Massachusetts Birth Defects 2000-2001

Massachusetts Birth Defects Monitoring Program
Bureau of Family and Community Health
Center for Community Health

Massachusetts Department of Public Health
March 2005

Massachusetts Birth Defects 2000-2001

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Sally Fogerty, Director, Center for Community Health; Assistant Commissioner, Bureau of Family and Community Health
Marlene Anderka, Principal Investigator, Massachusetts Birth Defects Prevention Study
Linda Casey, Director, Massachusetts Center for Birth Defects Research and Prevention

Prepared by: Massachusetts Birth Defects Monitoring Program staff
Project Director: Cathleen Higgins, Surveillance Coordinator

Massachusetts Department of Public Health
617-624-5510

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To obtain additional copies of this report, contact:

Massachusetts Department of Public Health
Bureau of Family and Community Health
Massachusetts Center for Birth Defects Research and Prevention
Massachusetts Birth Defects Monitoring Program
250 Washington Street
Boston, MA 02108
(617) 624-5510

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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% of all infant deaths result from a birth defect. The infant mortality rate due to birth defects in the US in 2001 was 137.6 per 100,000 live births (CDC).

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 $118 million in 2001 dollars (Harris, 1997; see Technical Notes for inflation adjustment). These figures include direct costs of medical treatment, development 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 seven 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 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 by the state newborn screening program.

This report presents statewide data on the prevalence of birth defects in live births and stillbirths in Massachusetts during the years 2000 and 2001. The first annual report presented Massachusetts data for birth defects for the year 1999. Our ability to find and identify infants born with birth defects to Massachusetts' residents has improved since that time. The 2000-2001 data are presented in combined form since the numbers are relatively small for individual defects and we have yet to establish a baseline for comparison. 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 2000-2001 was 140.47 per 10,000 live births. Four out of the ten most common defects were cardiovascular defects including Patent Ductus Arteriosus, Septal (Atrial and Ventricular) Defects and Tetralogy of Fallot. Common non-cardiovascular defects included Cleft Lip, Cleft Palate, Hypospadias, and Obstructive Genitourinary Defects.

Among the 162,596 live births to Massachusetts residents in 2000-2001, 2,224 had one or more birth defects. In addition, 60 stillbirths were identified as having a birth defect. Overall, 1.40% of births in the state had one or more birth defects.

This percentage was lower than the 3% to 5% estimated by CDC, perhaps due to the fact that Massachusetts currently has limited reporting. Counts for some defects (including Anencephaly, Trisomy 13, and Trisomy 18) were less than 60% of the expected counts based on 2000-2001 rates from the Metropolitan Atlanta Congenital Defects Program, which included cases among live births, stillbirths and elective terminations. These conditions are not reported in Massachusetts when they are prenatally diagnosed and the pregnancy is electively terminated. Stillbirth reports (limited to reports of fetal death older than 20 weeks or greater than 350 grams) from birthing hospitals may not indicate if there is a birth defect present. Other contributors to lower counts may be differences in defect criteria between the two surveillance systems and the fact that Massachusetts in 2000-2001 was in the early years of its surveillance system, when ascertainment and reporting were still improving.

Single vs. Multiple Defects

Of all 2,284 birth defect cases (infants and stillborns), 62.5% had a single defect and 37.5% 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, Gastroschisis, Hirschsprung Disease, Clubfoot, and Hypospadias appeared more often as a single defect rather than with other defects. The majority of Cardiovascular Defects, Limb Reductions, Hydrocephalus, Intestinal Atresias, and Obstructive Genitourinary Defects appeared more often in conjunction with other defects.

Plurality

Examining the birth defect rate by plurality is important since the number of multiple births has been increasing over time in Massachusetts. The birth defect prevalence rate was 135.19 for singletons and 255.91 for multiple births (more than one infant) per 10,000 live births. Birth defects that more commonly occurred in multiple births included Pulmonary Stenosis (Valvular), Rectal and Large Intestine Atresia, Esophageal Atresia/Tracheoesophageal Fistula, Septal Defects (Atrial and Ventricular), Hypospadias, and Lower Limb Reduction Defects.

Sex

The birth defect prevalence rate was 119.83 for females and 160.32 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. Common defects seen in both sexes included Septal Defects, Down Syndrome, Cleft Palate, Cleft Lip and Obstructive Genitourinary Defects. The most common defects seen in males were Atrial Septal Defects (Secundum and NOS), Hypospadias, Clubfoot, Down Syndrome and Obstructive Genitourinary Defects. The most common defects seen in females were Atrial Septal Defects (Secundum and NOS), Down Syndrome, Ventricular Septal Defects (Membranous and NOS), Polydactyly/Syndactyly and Cleft Palate without Cleft Lip.

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 152.12 for mothers younger than 20 years, 135.87 for those 20-24 years, 130.24 for those 25-29 years, 126.53 for those 30-34 years, and 155.57 for those 35 years and older. As expected, there was a strong association of Down Syndrome with advanced maternal age. Women 35 years and older had a live birth Down Syndrome rate of 26.73 per 10,000 births. This rate was three times that of any other maternal age group. Younger mothers (age 19 and under) had the highest rate (16.26%) of Gastroschisis. This association has 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 137.37 for Non-Hispanic Whites, 151.50 for Non-Hispanic Blacks, 139.36 for Hispanics, and 101.58 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, Down Syndrome, Polydactyly/Syndactyly, and Clubfoot. In Blacks, the most common defects included Septal Defects, Polydactyly/Syndactyly, Down Syndrome, and Hypospadias. The most common defects in Whites included Septal Defects, Down Syndrome, Clubfoot and Hypospadias. In Asians, the most common defects included Septal Defects, Cleft Lip, Cleft Palate, and Polydactyly/Syndactyly.

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. 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	2.6%
Serious, may be correctable, most have long-term needs	18.7%
Moderate, most correctable, many have long-term needs	71.1%
Mild, may be correctable, minimal long-term needs	7.6%

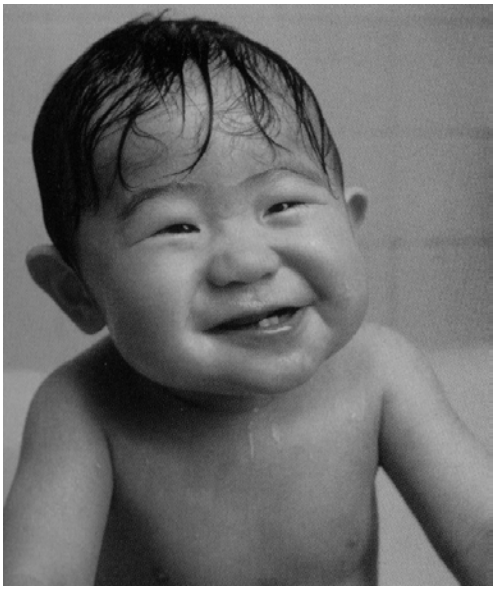
Nearly three percent of cases with birth defects were classified as “severe” and most did not survive. This percentage was an underestimate of cases due to limited data. For example, Cragan at CDC estimated that up to 80% of Anencephaly cases and 50% of any neural tube defect may be electively terminated after prenatal diagnosis (Cragan 2000). About 19% 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 71% 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, in the US, they are the leading cause of death in the first year of life. About 20% of all infant deaths result from a birth defect. The infant mortality rate due to birth defects in the United States in 2001 was 137.6 per 100,000 live births (CDC).

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 seven 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 Hospital Reporting Form (HRF) to capture essential data for each birth defect

case. The HRFs 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 2001 dollars.

Adjusting for inflation, the Massachusetts combined lifetime costs for babies born with 12 major structural birth defects were an estimated $118 million in 2001 dollars (see Technical Notes). These figures included direct costs of medical treatment, development 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.

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 recent 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 2000-2001 Surveillance Report

This report presents statewide data on the prevalence of birth defects in live births and stillbirths in Massachusetts during the years 2000 and 2001. The first annual report presented Massachusetts data for birth defects for the year 1999. Our ability to find and identify infants born with birth defects to Massachusetts' residents has improved since that time. There is about a 25% increase in cases from 1999-2001 that is attributable to this improved case ascertainment. This report presents two years of data, 2000 and 2001. The data is presented in combined form since the numbers are relatively small for individual defects and we have yet to establish a baseline for comparison.

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.

Milestones/History of Birth Defect Monitoring in Massachusetts

- **Early 1960s…..A national epidemic of limb reduction defects associated with women's prenatal use of thalidomide drew attention to birth defects.**

- **1963……Massachusetts passed legislation mandating hospitals to report birth defects to MDPH.**

- **1970……A trend developed to transport acutely ill infants to non-birthing tertiary care facilities that did not report to MDPH. Birth defects were underreported.**

- **1984……The High Risk Infant Identification System (HRIIS) was established. Obstetric nurses in birth hospitals captured data on birth defects. The program was phased out in the early 1990s.**

- **1990s…….Birth defects data were collected from administrative review of birth, death, and fetal death certificates from the Registry of Vital Records and Statistics and Uniform Hospital Discharge Data. Birth defects were underreported because of the passive nature of surveillance during this period.**

- **1995……The CDC sponsored a pilot study to analyze the administrative review data. This showed that birth defect reporting on birth certificates was underreported while it was reported more completely and accurately in hospital discharge data.**

- **1996……The CDC awarded funding to MDPH to establish the Center for Birth Defects Research and Prevention in collaboration with Slone Epidemiology Center at Boston University and the Genetics and Teratology Department at Massachusetts General Hospital.**

- **1997…….An active surveillance program was initiated. Trained personnel validated reports of birth defects, actively seeking cases in hospitals, nurseries, and neonatal intensive care units. Data were collected in the eastern part of the state.**

- **1998…….By the end of this year, the Birth Defects Monitoring Program (BDMP) had begun collecting cases from the all birthing hospitals in the state and one major tertiary care hospital.**

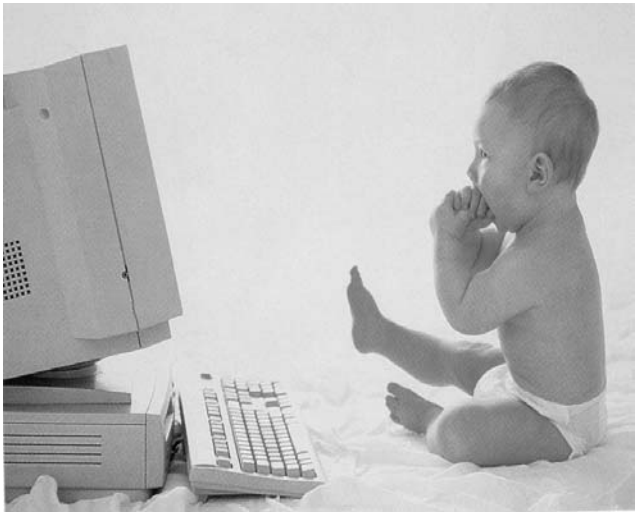
- **2001…….First BDMP Surveillance Report produced containing statewide data from the population-based, active BDMP.**

- **2002…….The Massachusetts legislature expanded case ascertainment up to 3 years of age and expanded reporting sources to include physicians, outpatient clinics and genetic services.**

- **Presently…….The BDMP collects data from 53 birthing hospitals, 1 tertiary care and 1 specialty hospital in Massachusetts, and 1 Rhode Island birth hospital and 1 Rhode Island tertiary care hospital near the MA/RI border. Data sources used to ascertain and verify cases include: hospital discharge reports, hospital nurseries and neonatal units, birth certificates, fetal and infant death certificates, hospital surgical and pathology departments.**

Chapter 2

Methods



Case Definition

This report presents data on selected birth defects present in births occurring during the calendar years 2000 and 2001 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 in Appendices).
- The diagnosis was made before the infant reached one year of age.

Data Collection

The Massachusetts Birth Defects Monitoring Program 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 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 a prevalence. Prevalence is calculated as the number of birth defect cases born at a point in time per 10,000 live births. 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 2000 and 2001. 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 80% 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. While reporting of such postnatal results can be made to the MDPH as a result of an amendment to the birth defects reporting law in 2002, lack of resources prevents the Monitoring Program from obtaining all of these results.

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 fetal demise prior to 20 weeks gestation (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. The inclusion of birth defect data from the RI hospitals resulted in 38 cases that were newly diagnosed, confirmed or received a different diagnosis.

Deliveries and diagnoses that occurred in other out of state facilities are not included at this time.

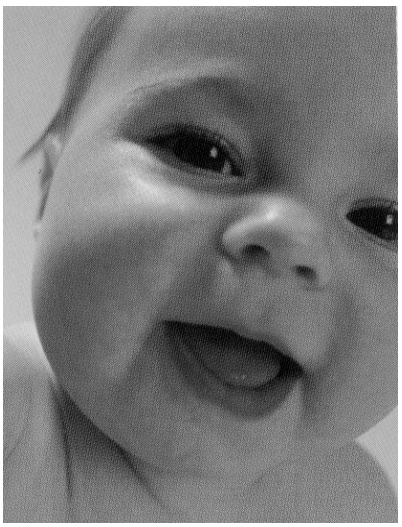
8. There are limitations in comparing data from the Metropolitan Atlanta Congenital Defects Program, which is considered the "gold standard" of birth defect surveillance, and the Massachusetts Birth Defects Monitoring Program. Factors such as differences in the demographics of the two populations, the 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 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 162,596 live births to Massachusetts residents in 2000-2001, 2,224 had one or more structural birth defects that were ascertained by MBDMP. In addition, 60 stillbirths were identified with a birth defect. Overall, 1.40% of births in the state were identified as having one or more birth defects. This was lower than the 3% to 5% estimated by the Metropolitan Atlanta Congenital Defects Program (MACDP). This difference may have been due to differences in criteria and/or scope between the MACDP and Massachusetts surveillance systems. Nevertheless, comparing expected birth defect counts from MACDP to the observed counts in Massachusetts helps to evaluate how our surveillance is doing in capturing cases. The MACDP collects defects in live births, stillbirths and elective terminations, using active surveillance methods. It is considered the "gold standard" of birth defects surveillance systems.

Table 2 shows the comparison of 2000-2001 observed counts for Massachusetts to expected counts generated by 2000-2001 rates from MACDP. Expected counts were calculated from Atlanta rates which included live births, stillbirths, and elective terminations. Each birth defect specific rate from Atlanta was multiplied by the number of total 2000-2001 Massachusetts births (162,596) to generate the expected numbers for that birth defect. A ratio of observed counts over expected counts (O/E) less than 1.0 indicates observed counts were less than expected counts. A ratio more than 1.0 indicates observed counts that were greater than expected counts. Overall, the O/E ratio ranges between 0.2 to 1.3. If the confidence interval of the O/E ratio does not include 1.00, the observed counts from Massachusetts show a significant elevation or deficit compared to the Atlanta expected counts.

Massachusetts counts fell below 60% of the expected counts for Anencephaly, Trisomy 13 and Trisomy 18. In general, these cases were prenatally diagnosed and therefore may not have been ascertained at a birthing hospital.
Brigham and Women's Hospital, where 12% of resident births occurred in 2000-2001, researched trends in elective termination in Massachusetts. For years 1994 and 1999, 81% of pregnancies with defects that the MA Birth Defects Monitoring Program considers severe and incompatible with life (which includes anencephaly, bilateral renal agenesis and Trisomy 18), were electively terminated. For the same years, 40% of pregnancies with defects we consider serious and with long-term needs, were terminated. Spina Bifida and Down Syndrome were included in this category (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). Spontaneous deliveries of stillbirths were reported by birthing hospitals but limited information about the stillbirth is included in the maternal record. Other contributors to lower counts may have been differences in populations, defect criteria between the two surveillance systems, and the fact that Massachusetts in 2000-2001 was in the early years of its surveillance system, when ascertainment and reporting were still improving.

The overall prevalence of reported birth defects in Massachusetts in 2000-2001 was 140.47 per 10,000 live births. This represents a 25% increase from 1999, when the prevalence rate was 111.79. This increase was due to better reporting from hospitals and improved ascertainment of cases. The majority of defects fell into Cardiovascular (34.2%) and Musculoskeletal (25.7%) categories. Figure 1 shows the percentage of reported birth defects by defect categories.

Table 3 shows the most common birth defects in the state. Four out of the ten most common defects were Cardiovascular Defects including Patent Ductus Arteriosus, Septal (Atrial and Ventricular) Defects, and Tetralogy of Fallot with and without Pulmonary Atresia. Common Non-Cardiovascular Defects included Cleft Lip, Cleft Palate, Down Syndrome, and Obstructive Genitourinary Defects. Cardiovascular Defects were the most commonly occurring birth defects in 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, 62.5% had single defects and 37.5% had multiple defects. Figure 2 shows counts for selected birth defects by single and multiple defect categories.

Anencephaly, Cleft Lip, Gastroschisis, Hirschsprung Disease, Clubfoot, and Hypospadias appeared more often as a single defect rather than in combination with other defects. Limb Reductions, Hydrocephalus, Intestinal Atresias, and Obstructive Genitourinary Defects 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.

Table 1 Prevalence of Birth Defects, Massachusetts: 2000-2001

Defect[1]	Livebirth Count	Stillbirth Count	Total Count	Rate per 10,000 Births	95% Confidence Interval
Central Nervous System: 219 cases					
Anencephaly	7	0	7	0.43	0.17—0.89
Encephalocele	7	0	7	0.43	0.17—0.89
Holoprosencephaly	9	0	9	0.55	0.25—1.05
Hydrocephaly w/o Spina Bifida	45	2	47	2.89	2.12—3.84
Microcephaly	42	0	42	2.58	1.86—3.49
Spina Bifida w/ and w/o Hydrocephaly	29	0	29	1.78	1.19—2.56
Tethered Cord	28	0	28	1.72	1.14—2.49
Other CNS	87	1	88		
Eye: 62 cases					
Aniridia	8	0	8	0.49	0.21—0.97
Anophthalmia/Microphthalmia	17	0	17	1.05	0.61—1.67
Congenital Glaucoma, Congenital Cataract	30	0	30	1.85	1.24—2.63
Other Eye	21	0	21		
Ear: 23 cases					
Anotia/Microtia	20	0	20	1.23	0.75—1.9
Other Ear	6	0	6		

Table 1 Prevalence of Birth Defects, Massachusetts: 2000-2001 (cont'd)

Defect[1]	Livebirth Count	Stillbirth Count	Total Count	Rate per 10,000 Births	95% Confidence Interval
Cardiovascular: 782 cases					
Anomalous Pulmonary Venous Connection					
Total/Partial Anomalous Pulmonary Venous Connection	24	0	24	1.48	0.95—2.2
Atrioventricular Canal Defects					
ASD Primum	16	0	16	0.98	0.56—1.6
Common Atrium	3	1	4	0.25	0.07—0.63
Complete Atrioventricular Canal Defect	43	2	45	2.77	2.02—3.7
Endocardial Cushion (OS and NOS)	9	0	9	0.55	0.25—1.05
VSD, Canal Type	11	0	11	0.68	0.34—1.21
Conotruncal (Outlet) and Aortic Arch					
Double Outlet Right Ventricle	19	0	19	1.17	0.7—1.82
d-Transposition of the Great Arteries	24	1	25	1.54	1—2.27
Interrupted Aortic Arch, Type B	4	0	4	0.25	0.07—0.63
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	69	3	72	4.43	3.46—5.58
Truncus	6	0	6	0.37	0.14—0.8
Ebstein Anomaly					
Ebstein Anomaly	4	2	6	0.37	0.14—0.8
Heterotaxy (Laterality Defects)					
Heterotaxy	16	1	17	1.05	0.61—1.67
Left-Sided Obstruction					
Aortic Valve Stenosis	16	1	17	1.05	0.61—1.67
Coarctation of Aorta	65	2	67	4.12	3.19—5.23
Hypoplastic Left Heart Syndrome	23	0	23	1.41	0.9—2.12
Interrupted Aortic Arch (Type A and NOS)	3	0	3	0.18	0.04—0.54
Patent Ductus Arteriosus					
Patent Ductus Arteriosus	265	1	266	16.36	14.45—18.45
Right-Sided Obstruction					
Pulmonary Stenosis, Valvular	67	0	67	4.12	3.19—5.23
Pulmonary Valve Atresia w/intact septum	11	1	12	0.74	0.38—1.29
Pulmonary Valve Atresia with VSD	5	0	5	0.31	0.1—0.72

Table 1 Prevalence of Birth Defects, Massachusetts: 2000-2001 (cont'd)

Defect[1]	Livebirth Count	Stillbirth Count	Total Count	Rate per 10,000 Births	95% Confidence Interval
Tricuspid Valve Atresia	9	2	11	0.68	0.34—1.21
Septal Defects					
ASD (Secundum and NOS)	292	6	298	18.33	16.31—20.53
VSD (Membranous and NOS)	152	3	155	9.53	8.09—11.16
VSD, Conoventricular/Malalignment	28	0	28	1.72	1.14—2.49
Single Ventricle and L-TGA					
L-TGA	6	0	6	0.37	0.14—0.8
Single Ventricle	7	0	7	0.43	0.17—0.89
Other Cardiovascular					
Other Cardiovascular	198	7	205		

Respiratory: 45 cases

Choanal Atresia	8	0	8	0.49	0.21—0.97
Lung Anomalies	29	0	29	1.78	1.19—2.56
Other Respiratory	9	0	9		

Orofacial: 244 cases

Cleft Lip w/ and w/o Cleft Palate	124	3	127	7.81	6.51—9.29
Cleft Palate w/o Cleft Lip	96	1	97	5.97	4.84—7.28
Pierre Robin Sequence	22	0	22	1.35	0.85—2.05
Other Orofacial	22	0	22		

Gastrointestinal: 225 cases

Biliary Atresia	5	0	5	0.31	0.1—0.72
Esophageal Atresia/Tracheoesophageal Fistula	35	1	36	2.21	1.55—3.07
Hirschsprung Disease	28	0	28	1.72	1.14—2.49
Rectal and Large Intestinal Atresia/Stenosis	62	1	63	3.87	2.98—4.96
Small Intestinal Atresia	32	0	32	1.97	1.35—2.78
Other Gastrointestinal	82	0	82		

Table 1 Prevalence of Birth Defects, Massachusetts: 2000-2001 (cont'd)

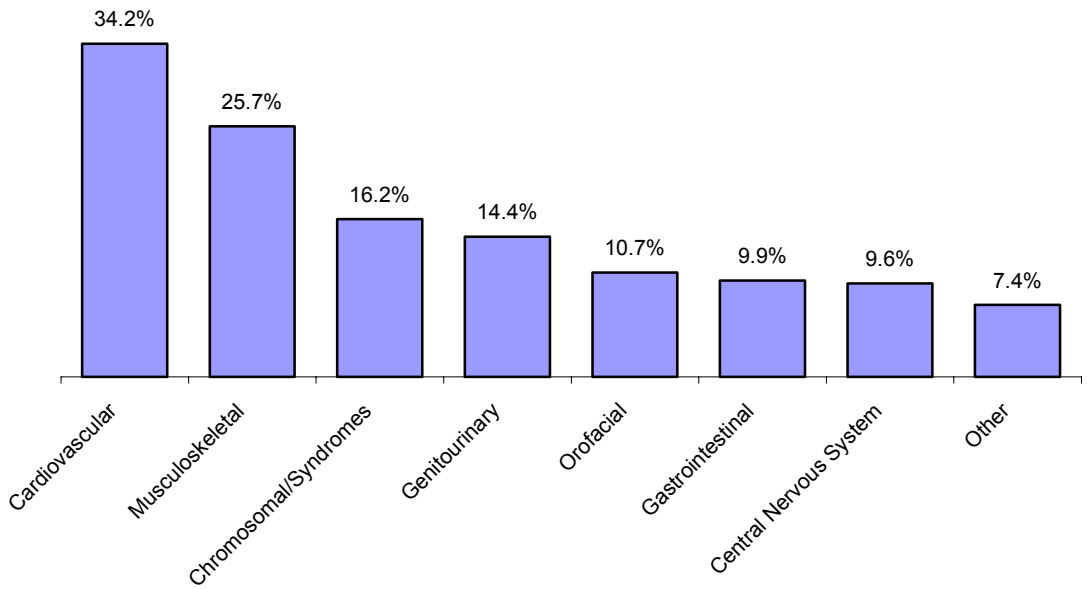
Defect[1]	Livebirth Count	Stillbirth Count	Total Count	Rate per 10,000 Births	95% Confidence Interval
Genitourinary: 329 cases					
Bladder Exstrophy	6	0	6	0.37	0.14—0.8
Cloacal Exstrophy	1	0	1	0.06	0—0.34
Hypospadias, 2nd or 3rd Degree	139	0	139	8.55	7.19—10.09
Obstructive Genitourinary Defect	125	0	125	7.69	6.4—9.16
Renal Agenesis/Hypoplasia	2	2	4	0.25	0.07—0.63
Other Genitourinary	111	2	113		
Musculoskeletal: 586 cases					
Club Foot	155	4	159	9.78	8.32—11.42
Craniosynostosis	72	0	72	4.43	3.46—5.58
Diaphragmatic Hernia	29	2	31	1.91	1.3—2.71
Gastroschisis	32	2	34	2.09	1.45—2.92
Omphalocele	21	3	24	1.48	0.95—2.2
Polydactyly/Syndactyly	143	2	145	8.92	7.53—10.49
Reduction Deformity, Lower Limbs	22	1	23	1.41	0.9—2.12
Reduction Deformity, Upper Limbs	45	1	46	2.83	2.07—3.77
Skeletal Dysplasia	16	0	16	0.98	0.56—1.6
Other Musculoskeletal	107	2	109		
Chromosomal and Other Syndromes: 369 cases					
Klinefelter Syndrome	7	0	7	0.43	0.17—0.89
Trisomy 13	8	3	11	0.68	0.34—1.21
Trisomy 18	10	6	16	0.98	0.56—1.6
Trisomy 21 (Down Syndrome)	182	11	193	11.87	10.25—13.67
Turner Syndrome	12	3	15	0.92	0.52—1.52
Other Chromosomal Syndromes/Other Syndromes	123	5	128		

Table 1 Prevalence of Birth Defects, Massachusetts: 2000-2001 (cont'd)

Defect[1]	Livebirth Count	Stillbirth Count	Total Count	Rate per 10,000 Births	95% Confidence Interval
Other: 40 cases					
Amniotic Bands	4	1	5	0.31	0.1—0.72
Skin Anomalies	11	2	13	0.8	0.43—1.37
Other, Specified	20	2	22		

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.

Figure 1 Percent of Cases by Birth Defect Category, Massachusetts: 2000-2001



Many cases have multiple defects, so percentages add up to more than 100%.
Total Number of Cases = 2,284

Table 2 Comparison of Selected Massachusetts 2000-2001 Observed Counts and Expected Counts Based on Atlanta 2000-2001 Rates

Defect	Atlanta Rate[1]	MA Observed Count[2]	MA Expected Count[3]	O/E Ratio	95% Confidence Interval[4]
Central Nervous System					
Anencephaly	2.08	7	34	0.21	0.12 — 0.30
Hydrocephaly w/o Spina Bifida	8.14	47	132	0.36	0.31 — 0.40
Microcephaly	6.55	42	106	0.40	0.35 — 0.44
Spina Bifida w/ and w/o Hydrocephaly	2.58	29	42	0.69	0.64 — 0.73
Eye					
Anophthalmia/Microphthalmia	2.98	17	48	0.35	0.28 — 0.43
Cardiovascular					
Conotruncal (Outlet) and Aortic Arch					
d-Transposition of the Great Arteries	6.45	25	105	0.24	0.19 — 0.29
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	3.47	72	56	1.29	1.33 — 1.25
Truncus	0.69	6	11	0.55	0.40 — 0.66
Ebstein Anomaly					
Ebstein Anomaly	0.40	6	6	1.00	1.00 — 1.00
Left-Sided Obstruction					
Aortic Valve Stenosis	1.89	17	31	0.55	0.47 — 0.62
Coarctation of Aorta	5.76	67	94	0.71	0.68 — 0.74
Hypoplastic Left Heart Syndrome	2.68	23	44	0.52	0.46 — 0.58
Right-Sided Obstruction					
Tricuspid Valve Atresia	1.49	11	24	0.46	0.36 — 0.55
Septal Defects					
ASD (Secundum and NOS)	24.81	298	403	0.74	0.73 — 0.75
Respiratory					
Choanal Atresia	1.29	8	21	0.38	0.27 — 0.49

Table 2 (cont'd) Comparison of Selected Massachusetts 2000-2001 Observed Counts and Expected Counts Based on Atlanta 2000-2001 Rates

Defect	Atlanta Rate[1]	MA Observed Count[2]	MA Expected Count[3]	O/E Ratio	95% Confidence Interval[4]
Orofacial					
Cleft Lip w/ and w/o Cleft Palate	7.84	127	127	1.00	1.00 — 1.00
Cleft Palate w/o Cleft Lip	8.14	97	132	0.73	0.71 — 0.76
Gastrointestinal					
Esophageal Atresia/Tracheoesophageal Fistula	2.98	36	48	0.75	0.71 — 0.78
Hirschsprung Disease	2.48	28	40	0.70	0.65 — 0.74
Rectal and Large Intestinal Atresia/Stenosis	3.97	63	65	0.97	0.97 — 0.97
Musculoskeletal					
Diaphragmatic Hernia	2.48	31	40	0.78	0.74 — 0.81
Gastroschisis	3.18	34	52	0.65	0.61 — 0.70
Omphalocele	2.58	24	42	0.57	0.51 — 0.63
Reduction Deformity, Lower Limbs	1.98	23	32	0.72	0.67 — 0.76
Reduction Deformity, Upper Limbs	4.76	46	77	0.60	0.55 — 0.64
Chromosomal and Other Syndromes					
Trisomy 13	1.19	11	19	0.58	0.48 — 0.66
Trisomy 18	1.89	16	31	0.52	0.43 — 0.59
Trisomy 21 (Down Syndrome)	13.79	193	224	0.86	0.85 — 0.87

1. Atlanta 2000-2001 rates were provided by the Metropolitan Atlanta Congenital Defects Program, and were reported per 10,000 live births.

2. Observed counts are among resident live births and stillbirths for Massachusetts, 2000-2001

3. Expected counts were calculated from Atlanta rates which include live births, stillbirths, and elective terminations. Each Atlanta rate was multiplied by the 2000-2001 Massachusetts births total (162,596) to generate expected counts.

4. If the 95% CI range does not include 1.00, the observed counts are significantly elevated or deficient from the expected counts.

Table 3 Selected Common Defects, among Live Births and Stillbirths, Massachusetts: 2000-2001

Defect[1]	Category	Count	Rate per 10,000 Births	95% Confidence Interval
ASD (Secundum and NOS)	Cardiovascular	298	18.33	16.31—20.53
Trisomy 21 (Down Syndrome)	Chromosomal and Other Syndromes	193	11.87	10.25—13.67
Club Foot	Musculoskeletal	159	9.78	8.32—11.42
VSD (Membranous and NOS)	Cardiovascular	155	9.53	8.09—11.16
Polydactyly/Syndactyly	Musculoskeletal	145	8.92	7.53—10.49
Hypospadias, 2nd or 3rd Degree	Genitourinary	139	8.55	7.19—10.09
Cleft Lip w/ and w/o Cleft Palate	Orofacial	127	7.81	6.51—9.29
Obstructive Genitourinary Defect	Genitourinary	125	7.69	6.40—9.16
Cleft Palate w/o Cleft Lip	Orofacial	97	5.97	4.84—7.28
Craniosynostosis	Musculoskeletal	72	4.43	3.46—5.58
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	Cardiovascular	72	4.43	3.46—5.58

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: 2000-2001

Defect[1]	Cases with one defect	Cases with two or more defects	Total Cases
Central Nervous System: 219 cases			
Anencephaly	7	0	7
Encephalocele	3	4	7
Holoprosencephaly	1	8	9
Hydrocephaly w/o Spina Bifida	16	31	47
Microcephaly	18	24	42
Spina Bifida w/ and w/o Hydrocephaly	14	15	29
Tethered Cord	10	18	28
Other CNS	26	62	88
Eye: 62 cases			
Aniridia	4	4	8
Anophthalmia/Microphthalmia	2	15	17
Congenital Glaucoma, Congenital Cataract	19	11	30
Other Eye	2	19	21
Ear: 23 cases			
Anotia/Microtia	9	11	20
Other Ear	1	5	6

Table 4 (cont'd) Single vs. Multiple Defects among Live Births and Stillbirths, Massachusetts: 2000-2001

Defect[1]	Cases with one defect	Cases with two or more defects	Total Cases
Cardiovascular: 782 cases			
Anomalous Pulmonary Venous Connection			
Total/Partial Anomalous Pulmonary Venous Connection	2	22	24
Atrioventricular Canal Defects			
ASD Primum	0	16	16
Common Atrium	0	4	4
Complete Atrioventricular Canal Defect	2	43	45
Endocardial Cushion (OS and NOS)	0	9	9
VSD, Canal Type	0	11	11
Conotruncal (Outlet) and Aortic Arch			
Double Outlet Right Ventricle	1	18	19
d-Transposition of the Great Arteries	1	24	25
Interrupted Aortic Arch, Type B	0	4	4
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	24	48	72
Truncus	0	6	6
Ebstein Anomaly			
Ebstein Anomaly	3	3	6
Heterotaxy (Laterality Defects)			
Heterotaxy	1	16	17
Left-Sided Obstruction			
Aortic Valve Stenosis	1	16	17
Coarctation of Aorta	7	60	67
Hypoplastic Left Heart Syndrome	1	22	23
Interrupted Aortic Arch (Type A and NOS)	0	3	3
Patent Ductus Arteriosus			
Patent Ductus Arteriosus	16	250	266
Right-Sided Obstruction			
Pulmonary Stenosis, Valvular	23	44	67
Pulmonary Valve Atresia w/intact septum	1	11	12
Pulmonary Valve Atresia with VSD	1	4	5

Table 4 (cont'd) Single vs. Multiple Defects among Live Births and Stillbirths, Massachusetts: 2000-2001

Defect[1]	Cases with one defect	Cases with two or more defects	Total Cases
Tricuspid Valve Atresia	1	10	11
Septal Defects			
ASD (Secundum and NOS)	69	229	298
VSD (Membranous and NOS)	56	99	155
VSD, Conoventricular/Malalignment	3	25	28
Single Ventricle and L-TGA			
L-TGA	0	6	6
Single Ventricle	0	7	7
Other Cardiovascular			
Other Cardiovascular	22	183	205
Respiratory: 45 cases			
Choanal Atresia	5	3	8
Lung Anomalies	12	17	29
Other Respiratory	2	7	9
Orofacial: 244 cases			
Cleft Lip w/ and w/o Cleft Palate	103	24	127
Cleft Palate w/o Cleft Lip	55	42	97
Pierre Robin Sequence	0	22	22
Other Orofacial	16	6	22

Table 4 (cont'd) Single vs. Multiple Defects among Live Births and Stillbirths, Massachusetts: 2000-2001

Defect[1]	Cases with one defect	Cases with two or more defects	Total Cases
Gastrointestinal: 225 cases			
Biliary Atresia	5	0	5
Esophageal Atresia/Tracheoesophageal Fistula	9	27	36
Hirschsprung Disease	19	9	28
Rectal and Large Intestinal Atresia/Stenosis	25	38	63
Small Intestinal Atresia	13	19	32
Other Gastrointestinal	36	46	82
Genitourinary: 329 cases			
Bladder Exstrophy	4	2	6
Cloacal Exstrophy	0	1	1
Hypospadias, 2nd or 3rd Degree	115	24	139
Obstructive Genitourinary Defect	16	109	125
Renal Agenesis/Hypoplasia	1	3	4
Other Genitourinary	18	95	113

Table 4 (cont'd) Single vs. Multiple Defects among Live Births and Stillbirths, Massachusetts: 2000-2001

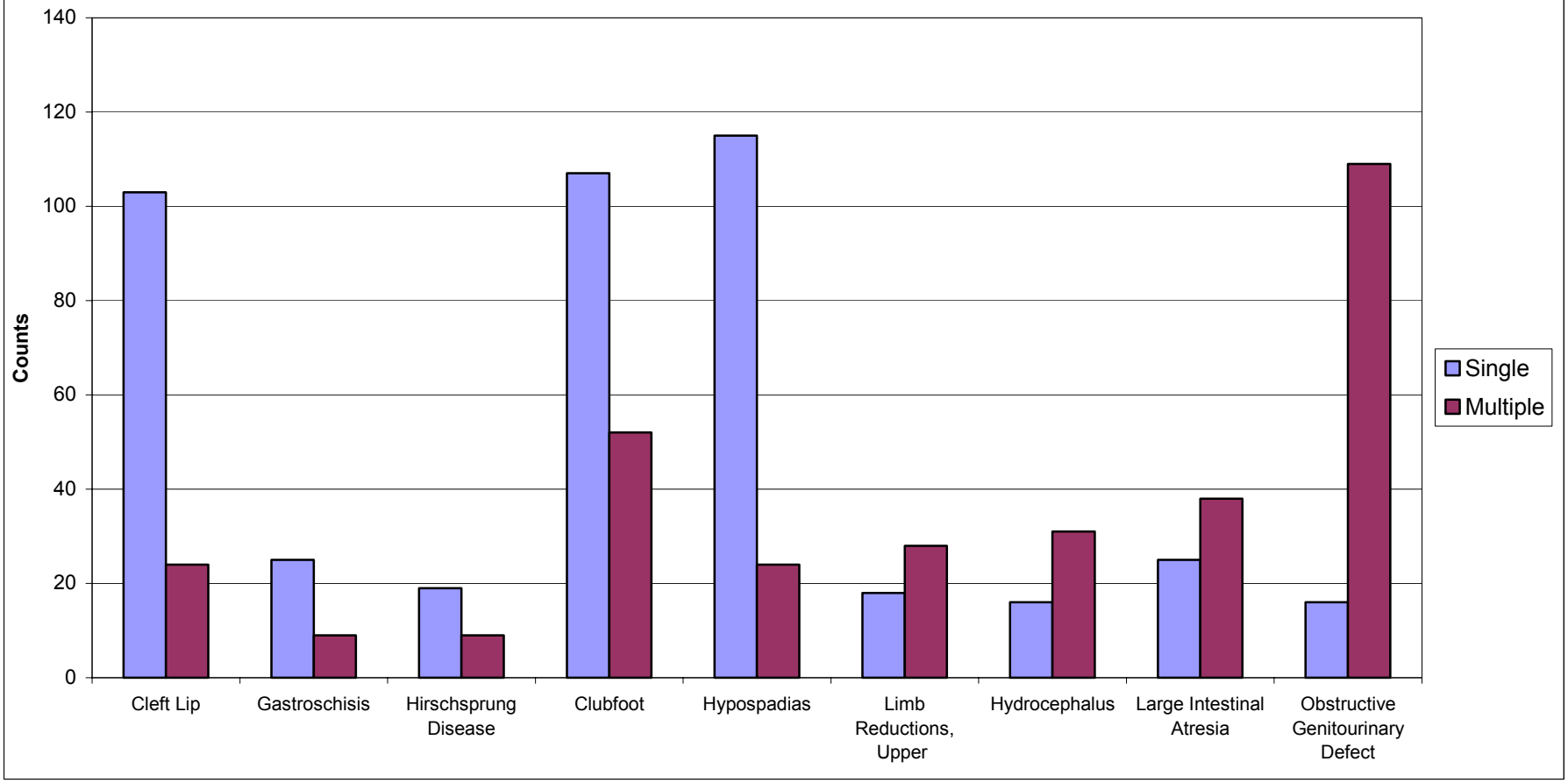
Defect[1]	Cases with one defect	Cases with two or more defects	Total Cases
Musculoskeletal: 586 cases			
Club Foot	107	52	159
Craniosynostosis	56	16	72
Diaphragmatic Hernia	15	16	31
Gastroschisis	25	9	34
Omphalocele	9	15	24
Polydactyly/Syndactyly	83	62	145
Reduction Deformity, Lower Limbs	10	13	23
Reduction Deformity, Upper Limbs	18	28	46
Skeletal Dysplasia	12	4	16
Other Musculoskeletal	10	99	109
Chromosomal and Other Syndromes: 369 cases			
Klinefelter Syndrome	6	1	7
Trisomy 13	1	10	11
Trisomy 18	3	13	16
Trisomy 21 (Down Syndrome)	87	106	193
Turner Syndrome	6	9	15
Other Chromosomal Syndromes/Other Syndromes	38	90	128
Other: 40 cases			
Amniotic Bands	0	5	5
Skin Anomalies	11	2	13
Other, Specified	5	17	22

Table 4 (cont'd) Single vs. Multiple Defects among Live Births and Stillbirths, Massachusetts: 2000-2001

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

Figure 2 Selected Birth Defects by Single and Multiple Defect Categories, Massachusetts: 2000-2001



Chapter 4

Prevalence of Birth Defects by Plurality and Sex

Plurality

Table 5 depicts the distribution of birth defects by plurality. The overall prevalence was 135.19 for singletons and 255.91 for multiple births (more than one infant) per 10,000 live births. While multiple births comprised 4.4% of all births, they comprised 8.0% of all birth defects cases (see Figure 3). Birth defects that more commonly occurred in multiple births included Pulmonary Stenosis (Valvular), Rectal and Large Intestine Atresia, Esophageal Atresia/Tracheoesophageal Fistula, Septal Defects (Atrial and Ventricular), Hypospadias, and Lower Limb Reduction Defects. Figure 4 depicts 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 119.83 for females and 160.32 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. Common defects seen in both sexes included Septal Defects, Down Syndrome, Cleft Palate, Cleft Lip, and Obstructive Genitourinary Defects. Figure 5 compares the prevalence of selected birth defects among males and females. The most common defects seen in males were Atrial Septal Defects (Secundum and NOS), Hypospadias, Clubfoot, Down Syndrome and Obstructive Genitourinary Defects. The most common defects seen in females were Atrial Septal Defects (Secundum and NOS), Down Syndrome, Ventricular Septal Defects (Membranous and NOS), Polydactyly/Syndactyly and Cleft Palate without Cleft Lip.

Table 5 Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2000-2001

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
Central Nervous System: 219 cases				
Anencephaly	singleton	6	0.39	0.14 — 0.84
	multiple	1	1.41	0.04 — 7.83
Encephalocele	singleton	7	0.45	0.18 — 0.93
	multiple	0	0.00	0.00 — 5.19
Holoprosencephaly	singleton	8	0.51	0.22 — 1.01
	multiple	1	1.41	0.04 — 7.83
Hydrocephaly w/o Spina Bifida	singleton	42	2.70	1.95 — 3.65
	multiple	5	7.03	2.28 — 16.41
Microcephaly	singleton	42	2.70	1.95 — 3.65
	multiple	0	0.00	0.00 — 5.19
Spina Bifida w/ and w/o Hydrocephaly	singleton	29	1.87	1.25 — 2.68
	multiple	0	0.00	0.00 — 5.19
Tethered Cord	singleton	26	1.67	1.09 — 2.45
	multiple	2	2.81	0.34 — 10.16
Other CNS	singleton	82	5.27	4.19 — 6.55
	multiple	6	8.44	3.10 — 18.36
Eye: 62 cases				
Aniridia	singleton	7	0.45	0.18 — 0.93
	multiple	1	1.41	0.04 — 7.83
Anophthalmia/Microphthalmia	singleton	16	1.03	0.59 — 1.67
	multiple	1	1.41	0.04 — 7.83
Congenital Glaucoma, Congenital Cataract	singleton	29	1.87	1.25 — 2.68
	multiple	1	1.41	0.04 — 7.83
Other Eye	singleton	21	1.35	0.84 — 2.06
	multiple	0	0.00	0.00 — 5.19
Ear: 23 cases				
Anotia/Microtia	singleton	17	1.09	0.64 — 1.75
	multiple	3	4.22	0.87 — 12.33
Other Ear	singleton	5	0.32	0.10 — 0.75

Table 5 (cont'd) Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2000-2001

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
Other Ear	multiple	1	1.41	0.04 — 7.83
Cardiovascular: 782 cases				
Anomalous Pulmonary Venous Connection				
Total/Partial Anomalous Pulmonary Venous Connect	singleton	22	1.41	0.89 — 2.14
	multiple	2	2.81	0.34 — 10.16
Atrioventricular Canal Defects				
ASD Primum	singleton	14	0.90	0.49 — 1.51
	multiple	2	2.81	0.34 — 10.16
Common Atrium	singleton	4	0.26	0.07 — 0.66
	multiple	0	0.00	0.00 — 5.19
Complete Atrioventricular Canal Defect	singleton	42	2.70	1.95 — 3.65
	multiple	3	4.22	0.87 — 12.33
Endocardial Cushion (OS and NOS)	singleton	7	0.45	0.18 — 0.93
	multiple	2	2.81	0.34 — 10.16
VSD, Canal Type	singleton	10	0.64	0.31 — 1.18
	multiple	1	1.41	0.04 — 7.83
Conotruncal (Outlet) and Aortic Arch				
Double Outlet Right Ventricle	singleton	19	1.22	0.74 — 1.91
	multiple	0	0.00	0.00 — 5.19
d-Transposition of the Great Arteries	singleton	25	1.61	1.04 — 2.37
	multiple	0	0.00	0.00 — 5.19
Interrupted Aortic Arch, Type B	singleton	4	0.26	0.07 — 0.66
	multiple	0	0.00	0.00 — 5.19
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	singleton	66	4.24	3.28 — 5.40
	multiple	6	8.44	3.10 — 18.36
Truncus	singleton	5	0.32	0.10 — 0.75
	multiple	1	1.41	0.04 — 7.83
Ebstein Anomaly				
Ebstein Anomaly	singleton	6	0.39	0.14 — 0.84
	multiple	0	0.00	0.00 — 5.19
Heterotaxy (Laterality Defects)				
Heterotaxy	singleton	18	1.16	0.69 — 1.83
	multiple	0	0.00	0.00 — 5.19
Left-Sided Obstruction				
Aortic Valve Stenosis	singleton	15	0.96	0.54 — 1.59
	multiple	2	2.81	0.34 — 10.16

Table 5 (cont'd) Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2000-2001

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
Coarctation of Aorta	singleton	63	4.05	3.11 — 5.18
	multiple	4	5.62	1.53 — 14.40
Hypoplastic Left Heart Syndrome	singleton	22	1.41	0.89 — 2.14
	multiple	1	1.41	0.04 — 7.83
Interrupted Aortic Arch (Type A and NOS)	singleton	3	0.19	0.04 — 0.56
	multiple	0	0.00	0.00 — 5.19
Patent Ductus Arteriosus				
Patent Ductus Arteriosus	singleton	254	16.34	14.39 — 18.47
	multiple	12	16.87	8.72 — 29.47
Right-Sided Obstruction				
Pulmonary Stenosis, Valvular	singleton	60	3.86	2.94 — 4.97
	multiple	7	9.84	3.96 — 20.28
Pulmonary Valve Atresia w/intact septum	singleton	11	0.71	0.35 — 1.27
	multiple	1	1.41	0.04 — 7.83
Pulmonary Valve Atresia with VSD	singleton	4	0.26	0.07 — 0.66
	multiple	1	1.41	0.04 — 7.83
Tricuspid Valve Atresia	singleton	10	0.64	0.31 — 1.18
	multiple	1	1.41	0.04 — 7.83
Septal Defects				
ASD (Secundum and NOS)	singleton	266	17.11	15.11 — 19.29
	multiple	32	44.99	30.78 — 63.52
VSD (Membranous and NOS)	singleton	130	8.36	6.99 — 9.93
	multiple	25	35.15	22.75 — 51.89
VSD, Conoventricular/Malalignment	singleton	27	1.74	1.14 — 2.53
	multiple	1	1.41	0.04 — 7.83
Single Ventricle and L-TGA				
L-TGA	singleton	4	0.26	0.07 — 0.66
	multiple	2	2.81	0.34 — 10.16
Single Ventricle	singleton	4	0.26	0.07 — 0.66
	multiple	3	4.22	0.87 — 12.33
Other Cardiovascular				
Other Cardiovascular	singleton	188	12.09	10.42 — 13.95
	multiple	18	25.31	15.00 — 40.00
Respiratory: 45 cases				
Choanal Atresia	singleton	7	0.45	0.18 — 0.93

Table 5 (cont'd) Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2000-2001

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
Choanal Atresia	multiple	1	1.41	0.04 — 7.83
Lung Anomalies	singleton	26	1.67	1.09 — 2.45
	multiple	3	4.22	0.87 — 12.33
Other Respiratory	singleton	9	0.58	0.26 — 1.10
	multiple	0	0.00	0.00 — 5.19
Orofacial: 244 cases				
Cleft Lip w/ and w/o Cleft Palate	singleton	122	7.85	6.52 — 9.37
	multiple	5	7.03	2.28 — 16.41
Cleft Palate w/o Cleft Lip	singleton	95	6.11	4.94 — 7.47
	multiple	2	2.81	0.34 — 10.16
Pierre Robin Sequence	singleton	21	1.35	0.84 — 2.06
	multiple	1	1.41	0.04 — 7.83
Other Orofacial	singleton	21	1.35	0.84 — 2.06
	multiple	1	1.41	0.04 — 7.83
Gastrointestinal: 225 cases				
Biliary Atresia	singleton	5	0.32	0.10 — 0.75
	multiple	0	0.00	0.00 — 5.19
Esophageal Atresia/Tracheoesophageal Fistula	singleton	27	1.74	1.14 — 2.53
	multiple	9	12.65	5.79 — 24.02
Hirschsprung Disease	singleton	28	1.80	1.20 — 2.60
	multiple	0	0.00	0.00 — 5.19
Rectal and Large Intestinal Atresia/Stenosis	singleton	56	3.60	2.72 — 4.68
	multiple	7	9.84	3.96 — 20.28
Small Intestinal Atresia	singleton	28	1.80	1.20 — 2.60
	multiple	4	5.62	1.53 — 14.40
Other Gastrointestinal	singleton	75	4.82	3.79 — 6.05
	multiple	7	9.84	3.96 — 20.28
Genitourinary: 329 cases				
Bladder Exstrophy	singleton	6	0.39	0.14 — 0.84

Table 5 (cont'd) Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2000-2001

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
Bladder Exstrophy	multiple	0	0.00	0.00 — 5.19
Cloacal Exstrophy	singleton	0	0.00	0.00 — 0.24
	multiple	1	1.41	0.04 — 7.83
Hypospadias, 2nd or 3rd Degree	singleton	124	7.98	6.63 — 9.51
	multiple	15	21.09	11.80 — 34.79
Obstructive Genitourinary Defect	singleton	114	7.33	6.05 — 8.81
	multiple	11	15.47	7.72 — 27.67
Renal Agenesis/Hypoplasia	singleton	2	0.13	0.02 — 0.46
	multiple	2	2.81	0.34 — 10.16
Other Genitourinary	singleton	100	6.43	5.23 — 7.82
	multiple	13	18.28	9.73 — 31.26
Musculoskeletal: 586 cases				
Club Foot	singleton	139	8.94	7.52 — 10.56
	multiple	20	28.12	17.18 — 43.43
Craniosynostosis	singleton	70	4.50	3.51 — 5.69
	multiple	2	2.81	0.34 — 10.16
Diaphragmatic Hernia	singleton	29	1.87	1.25 — 2.68
	multiple	2	2.81	0.34 — 10.16
Gastroschisis	singleton	33	2.12	1.46 — 2.98
	multiple	1	1.41	0.04 — 7.83
Omphalocele	singleton	22	1.41	0.89 — 2.14
	multiple	2	2.81	0.34 — 10.16
Polydactyly/Syndactyly	singleton	139	8.94	7.52 — 10.56
	multiple	6	8.44	3.10 — 18.36
Reduction Deformity, Lower Limbs	singleton	19	1.22	0.74 — 1.91
	multiple	4	5.62	1.53 — 14.40
Reduction Deformity, Upper Limbs	singleton	45	2.89	2.11 — 3.87
	multiple	1	1.41	0.04 — 7.83
Skeletal Dysplasia	singleton	16	1.03	0.59 — 1.67
	multiple	0	0.00	0.00 — 5.19
Other Musculoskeletal	singleton	95	6.11	4.94 — 7.47
	multiple	14	19.69	10.76 — 33.03

Table 5 (cont'd) Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2000-2001

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
Chromosomal and Other Syndromes: 369 cases				
Klinefelter Syndrome	singleton	7	0.45	0.18 — 0.93
	multiple	0	0.00	0.00 — 5.19
Trisomy 13	singleton	10	0.64	0.31 — 1.18
	multiple	1	1.41	0.04 — 7.83
Trisomy 18	singleton	16	1.03	0.59 — 1.67
	multiple	0	0.00	0.00 — 5.19
Trisomy 21 (Down Syndrome)	singleton	173	11.13	9.53 — 12.91
	multiple	20	28.12	17.18 — 43.43
Turner Syndrome	singleton	15	0.96	0.54 — 1.59
	multiple	0	0.00	0.00 — 5.19
Other Chromosomal Syndromes/Other Syndromes	singleton	123	7.91	6.57 — 9.44
	multiple	5	7.03	2.28 — 16.41
Other: 40 cases				
Amniotic Bands	singleton	5	0.32	0.10 — 0.75
	multiple	0	0.00	0.00 — 5.19
Skin Anomalies	singleton	12	0.77	0.40 — 1.35
	multiple	1	1.41	0.04 — 7.83
Other, Specified	singleton	22	1.41	0.89 — 2.14
	multiple	0	0.00	0.00 — 5.19

1. Plurality is the number of births to a woman from the same pregnancy. A singleton is the birth of one infant. Multiple birth 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.

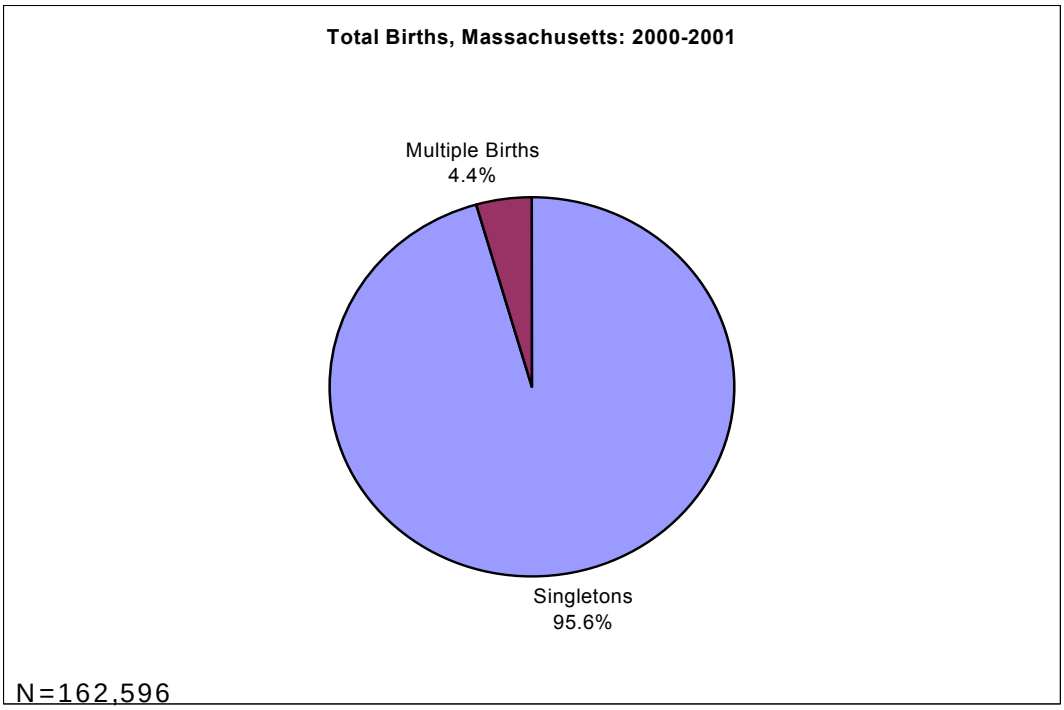
Table 6 Selected Common Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2000-2001

Defect [2]	Count	Rate per 10,000 Births	95% Confidence Interval
MULTIPLE			
ASD (Secundum and NOS)	32	44.99	30.78 — 63.52
VSD (Membranous and NOS)	25	35.15	22.75 — 51.89
Club Foot	20	28.12	17.18 — 43.43
Trisomy 21 (Down Syndrome)	20	28.12	17.18 — 43.43
Hypospadias, 2nd or 3rd Degree	15	21.09	11.80 — 34.79
Obstructive Genitourinary Defect	11	15.47	7.72 — 27.67
Esophageal Atresia/Tracheoesophageal Fistula	9	12.65	5.79 — 24.02
Rectal and Large Intestinal Atresia/Stenosis	7	9.84	3.96 — 20.28
Pulmonary Stenosis, Valvular	7	9.84	3.96 — 20.28
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	6	8.44	3.10 — 18.36
Polydactyly/Syndactyly	6	8.44	3.10 — 18.36
SINGLETON			
ASD (Secundum and NOS)	266	17.11	15.11 — 19.29
Trisomy 21 (Down Syndrome)	173	11.13	9.53 — 12.91
Polydactyly/Syndactyly	139	8.94	7.52 — 10.56
Club Foot	139	8.94	7.52 — 10.56
VSD (Membranous and NOS)	130	8.36	6.99 — 9.93
Hypospadias, 2nd or 3rd Degree	124	7.98	6.63 — 9.51
Cleft Lip w/ and w/o Cleft Palate	122	7.85	6.52 — 9.37
Obstructive Genitourinary Defect	114	7.33	6.05 — 8.81
Cleft Palate w/o Cleft Lip	95	6.11	4.94 — 7.47
Craniosynostosis	70	4.50	3.51 — 5.69

1. Plurality is the number of births to a woman from the same pregnancy. A singleton is the birth of one infant. Multiple birth 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



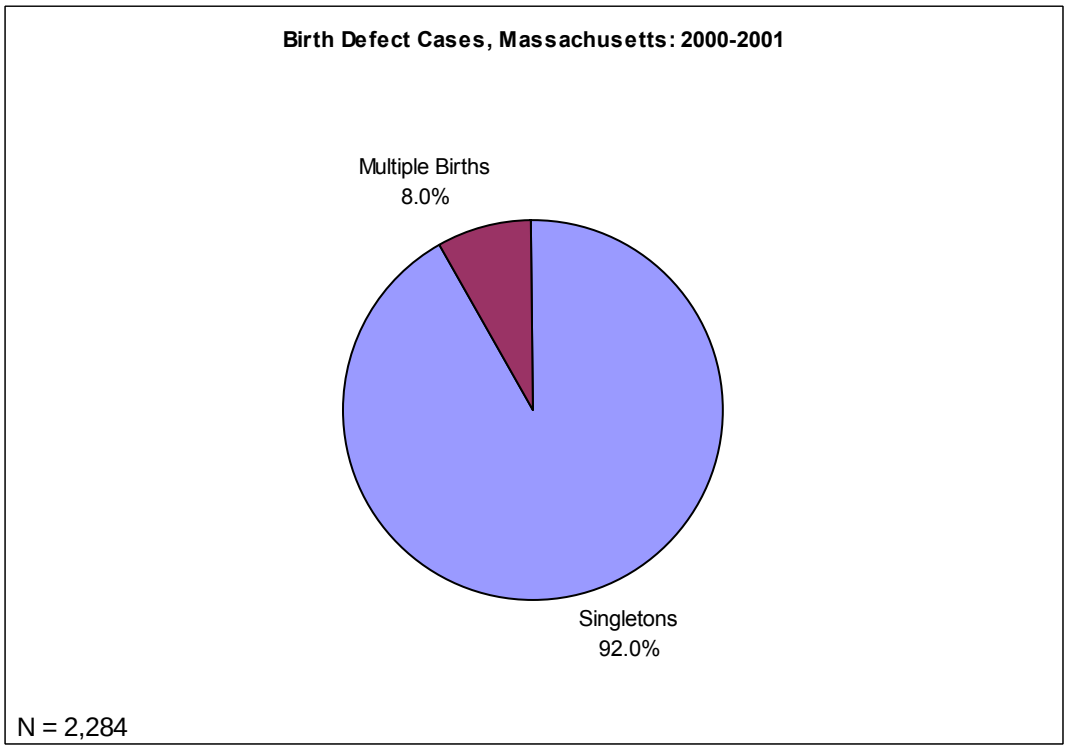
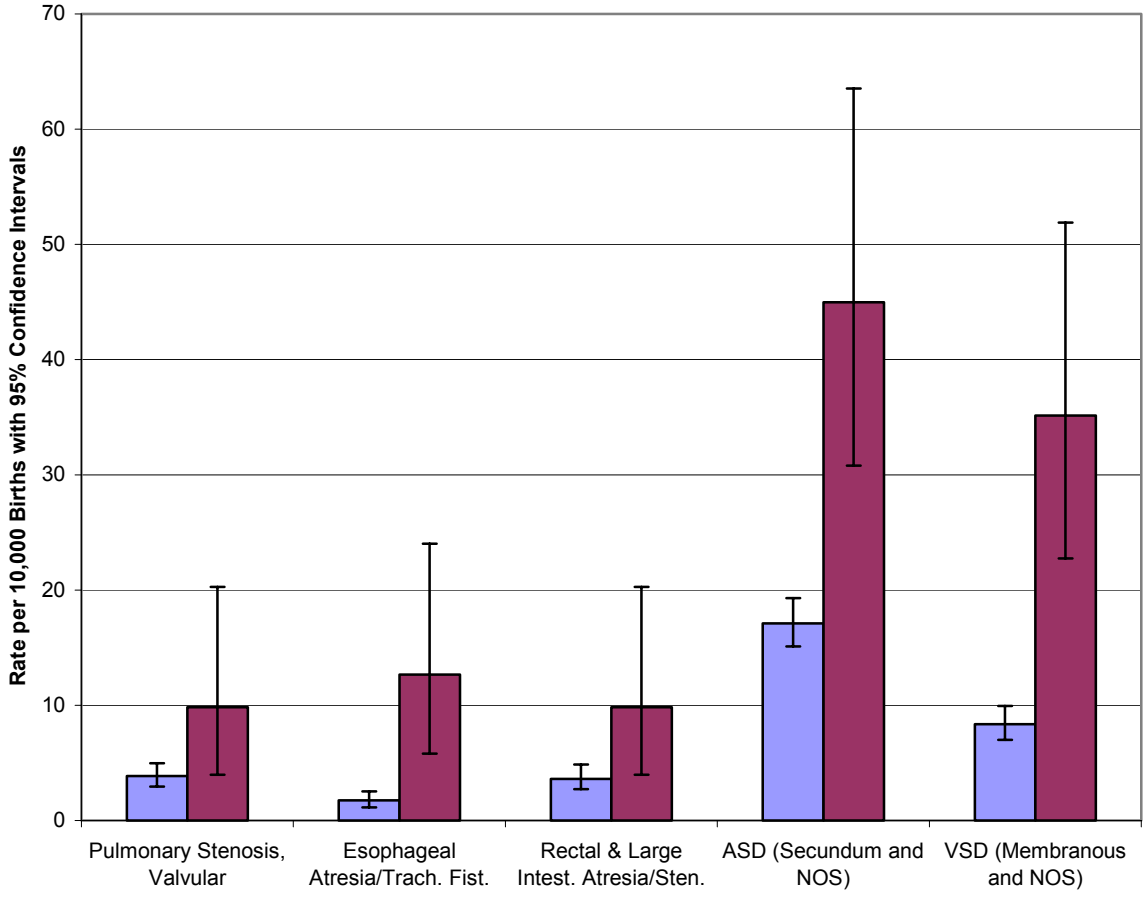


Figure 4 Prevalence of Selected Birth Defects by Plurality among Live Births and Stillbirths, Massachusetts: 2000-2001



Defects selected for reaching or almost reaching a statistically significant comparison.

Table 7 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2000-2001

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
Central Nervous System: 219 cases				
Anencephaly	male	1	0.12	0.00—0.67
	female	6	0.75	0.28—1.64
Encephalocele	male	4	0.48	0.13—1.24
	female	3	0.38	0.08—1.10
Holoprosencephaly	male	6	0.72	0.27—1.58
	female	3	0.38	0.08—1.10
Hydrocephaly w/o Spina Bifida	male	22	2.65	1.66—4.02
	female	25	3.14	2.03—4.63
Microcephaly	male	19	2.29	1.38—3.58
	female	23	2.89	1.83—4.33
Spina Bifida w/ and w/o Hydrocephaly	male	17	2.05	1.19—3.28
	female	12	1.51	0.78—2.63
Tethered Cord	male	17	2.05	1.19—3.28
	female	11	1.38	0.69—2.47
Other CNS	male	45	5.43	3.96—7.26
	female	43	5.40	3.90—7.27
Eye: 62 cases				
Aniridia	male	4	0.48	0.13—1.24
	female	4	0.50	0.14—1.29
Anophthalmia/Microphthalmia	male	9	1.09	0.50—2.06
	female	8	1.00	0.43—1.98
Congenital Glaucoma, Congenital Cataract	male	17	2.05	1.19—3.28
	female	13	1.63	0.87—2.79
Other Eye	male	8	0.97	0.42—1.90
	female	13	1.63	0.87—2.79
Ear: 23 cases				
Anotia/Microtia	male	13	1.57	0.84—2.68
	female	7	0.88	0.35—1.81
Other Ear	male	5	0.60	0.20—1.41
	female	1	0.13	0.00—0.70

Table 7 (cont'd) Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2000-2001

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
Cardiovascular: 782 cases				
Anomalous Pulmonary Venous Connection				
Total/Partial Anomalous Pulmonary Venous Connection	male	13	1.57	0.84—2.68
	female	11	1.38	0.69—2.47
Atrioventricular Canal Defects				
ASD Primum	male	4	0.48	0.13—1.24
	female	12	1.51	0.78—2.63
Common Atrium	male	2	0.24	0.03—0.87
	female	2	0.25	0.03—0.91
Complete Atrioventricular Canal Defect	male	23	2.77	1.76—4.16
	female	22	2.76	1.73—4.18
Endocardial Cushion (OS and NOS)	male	4	0.48	0.13—1.24
	female	5	0.63	0.20—1.46
VSD, Canal Type	male	4	0.48	0.13—1.24
	female	7	0.88	0.35—1.81
Conotruncal (Outlet) and Aortic Arch				
Double Outlet Right Ventricle	male	9	1.09	0.50—2.06
	female	10	1.25	0.60—2.31
d-Transposition of the Great Arteries	male	14	1.69	0.92—2.83
	female	11	1.38	0.69—2.47
Interrupted Aortic Arch, Type B	male	1	0.12	0.00—0.67
	female	3	0.38	0.08—1.10
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	male	40	4.83	3.45—6.57
	female	32	4.02	2.75—5.67
Truncus	male	4	0.48	0.13—1.24
	female	2	0.25	0.03—0.91
Ebstein Anomaly				
Ebstein Anomaly	male	4	0.48	0.13—1.24
	female	2	0.25	0.03—0.91
Heterotaxy (Laterality Defects)				
Heterotaxy	male	7	0.84	0.34—1.74
	female	10	1.25	0.60—2.31

Table 7 (cont'd) Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2000-2001

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
Left-Sided Obstruction				
Aortic Valve Stenosis	male	12	1.45	0.75—2.53
	female	5	0.63	0.20—1.46
Coarctation of Aorta	male	44	5.31	3.86—7.13
	female	23	2.89	1.83—4.33
Hypoplastic Left Heart Syndrome	male	17	2.05	1.19—3.28
	female	6	0.75	0.28—1.64
Interrupted Aortic Arch (Type A and NOS)	male	2	0.24	0.03—0.87
	female	1	0.13	0.00—0.70
Patent Ductus Arteriosus				
Patent Ductus Arteriosus	male	140	16.89	14.21—19.93
	female	126	15.81	13.17—18.82
Right-Sided Obstruction				
Pulmonary Stenosis, Valvular	male	39	4.70	3.35—6.43
	female	28	3.51	2.33—5.08
Pulmonary Valve Atresia w/intact septum	male	8	0.97	0.42—1.90
	female	4	0.50	0.14—1.29
Pulmonary Valve Atresia with VSD	male	3	0.36	0.07—1.06
	female	2	0.25	0.03—0.91
Tricuspid Valve Atresia	male	6	0.72	0.27—1.58
	female	5	0.63	0.20—1.46
Septal Defects				
ASD (Secundum and NOS)	male	164	19.78	16.87—23.05
	female	134	16.81	14.09—19.91
VSD (Membranous and NOS)	male	75	9.05	7.12—11.34
	female	80	10.04	7.96—12.49
VSD, Conoventricular/Malalignment	male	13	1.57	0.84—2.68
	female	15	1.88	1.05—3.10
Single Ventricle and L-TGA				
L-TGA	male	5	0.60	0.20—1.41
	female	1	0.13	0.00—0.70
Single Ventricle	male	7	0.84	0.34—1.74
	female	0	0.00	0.00—0.46
Other Cardiovascular				
Other Cardiovascular	male	118	14.23	11.78—17.05
	female	87	10.92	8.74—13.46

Table 7 (cont'd) Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2000-2001

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
Respiratory: 45 cases				
Choanal Atresia	male	2	0.24	0.03—0.87
	female	6	0.75	0.28—1.64
Lung Anomalies	male	17	2.05	1.19—3.28
	female	12	1.51	0.78—2.63
Other Respiratory	male	7	0.84	0.34—1.74
	female	2	0.25	0.03—0.91
Orofacial: 244 cases				
Cleft Lip w/ and w/o Cleft Palate	male	77	9.29	7.33—11.61
	female	50	6.27	4.66—8.27
Cleft Palate w/o Cleft Lip	male	46	5.55	4.06—7.40
	female	51	6.40	4.76—8.41
Pierre Robin Sequence	male	14	1.69	0.92—2.83
	female	8	1.00	0.43—1.98
Other Orofacial	male	12	1.45	0.75—2.53
	female	10	1.25	0.60—2.31
Gastrointestinal: 225 cases				
Biliary Atresia	male	3	0.36	0.07—1.06
	female	2	0.25	0.03—0.91
Esophageal Atresia/Tracheoesophageal Fistula	male	22	2.65	1.66—4.02
	female	14	1.76	0.96—2.95
Hirschsprung Disease	male	22	2.65	1.66—4.02
	female	6	0.75	0.28—1.64
Rectal and Large Intestinal Atresia/Stenosis	male	38	4.58	3.24—6.29
	female	25	3.14	2.03—4.63
Small Intestinal Atresia	male	10	1.21	0.58—2.22
	female	22	2.76	1.73—4.18
Other Gastrointestinal	male	39	4.70	3.35—6.43
	female	43	5.40	3.90—7.27

Table 7 (cont'd) Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2000-2001

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
Genitourinary: 329 cases				
Bladder Exstrophy	male	2	0.24	0.03—0.87
	female	4	0.50	0.14—1.29
Cloacal Exstrophy	male	0	0.00	0.00—0.45
	female	1	0.13	0.00—0.70
Hypospadias, 2nd or 3rd Degree	male	139	16.77	14.10—19.80
	female	0	0.00	0.00—0.46
Obstructive Genitourinary Defect	male	87	10.50	8.41—12.95
	female	38	4.77	3.37—6.54
Renal Agenesis/Hypoplasia	male	3	0.36	0.07—1.06
	female	1	0.13	0.00—0.70
Other Genitourinary	male	64	7.72	5.95—9.86
	female	49	6.15	4.55—8.13
Musculoskeletal: 586 cases				
Club Foot	male	110	13.27	10.91—15.99
	female	49	6.15	4.55—8.13
Craniosynostosis	male	44	5.31	3.86—7.13
	female	28	3.51	2.33—5.08
Diaphragmatic Hernia	male	19	2.29	1.38—3.58
	female	12	1.51	0.78—2.63
Gastroschisis	male	18	2.17	1.29—3.43
	female	16	2.01	1.15—3.26
Omphalocele	male	10	1.21	0.58—2.22
	female	14	1.76	0.96—2.95
Polydactyly/Syndactyly	male	82	9.89	7.87—12.28
	female	63	7.90	6.07—10.11
Reduction Deformity, Lower Limbs	male	10	1.21	0.58—2.22
	female	13	1.63	0.87—2.79
Reduction Deformity, Upper Limbs	male	28	3.38	2.24—4.88
	female	18	2.26	1.34—3.57
Skeletal Dysplasia	male	9	1.09	0.50—2.06
	female	7	0.88	0.35—1.81
Other Musculoskeletal	male	63	7.60	5.84—9.72
	female	46	5.77	4.23—7.70

Table 7 (cont'd) Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2000-2001

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
Chromosomal and Other Syndromes: 369 cases				
Klinefelter Syndrome	male	7	0.84	0.34—1.74
	female	0	0.00	0.00—0.46
Trisomy 13	male	5	0.60	0.20—1.41
	female	6	0.75	0.28—1.64
Trisomy 18	male	7	0.84	0.34—1.74
	female	9	1.13	0.52—2.14
Trisomy 21 (Down Syndrome)	male	110	13.27	10.91—15.99
	female	83	10.41	8.29—12.91
Turner Syndrome	male	0	0.00	0.00—0.45
	female	15	1.88	1.05—3.10
Other Chromosomal Syndromes/Other Syndromes	male	63	7.60	5.84—9.72
	female	65	8.16	6.29—10.40
Other: 40 cases				
Amniotic Bands	male	1	0.12	0.00—0.67
	female	4	0.50	0.14—1.29
Skin Anomalies	male	10	1.21	0.58—2.22
	female	3	0.38	0.08—1.10
Other, Specified	male	6	0.72	0.27—1.58
	female	16	2.01	1.15—3.26

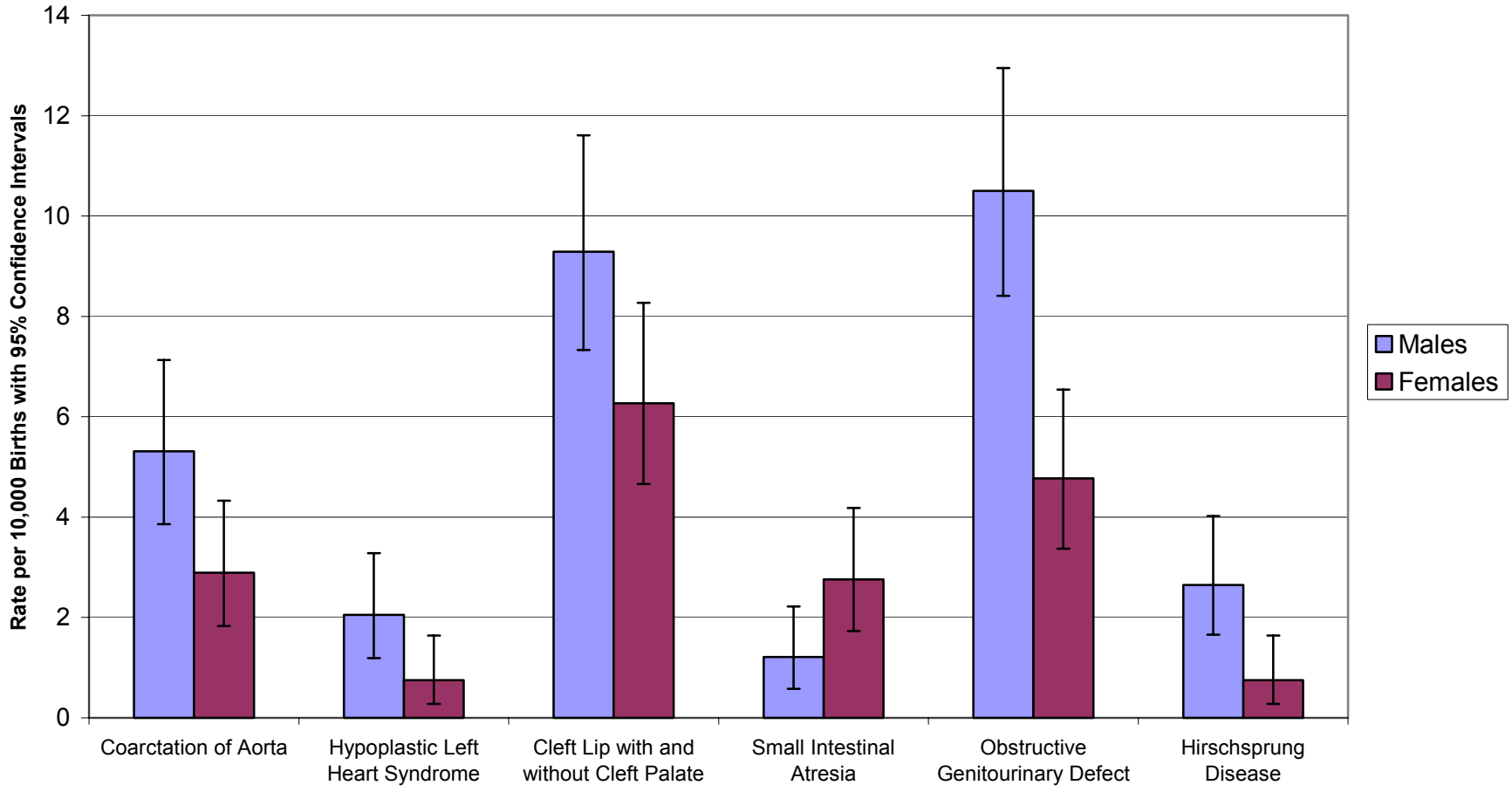
1. Cases can be included in more than one defect category. Cases are counted once within a defect category. Counts represent the number of defects, not the total number of cases with birth defects.

Table 8 Selected Common Defects by Sex of Live Births and Stillbirths, Massachusetts: 2000-2001

Defect[1]	Count	Rate per 10,000 Births	95% Confidence Interval
FEMALE			
ASD (Secundum and NOS)	134	16.81	14.09 — 19.91
Trisomy 21 (Down Syndrome)	83	10.41	8.29 — 12.91
VSD (Membranous and NOS)	80	10.04	7.96 — 12.49
Polydactyly/Syndactyly	63	7.90	6.07 — 10.11
Cleft Palate w/o Cleft Lip	51	6.40	4.76 — 8.41
Cleft Lip w/ and w/o Cleft Palate	50	6.27	4.66 — 8.27
Club Foot	49	6.15	4.55 — 8.13
Obstructive Genitourinary Defect	38	4.77	3.37 — 6.54
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	32	4.02	2.75 — 5.67
Craniosynostosis	28	3.51	2.33 — 5.08
Pulmonary Stenosis, Valvular	28	3.51	2.33 — 5.08
MALE			
ASD (Secundum and NOS)	164	19.78	16.87 — 23.05
Hypospadias, 2nd or 3rd Degree	139	16.77	14.10 — 19.80
Club Foot	110	13.27	10.91 — 15.99
Trisomy 21 (Down Syndrome)	110	13.27	10.91 — 15.99
Obstructive Genitourinary Defect	87	10.50	8.41 — 12.95
Polydactyly/Syndactyly	82	9.89	7.87 — 12.28
Cleft Lip w/ and w/o Cleft Palate	77	9.29	7.33 — 11.61
VSD (Membranous and NOS)	75	9.05	7.12 — 11.34
Cleft Palate w/o Cleft Lip	46	5.55	4.06 — 7.40
Craniosynostosis	44	5.31	3.86 — 7.13
Coarctation of Aorta	44	5.31	3.86 — 7.13

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

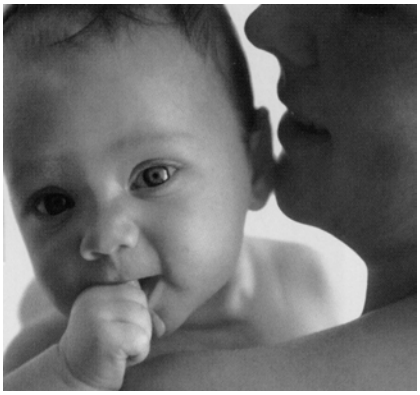
Figure 5 Prevalence of Selected Birth Defects By Sex of Infant among Live Births and Stillbirths, Massachusetts: 2000-2001



Defects selected for reaching or almost reaching a statistically significant comparison.

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 152.12 for mothers younger than 20 years, 135.87 for those 20-24 years, 130.24 for those 25-29 years, 126.53 for those 30-34 years, and 155.57 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). Women 35 years and older had a live birth Down Syndrome rate of 26.73 per 10,000 births. This rate was three times that of any other maternal age group. Figure 7 shows that younger mothers (aged 19 and under) had the highest rate (16.26%) 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 Hydrocephaly, Limb Reductions, and Polydactyly/Syndactyly than other age groups. Older mothers had higher rates for many heart defects, including Septal Defects, both Atrial and Ventricular Membranous, Atrioventricular Canal Defects and Coarctation of Aorta. 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 more common birth defects by maternal age groups. Atrial Septal Defects were common to all maternal age groups. Hypospadias and Polydactyly/Syndactyly were among the top five most common in every age group except mothers 35 and older.

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 throughout the 1990s. Births to every age group above 30 have increased since 1990 while births to age groups below 30 have decreased. (Massachusetts Births 2001)

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 137.37 for Non-Hispanic Whites, 151.50 for Non-Hispanic Blacks, 139.36 for Hispanics, and 101.58 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, Down Syndrome, Polydactyly/Syndactyly, and Clubfoot. In Blacks, the most common defects included Septal Defects, Polydactyly/Syndactyly, Down Syndrome, and Hypospadias. The most common defects in Whites included Septal Heart Defects, Down Syndrome, Clubfoot and Hypospadias. In Asians, the most common defects included Septal Defects, Cleft Lip, Cleft Palate, and Polydactyly/Syndactyly.

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: 2000-2001

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Central Nervous System				
Anencephaly	<20	0	0.00	0.00—3.53
	20-24	2	0.83	0.10—3.01
	25-29	1	0.26	0.01—1.44
	30-34	2	0.37	0.05—1.34
	35+	2	0.56	0.07—2.03
Encephalocele	<20	1	0.96	0.02—5.33
	20-24	1	0.42	0.01—2.32
	25-29	1	0.26	0.01—1.44
	30-34	2	0.37	0.05—1.34
	35+	2	0.56	0.07—2.03
Holoprosencephaly	<20	0	0.00	0.00—3.53
	20-24	1	0.42	0.01—2.32
	25-29	3	0.77	0.16—2.26
	30-34	2	0.37	0.05—1.34
	35+	3	0.84	0.17—2.47
Hydrocephaly w/o Spina Bifida	<20	7	6.70	2.69—13.80
	20-24	9	3.75	1.72—7.12
	25-29	10	2.58	1.24—4.74
	30-34	11	2.04	1.02—3.66
	35+	8	2.25	0.97—4.43
Microcephaly	<20	5	4.78	1.55—11.16
	20-24	7	2.92	1.17—6.01
	25-29	13	3.35	1.79—5.73
	30-34	11	2.04	1.02—3.66
	35+	6	1.69	0.62—3.67
Spina Bifida w/ and w/o Hydrocephaly	<20	4	3.83	1.04—9.80
	20-24	5	2.08	0.68—4.86
	25-29	10	2.58	1.24—4.74
	30-34	4	0.74	0.20—1.90
	35+	6	1.69	0.62—3.67
Tethered Cord	<20	0	0.00	0.00—3.53
	20-24	2	0.83	0.10—3.01
	25-29	9	2.32	1.06—4.41
	30-34	9	1.67	0.76—3.17
	35+	8	2.25	0.97—4.43

Table 9 (cont'd) Prevalence of Birth Defects by Maternal Age for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Other CNS	<20	10	9.57	4.59—17.60
	20-24	14	5.83	3.19—9.79
	25-29	23	5.93	3.76—8.90
	30-34	22	4.09	2.56—6.19
	35+	18	5.06	3.00—8.00
Eye				
Aniridia	<20	0	0.00	0.00—3.53
	20-24	0	0.00	0.00—1.54
	25-29	2	0.52	0.06—1.86
	30-34	2	0.37	0.05—1.34
	35+	4	1.13	0.31—2.88
Anophthalmia/Microphthalmia	<20	1	0.96	0.02—5.33
	20-24	2	0.83	0.10—3.01
	25-29	5	1.29	0.42—3.01
	30-34	7	1.30	0.52—2.68
	35+	2	0.56	0.07—2.03
Congenital Glaucoma, Congenital Cataract	<20	3	2.87	0.59—8.39
	20-24	7	2.92	1.17—6.01
	25-29	6	1.55	0.57—3.37
	30-34	11	2.04	1.02—3.66
	35+	3	0.84	0.17—2.47
Other Eye	<20	2	1.91	0.23—6.91
	20-24	3	1.25	0.26—3.65
	25-29	7	1.81	0.73—3.72
	30-34	6	1.11	0.41—2.43
	35+	3	0.84	0.17—2.47
Ear				
Anotia/Microtia	<20	2	1.91	0.23—6.91
	20-24	3	1.25	0.26—3.65
	25-29	7	1.81	0.73—3.72
	30-34	4	0.74	0.20—1.90
	35+	4	1.13	0.31—2.88

Table 9 (cont'd) Prevalence of Birth Defects by Maternal Age for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Other Ear	<20	1	0.96	0.02—5.33
	20-24	1	0.42	0.01—2.32
	25-29	1	0.26	0.01—1.44
	30-34	2	0.37	0.05—1.34
	35+	1	0.28	0.01—1.57
Cardiovascular				
Anomalous Pulmonary Venous Connection				
Total/Partial Anomalous Pulmonary Venous Connection	<20	1	0.96	0.02—5.33
	20-24	3	1.25	0.26—3.65
	25-29	5	1.29	0.42—3.01
	30-34	8	1.49	0.64—2.93
	35+	7	1.97	0.79—4.06
Atrioventricular Canal Defects				
ASD Primum	<20	0	0.00	0.00—3.53
	20-24	1	0.42	0.01—2.32
	25-29	2	0.52	0.06—1.86
	30-34	5	0.93	0.30—2.17
	35+	8	2.25	0.97—4.43
Common Atrium	<20	0	0.00	0.00—3.53
	20-24	0	0.00	0.00—1.54
	25-29	1	0.26	0.01—1.44
	30-34	1	0.19	0.00—1.04
	35+	1	0.28	0.01—1.57
Complete Atrioventricular Canal Defect	<20	1	0.96	0.02—5.33
	20-24	7	2.92	1.17—6.01
	25-29	8	2.06	0.89—4.07
	30-34	12	2.23	1.15—3.89
	35+	15	4.22	2.36—6.96
Endocardial Cushion (OS and NOS)	<20	0	0.00	0.00—3.53
	20-24	1	0.42	0.01—2.32
	25-29	2	0.52	0.06—1.86
	30-34	0	0.00	0.00—0.69
	35+	6	1.69	0.62—3.67

Table 9 (cont'd) Prevalence of Birth Defects by Maternal Age for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
VSD, Canal Type	<20	0	0.00	0.00—3.53
	20-24	1	0.42	0.01—2.32
	25-29	1	0.26	0.01—1.44
	30-34	1	0.19	0.00—1.04
	35+	8	2.25	0.97—4.43
Conotruncal (Outlet) and Aortic Arch				
Double Outlet Right Ventricle	<20	1	0.96	0.02—5.33
	20-24	5	2.08	0.68—4.86
	25-29	5	1.29	0.42—3.01
	30-34	5	0.93	0.30—2.17
	35+	3	0.84	0.17—2.47
d-Transposition of the Great Arteries	<20	1	0.96	0.02—5.33
	20-24	2	0.83	0.10—3.01
	25-29	4	1.03	0.28—2.64
	30-34	14	2.60	1.42—4.36
	35+	3	0.84	0.17—2.47
Interrupted Aortic Arch, Type B	<20	0	0.00	0.00—3.53
	20-24	1	0.42	0.01—2.32
	25-29	1	0.26	0.01—1.44
	30-34	2	0.37	0.05—1.34
	35+	0	0.00	0.00—1.04
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	<20	0	0.00	0.00—3.53
	20-24	12	5.00	2.58—8.74
	25-29	17	4.38	2.55—7.02
	30-34	21	3.90	2.42—5.96
	35+	19	5.35	3.22—8.35
Truncus	<20	1	0.96	0.02—5.33
	20-24	0	0.00	0.00—1.54
	25-29	3	0.77	0.16—2.26
	30-34	2	0.37	0.05—1.34
	35+	0	0.00	0.00—1.04
Ebstein Anomaly				
Ebstein Anomaly	<20	0	0.00	0.00—3.53
	20-24	0	0.00	0.00—1.54
	25-29	3	0.77	0.16—2.26
	30-34	0	0.00	0.00—0.69
	35+	1	0.28	0.01—1.57

Table 9 (cont'd) Prevalence of Birth Defects by Maternal Age for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Heterotaxy (Laterality Defects)				
Heterotaxy	<20	0	0.00	0.00—3.53
	20-24	3	1.25	0.26—3.65
	25-29	5	1.29	0.42—3.01
	30-34	6	1.11	0.41—2.43
	35+	2	0.56	0.07—2.03
Left-Sided Obstruction				
Aortic Valve Stenosis	<20	0	0.00	0.00—3.53
	20-24	4	1.67	0.45—4.27
	25-29	2	0.52	0.06—1.86
	30-34	4	0.74	0.20—1.90
	35+	6	1.69	0.62—3.67
Coarctation of Aorta	<20	4	3.83	1.04—9.80
	20-24	9	3.75	1.72—7.12
	25-29	11	2.84	1.42—5.08
	30-34	23	4.27	2.71—6.41
	35+	18	5.06	3.00—8.00
Hypoplastic Left Heart Syndrome	<20	0	0.00	0.00—3.53
	20-24	5	2.08	0.68—4.86
	25-29	6	1.55	0.57—3.37
	30-34	10	1.86	0.89—3.42
	35+	2	0.56	0.07—2.03
Interrupted Aortic Arch (Type A and NOS)	<20	1	0.96	0.02—5.33
	20-24	0	0.00	0.00—1.54
	25-29	0	0.00	0.00—0.95
	30-34	1	0.19	0.00—1.04
	35+	1	0.28	0.01—1.57
Patent Ductus Arteriosus				
Patent Ductus Arteriosus	<20	13	12.44	6.62—21.27
	20-24	39	16.25	11.56—22.22
	25-29	59	15.22	11.58—19.63
	30-34	90	16.72	13.45—20.55
	35+	64	18.00	13.87—22.99

Table 9 (cont'd) Prevalence of Birth Defects by Maternal Age for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Right-Sided Obstruction				
Pulmonary Stenosis, Valvular	<20	1	0.96	0.02—5.33
	20-24	7	2.92	1.17—6.01
	25-29	18	4.64	2.75—7.34
	30-34	29	5.39	3.61—7.74
	35+	12	3.38	1.74—5.90
Pulmonary Valve Atresia w/intact septum	<20	0	0.00	0.00—3.53
	20-24	1	0.42	0.01—2.32
	25-29	4	1.03	0.28—2.64
	30-34	3	0.56	0.11—1.63
	35+	3	0.84	0.17—2.47
Pulmonary Valve Atresia with VSD	<20	0	0.00	0.00—3.53
	20-24	1	0.42	0.01—2.32
	25-29	2	0.52	0.06—1.86
	30-34	2	0.37	0.05—1.34
	35+	0	0.00	0.00—1.04
Tricuspid Valve Atresia	<20	1	0.96	0.02—5.33
	20-24	0	0.00	0.00—1.54
	25-29	5	1.29	0.42—3.01
	30-34	1	0.19	0.00—1.04
	35+	2	0.56	0.07—2.03
Septal Defects				
ASD (Secundum and NOS)	<20	12	11.48	5.93—20.06
	20-24	38	15.84	11.21—21.74
	25-29	69	17.79	13.85—22.52
	30-34	95	17.65	14.28—21.58
	35+	78	21.94	17.34—27.39
VSD (Membranous and NOS)	<20	8	7.65	3.30—15.08
	20-24	15	6.25	3.50—10.31
	25-29	39	10.06	7.15—13.75
	30-34	43	7.99	5.78—10.76
	35+	47	13.22	9.71—17.58
VSD, Conoventricular/Malalignment	<20	1	0.96	0.02—5.33
	20-24	4	1.67	0.45—4.27
	25-29	5	1.29	0.42—3.01
	30-34	15	2.79	1.56—4.60
	35+	3	0.84	0.17—2.47

Table 9 (cont'd) Prevalence of Birth Defects by Maternal Age for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Single Ventricle and L-TGA				
L-TGA	<20	0	0.00	0.00—3.53
	20-24	1	0.42	0.01—2.32
	25-29	2	0.52	0.06—1.86
	30-34	1	0.19	0.00—1.04
	35+	2	0.56	0.07—2.03
Single Ventricle	<20	1	0.96	0.02—5.33
	20-24	0	0.00	0.00—1.54
	25-29	3	0.77	0.16—2.26
	30-34	2	0.37	0.05—1.34
	35+	1	0.28	0.01—1.57
Other Cardiovascular				
Other Cardiovascular	<20	7	6.70	2.69—13.80
	20-24	31	12.92	8.78—18.34
	25-29	48	12.38	9.13—16.41
	30-34	69	12.82	9.97—16.22
	35+	43	12.10	8.75—16.29
Respiratory				
Choanal Atresia	<20	0	0.00	0.00—3.53
	20-24	0	0.00	0.00—1.54
	25-29	1	0.26	0.01—1.44
	30-34	5	0.93	0.30—2.17
	35+	2	0.56	0.07—2.03
Lung Anomalies	<20	1	0.96	0.02—5.33
	20-24	4	1.67	0.45—4.27
	25-29	9	2.32	1.06—4.41
	30-34	10	1.86	0.89—3.42
	35+	5	1.41	0.46—3.28
Other Respiratory	<20	3	2.87	0.59—8.39
	20-24	2	0.83	0.10—3.01
	25-29	1	0.26	0.01—1.44
	30-34	2	0.37	0.05—1.34
	35+	1	0.28	0.01—1.57

Table 9 (cont'd) Prevalence of Birth Defects by Maternal Age for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Orofacial				
Cleft Lip w/ and w/o Cleft Palate	<20	8	7.65	3.30—15.08
	20-24	20	8.34	5.09—12.87
	25-29	34	8.77	6.07—12.25
	30-34	31	5.76	3.91—8.18
	35+	31	8.72	5.93—12.38
Cleft Palate w/o Cleft Lip	<20	7	6.70	2.69—13.80
	20-24	13	5.42	2.88—9.26
	25-29	22	5.67	3.56—8.59
	30-34	35	6.50	4.53—9.04
	35+	19	5.35	3.22—8.35
Pierre Robin Sequence	<20	1	0.96	0.02—5.33
	20-24	2	0.83	0.10—3.01
	25-29	4	1.03	0.28—2.64
	30-34	10	1.86	0.89—3.42
	35+	5	1.41	0.46—3.28
Other Orofacial	<20	1	0.96	0.02—5.33
	20-24	4	1.67	0.45—4.27
	25-29	2	0.52	0.06—1.86
	30-34	9	1.67	0.76—3.17
	35+	6	1.69	0.62—3.67
Gastrointestinal				
Biliary Atresia	<20	0	0.00	0.00—3.53
	20-24	0	0.00	0.00—1.54
	25-29	2	0.52	0.06—1.86
	30-34	3	0.56	0.11—1.63
	35+	0	0.00	0.00—1.04
Esophageal Atresia/Tracheoesophageal Fistula	<20	1	0.96	0.02—5.33
	20-24	2	0.83	0.10—3.01
	25-29	11	2.84	1.42—5.08
	30-34	12	2.23	1.15—3.89
	35+	9	2.53	1.16—4.81

Table 9 (cont'd) Prevalence of Birth Defects by Maternal Age for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Hirschsprung Disease	<20	1	0.96	0.02—5.33
	20-24	1	0.42	0.01—2.32
	25-29	5	1.29	0.42—3.01
	30-34	12	2.23	1.15—3.89
	35+	9	2.53	1.16—4.81
Rectal and Large Intestinal Atresia/Stenosis	<20	4	3.83	1.04—9.80
	20-24	10	4.17	2.00—7.66
	25-29	15	3.87	2.17—6.38
	30-34	17	3.16	1.84—5.06
	35+	16	4.50	2.57—7.31
Small Intestinal Atresia	<20	2	1.91	0.23—6.91
	20-24	5	2.08	0.68—4.86
	25-29	8	2.06	0.89—4.07
	30-34	10	1.86	0.89—3.42
	35+	7	1.97	0.79—4.06
Other Gastrointestinal	<20	9	8.61	3.94—16.35
	20-24	17	7.09	4.13—11.34
	25-29	8	2.06	0.89—4.07
	30-34	33	6.13	4.22—8.61
	35+	15	4.22	2.36—6.96
Genitourinary				
Bladder Exstrophy	<20	1	0.96	0.02—5.33
	20-24	0	0.00	0.00—1.54
	25-29	1	0.26	0.01—1.44
	30-34	3	0.56	0.11—1.63
	35+	1	0.28	0.01—1.57
Cloacal Exstrophy	<20	0	0.00	0.00—3.53
	20-24	0	0.00	0.00—1.54
	25-29	0	0.00	0.00—0.95
	30-34	1	0.19	0.00—1.04
	35+	0	0.00	0.00—1.04
Hypospadias, 2nd or 3rd Degree	<20	10	9.57	4.59—17.60
	20-24	20	8.34	5.09—12.87
	25-29	41	10.57	7.59—14.34
	30-34	44	8.18	5.94—10.97
	35+	24	6.75	4.33—10.05

Table 9 (cont'd) Prevalence of Birth Defects by Maternal Age for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Obstructive Genitourinary Defect	<20	9	8.61	3.94—16.35
	20-24	17	7.09	4.13—11.34
	25-29	31	7.99	5.43—11.35
	30-34	37	6.87	4.84—9.48
	35+	31	8.72	5.93—12.38
Renal Agenesis/Hypoplasia	<20	0	0.00	0.00—3.53
	20-24	2	0.83	0.10—3.01
	25-29	0	0.00	0.00—0.95
	30-34	0	0.00	0.00—0.69
	35+	0	0.00	0.00—1.04
Other Genitourinary	<20	7	6.70	2.69—13.80
	20-24	12	5.00	2.58—8.74
	25-29	21	5.42	3.35—8.28
	30-34	38	7.06	5.00—9.69
	35+	33	9.28	6.39—13.04
Musculoskeletal				
Club Foot	<20	10	9.57	4.59—17.60
	20-24	30	12.50	8.44—17.85
	25-29	34	8.77	6.07—12.25
	30-34	40	7.43	5.31—10.12
	35+	41	11.53	8.28—15.65
Craniosynostosis	<20	1	0.96	0.02—5.33
	20-24	11	4.58	2.29—8.20
	25-29	19	4.90	2.95—7.65
	30-34	22	4.09	2.56—6.19
	35+	19	5.35	3.22—8.35
Diaphragmatic Hernia	<20	3	2.87	0.59—8.39
	20-24	3	1.25	0.26—3.65
	25-29	9	2.32	1.06—4.41
	30-34	10	1.86	0.89—3.42
	35+	4	1.13	0.31—2.88
Gastroschisis	<20	17	16.26	9.47—26.04
	20-24	11	4.58	2.29—8.20
	25-29	3	0.77	0.16—2.26
	30-34	1	0.19	0.00—1.04
	35+	0	0.00	0.00—1.04

Table 9 (cont'd) Prevalence of Birth Defects by Maternal Age for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Omphalocele	<20	1	0.96	0.02—5.33
	20-24	4	1.67	0.45—4.27
	25-29	3	0.77	0.16—2.26
	30-34	11	2.04	1.02—3.66
	35+	2	0.56	0.07—2.03
Polydactyly/Syndactyly	<20	16	15.31	8.75—24.86
	20-24	21	8.75	5.42—13.38
	25-29	35	9.03	6.29—12.55
	30-34	44	8.18	5.94—10.97
	35+	27	7.60	5.01—11.05
Reduction Deformity, Lower Limbs	<20	3	2.87	0.59—8.39
	20-24	2	0.83	0.10—3.01
	25-29	6	1.55	0.57—3.37
	30-34	8	1.49	0.64—2.93
	35+	3	0.84	0.17—2.47
Reduction Deformity, Upper Limbs	<20	8	7.65	3.30—15.08
	20-24	7	2.92	1.17—6.01
	25-29	11	2.84	1.42—5.08
	30-34	10	1.86	0.89—3.42
	35+	9	2.53	1.16—4.81
Skeletal Dysplasia	<20	2	1.91	0.23—6.91
	20-24	4	1.67	0.45—4.27
	25-29	0	0.00	0.00—0.95
	30-34	7	1.30	0.52—2.68
	35+	3	0.84	0.17—2.47
Other Musculoskeletal	<20	17	16.26	9.47—26.04
	20-24	17	7.09	4.13—11.34
	25-29	27	6.96	4.59—10.13
	30-34	29	5.39	3.61—7.74
	35+	17	4.78	2.79—7.66
Chromosomal and Other Syndromes				
Klinefelter Syndrome	<20	0	0.00	0.00—3.53
	20-24	1	0.42	0.01—2.32
	25-29	0	0.00	0.00—0.95
	30-34	0	0.00	0.00—0.69
	35+	6	1.69	0.62—3.67

Table 9 (cont'd) Prevalence of Birth Defects by Maternal Age for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Trisomy 13	<20	0	0.00	0.00—3.53
	20-24	0	0.00	0.00—1.54
	25-29	1	0.26	0.01—1.44
	30-34	2	0.37	0.05—1.34
	35+	5	1.41	0.46—3.28
Trisomy 18	<20	0	0.00	0.00—3.53
	20-24	0	0.00	0.00—1.54
	25-29	3	0.77	0.16—2.26
	30-34	3	0.56	0.11—1.63
	35+	4	1.13	0.31—2.88
Trisomy 21 (Down Syndrome)	<20	7	6.70	2.69—13.80
	20-24	6	2.50	0.92—5.44
	25-29	29	7.48	5.01—10.74
	30-34	45	8.36	6.10—11.19
	35+	95	26.73	21.62—32.67
Turner Syndrome	<20	0	0.00	0.00—3.53
	20-24	1	0.42	0.01—2.32
	25-29	2	0.52	0.06—1.86
	30-34	3	0.56	0.11—1.63
	35+	6	1.69	0.62—3.67
Other Chromosomal Syndromes/Other Syndromes	<20	6	5.74	2.11—12.49
	20-24	19	7.92	4.77—12.37
	25-29	25	6.45	4.17—9.52
	30-34	44	8.18	5.94—10.97
	35+	29	8.16	5.46—11.72
Other				
Amniotic Bands	<20	0	0.00	0.00—3.53
	20-24	2	0.83	0.10—3.01
	25-29	0	0.00	0.00—0.95
	30-34	1	0.19	0.00—1.04
	35+	1	0.28	0.01—1.57
Skin Anomalies	<20	1	0.96	0.02—5.33
	20-24	0	0.00	0.00—1.54
	25-29	0	0.00	0.00—0.95
	30-34	5	0.93	0.30—2.17
	35+	5	1.41	0.46—3.28

Table 9 (cont'd) Prevalence of Birth Defects by Maternal Age for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Other, Specified	<20	3	2.87	0.59—8.39
	20-24	2	0.83	0.10—3.01
	25-29	3	0.77	0.16—2.26
	30-34	6	1.11	0.41—2.43
	35+	6	1.69	0.62—3.67

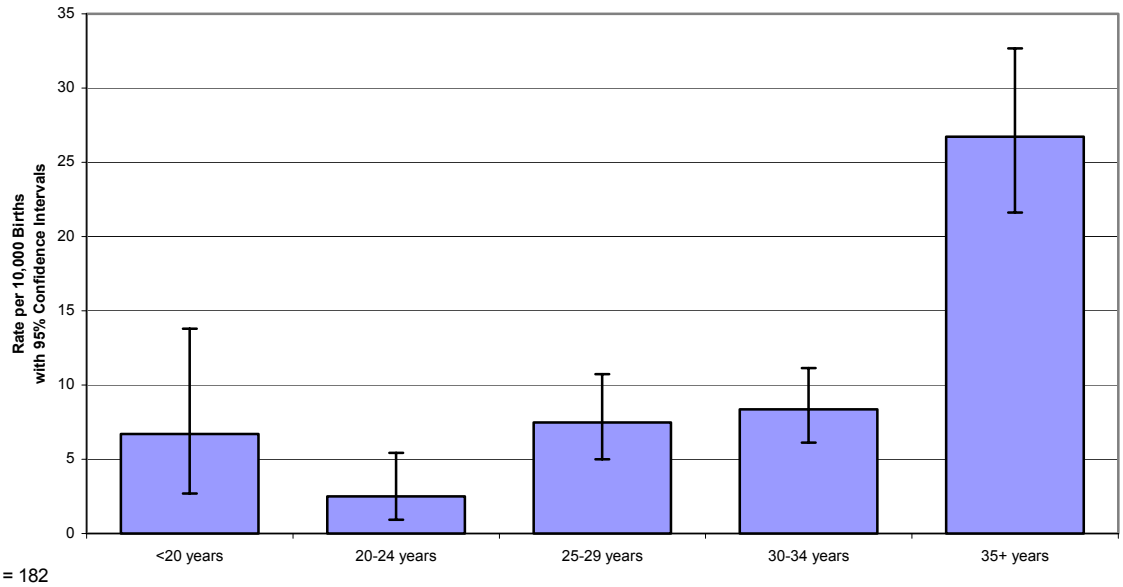
1. Cases can be included in more than one defect category. Cases are counted once within a defect category. Counts represent the number of defects, not the total number of cases with birth defects. Because only live births are presented on this table, case totals are not listed.

Table 10 Selected Common Defects for Live Births, by Maternal Age, Massachusetts: 2000-2001

Age	Defect [1]	Count	Rate per 10,000 Births	95% Confidence Interval
<20				
	Gastroschisis	17	16.26	9.47 — 26.04
	Polydactyly/Syndactyly	16	15.31	8.75 — 24.86
	ASD (Secundum and NOS)	12	11.48	5.93 — 20.06
	Club Foot	10	9.57	4.59 — 17.60
	Hypospadias, 2nd or 3rd Degree	10	9.57	4.59 — 17.60
20-24				
	ASD (Secundum and NOS)	38	15.84	11.21 — 21.74
	Club Foot	30	12.50	8.44 — 17.85
	Polydactyly/Syndactyly	21	8.75	5.42 — 13.38
	Cleft Lip w/ and w/o Cleft Palate	20	8.34	5.09 — 12.87
	Hypospadias, 2nd or 3rd Degree	20	8.34	5.09 — 12.87
25-29				
	ASD (Secundum and NOS)	69	17.79	13.85 — 22.52
	Hypospadias, 2nd or 3rd Degree	41	10.57	7.59 — 14.34
	VSD (Membranous and NOS)	39	10.06	7.15 — 13.75
	Polydactyly/Syndactyly	35	9.03	6.29 — 12.55
	Cleft Lip w/ and w/o Cleft Palate	34	8.77	6.07 — 12.25
	Club Foot	34	8.77	6.07 — 12.25
30-34				
	ASD (Secundum and NOS)	95	17.65	14.28 — 21.58
	Trisomy 21 (Down Syndrome)	45	8.36	6.10 — 11.19
	Hypospadias, 2nd or 3rd Degree	44	8.18	5.94 — 10.97
	Polydactyly/Syndactyly	44	8.18	5.94 — 10.97
	VSD (Membranous and NOS)	43	7.99	5.78 — 10.76
35+				
	Trisomy 21 (Down Syndrome)	95	26.73	21.62 — 32.67
	ASD (Secundum and NOS)	78	21.94	17.34 — 27.39
	VSD (Membranous and NOS)	47	13.22	9.71 — 17.58
	Club Foot	41	11.53	8.28 — 15.65
	Obstructive Genitourinary Defect	31	8.72	5.93 — 12.38
	Cleft Lip w/ and w/o Cleft Palate	31	8.72	5.93 — 12.38

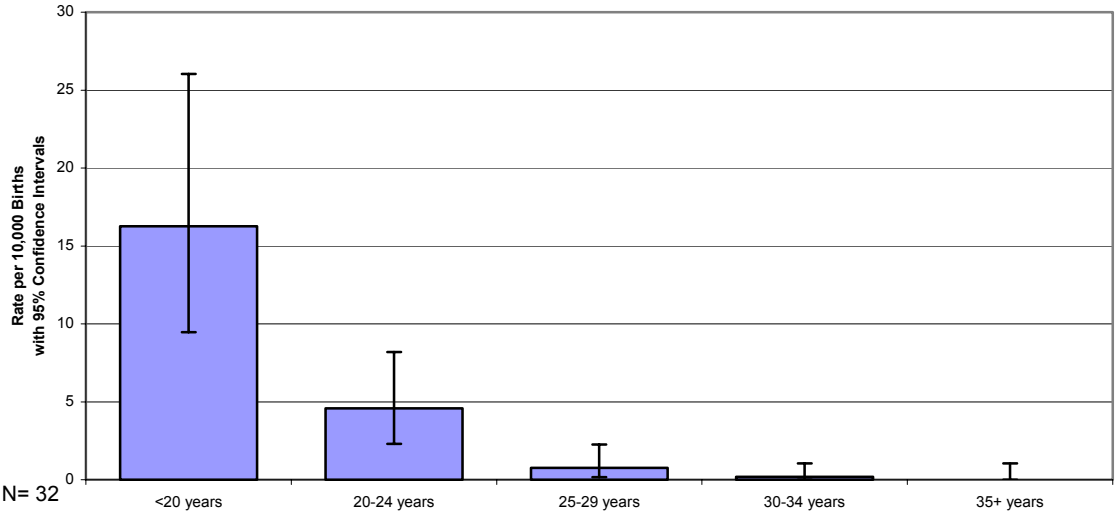
1. Excludes Patent Ductus Arteriosus (PDA) due to the high number of cases and the mild severity of the majority of these cases.

Figure 6 Rates of Down Syndrome among Maternal Age Groups, Massachusetts: 2000-2001



N = 182

Figure 7 Rates of Gastroschisis among Maternal Age Groups, Massachusetts: 2000-2001



N= 32

No Gastroschisis cases reported for the mother's age group 35 years and older

Table 11 Prevalence of Birth Defects by Maternal Race/ Hispanic Ethnicity for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
Central Nervous System				
Anencephaly	White, Non-Hispanic	6	0.50	0.18—1.10
	Black, Non-Hispanic	0	0.00	0.00—3.18
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	0	0.00	0.00—1.98
Encephalocele	White, Non-Hispanic	2	0.17	0.02—0.61
	Black, Non-Hispanic	4	3.44	0.94—8.82
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	0	0.00	0.00—1.98
Holoprosencephaly	White, Non-Hispanic	8	0.67	0.29—1.32
	Black, Non-Hispanic	1	0.86	0.02—4.80
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	0	0.00	0.00—1.98
Hydrocephaly w/o Spina Bifida	White, Non-Hispanic	26	2.18	1.43—3.20
	Black, Non-Hispanic	4	3.44	0.94—8.82
	Asian, Non-Hispanic	3	3.17	0.65—9.28
	Hispanic	10	5.36	2.57—9.86
Microcephaly	White, Non-Hispanic	29	2.43	1.63—3.50
	Black, Non-Hispanic	3	2.58	0.53—7.55
	Asian, Non-Hispanic	2	2.12	0.26—7.64
	Hispanic	7	3.75	1.51—7.73
Spina Bifida w/ and w/o Hydrocephaly	White, Non-Hispanic	19	1.59	0.96—2.49
	Black, Non-Hispanic	3	2.58	0.53—7.55
	Asian, Non-Hispanic	4	4.23	1.15—10.84
	Hispanic	2	1.07	0.13—3.87
Tethered Cord	White, Non-Hispanic	21	1.76	1.09—2.69
	Black, Non-Hispanic	2	1.72	0.21—6.22
	Asian, Non-Hispanic	1	1.06	0.03—5.90
	Hispanic	3	1.61	0.33—4.70
Other CNS	White, Non-Hispanic	66	5.54	4.28—7.05
	Black, Non-Hispanic	6	5.16	1.90—11.24
	Asian, Non-Hispanic	2	2.12	0.26—7.64
	Hispanic	11	5.90	2.94—10.55
Eye				
Aniridia	White, Non-Hispanic	8	0.67	0.29—1.32
	Black, Non-Hispanic	0	0.00	0.00—3.18
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	0	0.00	0.00—1.98

Table 11 (cont'd) Prevalence of Birth Defects by Maternal Race/ Hispanic Ethnicity for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
Anophthalmia/Microphthalmia	White, Non-Hispanic	10	0.84	0.40—1.54
	Black, Non-Hispanic	3	2.58	0.53—7.55
	Asian, Non-Hispanic	2	2.12	0.26—7.64
	Hispanic	2	1.07	0.13—3.87
Congenital Glaucoma, Congenital Cataract	White, Non-Hispanic	15	1.26	0.70—2.08
	Black, Non-Hispanic	5	4.30	1.40—10.04
	Asian, Non-Hispanic	2	2.12	0.26—7.64
	Hispanic	6	3.22	1.18—7.00
Other Eye	White, Non-Hispanic	17	1.43	0.83—2.28
	Black, Non-Hispanic	0	0.00	0.00—3.18
	Asian, Non-Hispanic	1	1.06	0.03—5.90
	Hispanic	3	1.61	0.33—4.70
Ear				
Anotia/Microtia	White, Non-Hispanic	13	1.09	0.58—1.87
	Black, Non-Hispanic	0	0.00	0.00—3.18
	Asian, Non-Hispanic	1	1.06	0.03—5.90
	Hispanic	5	2.68	0.87—6.25
Other Ear	White, Non-Hispanic	5	0.42	0.14—0.98
	Black, Non-Hispanic	1	0.86	0.02—4.80
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	0	0.00	0.00—1.98
Cardiovascular				
Anomalous Pulmonary Venous Connection				
Total/Partial Anomalous Pulmonary Venous	White, Non-Hispanic	17	1.43	0.83—2.28
	Black, Non-Hispanic	2	1.72	0.21—6.22
	Asian, Non-Hispanic	2	2.12	0.26—7.64
	Hispanic	3	1.61	0.33—4.70
Atrioventricular Canal Defects				
ASD Primum	White, Non-Hispanic	10	0.84	0.40—1.54
	Black, Non-Hispanic	3	2.58	0.53—7.55
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	2	1.07	0.13—3.87
Common Atrium	White, Non-Hispanic	3	0.25	0.05—0.74
	Black, Non-Hispanic	0	0.00	0.00—3.18
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	0	0.00	0.00—1.98

Table 11 (cont'd) Prevalence of Birth Defects by Maternal Race/ Hispanic Ethnicity for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
Complete Atrioventricular Canal Defect	White, Non-Hispanic	33	2.77	1.91—3.89
	Black, Non-Hispanic	2	1.72	0.21—6.22
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	3	1.61	0.33—4.70
Endocardial Cushion (OS and NOS)	White, Non-Hispanic	6	0.50	0.18—1.10
	Black, Non-Hispanic	2	1.72	0.21—6.22
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	1	0.54	0.01—2.99
VSD, Canal Type	White, Non-Hispanic	6	0.50	0.18—1.10
	Black, Non-Hispanic	3	2.58	0.53—7.55
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	2	1.07	0.13—3.87
Conotruncal (Outlet) and Aortic Arch				
Double Outlet Right Ventricle	White, Non-Hispanic	13	1.09	0.58—1.87
	Black, Non-Hispanic	2	1.72	0.21—6.22
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	2	1.07	0.13—3.87
d-Transposition of the Great Arteries	White, Non-Hispanic	21	1.76	1.09—2.69
	Black, Non-Hispanic	1	0.86	0.02—4.80
	Asian, Non-Hispanic	2	2.12	0.26—7.64
	Hispanic	0	0.00	0.00—1.98
Interrupted Aortic Arch, Type B	White, Non-Hispanic	4	0.34	0.09—0.86
	Black, Non-Hispanic	0	0.00	0.00—3.18
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	0	0.00	0.00—1.98
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	White, Non-Hispanic	48	4.03	2.97—5.34
	Black, Non-Hispanic	7	6.03	2.42—12.42
	Asian, Non-Hispanic	3	3.17	0.65—9.28
	Hispanic	9	4.82	2.21—9.16
Truncus	White, Non-Hispanic	6	0.50	0.18—1.10
	Black, Non-Hispanic	0	0.00	0.00—3.18
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	0	0.00	0.00—1.98
Ebstein Anomaly				
Ebstein Anomaly	White, Non-Hispanic	4	0.34	0.09—0.86
	Black, Non-Hispanic	0	0.00	0.00—3.18
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	0	0.00	0.00—1.98

Table 11 (cont'd) Prevalence of Birth Defects by Maternal Race/ Hispanic Ethnicity for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
Heterotaxy (Laterality Defects)				
Heterotaxy	White, Non-Hispanic	14	1.17	0.64—1.97
	Black, Non-Hispanic	1	0.86	0.02—4.80
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	1	0.54	0.01—2.99
Left-Sided Obstruction				
Aortic Valve Stenosis	White, Non-Hispanic	12	1.01	0.52—1.76
	Black, Non-Hispanic	1	0.86	0.02—4.80
	Asian, Non-Hispanic	1	1.06	0.03—5.90
	Hispanic	2	1.07	0.13—3.87
Coarctation of Aorta	White, Non-Hispanic	49	4.11	3.04—5.44
	Black, Non-Hispanic	5	4.30	1.40—10.04
	Asian, Non-Hispanic	2	2.12	0.26—7.64
	Hispanic	7	3.75	1.51—7.73
Hypoplastic Left Heart Syndrome	White, Non-Hispanic	20	1.68	1.03—2.59
	Black, Non-Hispanic	0	0.00	0.00—3.18
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	2	1.07	0.13—3.87
Interrupted Aortic Arch (Type A and NOS)	White, Non-Hispanic	1	0.08	0.00—0.47
	Black, Non-Hispanic	1	0.86	0.02—4.80
	Asian, Non-Hispanic	1	1.06	0.03—5.90
	Hispanic	0	0.00	0.00—1.98
Patent Ductus Arteriosus				
Patent Ductus Arteriosus	White, Non-Hispanic	188	15.78	13.60—18.20
	Black, Non-Hispanic	35	30.13	20.99—41.90
	Asian, Non-Hispanic	13	13.76	7.32—23.52
	Hispanic	22	11.79	7.39—17.85
Right-Sided Obstruction				
Pulmonary Stenosis, Valvular	White, Non-Hispanic	53	4.45	3.33—5.82
	Black, Non-Hispanic	9	7.75	3.54—14.71
	Asian, Non-Hispanic	1	1.06	0.03—5.90
	Hispanic	2	1.07	0.13—3.87
Pulmonary Valve Atresia w/intact septum	White, Non-Hispanic	7	0.59	0.24—1.21
	Black, Non-Hispanic	1	0.86	0.02—4.80
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	2	1.07	0.13—3.87
Pulmonary Valve Atresia with VSD	White, Non-Hispanic	4	0.34	0.09—0.86
	Black, Non-Hispanic	0	0.00	0.00—3.18
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	1	0.54	0.01—2.99

Table 11 (cont'd) Prevalence of Birth Defects by Maternal Race/ Hispanic Ethnicity for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
Tricuspid Valve Atresia	White, Non-Hispanic	5	0.42	0.14—0.98
	Black, Non-Hispanic	1	0.86	0.02—4.80
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	3	1.61	0.33—4.70
Septal Defects				
ASD (Secundum and NOS)	White, Non-Hispanic	206	17.29	15.01—19.82
	Black, Non-Hispanic	29	24.96	16.72—35.85
	Asian, Non-Hispanic	16	16.93	9.68—27.49
	Hispanic	28	15.01	9.97—21.69
VSD (Membranous and NOS)	White, Non-Hispanic	107	8.98	7.36—10.85
	Black, Non-Hispanic	14	12.05	6.59—20.22
	Asian, Non-Hispanic	8	8.46	3.65—16.68
	Hispanic	19	10.18	6.13—15.90
VSD, Conoventricular/Malalignment	White, Non-Hispanic	19	1.59	0.96—2.49
	Black, Non-Hispanic	2	1.72	0.21—6.22
	Asian, Non-Hispanic	3	3.17	0.65—9.28
	Hispanic	3	1.61	0.33—4.70
Single Ventricle and L-TGA				
L-TGA	White, Non-Hispanic	6	0.50	0.18—1.10
	Black, Non-Hispanic	0	0.00	0.00—3.18
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	0	0.00	0.00—1.98
Single Ventricle	White, Non-Hispanic	7	0.59	0.24—1.21
	Black, Non-Hispanic	0	0.00	0.00—3.18
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	0	0.00	0.00—1.98
Other Cardiovascular				
Other Cardiovascular	White, Non-Hispanic	151	12.67	10.73—14.86
	Black, Non-Hispanic	21	18.08	11.19—27.63
	Asian, Non-Hispanic	6	6.35	2.33—13.82
	Hispanic	13	6.97	3.71—11.92
Respiratory				
Choanal Atresia	White, Non-Hispanic	7	0.59	0.24—1.21
	Black, Non-Hispanic	0	0.00	0.00—3.18
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	1	0.54	0.01—2.99

Table 11 (cont'd) Prevalence of Birth Defects by Maternal Race/ Hispanic Ethnicity for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
Lung Anomalies	White, Non-Hispanic	20	1.68	1.03—2.59
	Black, Non-Hispanic	2	1.72	0.21—6.22
	Asian, Non-Hispanic	1	1.06	0.03—5.90
	Hispanic	5	2.68	0.87—6.25
Other Respiratory	White, Non-Hispanic	5	0.42	0.14—0.98
	Black, Non-Hispanic	0	0.00	0.00—3.18
	Asian, Non-Hispanic	2	2.12	0.26—7.64
	Hispanic	2	1.07	0.13—3.87
Orofacial				
Cleft Lip w/ and w/o Cleft Palate	White, Non-Hispanic	90	7.55	6.07—9.28
	Black, Non-Hispanic	4	3.44	0.94—8.82
	Asian, Non-Hispanic	13	13.76	7.32—23.52
	Hispanic	13	6.97	3.71—11.92
Cleft Palate w/o Cleft Lip	White, Non-Hispanic	73	6.13	4.80—7.70
	Black, Non-Hispanic	2	1.72	0.21—6.22
	Asian, Non-Hispanic	7	7.41	2.98—15.26
	Hispanic	13	6.97	3.71—11.92
Pierre Robin Sequence	White, Non-Hispanic	18	1.51	0.90—2.39
	Black, Non-Hispanic	0	0.00	0.00—3.18
	Asian, Non-Hispanic	1	1.06	0.03—5.90
	Hispanic	2	1.07	0.13—3.87
Other Orofacial	White, Non-Hispanic	13	1.09	0.58—1.87
	Black, Non-Hispanic	4	3.44	0.94—8.82
	Asian, Non-Hispanic	1	1.06	0.03—5.90
	Hispanic	4	2.14	0.58—5.49
Gastrointestinal				
Biliary Atresia	White, Non-Hispanic	4	0.34	0.09—0.86
	Black, Non-Hispanic	1	0.86	0.02—4.80
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	0	0.00	0.00—1.98
Esophageal Atresia/Tracheoesophageal Fistula	White, Non-Hispanic	25	2.10	1.36—3.10
	Black, Non-Hispanic	2	1.72	0.21—6.22
	Asian, Non-Hispanic	2	2.12	0.26—7.64
	Hispanic	5	2.68	0.87—6.25
Hirschsprung Disease	White, Non-Hispanic	22	1.85	1.16—2.80
	Black, Non-Hispanic	1	0.86	0.02—4.80
	Asian, Non-Hispanic	2	2.12	0.26—7.64
	Hispanic	3	1.61	0.33—4.70

Table 11 (cont'd) Prevalence of Birth Defects by Maternal Race/ Hispanic Ethnicity for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
Rectal and Large Intestinal Atresia/Stenosis	White, Non-Hispanic	46	3.86	2.83—5.15
	Black, Non-Hispanic	4	3.44	0.94—8.82
	Asian, Non-Hispanic	2	2.12	0.26—7.64
	Hispanic	9	4.82	2.21—9.16
Small Intestinal Atresia	White, Non-Hispanic	23	1.93	1.22—2.90
	Black, Non-Hispanic	3	2.58	0.53—7.55
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	6	3.22	1.18—7.00
Other Gastrointestinal	White, Non-Hispanic	64	5.37	4.14—6.86
	Black, Non-Hispanic	4	3.44	0.94—8.82
	Asian, Non-Hispanic	2	2.12	0.26—7.64
	Hispanic	11	5.90	2.94—10.55
Genitourinary				
Bladder Exstrophy	White, Non-Hispanic	5	0.42	0.14—0.98
	Black, Non-Hispanic	1	0.86	0.02—4.80
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	0	0.00	0.00—1.98
Cloacal Exstrophy	White, Non-Hispanic	0	0.00	0.00—0.31
	Black, Non-Hispanic	0	0.00	0.00—3.18
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	1	0.54	0.01—2.99
Hypospadias, 2nd or 3rd Degree	White, Non-Hispanic	104	8.73	7.13—10.57
	Black, Non-Hispanic	11	9.47	4.73—16.94
	Asian, Non-Hispanic	6	6.35	2.33—13.82
	Hispanic	14	7.50	4.10—12.59
Obstructive Genitourinary Defect	White, Non-Hispanic	96	8.06	6.53—9.84
	Black, Non-Hispanic	8	6.89	2.97—13.57
	Asian, Non-Hispanic	4	4.23	1.15—10.84
	Hispanic	17	9.11	5.31—14.59
Renal Agenesis/Hypoplasia	White, Non-Hispanic	1	0.08	0.00—0.47
	Black, Non-Hispanic	1	0.86	0.02—4.80
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	0	0.00	0.00—1.98
Other Genitourinary	White, Non-Hispanic	87	7.30	5.85—9.01
	Black, Non-Hispanic	11	9.47	4.73—16.94
	Asian, Non-Hispanic	2	2.12	0.26—7.64
	Hispanic	8	4.29	1.85—8.45

Table 11 (cont'd) Prevalence of Birth Defects by Maternal Race/ Hispanic Ethnicity for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
Musculoskeletal				
Club Foot	White, Non-Hispanic	121	10.15	8.43—12.13
	Black, Non-Hispanic	10	8.61	4.13—15.83
	Asian, Non-Hispanic	3	3.17	0.65—9.28
	Hispanic	20	10.72	6.55—16.56
Craniosynostosis	White, Non-Hispanic	59	4.95	3.77—6.39
	Black, Non-Hispanic	4	3.44	0.94—8.82
	Asian, Non-Hispanic	2	2.12	0.26—7.64
	Hispanic	6	3.22	1.18—7.00
Diaphragmatic Hernia	White, Non-Hispanic	26	2.18	1.43—3.20
	Black, Non-Hispanic	1	0.86	0.02—4.80
	Asian, Non-Hispanic	1	1.06	0.03—5.90
	Hispanic	1	0.54	0.01—2.99
Gastroschisis	White, Non-Hispanic	26	2.18	1.43—3.20
	Black, Non-Hispanic	0	0.00	0.00—3.18
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	5	2.68	0.87—6.25
Omphalocele	White, Non-Hispanic	14	1.17	0.64—1.97
	Black, Non-Hispanic	3	2.58	0.53—7.55
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	4	2.14	0.58—5.49
Polydactyly/Syndactyly	White, Non-Hispanic	86	7.22	5.77—8.91
	Black, Non-Hispanic	14	12.05	6.59—20.22
	Asian, Non-Hispanic	11	11.64	5.81—20.83
	Hispanic	25	13.40	8.67—19.78
Reduction Deformity, Lower Limbs	White, Non-Hispanic	19	1.59	0.96—2.49
	Black, Non-Hispanic	1	0.86	0.02—4.80
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	1	0.54	0.01—2.99
Reduction Deformity, Upper Limbs	White, Non-Hispanic	29	2.43	1.63—3.50
	Black, Non-Hispanic	3	2.58	0.53—7.55
	Asian, Non-Hispanic	1	1.06	0.03—5.90
	Hispanic	8	4.29	1.85—8.45
Skeletal Dysplasia	White, Non-Hispanic	9	0.76	0.35—1.43
	Black, Non-Hispanic	1	0.86	0.02—4.80
	Asian, Non-Hispanic	2	2.12	0.26—7.64
	Hispanic	4	2.14	0.58—5.49
Other Musculoskeletal	White, Non-Hispanic	65	5.45	4.21—6.95
	Black, Non-Hispanic	15	12.91	7.23—21.30
	Asian, Non-Hispanic	3	3.17	0.65—9.28
	Hispanic	20	10.72	6.55—16.56

Table 11 (cont'd) Prevalence of Birth Defects by Maternal Race/ Hispanic Ethnicity for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
Chromosomal and Other Syndromes				
Klinefelter Syndrome	White, Non-Hispanic	6	0.50	0.18—1.10
	Black, Non-Hispanic	1	0.86	0.02—4.80
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	0	0.00	0.00—1.98
Trisomy 13	White, Non-Hispanic	7	0.59	0.24—1.21
	Black, Non-Hispanic	1	0.86	0.02—4.80
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	0	0.00	0.00—1.98
Trisomy 18	White, Non-Hispanic	5	0.42	0.14—0.98
	Black, Non-Hispanic	3	2.58	0.53—7.55
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	1	0.54	0.01—2.99
Trisomy 21 (Down Syndrome)	White, Non-Hispanic	132	11.08	9.27—13.14
	Black, Non-Hispanic	14	12.05	6.59—20.22
	Asian, Non-Hispanic	2	2.12	0.26—7.64
	Hispanic	28	15.01	9.97—21.69
Turner Syndrome	White, Non-Hispanic	9	0.76	0.35—1.43
	Black, Non-Hispanic	1	0.86	0.02—4.80
	Asian, Non-Hispanic	2	2.12	0.26—7.64
	Hispanic	0	0.00	0.00—1.98
Other Chromosomal Syndromes/Other Syndromes	White, Non-Hispanic	88	7.38	5.92—9.10
	Black, Non-Hispanic	9	7.75	3.54—14.71
	Asian, Non-Hispanic	6	6.35	2.33—13.82
	Hispanic	16	8.58	4.90—13.93
Other				
Amniotic Bands	White, Non-Hispanic	1	0.08	0.00—0.47
	Black, Non-Hispanic	2	1.72	0.21—6.22
	Asian, Non-Hispanic	0	0.00	0.00—3.90
	Hispanic	1	0.54	0.01—2.99
Skin Anomalies	White, Non-Hispanic	8	0.67	0.29—1.32
	Black, Non-Hispanic	1	0.86	0.02—4.80
	Asian, Non-Hispanic	1	1.06	0.03—5.90
	Hispanic	1	0.54	0.01—2.99
Other, Specified	White, Non-Hispanic	16	1.34	0.77—2.18
	Black, Non-Hispanic	0	0.00	0.00—3.18
	Asian, Non-Hispanic	1	1.06	0.03—5.90
	Hispanic	3	1.61	0.33—4.70

Table 11 (cont'd) Prevalence of Birth Defects by Maternal Race/ Hispanic Ethnicity for Live Births, Massachusetts: 2000-2001

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval

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.

2. Due to small numbers races classified as "other" are excluded.

Table 12 Selected Common Defects by Maternal Race/Hispanic Ethnicity, among Live Births Massachusetts: 2000-2001

Race[1]	Defect[2]	Case	Rate per 10,000 Births	95% Confidence Interval
White, Non-Hispanic				
	ASD (Secundum and NOS)	206	17.29	15.01—19.82
	Trisomy 21 (Down Syndrome)	132	11.08	9.27—13.14
	Club Foot	121	10.15	8.43—12.13
	VSD (Membranous and NOS)	107	8.98	7.36—10.85
	Hypospadias, 2nd or 3rd Degree	104	8.73	7.13—10.57
	Obstructive Genitourinary Defect	96	8.06	6.53—9.84
	Cleft Lip w/ and w/o Cleft Palate	90	7.55	6.07—9.28
	Polydactyly/Syndactyly	86	7.22	5.77—8.91
	Cleft Palate w/o Cleft Lip	73	6.13	4.80—7.70
	Craniosynostosis	59	4.95	3.77—6.39
Black, Non-Hispanic				
	ASD (Secundum and NOS)	29	24.96	16.72—35.85
	Polydactyly/Syndactyly	14	12.05	6.59—20.22
	VSD (Membranous and NOS)	14	12.05	6.59—20.22
	Trisomy 21 (Down Syndrome)	14	12.05	6.59—20.22
	Hypospadias, 2nd or 3rd Degree	11	9.47	4.73—16.94
	Club Foot	10	8.61	4.13—15.83
	Pulmonary Stenosis, Valvular	9	7.75	3.54—14.71
	Obstructive Genitourinary Defect	8	6.89	2.97—13.57
	Tetralogy of Fallot w/ and w/o Pulmonary Atresia	7	6.03	2.42—12.42
	Congenital Glaucoma, Congenital Cataract	5	4.30	1.40—10.04
	Coarctation of Aorta	5	4.30	1.40—10.04
Asian, Non-Hispanic				
	ASD (Secundum and NOS)	16	16.93	9.68—27.49
	Cleft Lip w/ and w/o Cleft Palate	13	13.76	7.32—23.52
	Polydactyly/Syndactyly	11	11.64	5.81—20.83
	VSD (Membranous and NOS)	8	8.46	3.65—16.68
	Cleft Palate w/o Cleft Lip	7	7.41	2.98—15.26
	Hypospadias, 2nd or 3rd Degree	6	6.35	2.33—13.82
Hispanic				
	ASD (Secundum and NOS)	28	15.01	9.97—21.69
	Trisomy 21 (Down Syndrome)	28	15.01	9.97—21.69
	Polydactyly/Syndactyly	25	13.40	8.67—19.78
	Club Foot	20	10.72	6.55—16.56
	VSD (Membranous and NOS)	19	10.18	6.13—15.90
	Obstructive Genitourinary Defect	17	9.11	5.31—14.59
	Hypospadias, 2nd or 3rd Degree	14	7.50	4.10—12.59

Table 12 (cont'd) Selected Common Defects by Maternal Race/Hispanic Ethnicity, among Live Births Massachusetts: 2000-2001

Race[1]	Defect[2]	Case	Rate per 10,000 Births	95% Confidence Interval
	Cleft Lip w/ and w/o Cleft Palate	13	6.97	3.71–11.92
	Cleft Palate w/o Cleft Lip	13	6.97	3.71–11.92
	Hydrocephaly w/o Spina Bifida	10	5.36	2.57–9.86

1. Due to small numbers races classified as "other" are excluded.

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. 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	2.6%
Serious, may be correctable, most have long-term needs	18.7%
Moderate, most correctable, many have long-term needs	71.1%
Mild, may be correctable, minimal long-term needs	7.6%

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 19% 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 71% 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 some degree of variation. For example, children with Microtia (small ears) may have needed corrective surgery, hearing evaluations and family support services, whereas those with isolated Dextrocardia (heart in the right side of the chest instead of the left) and no associated heart defect have no clinical consequence.

Table 13 Birth Defects by Severity
Massachusetts: 2000-2001

Severe, Supportive Measures, Moderate, Most Usually Incompatible With Life	
Anencephaly	7
Bilateral renal agenesis	4
Trisomy 13	11
Trisomy 18	16
Severe syndrome or defect, not listed	21
TOTAL	59

Serious, May be Correctable, Most Have Long Term Needs	
Achondroplasia	9
Amniotic band complex	5
Aniridia	7
Anophthalmia	6
Anotia	0
Biliary atresia	5
Bladder exstrophy	6
Cloacal exstrophy	1
Congenital heart defect (CHD), multiple, moderate to severe, not listed elsewhere	10
Double outlet right ventricle	10
Encephalocele	5
Heterotaxy with CHD	10
Holoprosencephaly	9
Hypoplastic left heart syndrome	22
Limb reductions, moderate-severe	15
Osteogenesis imperfecta	2
Sacral agenesis, caudal regression	2
Single ventricle	3
Spina bifida	29
Trisomy 21	192
Serious syndrome or defect, not listed above	79
TOTAL	427

Moderate, Most Correctable, May Have Long Term Needs	
Aortic valve stenosis	7
Atrial septal defect	99
Atrioventricular canal (endocardinal cushion defect)	9
Choanal atresia	6
Cleft lip with/without cleft palate, cleft palate	192
Coarctation	38
Cataract, glaucoma	24
Clubfoot	116
Coloboma	4
Congenital heart defect, multiple, mild-moderate, not listed elsewhere	28
Craniosynostosis	64
Dandy-Walker malformation	2
Diaphragmatic hernia	24
Esophageal atresia/ tracheosophageal fistula	31
Ebstein anomaly	5
Gastroschisis	33
Genitourinary, obstructive	43
Hirschsprung disease	21
Holt-Oram syndrome	1
Hydrocephalus	28
Hypospadias, 2nd or 3rd degree	133
Intestinal atresia: duodenal, jejunal, Ileal	21
Imperforate anus/rectal atresia	47
Interrupted aortic arch	3
Klinefelter syndrome	7
Limb reductions, mild-moderate	34
Malrotation, intestinal	24
Microcephaly	22
Noonan syndrome	5
Omphalocele	15
Partial anomalous pulmonary venous return	2
Pulmonary atresia/stenosis	59
Tethered cord	11
Tetralogy of Fallot, TOF with pulmonary atresia	50
Total anomalous pulmonary venous return	8
Transposition great arteries	21
Trucuspid atresia/stenosis	6
Truncus	2
Turner syndrome	15
Ventricular septal defect	93
Moderate syndrome or defect, not listed above	272
TOTAL	1625

Mild, May be Correctable, Minimal Long Term Needs	
Bicuspid aortic valve	8
Meckel's diverticulum	5
Microphthalmia	0
Microtia	14
Patent ductus arteriosus	15
Poly/Syndactyly (except tags)	102
Situs inversus totalis without CHD, Situs inversus abdominis, isolated Dextrocardia	5
Mild defect, not listed above	24
TOTAL	173

N= 2,284

Appendices

Technical Notes

Definitions

2000-2001 Denominators Used in Calculating Rates

Birth Defects Codes and Exclusions by Defect Category

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

Glossary

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 at a specific time divided by the number of individuals at risk. 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 for 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 2001 Dollars

2001 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 1.1336 representing the inflation from 1994 to 2001. 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://www.jsc.nasa.gov/bu2/inflateGDP.html

Definitions

These definitions are derived from the Massachusetts Department of Public Health report titled Massachusetts Births, 2001.

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 reportable 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.

2000 and 2001 Populations Used in Calculating Rates

		Numbers of Live Births to MA Residents		
		2000	2001	Total
Overall		81,582	81,014	162,596
By Maternal Age	<20	5,395	5,057	10,452
	20-24	11,965	12,029	23,994
	25-29	19,761	19,015	38,776
	30-34	26,873	26,948	53,821
	35+	17,584	17,963	35,547
By Infant's Sex	Male	41,744	41,152	82,896
	Female	39,838	39,861	79,699
By Plurality	Singleton	78,075	77,409	155,484
	Multiple Birth	3,507	3,605	7,112
By Maternal Race/Ethnicity	White	60,051	59,115	119,166
	Black	5,755	5,862	11,617
	Hispanic	9,247	9,410	18,657
	Asian	4,667	4,784	9,451

Birth Defect Codes and Exclusions[1] Massachusetts 2000-2001		
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; hydrocephalus 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.
Tethered 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] Massachusetts 2000-2001 (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	
Heterotaxy (Laterality Defects)		
Heterotaxy	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	
Single Ventricle and L-TGA		
L-TGA	745.120	
Single Ventricle	745.300-745.380	

Birth Defect Codes and Exclusions[1] Massachusetts 2000-2001 (cont'd)		
Defect	**ICD-9 / BPA[2]**	**NOTES**
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	Exclude pulmonary/tricuspid/aortic valve insufficiency/regurgitation, mitral valve congenital insufficiency. Exclude peripheral pulmonary artery stenosis with physiologic PPS (ie <36 wks).
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, 748.310-748.385, 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, 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, 753.160, 753.310-753.480, 753.700-753.880	Exclude isolated undescended testicle(s), unspecified genitourinary anomalies.

Birth Defect Codes and Exclusions[1] Massachusetts 2000-2001 (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	
Polydactlyly/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	756.430-756.500	
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		
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, 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	225.200, 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 murmers 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.

Selected ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2000-2001

Central Nervous System

BPA Label	BPA Code	# of Defects
Agyria and lissencephaly	742240	2
Anencephaly	740020	7
Arrhinencephaly	742270	2
Brain cysts: Cerebral / subependymal / periventricular	742420	10
Brain cysts: Porencephaly / porencephalic	742410	3
Brain, reduction defect, OS	742280	12
Brain: Other specified anomalies / cortical atrophy / cranial nerve defects	742480	8
Cerebellum anomalies	742230	3
Cerebrum anomalies	742200	1
Corpus callosum anomalies	742210	39
Dandy-Walker Malformation	742310	6
Encephalocele, Frontal (including proencephalon)	742085	1
Encephalocele, Occipital	742000	3
Encephalocele, OS (including midline)	742080	1
Encephalocele, Parietal	742086	2
Enlarged brain and head / enlarged head / enlarged brain / megalencephaly	742400	19
Holoprosencephaly, Alobar	742265	1
Holoprosencephaly, Lobar	742267	3
Holoprosencephaly, NOS	742260	1
Holoprosencephaly, Semilobar	742266	4
Hydranencephaly	742320	1
Hydrocephaly, NOS	742390	27
Hydrocephaly, Anomalies of Aqueduct of Sylvius	742300	8
Hydrocephaly, OS	742380	10
Lipomeningocele, Highest level, lumbar, No mentioned hydrocephalus, unspecified open/closed	741953	1
Lipomeningomyelocele, Highest level, lumbar, No mentioned hydrocephalus, closed	741843	2
Lipomeningomyelocele, Highest level, lumbar, No mentioned hydrocephalus, unspecified open/closed	741943	1
Meningocele, Highest level, lumbar, Hydrocephalus, unspecified open/closed	741513	1
Meningocele, Highest level, sacral, No mentioned hydrocephalus, closed lesion	741814	3
Meningocele, Highest level, sacral, No mentioned hydrocephalus, unspecified open/closed	741914	1
Meningomyelocele/myelomeningocele, Highest level unspecified, Arnold Chiari ± hydrocephalus, open	741009	1
Meningomyelocele/myelomeningocele, Highest level, lumbar, Arnold Chiari ± hydrocephalus, closed	741103	1
Meningomyelocele/myelomeningocele, Highest level, lumbar, Arnold Chiari ± hydrocephalus, open	741003	7
Meningomyelocele/myelomeningocele, Highest level, lumbar, Hydrocephalus, open	741303	1
Meningomyelocele/myelomeningocele, Highest level, lumbar, Hydrocephalus, unspecified open/closed	741503	1
Meningomyelocele/myelomeningocele, Highest level, lumbar, No mentioned hydrocephalus, closed	741803	1
Meningomyelocele/myelomeningocele, Highest level, sacral, Arnold Chiari ± hydrocephalus, open	741004	1
Meningomyelocele/myelomeningocele, Highest level, sacral, No mentioned hydrocephalus, closed	741804	1
Meningomyelocele/myelomeningocele, Highest level, thoracic, Arnold Chiari ± hydrocephalus, closed	741102	1
Meningomyelocele/myelomeningocele, Highest level, thoracic, Arnold Chiari ± hydrocephalus, unspecified open/closed	741202	1
Meningomyelocele/myelomeningocele, Highest level, thoracic, Hydrocephalus, open	741302	1
Microcephaly	742100	42
Microgyria / polymicrogyria	742250	3
Myelocystocele, Highest level, lumbar, No mentioned hydrocephalus, closed	741833	2
Other specified spina bifida, Highest level unspecified, No mentioned hydrocephalus, unspecified open/closed	741989	1
Spinal cord: Other specified anomalies (Includes tethered cord)	742580	28
Unspecified spina bifida, Highest level, sacral, Arnold Chiari malformation ± hydrocephalus, open	741094	1

Selected ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2000-2001 (cont'd)

BPA Label	BPA Code	# of Defects
Eye		
Absence of iris / Aniridia, Bilateral	743424	7
Absence of iris / Aniridia, Right	743422	1
Anophthalmos, Bilateral	743004	1
Anophthalmos, Left	743001	2
Anophthalmos, Right	743002	1
Buphthalmos / Congenital Glaucoma, Bilateral	743204	4
Buphthalmos / Congenital Glaucoma, Laterality Unknown	743200	2
Buphthalmos / Congenital Glaucoma, Right	743202	1
Cataract, anterior polar, Left	743351	2
Cataract, anterior polar, Right	743352	1
Cataract, NOS, Left	743321	4
Cataract, NOS, Bilateral	743324	8
Cataract, NOS, Right	743322	5
Cataract, OS, Left	743361	1
Cataract, OS, Bilateral	743364	2
Coloboma of lens, Laterality Unknown	743340	1
Microphthalmos, Bilateral	743104	5
Microphthalmos, Left	743101	4
Microphthalmos, Right	743102	5
Peters Anomaly, Left	743461	1
Anterior segment: OS colobomas and anomalies	743480	1
Anterior segment: Unspecified colobomas and anomalies	743490	1
Choroid: Coloboma	743535	1
Eyelids: Coloboma	743636	1
Iris: Coloboma	743430	4
Optic disc: Specified anomalies / hypoplastic optic nerve / coloboma of the optic disc	743520	10
Retina: Specified anomalies / congenital retinal aneurysm	743510	2
Vitreous humor: Specified anomalies (includes PHPV)	743500	2
Ear		
Microtia, Bilateral	744214	1
Microtia, Left	744211	4
Microtia, Right	744212	15
Absence or stricture of auditory canal	744000	6

Selected ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2000-2001 (cont'd)

BPA Label	BPA Code	# of Defects
Cardiovascular		
Anomalous Pulmonary Venous		
Partial anomalous pulmonary venous return/connection/drainage	747430	9
Total anomalous pulmonary venous return/connection/drainage	747420	15
Atrioventricular Canal Defects		
Atrial septal defect, primum type (ASD1)	745600	16
Common Atrium	745610	4
Complete atrioventricular canal (CAVC)	745630	32
Complete atrioventricular canal (CAVC) with ventricular septal defect	745620	13
Endocardial cushion defect, NOS	745690	1
Endocardial cushion defect, OS	745680	8
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	11
Dextro-transposition of great arteries (dTGA, dTGV) w/ ventricular septal defect	745110	14
Double-outlet right ventricle (DORV) with normally related great arteries	745185	4
Double-outlet right ventricle (DORV) with transposed great arteries	745186	8
Double-outlet right ventricle (DORV), NOS	745189	3
Double-outlet right ventricle (DORV), OS	745188	4
Interrupted aortic arch, type B	747217	4
Pulmonary atresia with VSD (tetralogy of Fallot with pulmonary atresia)	747310	13
Tetralogy of Fallot	745200	59
Truncus Arteriosus	745000	6
Ebstein Anomaly		
Ebstein Malformation or Anomaly	746200	6
Heterotaxy (Laterality Defects)		
Complete situs inversus w/ dextrocardia	759300	6
Situs ambiguus, left; left isomerism	759360	3
Situs ambiguus, sidedness NOS	759380	3
Situs ambiguus, sidedness unclear	759370	2
Situs inversus w/ levocardia	759310	3
Left-Sided Obstruction		
Aortic stenosis, valvar	746300	17
Coarctation of the aorta (COA), postductal (distal)	747110	4
Coarctation of the aorta (COA), preductal (proximal)	747100	2
Coarctation of the aorta, juxtaductal	747120	3
Coarctation of the aorta, NOS	747190	58
Hypoplastic left heart syndrome	746700	23
Interrupted aortic arch, type A	747216	3
Patent Ductus Arteriosus		
Patent ductus arteriosus (PDA).	747000	266
Right-Sided Obstruction		
Pulmonary valve atresia with VSD (not TOF variant 747310)	746030	5
Pulmonary valve atresia/intact ventricular septum	746000	12
Pulmonic stenosis, valvar	746010	67
Tricuspid atresia	746100	11

Selected ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2000-2001 (cont'd)

BPA Label	BPA Code	# of Defects
Septal Defects		
Atrial septal defect, NOS	745599	79
Atrial septal defect, Secundum type (ASD2)	745510	219
Ventricular septal defect, NOS	745490	17
Ventricular septal defect, Malalignment-type (type I, subarterial) (VSDmal)	745487	28
Ventricular septal defect, Perimembranous (type II, membranous) (VSDmem)	745485	138
Single Ventricle and L-TGA		
L-TGA /Corrected transposition of great vessels / ventricular inversion.	745120	6
Single ventricle, Double Inlet Left Ventricle	745310	5
Single ventricle, OS (e.g., absent right or left AV connection)	745380	2
Other Cardiovascular		
"Pulmonic" or pulmonary atresia, stenosis, or hypoplasia, NOS w/ no mention of whether valve or artery	746995	2
Anomalies of coronary artery or sinus	746885	4
Aorta: Hypoplasia	747210	19
Aorta: Congenital aneurysm / dilatation	747270	3
Aorta: Persistent right aortic arch	747230	27
Aorta: Vascular ring / double aortic arch / vascular ring compression of trachea	747250	7
Aortic septal defect / aortopulmonary window	745010	1
Aortic valve: bicuspid BAV	746400	50
Aortic valve: Other specified anomalies / aortic valve atresia	746480	15
Cerebral vessels: Other anomalies / vein of Galen	747810	5
Great veins: Other specified anomalies (includes IVC interruption, bilateral SVC)	747480	11
Heart: Other specified anomalies / ectopia cordis / mesocardia / conduction defects, NOS	746880	36
Hypoplastic left ventricle. Excludes: hypoplastic left heart syndrome (746700)	746881	5
Hypoplastic right heart or right ventricle / Uhl's disease (parchment RV)	746882	1
Mitral valve: Absence, atresia, or hypoplasia	746505	15
Mitral valve: Congenital mitral stenosis	746500	6
Mitral valve: insufficiency or regurgitation, congenital	746600	9
Persistent left superior vena cava	747410	11
Pulmonary infundibular (subvalvular) stenosis	746830	9
Pulmonary valve: Other specified anomalies. Excludes: infundibular PS (746830)	746080	9
Peripheral arteries: Other anomalies / aberrant subclavian artery	747640	16
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	4
Pulmonary artery: stenosis. Use 746995 if artery or valve is not specified	747320	12
Situs: Dextrocardia without situs inversus / dextrocardia with situs solitus.	746800	5
Supra-aortic stenosis / supravalvular aortic stenosis. Excludes: aortic stenosis, congenital (746300)	747220	3
Respiratory		
Choanal atresia, Bilateral	748014	2
Choanal atresia, Left	748011	4
Choanal atresia, Right	748012	2
Choanal stenosis	748000	3
Congenital subglottic stenosis	748310	3
Larynx: Cleft / laryngotracheoesophageal cleft	748385	1
Other anomalies of trachea	748330	1
Hypoplasia of lung or pulmonary hypoplasia	748510	6
Agenesis or aplasia of lung	748500	2
Lung cysts: CCAM (congenital cystic adenomatoid malformation), OS	748480	8
Lung cysts: Single	748400	1
Other and unspecified anomalies of lung	748690	1
Other specified dysplasia of lung / fusion of lobes of lung	748580	4
Other specified respiratory system anomalies / congenital lobar emphysema / lymphangiectasia of lung	748880	4
Sequestration of lung	748520	6

Selected ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2000-2001 (cont'd)

BPA Label	BPA Code	# of Defects
Orofacial		
Cleft hard palate, Bilateral	749010	7
Cleft hard palate, Central	749020	7
Cleft hard palate, NOS	749030	22
Cleft hard palate, Unilateral, Left	749001	1
Cleft lip and palate, Bilateral cleft lip	749210	25
Cleft lip and palate, NOS	749290	5
Cleft lip and palate, Unilateral cleft lip, Left	749201	30
Cleft lip and palate, Unilateral cleft lip, Right	749202	17
Cleft lip, Bilateral	749110	3
Cleft lip, Central	749120	1
Cleft lip, NOS	749195	5
Cleft lip, Unilateral, Left	749101	27
Cleft lip, Unilateral, Right	749102	10
Cleft palate, NOS	749090	16
Cleft soft palate, Bilateral	749050	3
Cleft soft palate, Central	749060	8
Cleft soft palate, NOS	749070	34
Cleft soft palate, Unilateral, Right	749042	1
Cleft: Incomplete CL/ microform /pseudo / fused lip /healed lip	749190	4
Pierre Robin sequence	524080	22
Branchial cleft, sinus, fistula, cyst, or pit	744400	17
Other branchial cleft anomalies / dermal sinus of head	744480	1
Tongue: Dislocation or displacement / glossoptosis	750130	2
Tongue: large / macroglossia	750120	2
Gastrointestinal		
Meckel's diverticulum	751010	8
Anal atresia with fistula	751230	29
Anal atresia without mention of fistula	751240	27
Annular pancreas	751720	3
Biliary atresia, extrahepatic or NOS (use 751670 for intrahepatic)	751650	5
Duodenal web	751560	7
Duplication of anus, appendix, cecum, or intestine / enterogenous cyst	751500	4
Ectopic (displaced, anteriorly placed) anus	751530	2
Esophageal atresia with TE fistula	750310	32
Esophageal atresia without TE fistula	750300	1
Hirschsprung disease, NOS	751330	14
Hirschsprung disease: Long-segment (aganglionosis beyond rectum)	751310	7
Hirschsprung disease: Short-segment(aganglionosis involving no more than the anal	751320	7
Intestinal atresia/stenosis, Duodenum	751100	11
Intestinal atresia/stenosis, Ileum	751120	8
Intestinal atresia/stenosis, Jejunum	751110	15
Intestinal atresia/stenosis, Large Intestine, NOS	751200	2
Malrotation: cecum and/or colon	751400	9
Malrotation: Other specified and unspecified	751490	32
Malrotation: small intestine alone	751495	9
Microcolon	751520	4
Microgastria	750700	2
Other specified anomalies of intestine	751580	5
Persistent omphalomesenteric duct / persistent vitelline duct	751000	5
Rectal atresia/stenosis with fistula	751210	2
Rectal atresia/stenosis without mention of fistula	751220	3
Tracheoesophageal fistula without mention of esophageal atresia	750320	1
Tracheoesophageal fistula, "H" type	750325	2

Selected ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2000-2001 (cont'd)

Genitourinary

BPA Label	BPA Code	# of Defects
Urachus: Patent	753700	3
Absence of bladder or urethra	753800	1
Bladder exstrophy	753500	6
Cloacal exstrophy	751550	1
Genital organs: Other specified anomalies / microgenitalia / macrogenitalia	752880	1
Gyne: Cervix, absence, atresia or agenesis	752400	1
Gyne: OS anomalies of cervix, vagina, or external female genitalia /# VAGINAL tags	752480	5
Gyne: Ovaries absence or agenesis	752000	1
Gyne: Ovaries, Multiple cysts	752085	3
Gyne: Uterus absence or agenesis	752300	1
Gyne: Uterus, other anomalies / bicornuate/ unicornis	752380	1
Gyne: Vagina, absence or atresia complete or partial	752410	2
Hypospadias, Second Degree	752606	66
Hypospadias, Second Degree with Chordee	752626	44
Hypospadias, Third Degree	752607	13
Hypospadias, Third Degree with Chordee	752627	16
Indeterminate sex, NOS / ambiguous genitalia	752790	8
Kidney: Lobulated, fused, or horseshoe / crossed fused ectopia	753320	6
Kidneys: Multicystic renal dysplasia / multicystic kidney	753160	18
Kidneys: Polycystic, infantile type (IPKD)	753110	2
Obstruction, atresia or stenosis of anterior urethra	753620	1
Obstruction, atresia or stenosis of urinary meatus / meatal stenosis	753630	2
Other and unspecified atresia and stenosis of urethra and bladder neck	753690	2
Other atresia, or stenosis of bladder neck	753610	1
Other specified anomalies of bladder and urethra	753880	3
Penis: Other anomalies / concealed penis / absent or hooded foreskin	752860	9
Penis: Small / hypoplastic / micropenis	752865	7
Renal agenesis, bilateral	753000	4
Atresia, stricture, or stenosis of ureter / ureteropelvic junction obstruction or stenosis	753210	48
Congenital hydronephrosis / pyelocaliectasis	753200	95
Kidney: Double or triple, pelvis / pyelon duplex or triplex	753310	7
Kidney: Ectopic / pelvic	753330	6
Kidney: Enlarged, hyperplastic, or giant	753340	1
Kidney: Other specified anomalies	753380	2
Megaloureter, NOS / hydroureter	753220	25
Other and unspecified obstructive defects of renal pelvis and ureter	753290	2
Ureter: Accessory / double ureter / duplex collecting system	753410	28
Ureter: Ectopic	753420	7
Ureter: Other specified anomalies / ureterocele	753480	14
Testis and scrotum: Other anomalies / polyorchidism / bifid scrotum	752820	4
True hermaphroditism / ovotestis	752700	1
Urachus: Cyst	753710	1
Urethra: Congenital posterior urethral valves or posterior urethral obstruction	753600	16

Selected ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2000-2001 (cont'd)

Musculoskeletal

BPA Label	BPA Code	# of Defects
Achondroplasia	756430	9
Arthrogryposis multiplex congenita / distal arthrogryposis syndrome	755800	18
Osteogenesis imperfecta	756500	3
Other specified chondrodystrophy. Excludes: Conradi's (use 756575)	756480	3
Thanatophoric dwarfism	756447	1
Absence of foot or toes, Bilateral	755349	4
Absence of foot or toes, Left	755346	9
Absence of foot or toes, Right	755347	4
Absence of hand or fingers, Bilateral	755249	3
Absence of hand or fingers, Left	755246	17
Absence of hand or fingers, Right	755247	12
Absence of the forearm and hand, Right	755242	3
Absence of the lower leg and foot, Left	755341	1
Absence of thigh only (lower leg and foot present), Left	755331	1
Congenital postural scoliosis	754200	11
Craniosynostosis, Coronal, Bilateral	756014	7
Craniosynostosis, Coronal, Laterality Unknown	756010	3
Craniosynostosis, Coronal, Left	756011	3
Craniosynostosis, Coronal, Right	756012	5
Craniosynostosis, Lambdoidal, Bilateral	756024	1
Craniosynostosis, Lambdoidal, Laterality Unknown	756020	1
Craniosynostosis, Lambdoidal, Left	756021	2
Craniosynostosis, Metopic	756006	15
Craniosynostosis, Sagittal	756005	40
Craniosynostosis, Unspecified Type, Bilateral	756004	1
Diaphragmatic hernia, Bochdalek, Left	756611	1
Diaphragmatic hernia, Bochdalek, Right	756612	1
Diaphragmatic hernia, Morgagni, Laterality Unknown	756615	1
Diaphragmatic hernia, Morgagni, Left	756616	1
Diaphragmatic hernia, Morgagni, Right	756617	1
Diaphragmatic hernia, NOS (inc. absent/hemidiaphragm), Laterality Unknown	756600	1
Diaphragmatic hernia, NOS (inc. absent/hemidiaphragm), Left	756601	20
Diaphragmatic hernia, NOS (inc. absent/hemidiaphragm), Right	756602	5
Fibular aplasia/hypoplasia, Right	755372	2
Gastroschisis	756710	34
Klippel-Feil syndrome / Wildervanck syndrome	756110	1
Limb deficiencies, Upper Limb, Not Elsewhere Classifiable, Left	755281	1
Longitudinal deficiency of arm, NOS, Left	755251	1
Omphalocele	756700	24
Poland syndrome or anomaly	756800	5
Prune belly syndrome	756720	2
Radial aplasia/hypoplasia, Bilateral	755269	2
Radial aplasia/hypoplasia, Left	755266	3
Radial aplasia/hypoplasia, Right	755267	3
Anomalies of fingers / camptodactyly / macrodactylia / brachydactyly / triphalangeal	755500	21
Anomalies of forearm, NOS	755530	1

Selected ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2000-2001 (cont'd)

BPA Label	BPA Code	# of Defects
Bowing, femur	754400	1
Bowing, tibia and/or fibula	754410	1
Clubfoot, NOS / talipes, NOS	754730	65
Clubfoot: Metatarsus varus or adductus	754520	13
Clubfoot: Talipes calcaneovarus	754510	1
Clubfoot: Talipes equinovarus	754500	83
Eventration of diaphragm	756620	5
Knee: anomalies / hyperextended knee	755640	1
Lower limb: hypoplasia / Toes, feet, legs: hypoplasia	755685	4
Lower limb: other specified anomalies / hyperextended legs / shortening of legs	755680	1
Other absent or hypoplastic muscle / absent pectoralis major	756810	1
Polydactyly fingers / postaxial polydactyly, Type A	755005	20
Polydactyly: Accessory big toe (preaxial)	755030	11
Polydactyly: Accessory digits foot, NOS (preaxial, postaxial not specified)	755096	3
Polydactyly: Accessory digits hand, NOS (preaxial, postaxial not specified)	755095	5
Polydactyly: Accessory thumbs (preaxial polydactyly)	755010	41
Polydactyly: Accessory toes (postaxial)	755020	21
Ribs: Absence	756300	9
Ribs: Extra	756330	10
Ribs: Fused	756320	1
Ribs: Other anomalies	756340	1
Syndactyly: Fused fingers	755100	18
Syndactyly: Fused toes	755120	15
Syndactyly: Unspecified (see below for specified site)	755190	1
Syndactyly: Unspecified (webbed vs. fused) thumb and / or fingers, NOS	755193	13
Syndactyly: Unspecified toes	755194	16
Syndactyly: Webbed fingers	755110	10
Syndactyly: Webbed toes / Excludes webbing between the second and third toes	755130	7
Talipes calcaneovalgus	754600	3
Unspecified varus deformities of feet	754590	1
Upper limb: Hypoplasia / Fingers, hands, or arms: hypoplasia	755585	8
Upper limb: Other specified anomalies	755580	2
Sacral agenesis	756175	3
Spine: Kyphosis / kyphoscoliosis	756120	4
Split-Foot, Left	755356	1
Split-Hand, Left	755256	2
Split-Hand, Right	755257	1
Syndactyly: Unspecified, laterality not specified	755196	1
Thumb only missing or hypoplastic, Bilateral	755264	1
Thumb only missing or hypoplastic, Left	755261	4
Thumb only missing or hypoplastic, Right	755262	2
Tibial aplasia / hypoplasia, Right	755367	2
Transverse deficiency or amputation of the arm, NOS, Left	755201	1
Ulnar aplasia/hypoplasia, Bilateral	755274	1
Ulnar aplasia/hypoplasia, Left	755271	2
Vertebrae, cervical: anomalies	756140	1
Vertebrae, cervical: hemivertebrae	756145	3
Vertebrae, lumbar: anomalies	756160	5
Vertebrae, lumbar: hemivertebrae	756165	4
Vertebrae, sacrococcygeal: anomalies / agenesis of sacrum	756170	5
Vertebrae, thoracic: anomalies	756150	5
Vertebrae, thoracic: hemivertebrae	756155	13
Vertebrae: Hemivertebrae, NOS	756185	1
Vertebrae: Other specified anomalies	756180	5

Selected ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2000-2001 (cont'd)

Chromosomal and Other Syndromes

BPA Label	BPA Code	# of Defects
Apert syndrome / Acrocephalosyndactyly types I or II	756055	3
Autosome NOS: Other specified anomalies / marker / ring, derivative, mosaic, isochrome, inversions	758580	9
Deletion 13q / deletion of long arm of 13	758330	1
Deletion 17p or 18p / deletion of short arm chromosome 17 or 18	758350	1
Deletion 17q or 18q / deletion of the long arm of chromosome 17 or 18	758340	1
Deletion 4p / Wolff-Hirschorn syndrome	758320	1
Deletion: Autosome (not X or Y) (ie. #1-16, 4q,5q,19,20) / Include microdeletion	758380	13
Down syndrome: mosaic	758040	7
Down syndrome: translocation trisomy - duplication of a G group chromosome, NOS	758030	1
Down syndrome: translocation trisomy 21 - duplication of a 21q chromosome	758020	3
Down syndrome: trisomy 21	758000	182
Goldenhar syndrome / oculoauriculovertebral dysplasia	756060	4
Hemifacial microsomia	756065	5
Klinefelter syndrome: 47, XXY	758700	7
Mosaic XO/XY, 45X/46XY. Excludes: with Turner phenotype (use 758610)	758800	1
Mosaic XY/XXY, 46XY/47XXY. Excludes: Klinefelter phenotype (use 758710)	758820	1
Other craniofacial syndromes / Hallermann-Streiff syndrome	756046	3
Other specified acrocephalosyndactylies	756057	1
Sex chromosome: Other specified anomaly / fragile X	758880	2
Treacher-Collins syndrome / Mandibulofacial dysostosis	756045	1
Trisomy 13: Patau syndrome	758100	10
Trisomy 13: translocation trisomy with duplication of a 13q	758120	1
Trisomy 18: Edwards syndrome	758200	16
Trisomy, partial / 8/02 "partial trisomy" = "duplication". But, for "dup NOS" use 758930	758530	8
Trisomy: 6, 7, 9, 10, 11, 12 / Other trisomy C (archaic)	758510	2
Trisomy: Other total trisomy syndromes / trisomy 22 / trisomy, NOS	758520	1
Tuberous sclerosis / Bourneville's disease	759500	10
Turner syndrome: karyotype 45,X [XO]	758600	8
Turner syndrome: variant karyotypes, eg. isochromosome, mosaic (eg X, XX,XY),	758610	7
Unbalanced translocations, OS. Excludes: bal trans in normal (758400)	758540	2
Unspecified chromosome: Deletion of chromosome(s), NOS	758920	2
XXX female / 47XXX / Triple X syndrome	758850	5
XYY, male / 47,XYY / mosaic XYY male	758840	3
DiGeorge Syndrome	279110	12
Malformation OS: VATER/VACTERL/Acardia/Angelman/Bloom/CHARGE/hemihyper/Meckel-Gruber/ Neu-Laxova/PentalogyCantrell/Sotos/TownesBrock/WalkerWarburg/ Weaver/ VCFS,Shprintzen	759890	12
Malformation Syndromes / face: Aarskog /BOF/BOR/Fraser/FreemanSheldon/Kabuki/Miller-Dieker/Noonan/ Opitz G/oral-facial-digital/Oto-palato-digital/Septo-optic dysplasia/Waardenburg/Williams	759800	11
Malformation Syndromes / limbs: Baller-Gerold/Carpenter/caudal regression/Fryns/Holt-Oram/ Klippel-Trenaunay-Weber/LimbBodyWall/Roberts/Rubinstein-Taybi/sirenomelia/ thrombocytopenia-absent radius	759840	6
Malformation Syndromes / metabolic: Alagille/Alport/Beckwith-Wiedemann/Johansen-Blizzard/ leprechaunism/Lowe/Menkes(kinky hair)/Prader-Willi/Zellweger	759870	8
Malformation Syndromes / other skeletal: Marfan/Stickler/Beemer Langer	759860	7
Malformation Syndromes / short stature: Smith-Lemli-Optiz/de Lange/Cockayne/Laurence-Moon-Biedl/ Russell-Silver/Seckel	759820	4

Selected ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2000-2001 (cont'd)

BPA Label	BPA Code	# of Defects
Other		
Collodion baby	757110	1
Ectodermal dysplasia	757340	1
Epidermolysis bullosa	757330	2
Incontinentia pigmenti	757350	1
Other and unspecified ichthyosis	757190	3
X-linked ichthyosis	757196	3
Adrenogenital syndrome / adrenal hyperplasia	255200	2
Amniotic band sequence	658800	5
Anomalies of thymus / absent thymus	759240	3
Hamartomas: OS	759680	2
Ichthyosiform erythroderma	757197	2
Multiple congenital anomalies (=MCA NOS", not "MCA no specific diagnosis)	759700	4
Skin: Other specified anomalies / scalp defects	757800	1
Spleen: Absence / asplenia	759000	1
Spleen: Accessory / Use for polysplenia, though not exactly the same	759040	4
Spleen: Other specified anomalies	759080	3
Thyroglossal duct anomalies / thyroglossal cyst	759220	3

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.

Hydronephosis: 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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