	0.78	(0.21, 2.00)	4	0.78	(0.21, 2.00)
Bladder exstrophy	4	0.78	(0.21, 2.00)	4	0.78	(0.21, 2.00)
Obstructive genitourinary defect	34	6.65	(4.61, 9.29)	36	7.04	(4.93, 9.75)
Penile anomalies	36	7.04	(4.93, 9.75)	40	7.83	(5.59, 10.66)
Hypospadias/Epispadias ³	1	0.20	(0.00, 1.09)	2	0.39	(0.05, 1.41)
<i>Total Selected Genitourinary Defects</i>	78	15.26	(12.06, 19.04)	84	16.43	(13.11, 20.35)
I. MUSCULOSKELETAL						
Club foot	31	6.06	(4.12, 8.61)	33	6.46	(4.44, 9.07)
Reduction deformity, upper limbs	3	0.59	(0.12, 1.72)	3	0.59	(0.12, 1.72)
Reduction deformity, lower limbs	2	0.39	(0.05, 1.41)	4	0.78	(0.21, 2.00)
Abdominal wall defects	31	6.06	(4.12, 8.61)	31	6.06	(4.12, 8.61)
Gastroschisis/Omphalocele ³	0	0.00	(0.00, 0.72)	0	0.00	(0.00, 0.72)
Congenital hip dislocation	6	1.17	(0.43, 2.55)	7	1.37	(0.55, 2.82)
Diaphragmatic hernia	10	1.96	(0.94, 3.60)	10	1.96	(0.94, 3.60)
<i>Total Selected Musculoskeletal Defects</i>	83	16.24	(12.93, 20.13)	88	17.22	(13.81, 21.21)
J. CHROMOSOMAL						
Trisomy 13 (Patau syndrome)	0	0.00	(0.00, 0.72)	0	0.00	(0.00, 0.72)
Down syndrome	41	8.02	(5.76, 10.88)	42	8.22	(5.92, 11.11)
Trisomy 18 (Edward syndrome)	5	0.98	(0.32, 2.28)	6	1.17	(0.43, 2.55)
<i>Total Selected Chromosomal Defects</i>	46	9.00	(6.59, 12.00)	48	9.39	(6.92, 12.45)
<i>Total All Selected Defects</i>	675	132.05	(122.27 142.40)	747	146.14	(135.84 157.00)

¹ Rate per 10,000 live births

² 95% confidence interval for rate

³ Subset of previous birth defect group

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, 6/28/2001

**Table 8. Number and Rate of Selected Birth Defects for 1996
Chicago**

Selected Birth Defects Groups	Newborn Identification			Up to 1 year Identification		
	N	Rate ¹	95% CI ²	N	Rate ¹	95% CI ²
A. CENTRAL NERVOUS SYSTEM						
Anencephalus	5	0.95	(0.31, 2.21)	5	0.95	(0.31, 2.21)
Spina bifida without anencephalus	10	1.89	(0.91, 3.48)	13	2.46	(1.31, 4.21)
Hydrocephalus without spina bifida	27	5.11	(3.37, 7.44)	32	6.06	(4.14, 8.55)
Encephalocele	6	1.14	(0.42, 2.47)	7	1.32	(0.53, 2.73)
Microcephalus	41	7.76	(5.57, 10.53)	45	8.52	(6.21, 11.40)
<i>Total Selected CNS Defects</i>	89	16.85	(13.53, 20.73)	102	19.31	(15.74, 23.44)
B. EYE						
Coloboma of the eye	0	0.00	(0.00, 0.70)	0	0.00	(0.00, 0.70)
Anophthalmos/Microphthalmos	6	1.14	(0.42, 2.47)	6	1.14	(0.42, 2.47)
Congenital cataract	0	0.00	(0.00, 0.70)	1	0.19	(0.00, 1.05)
Aniridia	0	0.00	(0.00, 0.70)	0	0.00	(0.00, 0.70)
<i>Total Selected Eye Defects</i>	6	1.14	(0.42, 2.47)	7	1.32	(0.53, 2.73)
C. EAR						
Anotia/Microtia	0	0.00	(0.00, 0.70)	0	0.00	(0.00, 0.70)
D. CARDIOVASCULAR						
Common truncus	2	0.38	(0.05, 1.37)	2	0.38	(0.05, 1.37)
Transposition of great vessels	6	1.14	(0.42, 2.47)	9	1.70	(0.78, 3.23)
Tetralogy of fallot	5	0.95	(0.31, 2.21)	7	1.32	(0.53, 2.73)
Ventricular septal defect	54	10.22	(7.68, 13.34)	66	12.49	(9.66, 15.89)
Atrial septal defect	43	8.14	(5.89, 10.96)	60	11.36	(8.67, 14.62)
Endocardial cushion defect	8	1.51	(0.65, 2.98)	12	2.27	(1.17, 3.97)
Pulmonary valve stenosis and atresia	6	1.14	(0.42, 2.47)	20	3.79	(2.31, 5.85)
Tricuspid valve stenosis and atresia	5	0.95	(0.31, 2.21)	6	1.14	(0.42, 2.47)
Ebstein anomaly	2	0.38	(0.05, 1.37)	2	0.38	(0.05, 1.37)
Aortic valve stenosis	2	0.38	(0.05, 1.37)	2	0.38	(0.05, 1.37)
Hypoplastic left heart syndrome	2	0.38	(0.05, 1.37)	3	0.57	(0.12, 1.66)
Patent ductus arteriosus	139	26.31	(22.12, 31.07)	164	31.04	(26.47, 36.17)
Coarctation of aorta	4	0.76	(0.21, 1.94)	8	1.51	(0.65, 2.98)
Pulmonary artery anomalies	17	3.22	(1.87, 5.15)	32	6.06	(4.14, 8.55)
<i>Total Selected Cardiovascular Defects</i>	295	55.84	(49.65, 62.59)	393	74.39	(67.21, 82.12)
E. RESPIRATORY						
Lung agenesis/hypoplasia	5	0.95	(0.31, 2.21)	11	2.08	(1.04, 3.73)
F. OROFACIAL						
Cleft palate without cleft lip	17	3.22	(1.87, 5.15)	18	3.41	(2.02, 5.38)
Cleft lip with and without cleft palate	22	4.16	(2.61, 6.30)	28	5.30	(3.52, 7.66)
Choanal atresia	4	0.76	(0.21, 1.94)	5	0.95	(0.31, 2.21)
<i>Total Selected Orofacial Defects</i>	43	8.14	(5.89, 10.96)	51	9.65	(7.19, 12.69)
G. GASTROINTESTINAL						
Esophageal atresia/Tracheoesophageal fistula	13	2.46	(1.31, 4.21)	14	2.65	(1.45, 4.45)
Rectal and large intestinal atresia/stenosis	15	2.84	(1.59, 4.68)	16	3.03	(1.73, 4.92)
Pyloric stenosis	3	0.57	(0.12, 1.66)	17	3.22	(1.87, 5.15)
Hirschsprung disease (congenital megacolon)	9	1.70	(0.78, 3.23)	10	1.89	(0.91, 3.48)
Biliary atresia	1	0.19	(0.00, 1.05)	1	0.19	(0.00, 1.05)
<i>Total Selected Gastrointestinal Defects</i>	41	7.76	(5.57, 10.53)	58	10.98	(8.34, 14.19)

Selected Birth Defects Groups	Newborn Identification			Up to 1 year Identification		
	N	Rate ¹	95% CI ²	N	Rate ¹	95% CI ²
H. GENITOURINARY						
Renal agenesis/hypoplasia	5	0.95	(0.31, 2.21)	6	1.14	(0.42, 2.47)
Bladder exstrophy	1	0.19	(0.00, 1.05)	1	0.19	(0.00, 1.05)
Obstructive genitourinary defect	43	8.14	(5.89, 10.96)	46	8.71	(6.37, 11.61)
Penile anomalies	34	6.44	(4.46, 8.99)	38	7.19	(5.09, 9.87)
Hypospadias/Epispadias ³	0	0.00	(0.00, 0.70)	1	0.19	(0.00, 1.05)
<i>Total Selected Genitourinary Defects</i>	<i>83</i>	<i>15.71</i>	<i>(12.51, 19.48)</i>	<i>91</i>	<i>17.22</i>	<i>(13.87, 21.15)</i>
I. MUSCULOSKELETAL						
Club foot	39	7.38	(5.25, 10.09)	45	8.52	(6.21, 11.40)
Reduction deformity, upper limbs	6	1.14	(0.42, 2.47)	8	1.51	(0.65, 2.98)
Reduction deformity, lower limbs	2	0.38	(0.05, 1.37)	3	0.57	(0.12, 1.66)
Abdominal wall defects	19	3.60	(2.17, 5.62)	19	3.60	(2.17, 5.62)
Gastroschisis/Omphalocele ³	0	0.00	(0.00, 0.70)	0	0.00	(0.00, 0.70)
Congenital hip dislocation	4	0.76	(0.21, 1.94)	5	0.95	(0.31, 2.21)
Diaphragmatic hernia	10	1.89	(0.91, 3.48)	12	2.27	(1.17, 3.97)
<i>Total Selected Musculoskeletal Defects</i>	<i>80</i>	<i>15.14</i>	<i>(12.01, 18.85)</i>	<i>92</i>	<i>17.41</i>	<i>(14.04, 21.36)</i>
J. CHROMOSOMAL						
Trisomy 13 (Patau syndrome)	1	0.19	(0.00, 1.05)	1	0.19	(0.00, 1.05)
Down syndrome	43	8.14	(5.89, 10.96)	48	9.09	(6.70, 12.05)
Trisomy 18 (Edward syndrome)	8	1.51	(0.65, 2.98)	8	1.51	(0.65, 2.98)
<i>Total Selected Chromosomal Defects</i>	<i>52</i>	<i>9.84</i>	<i>(7.35, 12.91)</i>	<i>57</i>	<i>10.79</i>	<i>(8.17, 13.98)</i>
<i>Total All Selected Defects</i>	<i>694</i>	<i>131.36</i>	<i>(121.77, 144.51)</i>	<i>862</i>	<i>163.16</i>	<i>(152.45, 174.43)</i>

¹ Rate per 10,000 live births

² 95% confidence interval for rate

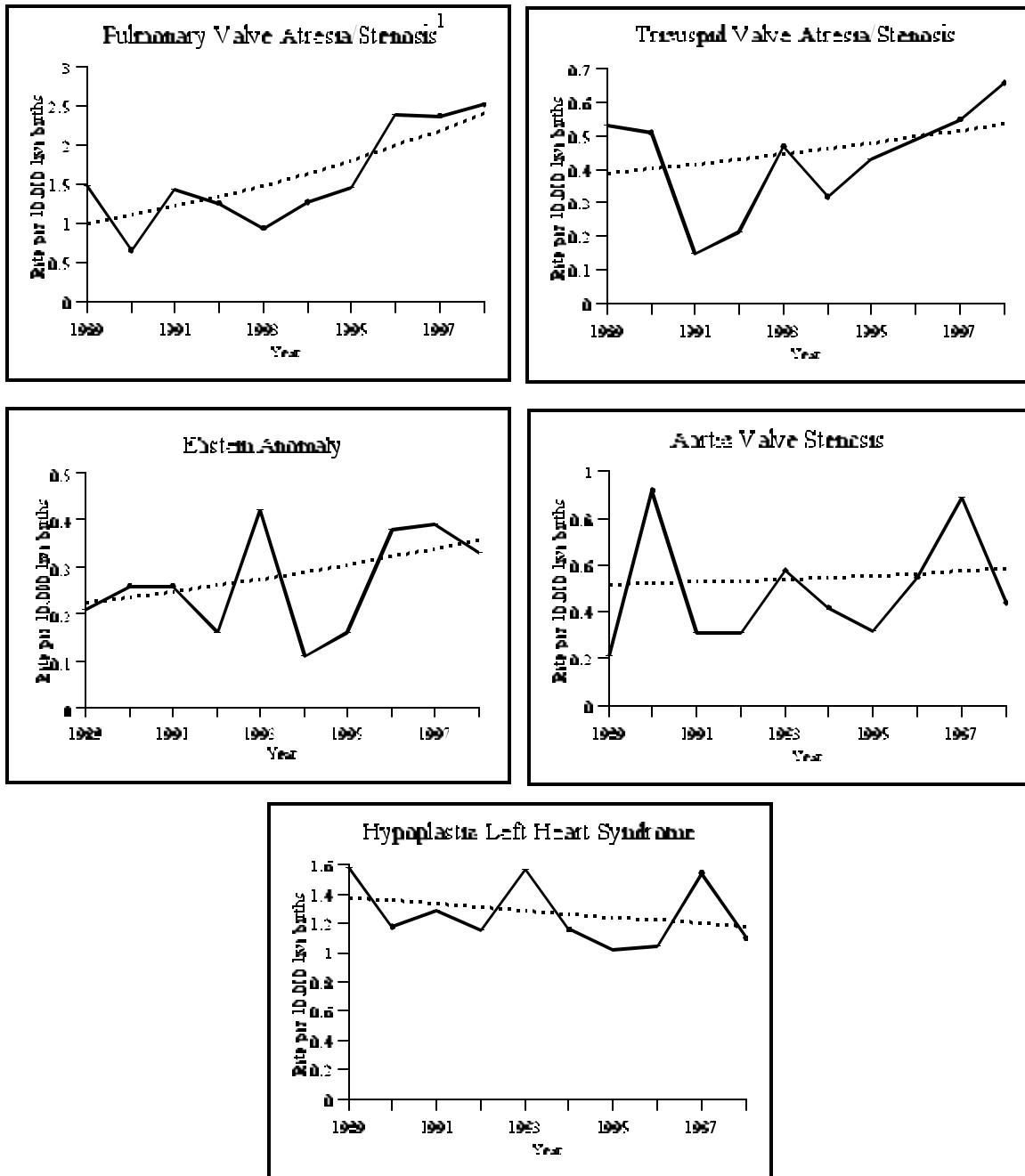
³ Subset of previous birth defect group

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, 6/28/2001

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

1989-1998

FIGURE 3b. TRENDS IN THE REPORTED PREVALENCE RATES OF CARDIAC DEFECTS IDENTIFIED DURING THE NEWBORN STAY, PER 10,000 LIVE BIRTHS 1989-1998

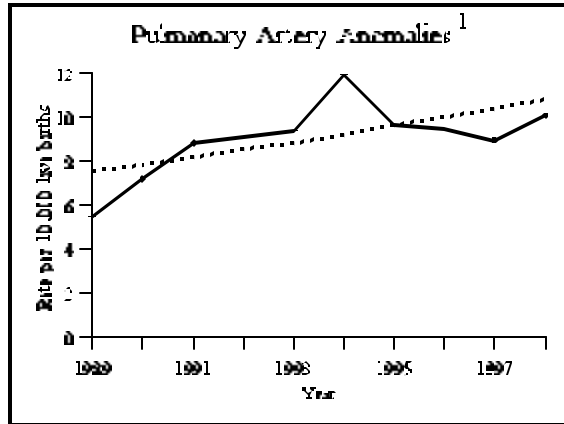
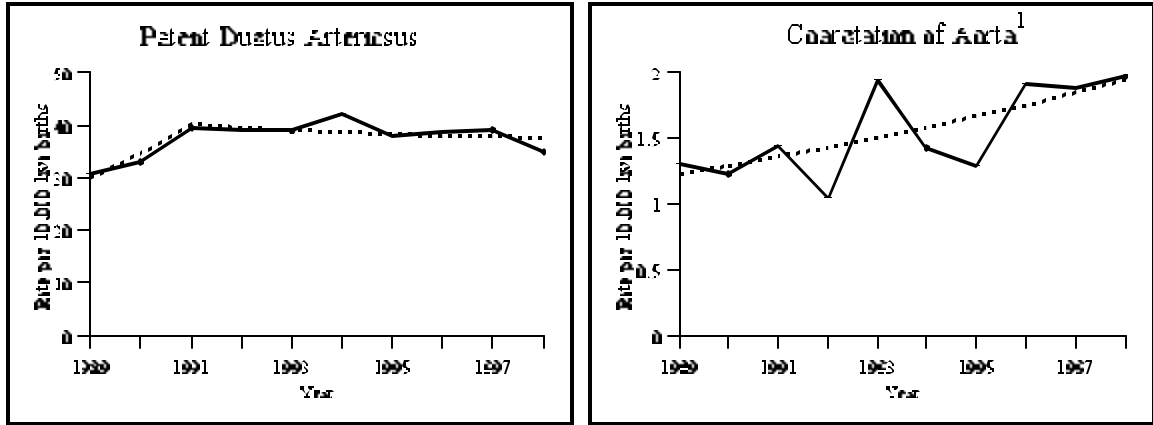


— Observed Rates Regression Line

¹Trend is significant; details are given in Table 2.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, 6/28/2001

FIGURE 4. TRENDS IN THE REPORTED PREVALENCE RATES OF CIRCULATORY DEFECTS IDENTIFIED DURING THE NEWBORN STAY, PER 10,000 LIVE BIRTHS 1989-1998

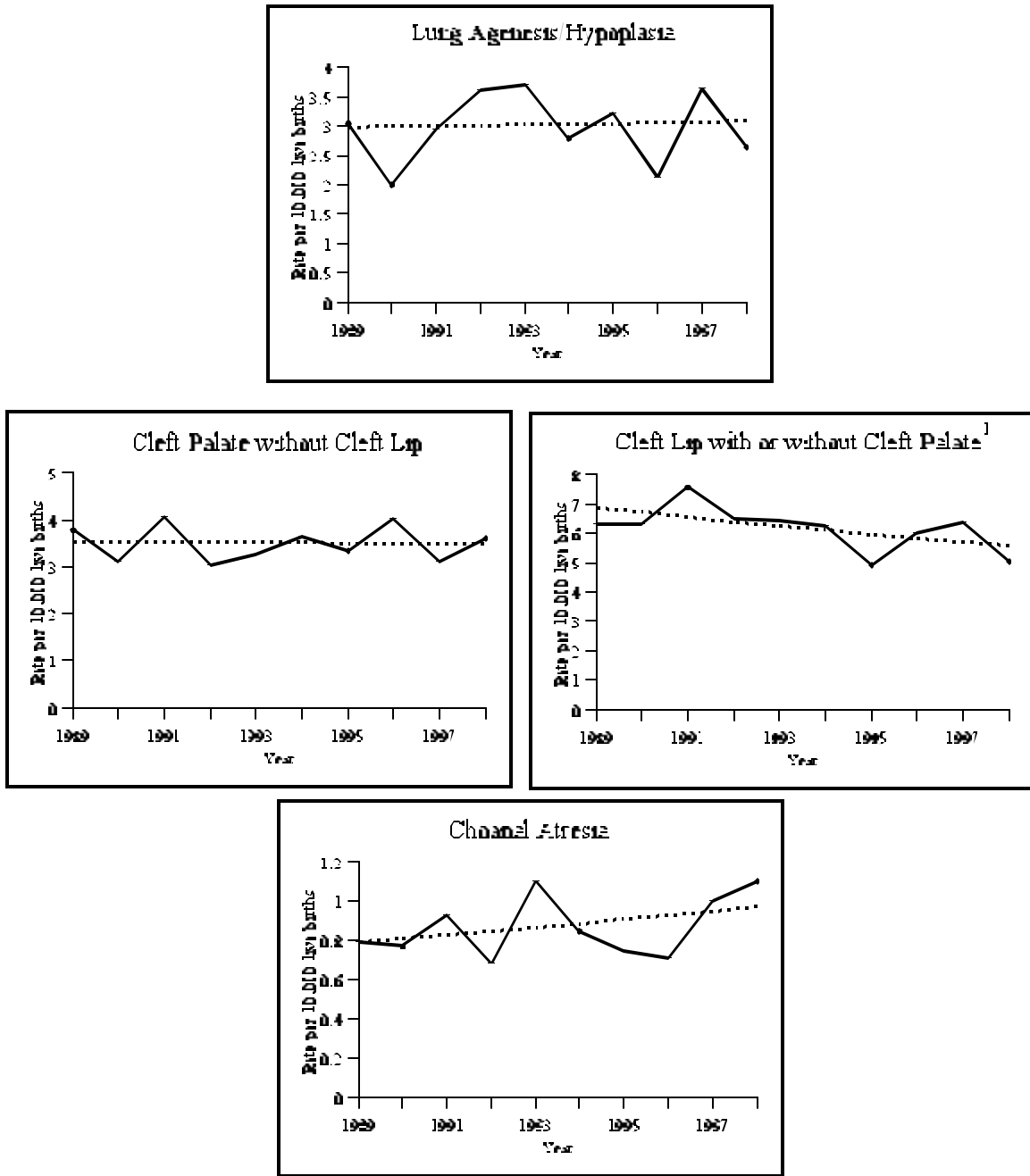


————— Observed Rates Regression Line

¹Trend is significant; details are given in Table 2.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, 6/28/2001

FIGURE 5. TRENDS IN THE REPORTED PREVALENCE RATES OF RESPIRATORY AND ORAL DEFECTS IDENTIFIED DURING THE NEWBORN STAY, PER 10,000 LIVE BIRTHS 1989-1998

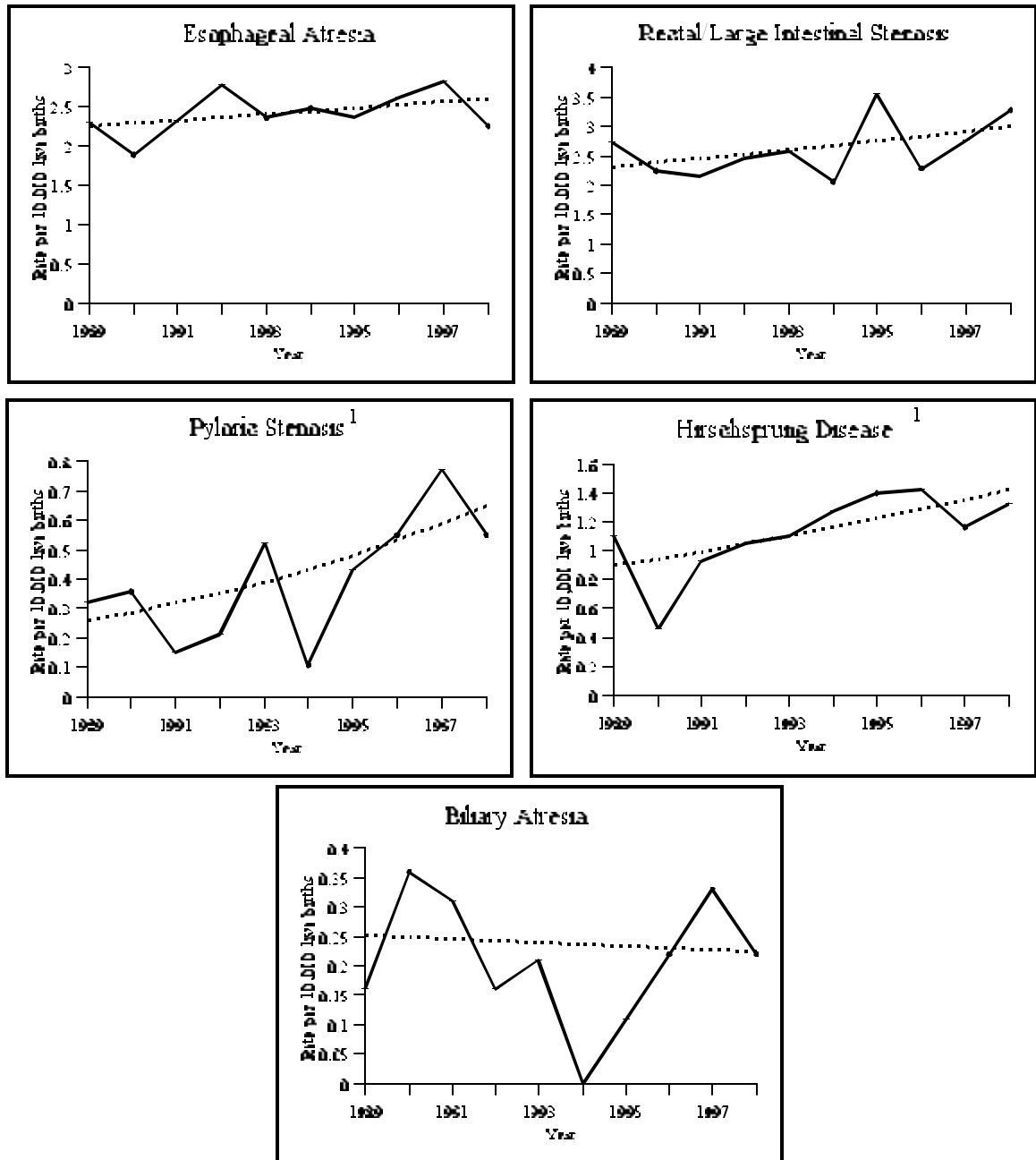


— Observed Rates Regression Line

¹Trend is significant; details are given in Table 1.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, 6/28/2001

**FIGURE 6. TRENDS IN THE REPORTED PREVALENCE RATES OF
GASTROINTESTINAL DEFECTS
IDENTIFIED DURING THE NEWBORN STAY, PER 10,000 LIVE BIRTHS
1989-1998**

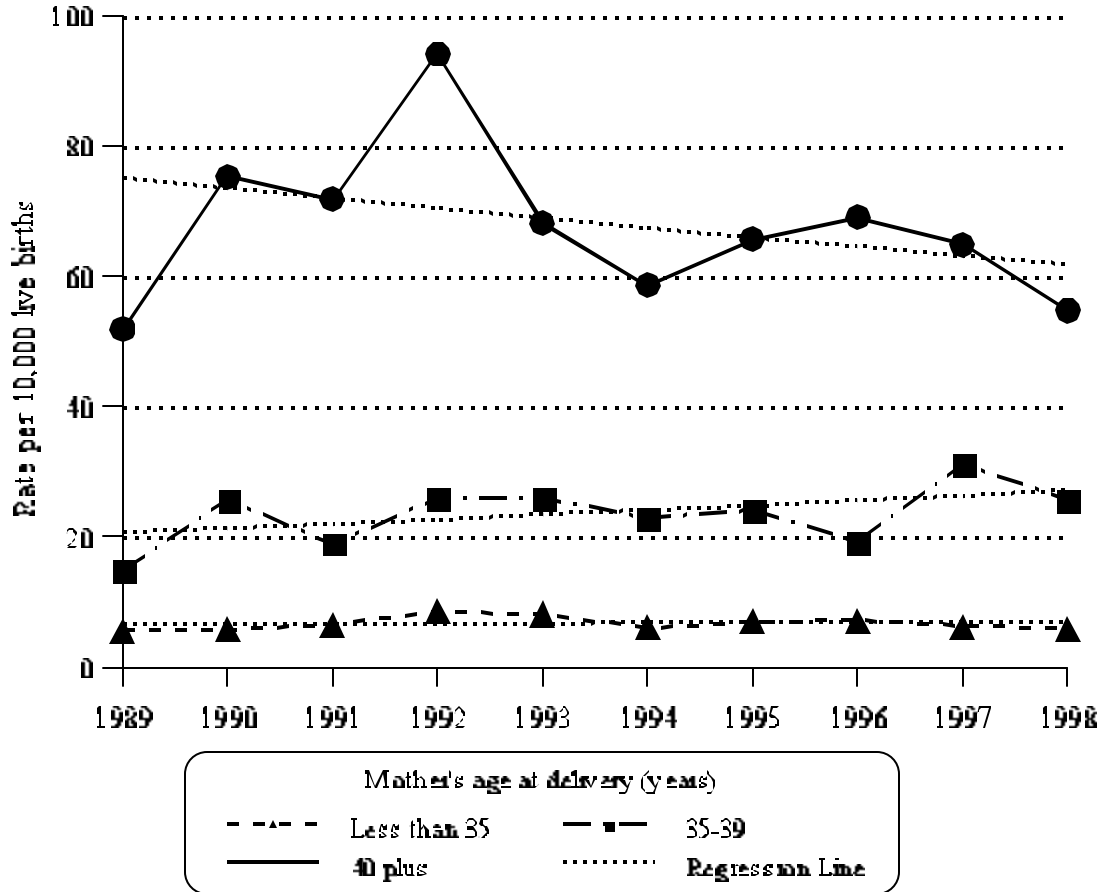


————— Observed Rates ··········· Regression Line

¹Trend is significant; details are given in tables 1 and 2.

Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, 6/28/2001

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 1989-1998



Source: Illinois Department of Public Health, Adverse Pregnancy Outcomes Reporting System, 6/28/2001

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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.

Birth Defect	ICD-9-CM Codes	Description
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	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.

Birth Defect	ICD-9-CM Codes	Description
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.
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.

Birth Defect	ICD-9-CM Codes	Description
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.
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.

Birth Defect	ICD-9-CM Codes	Description
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).
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.)

Birth Defect	ICD-9-CM Codes	Description
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.
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.

Birth Defect	ICD-9-CM Codes	Description
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.
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.

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