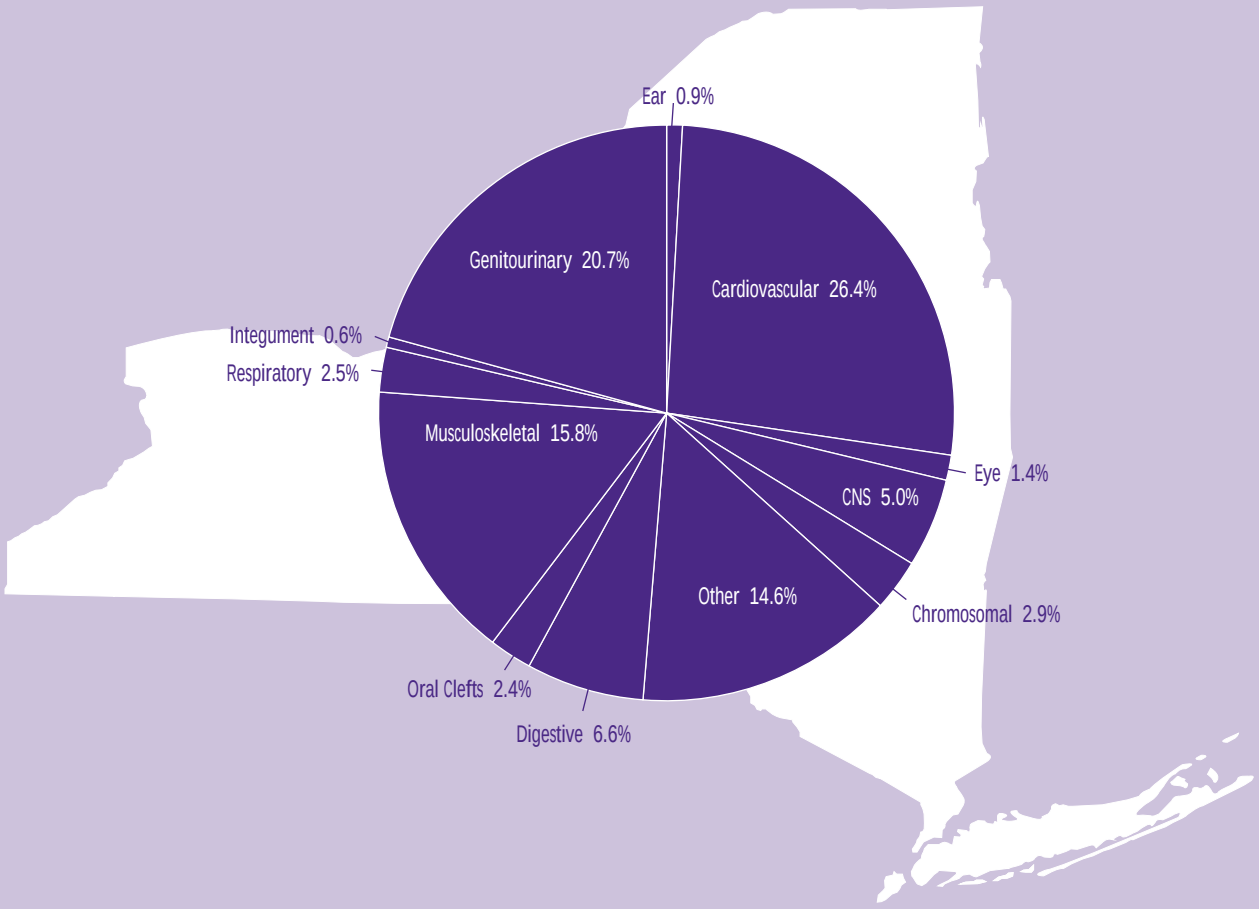
New York State Department of Health

Congenital Malformations Registry Annual Report



Statistical Summary of Children Born in 1995 and Diagnosed Through 1997

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New York State Department of Health

Congenital Malformations Registry

Annual Report

Statistical Summary of Children Born in 1995 and Diagnosed Through 1997

TABLE OF CONTENTS

Summary

This report presents rates of congenital malformations occurring among the 271,042 children who were born alive to New York State residents in 1995. The children reported with a major congenital malformation represent 3.9% of live births. Males had a higher rate of major congenital malformations than females (4.6% versus 3.1%), and black children had a higher major malformation rate than white children (4.4% versus 3.8%).

Section I of this report presents demographic characteristics of children reported to the registry, number of malformations and age at diagnosis. Other sections present the distribution of anomalies by organ system, rates for selected malformations by race and sex and the most common malformations for each county in the state.

This is the twelfth cohort report from the Congenital Malformations Registry. Reports are also available for the 1983-1994 birth cohorts. The statistics in this report are **not** comparable to reports before 1992. Cohort reports for the years 1989 to 1991 were not based on birth certificate matched cases. For this report, birth certificates were used to supplement or correct reported data. Birth certificate matching also helped eliminate duplicate cases reported under different names and nonresident births. In addition, 1992 was the first year that the registry used a new coding system, which allows for more specificity. For previous years, ICD-9 codes were used.

PROGRAM OVERVIEW

Background

Congenital malformations are the leading cause of infant mortality in the United States[1]. They are the fifth leading cause of years of potential life lost and a major cause of morbidity and mortality throughout childhood[1,2]. Twenty percent of infant deaths are attributed to congenital malformations[2], a percentage that has increased over time[1,2]. Approximately 25% of pediatric hospital admissions and about one-third of the total number of pediatric hospital days are for congenital malformations of various types[3]. Little is known about the causes of congenital malformations. Twenty percent may be due to a combination of heredity and other factors; 7.5% may be due to single gene mutations; 6% to chromosome abnormalities; and 5% to maternal illnesses, such as diabetes, infections or anticonvulsant drugs[4]. Approximately 40% to 60% of congenital malformations are of unknown origin[4,5].

Although radiation and rubella had been linked to birth defects, not until the thalidomide tragedy of the early 1960s was there a widespread interest in possible associations between congenital malformations and environmental agents. During the 1970s, interest continued to grow in birth defects and birth defects surveillance as a result of the growing recognition of the problems of toxic waste dumps such as Love Canal and accidents such as Three Mile Island and Seveso. In response, many states began to develop birth defects registries in order to have data for tracking trends in malformation rates[6,7]. A birth defects registry also makes it possible to respond to public concerns about possible excess occurrence of malformations with timely, objective investigations. A birth defects registry can provide cases for traditional epidemiologic studies of specific congenital malformations and provide information for the planning, provision and evaluation of health services[6,7].

New York State
Congenital Malformations Registry

The New York State Department of Health Congenital Malformations Registry (CMR) is one of the largest statewide, population-based birth defects registries in the nation. The concept of the Congenital Malformations Registry arose out of recognition of the environment as a potential etiologic factor in the occurrence of congenital malformations. Health studies during the Love Canal crisis revealed the inadequacies of relying on birth certificates to monitor and evaluate birth defects.

New York's Congenital Malformations Registry was established by enactment of Part 22 of the State Sanitary Code in 1981. Reporting to the registry began in October 1982. Hospitals and physicians are required to report children under two years of age diagnosed with a malformation. The majority of reports are sent by hospitals, primarily from their medical records departments. A small number are sent by individual physicians. The CMR relies upon physicians to verify diagnoses initially suspected in the hospital but confirmed on an outpatient basis, and to clarify nonspecific diagnoses reported by hospitals.

The Congenital Malformations Registry receives case reports on children diagnosed up to two years of age who were born or reside in New York State with a congenital malformation, chromosomal anomaly or persistent metabolic defect. For purposes of this registry and report, a congenital malformation is defined as any structural, functional or biochemical abnormality, determined genetically or induced during gestation and not due to birthing events.

Case reports are received on forms (see Appendix 1) provided by the Department of Health (DOH). Pertinent fields are coded and the narrative description of the malformation is converted to a code. The case report is matched to existing registry reports for possible duplicates. Data from reporting forms are entered on microcomputers and then transferred to the DOH mainframe for updating of the master files.

All information reported to the registry is held in strict confidence. Records and computer files are maintained in accordance with DOH regulations concerning data containing individual identifiers. Access to the data by anyone other than registry personnel is restricted and carefully monitored to

ensure that confidentiality is maintained. Families of children reported to the registry are never contacted without prior consent of the DOH's Institutional Review Board and notification of the child's physician.

1995 Report

This current report presents statistics for major anomalies only (see Appendix 2). This is in accordance with the practices of other state birth defects registries and allows better comparison between New York State rates and rates in other states. Minor anomalies may cause problems in the determination of malformation rates because they are common and variably reported. They may not even be recorded in the medical chart. Some studies have found rates of minor congenital anomalies as high as 21%[8].

The statistics in this report are **not** comparable to reports prior to 1992. The 1995 report is based on birth certificate matched cases with resident live births from the vital records file used as the denominator. The available birth certificate fields are used to supplement or correct reported data. Birth certificate data are used to establish maternal residence at birth. Birth certificate matching helps eliminate duplicate cases reported under different names. Racial data are not comparable because race is defined by maternal race from the birth certificate. In earlier years, race was defined by what was reported on the CMR form, which may differ from what is recorded on the birth certificate. In 1992, the registry began using a new coding system, the modified British Pediatric Association code (BPA). This coding scheme is used by a number of other congenital malformations registries and allows for greater specificity than does the ICD-9 system. Since 1992, the list of major malformations has been revised (see Appendix 4) changing the list of major malformations used in Sections I and II and the number of specific malformation prevalences in Section III.

CMR Birth Cohort reports are intended as a resource for programs providing primary, secondary and tertiary preventive health care and for public officials concerned with reducing overall mortality and morbidity. The first annual cohort included children born in 1983 and reported with a malformation diagnosed before their second birthday[9]. This report describes children born in 1995 and diagnosed before their second birthday. Reports are also available for the 1984 through 1994 birth cohorts. Some reports and additional information are available through the DOH Web site at http://www.health.state.ny.us.

Limitations

Care should be taken in the use of these data. Virtually all reports are abstracted from inpatient hospital records, since malformations diagnosed on an outpatient basis are not well reported. Accurate hospital clinical recognition of malformations depends on clinical acumen and interest. This is particularly true of conditions more difficult to diagnose, such as fetal alcohol syndrome. Consequently, identification of malformations may vary by area and by time. The abstracting of records requires well-trained medical records professionals who are fastidious in their reporting of such findings. Areas with hospitals that provide higher levels of care may have more thorough diagnoses and, thus, apparently higher rates. Similarly, areas with hospitals that report cases more completely will also appear to have higher rates. In regions with low numbers of births, small variations in incidence may produce large statistical fluctuations.

New York State Population

Based on population projections from the 1990 census, the 1995 population of New York State was about 18.3 million; more than 40% of the population lived in New York City. An additional 23% of the population lived in the six counties closest to New York City. In 1995, there were 271,042 resident live births reported to the state's vital registration, 21% to black mothers, and 19.2% to Hispanic mothers. The race of the child is based on race of the mother only. Nearly 47% of live births were to New York City residents.

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8. Merlob P, Papier CM, Klingberg MA, Reisner SH. Incidence of congenital malformations in the newborn, particularly minor abnormalities. In: Marois, ed. *Prevention of physical and mental congenital defects, Part C: Basic and medical sciences, education and future strategies. Proceedings of a conference of the Institut de la Vie*. New York: Alan R. Liss, 1985: 51-53.

9. New York State Department of Health. *Congenital Malformations Registry Annual Report: 1983 Birth Cohort.*

NOTES

Section I
Demographic Characteristics of Children Reported with Major Malformations

Introduction to Tables

These tables are based on children resident in New York State who were live born in 1995 and reported to the registry with major malformations. Since a new coding system began to be used in 1992, the list of major malformations has been revised (see Appendix 4). Thus, the prevalence in this report are not comparable to reports prior to 1992.

About 77% of children reported with major malformations have only one major malformation (Table 1). The overall occurrences of major malformations are 3.9% of live births. Male children have a higher rate of major malformations than female children (4.6% versus 3.1%, Table 2). This difference is consistent within different racial groups. The rates for major malformations are somewhat higher for black than for white children (4.4% versus 3.8%). The major malformation rate among children with residence at birth in New York State excluding New York City was approximately the same as children with residence at birth in New York City (3.9%). The smaller number of births in the "other" racial category makes these rates difficult to interpret.

Since most children had one major malformation, the race-sex patterns seen for all major malformations are similar to the patterns seen in children with a single major malformation (Table 3). All race-sex groups for children with multiple major malformations showed little variation (Table 4).

Most major malformations were diagnosed within the first three days of life for all races (76.0%, Table 5). The percentage diagnosed within the first three days was slightly higher for white (76.5%) than for black children (74.9%). The proportion of diagnoses made at age six months or later is somewhat greater for black children (5.7%) than for white children (5.2% Table 5).

Section I - Table 1

1995 Births - New York State Residents

Number of Major Malformations Per Child

Number of Malformations	Number of Children	Percent
All Children	10,484	100.0**
1	8,109	77.3
2	1,447	13.8
3	456	4.3
4	207	2.0
5	100	1.0
6	68	0.6
7	41	0.4
8	20	0.2
9	15	0.1
10	10	0.1
11	5	*
12	3	*
13	1	*
14	2	*

* Less than 0.05%

** Total may not add to 100 due to rounding

Section I - Table 2
1995 Births - New York State Residents
Percent of Live Births With One Or More Major Malformations
Sex by Race and Residence

Race and Residence	Both Sexes Infants	Both Sexes Total Births	Both Sexes %	Males Infants	Males Total Births	Males %	Females Infants	Females Total Births	Females %
New York State									
All Races‡	10,484	271,042	3.9	6,384	138,583	4.6	4,100	132,457	3.1
White	7,438	196,558	3.8	4,611	100,784	4.6	2,827	95,774	3.0
Black	2,463	55,556	4.4	1,429	28,070	5.1	1,034	27,484	3.8
Other	495	17,118	2.9	295	8,828	3.3	200	8,290	2.4
NYS Excluding NYC									
All Races‡	5,593	144,879	3.9	3,469	73,984	4.7	2,124	70,893	3.0
White	4,768	124,688	3.8	2,985	63,700	4.7	1,783	60,988	2.9
Black	645	14,711	4.4	386	7,452	5.2	259	7,257	3.6
Other	123	4,418	2.8	70	2,301	3.0	53	2,117	2.5
New York City									
All Races‡	4,891	126,163	3.9	2,915	64,599	4.5	1,976	61,564	3.2
White	2,670	71,870	3.7	1,626	37,084	4.4	1,044	34,786	3.0
Black	1,818	40,845	4.5	1,043	20,618	5.1	775	20,227	3.8
Other	372	12,700	2.9	225	6,527	3.4	147	6,173	2.4

‡Total includes unknowns within each category, thus row and column figures may not sum to totals.

Section I - Table 3
1995 Births - New York State Residents
Percent of Live Births With One Major Malformations
Sex by Race and Residence

Race and Residence	Both Sexes Infants	Both Sexes Total Births	Both Sexes %	Males Infants	Males Total Births	Males %	Females Infants	Females Total Births	Females %
New York State									
All Races‡	8,109	271,042	3.0	5,007	138,583	3.6	3,102	132,457	2.3
White	5,738	196,558	2.9	3,622	100,784	3.6	2,116	95,774	2.2
Black	1,939	55,556	3.5	1,132	28,070	4.0	807	27,484	2.9
Other	371	17,118	2.2	219	8,828	2.5	152	8,290	1.8
NYS Excluding NYC									
All Races‡	4,281	144,879	3.0	2,681	73,984	3.6	1,600	70,893	2.3
White	3,649	124,688	2.9	2,314	63,700	3.6	1,335	60,988	2.2
Black	497	14,711	3.4	296	7,452	4.0	201	7,257	2.8
Other	96	4,418	2.2	51	2,301	2.2	45	2,117	2.1
New York City									
All Races‡	3,828	126,163	3.0	2,326	64,599	3.6	1,502	61,564	2.4
White	2,089	71,870	2.9	1,308	37,084	3.5	781	34,786	2.2
Black	1,442	40,845	3.5	836	20,618	4.1	606	20,227	3.0
Other	275	12,700	2.2	168	6,527	2.6	107	6,173	1.7

‡Total includes unknowns within each category, thus row and column figures may not sum to totals.

Section I - Table 4
1995 Births - New York State Residents
Percent of Live Births With Two Or More Major Malformations
Sex by Race and Residence

Race and Residence	Both Sexes			Males			Females		
	Infants	Total Births	%	Infants	Total Births	%	Infants	Total Births	%
New York State									
All Races‡	2,375	271,042	0.9	1,377	138,583	1.0	998	132,457	0.8
White	1,700	196,558	0.9	989	100,784	1.0	711	95,774	0.7
Black	524	55,556	0.9	297	28,070	1.1	227	27,484	0.8
Other	124	17,118	0.7	76	8,828	0.9	48	8,290	0.6
NYS Excluding NYC									
All Races‡	1,312	144,879	0.9	788	73,984	1.1	524	70,893	0.7
White	1,119	124,688	0.9	671	63,700	1.1	448	60,988	0.7
Black	148	14,711	1.0	90	7,452	1.2	58	7,257	0.8
Other	27	4,418	0.6	19	2,301	0.8	8	2,117	0.4
New York City									
All Races‡	1,063	126,163	0.8	589	64,599	0.9	474	61,564	0.8
White	581	71,870	0.8	318	37,084	0.9	263	34,786	0.8
Black	376	40,845	0.9	207	20,618	1.0	169	20,227	0.8
Other	97	12,700	0.8	57	6,527	0.9	40	6,173	0.6

‡Total includes unknowns within each category, thus row and column figures may not sum to totals.

Section I - Table 5
1995 Births - New York State Residents
Age at Earliest Diagnosis in Children With Major Malformations
Race and Residence

Age at Diagnosis and Residence	All Races		White		Black		Other	
	Number	%	Number	%	Number	%	Number	%
New York State								
All Ages‡	10,484	100.0	7,438	100.0	2,463	100.0	495	100.0
Less than 3 days	7,966	76.0	5,692	76.5	1,844	74.9	361	72.9
3 to 29 days	1,073	10.2	773	10.4	267	10.8	70	14.1
30 to 182 days	888	8.5	629	8.5	212	8.6	39	7.9
6 months to 2 yrs.	557	5.3	384	5.2	140	5.7	25	5.1
NYS Excluding NYC								
All Ages‡	5,593	100.0	4,768	100.0	645	100.0	123	100.0
Less than 3 days	4,296	76.8	3,666	76.9	486	75.3	97	78.9
3 to 29 days	504	9.0	429	9.0	63	9.8	11	8.9
30 to 182 days	493	8.8	415	8.7	64	9.9	9	7.3
6 months to 2 yrs.	300	5.4	258	5.4	32	5.0	6	4.9
New York City								
All Ages‡	4,891	100.0	2,670	100.0	1,818	100.0	372	100.0
Less than 3 days	3,670	75.0	2,026	75.9	1,358	74.7	264	71.0
3 to 29 days	569	11.6	304	11.4	204	11.2	59	15.9
30 to 182 days	395	8.1	214	8.0	148	8.1	30	8.1
6 months to 2 yrs.	257	5.3	126	4.7	108	5.9	19	5.1

‡Total includes unknowns within each category, thus row and column figures may not sum to totals.

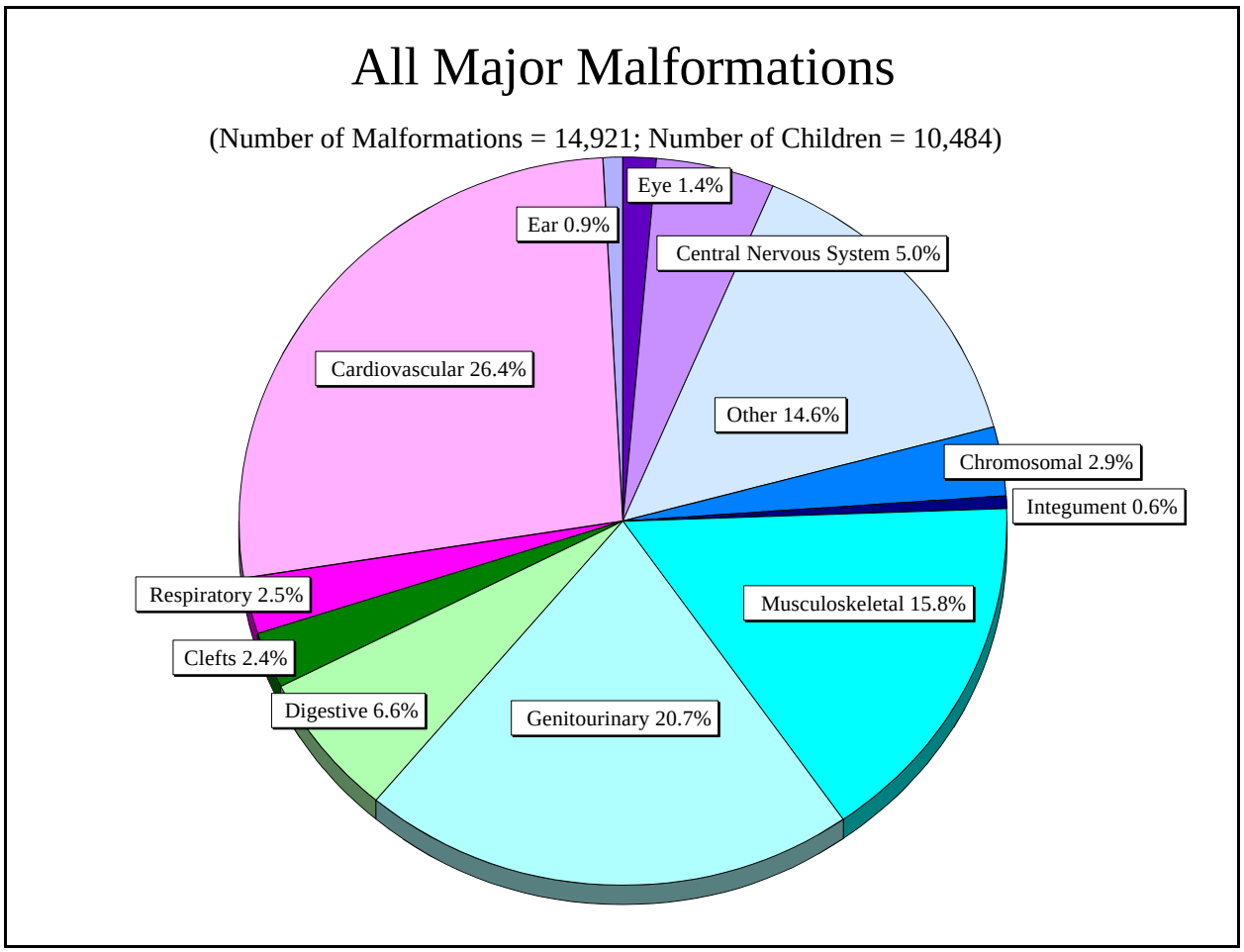
NOTES

Section II
Major Congenital Malformations by Organ System

Introduction to Figures

The organ system figures in this section present the distribution of 12 categories of major malformations, the relative contribution of each category to overall incidence of major malformations in New York State, and the contribution of type of malformation within each subset category. Some of these percentages may differ from previous reports because of the new malformation coding system described in the Program Overview.

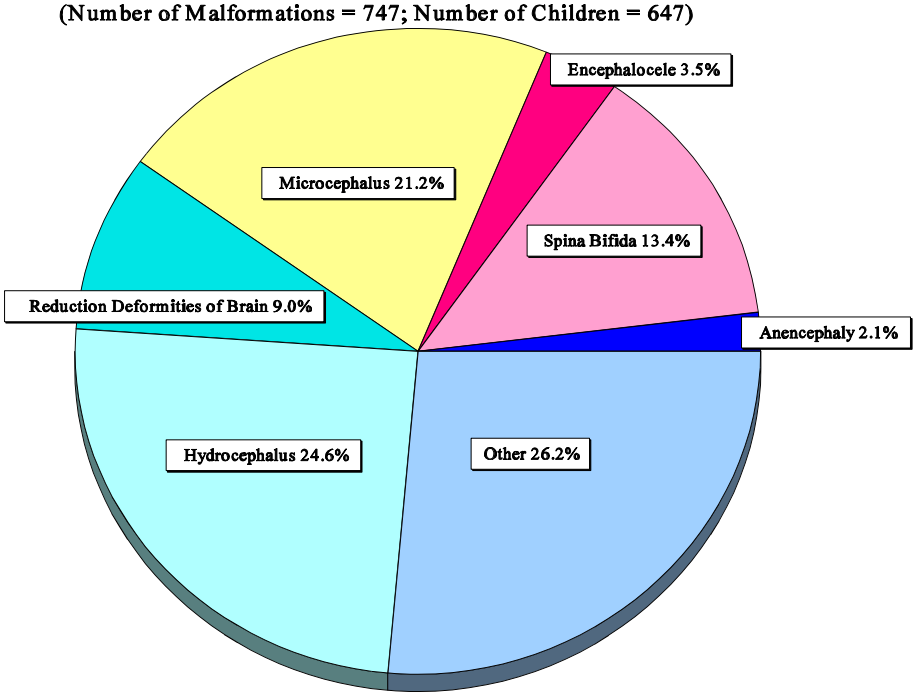
Major Malformations by Organ System
1995 Births - New York State Residents



Major Malformations by Organ System
1995 Births - New York State Residents

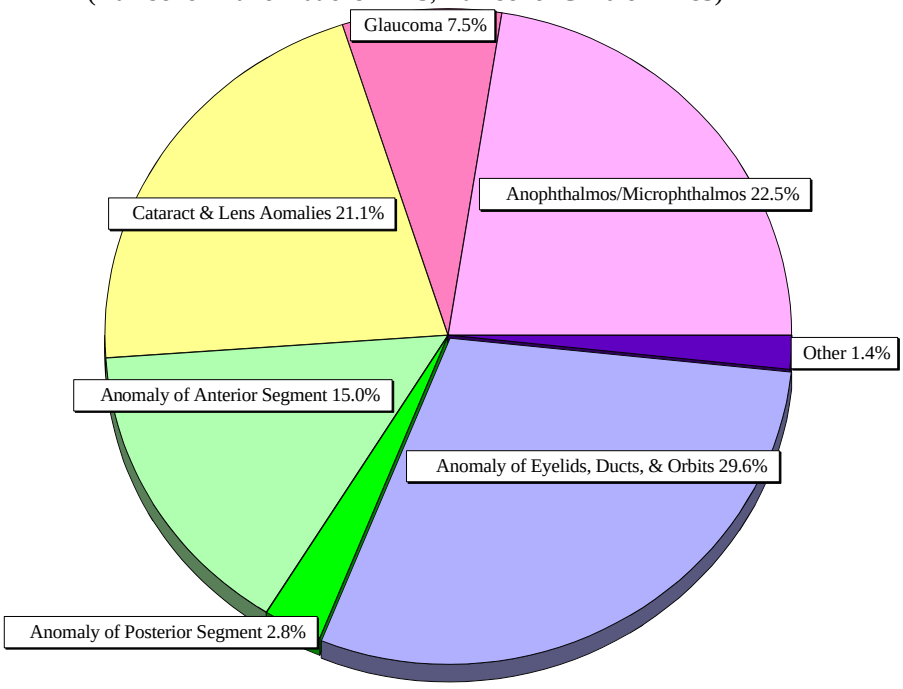
Central Nervous System Subset Category

(Number of Malformations = 747; Number of Children = 647)

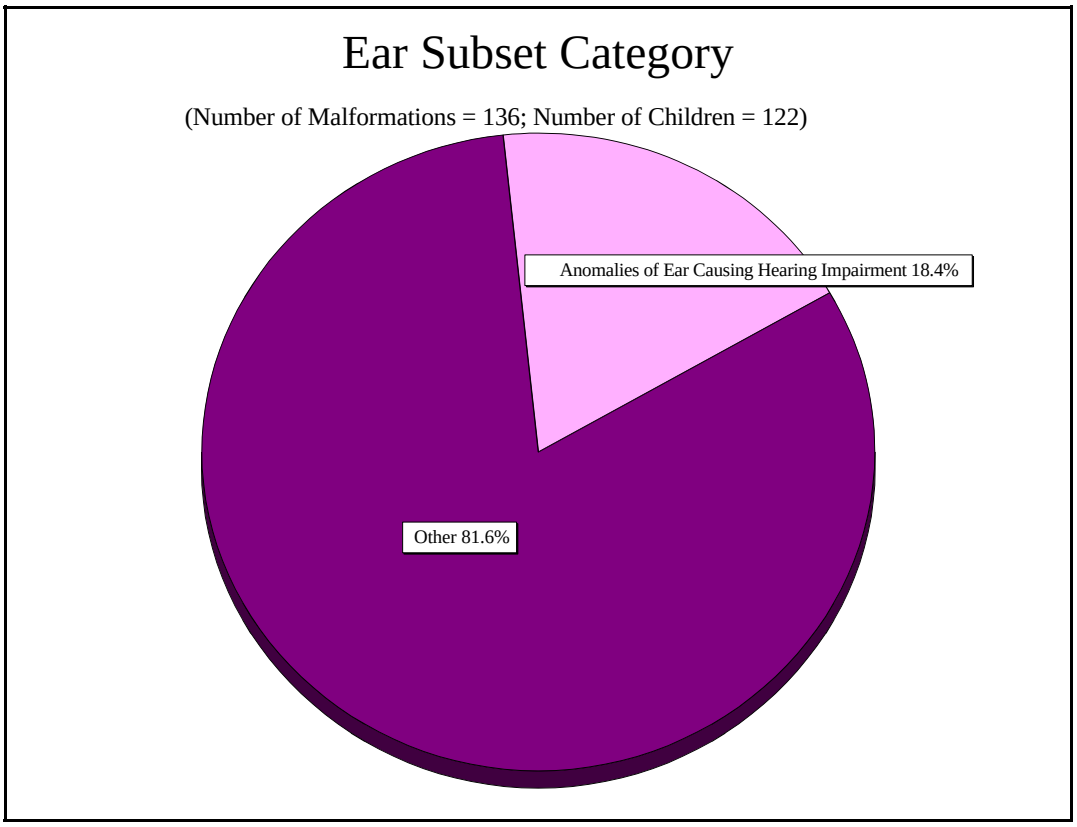


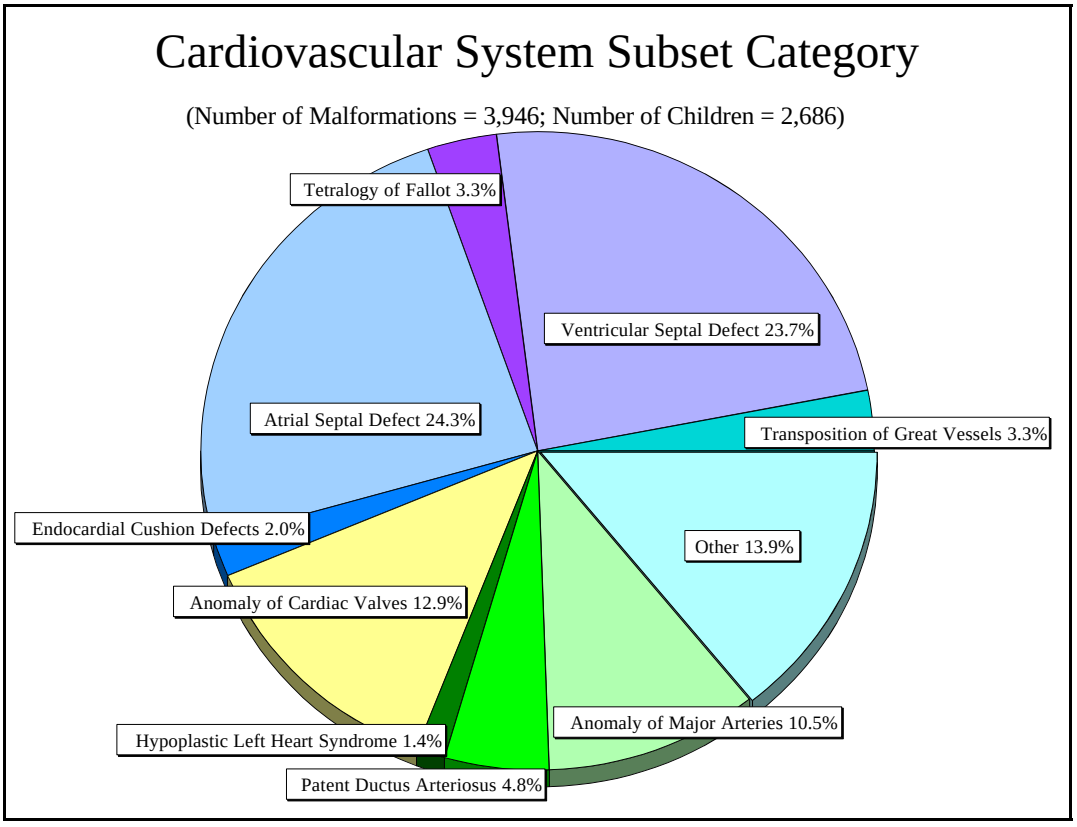
Eye Subset Category

(Number of Malformations = 213; Number of Children = 185)



Major Malformations by Organ System
1995 Births - New York State Residents

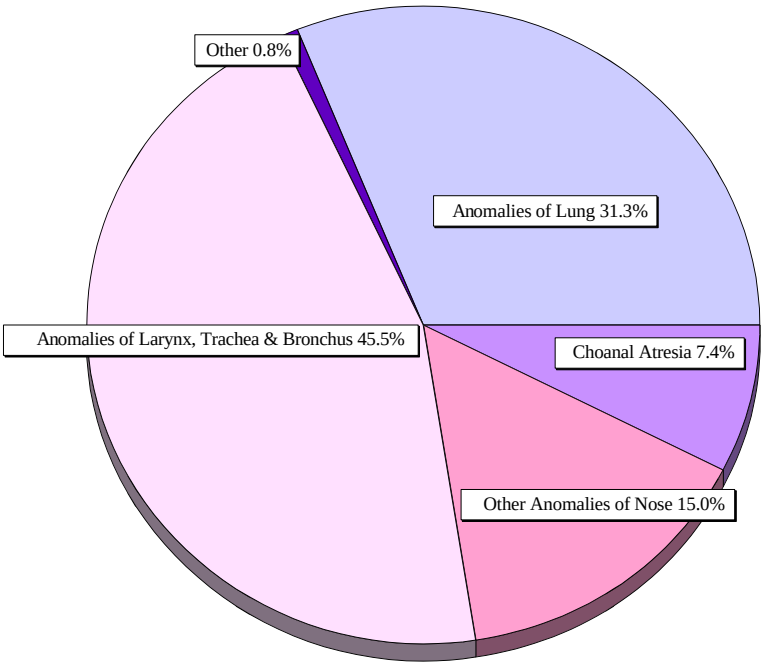




Major Malformations by Organ System
1995 Births - New York State Residents

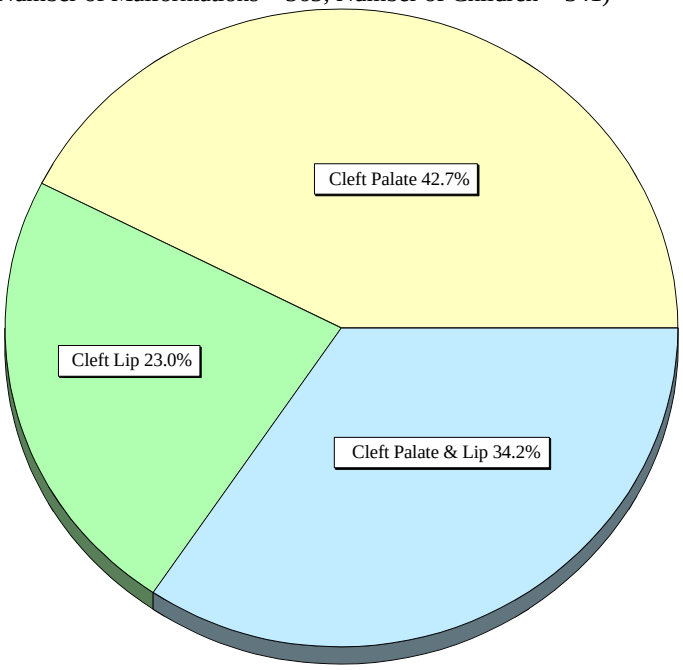
Respiratory System Subset Category

(Number of Malformations = 380; Number of Children = 362)

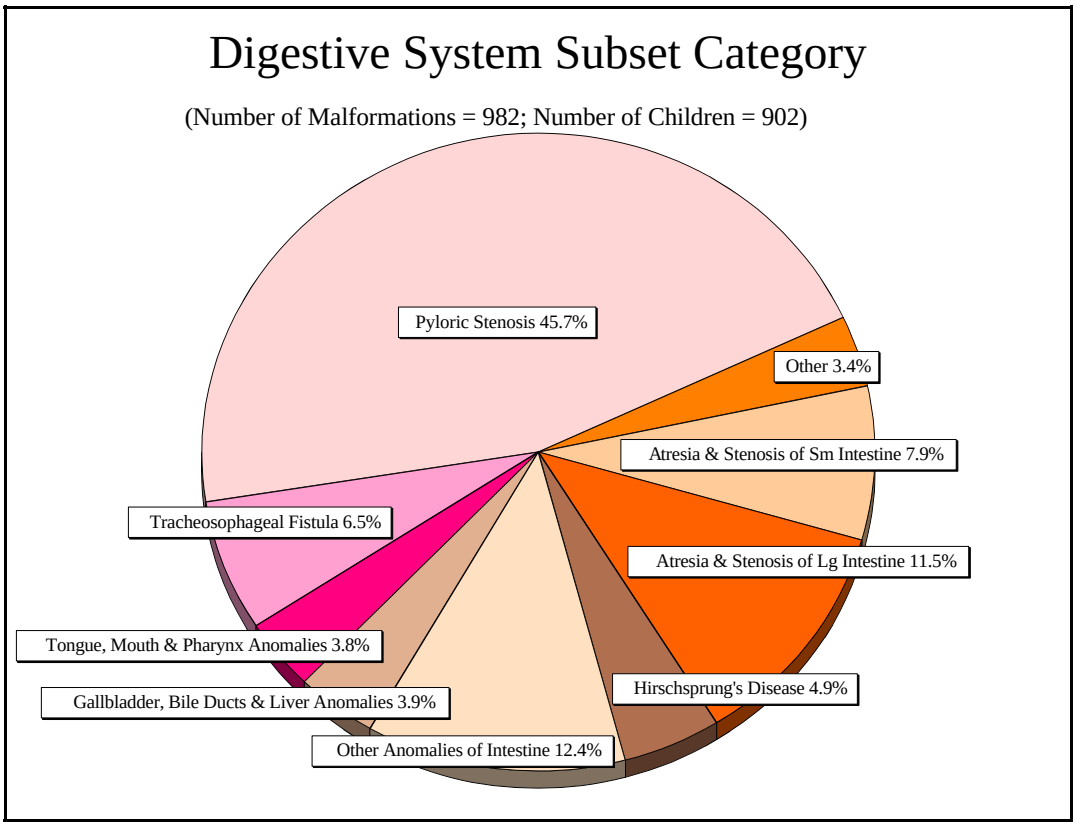


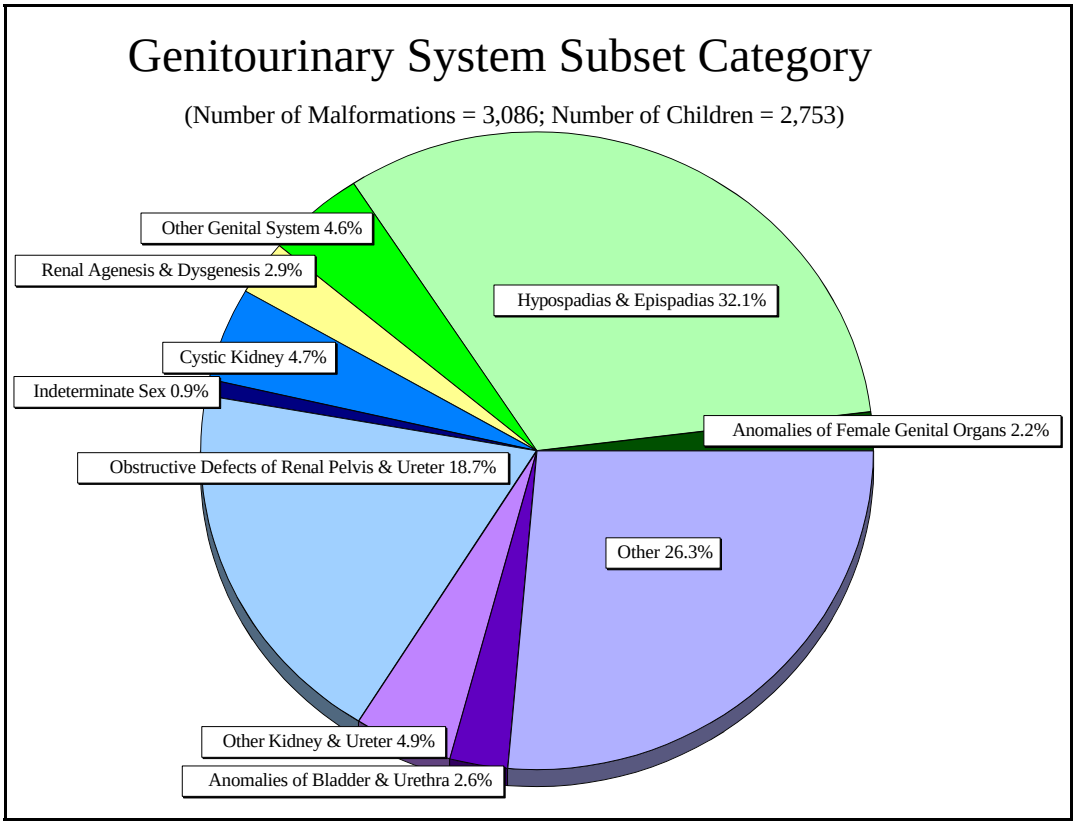
Oral Clefts Subset Category

(Number of Malformations = 365; Number of Children = 341)



Major Malformations by Organ System
1995 Births - New York State Residents

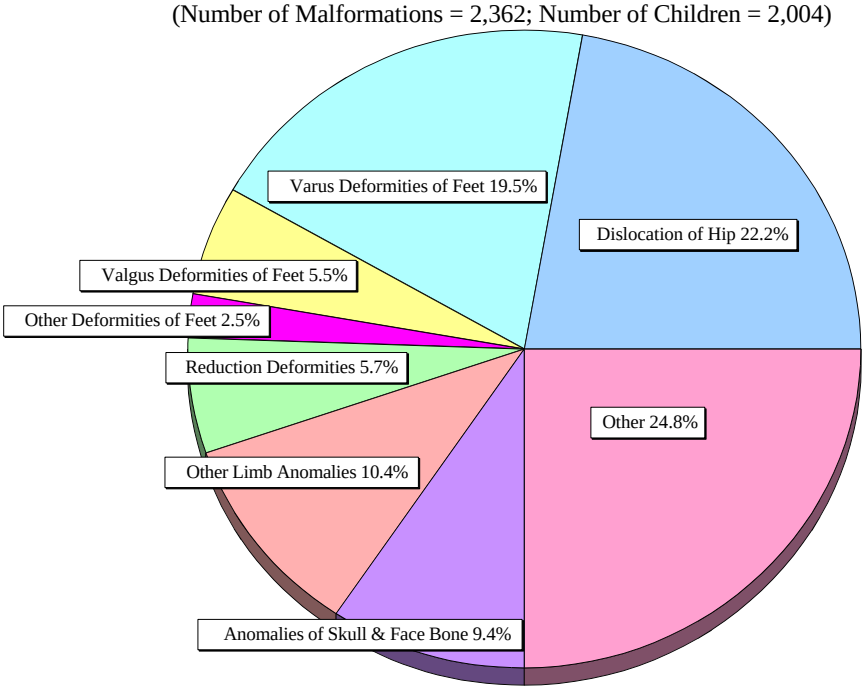




Major Malformations by Organ System
1995 Births - New York State Residents

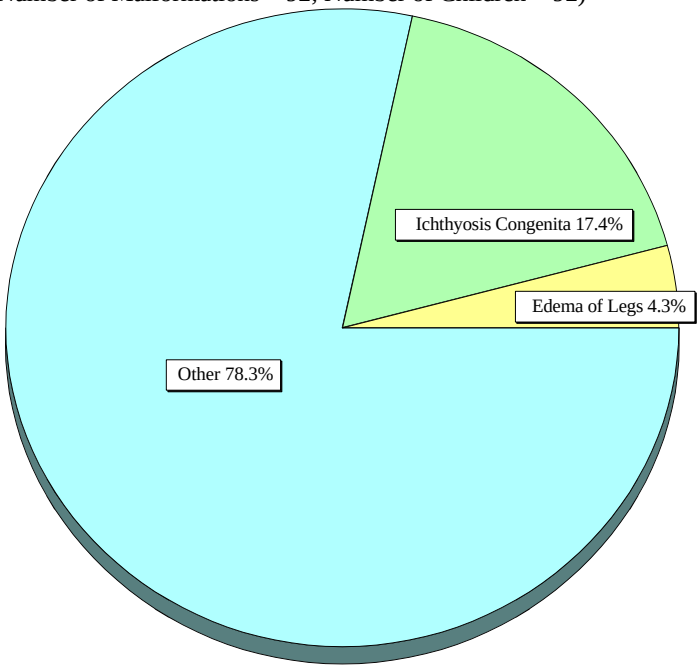
Musculoskeletal System Subset Category

(Number of Malformations = 2,362; Number of Children = 2,004)

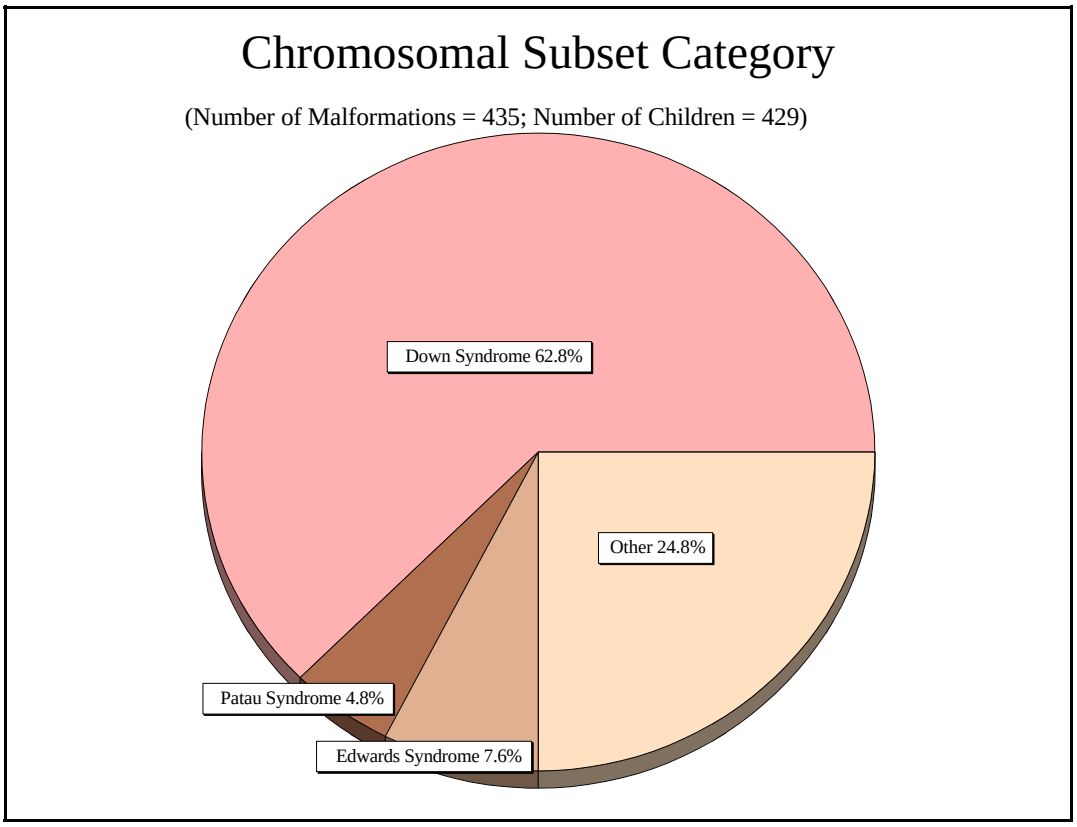


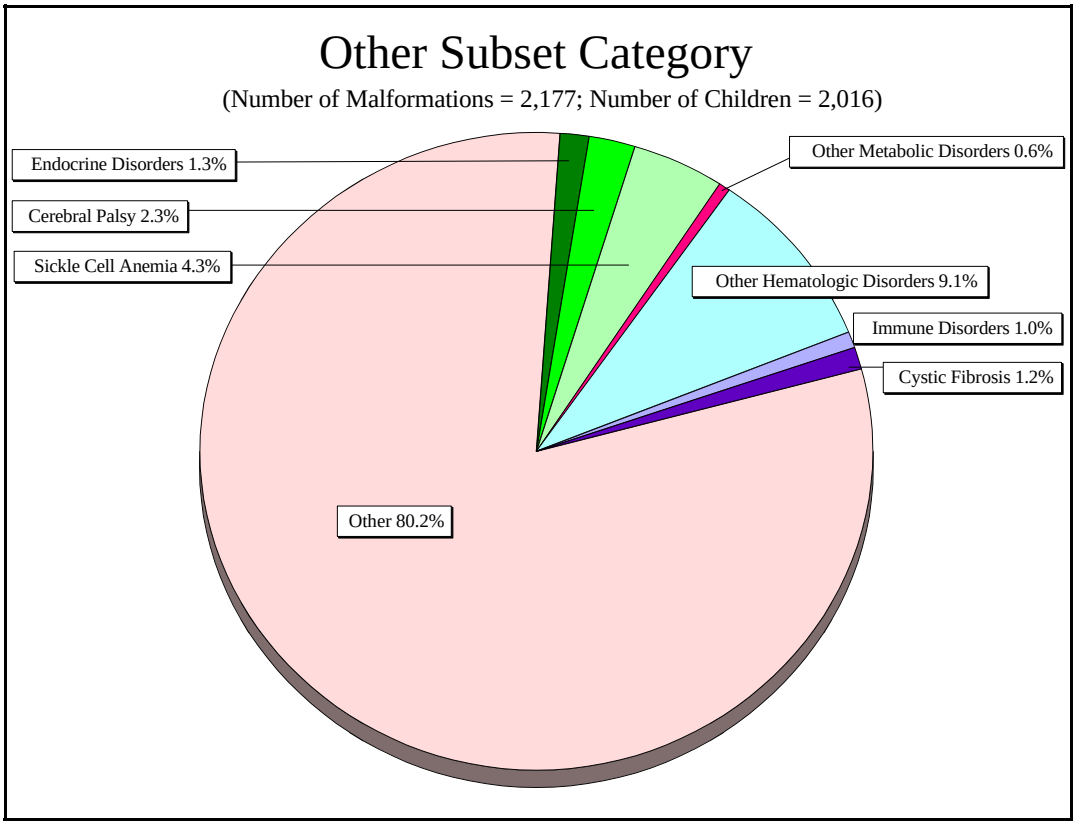
Integument Subset Category

(Number of Malformations = 92; Number of Children = 92)



Major Malformations by Organ System
1995 Births - New York State Residents





NOTES

Section III
Prevalence of Selected Malformations
Rates by Sex and Race

Introduction to Tables

The malformations presented in this section were selected because of the frequency with which they were reported and/or their clinical significance. Rates are per 10,000 live births. The sex ratio is calculated by dividing the rate in males by the rate in females. The malformation rates presented in this report may not be comparable to earlier reports. Previous reports from 1989 to 1991 did not use birth certificate matched cases, thus, the race and birthweight from the birth certificate were not available. Birthweight data are useful to calculate the rate of some malformations such as patent ductus arteriosus. In some cases, these conditions can result from being preterm rather than actually having a malformation. Racial data in this report also may not be comparable because race is defined by maternal race from the birth certificate. In the earlier reports, race was defined by what was reported on the CMR form, which may differ from what is recorded on the birth certificate.

Fluctuations in specific malformation prevalence should be interpreted with caution, especially differences in the "other" race category since the numbers in this group are small. In addition, several malformations were added in 1992 as a result of the change to the BPA code. Previously, these could not be distinguished using the ICD-9 codes. However, since ICD-9 codes are more familiar to most vendors, the ICD-9 code is given on the table with the named malformation. See Appendix 4 for further information on the BPA codes.

Section III
Children with Selected Major Malformations
Prevalences per 10,000 Live Births by Sex & Race

1995 Births - New York State Residents

ICD-9 Code	Malformations	Total Number	Total Prevalence	Male	Female	Ratio (M/F)	White	Black	Other
090	Congenital syphilis	619	22.8	23.2	22.5	1.0	10.3	71.3	11.7
243	Congenital hypothyroidism	81	3.0	3.2	2.8	1.1	2.4	4.9	3.5
270.1	Phenylketonuria	3	0.1	0.1	0.2	0.5	0.2	0.0	0.0
277.0	Cystic fibrosis	26	1.0	1.2	0.8	1.5	1.2	0.4	0.0
282.6	Sickle-cell anemia	94	3.5	3.5	3.4	1.0	0.4	15.7	0.0
658.8	Amniotic bands	8	0.3	0.1	0.5	0.3	0.4	0.2	0.0
740.0	Anencephalus	16	0.6	0.5	0.7	0.7	0.7	0.2	1.2
741.0	Spina bifida with hydrocephalus	52	1.9	1.8	2.0	0.9	1.7	2.7	2.3
741.9	Spina bifida without hydrocephalus	48	1.8	1.7	1.9	0.9	1.7	1.3	4.1
742.0	Encephalocele	26	1.0	0.8	1.1	0.7	0.8	1.1	2.3
742.1	Microcephalus	158	5.8	5.1	6.6	0.8	4.3	11.5	5.3
742.2	Agyria & lissencephaly	5	0.2	0.2	0.2	1.4	0.2	0.2	0.0
742.2	Anomalies of corpus callosum	42	1.5	1.9	1.1	1.7	1.5	2.0	1.2
742.2	Holoprosencephaly	10	0.4	0.4	0.3	1.4	0.4	0.2	1.2
742.3	Congenital hydrocephalus	184	6.8	7.6	5.9	1.3	6.5	7.7	7.6
742.4	Porencephaly	12	0.4	0.5	0.4	1.3	0.5	0.5	0.0
742.5	Congenital tethered cord	26	1.0	0.6	1.4	0.4	1.2	0.4	0.0
743.0	Anophthalmos	12	0.4	0.5	0.4	1.3	0.4	0.5	0.6
743.1	Microphthalmos	36	1.3	1.4	1.2	1.2	1.2	1.4	2.3
743.2	Glaucoma	16	0.6	0.7	0.5	1.6	0.5	0.9	0.6
743.3	Absence of lens	3	0.1	0.1	0.1	1.9	0.2	0.0	0.0
743.3	Congenital cataract	41	1.5	1.6	1.4	1.1	1.6	1.6	0.6
743.45	Aniridia	2	0.1	0.1	0.0	-.-	0.1	0.0	0.0
743.46	Coloboma of iris	5	0.2	0.3	0.1	3.8	0.3	0.0	0.0
744.0	Anotia/microtia	31	1.1	1.2	1.1	1.2	1.3	0.5	1.2
745.0	Common truncus	20	0.7	0.8	0.7	1.2	0.7	1.1	0.6
745.1	Transposition of great vessels	130	4.8	5.7	3.9	1.5	4.7	5.2	5.3
745.2	Tetralogy of Fallot	129	4.8	4.9	4.6	1.1	4.6	5.6	4.7
745.3	Common ventricle	28	1.0	1.6	0.5	3.5	0.9	1.6	0.6
745.4	Ventricular septal defect	936	34.5	33.2	35.9	0.9	36.3	29.7	33.9
745.5	Ostium secundum type atrial septal def.	959	35.4	34.3	36.5	0.9	32.9	47.0	30.4
745.6	Endocardial cushion defects	77	2.8	2.7	2.9	0.9	3.2	1.8	2.3
746.0	Atresia/stenosis of pulmonary valve	290	10.7	11.2	10.2	1.1	9.4	16	9.9
746.1	Tricuspid atresia/stenosis/hypoplasia	40	1.5	1.9	1.1	1.8	1.4	2.0	0.6
746.2	Ebstein's anomaly	11	0.4	0.4	0.4	1.1	0.5	0.2	0.0
746.3	Congenital stenosis of aortic valve	43	1.6	2.3	0.8	2.8	1.9	0.9	0.0
746.7	Hypoplastic left heart syndrome	55	2.0	2.2	1.8	1.2	2.0	2.2	1.8
746.85	Anomalies of coronary artery	21	0.8	0.9	0.7	1.3	0.7	1.3	0.6
747.0	Patent ductus arteriosis	188	6.9	6.1	7.9	0.8	6.4	8.1	10.5
747.1	Coartation of aorta	110	4.1	5.1	2.9	1.7	4.6	2.7	2.3
747.41	Total anomalous pulmonary venus connect.	17	0.6	0.6	0.6	1.1	0.8	0.4	0.0
748.0	Choanal atresia	28	1.0	1.2	0.8	1.5	1.2	0.5	0.6
748.5	Agenesis/hypoplasia of lung	98	3.6	3.9	3.3	1.2	3.3	4.7	4.7
749.0	Cleft palate	156	5.8	4.8	6.8	0.7	6.2	4.5	5.3

Section III
Children with Selected Major Malformations
Prevalences per 10,000 Live Births by Sex & Race

1995 Births - New York State Residents

ICD-9 Code	Malformation	Total Number	Total Prevalence	Male	Female	Ratio (M/F)	White	Black	Other
749.1	Cleft lip	84	3.1	4.0	2.2	1.8	3.6	1.6	2.9
749.2	Cleft palate & lip	125	4.6	5.3	3.9	1.4	5.1	3.2	3.5
750.3	Tracheoesophageal fistula etc.	64	2.4	2.9	1.8	1.6	2.9	0.9	1.2
750.5	Congenital hypertrophic pyloric stenosis	449	16.6	26.8	5.9	4.5	20.3	7.4	5.3
751.1	Atresia and stenosis of small intestine	78	2.9	2.8	2.9	1.0	2.4	4.5	2.9
751.2	Atresia and stenosis of rectum or anus	113	4.2	4.2	4.2	1.0	4.1	4.1	5.8
751.3	Hirschsprungs disease	48	1.8	2.3	1.2	1.9	1.7	1.6	2.9
751.4	Anomalies of intestinal fixation	50	1.8	2.2	1.4	1.6	1.6	1.4	5.8
751.61	Biliary atresia	18	0.7	0.7	0.6	1.2	0.6	0.9	0.6
752.6	Epispadias	28	1.0	2.0	0.0	-.-	0.7	2.2	1.2
752.6	Hypospadias	886	32.7	63.8	0.2	422.5	35.6	27	21
753.0	Renal agenesis and dysgenesis	89	3.3	3.9	2.6	1.5	3.9	1.8	1.8
753.1	Cystic kidney disease	146	5.4	6.9	3.9	1.8	5.3	6.3	4.1
753.2	Obstructive defect renal pelvis & ureter	577	21.3	31.5	10.6	3.0	24	12.1	22.8
753.5	Extrophy of urinary bladder	5	0.2	0.2	0.2	1.4	0.2	0.4	0.0
753.6	Atresia & stenosis of urethra & bladder	38	1.4	2.6	0.2	17.2	1.1	2.2	2.9
754.3	Congenital dislocation of hip	395	14.6	7.1	22.3	0.3	16.9	6.1	16.4
754.51	Talipes equinovarus	315	11.6	14.6	8.5	1.7	11.8	11.5	11.1
755.2	Reduction deformities of upper limb	81	3.0	2.8	3.2	0.9	3.7	1.1	1.2
755.3	Reduction deformities of lower limb	51	1.9	2.1	1.7	1.3	1.7	2.3	2.3
755.8	Arthrogryposis multiplex congenita	42	1.5	1.4	1.7	0.9	1.4	2.5	0.6
756.0	Craniosynostosis	109	4.0	5.4	2.6	2.1	4.6	1.8	4.7
756.0	Goldenhar syndrome	11	0.4	0.2	0.6	0.4	0.5	0.4	0.0
756.4	Chonodrodystrophy	31	1.1	1.2	1.1	1.0	1.2	0.7	2.3
756.51	Osteogenesis imperfecta	7	0.3	0.1	0.4	0.4	0.3	0.2	0.0
756.6	Diaphragmatic hernia	54	2.0	2.4	1.6	1.5	1.8	2.0	4.1
756.7	Gastroschisis	33	1.2	1.3	1.1	1.1	1.5	0.5	0.6
756.7	Omphalocele	33	1.2	1.3	1.1	1.1	1.2	1.1	1.8
756.7	Prune belly	4	0.1	0.3	0.0	-.-	0.2	0.0	0.0
758.0	Down syndrome	273	10.1	10.2	9.9	1.0	10.3	8.8	12.3
758.1	Patau syndrome	21	0.8	0.9	0.7	1.3	0.9	0.5	0.6
758.2	Edwards syndrome	33	1.2	0.8	1.7	0.5	1.0	1.8	1.8
758.6	Gonadal dysgenesis	20	0.7	0.3	1.2	0.2	0.8	0.4	1.8
758.7	Klinefelter syndrome	15	0.6	1.1	0.0	-.-	0.6	0.5	0.6
759.3	Situs inversus	15	0.6	0.4	0.8	0.5	0.6	0.5	0.6
760.71	Fetal alcohol syndrome	56	2.1	2.4	1.7	1.4	1.1	6.1	0.6
771.0	Congenital rubella	2	0.1	0.1	0.1	1.0	0.1	0.2	0.0
771.1	Congenital cytomegalovirus infection	37	1.4	1.4	1.3	1.1	1.1	2.3	1.8
771.2	Other congenital infections	61	2.3	2.0	2.5	0.8	1.7	4.9	0.0

NOTES

Section IV
Most Frequently Reported Selected Major Malformations by County

Introduction to Tables

Congenital Malformations Registry data were tabulated by county of residence at the time of diagnosis and four digit ICD-9-CM codes for major malformations. Certain codes for rare disorders and nonspecific codes are not included. The table on this page presents the number of children with major malformations by county, and the total number of live births for comparison.

For each county, the 10 most frequently reported codes are listed, except those instances in which the tenth and subsequent codes were equal in number. In this circumstance, the additional codes of equal number are listed. Some counties may have fewer than 10 codes reported. Children reported with more than one malformation may be represented more than once in these tables. These are presented on the following pages.

These county listings are not designed to be used for comparisons among counties or for analytic studies. They are most useful to assist in county planning, education, counseling and other health care service programs.

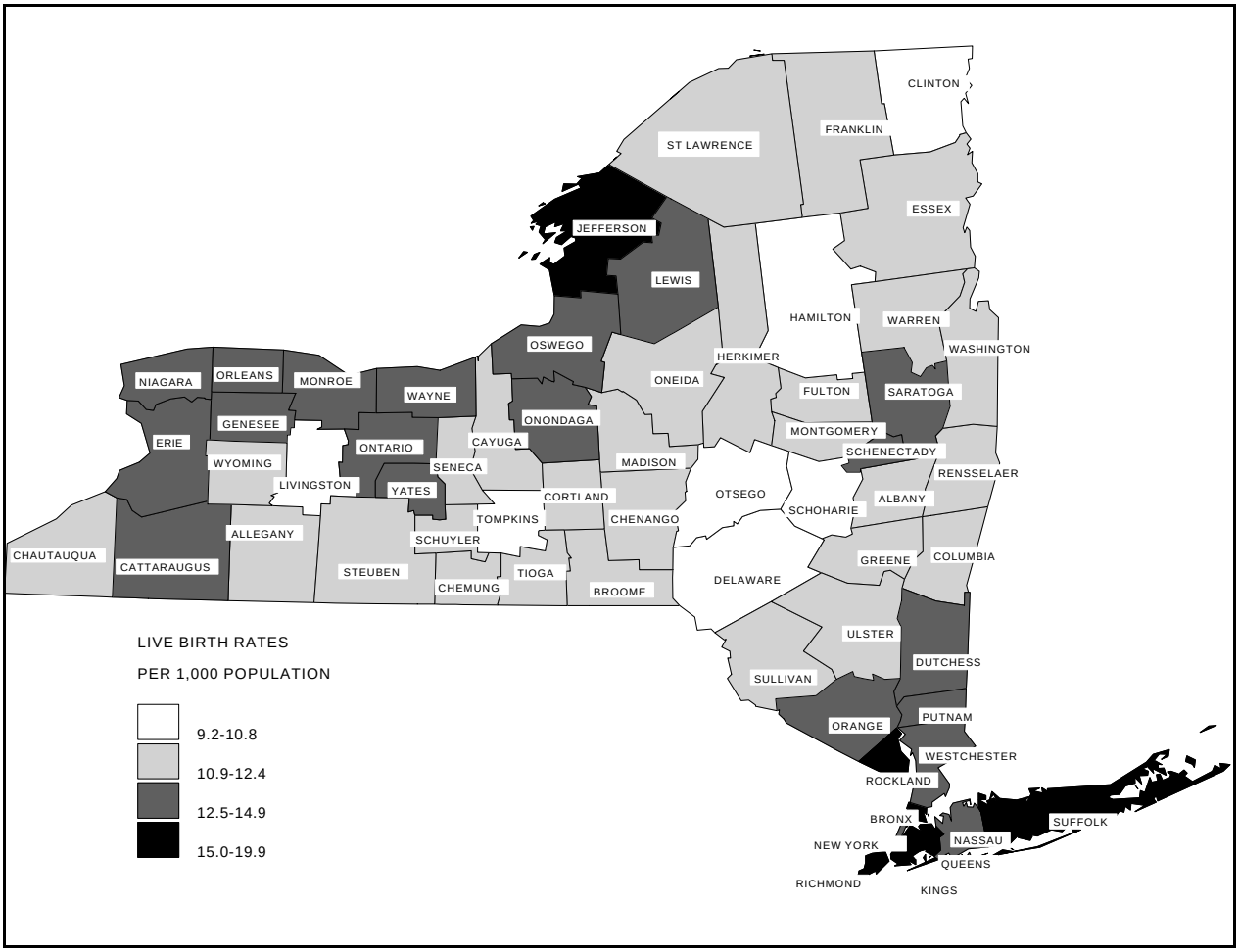
The map of New York State showing county live birth rates is part of the Bureau of Biometrics annual report, "Vital Statistics of New York State 1995." The Bureau of Biometrics supplies the CMR with the number of live births in a specific year, which is used as a denominator in the calculation of prevalences of malformations.

For information about vital statistics, contact:
New York State Department of Health
Bureau of Biometrics
ESP Concourse - C144
Albany, NY 12237-0044.

Children with Major Congenital Malformations and Births by County, 1995

County	Number of Children	Number of Live Births	Percent of Live Births
Albany	130	3,530	3.7
Allegany	16	594	2.7
Bronx	983	24,406	4.0
Broome	102	2,501	4.1
Cattaraugus	45	1,062	4.2
Cayuga	27	1,008	2.7
Chautauqua	62	1,659	3.7
Chemung	51	1,146	4.5
Chenango	23	617	3.7
Clinton	28	951	2.9
Columbia	15	731	2.1
Cortland	16	631	2.5
Delaware	15	466	3.2
Dutchess	108	3,451	3.1
Erie	629	12,364	5.1
Essex	7	429	1.6
Franklin	16	581	2.8
Fulton	40	654	6.1
Genesee	33	782	4.2
Greene	11	518	2.1
Hamilton	3	54	5.6
Herkimer	37	754	4.9
Jefferson	51	1,882	2.7
Kings	1,663	42,131	3.9
Lewis	13	367	3.5
Livingston	23	706	3.3
Madison	25	885	2.8
Monroe	290	10,010	2.9
Montgomery	27	614	4.4
Nassau	869	18,084	4.8
New York	786	20,611	3.8
Niagara	128	2,807	4.6
Oneida	110	2,881	3.8
Onondaga	232	6,478	3.6
Ontario	50	1,293	3.9
Orange	154	4,914	3.1
Orleans	26	575	4.5
Oswego	55	1,614	3.4
Otsego	25	645	3.9
Putnam	27	1,218	2.2
Queens	1,211	32,973	3.7
Rensselaer	81	1,956	4.1
Richmond	248	6,042	4.1
Rockland	130	4,168	3.1
St. Lawrence	48	1,266	3.8
Saratoga	99	2,561	3.9
Schenectady	84	1,962	4.3
Schoharie	4	334	1.2
Schuyler	8	237	3.4
Seneca	17	392	4.3
Steuben	48	1,257	3.8
Suffolk	813	20,302	4.0
Sullivan	34	870	3.9
Tioga	19	632	3.0
Tompkins	31	922	3.4
Ulster	81	2,085	3.9
Warren	27	724	3.7
Washington	25	726	3.4
Wayne	33	1,248	2.6
Westchester	458	12,980	3.5
Wyoming	23	488	4.7
Yates	11	313	3.5

Live Birth Rates, New York State 1995



Section IV
Ten Most Frequently Reported Major Malformations
By County, 1995
Congenital Malformations Registry

County	ICD-9 Code	Description	Number Reported
Albany			
	745.4	Ventricular septal defect	19
	745.5	Ostium secundum atrial septal defect	15
	752.6	Hypospadias & epispadias	13
	752.5	Undescended testicle	9
	750.5	Congenital hypertrophic pyloric stenosis	7
	753.2	Obstructive defects of renal pelvis & ureter	7
	753.1	Cystic kidney disease	5
	754.3	Congenital dislocation of hip	5
	758.0	Down syndrome	5
	746.0	Anomalies of pulmonary valve	4
	747.0	Patent ductus arteriosus	4
	754.5	Varus deformities of feet	4
	756.0	Anomalies of skull and face bones	4
Allegany			
	750.5	Congenital hypertrophic pyloric stenosis	4
	745.5	Ostium secundum atrial septal defect	3
	752.5	Undescended testicle	2
	286.0	Congenital factor VIII disorder	1
	746.7	Hypoplastic left heart syndrome	1
	746.8	Other specified anomalies of heart	1
	747.0	Patent ductus arteriosus	1
	747.3	Anomalies of pulmonary artery	1
	748.2	Web of larynx	1
	752.6	Hypospadias & epispadias	1
	752.8	Other specified anomalies of genital organs	1
	753.0	Renal agenesis & dysgenesis	1
	753.1	Cystic kidney disease	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	755.2	Reduction deformities of upper limb	1
	756.0	Anomalies of skull and face bones	1
	756.6	Anomalies of diaphragm	1
Bronx			
	752.6	Hypospadias & epispadias	76
	745.4	Ventricular septal defect	71
	752.5	Undescended testicle	57
	745.5	Ostium secundum atrial septal defect	50
	753.2	Obstructive defects of renal pelvis & ureter	50
	754.5	Varus deformities of feet	50
	754.3	Congenital dislocation of hip	36
	750.5	Congenital hypertrophic pyloric stenosis	35
	746.0	Anomalies of pulmonary valve	30
	742.1	Microcephalus	29
Broome			
	745.5	Ostium secundum atrial septal defect	28
	745.4	Ventricular septal defect	13
	750.5	Congenital hypertrophic pyloric stenosis	8
	752.5	Undescended testicle	7
	753.2	Obstructive defects of renal pelvis & ureter	7
	752.6	Hypospadias & epispadias	6
	754.3	Congenital dislocation of hip	5
	758.0	Down syndrome	5
	746.8	Other specified anomalies of heart	4

Section IV
Ten Most Frequently Reported Major Malformations
By County, 1995
Congenital Malformations Registry

County	ICD-9 Code	Description	Number Reported
Broome (cont.)	745.2	Tetralogy of Fallot	3
	746.6	Congenital mitral insufficiency	3
	747.3	Anomalies of pulmonary artery	3
	748.3	Other anomalies of larynx, trachea, & bronchus	3
	749.2	Cleft palate with cleft lip	3
Cattaraugus			
	745.5	Ostium secundum atrial septal defect	6
	746.8	Other specified anomalies of heart	5
	752.5	Undescended testicle	4
	745.4	Ventricular septal defect	3
	746.0	Anomalies of pulmonary valve	3
	749.0	Cleft palate	3
	750.5	Congenital hypertrophic pyloric stenosis	3
	752.6	Hypospadias & epispadias	3
	753.2	Obstructive defects of renal pelvis & ureter	3
	758.0	Down syndrome	3
Cayuga			
	742.1	Microcephalus	4
	753.2	Obstructive defects of renal pelvis & ureter	4
	756.0	Anomalies of skull and face bones	3
	742.3	Congenital hydrocephalus	2
	742.4	Other specified anomalies of brain	2
	745.4	Ventricular septal defect	2
	746.8	Other specified anomalies of heart	2
	748.3	Other anomalies of larynx, trachea, & bronchus	2
	749.2	Cleft palate with cleft lip	2
	752.5	Undescended testicle	2
	752.6	Hypospadias & epispadias	2
	754.5	Varus deformities of feet	2
	756.7	Anomalies of abdominal wall	2
Chautauqua			
	752.5	Undescended testicle	9
	750.5	Congenital hypertrophic pyloric stenosis	7
	745.5	Ostium secundum atrial septal defect	6
	746.8	Other specified anomalies of heart	6
	754.3	Congenital dislocation of hip	6
	754.5	Varus deformities of feet	4
	742.3	Congenital hydrocephalus	3
	745.4	Ventricular septal defect	3
	746.0	Anomalies of pulmonary valve	3
	747.3	Anomalies of pulmonary artery	3
	752.6	Hypospadias & epispadias	3
	755.6	Other anomalies of lower limb including pelvic girdle	3
Chemung			
	745.4	Ventricular septal defect	8
	752.6	Hypospadias & epispadias	6
	750.5	Congenital hypertrophic pyloric stenosis	5
	752.5	Undescended testicle	5
	746.8	Other specified anomalies of heart	3
	747.3	Anomalies of pulmonary artery	3
	754.5	Varus deformities of feet	3
	753.2	Obstructive defects of renal pelvis & ureter	2
	754.3	Congenital dislocation of hip	2

Section IV
Ten Most Frequently Reported Major Malformations
By County, 1995
Congenital Malformations Registry

County	ICD-9 Code	Description	Number Reported
Chemung (cont.)	758.8	Other conditions due to sex chromosome anomalies	2
Chenango			
	745.5	Ostium secundum atrial septal defect	3
	754.3	Congenital dislocation of hip	3
	745.4	Ventricular septal defect	2
	746.3	Congenital stenosis of aortic arch	2
	752.6	Hypospadias & epispadias	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	754.5	Varus deformities of feet	2
	228.1	Lymphangioma, any site	1
	741.0	Spina bifida with hydrocephalus	1
	741.9	Spina bifida w/o hydrocephalus	1
	742.1	Microcephalus	1
	742.3	Congenital hydrocephalus	1
	742.5	Other specified anomalies of spinal cord	1
	746.0	Anomalies of pulmonary valve	1
	746.1	Tricuspid atresia & stenosis	1
	746.2	Ebstein's anomaly	1
	747.2	Other anomalies of aorta	1
	747.3	Anomalies of pulmonary artery	1
	748.3	Other anomalies of larynx, trachea, & bronchus	1
	749.0	Cleft palate	1
	749.2	Cleft palate with cleft lip	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	751.1	Atresia and stenosis of small intestine	1
	751.2	Atresia and stenosis of large intestine, rectum, & anal canal	1
	751.7	Anomalies of pancreas	1
	752.8	Other specified anomalies of genital organs	1
	753.1	Cystic kidney disease	1
	755.6	Other anomalies of lower limb including pelvic girdle	1
	757.3	Other specified anomalies of skin	1
	758.0	Down syndrome	1
	759.8	Other specified anomalies	1
Clinton			
	752.6	Hypospadias & epispadias	8
	753.2	Obstructive defects of renal pelvis & ureter	6
	745.4	Ventricular septal defect	5
	754.3	Congenital dislocation of hip	5
	752.8	Other specified anomalies of genital organs	3
	745.5	Ostium secundum atrial septal defect	2
	753.1	Cystic kidney disease	2
	758.0	Down syndrome	2
	747.0	Patent ductus arteriosus	1
	748.5	Agenesis, hypoplasia & dysplasia, lung	1
	755.5	Other anomalies of upper limb including shoulder girdle	1
	756.0	Anomalies of skull and face bones	1
	756.4	Chondrodystrophy	1
	771.1	Congenital cytomegalovirus infection	1
Columbia			
	752.6	Hypospadias & epispadias	2
	742.3	Congenital hydrocephalus	1
	742.4	Other specified anomalies of brain	1
	743.8	Other specified anomalies of eye	1

Section IV
Ten Most Frequently Reported Major Malformations
By County, 1995
Congenital Malformations Registry

County	ICD-9 Code	Description	Number Reported
Columbia (cont.)	745.1	Transposition of great vessels	1
	745.5	Ostium secundum atrial septal defect	1
	747.0	Patent ductus arteriosus	1
	748.3	Other anomalies of larynx, trachea, & bronchus	1
	749.2	Cleft palate with cleft lip	1
	750.2	Other specified anomalies, mouth and pharynx	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	751.1	Atresia and stenosis of small intestine	1
	752.5	Undescended testicle	1
	755.6	Other anomalies of lower limb including pelvic girdle	1
	759.8	Other specified anomalies	1
Cortland			
	745.4	Ventricular septal defect	3
	752.6	Hypospadias & epispadias	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	771.2	Other congenital infections	2
	243.0	Congenital hypothyroidism	1
	275.4	Disorders of calcium metabolism	1
	448.0	Hereditary hemorrhagic telangiectasia	1
	747.0	Patent ductus arteriosus	1
	748.3	Other anomalies of larynx, trachea, & bronchus	1
	749.0	Cleft palate	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	753.4	Other specified anomalies of ureter	1
	753.8	Other specified anomalies of bladder & urethra	1
	757.3	Other specified anomalies of skin	1
	759.2	Anomalies of other endocrine glands	1
Delaware			
	750.5	Congenital hypertrophic pyloric stenosis	2
	752.6	Hypospadias & epispadias	2
	270.7	Disturbances of amino-acid metabolims	1
	275.4	Disorders of calcium metabolism	1
	745.0	Common truncus	1
	745.2	Tetralogy of Fallot	1
	745.4	Ventricular septal defect	1
	746.0	Anomalies of pulmonary valve	1
	746.8	Other specified anomalies of heart	1
	747.0	Patent ductus arteriosus	1
	749.1	Cleft lip	1
	751.3	Hirschprung's disease & other functional disorders of colon	1
	751.6	Anomalies of gallbladder, bile ducts, and liver	1
	752.5	Undescended testicle	1
	753.1	Cystic kidney disease	1
	754.5	Varus deformities of feet	1
	756.0	Anomalies of skull and face bones	1
	759.0	Anomalies of spleen	1
	759.8	Other specified anomalies	1
	771.1	Congenital cytomegalovirus infection	1
Dutchess			
	745.4	Ventricular septal defect	14
	752.6	Hypospadias & epispadias	10
	752.5	Undescended testicle	9
	754.5	Varus deformities of feet	7

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County	ICD-9 Code	Description	Number Reported
Dutchess (cont.)	745.5	Ostium secundum atrial septal defect	6
	747.1	Coarctation of aorta	5
	750.5	Congenital hypertrophic pyloric stenosis	5
	753.2	Obstructive defects of renal pelvis & ureter	5
	754.3	Congenital dislocation of hip	5
	751.1	Atresia and stenosis of small intestine	4
	755.6	Other anomalies of lower limb including pelvic girdle	4
	758.0	Down syndrome	4
	759.8	Other specified anomalies	4
Erie			
	752.5	Undescended testicle	66
	745.4	Ventricular septal defect	56
	752.6	Hypospadias & epispadias	54
	745.5	Ostium secundum atrial septal defect	45
	747.3	Anomalies of pulmonary artery	39
	750.5	Congenital hypertrophic pyloric stenosis	32
	754.5	Varus deformities of feet	25
	746.8	Other specified anomalies of heart	23
	748.3	Other anomalies of larynx, trachea, & bronchus	23
	746.0	Anomalies of pulmonary valve	20
Essex			
	282.0	Hereditary spherocytosis	1
	741.9	Spina bifida w/o hydrocephalus	1
	743.4	Coloboma & other anomalies of anterior segment	1
	745.4	Ventricular septal defect	1
	745.5	Ostium secundum atrial septal defect	1
	746.7	Hypoplastic left heart syndrome	1
	748.0	Choanal atresia	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	752.6	Hypospadias & epispadias	1
	753.2	Obstructive defects of renal pelvis & ureter	1
Franklin			
	745.4	Ventricular septal defect	4
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	1
	751.2	Atresia and stenosis of large intestine, rectum, & anal canal	1
	751.3	Hirschprung's disease & other functional disorders of colon	1
	752.5	Undescended testicle	1
	752.6	Hypospadias & epispadias	1
	754.3	Congenital dislocation of hip	1
	754.5	Varus deformities of feet	1
	756.1	Anomalies of spine	1
	756.8	Other specified anomalies of muscle, tendon, fascia, etc.	1
	757.3	Other specified anomalies of skin	1
	771.1	Congenital cytomegalovirus infection	1
Fulton			
	745.5	Ostium secundum atrial septal defect	7
	746.8	Other specified anomalies of heart	7
	745.4	Ventricular septal defect	5
	750.5	Congenital hypertrophic pyloric stenosis	4
	747.3	Anomalies of pulmonary artery	3
	748.3	Other anomalies of larynx, trachea, & bronchus	3
	754.3	Congenital dislocation of hip	3
	752.5	Undescended testicle	2

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County	ICD-9 Code	Description	Number Reported
Fulton (cont.)	752.6	Hypospadias & epispadias	2
	754.5	Varus deformities of feet	2
Genesee			
	745.5	Ostium secundum atrial septal defect	6
	750.5	Congenital hypertrophic pyloric stenosis	5
	752.6	Hypospadias & epispadias	5
	745.4	Ventricular septal defect	4
	754.5	Varus deformities of feet	3
	746.8	Other specified anomalies of heart	2
	756.0	Anomalies of skull and face bones	2
	277.0	Cystic fibrosis	1
	282.7	Other hemoglobinopathies	1
	746.3	Congenital stenosis of aortic arch	1
	747.3	Anomalies of pulmonary artery	1
	747.4	Anomalies of great veins	1
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	1
	751.1	Atresia and stenosis of small intestine	1
	751.3	Hirschprung's disease & other functional disorders of colon	1
	752.5	Undescended testicle	1
	752.7	Indeterminate sex & pseudo-hermaphroditism	1
	752.8	Other specified anomalies of genital organs	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	758.0	Down syndrome	1
	759.8	Other specified anomalies	1
Greene			
	745.5	Ostium secundum atrial septal defect	6
	750.5	Congenital hypertrophic pyloric stenosis	2
	745.4	Ventricular septal defect	1
	746.4	Congenital insufficiency of aortic arch	1
	746.8	Other specified anomalies of heart	1
	752.5	Undescended testicle	1
	752.6	Hypospadias & epispadias	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	753.3	Other specified anomalies of kidney	1
	753.4	Other specified anomalies of ureter	1
	756.7	Anomalies of abdominal wall	1
	758.0	Down syndrome	1
Hamilton			
	745.4	Ventricular septal defect	1
	752.6	Hypospadias & epispadias	1
	754.3	Congenital dislocation of hip	1
Herkimer			
	752.6	Hypospadias & epispadias	6
	754.5	Varus deformities of feet	6
	745.4	Ventricular septal defect	5
	755.6	Other anomalies of lower limb including pelvic girdle	4
	745.5	Ostium secundum atrial septal defect	3
	754.3	Congenital dislocation of hip	3
	747.0	Patent ductus arteriosus	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	752.5	Undescended testicle	2
	755.5	Other anomalies of upper limb including shoulder girdle	2

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County	ICD-9 Code	Description	Number Reported
Jefferson			
	752.6	Hypospadias & epispadias	12
	745.4	Ventricular septal defect	6
	753.2	Obstructive defects of renal pelvis & ureter	6
	745.5	Ostium secundum atrial septal defect	4
	752.5	Undescended testicle	3
	754.5	Varus deformities of feet	3
	742.1	Microcephalus	2
	744.0	Anomalies of ear causing impairment of hearing	2
	746.0	Anomalies of pulmonary valve	2
	747.3	Anomalies of pulmonary artery	2
	750.5	Congenital hypertrophic pyloric stenosis	2
Kings			
	745.5	Ostium secundum atrial septal defect	218
	745.4	Ventricular septal defect	145
	752.6	Hypospadias & epispadias	126
	752.5	Undescended testicle	103
	753.2	Obstructive defects of renal pelvis & ureter	72
	746.8	Other specified anomalies of heart	67
	754.3	Congenital dislocation of hip	61
	754.5	Varus deformities of feet	58
	746.0	Anomalies of pulmonary valve	53
	758.0	Down syndrome	50
Lewis			
	752.6	Hypospadias & epispadias	4
	754.3	Congenital dislocation of hip	3
	745.5	Ostium secundum atrial septal defect	2
	745.6	Endocardial cushion defects	2
	752.5	Undescended testicle	2
	758.0	Down syndrome	2
	740.0	Anencephalus	1
	745.4	Ventricular septal defect	1
	747.1	Coarctation of aorta	1
	747.3	Anomalies of pulmonary artery	1
	753.2	Obstructive defects of renal pelvis & ureter	1
Livingston			
	752.6	Hypospadias & epispadias	5
	745.4	Ventricular septal defect	3
	750.5	Congenital hypertrophic pyloric stenosis	3
	752.5	Undescended testicle	3
	743.4	Coloboma & other anomalies of anterior segment	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	754.5	Varus deformities of feet	2
	745.2	Tetralogy of Fallot	1
	746.0	Anomalies of pulmonary valve	1
	747.3	Anomalies of pulmonary artery	1
	749.1	Cleft lip	1
	752.8	Other specified anomalies of genital organs	1
	753.3	Other specified anomalies of kidney	1
	754.3	Congenital dislocation of hip	1
	755.6	Other anomalies of lower limb including pelvic girdle	1
	756.1	Anomalies of spine	1

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County	ICD-9 Code	Description	Number Reported
Madison			
	745.4	Ventricular septal defect	3
	745.5	Ostium secundum atrial septal defect	3
	752.5	Undescended testicle	3
	752.6	Hypospadias & epispadias	3
	742.3	Congenital hydrocephalus	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	754.5	Varus deformities of feet	2
	756.0	Anomalies of skull and face bones	2
	359.0	Congenital Hereditary Muscular Dystrophy	1
	742.1	Microcephalus	1
	742.2	Reduction deformities of brain	1
	742.5	Other specified anomalies of spinal cord	1
	745.0	Common truncus	1
	745.1	Transposition of great vessels	1
	746.0	Anomalies of pulmonary valve	1
	746.8	Other specified anomalies of heart	1
	747.1	Coarctation of aorta	1
	748.3	Other anomalies of larynx, trachea, & bronchus	1
	749.0	Cleft palate	1
	749.2	Cleft palate with cleft lip	1
	751.2	Atresia and stenosis of large intestine, rectum, & anal canal	1
	752.4	Anomalies of cervix, vagina & external female genitalia	1
	754.3	Congenital dislocation of hip	1
	755.6	Other anomalies of lower limb including pelvic girdle	1
	756.1	Anomalies of spine	1
	756.4	Chondrodystrophy	1
	756.7	Anomalies of abdominal wall	1
	758.3	Autosomal deletion syndromes	1
Monroe			
	752.6	Hypospadias & epispadias	31
	752.5	Undescended testicle	27
	745.4	Ventricular septal defect	25
	754.5	Varus deformities of feet	22
	754.3	Congenital dislocation of hip	14
	750.5	Congenital hypertrophic pyloric stenosis	13
	753.2	Obstructive defects of renal pelvis & ureter	13
	745.5	Ostium secundum atrial septal defect	12
	746.8	Other specified anomalies of heart	11
	747.3	Anomalies of pulmonary artery	10
	758.0	Down syndrome	10
Montgomery			
	752.5	Undescended testicle	5
	753.2	Obstructive defects of renal pelvis & ureter	3
	754.3	Congenital dislocation of hip	3
	754.5	Varus deformities of feet	3
	750.5	Congenital hypertrophic pyloric stenosis	2
	243.0	Congenital hypothyroidism	1
	448.0	Hereditary hemorrhagic telangiectasia	1
	742.3	Congenital hydrocephalus	1
	742.4	Other specified anomalies of brain	1
	745.1	Transposition of great vessels	1
	745.5	Ostium secundum atrial septal defect	1

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County	ICD-9 Code	Description	Number Reported
Montgomery (cont.)	747.0	Patent ductus arteriosus	1
	747.4	Anomalies of great veins	1
	749.2	Cleft palate with cleft lip	1
	750.2	Other specified anomalies, mouth and pharynx	1
	750.4	Other specified anomalies of esophagus	1
	751.2	Atresia and stenosis of large intestine, rectum, & anal canal	1
	752.6	Hypospadias & epispadias	1
	753.3	Other specified anomalies of kidney	1
	753.6	Atresia and stenosis of urethra & bladder neck	1
	755.2	Reduction deformities of upper limb	1
Nassau			
	752.6	Hypospadias & epispadias	109
	745.5	Ostium secundum atrial septal defect	88
	745.4	Ventricular septal defect	84
	753.2	Obstructive defects of renal pelvis & ureter	72
	752.5	Undescended testicle	69
	754.3	Congenital dislocation of hip	38
	754.5	Varus deformities of feet	37
	750.5	Congenital hypertrophic pyloric stenosis	36
	746.8	Other specified anomalies of heart	34
	752.8	Other specified anomalies of genital organs	30
New York			
	745.4	Ventricular septal defect	73
	745.5	Ostium secundum atrial septal defect	69
	752.6	Hypospadias & epispadias	63
	752.5	Undescended testicle	51
	754.3	Congenital dislocation of hip	42
	753.2	Obstructive defects of renal pelvis & ureter	37
	746.0	Anomalies of pulmonary valve	31
	750.5	Congenital hypertrophic pyloric stenosis	23
	746.8	Other specified anomalies of heart	21
	758.0	Down syndrome	21
Niagara			
	745.5	Ostium secundum atrial septal defect	15
	752.6	Hypospadias & epispadias	13
	745.4	Ventricular septal defect	12
	752.5	Undescended testicle	12
	746.8	Other specified anomalies of heart	10
	750.5	Congenital hypertrophic pyloric stenosis	10
	756.0	Anomalies of skull and face bones	6
	747.0	Patent ductus arteriosus	5
	747.3	Anomalies of pulmonary artery	5
	753.2	Obstructive defects of renal pelvis & ureter	5
Oneida			
	752.6	Hypospadias & epispadias	10
	754.3	Congenital dislocation of hip	10
	745.4	Ventricular septal defect	9
	752.5	Undescended testicle	9
	745.5	Ostium secundum atrial septal defect	8
	753.2	Obstructive defects of renal pelvis & ureter	6
	754.5	Varus deformities of feet	6
	745.1	Transposition of great vessels	4
	746.0	Anomalies of pulmonary valve	4

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County	ICD-9 Code	Description	Number Reported
Oneida (cont.)	755.2	Reduction deformities of upper limb	4
	759.8	Other specified anomalies	4
Onondaga			
	752.6	Hypospadias & epispadias	33
	752.5	Undescended testicle	21
	745.4	Ventricular septal defect	16
	745.5	Ostium secundum atrial septal defect	15
	753.2	Obstructive defects of renal pelvis & ureter	15
	754.5	Varus deformities of feet	15
	754.3	Congenital dislocation of hip	9
	748.3	Other anomalies of larynx, trachea, & bronchus	8
	755.6	Other anomalies of lower limb including pelvic girdle	8
	746.0	Anomalies of pulmonary valve	7
	746.8	Other specified anomalies of heart	7
	758.0	Down syndrome	7
Ontario			
	752.5	Undescended testicle	8
	754.3	Congenital dislocation of hip	8
	745.4	Ventricular septal defect	6
	752.6	Hypospadias & epispadias	5
	750.5	Congenital hypertrophic pyloric stenosis	4
	754.5	Varus deformities of feet	4
	755.6	Other anomalies of lower limb including pelvic girdle	4
	748.3	Other anomalies of larynx, trachea, & bronchus	3
	742.1	Microcephalus	2
	745.1	Transposition of great vessels	2
	745.5	Ostium secundum atrial septal defect	2
	749.2	Cleft palate with cleft lip	2
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	755.2	Reduction deformities of upper limb	2
Orange			
	752.5	Undescended testicle	16
	745.4	Ventricular septal defect	14
	752.6	Hypospadias & epispadias	13
	745.5	Ostium secundum atrial septal defect	9
	754.3	Congenital dislocation of hip	9
	754.5	Varus deformities of feet	9
	750.5	Congenital hypertrophic pyloric stenosis	7
	749.0	Cleft palate	6
	747.1	Coarctation of aorta	5
	747.3	Anomalies of pulmonary artery	5
	753.1	Cystic kidney disease	5
Orleans			
	752.6	Hypospadias & epispadias	5
	754.5	Varus deformities of feet	4
	747.3	Anomalies of pulmonary artery	3
	746.0	Anomalies of pulmonary valve	2
	448.0	Hereditary hemorrhagic telangiectasia	1
	742.3	Congenital hydrocephalus	1
	745.2	Tetralogy of Fallot	1
	745.4	Ventricular septal defect	1
	746.6	Congenital mitral insufficiency	1

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County	ICD-9 Code	Description	Number Reported
Orleans (cont.)	746.8	Other specified anomalies of heart	1
	750.2	Other specified anomalies, mouth and pharynx	1
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	752.5	Undescended testicle	1
	755.2	Reduction deformities of upper limb	1
	756.0	Anomalies of skull and face bones	1
	756.1	Anomalies of spine	1
	756.3	Other anomalies of ribs and sternum	1
	758.0	Down syndrome	1
Oswego			
	752.6	Hypospadias & epispadias	8
	745.4	Ventricular septal defect	5
	750.5	Congenital hypertrophic pyloric stenosis	5
	752.5	Undescended testicle	5
	745.5	Ostium secundum atrial septal defect	4
	746.8	Other specified anomalies of heart	4
	753.2	Obstructive defects of renal pelvis & ureter	4
	754.3	Congenital dislocation of hip	3
	747.1	Coarctation of aorta	2
	747.2	Other anomalies of aorta	2
	748.3	Other anomalies of larynx, trachea, & bronchus	2
	753.1	Cystic kidney disease	2
	755.6	Other anomalies of lower limb including pelvic girdle	2
	756.0	Anomalies of skull and face bones	2
Otsego			
	754.5	Varus deformities of feet	4
	742.2	Reduction deformities of brain	3
	746.8	Other specified anomalies of heart	3
	749.1	Cleft lip	3
	243.0	Congenital hypothyroidism	2
	749.0	Cleft palate	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	753.3	Other specified anomalies of kidney	2
	756.0	Anomalies of skull and face bones	2
Putnam			
	752.6	Hypospadias & epispadias	5
	753.2	Obstructive defects of renal pelvis & ureter	4
	754.3	Congenital dislocation of hip	3
	277.0	Cystic fibrosis	2
	745.4	Ventricular septal defect	2
	745.5	Ostium secundum atrial septal defect	2
	746.8	Other specified anomalies of heart	2
	749.0	Cleft palate	2
	752.5	Undescended testicle	2
	270.1	Phenylketonuria	1
	742.1	Microcephalus	1
	744.0	Anomalies of ear causing impairment of hearing	1
	745.2	Tetralogy of Fallot	1
	751.1	Atresia and stenosis of small intestine	1
	751.4	Anomalies of intestinal fixation	1
	751.6	Anomalies of gallbladder, bile ducts, and liver	1

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County	ICD-9 Code	Description	Number Reported
Putnam (cont.)	752.8	Other specified anomalies of genital organs	1
	753.3	Other specified anomalies of kidney	1
	756.0	Anomalies of skull and face bones	1
	756.4	Chondrodystrophy	1
	758.5	Other conditions due autosomal anomalies	1
	758.8	Other conditions due to sex chromosome anomalies	1
Queens			
	745.5	Ostium secundum atrial septal defect	130
	752.5	Undescended testicle	106
	745.4	Ventricular septal defect	91
	752.6	Hypospadias & epispadias	88
	754.3	Congenital dislocation of hip	61
	753.2	Obstructive defects of renal pelvis & ureter	56
	754.5	Varus deformities of feet	49
	746.8	Other specified anomalies of heart	48
	448.0	Hereditary hemorrhagic telangiectasia	41
	746.0	Anomalies of pulmonary valve	37
Rensselaer			
	750.5	Congenital hypertrophic pyloric stenosis	14
	745.5	Ostium secundum atrial septal defect	9
	752.6	Hypospadias & epispadias	7
	754.3	Congenital dislocation of hip	7
	745.4	Ventricular septal defect	5
	746.8	Other specified anomalies of heart	4
	749.2	Cleft palate with cleft lip	4
	757.3	Other specified anomalies of skin	4
	746.0	Anomalies of pulmonary valve	3
	753.1	Cystic kidney disease	3
	753.2	Obstructive defects of renal pelvis & ureter	3
Richmond			
	752.6	Hypospadias & epispadias	30
	745.5	Ostium secundum atrial septal defect	24
	745.4	Ventricular septal defect	21
	754.5	Varus deformities of feet	20
	753.2	Obstructive defects of renal pelvis & ureter	17
	746.8	Other specified anomalies of heart	14
	750.5	Congenital hypertrophic pyloric stenosis	14
	752.5	Undescended testicle	11
	754.3	Congenital dislocation of hip	9
	746.0	Anomalies of pulmonary valve	7
Rockland			
	752.6	Hypospadias & epispadias	19
	752.5	Undescended testicle	18
	750.5	Congenital hypertrophic pyloric stenosis	15
	745.4	Ventricular septal defect	13
	753.2	Obstructive defects of renal pelvis & ureter	10
	754.5	Varus deformities of feet	10
	745.5	Ostium secundum atrial septal defect	9
	754.3	Congenital dislocation of hip	8
	758.0	Down syndrome	6
	742.3	Congenital hydrocephalus	5
	746.0	Anomalies of pulmonary valve	5

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County	ICD-9 Code	Description	Number Reported
Saratoga			
	752.6	Hypospadias & epispadias	11
	754.3	Congenital dislocation of hip	11
	752.5	Undescended testicle	9
	745.4	Ventricular septal defect	6
	745.5	Ostium secundum atrial septal defect	6
	747.0	Patent ductus arteriosus	6
	754.5	Varus deformities of feet	6
	742.3	Congenital hydrocephalus	4
	750.5	Congenital hypertrophic pyloric stenosis	4
	746.8	Other specified anomalies of heart	3
	753.1	Cystic kidney disease	3
	758.0	Down syndrome	3
Schenectady			
	752.6	Hypospadias & epispadias	9
	745.4	Ventricular septal defect	8
	752.5	Undescended testicle	8
	754.3	Congenital dislocation of hip	8
	750.5	Congenital hypertrophic pyloric stenosis	6
	745.5	Ostium secundum atrial septal defect	5
	754.5	Varus deformities of feet	5
	742.3	Congenital hydrocephalus	4
	753.2	Obstructive defects of renal pelvis & ureter	4
	758.0	Down syndrome	4
Schoharie			
	746.8	Other specified anomalies of heart	2
	277.0	Cystic fibrosis	1
	745.4	Ventricular septal defect	1
	745.6	Endocardial cushion defects	1
	746.0	Anomalies of pulmonary valve	1
	747.3	Anomalies of pulmonary artery	1
	747.4	Anomalies of great veins	1
	752.6	Hypospadias & epispadias	1
	759.0	Anomalies of spleen	1
	759.3	Situs inversus	1
Schuyler			
	752.6	Hypospadias & epispadias	2
	343.9	Infantile cerebral palsy unspecified	1
	741.9	Spina bifida w/o hydrocephalus	1
	742.1	Microcephalus	1
	747.3	Anomalies of pulmonary artery	1
	754.5	Varus deformities of feet	1
	771.1	Congenital cytomegalovirus infection	1
Seneca			
	745.4	Ventricular septal defect	3
	752.6	Hypospadias & epispadias	3
	746.8	Other specified anomalies of heart	2
	747.0	Patent ductus arteriosus	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	758.0	Down syndrome	2
	745.5	Ostium secundum atrial septal defect	1
	749.0	Cleft palate	1
	751.1	Atresia and stenosis of small intestine	1

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County	ICD-9 Code	Description	Number Reported
Seneca (cont.)	752.8	Other specified anomalies of genital organs	1
	754.3	Congenital dislocation of hip	1
	754.5	Varus deformities of feet	1
	755.1	Syndactyly	1
	755.3	Reduction deformities of lower limb	1
St. Lawrence			
	752.6	Hypospadias & epispadias	8
	752.5	Undescended testicle	7
	745.5	Ostium secundum atrial septal defect	5
	754.5	Varus deformities of feet	5
	745.4	Ventricular septal defect	3
	746.8	Other specified anomalies of heart	3
	749.0	Cleft palate	3
	752.8	Other specified anomalies of genital organs	3
	753.2	Obstructive defects of renal pelvis & ureter	3
	755.2	Reduction deformities of upper limb	3
Steuben			
	752.6	Hypospadias & epispadias	11
	745.4	Ventricular septal defect	5
	745.5	Ostium secundum atrial septal defect	5
	746.8	Other specified anomalies of heart	5
	754.3	Congenital dislocation of hip	5
	749.2	Cleft palate with cleft lip	3
	741.9	Spina bifida w/o hydrocephalus	2
	749.1	Cleft lip	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	752.5	Undescended testicle	2
	756.0	Anomalies of skull and face bones	2
	758.0	Down syndrome	2
Suffolk			
	745.4	Ventricular septal defect	84
	752.6	Hypospadias & epispadias	76
	753.2	Obstructive defects of renal pelvis & ureter	67
	745.5	Ostium secundum atrial septal defect	65
	754.3	Congenital dislocation of hip	63
	752.5	Undescended testicle	55
	750.5	Congenital hypertrophic pyloric stenosis	42
	754.5	Varus deformities of feet	33
	756.0	Anomalies of skull and face bones	30
	746.8	Other specified anomalies of heart	24
Sullivan			
	752.6	Hypospadias & epispadias	4
	745.4	Ventricular septal defect	3
	752.5	Undescended testicle	3
	752.8	Other specified anomalies of genital organs	3
	742.4	Other specified anomalies of brain	2
	745.1	Transposition of great vessels	2
	745.2	Tetralogy of Fallot	2
	745.6	Endocardial cushion defects	2
	754.3	Congenital dislocation of hip	2
	754.5	Varus deformities of feet	2
	758.0	Down syndrome	2

Section IV
Ten Most Frequently Reported Major Malformations
By County, 1995
Congenital Malformations Registry

County	ICD-9 Code	Description	Number Reported
Tioga			
	745.4	Ventricular septal defect	4
	745.5	Ostium secundum atrial septal defect	4
	747.3	Anomalies of pulmonary artery	4
	747.4	Anomalies of great veins	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	756.0	Anomalies of skull and face bones	2
	758.0	Down syndrome	2
	741.0	Spina bifida with hydrocephalus	1
	745.6	Endocardial cushion defects	1
	746.0	Anomalies of pulmonary valve	1
	746.7	Hypoplastic left heart syndrome	1
	746.8	Other specified anomalies of heart	1
	749.2	Cleft palate with cleft lip	1
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	751.1	Atresia and stenosis of small intestine	1
	751.2	Atresia and stenosis of large intestine, rectum, & anal canal	1
	751.3	Hirschprung's disease & other functional disorders of colon	1
	751.4	Anomalies of intestinal fixation	1
	751.7	Anomalies of pancreas	1
	753.4	Other specified anomalies of ureter	1
	754.5	Varus deformities of feet	1
	759.8	Other specified anomalies	1
Tompkins			
	745.4	Ventricular septal defect	3
	755.6	Other anomalies of lower limb including pelvic girdle	3
	750.5	Congenital hypertrophic pyloric stenosis	2
	752.5	Undescended testicle	2
	754.3	Congenital dislocation of hip	2
	282.0	Hereditary spherocytosis	1
	286.0	Congenital factor VIII disorder	1
	343.9	Infantile cerebral palsy unspecified	1
	359.0	Congenital Hereditary Muscular Dystrophy	1
	741.9	Spina bifida w/o hydrocephalus	1
	742.3	Congenital hydrocephalus	1
	745.5	Ostium secundum atrial septal defect	1
	747.3	Anomalies of pulmonary artery	1
	749.1	Cleft lip	1
	749.2	Cleft palate with cleft lip	1
	751.1	Atresia and stenosis of small intestine	1
	751.3	Hirschprung's disease & other functional disorders of colon	1
	752.6	Hypospadias & epispadias	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	753.4	Other specified anomalies of ureter	1
	754.5	Varus deformities of feet	1
	756.8	Other specified anomalies of muscle, tendon, fascia, etc.	1
	757.1	Ichthyosis congenita	1
	758.0	Down syndrome	1
	758.7	Klinefelters syndrome	1
Ulster			
	752.6	Hypospadias & epispadias	9
	754.3	Congenital dislocation of hip	7

Section IV
Ten Most Frequently Reported Major Malformations
By County, 1995
Congenital Malformations Registry

County	ICD-9 Code	Description	Number Reported
Ulster (cont.)	752.5	Undescended testicle	6
	745.5	Ostium secundum atrial septal defect	5
	753.2	Obstructive defects of renal pelvis & ureter	5
	742.1	Microcephalus	4
	745.4	Ventricular septal defect	4
	749.2	Cleft palate with cleft lip	4
	750.5	Congenital hypertrophic pyloric stenosis	4
	754.5	Varus deformities of feet	4
	756.0	Anomalies of skull and face bones	4
Warren			
	752.5	Undescended testicle	4
	745.4	Ventricular septal defect	3
	753.2	Obstructive defects of renal pelvis & ureter	3
	745.5	Ostium secundum atrial septal defect	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	753.0	Renal agenesis & dysgenesis	2
	754.3	Congenital dislocation of hip	2
	755.2	Reduction deformities of upper limb	2
	756.0	Anomalies of skull and face bones	2
	758.0	Down syndrome	2
Washington			
	745.4	Ventricular septal defect	5
	753.2	Obstructive defects of renal pelvis & ureter	5
	752.5	Undescended testicle	4
	752.6	Hypospadias & epispadias	4
	743.4	Coloboma & other anomalies of anterior segment	2
	746.0	Anomalies of pulmonary valve	2
	746.1	Tricuspid atresia & stenosis	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	751.3	Hirschprung's disease & other functional disorders of colon	2
	754.5	Varus deformities of feet	2
	756.0	Anomalies of skull and face bones	2
Wayne			
	745.4	Ventricular septal defect	6
	758.0	Down syndrome	3
	746.0	Anomalies of pulmonary valve	2
	746.8	Other specified anomalies of heart	2
	747.0	Patent ductus arteriosus	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	752.6	Hypospadias & epispadias	2
	753.0	Renal agenesis & dysgenesis	2
	754.3	Congenital dislocation of hip	2
	754.5	Varus deformities of feet	2
	755.1	Syndactyly	2
	756.0	Anomalies of skull and face bones	2
Westchester			
	752.6	Hypospadias & epispadias	45
	745.4	Ventricular septal defect	38
	752.5	Undescended testicle	38
	745.5	Ostium secundum atrial septal defect	36
	753.2	Obstructive defects of renal pelvis & ureter	32
	754.3	Congenital dislocation of hip	27
	750.5	Congenital hypertrophic pyloric stenosis	17

Section IV
Ten Most Frequently Reported Major Malformations
By County, 1995
Congenital Malformations Registry

County	ICD-9 Code	Description	Number Reported
Westchester (cont.)	754.5	Varus deformities of feet	15
	746.8	Other specified anomalies of heart	13
	748.3	Other anomalies of larynx, trachea, & bronchus	13
	758.0	Down syndrome	13
Wyoming			
	745.5	Ostium secundum atrial septal defect	3
	752.5	Undescended testicle	3
	752.6	Hypospadias & epispadias	3
	753.2	Obstructive defects of renal pelvis & ureter	3
	755.6	Other anomalies of lower limb including pelvic girdle	3
	743.4	Coloboma & other anomalies of anterior segment	2
	745.4	Ventricular septal defect	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	753.1	Cystic kidney disease	2
	753.3	Other specified anomalies of kidney	2
	758.0	Down syndrome	2
Yates			
	746.8	Other specified anomalies of heart	3
	754.3	Congenital dislocation of hip	3
	753.1	Cystic kidney disease	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	745.1	Transposition of great vessels	1
	745.2	Tetralogy of Fallot	1
	745.4	Ventricular septal defect	1
	747.1	Coarctation of aorta	1
	747.3	Anomalies of pulmonary artery	1
	750.2	Other specified anomalies, mouth and pharynx	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	753.4	Other specified anomalies of ureter	1
	759.8	Other specified anomalies	1

NOTES

Section V
Comparison of Selected Malformation Prevalence with Other Birth Defects Registries

Introduction to Table

The CMR relies on reports from hospitals and physicians for case ascertainment. Underreporting is an obvious concern, and the CMR monitors hospital reporting and follows up if there appears to be underreporting. In this section, CMR live birth prevalence are compared with the prevalences of two other registries, the Metropolitan Atlanta Congenital Defects Program (MACDP)[1] and the California Birth Defects Monitoring Program (CBDMP)[2]. These two registries send data collection specialists to hospitals to identify and abstract records of children with malformations. To help evaluate possible underreporting, CMR prevalences of selected malformations, defined using BPA codes, were compared with prevalence from these two registries. (See Appendix 4 for further information on these BPA codes.) These two programs follow children through one year of age. The CMR follows children through two years; however, more than 95% of cases are reported in the first year. Most of the malformations in this table are recognized at birth. The exceptions are fetal alcohol syndrome and some cardiac malformations.

The most striking difference among the registries is the low prevalence of anencephaly in New York State. This probably is largely due to the inclusion of stillborn infants in the MACDP and the CBDMP. The CMR includes only live born children (see Section VI, Current Topics). Underreporting is not the only reason for possible differences. The prevalences of some birth defects differ by race. True geographic differences may also exist[3].

Section V
Comparison of Selected Malformation Prevalence with two other Birth Defects Registries

MACDP Code	Malformation	CMR 1995	MACDP[4] 1995	CBDMP[4] 1995
A01	Anencephalus	0.6	2.2	2.9
A04	Spina bifida	3.7	1.2	3.6
A15	Hydrocephalus	7.8	5.0	5.9
A13	Encephalocele	1.0	1.2	0.9
A16	Microcephalus	5.8	5.2	-.-
B01	An/Microphthalmos	1.7	3.5	2.7
D01	Common truncus	0.7	0.7	1.1
D02	Trans of great vessels	4.8	4.7	4.6
D03	Tetralogy of Fallot	4.8	2.7	2.9
D05	Ventricular septal defect	34.5	21.6	14.8
D26	Coarctation of aorta	4.1	4.2	4.0
E01	Choanal atresia	1.0	1.2	1.3
E06	Lung agenesis/hypoplasia	3.6	4.5	-.-
F01	Cleft palate	5.8	4.7	5.4
F02	Cleft lip ± cleft palate	7.7	9.9	10.9
F09	Esophageal/tracheoesophageal atresia	2.4	2.5	2.3
F16	Rectal/large intestine atresia	4.2	2.7	4.2
F08	Pyloric stenosis	16.6	7.7	14.2
F17	Hirschsprung's disease	1.8	1.2	1.7
F21	Biliary atresia	0.7	1.0	0.9
H01	Renal agenesis/hypoplasia	3.3	2.0	-.-
H08	Bladder exstrophy	0.2	-.-	0.3
G02	Hypo/epispadias	33.7	33.5	12.9
K01	Reduct deform of upper limb	3.0	4.2	3.1
K02	Reduct deform of lower limb	1.9	2.5	1.2
N01	Diaphragmatic hernia	2.0	1.5	2.7
N02	Omphalocele	1.2	1.5	1.8
N04	Gastroschisis	1.2	2.2	2.2
R01	Down syndrome	10.1	10.7	13.1
R02	Trisomy 13	0.8	2.0	1.1
R03	Trisomy 18	1.0	2.2	2.0
S02	Fetal alcohol syndrome	2.0	3.0	0.9
K05	Amniotic bands	0.3	1.0	1.6

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NOTES

Section VI
Current Topics

NTD Surveillance and Prevention

The prevention of neural tube defects (NTD) by folic acid has been one of the most exciting events in the area of birth defects. The primary prevention of the occurrence of a birth defect rather than secondary prevention through termination is very gratifying. However, tracking the effects of folic acid on trends in NTDs is difficult and cannot be done easily by most registries at present. Many NTD affected pregnancies are detected prenatally as a result of ultrasound or AFP screening. One study from the Centers for Disease Control and Prevention (CDC) found that about 40% of NTD cases are detected prenatally and terminated and the percent of terminations increased over time[2]. Tracking trends in livebirths of NTD cases will not be very useful for surveillance of trends in the actual occurrence of NTDs. As discussed in our report for the 1992 birth cohort, NTDs appear to be declining in New York State but the CMR receives reports primarily on livebirths.

CMR Neural Tube Defect Surveillance Project

New York State has in place a regional NTD surveillance project using active case ascertainment. This project was developed through funds from a three-year CDC cooperative agreement. Beginning in 1996, active surveillance of NTDs has been carried out in the seven counties of the Lower Hudson Valley (HSA 6), an area with about 30,000 annual births. A surveillance system has been established using hospitals, radiology facilities, health care providers and termination facilities. Cases were collected retrospectively back to 1990 and prospective case ascertainment began in the spring of 1996.

Active collection of NTD cases in HSA 6, including prenatally diagnosed cases, has clearly shown the advantages over the passive ascertainment of the CMR (see Table 1). For 1990 through 1997, 91 cases were reported to the CMR, while the surveillance found 167. More than 18% of liveborn cases from 1990 to present were not reported to the CMR. None of the stillborn/terminated cases were reported to the CMR. Further, there were a significant number of cases reported through the CMR which, upon review, were excluded because they did not meet case criteria. These included cases erroneously coded with ICD codes corresponding to NTDs, which were clearly not NTDs upon review of medical records.

Table 1
Number of cases of NTD by Year of Birth, 1990-1997, HSA 6
CMR Neural Tube Defect Surveillance Project
(Number of cases of NTD reported to the CMR from HSA 6)

	1990	**1991**	**1992**	**1993**	**1994**	**1995**	**1996*†**	**1997†**
Anencephaly	6 *(1)*	6 *(2)*	5 *(2)*	9 *(4)*	9 *(2)*	4 *(0)*	11 *(0)*	5 *(0)*
Spina bifida	12 (*11*)	13 (*12*)	13 *(11)*	8 *(4)*	9 *(8)*	10 *(6)*	10 *(8)*	7 *(2)*
Encephalocele	7 *(5)*	7 *(7)*	3 *(1)*	1 *(1)*	3 *(2)*	3 *(0)*	4 *(2)*	2 *(1)*
Total	**25 *(17)***	**26 *(21)***	**21 *(13)***	**18 *(9)***	**21 *(12)***	**17 *(6)***	**25 *(10)***	**14 *(3)***

* Prospective surveillance began May 1996
† CMR reporting incomplete

Table 2
Annual Occurrence of Neural Tube Defects*
CMR Neural Tube Defects Surveillance Project
Lower Hudson Valley, 1990-1996

Year	Anencephaly	Encephalocele	Spina bifida	Total
90	0.19	0.22	0.38	0.78
91	0.19	0.22	0.41	0.83
92	0.16	0.10	0.42	0.68
93	0.30	0.03	0.26	0.60
94	0.30	0.10	0.30	0.70
95	0.13	0.10	0.33	0.57
96**	0.37	0.14	0.34	0.85
Total 90-96	**0.23**	**0.13**	**0.35**	**0.72**

* Per 1,000 live births.
** Prospective surveillance began May 1996.

Although there is some variation from year to year, probably due to small numbers, overall the occurrence of NTDs appears to be stable (Table 2). The occurrence of the individual malformations also appears to be stable with a possible decline in encephalocele and increase in anencephaly. However, both of these apparent variations could result from small numbers or variable case ascertainment.

Table 3
Percent of NTD Cases Terminated
CMR Neural Tube Defect Surveillance Project
Lower Hudson Valley, 1990 to 1998

	Anencephaly	Encephalocele	Spina Bifida	Total
90	50.0	14.3	8.3	20.0
91	67.0	14.3	7.7	23.1
92	20.0	33.3	7.7	14.3
93	55.0	0.0	37.5	44.4
94	33.0	33.3	11.1	23.8
95	100.0	66.7	20.0	47.0
96*	36.0	25.0	30.0	40.0
97	100.0	50.0	42.8	64.3
98**	100.0	0.0	33.0	60.0
Total**	**54.0**	**26.7**	**21.2**	**33.1**

* Prospective Surveillance began May 1996.
** Includes first 3/4 of 1998.

The overall percent of NTD cases terminated increased from 20% in the early 1990s to 40 to 60% in the later years. This was most marked for anencephalic cases which went from about 50 to 100%. However, there has been a noticeable increase in termination of spina bifida cases from less than 10% in the early 1990s to about 33% in the late 1990s. This again illustrates the need for surveillance to include terminated cases to be able to see true declines in occurrence of NTDs.

As expected, prenatal diagnosis of NTDs has substantially affected the birth prevalence. Between 1990 and 1996, 31% of all NTDs were electively terminated, and an additional 8% were stillborn or spontaneously aborted. The live birth prevalence of NTDs during this period was 0.45/1,000 live births. However, when including the cases that have been electively terminated and those that were stillborn and spontaneously aborted, the total prevalence rises to 0.72/1,000 pregnancies. No cases of anencephaly have been reported to the CMR from HSA 6 since 1994. Tables 2 and 3 present data from this surveillance system. Using this surveillance system the CMR will continue to monitor the occurrence of NTDs.

Knowledge and Use of Folic Acid

The major factor in primary prevention of NTDs is whether or not women of childbearing age know about and use folic acid before they become pregnant[3]. National data suggests that more education is necessary[4]. In a March of Dimes survey carried out by the Gallup Organization, 32% of women of childbearing age took folic acid supplement daily. However, only 13% knew folic acid helps prevent birth defects and 7% knew it should be taken before pregnancy.

The Department of Health's Bureau of Community Relations has an active folic acid campaign. In the Fall of 1996, this Bureau launched a professional educational campaign on folic acid. This was repeated in fall 1997. By spring 1998, nearly one million pieces of educational materials including brochures available in English, Spanish, Chinese and French, as well as grocery list pads, fact sheets, resource lists, tray inserts, etc. were distributed. DOH has also forged partnerships with other health-related professional groups, as well as grocery market chains, who have distributed their own educational materials on folic acid, as well as reprinting and distributing DOH-produced folic acid materials. The state's folic acid campaign has drawn praise from the CDC, FDA, and other states. DOH has also advised other states concerning their creation of folic acid campaigns. Despite these efforts, data from two systems suggest that educational efforts are still needed.

PRAMS

The Pregnancy Risk Assessment Monitoring system (PRAMS) is a CDC sponsored program which supplements birth certificate data by conducting surveillance of maternal behaviors during pregnancy[5]. Table 4 shows responses from pregnant women who had a live birth in 1996.

Table 4
Percent of Women Who Had Heard that Taking Folic Acid Reduces the Risk of Some Birth Defects by Maternal Characteristics
PRAMS 1996

Maternal Characteristics	Percent
Race	
White	69.9
Black	53.5
Age	
<20	36.4
20-29	64.3
=> 30	75.6
Education	
<High School	46.2
High School	54.2
>High School	81.2
Medicaid Status	
On Medicaid	46.2
No Medicaid	76.1
Total	**67.9**

Many minority women, younger women, women with less education and lower SES women have not heard about folic acid reducing the risk of birth defects. This does not indicate how many women actually took folic acid before the pregnancy.

BRFSS

Another source of information on the knowledge and use of folic acid among New Yorkers is the Behavioral Risk Factor Surveillance System (BRFSS). The BRFSS is a CDC sponsored program.

A random sample of New Yorkers are surveyed regarding their health behaviors.

These tables show BRFSS data collected in 1997 for women of childbearing age. Table 5 shows the percent of women 18-45 who say that they take vitamins and know that the vitamins contain folic acid. Overall, about 30% of women reported that they knew they were taking vitamins with folic acid. Since almost 55% of women reported they took some vitamins, the percent of women taking folic acid might actually be higher. The percent of women who knew that they took folic acid varied by maternal characteristics. The percent was lower for minority women and women under 24 years. Women with higher levels of education were more likely to know they were taking folic acid. Women with health insurance other than Medicaid were also more likely to know they were taking folic acid. Only about 19% of women on Medicaid know if they were taking folic acid.

Table 5
Percentage of Women Who Take Vitamins and Folic Acid
BRFSS, 1997

Age	Yes, take Vitamins	Yes, take Folic Acid
18-24	51	20
25-29	51	28
30-34	58	35
35-39	60	35
40-44	54	29
Race		
White, Non-hispanic	57	32
Black	48	22
Hispanic	56	22
Education		
<12 years	47	18
12 years	50	24
13-15 years	56	29
16+ years	59	37
Insurance		
Medicaid	50	21
Other	55	30
TOTAL	**55**	**29**

Table 6 shows the percent of women who know that folic acid prevents some birth defects. The overall percent, 36, is higher than the national percent of 13. Again, younger and minority women were less likely to know this. Women with more education and health insurance other than Medicaid were more likely to know that folic acid prevents some birth defects.

Table 6
Percent of Women Who Know That Folic Acid Prevents Some Birth Defects
BRFSS, 1997

Age	Yes, Know Folic Acid
18-24	32
25-29	36
30-34	44
35-39	37
40-44	32
Race	
White, Non-Hispanic	42
Black	28
Hispanic	23
Education	
<12 years	17
12 years	27
13-15 years	35
16+ years	49
Insurance	
Medicaid	28
Other	37
TOTAL	**36**

Both these surveys indicate that we still need to educate women about the importance of folic acid and that there are specific groups of women who need to be especially targeted to receive the information.

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Abstracts of CMR Recent Publications

1. Applegate MS, Druschel CM. The epidemiology of infantile hypertrophic pyloric stenosis in New York State: 1983-1990. *Arch Pediatric Adolesc Med* 1995; 149:1123-1129.

Abstract. *Objective:* To investigate an apparent decline in infantile hypertrophic pyloric stenosis (IHPS) and to examine the characteristics of children with IHPS and any associated malformations. *Design.* Cohort study, children with IHPS are compared with the population of live births. Trends of IHPS compared in two data sets: a population-based birth defects registry and hospital discharge data. *Participants:* Children with IHPS identified from a birth defects registry and the population of infants live born to residents of New York State from 1983 to 1990. *Main Outcome Measure:* Trends in the incidence of IHPS in two data sets, demographic characteristics and malformations associated with IHPS. *Results:* IHPS declined from 2.4 per 1,000 live births in 1984 to 1.7 in 1990. White race and male gender were associated with a higher occurrence of IHPS; high birth order, older maternal age, higher maternal education and low birthweight were associated with lower occurrence. Seven percent of children with IHPS had a major malformation compared with 3.7% of the general population. Three major malformations occurred more frequently in children with IHPS: intestinal malrotation, obstructive defects of the urinary tract and esophageal atresia. Fewer cases were found in the birth defects registry compared with hospital discharge data. *Conclusions:* There is underreporting of IHPS to the birth defects registry accounting for some of the decline. Children with IHPS have more major malformations compared with the general population, although some of the excess could be attributed to increased detection. Further investigation is needed into the environmental and socioeconomic factors associated with IHPS.

2. Druschel CM. A descriptive study of prune belly in New York State, 1983-1989. *Arch Pediatr Adolesc Med* 1995; 149:70-76.

Abstract. *Objective:* To determine the prevalence and spectrum of prune belly in a defined population. *Design:* Population-based descriptive study using New York State's Congenital Malformations Registry (CMR). *Setting:* The CMR is a statewide registry of children diagnosed with congenital anomalies before the age of two years. Cases were children with prune belly born during the years 1983 to 1989 to women resident in New York State and verified by medical record review. *Main Outcome Measures:* The live birth prevalence of prune belly for the total population and for population subgroups, such as race, sex, plurality, maternal age. The occurrence of other malformations with prune belly. *Results:* Sixty cases were ascertained, 50 male and 10 female. The live birth prevalence was 3.2 per 100,000 and declined over the time period. The prevalence was higher in males, 5.1 per 100,000 than females, 1.1; and higher in blacks, 5.8 than whites, 2.6. The live birth prevalence in twins, 12.2 per 100,000, was four times higher than in singletons, 3.0. More than 60% of cases died, most in the first week. Seventy percent of cases had one of the commonly described associated defects. Pulmonary hypoplasia was the most common. Almost one-third of the cases had defects other than those typically associated with prune belly. *Conclusions:* Twins, black and younger mothers appear to be at higher risk. Mortality remains high, especially early, with many deaths due to pulmonary hypoplasia. Further studies should include stillborn and terminated cases.

3. Druschel CM, Hughes J, Olsen CL. Mortality among infants with congenital malformations in New York State, 1983-1988. *Public Health Rep* 1996; 111:359-365.

Abstract. While the majority of infant deaths are due to congenital anomalies, few studies specifically look at their mortality experience. This study examines the mortality of infants born from 1983 to 1988 ascertained from a statewide population-based Congenital Malformations Registry and risk factors for first-year mortality. Variables analyzed were the year of birth, birthweight, gestational age, infant sex, number of malformations, number of organ systems, level of care of the birth hospital, maternal age, maternal race and maternal education. Infants with major malformations had a risk of death 6.3 times higher than the general population of live births. The risk declined from 6.4 in 1983 to 5.9 in 1988. The survival for whites and blacks after adjusting for other factors was similar. Having a malformation outweighs most of the other usual risks for infant mortality.

4. Druschel CM, Hughes JP, Olsen CL. First year of life mortality among infants with oral clefts: New York State, 1983-1990. *Cleft Palate Craniofacial J* 1996; 33(5):400-405.

Abstract. This study examined the mortality experience of children with oral clefts using the New York State Congenital Malformations Registry. Infants born in the years 1983 to 1990 to New York residents, diagnosed with an oral cleft and matched to their birth certificate were included in the analysis. Children with oral clefts were compared with a sample of live births from the years 1983-1990 without malformation. Children with cleft palate without additional malformations had a statistically nonsignificant adjusted risk of 1.2 when compared with children with no malformations. Children with cleft lip with or without cleft palate had a 1.1 adjusted risk. However, 35% of children with oral clefts had associated malformations and experienced much higher mortality. Children with oral clefts should be carefully evaluated for additional malformations. If none are found, their mortality appears not to be elevated.

5. Olsen CL, Polan AK, Cross PK. Case ascertainment for state-based birth defects registries: characteristics of unreported infants ascertained through birth certificates and their impact on registry statistics in New York State. *Paediatr Perinat Epidemiol* 1996; 10:161-174.

Abstract. Cases in the New York State Congenital Malformations Registry are reported by hospitals and physicians. This study was undertaken to determine whether case finding should be expanded to include routine matching of vital records files to the registry to identify unreported children. Matching of children who were born in 1983-1986 and who had a congenital malformation noted on their birth certificate yielded 2,837 children who were not in the registry. The hospital of record was asked to submit a registry report if the child's medical record contained a congenital malformation. Medical records for 1,267 (45%) of these children indicated that the child was normal, with no mention of a malformation. Medical records could not be located for 137. Registry reports were submitted for 1,433; 67 of whom were subsequently found in the registry, leaving 1,366 bona fide new cases. These new cases differ significantly from registry cases for a number of birth certificate variables and type of congenital malformation. The birth certificate cases were more likely than registry cases to have only one malformation and to have only a minor malformation. The 1,366 new cases comprised 2.1% of all registry cases for 1983-1986. Their addition increased the statewide prevalence of major malformations by 1.7%, from 416.5 to 423.4 per 10,000 live births. Except for anencephaly, the prevalence of specific malformations was not altered measurably by the addition of these cases. Lengthy and continuous follow-up was required to obtain registry reports. The small number of cases found does not seem to justify the amount of resources that would be required to routinely use birth certificates to augment case finding in New York State.

6. Olsen CL, Cross PK, Gensburg LJ, Hughes JP. The effects of prenatal diagnosis, population aging, and changing fertility rates on the live birth prevalence of Down Syndrome in New York State, 1983-1992. *Prenatal Diagnosis* 1996; 16:991-1002.

Abstract. The incidence of Down syndrome (DS) at conception is highly dependent upon the maternal age distribution and age-specific pregnancy rates. Live birth prevalence of DS reflects these factors and fetal deaths. This study examined DS live birth prevalence from 1983-1992 in New York State and analyzed the effects of demographic changes and prenatal diagnosis use on the observed live birth prevalence. Expected DS live birth prevalence without prenatal diagnosis was calculated and compared to the observed prevalence. Data were obtained from the birth defects registry, vital records and population data maintained by the New York State Department of Health. Over time, DS live birth prevalence was stable at about 10.4 per 10,000 live births. The percentage and number of women in the population above age 30 increased, as did birth rates among these women. Birth rates among younger women decreased. The proportion of DS babies born to women aged 35 and over increased from 27.1% to 34.1%. Use of prenatal diagnosis by this age group ranged from 39.6% to 43.2%, and increased steadily from 1.8% to 4.3% among women under 35. Detection of DS fetuses increased from 82 in 1985 to 233 in 1992. Without prenatal diagnosis, DS live birth prevalence in 1992 would have reached 15.3 per 10,000 live births compared with the 10.2 observed. Prenatal diagnosis has prevented an increase in DS live birth prevalence but has not been sufficient to reduce live birth prevalence significantly.

7. Olsen C. L., Hughes, J. P., Youngblood, L. G., and Sharpe-Stimac, M. Epidemiology of holoprosencephaly and phenotypic characteristics of affected children: New York State, 1984-1989. *American Journal of Medical Genetics* 1997; 73:217-226.

Abstract. Holoprosencephaly is a congenital defect of the median structures of the brain and face. The epidemiology is poorly known due to the paucity of population-based studies. This study describes the epidemiology of holoprosencephaly in a large population, using cases identified through the New York State Congenital Malformations Registry. The authors describe the range and co-occurrence of craniofacial abnormalities present, and examine the correspondence between the severity of craniofacial abnormalities, chromosomal abnormalities and severity of the brain defect. Liveborn cases totaled 78, yielding a prevalence of 4.8 per 100,000 live births. Prevalence among females was nearly double that in males, and was 4.2 times higher among infants of mothers under age 18 compared with infants of older mothers. Only 9.8% of all cases had no craniofacial abnormalities other than the brain defect. Eye malformations were present in 76.8%, nose malformations in 69.5%, ear malformations in 50% and oral clefts in 41.5%. These malformations arise at different times during gestation. The variability in patterns of their co-occurrence suggests variability in the time during gestation when holoprosencephaly arises, which in turn supports a model of multiple causality. Children with alobar holoprosencephaly tended to have the most severe craniofacial anomalies, but the correspondence was not 100%. Similarly, craniofacial phenotype does not discriminate between cytogenetically normal and abnormal cases.

NOTES

APPENDICES

Appendix 1

Reporting Card, Congenital Malformations Registry

Appendix 1
Reporting Card, Congenital Malformations Registry

Type or print clearly using only blue or black ink.

NEW YORK STATE DEPARTMENT OF HEALTH
Bureau of Environmental and Occupational Epidemiology

CONGENITAL MALFORMATIONS REGISTRY
CONFIDENTIAL REPORT FORM

Reporting Source (STAMP ACCEPTABLE)
NAME
STREET ADDRESS
CITY STATE ZIP CODE

Medical Record Number (1)
Adm. Type Code (see over) (2)
PFI No. (3)
Child's Name LAST FIRST M. I. (4)
Is this birth (5) SINGLE 1 TWIN 2 OTHER (specify)
If the child has been identified by another name(s), enter the name(s) (6)
Birthweight/grams (7)
Head Circum. cm (8)
If not a single birth, specify (9) FIRST 1 SECOND 2 OTHER
Patient's Street Address CITY STATE ZIP CODE (10)
Date of Birth MONTH DAY YEAR (11)
Birth (12) LIVE 1 STILL 2
Sex (13) MALE 1 FEMALE 2 UNDES. 3
Race (14) WHITE 1 BLACK 2 NATIVE AMERICAN 81 PACIFIC ISLANDER 82 ASIAN (specify) 83 OTHER (specify) 85
Hispanic (15) YES 1 NO 2
Deceased (16) YES 1 NO 2
Foster/Adopted (17) FOSTER 1 ADOPTED 2 NO 3
Place of Birth (18) HOSPITAL CITY STATE
Date of Discharge (19) MONTH DAY YEAR
Mother's Name LAST FIRST M. I. MAIDEN NAME (20)
Age (21)
Date of Birth (22) MONTH DAY YEAR
Father's Name LAST FIRST M. I. (23)
Age (24)
Date of Birth (25) MONTH DAY YEAR
Date of Diagnosis (26) MONTH DAY YEAR
Child transferred to another facility (27) TRANSFERRED TO:
Pediatrician's Name (28) LAST FIRST
Mother's S.S. No. (29)
Father's S.S. No. (30)
Chromosome Studies? (31) YES 1 NO 2 PENDING 3 If yes/pending, Karyotype or name of chrom. testing facility is requested
Attending Physician (32) LAST FIRST
Diagnoses (CONTINUE ON BACK) (33)

DEPARTMENT OF HEALTH USE ONLY

DATE RECEIVED (34)
SOUNDEX (35)
CASE NUMBER (36)
BC NUMBER (37)
DC NUMBER (38)
R.A.B. (39)
PFI TRANS. TO (40)
RESIDENCE (41)
P.O.B. (42)
CHROM. REG. NO. (43)
CODES (44)
(45)
(46)
(47)
(48)
(49)
(50)
(51)

See other side of card

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Additional Diagnoses and Comments

Patient transferred from:

Mail completed form in sealed envelope to:

New York State Department of Health
Bureau of Environmental and Occuptaional Epidemiology
Congenital Malformations Registry
Flanigan Square
547 River Street, Room 200
Troy, NY 12180-2216
Telephone: (518) 402-7990

Admission Type:

1 Newborn (age 0-30 days) born at this facility
2 Newborn (age 0-30 days) transferred from another facility
3 Older child (age 31 days - 2 years) transferred from another facility
4 Older child (age 31 days - 2 years) originally admitted at this facility

Check here if you need more:

☐ Forms
☐ Envelopes

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Appendix 2

Classification of Codes

Congenital malformations have traditionally been divided into categories of "major" and "minor". A major anomaly has an adverse effect on the individual's health, functioning or social acceptability. A minor anomaly is generally considered of limited social or medical significance. While minor anomalies in themselves do not greatly affect the child, they can be related to major anomalies or be indications of certain syndromes[1,2].

The division between major and minor is far from perfect. No standard lists or definitions exist. We used several sources, including the practices of other registries, to develop a list of minor anomalies[3, 4, 5]. One serious problem in making this distinction is that some ICD-9-CM codes include major and minor malformations under the same code. A more specific coding scheme that eliminates most of these problems has been adopted.

Following is a general listing of conditions included in this report and their classification. A few codes are not listed since they contain only a very few cases. Reporting hospitals receive a CMR Handbook with a complete, detailed list of reportable anomalies.

Major Malformations

090.0 -	090.9	Congenital Syphilis
	658.8	Amniotic Bands
740 -	759*	Congenital Anomalies
	760.71	Fetal Alcohol Syndrome
771.0 -	771.2	Congenital Infections: including rubella, cytomegalovirus, toxoplasmosis and herpes simplex

*See list of minor and excluded codes

Minor Malformations

214	Lipoma
216	Benign neoplasm of skin
228.01	Hemangioma of skin
550	Inguinal hernia in males
553.1	Umbilical hernia
743.65	Specified congenital anomalies of lacrimal passages
744.1	Accessory auricle
744.29	Other specified anomalies of ear
744.3	Unspecified anomaly of ear
744.4	Branchial cleft cyst
744.89	Other specified anomalies of face and neck
744.9	Other unspecified anomalies of face and neck
747.0	Patent ductus arteriosis, if birth weight <1500 grams
747.5	Single umbilical artery
752.41	Embryonic cyst of cervix, vagina and external female genitalia
752.42	Imperforate hymen
752.5	Undescended testicle, if birth weight < 2500 grams
754.61	Congenital pes planus
755.0	Polydactyly
755.11, 755.13	Syndactyly without fusion of bone
757.2	Dermatoglyphic anomalies
757.32	Vascular hamartomas
757.33	Congenital pigmentation anomalies of skin
757.39	Other anomalies of skin
757.4	Specified anomalies of hair
757.5	Specified anomalies of nails
757.6	Specified anomalies of breast
757.8	Other specified anomalies of integument
757.9	Unspecified anomalies of the integument

Exclusions

750.0	Tongue tie
758.4	Balanced autosomal translocation in normal individual
778.6	Congenital hydrocele

References

1. Marden PM, Smith DW, McDonald MJ. Congenital anomalies in the newborn infant including minor variations. *J Pediat* 1964; 64:357-371.

2. Lippig KA, Werler MM, Caron CI, Cook CA, Holmes LB. Predictive value of minor abnormalities: association with major malformations. *J Pediatr* 1987; 110:530-537.

3. Merlob P, Papier CM, Klingberg MA, Reisner SH. Incidence of congenital malformations in the newborn, particularly minor abnormalities. In: Marois, ed. *Prevention of physical and mental congenital defects, Part C: Basic and medical sciences, education and future strategies. Proceedings of a conference of the Institut de la Vie***.** New York: Alan R. Liss, 1985:51-53.

4. Myrianthopoulos NC, Chung CS. Congenital malformations in singletons: epidemiologic survey. Birth Defects: *Original Article Series*, 1974; X: 2-3, 51-58.

5. Jones KL, *Smith's Recognizable Patterns of Human Malformation*. 4th ed. Philadelphia: W.B. Saunders Co., 1988:662-681.

Appendix 3

Birth Certificate Matching

Birth certificate matching is a vital part of registry activities. This serves to verify the individual's identity and distinguish him or her from all others and provides additional information about the baby and the mother. The matching is used to determine maternal residence at birth and to verify race and birthweight. Matched cases provide a basis to calculate population-based rates. It is critical to match a high percentage of cases to calculate rates accurately and to conduct meaningful surveillance.

Birth certificate matching is carried out by a computer program that compares the birth certificate tape for a given year to the CMR file of cases who were born in that year. The files are compared on several variables until (1) a match is found, (2) a possible match is found or (3) the list is exhausted without finding a match.

Possible matches are reviewed by CMR staff and a decision made about whether there is a match. Unmatched cases are checked further to see if data items have been correctly keyed and all possible aliases have been identified. An online search of the birth certificate files is done and certificates on file at the Vital Records office are reviewed to find unmatched cases. However, review of actual certificates is possible only for children born outside New York City since New York City birth certificates are not on file in Albany. New York City maintains its own vital records files.

The matching process is repeated until about 95% of reported cases are matched. This is a compromise between completeness and efficiency. After about 90% of cases are matched, each additional percentage requires greater and greater effort. The ability to review a copy of the birth certificate greatly enhances the chance of making a match. Matching is more complete for cases born in the state outside New York City than for New York City cases.

Table 1

Birth Certificate Matching by Place of Birth
1995 Births

Region	Matched	Not Matched
Outside New York City	95.7	4.3
New York City	92.8	7.2
New York State	**94.3**	**5.7**

Appendix 4

BPA Codes

Many birth defects registries use a coding system modified from the British Pediatric Association (BPA). This coding system provides more specificity than the ICD-9 system. The Centers for Disease Control and Prevention Metropolitan Atlanta Congenital Defects Program (MACDP) has developed codes that group conditions. The table below shows the MACDP codes (used in Section V) and the corresponding BPA and ICD-9 codes. The ICD-9 code may include conditions others than those specified by the BPA code. For example, ICD-9 code 756.7 includes both gastroschisis and omphalocele, but the BPA code allows these conditions to be distinguished.

MACDP Code	Condition	ICD-9	BPA 5-Digit Code
CENTRAL NERVOUS SYSTEM			
A01	Anencephaly	740.0, 740.1, 740.2	740.00, 740.02, 740.03, 740.10, 740.20, 740.21, 740.29
A02	Spina Bifida with Hydrocephaly	741.0	741.00, 741.01, 741.02, 741.03, 741.04, 741.05, 741.06, 741.07, 741.08, 741.09
A03	Spina Bifida without Hydrocephaly	741.9	741.90, 741.91, 741.92, 741.93, 741.94, 741.98, 741.99, 742.00, 742.08, 742.09
A13	Encephalocele	742.0	742.00, 742.08, 742.09
A15	Hydrocephaly	742.3	742.30, 742.31, 742.38, 742.39
A16	Microcephalus	742.1	742.10
EYE / EAR			
B01	Anophthalmia, Microphthalmia	743.0, 743.1	743.00, 743.10
B03	Glaucoma	743.2	743.20, 743.21, 743.22
B04	Cataract		743.32
B54	Ear anomaly with hearing loss	744.0	744.00, 744.01, 744.02, 744.03, 744.09, 744.21
CARDIAC			
D01	Truncus arteriosus	745.0	745.00, 745.01
D02	Transposition of great vessels	745.1	745.10, 745.11, 745.12, 745.18, 745.19
D03	Tetralogy of Fallot	745.2	745.20, 745.21, 746.84
D04	Single ventricle	745.3	745.30
D05	VSD	745.4	745.40, 745.41, 745.48, 745.49
D52	Hypoplastic left heart	746.7	746.70
D53	Total anomalous pulmonary venous return	747.41	747.42
RESPIRATORY			
E01	Choanal atresia	748.0	748.00
E06	Agenesis of lung	748.5	748.50, 748.51
CLEFTS			
F01	Cleft palate	749.0	749.00, 749.01, 749.02, 749.03, 749.04, 749.05, 749.06, 749.07, 749.09

F02	Cleft lip with or without cleft palate	749.0, 749.2, 750.5	749.10, 749.11, 749.12, 749.19, 749.20, 749.21, 749.22, 749.29, 749.51

MACDP Code	Condition	ICD-9	BPA 5-Digit Code
GASTRO-INTESTINAL -			
F14	Stenosis or atresia of duodenum	751.1	751.10
F15	Other stenosis or atresia of small intestine	751.1	751.11, 751.12, 751.19
F16	Stenosis or atresia of rectum or anus	751.2	751.21, 751.22, 751.23, 751.24
F17	Hirschsprung's Disease	751.3	751.30, 751.31, 751.32, 751.33
F18	Malrotation of intestine	751.4	751.40, 751.41, 751.42, 751.49
F21	Biliary atresia	751.61	751.65
GENITO-URINARY -			
H01	Renal agenesis	753.0	753.00, 753.01
H06	Obstruction of kidney or ureter	753.3	753.20, 753.21, 753.22, 753.29, 753.40, 753.42
H09	Bladder or urethra obstruction	753.6	753.60, 753.61, 753.62, 753.63
MUSCULOSKELETAL -			
J02	Curvature of spine (scoliosis or lordosis)	754.2	754.20, 754.21, 754.22
J03	Dislocation of hip	754.3	754.30
J11	Arthrogryposis multiplex congenita	754.89	755.80
K01	Reduction deformity - upper limb	755.2	755.20, 755.21, 755.22, 755.23, 755.24, 755.25, 755.26, 755.27, 755.28, 755.29
K02	Reduction deformity - lower limb		755.30, 755.31, 755.32, 755.33, 755.34, 755.35, 755.36, 755.37, 755.38, 755.39
K05	Amniotic bands	658.8	658.80
N01	Diaphragmatic hernia	756.6	756.61
N02	Omphalocele	756.7	756.70
N04	Gastroschisis	756.7	756.71
SYNDROMES -			
R01	Down Syndrome	758.0	758.00, 758.01, 758.02, 758.03, 758.04, 758.09
R02	Patau Syndrome (Trisomy 13)	758.1	758.10, 758.11, 758.12, 758.13, 758.19
R03	Edwards Syndrome (Trisomy 18)	758.2	758.20, 758.21, 758.23, 758.29
S02	Fetal Alcohol Syndrome	760.71	760.71
W03	Conjoined twins	759.4	759.40, 759.41, 759.42, 759.43, 759.44, 759.48, 759.49

Appendix 5

Glossary of Terms*

Agenesis Absence of part(s) of the body.

Agenesis, aplasia, or hypoplasia of the lung The absence or incomplete development of a lung or lung tissue.

Anencephaly Congenital absence of the skull, with cerebral hemispheres completely missing or reduced to small masses attached to the base of the skull. Anencephaly is not compatible with life.

Aniridia The complete absence of the iris of the eye or a defect of the iris. Can be congenital or traumatically induced.

Anopthalmia A developmental defect characterized by complete absence of the eyes, or by the presence of vestigial eyes.

Anotia A congenital absence of one or both ears.

Aortic valve stenosis A cardiac anomaly characterized by a narrowing or stricture of the aortic valve. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. This condition can be repaired surgically in some cases.

Atresia Imperforation; absence or closure of a normal opening.

Atrial Septal Defect A congenital cardiac malformation in which there are one or several openings in the atrial septum (muscular and fibrous wall between the right and left atria) allowing a mixing of oxygenated and unoxygenated blood. The openings vary in size and may resolve without treatment or may require surgical treatment. Also called *ostium secundum defect.*

Biliary atresia A congenital absence or underdevelopment of one or more of the ducts in the biliary tract. Correctable surgically.

Bladder extrophy Incomplete closure of the anterior wall of the bladder and the abdominal cavity. The upper urinary tract is generally normal. Often associated with anorectal and genital malformations, and epispadias. Affected persons are at a markedly increased risk of bladder carcinoma (squamous cell). This condition is usually corrected surgically after birth.

Cataract An opacity (clouding) of the lens of the eye.

Choanal atresia or stenosis A congenital anomaly in which a bony or membranous formation blocks the passageway between the nose and the pharynx. This defect is usually repaired surgically after birth. Bilateral Choanal atresia is a surgical emergency.

Cleft lip The congenital failure of the fetal components of the lip to fuse or join, forming a groove or fissure in the lip. Infants with this condition can have difficulty feeding, and may use assistive devices for feeding. This condition is corrected when the infant can tolerate surgery.

Cleft palate The congenital failure of the palate to fuse properly, forming a grooved depression or fissure in the roof of the mouth. This defect varies in degree of severity. The fissure can extend into the hard and soft palate and into the nasal cavities. Infants with this condition have difficulty feeding, and may use assistive devices for feeding. Surgical correction is begun as soon as possible. Children with cleft palates are at high risk for hearing problems due to ear infections.

Coarctation of the aorta Localized narrowing of the aorta. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. This condition can vary from mild to severe. Surgical correction is recommended even for mild defects.

Common Truncus Ateriosus A congenital heart defect in which the common arterial trunk fails to divide into pulmonary artery and aorta. This is corrected surgically.

Confidence interval (95%) The interval that contains the true prevalence (which we can only estimate) 95% of the time. [See Methods for more explanation.]

Congenital Existing at or dating from birth.

Congenital hip dislocation 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. Treatment in early infancy consists of bracing of the joint to cause a deepening of the acetabulum.

Conjoined Twins Monozygotic twins who are physically united at birth. The defect can range from a superficial connection to one in which only a single body part is duplicated. Classified as symmetrical or asymmetrical by the degree of separation and development.

Craniosynostosis A premature ossification (closing) of the cranial sutures before birth or soon after birth. This condition is occasionally associated with other skeletal defects. If no surgical correction is made, the growth of the skull is inhibited, and the head is deformed. The eyes and the brain are often damaged.

Diaphragmatic hernia 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. Usually life-threatening and requires emergent surgery.

Down syndrome (Trisomy 21) The chromosomal abnormality characterized by an extra copy of chromosome 21. In rare cases this syndrome is caused by *translocation.* The extra copy can be free-lying, or can be attached to some other chromosome, most frequently number 14. Down syndrome can occur in *mosaic*. So that there is a population of normal cells and a population of trisomy 21 cells. Down syndrome is characterized by moderate to severe mental retardation, sloping forehead, small ear canals, flat bridged nose and short fingers and toes. One third of infants have congenital heart disease, and one third have duodenal atresia. (Both can be present in the same infant.) Affected people can survive to middle or old age. There is an increased incidence of Alzheimer disease in adults with Down syndrome.

Ebstein anomaly A congenital heart defect in which the tricuspid valve is displaced downward into the right ventricle causing abnormal patterns of cardiac circulation.

Edwards syndrome (Trisomy 18) The chromosomal abnormality characterized by an extra copy of chromosome 18. The extra chromosome can be free lying or attached to another chromosome. Trisomy 18 can occur in mosaic. Edwards syndrome is characterized by mental retardation, neonatal hepatitis, low-set ears, skull malformation and short digits. Cardiac and renal anomalies are also common. Survival for more than a few months is rare.

Encephalocele The protrusion of the brain substance through a defect in the skull.

Endocardial cushion defect A variety of septal defects (malformations of the walls separating the two atria and two ventricles of the heart) resulting from imperfect fusion of the endocardial cushions in the embryonic heart.

Epispadias A congenital defect in which the urinary meatus (urinary outlet) opens above (dorsal to) the normal position. The urinary sphincters are defective, so incontinence does occur. Surgical correction is aimed at correcting incontinence and permitting sexual functioning. The corresponding defect in females is rare. See also Hypospadias.

Esophageal Stenosis or Atresia: A narrowing or incomplete formation of the esophagus. Usually a surgical emergency. Frequently associated with a Tracheoesophageal Fistula.

Fetal alcohol syndrome A constellation of physical abnormalities (including characteristic abnormal facial features and growth retardation), and problems of behavior and cognition in children born to mothers who drank alcohol during pregnancy.

Fistula An abnormal passage from an internal organ to the body surface or between two internal organs or structures.

Gastroschisis A congenital opening of the abdominal wall with protrusion of the intestines. This condition is surgically treated. Contrast with Omphalocele, below.

Hirschsprung disease The congenital absence of autonomic ganglia (nerves controlling involuntary and reflexive movement) in the muscles of the colon. This results in immobility of the intestines and may cause obstruction or stretching of the intestines. This condition is repaired surgically in early childhood by the removal of the affected portion of the intestine.

Holoprosencephaly Failure of the brain to develop into two equal halves, so there is structural abnormality of the brain. There may be associated midline facial defects including cyclopia (fusion of the eye orbits into a single cavity containing one eye) in severe cases. About half the cases are probably due to a single gene defect (the HPE gene). Frequently occurs with Trisomy 13.

Hydrocephalus The abnormal accumulation of fluid within the spaces of the brain.

Hyperplasia Overgrowth characterized by an increase in the number of cells of a tissue.

Hypoplasia A condition of arrested development in which an organ or part remains below the normal size or in an immature state.

Hypoplastic left heart syndrome Atresia, or marked hypoplasia, of the aortic opening or valve, with hypoplasia of the ascending aorta and defective development of the left ventricle (with mitral valve atresia). This condition can be surgically repaired in a series of three procedures over a period of one year. Transplantation is also a treatment. This condition is usually fatal in the first month of life if not treated.

Hypospadias A congenital defect in which the urinary meatus (urinary outlet) is on the underside of the penis or on the perineum (area between the genitals and the anus). The urinary sphincters are not defective so incontinence does not occur. The condition may be surgically corrected if needed for cosmetic, urologic, or reproductive reasons. The corresponding defect in women is rare. *See also epispadias.*

Limb defects See Reduction deformities.

Meninges Membranes that cover the brain and spinal cord.

Microcephaly The congenital smallness of the head, with corresponding smallness of the brain.

Microphthalmia The congenital abnormal smallness of one or both eyes. Can occur in the presence of other ocular defects.

Microtia A small or maldeveloped external ear and atretic or stenotic external auditory canal.

Mosaic In genetics, this refers to an individual organism that has two or more kinds of genetically different cell types. The degree of abnormality depends on the type of tissue containing affected cells. Individuals may vary from near normal to full manifestation of the genetic syndrome. Can occur in any chromosome abnormality syndrome.

Neural tube defect A defect resulting from failure of the neural tube to close in the first month of pregnancy. The major conditions include anencephaly, spina bifida, and encephalocele.

Obstructive Genitourinary Defect Stenosis or atresia of the urinary tract at any level. Severity of the defect depends largely upon the level of the obstruction. Urine accumulates behind the obstruction and damages the organs.

Omphalocele The protrusion of an organ into the umbilicus. The defect is usually closed surgically soon after birth. Contrast with Gastroschisis.

Patau Syndrome (Trisomy 13) The chromosomal abnormality caused by a extra chromosome 13. The extra copy can be free-lying, or can be attached to some other chromosome. Patau syndrome can occur in *mosaic* so that there is a population of normal cells and a population of trisomy 13 cells. Patau syndrome is characterized by impaired midline facial development, cleft lip and palate, polydactyly and mental retardation. Most infants do not survive beyond 6 months of life.

Patent ductus arteriosus A blood vessel between the pulmonary artery and the aorta. This is normal in fetal life, but can cause problems after birth, particularly in premature infants. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. The vast majority close spontaneously and cause no problems. Medical or surgical correction may be done. This is only an abnormality if it causes significant medical problems.

Pulmonary artery anomaly Abnormality in the formation of the pulmonary artery such as stenosis or atresia. See also common truncus.

Pulmonary valve atresia or stenosis A congenital heart condition characterized by absence or constriction of the pulmonary valve. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. This condition can vary from mild to severe. Mild forms are relatively well tolerated and require no intervention. More severe forms are surgically corrected.

Pyloric stenosis A narrowing of the pyloric sphincter at the outlet of the stomach. This causes a blockage of food from the stomach into the small intestine. Usually treated surgically.

Reduction defects of the lower limbs The congenital absence of a portion of the lower limb. There are two general types of defect, transverse and longitudinal. Transverse defects appear like amputations, or like missing segments of the limb. Longitudinal defects are missing rays of the limb (for example, a missing tibia and great toe).

Reduction defects of the upper limbs The congenital absence of a portion of the upper limb. There are two general types of defect, transverse and longitudinal. Transverse defects appear like amputations, or like missing segments of the limb. Longitudinal defects are missing rays of the limb (for example, a missing radius and thumb).

Renal agenesis or dysgenesis The failure, or deviation, of embryonic development of the kidney.

Spina bifida A neural tube defect resulting from failure of the spinal neural tube to close. The spinal cord and/or meninges may or may not protrude. This usually results in damage to the spinal cord with paralysis of the involved limbs. Includes myelomeningocele (involving both spinal cord and meninges) and meningocele (involving just the meninges).

Stenosis A narrowing or constriction of the diameter of a bodily passage or orifice.

Stenosis or atresia of large intestine, rectum and anus The absence, closure or constriction of the large intestine, rectum or anus. Can be surgically corrected or bypassed.

Stenosis or atresia of the small intestine A narrowing or incomplete formation of the small intestine obstructing movement of food through the digestive tract.

Tetralogy of Fallot A congenital cardiac anomaly consisting of four defects: ventricular septal defect, pulmonary valve stenosis or atresia, displacement of the aorta to the right, and hypertrophy of right ventricle. The condition is corrected surgically.

Tracheoesophageal fistula An abnormal passage between the esophagus and trachea. Leads to pneumonia. Corrected surgically. It is frequently associated with esophageal atresia.

Translocation The rearrangement of genetic material within the same chromosome or the transfer of a segment of one chromosome to another one. People with balanced translocations do not always manifest genetic syndromes, but may be carriers of genetic syndromes and can have children with unbalanced translocations. Can occur with any chromosomal anomaly syndrome.

Transposition of the great vessels A congenital malformation in which the aorta arises from the right ventricle and the pulmonary artery from the left ventricle (opposite of normal), so that the venous return from the peripheral circulation is recirculated- without being oxygenated in the lungs. Immediate surgical correction is needed. When this is not associated with other cardiac defects, and not corrected, it is fatal.

Tricuspid valve atresia or stenosis A congenital

cardiac condition characterized by the absence or constriction of the tricuspid valve. The opening between the right atrium and right ventricle is absent or restricted, and normal circulation is not possible. This condition is often associated with other cardiac defects. This condition is surgically corrected depending on the severity.

Trisomy A chromosomal abnormality characterized by one more than the normal number of chromosomes. Normally, cells contain two of each chromosome. In trisomy, cells contain three copies of a specific chromosome.

Trisomy 13 See Patau Syndrome.

Trisomy 18 See Edwards Syndrome.

Trisomy 21 See Down Syndrome.

Truncus Arteriosus See Common Truncus.

Ventricular Septal Defect (VSD) A congenital cardiac malformation in which there are one or several openings in the ventricular septum (muscular and fibrous wall between the right and left ventricle *or right and left lower chambers of the heart*) allowing a mixing of oxygenated and unoxygenated blood. The openings vary in size and may resolve without treatment or require surgical treatment.

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