NEW YORK STATE DEPARTMENT OF HEALTH

Congenital Malformations Registry

Statistical Summary of Children
Born in 2007 and Diagnosed Through 2009

Additional and related information is also available from the New York State Department of Health Web site on the Internet: http://www.health.state.ny.us

Comments regarding the format or content of this report are welcome.

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Year of Publication 2012

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Summary

This Congenital Malformations Registry Summary Report presents rates of congenital malformations occurring among the 245,338 children who were born alive to New York residents in 2007. The children reported with a major congenital malformation represent 5.0 percent of live births. Males had a higher rate of major congenital malformations than females (6.0 percent versus 3.9 percent), and black children had a higher major malformation rate than white children (6.0 percent versus 5.0 percent). This information is provided through mandated reporting by hospitals and physicians.

Demographic characteristics of those children reported to the Congenital Malformations Registry (CMR) and the number of malformations are included in section I of the report. 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 are also included.

This is the nineteenth report from the CMR. Reports are also available by request for the 1983 to 2006 birth cohorts. This report and the reports for 1994-2006 are also available on the Department of Health website. The statistics in this report are **not** comparable to reports before 1992. In 1992, the CMR began to use a new coding system that allows for greater detail in coding. For previous years, ICD-9 codes were used. Information from birth certificates was used to supplement or correct reported data. Birth certificate matching also helps eliminate duplicate cases reported under different names and nonresident births. Reports produced for 1989 to 1991 did not use birth certificate matching.

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 percent 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 percent may be due to single gene mutations; 6 percent to chromosome abnormalities; and 5 percent to maternal illnesses, such as diabetes, infections or anticonvulsant drugs.[4] Approximately 40 percent to 60 percent 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 in 1978 to 1983 confirmed 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 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 electronically on the Internet using the Health Commerce System (HCS). The Department of Health developed the HCS as a secure system for electronically collecting and distributing health-related data. 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 submitted on the HCS using either online data entry forms or file upload facility are transferred to a DOH UNIX server for updating of the CMR database.

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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.

2007 Report

This current report presents statistics for major anomalies only (see Appendix 1 and the glossary of birth defects in Appendix 5). This is in accordance with the practices of other state birth defects registries and allows 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.

The statistics in this report are **not** comparable to reports prior to 1992. The 2007 report is based on birth certificate matched cases (Appendix 2) 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. Using maternal race is a common practice among birth defects registries nationwide as the race of the father is poorly reported. 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.[8] This report describes

children born in 2007 and diagnosed before their second birthday. Reports are also available for the 1984 through 2006 birth cohorts. Some reports and additional information are available through the DOH Web site at http://www.health.state.ny.us/diseases/congenital_malformations/cmrhome.htm.

Limitations

Care should be taken in the use of these data. 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 the U.S. 2010 census, the population of New York State was about 19.4 million; more than 42 percent of the population lived in New York City. An additional 24 percent of the population lived in the six counties closest to New York City. In 2007, there were 245,338 resident live births reported to the Bureau of Biometrics and Health Statistics of the New York State Department of Health, 16.6 percent to black mothers, and 23.7 percent to Hispanic mothers. In accordance with the practices of other state birth defects registries, the race of the child is based on race of the mother only. Approximately 48.1 percent of live births were to New York City residents.

References

1. Kochanek KD, Hudson BC. Advanced report of final mortality statistics, 1992. *Monthly Vital Statistics Report* 1995; 43(6 suppl.). Hyattsville (MD):National Center for Health Statistics, 1995.

2. Centers for Disease Control. Contribution of birth defects to infant mortality - United States 1986. *MMWR* 1989; 38:633-635.

3. Epstein CJ. Genetic disorders and birth defects. In: *Pediatrics*, Rudolph AM, Hoffman JIE, Axelrod S, eds. Norwalk: Appleton & Lange, 1987:209-210.

4. Kalter IT, Warkany J. Congenital malformation etiologic factors and their role in prevention. Parts I and II. *N Engl J Med* 1983; 308:424-431, 491-497.

5. Nelson K, Holmes LB. Malformations due to presumed spontaneous mutations in newborn infants. *N Engl J Med* 1989; 320:19-23.

6. Holtzman NA, Khoury MJ. Monitoring for congenital malformations. *Ann Rev Public Health* 1986; 7:237-266.

7. Lynberg MC, Edmonds LD. Surveillance of birth defects. In: *Public Health Surveillance*, W Halpern and E Baker, eds. Van Nostrand Reinhold, NY, 1992:157-176.

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

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 2007 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.

The overall occurrence of major malformations was 5.0% of live births. Male children have a higher rate of major malformations than female children (6.0% versus 3.9%, Table 1). This difference is consistent within different racial groups. The rates for major malformations are somewhat higher for black than for white children (6.0 % versus 5.0%). The major malformation rate among children with residence at birth in New York State excluding New York City was comparable to that among children with residence at birth in New York City (5.1% versus 4.8%). The smaller number of births in the "other" racial category makes these rates difficult to interpret.

About 78.4% of children reported with major malformations have only one major malformation (Table 2). 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).

Section 1 - Table 1
2007 Births - New York State Residents
Percent of Live Births with One or More Major Malformations
Sex by Race/Ethnicity and Residence

	Both Sexes			Males			Females		
Race and Residence	Infants	Total Births	%	Infants	Total Births	%	Infants	Total Births	%
New York State									
- All Races	12,220	245,338	5.0	7,505	125,245	6.0	4,715	120,093	3.9
- Non-Hispanic White	6,010	119,779	5.0	3,790	61,438	6.2	2,220	58,341	3.8
- Non-Hispanic Black	2,455	40,680	6.0	1,432	20,641	6.9	1,023	20,039	5.1
- Hispanic	2,715	58,244	4.7	1,637	29,495	5.6	1,078	28,749	3.7
- Others/Unknown	1,040	26,635	3.9	646	13,671	4.7	394	12,964	3.0
NYS Excluding NYC									
- All Races	6,498	127,308	5.1	4,088	65,078	6.3	2,410	62,230	3.9
- Non-Hispanic White	4,430	87,266	5.1	2,832	44,845	6.3	1,598	42,421	3.8
- Non-Hispanic Black	786	12,671	6.2	464	6,392	7.3	322	6,279	5.1
- Hispanic	919	19,713	4.7	572	9,967	5.7	347	9,746	3.6
- Others/Unknown	363	7,658	4.7	220	3,874	5.7	143	3,784	3.8
New York City									
- All Races	5,722	118,030	4.8	3,417	60,167	5.7	2,305	57,863	4.0
- Non-Hispanic White	1,580	32,513	4.9	958	16,593	5.8	622	15,920	3.9
- Non-Hispanic Black	1,669	28,009	6.0	968	14,249	6.8	701	13,760	5.1
- Hispanic	1,796	38,531	4.7	1,065	19,528	5.5	731	19,003	3.8
- Others/Unknown	677	18,977	3.6	426	9,797	4.3	251	9,180	2.7

Section 1 - Table 2
2007 Births - New York State Residents
Number of Major Malformations Per Child

Number of Malformations	Number of Children	Percent
1	9,584	78.4
2	1,607	13.2
3	553	4.5
4	233	1.9
5	118	1.0
6	61	0.5
7	29	0.2
8	17	0.1
9	10	0.1
10	5	*
11	2	*
12	1	*
All Children	12,220	100.0

* - Less than 0.05%
Note: Total percent may not add to 100% due to rounding

Section 1 - Table 3
2007 Births - New York State Residents
Percent of Live Births with One Major Malformation
Sex by Race/Ethnicity and Residence

	Both Sexes			Males			Females		
Race and Residence	Infants	Total Births	%	Infants	Total Births	%	Infants	Total Births	%
New York State									
- All Races	9,584	245,338	3.9	5,955	125,245	4.8	3,629	120,093	3.0
- Non-Hispanic White	4,758	119,779	4.0	3,053	61,438	5.0	1,705	58,341	2.9
- Non-Hispanic Black	1,920	40,680	4.7	1,130	20,641	5.5	790	20,039	3.9
- Hispanic	2,106	58,244	3.6	1,266	29,495	4.3	840	28,749	2.9
- Others/Unknown	800	26,635	3.0	506	13,671	3.7	294	12,964	2.3
NYS Excluding NYC									
- All Races	5,100	127,308	4.0	3,242	65,078	5.0	1,858	62,230	3.0
- Non-Hispanic White	3,508	87,266	4.0	2,279	44,845	5.1	1,229	42,421	2.9
- Non-Hispanic Black	602	12,671	4.8	348	6,392	5.4	254	6,279	4.0
- Hispanic	710	19,713	3.6	447	9,967	4.5	263	9,746	2.7
- Others/Unknown	280	7,658	3.7	168	3,874	4.3	112	3,784	3.0
New York City									
- All Races	4,484	118,030	3.8	2,713	60,167	4.5	1,771	57,863	3.1
- Non-Hispanic White	1,250	32,513	3.8	774	16,593	4.7	476	15,920	3.0
- Non-Hispanic Black	1,318	28,009	4.7	782	14,249	5.5	536	13,760	3.9
- Hispanic	1,396	38,531	3.6	819	19,528	4.2	577	19,003	3.0
- Others/Unknown	520	18,977	2.7	338	9,797	3.5	182	9,180	2.0

Section 1 - Table 4
2007 Births - New York State Residents
Percent of Live Births with Two or More Major Malformations
Sex by Race/Ethnicity and Residence

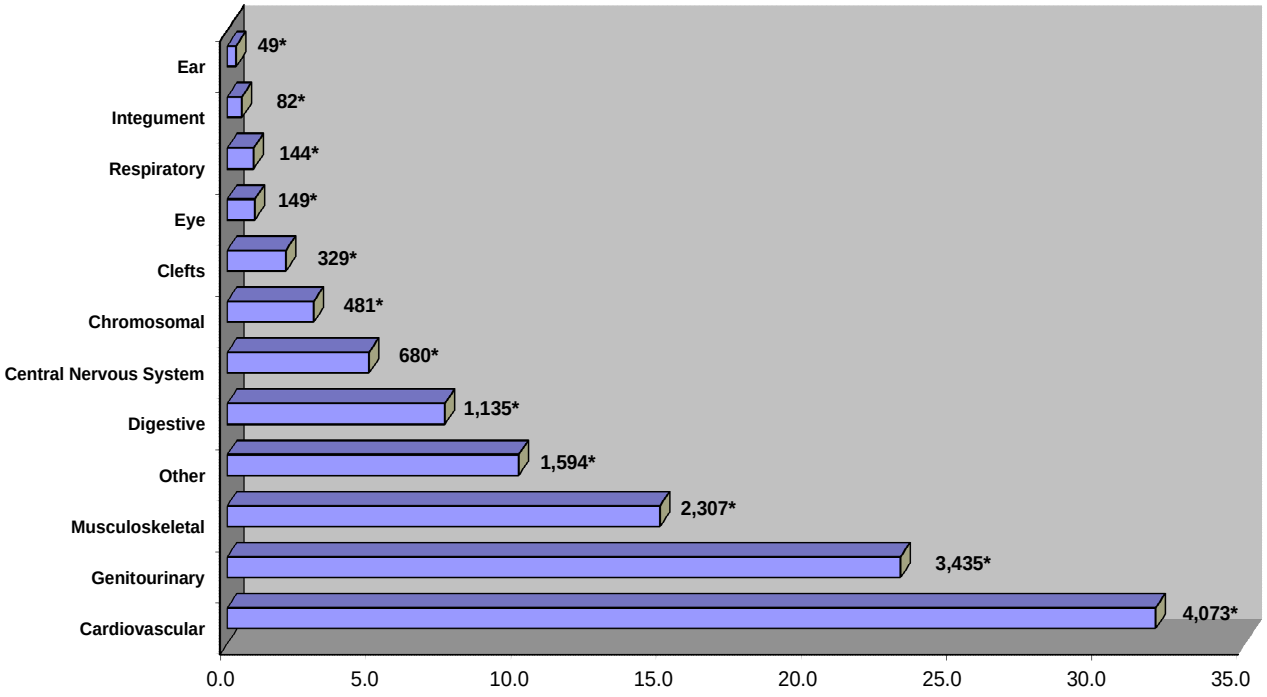
	Both Sexes			Males			Females		
Race and Residence	Infants	Total Births	%	Infants	Total Births	%	Infants	Total Births	%
New York State									
- All Races	2,636	245,338	1.1	1,550	125,245	1.2	1,086	120,093	0.9
- Non-Hispanic White	1,252	119,779	1.0	737	61,438	1.2	515	58,341	0.9
- Non-Hispanic Black	535	40,680	1.3	302	20,641	1.5	233	20,039	1.2
- Hispanic	609	58,244	1.0	371	29,495	1.3	238	28,749	0.8
- Others/Unknown	240	26,635	0.9	140	13,671	1.0	100	12,964	0.8
NYS Excluding NYC									
- All Races	1,398	127,308	1.1	846	65,078	1.3	552	62,230	0.9
- Non-Hispanic White	922	87,266	1.1	553	44,845	1.2	369	42,421	0.9
- Non-Hispanic Black	184	12,671	1.5	116	6,392	1.8	68	6,279	1.1
- Hispanic	209	19,713	1.1	125	9,967	1.3	84	9,746	0.9
- Others/Unknown	83	7,658	1.1	52	3,874	1.3	31	3,784	0.8
New York City									
- All Races	1,238	118,030	1.0	704	60,167	1.2	534	57,863	0.9
- Non-Hispanic White	330	32,513	1.0	184	16,593	1.1	146	15,920	0.9
- Non-Hispanic Black	351	28,009	1.3	186	14,249	1.3	165	13,760	1.2
- Hispanic	400	38,531	1.0	246	19,528	1.3	154	19,003	0.8
- Others/Unknown	157	18,977	0.8	88	9,797	0.9	69	9,180	0.8

Section II
Major Congenital Malformations by Organ System, 2007

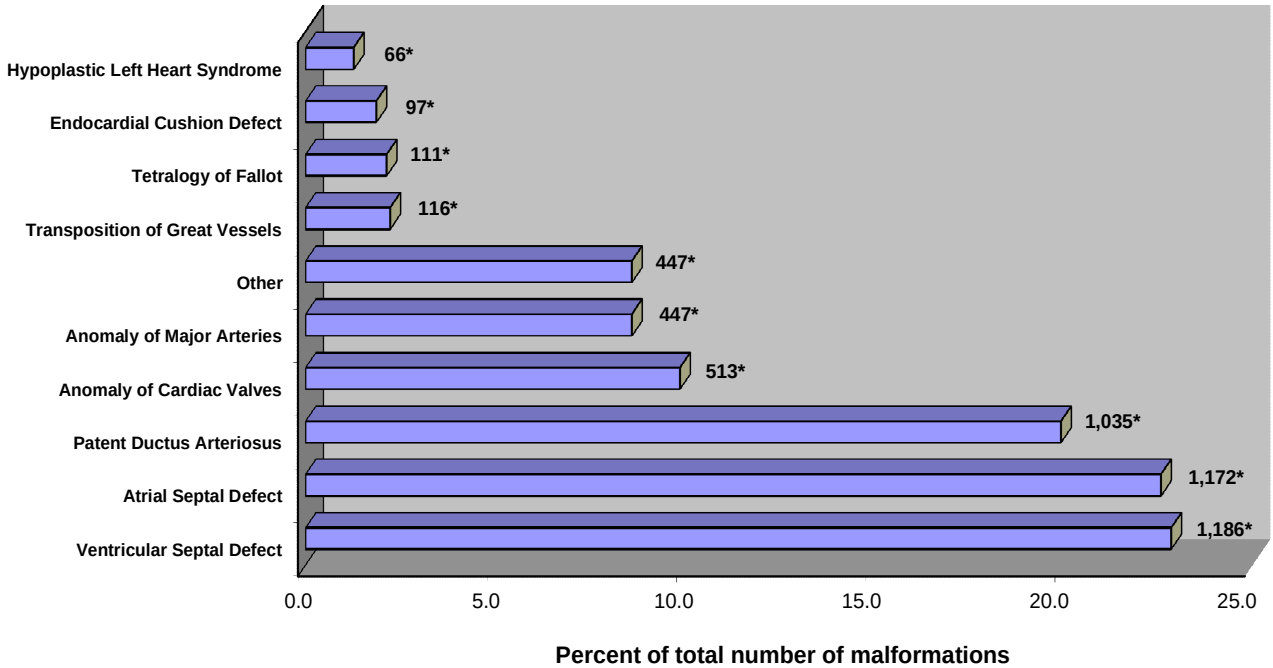
Introduction to Figures

The organ system figures in this section present the distribution of 12 categories of major malformations, the relative contribution of each category to overall prevalence 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.

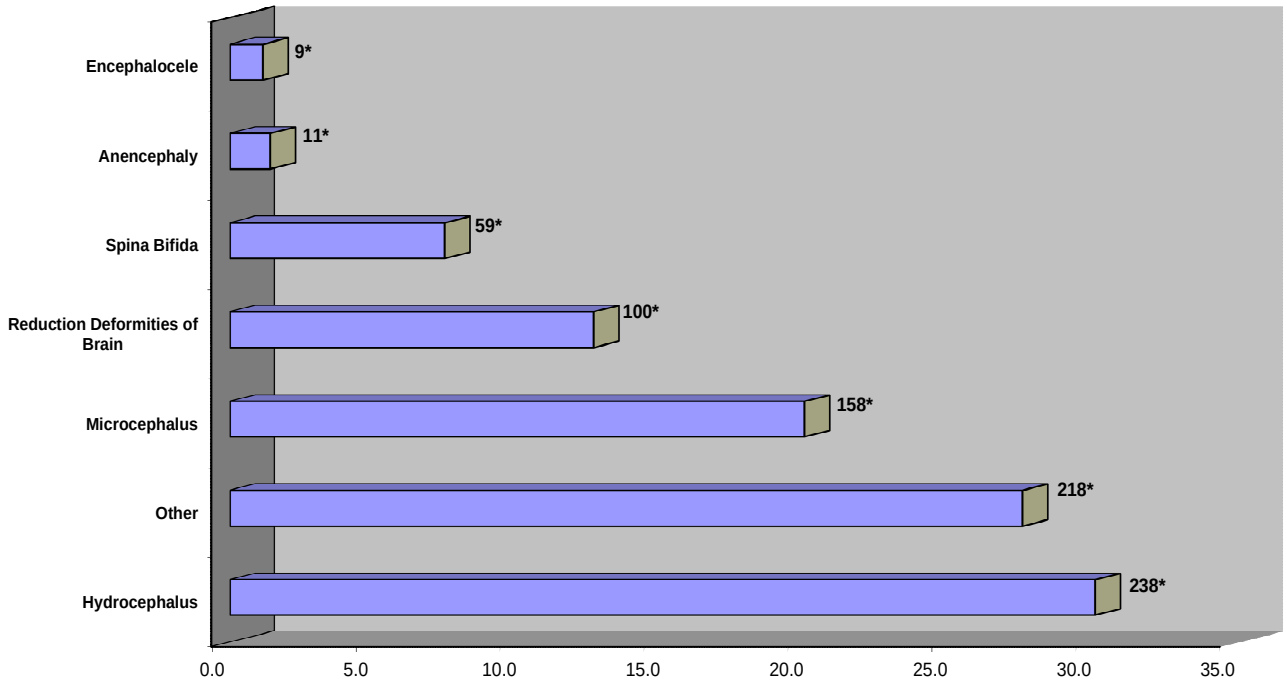
Major Malformations by Organ System
2007 Births - New York State Residents
(Number of Children = 12,220)
(* - Number of malformations in each organ system)



Major Malformations by Organ System
2007 Births - New York State Residents
Cardiovascular System Subset Category
(Number of Children = 4,073)
(* - Number of malformations in each category)

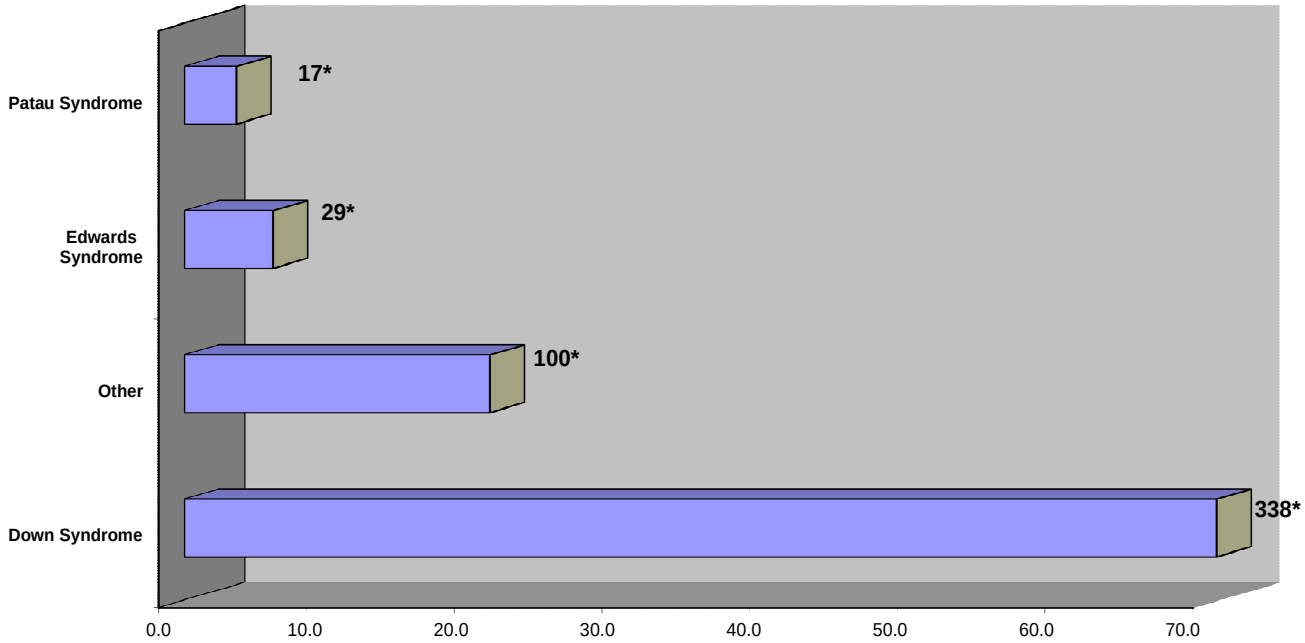


Major Malformations by Organ System
2007 Births - New York State Residents
Central Nervous System Subset Category
(Number of Children = 680)
(* - Number of malformations in each organ system)



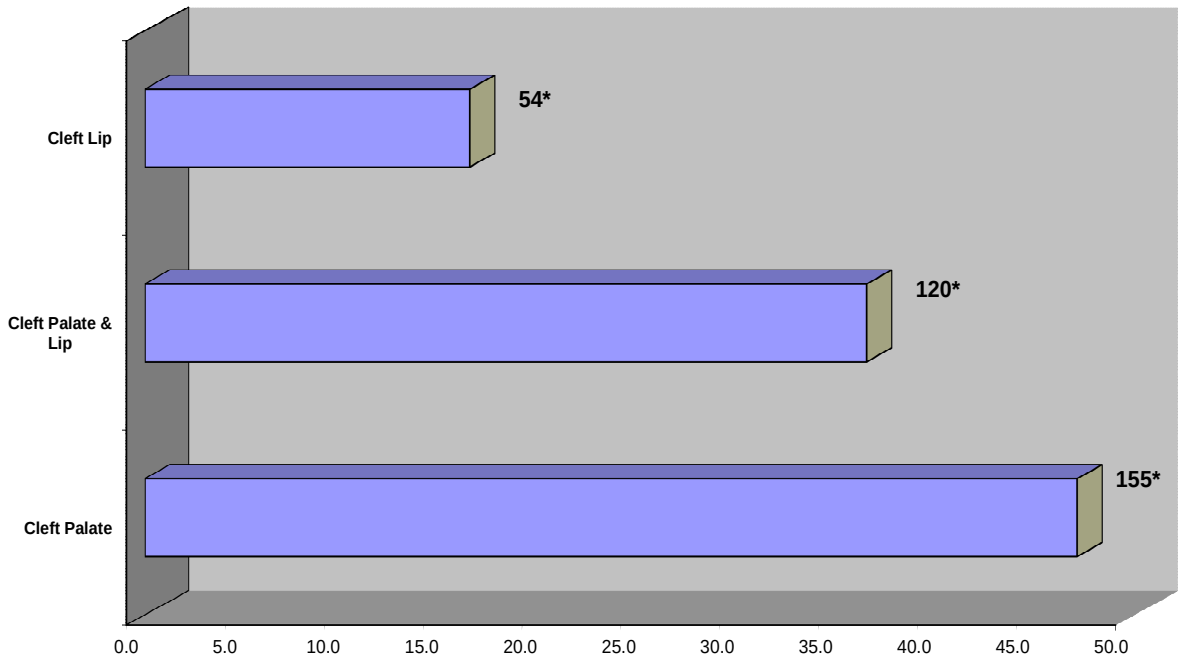
Percent of total number of malformations

Major Malformations by Organ System
2007 Births - New York State Residents
Chromosomal Subset Category
(Number of Children = 481)
(* - Number of malformations in each organ system)



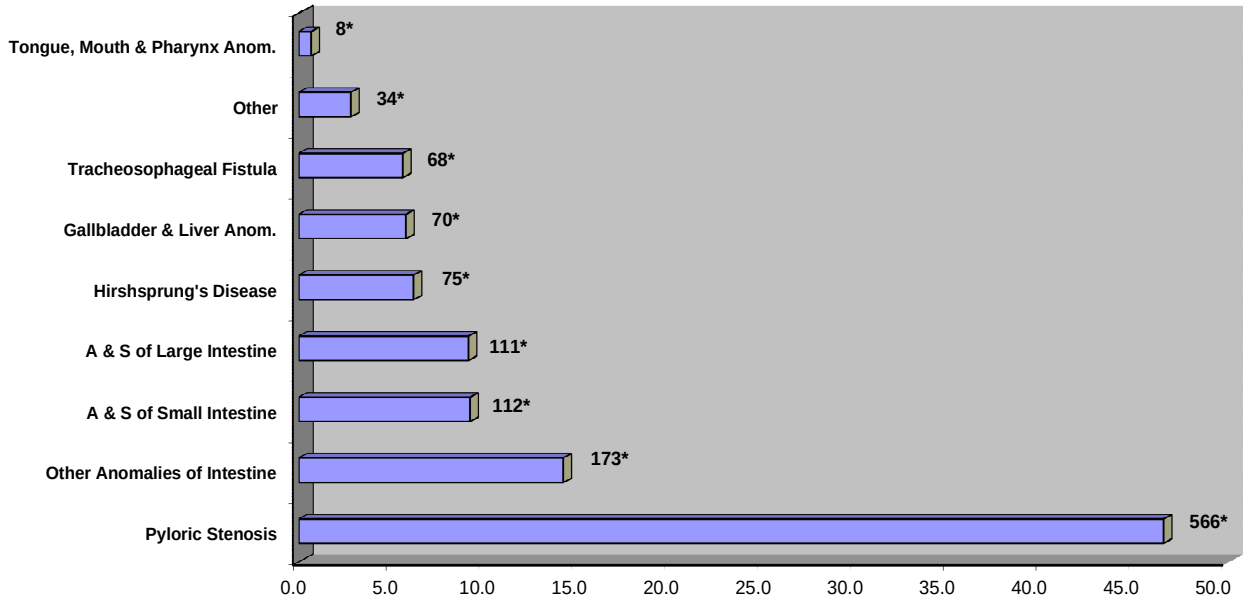
Percent of total number of malformations

Major Malformations by Organ System
2007 Births - New York State Residents
Oral Clefts Subset Category
(Number of Children = 329)
(* - Number of malformations in each organ system)



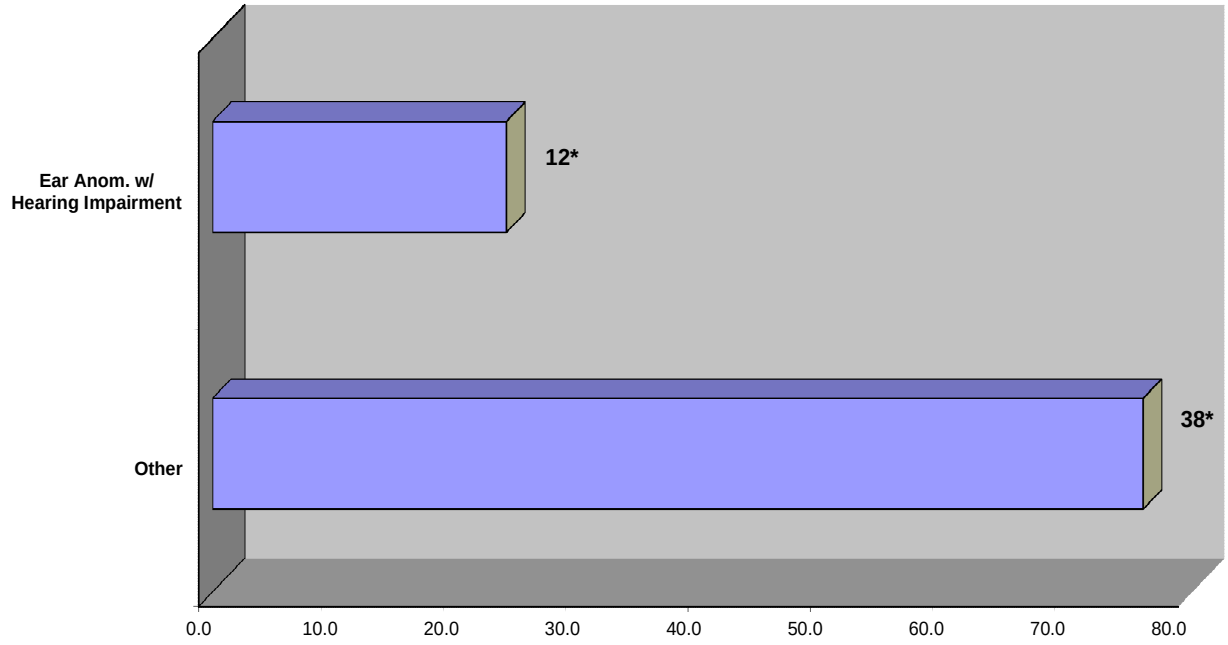
Percent of total number of malformations

Major Malformations by Organ System
2007 Births - New York State Residents
Digestive System Subset Category
(Number of Children = 1,135)
(* - Number of malformations in each organ system)

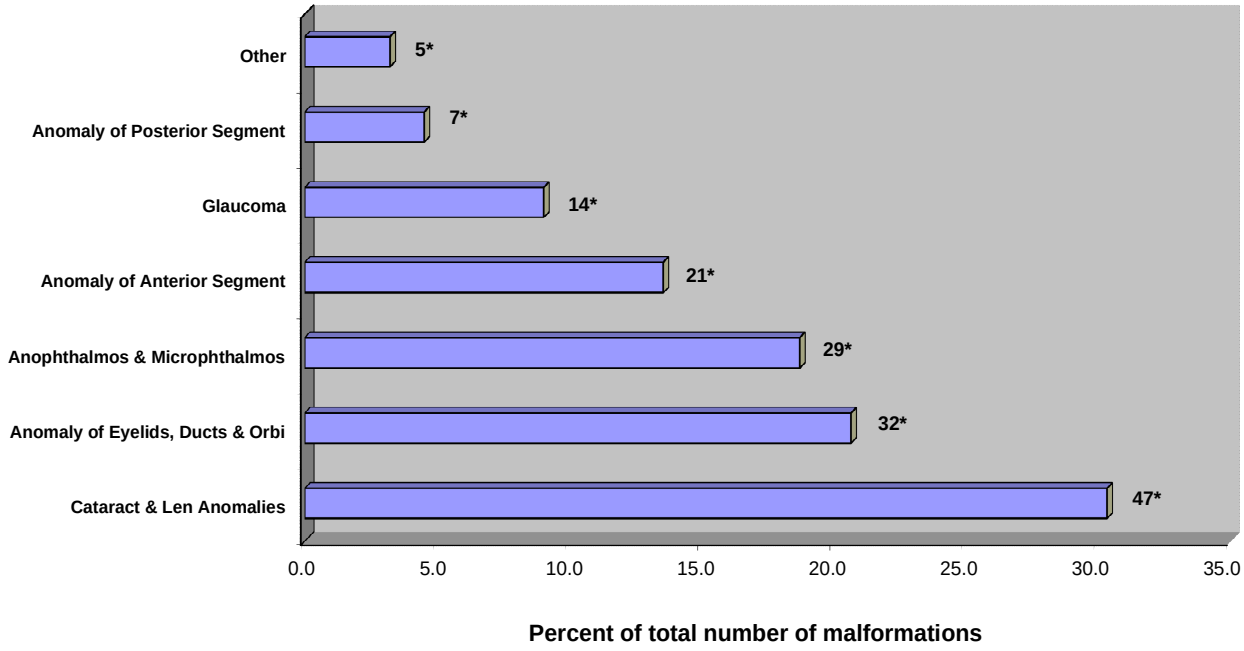


Percent of total number of malformations

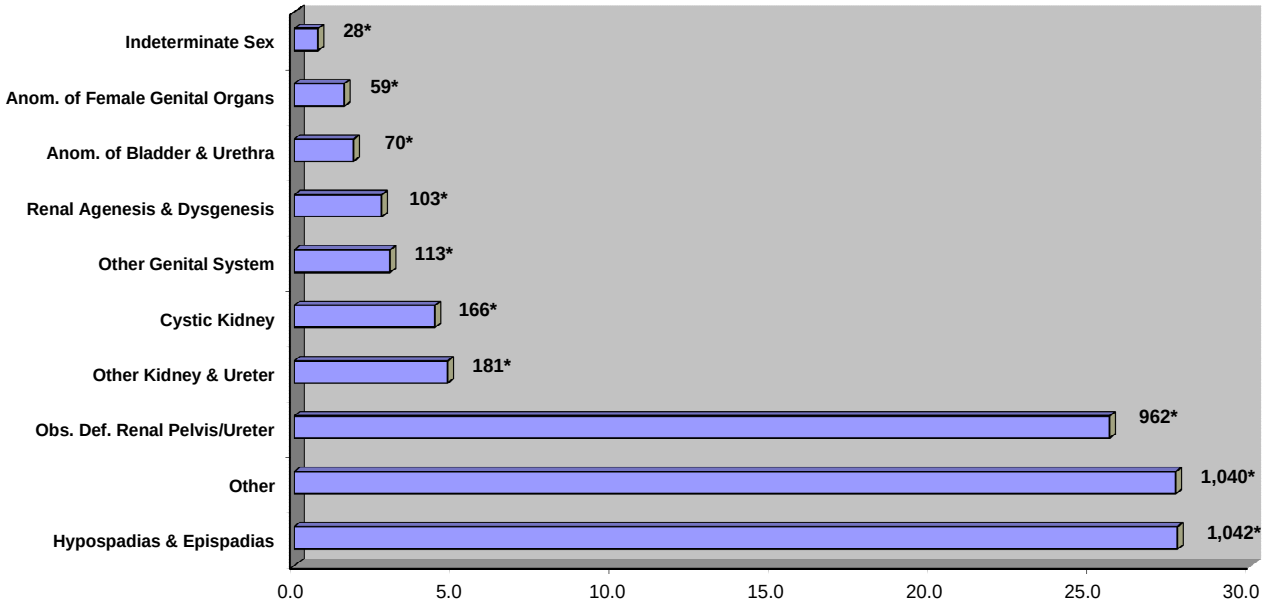
Major Malformations by Organ System
2007 Births - New York State Residents
Ear Subset Category
(Number of Children =49)
(* - Number of malformations in each organ system)



Major Malformations by Organ System
2007 Births - New York State Residents
Eye Subset Category
(Number of Children = 149)
(* - Number of malformations in each organ system)

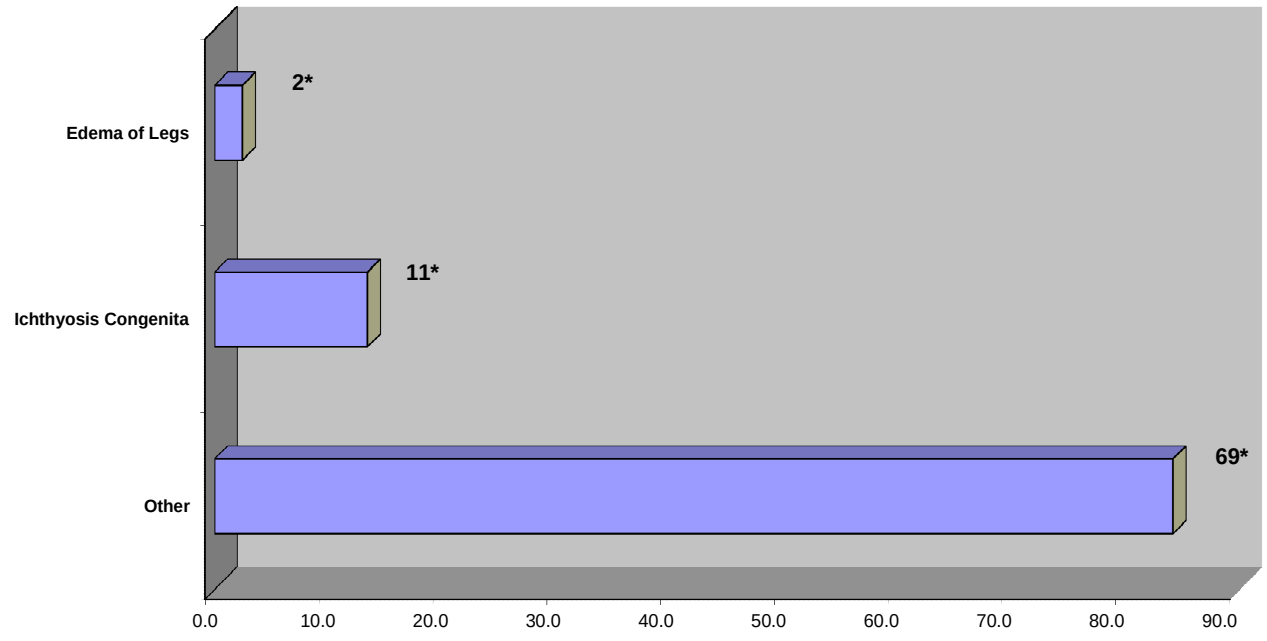


Major Malformations by Organ System
2007 Births - New York State Residents
Genitourinary System Subset Category
(Number of Children = 3,435)
(* - Number of malformations in each organ system)



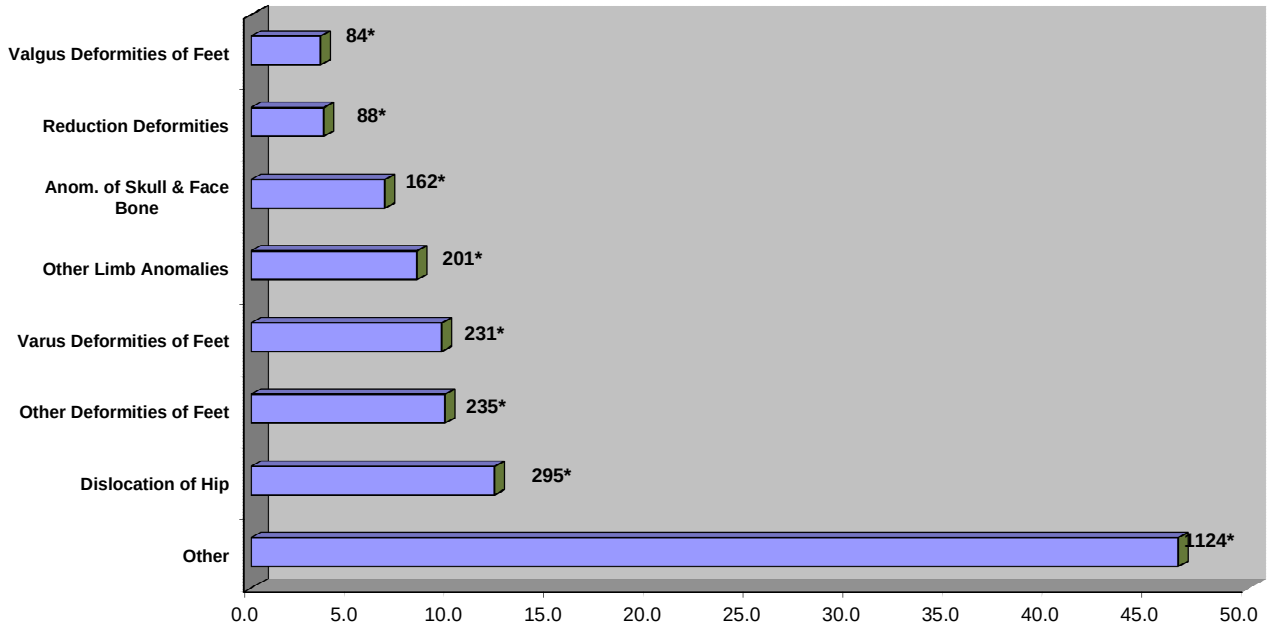
Percent of total number of malformations

Major Malformations by Organ System
2007 Births - New York State Residents
Integument System Subset Category
(Number of Children = 82)
(* - Number of malformations in each organ system)



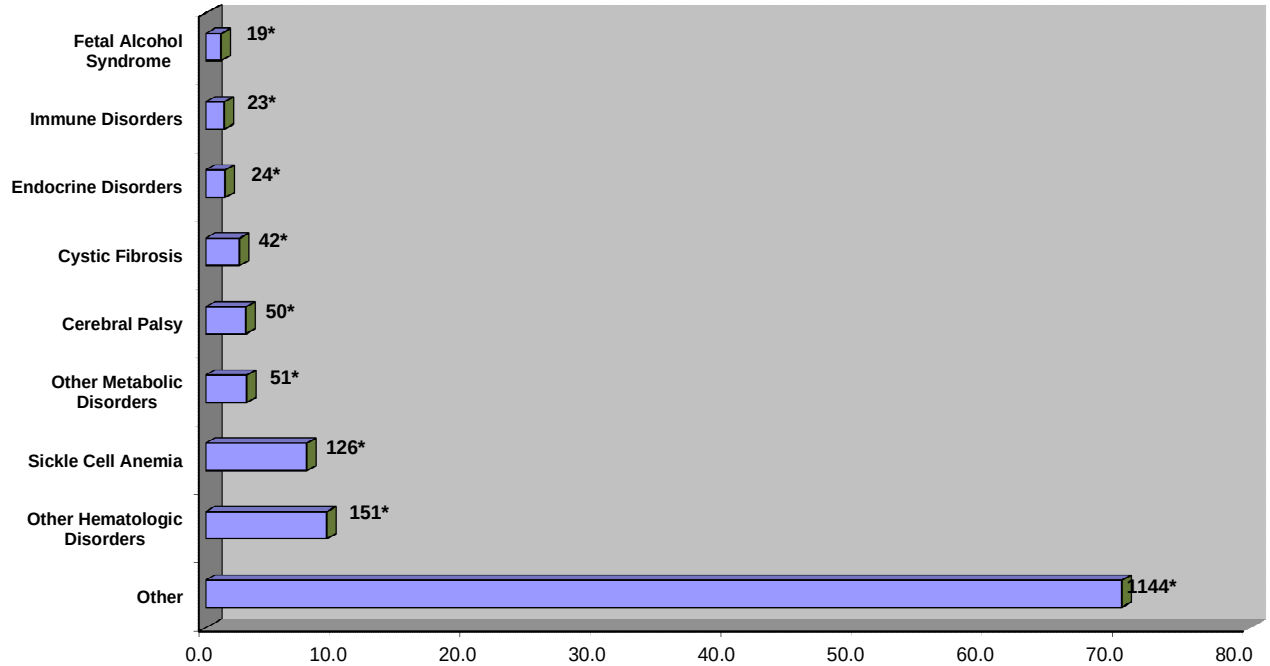
Percent of total number of malformations

Major Malformations by Organ System
2007 Births - New York State Residents
Musculoskeletal System Subset Category
(Number of Children =2,307)
(* - Number of malformations in each organ system)



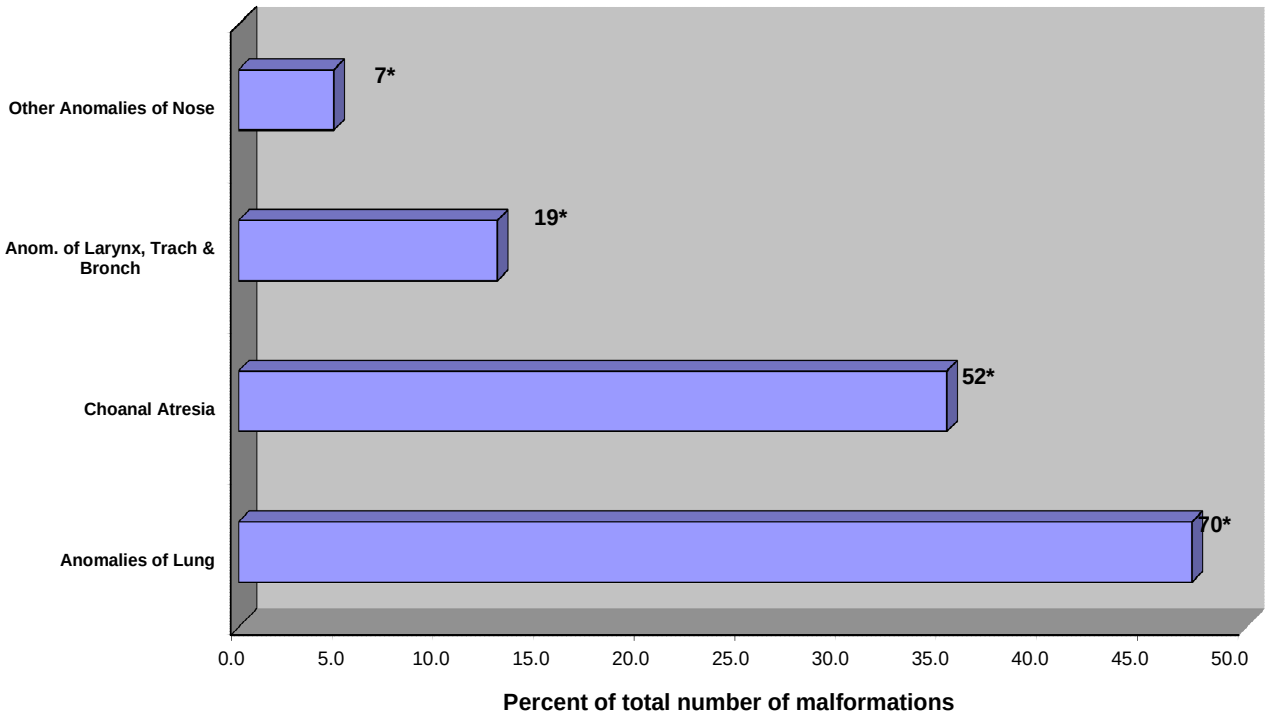
Percent of total number of malformations

Major Malformations by Organ System
2007 Births - New York State Residents
All Others Subset Category
(Number of Children = 1,594)
(* - Number of malformations in each organ system)



Percent of total number of malformations

Major Malformations by Organ System
2007 Births - New York State Residents
Respiratory System Subset Category
(Number of Children = 144)
(* - Number of malformations in each organ system)



Section III
Prevalence of Selected Malformations by Sex and Race/Ethnicity

Introduction to Table

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 birth weight from the birth certificate were not available. Birth weight 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 coding system. 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 Malformations
Prevalence per 10,000 Live Births by Sex and Race/Ethnicity

2007 Births– New York State Residents

ICD-9 Code	Malformation	Total Number	Total Prevalence	Male	Female	Ratio (M/F)	Non-Hispanic White	Non-Hispanic Black	His-panic	Other/ Unknown Race
243	Congenital hypothyroidism	114	4.6	4.7	4.6	1.0	4.5	6.6	3.8	4.1
270.1	Phenylketonuria	10	0.4	0.5	0.3	1.4	0.6	0.5	0.0	0.4
277.0	Cystic fibrosis	42	1.7	1.7	1.7	1.0	2.1	2.0	0.9	1.5
282.6	Sickle-cell anemia	126	5.1	5.4	4.8	1.1	0.4	26.3	1.7	1.5
740.0	Anencephalus	11	0.4	0.4	0.5	0.8	0.6	0.2	0.5	0.0
741.0	Spina bifida with hydrocephalus	25	1.0	1.0	1.1	0.9	1.1	1.0	1.0	0.8
741.9	Spina bifida without hydrocephalus	34	1.4	1.9	0.8	2.3	1.3	1.5	1.5	1.5
742.0	Encephalocele	9	0.4	0.3	0.4	0.8	0.6	0.5	0.0	0.0
742.1	Microcephalus	158	6.4	5.9	7.0	0.8	5.8	8.8	7.4	3.8
742.2	Agyria & lissencephaly	7	0.3	0.4	0.2	2.4	0.3	0.2	0.3	0.4
742.2	Anomalies of corpus callosum	56	2.3	2.7	1.8	1.5	2.5	2.2	2.2	1.5
742.2	Holoprosencephaly	14	0.6	0.4	0.7	0.5	0.8	0.2	0.5	0.0
742.3	Congenital hydrocephalus	238	9.7	12.0	7.3	1.6	8.5	14.3	10.0	7.5
742.4	Porencephaly	10	0.4	0.5	0.3	1.4	0.3	0.7	0.3	0.4
742.5	Congenital tethered cord	49	2.0	1.9	2.1	0.9	2.0	1.2	2.1	3.0
743.0	Anophthalmos	4	0.2	0.2	0.2	1.0	0.2	0.0	0.3	0.0
743.1	Microphthalmos	26	1.1	1.0	1.1	1.0	0.6	1.2	1.9	1.1
743.2	Glaucoma	14	0.6	0.6	0.5	1.3	0.8	0.7	0.3	0.0
743.3	Absence of lens	21	0.9	0.6	1.1	0.6	0.7	2.0	0.9	0.0
743.3	Congenital cataract	15	0.6	0.8	0.4	1.9	0.7	0.7	0.5	0.4
743.45	Aniridia	2	0.1	0.2	0.0	0.0	0.1	0.0	0.0	0.4
743.46	Coloboma of iris	2	0.1	0.2	0.0	0.0	0.1	0.0	0.0	0.4
744.0	Anotia/microtia	28	1.1	1.7	0.6	2.9	0.9	0.2	2.2	1.1
745.0	Common truncus	12	0.5	0.5	0.5	1.0	0.7	0.5	0.2	0.4
745.1	Transposition of great vessels	116	4.7	5.7	3.7	1.6	5.0	5.2	4.1	4.1
745.2	Tetralogy of Fallot	111	4.5	5.2	3.8	1.4	5.1	5.2	2.7	4.9
745.3	Common ventricle	12	0.5	0.2	0.7	0.3	0.5	0.7	0.5	0.0
745.4	Ventricular septal defect	1,186	48.3	43.4	53.5	0.8	48.2	46.5	53.4	40.9
745.5	Ostium secundum type atrial septal def.	1,172	47.8	47.9	47.6	1.0	39.2	67.8	49.6	51.8
745.6	Endocardial cushion defects	97	4.0	3.4	4.6	0.7	3.8	5.9	2.7	4.5
746.0	Atresia/stenosis of pulmonary valve	213	8.7	8.4	9.0	0.9	8.5	11.6	7.7	7.1
746.1	Tricuspid atresia/stenosis/hypoplasia	13	0.5	0.6	0.4	1.5	0.4	0.7	0.7	0.4
746.2	Ebstein's anomaly	12	0.5	0.6	0.3	1.9	0.5	0.5	0.5	0.4
746.3	Congenital stenosis of aortic valve	42	1.7	1.8	1.7	1.1	2.3	1.5	1.2	0.4
746.7	Hypoplastic left heart syndrome	66	2.7	2.8	2.6	1.1	2.7	3.2	3.1	1.1
746.85	Anomalies of coronary artery	12	0.5	0.5	0.5	1.0	0.5	0.7	0.3	0.4
747.0	Patent ductus arteriosis	1,035	42.2	45.9	38.3	1.2	39.8	66.6	33.8	33.8
747.10	Coartation of aorta	162	6.6	7.3	5.9	1.2	6.0	7.9	7.7	4.9
747.41	Total anomalous pulmonary venus connect.	22	0.9	1.1	0.7	1.7	0.5	2.0	1.0	0.8

2007 Births– New York State Residents (continued)

ICD-9 Code	Malformation	Total Number	Total Prevalence	Male	Female	Ratio (M/F)	Non-Hispanic White	Non-Hispanic Black	His-panic	Other/ Unknown Race
748.0	Choanal atresia	52	2.1	2.3	1.9	1.2	2.6	2.0	1.9	0.8
748.5	Agenesis/hypoplasia of lung	41	1.7	1.8	1.6	1.1	2.0	2.5	0.7	1.1
749.0	Cleft palate	155	6.3	5.4	7.2	0.7	7.0	5.4	5.8	5.6
749.1	Cleft lip	54	2.2	2.4	2.0	1.2	2.7	0.7	2.1	2.6
749.2	Cleft palate & lip	120	4.9	6.3	3.4	1.8	5.1	2.5	8.1	0.8
750.3	Tracheoesophageal fistula etc.	68	2.8	3.0	2.5	1.2	3.2	2.0	2.6	2.6
750.5	Congenital hypertrophic pyloric stenosis	566	23.1	37.8	7.7	4.9	27.2	13.8	24.9	14.6
751.1	Atresia and stenosis of small intestine	112	4.6	5.0	4.1	1.2	4.4	5.9	3.9	4.5
751.2	Atresia and stenosis of rectum or anus	111	4.5	5.9	3.1	1.9	4.2	2.7	5.3	7.1
751.3	Hirschsprungs disease	75	3.1	4.5	1.6	2.8	3.5	4.4	1.7	1.9
751.4	Anomalies of intestinal fixation	67	2.7	2.7	2.7	1.0	2.9	4.7	1.5	1.5
751.61	Biliary atresia	30	1.2	0.8	1.7	0.5	0.5	1.2	2.2	2.3
752.6	Epispadias	43	1.8	3.3	0.2	19.7	1.2	2.7	2.7	0.8
752.6	Hypospadias	967	39.4	77.0	0.2	308.1	53.2	34.9	21.3	24.0
753.0	Renal agenesis and dysgenesis	103	4.2	5.2	3.2	1.6	5.4	3.4	2.9	2.6
753.1	Cystic kidney disease	166	6.8	8.1	5.4	1.5	6.1	7.9	8.2	4.9
753.2	Obstructive defect renal pelvis & ureter	962	39.2	54.6	23.1	2.4	40.7	32.4	39.3	42.8
753.5	Extrophy of urinary bladder	5	0.2	0.2	0.2	1.4	0.3	0.2	0.0	0.0
753.6	Atresia & stenosis of urethra & bladder	43	1.8	3.4	0.1	40.3	1.9	1.7	1.4	1.9
754.3	Congenital dislocation of hip	212	8.6	3.6	13.9	0.3	9.9	4.4	10.1	6.4
754.51	Talipes equinovarus	140	5.7	7.5	3.8	2.0	5.9	5.9	6.2	3.4
755.2	Reduction deformities of upper limb	61	2.5	2.3	2.7	0.9	3.1	1.7	2.2	1.5
755.3	Reduction deformities of lower limb	29	1.2	0.9	1.5	0.6	1.3	1.0	1.0	1.5
755.8	Arthrogryposis multiplex congenita	10	0.4	0.4	0.4	1.0	0.3	0.7	0.3	0.4
756.0	Craniosynostosis	93	3.8	4.6	2.9	1.6	5.3	2.0	3.1	1.5
756.0	Goldenhar syndrome	2	0.1	0.0	0.2	0.0	0.2	0.0	0.0	0.0
756.4	Chonodrodystrophy	23	0.9	1.0	0.9	1.0	0.8	1.2	1.0	0.8
756.51	Osteogenesis imperfecta	14	0.6	0.5	0.7	0.7	0.7	0.7	0.5	0.0
756.6	Diaphragmatic hernia	67	2.7	3.0	2.5	1.2	3.5	1.5	1.9	3.0
756.7	Gastroschisis	58	2.4	2.3	2.4	1.0	2.7	2.5	2.2	1.1
756.7	Omphalocele	25	1.0	1.4	0.6	2.5	0.7	2.0	1.2	0.8
756.7	Prune belly	6	0.2	0.4	0.1	4.8	0.2	0.7	0.2	0.0
758.0	Down syndrome	338	13.8	14.1	13.4	1.1	14.6	12.8	14.9	9.0
758.1	Patau syndrome	17	0.7	0.7	0.7	1.1	0.7	0.5	1.0	0.4
758.2	Edwards syndrome	29	1.2	1.1	1.2	0.9	1.0	2.2	1.2	0.4
758.6	Gonadal dysgenesis	24	1.0	0.0	2.0	0.0	1.0	1.2	0.7	1.1
758.7	Klinefelter syndrome	9	0.4	0.7	0.0	0.0	0.2	0.7	0.5	0.4
759.3	Situs inversus	17	0.7	0.6	0.8	0.7	0.5	1.0	0.9	0.8
760.71	Fetal alcohol syndrome	19	0.8	0.7	0.8	0.9	0.8	2.2	0.2	0.0
762.8	Amniotic bands	15	0.6	0.6	0.6	1.1	0.5	0.5	1.2	0.0
771.1	Congenital cytomegalovirus infection	22	0.9	0.9	0.9	1.0	0.8	2.2	0.7	0.0
771.2	Other congenital infections	36	1.5	1.5	1.4	1.1	1.3	2.0	2.1	0.4

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 birth and four digit ICD-9-CM codes for major malformations. Certain codes for rare disorders and nonspecific codes are not included. Table 1 on the next page presents the number of children with major malformations by county and the percent of live births for comparison.

In table 2, the 10 most frequently reported codes for each county 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 county listings are not designed to be used for comparison among counties or for analytical studies. They are most useful to assist in county planning, education, counseling and other health care services programs.

Section IV – Table 1
2007 Births - New York State Residents
Children with Major Congenital Malformations & Percent of Live Births by County

County	Number of Children	Number of Live Births	Percent of Live Births
Albany	164	3,156	5.2
Allegany	22	505	4.4
Bronx	1,170	21,625	5.4
Broome	99	2,109	4.7
Cattaraugus	60	966	6.2
Cayuga	27	801	3.4
Chautauqua	92	1,434	6.4
Chemung	60	1,045	5.7
Chenango	18	547	3.3
Clinton	20	793	2.5
Columbia	28	548	5.1
Cortland	13	536	2.4
Delaware	20	453	4.4
Dutchess	147	2,936	5.0
Erie	665	10,031	6.6
Essex	15	283	5.3
Franklin	12	532	2.3
Fulton	29	580	5.0
Genesee	35	711	4.9
Greene	13	465	2.8
Hamilton	2	33	6.1
Herkimer	37	712	5.2
Jefferson	100	1,787	5.6
Kings	2,121	41,580	5.1
Lewis	9	291	3.1
Livingston	23	617	3.7
Madison	48	784	6.1
Monroe	387	8,677	4.5
Montgomery	36	632	5.7
Nassau	885	15,346	5.8
New York	877	20,517	4.3
Niagara	133	2,238	5.9
Oneida	152	2,595	5.9
Onondaga	299	5,516	5.4
Ontario	61	1,110	5.5
Orange	239	5,173	4.6
Orleans	21	439	4.8
Oswego	78	1,378	5.7
Otsego	27	550	4.9
Putnam	42	981	4.3
Queens	1,337	28,463	4.7
Rensselear	77	1,748	4.4
Richmond	217	5,845	3.7

Section IV – Table 1
2007 Births - New York State Residents
Children with Major Congenital Malformations & Percent of Live Births by County

County	Number of Children	Number of Live Births	Percent of Live Births
Rockland	198	4,414	4.5
Saratoga	90	2,333	3.9
Schenectady	96	1,846	5.2
Schoharie	14	290	4.8
Schuyler	5	175	2.9
Seneca	8	375	2.1
St Lawrence	69	1,214	5.7
Steuben	65	1,160	5.6
Suffolk	978	18,515	5.3
Sullivan	52	923	5.6
Tioga	9	367	2.5
Tompkins	27	943	2.9
Ulster	79	1,845	4.3
Warren	34	621	5.5
Washington	20	556	3.6
Wayne	45	1,080	4.2
Westchester	477	10,870	4.4
Wyoming	25	425	5.9
Yates	12	318	3.8

Section IV - Table 2
Most Frequently Reported Major Malformations by County

Birth Year: 2007

County	ICD-9 Code	Description	Number
ALBANY	752.6	Hypospadias & epispadias	15
	752.5	Undescended testicle	14
	745.5	Ostium secundum atrial septal defect	13
	745.4	Ventricular septal defect	12
	753.2	Obstructive defects of renal pelvis & ureter	12
	747.0	Patent ductus arteriosus	10
	755.0	Polydactyly	10
	750.5	Congenital hypertrophic pyloric stenosis	8
	747.3	Anomalies of pulmonary artery	5
	754.5	Varus deformities of feet	5
	754.7	Other deformities of feet	5
ALLEGANY	745.4	Ventricular septal defect	4
	752.6	Hypospadias & epispadias	3
	243.	Congenital hypothyroidism	2
	747.0	Patent ductus arteriosus	2
	749.0	Cleft palate	2
	752.5	Undescended testicle	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	755.6	Other anomalies of lower limb including pelvic girdle	2
	756.0	Anomalies of skull and face bones	2
	273.8	Other disorders of plasma protien	1
	277.0	Cystic fibrosis	1
	743.4	Coloboma & other anomalies of anterior segment	1
	745.1	Transposition of great vessels	1
	745.5	Ostium secundum atrial septal defect	1
	745.6	Endocardial cushion defects	1
	746.3	Congenital stenosis of aortic valve	1
	746.4	Congenital insufficiency of aortic valve	1
	746.6	Congenital mitral insufficiency	1
	746.7	Hypoplastic left heart syndrome	1
	747.1	Coarctation of aorta	1
	747.4	Anomalies of great veins	1
	748.0	Choanal atresia	1
	749.1	Cleft lip	1
	750.6	Congenital hiatus hernia	1
	751.5	Other anomalies of intestine	1
	755.0	Polydactyly	1
	756.6	Anomalies of diaphragm	1
	756.7	Anomalies of abdominal wall	1
	757.1	Ichthyosis congenita	1
	759.0	Anomalies of spleen	1
	759.8	Other specified anomalies	1
BRONX	755.0	Polydactyly	128
	745.5	Ostium secundum atrial septal defect	110
	752.5	Undescended testicle	96

Section IV - Table 2
Most Frequently Reported Major Malformations by County

Birth Year: 2007

County	ICD-9 Code	Description	Number
BRONX	753.2	Obstructive defects of renal pelvis & ureter	96
	752.6	Hypospadias & epispadias	95
	745.4	Ventricular septal defect	90
	747.0	Patent ductus arteriosus	54
	754.5	Varus deformities of feet	41
	282.6	Sickle-cell anemia	34
	750.5	Congenital hypertrophic pyloric stenosis	34
BROOME	745.4	Ventricular septal defect	10
	750.5	Congenital hypertrophic pyloric stenosis	10
	752.6	Hypospadias & epispadias	10
	752.5	Undescended testicle	9
	753.2	Obstructive defects of renal pelvis & ureter	8
	755.0	Polydactyly	6
	747.0	Patent ductus arteriosus	5
	747.1	Coarctation of aorta	4
	742.3	Congenital hydrocephalus	3
	754.5	Varus deformities of feet	3
	756.7	Anomalies of abdominal wall	3
CATTARAUGUS	745.4	Ventricular septal defect	7
	747.0	Patent ductus arteriosus	7
	752.6	Hypospadias & epispadias	6
	750.5	Congenital hypertrophic pyloric stenosis	4
	752.5	Undescended testicle	4
	753.2	Obstructive defects of renal pelvis & ureter	4
	755.0	Polydactyly	3
	273.8	Other disorders of plasma protien	2
	742.3	Congenital hydrocephalus	2
	742.4	Other specified anomalies of brain	2
	745.2	Tetralogy of Fallot	2
	746.8	Other specified anomalies of heart	2
	749.2	Cleft palate with cleft lip	2
	754.5	Varus deformities of feet	2
	754.7	Other deformities of feet	2
	756.0	Anomalies of skull and face bones	2
	756.7	Anomalies of abdominal wall	2
	758.0	Down syndrome	2
CAYUGA	752.6	Hypospadias & epispadias	5
	745.5	Ostium secundum atrial septal defect	2
	746.6	Congenital mitral insufficiency	2
	747.0	Patent ductus arteriosus	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	751.3	Hirschprung's disease & other functional disorders of colon	2
	752.5	Undescended testicle	2
	756.0	Anomalies of skull and face bones	2

Section IV - Table 2
Most Frequently Reported Major Malformations by County

Birth Year: 2007

County	ICD-9 Code	Description	Number
CAYUGA	758.0	Down syndrome	2
	228.0	Hemangioma, any site	1
	243.	Congenital hypothyroidism	1
	253.2	Panhypopituitarism	1
	343.9	Infantile cerebral palsy unspecified	1
	524.0	Major anomalies of jaw size	1
	742.2	Reduction deformities of brain	1
	742.3	Congenital hydrocephalus	1
	745.2	Tetralogy of Fallot	1
	745.4	Ventricular septal defect	1
	746.3	Congenital stenosis of aortic valve	1
	746.7	Hypoplastic left heart syndrome	1
	746.8	Other specified anomalies of heart	1
	747.1	Coarctation of aorta	1
	751.1	Atresia & stenosis of small intestine	1
	751.4	Anomalies of intestinal fixation	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	754.2	Deformities of spine	1
	756.1	Anomalies of spine	1
	756.7	Anomalies of abdominal wall	1
	757.3	Other specified anomalies of skin	1
CHAUTAUQUA	745.5	Ostium secundum atrial septal defect	25
	750.5	Congenital hypertrophic pyloric stenosis	10
	747.0	Patent ductus arteriosus	9
	745.4	Ventricular septal defect	6
	752.5	Undescended testicle	6
	746.8	Other specified anomalies of heart	4
	752.6	Hypospadias & epispadias	4
	754.7	Other deformities of feet	4
	333.2	Myoclonus	3
	742.1	Microcephalus	3
	742.4	Other specified anomalies of brain	3
	746.6	Congenital mitral insufficiency	3
	755.2	Reduction deformities of upper limb	3
	755.6	Other anomalies of lower limb including pelvic girdle	3
	758.0	Down syndrome	3
CHEMUNG	752.5	Undescended testicle	9
	753.2	Obstructive defects of renal pelvis & ureter	8
	750.5	Congenital hypertrophic pyloric stenosis	6
	752.6	Hypospadias & epispadias	6
	746.8	Other specified anomalies of heart	4
	754.3	Congenital dislocation of hip	4
	755.0	Polydactyly	4
	742.3	Congenital hydrocephalus	3
	754.5	Varus deformities of feet	3
	756.0	Anomalies of skull and face bones	3

Section IV - Table 2
Most Frequently Reported Major Malformations by County

Birth Year: 2007

County	ICD-9 Code	Description	Number
CHENANGO	752.6	Hypospadias & epispadias	3
	426.7	Anamalous atrioventricular excitation	2
	745.4	Ventricular septal defect	2
	754.5	Varus deformities of feet	2
	754.7	Other deformities of feet	2
	755.0	Polydactyly	2
	745.5	Ostium secundum atrial septal defect	1
	746.8	Other specified anomalies of heart	1
	747.0	Patent ductus arteriosus	1
	747.1	Coarctation of aorta	1
	748.1	Other anomalies of nose	1
	749.2	Cleft palate with cleft lip	1
	752.8	Other specified anomalies of genital organs	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	755.6	Other anomalies of lower limb including pelvic girdle	1
	756.7	Anomalies of abdominal wall	1
	758.0	Down syndrome	1
CLINTON	752.6	Hypospadias & epispadias	5
	753.2	Obstructive defects of renal pelvis & ureter	3
	745.5	Ostium secundum atrial septal defect	2
	751.5	Other anomalies of intestine	2
	742.3	Congenital hydrocephalus	1
	745.1	Transposition of great vessels	1
	745.4	Ventricular septal defect	1
	745.8	Other defect of septal closure	1
	747.0	Patent ductus arteriosus	1
	747.3	Anomalies of pulmonary artery	1
	747.4	Anomalies of great veins	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	1
	751.6	Anomalies of gallbladder, bile ducts, and liver	1
	752.5	Undescended testicle	1
	752.8	Other specified anomalies of genital organs	1
	753.1	Cystic kidney disease	1
	757.3	Other specified anomalies of skin	1
	758.5	Other conditions due autosomal anomalies	1
COLUMBIA	750.5	Congenital hypertrophic pyloric stenosis	7
	753.2	Obstructive defects of renal pelvis & ureter	5
	752.6	Hypospadias & epispadias	4
	746.8	Other specified anomalies of heart	3
	752.5	Undescended testicle	3
	745.5	Ostium secundum atrial septal defect	2
	746.0	Anomalies of pulmonary valve	2

Section IV - Table 2
Most Frequently Reported Major Malformations by County

Birth Year: 2007

County	ICD-9 Code	Description	Number
COLUMBIA	747.0	Patent ductus arteriosus	2
	228.0	Hemangioma, any site	1
	255.2	Adrenogenital disorders	1
	745.4	Ventricular septal defect	1
	746.4	Congenital insufficiency of aortic valve	1
	747.3	Anomalies of pulmonary artery	1
	747.4	Anomalies of great veins	1
	752.8	Other specified anomalies of genital organs	1
	753.0	Renal agenesis & dysgenesis	1
	753.4	Other specified anomalies of ureter	1
	754.1	Deformities of sternocleidomastoid muscle	1
	754.5	Varus deformities of feet	1
	756.0	Anomalies of skull and face bones	1
	756.7	Anomalies of abdominal wall	1
	759.8	Other specified anomalies	1
CORTLAND	752.5	Undescended testicle	3
	747.0	Patent ductus arteriosus	2
	747.1	Coarctation of aorta	2
	745.4	Ventricular septal defect	1
	745.5	Ostium secundum atrial septal defect	1
	746.8	Other specified anomalies of heart	1
	748.3	Other anomalies of larynx, trachea, & bronchus	1
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	1
	752.0	Anomalies of ovaries	1
	752.6	Hypospadias & epispadias	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	758.0	Down syndrome	1
	758.1	Patau syndrome	1
	759.5	Tuberous sclerosis	1
DELAWARE	747.0	Patent ductus arteriosus	4
	746.8	Other specified anomalies of heart	3
	745.4	Ventricular septal defect	2
	746.0	Anomalies of pulmonary valve	2
	752.6	Hypospadias & epispadias	2
	277.0	Cystic fibrosis	1
	524.0	Major anomalies of jaw size	1
	743.4	Coloboma & other anomalies of anterior segment	1
	743.6	Congenital anomalies of eyelids, lacrimal system & orbit	1
	746.6	Congenital mitral insufficiency	1
	746.7	Hypoplastic left heart syndrome	1
	749.2	Cleft palate with cleft lip	1
	752.5	Undescended testicle	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	754.3	Congenital dislocation of hip	1
	757.3	Other specified anomalies of skin	1

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Most Frequently Reported Major Malformations by County

Birth Year: 2007

County	ICD-9 Code	Description	Number
DUTCHESS	752.6	Hypospadias & epispadias	19
	753.2	Obstructive defects of renal pelvis & ureter	18
	745.4	Ventricular septal defect	15
	745.5	Ostium secundum atrial septal defect	13
	752.5	Undescended testicle	13
	750.5	Congenital hypertrophic pyloric stenosis	8
	749.0	Cleft palate	5
	753.1	Cystic kidney disease	5
	754.3	Congenital dislocation of hip	5
	754.5	Varus deformities of feet	5
	758.0	Down syndrome	5
ERIE	747.0	Patent ductus arteriosus	73
	745.4	Ventricular septal defect	60
	752.5	Undescended testicle	57
	752.6	Hypospadias & epispadias	45
	755.0	Polydactyly	44
	745.5	Ostium secundum atrial septal defect	38
	750.5	Congenital hypertrophic pyloric stenosis	27
	747.3	Anomalies of pulmonary artery	25
	753.2	Obstructive defects of renal pelvis & ureter	24
	746.8	Other specified anomalies of heart	20
	758.0	Down syndrome	20
ESSEX	752.6	Hypospadias & epispadias	3
	753.2	Obstructive defects of renal pelvis & ureter	3
	745.4	Ventricular septal defect	2
	745.6	Endocardial cushion defects	1
	746.6	Congenital mitral insufficiency	1
	747.0	Patent ductus arteriosus	1
	747.1	Coarctation of aorta	1
	749.0	Cleft palate	1
	751.6	Anomalies of gallbladder, bile ducts, and liver	1
	753.0	Renal agenesis & dysgenesis	1
	754.5	Varus deformities of feet	1
	755.0	Polydactyly	1
	759.8	Other specified anomalies	1
FRANKLIN	752.6	Hypospadias & epispadias	4
	750.5	Congenital hypertrophic pyloric stenosis	2
	748.0	Choanal atresia	1
	749.0	Cleft palate	1
	749.2	Cleft palate with cleft lip	1
	753.0	Renal agenesis & dysgenesis	1
	754.7	Other deformities of feet	1
	755.2	Reduction deformities of upper limb	1

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Most Frequently Reported Major Malformations by County

Birth Year: 2007

County	ICD-9 Code	Description	Number
FULTON	745.4	Ventricular septal defect	5
	752.6	Hypospadias & epispadias	5
	750.5	Congenital hypertrophic pyloric stenosis	3
	753.2	Obstructive defects of renal pelvis & ureter	3
	255.2	Adrenogenital disorders	1
	359.0	Congenital Hereditary Muscular Dystrophy	1
	742.4	Other specified anomalies of brain	1
	745.1	Transposition of great vessels	1
	745.3	Common ventricle	1
	746.0	Anomalies of pulmonary valve	1
	746.8	Other specified anomalies of heart	1
	747.0	Patent ductus arteriosus	1
	747.1	Coarctation of aorta	1
	748.0	Choanal atresia	1
	749.2	Cleft palate with cleft lip	1
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	1
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	1
	751.3	Hirschprung's disease & other functional disorders of colon	1
	751.5	Other anomalies of intestine	1
	753.3	Other specified anomalies of kidney	1
	753.8	Other specified anomalies of bladder & urethra	1
	754.5	Varus deformities of feet	1
	756.3	Other anomalies of ribs and sternum	1
	758.0	Down syndrome	1
	759.8	Other specified anomalies	1
GENESEE	752.5	Undescended testicle	5
	745.4	Ventricular septal defect	4
	747.0	Patent ductus arteriosus	4
	742.4	Other specified anomalies of brain	3
	750.5	Congenital hypertrophic pyloric stenosis	3
	243.	Congenital hypothyroidism	2
	758.0	Down syndrome	2
	255.2	Adrenogenital disorders	1
	742.3	Congenital hydrocephalus	1
	744.0	Anomalies of ear causing impairment of hearing	1
	746.8	Other specified anomalies of heart	1
	747.2	Other anomalies of aorta	1
	748.3	Other anomalies of larynx, trachea, & bronchus	1
	749.2	Cleft palate with cleft lip	1
	752.6	Hypospadias & epispadias	1
	753.1	Cystic kidney disease	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	753.7	Anomalies of urachus	1
	754.3	Congenital dislocation of hip	1
	754.6	Valgus deformities of feet	1
	756.4	Chondrodystrophy	1
	756.5	Osteodystrophies	1
	757.1	Ichthyosis congenita	1
	757.3	Other specified anomalies of skin	1

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Birth Year: 2007

County	ICD-9 Code	Description	Number
GREENE	752.5	Undescended testicle	4
	750.5	Congenital hypertrophic pyloric stenosis	2
	752.6	Hypospadias & epispadias	2
	743.4	Coloboma & other anomalies of anterior segment	1
	745.2	Tetralogy of Fallot	1
	747.0	Patent ductus arteriosus	1
	755.1	Syndactyly	1
	756.3	Other anomalies of ribs and sternum	1
	759.8	Other specified anomalies	1
	771.1	Congenital cytomegalovirus infection	1
HAMILTON	333.2	Myoclonus	1
	752.5	Undescended testicle	1
HERKIMER	752.5	Undescended testicle	4
	745.4	Ventricular septal defect	3
	747.0	Patent ductus arteriosus	3
	277.0	Cystic fibrosis	2
	745.5	Ostium secundum atrial septal defect	2
	749.0	Cleft palate	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	751.1	Atresia & stenosis of small intestine	2
	752.6	Hypospadias & epispadias	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	754.3	Congenital dislocation of hip	2
	758.0	Down syndrome	2
JEFFERSON	752.6	Hypospadias & epispadias	10
	750.5	Congenital hypertrophic pyloric stenosis	9
	754.5	Varus deformities of feet	9
	752.5	Undescended testicle	8
	753.2	Obstructive defects of renal pelvis & ureter	7
	754.3	Congenital dislocation of hip	6
	745.4	Ventricular septal defect	5
	745.5	Ostium secundum atrial septal defect	5
	747.0	Patent ductus arteriosus	4
	749.2	Cleft palate with cleft lip	4
KINGS	747.0	Patent ductus arteriosus	308
	745.5	Ostium secundum atrial septal defect	284
	745.4	Ventricular septal defect	215
	752.5	Undescended testicle	172
	755.0	Polydactyly	161

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Birth Year: 2007

County	ICD-9 Code	Description	Number
KINGS	753.2	Obstructive defects of renal pelvis & ureter	146
	752.6	Hypospadias & epispadias	125
	746.8	Other specified anomalies of heart	83
	750.5	Congenital hypertrophic pyloric stenosis	71
	758.0	Down syndrome	58
LEWIS	524.0	Major anomalies of jaw size	2
	745.4	Ventricular septal defect	2
	749.0	Cleft palate	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	228.1	Lymphangioma, any site	1
	282.0	Hereditary spherocytosis	1
	745.5	Ostium secundum atrial septal defect	1
	752.5	Undescended testicle	1
	754.7	Other deformities of feet	1
	756.0	Anomalies of skull and face bones	1
	758.0	Down syndrome	1
	759.8	Other specified anomalies	1
LIVINGSTON	747.0	Patent ductus arteriosus	3
	752.5	Undescended testicle	3
	745.4	Ventricular septal defect	2
	749.0	Cleft palate	2
	752.6	Hypospadias & epispadias	2
	190.5	Malignant neoplasm of the retina	1
	524.0	Major anomalies of jaw size	1
	745.2	Tetralogy of Fallot	1
	747.2	Other anomalies of aorta	1
	748.0	Choanal atresia	1
	748.5	Agenesis, hypoplasia & dysplasia, lung	1
	749.1	Cleft lip	1
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	1
	752.0	Anomalies of ovaries	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	754.5	Varus deformities of feet	1
	756.0	Anomalies of skull and face bones	1
	758.0	Down syndrome	1
	758.5	Other conditions due autosomal anomalies	1
MADISON	752.6	Hypospadias & epispadias	6
	753.2	Obstructive defects of renal pelvis & ureter	5
	745.4	Ventricular septal defect	3
	747.0	Patent ductus arteriosus	3
	752.5	Undescended testicle	3
	750.5	Congenital hypertrophic pyloric stenosis	2

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Birth Year: 2007

County	ICD-9 Code	Description	Number
MADISON	752.4	Anomalies of cervix, vagina & external female genitalia	2
	752.8	Other specified anomalies of genital organs	2
	753.3	Other specified anomalies of kidney	2
	754.3	Congenital dislocation of hip	2
MONROE	753.2	Obstructive defects of renal pelvis & ureter	50
	752.6	Hypospadias & epispadias	46
	752.5	Undescended testicle	32
	755.0	Polydactyly	21
	745.4	Ventricular septal defect	19
	745.5	Ostium secundum atrial septal defect	14
	747.0	Patent ductus arteriosus	14
	742.3	Congenital hydrocephalus	11
	750.5	Congenital hypertrophic pyloric stenosis	11
	755.6	Other anomalies of lower limb including pelvic girdle	11
	758.0	Down syndrome	11
MONTGOMERY	752.6	Hypospadias & epispadias	5
	750.5	Congenital hypertrophic pyloric stenosis	4
	752.5	Undescended testicle	3
	755.0	Polydactyly	3
	742.4	Other specified anomalies of brain	2
	745.4	Ventricular septal defect	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	524.0	Major anomalies of jaw size	1
	740.0	Anencephalus	1
	742.1	Microcephalus	1
	742.3	Congenital hydrocephalus	1
	742.5	Other specified anomalies of spinal cord	1
	743.1	Microphthalmos	1
	745.1	Transposition of great vessels	1
	745.2	Tetralogy of Fallot	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
	747.0	Patent ductus arteriosus	1
	750.6	Congenital hiatus hernia	1
	751.3	Hirschprung's disease & other functional disorders of colon	1
	751.4	Anomalies of intestinal fixation	1
	752.0	Anomalies of ovaries	1
	752.4	Anomalies of cervix, vagina & external female genitalia	1
	753.1	Cystic kidney disease	1
	754.5	Varus deformities of feet	1
	754.6	Valgus deformities of feet	1
	754.7	Other deformities of feet	1
	754.8	Other specified nonteratogenic anomalies	1

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Birth Year: 2007

County	ICD-9 Code	Description	Number
MONTGOMERY	755.2	Reduction deformities of upper limb	1
	755.5	Other anomalies of upper limb including shoulder girdle	1
	755.6	Other anomalies of lower limb including pelvic girdle	1
	756.4	Chondrodystrophy	1
	756.6	Anomalies of diaphragm	1
	758.5	Other conditions due autosomal anomalies	1
	758.8	Other conditions due to sex chromosome anomalies	1
	759.0	Anomalies of spleen	1
	762.8	Amniotic bands	1
NASSAU	747.0	Patent ductus arteriosus	121
	745.4	Ventricular septal defect	99
	753.2	Obstructive defects of renal pelvis & ureter	92
	752.6	Hypospadias & epispadias	85
	752.5	Undescended testicle	59
	745.5	Ostium secundum atrial septal defect	47
	750.5	Congenital hypertrophic pyloric stenosis	37
	752.8	Other specified anomalies of genital organs	34
	755.0	Polydactyly	33
	746.8	Other specified anomalies of heart	22
NEW YORK	745.5	Ostium secundum atrial septal defect	125
	745.4	Ventricular septal defect	115
	752.5	Undescended testicle	101
	747.0	Patent ductus arteriosus	77
	752.6	Hypospadias & epispadias	66
	753.2	Obstructive defects of renal pelvis & ureter	64
	755.0	Polydactyly	52
	754.3	Congenital dislocation of hip	35
	746.8	Other specified anomalies of heart	29
	746.0	Anomalies of pulmonary valve	22
NIAGARA	745.4	Ventricular septal defect	16
	752.5	Undescended testicle	14
	752.6	Hypospadias & epispadias	11
	745.5	Ostium secundum atrial septal defect	10
	747.0	Patent ductus arteriosus	8
	746.8	Other specified anomalies of heart	7
	746.0	Anomalies of pulmonary valve	6
	750.5	Congenital hypertrophic pyloric stenosis	5
	753.2	Obstructive defects of renal pelvis & ureter	5
	755.0	Polydactyly	5
ONEIDA	752.5	Undescended testicle	14
	750.5	Congenital hypertrophic pyloric stenosis	13
	752.6	Hypospadias & epispadias	12
	745.4	Ventricular septal defect	11

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Birth Year: 2007

County	ICD-9 Code	Description	Number
ONEIDA	755.0	Polydactyly	11
	756.0	Anomalies of skull and face bones	7
	746.4	Congenital insufficiency of aortic valve	6
	754.7	Other deformities of feet	6
	746.8	Other specified anomalies of heart	5
	749.0	Cleft palate	5
	755.6	Other anomalies of lower limb including pelvic girdle	5
ONONDAGA	752.5	Undescended testicle	33
	745.4	Ventricular septal defect	32
	752.6	Hypospadias & epispadias	32
	753.2	Obstructive defects of renal pelvis & ureter	23
	750.5	Congenital hypertrophic pyloric stenosis	18
	758.0	Down syndrome	11
	745.5	Ostium secundum atrial septal defect	9
	753.1	Cystic kidney disease	9
	754.3	Congenital dislocation of hip	9
	755.0	Polydactyly	9
ONTARIO	753.2	Obstructive defects of renal pelvis & ureter	8
	745.4	Ventricular septal defect	6
	750.5	Congenital hypertrophic pyloric stenosis	5
	752.6	Hypospadias & epispadias	5
	751.5	Other anomalies of intestine	4
	754.5	Varus deformities of feet	4
	747.0	Patent ductus arteriosus	3
	752.5	Undescended testicle	3
	754.3	Congenital dislocation of hip	3
	754.7	Other deformities of feet	3
	755.6	Other anomalies of lower limb including pelvic girdle	3
ORANGE	752.6	Hypospadias & epispadias	29
	745.5	Ostium secundum atrial septal defect	25
	745.4	Ventricular septal defect	24
	747.0	Patent ductus arteriosus	20
	750.5	Congenital hypertrophic pyloric stenosis	20
	753.2	Obstructive defects of renal pelvis & ureter	15
	752.5	Undescended testicle	13
	742.3	Congenital hydrocephalus	9
	754.7	Other deformities of feet	9
	746.8	Other specified anomalies of heart	8
ORLEANS	742.1	Microcephalus	2
	752.5	Undescended testicle	2
	755.0	Polydactyly	2
	255.2	Adrenogenital disorders	1
	273.8	Other disorders of plasma protien	1

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Birth Year: 2007

County	ICD-9 Code	Description	Number
ORLEANS	282.6	Sickle-cell anemia	1
	335.0	Werdnig-Hoffmann disease	1
	742.2	Reduction deformities of brain	1
	742.3	Congenital hydrocephalus	1
	743.3	Congenital cataract & lens anomalies	1
	745.4	Ventricular septal defect	1
	745.5	Ostium secundum atrial septal defect	1
	746.0	Anomalies of pulmonary valve	1
	746.7	Hypoplastic left heart syndrome	1
	747.0	Patent ductus arteriosus	1
	747.3	Anomalies of pulmonary artery	1
	747.4	Anomalies of great veins	1
	748.5	Agenesis, hypoplasia & dysplasia, lung	1
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	1
	751.5	Other anomalies of intestine	1
	751.6	Anomalies of gallbladder, bile ducts, and liver	1
	752.7	Indeterminate sex & pseudo-hermaphroditism	1
	753.0	Renal agenesis & dysgenesis	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	754.1	Deformities of sternocleidomastoid muscle	1
	754.4	Congenital genu recurvatum & bowing of long bones of leg	1
	754.7	Other deformities of feet	1
	758.0	Down syndrome	1
OSWEGO	752.6	Hypospadias & epispadias	9
	752.5	Undescended testicle	8
	750.5	Congenital hypertrophic pyloric stenosis	7
	745.4	Ventricular septal defect	6
	753.2	Obstructive defects of renal pelvis & ureter	5
	745.1	Transposition of great vessels	4
	745.5	Ostium secundum atrial septal defect	4
	754.7	Other deformities of feet	4
	746.8	Other specified anomalies of heart	3
	747.0	Patent ductus arteriosus	3
	749.0	Cleft palate	3
	755.0	Polydactyly	3
	755.2	Reduction deformities of upper limb	3
	756.0	Anomalies of skull and face bones	3
OTSEGO	753.2	Obstructive defects of renal pelvis & ureter	4
	758.0	Down syndrome	4
	747.1	Coarctation of aorta	3
	752.5	Undescended testicle	3
	745.4	Ventricular septal defect	2
	745.5	Ostium secundum atrial septal defect	2
	746.4	Congenital insufficiency of aortic valve	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	752.6	Hypospadias & epispadias	2
	753.1	Cystic kidney disease	2

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Birth Year: 2007

County	ICD-9 Code	Description	Number
PUTNAM	753.2	Obstructive defects of renal pelvis & ureter	7
	745.5	Ostium secundum atrial septal defect	6
	745.4	Ventricular septal defect	5
	752.6	Hypospadias & epispadias	5
	746.8	Other specified anomalies of heart	3
	742.2	Reduction deformities of brain	2
	745.6	Endocardial cushion defects	2
	747.1	Coarctation of aorta	2
	754.5	Varus deformities of feet	2
	756.0	Anomalies of skull and face bones	2
	756.6	Anomalies of diaphragm	2
	757.3	Other specified anomalies of skin	2
QUEENS	745.4	Ventricular septal defect	160
	745.5	Ostium secundum atrial septal defect	144
	753.2	Obstructive defects of renal pelvis & ureter	120
	752.5	Undescended testicle	118
	747.0	Patent ductus arteriosus	104
	752.6	Hypospadias & epispadias	88
	755.0	Polydactyly	64
	750.5	Congenital hypertrophic pyloric stenosis	62
	754.3	Congenital dislocation of hip	46
	758.0	Down syndrome	35
RENSSELAER	750.5	Congenital hypertrophic pyloric stenosis	12
	745.4	Ventricular septal defect	8
	752.6	Hypospadias & epispadias	7
	747.0	Patent ductus arteriosus	6
	752.5	Undescended testicle	6
	755.0	Polydactyly	6
	753.2	Obstructive defects of renal pelvis & ureter	5
	751.1	Atresia & stenosis of small intestine	3
	758.0	Down syndrome	3
	228.0	Hemangioma, any site	2
	746.0	Anomalies of pulmonary valve	2
	747.3	Anomalies of pulmonary artery	2
	749.0	Cleft palate	2
	754.5	Varus deformities of feet	2
	754.6	Valgus deformities of feet	2
RICHMOND	747.0	Patent ductus arteriosus	23
	752.6	Hypospadias & epispadias	20
	745.5	Ostium secundum atrial septal defect	18
	752.5	Undescended testicle	18

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Birth Year: 2007

County	ICD-9 Code	Description	Number
RICHMOND	745.4	Ventricular septal defect	17
	750.5	Congenital hypertrophic pyloric stenosis	12
	753.2	Obstructive defects of renal pelvis & ureter	11
	746.8	Other specified anomalies of heart	9
	758.0	Down syndrome	8
	742.1	Microcephalus	6
	755.0	Polydactyly	6
ROCKLAND	752.6	Hypospadias & epispadias	29
	752.5	Undescended testicle	22
	745.4	Ventricular septal defect	21
	745.5	Ostium secundum atrial septal defect	21
	747.0	Patent ductus arteriosus	11
	755.0	Polydactyly	11
	758.0	Down syndrome	10
	753.2	Obstructive defects of renal pelvis & ureter	9
	742.3	Congenital hydrocephalus	6
	746.8	Other specified anomalies of heart	6
SARATOGA	750.5	Congenital hypertrophic pyloric stenosis	9
	747.0	Patent ductus arteriosus	8
	754.3	Congenital dislocation of hip	8
	752.5	Undescended testicle	7
	753.2	Obstructive defects of renal pelvis & ureter	7
	752.6	Hypospadias & epispadias	4
	753.4	Other specified anomalies of ureter	4
	745.4	Ventricular septal defect	3
	752.8	Other specified anomalies of genital organs	3
	753.0	Renal agenesis & dysgenesis	3
	757.3	Other specified anomalies of skin	3
	759.8	Other specified anomalies	3
SCHENECTADY	752.6	Hypospadias & epispadias	18
	752.5	Undescended testicle	10
	753.2	Obstructive defects of renal pelvis & ureter	9
	745.4	Ventricular septal defect	5
	750.5	Congenital hypertrophic pyloric stenosis	5
	751.5	Other anomalies of intestine	4
	754.5	Varus deformities of feet	4
	754.7	Other deformities of feet	4
	755.0	Polydactyly	4
	758.0	Down syndrome	4
SCHOHARIE	752.6	Hypospadias & epispadias	4
	750.5	Congenital hypertrophic pyloric stenosis	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	756.0	Anomalies of skull and face bones	2

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Most Frequently Reported Major Malformations by County

Birth Year: 2007

County	ICD-9 Code	Description	Number
SCHOHARIE	745.4	Ventricular septal defect	1
	747.0	Patent ductus arteriosus	1
	751.4	Anomalies of intestinal fixation	1
	753.1	Cystic kidney disease	1
	756.1	Anomalies of spine	1
	756.8	Other specified anomalies of muscle, tendon, fascia, etc.	1
	759.2	Anomalies of other endocrine glands	1
SCHUYLER	749.2	Cleft palate with cleft lip	2
	745.2	Tetralogy of Fallot	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	753.0	Renal agenesis & dysgenesis	1
SENECA	745.4	Ventricular septal defect	2
	743.3	Congenital cataract & lens anomalies	1
	745.0	Common truncus	1
	745.1	Transposition of great vessels	1
	747.3	Anomalies of pulmonary artery	1
	749.2	Cleft palate with cleft lip	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	755.6	Other anomalies of lower limb including pelvic girdle	1
	758.0	Down syndrome	1
ST LAWRENCE	752.6	Hypospadias & epispadias	10
	750.5	Congenital hypertrophic pyloric stenosis	7
	752.5	Undescended testicle	7
	753.2	Obstructive defects of renal pelvis & ureter	6
	745.4	Ventricular septal defect	5
	758.0	Down syndrome	5
	745.1	Transposition of great vessels	4
	747.0	Patent ductus arteriosus	4
	742.4	Other specified anomalies of brain	3
	746.8	Other specified anomalies of heart	3
	751.5	Other anomalies of intestine	3
	754.6	Valgus deformities of feet	3
STEUBEN	752.6	Hypospadias & epispadias	9
	758.0	Down syndrome	6
	745.4	Ventricular septal defect	5
	750.5	Congenital hypertrophic pyloric stenosis	5
	752.5	Undescended testicle	5
	753.2	Obstructive defects of renal pelvis & ureter	5
	754.7	Other deformities of feet	4
	755.0	Polydactyly	4
	747.0	Patent ductus arteriosus	3
SUFFOLK	745.5	Ostium secundum atrial septal defect	182
	752.6	Hypospadias & epispadias	96
	745.4	Ventricular septal defect	93

Section IV - Table 2
Most Frequently Reported Major Malformations by County

Birth Year: 2007

County	ICD-9 Code	Description	Number
SUFFOLK	753.2	Obstructive defects of renal pelvis & ureter	73
	747.0	Patent ductus arteriosus	71
	752.5	Undescended testicle	61
	750.5	Congenital hypertrophic pyloric stenosis	55
	747.3	Anomalies of pulmonary artery	43
	755.0	Polydactyly	42
	746.0	Anomalies of pulmonary valve	32
SULLIVAN	753.2	Obstructive defects of renal pelvis & ureter	6
	745.4	Ventricular septal defect	5
	752.6	Hypospadias & epispadias	5
	747.0	Patent ductus arteriosus	4
	755.0	Polydactyly	4
	742.3	Congenital hydrocephalus	2
	745.5	Ostium secundum atrial septal defect	2
	751.6	Anomalies of gallbladder, bile ducts, and liver	2
	754.3	Congenital dislocation of hip	2
	754.7	Other deformities of feet	2
	755.6	Other anomalies of lower limb including pelvic girdle	2
TIOGA	752.5	Undescended testicle	2
	752.6	Hypospadias & epispadias	2
	747.0	Patent ductus arteriosus	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	755.0	Polydactyly	1
	756.6	Anomalies of diaphragm	1
	759.8	Other specified anomalies	1
TOMPKINS	753.2	Obstructive defects of renal pelvis & ureter	5
	745.4	Ventricular septal defect	3
	752.5	Undescended testicle	3
	750.5	Congenital hypertrophic pyloric stenosis	2
	758.0	Down syndrome	2
	228.0	Hemangioma, any site	1
	243.	Congenital hypothyroidism	1
	282.7	Other hemoglobinopathies	1
	742.2	Reduction deformities of brain	1
	743.6	Congenital anomalies of eyelids, lacrimal system & orbit	1
	745.2	Tetralogy of Fallot	1
	745.6	Endocardial cushion defects	1
	747.0	Patent ductus arteriosus	1
	749.1	Cleft lip	1
	749.2	Cleft palate with cleft lip	1
	751.3	Hirschprung's disease & other functional disorders of colon	1

Section IV - Table 2
Most Frequently Reported Major Malformations by County

Birth Year: 2007

County	ICD-9 Code	Description	Number
TOMPKINS	752.6	Hypospadias & epispadias	1
	753.6	Atresia and stenosis of urethra & bladder neck	1
	754.7	Other deformities of feet	1
	755.0	Polydactyly	1
	755.6	Other anomalies of lower limb including pelvic girdle	1
ULSTER	745.4	Ventricular septal defect	11
	752.6	Hypospadias & epispadias	10
	753.2	Obstructive defects of renal pelvis & ureter	7
	745.5	Ostium secundum atrial septal defect	6
	758.0	Down syndrome	6
	752.5	Undescended testicle	5
	754.3	Congenital dislocation of hip	5
	746.4	Congenital insufficiency of aortic valve	3
	749.0	Cleft palate	3
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	3
	752.8	Other specified anomalies of genital organs	3
	759.8	Other specified anomalies	3
WARREN	752.5	Undescended testicle	5
	750.5	Congenital hypertrophic pyloric stenosis	4
	752.6	Hypospadias & epispadias	3
	753.2	Obstructive defects of renal pelvis & ureter	3
	758.0	Down syndrome	3
	746.3	Congenital stenosis of aortic valve	2
	746.4	Congenital insufficiency of aortic valve	2
	747.0	Patent ductus arteriosus	2
	228.1	Lymphangioma, any site	1
	745.0	Common truncus	1
	745.4	Ventricular septal defect	1
	745.5	Ostium secundum atrial septal defect	1
	746.0	Anomalies of pulmonary valve	1
	746.6	Congenital mitral insufficiency	1
	747.3	Anomalies of pulmonary artery	1
	749.0	Cleft palate	1
	749.2	Cleft palate with cleft lip	1
	751.1	Atresia & stenosis of small intestine	1
	751.4	Anomalies of intestinal fixation	1
	751.6	Anomalies of gallbladder, bile ducts, and liver	1
	754.3	Congenital dislocation of hip	1
	755.0	Polydactyly	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
	756.7	Anomalies of abdominal wall	1
	757.3	Other specified anomalies of skin	1

Section IV - Table 2
Most Frequently Reported Major Malformations by County

Birth Year: 2007

County	ICD-9 Code	Description	Number
WASHINGTON	750.5	Congenital hypertrophic pyloric stenosis	3
	752.5	Undescended testicle	3
	752.6	Hypospadias & epispadias	3
	754.3	Congenital dislocation of hip	3
	742.2	Reduction deformities of brain	1
	744.2	Other specified anomalies of ear	1
	745.1	Transposition of great vessels	1
	745.4	Ventricular septal defect	1
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	1
	753.1	Cystic kidney disease	1
	754.7	Other deformities of feet	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
WAYNE	752.5	Undescended testicle	6
	753.2	Obstructive defects of renal pelvis & ureter	6
	752.6	Hypospadias & epispadias	5
	745.4	Ventricular septal defect	3
	754.5	Varus deformities of feet	3
	742.1	Microcephalus	2
	745.5	Ostium secundum atrial septal defect	2
	745.6	Endocardial cushion defects	2
	746.0	Anomalies of pulmonary valve	2
	747.2	Other anomalies of aorta	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	751.4	Anomalies of intestinal fixation	2
	755.0	Polydactyly	2
	757.3	Other specified anomalies of skin	2
	758.0	Down syndrome	2
WESTCHESTER	753.2	Obstructive defects of renal pelvis & ureter	53
	745.4	Ventricular septal defect	44
	745.5	Ostium secundum atrial septal defect	38
	752.5	Undescended testicle	34
	752.6	Hypospadias & epispadias	33
	747.0	Patent ductus arteriosus	27
	750.5	Congenital hypertrophic pyloric stenosis	24
	755.0	Polydactyly	23
	754.3	Congenital dislocation of hip	15
	758.0	Down syndrome	12
WYOMING	745.4	Ventricular septal defect	4
	752.6	Hypospadias & epispadias	4
	742.1	Microcephalus	2
	745.5	Ostium secundum atrial septal defect	2
	754.3	Congenital dislocation of hip	2

Section IV - Table 2
Most Frequently Reported Major Malformations by County

Birth Year: 2007

County	ICD-9 Code	Description	Number
WYOMING	228.1	Lymphangioma, any site	1
	270.2	Other disturbances of aromatic amino acid metabolism	1
	742.4	Other specified anomalies of brain	1
	746.8	Other specified anomalies of heart	1
	747.0	Patent ductus arteriosus	1
	750.6	Congenital hiatus hernia	1
	752.5	Undescended testicle	1
	753.1	Cystic kidney disease	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	754.2	Deformities of spine	1
	755.0	Polydactyly	1
	755.2	Reduction deformities of upper limb	1
	755.6	Other anomalies of lower limb including pelvic girdle	1
	756.0	Anomalies of skull and face bones	1
	771.2	Other congenital infections	1
YATES	742.1	Microcephalus	2
	749.2	Cleft palate with cleft lip	2
	524.0	Major anomalies of jaw size	1
	745.4	Ventricular septal defect	1
	745.5	Ostium secundum atrial septal defect	1
	747.3	Anomalies of pulmonary artery	1
	749.0	Cleft palate	1
	751.1	Atresia & stenosis of small intestine	1
	752.6	Hypospadias & epispadias	1
	753.1	Cystic kidney disease	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	754.5	Varus deformities of feet	1
	754.7	Other deformities of feet	1
	755.6	Other anomalies of lower limb including pelvic girdle	1
	756.0	Anomalies of skull and face bones	1
	757.0	Hereditary edema of legs	1
	758.8	Other conditions due to sex chromosome anomalies	1
	759.8	Other specified anomalies	1

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 over the years has developed methods to monitor hospital reporting. In this section, CMR live birth prevalences are compared with the national prevalence estimates for 21 selected defects developed by the Centers for Disease Control and Prevention (CDC) and the National Birth Defects Prevention Network (NBDPN).[1] The 21 defects were selected as they are generally diagnosed soon after birth and the accuracy of diagnosis should be similar across sites[1].These estimates were based on 11 registries which use active case-finding. Active case-finding uses data collection specialists who go to hospitals to identify and abstract records of children with malformations. The active case-finding systems were chosen as they have similar methodology and prevalence estimates are usually higher in systems using active case finding, although variation was observed even among the 11 active case finding systems (See Figure 2 in Parker [1]).

As can be seen from Table 1, the CMR prevalences are equal to or higher than the lower boundary of the actual range of the 11 registries for 16 of the 21 defects (bold prevalences). The prevalences are generally higher for New York State excluding New York City than for New York City (17 defect prevalences are equal to or higher than the lower boundary of the actual range of the 11 registries compared to prevalence of 12 defects for New York City).

The interpretation of differences among registry prevalences is difficult. The lower prevalences of the CMR for neural tube defects (spina bifida with anencephalus), and trisomy 18 is most likely due to the lack of reports on terminations as termination rates for these conditions are high. The lower rates in limb reduction and gastroschisis are more difficult to explain as these are also easily recognizable defects. We have noted that the rate for lower limb reduction deformity has been declining over several years and plan to examine defect trends in a future report.

Several registries would have the highest prevalence for one defect and the lowest prevalence for others. Variation among the registries in the rates of specific defects could reflect demographic differences in the populations as there are racial and ethnic differences in the rates of specific birth defects[1]. The prevalence of Down syndrome, trisomy 18 and trisomy 13 is highly dependent upon the maternal age distribution, age-specific pregnancy rates and women's use of prenatal diagnosis and pregnancy termination. The lower live birth prevalence rates of these chromosomal abnormalities in the CMR may be partially attributable to one or more of these factors. However, the source(s) of much of the variation is unclear and there may be true geographic differences. A comparison of birth defect prevalences between the Metropolitan Atlanta Congenital Defects Program (MACDP) and California Birth Defects Monitoring

program (CBDMP) for the years 1983-1988 that adjusted for race, sex and maternal age showed regional differences in arm, hand and limb reduction defects [2].

CMR staff will continue their efforts to improve reporting (See Appendix 3) and will track our progress using the NBDPN national prevalence estimates.

Section V - Table 1

Prevalence* of Selected Major Birth Defects in New York State (Birth years: 2005-2007)

Birth Defect Category	New York City	Upstate NY	New York State	NBDPN 2004-2006
Central nervous system defects				
Anencephalus	0.4	0.5	0.5	2.2
Spina bifida without anencephalus	**1.9**	**2.6**	**2.3**	3.7
Encephalocele	**0.5**	**0.6**	**0.5**	0.8
Eye defects				
Anophthalmia/ microphthalmia	**1.1**	**1.7**	**1.4**	2.1
Cardiovascular defects				
Common truncus	**0.6**	**0.9**	**0.8**	0.7
Transposition of great arteries	**4.2**	**5.0**	**4.6**	3.0
Tetralogy of Fallot	**4.8**	**4.8**	**4.8**	4.1
Atrioventricular septal defect, AVSD (Endocardial cushion defect)	3.3	3.5	3.4	4.7
Hypoplastic left heart syndrome	**2.3**	**3.0**	**2.7**	2.3
Orofacial defects				
Cleft palate without cleft lip	5.3	**7.0**	**6.2**	6.5
Cleft lip with and without cleft palate	6.7	**8.5**	**7.6**	10.9
Gastrointestinal defects				
Esophageal atresia/ tracheosophageal fistula	**2.4**	**2.8**	**2.6**	2.1
Rectal and large intestinal atresia/stenosis	**4.4**	**4.1**	**4.2**	4.9
Musculoskeletal defects				
Reduction deformity, upper limbs	1.5	**2.6**	2.1	3.6
Reduction deformity, lower limbs	0.7	**1.1**	**0.9**	1.7
Gastroschisis	1.6	3.3	2.5	4.7
Omphalocele	**1.1**	**1.4**	**1.3**	1.9
Diaphragmatic hernia	**2.3**	**3.5**	**2.9**	2.6
Chromosomal defects				
Trisomy 13	**1.1**	**0.8**	**0.9**	1.2
Down syndrome(trisomy 21)	11.8	**13.7**	**12.8**	13.5
Trisomy 18	1.1	1.1	1.1	2.6

[a] - Prevalence (number of defects per 10,000 live births)

Bold prevalences are within the range or higher than the 11 active registries (Figure 2 in Reference 1)

References

1. Parker, S. E., Mai, C. T., Canfield, M. A., Rickard, R., Wang, Y., Meyer, R. E., Anderson, P., Mason, C. A., Collins, J. S., Kirby, R. S. and Correa, A. , Updated national birth prevalence estimates for selected birth defects in the United States, 2004–2006. Birth Defects Research Part A: Clinical and Molecular Teratology, n/a. doi: 10.1002/bdra.20735

2. Schulman J, Edmonds LD, McClern AB, et al. Surveillance for and comparison of birth defect prevelences in two geographic areas - United States 1983-1988. In: CDC Surveillance Summaries; March 19, 1993. *Morbidity and Mortality Weekly Report* 1993; 42(No. SS-1):1-7.

Section VI
Current Topics

1. Case Confirmation and Ascertainment using Cytogenetic Testing Data Obtained from Electronic Clinical Laboratory Reporting System

Background: Hospitals and physicians are required to submit case reports, as well as confirmatory diagnosis information to the New York State Congenital Malformations Registry (CMR) on children who are born or reside in New York State and are diagnosed before the age of two years with major birth defects. However, the majority of the cases with chromosomal anomalies indicated in the hospital discharge files are reported to the CMR without confirmatory testing data, which are usually not available at the time of reporting. The objective of this project was to link the cytogenetic test reports, submitted by cytogenetic testing laboratories via the New York State Department of Health (NYSDOH)'s Electronic Clinical Laboratory Reporting System (ECLRS), to CMR cases to obtain confirmatory diagnoses and identify unreported cases with chromosomal anomalies.

Methods: Cytogenetic testing data submitted by the New York State licensed laboratories and stored on the ECLRS Sybase server were retrieved and matched to CMR cases. Several identifying variables such as the child's name and date of birth and parent's name and address were used in data matching. The laboratory testing results were used to confirm diagnoses of CMR cases for matched reports and to ascertain new birth defects cases by auditing hospitals and physicians using unmatched reports with abnormal testing results.

Results: By the end of 2010, a total of 927 reports on 747 children were submitted to the CMR by 14 cytogenetic testing laboratories via the NYSDOH ECLRS. Among the 747 children reported, 398 (53%) had abnormal test results and 412 (55%) were matched to the CMR cases. From these laboratory reports, 151 new cases with chromosomal anomalies were identified, confirmed and added to the CMR. The additional cases accounted for about 7.8% of all cases with chromosomal anomalies in the CMR for the reporting years 2008-2010.

Conclusions: Cytogenetic laboratory reports can serve as an important source for ascertaining and confirming cases diagnosed with Down syndrome, autosomal deletion syndrome and other chromosomal anomalies. Acquiring molecular genetics testing data directly from cytogenetic testing laboratories enables CMR staff to confirm diagnoses and improve the accuracy and efficiency of case reporting.

2. Linking Children With Congenital Disorders Identified Through Newborn Screening to the Birth Defects Surveillance Program in New York State

Background: The CMR received funds from the U.S. Centers for Disease Control and Prevention in 2008 to develop a population-based surveillance and tracking system in New York State for the long-term follow-up of children identified by the newborn screening program (NBS) through enhanced collaboration between the established NYSDOH newborn screening and birth defects surveillance programs. The objective of the current project was to link children with congenital disorders identified through the NBS program to the CMR to examine their birth defects and identify unreported birth defects cases.

Methods: The NBS Program performs more than 11 million tests annually on approximately 250,000 newborns for more than 40 congenital diseases and the human immunodeficiency virus. Newborns with positive screening results for congenital disorders were obtained from the NBS program for the birth years 2006-2007 and were matched to the CMR database. The Statistical Analysis System (SAS, North Carolina, USA) was used to develop programs for data matching. Deterministic data linkage methods were used with multiple criteria for establishing a match between records using appropriate combinations of personal identifiers. The birth defect information in the CMR for the NBS children identified through data matching were reviewed and analyzed.

Results: A total of 16,781 children with an abnormal screening test were identified from the NBS program for 2006-2007 births. Of these, 1,268 (7.6%) had congenital disorders confirmed by follow-up diagnostic testing. The results from data matching showed that only 35.3% of NBS children with confirmed congenital conditions were matched to the CMR. A majority of the confirmed NBS children were not matched (i.e., not reported) to the CMR. Among the NBS children who had confirmed NBS conditions, 18.8% were found in the CMR having malformations in the cardiovascular or central nervous system. More than 60% of the NBS children with abnormal screening test who were deceased or lost to follow-up or had normal follow-up confirmatory diagnostic tests were found in the CMR having malformations in the cardiovascular or central nervous system.

Conclusions: Linking data from population-based surveillance programs, birth defects registries and newborn screening programs is helpful in understanding the epidemiology of some confirmed screening conditions and associated birth defects, and identifying possible causes of these congenital disorders. The NBS database containing the confirmatory diagnostic testing results can serve as an additional data source for identifying unreported birth defects cases. The project team plans to contact and work with genetic services centers and hospitals to ascertain unreported children with congenital disorders identified through NBS program.

References

Wang Y, Caggana M, Sango-Jordan M, Sun M, Druschel CM. Long-term follow-up of children with confirmed newborn screening disorders using record linkage. *Genet Med.* 2011;13:881-6.

Section VII

Current Publications

1. Browne ML, Hoyt AT, Feldkamp ML, Rasmussen SA, Marshall EG, Druschel CM, Romitti PA. Maternal caffeine intake and risk of selected birth defects in the National Birth Defects Prevention Study. *Birth Defects Res A Clin Mol Teratol*. 2011; 91(2):93-101.

   Convincing evidence of an association between maternal caffeine intake and the birth defects included in this study (anotia/ microtia, esophageal atresia, small intestinal atresia, craniosynostosis, diaphragmatic hernia, omphalocele, and gastroschisis) was not observed. Small elevations were noted in odds ratios for total maternal dietary caffeine intake or specific types of caffeinated beverages and several types of defects; however, dose-response patterns were absent.

2. Carter TC, Olney RS, Mitchell AA, Romitti PA, Bell EM, Druschel CM; National Birth Defects Prevention Study. Maternal self-reported genital tract infections during pregnancy and the risk of selected birth defects. *Birth Defects Res A Clin Mol Teratol*. 2011; 91(2):108-16.

   Of over 50 specific birth defect types examined, genital tract infections during the periconceptional period were associated with small to modest increases in the risk of bilateral renal agenesis/hypoplasia, cleft lip with or without cleft palate, and transverse limb deficiency and chlamydia/gonorrhea/pelvic inflammatory disease was associated with cleft lip only. Considering the number of birth defects studied, this study found little evidence of a causal association between genital tract infections and birth defects.

3. Holtzer C, Meaney FJ, Andrews J, Ciafaloni E, Fox DJ, James KA, Lu Z, Miller L, Pandya S, Ouyang L, Cunniff C, and the MD STAR*net* group. Disparities in the Diagnosis of Duchenne and Becker Muscular Dystrophy. *Genetics in Medicine*. 2011; 13(11):942-7.

   Racial and ethnic disparities in the diagnostic process for Duchenne and Becker Muscular Dystrophy (DBMD) were found even after adjustment for family history of DBMD and changes in the diagnostic process over time. Black and Hispanic children were initially evaluated at older ages than White children, and the gap widened at later steps in the diagnostic process.

4. Matthews DJ, James KA, Miller LA, Pandya S, Campbell KA, Ciafaloni E, Mathews KD, Miller TM, Cunniff C, Meaney FJ, Druschel CM, Romitti P, Fox DJ, and the MD STAR*net* group. Use of corticosteroids in a population-based cohort of boys with Duchenne and Becker muscular dystrophy. *J Child Neurol.* 2010; 25(11):1319-24.

   The use of corticosteroids for treatment of Duchenne/Becker muscular dystrophy (DBMD) was examined. Corticosteroid use increased from 20% of individuals in 1991 to 44% in 2005. Average use varied by across 4 U.S. sites, ranging from 15% to 49%, with 6.9 years old as

the median age of treatment. Median dose for prednisone was 0.729 mg/kg; for deflazacort was 0.831 mg/kg. The most common discontinuation reasons were weight gain, behavioral side effects and loss of ambulation resulting in full-time wheelchair use.

5. Moxley RT 3rd, Pandya S, Ciafaloni E, Fox DJ, and Campbell K. Change in natural history of Duchenne muscular dystrophy with long-term corticosteroid treatment: Implications for management. *J Child Neurol.* 2010; 25(9):1116-29.

   In 2005, the American Academy of Neurology published a practice parameter indicating that prednisone has a beneficial effect on muscle strength and function in patients with Duchenne muscular dystrophy (DMD). Recent reports emphasize that longer term treatment with corticosteroids (greater than 3 years) produces important sustained benefits without causing major side effects. This review indicates that long-term corticosteroid therapy (1) prolongs ambulation by 2 to 5 years, (2) reduces the need for spinal stabilization surgery, (3) improves cardiopulmonary function, (4) delays use of noninvasive nasal ventilation, and (5) increases survival and quality of life of DMD patients.

6. Munsie JW, Lin S, Browne ML, Campbell KA, Caton AR, Bell EM, Rasmussen SA, Romitti PA, Druschel CM; National Birth Defects Prevention Study. Maternal bronchodilator use and the risk of orofacial clefts. *Hum Reprod*. 2011; 26(11):3147-54.

   Periconceptional maternal bronchodilator use was associated with a modest increase in the risk of cleft lip only and a nonsignificant increase in the risk of cleft palate only. No association was observed for maternal bronchodilator use and the risk of cleft lip with cleft palate. It is unclear whether the increased risk estimates observed in this study are due to bronchodilator use, the severity of asthma, or both, or to chance alone.

7. Nabukera SK, Romitti PA, Campbell KA, Meaney FJ, Hockett Sherlock S, Puzhankara S, Cunniff C, Druschel CM, Pandya S, Matthews D, Ciafaloni E, Mathews K, and the MD STAR*net* group. Use of Complementary and Alternative Medicines in the Management of Duchenne/Becker Muscular Dystrophy in the MD STARnet Cohort. Accepted for publication at *J Child Neurol.* 2011.

   Use of complementary and alternative medicine for males with Duchenne/Becker muscular dystrophy (DBMD) was examined. Through caregiver interviews, 80% reported ‘‘ever’’ using complementary and alternative medicines for their affected children (mind-body medicine/ 61.5%; biologically-based practices/48.0%; manipulative and body-based practices/29.0%; whole medical systems/8.5%). Overall, complementary and alternative medicines use was high; disease duration, education, and income levels influenced use.

8. Richardson S, Browne ML, Rasmussen SA, Druschel CM, Sun L, Jabs EW, Romitti PA; National Birth Defects Prevention Study. Associations between periconceptional alcohol consumption and craniosynostosis, omphalocele, and gastroschisis**.** *Birth Defects Res A Clin Mol Teratol*. 2011; 91(7):623-30.

   Maternal periconceptional alcohol consumption and in particular binge drinking was associated with increases in the risk of omphalocele and gastroschisis. A decreased risk of craniosynostosis was observed following second and third trimester alcohol consumption.

9. Wang Y, Hu J, Druschel CM, Kirby RS. 25-year Survival of Children with Birth Defects in New York State: A Population-based Study. *Birth Defects Res A Clin Mol Teratol*. 2011; 91(12):995-1003.

   Using the statewide, population-based birth defects surveillance data in New York State, the survival experience of the study cohort was examined across all survival time periods by individual birth defect of interest. Several risk factors that affect survival were identified.

10. Wang Y, Caggana M, Sango-Jordan M, Sun M, Druschel CM. Long-term follow-up of children with confirmed newborn screening disorders using record linkage. *Genet Med.* 2011; 13:881-6.

    The mortality and survival experience of the children with birth defects during infancy and childhood period by individual birth defects of interest were examined using the state-wide, population-based birth defects surveillance data. As expected, children born with birth defects had a higher risk of mortality compared to children without birth defects. The magnitude of the risk varied by children's age, birth and maternal characteristics and the lethality of the specific birth defects involved.

APPENDICES

Appendix 1

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

740 - 759*	Congenital Anomalies
760.71	Fetal Alcohol Syndrome
762.8	Amniotic Bands
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
553.1	Umbilical hernia
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
747.5	Single umbilical artery
752.41	Embryonic cyst of cervix, vagina and external female genitalia
752.42	Imperforate hymen
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 2

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 birth weight. 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 records for a given year to the CMR file of cases who were born in that year. A deterministic matching method is applied to identify all possible matches, using combinations of identifying variables such as name, date of birth, medical record number and mother's name and address information. Matching scores are assigned to each criterion. Assigning different points to different identifiers provides a way to recognize variations in quality or reliability of different data items. The records are compared on identifying variables that are available 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.

The matching process is repeated until about 95 percent of reported cases are matched. This is a compromise between completeness and efficiency. After about 90 percent 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.

Appendix 3

Case Ascertainments and Data Quality Assurance

The CMR uses the method of passive case ascertainment of birth defects that occur among live births, with an active follow-up for assuring the accuracy and completeness of case reporting. Birth defect cases reported from hospitals and physicians are reviewed and the diagnoses are coded by the registry's trained staff. Reporting hospitals and physicians are contacted for cases that have insufficient diagnostic information for coding. CMR staff recognize that completeness, accuracy and timeliness are the hallmarks of a good surveillance system. However, these attributes exist in tension, "conflicting principles" (Kallen 1988). Steps taken to improve completeness and accuracy may actually reduce timeliness. From the very beginning, the CMR has built in procedures to improve the quality of the data in the CMR. These systems have changed over time (Sekhobo and Druschel 2001; Druschel et al, 2001) and the CMR now has three major approaches to improving data quality: 1) matching to hospital discharge data, the Statewide Planning and Research Cooperative System (SPARCS) for completeness; 2) the web-based reporting system, the Health Commerce System (HCS) for timeliness and completeness; 3) on-site hospital audits for completeness and accuracy. In addition, we also periodically request medical records and compare them to the hospital's report for an additional review of accuracy.

SPARCS Audits For the SPARCS audit, children age 2 years or younger and diagnosed with reportable birth defects are selected from SPARCS files of all reporting hospitals and matched to the CMR database for the same birth year period. As about 90 percent of children reported to the CMR were diagnosed in the first six months of life, CMR staff begin to audit hospitals 12 to 24 months after the reporting period for each year of birth. Unmatched reports from the SPARCS hospital discharge files are sent to the hospital, requesting submission of the missed reports. A study (Wang et al, 2005) demonstrated that using hospital discharge data to improve case ascertainment is a valuable and effective method of enhancing birth defect surveillance, particularly for those hospitals with low reporting rates. Hospital audits resulted in not only added new reports (comprised 21.4 percent of all CMR reports) to the CMR but also improved reporting for subsequent years, probably due to hospitals' positively reacting to the audits. Auditing hospitals by CMR staff sent a message to reporting hospitals that both the quality and the quantity of their reports are closely monitored.

HCS Reporting A web-based reporting, data management and communication system has been successfully developed and implemented by CMR staff (Wang et al, 2007a, Steen et al, 2008). After pilot testing with two hospitals in 2001, the system was phased in for reporting in 2003. By January 2006, the CMR had converted all reporting hospitals statewide from a manual, paper-based reporting system to the web-based system. This new system provides a platform-independent environment for data submission, retrieval and analysis and offers a secure, cost-effective solution for participating hospitals. An authorized user can submit/edit data and view, update or query their case information dynamically from the CMR's database using any personal computer equipped with an internet browser from any geographic area throughout the state. This innovative system enables CMR staff to review and perform quality assurance on every report submitted and to query hospitals quickly about submitted reports. A study that evaluated the

completeness of submitted case information and timeliness of reporting to the CMR and the effectiveness of the HCS communication and query system when compared to the previous manual, paper-based system found that the implementation of the HCS system has resulted in more timely submission of cases and promoted effective communication between the CMR and reporting hospitals. There was a nearly 50 percent reduction in median days used for reporting. (Wang et al, 2007b).

Monitoring Hospital Reporting CMR staff have developed on-line SAS/IntrNet applications which empower the users to search and retrieve hospital submitted cases, generate real-time reports and perform simple statistical analysis using the CMR's database (Wang et al, 2008). For instance, CMR staff can select a reporting hospital and discharge years of interest and then, generate a real-time report table which lists the number of cases by discharge year and month. By reviewing this report, CMR staff are able to identify hospitals with unusual reporting patterns or problems, for instance, if they stopped or skipped reporting for certain months or years.

On-site Hospital Audits On-site hospital audits began in August of 2003 as an additional surveillance tool. CMR staff needed to know if all malformations were being captured from medical records, and if the reports were complete and accurate. This was piloted in 2002 and implemented in 2003. The procedure begins when the CMR announces to the hospital that they will be making an "in-house chart review or audit" and requests the hospital in question to send a discharge summary for all children 2 years of age and younger for a specific discharge period, usually one year. The list includes all children discharged in that given year, not just those with a congenital code. This is done so that reportable conditions that may have been miscoded can be identified. CMR staff review the discharge list, comparing it to the list of children who have already been reported to the CMR. A list of reported, not reported and partially reported cases is made. Depending on the time frame and number of auditors available, the entire list or a subset of this list will be sent to the hospital and they will be requested to produce the charts so that CMR staff can review them. CMR staff will spend between 1 and 2 days at a facility reviewing records. At the completion of the review, the facility will be asked to report any case that is considered by the CMR staff as reportable but not previously reported as well as any partially reported cases that need to be completed. A written summary of the audit findings is sent to the Director of Health Information Management including comments that may indicate what chronic reporting problems were evident. Since 2003, 95 hospitals have had an "in-house" audit; 5023 charts have been reviewed; 1915 cases that were not previously reported were flagged and subsequently reported, 436 cases that were partially reported were completed and 189 cases with incorrect diagnoses reported were corrected or deleted.

Hospital Report Card In order to improve the completeness of case reporting and the accuracy of reported cases, CMR staff have developed an on-line application to generate report cards for hospitals to track their reporting progress in 2008. The first report card summarizing reporting status and progress of hospitals for the reporting period of June 1 - December 31, 2007 was sent to each individual hospital in April 2008. The report cards for all reporting hospitals are generated bi-annually and made available online for the hospital officials.

Summary Surveillance requires on-going efforts to respond to changes in resources and technologies. There must also be constant communication and feedback between the reporting

sources and the surveillance system. The CMR has developed several methods to monitor and improve the system's completeness, accuracy and timeliness. CMR staff recognize that as a 'passive' reporting system much additional work must be done to be able to provide data of good quality. While 'active' case ascertainment systems seem to provide more completeness and accuracy, they require much higher funding levels and many more staff. In this era of cutbacks, these funding levels can be difficult to maintain and some of these systems have been forced to reduce their activities or decrease their areas of coverage. The CMR has seen many staff reductions over the years but by making use of new technologies has been able to improve the system. However, further improvements are needed and the CMR will continue to review procedures and develop new methods. The CMR is currently investigating ways to use hospital discharge summaries (most of which are electronic) as an additional source of case finding. As more and more hospitals go to electronic medical records, these might also assist us in case finding and confirmation of diagnoses. Birth defects are a serious health issue for affected infants and children and their families. With so many different conditions, surveillance of birth defects can be challenging but must be done so that they can be tracked and studied.

References

Druschel C, Sharpe-Stimac M, Cross P. Process of and Problems in Changing a Birth Defects Registry Reporting System. Teratology 2001;64:S30-S36.

Kallen B. Epidemiology of Human Reproduction. CRC Press, Boca Raton.

Sekhobo JP, Druschel CM. An Evaluation of Congenital Malformations Surveillance in New York State: An application of Centers for Disease Control and Prevention Guidelines for Evaluation Surveillance Systems. Public Health Reports 2001;116:296-302.

Steen PK, Wang Y, Tao Z, Cross PK, Druschel CM. Implementing a Web-based Case Reporting and Communication System Among Hospitals Reporting to the Birth Defects Registry in New York State. *J Public Health Manag Pract*. 2008; 14(6):E11-E16.

Wang Y, Sharpe-Stimac M, Cross PK, Druschel CM, Hwang SA. Improving Case Ascertainment of a Population-Based Birth Defects Registry in New York State Using Hospital Discharge Data. *Birth Defect Research Part A*, 2005, 73:663-668.

Wang Y, Cross PK, Steen PK, Tao Z, Druschel CM, Cukrovany JL, Marion DR, Hwang SA. Development of a Web-based Case Reporting, Management and Communication System for the Statewide Birth Defects Registry in New York State. *J Registry Management*. 2007a; 34(2):45-52.

Wang Y, Tao Z, Cross PK, Hwang SA. Evaluating the Timeliness and Completeness of a Web-based Reporting and Communication System of the New York State Congenital Malformations Registry. *J Registry Management*. 2007b; 34(4): 93-98.

Wang Y, Tao Z, Cross PK, Le LH, Steen PK, Babcock GD, Druschel CM, Hwang SA. Development of a Web-based Integrated Birth Defects Surveillance System in New York State. *J Public Health Manag Pract*. 2008; 14(6):E1-10.

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 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 Code
CENTRAL NERVOUS SYSTEM			
A01	Anencephaly	740.0, 740.1, 740.2	740.00, 740.01, 740.02, 740.03, 740.08, 740.10, 740.20, 740.21, 740.29
A02	Spina Bifida with Hydrocephaly	741.00, 741.01, 741.02, 741.03	741.000, 741.001, 741.002, 741.003, 741.004, 741.008, 741.009, 741.011, 741.012, 741.013, 741.014, 741.018, 741.019, 741.021, 741.022, 741.023, 741.024, 741.028, 741.029-741.599
A03	Spina Bifida without Hydrocephaly	741.90, 741.91, 741.92, 741.93	741.701, 741.702, 741.703, 741.704, 741.708, 741.709- 741.999
A13	Encephalocele	742.0	742.000, 742.080, 742.085, 742.086, 742.090
A15	Hydrocephaly	742.3	742.300, 742.310, 742.320, 742.380, 742.390
A16	Microcephalus	742.1	742.100, 742.150
EYE / EAR			
B01	Anophthalmia, Microphthalmia	743.00, 743.10, 743.11, 743.12	743.0000, 743.1000, 743.1009, 743.0003, 743.0006, 743.1001, 743.1002
B03	Glaucoma	743.20, 743.21, 743.22	743.2000, 743.210, 743.2001, 743.220
B04	Cataract	743.30, 743.31, 743.32, 743.33, 743.34, 743.35, 743.36, 743.37, 743.39	743.320, 743.325, 743.3261, 743.3262, 743.3263, 743.3264, 743.300, 743.310, 743.340, 743.3806, 743.330, 743.3269, 743.3809, 743.390
B54	Ear anomaly with hearing loss	744.00, 744.01, 744.02, 744.03, 744.04, 744.05, 744.09	744.0001, 744.0101, 744.0002, 744.0902, 744.0203, 744.0204 744.030, 744.0109, 744.0900
CARDIAC			
D01	Truncus arteriosus	745.0	745.000, 745.010
D02	Transposition of great vessels	745.10, 745.11, 745.12, 745.19	745.1001, 745.110, 745.1801, 745.120, 745.1809, 745.190
D03	Tetralogy of Fallot	745.2	745.200, 745.210
D04	Single ventricle	745.3	745.300
D05	VSD	745.4	745.480, 745.485, 745.486, 745.487, 745.490
D52	Hypoplastic left heart	746.7	746.700
D53	Total anomalous pulmonary venous return	747.41	747.420
RESPIRATORY			
E01	Choanal atresia	748.0	748.000
E06	Agenesis of lung	748.5	748.500, 748.510, 748.520, 748.580, 748.590

MACDP Code	Condition	ICD-9	BPA Code
CLEFTS			
F01	Cleft palate	749.00, 749.01, 749.02, 749.03, 749.04	749.010, 749.020, 749.030, 749.050, 749.060, 749.070, 749.090, 749.001, 749.002, 749.003, 749.041, 749.042, 749.043, 749.080
F02	Cleft lip with or without cleft palate	749.10, 749.11, 749.12, 749.13, 749.14, 749.20, 749.21, 749.22, 749.23, 749.24, 749.25	749.1010, 749.1020, 749.1030, 749.1100, 749.120, 749.1901, 749.1011, 749.1021, 749.1031, 749.1012, 749.1022, 749.1032, 749.1103, 749.1104, 749.2900, 749.2011, 749.2021, 749.2031, 749.2012, 749.2022, 749.2032, 749.2103, 749.2104, 749.2015, 749.2025, 749.2035, 749.2105, 749.2203, 749.2905
GASTRO-INTESTINAL			
F14	Stenosis or atresia of duodenum	751.1	751.100
F15	Other stenosis or atresia of small intestine	751.1	751.110, 751.120, 751.190, 751.195
F16	Stenosis or atresia of rectum or anus	751.2	751.210, 751.220, 751.230, 751.240
F17	Hirschsprung's Disease	751.3	751.300, 751.310, 751.320, 751.303
F18	Malrotation of intestine	751.4	751.400, 751.410, 751.420, 751.490, 751.495
F21	Biliary atresia	751.61	751.6501
GENITO-URINARY			
H01	Renal agenesis	753.0	753.000, 753.009, 753.010
H06	Obstruction of kidney or ureter	753.20, 753.21, 753.22	753.220, 753.221, 753.240, 753.241, 753.242, 753.243, 753.244, 753.290, 753.299
H09	Bladder or urethra obstruction	753.6	753.600, 753.610, 753.620, 753.630, 753.690
MUSCULOSKELETAL			
J02	Curvature of spine (scoliosis or lordosis)	754.2	754.200, 754.210, 754.220
J03	Dislocation of hip	754.30, 754.31	754.3000, 754.3010, 754.3020, 754.3030
J11	Arthrogryposis multiplex congenita	754.89	755.800
K01	Reduction deformity - upper limb	755.20, 755.21, 755.22, 755.23, 755.24, 755.25, 755.26, 755.27, 755.28, 755.29	755.200, 755.230, 755.240, 755.2901, 755.5851, 755.2602, 755.265, 755.2702, 755.280, 755.2902, 755.210, 755.218, 755.220, 755.2606, 755.2707, 755.2801, 755.247, 755.2609, 755.2709, 755.2900, 755.5800, 755.5850, 755.59859
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	755.300, 755.330, 755.3401, 755.33901, 755.6851, 755.360, 755.380, 755.3103, 755.3104, 755.318, 755.3801, 755.320, 755.365, 755.366, 755.3802, 755.3409, 755.3900, 755.6859
K05	Amniotic bands	762.8	658.800
N01	Diaphragmatic hernia	756.6	756.610, 756.615, 756.616
N02	Omphalocele	756.79	756.700
N04	Gastroschisis	756.79	756.710
SYNDROMES			
R01	Down Syndrome	758.0	758.000, 758.010, 758.020, 758.030, 758.040, 758.050, 758.09
R02	Patau Syndrome (Trisomy 13)	758.1	758.100, 758.110, 758.120, 758.130, 758.140, 758.150, 758.190
R03	Edwards Syndrome (Trisomy 18)	758.2	758.200, 758.210, 758.220, 758.230, 758.290, 758.295, 758.296
S02	Fetal Alcohol Syndrome	760.71	760.710, 760.715, 760.718
W03	Conjoined twins	759.4	759.400, 759.410, 759.420, 759.430, 759.440, 759.480, 759.490

Appendix 5

Glossary of Birth Defects and Related Terms

(Courtesy of the Texas Birth Defects Monitoring Division, August 2008)

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.

Anomalies of the tricuspid valve Includes tricuspid valve atresia or stenosis, as well as enlargement, dilation, or aneurysm of the tricuspid valve. See also tricuspid valve atresia or stenosis.

Anophthalmia 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.

Aorta The large arterial trunk that carries blood from the heart to be distributed by branch arteries through the body

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.*

Atrium One of the two upper chambers of the heart (plural atria). The right atrium receives unoxygenated blood from the body. The left atrium receives oxygenated blood from the lungs.

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

Birth prevalence

$$\frac{\text{\# of cases with birth defect A in an area and time period}}{\text{\# of live births in that area and time period}} \times 10{,}000$$

Bladder exstrophy 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.

Cluster An apparently unusual concentration of a health condition in a particular area and time period.

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 percent) The interval that contains the true prevalence (which we can only estimate) 95 percent 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.

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.

Dysgenesis Impaired or faulty development of part(s) of the body.

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.

Embryogenesis The development and growth of an embryo, especially the period from the second through the eighth week after conception.

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.

Folate B vitamin necessary for red blood cell production; folate deficiency can lead to anemia and, during embryogenesis, can affect the normal development of the fetus' neural tube; found in liver, green leafy vegetables, beans, beets, broccoli, cauliflower, citrus fruits, and sweet potatoes. *See folic acid.*

Folic acid One of the B vitamins especially important for a woman to take before conception to help prevent neural tube defects in a fetus; essential for DNA synthesis and therefore the growth and division of cells; obtained from fortified foods or from a multivitamin containing at least 4mg; also found in natural sources including liver, beans, and leafy green vegetables. While folate and folic acid are both forms of water-soluble B vitamins, folic acid refers to the synthetic vitamin used in supplements, whereas folate is the form found in foods.

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

Hernia A protrusion of an organ or part through connective tissue or through a wall of the cavity in which it is normally enclosed.

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.

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

Hyperplasia Overgrowth characterize 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 defects.

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.

Ostium secundum defect See atrial septal defect.

Patau syndrome (Trisomy 13) The chromosomal abnormality caused by an 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.

Poisson regression a type of statistical analysis based on the Poisson distribution used to compare rates of rare occurrences such as birth defects between different population groups, different areas, or different times.

Prevalence With respect to the prevalence of birth defects, see "*Birth prevalence*".

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 septaldefect, 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 (Patau syndrome) The chromosomal abnormality caused by an extra chromosome 13. The extra copy can be free-lying, or can be attached to some other chromosome. Trisomy 13 can occur in mosaic so that there is a population of normal cells and
a population of trisomy 13 cells. Trisomy 13 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.

Trisomy 18 (Edwards Syndrome) 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 so that there is a population of normal cells and a population of trisomy 18 cells. Trisomy 18 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.

Trisomy 21 (Down Syndrome) 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. Trisomy 21 can occur in mosaic, so that there is a population of normal cells and a population of trisomy 21 cells. Trisomy 21 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 Trisomy 21.

Truncus arteriosus *See Common truncus*.

Ventricle One of the two lower chambers of the heart (plural ventricles). The right ventricle sends blood to the lungs, and the left ventricle passes oxygen-rich blood to the rest of the body.

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