Massachusetts Birth Defects 2002-2003

Massachusetts Birth Defects Monitoring Program
Bureau of Family Health and Nutrition

Massachusetts Department of Public Health

January 2008

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Executive Summary

Although birth defects are rare when compared to other adverse birth outcomes, such as low birth weight or prematurity, they are the leading cause of death in the first year of life in the United States. About 20% nationally of all infant deaths result from a birth defect. In Massachusetts, 16.4% of all infant deaths were attributable to birth defects. (Massachusetts Deaths, 2002) The overall infant mortality rate for MA in 2002 was 4.9, for the U.S. it was 6.9. The infant mortality rate due to birth defects in the U.S. in 2002 was 13.99 per 10,000 live births (Callaghan et al., 2006) and for MA it was 8.06.

The causes of birth defects are poorly understood. For 60-70% of major birth defects no known cause has been identified. Researchers are looking at a wide variety of environmental exposures and risk factors as possible causes. Because most of the structural development of the fetus occurs during early pregnancy, studies usually focus on the "periconceptional" period, the month before and three months after conception. For the developing pregnancy, the environment includes any exposures to the fetus as well as any exposures to the mother.

The Massachusetts combined lifetime costs for babies born with any of 12 major structural birth defects are an estimated $122 million in 2003 dollars (Harris, 1997; see Technical Notes for inflation adjustment). These figures include direct costs of medical treatment, developmental services and special education, as well as indirect costs to society for lost wages due to early death or occupational limitations. The psychosocial costs cannot be calculated.

Over the past ten years, the Massachusetts Center for Birth Defects Research and Prevention has developed and refined its surveillance program. The first full year of population-based, active statewide surveillance data was 1999. The primary focus of the state surveillance system is the identification of major structural birth defects, with or without a chromosomal abnormality, and non-chromosomal malformation syndromes. Inborn errors of metabolism are monitored separately by the state newborn screening program.

This report presents statewide data on the prevalence of birth defects in live births and stillbirths in Massachusetts primarily during the years 2002 and 2003. The first annual report presented Massachusetts birth defects data for the year 1999. Our second report compiled data from 2000-2001. Our ability to find and identify infants born with birth defects to Massachusetts' residents has improved over time and is reflected in increased prevalence rates. The 2002-2003 data are presented in combined form since the numbers are relatively small for individual defects. Interpretations of these data must be made with caution until a multi-year estimate establishes a stable, baseline rate.

Prevalence

The overall prevalence of birth defects among births to Massachusetts residents in 2002-2003 was 157.41 per 10,000 live births. Among the 160,791 live births to Massachusetts residents in 2002-2003, 2,476 had one or more birth defects. In addition, 55 stillbirths were identified as having a birth defect. Three of the ten most common defects were cardiovascular defects: Atrial Septal Defects, Ventricular Septal Defects and Pulmonary Stenosis (Valvular). Common non-cardiovascular defects included Trisomy 21, Polydactyly/Syndactyly, Hypospadias, Clubfoot, Cleft Lip with and without Cleft Palate, Cleft Palate alone, and Obstructive Genitourinary Defects.

The CDC published improved national prevalence estimates for 18 selected major defects. Massachusetts was one of 11 states with population-based monitoring programs to contribute birth defect data. These average prevalence rates cover deliveries from 1999-2001. (MMWR, 2006) Massachusetts rates for 2002-2003 were significantly lower than the US rates for about half of the defects and were about the same as the national estimates for the other half. The lower rates for the other defects may reflect differences in defect criteria between surveillance systems and regional differences. Also, birth defects are not reported in Massachusetts when they are prenatally diagnosed and the pregnancy is subsequently electively terminated, which would tend to result in lower rates for MA for certain defects. Spontaneous deliveries of stillbirths >= 20 weeks of gestation were reported by birthing hospitals but limited information about the stillbirth is included in the maternal record. Thus some birth defects are not well documented and are unable to be confirmed for inclusion in state surveillance.

Single vs. Multiple Defects

Of all 2,531 birth defect cases (infants and stillborns) 56.2% had a single defect and 43.8% had multiple defects. (A case was defined as having multiple defects, if it had more than one defect from among those that were included in this surveillance report.) Anencephaly, Cleft Lip with and without Cleft Palate, Gastroschisis, Hirschsprung Disease, Clubfoot, and Hypospadias appeared more often as a single defect rather than with other defects. Birth defects which appeared more often in conjunction with other defects included the majority of Cardiovascular Defects, Limb Reductions, Hydrocephalus, Esophageal Atresia/Tracheoesophagela Fistula, Intestinal Atresias, and Obstructive Genitourinary Defects.

Selected Pregnancy Outcomes

We compared selected pregnancy outcomes (C-sections, birthweight, gestational age, multiple birth and infant death) among infants born with birth defects to those born without birth defects in 2002-2003. Of infants born with birth defects, 40.3% were C-section deliveries, compared to 28.4% of non-birth defect births; 21.4% of low birthweight births (<2500 grams) had a birth defect as opposed to 7.4 % of those without a birth defect; 4.8% of infants with a birth defect died before their 1st birthday, compared to 0.4% of those without a birth defect. While numbers of

infants with birth defects are relatively small, it is important to recognize the impact of these outcomes when diagnosing and treating a baby with a birth defect.

Plurality

Examining the birth defect rate by plurality is important since birth defects are more common among multiple births and the number of multiple births has been increasing over time in Massachusetts. The birth defect prevalence rate was 152.77 for singletons and 247.71 for multiple births (more than one infant) per 10,000 live births. Birth defects that more commonly occurred in multiple births included Esophageal Atresia/Tracheoesophageal Fistula, Hypospadias, Coarctation of Aorta, Diaphragmatic Hernia and Polydactyly/Syndactyly.

Sex

The birth defect prevalence rate was 128.79 for females and 184.71 for males per 10,000 live births. While the prevalence of most types of birth defects did not substantially differ by sex of the infant/fetus, some conditions were associated with sex. The most common defects seen in males were Hypospadias, Atrial Septal Defects (Secundum and NOS), Polydactyly/Syndactyly, Clubfoot, Down Syndrome and Obstructive Genitourinary Defects. The most common defects seen in females were Atrial Septal Defects (Secundum and NOS), Down Syndrome, Pulmonary Stenosis Valvular, Polydactyly/Syndactyly and Clubfoot.

Maternal Age

Monitoring birth defects by maternal age is important since the number of births to older mothers has been increasing over time in Massachusetts. The prevalence of birth defects varied by maternal age group. For live births only, rates per 10,000 live births were 143.46 for mothers younger than 20 years, 147.64 for those 20-24 years, 141.66 for those 25-29 years, 146.69 for those 30-34 years, and 183.98 for those 35 years and older. As expected, there was a strong association of Down Syndrome with advanced maternal age. Although more babies with Down Syndrome were born to women under 35, the Down Syndrome rate of 29.16 per 10,000 births for women 35 years and older was four times that of any other maternal age group. Younger mothers (age 19 and under) had the highest rate (12.75) of Gastroschisis. These associations have been shown in previous studies.

Maternal Race / Hispanic Ethnicity

The prevalence of birth defects varied by maternal race and Hispanic ethnicity. The rate per 10,000 live births was 157.85 for Non-Hispanic Whites, 142.62 for Non-Hispanic Blacks, 155.38 for Hispanics, and 114.03 for Asians/Pacific Islanders. Due to small numbers, the rates for other races were not calculated. The most common defects in Hispanics included Septal Defects, Polydactyly/Syndactyly, Clubfoot and Down Syndrome. In Blacks, the most common defects included Septal Defects, Down Syndrome, Polydactyly/Syndactyly, Pulmonary Stenosis (Valvular) and Hypospadias. The most common defects in Whites included Septal Defects, Hypospadias, Down

Syndrome, Polydactyly/Syndactyly and Clubfoot. In Asians, the most common defects included Septal Defects, Clubfoot, Cleft Lip, Cleft Palate, and Down Syndrome.

Severity

A severity scale was developed by the Center in collaboration with our partners at Boston University and the Massachusetts General Hospital. This scale was based on the usual outcome for a specific birth defect including its typical compatibility with survival, the need for immediate treatment, the need for long-term care, and the amenability of the defect to correction. A severity score was assigned to each case based on the most severe defect for that infant/fetus. If a case had multiple defects with equal severity, it was reviewed in detail by the Center Clinical Geneticist to determine severity category. Cases with a syndrome plus defect(s) were listed by syndrome only. (Syndromes are defined as a group of malformations that occur together frequently enough to be recognized collectively as a distinct abnormal condition.) Specific severity category definitions used in this report were as follows:

SEVERITY CATEGORIES	PERCENTAGE OF BIRTH DEFECTS CASES
Severe, supportive measures, usually incompatible with life	2.8%
Serious, may be correctable, most have long-term needs	16.6%
Moderate, most correctable, many have long-term needs	72.7%
Mild, may be correctable, minimal long-term needs	7.9%

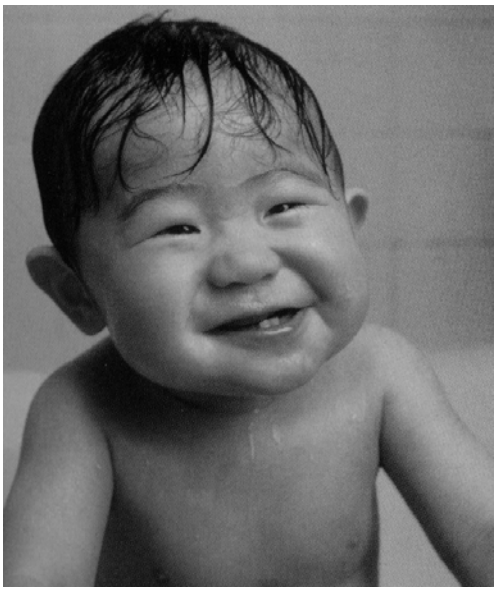
Nearly 3% of cases with birth defects were classified as “severe” and most did not survive. This percentage was an underestimate of cases due to limited data. For example, Cragan at CDC estimated that up to 80% of Anencephaly cases and 50% of any neural tube defect may be electively terminated after prenatal diagnosis (Cragan 2000). About 17% of cases were affected with a “serious” birth defect. These cases needed intensive medical care and planning for continuing care and long-term disability. “Moderately severe” birth defects comprised 73% of the total cases; all of these needed medical follow up; many may have required a number of surgeries and extensive treatment. “Mild” birth defects comprised nearly 8% of the affected infants. These defects may or may not have required corrective treatment.

Planning for children with special health care needs is essential to support affected infants and families. Coordination between the Birth Defect Monitoring Program and maternal and child health programs helps to ensure services for identified children and to provide population-based information to inform program planning and prevention strategies.

Two resource lists: "Selected National Resources" and "Public Health Resources in Massachusetts" accompany this report.

Chapter 1

Introduction



The Public Health Importance of Birth Defects

Each year in the United States, approximately 150,000 babies are born with birth defects. One in every 28 families of a newborn is forced to deal with the reality that their baby has a birth defect (March of Dimes). Birth defects, sometimes called congenital anomalies, are abnormalities of structure, function or metabolism present before birth. These abnormalities may be fatal or may result in physical or mental disability. Several thousands of defects have been identified. Some are life threatening while others are less significant.

Birth defects can lead to lifelong disability, require costly medical care and cause great distress in families. The economic, emotional and social impact on families is catastrophic.

Although birth defects are rare when compared to other adverse birth outcomes, such as low birth weight or prematurity, they are the leading cause of death in the first year of life in the United States. About 20% nationally of all infant deaths result from a birth defect. In Massachusetts, 16.4% of all infant deaths were attributable to birth defects. (Massachusetts Deaths, 2002) The overall infant mortality rate for MA in 2002 was 4.9, for the US it was 6.9. The infant mortality rate due to birth defects in the US in 2002 was 13.99 per 10,000 live births (Callaghan et al., 2006) and for MA it was 8.06.

Causes of Birth Defects

The causes of birth defects are poorly understood. Certain genetic and environmental factors have been implicated in selected defects. These include: prenatal environmental factors, such as infections, e.g., rubella, exposures to medications or other chemicals, drug or alcohol abuse or nutritional deficiencies. It is also known that a single abnormal gene can cause certain birth defects. The gene may have an error in its code, a missing piece or extra genetic material, all of which can result in malformations. Other birth defects may be caused by a combination of factors, such as genes interacting with environmental factors. For 60-70% of major birth defects, no known cause has been identified.

Researchers are looking at a wide variety of environmental exposures and risk factors as causes. Because most of the structural development of the fetus occurs during early pregnancy, studies usually focus on the “periconceptional” period, the month before and three months after conception. For the developing pregnancy, the environment includes any exposure to the fetus as well as any exposure to the mother.

Research has already provided some valuable information. Studies have shown that the presence of adequate amounts of folic acid (vitamin B9) in the mother’s system during the “periconceptional” period may help prevent defects of the brain

and spinal cord known as neural tube defects. Mandatory fortification of cereal grains with folic acid has resulted in a 26% reduction in the number of babies born with these neural tube defects (Mills, 2004).

Healthy People 2010 Challenges

Healthy People 2010 established the objectives of reducing the fetal and infant death rates by 40%, developmental disabilities rates by 50%, and neural tube defect rates by 50%. Birth defects surveillance is a critical component of the public health strategy to achieve these objectives. The active surveillance program in Massachusetts allows the Department of Public Health to monitor the extent and occurrence of birth defects within the Commonwealth. These data make it possible to identify:

- Changes in birth defects rates over time that may indicate a change in environmental conditions affecting the health of the population;
- Geographical areas with consistently high or unusual rates;
- Clusters of birth defects;
- Families of affected children who may benefit from services or who may be interested in participating in research studies; and
- Key data for preventive strategy planning by the Department of Public Health.

Birth Defects Surveillance in Massachusetts

Over the past ten years, the Center for Birth Defects Research and Prevention has developed and refined its surveillance program. The first full year of population-based, active surveillance statewide was 1999.

The primary focus of the state surveillance system is the identification of major structural birth defects, with or without a chromosomal abnormality and non-chromosomal malformation syndromes. This includes ICD-9 CM codes ranging from 740.0 to 759.9 and a few selected codes outside this range for defects such as DiGeorge Syndrome, Pierre Robin Sequence and Amniotic Bands Sequence.

The Center's active surveillance system uses multiple sources of ascertainment. Birth, tertiary (Level III nurseries) and specialty care hospitals in Massachusetts routinely submit discharge lists and nursery data on infants born with birth defects. Since over 70% of out-of-state births to Massachusetts' mothers occur in Rhode Island, two Rhode Island hospitals, the Women and Infants' Hospital and the Rhode Island Hospital, were added in 2000. In 2001, the Massachusetts Eye and Ear Infirmary was included in order to increase ascertainment of eye and ear anomalies that come to their attention. Vital records also serve as an additional source. Fetal death reports and infant death certificates are reviewed. Birth certificates are checked for additional information such as residency of the mother.

Potential birth defect cases, reported from these varied sources, are assigned to medical record abstractors who make field visits to hospital records departments. Abstractors have specialized training and ongoing education to abstract medical records of potential cases. Abstraction is conducted on a regular basis using a Confidential Reporting and Abstraction Form (CRAF) to capture essential data for each birth defect case. The CRAFs are submitted to the Center for review of completeness and accuracy. Surveillance data are entered and maintained in a confidential electronic database.

Economic Impact on Massachusetts

Estimating the economic impact of birth defects on the state of Massachusetts is challenging. The California Birth Defects Monitoring Program and the Metropolitan Atlanta Congenital Defects Program, using 1992 data, calculated the lifetime costs for families dealing with a baby with birth defects to be between $75,000 and $503,000 (Waitzman et al., 1994). Their estimated lifetime costs for a baby born with Spina Bifida would be $364,560 in 2003 dollars.

Adjusting for inflation, the Massachusetts combined lifetime costs for babies born with 12 major structural birth defects were an estimated $122 million in 2003 dollars (see Technical Notes). These figures included direct costs of medical treatment, developmental services and special education, as well as indirect costs to society for lost wages due to early death or occupational limitations. There are also social and emotional impacts.

Legislative Changes Regarding Birth Defects Surveillance

In March 2002, the Massachusetts Legislature amended the state birth defects monitoring statute (Chapter 111, section 67E) to allow expansion of the surveillance system to capture diagnoses through age three. It also extends mandated reporters to include attending physicians, primary care and specialist physicians who may diagnose birth defects. These physicians will now have a statutory duty to report within 30 days of making such a diagnosis. The amended statute also permits researchers to access state surveillance data after obtaining IRB approval and approval of the MDPH Commissioner pursuant to M.G.L.c.111s.24A/B/67E.

The 2002-2003 Surveillance Report

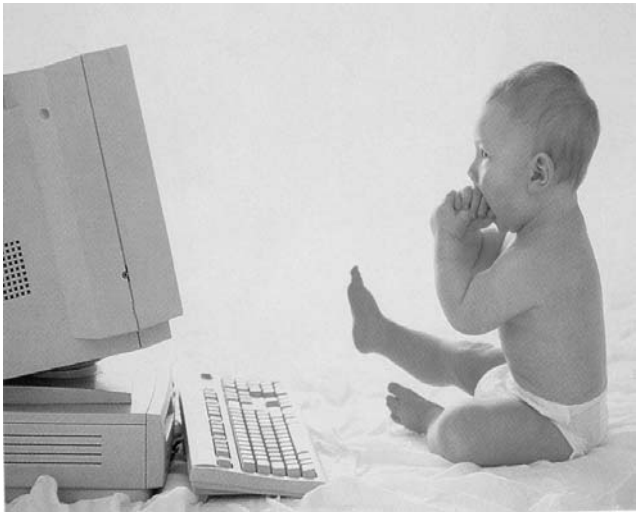
This report presents statewide data on the prevalence of birth defects in live births and stillbirths in Massachusetts during the years 2002 and 2003. The data are presented in combined form since the numbers are relatively small for individual defects. The first annual report presented Massachusetts data for birth defects for the year 1999. Our second report compiled data from 2000-2001. Our ability to find and identify infants born with birth defects to Massachusetts' residents has improved over time. There was about a 12% increase in cases from 2000-2001 to

2002-2003 that is attributable to this improved case ascertainment. Interpretations of these data must be made with caution until a multi-year estimate establishes a stable, baseline rate.

Unless otherwise indicated the report uses the term “births” to mean live births plus stillbirths. A stillbirth was defined as the delivery of a fetus that was not alive, and was greater than or equal to 20 weeks gestational age, or weighed at least 350 grams.

Chapter 2

Methods



Case Definition

This report presents data on selected birth defects present in births occurring during the calendar years 2002 through 2003 to Massachusetts residents. Cases met the following criteria:

- The infant was live born or, the fetus was stillborn with a gestational age greater than or equal to 20 weeks or with a weight of at least 350 grams.
- The infant or fetus had a structural birth defect that met diagnostic criteria (see Birth Defects Codes and Exclusions by Defect Category in Appendices).
- The diagnosis was made before the infant reached one year of age.

Data Collection

The Massachusetts Birth Defects Monitoring Program (BDMP) used active surveillance methods for population-based, statewide case ascertainment. Hospitals across the state submitted monthly discharge lists with birth defect diagnoses to the Center. Nursery and neonatal intensive care liaisons phoned in reports of birth defects. Abstractors reviewed medical charts for each potential case. If the infant or fetus had a birth defect that met the case definition criteria, detailed demographic and diagnostic information was recorded on a hospital reporting form. This information was entered into a confidential surveillance database for analysis.

Confidentiality

Great care was taken to protect the confidentiality of data. The Center has developed extensive procedures to guarantee the confidentiality of data and protect the privacy of families. These procedures uphold our ethical and legal obligations to safeguard confidentiality and fully comply with the strict requirements of state and federal laws.

Data Analysis

A defect may have occurred as a single event or with other defects. If the case had more than one defect within the same defect category, only one of these defects was counted in the category total. If the case had more than one defect in different defect categories, the case was listed in the total for each of these defect categories. Thus the counts in the defect categories presented in the prevalence tables represent the total number of defects, not the total number of cases with birth defects. Each case in the BDMP was linked to a Registry of Vital Records record. In this report, maternal age and race/ethnicity are drawn from the birth certificate. Because birth certificate data are more accurate for these fields than fetal death records, analyses of maternal age and race/ethnicity are limited to live births.

The occurrence of birth defects is reported as prevalence. Prevalence is calculated as the number of birth defect cases born during the period 2002-2003 per 10,000 live births born during the same period. Prevalence tables include the number of cases found, the estimated prevalence rate per 10,000 live births, and the 95% confidence interval for that rate. The incidence (new cases) of birth defects (based upon the number of embryos conceived within a year) is not fully measured because both the total number of conceptions that occur and the number of these conceptions resulting in a defect are not known (Sever 1996).

The confidence interval (CI) can be used to assess the magnitude and stability of a rate or ratio. The confidence interval (CI) for the rates in the tables is a range of values that has a 95% chance of including the underlying risk of an infant being born with a birth defect. Wide confidence intervals reflect the large variation due to small numbers (see Technical Notes).

Data Limitations

1. Birth defect counts for this report are only for calendar years 2002 through 2003. Due to the small numbers of birth defects, conclusions from these results are not valid until a more extensive multi-year estimate establishes a stable, baseline rate.

2. Currently, the Massachusetts Birth Defects Monitoring Program ascertains cases only at birthing hospitals, two non-birthing tertiary care centers and one specialty care hospital. Thus, defects that are not diagnosed at birth and that do not need hospitalization may be underreported (e.g., cardiac defects that are detected in an outpatient setting after the immediate newborn period).

3. Misclassification of birth defects may occur through coding errors or vague diagnoses. Quality control measures such as careful abstraction of the medical records minimize this error.

4. As medical diagnostic technology has improved, many prenatal and postnatal tests are now performed outside the traditional hospital setting. Prenatal diagnosis enables physicians to identify some birth defects well before the expected date of delivery, and offers women alternatives in the management of their affected pregnancies. These decisions have significant implications for monitoring birth defects. For example, it is estimated that up to 50% of all pregnancies affected with a neural tube defect may be discontinued and would thus not be included in hospital records (Cragan 2000). In addition, postnatal tests such as echocardiograms and ultrasounds may identify internal organ defects not diagnosed in the birthing hospital.

5. Spontaneous abortions that are delivered prior to 20 weeks of gestation and less than 350 grams are not included in the case definition. It has been estimated that about 29% of birth defects cases are missed by not monitoring early fetal loss (Forrester 1998, TBDR 2000).

6. Only diagnoses confirmed before one year of age are currently included. The frequency of diagnosed malformations can be higher among older children due to 'hidden' abnormalities such as kidney malformations or certain heart defects which are detected by accident or when a child is symptomatic (Holmes 1994). Another example, Fetal Alcohol Syndrome, may not be detected until developmental delays become evident when a child is much older.

7. In 2000, 1318 births occurred to MA residents at out-of-state hospitals. Of these births, 68.9% occurred in Rhode Island (RI) hospitals. In order to capture data on infants with birth defects residing in the southeastern region of Massachusetts that were born or treated at RI hospitals, we began receiving hospital discharge lists and abstracting medical records on infants with birth defects at two RI hospitals. Deliveries and diagnoses that occurred in other out-of-state facilities are not included at this time.

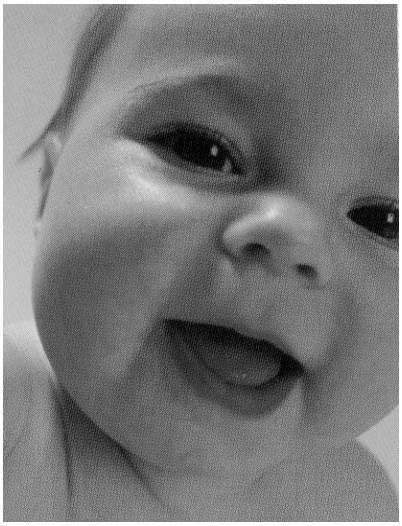
8. There are limitations when comparing the MA BDMP data to data from other states and national estimates. Factors such as differences in the demographics of the two populations, the environments in which they live, and the methods of surveillance conducted by the two programs may contribute to differences in the prevalence of birth defects.

Glossary

A glossary of selected birth defect terms is included in the appendices of this report.

Chapter 3

Prevalence of Birth Defects



Overall Prevalence of Birth Defects

Table 1 shows the prevalence of defects for all births and for live births and stillbirths separately. Among the 160,791 live births to Massachusetts residents in 2002-2003, 2,476 had one or more structural birth defects that were ascertained by MBDMP. In addition, 55 stillbirths were identified with a birth defect. Overall, 1.57% of births in the state were identified as having one or more birth defects.

The CDC published improved national prevalence estimates for 18 selected major defects. Massachusetts was one of 11 states with population-based monitoring programs to contribute birth defect data. These average prevalence rates cover deliveries from 1999-2001 (MMWR, 2006). Massachusetts rates for 2002-2003 were significantly lower than the US rates for about half of the defects and were about the same as the national estimates for the other half (see Table 2). The lower rates for the other defects may reflect differences in defect criteria between surveillance systems as well as regional differences. Also, birth defects are not reported in Massachusetts when they are prenatally diagnosed and the pregnancy is electively terminated, which would tend to result in lower rates for MA for certain defects. Spontaneous deliveries of stillbirths equal to or greater than 20 weeks of gestation were reported by birthing hospitals but limited information about the stillbirth is included in the maternal record. Thus, some birth defects are not well documented and are unable to be confirmed for inclusion in state surveillance.

Another data source to which we can compare MA rates is the Neural Tube Defect (NTD) Ascertainment project of the National Birth Defects Prevention Network at CDC. Massachusetts has submitted data quarterly since 1999. Using data from 1999-2000, researchers from CDC calculated prevalence rates for Spina Bifida and Anencephaly, two serious birth defects that occur early in pregnancy. (MMWR, 2004) Birth defect programs which included prenatally diagnosed cases of Spina Bifida that are subsequently electively terminated had a prevalence rate of 4.1. Massachusetts' rate of 1.6 for 2000-2003 is nearly 60% lower (see Table 2).

We can estimate cases missed if we compare our data to Brigham and Women's Hospital (BWH), where 12% of resident births occurred in 2002-2003. BWH researchers looked at trends in elective termination in Massachusetts. For the two years 1994 and 1999, 40-80% of pregnancies with either lethal or very severe defects were terminated (Peller 2004). CDC estimates that 80% of all Anencephaly and 50% of all neural tube defect cases may not be reported due to prenatal diagnosis and subsequent elective terminations (Cragan 2000).

The overall prevalence of reported birth defects in Massachusetts in 2002-2003 was 157.41 per 10,000 live births. This represents a 12.6% increase from 2000-2001, when the prevalence rate was 136.8. This increase was due to better reporting from hospitals and improved ascertainment of cases. The majority of defects occurred in the Cardiovascular (34.5%) and Musculoskeletal (26.8%)

categories. Figure 1 shows the percentage of reported birth defects by defect categories.

Table 3 shows the most common birth defects in the state. Three of the ten most common defects were cardiovascular defects: Atrial Septal Defects, Ventricular Septal Defects and Pulmonary Stenosis (Valvular). Common non-cardiovascular defects included Trisomy 21, Polydactyly/Syndactyly, Hypospadias, Clubfoot, Cleft Lip with and without Cleft Palate, Cleft Palate alone, and Obstructive Genitourinary Defects. Cardiovascular Defects were the most commonly occurring birth defects in both Massachusetts and in the nation. They also contribute more to infant deaths than any other defect category (Petrini 1998).

Single vs. Multiple Defects

Table 4 shows the distribution of birth defects by whether they appeared as a single diagnosis or in combination (multiple) with other defects. A case was defined as having multiple defects, if it had more than one defect from among those that are included in this surveillance report.

Among birth defect cases, 56.2% had single defects and 43.8% had multiple defects. Anencephaly, Cleft Lip with and without Cleft Palate, Gastroschisis, Hirschsprung Disease, Clubfoot, and Hypospadias appeared more often as a single defect rather than in combination with other defects. Limb Reductions, Hydrocephalus, Esophageal Atresia/Tracheoesophageal Fistula, Intestinal Atresias, and Obstructive Genitourinary Defects were some of the birth defects that appeared more often with other defects. Overall, Cardiovascular Defects were five times more likely to occur as one of multiple defects than as a single defect.

Selected Pregnancy Outcomes

Figure 2 compares selected pregnancy outcomes (C-sections, birthweight, gestational age, multiple birth and infant death) among infants born with birth defects to those born without birth defects in 2002-2003 by percentage. Of infants born with birth defects, 40.3% were C-section deliveries, compared to 28.4% of non-birth defect births; 21.4% of low birthweight births (<2500 grams) also had a birth defect as opposed to 7.4 % of those without a birth defect; 4.8% of infants with a birth defect died before their 1st birthday, compared to 0.4% of those without a birth defect. While numbers of infants with birth defects are relatively small, it is important to recognize the impact of these outcomes when diagnosing and treating a baby with a birth defect.

Table 1 Prevalence of Birth Defects, Massachusetts: 2002-2003

Defect[1]	Livebirth Count	Stillbirth Count	Total Count	Rate per 10,000 Births	95% Confidence Interval
Central Nervous System: 220 cases					
Anencephaly	3	3	6	0.37	0.14—0.81
Encephalocele	6	0	6	0.37	0.14—0.81
Holoprosencephaly	7	1	8	0.50	0.21—0.98
Hydrocephaly w/o Spina Bifida	53	2	55	3.42	2.58—4.45
Microcephaly	32	0	32	1.99	1.36—2.81
Spina Bifida w/ and w/o Hydrocephaly	25	1	26	1.62	1.06—2.37
Spinal Cord	53	1	54	3.36	2.52—4.38
Other CNS	87	3	90		
Eye: 62 cases					
Aniridia	4	0	4	0.25	0.07—0.64
Anophthalmia/Microphthalmia	13	2	15	0.93	0.52—1.54
Congenital Glaucoma, Congenital Cataract	31	0	31	1.93	1.31—2.74
Other Eye	25	0	25		
Ear: 19 cases					
Anotia/Microtia	18	1	19	1.18	0.71—1.85

Table 1 Prevalence of Birth Defects, Massachusetts: 2002-2003 (cont'd)

Defect[1]	Livebirth Count	Stillbirth Count	Total Count	Rate per 10,000 Births	95% Confidence Interval
Cardiovascular: 874 cases					
Anomalous Pulmonary Venous Connection					
Total/Partial Anomalous Pulmonary Venous Connection	21	0	21	1.31	0.81—2.0
Atrioventricular Canal Defects					
ASD Primum	14	0	14	0.87	0.48—1.46
Common Atrium	4	0	4	0.25	0.07—0.64
Complete Atrioventricular Canal Defect	47	0	47	2.92	2.15—3.89
Endocardial Cushion (OS and NOS)	9	0	9	0.56	0.26—1.06
VSD, Canal Type	10	1	11	0.68	0.34—1.22
Conotruncal (Outlet) and Aortic Arch					
Double Outlet Right Ventricle	19	1	20	1.24	0.76—1.92
d-Transposition of the Great Arteries	51	0	51	3.17	2.36—4.17
Interrupted Aortic Arch, Type B	4	0	4	0.25	0.07—0.64
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	63	2	65	4.04	3.12—5.15
Truncus	3	0	3	0.19	0.04—0.55
Ebstein Anomaly					
Ebstein Anomaly	6	1	7	0.44	0.18—0.90
Laterality Defects					
Heterotaxy, Situs Inversus	13	0	13	0.81	0.43—1.38
Left-Sided Obstruction					
Aortic Valve Stenosis	32	2	34	2.11	1.46—2.95
Coarctation of Aorta	74	0	74	4.60	3.61—5.78
Hypoplastic Left Heart Syndrome	20	0	20	1.24	0.76—1.92
Interrupted Aortic Arch (Type A and NOS)	4	0	4	0.25	0.07—0.64
Patent Ductus Arteriosus					
Patent Ductus Arteriosus	269	0	269	16.73	14.79—18.85
Right-Sided Obstruction					
Pulmonary Stenosis, Valvular	105	0	105	6.53	5.34—7.91
Pulmonary Valve Atresia w/intact septum	14	0	14	0.87	0.48—1.46
Pulmonary Valve Atresia with VSD	6	0	6	0.37	0.14—0.81

Table 1 Prevalence of Birth Defects, Massachusetts: 2002-2003 (cont'd)

Defect[1]	Livebirth Count	Stillbirth Count	Total Count	Rate per 10,000 Births	95% Confidence Interval
Tricuspid Valve Atresia	12	1	13	0.81	0.43—1.38
Septal Defects					
ASD (Secundum and NOS)	310	3	313	19.47	17.37—21.74
VSD (Membranous and NOS)	177	2	179	11.13	9.56—12.89
VSD, Conoventricular/Malalignment	30	0	30	1.87	1.26—2.66
Single Ventricle and L-TGA					
L-TGA	8	0	8	0.50	0.21—0.98
Single Ventricle	2	0	2	0.12	0.02—0.45
Other Cardiovascular					
Other Cardiovascular	254	6	260		
Respiratory: 43 cases					
Choanal Atresia	4	0	4	0.25	0.07—0.64
Lung Anomalies	24	2	26	1.62	1.06—2.37
Other Respiratory	14	0	14		
Orofacial: 251 cases					
Cleft Lip w/ and w/o Cleft Palate	110	1	111	6.90	5.68—8.31
Cleft Palate w/o Cleft Lip	94	1	95	5.91	4.78—7.22
Pierre Robin Sequence	35	0	35	2.18	1.52—3.03
Other Orofacial	49	3	52		
Gastrointestinal: 245 cases					
Biliary Atresia	10	0	10	0.62	0.3—1.14
Esophageal Atresia/Tracheoesophageal Fistula	38	1	39	2.43	1.72—3.32
Hirschsprung Disease	18	0	18	1.12	0.66—1.77
Rectal and Large Intestinal Atresia/Stenosis	61	3	64	3.98	3.07—5.08
Small Intestinal Atresia	43	2	45	2.80	2.04—3.74
Other Gastrointestinal	90	0	90		

Table 1 Prevalence of Birth Defects, Massachusetts: 2002-2003 (cont'd)

Defect[1]	Livebirth Count	Stillbirth Count	Total Count	Rate per 10,000 Births	95% Confidence Interval
Genitourinary: 455 cases					
Bladder Exstrophy	6	0	6	0.37	0.14—0.81
Cloacal Exstrophy	2	0	2	0.12	0.02—0.45
Hypospadias, 2nd or 3rd Degree	181	0	181	11.26	9.68—13.02
Obstructive Genitourinary Defect	147	2	149	9.27	7.84—10.88
Renal Agenesis/Hypoplasia	2	1	3	0.19	0.04—0.55
Other Genitourinary	191	3	194		
Musculoskeletal: 679 cases					
Club Foot	168	4	172	10.70	9.16—12.42
Craniosynostosis	88	0	88	5.47	4.39—6.74
Diaphragmatic Hernia	36	1	37	2.30	1.62—3.17
Gastroschisis	37	2	39	2.43	1.72—3.32
Omphalocele	24	7	31	1.93	1.31—2.74
Polydactyly/Syndactyly	190	2	192	11.94	10.31—13.75
Reduction Deformity, Lower Limbs	16	0	16	1.00	0.57—1.62
Reduction Deformity, Upper Limbs	40	5	45	2.80	2.04—3.74
Skeletal Dysplasia	27	1	28	1.74	1.16—2.52
Other Musculoskeletal	135	5	140		
Chromosomal and Other Syndromes: 439 cases					
Klinefelter Syndrome	7	1	8	0.50	0.21—0.98
Trisomy 13	8	1	9	0.56	0.26—1.06
Trisomy 18	17	5	22	1.37	0.86—2.07
Trisomy 21 (Down Syndrome)	194	6	200	12.44	10.77—14.29
Turner Syndrome	11	2	13	0.81	0.43—1.38
Other Chromosomal Syndromes/Other Syndromes	176	15	191		

Table 1 Prevalence of Birth Defects, Massachusetts: 2002-2003 (cont'd)

Defect[1]	Livebirth Count	Stillbirth Count	Total Count	Rate per 10,000 Births	95% Confidence Interval
Other: 30 cases					
Amniotic Bands	9	1	10	0.62	0.3—1.14
Skin Anomalies	7	0	7	0.44	0.18 - 0.90
Other, Specified	12	1	13		

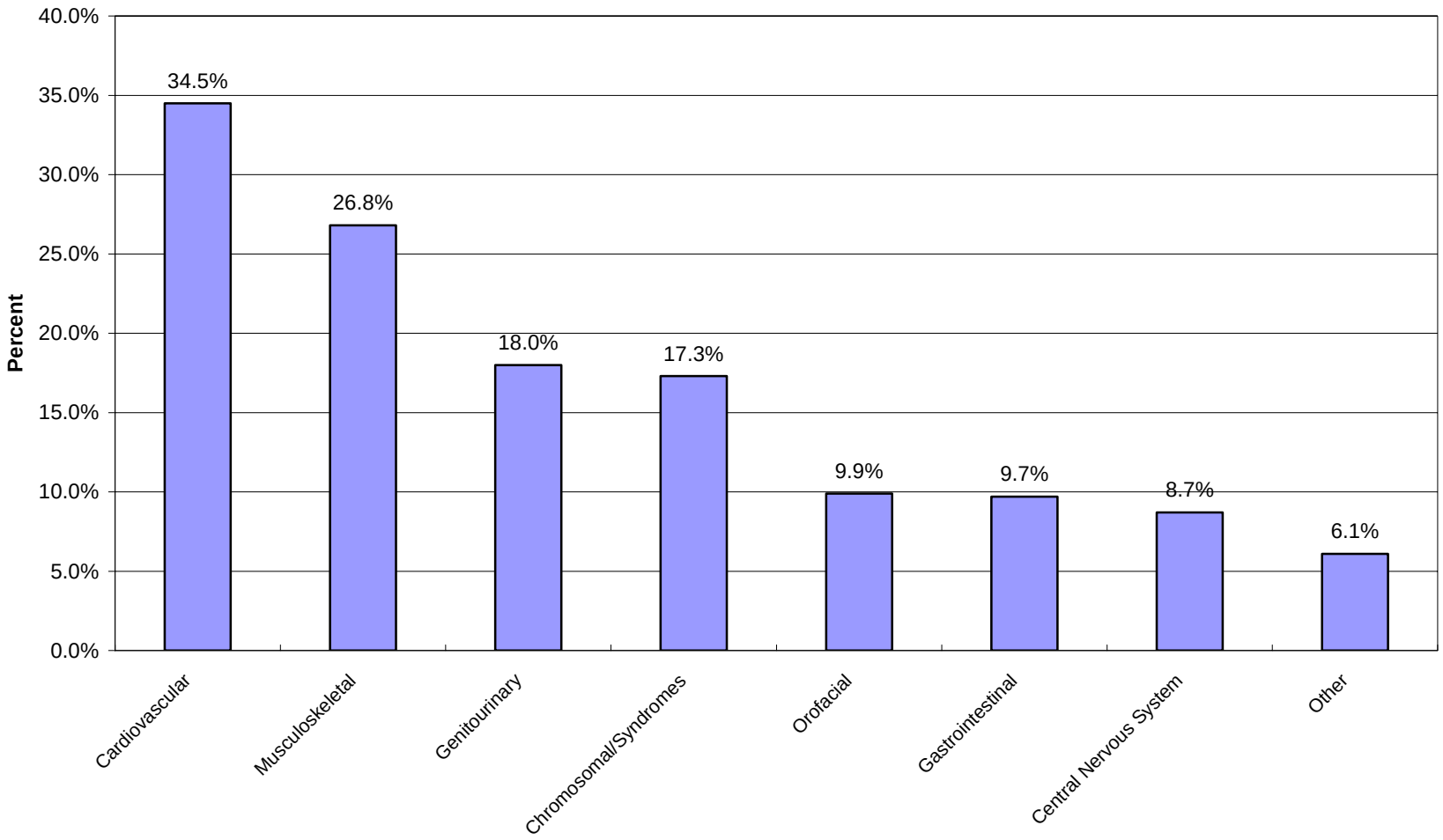
1. Cases can be included in more than one defect category. Cases are counted once within a defect category. Counts represent the number of defects, not the total number of cases with birth defects.

Table 2 Comparison of Selected Massachusetts 2002-2003 Birth Defect Rates to National Estimates[1]

Defect	Count	Rate per 10,000 Births MA	95% Confidence Interval	Rate per 10,000 Births US	95% Confidence Interval
Anencephaly[2]	6	0.37	0.14-0.81	3.45	3.27-3.64
Spina Bifida[2]	26	1.62	1.06-2.37	4.10	3.90-4.30
Anophthalmia/Microphthalmia	15	0.93	0.52-1.54	2.08	1.90-2.27
Truncus Arteriosus (common truncus)	3	0.19	0.04-0.55	0.82	0.71-0.93
Transposition of the Great Arteries[3]	59	3.67	2.79-4.73	4.73	4.47-5.00
Tetralogy of Fallot	65	4.04	3.12-5.15	3.92	3.67-4.17
Atrioventricular Septal Defect (Endocardial Cushion Defect)[4]	85	5.29	4.22-6.54	4.35	4.10-4.62
Hypoplastic Left Heart Syndrome	20	1.24	0.76-1.92	2.43	2.24-2.63
Cleft Palate without Cleft Lip	95	5.90	4.78-7.22	6.39	6.08-6.71
Cleft Lip with and without Cleft Palate	111	6.90	5.68-8.31	10.48	10.08-10.88
Esophageal Atresia/Tracheoesophageal Fistula	39	2.43	1.72-3.32	2.37	2.18-2.56
Rectal and Large Intestinal Atresia/Stenosis	64	3.98	3.07-5.08	4.81	4.54-5.08
Reduction Deformity, Upper Limbs	45	2.80	2.04-3.74	3.79	3.55-4.03
Reduction Deformity, Lower Limbs	16	1.00	0.57-1.62	1.90	1.73-2.07
Gastroschisis	39	2.43	1.72-3.32	3.73	3.49-3.97
Omphalocele	31	1.93	1.31-2.74	2.09	1.91-2.27
Diaphragmatic Hernia	37	2.30	1.62-3.17	2.94	2.73-3.15
Trisomy 21 (Down Syndrome)	200	12.44	10.77-14.29	13.65	13.19-14.12
Trisomy 13	9	0.56	0.26-1.06	1.33	1.18-1.47
Trisomy 18	22	1.37	0.86-2.07	2.41	2.22-2.61

1. All US rates except for Anencephaly and Spina Bifida are from MMWR report presenting 18 defects with improved national prevalence estimates. (MMWR, 2006).
2. US rates for Anencephaly and Spina Bifida are based on MMWR report detailing average prevalence rates from 23 active surveillance systems. These rates are from surveillance systems that include prenatally diagnosed and terminated pregnancies. (MMWR, 2004)
3. Includes d-TGA and L-TGA.
4. Includes ASD Primum, Common Atrium, CAVC, Endocardial Cushion Defect OS and NOS, and VSD canal type.

Figure 1 Birth Defect Cases by Category, Massachusetts: 2002-2003



Many cases have multiple defects, so percentages add up to more than 100%.
Total Number of Cases = 2,531.

Table 3 Most Common Defects, among Live Births and Stillbirths, Massachusetts: 2002-2003

Defect[1]	Category	Count	Rate per 10,000 Births	95% Confidence Interval
ASD (Secundum and NOS)	Cardiovascular	313	19.47	17.37-21.74
Trisomy 21 (Down Syndrome)	Chromosomal and Other Syndromes	200	12.44	10.77-14.29
Polydactyly/Syndactyly	Musculoskeletal	192	11.94	10.31-13.75
Hypospadias, 2nd or 3rd Degree	Genitourinary	181	11.26	9.68-13.02
VSD (Membranous and NOS)	Cardiovascular	179	11.13	9.56-12.89
Club Foot	Musculoskeletal	172	10.70	9.16-12.42
Obstructive Genitourinary Defect	Genitourinary	149	9.27	7.84-10.88
Cleft Lip w/ and w/o Cleft Palate	Orofacial	111	6.90	5.68 - 8.31
Pulmonary Stenosis, Valvular	Cardiovascular	105	6.53	5.34 - 7.91
Cleft Palate w/o Cleft Lip	Orofacial	95	5.91	4.78 - 7.22

1. Excludes Patent Ductus Arteriosus (PDA) due to the high number of cases and the mild severity of the majority of these cases.

Table 4 Single vs. Multiple Defects among Live Births and Stillbirths, Massachusetts: 2002-2003

Defect[1]	Cases with one defect	Cases with two or more defects[2]	Total Cases
Central Nervous System: 220 cases			
Anencephaly	5	1	6
Encephalocele	2	4	6
Holoprosencephaly	1	7	8
Hydrocephaly w/o Spina Bifida	17	38	55
Microcephaly	8	24	32
Spina Bifida w/ and w/o Hydrocephaly	10	16	26
Spinal Cord	15	39	54
Other CNS	18	72	90
Eye: 62 cases			
Aniridia	2	2	4
Anophthalmia/Microphthalmia	2	13	15
Congenital Glaucoma, Congenital Cataract	18	13	31
Other Eye	4	21	25
Ear: 19 cases			
Anotia/Microtia	10	9	19

Table 4 (cont'd) Single vs. Multiple Defects among Live Births and Stillbirths, Massachusetts: 2002-2003

Defect[1]	Cases with one defect	Cases with two or more defects[2]	Total Cases
Cardiovascular: 874 cases			
Anomalous Pulmonary Venous Connection			
Total/Partial Anomalous Pulmonary Venous Connection	5	16	21
Atrioventricular Canal Defects			
ASD Primum	0	14	14
Common Atrium	0	4	4
Complete Atrioventricular Canal Defect	3	44	47
Endocardial Cushion (OS and NOS)	0	9	9
VSD, Canal Type	1	10	11
Conotruncal (Outlet) and Aortic Arch			
Double Outlet Right Ventricle	4	16	20
d-Transposition of the Great Arteries	18	33	51
Interrupted Aortic Arch, Type B	0	4	4
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	17	48	65
Truncus	0	3	3
Ebstein Anomaly			
Ebstein Anomaly	2	5	7
Laterality Defects			
Heterotaxy, Situs Inversus	2	11	13
Left-Sided Obstruction			
Aortic Valve Stenosis	2	32	34
Coarctation of Aorta	12	62	74
Hypoplastic Left Heart Syndrome	5	15	20
Interrupted Aortic Arch (Type A and NOS)	0	4	4
Patent Ductus Arteriosus			
Patent Ductus Arteriosus	10	259	269
Right-Sided Obstruction			
Pulmonary Stenosis, Valvular	33	72	105
Pulmonary Valve Atresia w/intact septum	4	10	14
Pulmonary Valve Atresia with VSD	1	5	6

Table 4 Single vs. Multiple Defects among Live Births and (cont'd) Stillbirths, Massachusetts: 2002-2003

Defect[1]	Cases with one defect	Cases with two or more defects[2]	Total Cases
Tricuspid Valve Atresia	0	13	13
Septal Defects			
ASD (Secundum and NOS)	60	253	313
VSD (Membranous and NOS)	61	118	179
VSD, Conoventricular/Malalignment	8	22	30
Single Ventricle and L-TGA			
L-TGA	0	8	8
Single Ventricle	0	2	2
Other Cardiovascular			
Other Cardiovascular	24	236	260
Respiratory: 43 cases			
Choanal Atresia	3	1	4
Lung Anomalies	14	12	26
Other Respiratory	4	10	14
Orofacial: 251 cases			
Cleft Lip w/ and w/o Cleft Palate	81	30	111
Cleft Palate w/o Cleft Lip	32	63	95
Pierre Robin Sequence	0	35	35
Other Orofacial	20	32	52

Table 4 (cont'd) Single vs. Multiple Defects among Live Births and Stillbirths, Massachusetts: 2002-2003

Defect[1]	Cases with one defect	Cases with two or more defects[2]	Total Cases
Gastrointestinal: 245 cases			
Biliary Atresia	5	5	10
Esophageal Atresia/Tracheoesophageal Fistula	10	29	39
Hirschsprung Disease	13	5	18
Rectal and Large Intestinal Atresia/Stenosis	27	37	64
Small Intestinal Atresia	18	27	45
Other Gastrointestinal	42	48	90
Genitourinary: 455 cases			
Bladder Exstrophy	4	2	6
Cloacal Exstrophy	0	2	2
Hypospadias, 2nd or 3rd Degree	155	26	181
Obstructive Genitourinary Defect	17	132	149
Renal Agenesis/Hypoplasia	1	2	3
Other Genitourinary	68	126	194

Table 4 (cont'd) Single vs. Multiple Defects among Live Births and Stillbirths, Massachusetts: 2002-2003

Defect[1]	Cases with one defect	Cases with two or more defects[2]	Total Cases
Musculoskeletal: 679 cases			
Club Foot	107	65	172
Craniosynostosis	71	17	88
Diaphragmatic Hernia	18	19	37
Gastroschisis	25	14	39
Omphalocele	5	26	31
Polydactyly/Syndactyly	80	112	192
Reduction Deformity, Lower Limbs	3	13	16
Reduction Deformity, Upper Limbs	19	26	45
Skeletal Dysplasia	23	5	28
Other Musculoskeletal	15	125	140
Chromosomal and Other Syndromes: 439 cases			
Klinefelter Syndrome	6	2	8
Trisomy 13	1	8	9
Trisomy 18	3	19	22
Trisomy 21 (Down Syndrome)	82	118	200
Turner Syndrome	6	7	13
Other Chromosomal Syndromes/Other Syndromes	53	138	191
Other: 30 cases			
Amniotic Bands	1	9	10
Skin Anomalies	4	3	7
Other, Specified	2	11	13

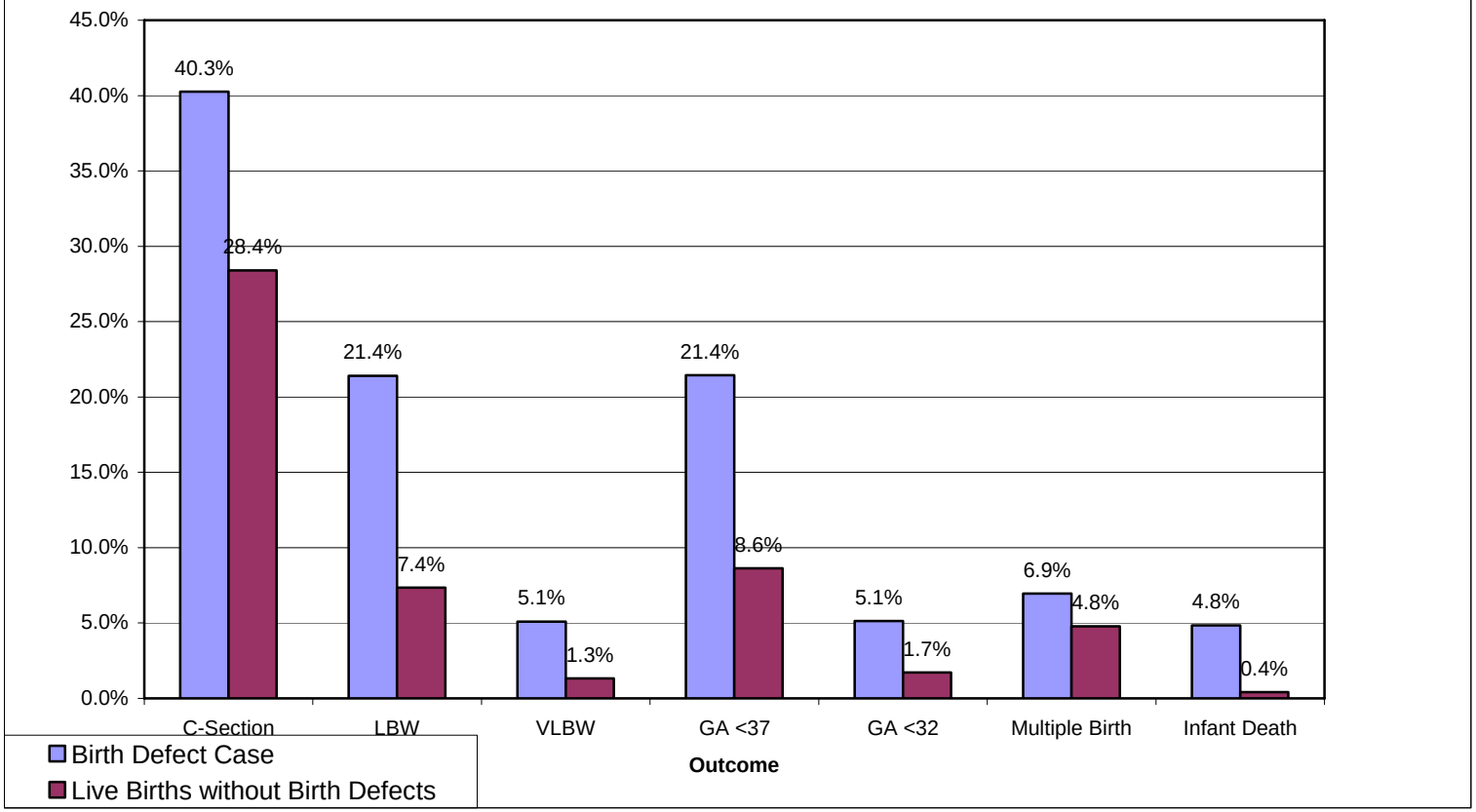
Table 4 (cont'd) Single vs. Multiple Defects among Live Births and Stillbirths, Massachusetts: 2002-2003

Defect[1]	Cases with one defect	Cases with two or more defects[2]	Total Cases

1. Cases can be included in more than one defect category. Cases are counted once within a defect category. Counts represent the number of defects, not the total number of cases with birth defects.

2. A case was defined as having multiple defects, if it had more than one defect from among those that were included in this surveillance report.

Figure 2 Pregnancy Outcomes: Birth Defect Cases Compared to Unaffected Live Births, Massachusetts: 2002-2003



Chapter 4

Prevalence of Birth Defects by Plurality and Sex

Plurality

Table 5 shows the distribution of birth defects by plurality. The overall prevalence was 152.77 for singletons and 247.71 for multiple births (more than one infant) per 10,000 live births. While multiple births comprised 4.8% of all live births, they comprised 7.4% of birth defects cases among live births (see Figure 3). Birth defects that occurred more frequently in multiple births included Esophageal Atresia/Tracheoesophageal Fistula, Hypospadias, Coarctation of Aorta, Diaphragmatic Hernia and Polydactyly/Syndactyly. Figure 4 presents rates for selected birth defects for singletons and multiples. Table 6 lists the most common defects among singletons and multiples. Examining birth defects by plurality is important since the number of multiple births has been increasing over time in Massachusetts.

Sex

Table 7 presents the prevalence of birth defects by sex of infant. The overall prevalence was 128.79 for females and 184.71 for males per 10,000 live births. While the prevalence of most types of birth defects did not substantially differ by sex of the infant/fetus, some conditions were associated with sex. Table 8 shows the most common birth defects for females and males. The most common defects seen in males were Hypospadias, Atrial Septal Defects (Secundum and NOS), Polydactyly/Syndactyly, Clubfoot, Down Syndrome and Obstructive Genitourinary Defects. The most common defects seen in females were Atrial Septal Defects (Secundum and NOS), Down Syndrome, Pulmonary Stenosis (Valvular), Polydactyly/Syndactyly and Clubfoot. Selected birth defects by sex of infant are presented in Figure 5.

Table 5 Prevalance of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2002-2003

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
Central Nervous System: 220 cases				
Anencephaly	singleton	4	0.26	0.07 — 0.67
	multiple	2	2.58	0.31 — 9.32
Encephalocele	singleton	5	0.33	0.11 — 0.76
	multiple	1	1.29	0.03 — 7.19
Holoprosencephaly	singleton	7	0.46	0.18 — 0.94
	multiple	1	1.29	0.03 — 7.19
Hydrocephaly w/o Spina Bifida	singleton	52	3.40	2.54 — 4.46
	multiple	3	3.87	0.80 — 11.31
Microcephaly	singleton	30	1.96	1.32 — 2.80
	multiple	2	2.58	0.31 — 9.32
Spina Bifida w/ and w/o Hydrocephaly	singleton	24	1.57	1.00 — 2.33
	multiple	2	2.58	0.31 — 9.32
Spinal Cord	singleton	52	3.40	2.54 — 4.46
	multiple	2	2.58	0.31 — 9.32
Other CNS	singleton	83	5.42	4.32 — 6.72
	multiple	7	9.03	3.63 — 18.61
Eye: 62 cases				
Aniridia	singleton	4	0.26	0.07 — 0.67
	multiple	0	0.00	0.00 — 4.76
Anophthalmia/Microphthalmia	singleton	13	0.85	0.45 — 1.45
	multiple	2	2.58	0.31 — 9.32
Congenital Glaucoma, Congenital Cataract	singleton	28	1.83	1.22 — 2.64
	multiple	3	3.87	0.80 — 11.31
Other Eye	singleton	23	1.50	0.95 — 2.26
	multiple	2	2.58	0.31 — 9.32
Ear: 19 cases				
Anotia/Microtia	singleton	15	0.98	0.55 — 1.62
	multiple	4	5.16	1.41 — 13.21

Table 5 Prevalance of Birth Defects by Plurality[1] of Live (cont'd) Births and Stillbirths, Massachusetts: 2002-2003

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
Cardiovascular: 874 cases				
Anomalous Pulmonary Venous Connection				
Total/Partial Anomalous Pulmonary Venous Connection	singleton	20	1.31	0.80 — 2.02
	multiple	1	1.29	0.03 — 7.19
Atrioventricular Canal Defects				
ASD Primum	singleton	14	0.91	0.50 — 1.53
	multiple	0	0.00	0.00 — 4.76
Common Atrium	singleton	4	0.26	0.07 — 0.67
	multiple	0	0.00	0.00 — 4.76
Complete Atrioventricular Canal Defect	singleton	45	2.94	2.14 — 3.93
	multiple	2	2.58	0.31 — 9.32
Endocardial Cushion (OS and NOS)	singleton	9	0.59	0.27 — 1.12
	multiple	0	0.00	0.00 — 4.76
VSD, Canal Type	singleton	8	0.52	0.23 — 1.03
	multiple	3	3.87	0.80 — 11.31
Conotruncal (Outlet) and Aortic Arch				
Double Outlet Right Ventricle	singleton	20	1.31	0.80 — 2.02
	multiple	0	0.00	0.00 — 4.76
d-Transposition of the Great Arteries	singleton	49	3.20	2.37 — 4.23
	multiple	2	2.58	0.31 — 9.32
Interrupted Aortic Arch, Type B	singleton	4	0.26	0.07 — 0.67
	multiple	0	0.00	0.00 — 4.76
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	singleton	64	4.18	3.22 — 5.34
	multiple	1	1.29	0.03 — 7.19
Truncus	singleton	2	0.13	0.02 — 0.47
	multiple	1	1.29	0.03 — 7.19
Ebstein Anomaly				
Ebstein Anomaly	singleton	7	0.46	0.18 — 0.94
	multiple	0	0.00	0.00 — 4.76
Laterality Defects				
Heterotaxy, Situs Inversus	singleton	13	0.85	0.45 — 1.45
	multiple	0	0.00	0.00 — 4.76
Left-Sided Obstruction				
Aortic Valve Stenosis	singleton	32	2.09	1.43 — 2.95
	multiple	2	2.58	0.31 — 9.32

Table 5 Prevalance of Birth Defects by Plurality[1] of Live (cont'd) Births and Stillbirths, Massachusetts: 2002-2003

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
Coarctation of Aorta	singleton	64	4.18	3.22 — 5.34
	multiple	10	12.90	6.19 — 23.73
Hypoplastic Left Heart Syndrome	singleton	19	1.24	0.75 — 1.94
	multiple	1	1.29	0.03 — 7.19
Interrupted Aortic Arch (Type A and NOS)	singleton	3	0.20	0.04 — 0.57
	multiple	1	1.29	0.03 — 7.19
Patent Ductus Arteriosus				
Patent Ductus Arteriosus	singleton	258	16.86	14.86 — 19.05
	multiple	11	14.19	7.08 — 25.39
Right-Sided Obstruction				
Pulmonary Stenosis, Valvular	singleton	94	6.14	4.96 — 7.52
	multiple	11	14.19	7.08 — 25.39
Pulmonary Valve Atresia w/intact septum	singleton	11	0.72	0.36 — 1.29
	multiple	3	3.87	0.80 — 11.31
Pulmonary Valve Atresia with VSD	singleton	6	0.39	0.14 — 0.85
	multiple	0	0.00	0.00 — 4.76
Tricuspid Valve Atresia	singleton	13	0.85	0.45 — 1.45
	multiple	0	0.00	0.00 — 4.76
Septal Defects				
ASD (Secundum and NOS)	singleton	294	19.21	17.08 — 21.54
	multiple	19	24.51	14.76 — 38.28
VSD (Membranous and NOS)	singleton	167	10.91	9.32 — 12.70
	multiple	12	15.48	8.00 — 27.04
VSD, Conoventricular/Malalignment	singleton	28	1.83	1.22 — 2.64
	multiple	2	2.58	0.31 — 9.32
Single Ventricle and L-TGA				
L-TGA	singleton	7	0.46	0.18 — 0.94
	multiple	1	1.29	0.03 — 7.19
Single Ventricle	singleton	2	0.13	0.02 — 0.47
	multiple	0	0.00	0.00 — 4.76
Other Cardiovascular				
Other Cardiovascular	singleton	236	15.42	13.52 — 17.52
	multiple	24	30.96	19.84 — 46.07

Table 5 Prevalance of Birth Defects by Plurality[1] of Live (cont'd) Births and Stillbirths, Massachusetts: 2002-2003

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
Respiratory: 43 cases				
Choanal Atresia	singleton	3	0.20	0.04 — 0.57
	multiple	1	1.29	0.03 — 7.19
Lung Anomalies	singleton	25	1.63	1.06 — 2.41
	multiple	1	1.29	0.03 — 7.19
Other Respiratory	singleton	13	0.85	0.45 — 1.45
	multiple	1	1.29	0.03 — 7.19
Orofacial: 251 cases				
Cleft Lip w/ and w/o Cleft Palate	singleton	107	6.99	5.73 — 8.45
	multiple	4	5.16	1.41 — 13.21
Cleft Palate w/o Cleft Lip	singleton	87	5.68	4.55 — 7.01
	multiple	8	10.32	4.46 — 20.34
Pierre Robin Sequence	singleton	32	2.09	1.43 — 2.95
	multiple	3	3.87	0.80 — 11.31
Other Orofacial	singleton	48	3.14	2.31 — 4.16
	multiple	4	5.16	1.41 — 13.21
Gastrointestinal: 245 cases				
Biliary Atresia	singleton	9	0.59	0.27 — 1.12
	multiple	1	1.29	0.03 — 7.19
Esophageal Atresia/Tracheoesophageal Fistula	singleton	35	2.29	1.59 — 3.18
	multiple	4	5.16	1.41 — 13.21
Hirschsprung Disease	singleton	17	1.11	0.65 — 1.78
	multiple	1	1.29	0.03 — 7.19
Rectal and Large Intestinal Atresia/Stenosis	singleton	61	3.99	3.05 — 5.12
	multiple	3	3.87	0.80 — 11.31
Small Intestinal Atresia	singleton	39	2.55	1.81 — 3.48
	multiple	6	7.74	2.84 — 16.85
Other Gastrointestinal	singleton	86	5.62	4.49 — 6.94
	multiple	4	5.16	1.41 — 13.21

Table 5 Prevalance of Birth Defects by Plurality[1] of Live (cont'd) Births and Stillbirths, Massachusetts: 2002-2003

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
Genitourinary: 455 cases				
Bladder Exstrophy	singleton	6	0.39	0.14 — 0.85
	multiple	0	0.00	0.00 — 4.76
Cloacal Exstrophy	singleton	2	0.13	0.02 — 0.47
	multiple	0	0.00	0.00 — 4.76
Hypospadias, 2nd or 3rd Degree	singleton	160	10.45	8.90 — 12.21
	multiple	21	27.09	16.77 — 41.41
Obstructive Genitourinary Defect	singleton	140	9.15	7.70 — 10.79
	multiple	9	11.61	5.31 — 22.04
Renal Agenesis/Hypoplasia	singleton	3	0.20	0.04 — 0.57
	multiple	0	0.00	0.00 — 4.76
Other Genitourinary	singleton	175	11.43	9.80 — 13.26
	multiple	19	24.51	14.76 — 38.28
Musculoskeletal: 679 cases				
Club Foot	singleton	158	10.32	8.78 — 12.07
	multiple	14	18.06	9.87 — 30.31
Craniosynostosis	singleton	84	5.49	4.38 — 6.80
	multiple	4	5.16	1.41 — 13.21
Diaphragmatic Hernia	singleton	30	1.96	1.32 — 2.80
	multiple	7	9.03	3.63 — 18.61
Gastroschisis	singleton	38	2.48	1.76 — 3.41
	multiple	1	1.29	0.03 — 7.19
Omphalocele	singleton	26	1.70	1.11 — 2.49
	multiple	5	6.45	2.09 — 15.05
Polydactyly/Syndactyly	singleton	176	11.50	9.86 — 13.33
	multiple	15	19.35	10.83 — 31.92
Reduction Deformity, Lower Limbs	singleton	15	0.98	0.55 — 1.62
	multiple	1	1.29	0.03 — 7.19
Reduction Deformity, Upper Limbs	singleton	40	2.61	1.87 — 3.56
	multiple	5	6.45	2.09 — 15.05
Skeletal Dysplasia	singleton	27	1.76	1.16 — 2.57
	multiple	1	1.29	0.03 — 7.19

Table 5 Prevalance of Birth Defects by Plurality[1] of Live (cont'd) Births and Stillbirths, Massachusetts: 2002-2003

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
Other Musculoskeletal	singleton	127	8.30	6.92 — 9.87
	multiple	13	16.77	8.93 — 28.68
Chromosomal and Other Syndromes: 439 cases				
Klinefelter Syndrome	singleton	8	0.52	0.23 — 1.03
	multiple	0	0.00	0.00 — 4.76
Trisomy 13	singleton	9	0.59	0.27 — 1.12
	multiple	0	0.00	0.00 — 4.76
Trisomy 18	singleton	18	1.18	0.70 — 1.86
	multiple	4	5.16	1.41 — 13.21
Trisomy 21 (Down Syndrome)	singleton	194	12.68	10.96 — 14.59
	multiple	6	7.74	2.84 — 16.85
Turner Syndrome	singleton	12	0.78	0.41 — 1.37
	multiple	1	1.29	0.03 — 7.19
Other Chromosomal Syndromes/Other Syndromes	singleton	175	11.43	9.80 — 13.26
	multiple	16	20.64	11.80 — 33.52
Other: 30 cases				
Amniotic Bands	singleton	9	0.59	0.27 — 1.12
	multiple	1	1.29	0.03 — 7.19
Skin Anomalies	singleton	6	0.39	0.14 — 0.85
	multiple	1	1.29	0.03 — 7.19
Other, Specified	singleton	11	0.72	0.36 — 1.29
	multiple	2	2.58	0.31 — 9.32

1. Plurality is the number of births to a woman from the same pregnancy. A singleton is the birth of one infant; multiple represents more than one infant.

2. Cases can be included in more than one defect category. Cases are counted once within a defect category. Counts represent the number of defects, not the total number of cases with birth defects.

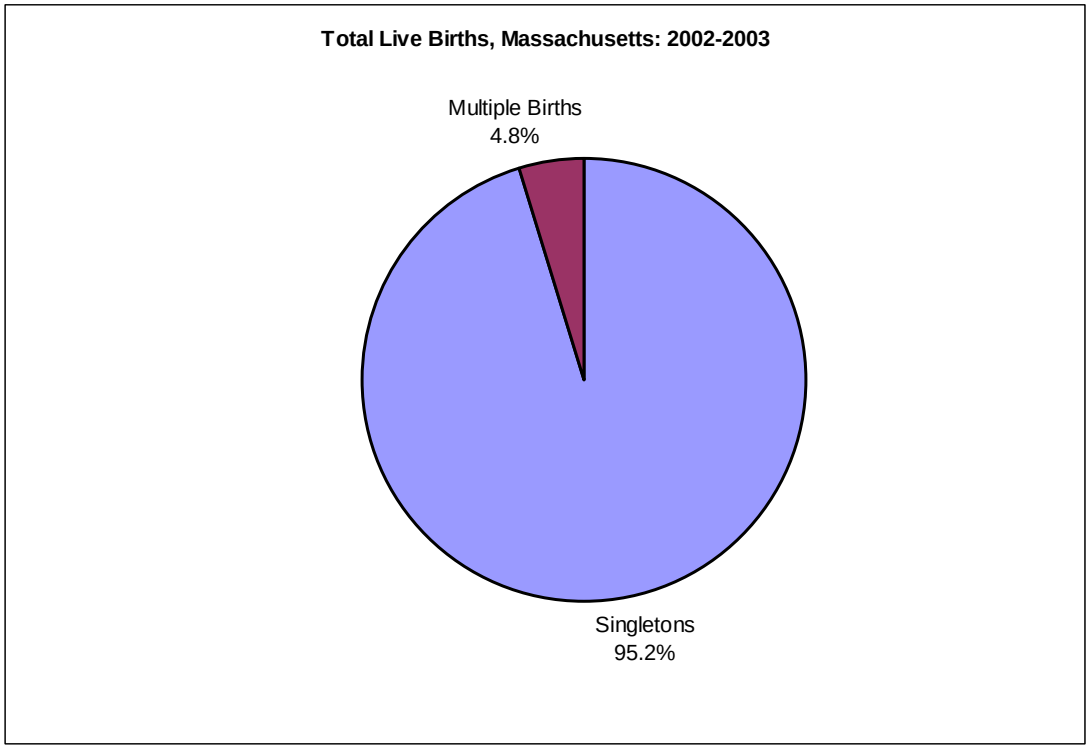
Table 6 Most Common Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts:2002-2003

Defect[2]	Count	Rate per 10,000 Births	95% Confidence Interval
MULTIPLE			
Hypospadias, 2nd or 3rd Degree	21	27.09	16.77 — 41.41
ASD (Secundum and NOS)	19	24.51	14.76 — 38.28
Polydactyly/Syndactyly	15	19.35	10.83 — 31.92
Club Foot	14	18.06	9.87 — 30.31
VSD (Membranous and NOS)	12	15.48	8.00 — 27.04
Pulmonary Stenosis, Valvular	11	14.19	7.08 — 25.39
Coarctation of Aorta	10	12.90	6.19 — 23.73
Obstructive Genitourinary Defect	9	11.61	5.31 — 22.04
Cleft Palate w/o Cleft Lip	8	10.32	4.46 — 20.34
Diaphragmatic Hernia	7	9.03	3.63 — 18.61
SINGLETON			
ASD (Secundum and NOS)	294	19.21	17.08 — 21.54
Trisomy 21 (Down Syndrome)	194	12.68	10.96 — 14.59
Polydactyly/Syndactyly	176	11.50	9.86 — 13.33
VSD (Membranous and NOS)	167	10.91	9.32 — 12.70
Hypospadias, 2nd or 3rd Degree	160	10.45	8.90 — 12.21
Club Foot	158	10.32	8.78 — 12.07
Obstructive Genitourinary Defect	140	9.15	7.70 — 10.79
Cleft Lip w/ and w/o Cleft Palate	107	6.99	5.73 — 8.45
Pulmonary Stenosis, Valvular	94	6.14	4.96 — 7.52
Cleft Palate w/o Cleft Lip	87	5.68	4.55 — 7.01

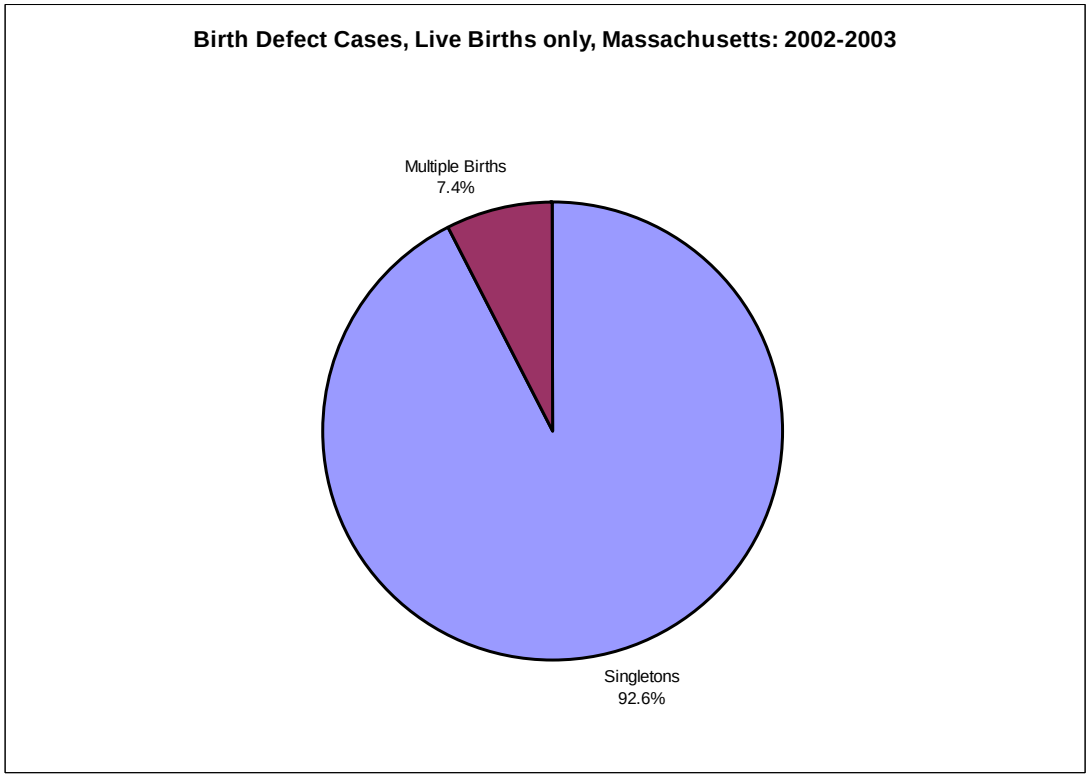
1. Plurality is the number of births to a woman from the same pregnancy. A singleton is the birth of one infant; multiple represents more than one infant.

2. Excludes Patent Ductus Arteriosus (PDA) due to the high number of cases and the mild severity of the majority of these cases.

Figure 3 Plurality of All Live Births and Birth Defect Cases, Live Births Only



N=160,791



N=2,476

Figure 4 Prevalence of Selected Birth Defects by Plurality among Live Births and Stillbirths, Massachusetts: 2002-2003

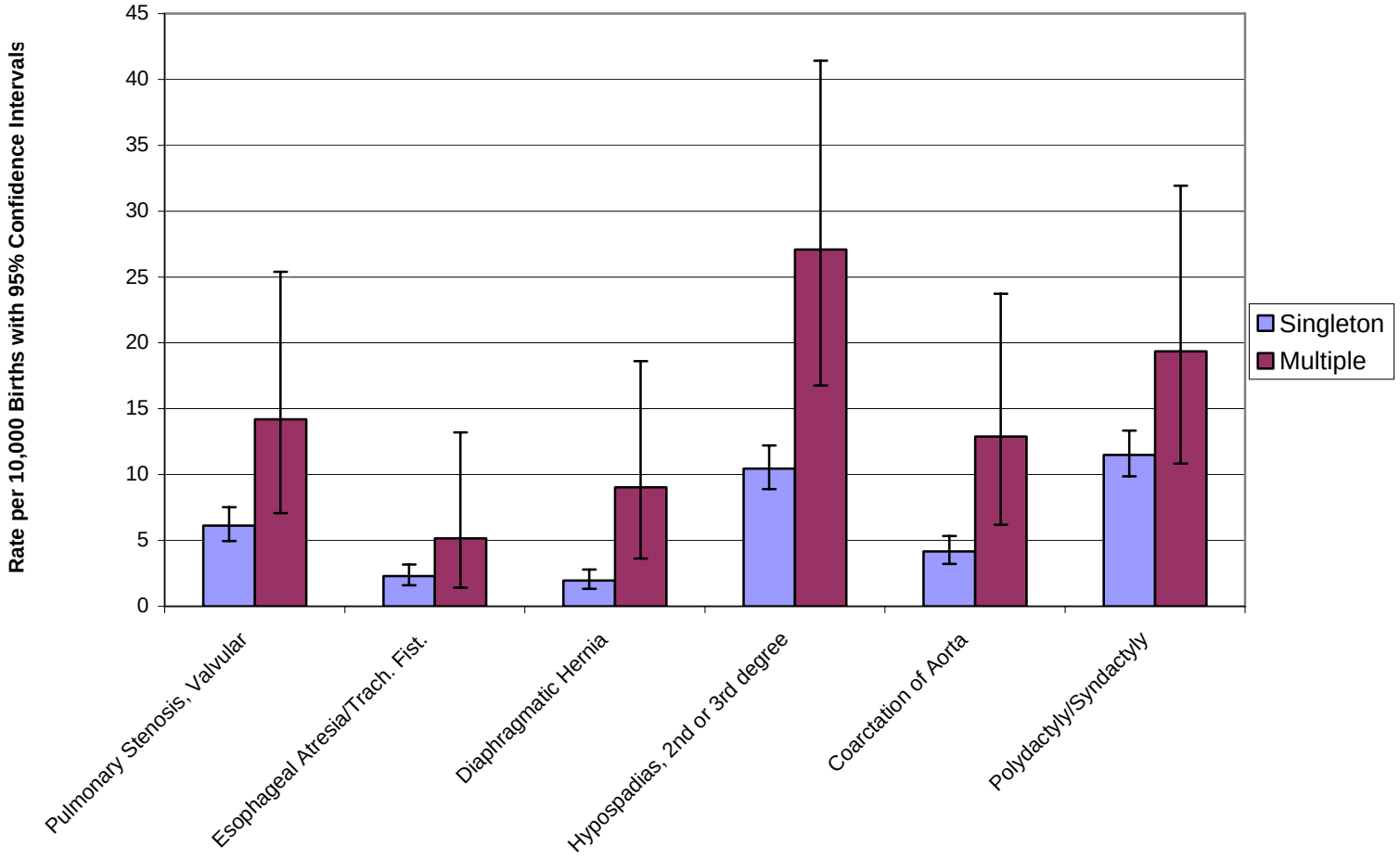


Table 7 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2002-2003

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
Central Nervous System: 220 cases				
Anencephaly	male	2	0.24	0.03 – 0.88
	female	4	0.51	0.14 – 1.30
Encephalocele	male	2	0.24	0.03 – 0.88
	female	4	0.51	0.14 – 1.30
Holoprosencephaly	male	3	0.36	0.08 – 1.07
	female	5	0.64	0.21 – 1.49
Hydrocephaly w/o Spina Bifida	male	33	4.01	2.76 – 5.63
	female	22	2.80	1.76 – 4.24
Microcephaly	male	15	1.82	1.02 – 3.01
	female	17	2.17	1.26 – 3.47
Spina Bifida w/ and w/o Hydrocephaly	male	15	1.82	1.02 – 3.01
	female	11	1.40	0.70 – 2.51
Spinal Cord	male	31	3.77	2.56 – 5.35
	female	23	2.93	1.86 – 4.40
Other CNS	male	54	6.56	4.93 – 8.56
	female	36	4.59	3.21 – 6.35
Eye: 62 cases				
Aniridia	male	2	0.24	0.03 – 0.88
	female	2	0.25	0.03 – 0.92
Anophthalmia/Microphthalmia	male	8	0.97	0.42 – 1.92
	female	7	0.89	0.36 – 1.84
Congenital Glaucoma, Congenital Cataract	male	13	1.58	0.84 – 2.70
	female	18	2.29	1.36 – 3.62
Other Eye	male	14	1.70	0.93 – 2.85
	female	11	1.40	0.70 – 2.51
Ear: 19 cases				
Anotia/Microtia	male	14	1.70	0.93 – 2.85
	female	5	0.64	0.21 – 1.49

Table 7 Prevalence of Birth Defects by Sex of Infant among Live (cont'd) Births and Stillbirths, Massachusetts: 2002-2003

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Inteval
Cardiovascular: 874 cases				
Anomalous Pulmonary Venous Connection				
Total/Partial Anomalous Pulmonary Venous Connection	male	14	1.70	0.93 – 2.85
	female	7	0.89	0.36 – 1.84
Atrioventricular Canal Defects				
ASD Primum	male	7	0.85	0.34 – 1.75
	female	7	0.89	0.36 – 1.84
Common Atrium	male	1	0.12	0.00 – 0.68
	female	3	0.38	0.08 – 1.12
Complete Atrioventricular Canal Defect	male	23	2.79	1.77 – 4.19
	female	24	3.06	1.96 – 4.55
Endocardial Cushion (OS and NOS)	male	3	0.36	0.08 – 1.07
	female	6	0.76	0.28 – 1.66
VSD, Canal Type	male	4	0.49	0.13 – 1.24
	female	7	0.89	0.36 – 1.84
Conotruncal (Outlet) and Aortic Arch				
Double Outlet Right Ventricle	male	12	1.46	0.75 – 2.55
	female	8	1.02	0.44 – 2.01
d-Transposition of the Great Arteries	male	32	3.89	2.66 – 5.49
	female	19	2.42	1.46 – 3.78
Interrupted Aortic Arch, Type B	male	1	0.12	0.00 – 0.68
	female	3	0.38	0.08 – 1.12
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	male	39	4.74	3.37 – 6.48
	female	26	3.31	2.16 – 4.85
Truncus	male	2	0.24	0.03 – 0.88
	female	1	0.13	0.00 – 0.71
Ebstein Anomaly				
Ebstein Anomaly	male	5	0.61	0.20 – 1.42
	female	2	0.25	0.03 – 0.92
Laterality Defects				
Heterotaxy, Situs Inversus	male	6	0.73	0.27 – 1.59
	female	7	0.89	0.36 – 1.84

Table 7 Prevalence of Birth Defects by Sex of Infant among Live (cont'd) Births and Stillbirths, Massachusetts: 2002-2003

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
Left-Sided Obstruction				
Aortic Valve Stenosis	male	19	2.31	1.39 – 3.61
	female	15	1.91	1.07 – 3.15
Coarctation of Aorta	male	43	5.23	3.78 – 7.04
	female	31	3.95	2.68 – 5.61
Hypoplastic Left Heart Syndrome	male	11	1.34	0.67 – 2.39
	female	9	1.15	0.52 – 2.18
Interrupted Aortic Arch (Type A and NOS)	male	2	0.24	0.03 – 0.88
	female	2	0.25	0.03 – 0.92
Patent Ductus Arteriosus				
Patent Ductus Arteriosus	male	127	15.43	12.87 – 18.36
	female	142	18.09	15.24 – 21.32
Right-Sided Obstruction				
Pulmonary Stenosis, Valvular	male	33	4.01	2.76 – 5.63
	female	72	9.17	7.18 – 11.55
Pulmonary Valve Atresia w/intact septum	male	9	1.09	0.50 – 2.08
	female	5	0.64	0.21 – 1.49
Pulmonary Valve Atresia with VSD	male	3	0.36	0.08 – 1.07
	female	3	0.38	0.08 – 1.12
Tricuspid Valve Atresia	male	7	0.85	0.34 – 1.75
	female	6	0.76	0.28 – 1.66
Septal Defects				
ASD (Secundum and NOS)	male	164	19.93	17.00 – 23.22
	female	149	18.98	16.06 – 22.29
VSD (Membranous and NOS)	male	86	10.45	8.36 – 12.91
	female	93	11.85	9.56 – 14.51
VSD, Conoventricular/Malalignment	male	14	1.70	0.93 – 2.85
	female	16	2.04	1.17 – 3.31
Single Ventricle and L-TGA				
L-TGA	male	4	0.49	0.13 – 1.24
	female	4	0.51	0.14 – 1.30
Single Ventricle	male	1	0.12	0.00 – 0.68
	female	1	0.13	0.00 – 0.71
Other Cardiovascular				
Other Cardiovascular	male	128	15.55	12.98 – 18.49
	female	132	16.82	14.07 – 19.94

Table 7 Prevalence of Birth Defects by Sex of Infant among Live (cont'd) Births and Stillbirths, Massachusetts: 2002-2003

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
Respiratory: 43 cases				
Choanal Atresia	male	2	0.24	0.03 – 0.88
	female	2	0.25	0.03 – 0.92
Lung Anomalies	male	15	1.82	1.02 – 3.01
	female	11	1.40	0.70 – 2.51
Other Respiratory	male	6	0.73	0.27 – 1.59
	female	8	1.02	0.44 – 2.01
Orofacial: 251 cases				
Cleft Lip w/ and w/o Cleft Palate	male	69	8.38	6.52 – 10.61
	female	42	5.35	3.86 – 7.23
Cleft Palate w/o Cleft Lip	male	46	5.59	4.09 – 7.46
	female	49	6.24	4.62 – 8.25
Pierre Robin Sequence	male	18	2.19	1.30 – 3.46
	female	17	2.17	1.26 – 3.47
Other Orofacial	male	25	3.04	1.97 – 4.48
	female	27	3.44	2.27 – 5.00
Gastrointestinal: 245 cases				
Biliary Atresia	male	3	0.36	0.08 – 1.07
	female	7	0.89	0.36 – 1.84
Esophageal Atresia/Tracheoesophageal Fistula	male	14	1.70	0.93 – 2.85
	female	25	3.18	2.06 – 4.70
Hirschsprung Disease	male	10	1.22	0.58 – 2.23
	female	8	1.02	0.44 – 2.01
Rectal and Large Intestinal Atresia/Stenosis	male	36	4.37	3.06 – 6.06
	female	28	3.57	2.37 – 5.16
Small Intestinal Atresia	male	26	3.16	2.06 – 4.63
	female	19	2.42	1.46 – 3.78
Other Gastrointestinal	male	47	5.71	4.20 – 7.59
	female	43	5.48	3.96 – 7.38

Table 7 Prevalence of Birth Defects by Sex of Infant among Live (cont'd) Births and Stillbirths, Massachusetts: 2002-2003

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
Genitourinary: 455 cases				
Bladder Exstrophy	male	4	0.49	0.13 – 1.24
	female	2	0.25	0.03 – 0.92
Cloacal Exstrophy	male	0	0.00	0.00 – 0.45
	female	2	0.25	0.03 – 0.92
Hypospadias, 2nd or 3rd Degree	male	181	21.99	18.91 – 25.44
	female	0	0.00	0.00 – 0.47
Obstructive Genitourinary Defect	male	107	13.00	10.66 – 15.71
	female	42	5.35	3.86 – 7.23
Renal Agenesis/Hypoplasia	male	2	0.24	0.03 – 0.88
	female	1	0.13	0.00 – 0.71
Other Genitourinary	male	128	15.55	12.98 – 18.49
	female	66	8.41	6.50 – 10.70
Musculoskeletal: 679 cases				
Club Foot	male	111	13.49	11.10 – 16.24
	female	61	7.77	5.94 – 9.98
Craniosynostosis	male	61	7.41	5.67 – 9.52
	female	27	3.44	2.27 – 5.00
Diaphragmatic Hernia	male	22	2.67	1.68 – 4.05
	female	15	1.91	1.07 – 3.15
Gastroschisis	male	32	3.89	2.66 – 5.49
	female	7	0.89	0.36 – 1.84
Omphalocele	male	17	2.07	1.20 – 3.31
	female	14	1.78	0.98 – 2.99
Polydactyly/Syndactyly	male	127	15.43	12.87 – 18.36
	female	65	8.28	6.39 – 10.55
Reduction Deformity, Lower Limbs	male	6	0.73	0.27 – 1.59
	female	10	1.27	0.61 – 2.34
Reduction Deformity, Upper Limbs	male	26	3.16	2.06 – 4.63
	female	19	2.42	1.46 – 3.78
Skeletal Dysplasia	male	15	1.82	1.02 – 3.01
	female	13	1.66	0.88 – 2.83
Other Musculoskeletal	male	73	8.87	6.95 – 11.15
	female	67	8.54	6.61 – 10.84

Table 7 Prevalence of Birth Defects by Sex of Infant among Live (cont'd) Births and Stillbirths, Massachusetts: 2002-2003

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
Chromosomal and Other Syndromes: 439 cases				
Klinefelter Syndrome	male	8	0.97	0.42 – 1.92
	female	0	0.00	0.00 – 0.47
Trisomy 13	male	7	0.85	0.34 – 1.75
	female	2	0.25	0.03 – 0.92
Trisomy 18	male	9	1.09	0.50 – 2.08
	female	13	1.66	0.88 – 2.83
Trisomy 21 (Down Syndrome)	male	107	13.00	10.66 – 15.71
	female	93	11.85	9.56 – 14.51
Turner Syndrome	male	1	0.12	0.00 – 0.68
	female	12	1.53	0.79 – 2.67
Other Chromosomal Syndromes/Other Syndromes	male	94	11.42	9.23 – 13.98
	female	97	12.36	10.02 – 15.07
Other: 30 cases				
Amniotic Bands	male	5	0.61	0.20 – 1.42
	female	5	0.64	0.21 – 1.49
Skin Anomalies	male	3	0.36	0.08 – 1.07
	female	4	0.51	0.14 – 1.30
Other, Specified	male	7	0.85	0.34 – 1.75
	female	6	0.76	0.28 – 1.66

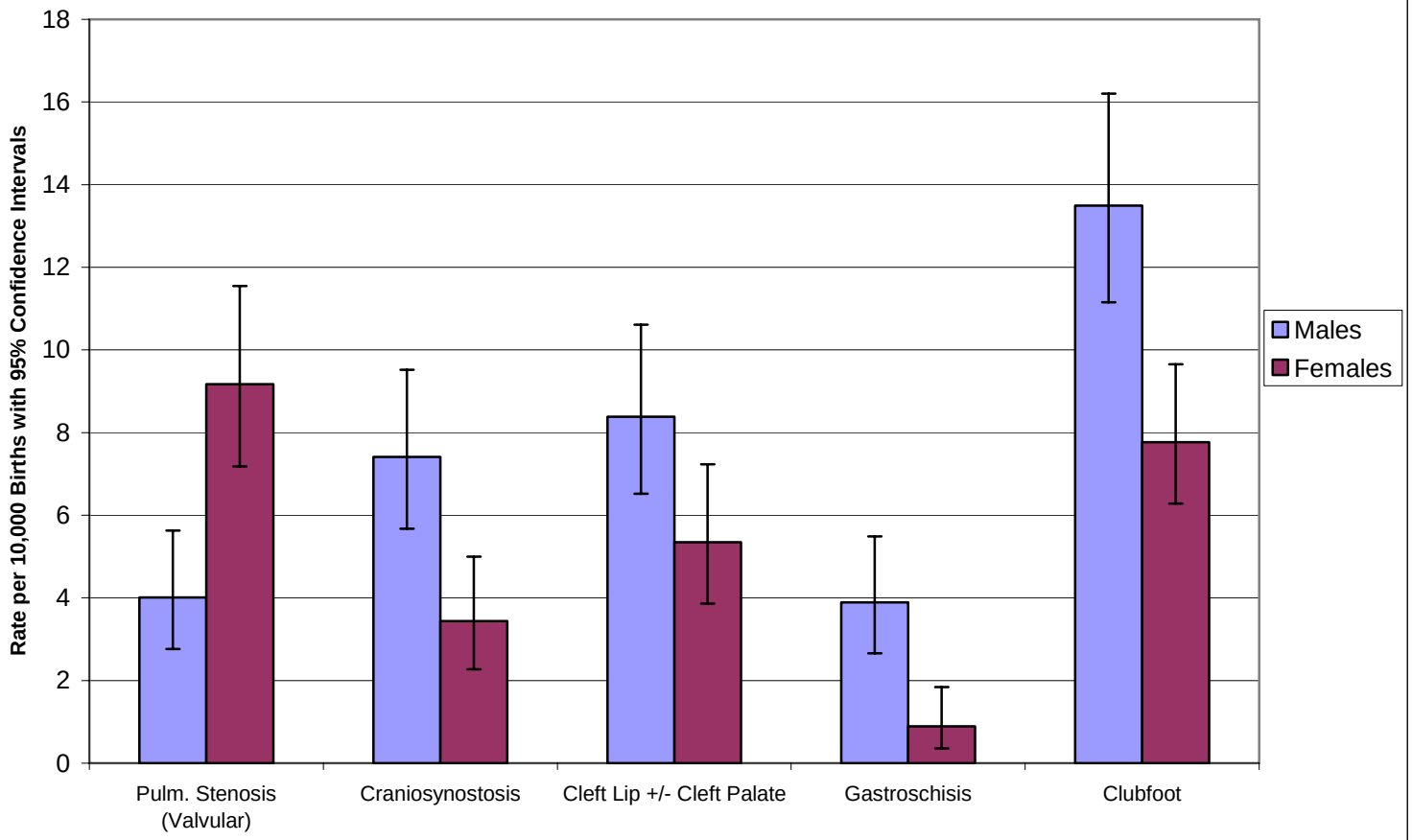
1. Cases can be included in more than one defect category. Cases are counted once within a defect category. Counts represent the number of defects, not the total number of cases with birth defects.

Table 8 Most Common Defects by Sex of Live Births and Stillbirths, Massachusetts: 2002-2003

Defect[1]	Count	Rate per 10,000 Births	95% Confidence Interval
FEMALE			
ASD (Secundum and NOS)	149	18.98	16.06 — 22.29
VSD (Membranous and NOS)	93	11.85	9.56 — 14.51
Trisomy 21 (Down Syndrome)	93	11.85	9.56 — 14.51
Pulmonary Stenosis, Valvular	72	9.17	7.18 — 11.55
Polydactyly/Syndactyly	65	8.28	6.39 — 10.55
Club Foot	61	7.77	5.94 — 9.98
Cleft Palate w/o Cleft Lip	49	6.24	4.62 — 8.25
Obstructive Genitourinary Defect	42	5.35	3.86 — 7.23
Cleft Lip w/ and w/o Cleft Palate	42	5.35	3.86 — 7.23
Coarctation of Aorta	31	3.95	2.68 — 5.61
MALE			
Hypospadias, 2nd or 3rd Degree	181	21.99	18.91 — 25.44
ASD (Secundum and NOS)	164	19.93	17.00 — 23.22
Polydactyly/Syndactyly	127	15.43	12.87 — 18.36
Club Foot	111	13.49	11.10 — 16.24
Obstructive Genitourinary Defect	107	13.00	10.66 — 15.71
Trisomy 21 (Down Syndrome)	107	13.00	10.66 — 15.71
VSD (Membranous and NOS)	86	10.45	8.36 — 12.91
Cleft Lip w/ and w/o Cleft Palate	69	8.38	6.52 — 10.61
Craniosynostosis	61	7.41	5.67 — 9.52
Cleft Palate w/o Cleft Lip	46	5.59	4.09 — 7.46

1. Excludes Patent Ductus Arteriosus (PDA) due to the high number of cases and the mild severity of the majority of these cases.

Figure 5 Prevalence of Selected Birth Defects By Sex of Infant among Live Births and Stillbirths, Massachusetts: 2002-2003



Chapter 5

Prevalence of Birth Defects by Maternal Age and Race / Ethnicity

Maternal Age

The prevalence of birth defects varied by maternal age. For live births only, rates per 10,000 live births were 143.46 for mothers younger than 20 years, 147.64 for those 20-24 years, 141.66 for those 25-29 years, 146.69 for those 30-34 years, and 183.98 for those 35 years and older. Table 9 shows the rates for birth defects by maternal age.

As expected, there was a strong association of Down Syndrome with advanced maternal age (see Figure 6). Although more babies with Down Syndrome are born to women under 35, the Down Syndrome rate of 29.16 per 10,000 births for women 35 years and older was four times that of any other maternal age group. Figure 7 shows that younger mothers (aged 19 and under) had the highest rate (12.75) of Gastroschisis cases. This association has been shown in previous studies (Forrester 1997). Mothers younger than 25 years of age had babies with higher rates of Gastroschisis, Double Outlet Right Ventricle, Heterotaxy, and Cleft Lip with and without Cleft Palate than other age groups. Older mothers had higher rates for many defects including Esophageal Atresia/Tracheoesophageal Fistula, Hypospadias, Tetratology of Fallot, and many Syndromes. While results for other defects also differed by age group, the small numbers from two years of surveillance were not sufficient for interpretation.

Table 10 displays the most common birth defects for live births by maternal age groups. Atrial Septal Defects and Ventricular Septal Defects were common to all maternal age groups. Polydactyly/Syndactyly and Club Foot (except for mothers 25-29 years) were among the top five most common in every age group.

Monitoring birth defects by maternal age is important since the number of births to older mothers has been increasing over time in Massachusetts. Birth rates for women ages 30+ have increased steadily from 1980 to 2003. Births to every age group above 30 have increased since 1990 while births to age groups below 30 have decreased. (Massachusetts Births 2003)

Maternal Race / Hispanic Ethnicity

Table 11 shows the variation in prevalence of birth defects by maternal race and Hispanic ethnicity. The rate per 10,000 live births was 157.85 for Non-Hispanic Whites, 142.62 for Non-Hispanic Blacks, 155.38 for Hispanics, and 114.03 for Asians/Pacific Islanders. The rates for other races were not calculated due to small numbers. Table 12 shows the more common defects by maternal race and Hispanic ethnicity. The most common defects in Hispanics included Septal Defects, Polydactyly/Syndactyly, Clubfoot and Down Syndrome. In Blacks, the most common defects included Septal Defects, Down Syndrome, Polydactyly/Syndactyly, Pulmonary Stenosis (Valvular) and Hypospadias. The most common defects in Whites included Septal Defects, Hypospadias, Down Syndrome, Polydactyly/Syndactyly and Clubfoot. In Asians, the most common defects included Septal Defects, Clubfoot, Cleft Lip, Cleft Palate, and Down Syndrome.

Multiple factors likely contribute to differences in prevalence by racial and ethnic groups including genetic variation, diet and lifestyle differences, differential access or use of health care services, or socioeconomic differences. More years of data and in-depth studies are needed to affirm the stability of these rates and to understand racial and ethnic patterns.

Table 9 Prevalence of Birth Defects by Maternal Age for Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Central Nervous System				
Anencephaly	<20	0	0.00	0.00–3.92
	20-24	1	0.42	0.01–2.34
	25-29	1	0.27	0.01–1.51
	30-34	1	0.19	0.00–1.03
	35+	0	0.00	0.00–1.01
Encephalocele	<20	0	0.00	0.00–3.92
	20-24	0	0.00	0.00–1.55
	25-29	1	0.27	0.01–1.51
	30-34	3	0.56	0.11–1.62
	35+	2	0.55	0.07–1.97
Holoprosencephaly	<20	0	0.00	0.00–3.92
	20-24	1	0.42	0.01–2.34
	25-29	1	0.27	0.01–1.51
	30-34	3	0.56	0.11–1.62
	35+	2	0.55	0.07–1.97
Hydrocephaly w/o Spina Bifida	<20	2	2.13	0.26–7.68
	20-24	11	4.63	2.31–8.28
	25-29	9	2.44	1.11–4.63
	30-34	24	4.45	2.85–6.61
	35+	7	1.91	0.77–3.93
Microcephaly	<20	2	2.13	0.26–7.68
	20-24	5	2.10	0.68–4.91
	25-29	11	2.98	1.49–5.33
	30-34	7	1.30	0.52–2.67
	35+	7	1.91	0.77–3.93

Table 9 Prevalence of Birth Defects by Maternal Age for (cont'd) Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Spina Bifida w/ and w/o Hydrocephaly	<20	2	2.13	0.26–7.68
	20-24	2	0.84	0.10–3.04
	25-29	9	2.44	1.11–4.63
	30-34	6	1.11	0.41–2.42
	35+	6	1.64	0.60–3.56
Spinal Cord	<20	5	5.31	1.73–12.40
	20-24	8	3.37	1.45–6.63
	25-29	14	3.79	2.07–6.36
	30-34	12	2.22	1.15–3.88
	35+	14	3.82	2.09–6.40
Other CNS	<20	11	11.69	5.84–20.92
	20-24	21	8.83	5.47–13.50
	25-29	20	5.42	3.31–8.37
	30-34	17	3.15	1.83–5.04
	35+	18	4.91	2.91–7.75
Eye				
Aniridia	<20	0	0.00	0.00–3.92
	20-24	1	0.42	0.01–2.34
	25-29	1	0.27	0.01–1.51
	30-34	2	0.37	0.04–1.34
	35+	0	0.00	0.00–1.01
Anophthalmia/Microphthalmia	<20	1	1.06	0.03–5.92
	20-24	4	1.68	0.46–4.31
	25-29	1	0.27	0.01–1.51
	30-34	6	1.11	0.41–2.42
	35+	1	0.27	0.01–1.52

Table 9 Prevalence of Birth Defects by Maternal Age for (cont'd) Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Congenital Glaucoma, Congenital Cataract	<20	0	0.00	0.00–3.92
	20-24	7	2.94	1.18–6.07
	25-29	6	1.63	0.60–3.54
	30-34	8	1.48	0.64–2.92
	35+	10	2.73	1.31–5.01
Other Eye	<20	0	0.00	0.00–3.92
	20-24	3	1.26	0.26–3.69
	25-29	7	1.90	0.76–3.91
	30-34	7	1.30	0.52–2.67
	35+	8	2.18	0.94–4.30
Ear				
Anotia/Microtia	<20	2	2.13	0.26–7.68
	20-24	3	1.26	0.26–3.69
	25-29	4	1.08	0.30–2.77
	30-34	8	1.48	0.64–2.92
	35+	1	0.27	0.01–1.52
Cardiovascular				
Anomalous Pulmonary Venous Connection				
Total/Partial Anomalous Pulmonary Venous Connection	<20	2	2.13	0.26–7.68
	20-24	2	0.84	0.10–3.04
	25-29	5	1.35	0.44–3.16
	30-34	10	1.85	0.89–3.41
	35+	2	0.55	0.07–1.97

Table 9 Prevalence of Birth Defects by Maternal Age for (cont'd) Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Atrioventricular Canal Defects				
ASD Primum	<20	2	2.13	0.26–7.68
	20-24	2	0.84	0.10–3.04
	25-29	4	1.08	0.30–2.77
	30-34	2	0.37	0.04–1.34
	35+	4	1.09	0.30–2.79
Common Atrium	<20	1	1.06	0.03–5.92
	20-24	2	0.84	0.10–3.04
	25-29	0	0.00	0.00–1.00
	30-34	1	0.19	0.00–1.03
	35+	0	0.00	0.00–1.01
Complete Atrioventricular Canal Defect	<20	4	4.25	1.16–10.88
	20-24	5	2.10	0.68–4.91
	25-29	9	2.44	1.11–4.63
	30-34	10	1.85	0.89–3.41
	35+	19	5.18	3.12–8.09
Endocardial Cushion (OS and NOS)	<20	0	0.00	0.00–3.92
	20-24	1	0.42	0.01–2.34
	25-29	0	0.00	0.00–1.00
	30-34	2	0.37	0.04–1.34
	35+	6	1.64	0.60–3.56
VSD, Canal Type	<20	0	0.00	0.00–3.92
	20-24	2	0.84	0.10–3.04
	25-29	2	0.54	0.07–1.96
	30-34	3	0.56	0.11–1.62
	35+	3	0.82	0.17–2.39

Table 9 Prevalence of Birth Defects by Maternal Age for (cont'd) Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Conotruncal (Outlet) and Aortic Arch				
Double Outlet Right Ventricle	<20	2	2.13	0.26–7.68
	20-24	8	3.37	1.45–6.63
	25-29	3	0.81	0.17–2.37
	30-34	3	0.56	0.11–1.62
	35+	3	0.82	0.17–2.39
d-Transposition of the Great Arteries	<20	1	1.06	0.03–5.92
	20-24	8	3.37	1.45–6.63
	25-29	12	3.25	1.68–5.68
	30-34	17	3.15	1.83–5.04
	35+	13	3.54	1.89–6.06
Interrupted Aortic Arch, Type B	<20	0	0.00	0.00–3.92
	20-24	0	0.00	0.00–1.55
	25-29	1	0.27	0.01–1.51
	30-34	1	0.19	0.00–1.03
	35+	2	0.55	0.07–1.97
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	<20	3	3.19	0.66–9.32
	20-24	8	3.37	1.45–6.63
	25-29	10	2.71	1.30–4.98
	30-34	20	3.70	2.26–5.72
	35+	22	6.00	3.76–9.08
Truncus	<20	0	0.00	0.00–3.92
	20-24	0	0.00	0.00–1.55
	25-29	0	0.00	0.00–1.00
	30-34	3	0.56	0.11–1.62
	35+	0	0.00	0.00–1.01

Table 9 Prevalence of Birth Defects by Maternal Age for (cont'd) Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Ebstein Anomaly				
Ebstein Anomaly	<20	1	1.06	0.03–5.92
	20-24	2	0.84	0.10–3.04
	25-29	0	0.00	0.00–1.00
	30-34	1	0.19	0.00–1.03
	35+	2	0.55	0.07–1.97
Laterality Defects				
Heterotaxy, Situs Inversus	<20	4	4.25	1.16–10.88
	20-24	4	1.68	0.46–4.31
	25-29	1	0.27	0.01–1.51
	30-34	2	0.37	0.04–1.34
	35+	2	0.55	0.07–1.97
Left-Sided Obstruction				
Aortic Valve Stenosis	<20	1	1.06	0.03–5.92
	20-24	4	1.68	0.46–4.31
	25-29	9	2.44	1.11–4.63
	30-34	12	2.22	1.15–3.88
	35+	6	1.64	0.60–3.56
Coarctation of Aorta	<20	3	3.19	0.66–9.32
	20-24	8	3.37	1.45–6.63
	25-29	17	4.60	2.68–7.37
	30-34	30	5.56	3.75–7.93
	35+	16	4.36	2.49–7.08
Hypoplastic Left Heart Syndrome	<20	1	1.06	0.03–5.92
	20-24	2	0.84	0.10–3.04
	25-29	6	1.63	0.60–3.54
	30-34	5	0.93	0.30–2.16
	35+	6	1.64	0.60–3.56

Table 9 Prevalence of Birth Defects by Maternal Age for (cont'd) Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Interrupted Aortic Arch (Type A and NOS)	<20	1	1.06	0.03–5.92
	20-24	1	0.42	0.01–2.34
	25-29	1	0.27	0.01–1.51
	30-34	0	0.00	0.00–0.68
	35+	1	0.27	0.01–1.52
Patent Ductus Arteriosus				
Patent Ductus Arteriosus	<20	13	13.82	7.36–23.62
	20-24	48	20.19	14.89–26.77
	25-29	49	13.27	9.82–17.55
	30-34	86	15.93	12.74–19.67
	35+	73	19.90	15.60–25.02
Right-Sided Obstruction				
Pulmonary Stenosis, Valvular	<20	7	7.44	2.99–15.33
	20-24	20	8.41	5.14–12.99
	25-29	27	7.31	4.82–10.64
	30-34	29	5.37	3.60–7.71
	35+	22	6.00	3.76–9.08
Pulmonary Valve Atresia w/intact septum	<20	0	0.00	0.00–3.92
	20-24	2	0.84	0.10–3.04
	25-29	5	1.35	0.44–3.16
	30-34	4	0.74	0.20–1.90
	35+	3	0.82	0.17–2.39
Pulmonary Valve Atresia with VSD	<20	0	0.00	0.00–3.92
	20-24	1	0.42	0.01–2.34
	25-29	2	0.54	0.07–1.96
	30-34	2	0.37	0.04–1.34
	35+	1	0.27	0.01–1.52

Table 9 Prevalence of Birth Defects by Maternal Age for (cont'd) Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Tricuspid Valve Atresia	<20	0	0.00	0.00–3.92
	20-24	4	1.68	0.46–4.31
	25-29	4	1.08	0.30–2.77
	30-34	3	0.56	0.11–1.62
	35+	1	0.27	0.01–1.52
Septal Defects				
ASD (Secundum and NOS)	<20	16	17.00	9.72–27.61
	20-24	54	22.71	17.06–29.64
	25-29	68	18.42	14.30–23.35
	30-34	82	15.19	12.08–18.85
	35+	90	24.53	19.73–30.15
VSD (Membranous and NOS)	<20	13	13.82	7.36–23.62
	20-24	26	10.94	7.14–16.02
	25-29	34	9.21	6.38–12.87
	30-34	54	10.00	7.51–13.05
	35+	50	13.63	10.12–17.97
VSD, Conoventricular/Malalignment	<20	2	2.13	0.26–7.68
	20-24	6	2.52	0.93–5.49
	25-29	3	0.81	0.17–2.37
	30-34	12	2.22	1.15–3.88
	35+	7	1.91	0.77–3.93
Single Ventricle and L-TGA				
L-TGA	<20	2	2.13	0.26–7.68
	20-24	0	0.00	0.00–1.55
	25-29	1	0.27	0.01–1.51
	30-34	1	0.19	0.00–1.03
	35+	4	1.09	0.30–2.79

Table 9 Prevalence of Birth Defects by Maternal Age for (cont'd) Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Single Ventricle	<20	0	0.00	0.00–3.92
	20-24	0	0.00	0.00–1.55
	25-29	2	0.54	0.07–1.96
	30-34	0	0.00	0.00–0.68
	35+	0	0.00	0.00–1.01
Other Cardiovascular				
Other Cardiovascular	<20	15	15.94	8.92–26.29
	20-24	39	16.40	11.67–22.43
	25-29	48	13.00	9.59–17.24
	30-34	91	16.85	13.57–20.69
	35+	61	16.63	12.72–21.36
Respiratory				
Choanal Atresia	<20	0	0.00	0.00–3.92
	20-24	0	0.00	0.00–1.55
	25-29	1	0.27	0.01–1.51
	30-34	3	0.56	0.11–1.62
	35+	0	0.00	0.00–1.01
Lung Anomalies	<20	0	0.00	0.00–3.92
	20-24	1	0.42	0.01–2.34
	25-29	6	1.63	0.60–3.54
	30-34	9	1.67	0.76–3.16
	35+	8	2.18	0.94–4.30
Other Respiratory	<20	1	1.06	0.03–5.92
	20-24	2	0.84	0.10–3.04
	25-29	3	0.81	0.17–2.37
	30-34	4	0.74	0.20–1.90
	35+	4	1.09	0.30–2.79

Table 9 Prevalence of Birth Defects by Maternal Age for (cont'd) Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Orofacial				
Cleft Lip w/ and w/o Cleft Palate	<20	7	7.44	2.99–15.33
	20-24	24	10.10	6.47–15.02
	25-29	22	5.96	3.73–9.02
	30-34	32	5.93	4.05–8.37
	35+	25	6.81	4.41–10.06
Cleft Palate w/o Cleft Lip	<20	9	9.56	4.37–18.16
	20-24	14	5.89	3.22–9.88
	25-29	21	5.69	3.52–8.69
	30-34	29	5.37	3.60–7.71
	35+	21	5.72	3.54–8.75
Pierre Robin Sequence	<20	3	3.19	0.66–9.32
	20-24	4	1.68	0.46–4.31
	25-29	6	1.63	0.60–3.54
	30-34	13	2.41	1.28–4.12
	35+	9	2.45	1.12–4.66
Other Orofacial	<20	2	2.13	0.26–7.68
	20-24	6	2.52	0.93–5.49
	25-29	10	2.71	1.30–4.98
	30-34	18	3.33	1.98–5.27
	35+	13	3.54	1.89–6.06
Gastrointestinal				
Biliary Atresia	<20	0	0.00	0.00–3.92
	20-24	2	0.84	0.10–3.04
	25-29	1	0.27	0.01–1.51
	30-34	0	0.00	0.00–0.68
	35+	7	1.91	0.77–3.93

Table 9 Prevalence of Birth Defects by Maternal Age for (cont'd) Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Esophageal Atresia/Tracheoesophageal Fistula	<20	0	0.00	0.00–3.92
	20-24	3	1.26	0.26–3.69
	25-29	8	2.17	0.94–4.27
	30-34	10	1.85	0.89–3.41
	35+	17	4.63	2.70–7.42
Hirschsprung Disease	<20	1	1.06	0.03–5.92
	20-24	3	1.26	0.26–3.69
	25-29	5	1.35	0.44–3.16
	30-34	4	0.74	0.20–1.90
	35+	5	1.36	0.44–3.18
Rectal and Large Intestinal Atresia/Stenosis	<20	2	2.13	0.26–7.68
	20-24	7	2.94	1.18–6.07
	25-29	15	4.06	2.27–6.70
	30-34	24	4.45	2.85–6.61
	35+	13	3.54	1.89–6.06
Small Intestinal Atresia	<20	5	5.31	1.73–12.40
	20-24	6	2.52	0.93–5.49
	25-29	10	2.71	1.30–4.98
	30-34	6	1.11	0.41–2.42
	35+	16	4.36	2.49–7.08
Other Gastrointestinal	<20	6	6.38	2.34–13.88
	20-24	18	7.57	4.49–11.97
	25-29	18	4.88	2.89–7.71
	30-34	28	5.19	3.45–7.50
	35+	20	5.45	3.33–8.42

Table 9 Prevalence of Birth Defects by Maternal Age for (cont'd) Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Genitourinary				
Bladder Exstrophy	<20	0	0.00	0.00–3.92
	20-24	1	0.42	0.01–2.34
	25-29	0	0.00	0.00–1.00
	30-34	4	0.74	0.20–1.90
	35+	1	0.27	0.01–1.52
Cloacal Exstrophy	<20	0	0.00	0.00–3.92
	20-24	1	0.42	0.01–2.34
	25-29	1	0.27	0.01–1.51
	30-34	0	0.00	0.00–0.68
	35+	0	0.00	0.00–1.01
Hypospadias, 2nd or 3rd Degree	<20	7	7.44	2.99–15.33
	20-24	15	6.31	3.53–10.41
	25-29	41	11.11	7.97–15.07
	30-34	69	12.78	9.94–16.17
	35+	49	13.36	9.88–17.66
Obstructive Genitourinary Defect	<20	9	9.56	4.37–18.16
	20-24	24	10.10	6.47–15.02
	25-29	34	9.21	6.38–12.87
	30-34	46	8.52	6.24–11.36
	35+	34	9.27	6.42–12.95
Renal Agenesis/Hypoplasia	<20	0	0.00	0.00–3.92
	20-24	0	0.00	0.00–1.55
	25-29	0	0.00	0.00–1.00
	30-34	2	0.37	0.04–1.34
	35+	0	0.00	0.00–1.01

Table 9 Prevalence of Birth Defects by Maternal Age for (cont'd) Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Other Genitourinary	<20	6	6.38	2.34 – 13.88
	20-24	28	11.78	7.83 – 17.02
	25-29	43	11.65	8.43 – 15.69
	30-34	56	10.37	7.83 – 13.47
	35+	58	15.81	12.00 – 20.44
Musculoskeletal				
Club Foot	<20	14	14.88	8.13 – 24.96
	20-24	31	13.04	8.86 – 18.51
	25-29	33	8.94	6.15 – 12.55
	30-34	52	9.63	7.19 – 12.63
	35+	38	10.36	7.33 – 14.22
Craniosynostosis	<20	1	1.06	0.03 – 5.92
	20-24	7	2.94	1.18 – 6.07
	25-29	21	5.69	3.52 – 8.69
	30-34	28	5.19	3.45 – 7.50
	35+	31	8.45	5.74 – 11.99
Diaphragmatic Hernia	<20	3	3.19	0.66 – 9.32
	20-24	5	2.10	0.68 – 4.91
	25-29	6	1.63	0.60 – 3.54
	30-34	11	2.04	1.02 – 3.65
	35+	11	3.00	1.50 – 5.36
Gastroschisis	<20	12	12.75	6.59 – 22.28
	20-24	15	6.31	3.53 – 10.41
	25-29	8	2.17	0.94 – 4.27
	30-34	1	0.19	0.00 – 1.03
	35+	1	0.27	0.01 – 1.52

Table 9 Prevalence of Birth Defects by Maternal Age for (cont'd) Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Omphalocele	<20	2	2.13	0.26–7.68
	20-24	5	2.10	0.68–4.91
	25-29	5	1.35	0.44–3.16
	30-34	5	0.93	0.30–2.16
	35+	7	1.91	0.77–3.93
Polydactyly/Syndactyly	<20	10	10.63	5.10–19.54
	20-24	29	12.20	8.17–17.52
	25-29	46	12.46	9.12–16.62
	30-34	68	12.59	9.78–15.97
	35+	37	10.08	7.10–13.90
Reduction Deformity, Lower Limbs	<20	1	1.06	0.03–5.92
	20-24	4	1.68	0.46–4.31
	25-29	3	0.81	0.17–2.37
	30-34	3	0.56	0.11–1.62
	35+	5	1.36	0.44–3.18
Reduction Deformity, Upper Limbs	<20	2	2.13	0.26–7.68
	20-24	7	2.94	1.18–6.07
	25-29	12	3.25	1.68–5.68
	30-34	13	2.41	1.28–4.12
	35+	6	1.64	0.60–3.56
Skeletal Dysplasia	<20	1	1.06	0.03–5.92
	20-24	2	0.84	0.10–3.04
	25-29	3	0.81	0.17–2.37
	30-34	12	2.22	1.15–3.88
	35+	9	2.45	1.12–4.66
Other Musculoskeletal	<20	7	7.44	2.99–15.33
	20-24	24	10.10	6.47–15.02
	25-29	29	7.85	5.26–11.28
	30-34	39	7.22	5.14–9.87
	35+	36	9.81	6.87–13.58

Table 9 Prevalence of Birth Defects by Maternal Age for (cont'd) Live Births, Massachusetts: 2002-2003

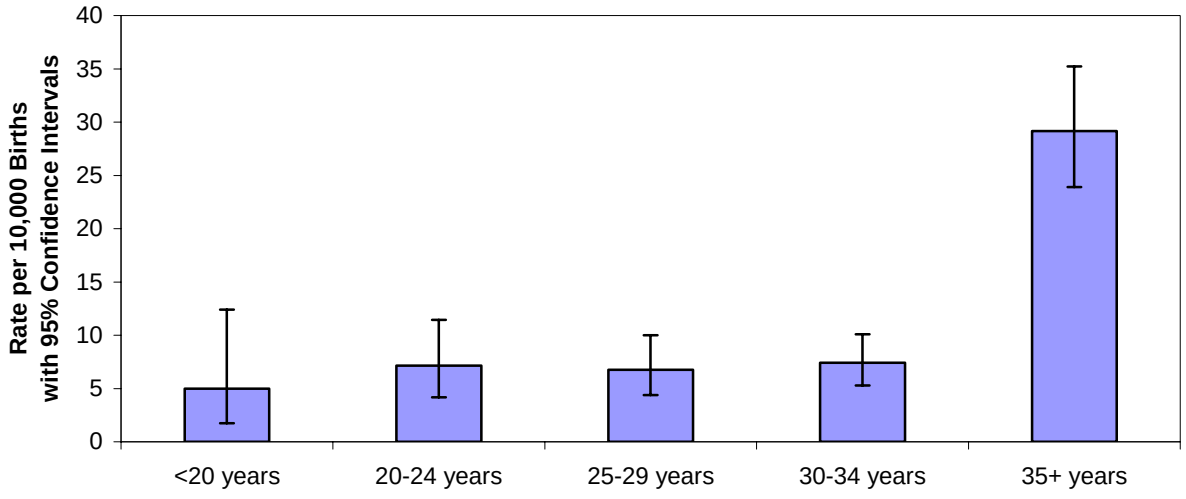
Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Chromosomal and Other Syndromes				
Klinefelter Syndrome	<20	1	1.06	0.03–5.92
	20-24	0	0.00	0.00–1.55
	25-29	1	0.27	0.01–1.51
	30-34	0	0.00	0.00–0.68
	35+	5	1.36	0.44–3.18
Trisomy 13	<20	0	0.00	0.00–3.92
	20-24	2	0.84	0.10–3.04
	25-29	1	0.27	0.01–1.51
	30-34	1	0.19	0.00–1.03
	35+	4	1.09	0.30–2.79
Trisomy 18	<20	0	0.00	0.00–3.92
	20-24	2	0.84	0.10–3.04
	25-29	3	0.81	0.17–2.37
	30-34	4	0.74	0.20–1.90
	35+	8	2.18	0.94–4.30
Trisomy 21 (Down Syndrome)	<20	5	5.31	1.73–12.40
	20-24	17	7.15	4.17–11.45
	25-29	25	6.77	4.38–10.00
	30-34	40	7.41	5.29–10.09
	35+	107	29.16	23.90–35.24
Turner Syndrome	<20	0	0.00	0.00–3.92
	20-24	0	0.00	0.00–1.55
	25-29	2	0.54	0.07–1.96
	30-34	5	0.93	0.30–2.16
	35+	4	1.09	0.30–2.79

Table 9 Prevalence of Birth Defects by Maternal Age for (cont'd) Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Other Chromosomal Syndromes/Other Syndromes	<20	8	8.50	3.67–16.75
	20-24	19	7.99	4.81–12.48
	25-29	39	10.56	7.51–14.44
	30-34	55	10.19	7.67–13.26
	35+	55	14.99	11.29–19.51
Other				
Amniotic Bands	<20	1	1.06	0.03–5.92
	20-24	3	1.26	0.26–3.69
	25-29	2	0.54	0.07–1.96
	30-34	2	0.37	0.04–1.34
	35+	1	0.27	0.01–1.52
Skin Anomalies	<20	0	0.00	0.00–3.92
	20-24	2	0.84	0.10–3.04
	25-29	1	0.27	0.01–1.51
	30-34	3	0.56	0.11–1.62
	35+	1	0.27	0.01–1.52
Other, Specified	<20	0	0.00	0.00–3.92
	20-24	5	2.10	0.68–4.91
	25-29	2	0.54	0.07–1.96
	30-34	3	0.56	0.11–1.62
	35+	2	0.55	0.07–1.97

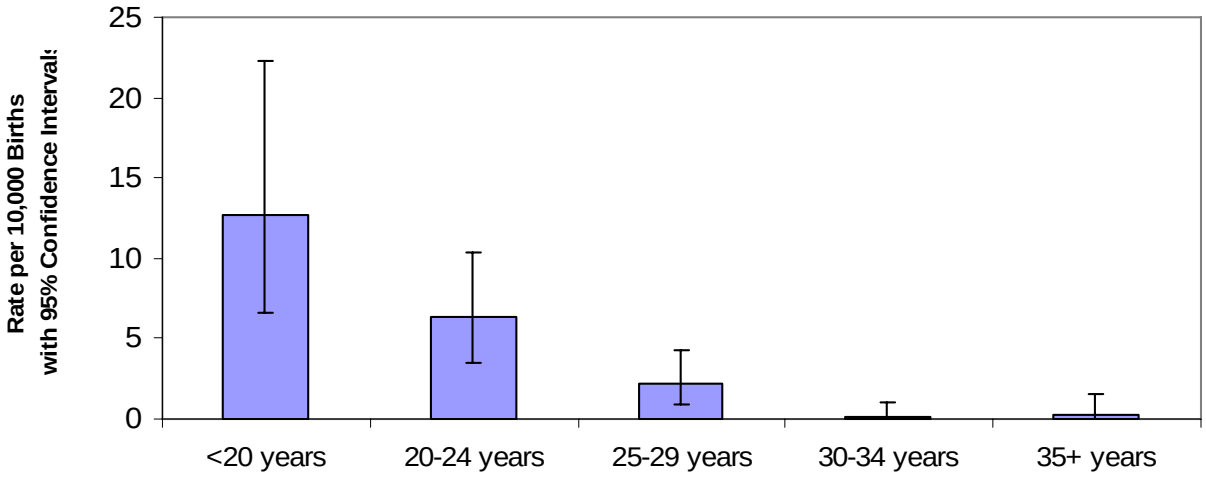
1. Cases can be included in more than one defect category. Cases are counted once within a defect category. Counts represent the number of defects, not the total number of cases with birth defects. Because only live births are presented on this table, case totals are not listed. Maternal age is drawn from the birth certificate. Because birth certificate data are more accurate for this field than fetal death records, analyses of maternal age are limited to live births.

Figure 6 Rates of Down Syndrome among Maternal Age Groups, Massachusetts: 2002-2003



N=194

Figure 7 Rates of Gastroschisis among Maternal Age Groups, Massachusetts: 2002-2003



N=37

Table 10 Most Common Defects for Live Births, by Maternal Age, Massachusetts: 2002-2003

Age	Defect[1]	Count	Rate per 10,000 Births	95% Confidence Interval
<20				
	ASD (Secundum and NOS)	16	17.00	9.72 — 27.61
	Club Foot	14	14.88	8.13 — 24.96
	VSD (Membranous and NOS)	13	13.82	7.36 — 23.62
	Gastroschisis	12	12.75	6.59 — 22.28
	Polydactyly/Syndactyly	10	10.63	5.10 — 19.54
20-24				
	ASD (Secundum and NOS)	54	22.71	17.06 — 29.64
	Club Foot	31	13.04	8.86 — 18.51
	Polydactyly/Syndactyly	29	12.20	8.17 — 17.52
	VSD (Membranous and NOS)	26	10.94	7.14 — 16.02
	Cleft Lip w/ and w/o Cleft Palate	24	10.10	6.47 — 15.02
	Obstructive Genitourinary Defect	24	10.10	6.47 — 15.02
25-29				
	ASD (Secundum and NOS)	68	18.42	14.30 — 23.35
	Polydactyly/Syndactyly	46	12.46	9.12 — 16.62
	Hypospadias, 2nd or 3rd Degree	41	11.11	7.97 — 15.07
	VSD (Membranous and NOS)	34	9.21	6.38 — 12.87
	Obstructive Genitourinary Defect	34	9.21	6.38 — 12.87
30-34				
	ASD (Secundum and NOS)	82	15.19	12.08 — 18.85
	Hypospadias, 2nd or 3rd Degree	69	12.78	9.94 — 16.17
	Polydactyly/Syndactyly	68	12.59	9.78 — 15.97
	VSD (Membranous and NOS)	54	10.00	7.51 — 13.05
	Club Foot	52	9.63	7.19 — 12.63
35+				
	Trisomy 21 (Down Syndrome)	107	29.16	23.90 — 35.24
	ASD (Secundum and NOS)	90	24.53	19.73 — 30.15
	VSD (Membranous and NOS)	50	13.63	10.12 — 17.97
	Hypospadias, 2nd or 3rd Degree	49	13.36	9.88 — 17.66
	Club Foot	38	10.36	7.33 — 14.22

1. Excludes Patent Ductus Arteriosus (PDA) due to the high number of cases and the mild severity of the majority of these cases. Maternal age is drawn from the birth certificate. Because birth certificate data are more accurate for this field than fetal death records, analyses of maternal age are limited to live births.

Table 11 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
Central Nervous System				
Anencephaly	White, Non-Hispanic	1	0.09	0.00 – 0.48
	Black, Non-Hispanic	0	0.00	0.00 – 3.11
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	2	1.04	0.13 – 3.74
Encephalocele	White, Non-Hispanic	4	0.35	0.09 – 0.88
	Black, Non-Hispanic	1	0.84	0.02 – 4.70
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	1	0.52	0.01 – 2.89
Holoprosencephaly	White, Non-Hispanic	5	0.43	0.14 – 1.01
	Black, Non-Hispanic	0	0.00	0.00 – 3.11
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	1	0.52	0.01 – 2.89
Hydrocephaly w/o Spina Bifida	White, Non-Hispanic	38	3.28	2.32 – 4.51
	Black, Non-Hispanic	5	4.22	1.37 – 9.85
	Asian, Non-Hispanic	4	3.80	1.04 – 9.73
	Hispanic	4	2.07	0.56 – 5.30
Microcephaly	White, Non-Hispanic	19	1.64	0.99 – 2.56
	Black, Non-Hispanic	3	2.53	0.52 – 7.40
	Asian, Non-Hispanic	5	4.75	1.54 – 11.09
	Hispanic	5	2.59	0.84 – 6.04
Spina Bifida w/ and w/o Hydrocephaly	White, Non-Hispanic	17	1.47	0.86 – 2.35
	Black, Non-Hispanic	4	3.38	0.92 – 8.64
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	3	1.55	0.32 – 4.54
Spinal Cord	White, Non-Hispanic	38	3.28	2.32 – 4.51
	Black, Non-Hispanic	3	2.53	0.52 – 7.40
	Asian, Non-Hispanic	1	0.95	0.02 – 5.29
	Hispanic	7	3.63	1.46 – 7.47
Other CNS	White, Non-Hispanic	62	5.36	4.11 – 6.87
	Black, Non-Hispanic	5	4.22	1.37 – 9.85
	Asian, Non-Hispanic	5	4.75	1.54 – 11.09
	Hispanic	13	6.73	3.59 – 11.51
Eye				
Aniridia	White, Non-Hispanic	3	0.26	0.05 – 0.76
	Black, Non-Hispanic	1	0.84	0.02 – 4.70
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	0	0.00	0.00 – 1.91

Table 11 (cont'd) Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
Anophthalmia/Microphthalmia	White, Non-Hispanic	7	0.60	0.24 – 1.25
	Black, Non-Hispanic	0	0.00	0.00 – 3.11
	Asian, Non-Hispanic	2	1.90	0.23 – 6.86
	Hispanic	3	1.55	0.32 – 4.54
Congenital Glaucoma, Congenital Cataract	White, Non-Hispanic	25	2.16	1.40 – 3.19
	Black, Non-Hispanic	3	2.53	0.52 – 7.40
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	3	1.55	0.32 – 4.54
Other Eye	White, Non-Hispanic	17	1.47	0.86 – 2.35
	Black, Non-Hispanic	1	0.84	0.02 – 4.70
	Asian, Non-Hispanic	1	0.95	0.02 – 5.29
	Hispanic	6	3.11	1.14 – 6.76
Ear				
Anotia/Microtia	White, Non-Hispanic	13	1.12	0.60 – 1.92
	Black, Non-Hispanic	2	1.69	0.20 – 6.10
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	3	1.55	0.32 – 4.54
Cardiovascular				
Anomalous Pulmonary Venous Connection				
Total/Partial Anomalous Pulmonary Venous	White, Non-Hispanic	15	1.30	0.73 – 2.14
	Black, Non-Hispanic	2	1.69	0.20 – 6.10
	Asian, Non-Hispanic	1	0.95	0.02 – 5.29
	Hispanic	3	1.55	0.32 – 4.54
Atrioventricular Canal Defects				
ASD Primum	White, Non-Hispanic	10	0.86	0.41 – 1.59
	Black, Non-Hispanic	2	1.69	0.20 – 6.10
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	2	1.04	0.13 – 3.74
Common Atrium	White, Non-Hispanic	1	0.09	0.00 – 0.48
	Black, Non-Hispanic	1	0.84	0.02 – 4.70
	Asian, Non-Hispanic	1	0.95	0.02 – 5.29
	Hispanic	1	0.52	0.01 – 2.89
Complete Atrioventricular Canal Defect	White, Non-Hispanic	33	2.85	1.96 – 4.00
	Black, Non-Hispanic	8	6.75	2.91 – 13.30
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	4	2.07	0.56—5.30

Table 11 (cont'd) Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
Endocardial Cushion (OS and NOS)	White, Non-Hispanic	4	0.35	0.09–0.88
	Black, Non-Hispanic	2	1.69	0.20–6.10
	Asian, Non-Hispanic	2	1.90	0.23–6.86
	Hispanic	1	0.52	0.01–2.89
VSD, Canal Type	White, Non-Hispanic	9	0.78	0.36–1.48
	Black, Non-Hispanic	1	0.84	0.02–4.70
	Asian, Non-Hispanic	0	0.00	0.00–3.51
	Hispanic	0	0.00	0.00–1.91
Conotruncal (Outlet) and Aortic Arch				
Double Outlet Right Ventricle	White, Non-Hispanic	8	0.69	0.30–1.36
	Black, Non-Hispanic	4	3.38	0.92–8.64
	Asian, Non-Hispanic	4	3.80	1.04–9.73
	Hispanic	2	1.04	0.13–3.74
d-Transposition of the Great Arteries	White, Non-Hispanic	44	3.80	2.76–5.10
	Black, Non-Hispanic	2	1.69	0.20–6.10
	Asian, Non-Hispanic	2	1.90	0.23–6.86
	Hispanic	2	1.04	0.13–3.74
Interrupted Aortic Arch, Type B	White, Non-Hispanic	2	0.17	0.02–0.62
	Black, Non-Hispanic	1	0.84	0.02–4.70
	Asian, Non-Hispanic	1	0.95	0.02–5.29
	Hispanic	0	0.00	0.00–1.91
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	White, Non-Hispanic	46	3.97	2.91–5.30
	Black, Non-Hispanic	3	2.53	0.52–7.40
	Asian, Non-Hispanic	2	1.90	0.23–6.86
	Hispanic	7	3.63	1.46–7.47
Truncus	White, Non-Hispanic	2	0.17	0.02–0.62
	Black, Non-Hispanic	1	0.84	0.02–4.70
	Asian, Non-Hispanic	0	0.00	0.00–3.51
	Hispanic	0	0.00	0.00–1.91
Ebstein Anomaly				
Ebstein Anomaly	White, Non-Hispanic	3	0.26	0.05–0.76
	Black, Non-Hispanic	0	0.00	0.00–3.11
	Asian, Non-Hispanic	0	0.00	0.00–3.51
	Hispanic	3	1.55	0.32–4.54
Laterality Defects				
Heterotaxy, Situs Inversus	White, Non-Hispanic	7	0.60	0.24–1.25
	Black, Non-Hispanic	1	0.84	0.02–4.70
	Asian, Non-Hispanic	0	0.00	0.00–3.51
	Hispanic	3	1.55	0.32–4.54

Table 11 (cont'd) Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
Left-Sided Obstruction				
Aortic Valve Stenosis	White, Non-Hispanic	23	1.99	1.26 – 2.98
	Black, Non-Hispanic	4	3.38	0.92 – 8.64
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	5	2.59	0.84 – 6.04
Coarctation of Aorta	White, Non-Hispanic	53	4.58	3.43 – 5.99
	Black, Non-Hispanic	6	5.06	1.86 – 11.02
	Asian, Non-Hispanic	3	2.85	0.59 – 8.33
	Hispanic	8	4.14	1.79 – 8.16
Hypoplastic Left Heart Syndrome	White, Non-Hispanic	14	1.21	0.66 – 2.03
	Black, Non-Hispanic	6	5.06	1.86 – 11.02
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	0	0.00	0.00 – 1.91
Interrupted Aortic Arch (Type A and NOS)	White, Non-Hispanic	2	0.17	0.02 – 0.62
	Black, Non-Hispanic	0	0.00	0.00 – 3.11
	Asian, Non-Hispanic	1	0.95	0.02 – 5.29
	Hispanic	1	0.52	0.01 – 2.89
Patent Ductus Arteriosus				
Patent Ductus Arteriosus	White, Non-Hispanic	190	16.42	14.16 – 18.92
	Black, Non-Hispanic	25	21.10	13.65 – 31.14
	Asian, Non-Hispanic	13	12.35	6.58 – 21.12
	Hispanic	34	17.61	12.20 – 24.61
Right-Sided Obstruction				
Pulmonary Stenosis, Valvular	White, Non-Hispanic	79	6.83	5.40 – 8.51
	Black, Non-Hispanic	11	9.28	4.63 – 16.61
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	13	6.73	3.59 – 11.51
Pulmonary Valve Atresia w/intact septum	White, Non-Hispanic	10	0.86	0.41 – 1.59
	Black, Non-Hispanic	1	0.84	0.02 – 4.70
	Asian, Non-Hispanic	1	0.95	0.02 – 5.29
	Hispanic	2	1.04	0.13 – 3.74
Pulmonary Valve Atresia with VSD	White, Non-Hispanic	3	0.26	0.05 – 0.76
	Black, Non-Hispanic	1	0.84	0.02 – 4.70
	Asian, Non-Hispanic	1	0.95	0.02 – 5.29
	Hispanic	0	0.00	0.00 – 1.91
Tricuspid Valve Atresia	White, Non-Hispanic	7	0.60	0.24 – 1.25
	Black, Non-Hispanic	3	2.53	0.52 – 7.40
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	1	0.52	0.01—2.89

Table 11 (cont'd) Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
Septal Defects				
ASD (Secundum and NOS)	White, Non-Hispanic	203	17.54	15.21—20.13
	Black, Non-Hispanic	41	34.60	24.83—46.94
	Asian, Non-Hispanic	14	13.30	7.27—22.32
	Hispanic	43	22.27	16.12—30.00
VSD (Membranous and NOS)	White, Non-Hispanic	113	9.76	8.05—11.74
	Black, Non-Hispanic	11	9.28	4.63—16.61
	Asian, Non-Hispanic	15	14.25	7.98—23.51
	Hispanic	33	17.09	11.77—24.00
VSD, Conoventricular/Malalignment	White, Non-Hispanic	20	1.73	1.06—2.67
	Black, Non-Hispanic	4	3.38	0.92—8.64
	Asian, Non-Hispanic	2	1.90	0.23—6.86
	Hispanic	2	1.04	0.13—3.74
Single Ventricle and L-TGA				
L-TGA	White, Non-Hispanic	5	0.43	0.14—1.01
	Black, Non-Hispanic	1	0.84	0.02—4.70
	Asian, Non-Hispanic	0	0.00	0.00—3.51
	Hispanic	2	1.04	0.13—3.74
Single Ventricle	White, Non-Hispanic	2	0.17	0.02—0.62
	Black, Non-Hispanic	0	0.00	0.00—3.11
	Asian, Non-Hispanic	0	0.00	0.00—3.51
	Hispanic	0	0.00	0.00—1.91
Other Cardiovascular				
Other Cardiovascular	White, Non-Hispanic	183	15.81	13.60—18.28
	Black, Non-Hispanic	26	21.94	14.33—32.15
	Asian, Non-Hispanic	11	10.45	5.22—18.70
	Hispanic	26	13.47	8.80—19.73
Respiratory				
Choanal Atresia	White, Non-Hispanic	4	0.35	0.09—0.88
	Black, Non-Hispanic	0	0.00	0.00—3.11
	Asian, Non-Hispanic	0	0.00	0.00—3.51
	Hispanic	0	0.00	0.00—1.91
Lung Anomalies	White, Non-Hispanic	16	1.38	0.79—2.24
	Black, Non-Hispanic	3	2.53	0.52—7.40
	Asian, Non-Hispanic	3	2.85	0.59—8.33
	Hispanic	1	0.52	0.01—2.89

Table 11 (cont'd) Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
Other Respiratory	White, Non-Hispanic	11	0.95	0.47 – 1.70
	Black, Non-Hispanic	0	0.00	0.00 – 3.11
	Asian, Non-Hispanic	1	0.95	0.02 – 5.29
	Hispanic	1	0.52	0.01 – 2.89
Orofacial				
Cleft Lip w/ and w/o Cleft Palate	White, Non-Hispanic	79	6.83	5.40 – 8.51
	Black, Non-Hispanic	9	7.59	3.47 – 14.42
	Asian, Non-Hispanic	7	6.65	2.67 – 13.70
	Hispanic	14	7.25	3.96 – 12.17
Cleft Palate w/o Cleft Lip	White, Non-Hispanic	66	5.70	4.41 – 7.25
	Black, Non-Hispanic	4	3.38	0.92 – 8.64
	Asian, Non-Hispanic	9	8.55	3.91 – 16.23
	Hispanic	10	5.18	2.48 – 9.53
Pierre Robin Sequence	White, Non-Hispanic	26	2.25	1.47 – 3.29
	Black, Non-Hispanic	2	1.69	0.20 – 6.10
	Asian, Non-Hispanic	3	2.85	0.59 – 8.33
	Hispanic	3	1.55	0.32 – 4.54
Other Orofacial	White, Non-Hispanic	37	3.20	2.25 – 4.41
	Black, Non-Hispanic	2	1.69	0.20 – 6.10
	Asian, Non-Hispanic	2	1.90	0.23 – 6.86
	Hispanic	8	4.14	1.79 – 8.16
Gastrointestinal				
Biliary Atresia	White, Non-Hispanic	7	0.60	0.24 – 1.25
	Black, Non-Hispanic	2	1.69	0.20 – 6.10
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	1	0.52	0.01 – 2.89
Esophageal Atresia/Tracheoesophageal Fistula	White, Non-Hispanic	32	2.76	1.89 – 3.90
	Black, Non-Hispanic	1	0.84	0.02 – 4.70
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	3	1.55	0.32 – 4.54
Hirschsprung Disease	White, Non-Hispanic	14	1.21	0.66 – 2.03
	Black, Non-Hispanic	1	0.84	0.02 – 4.70
	Asian, Non-Hispanic	1	0.95	0.02 – 5.29
	Hispanic	2	1.04	0.13 – 3.74
Rectal and Large Intestinal Atresia/Stenosis	White, Non-Hispanic	41	3.54	2.54 – 4.81
	Black, Non-Hispanic	7	5.91	2.38 – 12.17
	Asian, Non-Hispanic	1	0.95	0.02 – 5.29
	Hispanic	12	6.22	3.21 — 10.86

Table 11 (cont'd) Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
Small Intestinal Atresia	White, Non-Hispanic	28	2.42	1.61 – 3.50
	Black, Non-Hispanic	3	2.53	0.52 – 7.40
	Asian, Non-Hispanic	1	0.95	0.02 – 5.29
	Hispanic	11	5.70	2.84 – 10.19
Other Gastrointestinal	White, Non-Hispanic	60	5.18	3.96 – 6.67
	Black, Non-Hispanic	11	9.28	4.63 – 16.61
	Asian, Non-Hispanic	1	0.95	0.02 – 5.29
	Hispanic	16	8.29	4.74 – 13.46
Genitourinary				
Bladder Exstrophy	White, Non-Hispanic	6	0.52	0.19 – 1.13
	Black, Non-Hispanic	0	0.00	0.00 – 3.11
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	0	0.00	0.00 – 1.91
Cloacal Exstrophy	White, Non-Hispanic	0	0.00	0.00 – 0.32
	Black, Non-Hispanic	0	0.00	0.00 – 3.11
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	2	1.04	0.13 – 3.74
Hypospadias, 2nd or 3rd Degree	White, Non-Hispanic	150	12.96	10.97 – 15.21
	Black, Non-Hispanic	8	6.75	2.91 – 13.30
	Asian, Non-Hispanic	5	4.75	1.54 – 11.09
	Hispanic	17	8.81	5.13 – 14.10
Obstructive Genitourinary Defect	White, Non-Hispanic	108	9.33	7.65 – 11.27
	Black, Non-Hispanic	9	7.59	3.47 – 14.42
	Asian, Non-Hispanic	5	4.75	1.54 – 11.09
	Hispanic	17	8.81	5.13 – 14.10
Renal Agenesis/Hypoplasia	White, Non-Hispanic	1	0.09	0.00 – 0.48
	Black, Non-Hispanic	1	0.84	0.02 – 4.70
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	0	0.00	0.00 – 1.91
Other Genitourinary	White, Non-Hispanic	147	12.70	10.73 – 14.93
	Black, Non-Hispanic	10	8.44	4.05 – 15.52
	Asian, Non-Hispanic	11	10.45	5.22 – 18.70
	Hispanic	18	9.32	5.53 – 14.73
Musculoskeletal				
Club Foot	White, Non-Hispanic	125	10.80	8.99 – 12.87
	Black, Non-Hispanic	8	6.75	2.91 – 13.30
	Asian, Non-Hispanic	9	8.55	3.91 – 16.23
	Hispanic	19	9.84	5.92 – 15.37

Table 11 (cont'd) Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
Craniosynostosis	White, Non-Hispanic	79	6.83	5.40 – 8.51
	Black, Non-Hispanic	0	0.00	0.00 – 3.11
	Asian, Non-Hispanic	2	1.90	0.23 – 6.86
	Hispanic	6	3.11	1.14 – 6.76
Diaphragmatic Hernia	White, Non-Hispanic	26	2.25	1.47 – 3.29
	Black, Non-Hispanic	4	3.38	0.92 – 8.64
	Asian, Non-Hispanic	2	1.90	0.23 – 6.86
	Hispanic	4	2.07	0.56 – 5.30
Gastroschisis	White, Non-Hispanic	20	1.73	1.06 – 2.67
	Black, Non-Hispanic	2	1.69	0.20 – 6.10
	Asian, Non-Hispanic	3	2.85	0.59 – 8.33
	Hispanic	11	5.70	2.84 – 10.19
Omphalocele	White, Non-Hispanic	19	1.64	0.99 – 2.56
	Black, Non-Hispanic	2	1.69	0.20 – 6.10
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	2	1.04	0.13 – 3.74
Polydactyly/Syndactyly	White, Non-Hispanic	135	11.66	9.78 – 13.81
	Black, Non-Hispanic	13	10.97	5.84 – 18.76
	Asian, Non-Hispanic	6	5.70	2.09 – 12.41
	Hispanic	33	17.09	11.77 – 24.00
Reduction Deformity, Lower Limbs	White, Non-Hispanic	9	0.78	0.36 – 1.48
	Black, Non-Hispanic	1	0.84	0.02 – 4.70
	Asian, Non-Hispanic	1	0.95	0.02 – 5.29
	Hispanic	3	1.55	0.32 – 4.54
Reduction Deformity, Upper Limbs	White, Non-Hispanic	27	2.33	1.54 – 3.39
	Black, Non-Hispanic	3	2.53	0.52 – 7.40
	Asian, Non-Hispanic	1	0.95	0.02 – 5.29
	Hispanic	5	2.59	0.84 – 6.04
Skeletal Dysplasia	White, Non-Hispanic	21	1.81	1.12 – 2.77
	Black, Non-Hispanic	2	1.69	0.20 – 6.10
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	3	1.55	0.32 – 4.54
Other Musculoskeletal	White, Non-Hispanic	90	7.78	6.25 – 9.56
	Black, Non-Hispanic	11	9.28	4.63 – 16.61
	Asian, Non-Hispanic	8	7.60	3.28 – 14.98
	Hispanic	20	10.36	6.33 – 16.00

Table 11 (cont'd) Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2002-2003

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
Chromosomal and Other Syndromes				
Klinefelter Syndrome	White, Non-Hispanic	5	0.43	0.14 – 1.01
	Black, Non-Hispanic	1	0.84	0.02 – 4.70
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	1	0.52	0.01 – 2.89
Trisomy 13	White, Non-Hispanic	6	0.52	0.19 – 1.13
	Black, Non-Hispanic	1	0.84	0.02 – 4.70
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	0	0.00	0.00 – 1.91
Trisomy 18	White, Non-Hispanic	12	1.04	0.54 – 1.81
	Black, Non-Hispanic	2	1.69	0.20 – 6.10
	Asian, Non-Hispanic	1	0.95	0.02 – 5.29
	Hispanic	2	1.04	0.13 – 3.74
Trisomy 21 (Down Syndrome)	White, Non-Hispanic	145	12.53	10.57 – 14.74
	Black, Non-Hispanic	16	13.50	7.72 – 21.93
	Asian, Non-Hispanic	9	8.55	3.91 – 16.23
	Hispanic	19	9.84	5.92 – 15.37
Turner Syndrome	White, Non-Hispanic	10	0.86	0.41 – 1.59
	Black, Non-Hispanic	0	0.00	0.00 – 3.11
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	0	0.00	0.00 – 1.91
Other Chromosomal Syndromes/Other Syndromes	White, Non-Hispanic	133	11.49	9.62 – 13.62
	Black, Non-Hispanic	13	10.97	5.84 – 18.76
	Asian, Non-Hispanic	11	10.45	5.22 – 18.70
	Hispanic	17	8.81	5.13 – 14.10
Other				
Amniotic Bands	White, Non-Hispanic	6	0.52	0.19 – 1.13
	Black, Non-Hispanic	1	0.84	0.02 – 4.70
	Asian, Non-Hispanic	1	0.95	0.02 – 5.29
	Hispanic	0	0.00	0.00 – 1.91
Skin Anomalies	White, Non-Hispanic	6	0.52	0.19 – 1.13
	Black, Non-Hispanic	0	0.00	0.00 – 3.11
	Asian, Non-Hispanic	1	0.95	0.02 – 5.29
	Hispanic	0	0.00	0.00 – 1.91
Other, Specified	White, Non-Hispanic	9	0.78	0.36 – 1.48
	Black, Non-Hispanic	0	0.00	0.00 – 3.11
	Asian, Non-Hispanic	0	0.00	0.00 – 3.51
	Hispanic	2	1.04	0.13 – 3.74

Table 11 (cont'd) Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2002-2003

1. Cases can be included in more than one defect category. Cases are counted once within a defect category. Counts represent the number of defects, not the total number of cases with birth defects. Because only live births are presented on this table, case totals are not listed. Race/ethnicity is drawn from the birth certificate. Because birth certificate data are more accurate for this field than fetal death records, analyses of race/ethnicity are limited to live births.
2. Due to small numbers, races classified as "other" are excluded.

Table 12 Most Common Defects by Maternal Race/Hispanic Ethnicity, among Live Births Massachusetts: 2002-2003

Race[1] Defect[2]	Count	Rate per 10,000 Births	95% Confidence Interval
White, Non-Hispanic			
ASD (Secundum and NOS)	203	17.54	15.21—20.13
Hypospadias, 2nd or 3rd Degree	150	12.96	10.97—15.21
Trisomy 21 (Down Syndrome)	145	12.53	10.57—14.74
Polydactyly/Syndactyly	135	11.66	9.78—13.81
Club Foot	125	10.80	8.99—12.87
VSD (Membranous and NOS)	113	9.76	8.05—11.74
Obstructive Genitourinary Defect	108	9.33	7.65—11.27
Cleft Lip w/ and w/o Cleft Palate	79	6.83	5.40— 8.51
Pulmonary Stenosis, Valvular	79	6.83	5.40— 8.51
Craniosynostosis	79	6.83	5.40— 8.51
Black, Non-Hispanic			
ASD (Secundum and NOS)	41	34.60	24.83—46.94
Trisomy 21 (Down Syndrome)	16	13.50	7.72—21.93
Polydactyly/Syndactyly	13	10.97	5.84—18.76
VSD (Membranous and NOS)	11	9.28	4.63—16.61
Pulmonary Stenosis, Valvular	11	9.28	4.63—16.61
Obstructive Genitourinary Defect	9	7.59	3.47—14.42
Cleft Lip w/ and w/o Cleft Palate	9	7.59	3.47—14.42
Hypospadias, 2nd or 3rd Degree	8	6.75	2.91—13.30
Club Foot	8	6.75	2.91—13.30
Complete Atrioventricular Canal Defect	8	6.75	2.91—13.30
Asian, Non-Hispanic			
VSD (Membranous and NOS)	15	14.25	7.98—23.51
ASD (Secundum and NOS)	14	13.30	7.27—22.32
Club Foot	9	8.55	3.91—16.23
Cleft Palate w/o Cleft Lip	9	8.55	3.91—16.23
Trisomy 21 (Down Syndrome)	9	8.55	3.91—16.23
Cleft Lip w/ and w/o Cleft Palate	7	6.65	2.67—13.70
Polydactyly/Syndactyly	6	5.70	2.09—12.41
Microcephaly	5	4.75	1.54—11.09
Hypospadias, 2nd or 3rd Degree	5	4.75	1.54—11.09
Obstructive Genitourinary Defect	5	4.75	1.54—11.09
Hispanic			
ASD (Secundum and NOS)	43	22.27	16.12—30.00
VSD (Membranous and NOS)	33	17.09	11.77—24.00
Polydactyly/Syndactyly	33	17.09	11.77—24.00
Club Foot	19	9.84	5.92—15.37
Trisomy 21 (Down Syndrome)	19	9.84	5.92—15.37
Hypospadias, 2nd or 3rd Degree	17	8.81	5.13—14.10
Obstructive Genitourinary Defect	17	8.81	5.13—14.10
Cleft Lip w/ and w/o Cleft Palate	14	7.25	3.96—12.17
Pulmonary Stenosis, Valvular	13	6.73	3.59—11.51
Rectal and Large Intestinal Atresia/Stenosis	12	6.22	3.21—10.86

1. Due to small numbers, races classified as "other" are excluded. Race/ethnicity is drawn from the birth certificate. Because birth certificate data are more accurate for this field than fetal death records, analyses of race/ethnicity are limited to live births.
2. Excludes Patent Ductus Arteriosus (PDA) due to the high number of cases and the mild severity of the majority of these cases.

Chapter 6

Prevalence of Birth Defects by Severity

Prevalence of Birth Defects by Severity

Cases with birth defects were categorized by their level of severity. The severity scale was developed by the Center in collaboration with our partners at Boston University and the Massachusetts General Hospital. This scale was based on the usual outcome for a specific birth defect including its typical compatibility with survival, the need for immediate treatment, the need for long-term care, and the amenability of the defect to correction. A severity score was assigned to each case based on the most severe defect for that infant/fetus. If a case had multiple defects with equal severity, it was reviewed in detail by the Center Clinical Geneticist. Cases with a syndrome plus defect(s) are listed by syndrome only. (Syndromes are defined as a group of malformations that occur together frequently enough to be recognized collectively as a distinct abnormal condition.) Specific severity category definitions used in this report are as follows:

SEVERITY CATEGORIES	PERCENTAGE OF BIRTH DEFECTS CASES
Severe, supportive measures, usually incompatible with life	2.8%
Serious, may be correctable, most have long-term needs	16.6%
Moderate, most correctable, many have long-term needs	72.7%
Mild, may be correctable, minimal long-term needs	7.9%

Table 13 shows the distribution of cases by severity groups.

Nearly three percent of cases had birth defects classified as “severe,” and most did not survive the neonatal period. This percentage was an underestimate of these most “severe” cases due to limitations of the data, and because we are missing many “severe” defects including the estimated 80% of Anencephaly cases and 50% of any neural tube defects that are electively terminated after prenatal diagnosis (Cragan 2000).

About 17% of cases were affected with a “serious” birth defect. These cases typically require intensive medical care and planning for continuing care, and experience long-term disability.

“Moderately severe” birth defects comprised 73% of the total cases. All of these children needed medical follow up, and many needed surgeries and extensive treatment.

“Mild” birth defects comprised nearly 8% of the cases. Within the classification of “mild severity,” there was variability. For example, children with Microphthalmia (small eyes) could have mild reduction in the size of the globe or a more severe reduction resulting in visual loss or the need for intrusive ophthalmologic medical care. In contrast, babies with isolated Dextrocardia (heart in the right side of the chest instead of the left) and no other heart defect have no clinical consequence.

Table 13 Birth Defect Cases[1] by Severity
Massachusetts: 2002-2003

Severe, supportive measures, usually incompatible with life	**Count**
Anencephaly	6
Bilateral renal agenesis	3
Trisomy 13	9
Trisomy 18	22
Severe identifiable syndrome or condition, not elsewhere classified	17
Severe isolated defects, not elsewhere classified	11
Multiple severe defects, (Severe MCA,NEC)	2
Total	70

Serious, may be correctable, most have long-term needs	
Achondroplasia	12
Aniridia	3
Anophthalmia	1
Arthrogryposis	9
Biliary atresia	9
Bladder exstrophy	6
Cloacal exstrophy	2
CHD, multiple mod-severe,not elsewhere classified	1
Double outlet right ventricle	9
Encephalocele	5
Heterotaxy with CHD	7
Holoprosencephaly	4
Hypoplastic left heart syndrome	19
Limb reductions, mod-severe	0
Osteogenesis imperfecta	6
Sacral agenesis, caudal regression, sirenomelia	3
Single ventricle	1
Spina bifida	22
Amniotic band complex	10
Down syndrome	200
Mod serious syndrome/condition, not elsewhere classified	68
Mod serious defect, not elsewhere classified	9
Mod-severe multiple defects, (Mod-severe MCA,NEC)	15
Total	421

Moderate, most correctable, many have long-term needs	
Aortic valve stenosis	12
Atrial septal defect	83
Atrioventricular canal AVC / AVSD / ECD	8
Choanal atresia	3
Cleft lip/ palate	152
Coarctation	43
Cataract, glaucoma	25
Clubfoot	116
Coloboma	3
CHD, Mult mild-mod not, listed elsewhere	67
Craniosynostosis	78
Dandy-Walker malformation	16
Diaphragmatic hernia	25
Esophageal atresia/ TEF	20
Ebstein anomaly	5
Gastroschisis	33
Genitourinary, obstructive	127
Hirschsprung disease	13
Penis, buried, hidden	39
Hydrocephalus	17
Hypospadias, 2nd or 3rd degree	169
Intestinal atresia: duod, jejunal, ileal	27
Imperforate anus/rectal atresia and stenosis	40
Interrupted aortic arch	2
Klinefelter syndrome	8
Limb reductions, mild-mod	29
Malrotation	26
Microcephaly	10
Microtia	11
Omphalocele	10
Pulm sequestration/ CCAM	23
Pulmonary atresia/stenosis	86
Tethered cord	18
Tetralogy of Fallot	43
Total /partial anom. pulm venous return	14
Transposition great arteries	45
Tricuspid atresia/stenosis	4
Turner syndrome	13
Ventricular septal defect	102
Moderate syndrome/condition, not elsewhere classified	96
Moderate defect, not elsewhere classified	61
Moderate multiple severe defects, (Moderate MCA,NEC)	101
DiGeorge/ VCF/ 22q11 del spectrum	8
Goldenhar/FAVS/ OAVD	10
Total	1841

Mild, may be correctable, minimal long-term needs	
Bicuspid aortic valve	6
Meckel's diverticulum	1
Microphthalmia	2
CHD, OS, asymptomatic	4
Patent ductus arteriosus	7
Polydactyly, accessory thumbs, syndactyly	130
Heterotaxy without CHD, Situs inversus totalis without CHD, Situs inversus abdominis, isolated dextrocardia	7
Mild defect, not listed above	42
Total	199

N=2,531

[1]Birth defect totals are different from totals in Table 1 because cases with multiple defects and/or a syndrome were assigned to only one severity group.

Appendices

Technical Notes

Definitions

2002-2003 Denominators Used in Calculating Rates

Birth Defects Codes and Exclusions by Defect Category

All ICD9/BPA Codes with Counts-Live Births and Stillbirths

Glossary of Selected Birth Defect Terms

References

Technical Notes

Data Sources
Surveillance records were matched to records from the Registry of Vital Records and Statistics to gain supplemental information or to verify information on the cases. All records were matched. Birth certificate data were used as the source of information for mother's date of birth and race/ethnicity. Surveillance records provided all diagnostic and the remaining demographic information.

Prevalence, Rates and Confidence Intervals
Prevalence is defined as the number of individuals with a disease or condition over a specified period of time divided by the number of individuals at risk during the same period. The numerator is the number of cases of birth defects. Since the preferred denominator is all pregnancies and since the number of pregnancies cannot be determined, the number of total births is used as an approximation.

The rates provided in the tables are estimations of the proportion of infants born with birth defects. This rate is expressed as birth defect births per 10,000 births and is calculated by the formula:

Cases/total number live births x 10,000

Since fetal deaths are included in the numerator but not in the denominator, the result is technically a ratio, not a rate. This method of calculating rates is consistent with the national "Guidelines for Conducting Birth Defects Surveillance." (National Birth Defects Prevention Network, June 2004) Because the number of fetal deaths is so small, the inclusion of fetal deaths in the denominator does not substantially change the ratio.

The confidence interval (CI) is a method of assessing the magnitude and stability of a rate or ratio. The CI represents a range of values that has a 95% probability of including the true rate or ratio. Observed rates are subject to statistical variation. Thus, even if the underlying risk of an infant being born with a birth defect is identical in two subpopulations, the observed rates for the subpopulations may differ because of random variation. The confidence interval describes the precision of the observed rate as an estimate of the underlying risk of being born with a birth defect, with a wider interval indicating less certainty about this estimate. The width of the interval reflects the size of the subpopulation and the number of cases of birth defects. Smaller subpopulations with fewer defects lead to wider confidence intervals. The 95% confidence intervals used in the report are based on the Poisson distribution.

Assignment of Race/Ethnicity
The Center follows the recommendation of the National Center for Health Statistics of classifying births according to the self-reported race/ethnicity of the mother. The Massachusetts birth certificate records mother's race and ethnicity, including Hispanic ethnicity and was used to more accurately calculate Hispanic-specific rates of birth defect prevalence. Race/ethnicity is a self-reported item and is subject to the usual limitations of this type of information.

Calculation of 2003 Dollars
2003 dollars were calculated from the Gross Domestic Product Deflator Inflation Index, an inflation calculator for adjusting costs from one year to another using the Gross Domestic Product (GDP) Deflator inflation index of 1.0343 representing the inflation from 2001 to 2003. (Based on this calculator, costs increased from 118 million to 122 million during this period.) This inflation calculator is based on the inflation rate during the US Government Fiscal Year, which begins on October 1 and ends on September 30. http://www1.jsc.nasa.gov/bu2/inflateGDP.html.

Definitions

These definitions are derived from the Massachusetts Department of Public Health report titled Massachusetts Births, 2003.

Birthweight
The weight of an infant recorded at the time of delivery. It may be recorded in either pounds/ounces or grams.

1 pound = 453.6 grams
1,000 grams = 2 pounds and 3 ounces

Infant
A child whose age is less than one year (365 days).

Infant Death
Death of a child whose age is less than one year.

Live Birth
A live birth is any infant who breathes or shows any other evidence of life (such as beating of the heart, pulsation of the umbilical cord, or definite movement of voluntary muscles) after separation from the mother's uterus, regardless of the duration of gestation.

Neonatal
Infant under 28 days of age.

Neonatal Death
Death of a child whose age is less than 28 days.

Plurality
The number of births to a woman produced in the same gestational period. A singleton is the birth of one infant; twins represent the births of two infants, etc.

Resident Birth
The birth of an infant whose mother reports that her usual place of residence is in Massachusetts. In Massachusetts, a resident is a person with a permanent address in one of the 351 cities or towns.

Stillbirth
A stillbirth is the birth of a fetus at greater than or equal to 20 weeks gestation, or with a weight of at least 350 grams.

2002 and 2003 Populations Used in Calculating Rates

		Numbers of Live Births to MA Residents		
		2002	**2003**	**Total**
Overall		80,624	80,167	160,791
By Maternal Age	<20	4715	4695	9,410
	20-24	11,880	11,894	23,774
	25-29	18,484	18,436	36,920
	30-34	27,163	26,829	53,992
	35+	18,381	18,308	36,689
By Infant's Sex	Male	41,224	41,069	82,293
	Female	39,400	39,097	78,497
By Plurality	Singleton	76,673	76,367	153,040
	Multiple Birth	3,951	3,800	7,751
By Maternal Race/Ethnicity	White	58,136	57,604	115,740
	Black	5,948	5,902	11,850
	Hispanic	9,543	9,764	19,307
	Asian	5,300	5,224	10,524

Birth Defect Codes and Exclusions[1] by Defect Category		
Defect	ICD-9 / BPA[2]	NOTES
Central Nervous System		
Anencephaly	740.020-740.100	
Encephalocele	742.000-742.090	
Holoprosencephaly	742.260-742.267	
Hydrocephaly	742.300, 742.310, 742.380, 742.390	Postnatal diagnosis required. Exclude mild or transient hydrocephaly due to intraventricular hemorrhage; ventriculomegaly. Include if associated with prenatal infection.
Microcephaly	742.100	Include if 2 SD below the mean, adjusted for gestational age and length.
Spina Bifida	741.001-741.999	Include cases with and without associated hydrocephaly.
Spinal Cord	742.580	
Other CNS	742.200-742.250, 742.270-742.290, 742.320, 742.400-742.480, 742.900	Postnatal diagnosis required. Exclude cysts due to IVH, anoxia, postnatal infection.
Eye		
Aniridia	743.420-743.424	
Anophthalmia/Microphthalmia	743.000-743.104	Include all truly small eyes/globes, more than short palpebal fissures.
Congenital Glaucoma, Congenital Cataract	743.200-743.204, 743.320-743.326, 743.350-743.364	Exclude minor lens opacities.
Other Eye[3]	743.300-743.314, 743.340-743.344, 743.410, 743.430-743.636	Exclude blue sclera corneal opacity. Exclude long eyelashes, small palpebal fissures, tear duct cysts, blocked tear ducts; eyelid, lacrimal system and orbit anomalies.
Ear		
Anotia/Microtia	744.010-744.214	Exclude microtia Type I mild.
Other Ear[3]	744.000, 744.240, 744.250	Exclude low-set/rotated, absent ear lobes, minor anomalies.

Birth Defect Codes and Exclusions[1] by Defect Category (cont'd)		
Defect	ICD-9 / BPA[2]	NOTES
Cardiovascular		
Anomalous Pulmonary Venous Connection		
Total/Partial Anomalous Pulmonary Venous Connection	747.420, 747.430	
Atrioventricular Canal Defects		
ASD Primum	745.600	
Common Atrium	745.610	
Complete Atrioventricular Canal Defect	745.620, 745.630	
Endocardial Cushion (OS and NOS)	745.680, 745.690	
VSD, Canal Type	745.685	
Conotruncal (Outlet) and Aortic Arch		
Double Outlet Right Ventricle	745.185-745.189	
d-Transposition of the Great Arteries	745.100, 745.110	
Interrupted Aortic Arch, Type B	747.217	
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	745.200, 747.310	
Truncus	745.000	
Ebstein Anomaly		
Ebstein Anomaly	746.200	
Laterality Defects		
Heterotaxy, Situs Inversus	759.300-759.395	
Left-Sided Obstruction		
Aortic Valve Stenosis	746.300	
Coarctation of Aorta	747.100-747.190	
Hypoplastic Left Heart Syndrome	746.700, 747.200	
Interrupted Aortic Arch (Type A and NOS)	747.215, 747.216	

Birth Defect Codes and Exclusions[1] by Defect Category (cont'd)		
Defect	ICD-9 / BPA[2]	NOTES
Patent Ductus Arteriosus		
Patent Ductus Arteriosus	747.000	Exclude if on prostaglandin or gestational age <37 weeks. Include if >=37 weeks and >=6 wks when last noted or <6 wks if treated with indocin or surgery or associated with other codable defect.
Right-Sided Obstruction		
Pulmonary Stenosis, Valvular	746.010	
Pulmonary Valve Atresia w/Intact Septum	746.000	
Pulmonary Valve Atresia with VSD	746.030	
Tricuspid Valve Atresia	746.100	
Septal Defects		
ASD (Secundum, OS and NOS)	745.510, 745.580, 745.599	
VSD (Membranous and NOS)	745.485, 745.490	
VSD, Conoventricular/Malalignment	745.487	
Single Ventricle and L-TGA		
L-TGA	745.120	
Single Ventricle	745.300-745.380	
Other Cardiovascular		
Other Cardiovascular[3]	745.010, 746.080, 746.090, 746.400-746.600, 746.800-746.995, 747.210, 747.220-747.300, 747.320-747.410, 747.480-747.810, 747.880	Exclude pulmonary/tricuspid/aortic valve insufficiency/regurgitation, mitral valve congenital insufficiency. Exclude peripheral pulmonary artery stenosis with physiologic PPS (i.e. <36 wks).

Birth Defect Codes and Exclusions[1] by Defect Category (cont'd)		
Defect	ICD-9 / BPA[2]	NOTES
Respiratory		
Choanal Atresia	748.010-748.014	
Lung Anomalies[3]	748.400-748.580, 748.880	Exclude hypoplasia of lung if GA<36 weeks, or associated with space occupying lesion, diaphragmatic hernia, skeletal dysplasia, bilateral renal agenesis/oligohydramnios.
Other Respiratory[3]	748.000, 748.205, 748.310-748.385, 748.690	Exclude laryngo-tracheomalacia.
Orofacial		
Cleft Lip w/ and w/o Cleft Palate	749.101-749.290	Exclude isolated alveolar ridge, cleft gum.
Cleft Palate w/o Cleft Lip	749.001-749.090	Exclude isolated submucous cleft, bifid uvula.
Pierre Robin Sequence	524.080	
Other Orofacial[3]	744.400, 744.480, 748.120, 748.180, 750.120, 750.130	
Gastrointestinal		
Biliary Atresia	751.650	
Esophageal Atresia/Tracheoesophageal Fistula	750.300-750.330	
Hirschsprung Disease	751.300-751.340	
Rectal and Large Intestinal Atresia/Stenosis	751.200-751.240	
Small Intestinal Atresia	751.100-751.195	
Other Gastrointestinal[3]	750.600-751.010, 751.400-751.540, 751.560, 751.580, 751.660-751.800	Exclude isolated anal fistula, pyloric stenosis, unspecified anomalies of upper alimentary tract, superficial rectal fissure, tongue tie, protruding tongue.
Genitourinary		
Bladder Exstrophy	753.500	
Cloacal Exstrophy	751.550	
Hypospadias, 2nd or 3rd Degree	752.606-752.627	Exclude 1st degree hypospadias and epispadias.
Obstructive Genitourinary Defect[3]	753.200-753.290, 753.600-753.690	Include primary diagnosis with surgical intervention and secondary diagnosis with postnatal confirmation.
Renal Agenesis/Hypoplasia	753.000-753.008	Exclude isolated renal agenesis/hypoplasia.
Other Genitourinary[3]	752.000-752.480, 752.700-752.880, 753.110, 753.120, 753.160, 753.180, 753.310-753.480, 753.485, 753.700-753.880	Exclude isolated undescended testicle(s), unspecified genitourinary anomalies.

Birth Defect Codes and Exclusions[1] by Defect Category (cont'd)		
Defect	ICD-9 / BPA[2]	NOTES
Musculoskeletal		
Clubfoot	754.500, 754.520-754.735	Exclude positional, flexible, untreated (casting, surgery).
Craniosynostosis	756.000-756.024, 756.050, 756.056, 756.410	Exclude deformational plagiocephaly and other abnormal head shape w/o craniosynostosis.
Diaphragmatic Hernia	756.600-756.619	
Gastroschisis	756.710	
Omphalocele	756.700	
Polydactyly/Syndactyly	755.005-755.199	Exclude postaxial polydactyly: Type B. Exclude extra digit, NOS. Exclude accessory digits, NOS: hand/foot not specified, hand/hoot pre/postaxial not specified. Exclude isolated 2-3 toe syndactyly.
Reduction Deformity, Lower Limbs	755.300-755.390	
Reduction Deformity, Upper Limbs	755.200-755.290	
Skeletal Dysplasia	755.555, 756.430-756.590	
Other Musculoskeletal[3]	754.200-754.410, 754.510, 754.880, 755.440-755.800, 756.080-756.340,756.620, 756.680, 756.720-756.880	Exclude if flexible, untreated, positional. Exclude congenital dislocation hip. Exclude supernumerary rib in cervical region, deviated septum.
Chromosomal and Other Syndromes		
Klinefelter Syndrome	758.700-758.790	
Trisomy 13	758.100-758.190	
Trisomy 18	758.200-758.290	
Trisomy 21 (Down Syndrome)	758.000-758.090	
Turner Syndrome	758.600-758.690	
Other Chromosomal Syndromes/Other Syndromes	279.110, 756.045, 756.046, 756.055, 756.057-756.065, 756.525, 756.830, 756.850, 758.300-758.590, 758.800-758.990, 759.500, 759.610, 759.800-759.890	Exclude balanced autosomal translocation.
Other		
Amniotic Bands	658.800	
Skin Anomalies[3]	757.110-757.800	Exclude other specified, unspecified congenital anomalies of the integument. Exclude skin tags, urticaria pigmentosa, nevus not elsewhere classified (port wine, nevus flammeus, stork bite), specified anomalies of hair or nails, hypoplastic breast/nipple, absent nipple, small nipple.
Other, Specified	759.000-759.240, 759.680, 759.700	Exclude ectopic, lobulation, hyperplasia, splenomegaly, hypoplasia, misshapen, and other specified or unspecified anomalies of spleen. Exclude hypoplasia and other specified or unspecified anomalies of the adrenal gland.

[1] Other ICD 9 codes and diagnoses outside of the 740.0 - 759.9 range which are also excluded are: Syringomyelia, isolated; inguinal hernia, umbilical hernia, testicular torsion, sacral/pilonidal dimple, tibial torsion, hydroceles, webbing of neck and associated abnormalities, heart murmurs without confirmation of a structural defect.

[2] Coding scheme derives from International Classification of Diseases (ICD) 9th Revision/British Pediatric Association (BPA), 1979.

[3] Some defect(s) in this category are included only with surgical intervention or other treatment, if isolated; otherwise they require a codable defect.

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2002-2003

BPA Label	BPA Code	# of Defects
Central Nervous System		
Agyria and lissencephaly	742240	9
Anencephaly	740020	6
Brain cysts: Cerebral / subependymal / periventricular	742420	6
Brain cysts: Porencephaly / porencephalic	742410	1
Brain, reduction defect OS (8/02 Includes colpocephaly, pachygyria, schizencephaly)	742280	20
Brain, unspecified anomalies	742900	1
Brain: Other specified anomalies / cortical atrophy / cranial nerve defects	742480	6
Cerebellar Hypoplasia	742235	3
Cerebellum anomalies	742230	5
Cerebrum anomalies	742200	2
Corpus callosum anomalies	742210	44
Dandy-Walker Malformation	742310	18
Encephalocele, Occipital	742000	3
Encephalocele, Parietal	742086	3
Enlarged brain and head / enlarged head / enlarged brain / megalencephaly / macrocephaly	742400	16
Holoprosencephaly, Alobar	742265	1
Holoprosencephaly, Lobar	742267	5
Holoprosencephaly, NOS	742260	2
Hydranencephaly	742320	3
Hydrocephaly, NOS	742390	22
Hydrocephaly, Anomalies of Aqueduct of Sylvius	742300	11
Hydrocephaly, Other Specified	742380	9
Lipomeningocele, Highest level, lumbar, No mentioned hydrocephalus, open	741753	1
Lipomeningocele, Highest level, sacral, No mentioned hydrocephalus, closed	741854	1
Lipomeningomyelocele, Highest level, lumbar, No mentioned hydrocephalus, closed	741843	3
Meningocele, Highest level unspecified, No mentioned hydrocephalus, open	741719	1
Meningocele, Highest level, sacral, Arnold Chiari malformation ± hydrocephalus, open	741014	1
Meningocele, Highest level, thoracic, No mentioned hydrocephalus, closed	741812	1
Meningomyelocele/myelomeningocele, Highest level unspecified, Arnold Chiari malformation ± hydrocephalus, open	741009	1
Meningomyelocele/myelomeningocele, Highest level unspecified, No mentioned hydrocephalus, unspecified open/closed	741909	1
Meningomyelocele/myelomeningocele, Highest level, lumbar, Arnold Chiari malformation ± hydrocephalus, closed	741103	1
Meningomyelocele/myelomeningocele, Highest level, lumbar, Arnold Chiari malformation ± hydrocephalus, open	741003	9
Meningomyelocele/myelomeningocele, Highest level, lumbar, Hydrocephalus, other (aqueduct of Sylvius) or NOS, open	741303	1
Meningomyelocele/myelomeningocele, Highest level, sacral, No mentioned hydrocephalus, closed	741804	1
Meningomyelocele/myelomeningocele, Highest level, sacral, No mentioned hydrocephalus, open	741704	1
Meningomyelocele/myelomeningocele, Highest level, thoracic, Hydrocephalus, other (aqueduct of Sylvius) or NOS, open	741302	1
Microcephalus	742100	32
Microgyria / polymicrogyria	742250	6
Myelocele, Highest level, lumbar, Arnold Chiari malformation ± hydrocephalus, unspec. open/closed	741223	1
Spinal cord: Other specified anomalies (Includes tethered cord)	742580	54
Unspecified spina bifida, Highest level unspecified, No mentioned hydrocephalus, closed	741899	1

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2002-2003

BPA Label	BPA Code	# of Defects
Eye		
Absence of iris/Aniridia, Bilateral	743424	4
Anophthalmos, Bilateral	743004	1
Anophthalmos, Left	743001	1
Anophthalmos, Right	743002	2
Buphthalmos/Congenital Glaucoma, Bilateral	743204	5
Buphthalmos/Congenital Glaucoma, Left	743201	2
Cataract, anterior polar, Bilateral	743354	1
Cataract, anterior polar, Right	743352	1
Cataract, NOS, Laterality Unk	743320	1
Cataract, NOS, Left	743321	4
Cataract, NOS, Bilateral	743324	8
Cataract, NOS, Right	743322	6
Cataract, other specified, Bilateral	743364	4
Cataract, other specified, Right	743362	1
Microphthalmos, Bilateral	743104	7
Microphthalmos, Left	743101	1
Microphthalmos, Right	743102	4
Anterior segment: OS colobomas and anomalies	743480	2
Choroid: Coloboma	743535	4
Cornea, other anomalies. Excludes: megalocornea (use 743220)	743410	1
Eyelids: Coloboma	743636	5
Iris: Coloboma	743430	5
Optic disc: Specified anomalies / hypoplastic optic nerve / coloboma of the optic disc	743520	5
Retina: Specified anomalies / congenital retinal aneurysm.	743510	3
Vitreous humor: Specified anomalies (includes PHPV)	743500	7
Ear		
Anotia, Right	744012	1
Microtia, Bilateral	744214	6
Microtia, Left	744211	7
Microtia, Right	744212	5
Cardiovascular		
Anomalous Pulmonary Venous Connection		
Partial anomalous pulmonary venous return/connection/drainage	747430	5
Total anomalous pulmonary venous return/connection/drainage	747420	16
Atrioventricular Canal Defects		
Atrial septal defect, primum type (ASD1)	745600	14
Common Atrium	745610	4

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2002-2003

BPA Label	BPA Code	# of Defects
Complete atrioventricular canal (CAVC)	745630	37
Complete atrioventricular canal (CAVC) with ventricular septal defect	745620	10
Endocardial cushion defect, NOS	745690	2
Endocardial cushion defect, Other specified	745680	7
Ventricular septal defect, inflow type (subtricuspid, canal-type) (VSDavc)	745685	11
Conotruncal (Outlet) and Aortic Arch		
Dextro-transposition of great arteries (dTGA, dTGV) w/ intact ventricular septum	745100	24
Dextro-transposition of great arteries (dTGA, dTGV) w/ ventricular septal defect	745110	27
Double-outlet right ventricle (DORV) with normally related great arteries	745185	8
Double-outlet right ventricle (DORV) with transposed great arteries	745186	4
Double-outlet right ventricle (DORV), NOS	745189	5
Double-outlet right ventricle (DORV), Other Specified	745188	3
Interrupted aortic arch, type B	747217	4
Pulmonary atresia with VSD (tetralogy of Fallot with pulmonary atresia)	747310	12
Tetralogy of Fallot	745200	53
Truncus Arteriosus	745000	3
Ebstein Anomaly		
Ebstein Malformation or Anomaly	746200	7
Heterotaxy (Laterality Defects)		
Complete situs inversus w/ dextrocardia	759300	5
Situs ambiguus, right; right isomerism	759350	1
Situs ambiguus, sidedness NOS	759380	2
Situs ambiguus, sidedness unclear	759370	2
Situs inversus abdominis	759330	1
Situs inversus w/ levocardia	759310	1
Left-Sided Obstruction		
Aortic stenosis, valvar	746300	34
Coarctation of the aorta (COA), postductal (distal)	747110	3
Coarctation of the aorta (COA), preductal (proximal)	747100	2
Coarctation of the aorta, juxtaductal	747120	7
Coarctation of the aorta, NOS	747190	62
Hypoplastic left heart syndrome	746700	20
Interrupted aortic arch, type A	747216	4
Patent Ductus Arteriosus		
Patent ductus arteriosus (PDA)	747000	269
Right-sided Obstruction		
Pulmonary valve atresia with VSD (not TOF variant 747310)	746030	6

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2002-2003

BPA Label	BPA Code	# of Defects
Pulmonary valve atresia/intact ventricular septum	746000	14
Pulmonic stenosis, valvar	746010	105
Tricuspid atresia	746100	13
Septal Defects		
Atrial septal defect, NOS	745599	96
Atrial septal defect, OS	745580	2
Atrial septal defect, Secundum type (ASD2)	745510	215
Ventricular septal defect, NOS	745490	13
Ventricular septal defect, Malalignment-type (type I, subarterial) (VSDmal)	745487	30
Ventricular septal defect, Perimembranous (type II, membranous) (VSDmem)	745485	166
Single Ventricle and L-TGA		
L-TGA /Corrected transposition of great vessels / ventricular inversion	745120	8
Single ventricle, Double Inlet Left Ventricle	745310	2
Other Cardiovascular		
"Pulmonic" or pulmonary atresia, stenosis, or hypoplasia, NOS w/ no mention of whether valve of artery	746995	2
Anomalies of coronary artery or sinus	746885	10
Aorta: Hypoplasia	747210	14
Aorta: Congenital aneurysm / dilatation	747270	1
Aorta: Persistent right aortic arch	747230	32
Aorta: Vascular ring / double aortic arch / vascular ring compression of trachea	747250	10
Aortic septal defect / aortopulmonary window	745010	1
Aortic valve: bicuspid BAV	746400	66
Aortic valve: Other specified anomalies / aortic valve atresia	746480	18
Cerebral vessels: Other anomalies / vein of Galen	747810	1
Cor triatriatum	746820	1
Great veins: Other specified anomalies (includes IVC interruption, bilateral SVC)	747480	13
Heart: Other specified anomalies / ectopia cordis / mesocardia / conduction defects, NOS	746880	47
Hypoplastic left ventricle. Excludes: hypoplastic left heart syndrome (746700)	746881	6
Hypoplastic right heart or right ventricle / Uhl's disease (parchment RV)	746882	3
Mitral valve: Absence, atresia, or hypoplasia	746505	9
Mitral valve: Congenital mitral stenosis	746500	10
Mitral valve: insufficiency or regurgitation, congenital	746600	1
Persistent left superior vena cava	747410	32
Pulmonary infundibular (subvalvular) stenosis	746830	12
Pulmonary valve: Other specified anomalies. Excludes: infundibular PS (746830)	746080	19
Circulatory system: Other specified anomalies	747880	2
Peripheral arteries: Other anomalies / aberrant subclavian artery	747640	21
Pulmonary artery: atresia, absence or agenesis. Use 746995 if artery or valve is not specified	747300	2
Pulmonary artery: other specified / pulmonary artery hypoplasia	747380	5
Pulmonary artery: stenosis. Use 746.995 if artery or valve is not specified	747320	16
Situs: Dextrocardia without situs inversus / dextrocardia with situs solitus	746800	6
Supra-aortic stenosis / supravalvular aortic stenosis. Excludes: aortic stenosis, congenital (see 746300)	747220	1
Valves: Unspecified anomalies	746900	1

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2002-2003

BPA Label	BPA Code	# of Defects
Respiratory		
Choanal atresia, Bilateral	748014	2
Choanal atresia, Right	748012	2
Choanal stenosis	748000	5
Congenital subglottic stenosis	748310	1
Larynx: Cleft / laryngotracheoesophageal cleft	748385	3
Larynx: Web, glottic	748205	1
Other anomalies of trachea	748330	3
Hypoplasisa of lung or pulmonary hypoplasia	748510	2
Agenesis or aplasia of lung	748500	1
Lung cysts: CCAM (cong cystic adenomatoid malf), OS	748480	14
Other anomalies of bronchus	748350	1
Other specified dysplasia of lung / fusion of lobes of lung	748580	1
Other specified respiratory system anomalies / congenital lobar emphysema / lymphangiectasia of lung	748880	2
Sequestration of lung	748520	9
Orofacial		
Cleft hard palate, Bilateral	749010	6
Cleft hard palate, Central	749020	5
Cleft hard palate, NOS	749030	18
Cleft lip and palate, Bilateral cleft lip	749210	13
Cleft lip and palate, NOS	749290	5
Cleft lip and palate, Unilateral cleft lip, Left	749201	32
Cleft lip and palate, Unilateral cleft lip, Right	749202	14
Cleft lip, Bilateral	749110	6
Cleft lip, NOS	749195	4
Cleft lip, Unilateral, Left	749101	16
Cleft lip, Unilateral, Right	749102	17
Cleft palate, NOS	749090	19
Cleft soft palate, Bilateral	749050	2
Cleft soft palate, Central	749060	2
Cleft soft palate, NOS	749070	40
Cleft soft palate, Unilateral, Left	749041	2
Cleft soft palate, Unilateral, Right	749042	1
Cleft: Incomplete CL/ microform /pseudo / fused lip /healed lip	749190	4
Face or neck: Other specified anomalies (6/03 eg. facial cleft)	744880	6
Nose: Fissured, notched, or cleft	748120	1
Nose: OS anomalies: small nose and nostril / absent nasal septum / flat or wide nasal bridge/ beaked nose	748180	14
Pierre Robin sequence	524080	35
Branchial cleft, sinus, fistula, cyst, or pit	744400	20
Other branchial cleft anomalies / dermal sinus of head	744480	1
Tongue: Dislocation or displacement / glossoptosis	750130	7
Tongue: large / macroglossia	750120	4

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2002-2003

BPA Label	BPA Code	# of Defects
Gastrointestinal		
Meckel's diverticulum	751010	8
Anal atresia with fistula	751230	36
Anal atresia without mention of fistula	751240	21
Annular pancreas	751720	7
Biliary atresia, extrahepatic or NOS (use 751670 for intrahepatic)	751650	10
Choledochal cysts	751660	3
Duodenal web	751560	5
Duplication of anus, appendix, cecum, or intestine / enterogenous cyst	751500	2
Ectopic (displaced, anteriorly placed) anus	751530	12
Esophageal atresia with TE fistula	750310	31
Esophageal atresia without TE fistula	750300	3
Hirschsprung disease, NOS	751330	5
Hirschsprung disease: Long-segment (aganglionosis beyond rectum)	751310	8
Hirschsprung disease: Short-segment (aganglionosis involving no more than the anal sphincter and the rectum)	751320	5
Intestinal atresia/stenosis, Duodenum	751100	20
Intestinal atresia/stenosis, Ileum	751120	9
Intestinal atresia/stenosis, Jejunum	751110	16
Intestinal atresia/stenosis, Large Intestine, NOS	751200	1
Intestinal atresia/stenosis, Small Intestine, NOS	751190	1
Malrotation: cecum and/or colon	751400	1
Malrotation: Other specified and unspecified	751490	44
Malrotation: small intestine alone	751495	8
Microcolon	751520	1
Other specified anomalies of intestine / Rectal fissures	751580	1
Other specified anomalies of upper alimentary tract	750800	1
Persistent omphalomesenteric duct / persistent vitelline duct	751000	4
Rectal atresia/stenosis with fistula	751210	1
Rectal atresia/stenosis without mention of fistula	751220	5
Tracheoesophageal fistula without mention of esophageal atresia	750320	1
Tracheoesophageal fistula, "H" type	750325	4
Genitourinary		
Urachus: Patent	753700	1
Absence of bladder or urethra	753800	1
Bladder exstrophy	753500	6
Cloacal exstrophy	751550	2
Double urethra or urinary meatus	753840	2
Genital organs: Other specified anomalies / microgenitalia / macrogenitalia	752880	1
Gyne: Hymen Imperforate	752430	2
Gyne: OS anomalies of cervix, vagina, or external female genitalia / Vaginal tags / Hymenal tags	752480	4
Gyne: Ovaries, Multiple cysts	752085	2
Gyne: Uterus absence or agenesis	752300	2
Gyne: Uterus fistulae connecting with digestive or urinary tract/ uterointestinal fistula / uterovesical fistula	752320	1
Gyne: Uterus, other anomalies / bicornuate/ unicornis	752380	7
Gyne: Vagina, absence or atresia complete or partial	752410	2
Hypospadias, Second Degree	752606	64

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2002-2003

BPA Label	BPA Code	# of Defects
Hypospadias, Second Degree with Chordee	752626	76
Hypospadias, Third Degree	752607	11
Hypospadias, Third Degree with Chordee	752627	30
Indeterminate sex, NOS / ambiguous genitalia	752790	15
Kidney: Lobulated, fused, or horseshoe / crossed fused ectopia	753320	6
Kidney: Other specified disease / cystic NOS	753180	1
Kidneys: Multicystic renal dysplasia / multicystic kidney	753160	24
Kidneys: Polycystic, adult type (APKD)	753120	1
Kidneys: Polycystic, infantile type (IPKD)	753110	3
Obstruction, atresia or stenosis of urinary meatus / meatal stenosis	753630	1
Other and unspecified atresia and stenosis of urethra and bladder neck	753690	1
Other specified anomalies of bladder and urethra	753880	2
Penis: Other anomalies / concealed penis / absent or hooded foreskin	752860	54
Penis: Small / hypoplastic / micropenis	752865	9
Renal agenesis, bilateral	753000	1
Renal agenesis, left + renal hypoplasia, right	753006	1
Renal hypoplasia, bilateral	753005	1
Atresia, stricture, or stenosis of ureter / ureteropelvic junction obstruction or stenosis /ureterovesical junction obstruction or stenosis / hypoplastic ureter	753210	58
Congenital hydronephrosis / pyelocaliectasis	753200	122
Kidney: Double or triple, pelvis / pyelon duplex or triplex	753310	7
Kidney: Ectopic / pelvic	753330	7
Kidney: Enlarged, hyperplastic, or giant	753340	1
Megaloureter, NOS / hydroureter	753220	27
Other and unspecified obstructive defects of renal pelvis and ureter	753290	2
Ureter: Accessory / double ureter / duplex collecting system	753410	27
Ureter: Ectopic	753420	17
Ureter: Other specified anomalies / ureterocele	753480	17
Ureter: Variations of vesicoureteral reflux	753485	19
Testis and scrotum: Other anomalies / polyorchidism / bifid scrotum	752820	6
Urachus: Cyst	753710	9
Urethra: Congenital posterior urethral valves or posterior urethral obstruction	753600	14

Musculoskeletal

Achondroplasia	756430	12
Arthrogryposis multiplex congenita / distal arthrogryposis syndrome. Includes: one or more flexion contractures of individual joints	755800	22
Cleidocranial dysostosis	755555	1
Infantile cortical hyperostosis / Caffey syndrome	756530	1
Osteogenesis imperfecta	756500	8
Osteopetrosis / Albers-Schonberg syndrome / marble bones	756540	1
Other specified chondrodystrophy. Excludes: Conradi's (use 756575)	756480	2
Other specified osteodystrophies	756580	1
Spondyloepiphyseal dysplasia	756460	1
Thanatophoric dwarfism	756447	1
Absence of foot or toes, Left	755346	5
Absence of foot or toes, Right	755347	3
Absence of hand or fingers, Bilateral	755249	3
Absence of hand or fingers, Left	755246	19

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BPA Label	BPA Code	# of Defects
Absence of hand or fingers, Right	755247	3
Absence of the forearm and hand, Right	755242	2
Absence of thigh and lower leg only (foot present), Left	755321	1
Congenital postural scoliosis	754200	18
Craniosynostosis, Coronal, Bilateral	756014	6
Craniosynostosis, Coronal, Left	756011	4
Craniosynostosis, Coronal, Right	756012	9
Craniosynostosis, Lambdoidal, Bilateral	756024	1
Craniosynostosis, Lambdoidal, Left	756021	1
Craniosynostosis, Lambdoidal, Right	756022	2
Craniosynostosis, Metopic	756006	14
Craniosynostosis, Sagittal	756005	56
Craniosynostosis, Unspecified Type, Bilateral	756004	1
Diaghragmatic hernia, Morgagni, Bilateral	756619	1
Diaphragmatic hernia, Bochdalek, Left	756611	4
Diaphragmatic hernia, Esophageal	756605	1
Diaphragmatic hernia, Morgagni, Laterality Unk	756615	2
Diaphragmatic hernia, Morgagni, Left	756616	1
Diaphragmatic hernia, Morgagni, Right	756617	3
Diaphragmatic hernia, NOS (inc. absent/hemidiaphragm), Bilateral	756604	2
Diaphragmatic hernia, NOS (inc. absent/hemidiaphragm), Laterality Unk	756600	1
Diaphragmatic hernia, NOS (inc. absent/hemidiaphragm), Left	756601	20
Diaphragmatic hernia, NOS (inc. absent/hemidiaphragm), Right	756602	2
Fibular aplasia/hypoplasia, Left	755371	2
Gastroschisis	756710	39
Limb deficiencies, Lower Limb, Not Elsewhere Classifiable, Left	755381	1
Omphalocele	756700	31
Poland syndrome or anomaly	756800	5
Radial aplasia/hypoplasia, Bilateral	755269	1
Radial aplasia/hypoplasia, Left	755266	1
Radial aplasia/hypoplasia, Right	755267	2
Anomalies of fingers / camptodactyly/ macro- / brachy-/ clino-, triphalangeal thumb	755500	31
Bowing, femur	754400	1
Clubfoot, NOS / talipes, NOS	754730	62
Clubfoot: Metatarsus varus or adductus	754520	18
Clubfoot: Talipes calcaneovarus	754510	4
Clubfoot: Talipes equinovarus	754500	95
Eventration of diaphragm	756620	3
Lower limb: hypoplasia / Toes, feet, legs: hypoplasia. Excludes: aplasia of or absent lower limb	755685	7
Lower limb: other specified anomalies / hyperextended legs / shortening of legs	755680	8
Other absent or hypoplastic muscle / absent pectoralis major. Excludes: prune belly syndrome (use 756720)	756810	3
Other specified deformities of ankle and / or toes / dorsiflexion of foot. Excludes: widely spaced first and second toes (use 755600)	754780	3
Other specified deformity of hands (see 755.500 for specified anomalies of fingers)	754880	1
Polydactyly fingers / postaxial polydactyly, Type A	755005	27
Polydactyly: Accessory big toe (preaxial)	755030	15
Polydactyly: Accessory digits foot, NOS (preaxial, postaxial not specified)	755096	4
Polydactyly: Accessory digits hand, NOS (preaxial, postaxial not specified)	755095	5
Polydactyly: Accessory thumbs (preaxial polydactyly)	755010	45
Polydactyly: Accessory toes (postaxial)	755020	43

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BPA Label	BPA Code	# of Defects
Ribs: Absence	756300	8
Ribs: Extra	756330	5
Ribs: Fused	756320	7
Ribs: Other anomalies	756340	6
Syndactyly: Fused fingers	755100	18
Syndactyly: Fused toes	755120	41
Syndactyly: Unspecified (see below for specified site)	755190	2
Syndactyly: Unspecified (webbed vs. fused) thumb and / or fingers, NOS	755193	16
Syndactyly: Unspecified (webbed vs. fused), digits not known	755199	2
Syndactyly: Unspecified toes	755194	8
Syndactyly: Webbed fingers	755110	21
Syndactyly: Webbed toes / webbing between the second and third toes	755130	23
Talipes calcaneovalgus	754600	3
Upper limb: Hypoplasia / Fingers, hands, or arms: hypoplasia. Excludes: aplasia or absent upper limb (see 7552)	755585	14
Upper limb: Other specified anomalies / hyperextensibility of upper limb / shortening of upper limb	755580	9
Sacral agenesis	756175	4
Skull and face bone: Other specified anomalies / localized skull defects / mid-facial hypoplasia / prominent maxilla/hypotelorism / flatTocciput / prominent occiput	756080	2
Spine: Kyphosis / kyphoscoliosis	756120	3
Split-Foot, Left	755356	1
Split-Foot, Right	755357	2
Split-Hand, Bilateral	755259	5
Split-Hand, Left	755256	1
Split-Hand, Right	755257	2
Syndactyly: Unspecified, laterality not spec.	755196	1
Thumb only missing or hypoplastic, Left	755261	2
Thumb only missing or hypoplastic, Right	755262	1
Total absence of the arm, Bilateral	755209	1
Total absence of the arm, Right	755207	1
Transverse deficiency or amputation of the arm, NOS, Bilateral	755204	1
Transverse deficiency or amputation of the leg, NOS, Right	755302	1
Upper limb, Phocomelia, NOS, Left	755211	1
Vertebrae, cervical: anomalies	756140	3
Vertebrae, lumbar: anomalies	756160	3
Vertebrae, lumbar: hemivertebrae	756165	6
Vertebrae, sacrococcygeal: anomalies / agenesis of sacrum	756170	6
Vertebrae, thoracic: anomalies	756150	12
Vertebrae, thoracic: hemivertebrae	756155	12
Vertebrae: Other specified anomalies	756180	3

Chromosomal and Other Syndromes

Apert syndrome / Acrocephalosyndactyly types I or II	756055	1
Autosome NOS: Other spec anomalies / marker / 8/02: Ring, derivative, mosaic, isochrome, "additional" material / 3/03 inversions	758580	16
Balanced autosomal translocation in normal individual	758400	1
Deletion 13q / deletion of long arm of 13	758330	1
Deletion 17p or 18p / deletion of short arm chromosome 17 or 18	758350	3
Deletion 17q or 18q / deletion of the long arm of chromosome 17 or 18	758340	2
Deletion 4p / Wolff-Hirschorn syndrome	758320	1
Deletion 5p / Cri du chat syndrome	758310	4

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BPA Label	BPA Code	# of Defects
Deletion: Autosome (not X or Y)(ie. #1-16, 4q,5q,19,20) / 8/02 Include microdeletion / 10/02: use for 22q11 (also code phenotypes DGS and VCFS, if available)	758380	16
Down syndrome diagnosed clinically, but no karyotype report in medical record	758090	1
Down syndrome: mosaic	758040	1
Down syndrome: translocation trisomy - duplication of a G group chromosome, NOS	758030	2
Down syndrome: trisomy 21	758000	196
Ehlers-Danlos syndrome	756850	1
Ellis-van Creveld syndrome	756525	1
Goldenhar syndrome / oculoauriculovertebral dysplasia	756060	6
Hemifacial microsomia	756065	5
Kartagener syndrome or triad	759340	1
Klinefelter syndrome: 47, XXY	758700	8
Mosaic XO/XX. Excludes: with Turner phenotype (758610)	758810	3
Mosaic XO/XY, 45X/46XY. Excludes: with Turner phenotype (758610)	758800	2
Nail-patella syndrome	756830	1
Other craniofacial syndromes / Hallermann-Streiff syndrome	756046	2
Other specified acrocephalosyndactylies	756057	1
Sex chromosome: Other specified anomaly / fragile X	758880	4
Specified syndromes, not elsewhere classified, involving skin anomalies	757300	1
Treacher-Collins syndrome / Mandibulofacial dysostosis	756045	2
Triploidy	758586	6
Trisomy 13: Patau syndrome	758100	8
Trisomy 13: translocation trisomy with duplication of a 13q	758120	1
Trisomy 18: Edwards syndrome	758200	21
Trisomy 18: Translocation trisomy with duplication of 18q	758220	1
Trisomy, partial / 8/02 "partial trisomy" = "duplication". But, for "dup NOS" use 758930	758530	4
Trisomy: 6, 7, 9, 10, 11, 12 / Other trisomy C (archaic)	758510	4
Trisomy: Other total trisomy syndromes / trisomy 22 / trisomy, NOS	758520	2
Tuberous sclerosis / Bourneville's disease	759500	4
Turner syndrome: karyotype 45,X [XO] Note: The 7586xx code series that follows excludes pure gonadal dysgenesis(752720)	758600	5
Turner syndrome: variant karyotypes, eg. isochromosome, mosaic (eg X, XX,XY), partial X deletion, ring X chromosome. Excludes: Turner phenotype with normal karyotype	758610	8
Unbalanced translocations, OS. Excludes: bal trans in normal (75.400)	758540	6
Unspecified chromosome: Duplication of chromosome(s), NOS	758930	1
XXX female / 47XXX / Triple X syndrome	758850	10
XYY, male / 47,XYY / mosaic XYY male	758840	4
DiGeorge Syndrome: 10/02: Code specific phenotype + chrome/FISH 22, if available	279110	8
Malf OS: VATER/VACTERL/Acardia/ Angelman/Bloom/CHARGE/hemihyper/Meckel-Gruber/Neu-Laxova/PentalogyCantrell/ Sotos/ TownesBrock/ WalkerWarburg/ Weaver / 10/02 VCFS,Shprintzen	759890	27
Malf. Syndromes/face: Aarskog /BOF /BOR /Fraser /FreemanSheldon / Kabuki / Miller-Dieker/ Noonan /Opitz G / oral-facial-digita/ Oto-palato-digital / Septo-optic dysplasia / Waardenburg / Williams	759800	19
Malf. Syndromes/limbs: Baller-Gerold/ Carpenter / caudal regression /Fryns/ Holt-Oram / Klippel-Trenaunay-Webe/ LimbBodyWall /Roberts/ Rubinstein-Taybi / sirenomelia / thrombocytopenia-absent radius	759840	5
Malf. Syndromes/metabolic: Alagille /Alport / Beckwith-Wiedemann / Johansen-Blizzard/ leprechaunism / Lowe/ Menkes(kinky hair) /Prader-Willi/ Zellweger	759870	22
Malf. Syndromes/other skeletal: Marfan / Stickler/ Beemer Langer	759860	4
Malf. Syndromes/short stature: Smith-Lemli-Optiz /de Lange / Cockayne / Laurence-Moon-Biedl / Russell-Silver / Seckel	759820	4

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Other

BPA Label	BPA Code	# of Defects
Ectodermal dysplasia. Excludes: Ellis-van Creveld syndrome (756.525)	757340	1
Epidermolysis bullosa	757330	2
Other and unspecified ichthyosis	757190	1
Amniotic band sequence	658800	10
Anomalies of thymus / absent thymus / Thymichypertrophy	759240	3
Skin: Other specified anomalies / scalp defects. For specified anomalies of skin see 757390. For specified anomalies of hair, see 757480. For specified anomalies of nails 757580	757800	3
Spleen: Absence / asplenia	759000	3
Spleen: Accessory / 8/02 Use for polysplenia, though not exactly the same	759040	4
Spleen: Hypoplasia	759010	1
Thyroglossal duct anomalies / thyroglossal cyst	759220	2

Glossary of Selected Birth Defects Terms[1]

Agenesis, aplasia: Congenital absence of a body part or organ, implying that the structure never formed. Result of an error in development, as opposed to an external process.

Agenesis corpus callosum: Congenital absence of the part of the brain which connects the two cerebral hemispheres.

Amniotic band sequence: Highly variable group of defects (or single defect) due to encirclement (strangulation) of a body part by strands of a fragmented amniotic sac. Includes terminal transverse limb defects, clefts and body wall defects.

Anencephaly: Congenital absence of the skull and brain.

Aniridia: Congenital complete absence of the iris of the eye.

Anophthalmia: Congenital complete (or essentially complete) absence of the eye globe.

Anotia: Congenital absence of the ear.

Aortic valve stenosis: Congenital heart defect characterized by aortic valve narrowing reducing the flow of blood.

Arthrogryposis: Multiple congenital contractures of various joints.

Atresia / Imperforation: Congenital absence or closure of a normal opening (valve or lumen).

Atresia or stenosis of large intestine, rectum and anus: Congenital absence, closure or constriction of the large intestine, rectum or anus (commonly known as **imperforate anus**).

Atresia or stenosis of small intestine: Congenital absence, closure or constriction of the small intestine (**duodenal, jejunal, ileal atresia/stenosis**).

Atrial Septal Defect (ASD): Congenital heart defect characterized by one or more openings in the atrial septum (wall between the right and left atria). Most common type is called **ASD, secundum**.

Biliary atresia: Congenital absence of the ducts in the biliary tract.

Birth defect: Congenital abnormalities of structure, function or metabolism present before birth.

Bladder exstrophy: Congenital exposure of the bladder mucosa caused by incomplete closure of the anterior bladder wall and the abdominal cavity.

Branchial cleft, fistula, tag, cyst: Congenital abnormality of the neck or area just below the collarbone (clavicle). Includes skin pits (cleft), tissue tags, or cysts.

Cataract: Congenital opacity (clouding) of the lens of the eye.

Choanal atresia, choanal stenosis: Congenital absence (or narrowing) of the passageway between the nose and pharynx due to a thick bone or thin “membranous” bone.

Cleft lip: Congenital defect of the upper lip in which there is incomplete closure.

Cleft palate: Congenital defect in the closure of the palate; the structure which separates the nasal cavities and the back of the mouth. May involve the soft palate, hard palate or alveolus (gum).

Coarctation of the aorta: Congenital heart defect characterized by narrowing of the descending aorta. Usually occurs as an indentation at a specific location, less commonly diffuse narrowing.

Congenital: Abnormality or problem present at birth. Includes defects detected prenatally and those not recognized until after the newborn period.

Congenital heart defect (CHD), cardiovascular malformation (CVM): Abnormal heart structure present at birth. Includes defects detected prenatally, and those recognized after the newborn period.

Craniosynostosis: Congenital abnormality of skull shape due to premature fusion of the sutures between the skull bones. Head may be elongated, foreshortened, tower-like or asymmetrically flattened.

Dandy-Walker malformation: Congenital defect of the cerebellum involving a small cerebellar vermis and cystic dilation of the fourth ventricle.

Diaphragmatic hernia: Congenital defect of the muscular diaphragm resulting in herniation of the abdominal contents into the chest. Incomplete, asymptomatic variation is called eventration.

Down syndrome (Trisomy 21): Distinctive and common chromosome abnormality syndrome caused by an extra copy of chromosome 21. Can be complete (Trisomy 21), attached to another chromosome (translocation), or mixed with cells containing normal chromosomes (mosaic).

Dysplasia: Abnormal cell organization of an organ. Usually congenital, may be acquired.

Ebstein anomaly: Congenital heart defect characterized by downward displacement of the tricuspid valve into the right ventricle, associated with tricuspid valve regurgitation.

Encephalocele: Congenital defect of the skull resulting in herniation (protrusion) of the brain.

Endocardial cushion defect (ECD), atrioventricular canal (AVC) defect, atrioventricular septal defect (AVSD): Congenital heart defect characterized by a combined atrial and ventricular septal defect, and common atrioventricular valve (instead of distinct tricuspid and mitral valves). In contrast to complete AVC, the partial AVC includes an atrial septal defect, primum type, plus a cleft mitral valve.

Esophageal atresia: Congenital discontinuity of the lumen of the esophagus. Usually associated with a tracheoesophageal fistula (TEF) which is an abnormal connection between the esophagus and trachea.

Fistula: Abnormal connection between an internal organ and the body surface, or between two internal organs or structures. Can be congenital or acquired.

Gastroschisis: Congenital opening of the abdominal wall with protrusion of the abdominal contents. Can be distinguished from omphalocele by location usually to the right of the umbilicus.

Heterotaxy (situs anomalies): Congenital malposition of the abdominal organs often associated with a congenital heart defect.

Hirschsprung disease: Congenital aganglionic megacolon (enlarged colon) due to absent nerves in the wall of the colon.

Holoprosencephaly: Spectrum of congenital defects of the forebrain due to failure of the brain to develop into two equal halves. Includes alobar (single ventricle), semilobar and lobar types.

Hydrocephalus: Accumulation of fluid within the spaces of the brain. Can be congenital or acquired.

Hydronephrosis: Enlargement of the urine-filled chambers (pelves, calyces) of the kidney

Hyperplasia: Overgrowth due to an increase in the number of cells of tissue.

Hypertrophy: Overgrowth due to enlargement of existing cells.

Hypoplasia: Small size of organ or part due to arrested development.

Hypoplastic left heart syndrome (HLHS): Congenital heart defect characterized by extreme smallness of left-sided structures. Classically, aortic valve/mitral valve atresia or marked hypoplasia, ascending aorta and left ventricle hypoplasia.

Hypospadias: Congenital defect of the penis in which the urethral meatus (urinary outlet) is not on the glans (tip). Severity based on location from shaft to scrotum and perineum.

Limb deficiency, upper (arms) / lower (legs): Congenital absence of a portion or entire limb. Types include transverse (resembling an amputation), longitudinal (missing ray) and intercalary (missing bone in-between).

Macrocephaly: Large head due to extra fluid or extra volume.

Meninges: Membranes that cover the brain and spinal cord.

Microcephaly: Small head, with corresponding smallness of the brain.

Microphthalmia: Congenital smallness of the eye globe.

Microtia: Congenital smallness or maldevelopment of the external ear, with or without absence or narrowing of the external auditory canal.

Mosaic: In genetics, two or more different chromosome types in cell lines. Proportion of normal to abnormal cells usually correlated to severity.

Neural tube defect (NTD): Congenital opening from head to the base of the spine resulting from failure of the neural tube to close in the first month of pregnancy. Includes anencephaly, spina bifida, and encephalocele.

NOS: Not Otherwise Specified

Obstructive genitourinary defect: Congenital narrowing or absence of the urinary tract structure at any level. Severity often depends upon the level of the obstruction. Often accompanied by hydronephrosis.

Omphalocele: Congenital opening of the abdominal wall with protrusion of the abdominal contents. Can be distinguished from gastroschisis by location within umbilical ring.

Patent ductus arteriosus (PDA): Congenital heart defect characterized by persistence of the fetal blood vessel connecting the pulmonary artery and the aorta.

Polydactyly: Extra fingers or toes which may be medial (pre-axial) or lateral (postaxial).

Pulmonary atresia: Congenital heart defect characterized by absence of the pulmonary valve or pulmonary artery itself. May occur with an intact ventricular septum (PA/IVS) or with a ventricular septal defect, in which it is more properly called Tetralogy of Fallot with pulmonary atresia (TOF/PA).

Pulmonary stenosis (PS): Congenital heart defect characterized by narrowing of the pulmonary valve.

Renal agenesis: Congenital absence of the kidney.

Spina bifida: Neural tube defect with protrusion of the spinal cord and/or meninges. Includes myelomeningocele (involving both spinal cord and meninges) and meningocele (involving just the meninges).

Stenosis: Narrowing or constriction of the diameter of a bodily passage or orifice.

Tetralogy of Fallot (TOF): Congenital heart defect composed of ventricular septal defect, pulmonary stenosis or atresia, displacement of the aorta to the right, and hypertrophy of right ventricle.

Tracheoesophageal fistula (TEF): See **Esophageal atresia**.

Translocation: Chromosome rearrangement in which a piece of genetic material is transferred from one segment to another. May be balanced (no chromosome material gained or lost), or unbalanced (material has been gained or lost).

Transposition of the great vessels (arteries) (dTGA): Congenital heart defect in which the aorta arises from the right ventricle, and the pulmonary artery arises from the left ventricle (opposite of normal).

Tricuspid atresia: Congenital heart defect characterized by the absence of the tricuspid valve.

Trisomy: Chromosome abnormality characterized by a third copy of a chromosome. Includes complete and partial formation of an extra chromosome.

Trisomy 13: Chromosome abnormality caused by an extra chromosome 13.

Trisomy 18: Chromosomal abnormality caused by an extra chromosome 18.

Trisomy 21: See **Down Syndrome**.

Truncus arteriosus: Congenital heart defect characterized by a single great arterial trunk, instead of a separate aorta and pulmonary artery.

Ventricular Septal Defect (VSD): Congenital heart defect characterized by one or several openings in the ventricular septum. Includes subtypes based on location of the "hole" in the septum, ie. membranous, muscular, conoventricular, subtricuspid/canal.

[1] Adapted from the Texas Birth Defects Monitoring Division, Texas Department of Health, http://www.tdh.state.tx.us/tbdmd/glossary.htm. Modified 2/27/01, Accessed 4/2/01.

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