NEW YORK STATE DEPARTMENT OF HEALTH

Congenital Malformations Registry

Statistical Summary of Children
Born in 2008 and Diagnosed Through 2010

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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Summary

This Congenital Malformations Registry Summary Report presents rates of congenital malformations occurring among the 246,655 children who were born alive to New York residents in 2008. The children reported with a major congenital malformation represent 5.1 percent of live births. Males had a higher rate of major congenital malformations than females (6.0 percent versus 4.0 percent), and black children had a higher major malformation rate than white children (6.6 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 2007 birth cohorts. This report and the reports for 1994-2007 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.

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.

2008 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 2008 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 2008 and diagnosed before their second birthday. Reports are also available for the 1984 through 2007 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 2008, there were 246,655 resident live births reported to the Bureau of Biometrics and Health Statistics of the New York State Department of Health, 16.3 percent to black mothers, and 23.9 percent to Hispanic mothers. In accordance with the practices of other state birth defects registries, the race of the child is based on the race of the mother only. Approximately 49.3 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 2008 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 5.1% of live births. Male children have a higher rate of major malformations than female children (6.0% versus 4.0%, Table 1). This difference is consistent within different racial groups. The rates for major malformations are somewhat higher for black than for white children (6.6 % 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 (5.2% versus 4.9%). The smaller number of births in the "other" racial category makes these rates difficult to interpret.

About 79.0% 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 I - Table 1
2008 Births - New York State Residents
Percent of Live Births with One or More Major Malformation
Sex by Race/Ethnicity and Residence

	Both Sexes			Males			Females		
Race and Residence	Infants with birth defects	Total Births	%	Infants with birth defects	Total Births	%	Infants with birth defects	Total Births	%
New York State									
All Races	12,462	246,655	5.1	7,643	126,673	6.0	4,819	119,982	4.0
Non-Hispanic white	5,947	119,196	5.0	3,751	61,335	6.1	2,196	57,861	3.8
Non-Hispanic black	2,636	40,216	6.6	1,519	20,476	7.4	1,117	19,740	5.7
Hispanic	2,680	59,003	4.5	1,655	30,364	5.5	1,025	28,639	3.6
Others/Unknown	1,199	28,240	4.2	718	14,498	5.0	481	13,742	3.5
NYS Excluding NYC									
All Races	6,521	125,071	5.2	4,060	64,165	6.3	2,461	60,906	4.0
Non-Hispanic white	4,351	84,705	5.1	2,759	43,570	6.3	1,592	41,135	3.9
Non-Hispanic black	892	12,628	7.1	516	6,485	8.0	376	6,143	6.1
Hispanic	917	19,905	4.6	575	10,215	5.6	342	9,690	3.5
Others/Unknown	361	7,833	4.6	210	3,895	5.4	151	3,938	3.8
New York City									
All Races	5,941	121,584	4.9	3,583	62,508	5.7	2,358	59,076	4.0
Non-Hispanic white	1,596	34,491	4.6	992	17,765	5.6	604	16,726	3.6
Non-Hispanic black	1,744	27,588	6.3	1,003	13,991	7.2	741	13,597	5.4
Hispanic	1,763	39,098	4.5	1,080	20,149	5.4	683	18,949	3.6
Others/Unknown	838	20,407	4.1	508	10,603	4.8	330	9,804	3.4

Section I - Table 2
2008 Births - New York State Residents
Number of Major Malformations Per Child

Number of Malformations	Number of Children	Percent
1	9,848	79.0
2	1,637	13.1
3	520	4.2
4	221	1.8
5	108	0.9
6	51	0.4
7	40	0.3
8	16	0.1
9	11	0.1
10	6	*
11	2	*
12	2	*
All Children	12,462	100.0

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

Section I - Table 3
2008 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 with birth defects	Total Births	%	Infants with birth defects	Total Births	%	Infants with birth defects	Total Births	%
New York State									
All Races	9,848	246,655	4.0	6,155	126,673	4.9	3,693	119,982	3.1
Non-Hispanic white	4,682	119,196	3.9	3,026	61,335	4.9	1,656	57,861	2.9
Non-Hispanic black	2,118	40,216	5.3	1,239	20,476	6.1	879	19,740	4.5
Hispanic	2,090	59,003	3.5	1,310	30,364	4.3	780	28,639	2.7
Others/Unknown	958	28,240	3.4	580	14,498	4.0	378	13,742	2.8
NYS Excluding NYC									
All Races	5,127	125,071	4.1	3,262	64,165	5.1	1,865	60,906	3.1
Non-Hispanic white	3,410	84,705	4.0	2,226	43,570	5.1	1,184	41,135	2.9
Non-Hispanic black	723	12,628	5.7	422	6,485	6.5	301	6,143	4.9
Hispanic	721	19,905	3.6	452	10,215	4.4	269	9,690	2.8
Others/Unknown	273	7,833	3.5	162	3,895	4.2	111	3,938	2.8
New York City									
All Races	4,721	121,584	3.9	2,893	62,508	4.6	1,828	59,076	3.1
Non-Hispanic white	1,272	34,491	3.7	800	17,765	4.5	472	16,726	2.8
Non-Hispanic black	1,395	27,588	5.1	817	13,991	5.8	578	13,597	4.3
Hispanic	1,369	39,098	3.5	858	20,149	4.3	511	18,949	2.7
Others/Unknown	685	20,407	3.4	418	10,603	3.9	267	9,804	2.7

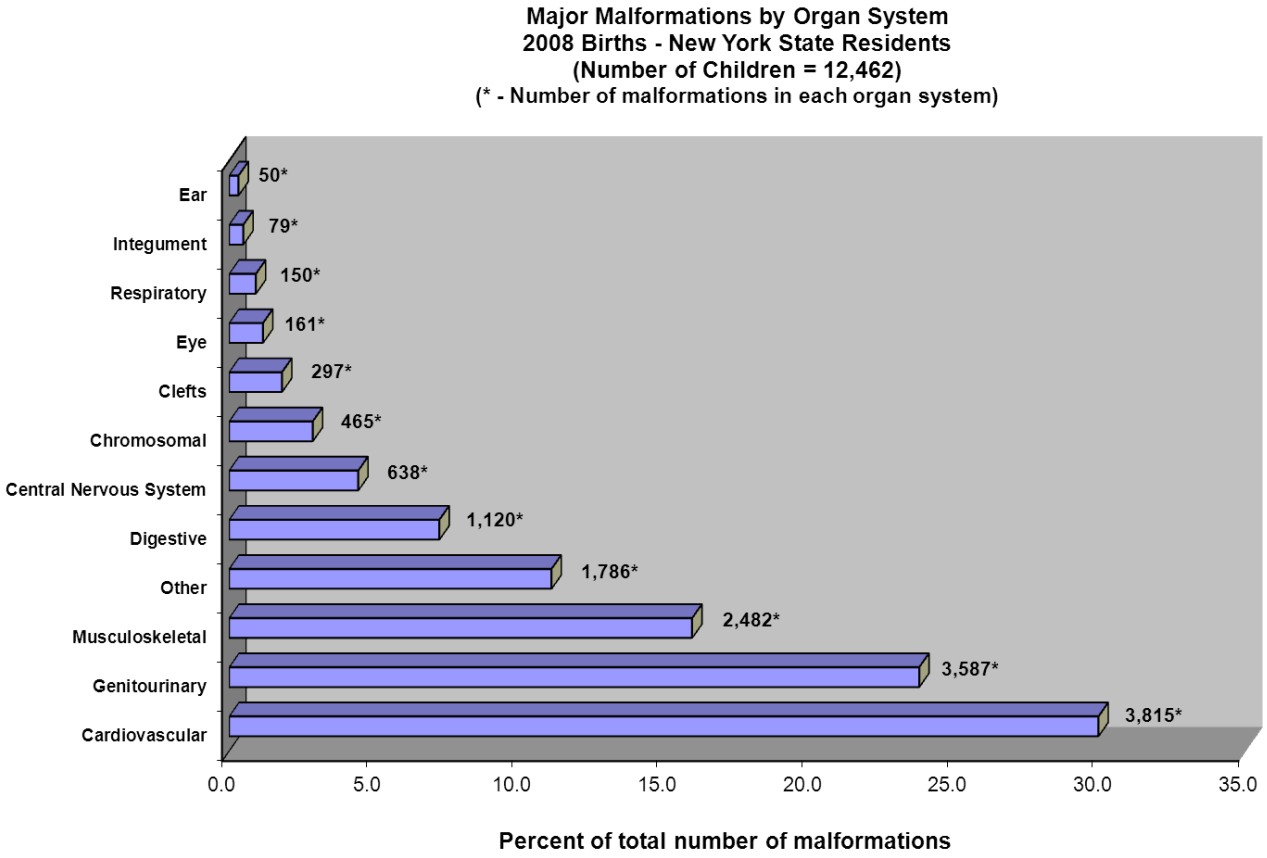
Section I - Table 4
2008 Births - New York State Residents
Percent of Live Births with Two or More Major Malformations
Sex by Race/Ethnicity and Residence

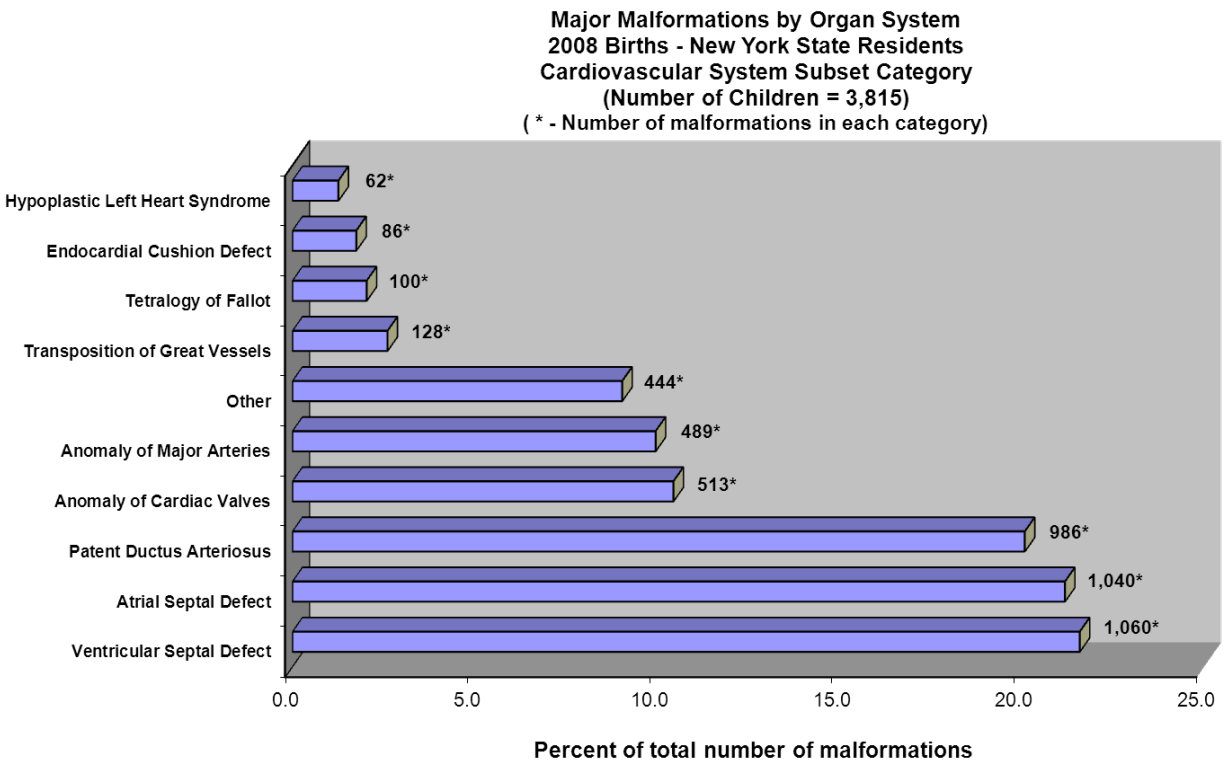
	Both Sexes			Males			Females		
Race and Residence	Infants with birth defects	Total Births	%	Infants with birth defects	Total Births	%	Infants with birth defects	Total Births	%
New York State									
All Races	2,614	246,655	1.1	1,488	126,673	1.2	1,126	119,982	0.9
Non-Hispanic white	1,265	119,196	1.1	725	61,335	1.2	540	57,861	0.9
Non-Hispanic black	518	40,216	1.3	280	20,476	1.4	238	19,740	1.2
Hispanic	590	59,003	1.0	345	30,364	1.1	245	28,639	0.9
Others/Unknown	241	28,240	0.9	138	14,498	1.0	103	13,742	0.7
NYS Excluding NYC									
All Races	1,394	125,071	1.1	798	64,165	1.2	596	60,906	1.0
Non-Hispanic white	941	84,705	1.1	533	43,570	1.2	408	41,135	1.0
Non-Hispanic black	169	12,628	1.3	94	6,485	1.4	75	6,143	1.2
Hispanic	196	19,905	1.0	123	10,215	1.2	73	9,690	0.8
Others/Unknown	88	7,833	1.1	48	3,895	1.2	40	3,938	1.0
New York City									
All Races	1,220	121,584	1.0	690	62,508	1.1	530	59,076	0.9
Non-Hispanic white	324	34,491	0.9	192	17,765	1.1	132	16,726	0.8
Non-Hispanic black	349	27,588	1.3	186	13,991	1.3	163	13,597	1.2
Hispanic	394	39,098	1.0	222	20,149	1.1	172	18,949	0.9
Others/Unknown	153	20,407	0.7	90	10,603	0.8	63	9,804	0.6

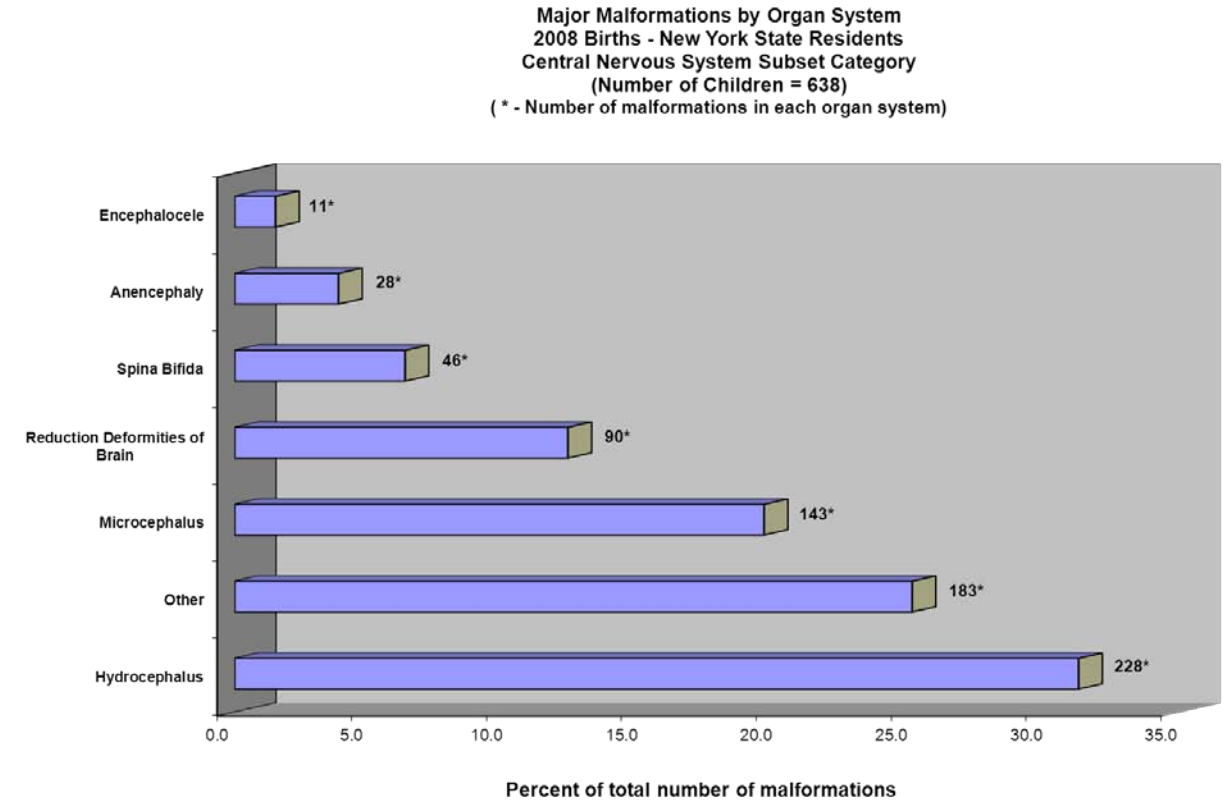
Section II
Major Congenital Malformations by Organ System, 2008

Introduction to Figures

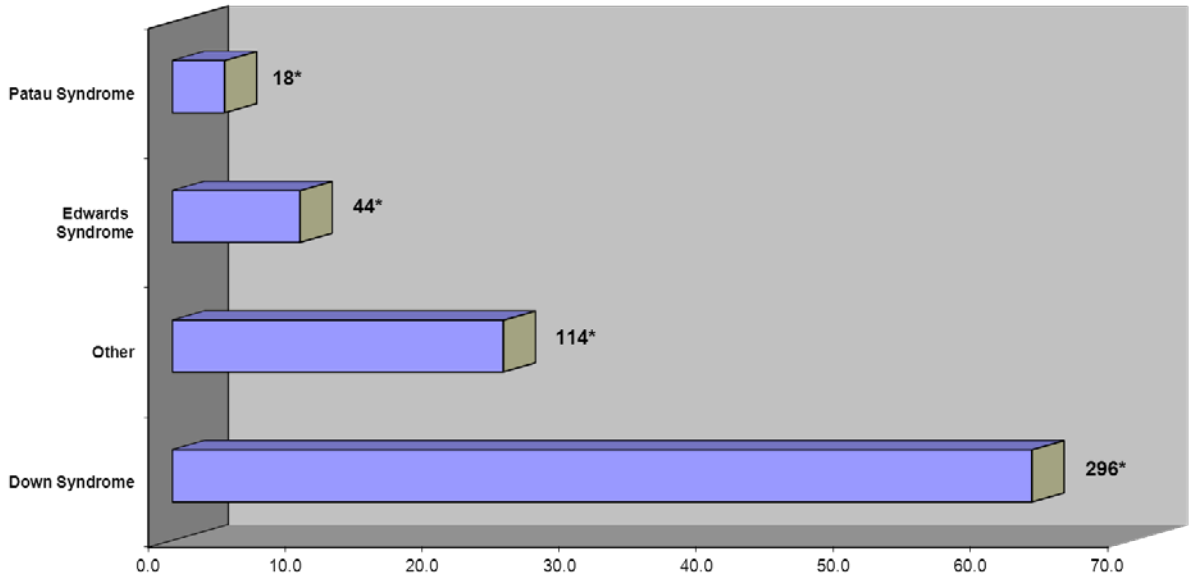
The organ system figures in this section present the distribution of 12 categories of major malformations, the relative contribution of each category to overall prevalence of major malformations in New York State, and the contribution of type of malformation within each subset category. Some of these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


Major Malformations by Organ System
2008 Births - New York State Residents
(Number of Children = 12,462)
(* - Number of malformations in each organ system)
Ear
Integument
Respiratory
Eye
Clefts
Chromosomal
Central Nervous System
Digestive
Other
Musculoskeletal
Genitourinary
Cardiovascular
50*
79*
150*
161*
297*
465*
638*
1,120*
1,786*
2,482*
3,587*
3,815*
0.0
5.0
10.0
15.0
20.0
25.0
30.0
35.0
Percent of total number of malformations


Major Malformations by Organ System
2008 Births - New York State Residents
Cardiovascular System Subset Category
(Number of Children = 3,815)
(* - Number of malformations in each category)
Hypoplastic Left Heart Syndrome
Endocardial Cushion Defect
Tetralogy of Fallot
Transposition of Great Vessels
Other
Anomaly of Major Arteries
Anomaly of Cardiac Valves
Patent Ductus Arteriosus
Atrial Septal Defect
Ventricular Septal Defect
62*
86*
100*
128*
444*
489*
513*
986*
1,040*
1,060*
0.0
5.0
10.0
15.0
20.0
25.0
Percent of total number of malformations

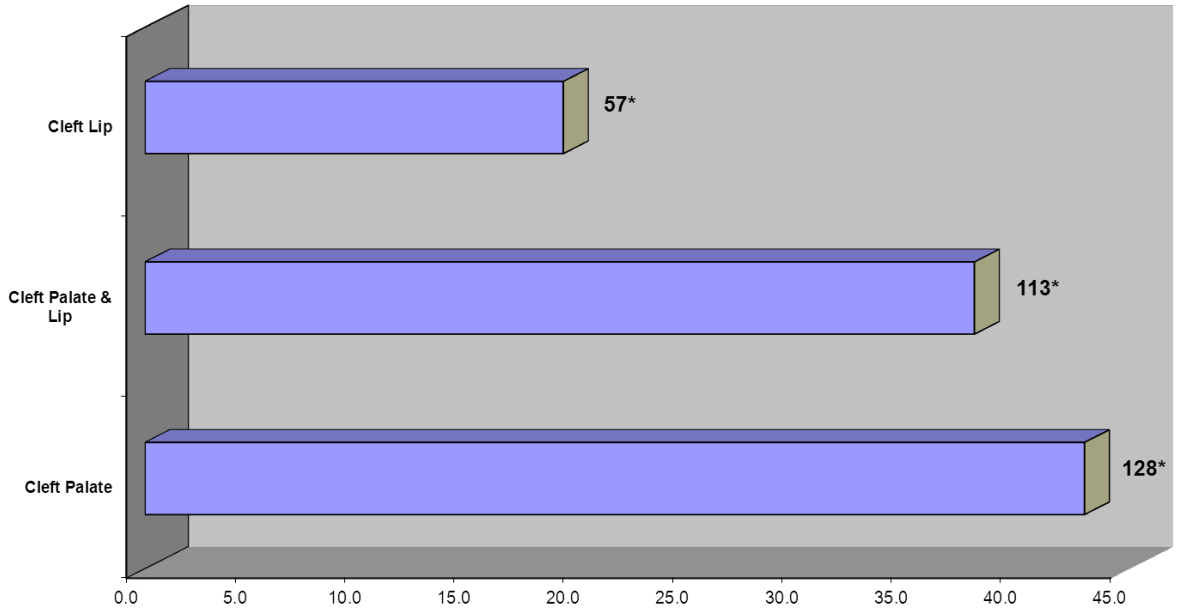

Major Malformations by Organ System
2008 Births - New York State Residents
Central Nervous System Subset Category
(Number of Children = 638)
(* - Number of malformations in each organ system)
Encephalocele
Anencephaly
Spina Bifida
Reduction Deformities of Brain
Microcephalus
Other
Hydrocephalus
11*
28*
46*
90*
143*
183*
228*
0.0
5.0
10.0
15.0
20.0
25.0
30.0
35.0
Percent of total number of malformations

Major Malformations by Organ System
2008 Births - New York State Residents
Chromosomal Subset Category
(Number of Children = 465)
(* - Number of malformations in each organ system)

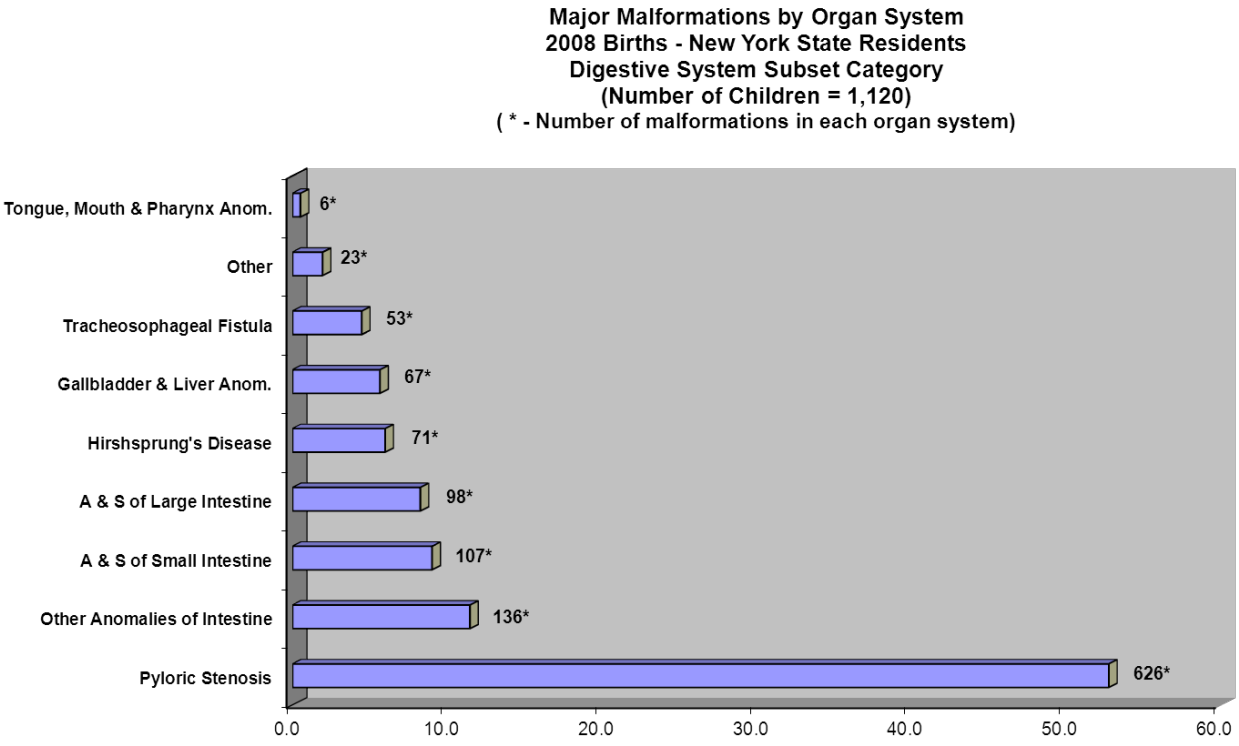


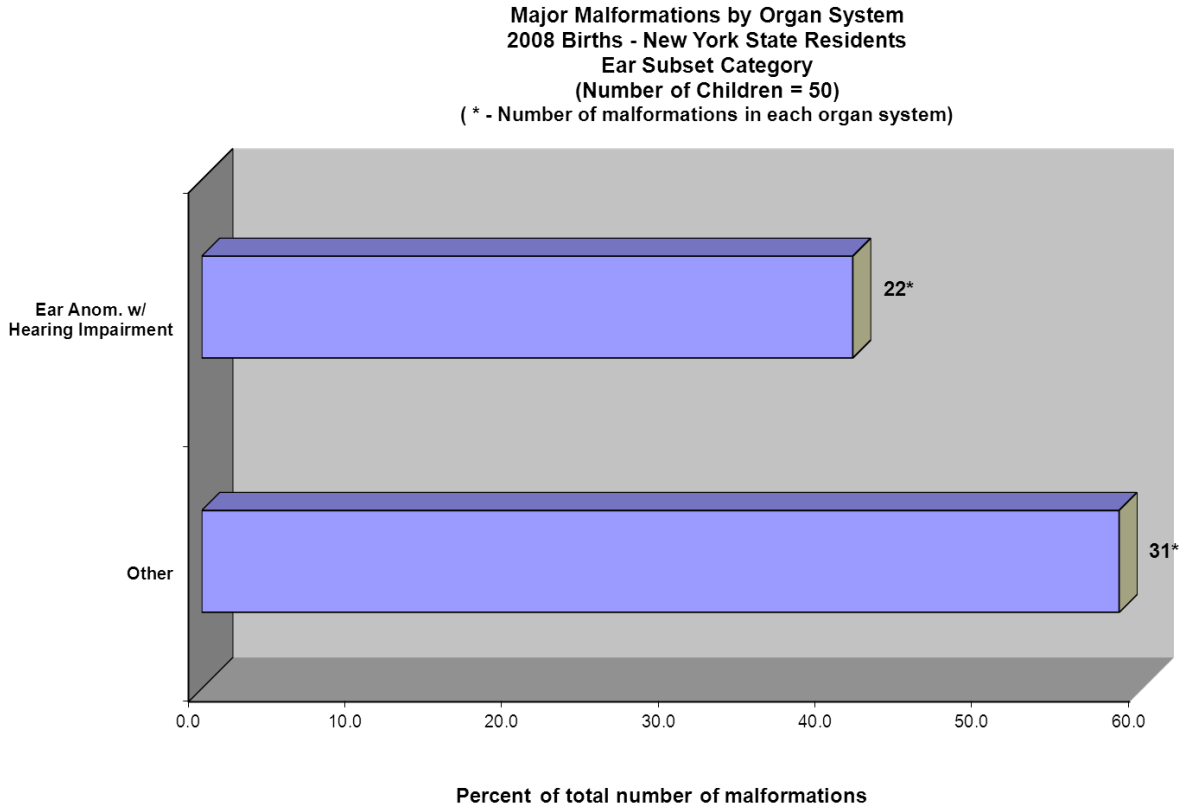
Percent of total number of malformations

Major Malformations by Organ System
2008 Births - New York State Residents
Oral Clefts Subset Category
(Number of Children = 297)
(* - Number of malformations in each organ system)

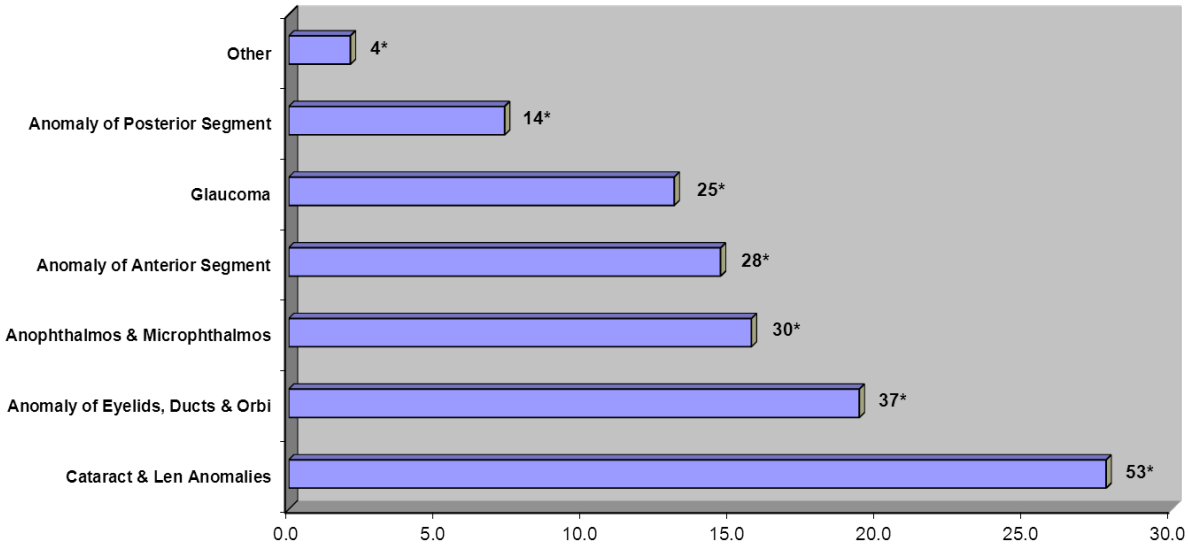


Percent of total number of malformations


Major Malformations by Organ System
2008 Births - New York State Residents
Digestive System Subset Category
(Number of Children = 1,120)
(* - Number of malformations in each organ system)
Tongue, Mouth & Pharynx Anom.
Other
Tracheosophageal Fistula
Gallbladder & Liver Anom.
Hirshsprung's Disease
A & S of Large Intestine
A & S of Small Intestine
Other Anomalies of Intestine
Pyloric Stenosis
6*
23*
53*
67*
71*
98*
107*
136*
626*
0.0
10.0
20.0
30.0
40.0
50.0
60.0
Percent of total number of malformations

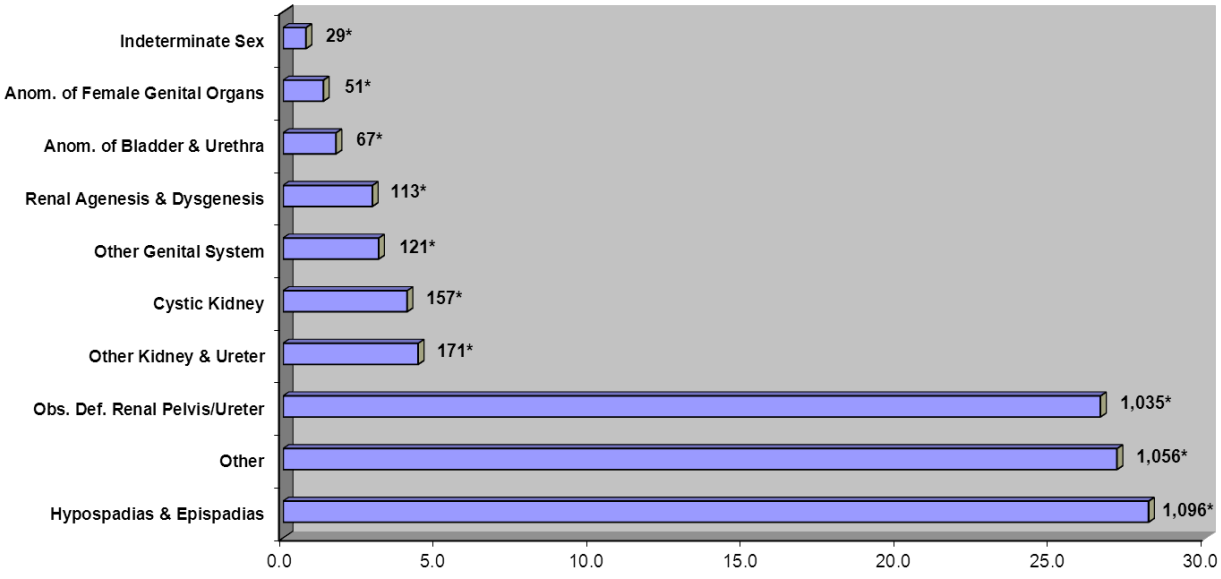

Major Malformations by Organ System
2008 Births - New York State Residents
Ear Subset Category
(Number of Children = 50)
(* - Number of malformations in each organ system)
Ear Anom. w/
Hearing Impairment
Other
22*
31*
0.0
10.0
20.0
30.0
40.0
50.0
60.0
Percent of total number of malformations

Major Malformations by Organ System
2008 Births - New York State Residents
Eye Subset Category
(Number of Children = 161)
(* - Number of malformations in each organ system)



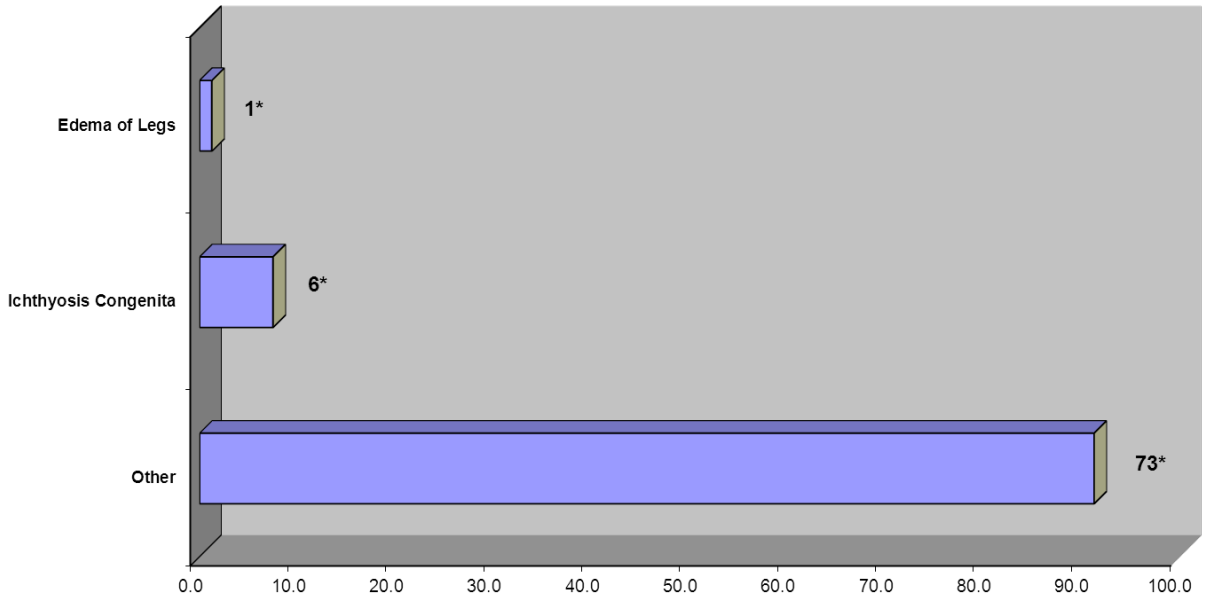
Percent of total number of malformations

Major Malformations by Organ System
2008 Births - New York State Residents
Genitourinary System Subset Category
(Number of Children = 3,587)
(* - Number of malformations in each organ system)

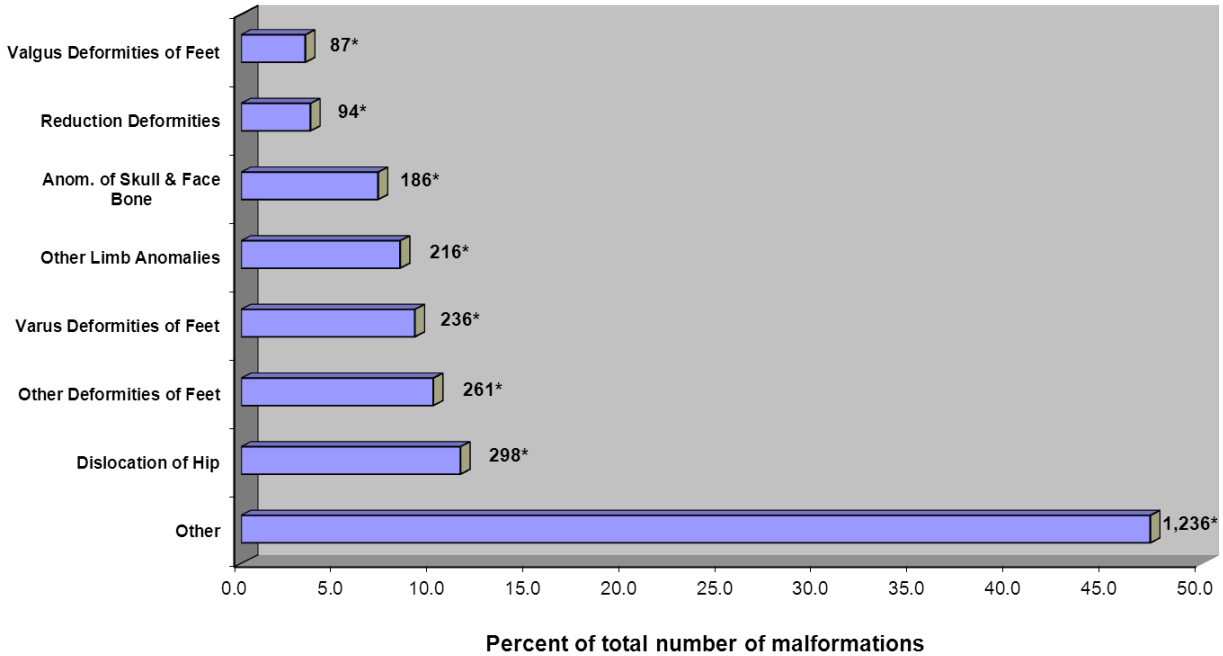


Percent of total number of malformations

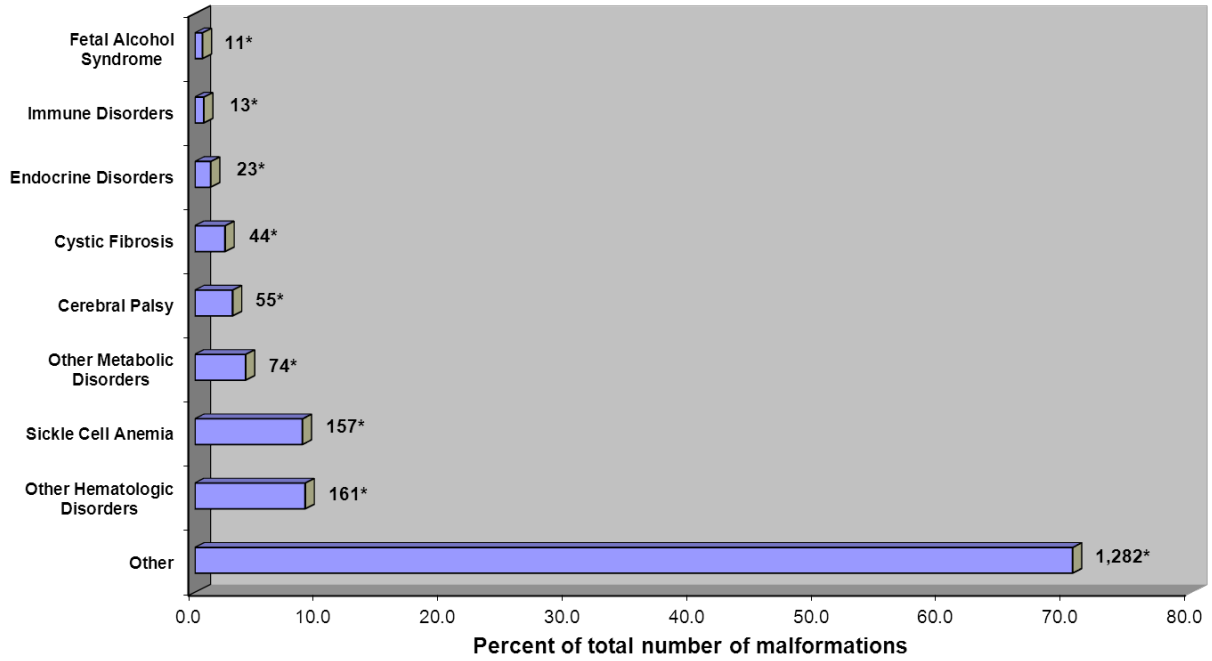
Major Malformations by Organ System
2008 Births - New York State Residents
Integument System Subset Category
(Number of Children = 79)
(* - Number of malformations in each organ system)



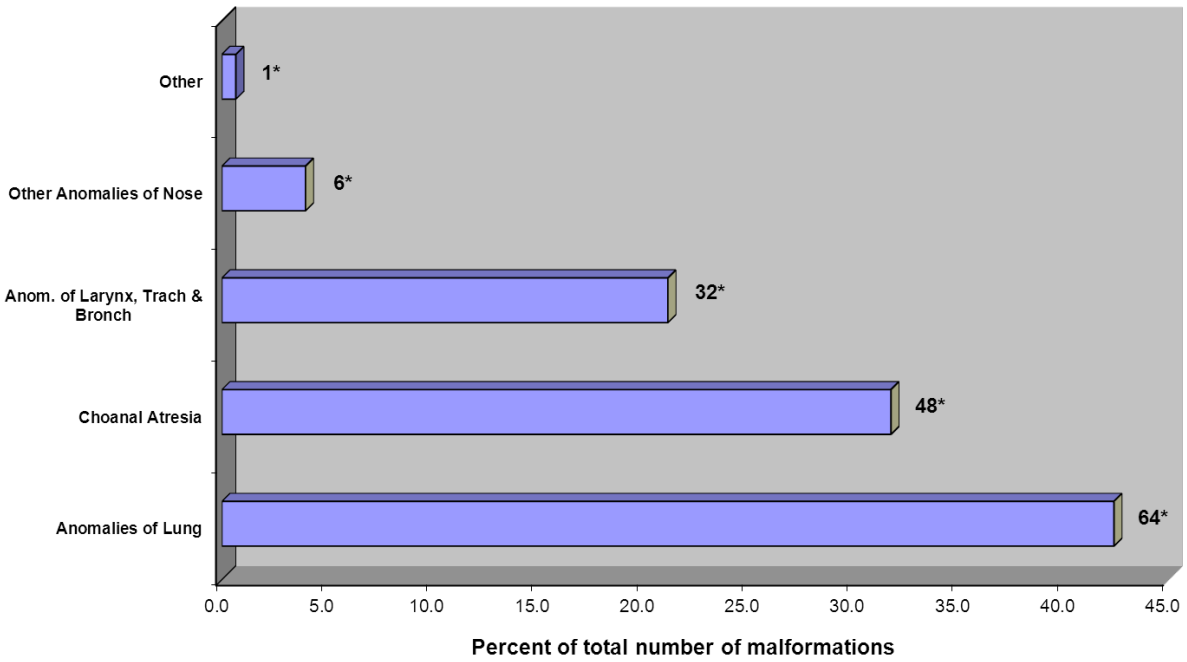
Major Malformations by Organ System
2008 Births - New York State Residents
Musculoskeletal System Subset Category
(Number of Children = 2,482)
(* - Number of malformations in each organ system)



Major Malformations by Organ System
2008 Births - New York State Residents
All Others Subset Category
(Number of Children = 1,786)
(* - Number of malformations in each organ system)



Major Malformations by Organ System
2008 Births - New York State Residents
Respiratory System Subset Category
(Number of Children = 150)
(* - 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

2008 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	133	5.4	5.3	5.5	1.0	4.2	9.2	5.1	5.7
270.1	Phenylketonuria	10	0.4	0.6	0.3	2.2	0.3	0.5	0.2	1.1
277.0	Cystic fibrosis	44	1.8	1.7	1.8	0.9	1.9	1.7	2.2	0.4
282.6	Sickle-cell anemia	161	6.5	6.3	6.8	0.9	0.4	31.1	3.9	2.8
740.0	Anencephalus	11	0.4	0.7	0.2	4.3	0.3	0.5	0.8	0.0
741.0	Spina bifida with hydrocephalus	24	1.0	0.8	1.2	0.7	1.2	1.5	0.5	0.4
741.9	Spina bifida without hydrocephalus	22	0.9	0.9	0.8	1.1	1.0	0.7	0.8	0.7
742.0	Encephalocele	28	1.1	1.2	1.1	1.1	0.8	1.0	1.0	2.8
742.1	Microcephalus	143	5.8	4.9	6.8	0.7	3.5	8.5	8.0	7.1
742.2	Agyria & lissencephaly	5	0.2	0.3	0.1	3.8	0.1	0.0	0.2	1.1
742.2	Anomalies of corpus callosum	58	2.4	2.4	2.3	1.1	2.3	2.2	2.9	1.8
742.2	Holoprosencephaly	13	0.5	0.6	0.5	1.1	0.7	0.5	0.5	0.0
742.3	Congenital hydrocephalus	183	7.4	8.6	6.2	1.4	6.9	9.4	7.3	7.1
742.4	Porencephaly	12	0.5	0.8	0.2	4.7	0.2	0.7	1.2	0.0
742.5	Congenital tethered cord	50	2.0	2.1	1.9	1.1	2.3	1.7	1.7	1.8
743.0	Anophthalmos	4	0.2	0.2	0.2	0.9	0.3	0.2	0.0	0.0
743.1	Microphthalmos	26	1.1	0.8	1.3	0.6	0.6	1.2	2.0	0.7
743.2	Glaucoma	25	1.0	1.1	0.9	1.2	0.4	2.0	1.4	1.4
743.3	Absence of lens	1	0.0	0.0	0.1	0.0	0.1	0.0	0.0	0.0
743.3	Congenital cataract	53	2.1	1.4	2.9	0.5	2.0	3.0	2.2	1.4
743.45	Aniridia	5	0.2	0.2	0.2	1.4	0.1	0.2	0.5	0.0
743.46	Coloboma of iris	5	0.2	0.2	0.2	1.4	0.1	0.0	0.5	0.4
744.0	Anotia/microtia	30	1.2	1.3	1.2	1.1	1.3	0.7	1.4	1.1
745.0	Common truncus	16	0.6	0.9	0.4	2.1	0.7	1.5	0.3	0.0
745.1	Transposition of great vessels	86	3.5	3.8	3.2	1.2	3.4	3.2	3.6	4.2
745.2	Tetralogy of Fallot	128	5.2	5.4	4.9	1.1	5.1	7.0	3.9	5.7
745.3	Common ventricle	20	0.8	0.9	0.8	1.2	0.5	2.0	0.7	0.7
745.4	Ventricular septal defect	1,060	43.0	38.9	47.3	0.8	45.8	44.0	40.7	34.3
745.5	Ostium secundum type atrial septal def.	1,040	42.2	43.0	41.3	1.0	32.7	74.3	37.8	45.3
745.6	Endocardial cushion defects	100	4.1	3.4	4.8	0.7	3.8	7.2	3.2	2.5
746.0	Atresia/stenosis of pulmonary valve	219	8.9	9.3	8.4	1.1	8.1	11.9	8.5	8.5
746.1	Tricuspid atresia/stenosis/hypoplasia	13	0.5	0.7	0.3	2.1	0.3	1.2	0.5	0.7
746.2	Ebstein's anomaly	14	0.6	0.6	0.6	0.9	0.6	0.2	0.5	1.1
746.3	Congenital stenosis of aortic valve	34	1.4	1.9	0.8	2.3	1.7	0.2	1.5	1.4
746.7	Hypoplastic left heart syndrome	62	2.5	3.3	1.7	2.0	3.1	2.2	2.0	1.4
746.85	Anomalies of coronary artery	24	1.0	0.9	1.0	0.9	0.8	2.0	1.0	0.4
747.0	Patent ductus arteriosis	986	40.0	40.3	39.6	1.0	39.0	60.4	30.5	34.7
747.10	Coartation of aorta	148	6.0	6.3	5.7	1.1	6.5	6.5	4.9	5.7
747.41	Total anomalous pulmonary venus connect.	27	1.1	1.3	0.8	1.6	0.8	1.0	1.9	1.1

2008 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	48	1.9	2.2	1.7	1.3	2.6	2.0	1.4	0.4
748.5	Agenesis/hypoplasia of lung	42	1.7	1.9	1.5	1.3	1.4	2.0	2.2	1.4
749.0	Cleft palate	128	5.2	4.2	6.3	0.7	6.2	4.0	4.4	4.2
749.1	Cleft lip	57	2.3	3.4	1.2	2.9	2.4	1.5	2.2	3.2
749.2	Cleft palate & lip	113	4.6	5.5	3.6	1.5	5.5	2.7	4.7	2.8
750.3	Tracheoesophageal fistula etc.	67	2.7	2.4	3.0	0.8	3.1	3.0	2.2	1.8
750.5	Congenital hypertrophic pyloric stenosis	626	25.4	40.3	9.7	4.2	29.7	15.4	28.8	14.2
751.1	Atresia and stenosis of small intestine	98	4.0	3.5	4.5	0.8	3.9	4.0	4.4	3.5
751.2	Atresia and stenosis of rectum or anus	107	4.3	4.7	4.0	1.2	4.4	3.2	5.4	3.2
751.3	Hirschsprungs disease	71	2.9	3.8	1.9	2.0	3.0	3.0	2.2	3.5
751.4	Anomalies of intestinal fixation	65	2.6	2.9	2.3	1.3	3.0	2.5	1.9	2.8
751.61	Biliary atresia	20	0.8	0.8	0.8	0.9	0.7	1.5	0.7	0.7
752.6	Epispadias	37	1.5	2.8	0.1	34.1	1.2	3.5	1.0	1.1
752.6	Hypospadias	1,021	41.4	80.4	0.3	321.4	55.3	33.6	27.8	22.3
753.0	Renal agenesis and dysgenesis	113	4.6	5.1	4.0	1.3	5.5	4.5	4.1	2.1
753.1	Cystic kidney disease	171	6.9	7.7	6.2	1.2	7.0	8.2	7.5	3.9
753.2	Obstructive defect renal pelvis & ureter	1,035	42.0	57.7	25.3	2.3	44.1	30.3	41.2	51.0
753.5	Extrophy of urinary bladder	4	0.2	0.2	0.2	0.9	0.3	0.0	0.0	0.0
753.6	Atresia & stenosis of urethra & bladder	41	1.7	3.2	0.1	37.9	1.1	3.5	1.4	2.1
754.3	Congenital dislocation of hip	203	8.2	3.6	13.1	0.3	9.6	4.0	8.1	8.5
754.51	Talipes equinovarus	139	5.6	5.8	5.5	1.0	5.9	5.5	6.8	2.5
755.2	Reduction deformities of upper limb	72	2.9	3.2	2.6	1.3	3.7	2.0	3.2	0.4
755.3	Reduction deformities of lower limb	29	1.2	1.2	1.2	1.0	1.2	1.7	1.0	0.7
755.8	Arthrogryposis multiplex congenita	19	0.8	0.8	0.8	1.1	0.4	2.2	0.5	0.7
756.0	Craniosynostosis	109	4.4	5.8	2.9	2.0	5.7	2.0	4.6	2.1
756.0	Goldenhar syndrome	13	0.5	0.6	0.4	1.5	0.5	0.0	1.2	0.0
756.4	Chonodrodystrophy	49	2.0	1.9	2.1	0.9	1.7	3.7	2.2	0.4
756.51	Osteogenesis imperfecta	22	0.9	0.9	0.9	0.9	0.7	1.2	0.3	2.5
756.6	Diaphragmatic hernia	65	2.6	2.6	2.7	1.0	3.1	2.5	2.2	1.8
756.7	Gastroschisis	82	3.3	3.6	3.1	1.2	3.4	2.2	4.2	2.8
756.7	Omphalocele	23	0.9	0.9	0.9	1.0	1.1	0.5	1.2	0.4
756.7	Prune belly	5	0.2	0.4	0.0	.	0.2	0.2	0.0	0.7
758.0	Down syndrome	296	12.0	11.5	12.5	0.9	12.0	15.2	12.7	6.0
758.1	Patau syndrome	18	0.7	0.9	0.5	1.9	1.0	0.2	0.8	0.0
758.2	Edwards syndrome	44	1.8	1.1	2.5	0.4	1.2	2.7	2.7	1.1
758.6	Gonadal dysgenesis	17	0.7	0.1	1.3	0.1	0.6	0.5	1.0	0.7
758.7	Klinefelter syndrome	22	0.9	1.7	0.0	.	0.7	0.7	1.5	0.7
759.3	Situs inversus	26	1.1	0.9	1.2	0.8	0.8	2.0	0.7	1.8
760.71	Fetal alcohol syndrome	11	0.4	0.6	0.3	1.7	0.2	0.7	0.8	0.4
762.8	Amniotic bands	11	0.4	0.6	0.3	1.7	0.4	0.7	0.5	0.0
771.1	Congenital cytomegalovirus infection	23	0.9	0.7	1.2	0.6	0.6	2.2	1.0	0.4
771.2	Other congenital infections	49	2.0	2.4	1.6	1.5	2.1	2.0	2.0	1.4

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
2008 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	145	3,071	4.7
Allegany	25	515	4.9
Bronx	1,189	22,998	5.2
Broome	90	2,062	4.4
Cattaraugus	56	988	5.7
Cayuga	43	827	5.2
Chautauqua	90	1,327	6.8
Chemung	60	902	6.7
Chenango	23	536	4.3
Clinton	19	739	2.6
Columbia	22	556	4.0
Cortland	25	574	4.4
Delaware	27	438	6.2
Dutchess	131	2,764	4.7
Erie	681	9,674	7.0
Essex	9	288	3.1
Franklin	12	520	2.3
Fulton	34	611	5.6
Genesee	36	634	5.7
Greene	16	444	3.6
Hamilton	1	31	3.2
Herkimer	31	644	4.8
Jefferson	118	2,129	5.5
Kings	2,231	41,591	5.4
Lewis	20	330	6.1
Livingston	30	592	5.1
Madison	48	710	6.8
Monroe	436	8,709	5.0
Montgomery	48	664	7.2
Nassau	809	15,007	5.4
New York	958	20,249	4.7
Niagara	124	2,262	5.5
Oneida	109	2,608	4.2
Onondaga	318	5,544	5.7
Ontario	59	1,105	5.3
Orange	240	5,173	4.6
Orleans	20	407	4.9
Oswego	78	1,381	5.6
Otsego	17	552	3.1
Putnam	62	933	6.6
Queens	1,335	31,005	4.3
Rensselear	86	1,663	5.2
Richmond	228	5,741	4.0
Rockland	233	4,433	5.3
Saratoga	106	2,287	4.6
Schenectady	99	1,853	5.3
Schoharie	17	328	5.2
Schuyler	7	176	4.0
Seneca	10	364	2.7
St Lawrence	54	1,228	4.4
Steuben	49	1,029	4.8
Suffolk	936	17,947	5.2
Sullivan	39	936	4.2

Section IV – Table 1
2008 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
Tioga	21	355	5.9
Tompkins	31	878	3.5
Ulster	80	1,787	4.5
Warren	37	657	5.6
Washington	23	587	3.9
Wayne	52	1,112	4.7
Westchester	494	10,521	4.7
Wyoming	27	388	7.0
Yates	8	291	2.7

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

Birth Year: 2008

County	ICD-9 Code	Description	Number
ALBANY	752.6	Hypospadias & epispadias	18
	753.2	Obstructive defects of renal pelvis & ureter	15
	752.5	Undescended testicle	13
	745.4	Ventricular septal defect	12
	745.5	Ostium secundum atrial septal defect	10
	747	Patent ductus arteriosus	10
	755	Polydactyly	9
	750.5	Congenital hypertrophic pyloric stenosis	7
	747.1	Coarctation of aorta	4
	754.3	Congenital dislocation of hip	4
ALLEGANY	745.4	Ventricular septal defect	3
	746	Anomalies of pulmonary valve	3
	747.4	Anomalies of great veins	3
	750.5	Congenital hypertrophic pyloric stenosis	2
	754.7	Other deformities of feet	2
	755.6	Other anomalies of lower limb including pelvic girdle	2
	243	Congenital hypothyroidism	1
	273.8	Other disorders of plasma protein	1
	286.3	Congenital deficiency of other clotting factors	1
	742.1	Microcephalus	1
	742.3	Congenital hydrocephalus	1
	745	Common truncus	1
	745.5	Ostium secundum atrial septal defect	1
	745.6	Endocardial cushion defects	1
	746.7	Hypoplastic left heart syndrome	1
	746.8	Other specified anomalies of heart	1
	747	Patent ductus arteriosus	1
	747.2	Other anomalies of aorta	1
	747.3	Anomalies of pulmonary artery	1
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	1
	751.5	Other anomalies of intestine	1
	752.5	Undescended testicle	1
	752.8	Other specified anomalies of genital organs	1
	753.1	Cystic kidney disease	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	754.3	Congenital dislocation of hip	1
	755.5	Other anomalies of upper limb including shoulder girdle	1
	756	Anomalies of skull and face bones	1
	756.7	Anomalies of abdominal wall	1
	759.8	Other specified anomalies	1
BRONX	755	Polydactyly	126
	752.5	Undescended testicle	119
	752.6	Hypospadias & epispadias	94
	745.5	Ostium secundum atrial septal defect	91

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

Birth Year: 2008

County	ICD-9 Code	Description	Number
BRONX	745.4	Ventricular septal defect	86
	753.2	Obstructive defects of renal pelvis & ureter	86
	754.5	Varus deformities of feet	75
	747	Patent ductus arteriosus	58
	282.6	Sickle-cell anemia	47
	750.5	Congenital hypertrophic pyloric stenosis	44
BROOME	745.4	Ventricular septal defect	8
	752.5	Undescended testicle	8
	747	Patent ductus arteriosus	6
	752.6	Hypospadias & epispadias	5
	753.2	Obstructive defects of renal pelvis & ureter	5
	745.5	Ostium secundum atrial septal defect	4
	746.8	Other specified anomalies of heart	4
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	4
	750.5	Congenital hypertrophic pyloric stenosis	4
	751.6	Anomalies of gallbladder, bile ducts, and liver	4
	756	Anomalies of skull and face bones	4
CATTARAUGUS	750.5	Congenital hypertrophic pyloric stenosis	9
	752.5	Undescended testicle	7
	753.2	Obstructive defects of renal pelvis & ureter	5
	745.4	Ventricular septal defect	4
	745.5	Ostium secundum atrial septal defect	4
	752.8	Other specified anomalies of genital organs	4
	747	Patent ductus arteriosus	3
	277	Cystic fibrosis	2
	742.4	Other specified anomalies of brain	2
	746.7	Hypoplastic left heart syndrome	2
	746.8	Other specified anomalies of heart	2
	747.3	Anomalies of pulmonary artery	2
	753	Renal agenesis & dysgenesis	2
	754.5	Varus deformities of feet	2
	756.7	Anomalies of abdominal wall	2
CAYUGA	745.4	Ventricular septal defect	6
	750.5	Congenital hypertrophic pyloric stenosis	6
	753.2	Obstructive defects of renal pelvis & ureter	6
	752.6	Hypospadias & epispadias	4
	742.4	Other specified anomalies of brain	2
	745.1	Transposition of great vessels	2
	746	Anomalies of pulmonary valve	2
	747	Patent ductus arteriosus	2
	749.2	Cleft palate with cleft lip	2
	755	Polydactyly	2
	755.2	Reduction deformities of upper limb	2

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

Birth Year: 2008

County	ICD-9 Code	Description	Number
CHAUTAUQUA	752.6	Hypospadias & epispadias	11
	745.5	Ostium secundum atrial septal defect	10
	750.5	Congenital hypertrophic pyloric stenosis	9
	752.5	Undescended testicle	7
	753.2	Obstructive defects of renal pelvis & ureter	7
	745.4	Ventricular septal defect	6
	752.8	Other specified anomalies of genital organs	5
	746.8	Other specified anomalies of heart	4
	524	Major anomalies of jaw size	3
	742.4	Other specified anomalies of brain	3
	746.6	Congenital mitral insufficiency	3
	747	Patent ductus arteriosus	3
	754.5	Varus deformities of feet	3
	754.7	Other deformities of feet	3
	756	Anomalies of skull and face bones	3
CHEMUNG	752.5	Undescended testicle	11
	753.2	Obstructive defects of renal pelvis & ureter	9
	745.4	Ventricular septal defect	8
	747	Patent ductus arteriosus	5
	243	Congenital hypothyroidism	4
	742.2	Reduction deformities of brain	3
	750.5	Congenital hypertrophic pyloric stenosis	3
	524	Major anomalies of jaw size	2
	745.5	Ostium secundum atrial septal defect	2
	746.8	Other specified anomalies of heart	2
	749.2	Cleft palate with cleft lip	2
	754.3	Congenital dislocation of hip	2
	755	Polydactyly	2
CHENANGO	747.1	Coarctation of aorta	2
	749	Cleft palate	2
	749.2	Cleft palate with cleft lip	2
	752.5	Undescended testicle	2
	752.6	Hypospadias & epispadias	2
	228	Hemangioma, any site	1
	243	Congenital hypothyroidism	1
	333.2	Myoclonus	1
	524	Major anomalies of jaw size	1
	745.4	Ventricular septal defect	1
	745.5	Ostium secundum atrial septal defect	1
	745.6	Endocardial cushion defects	1
	746	Anomalies of pulmonary valve	1
	746.3	Congenital stenosis of aortic valve	1
	746.4	Congenital insufficiency of aortic valve	1
	746.8	Other specified anomalies of heart	1
	747.3	Anomalies of pulmonary artery	1

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

Birth Year: 2008

County	ICD-9 Code	Description	Number
CHENANGO	747.4	Anomalies of great veins	1
	748.5	Agenesis, hypoplasia & dysplasia, lung	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	753	Renal agenesis & dysgenesis	1
	753.1	Cystic kidney disease	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	755	Polydactyly	1
	756	Anomalies of skull and face bones	1
	758	Down syndrome	1
	758.8	Other conditions due to sex chromosome anomalies	1
CLINTON	753.2	Obstructive defects of renal pelvis & ureter	3
	745.4	Ventricular septal defect	2
	747.1	Coarctation of aorta	2
	749	Cleft palate	2
	749.2	Cleft palate with cleft lip	2
	752.6	Hypospadias & epispadias	2
	742.1	Microcephalus	1
	745.1	Transposition of great vessels	1
	745.2	Tetralogy of Fallot	1
	746.4	Congenital insufficiency of aortic valve	1
	747	Patent ductus arteriosus	1
	755.6	Other anomalies of lower limb including pelvic girdle	1
	756.1	Anomalies of spine	1
	756.7	Anomalies of abdominal wall	1
COLUMBIA	750.5	Congenital hypertrophic pyloric stenosis	3
	755	Polydactyly	3
	745.5	Ostium secundum atrial septal defect	2
	747	Patent ductus arteriosus	2
	752.6	Hypospadias & epispadias	2
	243	Congenital hypothyroidism	1
	333.2	Myoclonus	1
	744	Anomalies of ear causing impairment of hearing	1
	747.3	Anomalies of pulmonary artery	1
	751.1	Atresia & stenosis of small intestine	1
	751.7	Anomalies of pancreas	1
	752.5	Undescended testicle	1
	753.7	Anomalies of urachus	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
	756.6	Anomalies of diaphragm	1
	758	Down syndrome	1
	759.5	Tuberous sclerosis	1
	759.8	Other specified anomalies	1

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Birth Year: 2008

County	ICD-9 Code	Description	Number
CORTLAND	752.6	Hypospadias & epispadias	5
	745.4	Ventricular septal defect	4
	747	Patent ductus arteriosus	3
	753.2	Obstructive defects of renal pelvis & ureter	3
	749.2	Cleft palate with cleft lip	2
	752.5	Undescended testicle	2
	754.7	Other deformities of feet	2
	746.4	Congenital insufficiency of aortic valve	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	751.3	Hirschprung's disease & other functional disorders of colon	1
	753	Renal agenesis & dysgenesis	1
	753.3	Other specified anomalies of kidney	1
	753.6	Atresia and stenosis of urethra & bladder neck	1
	755	Polydactyly	1
	756.6	Anomalies of diaphragm	1
	756.7	Anomalies of abdominal wall	1
	758	Down syndrome	1
	758.3	Autosomal deletion syndromes	1
	759.2	Anomalies of other endocrine glands	1
DELAWARE	745.4	Ventricular septal defect	4
	742.5	Other specified anomalies of spinal cord	2
	745.5	Ostium secundum atrial septal defect	2
	746.8	Other specified anomalies of heart	2
	747.1	Coarctation of aorta	2
	749.2	Cleft palate with cleft lip	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	754.7	Other deformities of feet	2
	255.2	Adrenogenital disorders	1
	742.1	Microcephalus	1
	743.1	Microphthalmos	1
	743.3	Congenital cataract & lens anomalies	1
	745.2	Tetralogy of Fallot	1
	746	Anomalies of pulmonary valve	1
	746.2	Ebstein's anomaly	1
	746.4	Congenital insufficiency of aortic valve	1
	747	Patent ductus arteriosus	1
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	1
	751.3	Hirschprung's disease & other functional disorders of colon	1
	751.4	Anomalies of intestinal fixation	1
	752.5	Undescended testicle	1
	752.6	Hypospadias & epispadias	1
	752.7	Indeterminate sex & pseudo-hermaphroditism	1
	752.8	Other specified anomalies of genital organs	1
	753.1	Cystic kidney disease	1
	753.4	Other specified anomalies of ureter	1
	753.7	Anomalies of urachus	1
	754.5	Varus deformities of feet	1

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Birth Year: 2008

County	ICD-9 Code	Description	Number
DELAWARE	756	Anomalies of skull and face bones	1
	756.1	Anomalies of spine	1
	756.3	Other anomalies of ribs and sternum	1
	756.6	Anomalies of diaphragm	1
	759.8	Other specified anomalies	1
DUTCHESS	750.5	Congenital hypertrophic pyloric stenosis	13
	752.6	Hypospadias & epispadias	13
	753.2	Obstructive defects of renal pelvis & ureter	11
	745.4	Ventricular septal defect	9
	755	Polydactyly	8
	752.5	Undescended testicle	7
	747	Patent ductus arteriosus	6
	754.3	Congenital dislocation of hip	6
	754.5	Varus deformities of feet	6
	754.7	Other deformities of feet	5
ERIE	747	Patent ductus arteriosus	74
	752.5	Undescended testicle	64
	752.6	Hypospadias & epispadias	54
	745.4	Ventricular septal defect	52
	750.5	Congenital hypertrophic pyloric stenosis	44
	755	Polydactyly	42
	746.8	Other specified anomalies of heart	31
	745.5	Ostium secundum atrial septal defect	25
	742.4	Other specified anomalies of brain	22
	753.2	Obstructive defects of renal pelvis & ureter	22
ESSEX	745.4	Ventricular septal defect	2
	747	Patent ductus arteriosus	2
	752.5	Undescended testicle	2
	524	Major anomalies of jaw size	1
	742.4	Other specified anomalies of brain	1
	745.1	Transposition of great vessels	1
	745.5	Ostium secundum atrial septal defect	1
	749	Cleft palate	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	754.3	Congenital dislocation of hip	1
	754.7	Other deformities of feet	1
	758.2	Edwards syndrome	1
	758.3	Autosomal deletion syndromes	1
FRANKLIN	752.6	Hypospadias & epispadias	4
	745.5	Ostium secundum atrial septal defect	2
	741	Spina bifida with hydrocephalus	1
	745.4	Ventricular septal defect	1
	746.3	Congenital stenosis of aortic valve	1
	746.6	Congenital mitral insufficiency	1
	747.6	Other anomalies of peripheral vascular system	1

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Birth Year: 2008

County	ICD-9 Code	Description	Number
FRANKLIN	749.2	Cleft palate with cleft lip	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	755.6	Other anomalies of lower limb including pelvic girdle	1
	759.8	Other specified anomalies	1
FULTON	745.4	Ventricular septal defect	4
	752.6	Hypospadias & epispadias	4
	747	Patent ductus arteriosus	2
	747.1	Coarctation of aorta	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	752.4	Anomalies of cervix, vagina & external female genitalia	2
	752.5	Undescended testicle	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	754.5	Varus deformities of feet	2
	754.6	Valgus deformities of feet	2
	771.2	Other congenital infections	2
GENESEE	747	Patent ductus arteriosus	6
	750.5	Congenital hypertrophic pyloric stenosis	4
	742.3	Congenital hydrocephalus	3
	752.5	Undescended testicle	3
	752.6	Hypospadias & epispadias	3
	753.2	Obstructive defects of renal pelvis & ureter	3
	742.2	Reduction deformities of brain	2
	745.5	Ostium secundum atrial septal defect	2
	754.3	Congenital dislocation of hip	2
	758	Down syndrome	2
GREENE	752.5	Undescended testicle	3
	228.1	Lymphangioma, any site	2
	745.5	Ostium secundum atrial septal defect	2
	255.2	Adrenogenital disorders	1
	742.3	Congenital hydrocephalus	1
	742.4	Other specified anomalies of brain	1
	743.6	Congenital anomalies of eyelids, lacrimal system & orbit	1
	745.4	Ventricular septal defect	1
	745.6	Endocardial cushion defects	1
	746.6	Congenital mitral insufficiency	1
	751.4	Anomalies of intestinal fixation	1
	752.6	Hypospadias & epispadias	1
	753.1	Cystic kidney disease	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	754.3	Congenital dislocation of hip	1
	754.7	Other deformities of feet	1
	756	Anomalies of skull and face bones	1
	756.7	Anomalies of abdominal wall	1
	758	Down syndrome	1

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Birth Year: 2008

County	ICD-9 Code	Description	Number
HAMILTON	754.7	Other deformities of feet	1
HERKIMER	745.4	Ventricular septal defect	7
	752.5	Undescended testicle	4
	752.6	Hypospadias & epispadias	3
	748	Choanal atresia	2
	752.7	Indeterminate sex & pseudo-hermaphroditism	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	754.3	Congenital dislocation of hip	2
	758	Down syndrome	2
	271.1	Galactosemia	1
	742.1	Microcephalus	1
	743.4	Coloboma & other anomalies of anterior segment	1
	745.2	Tetralogy of Fallot	1
	745.5	Ostium secundum atrial septal defect	1
	746.8	Other specified anomalies of heart	1
	749	Cleft palate	1
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	751.5	Other anomalies of intestine	1
	751.6	Anomalies of gallbladder, bile ducts, and liver	1
	753.1	Cystic kidney disease	1
	753.3	Other specified anomalies of kidney	1
	754.5	Varus deformities of feet	1
	756.7	Anomalies of abdominal wall	1
	759.3	Situs inversus	1
	759.6	Other hamartoses,nec	1
	759.8	Other specified anomalies	1
JEFFERSON	752.5	Undescended testicle	16
	754.5	Varus deformities of feet	15
	752.6	Hypospadias & epispadias	14
	750.5	Congenital hypertrophic pyloric stenosis	9
	745.4	Ventricular septal defect	8
	742.3	Congenital hydrocephalus	6
	754.3	Congenital dislocation of hip	6
	755	Polydactyly	5
	745.5	Ostium secundum atrial septal defect	3
	746.8	Other specified anomalies of heart	3
	747	Patent ductus arteriosus	3
	747.1	Coarctation of aorta	3
	754.6	Valgus deformities of feet	3
	758	Down syndrome	3
KINGS	747	Patent ductus arteriosus	282
	745.5	Ostium secundum atrial septal defect	280
	745.4	Ventricular septal defect	197
	755	Polydactyly	169
	753.2	Obstructive defects of renal pelvis & ureter	165

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Birth Year: 2008

County	ICD-9 Code	Description	Number
KINGS	752.5	Undescended testicle	164
	752.6	Hypospadias & epispadias	153
	750.5	Congenital hypertrophic pyloric stenosis	84
	746.8	Other specified anomalies of heart	77
	758	Down syndrome	64
LEWIS	752.6	Hypospadias & epispadias	5
	752.5	Undescended testicle	4
	270.1	Phenylketonuria	1
	743.5	Congenital anomalies of posterior segment of eye	1
	745.4	Ventricular septal defect	1
	745.5	Ostium secundum atrial septal defect	1
	746.8	Other specified anomalies of heart	1
	747	Patent ductus arteriosus	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	751.1	Atresia & stenosis of small intestine	1
	753.8	Other specified anomalies of bladder & urethra	1
	754.1	Deformities of sternocleidomastoid muscle	1
	755.5	Other anomalies of upper limb including shoulder girdle	1
	758	Down syndrome	1
LIVINGSTON	747	Patent ductus arteriosus	3
	752.5	Undescended testicle	3
	752.6	Hypospadias & epispadias	3
	753.2	Obstructive defects of renal pelvis & ureter	3
	755.6	Other anomalies of lower limb including pelvic girdle	3
	745.4	Ventricular septal defect	2
	751.1	Atresia & stenosis of small intestine	2
	277	Cystic fibrosis	1
	333.2	Myoclonus	1
	524	Major anomalies of jaw size	1
	744.2	Other specified anomalies of ear	1
	746	Anomalies of pulmonary valve	1
	746.7	Hypoplastic left heart syndrome	1
	746.8	Other specified anomalies of heart	1
	747.3	Anomalies of pulmonary artery	1
	748	Choanal atresia	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	751.3	Hirschprung's disease & other functional disorders of colon	1
	754.5	Varus deformities of feet	1
	756.4	Chondrodystrophy	1
	758.8	Other conditions due to sex chromosome anomalies	1
	762.8	Amniotic bands	1
MADISON	752.5	Undescended testicle	9
	752.6	Hypospadias & epispadias	7
	753.2	Obstructive defects of renal pelvis & ureter	7
	745.4	Ventricular septal defect	5
	524	Major anomalies of jaw size	3

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Birth Year: 2008

County	ICD-9 Code	Description	Number
MADISON	745.5	Ostium secundum atrial septal defect	3
	750.5	Congenital hypertrophic pyloric stenosis	3
	758	Down syndrome	3
	742.1	Microcephalus	2
	746.8	Other specified anomalies of heart	2
	747.1	Coarctation of aorta	2
	749.2	Cleft palate with cleft lip	2
	754.1	Deformities of sternocleidomastoid muscle	2
	754.3	Congenital dislocation of hip	2
	756.6	Anomalies of diaphragm	2
	759.8	Other specified anomalies	2
MONROE	753.2	Obstructive defects of renal pelvis & ureter	54
	752.6	Hypospadias & epispadias	43
	755	Polydactyly	41
	752.5	Undescended testicle	34
	745.4	Ventricular septal defect	29
	750.5	Congenital hypertrophic pyloric stenosis	25
	747	Patent ductus arteriosus	16
	754.7	Other deformities of feet	13
	243	Congenital hypothyroidism	11
	742.3	Congenital hydrocephalus	11
	758	Down syndrome	11
MONTGOMERY	750.5	Congenital hypertrophic pyloric stenosis	7
	752.5	Undescended testicle	5
	752.6	Hypospadias & epispadias	5
	754.5	Varus deformities of feet	5
	753.2	Obstructive defects of renal pelvis & ureter	4
	755	Polydactyly	4
	282.6	Sickle-cell anemia	2
	742.2	Reduction deformities of brain	2
	745.4	Ventricular septal defect	2
	745.5	Ostium secundum atrial septal defect	2
	746.8	Other specified anomalies of heart	2
	752.4	Anomalies of cervix, vagina & external female genitalia	2
	753.4	Other specified anomalies of ureter	2
NASSAU	753.2	Obstructive defects of renal pelvis & ureter	100
	745.4	Ventricular septal defect	96
	747	Patent ductus arteriosus	95
	752.6	Hypospadias & epispadias	71
	745.5	Ostium secundum atrial septal defect	62
	752.5	Undescended testicle	43
	755	Polydactyly	39
	750.5	Congenital hypertrophic pyloric stenosis	38
	752.8	Other specified anomalies of genital organs	26

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County	ICD-9 Code	Description	Number
NEW YORK	745.4	Ventricular septal defect	113
	745.5	Ostium secundum atrial septal defect	100
	752.5	Undescended testicle	85
	747	Patent ductus arteriosus	79
	752.6	Hypospadias & epispadias	75
	753.2	Obstructive defects of renal pelvis & ureter	75
	746.8	Other specified anomalies of heart	46
	755	Polydactyly	44
	750.5	Congenital hypertrophic pyloric stenosis	36
	754.3	Congenital dislocation of hip	26
NIAGARA	752.6	Hypospadias & epispadias	13
	750.5	Congenital hypertrophic pyloric stenosis	11
	752.5	Undescended testicle	11
	745.4	Ventricular septal defect	9
	746.8	Other specified anomalies of heart	6
	747	Patent ductus arteriosus	6
	747.3	Anomalies of pulmonary artery	5
	752.8	Other specified anomalies of genital organs	5
	753.2	Obstructive defects of renal pelvis & ureter	5
	742.4	Other specified anomalies of brain	4
	745.2	Tetralogy of Fallot	4
ONEIDA	752.6	Hypospadias & epispadias	15
	752.5	Undescended testicle	7
	745.4	Ventricular septal defect	6
	750.5	Congenital hypertrophic pyloric stenosis	6
	753.2	Obstructive defects of renal pelvis & ureter	6
	747	Patent ductus arteriosus	5
	754.3	Congenital dislocation of hip	5
	755	Polydactyly	5
	756	Anomalies of skull and face bones	5
	745.5	Ostium secundum atrial septal defect	4
	753.1	Cystic kidney disease	4
	754.7	Other deformities of feet	4
ONONDAGA	752.6	Hypospadias & epispadias	36
	753.2	Obstructive defects of renal pelvis & ureter	36
	745.4	Ventricular septal defect	32
	752.5	Undescended testicle	28
	750.5	Congenital hypertrophic pyloric stenosis	15
	755	Polydactyly	15
	524	Major anomalies of jaw size	10
	754.3	Congenital dislocation of hip	10
	754.7	Other deformities of feet	10
	753.1	Cystic kidney disease	9

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Birth Year: 2008

County	ICD-9 Code	Description	Number
ONTARIO	752.6	Hypospadias & epispadias	8
	752.5	Undescended testicle	7
	753.2	Obstructive defects of renal pelvis & ureter	6
	750.5	Congenital hypertrophic pyloric stenosis	5
	745.4	Ventricular septal defect	4
	754.3	Congenital dislocation of hip	4
	747	Patent ductus arteriosus	3
	755.6	Other anomalies of lower limb including pelvic girdle	3
	745.5	Ostium secundum atrial septal defect	2
	751.5	Other anomalies of intestine	2
	759.8	Other specified anomalies	2
ORANGE	745.4	Ventricular septal defect	29
	745.5	Ostium secundum atrial septal defect	29
	752.6	Hypospadias & epispadias	28
	753.2	Obstructive defects of renal pelvis & ureter	21
	747	Patent ductus arteriosus	20
	750.5	Congenital hypertrophic pyloric stenosis	16
	752.5	Undescended testicle	16
	755	Polydactyly	11
	742.3	Congenital hydrocephalus	10
	746.8	Other specified anomalies of heart	8
ORLEANS	742.5	Other specified anomalies of spinal cord	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	751.1	Atresia & stenosis of small intestine	2
	752.5	Undescended testicle	2
	756	Anomalies of skull and face bones	2
	756.7	Anomalies of abdominal wall	2
	243	Congenital hypothyroidism	1
	747	Patent ductus arteriosus	1
	749	Cleft palate	1
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	1
	751.3	Hirschprung's disease & other functional disorders of colon	1
	751.4	Anomalies of intestinal fixation	1
	751.7	Anomalies of pancreas	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	753.6	Atresia and stenosis of urethra & bladder neck	1
	754.5	Varus deformities of feet	1
	755	Polydactyly	1
	755.6	Other anomalies of lower limb including pelvic girdle	1
	756.5	Osteodystrophies	1
OSWEGO	745.4	Ventricular septal defect	9
	750.5	Congenital hypertrophic pyloric stenosis	8
	753.2	Obstructive defects of renal pelvis & ureter	8
	752.5	Undescended testicle	6
	754.7	Other deformities of feet	6

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County	ICD-9 Code	Description	Number
OSWEGO	752.6	Hypospadias & epispadias	5
	747	Patent ductus arteriosus	4
	758	Down syndrome	4
	746.8	Other specified anomalies of heart	3
	524	Major anomalies of jaw size	2
	745.5	Ostium secundum atrial septal defect	2
	745.6	Endocardial cushion defects	2
	749.2	Cleft palate with cleft lip	2
	753.1	Cystic kidney disease	2
	754.3	Congenital dislocation of hip	2
	755	Polydactyly	2
	771.1	Congenital cytomegalovirus infection	2
OTSEGO	752.6	Hypospadias & epispadias	4
	754.7	Other deformities of feet	3
	745.5	Ostium secundum atrial septal defect	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	282	Hereditary spherocytosis	1
	745.4	Ventricular septal defect	1
	745.6	Endocardial cushion defects	1
	746.6	Congenital mitral insufficiency	1
	751.1	Atresia & stenosis of small intestine	1
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	1
	751.4	Anomalies of intestinal fixation	1
	751.8	Other specified anomalies of digestive system	1
	753	Renal agenesis & dysgenesis	1
	754.5	Varus deformities of feet	1
	755	Polydactyly	1
	756.7	Anomalies of abdominal wall	1
	758.7	Klinefelters syndrome	1
PUTNAM	742.4	Other specified anomalies of brain	8
	752.6	Hypospadias & epispadias	8
	745.4	Ventricular septal defect	6
	747.3	Anomalies of pulmonary artery	5
	753.2	Obstructive defects of renal pelvis & ureter	5
	745.5	Ostium secundum atrial septal defect	4
	750.5	Congenital hypertrophic pyloric stenosis	4
	752.5	Undescended testicle	4
	746.8	Other specified anomalies of heart	3
	747.1	Coarctation of aorta	3
QUEENS	745.5	Ostium secundum atrial septal defect	131
	753.2	Obstructive defects of renal pelvis & ureter	129
	752.5	Undescended testicle	121
	745.4	Ventricular septal defect	91
	752.6	Hypospadias & epispadias	91
	747	Patent ductus arteriosus	85
	755	Polydactyly	76

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County	ICD-9 Code	Description	Number
QUEENS	750.5	Congenital hypertrophic pyloric stenosis	56
	746.8	Other specified anomalies of heart	43
	754.3	Congenital dislocation of hip	38
RENSSELAER	752.5	Undescended testicle	8
	752.6	Hypospadias & epispadias	8
	745.5	Ostium secundum atrial septal defect	7
	747	Patent ductus arteriosus	6
	750.5	Congenital hypertrophic pyloric stenosis	6
	753.2	Obstructive defects of renal pelvis & ureter	6
	746	Anomalies of pulmonary valve	5
	755	Polydactyly	5
	754.3	Congenital dislocation of hip	4
	745.4	Ventricular septal defect	3
	747.3	Anomalies of pulmonary artery	3
	755.6	Other anomalies of lower limb including pelvic girdle	3
	758	Down syndrome	3
RICHMOND	752.6	Hypospadias & epispadias	28
	752.5	Undescended testicle	27
	753.2	Obstructive defects of renal pelvis & ureter	20
	747	Patent ductus arteriosus	17
	745.4	Ventricular septal defect	15
	750.5	Congenital hypertrophic pyloric stenosis	12
	745.5	Ostium secundum atrial septal defect	10
	755	Polydactyly	9
	758	Down syndrome	9
	749.2	Cleft palate with cleft lip	5
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	5
ROCKLAND	745.5	Ostium secundum atrial septal defect	36
	745.4	Ventricular septal defect	32
	752.6	Hypospadias & epispadias	23
	747	Patent ductus arteriosus	18
	752.5	Undescended testicle	15
	753.2	Obstructive defects of renal pelvis & ureter	15
	755	Polydactyly	11
	758	Down syndrome	7
	746.8	Other specified anomalies of heart	6
	747.1	Coarctation of aorta	6
	749	Cleft palate	6
SARATOGA	752.6	Hypospadias & epispadias	18
	753.2	Obstructive defects of renal pelvis & ureter	12
	752.5	Undescended testicle	7
	745.4	Ventricular septal defect	6
	747	Patent ductus arteriosus	5
	750.5	Congenital hypertrophic pyloric stenosis	5
	754.3	Congenital dislocation of hip	5

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County	ICD-9 Code	Description	Number
SARATOGA	745.5	Ostium secundum atrial septal defect	4
	746.8	Other specified anomalies of heart	4
	757.3	Other specified anomalies of skin	4
	758	Down syndrome	4
SCHENECTADY	752.6	Hypospadias & epispadias	17
	747	Patent ductus arteriosus	8
	752.5	Undescended testicle	8
	753.2	Obstructive defects of renal pelvis & ureter	8
	755	Polydactyly	8
	750.5	Congenital hypertrophic pyloric stenosis	6
	754.3	Congenital dislocation of hip	5
	756	Anomalies of skull and face bones	5
	756.7	Anomalies of abdominal wall	5
	746	Anomalies of pulmonary valve	4
SCHOHARIE	745.4	Ventricular septal defect	3
	752.5	Undescended testicle	3
	752.6	Hypospadias & epispadias	3
	753.1	Cystic kidney disease	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	754.7	Other deformities of feet	2
	524	Major anomalies of jaw size	1
	742.1	Microcephalus	1
	742.3	Congenital hydrocephalus	1
	748	Choanal atresia	1
	749.2	Cleft palate with cleft lip	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	752.8	Other specified anomalies of genital organs	1
	757.6	Specified anomalies of breast	1
SCHUYLER	747	Patent ductus arteriosus	3
	228	Hemangioma, any site	2
	746.8	Other specified anomalies of heart	1
	752.6	Hypospadias & epispadias	1
	756.3	Other anomalies of ribs and sternum	1
	756.6	Anomalies of diaphragm	1
SENECA	750.5	Congenital hypertrophic pyloric stenosis	3
	753.2	Obstructive defects of renal pelvis & ureter	2
	746	Anomalies of pulmonary valve	1
	746.1	Tricuspid atresia & stenosis	1
	746.4	Congenital insufficiency of aortic valve	1
	747.1	Coarctation of aorta	1
	752.6	Hypospadias & epispadias	1
	755.2	Reduction deformities of upper limb	1
	771.2	Other congenital infections	1

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

Birth Year: 2008

County	ICD-9 Code	Description	Number
ST LAWRENCE	750.5	Congenital hypertrophic pyloric stenosis	8
	752.6	Hypospadias & epispadias	8
	754.5	Varus deformities of feet	6
	753.2	Obstructive defects of renal pelvis & ureter	5
	745.4	Ventricular septal defect	4
	747	Patent ductus arteriosus	3
	749	Cleft palate	3
	524	Major anomalies of jaw size	2
	746	Anomalies of pulmonary valve	2
	747.3	Anomalies of pulmonary artery	2
	754.7	Other deformities of feet	2
	756	Anomalies of skull and face bones	2
STEUBEN	752.5	Undescended testicle	9
	753.2	Obstructive defects of renal pelvis & ureter	7
	752.6	Hypospadias & epispadias	5
	750.5	Congenital hypertrophic pyloric stenosis	4
	745.4	Ventricular septal defect	3
	746	Anomalies of pulmonary valve	3
	749.2	Cleft palate with cleft lip	2
	754.3	Congenital dislocation of hip	2
	758	Down syndrome	2
SUFFOLK	745.5	Ostium secundum atrial septal defect	134
	752.6	Hypospadias & epispadias	96
	745.4	Ventricular septal defect	83
	747	Patent ductus arteriosus	76
	752.5	Undescended testicle	75
	753.2	Obstructive defects of renal pelvis & ureter	65
	750.5	Congenital hypertrophic pyloric stenosis	45
	755	Polydactyly	37
	747.3	Anomalies of pulmonary artery	35
	755.6	Other anomalies of lower limb including pelvic girdle	26
SULLIVAN	750.5	Congenital hypertrophic pyloric stenosis	7
	752.6	Hypospadias & epispadias	4
	753.2	Obstructive defects of renal pelvis & ureter	4
	745.4	Ventricular septal defect	3
	243	Congenital hypothyroidism	2
	746.8	Other specified anomalies of heart	2
	747	Patent ductus arteriosus	2
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	2
	756	Anomalies of skull and face bones	2
	228	Hemangioma, any site	1
	524	Major anomalies of jaw size	1
	742.1	Microcephalus	1
	742.2	Reduction deformities of brain	1
	744	Anomalies of ear causing impairment of hearing	1
	744.8	Other specified anomalies of face & neck	1

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

Birth Year: 2008

County	ICD-9 Code	Description	Number
SULLIVAN	745	Common truncus	1
	745.5	Ostium secundum atrial septal defect	1
	746.3	Congenital stenosis of aortic valve	1
	746.4	Congenital insufficiency of aortic valve	1
	746.5	Congenital mitral stenosis	1
	746.6	Congenital mitral insufficiency	1
	749	Cleft palate	1
	751.1	Atresia & stenosis of small intestine	1
	752.5	Undescended testicle	1
	753	Renal agenesis & dysgenesis	1
	753.1	Cystic kidney disease	1
	753.6	Atresia and stenosis of urethra & bladder neck	1
	754.3	Congenital dislocation of hip	1
	754.5	Varus deformities of feet	1
	754.7	Other deformities of feet	1
	754.8	Other specified nonteratogenic anomalies	1
	755.2	Reduction deformities of upper limb	1
	755.3	Reduction deformities of lower limb	1
	756.1	Anomalies of spine	1
	756.4	Chondrodystrophy	1
	758	Down syndrome	1
	758.1	Patau syndrome	1
	758.3	Autosomal deletion syndromes	1
	758.5	Other conditions due autosomal anomalies	1
TIOGA	752.5	Undescended testicle	4
	745.4	Ventricular septal defect	2
	747	Patent ductus arteriosus	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	755.2	Reduction deformities of upper limb	2
	243	Congenital hypothyroidism	1
	253.2	Panhypopituitarism	1
	273.8	Other disorders of plasma protien	1
	742.1	Microcephalus	1
	742.2	Reduction deformities of brain	1
	742.3	Congenital hydrocephalus	1
	747.1	Coarctation of aorta	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	751.4	Anomalies of intestinal fixation	1
	753.1	Cystic kidney disease	1
	754.5	Varus deformities of feet	1
	756.1	Anomalies of spine	1
	756.6	Anomalies of diaphragm	1
	756.7	Anomalies of abdominal wall	1
	758.5	Other conditions due autosomal anomalies	1
	759.8	Other specified anomalies	1

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

Birth Year: 2008

County	ICD-9 Code	Description	Number
TOMPKINS	745.4	Ventricular septal defect	4
	746.8	Other specified anomalies of heart	3
	750.5	Congenital hypertrophic pyloric stenosis	3
	753.2	Obstructive defects of renal pelvis & ureter	3
	745.5	Ostium secundum atrial septal defect	2
	746.4	Congenital insufficiency of aortic valve	2
	752.6	Hypospadias & epispadias	2
	754.6	Valgus deformities of feet	2
	755	Polydactyly	2
	756	Anomalies of skull and face bones	2
	756.1	Anomalies of spine	2
ULSTER	752.6	Hypospadias & epispadias	9
	753.2	Obstructive defects of renal pelvis & ureter	8
	742.4	Other specified anomalies of brain	5
	752.5	Undescended testicle	5
	742.3	Congenital hydrocephalus	3
	745.4	Ventricular septal defect	3
	749.2	Cleft palate with cleft lip	3
	750.5	Congenital hypertrophic pyloric stenosis	3
	754.3	Congenital dislocation of hip	3
	755	Polydactyly	3
	756.4	Chondrodystrophy	3
	756.7	Anomalies of abdominal wall	3
	758	Down syndrome	3
WARREN	752.6	Hypospadias & epispadias	6
	750.5	Congenital hypertrophic pyloric stenosis	4
	745.5	Ostium secundum atrial septal defect	2
	747	Patent ductus arteriosus	2
	750.6	Congenital hiatus hernia	2
	755	Polydactyly	2
	270.1	Phenylketonuria	1
	742.2	Reduction deformities of brain	1
	742.3	Congenital hydrocephalus	1
	742.5	Other specified anomalies of spinal cord	1
	745.2	Tetralogy of Fallot	1
	745.4	Ventricular septal defect	1
	746	Anomalies of pulmonary valve	1
	746.4	Congenital insufficiency of aortic valve	1
	747.3	Anomalies of pulmonary artery	1
	750.2	Other specified anomalies, mouth and pharynx	1
	751.4	Anomalies of intestinal fixation	1
	751.7	Anomalies of pancreas	1
	752.4	Anomalies of cervix, vagina & external female genitalia	1
	752.5	Undescended testicle	1
	752.8	Other specified anomalies of genital organs	1
	753.1	Cystic kidney disease	1

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

Birth Year: 2008

County	ICD-9 Code	Description	Number
WARREN	753.2	Obstructive defects of renal pelvis & ureter	1
	753.4	Other specified anomalies of ureter	1
	754.3	Congenital dislocation of hip	1
	754.4	Congenital genu recurvatum & bowing of long bones of leg	1
	754.5	Varus deformities of feet	1
	755.2	Reduction deformities of upper limb	1
	756.4	Chondrodystrophy	1
	757.3	Other specified anomalies of skin	1
	758	Down syndrome	1
	758.3	Autosomal deletion syndromes	1
WASHINGTON	752.6	Hypospadias & epispadias	5
	750.5	Congenital hypertrophic pyloric stenosis	3
	749	Cleft palate	2
	752.5	Undescended testicle	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	754.3	Congenital dislocation of hip	2
	741	Spina bifida with hydrocephalus	1
	742.1	Microcephalus	1
	742.3	Congenital hydrocephalus	1
	745.5	Ostium secundum atrial septal defect	1
	747	Patent ductus arteriosus	1
	747.3	Anomalies of pulmonary artery	1
	750.2	Other specified anomalies, mouth and pharynx	1
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	1
	752.4	Anomalies of cervix, vagina & external female genitalia	1
	752.8	Other specified anomalies of genital organs	1
	755	Polydactyly	1
	756.5	Osteodystrophies	1
	756.8	Other specified anomalies of muscle, tendon, fascia, etc.	1
WAYNE	752.6	Hypospadias & epispadias	5
	753.2	Obstructive defects of renal pelvis & ureter	5
	752.5	Undescended testicle	4
	745.2	Tetralogy of Fallot	3
	754.5	Varus deformities of feet	3
	754.7	Other deformities of feet	3
	228	Hemangioma, any site	2
	243	Congenital hypothyroidism	2
	742.3	Congenital hydrocephalus	2
	745.4	Ventricular septal defect	2
	745.5	Ostium secundum atrial septal defect	2
	746.8	Other specified anomalies of heart	2
	749.2	Cleft palate with cleft lip	2
	755	Polydactyly	2
	756.7	Anomalies of abdominal wall	2
	757.3	Other specified anomalies of skin	2
	758	Down syndrome	2

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

Birth Year: 2008

County	ICD-9 Code	Description	Number
WESTCHESTER	753.2	Obstructive defects of renal pelvis & ureter	51
	752.6	Hypospadias & epispadias	45
	747	Patent ductus arteriosus	37
	745.5	Ostium secundum atrial septal defect	35
	752.5	Undescended testicle	32
	745.4	Ventricular septal defect	30
	755	Polydactyly	27
	750.5	Congenital hypertrophic pyloric stenosis	25
	746.8	Other specified anomalies of heart	21
	757.3	Other specified anomalies of skin	17
WYOMING	747	Patent ductus arteriosus	4
	745.4	Ventricular septal defect	3
	752.5	Undescended testicle	3
	751.3	Hirschprung's disease & other functional disorders of colon	2
	756.7	Anomalies of abdominal wall	2
	743.3	Congenital cataract & lens anomalies	1
	746	Anomalies of pulmonary valve	1
	746.6	Congenital mitral insufficiency	1
	746.8	Other specified anomalies of heart	1
	747.2	Other anomalies of aorta	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	753.1	Cystic kidney disease	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	754.3	Congenital dislocation of hip	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
	755.6	Other anomalies of lower limb including pelvic girdle	1
	756.8	Other specified anomalies of muscle, tendon, fascia, etc.	1
	758.8	Other conditions due to sex chromosome anomalies	1
YATES	755.6	Other anomalies of lower limb including pelvic girdle	2
	745.5	Ostium secundum atrial septal defect	1
	745.6	Endocardial cushion defects	1
	748.4	Congenital cystic lung	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	754.8	Other specified nonteratogenic anomalies	1
	756.1	Anomalies of spine	1
	759.6	Other hamartoses,nec	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 prevalence for most defects are significantly different for New York State excluding New York City and for New York City, except hypoplastic left heart syndrome, cleft lip with and without cleft palate, upper limb deformity, gastroschisis, and diaphragmatic hernia. However, the CMR prevalences are equal to or higher than the lower boundary of the actual range of the 11 registries for 9 of the 21 defects (bold prevalences).

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 are 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.

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: 2006-2008)

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	2.2	3.7
Encephalocele	**0.8**	**0.7**	**0.8**	0.8
Eye defects				
Anophthalmia/ microphthalmia	1.1	1.6	1.3	2.1
Cardiovascular defects				
Common truncus	0.4	**0.9**	**0.7**	0.7
Transposition of great arteries	**4.1**	**4.6**	**4.4**	3.0
Tetralogy of Fallot	**5.1**	**4.9**	**5.0**	4.1
Atrioventricular septal defect, AVSD (Endocardial cushion defect)	3.5	3.7	3.6	4.7
Hypoplastic left heart syndrome	**2.0**	**3.3**	**2.7**	2.3
Orofacial defects				
Cleft palate without cleft lip	5.5	**6.7**	**6.1**	6.5
Cleft lip with and without cleft palate	6.5	8.3	7.5	10.9
Gastrointestinal defects				
Esophageal atresia/ tracheosophageal fistula	**2.3**	**3.0**	**2.7**	2.1
Rectal and large intestinal atresia/stenosis	4.1	4.2	4.2	4.9
Musculoskeletal defects				
Reduction deformity, upper limbs	1.6	2.8	2.2	3.6
Reduction deformity, lower limbs	0.7	1.1	0.9	1.7
Gastroschisis	1.9	3.5	2.8	4.7
Omphalocele	1.1	1.3	1.2	1.9
Diaphragmatic hernia	**2.2**	**3.4**	**2.8**	2.6
Chromosomal defects				
Trisomy 13	0.9	0.8	0.8	1.2
Trisomy 21 (Down syndrome)	12.2	**13.4**	**12.8**	13.5
Trisomy 18	1.4	1.4	1.4	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 Res A Clin Mol Teratol*, 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. *MMWR* 1993; 42(No. SS-1):1-7.

Section VI
Current Topics

1. A Retrospective Cohort Study of Mortality Among Children with Birth Defects in New York State, 1983-2006 [1]

Background: Birth defects, which occur in about 3% of live births in the United States each year, have remained the leading cause of infant mortality, although the infant mortality rate in the United States has decreased significantly over the past decades. The objective of this study was to conduct a retrospective cohort study with long-term follow-up of the children in New York State Congenital Malformations Registry (CMR) for up to 25 years on their mortality status to evaluate the mortality risk of children with birth defects by age at death, birth defect category and other possible contributing factors.

Methods: Children born in 1983-2006 with reportable birth defects were included in the study cohort. A sub-cohort was also constructed containing children born in 1983-2006 with selected major birth defects that are relatively severe congenital conditions identified at birth. New York State live births in 1983-2006 were used as the comparison population. The deaths among the study cohort were identified through linking CMR cases to the death certificate files. A Poisson regression model was used to calculate mortality rate ratios (mortality risk) adjusting for selected infant and maternal risk factors.

Results: Compared to children without birth defects, the overall mortality risk was 6.7 times higher for children with any of the reportable birth defects and 23.6 times higher for children with selected major birth defects. The mortality risk decreased with increasing age of children. The top 5 most significantly high mortality risks were found among infants in neonatal period with anencephaly (RR=200.5, 95% CI:180.6-222.5) and renal agenesis/dysgenesis (RR=69.5, 95% CI:64.2-75.2), and among children aged 1- 2 years with Trisomy 18 (RR=206.4, 95% CI:28.5-331.6), Trisomy 13 (RR=157.3, 95% CI:83.5-296.4), and hypoplastic left heart (RR=161.5, 95% CI:20.1-217.2). Low birth weight and gestational age, multiple malformations and high lethality of anomalies involved were the major contributors to the high risk of mortality among children with selected major birth defects compared to those without birth defects.

Conclusions: Using the state-wide, population-based birth defects surveillance data to conduct a long-term follow-up of a large cohort of New York children, we were able to examine mortality and survival experience of the affected children during infancy and childhood period by individual birth defects of interest. 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.

2. 25-year Survival of Children with Birth Defects in New York State: A Population-based Study [2]

Background: Few studies have been conducted on long-term survival of children with major birth defects due to lack of longitudinal birth defects surveillance data. The objective of this study was to conduct a 25-year survival analysis among New York children born with major defects by survival age, birth defect category and other possible contributing factors.

Methods: A cohort was constructed containing children born in 1983-2006 with selected major birth defects. Deaths among the study cohort were identified by matching the children to their death certificates. The survival probability was estimated by Kaplan-Meier methods. Cox proportional hazards regression was used to examine the effect of the risk factors on survival.

Results: A total of 9,112 deaths were identified among 57,002 live births during 1983-2006 with selected birth defects. The overall 25-year survival probability of the study cohort was 82.51% (95% CI, 82.11%-82.89%). The estimated survival probability was comparable to that reported from previous studies with regard to individual defects including spina bifida, encephalocele, atrioventricular septal defects, tracheoesophageal fistula and esophageal atresia/stenosis, renal agenesis or dysgenesis, lower limbs reduction, diaphragmatic hernia, abdominal wall defects and Down syndrome. Sex, low birth weight/gestational age, existence of multiple birth defects (non-isolated) and maternal age and nativity were identified as risk factors.

Conclusions: 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.

References

1. Wang Y., Hu J, Druschel CM. A Retrospective Cohort Study of Mortality Among Children with Birth Defects in New York State, 1983-2006. *Birth Defects Res A Clin Mol Teratol*. 2010; 88:1023-31.

2. 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; 91:995-1003.

Section VII

Current Publications

1. Agopian AJ, Lupo PJ, Herdt-Losavio ML, Langlois PH, Rocheleau CM, Mitchell LE, and the National Birth Defects Prevention Study. Differences in folic acid use, prenatal care, smoking, and drinking in early pregnancy by occupation. *Prev Med* 2012;55(4):341-345. DOI:10.1016/j.ypmed.2012.07.015.

   Several occupational groups were identified in which women were more likely to smoke, consume alcohol, not use folic acid, or not have prenatal care during early pregnancy. Discovery of these associations identifies high-risk occupational groups that could be targeted for health promotion activities (e.g., workplace interventions). Better understanding of characteristics that motivate high-risk behaviors could further inform workplace health promotion activities. The development of successful workplace health promotion strategies that reduce the prevalence of high-risk behaviors could both improve maternal and child health and reduce healthcare costs.

2. Browne ML, Carter TC, Kay DM, Kuehn D, Brody LC, Romitti PA, Liu A, Caggana M, Druschel CM, Mills JL. Evaluation of genes involved in limb development, angiogenesis, and coagulation as risk factors for congenital limb deficiencies. *Am J Med Genet A* 2012 Oct;158A(10):2463-72.

   Genotypes were obtained for 132 SNPs in genes involved in limb development (SHH, WNT7A, FGF4, FGF8, FGF10, TBX3, TBX5, SALL4, GREM1, GDF5, CTNNB1, EN1, CYP26A1, CYP26B1), angiogenesis (VEGFA, HIF1A, NOS3), and coagulation (F2, F5, MTHFR). Our data suggest that common variants in FGF10 increase the risk for a wide range of non-syndromic limb deficiencies. We also observed suggestive evidence for associations with SNPs in other genes including CYP26B1 and WNT7A.

3. Carter TC, Kay DM, Browne ML, Liu A, Romitti PA, Kuehn D, Conley MR, Caggana M, Druschel CM, Brody LC, Mills JL. Anorectal atresia and Variants at Predicted Regulatory Sites in Candidate Genes. *Ann Hum Gene* 2012; Epub ahead of print.

   Variants predicted to affect transcription factor binding, splicing, and DNA methylation in WNT3A, PCSK5, TCF4, MKKS, GLI2, HOXD12, and BMP4 were associated with anorectal atresia based on a nominal P value < 0.05. Our results for MKKS support previously suggested associations with anorectal malformations. Our findings suggest that more research is needed to determine whether altered GLI2 and BMP4 expression is important in anorectal atresia in humans.

4. Carter TC, Kay DM, Browne ML, Liu A, Romitti PA, Kuehn D, Conley MR, Caggana M, Druschel CM, Brody LC, Mills JL. Hirschsprung's disease and variants in genes that regulate enteric neural crest cell proliferation, migration and differentiation. *J Hum Genet* 2012 Aug; 57(8):485-93.

Hirschsprung's disease (HSCR) results from failed colonization of the embryonic gut by enteric neural crest cells (ENCCs); colonization requires RET proto-oncogene (RET) signaling. Our population-based study identified novel RET variants in HSCR cases and showed that common RET variants may not contribute to HSCR in all race/ethnic groups. The findings for HOXB5 and PHOX2B provide supportive evidence that genes regulating ENCC proliferation, migration and differentiation could be risk factors for HSCR.

5. Caton AR. Exploring the seasonality of birth defects in the New York State Congenital Malformations Registry. *Birth Defects Res A Clin Mol Teratol* 2012;94:424-37.

   We aimed to determine whether the occurrence of birth defects varied by month of conception using the population-based New York State Congenital Malformations Registry (CMR). We merged live birth certificates (n = 2,044,091) with CMR records for mothers residing in New York State, excluding New York City, for the years 1992 through 2006. Of 42 groups examined in the 15-year period, 24 (57%) had at least one statistically significant test result, suggesting a trend or seasonal variation. Congenital cataract, pulmonary valve atresia/stenosis, coarctation of aorta, biliary atresia, and renal agenesis or hypoplasia had at least three significant tests. These results may help to generate hypotheses about environmental factors that vary by season for further studies.

6. Chen L, Bell EM, Browne ML, Druschel CM, Romitti PA, Schmidt RJ, Burns TL, Moslehi R, Olney RS; and the National Birth Defects Prevention Study. Maternal caffeine consumption and risk of congenital limb deficiencies. *Birth Defects Res A Clin Mol Teratol* 2012: Epub ahead of print.

   A weak increased risk of congenital limb deficiencies was associated with maternal dietary caffeine consumption in this study; however, risk did not vary by amount of caffeine consumed.

7. Ciafaloni E, Fox DJ, Pandya S, Westfield CP, Puzhankara S, Romitti PA, Mathews KD, Miller TM, Matthews DJ, Miller LA, Cunniff C, Druschel CD, Moxley RT. Delayed Diagnosis in Duchenne Muscular Dystrophy: Data from the Muscular Dystrophy Surveillance, Tracking, and Research Network (MD STAR*net*). *J Pediatr* 2009;155:380-5.

   Multistate, population-based data was used to identify key factors contributing to delay in diagnosis of males with Duchenne muscular dystrophy (DMD) without known family history. In this group of boys, first signs or symptoms was noted at a mean age of 2.5 years, and concerns resulted in primary care provider evaluation of the child at a mean age of 3.6 years. The mean age for the first creatine kinase blood tests was 4.7 years and mean age at definitive diagnosis was 4.9 years. There is a delay of about 2.5 years between onset of DMD symptoms and time of diagnosis, unchanged over the previous two decades. This delay results in lost opportunities for timely genetic counseling and initiation of corticosteroid treatment. Based on these findings, recommendations were made that primary care practitioners check creatine kinase early when evaluating boys with unexplained developmental delays.

8. Desrosiers TA, Herring AH, Shapira SK, Luben TJ, Hooiveld M, Herdt-Losavio ML, Lin S, Olshan AF and the National Birth Defects. Paternal occupation and birth defects in offspring: Findings from the National Birth Defects Prevention Study. Accepted by *Occup Environ Med* 2012; 69:534-542. DOI:10.1136/oemed-2011-100372.

   This large population-based study was conducted to explore the relation between multiple paternal occupations and over 60 types of birth defects using Bayesian analytic methods that specifically address the statistical challenges associated with analysis of sparsely distributed data across numerous exposures and outcomes. Results from this study indicate that paternal work in a number of occupations may be associated with an increased prevalence of various birth defects in offspring. Findings can be used to inform future investigation of specific paternal occupations found to be associated with birth defects or to generate hypotheses about chemical or physical exposures and exposure mixtures common to such occupations.

9. Lin S, Munsie JP, Herst-Losavio M, Druschel CM, Campbell KC, Browne ML. Maternal asthma medication use and the risk of selected birth defects. *Pediatrics* 2012; 129: e317-e324. DOI: 10.1542/peds.2010-2660.

   This study found no significant associations between maternal periconceptional use of asthma medications and risks of neural tube defects, some musculoskeletal defects (limb deficiency and diaphragmatic hernia), and small intestinal atresia. It suggests, however, that maternal asthma medication use may increase the risk of esophageal atresia, anorectal atresia, and omphalocele. Although the magnitudes of the associations were moderate (ORs ranged from 2.14 to 4.26), we cannot disentangle the effects of asthma severity and related hypoxia from those attributable to medication use, nor can we rule out the possibility that these findings are attributable to bias or chance. The current clinical guidelines and specific recommendations for aggressive asthma management during pregnancy should remain unchanged. Additional studies with detailed data on asthma severity and treatment and a large sample are recommended to clarify whether our findings are attributable to maternal use of asthma medication, asthma severity, or chance alone.

10. Mills JL, Carter TC, Kay DM, Browne ML, Brody LC, Liu A, Romitti PA, Caggana M, Druschel CM. Folate and vitamin B12-related genes and risk for omphalocele. *Hum Genet* 2012 May;131(5):739-46.

    Both taking folic acid-containing vitamins around conception and consuming food fortified with folic acid have been reported to reduce omphalocele rates. We examined single nucleotide polymorphisms (SNPs) known to be important in folate, vitamin B12, or choline. We observed associations between omphalocele and transcobalamin receptor (TCblR) and transporter (TCN2) SNPs and a betaine-homocysteine S-methyltransferase (BHMT) SNP. These results suggests that disruption of methylation reactions, in which folate, vitamin B12, and homocysteine play critical parts, may be a risk factor for omphalocele. Our data, if confirmed, suggest that supplements containing both folic acid and vitamin B12 may be beneficial in preventing omphaloceles.

11. Van Zutphen AR, Lin S, Fletcher BA, Hwang SA. A population-based case-control study of extreme summer temperature and birth defects. *Environ Health Perspect* 2012 Oct;120:1443-9.

    To determine whether pregnancies are potentially vulnerable to the weather extremes anticipated with climate change, we evaluated the relationship between extreme summer temperature and birth defects in a population-based case-control study by linking the New York State Congenital Malformations Registry to birth certificates for the years 1992-2006. Among 6,422 cases and 59,328 controls that shared at least 1 week of the critical period in summer, we found positive and consistent associations between multiple heat indicators during the relevant developmental window and congenital cataracts which should be confirmed with other data sources.

12. Wang Y, Kennedy J, Caggana M, Zimmerman R, Thomas S, Berninger J, Harris K, Green NS, Oyeku S, Hulihan M, Grant AM, Grosse SD. Sickle cell disease incidence among newborns in New York State by maternal race/ethnicity and nativity. *Genet Med* 2012; published online on September 27, 2012.

    This study provides the first US estimates of sickle cell disease incidence by maternal nativity. Newborns of non-Hispanic black mothers accounted for 86% of sickle cell disease cases whereas newborns of Hispanic mothers accounted for 12% of cases. The estimated incidence was 1:230 live births for non-Hispanic black mothers, 1:2,320 births for Hispanic mothers, and 1:41,647 births for non-Hispanic white mothers. Newborns of foreign-born non-Hispanic black mothers had a twofold higher incidence of sickle cell disease than those born to US-born non-Hispanic black mothers ($P < 0.001$). Such findings identify at-risk populations and inform outreach activities that promote ongoing, high-quality medical management to affected children.

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."[1] 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 [2, 3] 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 [4] 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.[5, 6] 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.[7]

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.[8] 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

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

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

3. 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.

4. 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.

5. 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*. 2007; 34(2):45-52.

6. 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.

7. 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.

8. 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