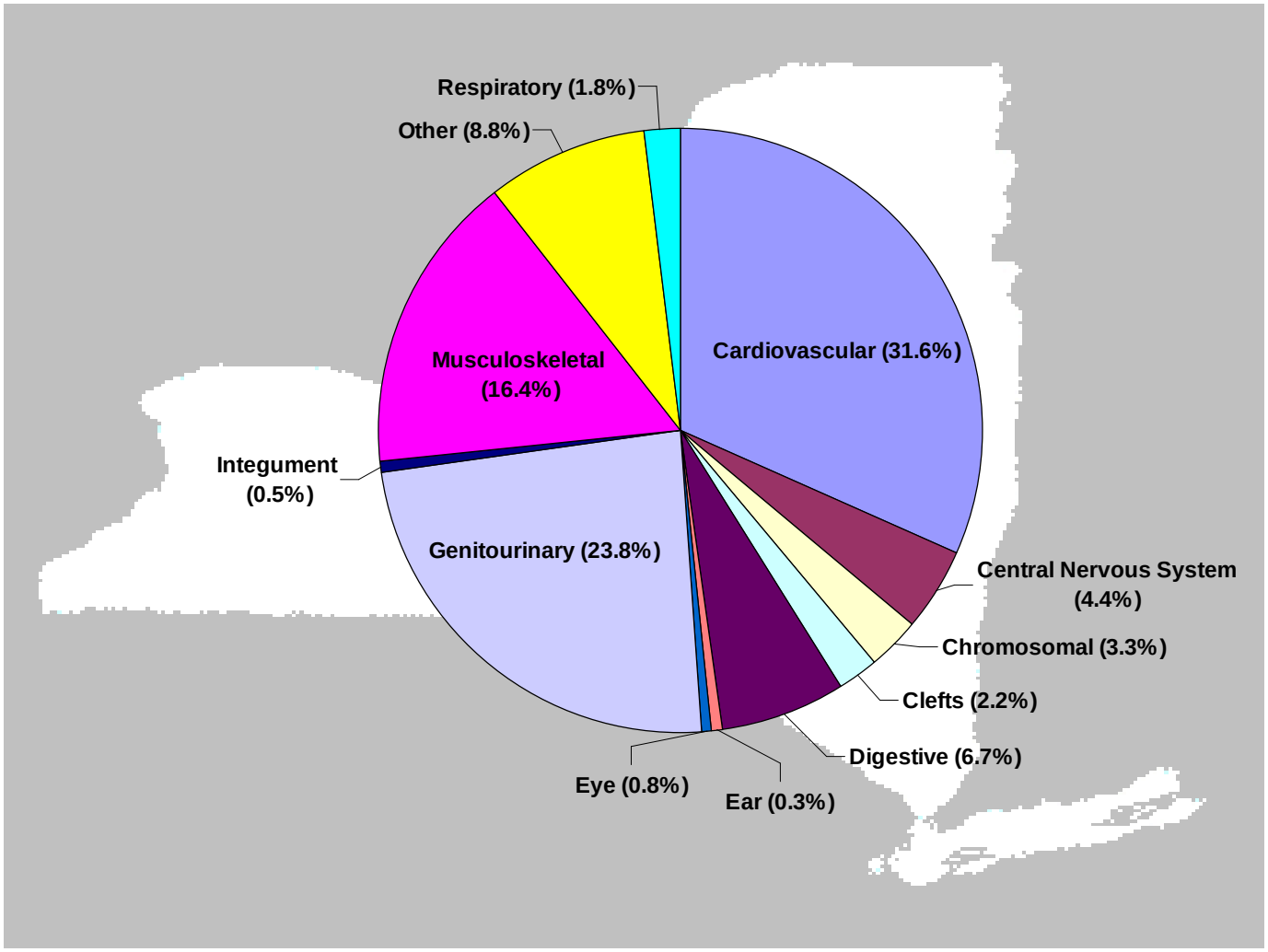
New York State Department of Health

Congenital Malformations Registry Summary Report



Statistical Summary of Children
Born in 2002-2004 and Diagnosed Through 2006

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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TABLE OF CONTENTS

Summary

This Congenital Malformations Registry Summary Report presents rates of congenital malformations occurring among the 503,799 children who were born alive to New York residents in 2002-2004. The children reported with a major congenital malformation represent 4.4% of live births. Males had a higher rate of major congenital malformations than females (5.3% versus 3.4%), and black children had a higher major malformation rate than white children (5.6% versus 4.2%). This information is provided through mandated reporting by hospitals and physicians.

Demographic characteristics of those children reported to the Congenital Malformations Registry (CMR), number of malformations and age at diagnosis 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 sixteenth report from the CMR. Reports are also available by request for the 1983 to 2001 birth cohorts. This report and the reports for 1994-2001 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% of pediatric hospital admissions and about one-third of the total number of pediatric hospital days are for congenital malformations of various types.[3] Little is known about the causes of congenital malformations. Twenty percent may be due to a combination of heredity and other factors; 7.5% may be due to single gene mutations; 6% to chromosome abnormalities; and 5% to maternal illnesses, such as diabetes, infections or anticonvulsant drugs.[4] Approximately 40% to 60% of congenital malformations are of unknown origin.[4,5]

Although radiation and rubella had been linked to birth defects, not until the thalidomide tragedy of the early 1960s was there a widespread interest in possible associations between congenital malformations and environmental agents. During the 1970s, interest continued to grow in birth defects and birth defects surveillance as a result of the growing recognition of the problems of toxic waste dumps such as Love Canal and accidents such as Three Mile Island and Seveso. In response, many states began to develop birth defects registries in order to have data for tracking trends in malformation rates.[6,7] A birth defects registry also makes it possible to respond to public concerns about a 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 from electronic reports is transferred to a DOH UNIX server for updating of the CMR database.

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

2002-2004 Report

This current report presents statistics for major anomalies only (see Appendix 1). 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 2002-2004 report is based on birth certificate matched cases with resident live births from the vital records file used as the denominator. The available birth certificate fields are used to supplement or correct reported data. Birth certificate data are used to establish maternal residence at birth. Birth certificate matching helps eliminate duplicate cases reported under different names. Racial data are not comparable because race is defined by maternal race from the birth certificate. 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 3) 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 2002-2004 and diagnosed before their second birthday. Reports are also available for the 1984 through 2001 birth cohorts. Some reports and additional information are available through the DOH Web site at http://www.health.state.ny.us.

Limitations

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

New York State Population

Based on the U.S. 2000 census, the population of New York State was about 19.0 million; more than 42% of the population lived in New York City. An additional 23% of the population lived in the six counties closest to New York City. In 2002-2004, there were 503,799 resident live births reported to the state's vital registration, 19.2% to black mothers, and 21.6% 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 47.1% of live births were to New York City residents.

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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 2002 to 2004 and reported to the registry with major malformations. Since a new coding system was instituted in 1992, the list of major malformations has been revised (see Appendix 3). Thus, the prevalence rates in this report are not comparable to reports prior to 1992.

The overall occurrences of major malformations for the two years ranged from 4.3% to 4.4% of live births. Male children have a higher rate of major malformations than female children (5.2% to 5.4% versus 3.4%, Tables 1a and 1b). This difference is consistent within different racial groups. The rates for major malformations are somewhat higher for black than for white children (5.5% to 5.6% versus 4.1% to 4.2%). 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 (4.3% to 4.5% versus 4.3%). The smaller number of births in the "other" racial category makes these rates difficult to interpret.

About 79% of children reported with major malformations have only one major malformation (Tables 2a and 2b). 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 (Tables 3a and 3b). All race-sex groups for children with multiple major malformations showed little variation (Tables 4a and 4b)

Section 1 - Table 1a
2002 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	11,008	250,793	4.4	6,882	128,472	5.4	4,126	122,321	3.4
- Non-Hispanic White	5,443	129,899	4.2	3,544	66,642	5.3	1,899	63,257	3.0
- Non-Hispanic Black	2,572	44,956	5.7	1,502	22,838	6.6	1,070	22,118	4.8
- Hispanic	2,206	54,264	4.1	1,340	27,825	4.8	866	26,439	3.3
- Others/Unknown	787	21,674	3.6	496	11,167	4.4	291	10,507	2.8
NYS Excluding NYC									
- All Races	5,742	132,857	4.3	3,683	68,133	5.4	2,059	64,724	3.2
- Non-Hispanic White	4,118	97,855	4.2	2,680	50,179	5.3	1,438	47,676	3.0
- Non-Hispanic Black	801	14,075	5.7	486	7,143	6.8	315	6,932	4.5
- Hispanic	570	14,817	3.8	361	7,690	4.7	209	7,127	2.9
- Others/Unknown	253	6,110	4.1	156	3,121	5.0	97	2,989	3.2
New York City									
- All Races	5,266	117,936	4.5	3,199	60,339	5.3	2,067	57,597	3.6
- Non-Hispanic White	1,325	32,044	4.1	864	16,463	5.2	461	15,581	3.0
- Non-Hispanic Black	1,771	30,881	5.7	1,016	15,695	6.5	755	15,186	5.0
- Hispanic	1,636	39,447	4.1	979	20,135	4.9	657	19,312	3.4
- Others/Unknown	534	15,564	3.4	340	8,046	4.2	194	7,518	2.6

Section 1 - Table 1b
2003 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	11,036	252,997	4.4	6,757	129,715	5.2	4,279	123,282	3.5
- Non-Hispanic White	5,528	131,762	4.2	3,505	67,837	5.2	2,023	63,925	3.2
- Non-Hispanic Black	2,484	43,733	5.7	1,432	22,235	6.4	1,052	21,498	4.9
- Hispanic	2,246	54,695	4.1	1,347	27,779	4.8	899	26,916	3.3
- Others/Unknown	778	22,807	3.4	473	11,864	4.0	305	10,943	2.8
NYS Excluding NYC									
- All Races	5,798	133,529	4.3	3,651	68,358	5.3	2,147	65,171	3.3
- Non-Hispanic White	4,142	98,086	4.2	2,651	50,444	5.3	1,491	47,642	3.1
- Non-Hispanic Black	855	13,862	6.2	500	7,010	7.1	355	6,852	5.2
- Hispanic	589	15,322	3.8	364	7,674	4.7	225	7,648	2.9
- Others/Unknown	212	6,259	3.4	136	3,230	4.2	76	3,029	2.5
New York City									
- All Races	5,238	119,468	4.4	3,106	61,357	5.1	2,132	58,111	3.7
- Non-Hispanic White	1,386	33,676	4.1	854	17,393	4.9	532	16,283	3.3
- Non-Hispanic Black	1,629	29,871	5.5	932	15,225	6.1	697	14,646	4.8
- Hispanic	1,657	39,373	4.2	983	20,105	4.9	674	19,268	3.5
- Others/Unknown	566	16,548	3.4	337	8,634	3.9	229	7,914	2.9

Section 1 - Table 1c
2004 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	11,678	246,169	4.7	7,239	125,982	5.7	4,439	120,187	3.7
- Non-Hispanic White	5,708	124,776	4.6	3,675	63,893	5.8	2,033	60,883	3.3
- Non-Hispanic Black	2,603	41,345	6.3	1,513	21,014	7.2	1,090	20,331	5.4
- Hispanic	2,436	56,426	4.3	1,475	28,763	5.1	961	27,663	3.5
- Others/Unknown	931	23,622	3.9	576	12,312	4.7	355	11,310	3.1
NYS Excluding NYC									
- All Races	6,120	127,612	4.8	3,918	65,380	6.0	2,202	62,232	3.5
- Non-Hispanic White	4,288	91,596	4.7	2,781	46,903	5.9	1,507	44,693	3.4
- Non-Hispanic Black	782	11,929	6.6	487	6,095	8.0	295	5,834	5.1
- Hispanic	775	17,168	4.5	483	8,809	5.5	292	8,359	3.5
- Others/Unknown	275	6,919	4.0	167	3,573	4.7	108	3,346	3.2
New York City									
- All Races	5,558	118,557	4.7	3,321	60,602	5.5	2,237	57,955	3.9
- Non-Hispanic White	1,420	33,180	4.3	894	16,990	5.3	526	16,190	3.2
- Non-Hispanic Black	1,821	29,416	6.2	1,026	14,919	6.9	795	14,497	5.5
- Hispanic	1,661	39,258	4.2	992	19,954	5.0	669	19,304	3.5
- Others/Unknown	656	16,703	3.9	409	8,739	4.7	247	7,964	3.1

Section 1 - Table 2a
2002 Births - New York State Residents
Number of Major Malformations Per Child

Number of Malformations	Number of Children	Percent
1	9,006	81.8
2	1,353	12.3
3	404	3.7
4	147	1.3
5	58	0.5
6	23	0.2
7	9	0.1
8	7	0.1
10	1	*
All Children	11,008	100.0

Section 1 - Table 2b
2003 Births - New York State Residents
Number of Major Malformations Per Child

Number of Malformations	Number of Children	Percent
1	8,966	81.2
2	1,387	12.6
3	421	3.8
4	150	1.4
5	65	0.6
6	23	0.2
7	14	0.1
8	6	0.1
9	3	*
10	1	*
All Children	11,036	100.0

Section 1 - Table 2c
2004 Births - New York State Residents
Number of Major Malformations Per Child

Number of Malformations	Number of Children	Percent
1	9,539	86.4
2	1,414	12.8
3	428	3.9
4	171	1.5
5	81	0.7
6	27	0.2
7	9	0.1
8	5	0.0
9	2	*
10	1	*
11	1	*
All Children	11,678	100.0

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

Section 1 - Table 3a
2002 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	9,006	250,793	3.6	5,718	128,472	4.5	3,288	122,321	2.7
- Non-Hispanic White	4,455	129,899	3.4	2,970	66,642	4.5	1,485	63,257	2.3
- Non-Hispanic Black	2,103	44,956	4.7	1,242	22,838	5.4	861	22,118	3.9
- Hispanic	1,809	54,264	3.3	1,109	27,825	4.0	700	26,439	2.6
- Others/Unknown	639	21,674	2.9	397	11,167	3.6	242	10,507	2.3
NYS Excluding NYC									
- All Races	4,673	132,857	3.5	3,048	68,133	4.5	1,625	64,724	2.5
- Non-Hispanic White	3,367	97,855	3.4	2,238	50,179	4.5	1,129	47,676	2.4
- Non-Hispanic Black	659	14,075	4.7	404	7,143	5.7	255	6,932	3.7
- Hispanic	451	14,817	3.0	288	7,690	3.7	163	7,127	2.3
- Others/Unknown	196	6,110	3.2	118	3,121	3.8	78	2,989	2.6
New York City									
- All Races	4,333	117,936	3.7	2,670	60,339	4.4	1,663	57,597	2.9
- Non-Hispanic White	1,088	32,044	3.4	732	16,463	4.4	356	15,581	2.3
- Non-Hispanic Black	1,444	30,881	4.7	838	15,695	5.3	606	15,186	4.0
- Hispanic	1,358	39,447	3.4	821	20,135	4.1	537	19,312	2.8
- Others/Unknown	443	15,564	2.8	279	8,046	3.5	164	7,518	2.2

Section 1 - Table 3b
2003 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	8,966	252,997	3.5	5,579	129,715	4.3	3,387	123,282	2.7
- Non-Hispanic White	4,496	131,762	3.4	2,905	67,837	4.3	1,591	63,925	2.5
- Non-Hispanic Black	2,030	43,733	4.6	1,163	22,235	5.2	867	21,498	4.0
- Hispanic	1,816	54,695	3.3	1,122	27,779	4.0	694	26,916	2.6
- Others/Unknown	624	22,807	2.7	389	11,864	3.3	235	10,943	2.1
NYS Excluding NYC									
- All Races	4,722	133,529	3.5	3,028	68,358	4.4	1,694	65,171	2.6
- Non-Hispanic White	3,375	98,086	3.4	2,204	50,444	4.4	1,171	47,642	2.5
- Non-Hispanic Black	713	13,862	5.1	419	7,010	6.0	294	6,852	4.3
- Hispanic	468	15,322	3.1	297	7,674	3.9	171	7,648	2.2
- Others/Unknown	166	6,259	2.7	108	3,230	3.3	58	3,029	1.9
New York City									
- All Races	4,244	119,468	3.6	2,551	61,357	4.2	1,693	58,111	2.9
- Non-Hispanic White	1,121	33,676	3.3	701	17,393	4.0	420	16,283	2.6
- Non-Hispanic Black	1,317	29,871	4.4	744	15,225	4.9	573	14,646	3.9
- Hispanic	1,348	39,373	3.4	825	20,105	4.1	523	19,268	2.7
- Others/Unknown	458	16,548	2.8	281	8,634	3.3	177	7,914	2.2

Section 1 - Table 3c
2004 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	9,539	246,169	3.9	5,979	125,982	4.7	3,560	120,187	3.0
- Non-Hispanic White	4,677	124,776	3.7	3,053	63,893	4.8	1,624	60,883	2.7
- Non-Hispanic Black	2,126	41,345	5.1	1,240	21,014	5.9	886	20,331	4.4
- Hispanic	1,980	56,426	3.5	1,219	28,763	4.2	761	27,663	2.8
- Others/Unknown	756	23,622	3.2	467	12,312	3.8	289	11,310	2.6
NYS Excluding NYC									
- All Races	4,993	127,612	3.9	3,232	65,380	4.9	1,761	62,232	2.8
- Non-Hispanic White	3,497	91,596	3.8	2,300	46,903	4.9	1,197	44,693	2.7
- Non-Hispanic Black	646	11,929	5.4	399	6,095	6.5	247	5,834	4.2
- Hispanic	624	17,168	3.6	397	8,809	4.5	227	8,359	2.7
- Others/Unknown	226	6,919	3.3	136	3,573	3.8	90	3,346	2.7
New York City									
- All Races	4,546	118,557	3.8	2,747	60,602	4.5	1,799	57,955	3.1
- Non-Hispanic White	1,180	33,180	3.6	753	16,990	4.4	427	16,190	2.6
- Non-Hispanic Black	1,480	29,416	5.0	841	14,919	5.6	639	14,497	4.4
- Hispanic	1,356	39,258	3.5	822	19,954	4.1	534	19,304	2.8
- Others/Unknown	530	16,703	3.2	331	8,739	3.8	199	7,964	2.5

Section 1 - Table 4a
2002 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	2,002	250,793	0.8	1,164	128,472	0.9	838	122,321	0.7
- Non-Hispanic White	988	129,899	0.8	574	66,642	0.9	414	63,257	0.7
- Non-Hispanic Black	469	44,956	1.0	260	22,838	1.1	209	22,118	0.9
- Hispanic	397	54,264	0.7	231	27,825	0.8	166	26,439	0.6
- Others/Unknown	148	21,674	0.7	99	11,167	0.9	49	10,507	0.5
NYS Excluding NYC									
- All Races	1,069	132,857	0.8	635	68,133	0.9	434	64,724	0.7
- Non-Hispanic White	751	97,855	0.8	442	50,179	0.9	309	47,676	0.6
- Non-Hispanic Black	142	14,075	1.0	82	7,143	1.1	60	6,932	0.9
- Hispanic	119	14,817	0.8	73	7,690	0.9	46	7,127	0.6
- Others/Unknown	57	6,110	0.9	38	3,121	1.2	19	2,989	0.6
New York City									
- All Races	933	117,936	0.8	529	60,339	0.9	404	57,597	0.7
- Non-Hispanic White	237	32,044	0.7	132	16,463	0.8	105	15,581	0.7
- Non-Hispanic Black	327	30,881	1.1	178	15,695	1.1	149	15,186	1.0
- Hispanic	278	39,447	0.7	158	20,135	0.8	120	19,312	0.6
- Others/Unknown	91	15,564	0.6	61	8,046	0.8	30	7,518	0.4

Section 1 - Table 4b
2003 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	2,070	252,997	0.8	1,178	129,715	0.9	892	123,282	0.7
- Non-Hispanic White	1,032	131,762	0.8	600	67,837	0.9	432	63,925	0.7
- Non-Hispanic Black	454	43,733	1.0	269	22,235	1.2	185	21,498	0.9
- Hispanic	430	54,695	0.8	225	27,779	0.8	205	26,916	0.8
- Others/Unknown	154	22,807	0.7	84	11,864	0.7	70	10,943	0.6
NYS Excluding NYC									
- All Races	1,076	133,529	0.8	623	68,358	0.9	453	65,171	0.7
- Non-Hispanic White	767	98,086	0.8	447	50,444	0.9	320	47,642	0.7
- Non-Hispanic Black	142	13,862	1.0	81	7,010	1.2	61	6,852	0.9
- Hispanic	121	15,322	0.8	67	7,674	0.9	54	7,648	0.7
- Others/Unknown	46	6,259	0.7	28	3,230	0.9	18	3,029	0.6
New York City									
- All Races	994	119,468	0.8	555	61,357	0.9	439	58,111	0.8
- Non-Hispanic White	265	33,676	0.8	153	17,393	0.9	112	16,283	0.7
- Non-Hispanic Black	312	29,871	1.0	188	15,225	1.2	124	14,646	0.8
- Hispanic	309	39,373	0.8	158	20,105	0.8	151	19,268	0.8
- Others/Unknown	108	16,548	0.7	56	8,634	0.6	52	7,914	0.7

Section 1 - Table 4c
2004 Births - New York State Residents
Percent of Live Births with Two or More Major Malformations
Sex by Race/Ethnicity and Residence

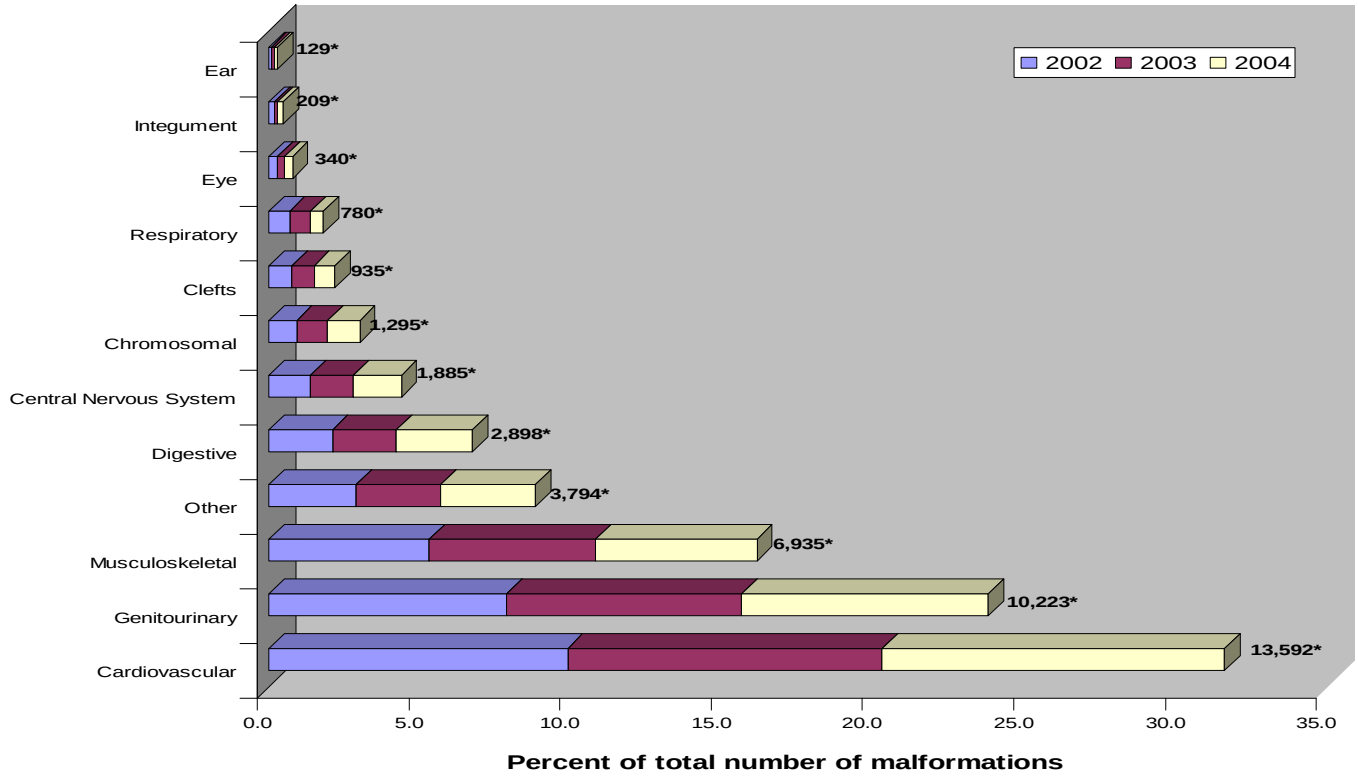
	Both Sexes			Males			Females		
Race and Residence	Infants	Total Births	%	Infants	Total Births	%	Infants	Total Births	%
New York State									
- All Races	2,139	246,169	0.9	1,260	125,982	1.0	879	120,187	0.7
- Non-Hispanic White	1,031	124,776	0.8	622	63,893	1.0	409	60,883	0.7
- Non-Hispanic Black	477	41,345	1.2	273	21,014	1.3	204	20,331	1.0
- Hispanic	456	56,426	0.8	256	28,763	0.9	200	27,663	0.7
- Others/Unknown	175	23,622	0.7	109	12,312	0.9	66	11,310	0.6
NYS Excluding NYC									
- All Races	1,127	127,612	0.9	686	65,380	1.0	441	62,232	0.7
- Non-Hispanic White	791	91,596	0.9	481	46,903	1.0	310	44,693	0.7
- Non-Hispanic Black	136	11,929	1.1	88	6,095	1.4	48	5,834	0.8
- Hispanic	151	17,168	0.9	86	8,809	1.0	65	8,359	0.8
- Others/Unknown	49	6,919	0.7	31	3,573	0.9	18	3,346	0.5
New York City									
- All Races	1,012	118,557	0.9	574	60,602	0.9	438	57,955	0.8
- Non-Hispanic White	240	33,180	0.7	141	16,990	0.8	99	16,190	0.6
- Non-Hispanic Black	341	29,416	1.2	185	14,919	1.2	156	14,497	1.1
- Hispanic	305	39,258	0.8	170	19,954	0.9	135	19,304	0.7
- Others/Unknown	126	16,703	0.8	78	8,739	0.9	48	7,964	0.6

Section II
Major Congenital Malformations by Organ System, 2002-2004

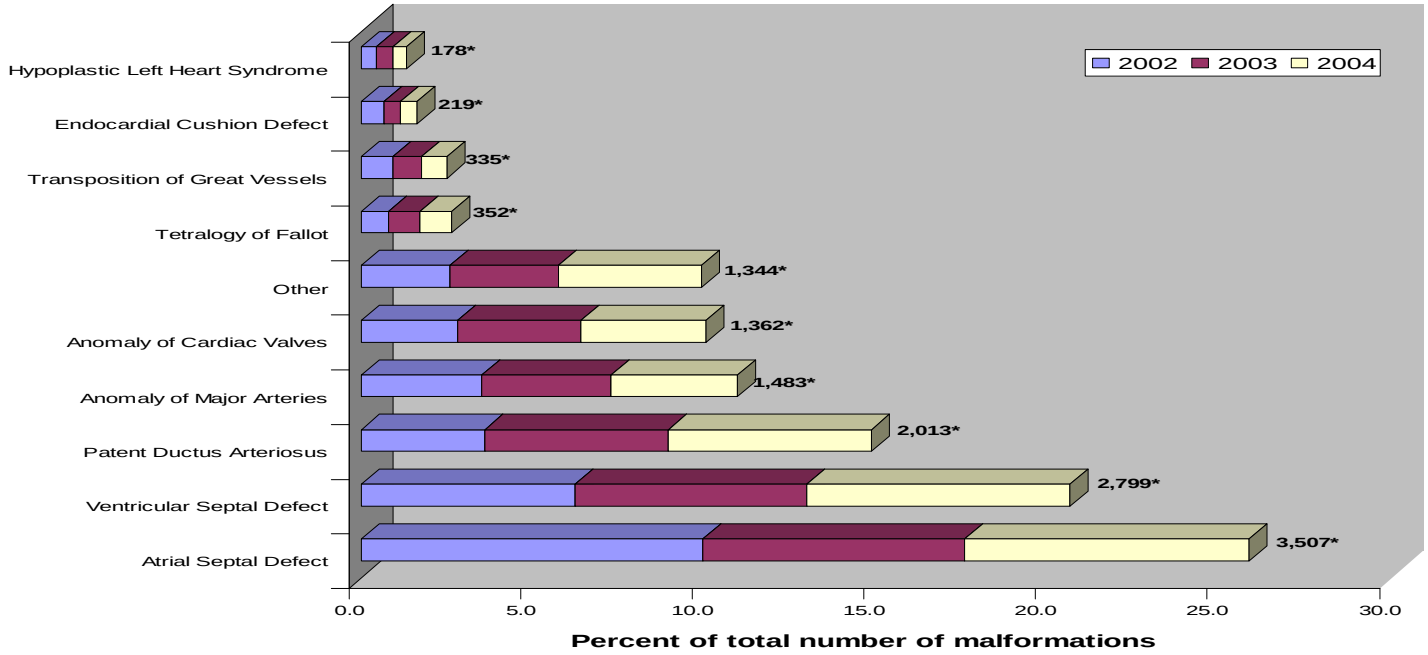
Introduction to Figures

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 the 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.

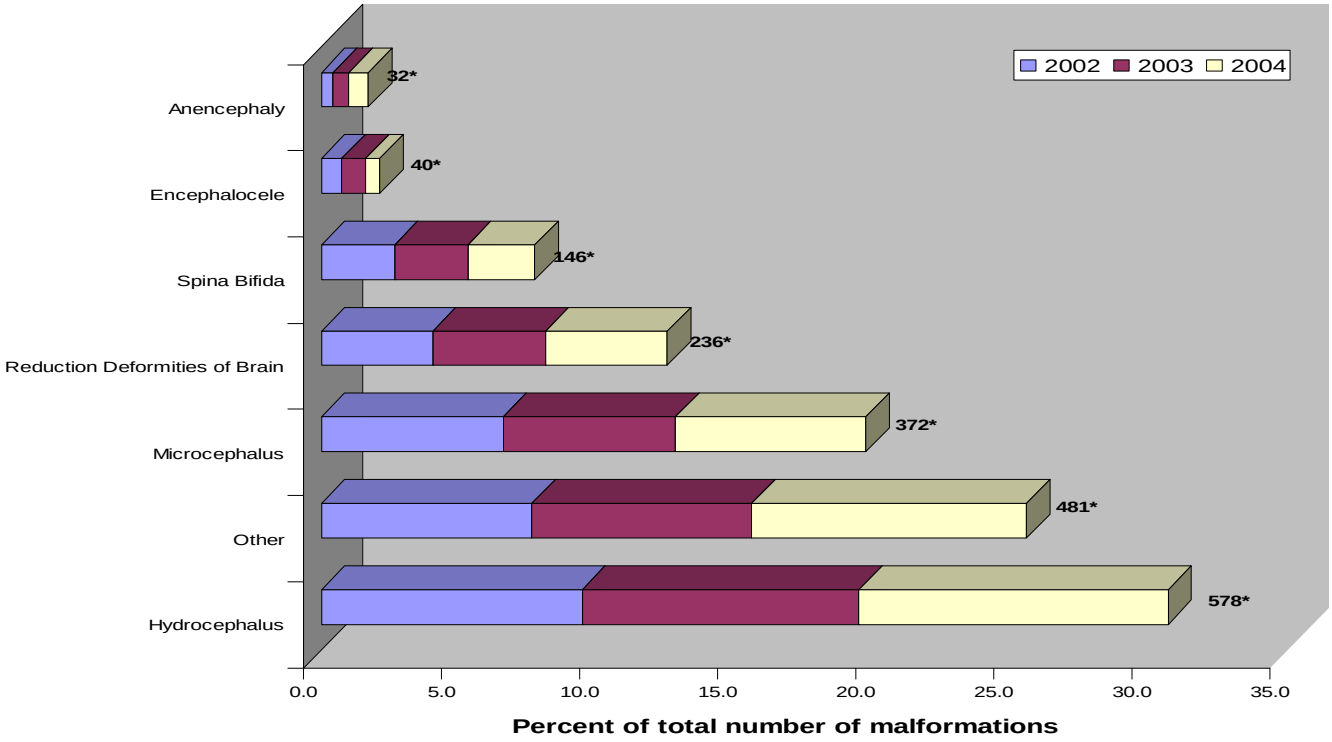
Major Malformations by Organ System
2002 - 2004 Births - New York State Residents
(Number of Children = 33,722)
(* - Number of malformations in each organ system)



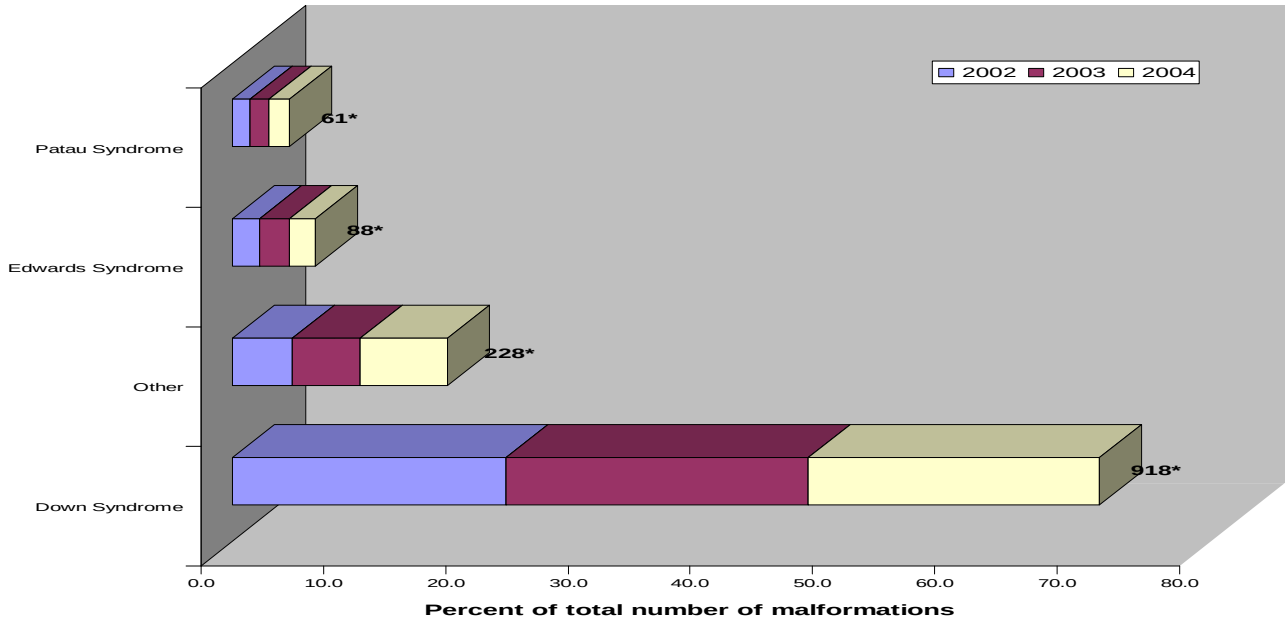
Major Malformations by Organ System
2002 - 2004 Births - New York State Residents
Cardovascular System Subset Category
(Number of Children = 10,579)
(* - Number of malformations in each category)



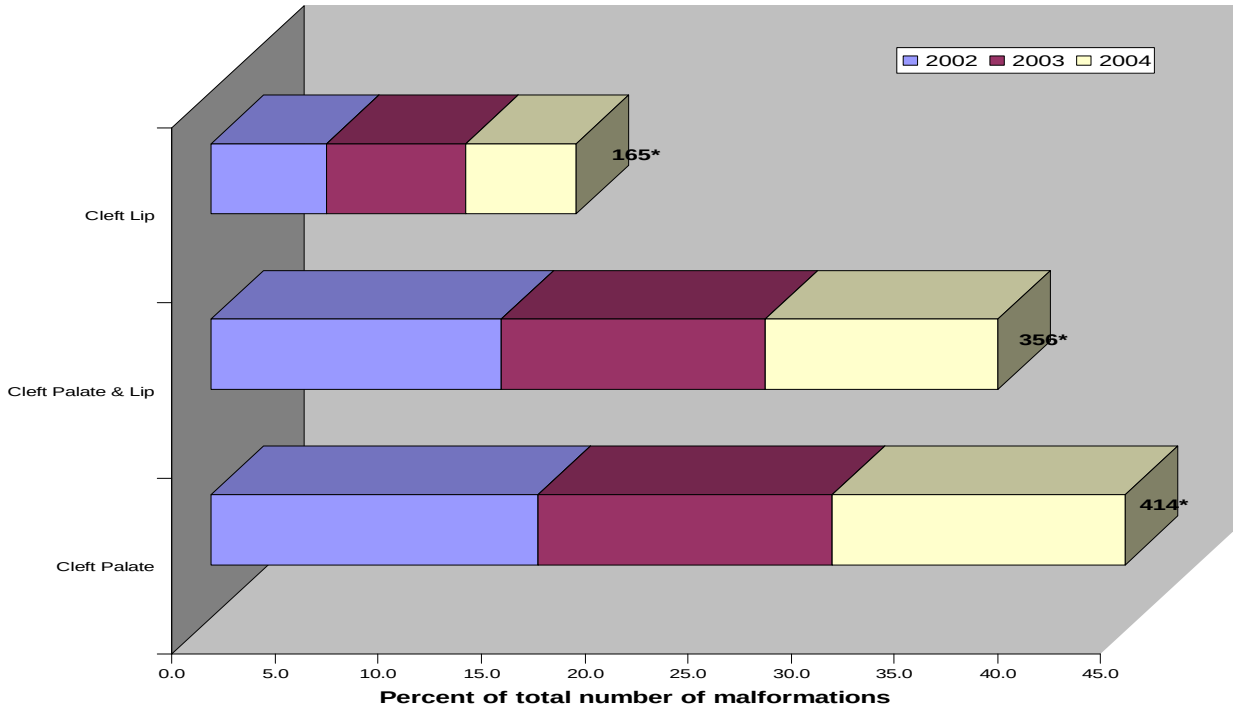
Major Malformations by Organ System
2002 - 2004 Births - New York State Residents
Central Nervous System Subset Category
(Number of Children = 1,690)
(* - Number of malformations in each organ system)



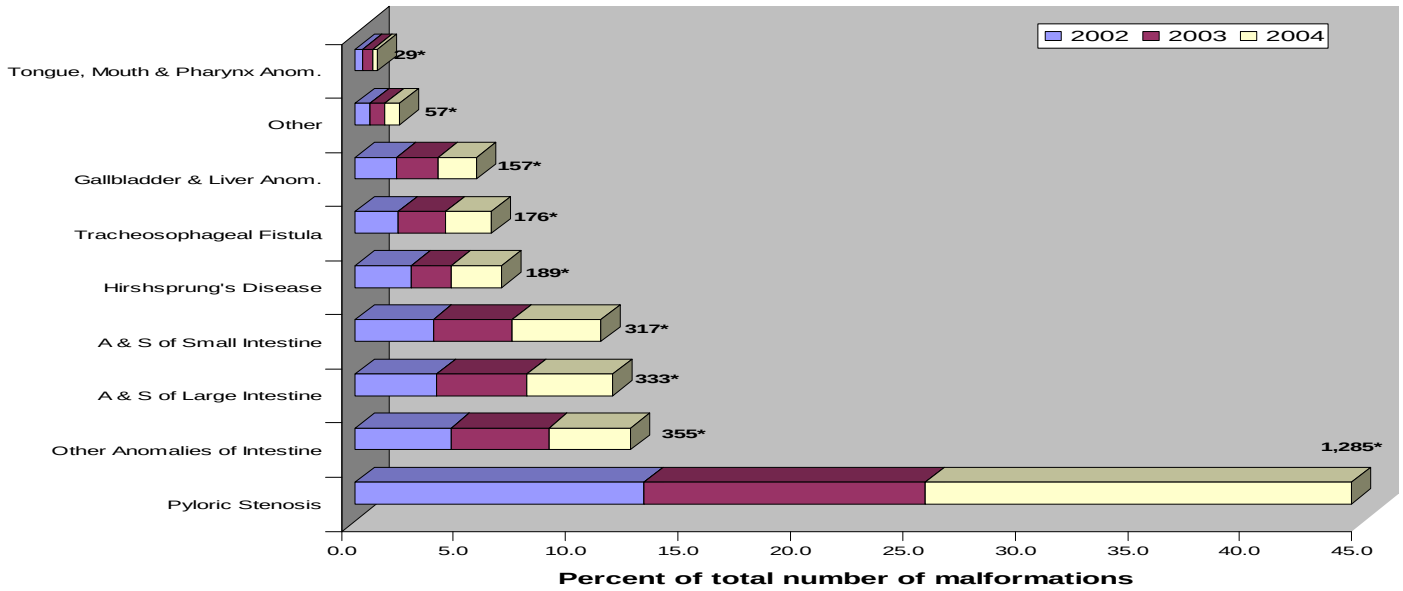
Major Malformations by Organ System
2002 - 2004 Births - New York State Residents
Chromosomal Subset Category
(Number of Children = 1,287)
(* - Number of malformations in each organ system)



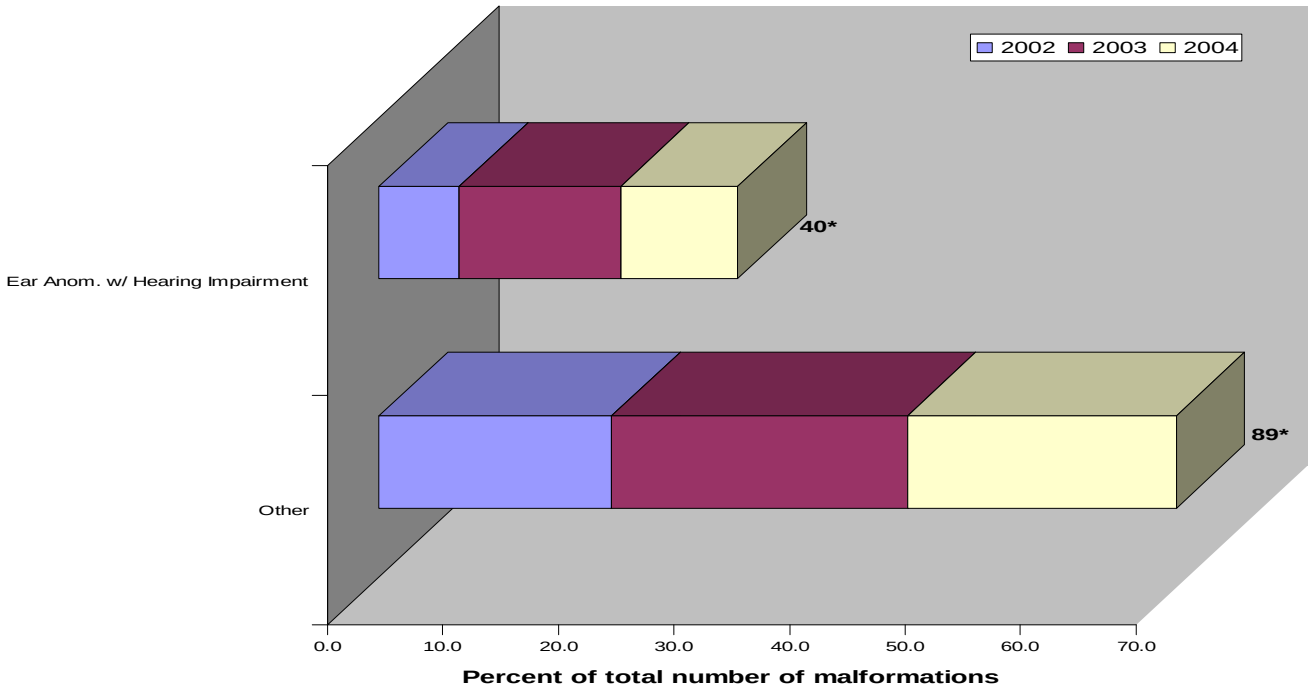
Major Malformations by Organ System
2002 - 2004 Births - New York State Residents
Oral Clefts Subset Category
(Number of Children = 934)
(* - Number of malformations in each organ system)



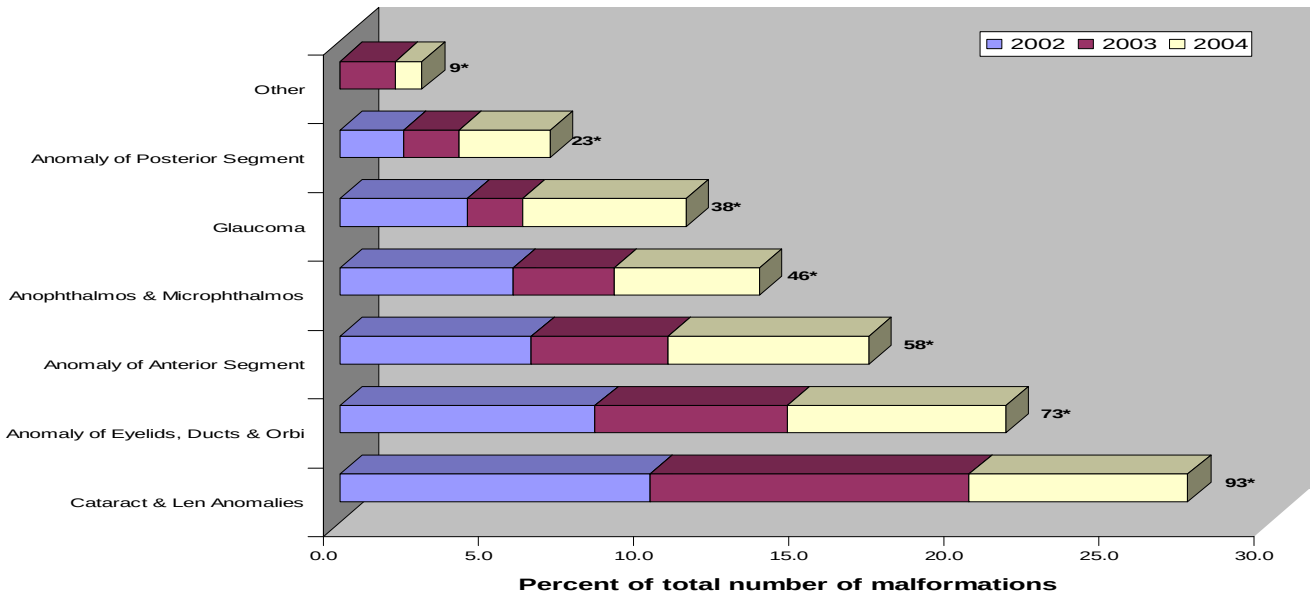
Major Malformations by Organ System
2002 - 2004 Births - New York State Residents
Digestive System Subset Category
(Number of Children = 2,790)
(* - Number of malformations in each organ system)



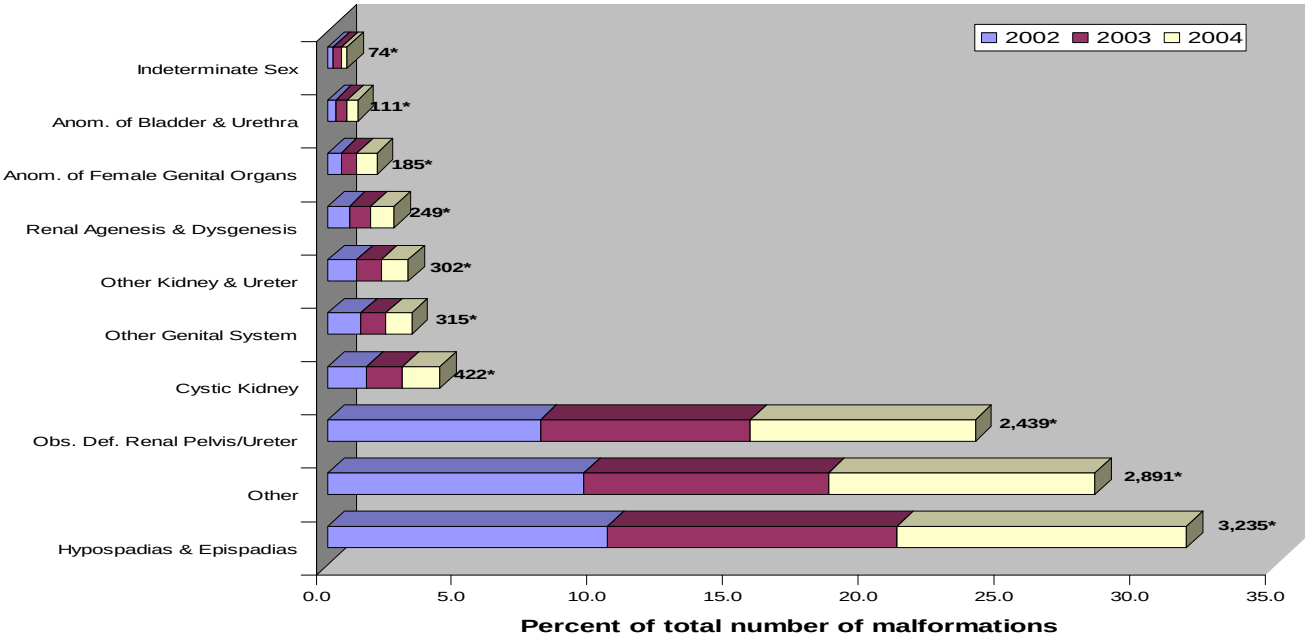
Major Malformations by Organ System
2002 - 2004 Births - New York State Residents
Ear Subset Category
(Number of Children = 126)
(* - Number of malformations in each organ system)



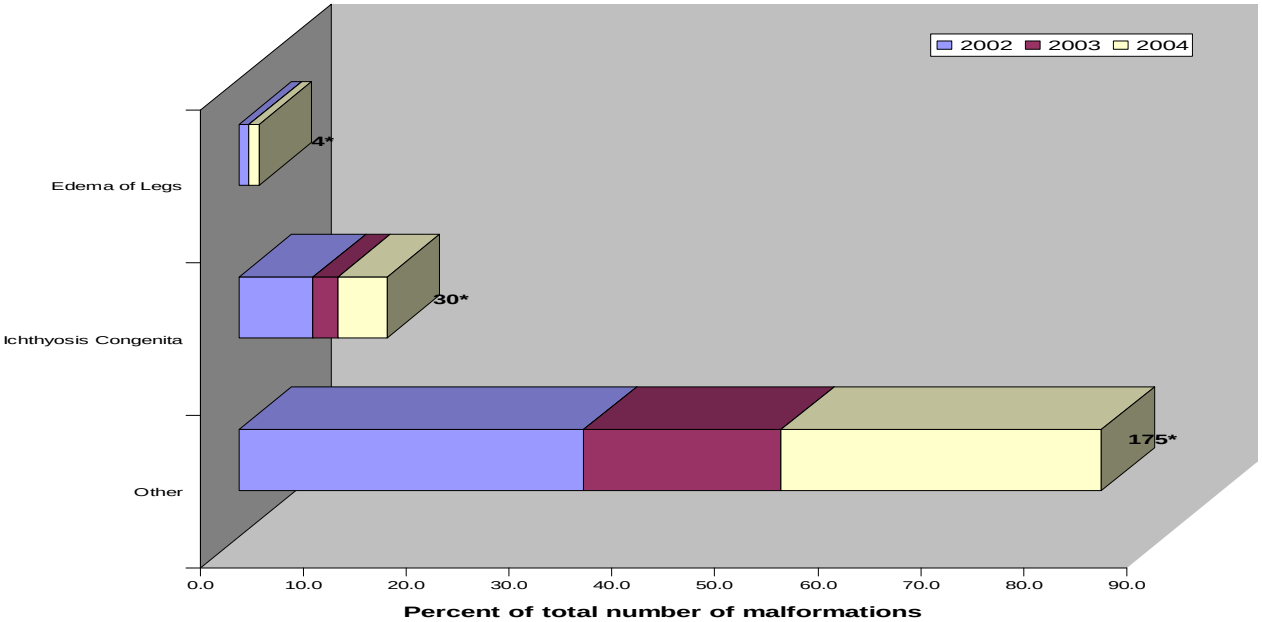
Major Malformations by Organ System
2002 - 2004 Births - New York State Residents
Eye Subset Category
(Number of Children = 340)
(* - Number of malformations in each organ system)



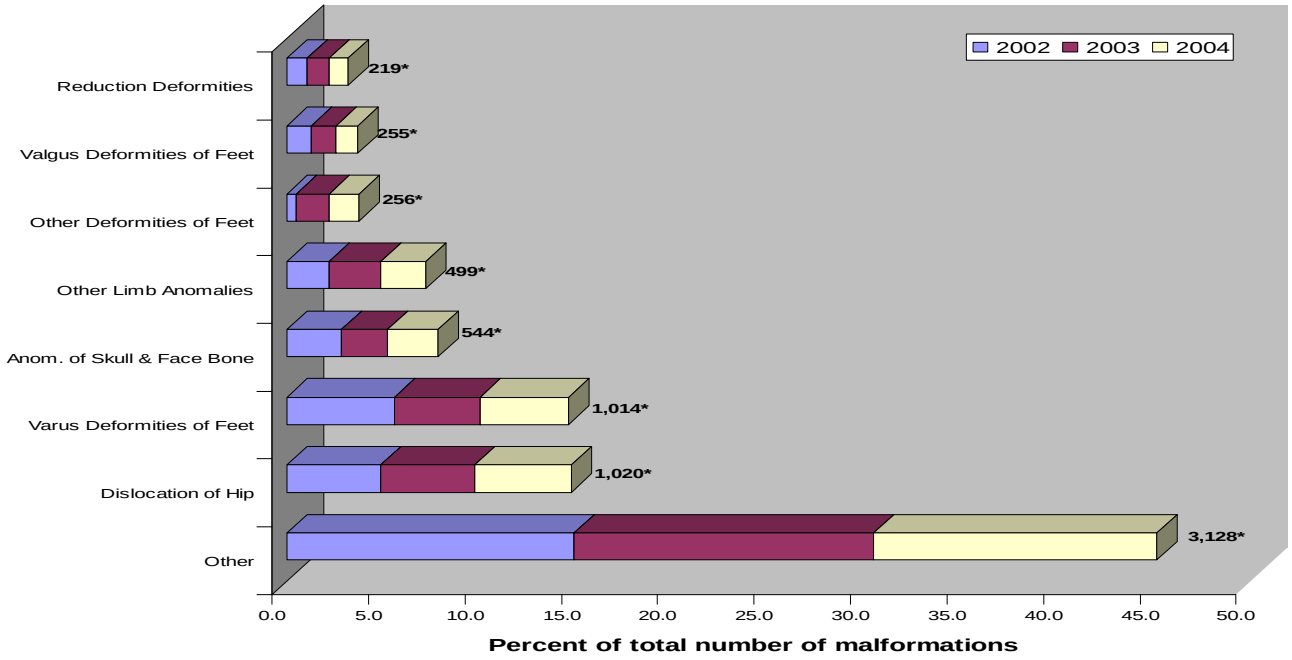
Major Malformations by Organ System
2002 - 2004 Births - New York State Residents
Genitourinary System Subset Category
(Number of Children = 9,666)
(* - Number of malformations in each organ system)



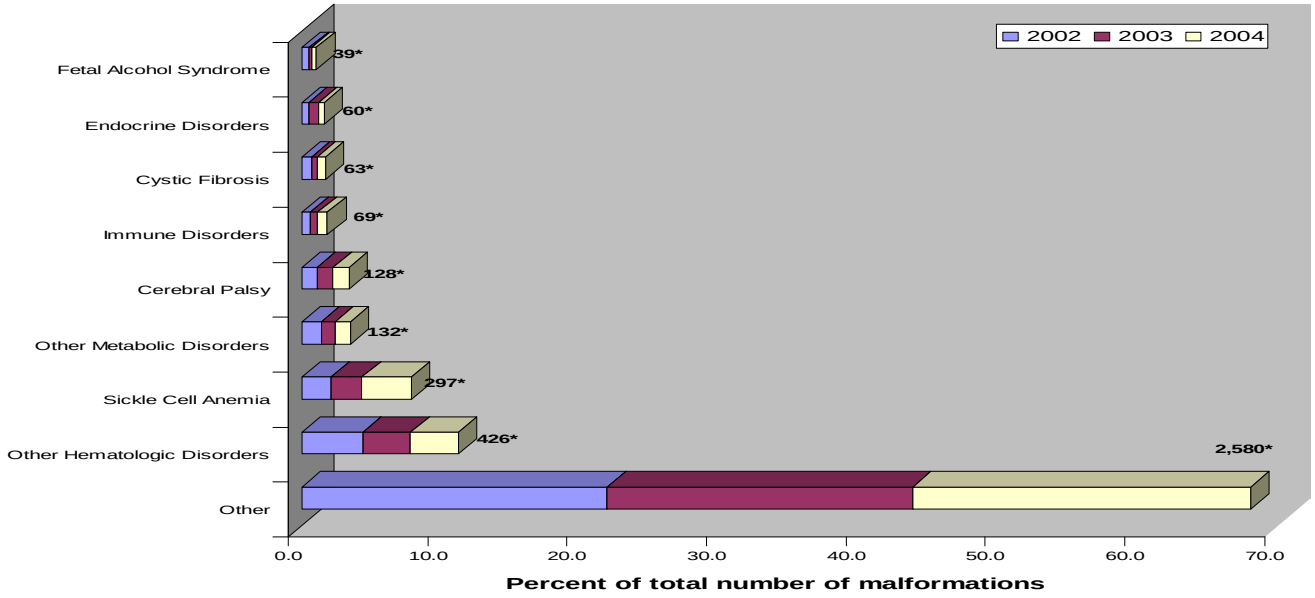
Major Malformations by Organ System
2002 - 2004 Births - New York State Residents
Integument System Subset Category
(Number of Children = 209)
(* - Number of malformations in each organ system)



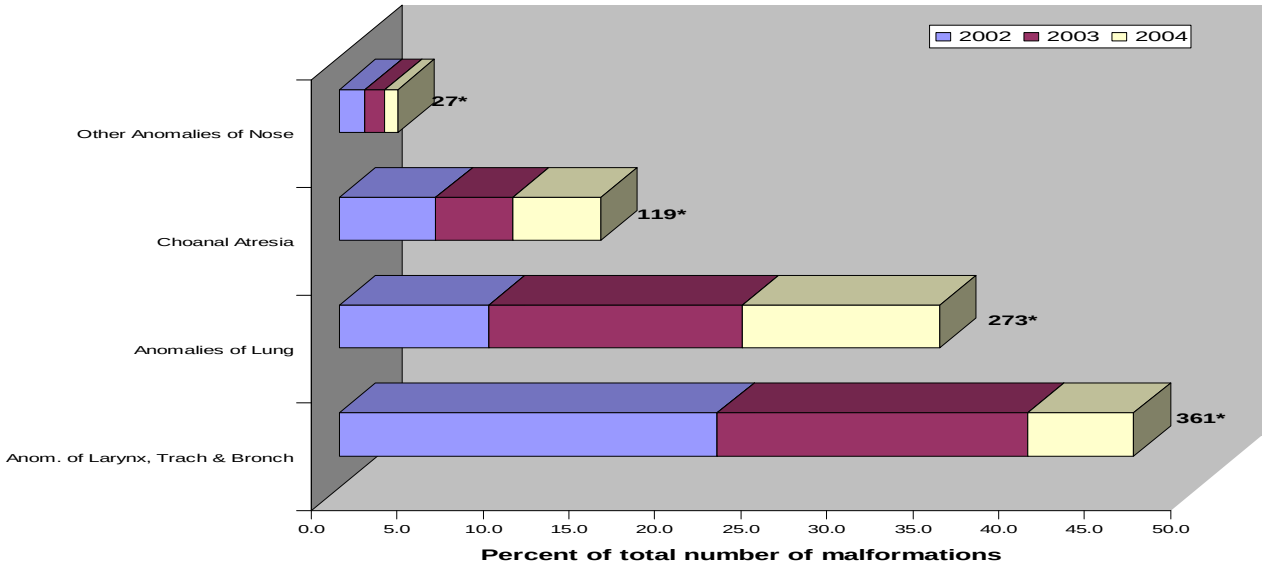
Major Malformations by Organ System
2002 - 2004 Births - New York State Residents
Musculoskeletal System Subset Category
(Number of Children = 6,804)
(* - Number of malformations in each organ system)



Major Malformations by Organ System
2002 - 2004 Births - New York State Residents
All Others Subset Category
(Number of Children = 3,789)
(* - Number of malformations in each organ system)



Major Malformations by Organ System
2002 - 2004 Births - New York State Residents
Respiratory System Subset Category
(Number of Children = 777)
(* - Number of malformations in each organ system)



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

Introduction to Tables

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

Fluctuations in specific malformation prevalence should be interpreted with caution, especially differences in the "other" race category since the numbers in this group are small. In addition, several malformations were added in 1992 as a result of the change to the BPA coding system. Previously, these could not be distinguished using the ICD-9 codes. However, since ICD-9 codes are more familiar to most vendors, the ICD-9 code is given on the table with the named malformation. See Appendix 3 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

2002 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	55	2.2	2.1	2.3	0.9	1.5	4.7	1.7	2.3
270.1	Phenylketonuria	3	0.1	0.2	0.0	0.0	0.2	0.0	0.2	0.0
277.0	Cystic fibrosis	26	1.0	0.9	1.1	0.8	1.5	0.4	0.7	0.0
282.6	Sickle-cell anemia	79	3.2	3.0	3.4	0.9	0.2	14.9	1.7	0.0
740.0	Anencephalus	8	0.3	0.3	0.3	1.0	0.2	0.9	0.2	0.0
741.0	Spina bifida with hydrocephalus	31	1.2	1.2	1.2	1.0	1.1	1.1	1.8	0.9
741.9	Spina bifida without hydrocephalus	19	0.8	0.7	0.8	0.9	0.8	0.4	1.1	0.5
742.0	Encephalocele	14	0.6	0.6	0.5	1.3	0.5	0.4	0.9	0.0
742.1	Microcephalus	124	4.9	4.4	5.6	0.8	1.9	10.7	8.3	2.8
742.2	Agyria & lissencephaly	4	0.2	0.2	0.1	2.9	0.2	0.4	0.0	0.0
742.2	Anomalies of corpus callosum	39	1.6	1.9	1.1	1.7	1.5	1.8	1.7	1.4
742.2	Holoprosencephaly	17	0.7	0.5	0.8	0.7	0.7	0.7	0.7	0.5
742.3	Congenital hydrocephalus	178	7.1	8.5	5.6	1.5	5.2	11.1	8.8	5.5
742.4	Porencephaly	10	0.4	0.5	0.2	2.2	0.5	0.4	0.2	0.0
742.5	Congenital tethered cord	15	0.6	0.5	0.7	0.6	0.6	0.2	0.9	0.5
743.0	Anophthalmos	1	0.0	0.0	0.1	0.0	0.1	0.0	0.0	0.0
743.1	Microphthalmos	18	0.7	1.0	0.4	2.5	0.6	1.1	0.9	0.0
743.2	Glaucoma	14	0.6	0.7	0.4	1.7	0.5	0.2	1.3	0.0
743.3	Absence of lens	8	0.3	0.4	0.2	1.6	0.2	0.7	0.2	0.5
743.3	Congenital cataract	22	0.9	0.9	0.9	1.0	0.6	1.3	1.1	0.9
743.45	Aniridia	1	0.0	0.0	0.1	0.0	0.0	0.0	0.2	0.0
743.46	Coloboma of iris	2	0.1	0.2	0.0	0.0	0.0	0.0	0.4	0.0
744.0	Anotia/microtia	15	0.6	0.5	0.7	0.8	0.6	0.2	0.6	1.4
745.0	Common truncus	15	0.6	0.2	1.0	0.2	0.4	0.9	0.9	0.5
745.1	Transposition of great vessels	124	4.9	5.8	4.1	1.4	4.8	4.4	5.7	5.1
745.2	Tetralogy of Fallot	104	4.1	4.1	4.2	1.0	4.0	4.2	3.1	7.4
745.3	Common ventricle	11	0.4	0.5	0.3	1.7	0.4	0.0	1.1	0.0
745.4	Ventricular septal defect	843	33.6	32.3	35.0	0.9	35.3	35.4	30.0	28.6
745.5	Ostium secundum type atrial septal def.	1,347	53.7	54.9	52.5	1.0	48.5	91.9	39.1	42.4
745.6	Endocardial cushion defects	84	3.3	3.0	3.8	0.8	2.8	5.1	3.3	2.8
746.0	Atresia/stenosis of pulmonary valve	210	8.4	8.3	8.5	1.0	7.9	11.1	8.1	6.0
746.1	Tricuspid Atresia/stenosis/hypoplasia	32	1.3	1.7	0.8	2.1	1.2	2.4	0.7	0.5
746.2	Ebstein's anomaly	18	0.7	0.7	0.7	1.0	0.6	0.7	0.7	1.4
746.3	Congenital stenosis of aortic valve	26	1.0	1.2	0.8	1.5	1.2	0.9	0.7	0.9
746.7	Hypoplastic left heart syndrome	55	2.2	2.5	1.9	1.3	1.5	2.9	3.7	1.4
746.85	Anomalies of coronary artery	9	0.4	0.4	0.3	1.2	0.2	0.4	0.6	0.9
747.0	Patent ductus arteriosis	488	19.5	19.9	19.0	1.1	14.8	35.6	16.2	22.1
747.10	Coarctation of aorta	119	4.7	5.1	4.3	1.2	5.2	5.8	3.9	2.3
747.41	Total anomalous pulmonary venus connect.	30	1.2	1.3	1.1	1.2	1.0	1.1	1.8	0.9

2002 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	Hispanic	Other/ Unknown Race
748.0	Choanal Atresia	44	1.8	1.5	2.0	0.7	1.8	2.0	1.1	2.3
748.5	Agenesis/hypoplasia of lung	54	2.2	2.6	1.6	1.6	2.5	1.8	2.4	0.0
749.0	Cleft palate	148	5.9	5.4	6.5	0.8	6.8	4.0	5.0	6.9
749.1	Cleft lip	52	2.1	2.3	1.8	1.3	2.5	0.9	2.4	1.4
749.2	Cleft palate & lip	131	5.2	7.0	3.4	2.1	4.9	4.9	5.3	7.4
750.3	Tracheoesophageal fistula etc.	56	2.2	2.4	2.0	1.2	2.6	2.0	2.0	0.9
750.5	Congenital hypertrophic pyloric stenosis	372	14.8	23.8	5.4	4.4	16.7	7.1	18.4	10.6
751.1	Atresia and stenosis of small intestine	100	4.0	3.7	4.3	0.9	3.6	5.1	4.4	2.8
751.2	Atresia and stenosis of rectum or anus	105	4.2	4.2	4.2	1.0	4.8	2.7	4.4	3.2
751.3	Hirschsprungs disease	72	2.9	4.3	1.4	3.1	2.9	3.3	2.8	1.8
751.4	Anomalies of intestinal fixation	42	1.7	1.9	1.4	1.4	1.9	2.7	0.7	0.5
751.61	Biliary Atresia	26	1.0	1.1	1.0	1.1	0.8	0.9	0.9	2.8
752.6	Epispadias	22	0.9	1.6	0.1	20.0	1.0	0.9	0.7	0.5
752.6	Hypospadias	881	35.1	68.1	0.5	138.9	44.4	32.3	20.1	23.1
753.0	Renal agenesis and dysgenesis	81	3.2	3.7	2.8	1.3	3.2	3.3	3.3	2.8
753.1	Cystic kidney disease	147	5.9	6.2	5.5	1.1	5.2	7.3	6.4	5.1
753.2	Obstructive defect renal pelvis & ureter	801	31.9	44.2	19.0	2.3	33.4	26.2	34.5	28.6
753.5	Extrophy of urinary bladder	5	0.2	0.2	0.2	1.4	0.2	0.0	0.0	0.9
753.6	Atresia & stenosis of urethra & bladder	12	0.5	0.9	0.0	0.0	0.8	0.2	0.2	0.0
754.3	Congenital dislocation of hip	251	10.0	4.0	16.3	0.2	11.9	4.2	11.2	7.4
754.51	Talipes equinovarus	298	11.9	14.5	9.2	1.6	10.8	13.6	12.7	12.9
755.2	Reduction deformities of upper limb	44	1.8	1.5	2.0	0.7	2.0	1.6	1.7	0.9
755.3	Reduction deformities of lower limb	27	1.1	1.5	0.7	2.3	1.2	1.1	0.7	0.9
755.8	Arthrogryposis multiplex congenita	14	0.6	0.5	0.6	1.0	0.6	0.9	0.4	0.0
756.0	Craniosynostosis	92	3.7	4.3	3.0	1.4	4.5	3.1	2.6	2.8
756.0	Goldenhar syndrome	9	0.4	0.3	0.4	0.8	0.5	0.2	0.2	0.5
756.4	Chonodrodystrophy	22	0.9	0.9	0.8	1.1	0.5	2.0	1.1	0.0
756.51	Osteogenesis imperfecta	6	0.2	0.3	0.2	1.9	0.3	0.4	0.0	0.0
756.6	Diaphragmatic hernia	41	1.6	1.2	2.0	0.6	1.8	1.3	1.5	1.8
756.7	Gastroschisis	50	2.0	2.0	2.0	1.0	2.1	1.6	2.6	0.9
756.7	Omphalocele	35	1.4	1.7	1.1	1.6	1.5	1.1	1.7	0.9
756.7	Prune belly	5	0.2	0.4	0.0	0.0	0.0	0.7	0.4	0.0
758.0	Down syndrome	290	11.6	12.2	10.9	1.1	11.8	14.2	10.1	8.3
758.1	Patau syndrome	19	0.8	0.9	0.6	1.6	0.6	1.1	0.9	0.5
758.2	Edwards syndrome	29	1.2	0.7	1.6	0.4	1.5	1.6	0.6	0.0
758.6	Gonadal dysgenesis	12	0.5	0.2	0.8	0.2	0.5	0.2	0.6	0.5
758.7	Klinefelter syndrome	17	0.7	1.3	0.0	0.0	0.6	0.2	1.3	0.5
759.3	Situs inversus	15	0.6	0.9	0.3	2.6	0.5	0.9	0.6	0.5
760.71	Fetal alcohol syndrome	20	0.8	0.8	0.8	1.0	0.6	1.8	0.6	0.5
771.0	Congenital rubella	1	0.0	0.1	0.0	0.0	0.1	0.0	0.0	0.0
771.1	Congenital cytomegalovirus infection	16	0.6	0.5	0.8	0.6	0.5	1.6	0.4	0.5
771.2	Other congenital infections	32	1.3	1.9	0.7	2.9	0.8	1.8	1.8	1.4

Section III - Table 2
Children with Selected Malformations
Prevalence per 10,000 Live Births by Sex and Race/Ethnicity

2003 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	87	3.4	4.2	2.7	1.6	2.5	5.9	2.9	5.3
270.1	Phenylketonuria	3	0.1	0.2	0.1	1.9	0.2	0.0	0.2	0.0
277.0	Cystic fibrosis	16	0.6	0.8	0.4	2.1	1.0	0.2	0.2	0.4
282.6	Sickle-cell anemia	84	3.3	3.3	3.3	1.0	0.1	16.7	1.6	0.4
740.0	Anencephalus	11	0.4	0.5	0.4	1.1	0.5	0.5	0.2	0.4
741.0	Spina bifida with hydrocephalus	29	1.1	0.5	1.8	0.3	1.1	1.1	1.3	1.3
741.9	Spina bifida without hydrocephalus	21	0.8	0.5	1.2	0.4	0.6	0.7	1.6	0.4
742.0	Encephalocele	16	0.6	0.6	0.6	1.0	0.8	0.9	0.4	0.0
742.1	Microcephalus	118	4.7	4.3	5.0	0.9	3.6	7.5	5.7	2.6
742.2	Agyria & lissencephaly	7	0.3	0.3	0.2	1.3	0.3	0.0	0.4	0.4
742.2	Anomalies of corpus callosum	41	1.6	1.7	1.5	1.1	1.4	3.0	0.7	2.2
742.2	Holoprosencephaly	10	0.4	0.2	0.6	0.4	0.2	0.2	1.1	0.0
742.3	Congenital hydrocephalus	189	7.5	8.0	6.9	1.2	5.8	12.1	7.9	7.5
742.4	Porencephaly	11	0.4	0.5	0.4	1.1	0.3	0.7	0.7	0.0
742.5	Congenital tethered cord	24	0.9	0.9	1.0	1.0	1.3	0.0	0.9	0.9
743.0	Anophthalmos	2	0.1	0.1	0.1	1.0	0.1	0.0	0.2	0.0
743.1	Microphthalmos	9	0.4	0.3	0.4	0.8	0.3	0.0	0.9	0.0
743.2	Glaucoma	6	0.2	0.4	0.1	4.8	0.4	0.2	0.0	0.0
743.3	Absence of lens	11	0.4	0.4	0.5	0.8	0.5	0.2	0.4	0.9
743.3	Congenital cataract	22	0.9	0.7	1.1	0.7	1.1	0.7	0.5	0.9
743.46	Coloboma of iris	5	0.2	0.3	0.1	3.8	0.3	0.2	0.0	0.0
744.0	Anotia/microtia	15	0.6	0.5	0.6	0.8	0.5	0.5	1.1	0.0
745.0	Common truncus	16	0.6	0.6	0.6	1.0	0.4	1.4	0.7	0.4
745.1	Transposition of great vessels	111	4.4	5.1	3.7	1.4	4.8	3.7	4.0	4.4
745.2	Tetralogy of Fallot	123	4.9	5.6	4.1	1.3	4.8	4.6	5.1	5.3
745.3	Common ventricle	17	0.7	0.6	0.7	0.8	0.5	1.1	0.7	0.9
745.4	Ventricular septal defect	917	36.2	32.2	40.5	0.8	36.9	34.3	36.6	35.5
745.5	Ostium secundum type atrial septal def.	1,036	40.9	40.2	41.7	1.0	33.9	59.2	43.1	41.2
745.6	Endocardial cushion defects	66	2.6	2.8	2.4	1.1	3.0	2.5	2.2	1.3
746.0	Atresia/stenosis of pulmonary valve	254	10.0	9.9	10.1	1.0	8.8	13.7	9.3	11.8
746.1	Tricuspid Atresia/stenosis/hypoplasia	28	1.1	1.2	1.1	1.1	0.7	2.5	0.9	1.3
746.2	Ebstein’s anomaly	13	0.5	0.5	0.6	0.8	0.5	0.0	0.5	1.8
746.3	Congenital stenosis of aortic valve	34	1.3	1.9	0.7	2.6	1.8	0.9	0.7	0.9
746.7	Hypoplastic left heart syndrome	70	2.8	3.0	2.5	1.2	3.0	3.0	2.6	1.8
746.85	Anomalies of coronary artery	10	0.4	0.3	0.5	0.6	0.5	0.5	0.2	0.4
747.0	Patent ductus arteriosis	722	28.5	27.3	29.9	0.9	26.2	48.0	19.9	25.4
747.10	Coarctation of aorta	121	4.8	4.9	4.6	1.1	5.2	5.5	3.8	3.5
747.41	Total anomalous pulmonary venus connect.	18	0.7	0.6	0.8	0.8	0.5	0.7	1.5	0.0
748.0	Choanal Atresia	35	1.4	1.5	1.2	1.3	1.7	0.9	1.3	0.4

2003 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	Hispanic	Other/ Unknown Race
748.5	Agenesis/hypoplasia of lung	74	2.9	3.0	2.8	1.1	2.9	4.1	2.9	0.9
749.0	Cleft palate	133	5.3	5.2	5.3	1.0	5.2	4.3	5.5	7.0
749.1	Cleft lip	63	2.5	2.8	2.2	1.3	3.7	1.1	1.5	0.4
749.2	Cleft palate & lip	120	4.7	5.6	3.9	1.4	4.7	4.1	5.7	3.9
750.3	Tracheoesophageal fistula etc.	60	2.4	2.6	2.1	1.2	3.1	1.8	1.5	1.3
750.5	Congenital hypertrophic pyloric stenosis	362	14.3	22.6	5.6	4.0	15.7	8.2	18.8	7.0
751.1	Atresia and stenosis of small intestine	103	4.1	4.5	3.6	1.3	3.7	5.7	3.8	3.5
751.2	Atresia and stenosis of rectum or anus	117	4.6	4.5	4.8	0.9	4.3	4.1	5.9	4.8
751.3	Hirschsprungs disease	52	2.1	3.1	1.0	3.2	2.4	2.3	1.3	1.8
751.4	Anomalies of intestinal fixation	32	1.3	1.6	0.9	1.8	1.7	0.7	0.5	1.3
751.61	Biliary Atresia	24	0.9	1.2	0.7	1.6	0.8	0.7	1.1	1.8
752.6	Epispadias	23	0.9	1.8	0.0	0.0	1.2	0.9	0.4	0.4
752.6	Hypospadias	871	34.4	67.0	0.2	413.0	40.5	35.0	24.5	22.4
753.0	Renal agenesis and dysgenesis	81	3.2	4.1	2.3	1.8	4.2	2.7	2.2	0.9
753.1	Cystic kidney disease	134	5.3	5.5	5.1	1.1	5.2	5.5	6.2	3.1
753.2	Obstructive defect renal pelvis & ureter	789	31.2	41.6	20.3	2.0	33.9	23.8	29.8	32.9
753.5	Extrophy of urinary bladder	5	0.2	0.2	0.2	0.6	0.2	0.2	0.2	0.0
753.6	Atresia & stenosis of urethra & bladder	18	0.7	1.3	0.1	16.2	0.6	1.4	0.7	0.0
754.3	Congenital dislocation of hip	254	10.0	5.6	14.7	0.4	11.1	5.9	10.4	11.0
754.51	Talipes equinovarus	207	8.2	9.9	6.4	1.5	8.4	6.6	9.9	5.7
755.2	Reduction deformities of upper limb	54	2.1	2.4	1.9	1.3	2.7	1.6	1.5	1.3
755.3	Reduction deformities of lower limb	25	1.0	1.3	0.6	2.0	1.1	1.1	0.9	0.4
755.8	Arthrogryposis multiplex congenita	11	0.4	0.2	0.6	0.4	0.5	0.2	0.7	0.0
756.0	Craniosynostosis	98	3.9	4.2	3.6	1.2	4.6	2.7	3.5	2.6
756.0	Goldenhar syndrome	5	0.2	0.3	0.1	3.8	0.2	0.2	0.4	0.0
756.4	Chonodrodystrophy	30	1.2	1.0	1.4	0.7	1.0	2.3	0.9	0.9
756.51	Osteogenesis imperfecta	9	0.4	0.2	0.5	0.5	0.4	0.5	0.4	0.0
756.6	Diaphragmatic hernia	59	2.3	2.8	1.9	1.5	2.7	2.1	2.6	0.4
756.7	Gastroschisis	44	1.7	1.9	1.6	1.1	1.5	2.3	2.0	1.3
756.7	Omphalocele	30	1.2	1.3	1.1	1.2	0.8	1.8	1.5	1.3
756.7	Prune belly	1	0.0	0.0	0.1	0.0	0.1	0.0	0.0	0.0
758.0	Down syndrome	319	12.6	12.9	12.3	1.0	13.1	12.3	11.5	12.7
758.1	Patau syndrome	20	0.8	1.0	0.6	1.8	0.6	1.6	0.5	0.9
758.2	Edwards syndrome	32	1.3	1.2	1.3	1.0	0.8	2.5	1.3	1.8
758.6	Gonadal dysgenesis	15	0.6	0.1	1.1	0.1	0.5	0.9	0.5	0.4
758.7	Klinefelter syndrome	9	0.4	0.7	0.0	0.0	0.3	0.2	0.7	0.0
759.3	Situs inversus	26	1.0	1.0	1.1	1.0	0.8	1.6	1.1	0.9
760.71	Fetal alcohol syndrome	6	0.2	0.3	0.2	1.9	0.1	0.7	0.4	0.0
771.1	Congenital cytomegalovirus infection	12	0.5	0.6	0.3	1.9	0.5	0.5	0.5	0.4
771.2	Other congenital infections	18	0.7	0.8	0.6	1.5	0.7	0.9	0.9	0.0

Section III - Table 3
Children with Selected Malformations
Prevalence per 10,000 Live Births by Sex and Race/Ethnicity

2004 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	101	4.1	4.3	3.9	1.1	3.3	7.0	3.4	5.1
270.1	Phenylketonuria	4	0.2	0.2	0.2	1.0	0.1	0.2	0.2	0.4
277.0	Cystic fibrosis	21	0.9	0.9	0.8	1.0	1.4	0.2	0.5	0.0
282.6	Sickle-cell anemia	134	5.4	5.2	5.7	0.9	0.4	27.8	2.1	0.8
740.0	Anencephalus	13	0.5	0.3	0.7	0.4	0.7	0.2	0.2	0.8
741.0	Spina bifida with hydrocephalus	26	1.1	0.9	1.2	0.7	0.8	1.2	1.4	1.3
741.9	Spina bifida without hydrocephalus	20	0.8	0.7	0.9	0.8	1.0	0.2	0.9	0.4
742.0	Encephalocele	10	0.4	0.5	0.3	1.4	0.4	0.2	0.7	0.0
742.1	Microcephalus	130	5.3	5.4	5.2	1.0	3.9	8.5	6.6	3.8
742.2	Agyria & lissencephaly	6	0.2	0.3	0.2	1.9	0.2	0.7	0.2	0.0
742.2	Anomalies of corpus callosum	50	2.0	2.4	1.7	1.4	1.8	2.7	2.1	2.1
742.2	Holoprosencephaly	11	0.4	0.6	0.3	1.7	0.3	0.7	0.7	0.0
742.3	Congenital hydrocephalus	211	8.6	9.3	7.8	1.2	6.5	14.0	10.1	6.4
742.4	Porencephaly	8	0.3	0.2	0.5	0.3	0.5	0.5	0.0	0.0
742.5	Congenital tethered cord	41	1.7	1.7	1.7	1.0	2.1	1.5	0.9	1.7
743.0	Anophthalmos	2	0.1	0.1	0.1	1.0	0.2	0.0	0.0	0.0
743.1	Microphthalmos	14	0.6	0.6	0.6	1.0	0.4	0.7	0.5	1.3
743.2	Glaucoma	18	0.7	0.8	0.7	1.2	0.6	1.5	0.4	1.3
743.3	Absence of lens	8	0.3	0.4	0.2	1.6	0.2	0.7	0.0	0.8
743.3	Congenital cataract	10	0.4	0.6	0.2	2.2	0.4	0.7	0.2	0.4
743.45	Aniridia	2	0.1	0.2	0.0	0.0	0.0	0.0	0.4	0.0
743.46	Coloboma of iris	2	0.1	0.1	0.1	1.0	0.2	0.0	0.0	0.0
744.0	Anotia/microtia	20	0.8	0.9	0.7	1.2	1.0	0.2	0.7	0.8
745.0	Common truncus	15	0.6	0.6	0.6	1.1	0.4	1.5	0.4	0.8
745.1	Transposition of great vessels	100	4.1	4.7	3.4	1.4	4.4	4.8	3.9	1.3
745.2	Tetralogy of Fallot	125	5.1	6.6	3.5	1.9	5.5	5.8	3.9	4.2
745.3	Common ventricle	26	1.1	1.0	1.2	0.8	1.0	1.2	1.4	0.4
745.4	Ventricular septal defect	1,039	42.2	39.8	44.8	0.9	42.2	43.1	42.9	39.4
745.5	Ostium secundum type atrial septal def.	1,124	45.7	45.9	45.4	1.0	32.6	86.1	41.8	52.9
745.6	Endocardial cushion defects	69	2.8	2.5	3.1	0.8	2.9	3.4	2.1	3.0
746.0	Atresia/stenosis of pulmonary valve	235	9.5	9.9	9.2	1.1	8.6	15.5	8.3	7.2
746.1	Tricuspid Atresia/stenosis/hypoplasia	21	0.9	0.7	1.0	0.7	0.7	1.7	0.7	0.4
746.2	Ebstein's anomaly	18	0.7	0.8	0.7	1.2	0.7	0.5	0.9	0.8
746.3	Congenital stenosis of aortic valve	31	1.3	1.9	0.6	3.3	1.5	1.0	1.1	0.8
746.7	Hypoplastic left heart syndrome	53	2.2	2.5	1.8	1.3	2.3	3.6	1.4	0.4
746.85	Anomalies of coronary artery	16	0.6	1.0	0.3	2.9	0.9	0.7	0.2	0.4
747.0	Patent ductus arteriosis	803	32.6	30.4	34.9	0.9	29.4	58.3	23.4	26.7
747.10	Coarctation of aorta	118	4.8	6.3	3.2	1.9	5.1	4.6	5.3	2.1
747.41	Total anomalous pulmonary venus connect.	26	1.1	1.2	0.9	1.3	0.6	1.2	2.1	0.4

2004 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	Hispanic	Other/ Unknown Race
748.0	Choanal Atresia	40	1.6	1.7	1.5	1.2	2.1	1.5	0.9	1.3
748.5	Agenesis/hypoplasia of lung	58	2.4	2.9	1.8	1.6	2.7	3.6	1.2	0.8
749.0	Cleft palate	133	5.4	4.8	6.0	0.8	5.5	4.4	5.1	7.2
749.1	Cleft lip	50	2.0	2.4	1.7	1.4	2.6	1.7	1.4	1.3
749.2	Cleft palate & lip	105	4.3	5.1	3.4	1.5	4.7	1.5	5.7	3.4
750.3	Tracheoesophageal fistula etc.	60	2.4	2.9	1.9	1.5	2.8	2.7	1.6	2.1
750.5	Congenital hypertrophic pyloric stenosis	549	22.3	35.2	8.7	4.0	25.6	12.3	26.9	11.0
751.1	Atresia and stenosis of small intestine	114	4.6	4.3	5.0	0.9	4.4	4.1	5.5	4.7
751.2	Atresia and stenosis of rectum or anus	111	4.5	5.5	3.5	1.6	4.5	4.8	3.9	5.5
751.3	Hirschsprungs disease	65	2.6	3.9	1.3	2.9	2.6	2.7	2.7	2.5
751.4	Anomalies of intestinal fixation	38	1.5	1.7	1.3	1.3	1.8	2.4	0.2	2.1
751.61	Biliary Atresia	22	0.9	1.1	0.7	1.7	0.6	1.2	1.1	1.3
752.6	Epispadias	25	1.0	1.9	0.1	22.9	1.1	1.9	0.4	0.4
752.6	Hypospadias	873	35.5	69.1	0.2	276.7	44.6	33.1	22.2	22.9
753.0	Renal agenesis and dysgenesis	87	3.5	4.5	2.5	1.8	3.7	4.4	2.5	3.8
753.1	Cystic kidney disease	141	5.7	6.4	5.0	1.3	5.0	7.5	6.6	4.7
753.2	Obstructive defect renal pelvis & ureter	849	34.5	48.6	19.7	2.5	36.9	28.1	35.4	30.9
753.5	Extrophy of urinary bladder	3	0.1	0.2	0.1	1.9	0.1	0.0	0.4	0.0
753.6	Atresia & stenosis of urethra & bladder	13	0.5	1.0	0.0	0.0	0.6	1.0	0.2	0.0
754.3	Congenital dislocation of hip	228	9.3	5.9	12.8	0.5	10.3	5.3	10.1	8.5
754.51	Talipes equinovarus	192	7.8	8.6	7.0	1.2	8.7	10.6	5.5	3.4
755.2	Reduction deformities of upper limb	44	1.8	1.7	1.8	1.0	1.8	1.9	2.1	0.4
755.3	Reduction deformities of lower limb	24	1.0	1.2	0.7	1.6	1.0	1.5	0.9	0.0
755.8	Arthrogryposis multiplex congenita	10	0.4	0.2	0.7	0.2	0.1	1.0	0.7	0.4
756.0	Craniosynostosis	95	3.9	5.1	2.6	2.0	5.3	1.5	3.4	1.7
756.0	Goldenhar syndrome	5	0.2	0.2	0.2	1.4	0.3	0.0	0.2	0.0
756.4	Chonodrodystrophy	34	1.4	1.7	1.0	1.7	1.1	2.2	1.8	0.4
756.51	Osteogenesis imperfecta	10	0.4	0.6	0.2	2.2	0.2	0.7	0.7	0.0
756.6	Diaphragmatic hernia	63	2.6	3.3	1.8	1.8	3.0	2.2	1.9	2.5
756.7	Gastroschisis	49	2.0	2.2	1.7	1.3	2.1	1.2	2.7	1.3
756.7	Omphalocele	33	1.3	1.7	0.9	1.9	1.3	2.2	1.1	0.8
756.7	Prune belly	8	0.3	0.6	0.1	6.7	0.1	1.2	0.2	0.4
758.0	Down syndrome	309	12.6	13.4	11.6	1.2	12.6	13.3	13.5	8.9
758.1	Patau syndrome	22	0.9	1.0	0.7	1.4	0.8	1.2	0.7	1.3
758.2	Edwards syndrome	27	1.1	0.9	1.3	0.7	0.9	2.7	0.5	0.8
758.6	Gonadal dysgenesis	18	0.7	0.1	1.4	0.1	0.9	0.5	0.7	0.4
758.7	Klinefelter syndrome	17	0.7	1.3	0.1	15.3	0.7	0.2	1.1	0.4
759.3	Situs inversus	13	0.5	0.5	0.6	0.8	0.6	0.2	0.5	0.8
760.71	Fetal alcohol syndrome	13	0.5	0.6	0.5	1.1	0.2	2.4	0.0	0.0
771.1	Congenital cytomegalovirus infection	13	0.5	0.6	0.4	1.5	0.2	1.9	0.4	0.4
771.2	Other congenital infections	42	1.7	1.0	2.4	0.4	1.4	2.4	1.8	1.7

Section IV
Most Frequently Reported Selected Major Malformations by County

Introduction to Tables

Congenital Malformation 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, 2002-2004

County	Birth Year = 2002			Birth Year = 2003			Birth Year = 2004		
	Number of Children	Number of Live Births	Percent of Live Births	Number of Children	Number of Live Births	Percent of Live Births	Number of Children	Number of Live Births	Percent of Live Births
Albany	123	3,226	3.8	161	3,220	5.0	135	3,062	4.4
Allegany	23	541	4.3	17	552	3.1	22	527	4.2
Bronx	1,020	22,449	4.5	1,072	22,601	4.7	1,077	22,188	4.9
Broome	95	2,062	4.6	91	2,075	4.4	88	2,017	4.4
Cattaraugus	41	983	4.2	39	1,009	3.9	48	861	5.6
Cayuga	44	825	5.3	42	838	5.0	36	839	4.3
Chautauqua	80	1,501	5.3	79	1,468	5.4	78	1,306	6.0
Chemung	46	1,068	4.3	50	1,081	4.6	46	923	5.0
Chenango	30	551	5.4	25	525	4.8	21	524	4.0
Clinton	31	783	4.0	15	808	1.9	18	721	2.5
Columbia	24	598	4.0	11	641	1.7	24	532	4.5
Cortland	15	560	2.7	18	528	3.4	20	506	4.0
Delaware	16	417	3.8	23	443	5.2	15	434	3.5
Dutchess	138	3,210	4.3	122	3,277	3.7	124	2,969	4.2
Erie	534	10,667	5.0	520	10,510	4.9	614	10,192	6.0
Essex	9	331	2.7	7	352	2.0	4	309	1.3
Franklin	12	491	2.4	9	438	2.1	16	508	3.1
Fulton	24	592	4.1	29	582	5.0	40	549	7.3
Genesee	25	645	3.9	29	625	4.6	36	625	5.8
Greene	12	454	2.6	24	441	5.4	14	444	3.2
Hamilton	1	35	2.9	1	48	2.1	2	38	5.3
Herkimer	20	682	2.9	29	702	4.1	37	698	5.3
Jefferson	71	1,545	4.6	96	1,704	5.6	121	1,499	8.1
Kings	2,036	39,386	5.2	2,003	39,521	5.1	2,190	39,702	5.5
Lewis	17	306	5.6	11	320	3.4	16	284	5.6
Livingston	31	665	4.7	31	627	4.9	30	634	4.7
Madison	37	710	5.2	39	740	5.3	24	702	3.4
Monroe	372	8,883	4.2	349	8,924	3.9	362	8,528	4.2
Montgomery	31	572	5.4	27	565	4.8	31	611	5.1
Nassau	811	16,335	5.0	787	16,015	4.9	817	15,604	5.2
New York	748	19,785	3.8	788	20,622	3.8	785	20,483	3.8
Niagara	123	2,405	5.1	108	2,408	4.5	110	2,317	4.7
Oneida	92	2,488	3.7	131	2,611	5.0	123	2,576	4.8
Onondaga	283	5,626	5.0	257	5,613	4.6	289	5,592	5.2
Ontario	61	1,142	5.3	52	1,162	4.5	45	1,071	4.2
Orange	166	5,040	3.3	187	5,133	3.6	234	5,091	4.6
Orleans	22	484	4.5	18	461	3.9	13	454	2.9
Oswego	60	1,357	4.4	74	1,391	5.3	66	1,355	4.9
Otsego	21	572	3.7	20	537	3.7	20	545	3.7
Putnam	33	1,195	2.8	54	1,165	4.6	52	1,082	4.8
Queens	1,249	30,497	4.1	1,142	30,787	3.7	1,281	30,345	4.2
Rensselear	76	1,670	4.6	60	1,767	3.4	85	1,647	5.2
Richmond	213	5,819	3.7	233	5,937	3.9	225	5,839	3.9
Rockland	161	4,532	3.6	178	4,492	4.0	189	4,228	4.5
Saratoga	98	2,370	4.1	100	2,470	4.0	105	2,404	4.4
Schenectady	48	1,739	2.8	80	1,841	4.3	100	1,833	5.5

Section IV – Table 1 (continued)
Children with Major Congenital Malformations & Percent of Live Births
by County and Birth Year, 2002-2004

	Birth Year = 2002			Birth Year = 2003			Birth Year = 2004		
County	Number of Children	Number of Live Births	Percent of Live Births	Number of Children	Number of Live Births	Percent of Live Births	Number of Children	Number of Live Births	Percent of Live Births
Schoharie	12	307	3.9	8	293	2.7	16	289	5.5
Schuyler	6	199	3.0	6	186	3.2	8	185	4.3
Seneca	10	369	2.7	15	375	4.0	12	336	3.6
St Lawrence	69	1,215	5.7	70	1,209	5.8	52	1,209	4.3
Steuben	55	1,141	4.8	47	1,153	4.1	60	1,070	5.6
Suffolk	902	19,853	4.5	824	19,986	4.1	861	19,608	4.4
Sullivan	33	788	4.2	32	911	3.5	37	811	4.6
Tioga	21	605	3.5	19	584	3.3	18	365	4.9
Tompkins	26	831	3.1	36	977	3.7	43	905	4.8
Ulster	57	1,793	3.2	55	1,826	3.0	75	1,835	4.1
Warren	33	662	5.0	25	639	3.9	32	611	5.2
Washington	15	603	2.5	20	622	3.2	29	545	5.3
Wayne	47	1,099	4.3	48	1,146	4.2	42	1,118	3.8
Westchester	471	12,807	3.7	559	12,789	4.4	527	11,417	4.6
Wyoming	20	446	4.5	22	424	5.2	29	429	6.8
Yates	8	281	2.8	12	300	4.0	9	238	3.8

Section IV - Table 2
Most Frequently Reported Major Malformations By County
Birth Years: 2002-2004

County	ICD-9 Code	Description	Number
ALBANY	745.4	Ventricular septal defect	50
	752.5	Undescended testicle	46
	752.6	Hypospadias & epispadias	39
	753.2	Obstructive defects of renal pelvis & ureter	36
	745.5	Ostium secundum atrial septal defect	35
	747.0	Patent ductus arteriosus	29
	746.8	Other specified anomalies of heart	18
	755.0	Polydactyly	17
	758.0	Down syndrome	16
	754.5	Varus deformities of feet	15
ALLEGANY	752.6	Hypospadias & epispadias	10
	745.4	Ventricular septal defect	5
	753.2	Obstructive defects of renal pelvis & ureter	5
	747.3	Anomalies of pulmonary artery	4
	750.5	Congenital hypertrophic pyloric stenosis	4
	747.0	Patent ductus arteriosus	3
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	3
	755.6	Other anomalies of lower limb including pelvic girdle	3
	756.0	Anomalies of skull and face bones	3
	742.2	Reduction deformities of brain	2
	745.5	Ostium secundum atrial septal defect	2
	748.3	Other anomalies of larynx, trachea, & bronchus	2
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	2
	752.4	Anomalies of cervix, vagina & external female genitalia	2
	752.5	Undescended testicle	2
	755.0	Polydactyly	2
	756.7	Anomalies of abdominal wall	2
	758.0	Down syndrome	2
	759.8	Other specified anomalies	2
BRONX	755.0	Polydactyly	346
	745.5	Ostium secundum atrial septal defect	274
	752.5	Undescended testicle	262
	753.2	Obstructive defects of renal pelvis & ureter	221
	752.6	Hypospadias & epispadias	216
	745.4	Ventricular septal defect	212
	754.3	Congenital dislocation of hip	144
	747.0	Patent ductus arteriosus	139
	754.5	Varus deformities of feet	120
	750.5	Congenital hypertrophic pyloric stenosis	106
BROOME	745.4	Ventricular septal defect	40
	745.5	Ostium secundum atrial septal defect	33
	752.6	Hypospadias & epispadias	22
	746.0	Anomalies of pulmonary valve	19

Section IV - Table 2
Most Frequently Reported Major Malformations By County
Birth Years: 2002-2004

County	ICD-9 Code	Description	Number
BROOME	752.5	Undescended testicle	19
	747.3	Anomalies of pulmonary artery	14
	750.5	Congenital hypertrophic pyloric stenosis	13
	747.0	Patent ductus arteriosus	12
	753.2	Obstructive defects of renal pelvis & ureter	10
	755.0	Polydactyly	10
	756.0	Anomalies of skull and face bones	10
	758.0	Down syndrome	10
CATTARAUGUS	745.5	Ostium secundum atrial septal defect	13
	752.6	Hypospadias & epispadias	13
	746.8	Other specified anomalies of heart	12
	745.4	Ventricular septal defect	11
	747.0	Patent ductus arteriosus	8
	747.3	Anomalies of pulmonary artery	8
	750.5	Congenital hypertrophic pyloric stenosis	8
	748.3	Other anomalies of larynx, trachea, & bronchus	7
	754.5	Varus deformities of feet	6
	753.2	Obstructive defects of renal pelvis & ureter	5
CAYUGA	752.6	Hypospadias & epispadias	20
	752.5	Undescended testicle	16
	750.5	Congenital hypertrophic pyloric stenosis	8
	756.0	Anomalies of skull and face bones	7
	745.5	Ostium secundum atrial septal defect	5
	754.3	Congenital dislocation of hip	5
	758.0	Down syndrome	5
	745.4	Ventricular septal defect	4
	749.2	Cleft palate with cleft lip	4
	755.0	Polydactyly	4
	755.6	Other anomalies of lower limb including pelvic girdle	4
CHAUTAUQUA	745.5	Ostium secundum atrial septal defect	43
	752.6	Hypospadias & epispadias	27
	745.4	Ventricular septal defect	22
	747.3	Anomalies of pulmonary artery	21
	752.5	Undescended testicle	21
	747.0	Patent ductus arteriosus	18
	746.8	Other specified anomalies of heart	16
	754.5	Varus deformities of feet	12
	750.5	Congenital hypertrophic pyloric stenosis	10
	754.3	Congenital dislocation of hip	9
CHEMUNG	752.5	Undescended testicle	16
	745.4	Ventricular septal defect	14
	752.6	Hypospadias & epispadias	13

Section IV - Table 2
Most Frequently Reported Major Malformations By County
Birth Years: 2002-2004

County	ICD-9 Code	Description	Number
CHEMUNG	753.2	Obstructive defects of renal pelvis & ureter	12
	745.5	Ostium secundum atrial septal defect	8
	746.0	Anomalies of pulmonary valve	7
	750.5	Congenital hypertrophic pyloric stenosis	7
	747.0	Patent ductus arteriosus	6
	746.8	Other specified anomalies of heart	5
	273.8	Other disorders of plasma protein	4
	749.2	Cleft palate with cleft lip	4
	754.5	Varus deformities of feet	4
	755.0	Polydactyly	4
	758.0	Down syndrome	4
CHENANGO	752.5	Undescended testicle	9
	752.6	Hypospadias & epispadias	9
	753.2	Obstructive defects of renal pelvis & ureter	6
	745.5	Ostium secundum atrial septal defect	5
	753.1	Cystic kidney disease	4
	745.4	Ventricular septal defect	3
	746.0	Anomalies of pulmonary valve	3
	754.5	Varus deformities of feet	3
	755.0	Polydactyly	3
	756.0	Anomalies of skull and face bones	3
CLINTON	745.4	Ventricular septal defect	14
	752.6	Hypospadias & epispadias	13
	745.5	Ostium secundum atrial septal defect	12
	753.2	Obstructive defects of renal pelvis & ureter	5
	747.0	Patent ductus arteriosus	4
	752.5	Undescended testicle	4
	756.0	Anomalies of skull and face bones	3
	748.3	Other anomalies of larynx, trachea, & bronchus	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	754.7	Other deformities of feet	2
COLUMBIA	752.6	Hypospadias & epispadias	11
	752.5	Undescended testicle	5
	747.0	Patent ductus arteriosus	4
	753.2	Obstructive defects of renal pelvis & ureter	4
	745.2	Tetralogy of Fallot	2
	745.4	Ventricular septal defect	2
	745.5	Ostium secundum atrial septal defect	2
	746.4	Congenital insufficiency of aortic valve	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	753.1	Cystic kidney disease	2
	754.5	Varus deformities of feet	2
	754.6	Valgus deformities of feet	2

Section IV - Table 2
Most Frequently Reported Major Malformations By County
Birth Years: 2002-2004

County	ICD-9 Code	Description	Number
COLUMBIA	755.0	Polydactyly	2
	756.0	Anomalies of skull and face bones	2
	758.0	Down syndrome	2
CORTLAND	750.5	Congenital hypertrophic pyloric stenosis	9
	753.2	Obstructive defects of renal pelvis & ureter	6
	746.8	Other specified anomalies of heart	3
	749.2	Cleft palate with cleft lip	3
	752.5	Undescended testicle	3
	745.1	Transposition of great vessels	2
	745.4	Ventricular septal defect	2
	746.0	Anomalies of pulmonary valve	2
	747.0	Patent ductus arteriosus	2
	747.1	Coarctation of aorta	2
	752.8	Other specified anomalies of genital organs	2
	754.5	Varus deformities of feet	2
	756.7	Anomalies of abdominal wall	2
DELAWARE	745.5	Ostium secundum atrial septal defect	8
	752.5	Undescended testicle	4
	752.6	Hypospadias & epispadias	4
	753.2	Obstructive defects of renal pelvis & ureter	4
	746.8	Other specified anomalies of heart	3
	747.0	Patent ductus arteriosus	3
	747.3	Anomalies of pulmonary artery	3
	749.0	Cleft palate	3
	750.5	Congenital hypertrophic pyloric stenosis	3
	524.0	Major anomalies of jaw size	2
	745.4	Ventricular septal defect	2
	754.5	Varus deformities of feet	2
	756.0	Anomalies of skull and face bones	2
	756.7	Anomalies of abdominal wall	2
	758.0	Down syndrome	2
DUTCHESS	752.6	Hypospadias & epispadias	39
	745.4	Ventricular septal defect	32
	745.5	Ostium secundum atrial septal defect	28
	752.5	Undescended testicle	27
	753.2	Obstructive defects of renal pelvis & ureter	23
	755.0	Polydactyly	21
	754.3	Congenital dislocation of hip	18
	750.5	Congenital hypertrophic pyloric stenosis	17
	755.6	Other anomalies of lower limb including pelvic girdle	15
	747.0	Patent ductus arteriosus	14
	754.5	Varus deformities of feet	14

Section IV - Table 2
Most Frequently Reported Major Malformations By County
Birth Years: 2002-2004

County	ICD-9 Code	Description	Number
ERIE	745.5	Ostium secundum atrial septal defect	170
	747.3	Anomalies of pulmonary artery	159
	747.0	Patent ductus arteriosus	146
	745.4	Ventricular septal defect	145
	752.5	Undescended testicle	144
	746.8	Other specified anomalies of heart	126
	752.6	Hypospadias & epispadias	120
	755.0	Polydactyly	93
	750.5	Congenital hypertrophic pyloric stenosis	79
	753.2	Obstructive defects of renal pelvis & ureter	78
ESSEX	745.5	Ostium secundum atrial septal defect	3
	752.6	Hypospadias & epispadias	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	745.4	Ventricular septal defect	1
	746.1	Tricuspid atresia & stenosis	1
	746.6	Congenital mitral insufficiency	1
	746.8	Other specified anomalies of heart	1
	747.3	Anomalies of pulmonary artery	1
	749.0	Cleft palate	1
	750.2	Other specified anomalies, mouth and pharynx	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	751.1	Atresia & stenosis of small intestine	1
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	1
	753.3	Other specified anomalies of kidney	1
	754.6	Valgus deformities of feet	1
	756.0	Anomalies of skull and face bones	1
	758.3	Autosomal deletion syndromes	1
	759.3	Situs inversus	1
FRANKLIN	752.6	Hypospadias & epispadias	6
	752.5	Undescended testicle	4
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	2
	754.5	Varus deformities of feet	2
	755.0	Polydactyly	2
	243.	Congenital hypothyroidism	1
	524.0	Major anomalies of jaw size	1
	743.2	Buphthalmos	1
	743.3	Congenital cataract & lens anomalies	1
	745.2	Tetralogy of Fallot	1
	745.4	Ventricular septal defect	1
	745.5	Ostium secundum atrial septal defect	1
	746.0	Anomalies of pulmonary valve	1
	746.7	Hypoplastic left heart syndrome	1
	747.0	Patent ductus arteriosus	1
	749.0	Cleft palate	1

Section IV - Table 2
Most Frequently Reported Major Malformations By County
Birth Years: 2002-2004

County	ICD-9 Code	Description	Number
FRANKLIN	749.1	Cleft lip	1
	749.2	Cleft palate with cleft lip	1
	750.4	Other specified anomalies of esophagus	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	754.0	Deformities of skull, face, & jaw	1
	754.3	Congenital dislocation of hip	1
	754.7	Other deformities of feet	1
	755.1	Syndactyly	1
	756.6	Anomalies of diaphragm	1
FULTON	754.5	Varus deformities of feet	12
	752.6	Hypospadias & epispadias	9
	756.0	Anomalies of skull and face bones	8
	750.5	Congenital hypertrophic pyloric stenosis	7
	754.3	Congenital dislocation of hip	7
	746.8	Other specified anomalies of heart	6
	745.4	Ventricular septal defect	5
	745.5	Ostium secundum atrial septal defect	4
	747.3	Anomalies of pulmonary artery	4
	749.0	Cleft palate	4
GENESEE	752.6	Hypospadias & epispadias	10
	746.8	Other specified anomalies of heart	9
	747.0	Patent ductus arteriosus	9
	752.5	Undescended testicle	9
	745.4	Ventricular septal defect	7
	745.5	Ostium secundum atrial septal defect	5
	753.2	Obstructive defects of renal pelvis & ureter	5
	754.3	Congenital dislocation of hip	5
	750.5	Congenital hypertrophic pyloric stenosis	4
	745.1	Transposition of great vessels	3
	747.3	Anomalies of pulmonary artery	3
GREENE	753.2	Obstructive defects of renal pelvis & ureter	7
	745.4	Ventricular septal defect	6
	752.5	Undescended testicle	6
	745.5	Ostium secundum atrial septal defect	4
	746.8	Other specified anomalies of heart	4
	741.0	Spina bifida with hydrocephalus	3
	742.3	Congenital hydrocephalus	3
	747.1	Coarctation of aorta	3
	750.5	Congenital hypertrophic pyloric stenosis	3
	752.6	Hypospadias & epispadias	3
	755.0	Polydactyly	3

Section IV - Table 2
Most Frequently Reported Major Malformations By County
Birth Years: 2002-2004

County	ICD-9 Code	Description	Number
HAMILTON	746.6	Congenital mitral insufficiency	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	752.5	Undescended testicle	1
	754.5	Varus deformities of feet	1
HERKIMER	752.6	Hypospadias & epispadias	14
	752.5	Undescended testicle	13
	753.2	Obstructive defects of renal pelvis & ureter	7
	745.4	Ventricular septal defect	5
	754.3	Congenital dislocation of hip	5
	754.5	Varus deformities of feet	5
	745.5	Ostium secundum atrial septal defect	4
	745.1	Transposition of great vessels	3
	745.2	Tetralogy of Fallot	3
	746.4	Congenital insufficiency of aortic valve	3
	746.8	Other specified anomalies of heart	3
	747.0	Patent ductus arteriosus	3
	749.2	Cleft palate with cleft lip	3
	756.7	Anomalies of abdominal wall	3
	758.0	Down syndrome	3
JEFFERSON	745.4	Ventricular septal defect	33
	752.6	Hypospadias & epispadias	33
	752.5	Undescended testicle	31
	754.5	Varus deformities of feet	28
	750.5	Congenital hypertrophic pyloric stenosis	14
	753.2	Obstructive defects of renal pelvis & ureter	12
	755.0	Polydactyly	12
	745.5	Ostium secundum atrial septal defect	10
	749.0	Cleft palate	10
	747.0	Patent ductus arteriosus	7
	748.3	Other anomalies of larynx, trachea, & bronchus	7
	749.2	Cleft palate with cleft lip	7
	754.3	Congenital dislocation of hip	7
	756.0	Anomalies of skull and face bones	7
KINGS	745.5	Ostium secundum atrial septal defect	1214
	747.0	Patent ductus arteriosus	562
	745.4	Ventricular septal defect	500
	752.5	Undescended testicle	459
	755.0	Polydactyly	453
	752.6	Hypospadias & epispadias	415
	753.2	Obstructive defects of renal pelvis & ureter	356
	746.8	Other specified anomalies of heart	203
	758.0	Down syndrome	161
	750.5	Congenital hypertrophic pyloric stenosis	158

Section IV - Table 2
Most Frequently Reported Major Malformations By County
Birth Years: 2002-2004

County	ICD-9 Code	Description	Number
LEWIS	745.5	Ostium secundum atrial septal defect	6
	754.5	Varus deformities of feet	6
	756.0	Anomalies of skull and face bones	4
	343.9	Infantile cerebral palsy unspecified	3
	752.5	Undescended testicle	3
	752.6	Hypospadias & epispadias	3
	745.4	Ventricular septal defect	2
	746.0	Anomalies of pulmonary valve	2
	746.4	Congenital insufficiency of aortic valve	2
	747.1	Coarctation of aorta	2
	752.8	Other specified anomalies of genital organs	2
	754.3	Congenital dislocation of hip	2
LIVINGSTON	753.2	Obstructive defects of renal pelvis & ureter	17
	752.6	Hypospadias & epispadias	9
	752.5	Undescended testicle	8
	755.6	Other anomalies of lower limb including pelvic girdle	6
	754.3	Congenital dislocation of hip	5
	758.0	Down syndrome	5
	748.3	Other anomalies of larynx, trachea, & bronchus	4
	751.4	Anomalies of intestinal fixation	4
	747.0	Patent ductus arteriosus	3
	754.5	Varus deformities of feet	3
	756.0	Anomalies of skull and face bones	3
MADISON	745.4	Ventricular septal defect	10
	752.6	Hypospadias & epispadias	9
	752.5	Undescended testicle	8
	753.2	Obstructive defects of renal pelvis & ureter	8
	747.0	Patent ductus arteriosus	5
	750.5	Congenital hypertrophic pyloric stenosis	5
	745.5	Ostium secundum atrial septal defect	4
	748.3	Other anomalies of larynx, trachea, & bronchus	4
	749.2	Cleft palate with cleft lip	4
	747.3	Anomalies of pulmonary artery	3
	754.3	Congenital dislocation of hip	3
	755.0	Polydactyly	3
	755.6	Other anomalies of lower limb including pelvic girdle	3
MONROE	752.6	Hypospadias & epispadias	151
	753.2	Obstructive defects of renal pelvis & ureter	116
	752.5	Undescended testicle	92
	755.0	Polydactyly	87
	745.4	Ventricular septal defect	63
	745.5	Ostium secundum atrial septal defect	44
	750.5	Congenital hypertrophic pyloric stenosis	44

Section IV - Table 2
Most Frequently Reported Major Malformations By County
Birth Years: 2002-2004

County	ICD-9 Code	Description	Number
MONROE	758.0	Down syndrome	39
	754.5	Varus deformities of feet	29
	747.3	Anomalies of pulmonary artery	28
MONTGOMERY	752.6	Hypospadias & epispadias	14
	750.5	Congenital hypertrophic pyloric stenosis	7
	752.5	Undescended testicle	7
	749.0	Cleft palate	5
	745.4	Ventricular septal defect	4
	745.5	Ostium secundum atrial septal defect	4
	747.0	Patent ductus arteriosus	4
	753.2	Obstructive defects of renal pelvis & ureter	4
	754.5	Varus deformities of feet	4
	755.0	Polydactyly	4
NASSAU	752.6	Hypospadias & epispadias	313
	745.4	Ventricular septal defect	226
	752.5	Undescended testicle	222
	753.2	Obstructive defects of renal pelvis & ureter	216
	747.0	Patent ductus arteriosus	174
	745.5	Ostium secundum atrial septal defect	161
	752.8	Other specified anomalies of genital organs	100
	750.5	Congenital hypertrophic pyloric stenosis	95
	755.0	Polydactyly	73
	746.8	Other specified anomalies of heart	69
NEW YORK	745.5	Ostium secundum atrial septal defect	251
	752.6	Hypospadias & epispadias	227
	745.4	Ventricular septal defect	221
	752.5	Undescended testicle	179
	753.2	Obstructive defects of renal pelvis & ureter	173
	755.0	Polydactyly	146
	754.3	Congenital dislocation of hip	115
	747.0	Patent ductus arteriosus	105
	754.5	Varus deformities of feet	69
	750.5	Congenital hypertrophic pyloric stenosis	68
NIAGARA	745.5	Ostium secundum atrial septal defect	33
	752.6	Hypospadias & epispadias	33
	745.4	Ventricular septal defect	27
	750.5	Congenital hypertrophic pyloric stenosis	26
	746.8	Other specified anomalies of heart	24
	747.3	Anomalies of pulmonary artery	24
	752.5	Undescended testicle	23
	747.0	Patent ductus arteriosus	19
	755.0	Polydactyly	17

Section IV - Table 2
Most Frequently Reported Major Malformations By County
Birth Years: 2002-2004

County	ICD-9 Code	Description	Number
NIAGARA	754.5	Varus deformities of feet	14
ONEIDA	752.5	Undescended testicle	39
	752.6	Hypospadias & epispadias	29
	754.3	Congenital dislocation of hip	24
	750.5	Congenital hypertrophic pyloric stenosis	19
	745.4	Ventricular septal defect	17
	755.0	Polydactyly	16
	746.0	Anomalies of pulmonary valve	14
	755.6	Other anomalies of lower limb including pelvic girdle	14
	745.5	Ostium secundum atrial septal defect	13
	746.8	Other specified anomalies of heart	12
ONONDAGA	745.4	Ventricular septal defect	87
	752.6	Hypospadias & epispadias	74
	753.2	Obstructive defects of renal pelvis & ureter	71
	745.5	Ostium secundum atrial septal defect	65
	752.5	Undescended testicle	64
	747.0	Patent ductus arteriosus	40
	755.0	Polydactyly	40
	750.5	Congenital hypertrophic pyloric stenosis	36
	758.0	Down syndrome	31
	746.8	Other specified anomalies of heart	23
ONTARIO	752.6	Hypospadias & epispadias	31
	752.5	Undescended testicle	14
	753.2	Obstructive defects of renal pelvis & ureter	13
	745.4	Ventricular septal defect	12
	754.3	Congenital dislocation of hip	11
	750.5	Congenital hypertrophic pyloric stenosis	7
	754.5	Varus deformities of feet	6
	749.2	Cleft palate with cleft lip	5
	755.0	Polydactyly	5
	745.2	Tetralogy of Fallot	4
	749.0	Cleft palate	4
	752.8	Other specified anomalies of genital organs	4
	755.6	Other anomalies of lower limb including pelvic girdle	4
	756.0	Anomalies of skull and face bones	4
ORANGE	752.6	Hypospadias & epispadias	63
	752.5	Undescended testicle	56
	745.4	Ventricular septal defect	51
	753.2	Obstructive defects of renal pelvis & ureter	49
	745.5	Ostium secundum atrial septal defect	42
	747.0	Patent ductus arteriosus	32
	750.5	Congenital hypertrophic pyloric stenosis	28

Section IV - Table 2
Most Frequently Reported Major Malformations By County
Birth Years: 2002-2004

County	ICD-9 Code	Description	Number
ORANGE	754.5	Varus deformities of feet	24
	755.0	Polydactyly	23
	758.0	Down syndrome	18
ORLEANS	750.5	Congenital hypertrophic pyloric stenosis	5
	752.5	Undescended testicle	5
	753.2	Obstructive defects of renal pelvis & ureter	5
	745.4	Ventricular septal defect	4
	747.3	Anomalies of pulmonary artery	4
	756.0	Anomalies of skull and face bones	4
	742.2	Reduction deformities of brain	3
	745.5	Ostium secundum atrial septal defect	3
	752.6	Hypospadias & epispadias	3
	743.4	Coloboma & other anomalies of anterior segment	2
	746.8	Other specified anomalies of heart	2
	749.2	Cleft palate with cleft lip	2
	755.1	Syndactyly	2
	755.6	Other anomalies of lower limb including pelvic girdle	2
	757.3	Other specified anomalies of skin	2
OSWEGO	752.5	Undescended testicle	25
	745.4	Ventricular septal defect	24
	752.6	Hypospadias & epispadias	17
	745.5	Ostium secundum atrial septal defect	12
	753.2	Obstructive defects of renal pelvis & ureter	11
	750.5	Congenital hypertrophic pyloric stenosis	10
	747.0	Patent ductus arteriosus	9
	749.0	Cleft palate	9
	746.0	Anomalies of pulmonary valve	7
	748.3	Other anomalies of larynx, trachea, & bronchus	5
OTSEGO	752.6	Hypospadias & epispadias	11
	750.5	Congenital hypertrophic pyloric stenosis	6
	742.3	Congenital hydrocephalus	4
	754.5	Varus deformities of feet	4
	745.5	Ostium secundum atrial septal defect	3
	753.2	Obstructive defects of renal pelvis & ureter	3
	758.0	Down syndrome	3
	759.8	Other specified anomalies	3
	243.	Congenital hypothyroidism	2
	742.5	Other specified anomalies of spinal cord	2
	745.4	Ventricular septal defect	2
	747.0	Patent ductus arteriosus	2
	747.3	Anomalies of pulmonary artery	2
	752.5	Undescended testicle	2
	754.3	Congenital dislocation of hip	2

Section IV - Table 2
Most Frequently Reported Major Malformations By County
Birth Years: 2002-2004

County	ICD-9 Code	Description	Number
PUTNAM	752.6	Hypospadias & epispadias	18
	753.2	Obstructive defects of renal pelvis & ureter	16
	745.4	Ventricular septal defect	13
	745.5	Ostium secundum atrial septal defect	11
	750.5	Congenital hypertrophic pyloric stenosis	8
	747.0	Patent ductus arteriosus	7
	752.5	Undescended testicle	6
	747.1	Coarctation of aorta	4
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	4
	754.3	Congenital dislocation of hip	4
QUEENS	745.5	Ostium secundum atrial septal defect	408
	745.4	Ventricular septal defect	328
	752.5	Undescended testicle	308
	753.2	Obstructive defects of renal pelvis & ureter	300
	752.6	Hypospadias & epispadias	288
	755.0	Polydactyly	207
	747.0	Patent ductus arteriosus	204
	750.5	Congenital hypertrophic pyloric stenosis	133
	754.3	Congenital dislocation of hip	115
	747.3	Anomalies of pulmonary artery	113
RENSSELAER	752.6	Hypospadias & epispadias	37
	745.4	Ventricular septal defect	21
	752.5	Undescended testicle	20
	745.5	Ostium secundum atrial septal defect	15
	747.0	Patent ductus arteriosus	14
	753.2	Obstructive defects of renal pelvis & ureter	14
	750.5	Congenital hypertrophic pyloric stenosis	12
	747.3	Anomalies of pulmonary artery	7
	754.5	Varus deformities of feet	7
	746.0	Anomalies of pulmonary valve	6
	754.3	Congenital dislocation of hip	6
RICHMOND	745.5	Ostium secundum atrial septal defect	86
	752.6	Hypospadias & epispadias	73
	752.5	Undescended testicle	61
	745.4	Ventricular septal defect	56
	747.0	Patent ductus arteriosus	41
	755.0	Polydactyly	35
	753.2	Obstructive defects of renal pelvis & ureter	30
	758.0	Down syndrome	24
	746.0	Anomalies of pulmonary valve	20
	754.5	Varus deformities of feet	19
ROCKLAND	752.6	Hypospadias & epispadias	68

Section IV - Table 2
Most Frequently Reported Major Malformations By County
Birth Years: 2002-2004

County	ICD-9 Code	Description	Number
ROCKLAND	745.4	Ventricular septal defect	53
	752.5	Undescended testicle	53
	753.2	Obstructive defects of renal pelvis & ureter	41
	745.5	Ostium secundum atrial septal defect	38
	755.0	Polydactyly	35
	747.0	Patent ductus arteriosus	28
	750.5	Congenital hypertrophic pyloric stenosis	21
	758.0	Down syndrome	17
	746.8	Other specified anomalies of heart	16
SARATOGA	752.6	Hypospadias & epispadias	30
	752.5	Undescended testicle	24
	745.5	Ostium secundum atrial septal defect	23
	753.2	Obstructive defects of renal pelvis & ureter	23
	747.0	Patent ductus arteriosus	22
	745.4	Ventricular septal defect	19
	754.3	Congenital dislocation of hip	16
	754.5	Varus deformities of feet	12
	750.5	Congenital hypertrophic pyloric stenosis	11
	746.8	Other specified anomalies of heart	10
SCHENECTADY	752.5	Undescended testicle	25
	752.6	Hypospadias & epispadias	25
	754.5	Varus deformities of feet	14
	745.4	Ventricular septal defect	12
	750.5	Congenital hypertrophic pyloric stenosis	12
	747.0	Patent ductus arteriosus	11
	753.2	Obstructive defects of renal pelvis & ureter	11
	745.5	Ostium secundum atrial septal defect	10
	755.0	Polydactyly	9
	758.0	Down syndrome	8
SCHOHARIE	752.5	Undescended testicle	6
	752.6	Hypospadias & epispadias	5
	745.4	Ventricular septal defect	3
	747.0	Patent ductus arteriosus	3
	745.5	Ostium secundum atrial septal defect	2
	748.0	Choanal atresia	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	753.3	Other specified anomalies of kidney	2
	756.1	Anomalies of spine	2
	190.5	Malignant neoplasm of the retina	1
	228.0	Hemangioma, any site	1
	286.0	Congenital factor VIII disorder	1
	333.2	Myoclonus	1
	742.4	Other specified anomalies of brain	1

Section IV - Table 2
Most Frequently Reported Major Malformations By County
Birth Years: 2002-2004

County	ICD-9 Code	Description	Number
SCHOHARIE	742.5	Other specified anomalies of spinal cord	1
	745.3	Common ventricle	1
	745.6	Endocardial cushion defects	1
	746.4	Congenital insufficiency of aortic valve	1
	746.8	Other specified anomalies of heart	1
	747.1	Coarctation of aorta	1
	748.3	Other anomalies of larynx, trachea, & bronchus	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	751.1	Atresia & stenosis of small intestine	1
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	1
	752.4	Anomalies of cervix, vagina & external female genitalia	1
	753.0	Renal agenesis & dysgenesis	1
	754.5	Varus deformities of feet	1
	754.6	Valgus deformities of feet	1
	755.6	Other anomalies of lower limb including pelvic girdle	1
	756.7	Anomalies of abdominal wall	1
SCHUYLER	752.6	Hypospadias & epispadias	4
	753.2	Obstructive defects of renal pelvis & ureter	3
	745.4	Ventricular septal defect	2
	752.5	Undescended testicle	2
	243.	Congenital hypothyroidism	1
	273.8	Other disorders of plasma protein	1
	742.1	Microcephalus	1
	745.5	Ostium secundum atrial septal defect	1
	746.8	Other specified anomalies of heart	1
	751.1	Atresia & stenosis of small intestine	1
	754.5	Varus deformities of feet	1
	755.6	Other anomalies of lower limb including pelvic girdle	1
	756.1	Anomalies of spine	1
	756.7	Anomalies of abdominal wall	1
	758.0	Down syndrome	1
SENECA	752.6	Hypospadias & epispadias	6
	752.5	Undescended testicle	4
	754.3	Congenital dislocation of hip	4
	746.0	Anomalies of pulmonary valve	3
	753.2	Obstructive defects of renal pelvis & ureter	3
	745.4	Ventricular septal defect	2
	746.8	Other specified anomalies of heart	2
	747.0	Patent ductus arteriosus	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	753.1	Cystic kidney disease	2
	755.0	Polydactyly	2
	756.0	Anomalies of skull and face bones	2
	758.0	Down syndrome	2

Section IV - Table 2
Most Frequently Reported Major Malformations By County
Birth Years: 2002-2004

County	ICD-9 Code	Description	Number
ST LAWRENCE	754.5	Varus deformities of feet	36
	752.6	Hypospadias & epispadias	25
	753.2	Obstructive defects of renal pelvis & ureter	14
	754.6	Valgus deformities of feet	13
	745.4	Ventricular septal defect	11
	752.5	Undescended testicle	8
	755.0	Polydactyly	8
	750.5	Congenital hypertrophic pyloric stenosis	7
	754.3	Congenital dislocation of hip	6
	747.3	Anomalies of pulmonary artery	5
	752.8	Other specified anomalies of genital organs	5
STEUBEN	753.2	Obstructive defects of renal pelvis & ureter	16
	752.5	Undescended testicle	13
	752.6	Hypospadias & epispadias	13
	745.4	Ventricular septal defect	12
	754.5	Varus deformities of feet	11
	750.5	Congenital hypertrophic pyloric stenosis	9
	747.3	Anomalies of pulmonary artery	7
	745.5	Ostium secundum atrial septal defect	6
	754.3	Congenital dislocation of hip	6
	756.0	Anomalies of skull and face bones	6
SUFFOLK	752.6	Hypospadias & epispadias	349
	752.5	Undescended testicle	236
	745.5	Ostium secundum atrial septal defect	222
	745.4	Ventricular septal defect	216
	753.2	Obstructive defects of renal pelvis & ureter	191
	747.0	Patent ductus arteriosus	132
	747.3	Anomalies of pulmonary artery	112
	750.5	Congenital hypertrophic pyloric stenosis	103
	758.0	Down syndrome	88
	755.0	Polydactyly	86
SULLIVAN	745.5	Ostium secundum atrial septal defect	11
	752.6	Hypospadias & epispadias	11
	745.4	Ventricular septal defect	10
	750.5	Congenital hypertrophic pyloric stenosis	9
	753.2	Obstructive defects of renal pelvis & ureter	8
	758.0	Down syndrome	8
	747.0	Patent ductus arteriosus	7
	752.5	Undescended testicle	6
	754.3	Congenital dislocation of hip	6
	755.0	Polydactyly	4
TIOGA	745.4	Ventricular septal defect	7

Section IV - Table 2
Most Frequently Reported Major Malformations By County
Birth Years: 2002-2004

County	ICD-9 Code	Description	Number
TIOGA	746.0	Anomalies of pulmonary valve	5
	747.3	Anomalies of pulmonary artery	5
	752.5	Undescended testicle	5
	746.8	Other specified anomalies of heart	4
	752.6	Hypospadias & epispadias	4
	745.5	Ostium secundum atrial septal defect	3
	747.0	Patent ductus arteriosus	3
	749.1	Cleft lip	3
	754.3	Congenital dislocation of hip	3
TOMPKINS	745.4	Ventricular septal defect	16
	752.6	Hypospadias & epispadias	11
	745.5	Ostium secundum atrial septal defect	6
	747.0	Patent ductus arteriosus	6
	750.5	Congenital hypertrophic pyloric stenosis	5
	752.5	Undescended testicle	5
	753.2	Obstructive defects of renal pelvis & ureter	5
	755.0	Polydactyly	4
	755.6	Other anomalies of lower limb including pelvic girdle	4
	742.3	Congenital hydrocephalus	3
	746.4	Congenital insufficiency of aortic valve	3
	749.0	Cleft palate	3
	756.0	Anomalies of skull and face bones	3
	758.0	Down syndrome	3
ULSTER	752.6	Hypospadias & epispadias	27
	745.4	Ventricular septal defect	16
	750.5	Congenital hypertrophic pyloric stenosis	14
	752.5	Undescended testicle	14
	755.0	Polydactyly	12
	745.5	Ostium secundum atrial septal defect	11
	747.0	Patent ductus arteriosus	10
	753.2	Obstructive defects of renal pelvis & ureter	10
	754.3	Congenital dislocation of hip	7
	746.0	Anomalies of pulmonary valve	6
WARREN	753.2	Obstructive defects of renal pelvis & ureter	12
	752.5	Undescended testicle	10
	754.3	Congenital dislocation of hip	8
	745.5	Ostium secundum atrial septal defect	7
	747.0	Patent ductus arteriosus	6
	750.5	Congenital hypertrophic pyloric stenosis	6
	752.6	Hypospadias & epispadias	6
	755.0	Polydactyly	5
	758.0	Down syndrome	3
	742.2	Reduction deformities of brain	2

Section IV - Table 2
Most Frequently Reported Major Malformations By County
Birth Years: 2002-2004

County	ICD-9 Code	Description	Number
WARREN	746.8	Other specified anomalies of heart	2
	749.0	Cleft palate	2
	749.2	Cleft palate with cleft lip	2
	751.1	Atresia & stenosis of small intestine	2
	753.1	Cystic kidney disease	2
	754.5	Varus deformities of feet	2
	756.7	Anomalies of abdominal wall	2
WASHINGTON	752.5	Undescended testicle	9
	750.5	Congenital hypertrophic pyloric stenosis	8
	752.6	Hypospadias & epispadias	5
	753.2	Obstructive defects of renal pelvis & ureter	5
	745.4	Ventricular septal defect	4
	747.0	Patent ductus arteriosus	3
	754.3	Congenital dislocation of hip	3
	756.0	Anomalies of skull and face bones	3
	742.3	Congenital hydrocephalus	2
	745.5	Ostium secundum atrial septal defect	2
	745.6	Endocardial cushion defects	2
	747.1	Coarctation of aorta	2
	752.7	Indeterminate sex & pseudo-hermaphroditism	2
	758.0	Down syndrome	2
WAYNE	752.6	Hypospadias & epispadias	23
	752.5	Undescended testicle	15
	753.2	Obstructive defects of renal pelvis & ureter	14
	745.4	Ventricular septal defect	12
	745.5	Ostium secundum atrial septal defect	5
	750.5	Congenital hypertrophic pyloric stenosis	5
	753.3	Other specified anomalies of kidney	5
	756.7	Anomalies of abdominal wall	5
	758.0	Down syndrome	5
	524.0	Major anomalies of jaw size	4
	755.6	Other anomalies of lower limb including pelvic girdle	4
WESTCHESTER	753.2	Obstructive defects of renal pelvis & ureter	191
	752.6	Hypospadias & epispadias	159
	752.5	Undescended testicle	150
	745.4	Ventricular septal defect	113
	745.5	Ostium secundum atrial septal defect	101
	747.0	Patent ductus arteriosus	94
	755.0	Polydactyly	72
	754.3	Congenital dislocation of hip	63
	750.5	Congenital hypertrophic pyloric stenosis	60
	754.5	Varus deformities of feet	45

Section IV - Table 2
Most Frequently Reported Major Malformations By County
Birth Years: 2002-2004

County	ICD-9 Code	Description	Number
WYOMING	752.6	Hypospadias & epispadias	10
	752.5	Undescended testicle	8
	745.5	Ostium secundum atrial septal defect	5
	745.4	Ventricular septal defect	4
	746.8	Other specified anomalies of heart	4
	747.0	Patent ductus arteriosus	4
	747.3	Anomalies of pulmonary artery	4
	750.5	Congenital hypertrophic pyloric stenosis	4
	753.2	Obstructive defects of renal pelvis & ureter	4
	754.3	Congenital dislocation of hip	3
	754.5	Varus deformities of feet	3
	755.6	Other anomalies of lower limb including pelvic girdle	3
YATES	752.5	Undescended testicle	5
	755.0	Polydactyly	3
	273.8	Other disorders of plasma protein	2
	745.4	Ventricular septal defect	2
	745.5	Ostium secundum atrial septal defect	2
	752.6	Hypospadias & epispadias	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	754.3	Congenital dislocation of hip	2
	742.0	Encephalocele	1
	742.1	Microcephalus	1
	746.7	Hypoplastic left heart syndrome	1
	747.0	Patent ductus arteriosus	1
	747.3	Anomalies of pulmonary artery	1
	747.8	Other specified anomalies of circulatory system	1
	748.6	Other anomalies of lung	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	752.8	Other specified anomalies of genital organs	1
	753.0	Renal agenesis & dysgenesis	1
	754.5	Varus deformities of feet	1
	754.7	Other deformities of feet	1
	755.2	Reduction deformities of upper limb	1
	755.6	Other anomalies of lower limb including pelvic girdle	1
	756.0	Anomalies of skull and face bones	1
	756.7	Anomalies of abdominal wall	1
	758.0	Down syndrome	1
	758.1	Patau syndrome	1

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

Introduction to Table

The CMR relies on reports from hospitals and physicians for case ascertainment. Underreporting is an obvious concern, and the CMR over the years has developed methods to monitor hospital reporting. In this section, CMR live birth prevalences are compared with the national prevalence estimates for 21 selected defects developed by the Centers for Disease Control and Prevention (CDC) and the National Birth Defects Prevention Network (NBDPN).[1] The 21 defects were selected because 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 because they have similar methodology and prevalence. Estimates are usually higher in systems using active case finding, although variations were observed even among the 11 active case finding systems (See Figure 1 in Canfield[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 13 of the 21 defects (bold prevalences) and six of the defects are equal to or higher than the lower 95% Confidence Interval (CI) (boxed prevalences). The prevalences are generally higher for New York State not including New York City than for New York City (16 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 (NTD) is most likely due to the lack of reports on terminations as termination rates for NTDs are high (See CMR report 1998-2001, Section 6, NTD Surveillance and Trends). Lower prevalences for Anophthalmia/Microphthalmia might occur as some of these children have multiple defects and other defects may be reported but not the Anophthalmia/Microphthalmia. (Preliminary data from 2005 shows an increase in Anophthalmia/Microphthalmia which could be the result of the on-site audits. The lower prevalence for cleft lip with/without cleft palate is difficult to explain. There has been little variation in the prevalence since 1983 and it is an easily identified condition. There is also a wide variation within New York State itself from 5.3 in New York City to 13.0 in another region.

None of the 11 registries had all 21 defect prevalences fall within the 95% confidence intervals. Several registries would have the highest prevalence for one defect and the lowest prevalence for others. Variation among the registries in the rates of specific defects could reflect demographic differences in the populations as there are racial and ethnic differences in the rates of specific birth defects[1]. The prevalence of Down syndrome, Trisomy 18 and Trisomy 13 is highly dependent upon the maternal age distribution, age-specific pregnancy rates and women's use of prenatal diagnosis and pregnancy termination. The lower live birth prevalence rates of these chromosomal abnormalities in the CMR may be partially attributable to one or more of these factors. However, the source(s) of much of the variation is unclear and there may be true geographic differences. A comparison of birth defect prevalences between the Metropolitan Atlanta Congenital Defects Program (MACDP) and the California Birth Defects Monitoring Program (CBDMP) for the years 1983-1988 that adjusted for race, sex and maternal age showed regional differences in arm, hand and limb reduction defects [2.]

CMR staff will continue their efforts to improve reporting (See Section 6) and will continue to 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: 2002-004)

Birth defect Category	NY City	Upstate NY	NYS	NBDPN 1999-2001	95% CI Range
Central nervous system defects					
Anencephalus	0.4	0.4	0.4	2.5	2.3-2.7
Spina Bifida without Anencephalus	**1.8**	**2.1**	**1.9**	3.7	3.4-3.9
Encephalocele	0.3	**0.7**	**0.5**	0.9	0.8-1.0
Eye defects					
Anophthalmia/ Microphthalmia	0.6	0.7	0.6	2.1	1.9-2.3
Cardiovascular defects					
Common Truncus	**0.9**	**0.3**	**0.6**	0.8	0.7-0.9
Transposition of Great Arteries	**4.0**	**4.9**	**4.5**	4.7	4.5-5.0
Tetralogy of Fallot	**4.7**	**4.7**	**4.7**	3.9	3.8-4.2
Endocardial Cushion Defect	**3.0**	**2.9**	**2.9**	4.4	4.1-4.6
Hypoplastic Left Heart Syndrome	**2.2**	**2.6**	**2.4**	2.4	2.2-2.6
Orofacial defects					
Cleft palate without cleft lip	**4.5**	**6.4**	**5.5**	6.4	6.1-6.7
Cleft lip with and without cleft palate	5.3	**8.5**	6.9	10.5	10.1-10.9
Gastrointestinal defects					
Esophageal atresia/ tracheosophageal fistula	**2.4**	**2.3**	**2.3**	2.4	2.2-2.6
Rectal and large intestinal atresia/stenosis	**4.2**	**4.7**	**4.5**	4.8	4.5-5.1
Musculoskeletal defects					
Reduction deformity, upper limbs	1.3	**2.2**	1.8	3.8	3.5-4.0
Reduction deformity, lower limbs	0.7	1.1	0.9	1.9	1.7-2.1
Gastroschisis	1.5	**2.2**	1.9	3.7	3.5-4.0
Omphalocele	**1.1**	**1.5**	**1.3**	2.1	1.9-2.3
Diaphragmatic hernia	1.5	1.9	1.7	2.9	2.7-3.1
Chromosomal defects					
Trisomy 13	**0.8**	**0.8**	**0.8**	1.3	1.2-1.5
Down syndrome(Trisomy 21)	**11.3**	**13.2**	**12.3**	13.7	13.2-14.1
Trisomy 18	1.0	1.2	1.1	2.4	2.2-2.6

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

Bold prevalences are within the range of the 11 active registries

Boxed prevalences are equal to or greater than the lower limit of the 95% CI range

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Section VI
Current Topics

Introduction

There are no national data on birth defects despite it being a leading cause of infant mortality and a major cause of mortality and morbidity throughout childhood (Mathews 2007, Hoyert 2006). Data on birth defect prevalences generally come from birth defects registries maintained by specific states. National prevalence estimates have been done in the past using hospital discharge data (Edmonds 1990). The National Birth Defects Prevention Network (NBDPN) is an organization of state birth defects surveillance programs which collects data from 34 state population-based surveillance systems. Recently the Centers for Disease Control and Prevention (CDC) and the NBDPN developed national prevalence estimates for 21 selected major birth defects (Canfield 2006). The defects were chosen because they are recognizable at or shortly after birth and they are likely to be ascertained similarly across states. After examining the data, the NBDPN chose to present numbers using only data from the 11 state registries which used 'active' ascertainment. 'Active' ascertainment systems use field staff to visit hospitals to ascertain and abstract information on cases. These registries were thought to provide more 'reliable and valid' data for the estimates. The national estimates are useful and can be used as a standard for comparisons among registries, for health care planning and to evaluate interventions such as folic acid.

The CMR relies on hospital reporting and thus is termed a 'passive' registry; therefore data from the CMR were not used in creating the national prevalence estimates. We will be using these estimates as a benchmark for our registry (see Section 5). 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, Druschel 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% of children reported to the CMR were diagnosed in the first six months of life, CMR staff begins 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 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% 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 2007a). 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.

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. 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 is able to see if the hospital has been submitting an appropriate number of cases routinely or if the hospital stopped or skipped reporting for certain months or years.

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% reduction in median days used for reporting. (Wang 2007b).

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. The CMR staff reviews the discharge list, comparing it to the list of children who have already been reported to the CMR. The CMR then creates a list of reported, not reported and partially reported cases. 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 medical records including comments that may indicate what chronic reporting problems were evident. Since 2003, 62 hospitals have had an "in-house" audit; 3598 charts have been reviewed; 1311 cases that were not previously reported were flagged and subsequently reported, 326 cases that were partially reported were completed and 154 cases with incorrect diagnoses reported were corrected or deleted.

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 recognizes 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.

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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

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

*See list of minor and excluded codes

Minor Malformations

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

Exclusions

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

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

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

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

Appendix 3

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 5-Digit Code
CENTRAL NERVOUS SYSTEM			
A01	Anencephaly	740.0, 740.1, 740.2	740.00, 740.02, 740.03, 740.10, 740.20, 740.21, 740.29
A02	Spina Bifida with Hydrocephaly	741.0	741.00, 741.01, 741.02, 741.03, 741.04, 741.05, 741.06, 741.07, 741.08, 741.09
A03	Spina Bifida without Hydrocephaly	741.9	741.90, 741.91, 741.92, 741.93, 741.94, 741.98, 741.99, 742.00, 742.08, 742.09
A13	Encephalocele	742.0	742.00, 742.08, 742.09
A15	Hydrocephaly	742.3	742.30, 742.31, 742.38, 742.39
A16	Microcephalus	742.1	742.10
EYE / EAR			
B01	Anophthalmia, Microphthalmia	743.0, 743.1	743.00, 743.10
B03	Glaucoma	743.2	743.20, 743.21, 743.22
B04	Cataract		743.32
B54	Ear anomaly with hearing loss	744.0	744.00, 744.01, 744.02, 744.03, 744.09, 744.21
CARDIAC			
D01	Truncus arteriosus	745.0	745.00, 745.01
D02	Transposition of great vessels	745.1	745.10, 745.11, 745.12, 745.18, 745.19
D03	Tetralogy of Fallot	745.2	745.20, 745.21, 746.84
D04	Single ventricle	745.3	745.30
D05	VSD	745.4	745.40, 745.41, 745.48, 745.49
D52	Hypoplastic left heart	746.7	746.70
D53	Total anomalous pulmonary venous return	747.41	747.42
RESPIRATORY			
E01	Choanal atresia	748.0	748.00
E06	Agenesis of lung	748.5	748.50, 748.51
CLEFTS			
F01	Cleft palate	749.0	749.00, 749.01, 749.02, 749.03, 749.04, 749.05, 749.06, 749.07, 749.09
F02	Cleft lip with or without cleft palate	749.0, 749.2, 750.5	749.10, 749.11, 749.12, 749.19, 749.20, 749.21, 749.22, 749.29, 749.51

MACDP Code	Condition	ICD-9	BPA 5-Digit Code
GASTRO-INTESTINAL			
F14	Stenosis or atresia of duodenum	751.1	751.10
F15	Other stenosis or atresia of small intestine	751.1	751.11, 751.12, 751.19
F16	Stenosis or atresia of rectum or anus	751.2	751.21, 751.22, 751.23, 751.24
F17	Hirschsprung's Disease	751.3	751.30, 751.31, 751.32, 751.33
F18	Malrotation of intestine	751.4	751.40, 751.41, 751.42, 751.49
F21	Biliary atresia	751.61	751.65
GENITO-URINARY			
H01	Renal agenesis	753.0	753.00, 753.01
H06	Obstruction of kidney or ureter	753.3	753.20, 753.21, 753.22, 753.29, 753.40, 753.42
H09	Bladder or urethra obstruction	753.6	753.60, 753.61, 753.62, 753.63
MUSCULOSKELETAL			
J02	Curvature of spine (scoliosis or lordosis)	754.2	754.20, 754.21, 754.22
J03	Dislocation of hip	754.3	754.30
J11	Arthrogryposis multiplex congenita	754.89	755.80
K01	Reduction deformity - upper limb	755.2	755.20, 755.21, 755.22, 755.23, 755.24, 755.25, 755.26, 755.27, 755.28, 755.29
K02	Reduction deformity - lower limb	755.3	755.30, 755.31, 755.32, 755.33, 755.34, 755.35, 755.36, 755.37, 755.38, 755.39
K05	Amniotic bands	658.8	658.80
N01	Diaphragmatic hernia	756.6	756.61
N02	Omphalocele	756.7	756.70
N04	Gastroschisis	756.7	756.71
SYNDROMES			
R01	Down Syndrome	758.0	758.00, 758.01, 758.02, 758.03, 758.04, 758.09
R02	Patau Syndrome (Trisomy 13)	758.1	758.10, 758.11, 758.12, 758.13, 758.19
R03	Edwards Syndrome (Trisomy 18)	758.2	758.20, 758.21, 758.23, 758.29
S02	Fetal Alcohol Syndrome	760.71	760.71
W03	Conjoined twins	759.4	759.40, 759.41, 759.42, 759.43, 759.44, 759.48, 759.49

Appendix 4

Glossary of Terms*

Agenesis Absence of part(s) of the body.

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

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

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

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

Anotia A congenital absence of one or both ears.

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

Atresia Imperforation; absence or closure of a normal opening.

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

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

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

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

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

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

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

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

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

Confidence interval (95%) The interval that contains the true prevalence (which we can only estimate) 95% of the time.

Congenital Existing at or dating from birth.

Congenital hip dislocation A congenital defect in which the head of the femur does not articulate with the acetabulum of the pelvis because of an abnormal shallowness of the acetabulum. Treatment in early infancy consists of bracing of the joint to cause a deepening of the acetabulum.

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

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

Diaphragmatic hernia A failure of the diaphragm to form completely, leaving a hole. Abdominal organs can protrude through the hole into the chest cavity and interfere with development of the heart and lungs. Usually life-threatening and requires emergent surgery.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Limb defects See Reduction deformities.

Meninges Membranes that cover the brain and spinal cord.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Trisomy 13 See Patau Syndrome.

Trisomy 18 See Edwards Syndrome.

Trisomy 21 See Down Syndrome.

Truncus Arteriosus See Common Truncus.

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

*Courtesy of the Texas Birth Defects Monitoring Division February 1999