NEW YORK STATE DEPARTMENT OF HEALTH

Congenital Malformations Registry

Summary Report

Statistical Summary of Children
Born in 2006 and Diagnosed Through 2008

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 242,365 children who were born alive to New York residents in 2006. 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 (6.1 percent versus 4.0 percent), and black children had a higher major malformation rate than white children (6.4 percent versus 4.9 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 number of malformations are included in 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 eighteenth report from the CMR. Reports are also available by request for the 1983 to 2005 birth cohorts. This report and the reports for 1994-2005 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 Provider Network (HPN). The Department of Health developed the HPN 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 HPN using either online data entry forms or file upload facility are transferred to a DOH UNIX server for updating of the CMR database.

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All information reported to the registry is held in strict confidence. Records and computer files are maintained in accordance with DOH regulations concerning data containing individual identifiers. Access to the data by anyone other than registry personnel is restricted and carefully monitored to ensure that confidentiality is maintained. Families of children reported to the registry are never contacted without prior consent of the DOH's Institutional Review Board and notification of the child's physician.

2006 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 2006 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.[9] This report describes children born in 2006 and diagnosed before their second birthday. Reports are also available for the 1984 through 2005 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. 2000 census, the population of New York State was about 19.0 million; more than 42 percent of the population lived in New York City. An additional 23 percent of the population lived in the six counties closest to New York City. In 2006, there were 242,365 resident live births reported to the state's vital registration, 16.7 percent to black mothers, and 23.7 percent to Hispanic mothers. In accordance with the practices of other state birth defects registries, the race of the child is based on race of the mother only. Nearly 47.4 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 2006 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 (6.1% 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 (6.4 % versus 4.9%). 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 (5.0% versus 5.2%). The smaller number of births in the "other" racial category makes these rates difficult to interpret.

About 78% of children reported with major malformations have only one major malformation (Table 2). Since most children had one major malformation, the race-sex patterns seen for all major malformations are similar to the patterns seen in children with a single major malformation (Table 3). All race-sex groups for children with multiple major malformations showed little variation (Table 4).

Section 1 - Table 1
2006 Births - New York State Residents
Percent of Live Births with One or More Major Malformations
Sex by Race/Ethnicity and Residence

	Both Sexes			Males			Females		
Race and Residence	Infants	Total Births	%	Infants	Total Births	%	Infants	Total Births	%
New York State									
- All Races	12,277	242,365	5.1	7,516	124,224	6.1	4,761	118,141	4.0
- Non-Hispanic White	5,922	119,734	4.9	3,737	61,406	6.1	2,185	58,328	3.7
- Non-Hispanic Black	2,566	40,367	6.4	1,487	20,533	7.2	1,079	19,834	5.4
- Hispanic	2,734	57,555	4.8	1,644	29,420	5.6	1,090	28,135	3.9
- Others/Unknown	1,055	24,709	4.3	648	12,865	5.0	407	11,844	3.4
NYS Excluding NYC									
- All Races	6,343	127,461	5.0	3,942	65,384	6.0	2,401	62,077	3.9
- Non-Hispanic White	4,391	88,253	5.0	2,782	45,241	6.1	1,609	43,012	3.7
- Non-Hispanic Black	778	12,539	6.2	438	6,402	6.8	340	6,137	5.5
- Hispanic	836	19,211	4.4	502	9,826	5.1	334	9,385	3.6
- Others/Unknown	338	7,458	4.5	220	3,915	5.6	118	3,543	3.3
New York City									
- All Races	5,934	114,904	5.2	3,574	58,840	6.1	2,360	56,064	4.2
- Non-Hispanic White	1,531	31,481	4.9	955	16,165	5.9	576	15,316	3.8
- Non-Hispanic Black	1,788	27,828	6.4	1,049	14,131	7.4	739	13,697	5.4
- Hispanic	1,898	38,344	4.9	1,142	19,594	5.8	756	18,750	4.0
- Others/Unknown	717	17,251	4.2	428	8,950	4.8	289	8,301	3.5

Section 1 - Table 2
2006 Births - New York State Residents
Number of Major Malformations Per Child

Number of Malformations	Number of Children	Percent
1	9,626	78.4
2	1,644	13.4
3	554	4.5
4	250	2.0
5	95	0.8
6	51	0.4
7	27	0.2
8	18	0.1
9	8	0.1
10	4	*
All Children	12,277	100.0

* - Less than 0.05%

Note: Total percent may not add to 100% due to rounding

Section 1 - Table 3
2006 Births - New York State Residents
Percent of Live Births with One Major Malformation
Sex by Race/Ethnicity and Residence

	Both Sexes			Males			Females		
Race and Residence	Infants	Total Births	%	Infants	Total Births	%	Infants	Total Births	%
New York State									
- All Races	9,626	242,365	4.0	6,025	124,224	4.9	3,601	118,141	3.0
- Non-Hispanic White	4,632	119,734	3.9	2,993	61,406	4.9	1,639	58,328	2.8
- Non-Hispanic Black	2,036	40,367	5.0	1,183	20,533	5.8	853	19,834	4.3
- Hispanic	2,157	57,555	3.7	1,346	29,420	4.6	811	28,135	2.9
- Others/Unknown	801	24,709	3.2	503	12,865	3.9	298	11,844	2.5
NYS Excluding NYC									
- All Races	4,943	127,461	3.9	3,164	65,384	4.8	1,779	62,077	2.9
- Non-Hispanic White	3,406	88,253	3.9	2,217	45,241	4.9	1,189	43,012	2.8
- Non-Hispanic Black	619	12,539	4.9	353	6,402	5.5	266	6,137	4.3
- Hispanic	658	19,211	3.4	417	9,826	4.2	241	9,385	2.6
- Others/Unknown	260	7,458	3.5	177	3,915	4.5	83	3,543	2.3
New York City									
- All Races	4,683	114,904	4.1	2,861	58,840	4.9	1,822	56,064	3.2
- Non-Hispanic White	1,226	31,481	3.9	776	16,165	4.8	450	15,316	2.9
- Non-Hispanic Black	1,417	27,828	5.1	830	14,131	5.9	587	13,697	4.3
- Hispanic	1,499	38,344	3.9	929	19,594	4.7	570	18,750	3.0
- Others/Unknown	541	17,251	3.1	326	8,950	3.6	215	8,301	2.6

Section 1 - Table 4
2006 Births - New York State Residents
Percent of Live Births with Two or More Major Malformations
Sex by Race/Ethnicity and Residence

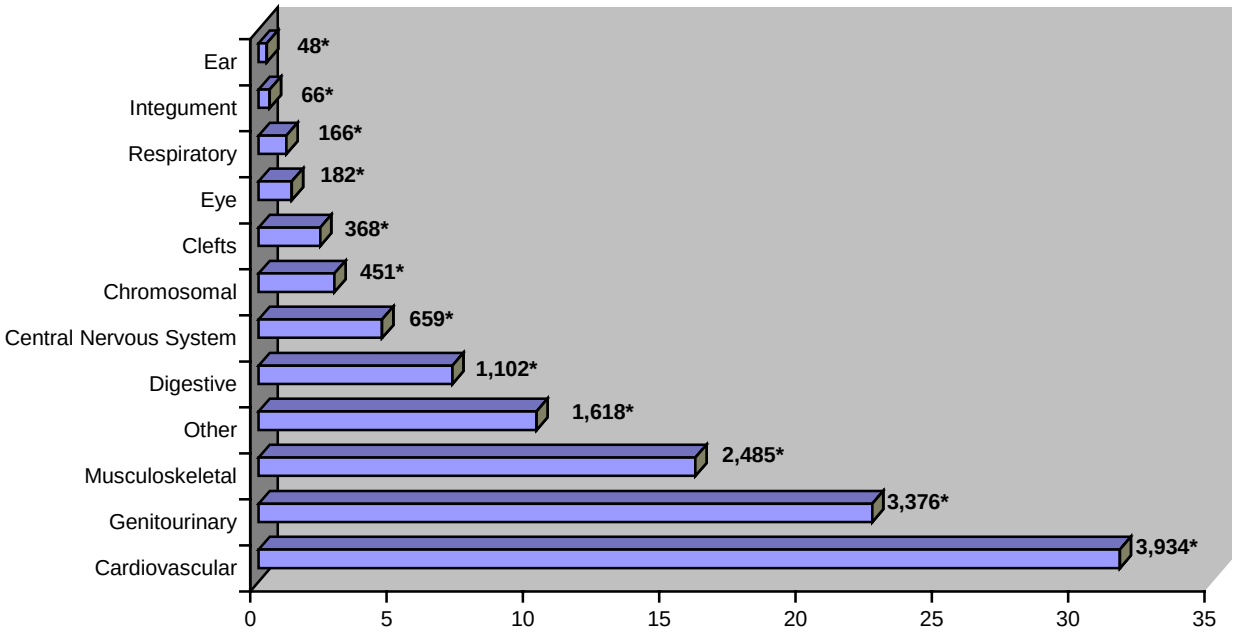
	Both Sexes			Males			Females		
Race and Residence	Infants	Total Births	%	Infants	Total Births	%	Infants	Total Births	%
New York State									
- All Races	2,651	242,365	1.1	1,491	124,224	1.2	1,160	118,141	1.0
- Non-Hispanic White	1,290	119,734	1.1	744	61,406	1.2	546	58,328	0.9
- Non-Hispanic Black	530	40,367	1.3	304	20,533	1.5	226	19,834	1.1
- Hispanic	577	57,555	1.0	298	29,420	1.0	279	28,135	1.0
- Others/Unknown	254	24,709	1.0	145	12,865	1.1	109	11,844	0.9
NYS Excluding NYC									
- All Races	1,400	127,461	1.1	778	65,384	1.2	622	62,077	1.0
- Non-Hispanic White	985	88,253	1.1	565	45,241	1.2	420	43,012	1.0
- Non-Hispanic Black	159	12,539	1.3	85	6,402	1.3	74	6,137	1.2
- Hispanic	178	19,211	0.9	85	9,826	0.9	93	9,385	1.0
- Others/Unknown	78	7,458	1.0	43	3,915	1.1	35	3,543	1.0
New York City									
- All Races	1,251	114,904	1.1	713	58,840	1.2	538	56,064	1.0
- Non-Hispanic White	305	31,481	1.0	179	16,165	1.1	126	15,316	0.8
- Non-Hispanic Black	371	27,828	1.3	219	14,131	1.5	152	13,697	1.1
- Hispanic	399	38,344	1.0	213	19,594	1.1	186	18,750	1.0
- Others/Unknown	176	17,251	1.0	102	8,950	1.1	74	8,301	0.9

Section II
Major Congenital Malformations by Organ System, 2006

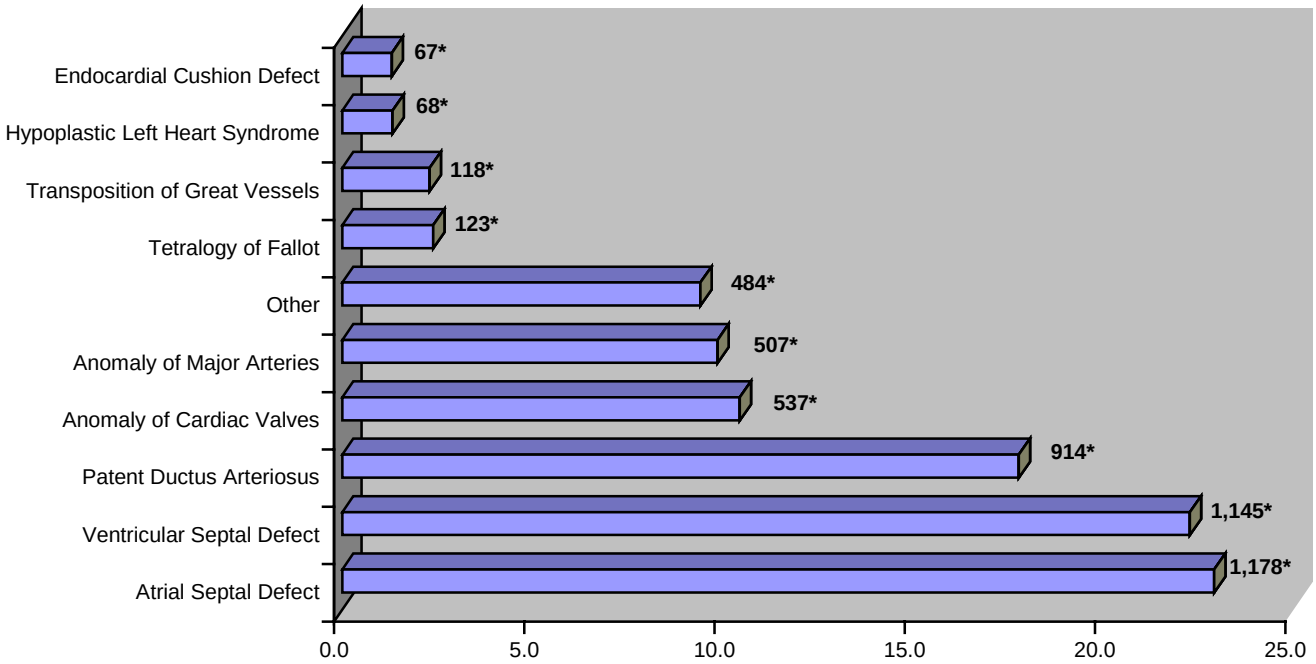
Introduction to Figures

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.

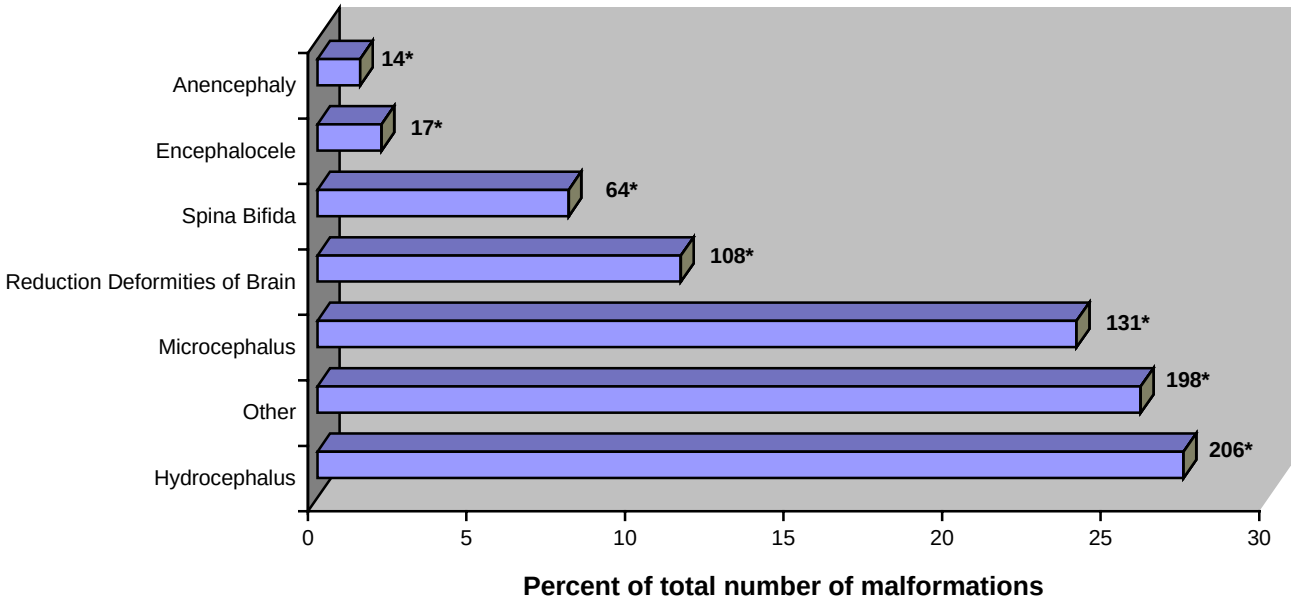
Major Malformations by Organ System
2006 Births - New York State Residents
(Number of Children = 12,277)
(* - Number of malformations in each organ system)



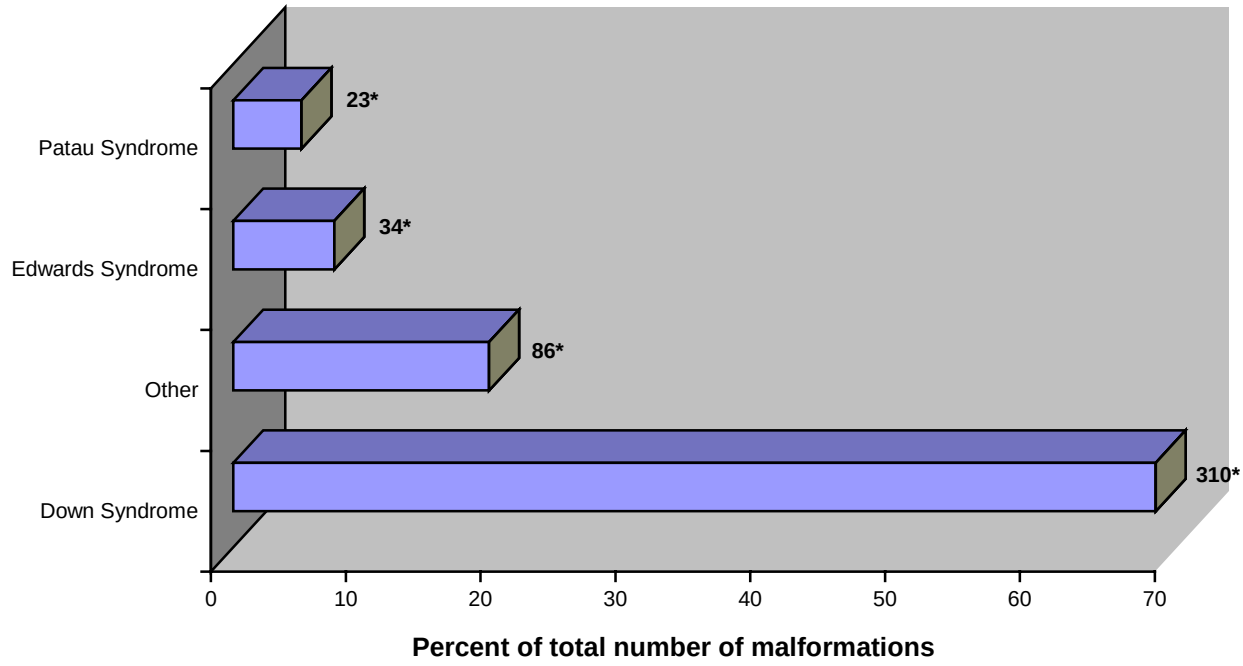
Major Malformations by Organ System
2006 Births - New York State Residents
Cardiovascular System Subset Category
(Number of Children = 3,934)
(* - Number of malformations in each category)



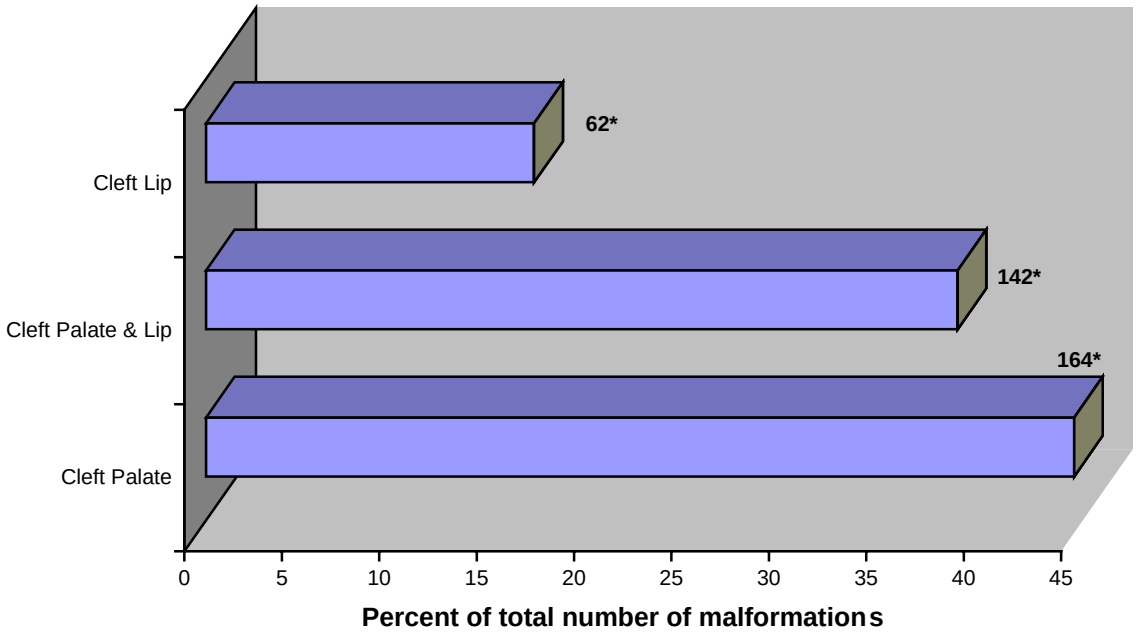
Major Malformations by Organ System
2006 Births - New York State Residents
Central Nervous System Subset Category
(Number of Children = 659)
(* - Number of malformations in each organ system)



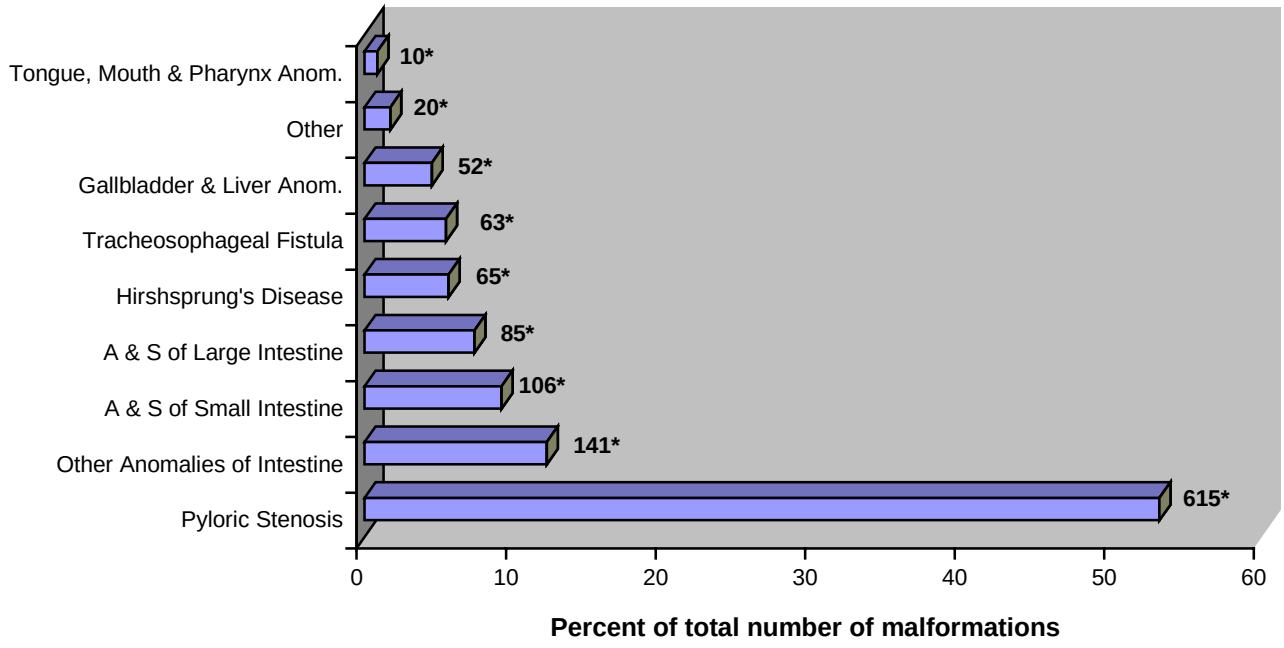
Major Malformations by Organ System
2006 Births - New York State Residents
Chromosomal Subset Category
(Number of Children = 451)
(* - Number of malformations in each organ system)



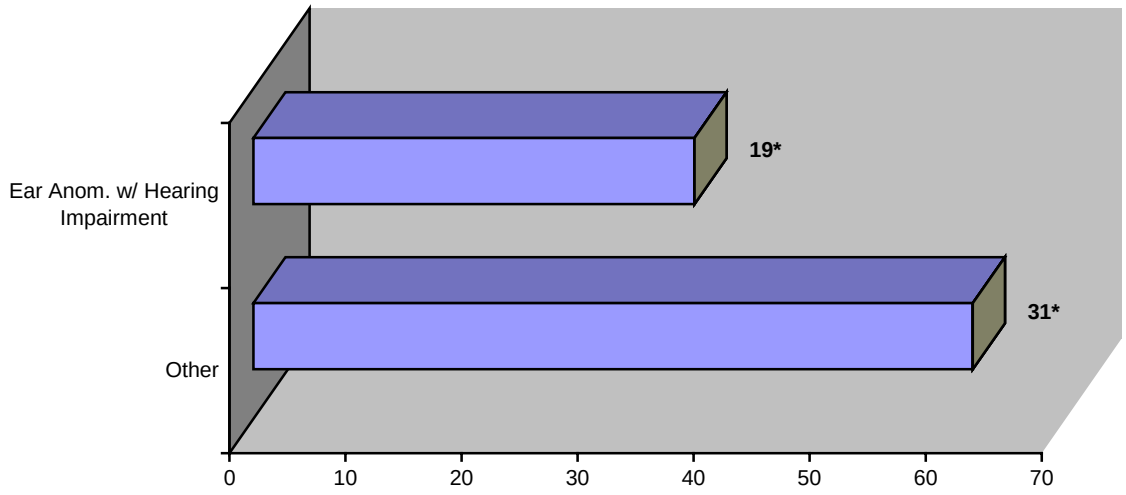
Major Malformations by Organ System
2006 Births - New York State Residents
Oral Clefts Subset Category
(Number of Children = 368)
(* - Number of malformations in each organ system)



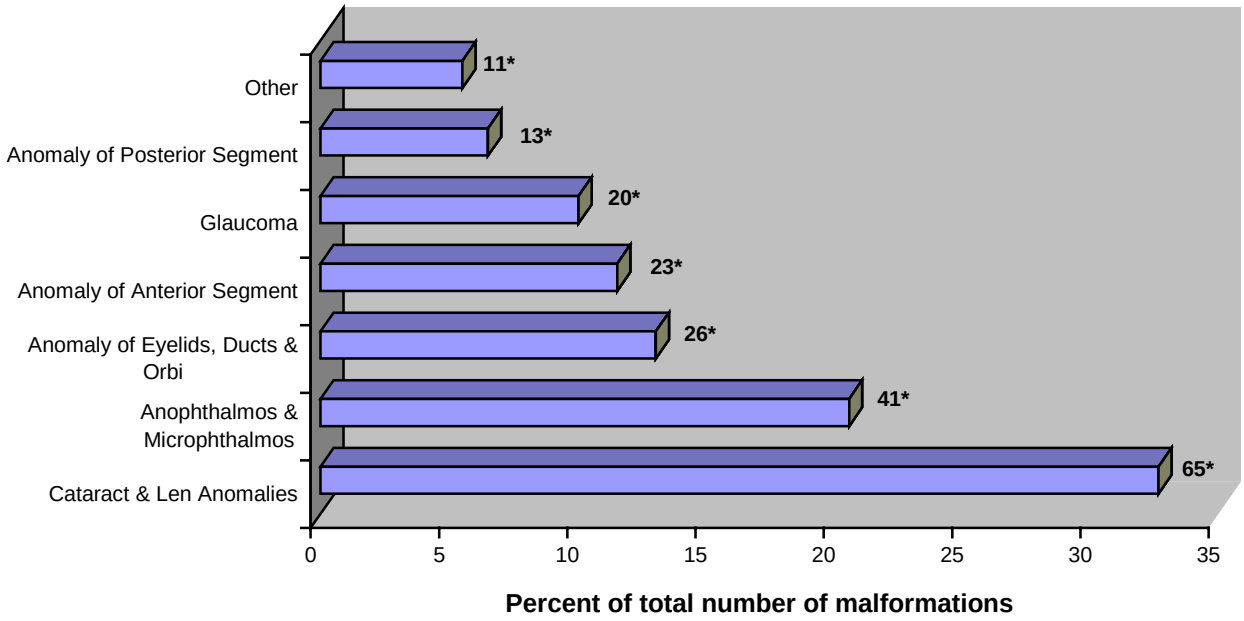
Major Malformations by Organ System
2006 Births - New York State Residents
Digestive System Subset Category
(Number of Children = 1102)
(* - Number of malformations in each organ system)



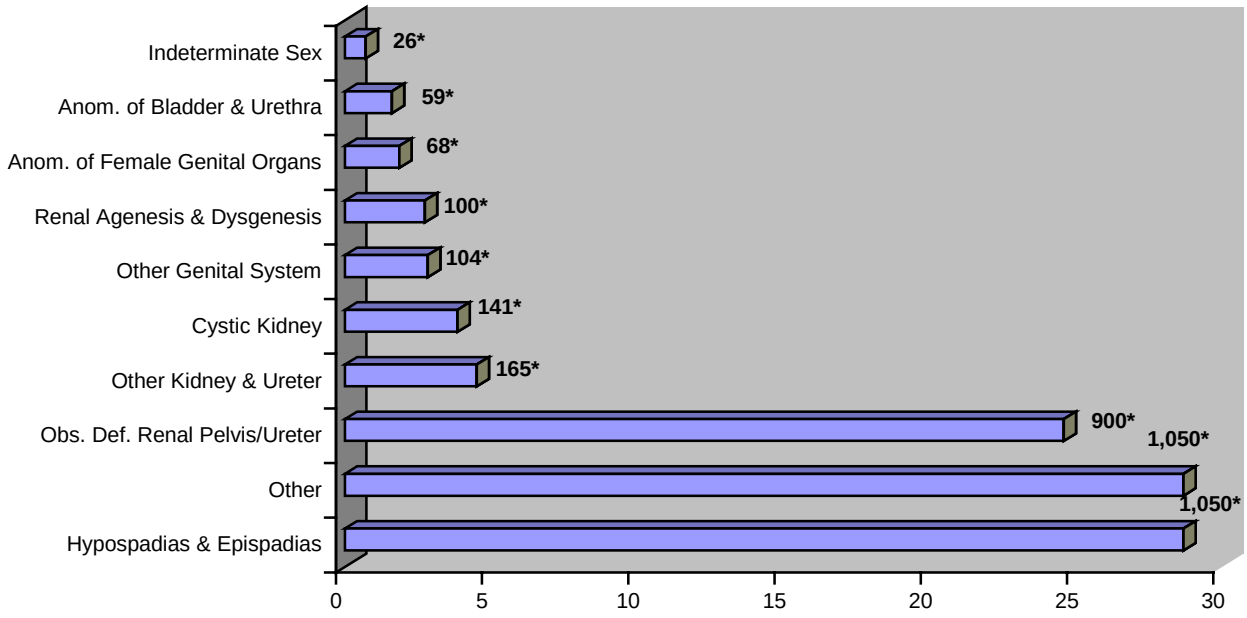
Major Malformations by Organ System
2006 Births - New York State Residents
Ear Subset Category
(Number of Children =48)
(* - Number of malformations in each organ system)



Major Malformations by Organ System
2006 Births - New York State Residents
Eye Subset Category
(Number of Children = 182)
(* - Number of malformations in each organ system)

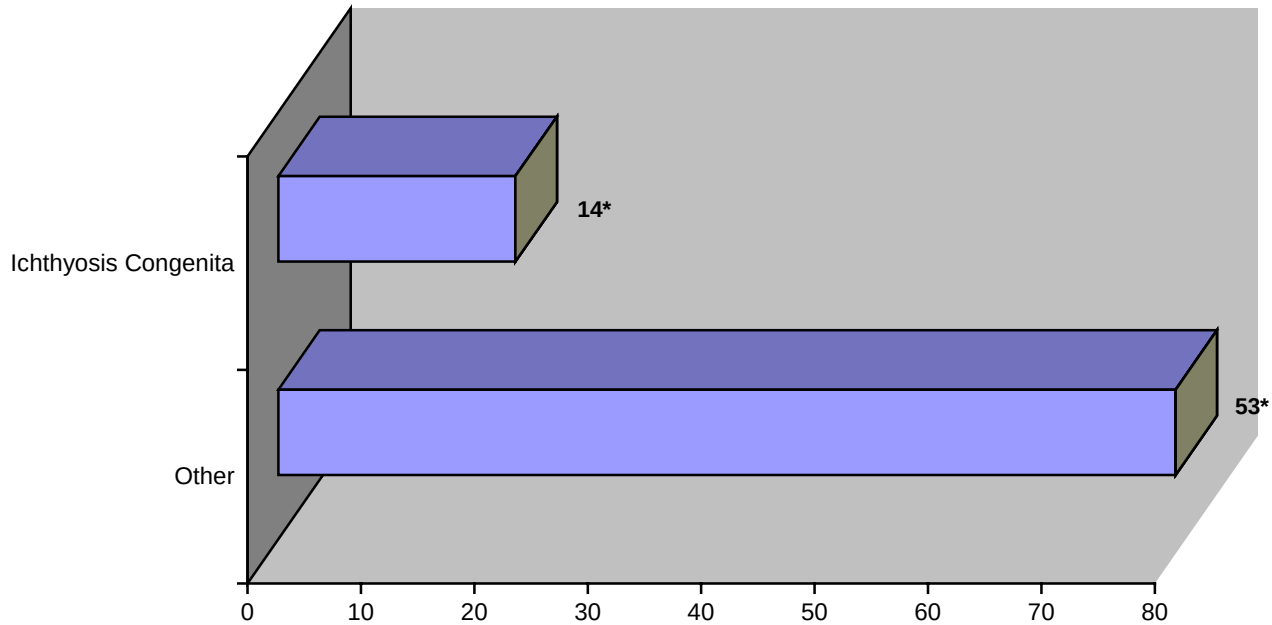


Major Malformations by Organ System
2006 Births - New York State Residents
Genitourinary System Subset Category
(Number of Children = 3,376)
(* - Number of malformations in each organ system)



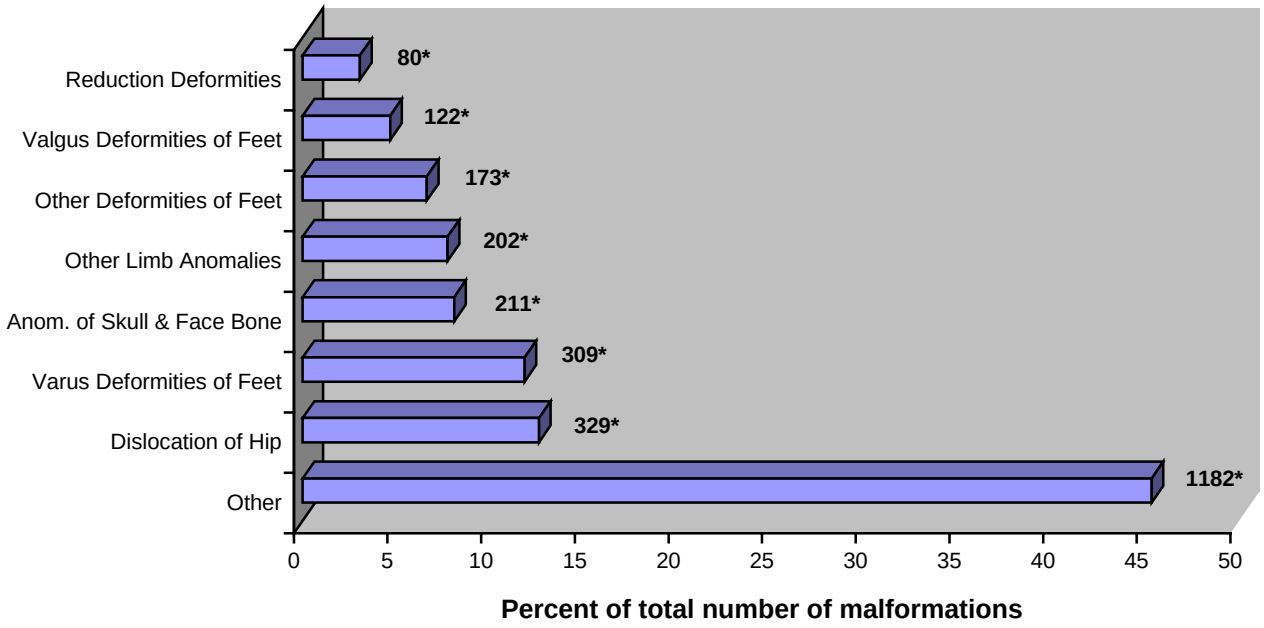
Percent of total number of malformations

Major Malformations by Organ System
2006 Births - New York State Residents
Integument System Subset Category
(Number of Children = 66)
(* - Number of malformations in each organ system)

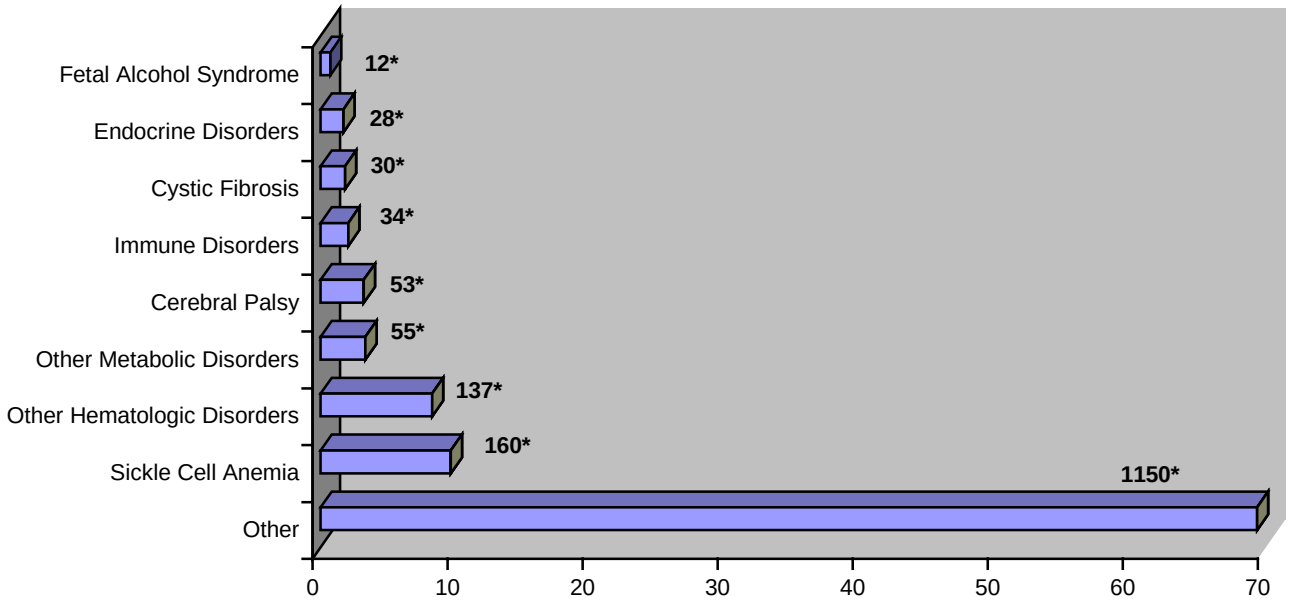


Percent of total number of malformations

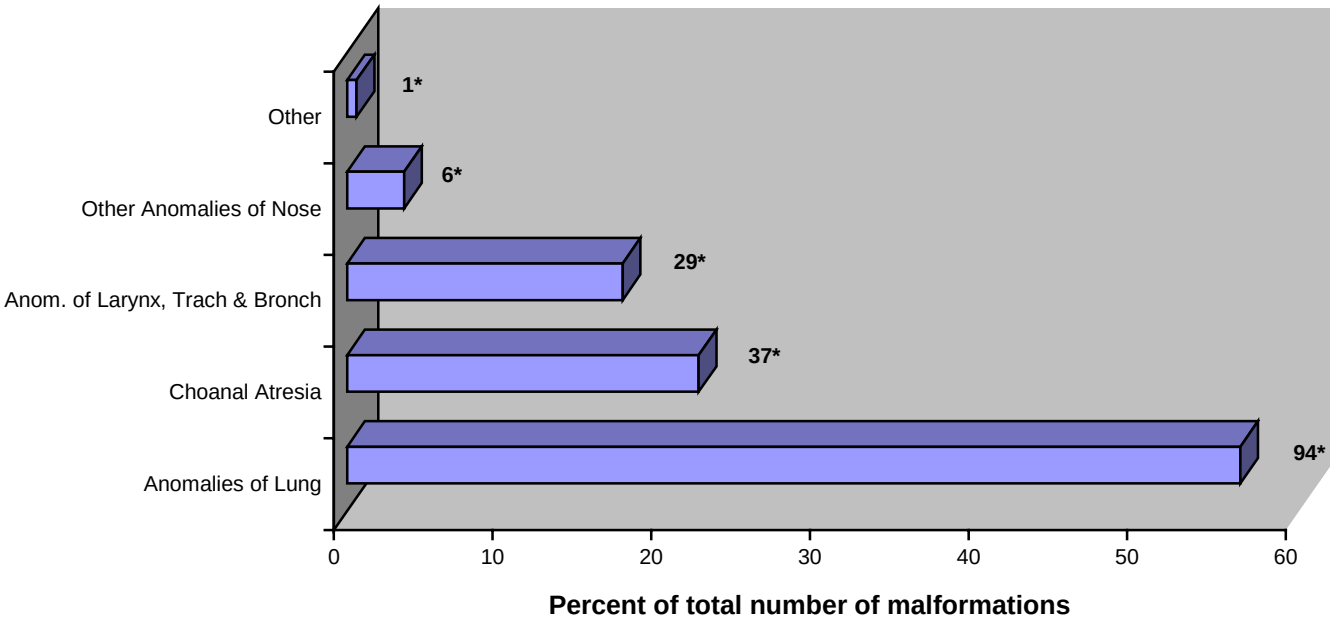
Major Malformations by Organ System
2006 Births - New York State Residents
Musculoskeletal System Subset Category
(Number of Children =2,485)
(* - Number of malformations in each organ system)



Major Malformations by Organ System
2006 Births - New York State Residents
All Others Subset Category
(Number of Children = 1,618)
(* - Number of malformations in each organ system)



Major Malformations by Organ System
2006 Births - New York State Residents
Respiratory System Subset Category
(Number of Children = 166)
(* - Number of malformations in each organ system)



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

Introduction

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

Fluctuations in specific malformation prevalence should be interpreted with caution, especially differences in the "other" race category since the numbers in this group are small. In addition, several malformations were added in 1992 as a result of the change to the BPA 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– Table 1
Children With Selected Malformations
Prevalence per 10,000 Live Births by Sex and Race/Ethnicity

2006 Births– New York State Residents

ICD-9 Code	Malformation	Total Number	Total Prevalence	Male	Female	Ratio (M/F)	Non-Hispanic White	Non-Hispanic Black	Hispanic	Other/ Unknown Race
243	Congenital hypothyroidism	123	5.1	5.9	4.2	1.4	3.7	6.4	6.9	5.3
270.1	Phenylketonuria	4	0.2	0.2	0.1	2.9	0.3	0.0	0.2	0.0
277.0	Cystic fibrosis	30	1.2	1.4	1.1	1.2	1.5	1.0	1.2	0.4
282.6	Sickle-cell anemia	160	6.6	6.7	6.5	1.0	0.3	33.2	3.8	0.0
740.0	Anencephalus	14	0.6	0.6	0.6	1.0	0.4	1.0	0.9	0.0
741.0	Spina bifida with hydrocephalus	31	1.3	1.3	1.3	1.0	1.6	0.5	1.7	0.0
741.9	Spina bifida without hydrocephalus	33	1.4	1.5	1.2	1.3	1.1	1.2	1.0	3.6
742.0	Encephalocele	17	0.7	0.8	0.6	1.4	0.5	1.2	0.9	0.4
742.1	Microcephalus	131	5.4	4.7	6.1	0.8	4.7	9.4	4.7	4.0
742.2	Agyria & lissencephaly	4	0.2	0.2	0.1	2.9	0.2	0.0	0.0	0.8
742.2	Anomalies of corpus callosum	72	3.0	2.9	3.0	1.0	3.3	3.0	2.6	2.4
742.2	Holoprosencephaly	13	0.5	0.5	0.6	0.8	0.4	0.7	0.9	0.0
742.3	Congenital hydrocephalus	206	8.5	9.7	7.3	1.3	7.4	11.9	9.0	6.9
742.4	Porencephaly	9	0.4	0.4	0.3	1.2	0.5	0.2	0.3	0.0
742.5	Congenital tethered cord	37	1.5	1.0	2.0	0.5	2.0	0.2	1.0	2.4
743.0	Anophthalmos	3	0.1	0.1	0.2	0.5	0.2	0.2	0.0	0.0
743.1	Microphthalmos	39	1.6	1.6	1.6	1.0	1.7	1.2	1.6	2.0
743.2	Glaucoma	20	0.8	1.0	0.6	1.8	0.7	2.0	0.5	0.4
743.3	Absence of lens	20	0.8	0.6	1.1	0.5	1.0	1.7	0.2	0.0
743.3	Congenital cataract	23	0.9	1.0	0.8	1.2	0.9	1.2	0.7	1.2
743.45	Aniridia	4	0.2	0.1	0.3	0.3	0.2	0.0	0.2	0.4
743.46	Coloboma of iris	5	0.2	0.1	0.3	0.2	0.3	0.2	0.0	0.4
744.0	Anotia/microtia	22	0.9	0.6	1.3	0.4	1.3	0.0	1.0	0.4
745.0	Common truncus	20	0.8	0.6	1.0	0.6	0.8	2.0	0.3	0.4
745.1	Transposition of great vessels	118	4.9	5.6	4.1	1.3	4.8	7.2	4.0	3.6
745.2	Tetralogy of Fallot	123	5.1	5.6	4.6	1.2	5.0	6.2	3.8	6.5
745.3	Common ventricle	10	0.4	0.4	0.4	1.0	0.3	0.7	0.7	0.0
745.4	Ventricular septal defect	1,145	47.2	42.4	52.3	0.8	47.7	44.8	49.3	44.1
745.5	Ostium secundum type atrial septal def.	1,178	48.6	48.3	48.9	1.0	41.6	70.1	47.4	50.2
745.6	Endocardial cushion defects	67	2.8	2.8	2.7	1.0	3.1	3.5	2.1	1.6
746.0	Atresia/stenosis of pulmonary valve	204	8.4	8.5	8.4	1.0	8.2	11.1	6.6	9.3
746.1	Tricuspid atresia/stenosis/hypoplasia	21	0.9	0.8	0.9	0.9	0.8	0.7	1.2	0.8
746.2	Ebstein's anomaly	13	0.5	0.6	0.4	1.5	0.8	0.5	0.3	0.0
746.3	Congenital stenosis of aortic valve	55	2.3	2.9	1.6	1.8	3.2	2.0	1.0	1.2
746.7	Hypoplastic left heart syndrome	68	2.8	3.0	2.6	1.1	3.3	2.7	2.1	2.4
746.85	Anomalies of coronary artery	22	0.9	0.9	0.9	1.0	0.7	1.2	1.2	0.8
747.0	Patent ductus arteriosis	914	37.7	39.8	35.5	1.1	34.4	59.7	29.4	37.2
747.10	Coartation of aorta	156	6.4	6.1	6.8	0.9	6.7	7.2	5.7	5.7
747.41	Total anomalous pulmonary venus conne	23	0.9	1.1	0.8	1.5	0.9	0.5	1.4	0.8

2006 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	37	1.5	2.0	1.0	2.0	1.6	1.7	1.6	0.8
748.5	Agenesis/hypoplasia of lung	54	2.2	2.7	1.8	1.5	2.3	2.0	2.4	1.6
749.0	Cleft palate	164	6.8	5.2	8.5	0.6	7.4	5.9	5.6	7.7
749.1	Cleft lip	62	2.6	2.7	2.5	1.1	3.3	2.0	1.7	1.6
749.2	Cleft palate & lip	142	5.9	6.4	5.3	1.2	5.8	4.0	6.8	7.3
750.3	Tracheoesophageal fistula etc.	63	2.6	2.3	2.9	0.8	3.0	2.0	2.3	2.4
750.5	Congenital hypertrophic pyloric stenosis	615	25.4	41.5	8.4	5.0	27.6	17.1	31.6	13.4
751.1	Atresia and stenosis of small intestine	106	4.4	4.7	4.1	1.1	4.2	4.2	5.0	4.0
751.2	Atresia and stenosis of rectum or anus	85	3.5	3.5	3.5	1.0	3.1	3.7	4.7	2.4
751.3	Hirschsprungs disease	65	2.7	3.4	1.9	1.7	2.8	4.7	1.7	1.2
751.4	Anomalies of intestinal fixation	67	2.8	2.6	3.0	0.9	2.7	4.7	1.7	2.4
751.61	Biliary atresia	24	1.0	1.0	0.9	1.1	0.7	2.0	0.9	1.2
752.6	Epispadias	36	1.5	2.8	0.1	33.3	1.5	2.5	1.2	0.4
752.6	Hypospadias	975	40.2	77.5	1.0	76.3	49.8	37.2	23.8	37.2
753.0	Renal agenesis and dysgenesis	100	4.1	4.8	3.4	1.4	4.4	5.7	2.8	3.2
753.1	Cystic kidney disease	141	5.8	5.8	5.8	1.0	4.5	9.4	6.3	5.3
753.2	Obstructive defect renal pelvis & ureter	900	37.1	50.8	22.8	2.2	39.4	30.0	36.3	39.7
753.5	Extrophy of urinary bladder	3	0.1	0.1	0.2	0.5	0.2	0.2	0.0	0.0
753.6	Atresia & stenosis of urethra & bladder	30	1.2	2.3	0.2	13.3	1.4	1.5	1.0	0.4
754.3	Congenital dislocation of hip	235	9.7	4.0	15.7	0.3	10.0	4.0	13.9	7.7
754.51	Talipes equinovarus	189	7.8	9.2	6.3	1.4	8.4	5.7	8.9	5.7
755.2	Reduction deformities of upper limb	56	2.3	2.3	2.4	1.0	2.8	1.5	1.9	2.0
755.3	Reduction deformities of lower limb	26	1.1	1.0	1.1	1.0	1.3	1.2	0.9	0.4
755.8	Arthrogryposis multiplex congenita	20	0.8	0.8	0.8	1.0	0.9	1.0	0.7	0.4
756.0	Craniosynostosis	116	4.8	5.4	4.1	1.3	6.0	1.5	5.2	3.2
756.0	Goldenhar syndrome	6	0.2	0.2	0.3	1.0	0.5	0.0	0.0	0.0
756.4	Chonodrodystrophy	36	1.5	1.0	2.0	0.5	1.2	2.7	1.4	1.2
756.51	Osteogenesis imperfecta	10	0.4	0.6	0.3	2.2	0.6	0.0	0.3	0.4
756.6	Diaphragmatic hernia	81	3.3	3.5	3.2	1.1	3.3	3.0	3.3	4.0
756.7	Gastroschisis	63	2.6	2.9	2.3	1.3	2.8	3.0	2.6	0.8
756.7	Omphalocele	39	1.6	1.5	1.7	0.9	1.4	2.7	0.9	2.4
756.7	Prune belly	10	0.4	0.7	0.1	8.6	0.3	1.2	0.3	0.0
758.0	Down syndrome	310	12.8	13.5	12.0	1.1	12.9	14.6	13.2	8.1
758.1	Patau syndrome	23	0.9	0.8	1.1	0.7	1.0	1.0	1.0	0.4
758.2	Edwards syndrome	34	1.4	1.0	1.8	0.6	1.2	1.5	1.7	1.6
758.6	Gonadal dysgenesis	13	0.5	0.0	1.1	0.0	0.8	0.0	0.5	0.0
758.7	Klinefelter syndrome	15	0.6	1.2	0.0	0.0	0.5	1.0	0.3	1.2
759.3	Situs inversus	19	0.8	0.8	0.8	1.1	0.6	0.7	1.0	1.2
760.71	Fetal alcohol syndrome	12	0.5	0.8	0.2	4.8	0.3	1.2	0.5	0.0
762.8	Amniotic bands	7	0.3	0.3	0.3	1.3	0.3	0.5	0.3	0.0
771.0	Congenital rubella	2	0.1	0.1	0.1	1.0	0.0	0.2	0.2	0.0
771.1	Congenital cytomegalovirus infection	17	0.7	0.8	0.6	1.4	0.3	1.0	1.0	1.2
771.2	Other congenital infections	42	1.7	1.8	1.7	1.0	1.6	2.2	1.7	1.6

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. The table on the next page presents the number of children with major malformations by county, and the percent of live births for comparison.

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

These county listings are not designed to be used for 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
Children with Major Congenital Malformations & Percent of Live Births
by County and Birth Year, 2006

County	Number of Children	Number of Live Births	Percent of Live Births
Albany	145	3,064	4.7
Allegany	20	492	4.1
Bronx	1,250	21,276	5.9
Broome	97	2,115	4.6
Cattaraugus	57	991	5.8
Cayuga	47	859	5.5
Chautauqua	86	1,377	6.2
Chemung	58	968	6.0
Chenango	24	538	4.5
Clinton	20	731	2.7
Columbia	42	569	7.4
Cortland	19	511	3.7
Delaware	23	468	4.9
Dutchess	116	2,877	4.0
Erie	645	9,957	6.5
Essex	5	284	1.8
Franklin	10	509	2.0
Fulton	33	569	5.8
Genesee	38	636	6.0
Greene	17	448	3.8
Hamilton	1	31	3.2
Herkimer	38	644	5.9
Jefferson	85	1,670	5.1
Kings	2,175	40,096	5.4
Lewis	16	327	4.9
Livingston	26	651	4.0
Madison	33	750	4.4
Monroe	364	8,887	4.1
Montgomery	30	623	4.8
Nassau	770	15,340	5.0
New York	930	20,239	4.6
Niagara	119	2,205	5.4
Oneida	152	2,592	5.9
Onondaga	327	5,471	6.0
Ontario	45	1,135	4.0
Orange	225	5,294	4.3
Orleans	23	462	5.0
Oswego	92	1,376	6.7
Otsego	29	549	5.3
Putnam	46	991	4.6
Queens	1,338	27,487	4.9
Rensselear	86	1,642	5.2
Richmond	241	5,806	4.2

Section IV – Table 1 (continued)
Children with Major Congenital Malformations & Percent of Live Births
by County and Birth Year, 2006

County	Number of Children	Number of Live Births	Percent of Live Births
Rockland	215	4,310	5.0
Saratoga	118	2,386	4.9
Schenectady	80	1,847	4.3
Schoharie	17	293	5.8
Schuyler	12	163	7.4
Seneca	15	353	4.2
St Lawrence	60	1,262	4.8
Steuben	38	1,087	3.5
Suffolk	968	19,097	5.1
Sullivan	37	943	3.9
Tioga	22	397	5.5
Tompkins	33	842	3.9
Ulster	75	1,869	4.0
Warren	28	585	4.8
Washington	42	551	7.6
Wayne	58	1,139	5.1
Westchester	489	11,038	4.4
Wyoming	19	382	5.0
Yates	8	314	2.5

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

Birth Year: 2006

County	ICD-9 Code	Description	Number
ALBANY	752.6	Hypospadias & epispadias	16
	747.0	Patent ductus arteriosus	14
	745.5	Ostium secundum atrial septal defect	13
	753.2	Obstructive defects of renal pelvis & ureter	13
	745.4	Ventricular septal defect	12
	752.5	Undescended testicle	11
	755.0	Polydactyly	9
	750.5	Congenital hypertrophic pyloric stenosis	7
	754.3	Congenital dislocation of hip	7
	756.7	Anomalies of abdominal wall	7
ALLEGANY	752.6	Hypospadias & epispadias	5
	746.8	Other specified anomalies of heart	2
	749.2	Cleft palate with cleft lip	2
	270.7	Disturbances of amino-acid metabolims	1
	333.2	Myoclonus	1
	745.1	Transposition of great vessels	1
	745.5	Ostium secundum atrial septal defect	1
	747.0	Patent ductus arteriosus	1
	747.3	Anomalies of pulmonary artery	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	754.1	Deformities of sternocleidomastoid muscle	1
	754.3	Congenital dislocation of hip	1
	754.5	Varus deformities of feet	1
	756.0	Anomalies of skull and face bones	1
	756.3	Other anomalies of ribs and sternum	1
	757.3	Other specified anomalies of skin	1
	758.0	Down syndrome	1
BRONX	755.0	Polydactyly	114
	745.5	Ostium secundum atrial septal defect	113
	752.5	Undescended testicle	111
	745.4	Ventricular septal defect	105
	752.6	Hypospadias & epispadias	91
	753.2	Obstructive defects of renal pelvis & ureter	73
	754.5	Varus deformities of feet	61
	750.5	Congenital hypertrophic pyloric stenosis	60
	747.0	Patent ductus arteriosus	54
	282.6	Sickle-cell anemia	46
BROOME	750.5	Congenital hypertrophic pyloric stenosis	12
	752.5	Undescended testicle	9
	745.5	Ostium secundum atrial septal defect	6
	747.0	Patent ductus arteriosus	5
	752.6	Hypospadias & epispadias	5
	745.2	Tetralogy of Fallot	4

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

Birth Year: 2006

County	ICD-9 Code	Description	Number
BROOME	745.4	Ventricular septal defect	4
	747.3	Anomalies of pulmonary artery	4
	748.5	Agenesis, hypoplasia & dysplasia, lung	4
	755.0	Polydactyly	4
CATTARAUGUS	745.4	Ventricular septal defect	8
	753.2	Obstructive defects of renal pelvis & ureter	8
	746.8	Other specified anomalies of heart	6
	747.0	Patent ductus arteriosus	6
	752.6	Hypospadias & epispadias	6
	745.5	Ostium secundum atrial septal defect	5
	747.3	Anomalies of pulmonary artery	5
	746.6	Congenital mitral insufficiency	3
	747.1	Coarctation of aorta	3
	750.5	Congenital hypertrophic pyloric stenosis	3
	754.5	Varus deformities of feet	3
CAYUGA	752.5	Undescended testicle	7
	752.6	Hypospadias & epispadias	6
	750.5	Congenital hypertrophic pyloric stenosis	3
	742.3	Congenital hydrocephalus	2
	743.3	Congenital cataract & lens anomalies	2
	745.4	Ventricular septal defect	2
	746.3	Congenital stenosis of aortic valve	2
	747.0	Patent ductus arteriosus	2
	754.5	Varus deformities of feet	2
	755.0	Polydactyly	2
	755.6	Other anomalies of lower limb including pelvic girdle	2
CHAUTAUQUA	745.5	Ostium secundum atrial septal defect	23
	745.4	Ventricular septal defect	9
	752.5	Undescended testicle	9
	747.0	Patent ductus arteriosus	8
	747.3	Anomalies of pulmonary artery	8
	750.5	Congenital hypertrophic pyloric stenosis	6
	746.8	Other specified anomalies of heart	5
	742.4	Other specified anomalies of brain	3
	742.2	Reduction deformities of brain	2
	746.4	Congenital insufficiency of aortic valve	2
	746.7	Hypoplastic left heart syndrome	2
	747.1	Coarctation of aorta	2
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	2
	752.6	Hypospadias & epispadias	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	754.5	Varus deformities of feet	2
	756.0	Anomalies of skull and face bones	2

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

Birth Year: 2006

County	ICD-9 Code	Description	Number
CHEMUNG	752.6	Hypospadias & epispadias	9
	753.2	Obstructive defects of renal pelvis & ureter	7
	752.5	Undescended testicle	6
	745.1	Transposition of great vessels	3
	745.5	Ostium secundum atrial septal defect	3
	747.0	Patent ductus arteriosus	3
	747.3	Anomalies of pulmonary artery	3
	748.3	Other anomalies of larynx, trachea, & bronchus	3
	750.5	Congenital hypertrophic pyloric stenosis	3
	754.5	Varus deformities of feet	3
CHENANGO	753.2	Obstructive defects of renal pelvis & ureter	3
	243.	Congenital hypothyroidism	2
	745.4	Ventricular septal defect	2
	745.5	Ostium secundum atrial septal defect	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	752.5	Undescended testicle	2
	753.1	Cystic kidney disease	2
	756.0	Anomalies of skull and face bones	2
	756.7	Anomalies of abdominal wall	2
	228.1	Lymphangioma, any site	1
	524.0	Major anomalies of jaw size	1
	742.2	Reduction deformities of brain	1
	746.0	Anomalies of pulmonary valve	1
	746.8	Other specified anomalies of heart	1
	747.0	Patent ductus arteriosus	1
	747.3	Anomalies of pulmonary artery	1
	749.0	Cleft palate	1
	749.2	Cleft palate with cleft lip	1
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	1
	752.6	Hypospadias & epispadias	1
	754.5	Varus deformities of feet	1
	756.6	Anomalies of diaphragm	1
	758.1	Patau syndrome	1
	759.5	Tuberous sclerosis	1
	759.8	Other specified anomalies	1
CLINTON	752.6	Hypospadias & epispadias	6
	745.4	Ventricular septal defect	3
	745.5	Ostium secundum atrial septal defect	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	333.2	Myoclonus	1
	747.0	Patent ductus arteriosus	1
	749.2	Cleft palate with cleft lip	1
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	1
	750.5	Congenital hypertrophic pyloric stenosis	1

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

Birth Year: 2006

County	ICD-9 Code	Description	Number
CLINTON	751.1	Atresia & stenosis of small intestine	1
	751.5	Other anomalies of intestine	1
	753.1	Cystic kidney disease	1
COLUMBIA	752.6	Hypospadias & epispadias	5
	753.2	Obstructive defects of renal pelvis & ureter	5
	747.0	Patent ductus arteriosus	4
	745.5	Ostium secundum atrial septal defect	3
	742.2	Reduction deformities of brain	2
	742.4	Other specified anomalies of brain	2
	745.4	Ventricular septal defect	2
	746.7	Hypoplastic left heart syndrome	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	752.5	Undescended testicle	2
	753.7	Anomalies of urachus	2
	754.6	Valgus deformities of feet	2
	755.0	Polydactyly	2
CORTLAND	742.1	Microcephalus	2
	747.0	Patent ductus arteriosus	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	753.0	Renal agenesis & dysgenesis	2
	756.6	Anomalies of diaphragm	2
	759.3	Situs inversus	2
	243.	Congenital hypothyroidism	1
	743.1	Microphthalmos	1
	745.4	Ventricular septal defect	1
	745.5	Ostium secundum atrial septal defect	1
	745.8	Other defect of septal closure	1
	747.3	Anomalies of pulmonary artery	1
	749.2	Cleft palate with cleft lip	1
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	1
	752.5	Undescended testicle	1
	752.6	Hypospadias & epispadias	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	754.3	Congenital dislocation of hip	1
	756.0	Anomalies of skull and face bones	1
	756.8	Other specified anomalies of muscle, tendon, fascia, etc.	1
	757.6	Specified anomalies of breast	1
	758.6	Gonadal dysgenesis	1
DELAWARE	752.6	Hypospadias & epispadias	4
	745.5	Ostium secundum atrial septal defect	3
	743.1	Microphthalmos	2
	746.0	Anomalies of pulmonary valve	2
	747.0	Patent ductus arteriosus	2

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

Birth Year: 2006

County	ICD-9 Code	Description	Number
DELAWARE	753.2	Obstructive defects of renal pelvis & ureter	2
	756.7	Anomalies of abdominal wall	2
	741.9	Spina bifida w/o hydrocephalus	1
	742.2	Reduction deformities of brain	1
	742.4	Other specified anomalies of brain	1
	745.1	Transposition of great vessels	1
	745.4	Ventricular septal defect	1
	746.4	Congenital insufficiency of aortic valve	1
	748.5	Agenesis, hypoplasia & dysplasia, lung	1
	749.1	Cleft lip	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	752.5	Undescended testicle	1
	752.8	Other specified anomalies of genital organs	1
	753.3	Other specified anomalies of kidney	1
	754.1	Deformities of sternocleidomastoid muscle	1
	755.2	Reduction deformities of upper limb	1
	756.0	Anomalies of skull and face bones	1
DUTCHESS	745.4	Ventricular septal defect	11
	752.6	Hypospadias & epispadias	10
	754.3	Congenital dislocation of hip	10
	753.2	Obstructive defects of renal pelvis & ureter	8
	745.5	Ostium secundum atrial septal defect	7
	749.0	Cleft palate	7
	750.5	Congenital hypertrophic pyloric stenosis	7
	747.0	Patent ductus arteriosus	6
	752.5	Undescended testicle	5
	756.0	Anomalies of skull and face bones	4
	756.4	Chondrodystrophy	4
	758.0	Down syndrome	4
ERIE	747.0	Patent ductus arteriosus	83
	745.4	Ventricular septal defect	63
	752.5	Undescended testicle	59
	752.6	Hypospadias & epispadias	48
	745.5	Ostium secundum atrial septal defect	46
	755.0	Polydactyly	40
	750.5	Congenital hypertrophic pyloric stenosis	38
	753.2	Obstructive defects of renal pelvis & ureter	30
	747.3	Anomalies of pulmonary artery	24
	746.8	Other specified anomalies of heart	22
ESSEX	746.3	Congenital stenosis of aortic valve	1
	747.0	Patent ductus arteriosus	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	751.1	Atresia & stenosis of small intestine	1

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County	ICD-9 Code	Description	Number
ESSEX	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	1
	755.0	Polydactyly	1
	756.7	Anomalies of abdominal wall	1
FRANKLIN	752.6	Hypospadias & epispadias	4
	754.6	Valgus deformities of feet	2
	749.2	Cleft palate with cleft lip	1
	752.5	Undescended testicle	1
	752.8	Other specified anomalies of genital organs	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	754.5	Varus deformities of feet	1
	756.7	Anomalies of abdominal wall	1
FULTON	745.4	Ventricular septal defect	7
	747.0	Patent ductus arteriosus	3
	752.5	Undescended testicle	3
	743.1	Microphthalmos	2
	745.5	Ostium secundum atrial septal defect	2
	746.8	Other specified anomalies of heart	2
	752.6	Hypospadias & epispadias	2
	758.0	Down syndrome	2
	270.2	Other disturbances of aromatic amino acid metabolism	1
	742.2	Reduction deformities of brain	1
	742.5	Other specified anomalies of spinal cord	1
	743.3	Congenital cataract & lens anomalies	1
	745.0	Common truncus	1
	745.2	Tetralogy of Fallot	1
	745.6	Endocardial cushion defects	1
	746.4	Congenital insufficiency of aortic valve	1
	747.1	Coarctation of aorta	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
	753.2	Obstructive defects of renal pelvis & ureter	1
	753.7	Anomalies of urachus	1
	754.3	Congenital dislocation of hip	1
	754.6	Valgus deformities of feet	1
	755.0	Polydactyly	1
	755.2	Reduction deformities of upper limb	1
	755.3	Reduction deformities of lower limb	1
	756.0	Anomalies of skull and face bones	1
	756.6	Anomalies of diaphragm	1
	756.7	Anomalies of abdominal wall	1
	758.1	Patau syndrome	1
	758.2	Edwards syndrome	1

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Birth Year: 2006

County	ICD-9 Code	Description	Number
GENESEE	745.4	Ventricular septal defect	8
	747.0	Patent ductus arteriosus	6
	758.0	Down syndrome	6
	750.5	Congenital hypertrophic pyloric stenosis	3
	752.6	Hypospadias & epispadias	3
	753.2	Obstructive defects of renal pelvis & ureter	3
	754.7	Other deformities of feet	3
	745.6	Endocardial cushion defects	2
	751.7	Anomalies of pancreas	2
	752.5	Undescended testicle	2
GREENE	747.0	Patent ductus arteriosus	3
	750.5	Congenital hypertrophic pyloric stenosis	3
	752.6	Hypospadias & epispadias	2
	758.0	Down syndrome	2
	742.4	Other specified anomalies of brain	1
	745.4	Ventricular septal defect	1
	746.0	Anomalies of pulmonary valve	1
	751.1	Atresia & stenosis of small intestine	1
	752.5	Undescended testicle	1
	754.5	Varus deformities of feet	1
	754.8	Other specified nonteratogenic anomalies	1
	756.7	Anomalies of abdominal wall	1
HAMILTON	754.3	Congenital dislocation of hip	1
HERKIMER	752.6	Hypospadias & epispadias	6
	754.5	Varus deformities of feet	6
	745.4	Ventricular septal defect	5
	752.5	Undescended testicle	4
	754.3	Congenital dislocation of hip	3
	745.5	Ostium secundum atrial septal defect	2
	746.8	Other specified anomalies of heart	2
	747.1	Coarctation of aorta	2
	755.0	Polydactyly	2
	270.0	Disturbances of amino-acid transport	1
	524.0	Major anomalies of jaw size	1
	741.9	Spina bifida w/o hydrocephalus	1
	742.1	Microcephalus	1
	742.4	Other specified anomalies of brain	1
	745.6	Endocardial cushion defects	1
	746.3	Congenital stenosis of aortic valve	1
	746.4	Congenital insufficiency of aortic valve	1
	746.7	Hypoplastic left heart syndrome	1
	747.3	Anomalies of pulmonary artery	1
	748.5	Agenesis, hypoplasia & dysplasia, lung	1

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Birth Year: 2006

County	ICD-9 Code	Description	Number
HERKIMER	749.0	Cleft palate	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	752.4	Anomalies of cervix, vagina & external female genitalia	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	753.3	Other specified anomalies of kidney	1
	753.6	Atresia and stenosis of urethra & bladder neck	1
	754.6	Valgus deformities of feet	1
	754.7	Other deformities of feet	1
	758.0	Down syndrome	1
	758.3	Autosomal deletion syndromes	1
JEFFERSON	754.5	Varus deformities of feet	14
	752.5	Undescended testicle	12
	752.6	Hypospadias & epispadias	9
	753.2	Obstructive defects of renal pelvis & ureter	5
	745.4	Ventricular septal defect	4
	754.3	Congenital dislocation of hip	4
	755.0	Polydactyly	4
	747.0	Patent ductus arteriosus	3
	746.0	Anomalies of pulmonary valve	2
	753.3	Other specified anomalies of kidney	2
	754.7	Other deformities of feet	2
	756.6	Anomalies of diaphragm	2
	759.1	Anomalies of adrenal gland	2
	771.2	Other congenital infections	2
KINGS	747.0	Patent ductus arteriosus	276
	745.5	Ostium secundum atrial septal defect	251
	745.4	Ventricular septal defect	215
	752.5	Undescended testicle	166
	752.6	Hypospadias & epispadias	150
	755.0	Polydactyly	149
	753.2	Obstructive defects of renal pelvis & ureter	142
	746.8	Other specified anomalies of heart	92
	750.5	Congenital hypertrophic pyloric stenosis	65
	747.3	Anomalies of pulmonary artery	62
LEWIS	749.0	Cleft palate	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	752.6	Hypospadias & epispadias	2
	524.0	Major anomalies of jaw size	1
	743.3	Congenital cataract & lens anomalies	1
	745.4	Ventricular septal defect	1
	746.6	Congenital mitral insufficiency	1
	749.1	Cleft lip	1
	751.3	Hirschprung's disease & other functional disorders of colon	1

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Birth Year: 2006

County	ICD-9 Code	Description	Number
LEWIS	751.6	Anomalies of gallbladder, bile ducts, and liver	1
	754.5	Varus deformities of feet	1
	754.7	Other deformities of feet	1
	754.8	Other specified nonteratogenic anomalies	1
	756.0	Anomalies of skull and face bones	1
LIVINGSTON	752.6	Hypospadias & epispadias	5
	745.4	Ventricular septal defect	3
	749.0	Cleft palate	3
	752.5	Undescended testicle	3
	750.5	Congenital hypertrophic pyloric stenosis	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	524.0	Major anomalies of jaw size	1
	742.2	Reduction deformities of brain	1
	743.1	Microphthalmos	1
	743.6	Congenital anomalies of eyelids, lacrimal system & orbit	1
	749.2	Cleft palate with cleft lip	1
	751.5	Other anomalies of intestine	1
	751.7	Anomalies of pancreas	1
	753.0	Renal agenesis & dysgenesis	1
	753.7	Anomalies of urachus	1
	754.3	Congenital dislocation of hip	1
	754.5	Varus deformities of feet	1
	755.0	Polydactyly	1
	755.1	Syndactyly	1
	756.1	Anomalies of spine	1
	758.0	Down syndrome	1
MADISON	742.1	Microcephalus	2
	745.4	Ventricular septal defect	2
	747.0	Patent ductus arteriosus	2
	749.0	Cleft palate	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	751.1	Atresia & stenosis of small intestine	2
	752.6	Hypospadias & epispadias	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	756.0	Anomalies of skull and face bones	2
	758.0	Down syndrome	2
MONROE	752.6	Hypospadias & epispadias	50
	753.2	Obstructive defects of renal pelvis & ureter	48
	750.5	Congenital hypertrophic pyloric stenosis	22
	755.0	Polydactyly	21
	745.4	Ventricular septal defect	20
	758.0	Down syndrome	16
	752.5	Undescended testicle	15

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Birth Year: 2006

County	ICD-9 Code	Description	Number
MONROE	754.5	Varus deformities of feet	14
	745.5	Ostium secundum atrial septal defect	13
	746.0	Anomalies of pulmonary valve	13
MONTGOMERY	752.6	Hypospadias & epispadias	5
	752.5	Undescended testicle	4
	754.3	Congenital dislocation of hip	4
	747.0	Patent ductus arteriosus	3
	750.5	Congenital hypertrophic pyloric stenosis	3
	753.2	Obstructive defects of renal pelvis & ureter	3
	746.0	Anomalies of pulmonary valve	2
	749.0	Cleft palate	2
	243.	Congenital hypothyroidism	1
	524.0	Major anomalies of jaw size	1
	742.1	Microcephalus	1
	742.3	Congenital hydrocephalus	1
	742.4	Other specified anomalies of brain	1
	745.4	Ventricular septal defect	1
	745.6	Endocardial cushion defects	1
	751.1	Atresia & stenosis of small intestine	1
	751.5	Other anomalies of intestine	1
	754.5	Varus deformities of feet	1
	754.6	Valgus deformities of feet	1
	755.0	Polydactyly	1
	758.0	Down syndrome	1
	759.8	Other specified anomalies	1
NASSAU	752.5	Undescended testicle	80
	753.2	Obstructive defects of renal pelvis & ureter	79
	752.6	Hypospadias & epispadias	72
	745.4	Ventricular septal defect	71
	747.0	Patent ductus arteriosus	60
	750.5	Congenital hypertrophic pyloric stenosis	44
	752.8	Other specified anomalies of genital organs	38
	745.5	Ostium secundum atrial septal defect	35
	755.0	Polydactyly	33
	747.3	Anomalies of pulmonary artery	19
NEW YORK	745.5	Ostium secundum atrial septal defect	114
	745.4	Ventricular septal defect	109
	752.5	Undescended testicle	64
	752.6	Hypospadias & epispadias	61
	753.2	Obstructive defects of renal pelvis & ureter	60
	747.0	Patent ductus arteriosus	56
	755.0	Polydactyly	53
	746.8	Other specified anomalies of heart	44

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Birth Year: 2006

County	ICD-9 Code	Description	Number
NEW YORK	750.5	Congenital hypertrophic pyloric stenosis	37
	754.3	Congenital dislocation of hip	35
NIAGARA	745.4	Ventricular septal defect	14
	752.5	Undescended testicle	13
	753.2	Obstructive defects of renal pelvis & ureter	11
	747.0	Patent ductus arteriosus	8
	752.6	Hypospadias & epispadias	8
	755.0	Polydactyly	6
	746.8	Other specified anomalies of heart	5
	758.0	Down syndrome	5
	745.2	Tetralogy of Fallot	4
	751.4	Anomalies of intestinal fixation	4
	754.6	Valgus deformities of feet	4
ONEIDA	752.6	Hypospadias & epispadias	16
	745.4	Ventricular septal defect	15
	752.5	Undescended testicle	14
	747.0	Patent ductus arteriosus	8
	750.5	Congenital hypertrophic pyloric stenosis	8
	754.3	Congenital dislocation of hip	7
	742.3	Congenital hydrocephalus	6
	742.4	Other specified anomalies of brain	6
	753.2	Obstructive defects of renal pelvis & ureter	6
	755.0	Polydactyly	6
	755.6	Other anomalies of lower limb including pelvic girdle	6
ONONDAGA	752.5	Undescended testicle	36
	752.6	Hypospadias & epispadias	35
	745.4	Ventricular septal defect	32
	755.0	Polydactyly	20
	750.5	Congenital hypertrophic pyloric stenosis	17
	747.0	Patent ductus arteriosus	16
	753.2	Obstructive defects of renal pelvis & ureter	14
	756.1	Anomalies of spine	13
	746.0	Anomalies of pulmonary valve	10
	746.8	Other specified anomalies of heart	10
ONTARIO	752.5	Undescended testicle	6
	746.0	Anomalies of pulmonary valve	3
	750.5	Congenital hypertrophic pyloric stenosis	3
	753.2	Obstructive defects of renal pelvis & ureter	3
	754.3	Congenital dislocation of hip	3
	756.0	Anomalies of skull and face bones	3
	243.	Congenital hypothyroidism	2
	749.1	Cleft lip	2

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County	ICD-9 Code	Description	Number
ONTARIO	752.6	Hypospadias & epispadias	2
ORANGE	753.2	Obstructive defects of renal pelvis & ureter	34
	745.4	Ventricular septal defect	25
	752.5	Undescended testicle	23
	752.6	Hypospadias & epispadias	22
	745.5	Ostium secundum atrial septal defect	21
	747.0	Patent ductus arteriosus	19
	750.5	Congenital hypertrophic pyloric stenosis	15
	758.0	Down syndrome	9
	755.0	Polydactyly	7
	742.3	Congenital hydrocephalus	6
	754.5	Varus deformities of feet	6
ORLEANS	750.5	Congenital hypertrophic pyloric stenosis	6
	747.0	Patent ductus arteriosus	5
	752.6	Hypospadias & epispadias	4
	745.4	Ventricular septal defect	2
	752.5	Undescended testicle	2
	277.0	Cystic fibrosis	1
	333.2	Myoclonus	1
	742.2	Reduction deformities of brain	1
	742.3	Congenital hydrocephalus	1
	743.8	Other specified anomalies of eye	1
	751.5	Other anomalies of intestine	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	753.6	Atresia and stenosis of urethra & bladder neck	1
	756.0	Anomalies of skull and face bones	1
	756.7	Anomalies of abdominal wall	1
	758.0	Down syndrome	1
OSWEGO	745.4	Ventricular septal defect	9
	752.6	Hypospadias & epispadias	9
	750.5	Congenital hypertrophic pyloric stenosis	8
	752.5	Undescended testicle	8
	753.2	Obstructive defects of renal pelvis & ureter	7
	747.0	Patent ductus arteriosus	6
	524.0	Major anomalies of jaw size	5
	754.7	Other deformities of feet	5
	753.0	Renal agenesis & dysgenesis	4
	746.8	Other specified anomalies of heart	3
OTSEGO	750.5	Congenital hypertrophic pyloric stenosis	3
	752.6	Hypospadias & epispadias	3
	742.3	Congenital hydrocephalus	2
	745.4	Ventricular septal defect	2

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County	ICD-9 Code	Description	Number
OTSEGO	747.0	Patent ductus arteriosus	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	756.7	Anomalies of abdominal wall	2
	273.8	Other disorders of plasma protien	1
	277.0	Cystic fibrosis	1
	745.5	Ostium secundum atrial septal defect	1
	748.0	Choanal atresia	1
	748.3	Other anomalies of larynx, trachea, & bronchus	1
	748.6	Other anomalies of lung	1
	749.1	Cleft lip	1
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	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
	753.3	Other specified anomalies of kidney	1
	753.4	Other specified anomalies of ureter	1
	754.5	Varus deformities of feet	1
	754.7	Other deformities of feet	1
	756.4	Chondrodystrophy	1
	758.0	Down syndrome	1
PUTNAM	753.2	Obstructive defects of renal pelvis & ureter	8
	745.5	Ostium secundum atrial septal defect	6
	745.4	Ventricular septal defect	5
	752.6	Hypospadias & epispadias	4
	747.0	Patent ductus arteriosus	3
	755.0	Polydactyly	3
	333.2	Myoclonus	2
	751.1	Atresia & stenosis of small intestine	2
	752.5	Undescended testicle	2
QUEENS	745.5	Ostium secundum atrial septal defect	170
	745.4	Ventricular septal defect	129
	753.2	Obstructive defects of renal pelvis & ureter	120
	752.5	Undescended testicle	105
	752.6	Hypospadias & epispadias	96
	747.0	Patent ductus arteriosus	79
	755.0	Polydactyly	75
	750.5	Congenital hypertrophic pyloric stenosis	68
	754.3	Congenital dislocation of hip	49
	746.8	Other specified anomalies of heart	42
RENSSELAER	747.0	Patent ductus arteriosus	11
	752.5	Undescended testicle	10
	752.6	Hypospadias & epispadias	8

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Birth Year: 2006

County	ICD-9 Code	Description	Number
RENSSELAER	750.5	Congenital hypertrophic pyloric stenosis	7
	745.4	Ventricular septal defect	5
	753.2	Obstructive defects of renal pelvis & ureter	5
	755.6	Other anomalies of lower limb including pelvic girdle	3
	756.0	Anomalies of skull and face bones	3
	333.2	Myoclonus	2
	745.5	Ostium secundum atrial septal defect	2
	746.0	Anomalies of pulmonary valve	2
	747.3	Anomalies of pulmonary artery	2
	749.2	Cleft palate with cleft lip	2
	754.7	Other deformities of feet	2
	755.2	Reduction deformities of upper limb	2
RICHMOND	752.6	Hypospadias & epispadias	26
	752.5	Undescended testicle	25
	745.5	Ostium secundum atrial septal defect	24
	753.2	Obstructive defects of renal pelvis & ureter	18
	750.5	Congenital hypertrophic pyloric stenosis	17
	745.4	Ventricular septal defect	15
	747.0	Patent ductus arteriosus	11
	755.0	Polydactyly	11
	745.2	Tetralogy of Fallot	5
	746.8	Other specified anomalies of heart	5
	751.1	Atresia & stenosis of small intestine	5
	751.6	Anomalies of gallbladder, bile ducts, and liver	5
	756.0	Anomalies of skull and face bones	5
ROCKLAND	745.5	Ostium secundum atrial septal defect	31
	752.6	Hypospadias & epispadias	25
	747.0	Patent ductus arteriosus	24
	745.4	Ventricular septal defect	21
	752.5	Undescended testicle	18
	753.2	Obstructive defects of renal pelvis & ureter	17
	755.0	Polydactyly	11
	747.3	Anomalies of pulmonary artery	9
	758.0	Down syndrome	9
	750.5	Congenital hypertrophic pyloric stenosis	7
SARATOGA	752.5	Undescended testicle	13
	745.4	Ventricular septal defect	12
	752.6	Hypospadias & epispadias	11
	753.2	Obstructive defects of renal pelvis & ureter	10
	750.5	Congenital hypertrophic pyloric stenosis	8
	745.5	Ostium secundum atrial septal defect	6
	747.0	Patent ductus arteriosus	6
	754.3	Congenital dislocation of hip	6

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County	ICD-9 Code	Description	Number
SARATOGA	754.5	Varus deformities of feet	6
	524.0	Major anomalies of jaw size	4
	742.3	Congenital hydrocephalus	4
	756.0	Anomalies of skull and face bones	4
SCHENECTADY	753.2	Obstructive defects of renal pelvis & ureter	12
	752.6	Hypospadias & epispadias	11
	758.0	Down syndrome	7
	745.4	Ventricular septal defect	6
	752.5	Undescended testicle	4
	333.2	Myoclonus	3
	747.0	Patent ductus arteriosus	3
	750.5	Congenital hypertrophic pyloric stenosis	3
	754.3	Congenital dislocation of hip	3
	755.0	Polydactyly	3
SCHOHARIE	750.5	Congenital hypertrophic pyloric stenosis	3
	752.6	Hypospadias & epispadias	3
	746.4	Congenital insufficiency of aortic valve	2
	747.0	Patent ductus arteriosus	2
	749.1	Cleft lip	2
	282.6	Sickle-cell anemia	1
	742.2	Reduction deformities of brain	1
	746.3	Congenital stenosis of aortic valve	1
	746.6	Congenital mitral insufficiency	1
	746.7	Hypoplastic left heart syndrome	1
	747.8	Other specified anomalies of circulatory system	1
	752.5	Undescended testicle	1
	754.5	Varus deformities of feet	1
	756.7	Anomalies of abdominal wall	1
SCHUYLER	747.0	Patent ductus arteriosus	2
	754.7	Other deformities of feet	2
	743.8	Other specified anomalies of eye	1
	745.1	Transposition of great vessels	1
	746.0	Anomalies of pulmonary valve	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	751.4	Anomalies of intestinal fixation	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	754.3	Congenital dislocation of hip	1
	754.5	Varus deformities of feet	1
SENECA	750.5	Congenital hypertrophic pyloric stenosis	5
	753.0	Renal agenesis & dysgenesis	2
	754.5	Varus deformities of feet	2
	755.6	Other anomalies of lower limb including pelvic girdle	2

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County	ICD-9 Code	Description	Number
SENECA	243.	Congenital hypothyroidism	1
	277.0	Cystic fibrosis	1
	333.2	Myoclonus	1
	745.4	Ventricular septal defect	1
	749.2	Cleft palate with cleft lip	1
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	1
	752.6	Hypospadias & epispadias	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	756.7	Anomalies of abdominal wall	1
ST LAWRENCE	754.6	Valgus deformities of feet	9
	752.5	Undescended testicle	8
	754.5	Varus deformities of feet	8
	752.6	Hypospadias & epispadias	6
	745.4	Ventricular septal defect	4
	753.2	Obstructive defects of renal pelvis & ureter	3
	754.7	Other deformities of feet	3
	742.4	Other specified anomalies of brain	2
	746.2	Ebstein's anomaly	2
	746.4	Congenital insufficiency of aortic valve	2
	747.1	Coarctation of aorta	2
	749.2	Cleft palate with cleft lip	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	754.3	Congenital dislocation of hip	2
	755.0	Polydactyly	2
	756.6	Anomalies of diaphragm	2
STEUBEN	745.5	Ostium secundum atrial septal defect	5
	752.5	Undescended testicle	5
	752.6	Hypospadias & epispadias	4
	753.2	Obstructive defects of renal pelvis & ureter	4
	745.4	Ventricular septal defect	3
	273.8	Other disorders of plasma protien	2
	746.4	Congenital insufficiency of aortic valve	2
	746.8	Other specified anomalies of heart	2
	747.1	Coarctation of aorta	2
	758.0	Down syndrome	2
SUFFOLK	745.5	Ostium secundum atrial septal defect	165
	745.4	Ventricular septal defect	96
	752.6	Hypospadias & epispadias	85
	752.5	Undescended testicle	77
	753.2	Obstructive defects of renal pelvis & ureter	55
	750.5	Congenital hypertrophic pyloric stenosis	51
	747.0	Patent ductus arteriosus	42
	755.0	Polydactyly	42

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

Birth Year: 2006

County	ICD-9 Code	Description	Number
SUFFOLK	747.3	Anomalies of pulmonary artery	35
	754.5	Varus deformities of feet	32
SULLIVAN	747.0	Patent ductus arteriosus	8
	745.5	Ostium secundum atrial septal defect	3
	752.6	Hypospadias & epispadias	3
	745.4	Ventricular septal defect	2
	751.5	Other anomalies of intestine	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	753.4	Other specified anomalies of ureter	2
	754.7	Other deformities of feet	2
	755.0	Polydactyly	2
	758.0	Down syndrome	2
TIOGA	754.8	Other specified nonteratogenic anomalies	4
	742.3	Congenital hydrocephalus	2
	745.5	Ostium secundum atrial septal defect	2
	747.1	Coarctation of aorta	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	228.1	Lymphangioma, any site	1
	741.0	Spina bifida with hydrocephalus	1
	746.4	Congenital insufficiency of aortic valve	1
	746.7	Hypoplastic left heart syndrome	1
	746.8	Other specified anomalies of heart	1
	747.0	Patent ductus arteriosus	1
	747.3	Anomalies of pulmonary artery	1
	749.0	Cleft palate	1
	750.2	Other specified anomalies, mouth and pharynx	1
	752.5	Undescended testicle	1
	752.6	Hypospadias & epispadias	1
	752.7	Indeterminate sex & pseudo-hermaphroditism	1
	753.1	Cystic kidney disease	1
	754.1	Deformities of sternocleidomastoid muscle	1
	754.3	Congenital dislocation of hip	1
	754.6	Valgus deformities of feet	1
	755.2	Reduction deformities of upper limb	1
TOMPKINS	745.4	Ventricular septal defect	3
	752.5	Undescended testicle	3
	752.6	Hypospadias & epispadias	3
	755.0	Polydactyly	3
	746.8	Other specified anomalies of heart	2
	747.0	Patent ductus arteriosus	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	758.0	Down syndrome	2
	243.	Congenital hypothyroidism	1

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

Birth Year: 2006

County	ICD-9 Code	Description	Number
TOMPKINS	742.2	Reduction deformities of brain	1
	743.1	Microphthalmos	1
	745.2	Tetralogy of Fallot	1
	745.5	Ostium secundum atrial septal defect	1
	745.6	Endocardial cushion defects	1
	747.3	Anomalies of pulmonary artery	1
	748.0	Choanal atresia	1
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	1
	750.4	Other specified anomalies of esophagus	1
	751.3	Hirschprung's disease & other functional disorders of colon	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	754.5	Varus deformities of feet	1
	754.7	Other deformities of feet	1
	754.8	Other specified nonteratogenic anomalies	1
	759.0	Anomalies of spleen	1
	759.6	Other hamartoses,nec	1
	759.8	Other specified anomalies	1
ULSTER	745.5	Ostium secundum atrial septal defect	9
	752.6	Hypospadias & epispadias	9
	752.5	Undescended testicle	7
	745.4	Ventricular septal defect	6
	749.0	Cleft palate	5
	755.0	Polydactyly	5
	756.0	Anomalies of skull and face bones	5
	746.0	Anomalies of pulmonary valve	4
	746.8	Other specified anomalies of heart	4
	747.1	Coarctation of aorta	4
	753.2	Obstructive defects of renal pelvis & ureter	4
WARREN	745.4	Ventricular septal defect	5
	753.2	Obstructive defects of renal pelvis & ureter	4
	752.5	Undescended testicle	3
	754.5	Varus deformities of feet	3
	742.3	Congenital hydrocephalus	2
	745.1	Transposition of great vessels	2
	747.0	Patent ductus arteriosus	2
	749.1	Cleft lip	2
	752.6	Hypospadias & epispadias	2
	754.3	Congenital dislocation of hip	2
WASHINGTON	752.6	Hypospadias & epispadias	6
	753.2	Obstructive defects of renal pelvis & ureter	4
	745.4	Ventricular septal defect	3
	745.5	Ostium secundum atrial septal defect	3
	747.0	Patent ductus arteriosus	3

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

Birth Year: 2006

County	ICD-9 Code	Description	Number
WASHINGTON	752.5	Undescended testicle	3
	756.0	Anomalies of skull and face bones	3
	758.0	Down syndrome	3
	749.2	Cleft palate with cleft lip	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	755.0	Polydactyly	2
	755.2	Reduction deformities of upper limb	2
	756.6	Anomalies of diaphragm	2
	756.7	Anomalies of abdominal wall	2
WAYNE	753.2	Obstructive defects of renal pelvis & ureter	9
	752.5	Undescended testicle	6
	750.5	Congenital hypertrophic pyloric stenosis	5
	752.6	Hypospadias & epispadias	5
	754.3	Congenital dislocation of hip	3
	754.5	Varus deformities of feet	3
	755.0	Polydactyly	3
	228.1	Lymphangioma, any site	2
	745.4	Ventricular septal defect	2
	745.5	Ostium secundum atrial septal defect	2
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	2
	753.0	Renal agenesis & dysgenesis	2
	753.3	Other specified anomalies of kidney	2
WESTCHESTER	745.5	Ostium secundum atrial septal defect	56
	745.4	Ventricular septal defect	53
	752.6	Hypospadias & epispadias	53
	752.5	Undescended testicle	42
	753.2	Obstructive defects of renal pelvis & ureter	40
	747.0	Patent ductus arteriosus	33
	750.5	Congenital hypertrophic pyloric stenosis	28
	755.0	Polydactyly	28
	746.8	Other specified anomalies of heart	14
	747.3	Anomalies of pulmonary artery	13
WYOMING	752.5	Undescended testicle	4
	745.4	Ventricular septal defect	3
	754.3	Congenital dislocation of hip	3
	756.0	Anomalies of skull and face bones	3
	740.0	Anencephalus	1
	741.9	Spina bifida w/o hydrocephalus	1
	744.2	Other specified anomalies of ear	1
	745.2	Tetralogy of Fallot	1
	746.6	Congenital mitral insufficiency	1
	747.0	Patent ductus arteriosus	1
	749.1	Cleft lip	1

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

Birth Year: 2006

County	ICD-9 Code	Description	Number
WYOMING	752.6	Hypospadias & epispadias	1
	753.1	Cystic kidney disease	1
	753.3	Other specified anomalies of kidney	1
	754.7	Other deformities of feet	1
	758.2	Edwards syndrome	1
YATES	755.6	Other anomalies of lower limb including pelvic girdle	2
	747.1	Coarctation of aorta	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	751.5	Other anomalies of intestine	1
	752.5	Undescended testicle	1
	754.3	Congenital dislocation of hip	1

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

Introduction

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 improve case ascertainment and monitor hospital reporting (Appendix 3). In this section, CMR live birth prevalences are compared with the national prevalence estimates for 21 selected defects developed by the Centers for Disease Control and Prevention (CDC) and the National Birth Defects Prevention Network (NBDPN).[1] The 21 defects were selected as they are generally diagnosed soon after birth and the accuracy of diagnosis should be similar across sites[1].These estimates were based on 11 registries which use active case-finding. Active case-finding uses data collection specialists who go to hospitals to identify and abstract records of children with malformations. The active case-finding systems were chosen as they have similar methodology and prevalence estimates are usually higher in systems using active case finding, although variation was observed even among the 11 active case finding systems (See Figure 2 in Parker[1]).

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

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

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

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: 2004-2006)

Birth Defect Category	New York City	Upstate NY	New York State	NBDPN 2004-2006
Central nervous system defects				
Anencephalus	0.4	0.6	0.5	2.2
Spina bifida without anencephalus	**2.1**	**2.2**	**2.2**	3.7
Encephalocele	**0.5**	**0.5**	**0.5**	0.8
Eye defects				
Anophthalmia/ microphthalmia	**0.9**	**1.5**	**1.3**	2.1
Cardiovascular defects				
Common truncus	**0.9**	**0.7**	**0.8**	0.7
Transposition of great arteries	**4.0**	**4.7**	**4.4**	3.0
Tetralogy of Fallot	**4.9**	**4.9**	**4.9**	4.1
Endocardial cushion defect	2.7	3.0	2.9	4.7
Hypoplastic left heart syndrome	**2.2**	**2.9**	**2.5**	2.3
Orofacial defects				
Cleft palate without cleft lip	5.1	**6.6**	**5.9**	6.5
Cleft lip with and without cleft palate	6.1	**8.7**	**7.4**	10.9
Gastrointestinal defects				
Esophageal atresia/ tracheosophageal	**2.5**	**2.4**	**2.4**	2.1
Rectal and large intestinal	**4.1**	**4.3**	**4.2**	4.9
Musculoskeletal defects				
Reduction deformity, upper limbs	1.4	**2.2**	1.8	3.6
Reduction deformity, lower limbs	0.8	**0.9**	**0.8**	1.7
Gastroschisis	1.5	3.1	2.3	4.7
Omphalocele	**1.2**	**1.6**	**1.4**	1.9
Diaphragmatic hernia	**2.0**	**3.4**	**2.7**	2.6
Chromosomal defects				
Trisomy 13	**1.1**	**1.0**	**1.0**	1.2
Down syndrome(trisomy 21)	11.4	**13.3**	**12.4**	13.5
Trisomy 18	1.0	1.1	1.0	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 SE, Mai CT, Canfield MA, Rickard R, Wang Y, Meyer RE, Anderson P, Mason CA, Collins JS, Kirby RS and Correa A Updated national birth prevalence estimates for selected birth defects in the United States, 2004–2006. Birth Defects Research Part A: Clinical and Molecular Teratology, n/a. doi: 10.1002/bdra.20735

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

Section VI
Current Topics

The National Centers for Birth Defects Research and Prevention

Background

To help reduce birth defects among U.S. babies, in 1996, Congress directed the Centers for Disease Control and Prevention (CDC) to establish the Centers of Excellence for Birth Defects Research and Prevention. That same year, the New York Center was established as part of the New York State Department of Health (NYSDOH) Congenital Malformations Registry. Currently there are centers in 9 states including New York.

The major research effort of the Centers is the National Birth Defects Prevention Study (NBDPS). The NBDPS is the largest study of birth defects causes ever undertaken in the United States. Researchers have gathered information from more than 36,000 participants and are using this information to look at key questions on potential causes of birth defects.

The study has three components. First, the Centers identify and collect information on infants who have at least one of 30 major birth defects. The Centers also collect data on selected infants who do not have any major malformations ("controls") and compare the data to that collected for infants with birth defects ("cases"). Second, the infants' mothers are interviewed using a computer-assisted telephone interview (CATI). The interview includes questions about pregnancy and medical histories, lifestyle habits, and possible exposure of the mother or her fetus to harmful substances in the mother's workplace. Third, the Centers collect cheek cell samples from the infants and their parents. Researchers will study the DNA (genetic material) from these cheek cells to find out genetic susceptibilities to birth defects

Topics Being Studied by The New York Center

- Untreated hypertension, thyroid disease and asthma and risk to the mother and baby. Our studies of maternal illness and medication use can help women and physicians make informed decisions about medication use during pregnancy. A future study will look at risks associated with a medication used to treat tension headaches.

- Effects of caffeine on the risk of various birth defects. A study that takes into account genetically determined differences in caffeine metabolism is being conducted.

- Alcohol exposure during pregnancy. We are researching the effects of alcohol use on the risk of several birth defects.

- Climate, air pollution and tap water use and birth defects. Since we are based in the Department of Health's Center for Environmental Health, we have access to resources and expertise that we will use to conduct studies of the effect of climate, air pollution and tap water use on birth defects.

- Home and Occupational Pesticide Exposure (HOPE) Study. New York is a rural state and issues of pesticide exposure are of concern. We have developed a pilot study of home and occupational pesticide exposure on birth defects by collecting additional information from NBDPS participants on pesticide exposure.

NBDPS Research Publications by New York Center Staff

- Maternal occupation and the risk of birth defects: an overview from the National Birth Defects Prevention Study. Occup Environ Med. 2010 Jan;67(1):58-66.

  This study looked at a wide range of occupations and birth defects. We found that one or more birth defects occurred with greater frequency among mothers who worked as janitors/cleaners, scientists and electronic equipment operators. A detailed study to follow up on these findings is being planned.

- Antihypertensive medication use during pregnancy and the risk of cardiovascular malformations. Hypertension. 2009 Jul;54(1):63-70.

  Treatment with antihypertensive medication early in pregnancy was associated with increased risk of having an infant with specific congenital heart defects. Some increases in risk were also observed for treatment that began after the first trimester and for untreated hypertension. We concluded that antihypertensive medication use and/or the underlying hypertension might increase the risk of having an infant with specific congenital heart defects.

- Maternal thyroid disease, thyroid medication use, and selected birth defects in the National Birth Defects Prevention Study. Birth Defects Res A Clin Mol Teratol. 2009 Jul;85(7):621-8.

  Consistent with previous studies, we found that maternal thyroid disease or thyroid medication use was associated with increased risk of hydrocephaly and hypospadias. We also observed associations with certain congenital heart defects and with anorectal atresia. As with other studies of medication use during pregnancy, additional research will be needed to find out whether it is the medical condition or particular medications that pose a risk to the fetus.

- Antifungal drugs and the risk of selected birth defects. Am J Obstet Gynecol. 2008 Feb;198(2):191.e1-7.

  Use of antifungal drugs during early pregnancy was not strongly associated with the risk of most birth defects. We did find an increased risk for a particular congenital heart defect, hypoplastic left heart syndrome. Since previous studies have not looked at antifungal drug use and the risk of specific defects, additional research will be needed to investigate this finding.

- Maternal hypertension, antihypertensive medication use, and the risk of severe hypospadias. Birth Defects Res A Clin Mol Teratol. 2008 Jan;82(1):34-40.

  We found an association between hypertension, antihypertensive medication use, and the risk of severe hypospadias, particularly when medication use began late in pregnancy. Because hypospadias occurs in early pregnancy, we suspect that the hypertension itself plays a role in the higher risk of hypospadias. We are conducting another study to look at individual types of medications and the risk of severe hypospadias.

- Maternal asthma medication use and the risk of gastroschisis. Am J Epidemiol. 2008 Jul 1;168(1):73-9.

  We observed that mothers who used certain asthma medications during pregnancy had an excess risk of giving birth to an infant with gastroschisis, an abdominal wall defect. [Additional studies will be needed to find out whether the increased risk was because of the medications or because of the asthmatic condition.]

- Maternal caffeine consumption and risk of cardiovascular malformations. Birth Defects Res A Clin Mol Teratol. 2007 Jul;79(7):533-43.

Our study found no evidence that moderate intake of caffeine during pregnancy increases the risk of congenital heart defects.

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

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

***See list of minor and excluded codes**

Minor Malformations

214	Lipoma
216	Benign neoplasm of skin
228.01	Hemangioma of skin
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 recognizes that completeness, accuracy and timeliness are the hallmarks of a good surveillance system. However, these attributes exist in tension, "conflicting principles" (Kallen 1988). Steps taken to improve completeness and accuracy may actually reduce timeliness. From the very beginning, the CMR has built in procedures to improve the quality of the data in the CMR. These systems have changed over time (Sekhobo and Druschel 2001; Druschel et al, 2001) and the CMR now has three major approaches to improving data quality: 1) matching to hospital discharge data, the Statewide Planning and Research Cooperative System (SPARCS) for completeness; 2) the web-based reporting system, the Health Provider Network (HPN) 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 recent study (Wang et al, 2005) demonstrated that using hospital discharge data to improve case ascertainment is a valuable and effective method of enhancing birth defect surveillance, particularly for those hospitals with low reporting rates. Hospital audits resulted in not only added new reports (comprised 21.4 percent of all CMR reports) to the CMR but also improved reporting for subsequent years, probably due to hospitals' positively reacting to the audits. Auditing hospitals by CMR staff sent a message to reporting hospitals that both the quality and the quantity of their reports are closely monitored.

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

reporting hospitals. There was a nearly 50 percent reduction in median days used for reporting.

(Wang et al, 2007b).

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

On-site Hospital Audits On-site hospital audits began in August of 2003 as an additional surveillance tool. CMR staff needed to know if all malformations were being captured from medical records, and if the reports were complete and accurate. This was piloted in 2002 and implemented in 2003. The procedure begins when the CMR announces to the hospital that they will be making an "in-house chart review or audit" and requests the hospital in question to send a discharge summary for all children 2 years of age and younger for a specific discharge period, usually one year. The list includes all children discharged in that given year, not just those with a congenital code. This is done so that reportable conditions that may have been miscoded can be identified. CMR staff review the discharge list, comparing it to the list of children who have already been reported to the CMR. A list of reported, not reported and partially reported cases is made. Depending on the time frame and number of auditors available, the entire list or a subset of this list will be sent to the hospital and they will be requested to produce the charts so that CMR staff can review them. CMR staff will spend between 1 and 2 days at a facility reviewing records. At the completion of the review, the facility will be asked to report any case that is considered by the CMR staff as reportable but not previously reported as well as any partially reported cases that need to be completed. A written summary of the audit findings is sent to the Director of Health Information Management including comments that may indicate what chronic reporting problems were evident. Since 2003, 87 hospitals have had an "in-house" audit; 4913 charts have been reviewed; 1835 cases that were not previously reported were flagged and subsequently reported, 430 cases that were partially reported were completed and 187 cases with incorrect diagnoses reported were corrected or deleted.

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

Summary Surveillance requires on-going efforts to respond to changes in resources and technologies. There must also be constant communication and feedback between the reporting sources and the surveillance system. The CMR has developed several methods to monitor and improve the system's completeness, accuracy and timeliness. CMR staff recognize that as a 'passive' reporting system much additional work must be done to be able to provide data of good quality. While 'active'case ascertainment systems seem to provide more completeness and

accuracy, they require much higher funding levels and many more staff. In this era of cutbacks, these funding levels can be difficult to maintain and some of these systems have been forced to

reduce their activities or decrease their areas of coverage. The CMR has seen many staff reductions over the years but by making use of new technologies has been able to improve the system. However, further improvements are needed and the CMR will continue to review procedures and develop new methods. The CMR is currently investigating ways to use hospital discharge summaries (most of which are electronic) as an additional source of case finding. As more and more hospitals go to electronic medical records, these might also assist us in case finding and confirmation of diagnoses. Birth defects are a serious health issue for affected infants and children and their families. With so many different conditions, surveillance of birth defects can be challenging but must be done so that they can be tracked and studied.

References

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

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

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

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

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

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

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

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

Appendix 4

BPA Codes

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

MACDP Code	Condition	ICD-9	BPA Code
CENTRAL NERVOUS SYSTEM			
A01	Anencephaly	740.0, 740.1, 740.2	740.00, 740.01, 740.02, 740.03, 740.08, 740.10, 740.20, 740.21, 740.29
A02	Spina Bifida with Hydrocephaly	741.00, 741.01, 741.02, 741.03	741.000, 741.001, 741.002, 741.003, 741.004, 741.008, 741.009, 741.011, 741.012, 741.013, 741.014, 741.018, 741.019, 741.021, 741.022, 741.023, 741.024, 741.028, 741.029-741.599
A03	Spina Bifida without Hydrocephaly	741.90, 741.91, 741.92, 741.93	741.701, 741.702, 741.703, 741.704, 741.708, 741.709- 741.999
A13	Encephalocele	742.0	742.000, 742.080, 742.085, 742.086, 742.090
A15	Hydrocephaly	742.3	742.300, 742.310, 742.320, 742.380, 742.390
A16	Microcephalus	742.1	742.100, 742.150
EYE / EAR			
B01	Anophthalmia, Microphthalmia	743.00, 743.10, 743.11, 743.12	743.0000, 743.1000, 743.1009, 743.0003, 743.0006, 743.1001, 743.1002
B03	Glaucoma	743.20, 743.21, 743.22	743.2000, 743.210, 743.2001, 743.220
B04	Cataract	743.30, 743.31, 743.32, 743.33, 743.34, 743.35, 743.36, 743.37, 743.39	743.320, 743.325, 743.3261, 743.3262, 743.3263, 743.3264, 743.300, 743.310, 743.340, 743.3806, 743.330, 743.3269, 743.3809, 743.390
B54	Ear anomaly with hearing loss	744.00, 744.01, 744.02, 744.03, 744.04, 744.05, 744.09	744.0001, 744.0101, 744.0002, 744.0902, 744.0203, 744.0204 744.030, 744.0109, 744.0900
CARDIAC			
D01	Truncus arteriosus	745.0	745.000, 745.010
D02	Transposition of great vessels	745.10, 745.11, 745.12, 745.19	745.1001, 745.110, 745.1801, 745.120, 745.1809, 745.190
D03	Tetralogy of Fallot	745.2	745.200, 745.210
D04	Single ventricle	745.3	745.300
D05	VSD	745.4	745.480, 745.485, 745.486, 745.487, 745.490
D52	Hypoplastic left heart	746.7	746.700
D53	Total anomalous pulmonary venous return	747.41	747.420
RESPIRATORY			
E01	Choanal atresia	748.0	748.000
E06	Agenesis of lung	748.5	748.500, 748.510, 748.520, 748.580, 748.590

MACDP Code	Condition	ICD-9	BPA Code
CLEFTS			
F01	Cleft palate	749.00, 749.01, 749.02, 749.03, 749.04	749.010, 749.020, 749.030, 749.050, 749.060, 749.070, 749.090, 749.001, 749.002, 749.003, 749.041, 749.042, 749.043, 749.080
F02	Cleft lip with or without cleft palate	749.10, 749.11, 749.12, 749.13, 749.14, 749.20, 749.21, 749.22, 749.23, 749.24, 749.25	749.1010, 749.1020, 749.1030, 749.1100, 749.120, 749.1901, 749.1011, 749.1021, 749.1031, 749.1012, 749.1022, 749.1032, 749.1103, 749.1104, 749.2900, 749.2011, 749.2021, 749.2031, 749.2012, 749.2022, 749.2032, 749.2103, 749.2104, 749.2015, 749.2025, 749.2035, 749.2105, 749.2203, 749.2905
GASTRO-INTESTINAL			
F14	Stenosis or atresia of duodenum	751.1	751.100
F15	Other stenosis or atresia of small intestine	751.1	751.110, 751.120, 751.190, 751.195
F16	Stenosis or atresia of rectum or anus	751.2	751.210, 751.220, 751.230, 751.240
F17	Hirschsprung's Disease	751.3	751.300, 751.310, 751.320, 751.303
F18	Malrotation of intestine	751.4	751.400, 751.410, 751.420, 751.490, 751.495
F21	Biliary atresia	751.61	751.6501
GENITO-URINARY			
H01	Renal agenesis	753.0	753.000, 753.009, 753.010
H06	Obstruction of kidney or ureter	753.20, 753.21, 753.22	753.220, 753.221, 753.240, 753.241, 753.242, 753.243, 753.244, 753.290, 753.299
H09	Bladder or urethra obstruction	753.6	753.600, 753.610, 753.620, 753.630, 753.690
MUSCULOSKELETAL			
J02	Curvature of spine (scoliosis or lordosis)	754.2	754.200, 754.210, 754.220
J03	Dislocation of hip	754.30, 754.31	754.3000, 754.3010, 754.3020, 754.3030
J11	Arthrogryposis multiplex congenita	754.89	755.800
K01	Reduction deformity - upper limb	755.20, 755.21, 755.22, 755.23, 755.24, 755.25, 755.26, 755.27, 755.28, 755.29	755.200, 755.230, 755.240, 755.2901, 755.5851, 755.2602, 755.265, 755.2702, 755.280, 755.2902, 755.210, 755.218, 755.220, 755.2606, 755.2707, 755.2801, 755.247, 755.2609, 755.2709, 755.2900, 755.5800, 755.5850, 755.59859
K02	Reduction deformity - lower limb	755.30, 755.31, 755.32, 755.33, 755.34, 755.35, 755.36, 755.37, 755.38, 755.39	755.300, 755.330, 755.3401, 755.33901, 755.6851, 755.360, 755.380, 755.3103, 755.3104, 755.318, 755.3801, 755.320, 755.365, 755.366, 755.3802, 755.3409, 755.3900, 755.6859
K05	Amniotic bands	658.80	658.801
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