Massachusetts Birth Defects 2004-2005

Massachusetts Birth Defects Monitoring Program
Bureau of Family Health and Nutrition

Massachusetts Department of Public Health

November 2009

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Ron Benham, Director, Bureau of Family Health and Nutrition
Marlene Anderka, Director, Massachusetts Center for Birth Defects Research and Prevention
Cathleen Higgins, Birth Defects Surveillance Coordinator

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Kyra S. Minihane, MBA, March of Dimes Massachusetts Chapter

Registry of Vital Records and Statistics staff
Bureau of Health Information, Statistics, Research and Evaluation
Massachusetts Department of Public Health

Coleen Boyle, PhD, and staff
Division of Birth Defects and Developmental Disabilities
National Center on Birth Defects and Developmental Disabilities
Centers for Disease Control and Prevention

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For more information, contact:

Massachusetts Department of Public Health
Bureau of Family Health and Nutrition
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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Note to Readers: Changes in this year's report

This report contains changes from previous reports. These are outlined by section.

Section	Change or addition
Chap 1	This chapter contains a brief discussion on folic acid awareness and behavior of women in Massachusetts.
Chap 3	Prevalence of selected birth defects in live births, stillbirths and missing pregnancy outcomes not included in Massachusetts surveillance in this reporting period has been estimated and compared to average national rates. This chapter contains some preliminary trend analyses for selected birth defects in Massachusetts.
Chap 4	We include a brief discussion on the potential impact of assisted reproductive technology (ART) on birth defect prevalence in Massachusetts.
Chap 5	Analyses are presented for prevalence of chromosomal and all other defects by maternal characteristics such as race/Hispanic ethnicity, birthplace and Massachusetts region.
Chap 6	We describe the use of a new automated algorithm created to classify cases by severity. In addition, a new section includes the breakdown of birth defect causes, with the birth defect cases with known causes being classified by etiology and those with unknown causes being classified by pattern.
Appendix	A list of birth defects classified by severity level and a map of Massachusetts regions are included. Also included are two tables describing folic acid awareness and behavior.

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Executive Summary

Although birth defects are rare when compared to other adverse birth outcomes, such as low birthweight or prematurity, they are the leading cause of death in the first year of life in the United States. Nationally, about 20% of all infant deaths result from a birth defect. In Massachusetts, 16.0% of all infant deaths were attributable to birth defects (MADPH 2007).

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.

Folic acid deficiency is related to certain birth defects. The awareness of folic acid intake to prevent birth defects may be increasing, although almost half of Massachusetts mothers with recent live births surveyed as part of the CDC Pregnancy Risk Assessment Monitoring System (PRAMS) did not take any multivitamins or prenatal vitamins during the month prior to pregnancy (Lu 2009).

The Massachusetts combined lifetime costs for babies born with any of 12 major structural birth defects are an estimated $125 million in 2005 dollars (see Technical Notes for inflation adjustment) (Harris and James 1997). These figures include direct costs of medical treatment, developmental services and special education, as well as indirect costs to society for lost wages due to early death or occupational limitations. The psychosocial costs cannot be calculated.

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

This report presents statewide data on the prevalence of birth defects in live births and stillbirths in Massachusetts during the years 2004 and 2005. The first annual report presented Massachusetts birth defects data for the year 1999. Two subsequent reports compiled data from 2000-2001 and 2002-2003, respectively. Our ability to find and identify infants born with birth defects to Massachusetts residents has improved over time and is reflected in increased prevalence rates. The 2004-2005 data are presented in combined form since the numbers are

relatively small for individual defects. Interpretations of these data must be made with caution until a multi-year estimate establishes a stable, baseline rate.

The data allow for some preliminary trend analyses and evaluation of the efficacy of public health treatment and prevention efforts such as folic acid awareness and intake monitoring. The BDMP continues to monitor and improve case ascertainment quality so that reliable and accurate data are available to inform policy planning of public health efforts.

Planning for children with special health care needs is essential to support affected infants and families. Coordination between the BDMP 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.

Prevalence

The overall prevalence of birth defects among births to Massachusetts residents in 2004-2005 was 166.8 per 10,000 live births. Among the 155,284 live births to Massachusetts residents in 2004-2005, 2,536 had one or more birth defects. In addition, 54 stillbirths were identified as having a birth defect. Population-based, active surveillance statewide from 1999 through 2005 provides baseline frequencies for birth defects, although more years of data will establish the stability of these preliminary baseline frequencies. Three of the ten most common defects were cardiovascular defects: atrial septal defects (secundum and NOS), ventricular septal defects (membranous and NOS) and pulmonary stenosis, valvular. Common non-cardiovascular defects included hypospadias, clubfoot, polydactyly/syndactyly, Down syndrome, obstructive genitourinary defects, cleft lip with and without cleft palate and cleft palate without cleft lip.

The CDC published improved national prevalence estimates for 18 selected major defects. Massachusetts was one of 11 states with population-based monitoring programs to contribute birth defect data. These average prevalence rates cover deliveries from 1999-2001 (CDC 2006). Massachusetts rates for 2004-2005 were significantly lower than the U.S. rates for approximately half of the defects and were about the same as the national estimates for the other half, although the Massachusetts rate for endocardial cushion defects was higher than the U.S. rate. The lower rates for the other defects may reflect differences in surveillance system methodology and regional variation. Also, birth defects are not reported in Massachusetts when they are prenatally diagnosed and the pregnancy is electively terminated, which would tend to result in lower rates for Massachusetts for certain defects. Based on previous studies of defects in association with prenatal screening and subsequent elective termination (Forrester, Merz et al. 1998; Cragan and Khoury 2000; Peller, Westgate et al. 2004), adjusted Massachusetts rates that include cases currently not included in surveillance become similar to or slightly higher than the average U.S. rates. In addition, spontaneous deliveries of stillbirths ≥ 20 weeks of gestation were reported by birthing hospitals, but limited

information about the stillbirth is included in the maternal record. Thus some birth defects are not well documented and cannot be confirmed for inclusion in state surveillance. Regulations promulgated in February 2009 allow for the collection of defects diagnosed prenatally and up to three years of age. Collecting these data will yield more comprehensive estimates of birth defects in the Commonwealth.

Cases with One Defect vs. Two or More Defects

Of all 2,590 birth defect cases (2,536 live births and 54 stillbirths) 60.9% had one defect and 39.1% had more than one (defined as having more than one defect among defects that were included in this surveillance report). Anencephaly, cleft lip with and without cleft palate, biliary atresia and hypospadias appeared more often as a single defect rather than with other defects. Birth defects which appeared more often in conjunction with other defects included holoprosencephaly, anophthalmia/microphthalmia, cloacal exstrophy, omphalocele, reduction deformity of lower limbs, trisomy 18 and the majority of cardiovascular defects.

Selected Pregnancy Outcomes

We compared selected pregnancy outcomes (C-sections, birthweight, gestational age, multiple birth and infant death) among infants born with birth defects to those born without birth defects in 2004-2005. Of infants born with birth defects, 43.5% were C-section deliveries, compared to 31.4% of non-birth defect births; 21.1% of low birthweight births (<2500 grams) had a birth defect as opposed to 7.6% of those without a birth defect; 5.0% of infants with a birth defect died before their 1st birthday, compared to 0.4% of those without a birth defect. While numbers of infants with birth defects are relatively small, it is important to recognize the impact of these outcomes when diagnosing and treating a baby with a birth defect.

Plurality

Examining the birth defect rate by plurality is important since birth defects are more common among multiple births, and the number of multiple births has been on the rise in Massachusetts. The birth defect prevalence was 164.3 for singletons and 216.4 for multiple births (more than one infant) per 10,000 live births. Birth defects that occurred more frequently in multiple births included tetralogy of Fallot with and without pulmonary atresia, valvular pulmonary stenosis, hypospadias (2nd or 3rd degree) and clubfoot.

Sex

The birth defect prevalence was 134.5 for females and 197.6 for males per 10,000 live births. While the prevalence of most types of birth defects did not substantially differ by sex of the infant/fetus, some conditions were associated with sex. The most common defects seen in males were hypospadias, atrial septal defects (secundum and NOS), obstructive genitourinary defects, polydactyly/syndactyly, clubfoot and Down syndrome. The most common defects seen in females were atrial septal defects (secundum and NOS), ventricular septal defects (membranous and NOS), Down syndrome, clubfoot, polydactyly/syndactyly and cleft palate.

Analysis of Trends

Preliminary trend analysis using data from this report and previous reports may be performed with the understanding that better ascertainment of cases and increased use of diagnostic technologies may be factors in any apparent increase in birth defect rates. Trends in select cardiovascular, orofacial and musculoskeletal birth defects for which we have large numbers of cases during each report interval allow for preliminary baseline prevalence rates to be estimated. Gastroschisis and hypospadias are two defects that display increases between 2000 to 2005, which may reflect the aggressive strategies and improvements in surveillance. The increased prevalence rate of gastroschisis parallels the national trend.

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 157.6 for mothers younger than 20 years, 161.5 for those 20-24 years, 154.7 for those 25-29 years, 158.4 for those 30-34 years and 181.1 for those 35 years and older. Mothers younger than the age of 20 had the highest rate (14.1 per 10,000) of gastroschisis. This association has been shown in previous studies (Fillingham and Rankin 2008; Vu, Nobuhara et al. 2008). As expected, there was a strong association between Down syndrome and advanced maternal age. Although 45% of babies with Down syndrome were born to women under 35, the Down syndrome rate of 27.0 per 10,000 births for women 35 years and older was about three times that of any other maternal age group. The next highest Down syndrome rate of 9.2 per 10,000 births occurred in the 30-34 age group. The pattern of higher Down syndrome rates among older women reflects the pattern of higher chromosomal defects in general among older women. In 2005 Massachusetts had the highest ratio of the number of assisted reproductive technology (ART) procedures among state residents at (1,340 per million residents). In 2005, Massachusetts was one of the five states with the highest frequencies of ART procedures performed (Wright, Chang et al. 2008). This high frequency of ART procedures may be due in part to more complete insurance coverage in Massachusetts, and may, in fact, be an underestimate (Zhang Z, Macaluso M et al). In the U.S., ART has also been associated with some birth defects such as septal heart defects and cleft lip with and without cleft palate (Reefhuis, Honein et al. 2009).

Maternal Race / Hispanic Ethnicity

The prevalence of birth defects varied by maternal race and Hispanic ethnicity. The rate per 10,000 live births was 161.9 for Non-Hispanic Whites, 180.5 for Non-Hispanic Blacks, 142.0 for Non-Hispanic Asians/Pacific Islanders and 166.7 for Hispanics. Due to small numbers, the rates for other races were not calculated. The most common defects in Hispanics included Down syndrome, septal defects, clubfoot, cleft lip with and without cleft palate and obstructive genitourinary defects.

In Blacks, the most common defects included atrial septal defects (secundum and NOS), Down syndrome, polydactyly/syndactyly, clubfoot, hypospadias and obstructive genitourinary defects. The most common defects in Whites included atrial septal defects, hypospadias, clubfoot, obstructive genitourinary defects, polydactyly/syndactyly and Down syndrome. In Asians, the most common defects included polydactyly/syndactyly, hypospadias, septal defects, obstructive genitourinary defects, Down syndrome and cleft palate. The age-adjusted rates of chromosomal defects in Blacks, Hispanics and the combined "other" races were about 2 times the rate in Whites and almost 3 times the rate in Asians. This may reflect differential health access and prenatal diagnoses. Maternal birthplace (U.S. versus non-U.S.) may be a contributing factor in group differences, as White and Black women born in the U.S. had slightly higher rates than those born outside the U.S., while the opposite was observed in Asian women. The prevalence of birth defects in children of Hispanic women born in the U.S. Territories (including Puerto Rico, U.S. Virgin Islands and Guam) was non-significantly higher than the rates of women born both in and outside the U.S.

Region

The birth defect rates among six Massachusetts regions in 2004-2005 were not statistically significantly different. The rates ranged from 148.7 per 10,000 in Western Massachusetts to 172.9 per 10,000 in Southeast Massachusetts.

Severity

A severity scale was developed by the MCBDRP 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. We developed a new automated algorithm to classify the cases. Nearly 3% of cases with birth defects were classified as "severe" and most of these cases did not survive. This percentage was an underestimate of severe cases due to limited data. For example, researchers 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 and Khoury 2000). About 17% of cases were affected with a "serious" birth defect. Many of these cases needed intensive medical care and planning for continuing care and long-term disability. "Moderately severe" birth defects comprised 73% of the total cases; all of these needed medical follow up; many may have required a number of surgeries and extensive treatment. "Mild" birth defects comprised nearly 7% of the affected infants. These defects may or may not have required corrective treatment.

Etiology and Pattern

The surveillance system in Massachusetts allowed for the collection of relevant etiology (cause) information. Cases with known etiology accounted for about 18% of the birth defects in Massachusetts in 2004-2005. Of the cases with known cause, "single gene" etiology accounts for almost 24%, "chromosomal" etiology

accounts for almost 67% and “maternal-fetal factors” accounted for about 8%. The majority of birth defects cases in Massachusetts in 2004-2005 have unknown etiology (82%). Among the cases with unknown etiology, the “multiple major” (2 or more major defects in different organs/body parts) pattern comprised the 11%, while the “isolated” pattern comprised the majority (89%). Within the “isolated” pattern, three subgroups further described the occurrence of the birth defect(s) of cases: 1) truly solitary defects, 2) two or more defects in the same organ or body part, or an isolated major and some minor defects in different body parts, and 3) a sequence (common primary defect with consistent, related anomalies).

Related Public Health Resources

Two resources include: "Public Health Resources in Massachusetts" and a list of "Selected National Resources." Both may be found through the Massachusetts Department of Public Health website: www.mass.gov/dph/birthdefects.

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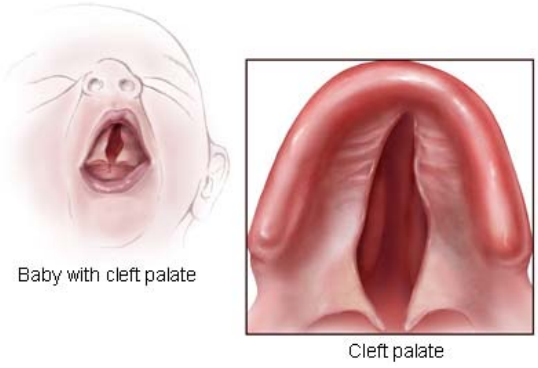
Chapter 1

Introduction



Courtesy of the Centers for Disease Control and Prevention

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 faces the reality that their baby has a birth defect (March of Dimes 2006). 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 severe.

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 may be catastrophic.

Although birth defects are rare when compared to other adverse birth outcomes, such as low birthweight or prematurity, they are the leading cause of death in the first year of life in the United States. Nationally, about 20% of all infant deaths result from a birth defect. In Massachusetts, 16.0% of all infant deaths were attributable to birth defects (MADPH 2007). The overall infant mortality rate for Massachusetts in 2004-2005 was 4.9 per 1,000 births, and for the U.S. it was 6.8 per 1,000 births.

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.

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 70% of major birth defects, no known cause has been identified (CDC 2006). We show in Chapter 6 that about 82% of birth defect cases in Massachusetts have unknown etiology. These cases were further classified by birth defect patterns.

Researchers are looking at a wide variety of environmental exposures and risk factors as causes of birth defects. 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.

Birth Defects and Folic Acid Awareness and Behavior

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 and Signore 2004). However, recent studies in certain populations have suggested that not all cases of neural tube defects are preventable by increasing folate intake (Heseker, Mason et al. 2008) and that periconceptional supplement use did not reduce the risk of neural tube defects possibly because folate fortification reduced the occurrence of only folic acid-sensitive neural tube defects (Mosley, Cleves et al. 2009).

The Behavioral Risk Factor Surveillance System (BRFSS), maintained by the Massachusetts Department of Public Health, Bureau of Health Information Statistics, Research and Evaluation, included questions about folic acid awareness and behavior in its 2000 and 2004 surveys. In the 2004 survey, 63.8% of female respondents ages 18-49 recognized that taking folic acid can prevent birth defects. Also, 51.6% of the participating women ages 18-49 responded that they take folic acid on a daily basis. The responses for both surveys by age group can be found in the Appendix.

An additional and potentially more relevant indicator of folic acid intake can be multivitamin use in women who recently had a pregnancy. Massachusetts conducted a 3-month pilot Pregnancy Risk Assessment Monitoring System (PRAMS) study among resident women who were 18 years or older and who had a liveborn infant between January and March 2005. Mothers of singletons, twins and triplets (higher order multiples were excluded) were asked about their weekly consumption of a multivitamin or prenatal vitamin the month prior to becoming pregnant (Lu 2009). 42.5% of the respondents (N=348) took no multivitamins, 12.4% took 1-3 multivitamins per week, 9.1% took 4-6 multivitamins per week and 35.8% took multivitamins on a daily basis (see table in Appendix).

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% (DHHS 2000). 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 defect rates over time that may indicate a change in environmental conditions affecting the health of the population;
- Geographical areas with consistently high or unusual rates;
- Clusters of birth defects;
- Families of affected children who may benefit from services or who may be interested in participating in research studies; and
- Key data for preventive strategy planning by the Department of Public Health.

Birth Defects Surveillance in Massachusetts

Over the past ten years, the BDMP has developed and refined its surveillance program. The first full year of population-based, active surveillance statewide was 1999.

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

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

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

Economic Impact on Massachusetts

Estimating the economic impact of birth defects on the state of Massachusetts is challenging. The California Birth Defects Monitoring Program and the Metropolitan Atlanta Congenital Defects Program, using 1992 data, calculated the lifetime costs for families dealing with a baby with birth defects to be between $75,000 and $503,000 (CDC 1995). Their estimated lifetime costs for a baby born with spina bifida would be $373,966 in 2005 dollars.

Adjusting for inflation, the Massachusetts combined lifetime costs for babies born with 12 major structural birth defects were an estimated $125 million in 2005 dollars (see Technical Notes). These figures included direct costs of medical treatment, developmental services and special education, as well as indirect costs to society for lost wages due to early death or occupational limitations. There are also social and emotional impacts.

Legislative Changes Regarding Birth Defects Surveillance

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

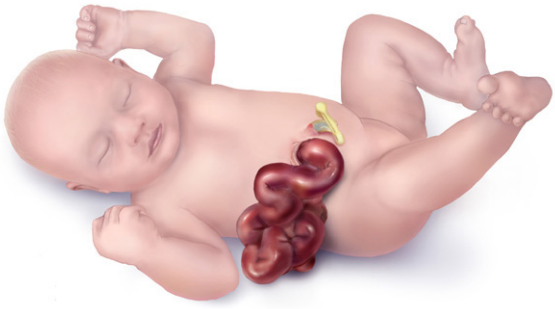
The 2004-2005 Surveillance Report

This report presents statewide data on the prevalence of birth defects in live births and stillbirths in Massachusetts during the years 2004 and 2005. The data are presented in combined form since the numbers are relatively small for individual defects. The first annual report presented Massachusetts data for birth defects for the year 1999. Our second and third reports compiled data from 2000-2001 and 2002-2003, respectively. Our ability to find and identify infants born with birth defects to Massachusetts residents has improved over time. The approximate 12% increase in prevalence from 2000-2001 to 2002-2003 and approximately 9% increase in prevalence from 2002-2003 to 2004-2005 are attributable to improved case ascertainment. Interpretations of these data must be made with caution until a multi-year estimate establishes a stable, baseline rate.

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

Chapter 2

Methods



Baby with gastroschisis

Courtesy of the Centers for Disease Control and Prevention

Case Definition

This report summarizes data on selected birth defects present in births occurring during the calendar years 2004 through 2005 to Massachusetts residents. Cases met the following criteria:

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

Data Collection

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

Confidentiality

Great care was taken to protect the confidentiality of data. The Center has developed extensive procedures to guarantee the confidentiality of the 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 in combination with other defects. If the case had more than one defect within the same defect category, only one of these defects was counted in the category total. If the case had more than one defect in different defect categories, the case was listed in the total for each of these defect categories. Thus the counts in the defect categories presented in the prevalence tables represent the total number of defects and not the total number of cases with birth defects. Each case in the BDMP was linked to a Registry of Vital Records and Statistics 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 reports, analyses of maternal age and race/ethnicity are limited to live births.

The occurrence of birth defects is reported as prevalence. Prevalence is calculated as the number of birth defect cases born during the period 2004-2005 per 10,000 live births born during the same period. Prevalence tables include the number of cases found, the estimated prevalence rate per 10,000 live births and the 95% confidence interval for that rate. The incidence (new cases) of birth defects (based upon the number of embryos conceived within a year) is not fully measured because both the total number of conceptions that occur and the number of these conceptions resulting in a defect are not known (Sever 2004).

The confidence interval (CI) can be used to assess the magnitude and stability of a rate or ratio. The 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 2004 through 2005. 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 BDMP ascertains cases only at birthing hospitals, two non-birthing tertiary care centers, two non-birthing community hospitals and one specialty care hospital. Thus, defects that are not diagnosed at birth, that do not need hospitalization, or that are diagnosed after age one may be underreported (e.g., cardiac defects that are detected in an outpatient setting after the immediate newborn period).

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

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

5. Spontaneous abortions that are delivered prior to 20 weeks of gestation and less than 350 grams are not included in the case definition. It has been estimated that about 29% of birth defects cases are missed by not monitoring early fetal loss (Forrester, Merz et al. 1998; Ethen and Canfield 2002).

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, 1,318 births occurred to Massachusetts residents at out-of-state hospitals. Of these births, 68.9% occurred in Rhode Island (RI) hospitals. In order to capture data on infants with birth defects residing in the southeastern region of Massachusetts that were born or treated at RI hospitals, we began receiving hospital discharge lists and abstracting medical records on infants with birth defects at two RI hospitals. Deliveries and diagnoses that occurred in other out-of-state facilities are not included at this time.

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

Glossary

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

Chapter 3

Prevalence of Birth Defects

Baby with anencephaly

Courtesy of the Centers for Disease Control and Prevention

Overall Prevalence of Birth Defects

Table 1 shows the prevalence of defects for all births and for live births and stillbirths separately. Among the 155,284 live births to Massachusetts residents in 2004-2005, 2,536 had one or more structural birth defects that were ascertained by Massachusetts BDMP. In addition, 54 stillbirths were identified with a birth defect. Overall, 1.7% of births in the state (166.8 per 10,000 live births) were identified as having one or more birth defects. This represents a 9.4% increase from 2002-2003, when the prevalence rate was 157.4 per 10,000. This increase was at least partly due to better reporting from hospitals and improved ascertainment of cases. The majority of defects occurred in the cardiovascular (33.3%) and musculoskeletal (26.9%) categories. Figure 1 shows the percentage of reported birth defects by defect categories. Cases can be included in more than one defect category.

The CDC published improved national prevalence estimates for 18 selected major defects. Massachusetts was one of 11 states with population-based monitoring programs to contribute birth defect data. These average prevalence rates cover deliveries from 1999-2001 (CDC 2006). Massachusetts rates for 2004-2005 were significantly lower than the US rates for approximately half of the defects and were about the same as the national estimates for the other half, although the Massachusetts rate for endocardial cushion defects combined was greater than the US rate (see Table 2). The different rates may reflect variation in defect criteria between surveillance systems as well as regions. Also, birth defects are not reported in Massachusetts when they are prenatally diagnosed and the pregnancy is electively terminated, which would tend to result in lower rates for Massachusetts for certain defects. Spontaneous deliveries of stillbirths equal to or greater than 20 weeks of gestation were reported by birthing hospitals but limited information about the stillbirth is included in the maternal record. Thus, some birth defects are not well documented and are unable to be confirmed for inclusion in state surveillance.

Another data source to which we can compare Massachusetts rates is the neural tube defect (NTD) Ascertainment project of the National Birth Defects Prevention Network at CDC. Massachusetts has submitted data quarterly since 1999. Using data from 1999-2000, researchers from CDC calculated prevalence rates for spina bifida and anencephaly, two serious birth defects that occur early in pregnancy (CDC 2004). Birth defect programs which included prenatally diagnosed cases of spina bifida that are subsequently electively terminated had a prevalence rate of 4.1. Massachusetts does not collect data on elective terminations and has the spina bifida prevalence rate of 1.7 per 10,000 for 2004-2005 which is nearly 60% lower than the national rate (see Table 2).

CDC estimates that up to 50% of pregnancies with neural tube defects and up to 80% of pregnancies with anencephaly may be electively terminated after prenatal diagnosis (Cragan and Khoury 2000). Substantial evidence from past studies have examined the effect of prenatal diagnoses and elective termination on the

prevalence of various birth defects (Forrester, Merz et al. 1998). Researchers at Brigham and Women Hospital (BWH), where 11% of resident births occurred in 2004-2005, looked at past trends in elective termination in Massachusetts. For the two years 1994 and 1999, 40-80% of pregnancies prenatally diagnosed with either lethal or very severe defects were terminated (Peller, Westgate et al. 2004), suggesting that epidemiologic studies of major malformations must include elective terminations to be complete.

We can estimate cases not included in the surveillance if we compare our data to several studies such as the aforementioned BWH study (Table 3). These studies provide the numbers of cases captured by surveillance with and without the inclusion of electively terminated cases. We calculate the difference in number of cases between the two surveillance systems as a percentage of possible cases missed by a surveillance system such as ours which does not include electively terminated cases. We then adjust the Massachusetts rates for the selected birth defects to include all cases (non-terminated cases and estimated terminated cases), assuming that the cited studies examined populations similar to the residents of Massachusetts who had prenatal health access similar to that found in Massachusetts.

The unadjusted 2004-2005 Massachusetts rates for anencephaly, spina bifida and trisomy 18 were lower than the national estimates. The rate for Down syndrome was similar to the national estimate (Table 2). An estimated 50-72% of anencephaly cases are missed through exclusion of terminated cases. Upon adjusting the Massachusetts rate of anencephaly, one out of the three adjusted Massachusetts rates of anencephaly may be similar to the national estimate while the other two adjusted rates are lower than the national estimate. Likewise, with an estimated 29-46% of spina bifida cases missed through exclusion of terminated cases in surveillance, one of the three adjusted Massachusetts rates of spina bifida may also be similar to the national estimate while the other two are lower than the national estimate. In the case of trisomy 18, where 49-57% of cases may be missed due to elective termination, the two adjusted Massachusetts rates are similar to the national estimate. The estimated 35-37% of Down syndrome cases missed due to elective termination based on two sources translates to adjusted Massachusetts rates exceeding the national estimate. Table 3 lists the adjusted rates. The adjusted rates of trisomy 18 and Down syndrome in Massachusetts are similar and higher, respectively, compared to national estimates, possibly reflecting the older population having children in Massachusetts.

The adjusted rates calculated here in Table 3 are based on previous studies examining data from various locations and dating as far back as 1974 through 1999. Therefore, the adjusted rates for these defects may be an underestimation, because the studies cited are not recent and may not reflect the current behaviors in prenatal diagnostic testing and elective termination decision-making processes in the state. In recent years, due to the increasing numbers of pregnancies in older women, different diagnostic testing behaviors may have developed. Further

evidence of these trends may be reflected in the 3-month pilot study for the CDC Pregnancy Risk Assessment Monitoring System (PRAMS) in Massachusetts women who were 18 years or older and who had a liveborn infant between January to March 2005. Mothers of singleton, twins and triplets (higher order multiples were excluded) were asked whether they had discussed with healthcare providers the availability of tests to screen for birth defects or diseases during prenatal care visits. 90% of the respondents (N=348) replied that a healthcare provider had discussed the issue with them, and the responses did not vary statistically significantly among the different racial and ethnic groups polled (Lu 2009).

Table 4 shows the most common birth defects in the state. Three of the ten most common defects were cardiovascular defects: atrial septal defects (secundum and NOS), ventricular septal defects (membranous and NOS) and pulmonary stenosis, valvular. Common non-cardiovascular defects included hypospadias, clubfoot, polydactyly/syndactyly, Down syndrome, obstructive genitourinary defect, cleft lip with and without cleft palate and cleft palate without cleft lip. Cardiovascular defects were the most commonly occurring birth defects in both Massachusetts and in the nation. They also contribute more to infant deaths than any other defect category (CDC 1998).

Cases with One Defect vs. Two or More Defects

Table 5 shows the distribution of birth defects by whether they appeared as a one diagnosis or in combination (more than one) with defects from other organs, organ systems, or parts of the body. A case was defined as having more than one defect if it had more than one defect from among those defects that are included in this surveillance report.

Among birth defect cases, 60.9% had one defect and 39.1% had more than one defect. Anencephaly, cleft lip with and without cleft palate, biliary atresia and hypospadias appeared more often as a one defect rather than with other defects. Birth defects that appeared more often in conjunction with other defects included holoprosencephaly, anophthalmia/microphthalmia, cloacal exstrophy, omphalocele, reduction deformity of lower limbs, trisomy 18 and the majority of cardiovascular defects. Overall, in 2004-2005, cardiovascular defects were 6.6 times more likely to occur with other defects than as a single defect, whereas in 2002-2003 they were 5 times more likely to occur with more than one defect than as one defect.

Selected Pregnancy Outcomes

Figure 2 compares selected pregnancy outcomes (Cesarean section, birthweight, gestational age, multiple birth and infant death) among infants born with birth defects to those born without birth defects in 2004-2005 by percentage. Of infants born with birth defects, 43.5% were Cesarean deliveries, compared to 31.4% of non-birth defect births; 21.1% of infants with birth defects had low birthweight (<2,500 grams) as opposed to 7.6% of those without a birth defect; 5.0% of infants

with a birth defect died before their 1st birthday, compared to 0.4% of those without a birth defect. While numbers of infants with birth defects are relatively small, it is important to recognize the impact of these outcomes when diagnosing and treating a baby with a birth defect.

Prevalence of Birth Defects by Sex

Table 6 presents the prevalence of birth defects by sex of the infant/fetus. The overall prevalence was 135.0 (126.95-143.51) for females and 197.6 (188.0-207.51) for males per 10,000 live births. While the prevalence of most types of birth defects did not differ by sex of the infant/fetus, some conditions were significantly associated with sex. Table 7 shows the most common birth defects for females and males. The most common defects seen in males were hypospadias, atrial septal defects (secundum and NOS), obstructive genitourinary defects, polydactyly/syndactyly, clubfoot and Down syndrome. The most common defects seen in females were atrial septal defects (secundum and NOS), ventricular septal defects (membranous and NOS), Down syndrome, clubfoot, polydactyly/syndactyly and cleft palate. Selected birth defects by sex of infant are presented in Figure 3.

Trend Analyses of Selected Birth Defects

The statewide data on the prevalence of birth defects in live births and stillbirths during multiple years from this surveillance report as well as three prior surveillance reports allow for some trend analysis. Data from the first few reports must be interpreted with caution since surveillance techniques were being established and executed. However, preliminary trend analysis with recent reports may be performed with the understanding that better ascertainment of cases and increased use of diagnostic technologies may be factors in any apparent increase in birth defect rates. A comparison of selected cardiovascular birth defects rates from 2004-2005 to rates in the previous two reports suggests that most rates have remained steady or slightly increased due to better case confirmation. Similar trends are found in selected orofacial and musculoskeletal birth defects as well.

Defects such as gastroschisis and hypospadias are some of the defects of interest due to increasing national prevalence trends. Surveillance in Massachusetts and elsewhere has suggested an increased risk of gastroschisis in pregnancies of very young women (under 20), although the mechanisms for this are unknown. A recent study has shown that although younger women had comparatively larger risk of gastroschisis and greater prevalence of smoking, cigarette smoking had no effect on risk in women under 20, but smoking (possibly the duration of smoking) increased the risk in older women (Werler, Mitchell et al. 2009). The rate of gastroschisis in Massachusetts has increased 72.4% between 2000 and 2005 (Figure 4). This increasing trend parallels the increasing national rates of gastroschisis, although a factor in this increase may be better ascertainment of cases by the surveillance system in Massachusetts. Hypospadias displayed an increase between 2000 to 2005 of about 46.8% (Figure 4). This may be due to aggressive strategies of case confirmation and improvements in surveillance,

although over time, increases have been observed in other locations, around the U.S. and internationally as well.

Table 1 Prevalence of Birth Defects, Massachusetts: 2004-2005

Defect[1]	Count among live births	Count among stillbirths	Total Count	Rate per 10,000 Births	95% Confidence Interval
Total Cases	2,536	54	2,590	166.8	160.6-173.4
Central Nervous System: 227 Total Cases					
Anencephaly	8	4	12	0.77	0.40 – 1.35
Encephalocele	3	0	3	0.19	0.04 – 0.56
Holoprosencephaly	6	1	7	0.45	0.18 – 0.93
Hydrocephaly w/o spina bifida	44	2	46	2.96	2.17 – 3.95
Microcephaly	31	0	31	2.00	1.36 – 2.83
Spina bifida w/ and w/o hydrocephaly	25	1	26	1.67	1.09 – 2.45
Spinal cord defect	45	0	45	2.90	2.11 – 3.88
Other CNS	101	2	103	6.63*	5.41– 8.04
Eye: 78 Total Cases					
Anophthalmia/microphthalmia	7	1	8	0.52	0.22 – 1.02
Congenital glaucoma, congenital cataract	46	0	46	2.96	2.17 – 3.95
Other eye	34	0	34	2.19*	1.52 – 3.06
Ear: 49 Total Cases					
Anotia/microtia	27	0	27	1.74	1.15 – 2.53
Other ear	24	2	26	1.67*	1.09 – 2.45
Cardiovascular: 862 Total Cases					
Anomalous Pulmonary Venous Connection					
Total/partial anomalous pulmonary venous connection	31	0	31	2.00	1.36 – 2.83
Atrioventricular Canal Defects					
ASD primum	9	0	9	0.58	0.27 – 1.10
Common atrium	5	0	5	0.32	0.10 – 0.75
Complete atrioventricular canal defect	57	2	59	3.80	2.89 – 4.90
Endocardial cushion defect (OS and NOS)	12	1	13	0.84	0.45 – 1.43
VSD, canal type	10	1	11	0.71	0.35 – 1.27
Conotruncal (Outlet) and Aortic Arch					
Double outlet right ventricle	29	0	29	1.87	1.25 – 2.68
d – Transposition of the great arteries	35	1	36	2.32	1.62 – 3.21
Interrupted aortic arch, type B	2	0	2	0.13	0.02 – 0.47

Table 1 Prevalence of Birth Defects, Massachusetts: 2004-2005

Defect[1]	Count among live births	Count among stillbirths	Total Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)					
Tetralogy of Fallot w/ and w/o pulmonary atresia	70	0	70	4.51	3.51 – 5.70
Truncus arteriosus	3	0	3	0.19	0.04 – 0.56
Ebstein Anomaly					
Ebstein anomaly	8	0	8	0.52	0.22 – 1.02
Laterality Defects					
Heterotaxy	29	0	29	1.87	1.25 – 2.68
Left-Sided Obstruction					
Aortic valve stenosis	23	0	23	1.48	0.94 – 2.22
Coarctation of aorta	76	0	76	4.89	3.86 – 6.13
Hypoplastic left heart syndrome	20	1	21	1.35	0.84 – 2.07
Interrupted aortic arch (type A and NOS)	2	0	2	0.13	0.02 – 0.47
Patent Ductus Arteriosus					
Patent ductus arteriosus	303	0	303	19.51	17.38 – 21.84
Right-Sided Obstruction					
Pulmonary stenosis, valvular	92	0	92	5.92	4.78 – 7.27
Pulmonary valve atresia w/intact septum	11	1	12	0.77	0.40 – 1.35
Pulmonary valve atresia with VSD	8	0	8	0.52	0.22 – 1.02
Tricuspid valve atresia	5	2	7	0.45	0.18 – 0.93
Septal Defects					
ASD (secundum and NOS)	284	1	285	18.35	16.28 – 20.61
VSD (membranous and NOS)	150	9	159	10.24	8.71 – 11.96
VSD, conoventricular/malalignment	18	0	18	1.16	0.69 – 1.83
Single Ventricle and L – TGA					
L – TGA	8	0	8	0.52	0.22 – 1.02
Single ventricle	9	0	9	0.58	0.27 – 1.10
Other Cardiovascular					
Other cardiovascular	297	3	300	19.32*	17.19 – 21.63
Respiratory: 76 Total Cases					
Choanal atresia	10	0	10	0.64	0.31 – 1.18
Lung anomalies	40	2	42	2.70	1.95 – 3.66
Other respiratory	25	1	26	1.67*	1.09 – 2.45
Orofacial: 279 Total Cases					

Table 1 Prevalence of Birth Defects, Massachusetts: 2004-2005

Defect[1]	Count among live births	Count among stillbirths	Total Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)					
Cleft lip w/ and w/o cleft palate	118	4	122	7.86	6.52 – 9.38
Cleft palate w/o cleft lip	93	1	94	6.05	4.89 – 7.41
Pierre Robin sequence	34	0	34	2.19	1.52 – 3.06
Other orofacial	72	2	74	4.77*	3.74 – 5.98
Gastrointestinal: 237 Total Cases					
Biliary atresia	5	0	5	0.32	0.10 – 0.75
Esophageal atresia/tracheoesophageal fistula	38	2	40	2.58	1.84 – 3.51
Hirschsprung disease	35	0	35	2.25	1.57 – 3.13
Rectal and large intestinal atresia/stenosis	46	1	47	3.03	2.22 – 4.02
Small intestinal atresia	42	1	43	2.77	2.00 – 3.73
Other gastrointestinal	89	2	91	5.86*	4.72 – 7.20
Genitourinary: 541 Total Cases					
Bladder exstrophy	4	0	4	0.26	0.07 – 0.66
Cloacal exstrophy	5	0	5	0.32	0.10 – 0.75
Hypospadias, 2nd or 3rd degree	211	1	212	13.65	11.88 – 15.62
Obstructive genitourinary defect	167	0	167	10.75	9.19 – 12.51
Renal agenesis/hypoplasia	6	1	7	0.45	0.18 – 0.93
Other genitourinary	236	3	239	15.39*	13.5 – 17.47
Musculoskeletal: 696 Cases					
Clubfoot	180	6	186	11.98	10.32 – 13.83
Craniosynostosis	68	0	68	4.38	3.40 – 5.55
Diaphragmatic hernia	42	3	45	2.90	2.11 – 3.88
Gastroschisis	42	6	48	3.09	2.28 – 4.10
Omphalocele	12	2	14	0.90	0.49 – 1.51
Polydactyly/syndactyly	183	2	185	11.91	10.26 – 13.76
Reduction deformity, lower limbs	21	1	22	1.42	0.89 – 2.14
Reduction deformity, upper limbs	47	1	48	3.09	2.28 – 4.10
Skeletal dysplasia	23	1	24	1.55	0.99 – 2.30
Other musculoskeletal	168	6	174	11.21*	9.60 – 13.00
Chromosomal and other Syndromes: 413 Total Cases					
Klinefelter syndrome	9	0	9	0.58	0.27 – 1.10

Table 1 Prevalence of Birth Defects, Massachusetts: 2004-2005

Defect[1]	Count among live births	Count among stillbirths	Total Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)					
Trisomy 13	10	1	11	0.71	0.35 – 1.27
Trisomy 18	12	10	22	1.42	0.89 – 2.14
Trisomy 21 (Down syndrome)	181	3	184	11.85	10.20 – 13.69
Turner syndrome	13	3	16	1.03	0.59 – 1.67
Other chromosomal syndromes/other syndromes	169	5	174	11.21*	9.60 – 13.00
Other: 49 Total Cases					
Amniotic bands	14	1	15	0.97	0.54 – 1.59
Skin anomalies	8	0	8	0.52	0.22 – 1.02
Other, specified	27	0	27	1.74*	1.15 – 2.53

[1.] Cases can be included in the count for more than one defect. Cases are counted once in the total for a defect category.

* Note that this rate may represent a heterogeneous group of defects.

Table 2 Comparison of Selected Massachusetts 2004-2005 Birth Defect Rates to National Estimates

Defect	Count	Rate per 10,000 Births MA[1]	95% Confidence Interval	Rate per 10,000 Births US[2]	95% Confidence Interval
Anencephaly[3]	12	0.77	0.40 – 1.35	3.45	3.27 – 3.64
Spina bifida[3]	26	1.67	1.09 – 2.45	4.10	3.90 – 4.30
Anophthalmia/microphthalmia	8	0.52	0.22 – 1.02	2.08	1.90 – 2.27
Truncus arteriosus (common truncus)	3	0.19	0.04 – 0.56	0.82	0.71 – 0.93
Transposition of the great arteries[4]	44	2.83	2.06 – 3.80	4.73	4.47 – 5.00
Tetralogy of Fallot	70	4.51	3.51 – 5.70	3.92	3.67 – 4.17
Endocardial cushion defect [5]	97	6.26	5.07 – 7.62	4.35	4.10 – 4.62
Hypoplastic left heart syndrome	21	1.35	0.84 – 2.07	2.43	2.24 – 2.63
Cleft palate without cleft lip	94	6.05	4.89 – 7.41	6.39	6.08 – 6.71
Cleft lip with and without cleft palate	122	7.86	6.52 – 9.38	10.48	10.08 – 10.88
Esophageal atresia/tracheoesophageal fistula	40	2.58	1.84 – 3.51	2.37	2.18 – 2.56
Rectal and large intestinal atresia/stenosis	47	3.03	2.22 – 4.02	4.81	4.54 – 5.08
Reduction deformity, upper limbs	48	3.09	2.28 – 4.10	3.79	3.55 – 4.03
Reduction deformity, lower limbs	22	1.42	0.89 – 2.14	1.90	1.73 – 2.07
Gastroschisis	48	3.09	2.28 – 4.10	3.73	3.49 – 3.97
Omphalocele	14	0.90	0.49 – 1.51	2.09	1.91 – 2.27
Diaphragmatic hernia	45	2.90	2.11 – 3.88	2.94	2.73 – 3.15
Trisomy 21 (Down syndrome)	184	11.85	10.20–13.69	13.65	13.19 – 14.12
Trisomy 13	11	0.71	0.35 – 1.27	1.33	1.18 – 1.47
Trisomy 18	22	1.42	0.89 – 2.14	2.41	2.22 – 2.61

1. MA rate is based on live births and stillbirths.
2. All US rates except for anencephaly and spina bifida are from the CDC MMWR report presenting improved national prevalence estimates based on the average prevalence from 11 states including MA (CDC 2006). Rates were adjusted for race–specific distribution of US live births during 1999–2001 for all defects except anencephaly, spina bifida, Down syndrome, trisomy 13 and trisomy 18. Rates for Down syndrome, trisomy 13 and trisomy 18 were adjusted for maternal age.
3. National estimates for anencephaly and spina bifida are based on the CDC MMWR report detailing average prevalence rates from 23 active surveillance systems including MA. These rates are from surveillance systems that include prenatally diagnosed and terminated pregnancies (CDC 2004).
4. Includes d–TGA and L–TGA.
5. Includes ASD primum, common atrium, CAVC, endocardial cushion defect OS and NOS and VSD canal type.

Table 3 Prevalence of Selected Birth Defects Adjusted for Cases Not Currently Included in Massachusetts Surveillance (i.e. Elective Termination)

Defect	MA rate	95% Confidence Interval	Estimated Missing %[1]	Estimate Source[2]	Adjusted MA rate[3]	95% Confidence Interval	US rate	95% Confidence Interval
Anencephaly	0.77	0.40 – 1.35	66.7	Peller, et al.	2.32	1.62 – 3.21	3.45[4]	3.27 – 3.64
			50.0	Cragan, et al.	1.55	0.99 – 2.3		
			72.0	Forrester, et al.	2.76	2.00 – 3.72		
Spina bifida	1.67	1.09 – 2.45	45.5	Peller, et al.	3.07	2.26 – 4.07	4.10[4]	3.90 – 4.30
			29.0	Cragan, et al.	2.36	1.66 – 3.25		
			29.8	Forrester, et al.	2.38	1.68 – 3.28		
Trisomy 21 (Down syndrome)	11.85	10.20–13.69	35.2	Peller, et al.	18.29	16.22–20.54	12.94[5]	12.51–13.39
			37.3	Forrester, et al.	18.90	16.80–21.19		
Trisomy 18	1.42	0.89 – 2.14	56.5	Peller, et al.	3.26	2.42 – 4.29	2.29[5]	2.11 – 2.48
			49.0	Forrester, et al.	2.78	2.01 – 3.74		

1. Missing were cases of all gestational ages for which the diagnosed defect was ascertained before or after an elective termination. Missing % is defined as the percentage of electively terminated cases divided by all cases liveborn, stillborn and electively terminated for each defect.
2. Studies on the effect of prenatal diagnosis and elective terminations on birth defect surveillance. Peller, et al. provides data on liveborn, stillborn and elective terminations from a large urban tertiary center in Boston, MA, for the years 1974, 1979, 1984, 1989, 1994 and 1999 (Peller, Westgate et al. 2004). Cragan, et al. provides data on liveborn, stillborn and elective terminations from multiple states; the California Birth Defects Monitoring Program (1989 – 1991) was used here because it contributed the largest overall sample size to the study (Cragan and Khoury 2000). Forrester, et al. provides data on liveborn, stillborn and elective terminations from Hawaii's population based, active surveillance system, 1987 – 1996 (Forrester, Merz et al. 1998).
3. Adjusted rates included cases from elective terminations estimated according to the respective sources.
4. US average prevalence rates for trisomy 21 and trisomy 18 are unadjusted pooled estimates from 11 states during 1999 – 2001. While all 11 states had active surveillance, 3 of the states did not include cases of <20 weeks gestation ascertained from specialized sources for prenatal ascertainment; four states routinely visit prenatal diagnostic centers to ascertain cases, and four states obtain some prenatal data from sources such as genetics laboratories (CDC 2006).
5. US average prevalence rates for anencephaly and spina bifida are calculated from eight population-based surveillance systems that collect data systematically from sources with diagnostic prenatal ascertainment, including live births, stillbirths, fetal deaths and elective terminations during 1999 – 2000 (CDC 2004).

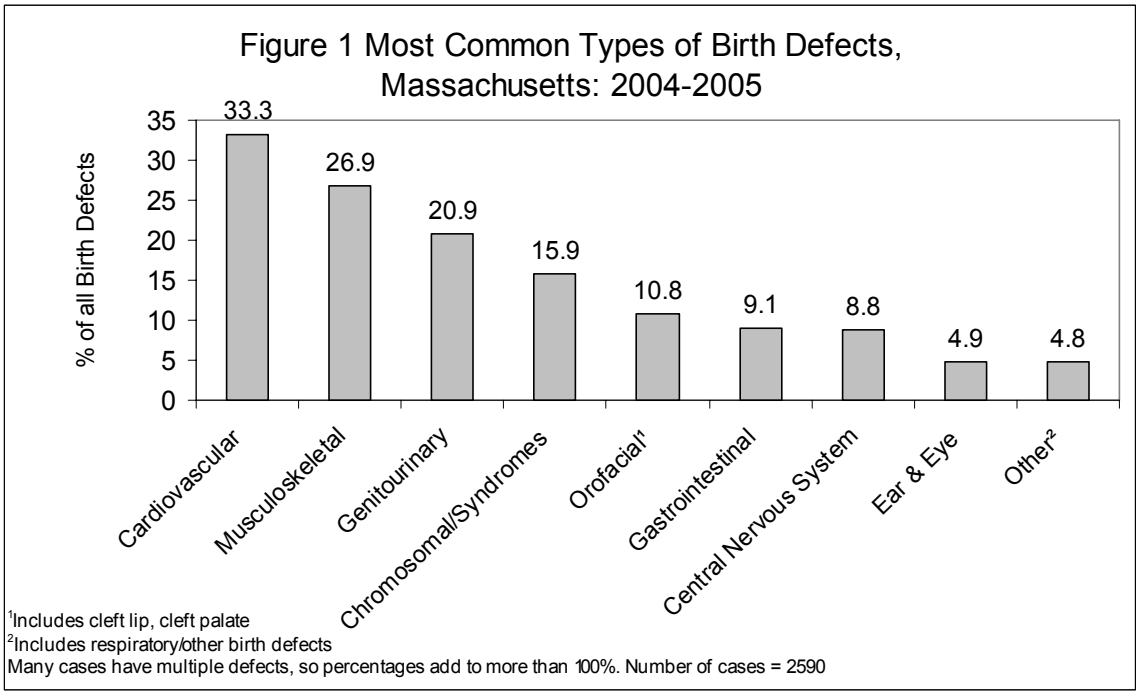


Table 4 Most Common Defects among Live Births and Stillbirths, Massachusetts: 2004 – 2005

Defect[1]	Category	Count	Rate per 10,000 Births	95% Confidence Interval
ASD (secundum and NOS)	Cardiovascular	285	18.35	16.28 – 20.61
Hypospadias, 2nd or 3rd degree	Genitourinary	212	13.65	11.88 – 15.62
Clubfoot	Musculoskeletal	186	11.98	10.32 – 13.83
Polydactyly/syndactyly	Musculoskeletal	185	11.91	10.26 – 13.76
Trisomy 21 (Down syndrome)	Chromosomal and other syndromes	184	11.85	10.20 – 13.69
Obstructive genitourinary defect	Genitourinary	167	10.75	9.19 – 12.51
VSD (membranous and NOS)	Cardiovascular	159	10.24	8.71 – 11.96
Cleft lip w/ and w/o cleft palate	Orofacial	122	7.86	6.52 – 9.38
Cleft palate w/o cleft lip	Orofacial	94	6.05	4.89 – 7.41
Pulmonary stenosis, valvular	Cardiovascular	92	5.92	4.78 – 7.27

1. Excludes patent ductus arteriosus (PDA) due to the high number of cases and the mild severity of the majority of these cases.

Table 5 Cases with One Defect vs. Two or More Defects among Live Births and Stillbirths, Massachusetts: 2004-2005

Defect[1]	Cases with one Defect	Cases with two or more defects[2]	Total Cases
Central Nervous System: 227 Total Cases			
Anencephaly	10	2	12
Encephalocele	1	2	3
Holoprosencephaly	0	7	7
Hydrocephaly w/o spina bifida	14	32	46
Microcephaly	15	16	31
Spina bifida w/ and w/o hydrocephaly	9	17	26
Spinal cord	15	30	45
Other CNS	40	63	103
Eye: 78 Total Cases			
Anophthalmia/microphthalmia	0	8	8
Congenital glaucoma, congenital cataract	33	13	46
Other eye	7	27	34
Ear: 49 Total Cases			
Anotia/microtia	14	13	27
Other ear	5	21	26
Cardiovascular: 862 Total Cases			
Anomalous Pulmonary Venous Connection			
Total/partial anomalous pulmonary venous connection	1	30	31
Atrioventricular Canal Defects			
ASD primum	0	9	9
Common atrium	0	5	5
Complete atrioventricular canal defect	3	56	59
Endocardial cushion (OS and NOS)	1	12	13
VSD, canal type	1	10	11
Conotruncal (Outlet) and Aortic Arch			
Double outlet right ventricle	1	28	29
d – Transposition of the great arteries	16	20	36
Interrupted aortic arch, type B	0	2	2
Tetralogy of Fallot w/ and w/o pulmonary atresia	14	56	70
Truncus arteriosus	0	3	3
Ebstein Anomaly			
Ebstein anomaly	3	5	8

Table 5 Cases with One Defect vs. Two or More Defects among Live Births and Stillbirths, Massachusetts: 2004-2005

Defect[1]	Cases with one Defect	Cases with two or more defects[2]	Total Cases
(cont'd)			
Laterality Defects			
Heterotaxy	1	28	29
Left – Sided Obstruction			
Aortic valve stenosis	0	23	23
Coarctation of aorta	8	68	76
Hypoplastic left heart syndrome	6	15	21
Interrupted aortic arch (type A and NOS)	0	2	2
Patent Ductus Arteriosus			
Patent ductus arteriosus	5	298	303
Right – Sided Obstruction			
Pulmonary stenosis, valvular	29	63	92
Pulmonary valve atresia w/intact septum	4	8	12
Pulmonary valve atresia with VSD	0	8	8
Tricuspid valve atresia	1	6	7
Septal Defects			
ASD (secundum and NOS)	45	240	285
VSD (membranous and NOS)	42	117	159
VSD, conoventricular/malalignment	3	15	18
Single Ventricle and L – TGA			
L – TGA	1	7	8
Single ventricle	0	9	9
Other Cardiovascular			
Other cardiovascular	28	272	300
Respiratory: 76 Total Cases			
Choanal atresia	7	3	10
Lung anomalies	31	11	42
Other respiratory	11	15	26
Orofacial: 279 Total Cases			
Cleft lip w/ and w/o cleft palate	97	25	122
Cleft palate w/o cleft lip	33	61	94
Pierre Robin sequence	0	34	34
Other orofacial	34	40	74

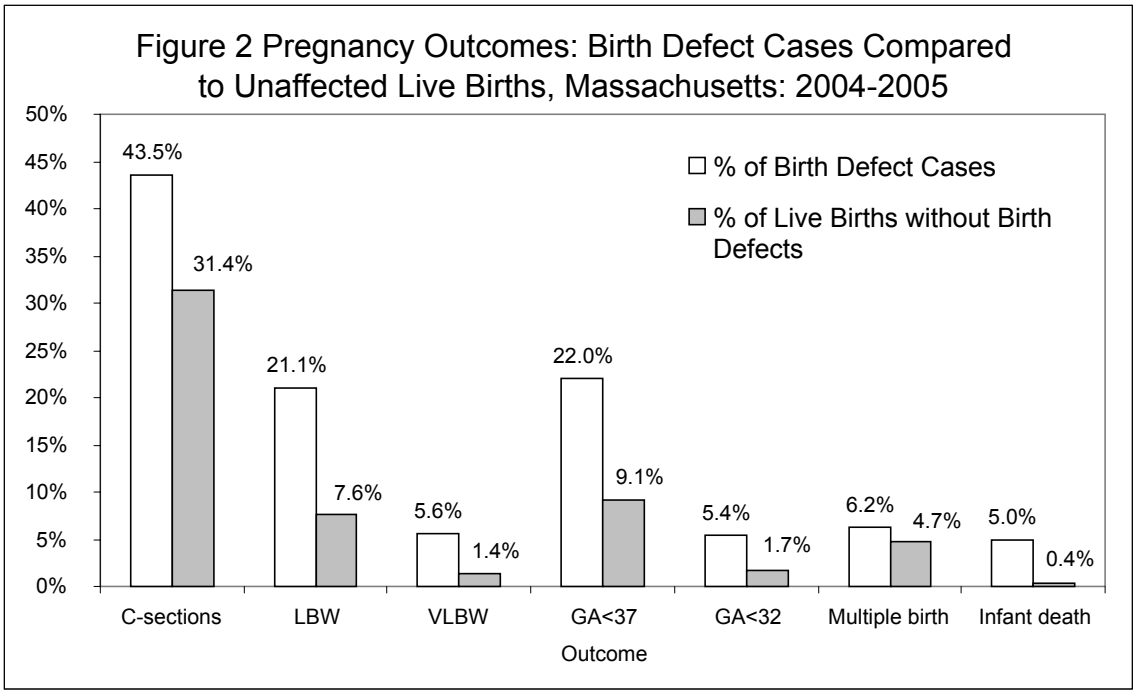
Table 5 Cases with One Defect vs. Two or More Defects among Live Births and Stillbirths, Massachusetts: 2004-2005

Defect[1]	Cases with one Defect	Cases with two or more defects[2]	Total Cases
(cont'd)			
Gastrointestinal: 237 Total Cases			
Biliary atresia	5	0	5
Esophageal atresia/tracheoesophageal fistula	15	25	40
Hirschsprung disease	24	11	35
Rectal and large intestinal atresia/stenosis	14	33	47
Small intestinal atresia	19	24	43
Other gastrointestinal	32	59	91
Genitourinary: 541 Total Cases			
Bladder exstrophy	2	2	4
Cloacal exstrophy	1	4	5
Hypospadias, 2nd or 3rd degree	184	28	212
Obstructive genitourinary defect	56	111	167
Renal agenesis/hypoplasia	2	5	7
Other genitourinary	106	133	239
Musculoskeletal: 696 Total Cases			
Clubfoot	124	62	186
Craniosynostosis	51	17	68
Diaphragmatic hernia	23	22	45
Gastroschisis	35	13	48
Omphalocele	3	11	14
Polydactyly/syndactyly	119	66	185
Reduction deformity, lower limbs	5	17	22
Reduction deformity, upper limbs	20	28	48
Skeletal dysplasia	14	10	24
Other musculoskeletal	20	154	174
Chromosomal and other Syndromes: 413 Total Cases			
Klinefelter syndrome	5	4	9
Trisomy 13	0	11	11
Trisomy 18	5	17	22
Trisomy 21 (Down syndrome)	48	136	184
Turner syndrome	10	6	16
Other chromosomal syndromes/other syndromes	40	134	174

Table 5 Cases with One Defect vs. Two or More Defects among Live Births and Stillbirths, Massachusetts: 2004-2005

Defect[1]	Cases with one Defect	Cases with two or more defects[2]	Total Cases
(cont'd)			
Other: 49 Total Cases			
Amniotic bands	0	15	15
Skin anomalies	5	3	8
Other, specified	2	25	27

1. Cases can be included in the count for more than one defect. Cases are counted once in the total for a defect category.
2. A case was defined as having multiple defects if it had more than one defect from among those that were included in this surveillance report.



LBW, low birthweight
VLBW, very low birthweight
GA<37, gestational age less than 37 weeks
GA<32, gestational age less than 32 weeks

Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2004-2005

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
Central Nervous System: 227 Total Cases				
Anencephaly	Male	2	0.25	0.03 – 0.91
	Female	10	1.32	0.63 – 2.43
Encephalocele	Male	1	0.13	0.00 – 0.70
	Female	2	0.26	0.03 – 0.95
Holoprosencephaly	Male	3	0.38	0.08 – 1.10
	Female	4	0.53	0.14 – 1.35
Hydrocephaly w/o spina bifida	Male	34	4.28	2.96 – 5.98
	Female	12	1.58	0.82 – 2.76
Microcephaly	Male	12	1.51	0.78 – 2.64
	Female	19	2.51	1.51 – 3.91
Spina bifida w/ and w/o hydrocephaly	Male	12	1.51	0.78 – 2.64
	Female	14	1.85	1.01 – 3.10
Spinal cord	Male	20	2.52	1.54 – 3.89
	Female	25	3.30	2.13 – 4.87
Other CNS	Male	51	6.42	4.78 – 8.44
	Female	52	6.86	5.12 – 8.99
Eye: 78 Total Cases				
Anophthalmia/microphthalmia	Male	2	0.25	0.03 – 0.91
	Female	6	0.79	0.29 – 1.72
Congenital glaucoma, congenital cataract	Male	24	3.02	1.94 – 4.49
	Female	22	2.90	1.82 – 4.39
Other eye	Male	19	2.39	1.44 – 3.73
	Female	15	1.98	1.11 – 3.26
Ear: 49 Total Cases				
Anotia/microtia	Male	18	2.27	1.34 – 3.58
	Female	9	1.19	0.54 – 2.25
Other ear	Male	15	1.89	1.06 – 3.11
	Female	11	1.45	0.72 – 2.60

Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2004-2005

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Cardiovascular: 862 Total Cases				
Anomalous Pulmonary Venous Connection				
Total/partial anomalous pulmonary venous connection	Male	16	2.01	1.15 – 3.27
	Female	15	1.98	1.11 – 3.26
Atrioventricular Canal Defects				
ASD primum	Male	2	0.25	0.03 – 0.91
	Female	7	0.92	0.37 – 1.90
Common atrium	Male	3	0.38	0.08 – 1.10
	Female	2	0.26	0.03 – 0.95
Complete atrioventricular canal defect	Male	26	3.27	2.14 – 4.79
	Female	33	4.35	3.00 – 6.11
Endocardial cushion (OS and NOS)	Male	8	1.01	0.43 – 1.98
	Female	5	0.66	0.21 – 1.54
VSD, canal type	Male	8	1.01	0.43 – 1.98
	Female	3	0.40	0.08 – 1.16
Conotruncal (Outlet) and Aortic Arch				
Double outlet right ventricle	Male	15	1.89	1.06 – 3.11
	Female	14	1.85	1.01 – 3.10
d – Transposition of the great arteries	Male	25	3.15	2.04 – 4.64
	Female	11	1.45	0.72 – 2.60
Interrupted aortic arch, type B	Male	2	0.25	0.03 – 0.91
	Female	0	0.00	0.00 – 0.49
Tetralogy of Fallot w/ and w/o pulmonary atresia	Male	39	4.91	3.49 – 6.71
	Female	31	4.09	2.78 – 5.80
Truncus arteriosus	Male	2	0.25	0.03 – 0.91
	Female	1	0.13	0.00 – 0.73
Ebstein Anomaly				
Ebstein anomaly	Male	5	0.63	0.20 – 1.47
	Female	3	0.40	0.08 – 1.16
Laterality Defects				

Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2004-2005

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Heterotaxy	Male	12	1.51	0.78 – 2.64
	Female	17	2.24	1.31 – 3.59
Left – Sided Obstruction				
Aortic valve stenosis	Male	15	1.89	1.06 – 3.11
	Female	8	1.06	0.46 – 2.08
Coarctation of aorta	Male	42	5.29	3.81 – 7.15
	Female	34	4.48	3.11 – 6.27
Hypoplastic left heart syndrome	Male	8	1.01	0.43 – 1.98
	Female	13	1.71	0.91 – 2.93
Interrupted aortic arch (type A and NOS)	Male	2	0.25	0.03 – 0.91
	Female	0	0.00	0.00 – 0.49
Patent Ductus Arteriosus				
Patent ductus arteriosus	Male	155	19.51	16.56 – 22.83
	Female	148	19.52	16.5 – 22.93
Right – Sided Obstruction				
Pulmonary stenosis, valvular	Male	45	5.66	4.13 – 7.58
	Female	47	6.20	4.55 – 8.24
Pulmonary valve atresia w/intact septum	Male	6	0.76	0.28 – 1.64
	Female	6	0.79	0.29 – 1.72
Pulmonary valve atresia with VSD	Male	0	0.00	0.00 – 0.46
	Female	8	1.06	0.46 – 2.08
Tricuspid valve atresia	Male	1	0.13	0.00 – 0.70
	Female	6	0.79	0.29 – 1.72
Septal Defects				
ASD (secundum and NOS)	Male	131	16.49	13.79 – 19.56
	Female	154	20.31	17.23 – 23.78
VSD (membranous and NOS)	Male	74	9.31	7.31 – 11.69
	Female	85	11.21	8.95 – 13.86
VSD, conoventricular/malalignment	Male	8	1.01	0.43 – 1.98
	Female	10	1.32	0.63 – 2.43
Single Ventricle and L – TGA				

Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2004-2005

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
L – TGA	Male	5	0.63	0.20 – 1.47
	Female	3	0.40	0.08 – 1.16
Single ventricle	Male	5	0.63	0.20 – 1.47
	Female	4	0.53	0.14 – 1.35
Other Cardiovascular				
Other cardiovascular	Male	160	20.14	17.14 – 23.51
	Female	140	18.46	15.53 – 21.79
Respiratory: 76 Total Cases				
Choanal atresia	Male	5	0.63	0.20 – 1.47
	Female	5	0.66	0.21 – 1.54
Lung anomalies	Male	25	3.15	2.04 – 4.64
	Female	17	2.24	1.31 – 3.59
Other respiratory	Male	16	2.01	1.15 – 3.27
	Female	10	1.32	0.63 – 2.43
Orofacial: 279 Total Cases				
Cleft lip w/ and w/o cleft palate	Male	84	10.57	8.43 – 13.09
	Female	38	5.01	3.55 – 6.88
Cleft palate w/o cleft lip	Male	35	4.41	3.07 – 6.13
	Female	59	7.78	5.92 – 10.04
Pierre Robin sequence	Male	16	2.01	1.15 – 3.27
	Female	18	2.37	1.41 – 3.75
Other orofacial	Male	48	6.04	4.45 – 8.01
	Female	26	3.43	2.24 – 5.02
Gastrointestinal: 237 Total Cases				
Biliary atresia	Male	4	0.50	0.14 – 1.29
	Female	1	0.13	0.00 – 0.73
Esophageal atresia/tracheoesophageal fistula	Male	21	2.64	1.64 – 4.04
	Female	19	2.51	1.51 – 3.91
Hirschsprung disease	Male	29	3.65	2.44 – 5.24
	Female	6	0.79	0.29 – 1.72

Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2004-2005

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Rectal and large intestinal atresia/stenosis	Male	27	3.40	2.24 – 4.94
	Female	20	2.64	1.61 – 4.07
Small intestinal atresia	Male	25	3.15	2.04 – 4.64
	Female	18	2.37	1.41 – 3.75
	Male	40	5.03	3.60 – 6.86
	Female	51	6.73	5.01 – 8.84
Genitourinary: 541 Total Cases				
Bladder exstrophy	Male	4	0.50	0.14 – 1.29
	Female	0	0.00	0.00 – 0.49
Cloacal exstrophy	Male	2	0.25	0.03 – 0.91
	Female	3	0.40	0.08 – 1.16
Hypospadias, 2nd or 3rd degree	Male	212	26.68	23.21 – 30.53
	Female	0	0.00	0.00 – 0.49
Obstructive genitourinary defect	Male	120	15.10	12.52 – 18.06
	Female	47	6.20	4.55 – 8.24
Renal agenesis/hypoplasia	Male	4	0.50	0.14 – 1.29
	Female	3	0.40	0.08 – 1.16
Other genitourinary	Male	179	22.53	19.35 – 26.08
	Female	60	7.91	6.04 – 10.19
Musculoskeletal: 696 Total Cases				
Clubfoot	Male	115	14.47	11.95 – 17.37
	Female	71	9.36	7.31 – 11.81
Craniosynostosis	Male	43	5.41	3.92 – 7.29
	Female	25	3.30	2.13 – 4.87
Diaphragmatic hernia	Male	31	3.90	2.65 – 5.54
	Female	14	1.85	1.01 – 3.10
Gastroschisis	Male	24	3.02	1.94 – 4.49
	Female	24	3.17	2.03 – 4.71
Omphalocele	Male	8	1.01	0.43 – 1.98
	Female	6	0.79	0.29 – 1.72
Polydactyly/syndactyly	Male	119	14.98	12.41 – 17.92
	Female	66	8.70	6.73 – 11.07

Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2004-2005

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Reduction deformity, lower limbs	Male	14	1.76	0.96 – 2.96
	Female	8	1.06	0.46 – 2.08
Reduction deformity, upper limbs	Male	20	2.52	1.54 – 3.89
	Female	28	3.69	2.45 – 5.34
Skeletal dysplasia	Male	14	1.76	0.96 – 2.96
	Female	10	1.32	0.63 – 2.43
Other musculoskeletal	Male	95	11.96	9.67 – 14.62
	Female	79	10.42	8.25 – 12.98
Chromosomal and other Syndromes: 413 Total Cases				
Klinefelter syndrome	Male	9	1.13	0.52 – 2.15
	Female	0	0.00	0.00 – 0.49
Trisomy 13	Male	8	1.01	0.43 – 1.98
	Female	3	0.40	0.08 – 1.16
Trisomy 18	Male	8	1.01	0.43 – 1.98
	Female	14	1.85	1.01 – 3.10
Trisomy 21 (Down syndrome)	Male	100	12.59	10.24 – 15.31
	Female	84	11.08	8.84 – 13.71
Turner syndrome	Male	2	0.25	0.03 – 0.91
	Female	14	1.85	1.01 – 3.10
Other chromosomal syndromes/other syndromes	Male	86	10.82	8.66 – 13.37
	Female	88	11.61	9.31 – 14.30
Other: 49 Total Cases				
Amniotic bands	Male	7	0.88	0.35 – 1.82
	Female	8	1.06	0.46 – 2.08
Skin anomalies	Male	3	0.38	0.08 – 1.10
	Female	5	0.66	0.21 – 1.54
Other, specified	Male	11	1.38	0.69 – 2.48
	Female	16	2.11	1.21 – 3.43

[1] Cases can be included in the count for more than one defect. Cases are counted once in the total for a defect category.

Table 7 Most Common Defects by Sex of Live Births and Stillbirths, Massachusetts: 2004-2005

Defect[1]	Count	Rate per 10,000 Births	95% Confidence Interval
FEMALE			
ASD (secundum and NOS)	154	20.31	17.23 – 23.78
VSD (membranous and NOS)	85	11.21	8.95 – 13.86
Trisomy 21 (Down syndrome)	84	11.08	8.84 – 13.71
Clubfoot	72	9.50	7.43 – 11.96
Polydactyly/syndactyly	66	8.70	6.73 – 11.07
Cleft palate w/o cleft lip	59	7.78	5.92 – 10.04
Obstructive genitourinary defect	47	6.20	4.55 – 8.24
Pulmonary stenosis, valvular	47	6.20	4.55 – 8.24
Cleft lip w/ and w/o cleft palate	38	5.01	3.55 – 6.88
Coarctation of aorta	34	4.48	3.11 – 6.27
MALE			
Hypospadias, 2nd or 3rd degree	212	26.68	23.21 – 30.53
ASD (secundum and NOS)	131	16.49	13.79 – 19.56
Obstructive genitourinary defect	120	15.10	12.52 – 18.06
Polydactyly/syndactyly	119	14.98	12.41 – 17.92
Clubfoot	115	14.47	11.95 – 17.37
Trisomy 21 (Down syndrome)	100	12.59	10.24 – 15.31
Cleft lip w/ and w/o cleft palate	84	10.57	8.43 – 13.09
VSD (membranous and NOS)	74	9.31	7.31 – 11.69
Pulmonary stenosis, valvular	45	5.66	4.13 – 7.58
Craniosynostosis	43	5.41	3.92 – 7.29

1. Excludes patent ductus arteriosus (PDA) due to the high number of cases and the mild severity of the majority of these cases.

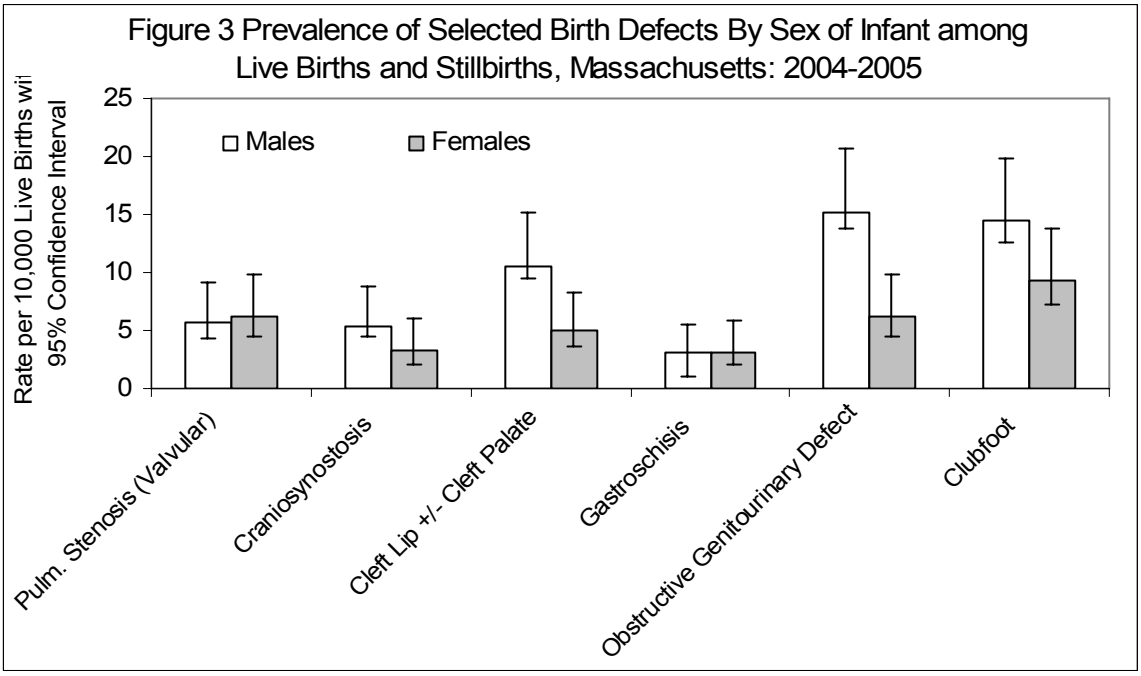


Figure 3 Prevalence of Selected Birth Defects By Sex of Infant among Live Births and Stillbirths, Massachusetts: 2004-2005
Rate per 10,000 Live Births wi
95% Confidence Interval
25
20
15
10
5
0
Males
Females
Pulm. Stenosis (Valvular)
Craniosynostosis
Cleft Lip +/- Cleft Palate
Gastroschisis
Obstructive Genitourinary Defect
Clubfoot

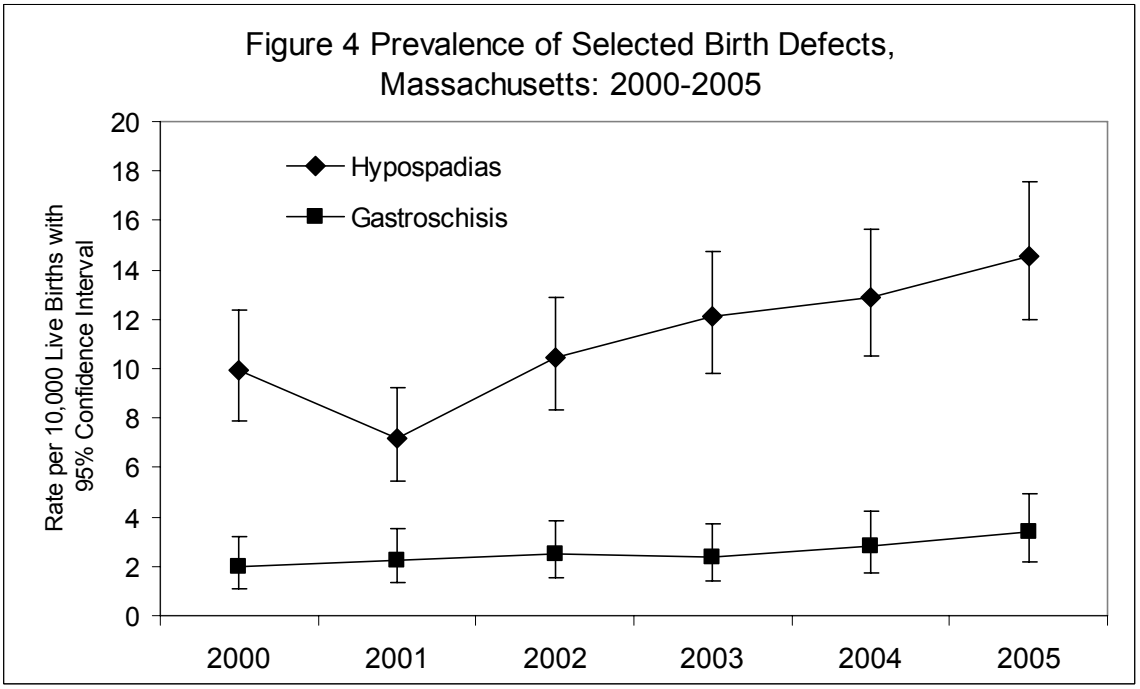


Figure 4 Prevalence of Selected Birth Defects, Massachusetts: 2000-2005
Rate per 10,000 Live Births with
95% Confidence Interval
20
18
16
14
12
10
8
6
4
2
0
Hypospadias
Gastroschisis
2000
2001
2002
2003
2004
2005

Chapter 4

Prevalence of Birth Defects by Plurality and Maternal Age

Baby with cleft lip

Courtesy of the Centers for Disease Control and Prevention

Plurality

Table 8 shows the distribution of birth defects by plurality. The overall prevalence was 164.3 (157.9-170.9) for singletons and 216.4 (184.1-252.8) for multiple births (more than one infant) per 10,000 live births. While multiple births comprised 4.7% of all live births, they comprised 6.2% of birth defects cases among live births (see Figure 5). Birth defects that occurred more frequently in multiple births included tetralogy of Fallot with and without pulmonary atresia, valvular pulmonary stenosis, hypospadias (2nd or 3rd degree) and clubfoot. Figure 6 presents rates for selected birth defects for singletons and multiples. Table 9 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.

Maternal Age

The prevalence of birth defects varied by maternal age. For live births only, rates per 10,000 live births were 157.6 (133.0-185.5) for mothers younger than 20 years, 161.5 (145.7-178.4) for those 20-24 years, 154.7 (142.2-168.0) for those 25-29 years, 158.4 (147.6-169.7) for those 30-34 years and 181.1 (167.7-195.3) for those 35 years and older. Table 10 shows the rates for birth defects by maternal age.

As expected, there was a strong association between Down syndrome and advanced maternal age (see Figure 7). Although 45% of babies with Down syndrome were born to women under 35, the Down syndrome rate of 27.0 per 10,000 births for women 35 years and older was at least two times that of any other maternal age group. The Down syndrome rate among mothers aged 30-34 was the second highest rate at 9.2 per 10,000 births. The pattern of higher Down syndrome rates among older women reflects the pattern of higher chromosomal defects in general among older women. Figure 8 shows the trend of increasing chromosomal defects as women age; the rate of chromosomal defects among women in the 35+ age group is significantly higher than all other age groups. The proportion of all birth defects that are chromosomal defects was higher in the 35+ age group than in other age groups.

Figure 9 shows that younger mothers (aged 19 and under) had the highest rate (14.13) of gastroschisis cases. This association has been shown in previous studies (Forrester and Merz 1999). Mothers younger than 25 years of age had infants with higher rates of gastroschisis and tetralogy of Fallot with and without pulmonary atresia than other age groups. Older mothers had higher rates for many defects including Down syndrome and ventricular septal defects (membranous and NOS). While results for other defects also differed by age group, the small numbers from two years of surveillance were not sufficient for interpretation.

Table 11 displays the most common birth defects for live births by maternal age groups. Atrial septal defects were one of the five most frequently occurring defects common to all maternal age groups. Polydactyly/syndactyly and clubfoot (except

for mothers 35 years and older) were among the five most common birth defects in every age group.

Monitoring birth defects by maternal age is important since the number of births to older mothers has been increasing over time in Massachusetts. Birth rates for women ages 30+ have increased steadily from 1980 to 2006. The number of births to women ages 30+ surpassed the number of births to women below age 30 in 1996. Recent data suggest that the numbers of births to women ages 30+ peaked in 2002 and has slightly decreased since, whereas the numbers of births to women ages below 30 reached a low point in 2004 and may be slightly increasing so that the numbers of births to women in the two age groups are converging (MADPH 2009). The number of births to women ages 30+ in 2006 is almost 2.3 times the number in 1980, and likewise, the number of births to women under age 30 in 2006 is 1.5 times less than the number in 1980 (MADPH 2008).

The percentage of women giving birth in the state who are over the age of 35 has more than doubled from 11.0% in 1989 to 24.0% in 2004-2005. In addition, multiple births occurred about twice as often in mothers giving birth at age 35 or over (7.6%) compared to mothers giving birth at an age under 35 (3.8%) (MADPH 2007).

A factor in both the increased percentage of women giving birth at an age of 35 or over and the disparity of multiple births among these older women giving birth may be the use of assisted reproductive technology (ART). Between 1997 to 2004, a steady 2% of Massachusetts births resulted from ART (CDC 2009) though this may be an underestimate (Zhang Z, Macaluso M et al). According to the CDC, Massachusetts ranked 5th highest in number of ART procedures performed (8,571), after California, New York, Illinois and New Jersey. In 2005 Massachusetts had the highest ratio of the number of ART procedures among state residents at 1,340 per million residents. Of the infants born in 2005 as a result of ART procedures, 43.5% were born in multiple birth deliveries (Wright, Chang et al. 2008). Figure 10 shows the disparity in percent of births that are single versus multiple from ART births and non-ART births in Massachusetts (CDC 2009). The vast majority of non-ART births are single deliveries, whereas almost half of ART births are multiple deliveries. ART poses many risks associated with multiple births that may lead to adverse maternal and infant outcomes such as low birthweight and preterm delivery. ART has also been associated with some birth defects such as septal heart defects and cleft lip with or without cleft palate (Reefhuis, Honein et al. 2009).

Table 8 Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2004-2005

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
Central Nervous System: 227 Total Cases				
Anencephaly	Singleton	11	0.74	0.37 – 1.33
	Multiple	1	1.36	0.03 – 7.58
Encephalocele	Singleton	3	0.20	0.04 – 0.59
	Multiple	0	0.00	0.00 – 5.02
Holoprosencephaly	Singleton	6	0.41	0.15 – 0.88
	Multiple	1	1.36	0.03 – 7.58
Hydrocephaly w/o spina bifida	Singleton	42	2.84	2.05 – 3.84
	Multiple	4	5.44	1.48 – 13.94
Microcephaly	Singleton	29	1.96	1.31 – 2.82
	Multiple	2	2.72	0.33 – 9.83
Spina bifida w/ and w/o hydrocephaly	Singleton	25	1.69	1.09 – 2.49
	Multiple	1	1.36	0.03 – 7.58
Spinal cord	Singleton	44	2.97	2.16 – 3.99
	Multiple	1	1.36	0.03 – 7.58
Other CNS	Singleton	98	6.62	5.38 – 8.07
	Multiple	5	6.80	2.21 – 15.88
Eye: 78 Total Cases				
Anophthalmia/microphthalmia	Singleton	8	0.54	0.23 – 1.07
	Multiple	0	0.00	0.00 – 5.02
Congenital glaucoma, congenital cataract	Singleton	43	2.91	2.10 – 3.92
	Multiple	3	4.08	0.84 – 11.93
Other eye	Singleton	33	2.23	1.54 – 3.13
	Multiple	1	1.36	0.03 – 7.58
Ear: 49 Total Cases				
Anotia/microtia	Singleton	25	1.69	1.09 – 2.49
	Multiple	2	2.72	0.33 – 9.83
Other ear	Singleton	25	1.69	1.09 – 2.49
	Multiple	1	1.36	0.03 – 7.58
Cardiovascular: 862 Total Cases				
Anomalous Pulmonary Venous Connection				
Total/partial anomalous pulmonary venous connection	Singleton	29	1.96	1.31 – 2.82
	Multiple	2	2.72	0.33 – 9.83

Table 8 Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2004-2005

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Atrioventricular Canal Defects				
ASD primum	Singleton	8	0.54	0.23 – 1.07
	Multiple	1	1.36	0.03 – 7.58
Common atrium	Singleton	5	0.34	0.11 – 0.79
	Multiple	0	0.00	0.00 – 5.02
Complete atrioventricular canal defect	Singleton	56	3.79	2.86 – 4.92
	Multiple	3	4.08	0.84 – 11.93
Endocardial cushion defect (OS and NOS)	Singleton	13	0.88	0.47 – 1.50
	Multiple	0	0.00	0.00 – 5.02
VSD, canal type	Singleton	11	0.74	0.37 – 1.33
	Multiple	0	0.00	0.00 – 5.02
Conotruncal (Outlet) and Aortic Arch				
Double outlet right ventricle	Singleton	27	1.83	1.20 – 2.66
	Multiple	2	2.72	0.33 – 9.83
d – Transposition of the great arteries	Singleton	36	2.43	1.70 – 3.37
	Multiple	0	0.00	0.00 – 5.02
Interrupted aortic arch, type B	Singleton	2	0.14	0.02 – 0.49
	Multiple	0	0.00	0.00 – 5.02
Tetralogy of Fallot w/ and w/o pulmonary atresia	Singleton	61	4.12	3.15 – 5.30
	Multiple	9	12.25	5.60 – 23.25
Truncus arteriosus	Singleton	3	0.20	0.04 – 0.59
	Multiple	0	0.00	0.00 – 5.02
Ebstein Anomaly				
Ebstein anomaly	Singleton	8	0.54	0.23 – 1.07
	Multiple	0	0.00	0.00 – 5.02
Laterality Defects				
Heterotaxy	Singleton	28	1.89	1.26 – 2.74
	Multiple	1	1.36	0.03 – 7.58

Table 8 Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2004-2005

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Left – Sided Obstruction				
Aortic valve stenosis	Singleton	21	1.42	0.88 – 2.17
	Multiple	2	2.72	0.33 – 9.83
Coarctation of aorta	Singleton	72	4.87	3.81 – 6.13
	Multiple	4	5.44	1.48 – 13.94
Hypoplastic left heart syndrome	Singleton	19	1.28	0.77 – 2.01
	Multiple	2	2.72	0.33 – 9.83
Interrupted aortic arch (type A and NOS)	Singleton	2	0.14	0.02 – 0.49
	Multiple	0	0.00	0.00 – 5.02
Patent Ductus Arteriosus				
Patent ductus arteriosus	Singleton	295	19.94	17.73 – 22.35
	Multiple	8	10.89	4.70 – 21.45
Right – Sided Obstruction				
Pulmonary stenosis, valvular	Singleton	83	5.61	4.47 – 6.96
	Multiple	9	12.25	5.60 – 23.25
Pulmonary valve atresia w/intact septum	Singleton	11	0.74	0.37 – 1.33
	Multiple	1	1.36	0.03 – 7.58
Pulmonary valve atresia with VSD	Singleton	8	0.54	0.23 – 1.07
	Multiple	0	0.00	0.00 – 5.02
Tricuspid valve atresia	Singleton	7	0.47	0.19 – 0.97
	Multiple	0	0.00	0.00 – 5.02
Septal Defects				
ASD (secundum and NOS)	Singleton	261	17.64	15.57 – 19.92
	Multiple	24	32.66	20.93 – 48.6
VSD (membranous and NOS)	Singleton	148	10.0	8.46 – 11.75
	Multiple	11	14.97	7.47 – 26.79
VSD, conoventricular/malalignment	Singleton	16	1.08	0.62 – 1.76
	Multiple	2	2.72	0.33 – 9.83
Single Ventricle and L – TGA				
L – TGA	Singleton	8	0.54	0.23 – 1.07
	Multiple	0	0.00	0.00 – 5.02

Table 8 Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2004-2005

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Single ventricle	Singleton	9	0.61	0.28 – 1.15
	Multiple	0	0.00	0.00 – 5.02
Other Cardiovascular				
Other cardiovascular	Singleton	279	18.86	16.71 – 21.21
	Multiple	21	28.58	17.69 – 43.69
Respiratory:76 Total Cases				
Choanal atresia	Singleton	10	0.68	0.32 – 1.24
	Multiple	0	0.00	0.00 – 5.02
Lung anomalies	Singleton	41	2.77	1.99 – 3.76
	Multiple	1	1.36	0.03 – 7.58
Other respiratory	Singleton	25	1.69	1.09 – 2.49
	Multiple	1	1.36	0.03 – 7.58
Orofacial: 279 Total Cases				
Cleft lip w/ and w/o cleft palate	Singleton	114	7.71	6.36 – 9.26
	Multiple	8	10.89	4.70 – 21.45
Cleft palate w/o cleft lip	Singleton	91	6.15	4.95 – 7.55
	Multiple	3	4.08	0.84 – 11.93
Pierre Robin sequence	Singleton	33	2.23	1.54 – 3.13
	Multiple	1	1.36	0.03 – 7.58
Other orofacial	Singleton	69	4.66	3.63 – 5.90
	Multiple	5	6.80	2.21 – 15.88
Gastrointestinal: 237 Total Cases				
Biliary atresia	Singleton	5	0.34	0.11 – 0.79
	Multiple	0	0.00	0.00 – 5.02
Esophageal atresia/tracheoesophageal fistula	Singleton	35	2.37	1.65 – 3.29
	Multiple	5	6.80	2.21 – 15.88
Hirschsprung disease	Singleton	34	2.30	1.59 – 3.21
	Multiple	1	1.36	0.03 – 7.58
Rectal and large intestinal atresia/stenosis	Singleton	41	2.77	1.99 – 3.76
	Multiple	6	8.17	3.00 – 17.77

Table 8 Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2004-2005

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Small intestinal atresia	Singleton	41	2.77	1.99 – 3.76
	Multiple	2	2.72	0.33 – 9.83
Other gastrointestinal	Singleton	85	5.75	4.59 – 7.10
	Multiple	6	8.17	3.00 – 17.77
Genitourinary: 541 Total Cases				
Bladder exstrophy	Singleton	4	0.27	0.07 – 0.69
	Multiple	0	0.00	0.00 – 5.02
Cloacal exstrophy	Singleton	5	0.34	0.11 – 0.79
	Multiple	0	0.00	0.00 – 5.02
Hypospadias, 2nd or 3rd degree	Singleton	197	13.32	11.52 – 15.31
	Multiple	15	20.41	11.43 – 33.67
Obstructive genitourinary defect	Singleton	161	10.88	9.27 – 12.7
	Multiple	6	8.17	3.00 – 17.77
Renal agenesis/hypoplasia	Singleton	5	0.34	0.11 – 0.79
	Multiple	2	2.72	0.33 – 9.83
Other genitourinary	Singleton	228	15.41	13.48 – 17.55
	Multiple	11	14.97	7.47 – 26.79
Musculoskeletal: 696 Total Cases				
Clubfoot	Singleton	171	11.56	9.89 – 13.43
	Multiple	15	20.41	11.43 – 33.67
Craniosynostosis	Singleton	65	4.39	3.39 – 5.60
	Multiple	3	4.08	0.84 – 11.93
Diaphragmatic hernia	Singleton	44	2.97	2.16 – 3.99
	Multiple	1	1.36	0.03 – 7.58
Gastroschisis	Singleton	48	3.24	2.39 – 4.30
	Multiple	0	0.00	0.00 – 5.02
Omphalocele	Singleton	12	0.81	0.42 – 1.42
	Multiple	2	2.72	0.33 – 9.83
Polydactyly/syndactyly	Singleton	175	11.83	10.14 – 13.72
	Multiple	10	13.61	6.53 – 25.03
Reduction deformity, lower limbs	Singleton	19	1.28	0.77 – 2.01
	Multiple	3	4.08	0.84 – 11.93

Table 8 Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2004-2005

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Reduction deformity, upper limbs	Singleton	44	2.97	2.16 – 3.99
	Multiple	4	5.44	1.48 – 13.94
Skeletal dysplasia	Singleton	24	1.62	1.04 – 2.41
	Multiple	0	0.00	0.00 – 5.02
Other musculoskeletal	Singleton	162	10.95	9.33 – 12.77
	Multiple	12	16.33	8.44 – 28.53
Chromosomal and other Syndromes: 413 Total Cases				
Klinefelter syndrome	Singleton	9	0.61	0.28 – 1.15
	Multiple	0	0.00	0.00 – 5.02
Trisomy 13	Singleton	11	0.74	0.37 – 1.33
	Multiple	0	0.00	0.00 – 5.02
Trisomy 18	Singleton	19	1.28	0.77 – 2.01
	Multiple	3	4.08	0.84 – 11.93
Trisomy 21 (Down syndrome)	Singleton	174	11.76	10.08 – 13.65
	Multiple	10	13.61	6.53 – 25.03
Turner syndrome	Singleton	16	1.08	0.62 – 1.76
	Multiple	0	0.00	0.00 – 5.02
Other chromosomal syndromes/other syndromes	Singleton	156	10.55	8.96 – 12.34
	Multiple	18	24.5	14.52 – 38.72
Other: 49 Total Cases				
Amniotic bands	Singleton	14	0.95	0.52 – 1.59
	Multiple	1	1.36	0.03 – 7.58
Skin anomalies	Singleton	6	0.41	0.15 – 0.88
	Multiple	2	2.72	0.33 – 9.83
Other, specified	Singleton	25	1.69	1.09 – 2.49
	Multiple	2	2.72	0.33 – 9.83

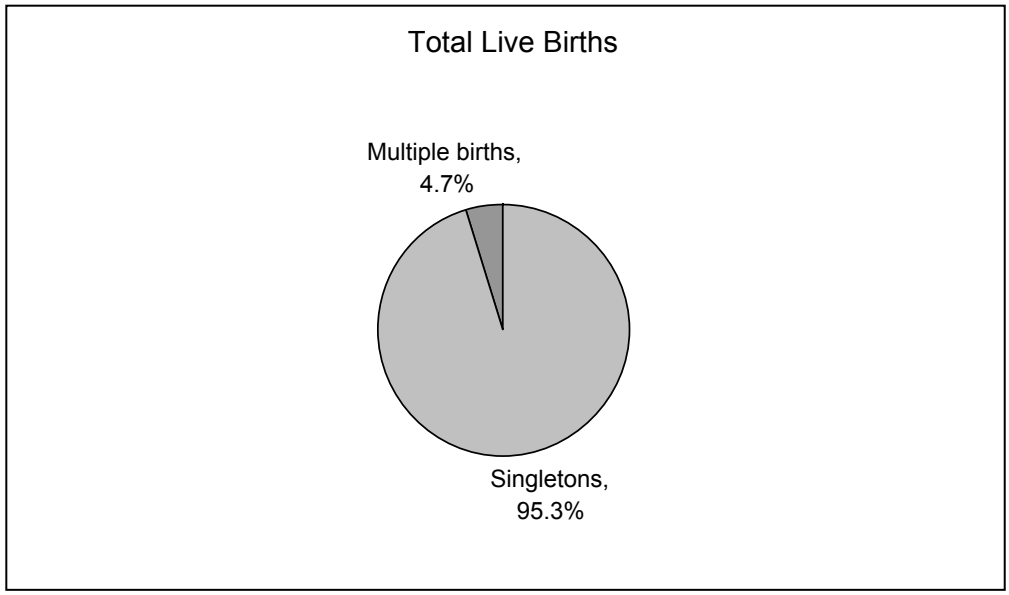
1. Plurality is the number of births to a woman from the same pregnancy. A singleton is the birth of one infant; multiple represents more than one infant.
2. Cases can be included in the count for more than one defect. Cases are counted once in the total for a defect category.

Table 9 Most Common Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2004-2005

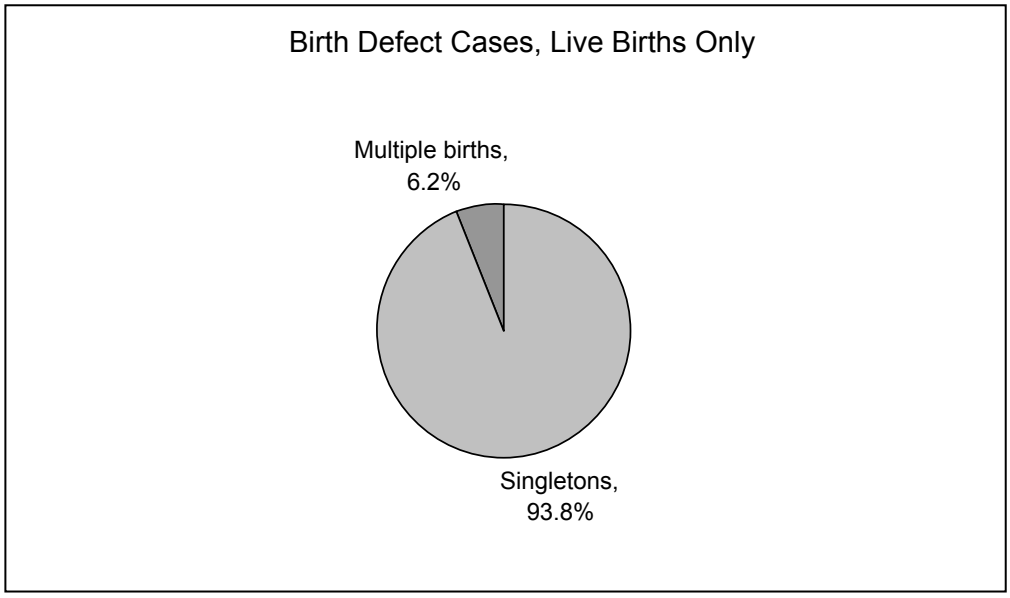
Defect[2]	Count	Rate per 10,000 Births	95% Confidence Interval
MULTIPLE			
ASD (secundum and NOS)	24	32.66	20.93 – 48.60
Hypospadias, 2nd or 3rd degree	15	20.41	11.43 – 33.67
Clubfoot	15	20.41	11.43 – 33.67
VSD (membranous and NOS)	11	14.97	7.47 – 26.79
Trisomy 21 (Down syndrome)	10	13.61	6.53 – 25.03
Polydactyly/syndactyly	10	13.61	6.53 – 25.03
Pulmonary stenosis, valvular	9	12.25	5.60 – 23.25
Tetralogy of Fallot w/ and w/o pulmonary atresia	9	12.25	5.60 – 23.25
Cleft lip w/ and w/o cleft palate	8	10.89	4.70 – 21.45
Rectal and large intestinal atresia/stenosis	6	8.17	3.00 – 17.77
SINGLETON			
ASD (secundum and NOS)	261	17.64	15.57 – 19.92
Hypospadias, 2nd or 3rd degree	197	13.32	11.52 – 15.31
Polydactyly/syndactyly	175	11.83	10.14 – 13.72
Trisomy 21 (Down syndrome)	174	11.76	10.08 – 13.65
Clubfoot	171	11.56	9.89 – 13.43
Obstructive genitourinary defect	161	10.88	9.27 – 12.70
VSD (membranous and NOS)	148	10.0	8.46 – 11.75
Cleft lip w/ and w/o cleft palate	114	7.71	6.36 – 9.26
Cleft palate w/o cleft lip	91	6.15	4.95 – 7.55
Pulmonary stenosis, valvular	83	5.61	4.47 – 6.96

1. Plurality is the number of births to a woman from the same pregnancy. A singleton is the birth of one infant; multiple represents more than one infant.
2. Excludes patent ductus arteriosus (PDA) due to the high number of cases and the mild severity of the majority of these cases.

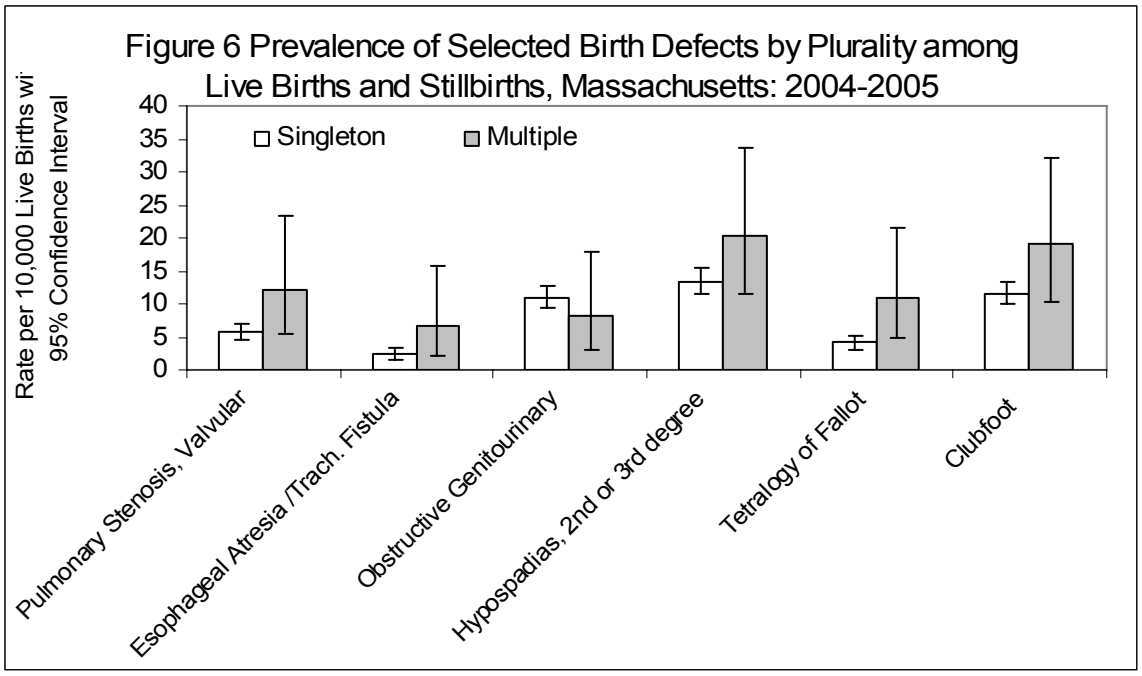
Figure 5 Plurality of All Live Births and Birth Defect Cases, Live Births Only, Massachusetts: 2004 – 2005



N=155,284



N=2,536



Number of Singleton cases = 2,431; rate = 164.3/10,000 births
Number of Multiple cases = 159; rate: 216.4

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Age Group (yrs)	Count	Rate per 10,000 Births	95% Confidence Interval
Central Nervous System				
Anencephaly	<20	4	4.35	1.18 – 11.13
	20 – 24	2	0.85	0.10 – 3.08
	25 – 29	0	0.00	0.00 – 1.03
	30 – 34	1	0.20	0.01 – 1.12
	35+	1	0.27	0.01 – 1.52
Encephalocele	<20	0	0.00	0.00 – 4.01
	20 – 24	0	0.00	0.00 – 1.57
	25 – 29	1	0.28	0.01 – 1.55
	30 – 34	1	0.20	0.01 – 1.12
	35+	1	0.27	0.01 – 1.52
Holoprosencephaly	<20	0	0.00	0.00 – 4.01
	20 – 24	1	0.43	0.01 – 2.37
	25 – 29	2	0.56	0.07 – 2.01
	30 – 34	3	0.60	0.12 – 1.76
	35+	0	0.00	0.00—1.00
Hydrocephaly w/o spina bifida	<20	2	2.17	0.26 – 7.85
	20 – 24	6	2.56	0.94 – 5.56
	25 – 29	14	3.90	2.13 – 6.54
	30 – 34	11	2.20	1.10 – 3.94
	35+	11	3.00	1.50 – 5.36
Microcephaly	<20	4	4.35	1.18 – 11.13
	20 – 24	9	3.83	1.75 – 7.28
	25 – 29	8	2.23	0.96 – 4.39
	30 – 34	4	0.80	0.22 – 2.05
	35+	6	1.63	0.60 – 3.56
Spina bifida w/ and w/o hydrocephaly	<20	1	1.09	0.03 – 6.06
	20 – 24	2	0.85	0.10 – 3.08
	25 – 29	11	3.06	1.53 – 5.48
	30 – 34	6	1.20	0.44 – 2.61
	35+	5	1.36	0.44 – 3.18

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Age Group (yrs)	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Spinal cord	<20	2	2.17	0.26 – 7.85
	20 – 24	4	1.70	0.46 – 4.36
	25 – 29	16	4.45	2.54 – 7.23
	30 – 34	16	3.20	1.83 – 5.20
	35+	7	1.91	0.77 – 3.93
Other CNS	<20	9	9.78	4.47 – 18.57
	20 – 24	16	6.82	3.90 – 11.07
	25 – 29	26	7.23	4.73 – 10.60
	30 – 34	28	5.61	3.73 – 8.10
	35+	22	5.99	3.75 – 9.07
Eye				
Anophthalmia/microphthalmia	<20	1	1.09	0.03 – 6.06
	20 – 24	2	0.85	0.10 – 3.08
	25 – 29	1	0.28	0.01 – 1.55
	30 – 34	0	0.00	0.00 – 0.74
	35+	3	0.82	0.17 – 2.39
Congenital glaucoma, congenital cataract	<20	4	4.35	1.18 – 11.13
	20 – 24	7	2.98	1.20 – 6.14
	25 – 29	14	3.90	2.13 – 6.54
	30 – 34	10	2.00	0.96 – 3.68
	35+	11	3.00	1.50 – 5.36
Other eye	<20	3	3.26	0.67 – 9.53
	20 – 24	6	2.56	0.94 – 5.56
	25 – 29	9	2.50	1.15 – 4.75
	30 – 34	6	1.20	0.44 – 2.61
	35+	10	2.72	1.31 – 5.01
Ear				
Anotia/microtia	<20	3	3.26	0.67 – 9.53
	20 – 24	4	1.70	0.46 – 4.36
	25 – 29	5	1.39	0.45 – 3.25
	30 – 34	7	1.40	0.56 – 2.89
	35+	8	2.18	0.94 – 4.29

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Age Group (yrs)	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Other ear	<20	1	1.09	0.03 – 6.06
	20 – 24	1	0.43	0.01 – 2.37
	25 – 29	4	1.11	0.30 – 2.85
	30 – 34	10	2.00	0.96 – 3.68
	35+	8	2.18	0.94 – 4.29
Cardiovascular				
Atrioventricular Canal Defects				
Total/partial anomalous pulmonary venous connection	<20	1	1.09	0.03 – 6.06
	20 – 24	5	2.13	0.69 – 4.97
	25 – 29	7	1.95	0.78 – 4.01
	30 – 34	12	2.40	1.24 – 4.20
	35+	6	1.63	0.60 – 3.56
Atrioventricular Canal Defects				
ASD primum	<20	0	0.00	0.00 – 4.01
	20 – 24	3	1.28	0.26 – 3.73
	25 – 29	2	0.56	0.07 – 2.01
	30 – 34	2	0.40	0.05 – 1.45
	35+	2	0.54	0.07 – 1.97
Common atrium	<20	0	0.00	0.00 – 4.01
	20 – 24	2	0.85	0.10 – 3.08
	25 – 29	1	0.28	0.01 – 1.55
	30 – 34	2	0.40	0.05 – 1.45
	35+	0	0.00	0.00—1.00
Complete atrioventricular canal defect	<20	2	2.17	0.26 – 7.85
	20 – 24	9	3.83	1.75 – 7.28
	25 – 29	5	1.39	0.45 – 3.25
	30 – 34	25	5.01	3.24 – 7.39
	35+	16	4.36	2.49 – 7.08

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Age Group (yrs)	Count	Rate per 10,000 Births	95% Confidence Interval
(*cont'd*)				
Endocardial cushion defect (OS and NOS)	<20	0	0.00	0.00 – 4.01
	20 – 24	2	0.85	0.10 – 3.08
	25 – 29	1	0.28	0.01 – 1.55
	30 – 34	6	1.20	0.44 – 2.61
	35+	3	0.82	0.17 – 2.39
VSD, canal type	<20	0	0.00	0.00 – 4.01
	20 – 24	3	1.28	0.26 – 3.73
	25 – 29	1	0.28	0.01 – 1.55
	30 – 34	1	0.20	0.01 – 1.12
	35+	5	1.36	0.44 – 3.18
Conotruncal (Outlet) and Aortic Arch				
Double outlet right ventricle	<20	0	0.00	0.00 – 4.01
	20 – 24	7	2.98	1.20 – 6.14
	25 – 29	4	1.11	0.30 – 2.85
	30 – 34	10	2.00	0.96 – 3.68
	35+	8	2.18	0.94 – 4.29
d – Transposition of the great arteries	<20	2	2.17	0.26 – 7.85
	20 – 24	6	2.56	0.94 – 5.56
	25 – 29	8	2.23	0.96 – 4.39
	30 – 34	10	2.00	0.96 – 3.68
	35+	9	2.45	1.12 – 4.65
Interrupted aortic arch, type B	<20	0	0.00	0.00 – 4.01
	20 – 24	0	0.00	0.00 – 1.57
	25 – 29	0	0.00	0.00 – 1.03
	30 – 34	1	0.20	0.01 – 1.12
	35+	1	0.27	0.01 – 1.52
Tetralogy of Fallot w/ and w/o pulmonary atresia	<20	7	7.61	3.06 – 15.68
	20 – 24	10	4.26	2.04 – 7.83
	25 – 29	13	3.62	1.93 – 6.19
	30 – 34	19	3.80	2.29 – 5.94
	35+	21	5.72	3.54 – 8.74

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Age Group (yrs)	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Truncus arteriosus	<20	0	0.00	0.00 – 4.01
	20 – 24	0	0.00	0.00 – 1.57
	25 – 29	1	0.28	0.01 – 1.55
	30 – 34	2	0.40	0.05 – 1.45
	35+	0	0.00	0.00—1.00
Ebstein Anomaly				
Ebstein anomaly	<20	1	1.09	0.03 – 6.06
	20 – 24	0	0.00	0.00 – 1.57
	25 – 29	3	0.83	0.17 – 2.44
	30 – 34	3	0.6	0.12 – 1.76
	35+	1	0.27	0.01 – 1.52
Laterality Defects				
Heterotaxy	<20	0	0.00	0.00 – 4.01
	20 – 24	5	2.13	0.69 – 4.97
	25 – 29	9	2.50	1.15 – 4.75
	30 – 34	10	2.00	0.96 – 3.68
	35+	5	1.36	0.44 – 3.18
Left – Sided Obstruction				
Aortic valve stenosis	<20	2	2.17	0.26 – 7.85
	20 – 24	0	0.00	0.00 – 1.57
	25 – 29	7	1.95	0.78 – 4.01
	30 – 34	10	2.00	0.96 – 3.68
	35+	4	1.09	0.30 – 2.79
Coarctation of aorta	<20	5	5.44	1.76 – 12.68
	20 – 24	11	4.69	2.34 – 8.38
	25 – 29	14	3.90	2.13 – 6.54
	30 – 34	31	6.21	4.22 – 8.81
	35+	15	4.08	2.29 – 6.74

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Age Group (yrs)	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Hypoplastic left heart syndrome	<20	5	5.44	1.76 – 12.68
	20 – 24	1	0.43	0.01 – 2.37
	25 – 29	4	1.11	0.30 – 2.85
	30 – 34	7	1.40	0.56 – 2.89
	35+	3	0.82	0.17 – 2.39
Interrupted aortic arch (type A and NOS)	<20	1	1.09	0.03 – 6.06
	20 – 24	1	0.43	0.01 – 2.37
	25 – 29	0	0.00	0.00 – 1.03
	30 – 34	0	0.00	0.00 – 0.74
	35+	0	0.00	0.00—1.00
Patent Ductus Arteriosus				
Patent ductus arteriosus	<20	17	18.48	10.77 – 29.59
	20 – 24	52	22.15	16.54 – 29.05
	25 – 29	49	13.63	10.09 – 18.02
	30 – 34	92	18.42	14.85 – 22.59
	35+	93	25.33	20.44 – 31.03
Right – Sided Obstruction				
Pulmonary stenosis, valvular	<20	5	5.44	1.76 – 12.68
	20 – 24	12	5.11	2.64 – 8.93
	25 – 29	16	4.45	2.54 – 7.23
	30 – 34	27	5.41	3.56 – 7.87
	35+	32	8.71	5.96 – 12.30
Pulmonary valve atresia w/intact septum	<20	1	1.09	0.03 – 6.06
	20 – 24	3	1.28	0.26 – 3.73
	25 – 29	1	0.28	0.01 – 1.55
	30 – 34	5	1.00	0.33 – 2.34
	35+	1	0.27	0.01 – 1.52
Pulmonary valve atresia with VSD	<20	0	0.00	0.00 – 4.01
	20 – 24	1	0.43	0.01 – 2.37
	25 – 29	1	0.28	0.01 – 1.55
	30 – 34	5	1.00	0.33 – 2.34
	35+	1	0.27	0.01 – 1.52

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Age Group (yrs)	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Tricuspid valve atresia	<20	0	0.00	0.00 – 4.01
	20 – 24	0	0.00	0.00 – 1.57
	25 – 29	1	0.28	0.01 – 1.55
	30 – 34	2	0.40	0.05 – 1.45
	35+	2	0.54	0.07 – 1.97
Septal Defects				
ASD (secundum and NOS)	<20	14	15.22	8.32 – 25.53
	20 – 24	41	17.47	12.53 – 23.69
	25 – 29	59	16.42	12.50 – 21.18
	30 – 34	88	17.62	14.13 – 21.71
	35+	82	22.33	17.76 – 27.72
VSD (membranous and NOS)	<20	4	4.35	1.18 – 11.13
	20 – 24	23	9.80	6.21 – 14.70
	25 – 29	32	8.90	6.09 – 12.57
	30 – 34	47	9.41	6.91 – 12.51
	35+	44	11.98	8.71 – 16.09
VSD, conoventricular/malalignment	<20	1	1.09	0.03 – 6.06
	20 – 24	2	0.85	0.10 – 3.08
	25 – 29	8	2.23	0.96 – 4.39
	30 – 34	2	0.40	0.05 – 1.45
	35+	5	1.36	0.44 – 3.18
Single Ventricle and L – TGA				
L – TGA	<20	2	2.17	0.26 – 7.85
	20 – 24	0	0.00	0.00 – 1.57
	25 – 29	1	0.28	0.01 – 1.55
	30 – 34	3	0.60	0.12 – 1.76
	35+	2	0.54	0.07 – 1.97
Single ventricle	<20	1	1.09	0.03 – 6.06
	20 – 24	2	0.85	0.10 – 3.08
	25 – 29	0	0.00	0.00 – 1.03
	30 – 34	3	0.60	0.12 – 1.76
	35+	3	0.82	0.17 – 2.39

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Age Group (yrs)	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Other Cardiovascular				
Other cardiovascular	<20	17	18.48	10.77 – 29.59
	20 – 24	41	17.47	12.53 – 23.69
	25 – 29	61	16.97	12.98 – 21.80
	30 – 34	103	20.62	16.83 – 25.01
	35+	75	20.42	16.07 – 25.60
Respiratory				
Choanal atresia	<20	2	2.17	0.26 – 7.85
	20 – 24	1	0.43	0.01 – 2.37
	25 – 29	2	0.56	0.07 – 2.01
	30 – 34	4	0.80	0.22 – 2.05
	35+	1	0.27	0.01 – 1.52
Lung anomalies	<20	2	2.17	0.26 – 7.85
	20 – 24	7	2.98	1.20 – 6.14
	25 – 29	14	3.90	2.13 – 6.54
	30 – 34	11	2.20	1.10 – 3.94
	35+	6	1.63	0.60 – 3.56
Other respiratory	<20	1	1.09	0.03 – 6.06
	20 – 24	7	2.98	1.20 – 6.14
	25 – 29	3	0.83	0.17 – 2.44
	30 – 34	9	1.80	0.82 – 3.42
	35+	5	1.36	0.44 – 3.18
Orofacial				
Cleft lip w/ and w/o cleft palate	<20	5	5.44	1.76 – 12.68
	20 – 24	18	7.67	4.54 – 12.12
	25 – 29	33	9.18	6.32 – 12.89
	30 – 34	33	6.61	4.55 – 9.28
	35+	29	7.90	5.29 – 11.34

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Age Group (yrs)	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Cleft palate w/o cleft lip	<20	3	3.26	0.67 – 9.53
	20 – 24	12	5.11	2.64 – 8.93
	25 – 29	27	7.51	4.95 – 10.93
	30 – 34	30	6.01	4.05 – 8.57
	35+	21	5.72	3.54 – 8.74
Pierre Robin sequence	<20	0	0.00	0.00 – 4.01
	20 – 24	2	0.85	0.10 – 3.08
	25 – 29	9	2.50	1.15 – 4.75
	30 – 34	14	2.80	1.53 – 4.70
	35+	9	2.45	1.12 – 4.65
Other orofacial	<20	4	4.35	1.18 – 11.13
	20 – 24	16	6.82	3.90 – 11.07
	25 – 29	16	4.45	2.54 – 7.23
	30 – 34	19	3.80	2.29 – 5.94
	35+	17	4.63	2.70 – 7.41
Gastrointestinal				
Biliary atresia	<20	0	0.00	0.00 – 4.01
	20 – 24	2	0.85	0.10 – 3.08
	25 – 29	1	0.28	0.01 – 1.55
	30 – 34	2	0.40	0.05 – 1.45
	35+	0	0.00	0.00—1.00
Esophageal atresia/tracheoesophageal fistula	<20	1	1.09	0.03 – 6.06
	20 – 24	5	2.13	0.69 – 4.97
	25 – 29	8	2.23	0.96 – 4.39
	30 – 34	13	2.60	1.39 – 4.45
	35+	11	3.00	1.50 – 5.36
Hirschsprung disease	<20	1	1.09	0.03 – 6.06
	20 – 24	8	3.41	1.47 – 6.71
	25 – 29	5	1.39	0.45 – 3.25
	30 – 34	14	2.80	1.53 – 4.70
	35+	7	1.91	0.77 – 3.93

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Age Group (yrs)	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Rectal and large intestinal atresia/stenosis	<20	5	5.44	1.76 – 12.68
	20 – 24	4	1.70	0.46 – 4.36
	25 – 29	10	2.78	1.33 – 5.12
	30 – 34	17	3.40	1.98 – 5.45
	35+	10	2.72	1.31 – 5.01
Small intestinal atresia	<20	4	4.35	1.18 – 11.13
	20 – 24	7	2.98	1.20 – 6.14
	25 – 29	12	3.34	1.73 – 5.83
	30 – 34	12	2.40	1.24 – 4.20
	35+	7	1.91	0.77 – 3.93
Other gastrointestinal	<20	3	3.26	0.67 – 9.53
	20 – 24	13	5.54	2.95 – 9.47
	25 – 29	23	6.40	4.06 – 9.60
	30 – 34	29	5.81	3.89 – 8.34
	35+	21	5.72	3.54 – 8.74
Genitourinary				
Bladder exstrophy	<20	0	0.00	0.00 – 4.01
	20 – 24	2	0.85	0.10 – 3.08
	25 – 29	0	0.00	0.00 – 1.03
	30 – 34	1	0.20	0.01 – 1.12
	35+	1	0.27	0.01 – 1.52
Cloacal exstrophy	<20	0	0.00	0.00 – 4.01
	20 – 24	0	0.00	0.00 – 1.57
	25 – 29	1	0.28	0.01 – 1.55
	30 – 34	3	0.60	0.12 – 1.76
	35+	1	0.27	0.01 – 1.52
Hypospadias, 2nd or 3rd degree	<20	7	7.61	3.06 – 15.68
	20 – 24	35	14.91	10.39 – 20.74
	25 – 29	42	11.69	8.42 – 15.80
	30 – 34	75	15.02	11.81 – 18.82
	35+	52	14.16	10.58 – 18.57

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Age Group (yrs)	Count	Rate per 10,000 Births	95% Confidence Interval
(*cont'd*)				
Obstructive genitourinary defect	<20	3	3.26	0.67 – 9.53
	20 – 24	29	12.35	8.27 – 17.74
	25 – 29	36	10.02	7.02 – 13.87
	30 – 34	52	10.41	7.78 – 13.65
	35+	47	12.80	9.40 – 17.02
Renal agenesis/hypoplasia	<20	1	1.09	0.03 – 6.06
	20 – 24	1	0.43	0.01 – 2.37
	25 – 29	0	0.00	0.00 – 1.03
	30 – 34	2	0.40	0.05 – 1.45
	35+	2	0.54	0.07 – 1.97
Other genitourinary	<20	13	14.13	7.52 – 24.17
	20 – 24	31	13.21	8.97 – 18.74
	25 – 29	46	12.80	9.37 – 17.07
	30 – 34	74	14.82	11.63 – 18.60
	35+	72	19.61	15.34 – 24.69
Musculoskeletal				
Clubfoot	<20	9	9.78	4.47 – 18.57
	20 – 24	37	15.76	11.1 – 21.73
	25 – 29	41	11.41	8.19 – 15.48
	30 – 34	56	11.21	8.47 – 14.56
	35+	37	10.08	7.09 – 13.89
Craniosynostosis	<20	1	1.09	0.03 – 6.06
	20 – 24	7	2.98	1.20 – 6.14
	25 – 29	14	3.90	2.13 – 6.54
	30 – 34	27	5.41	3.56 – 7.87
	35+	19	5.17	3.12 – 8.08
Diaphragmatic hernia	<20	5	5.44	1.76 – 12.68
	20 – 24	4	1.70	0.46 – 4.36
	25 – 29	4	1.11	0.30 – 2.85
	30 – 34	17	3.40	1.98 – 5.45
	35+	12	3.27	1.69 – 5.71

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Age Group (yrs)	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Gastroschisis	<20	13	14.13	7.52 – 24.17
	20 – 24	16	6.82	3.90 – 11.07
	25 – 29	8	2.23	0.96 – 4.39
	30 – 34	4	0.80	0.22 – 2.05
	35+	1	0.27	0.01 – 1.52
Omphalocele	<20	0	0.00	0.00 – 4.01
	20 – 24	2	0.85	0.10 – 3.08
	25 – 29	2	0.56	0.07 – 2.01
	30 – 34	4	0.80	0.22 – 2.05
	35+	4	1.09	0.30 – 2.79
Polydactyly/syndactyly	<20	12	13.04	6.74 – 22.79
	20 – 24	24	10.22	6.55 – 15.21
	25 – 29	41	11.41	8.19 – 15.48
	30 – 34	65	13.01	10.04 – 16.59
	35+	41	11.17	8.01 – 15.15
Reduction deformity, lower limbs	<20	2	2.17	0.26 – 7.85
	20 – 24	4	1.70	0.46 – 4.36
	25 – 29	3	0.83	0.17 – 2.44
	30 – 34	11	2.20	1.10 – 3.94
	35+	1	0.27	0.01 – 1.52
Reduction deformity, upper limbs	<20	1	1.09	0.03 – 6.06
	20 – 24	12	5.11	2.64 – 8.93
	25 – 29	6	1.67	0.61 – 3.63
	30 – 34	18	3.60	2.14 – 5.70
	35+	10	2.72	1.31 – 5.01
Skeletal dysplasia	<20	1	1.09	0.03 – 6.06
	20 – 24	5	2.13	0.69 – 4.97
	25 – 29	7	1.95	0.78 – 4.01
	30 – 34	5	1.00	0.33 – 2.34
	35+	5	1.36	0.44 – 3.18

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2004-2005

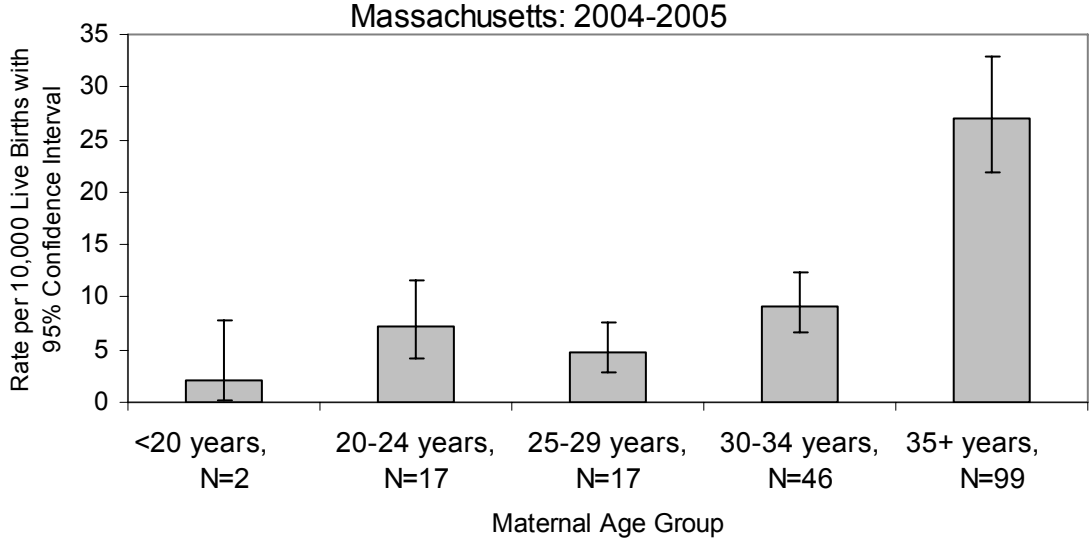
Defect[1]	Maternal Age Group (yrs)	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Other musculoskeletal	<20	8	8.70	3.75 – 17.14
	20 – 24	31	13.21	8.97 – 18.74
	25 – 29	41	11.41	8.19 – 15.48
	30 – 34	50	10.01	7.43 – 13.20
	35+	38	10.35	7.32 – 14.20
Chromosomal and other Syndromes				
Klinefelter syndrome	<20	0	0.00	0.00 – 4.01
	20 – 24	0	0.00	0.00 – 1.57
	25 – 29	2	0.56	0.07 – 2.01
	30 – 34	2	0.40	0.05 – 1.45
	35+	5	1.36	0.44 – 3.18
Trisomy 13	<20	0	0.00	0.00 – 4.01
	20 – 24	2	0.85	0.10 – 3.08
	25 – 29	3	0.83	0.17 – 2.44
	30 – 34	2	0.40	0.05 – 1.45
	35+	3	0.82	0.17 – 2.39
Trisomy 18	<20	1	1.09	0.03 – 6.06
	20 – 24	0	0.00	0.00 – 1.57
	25 – 29	1	0.28	0.01 – 1.55
	30 – 34	1	0.20	0.01 – 1.12
	35+	9	2.45	1.12 – 4.65
Trisomy 21 (Down syndrome)	<20	2	2.17	0.26 – 7.85
	20 – 24	17	7.24	4.22 – 11.59
	25 – 29	17	4.73	2.76 – 7.57
	30 – 34	46	9.21	6.74 – 12.29
	35+	99	26.96	21.91 – 32.82
Turner syndrome	<20	0	0.00	0.00 – 4.01
	20 – 24	1	0.43	0.01 – 2.37
	25 – 29	0	0.00	0.00 – 1.03
	30 – 34	4	0.80	0.22 – 2.05
	35+	8	2.18	0.94 – 4.29

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Age Group (yrs)	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Other chromosomal syndromes/other syndromes	<20	6	6.52	2.39 – 14.20
	20 – 24	28	11.93	7.93 – 17.24
	25 – 29	30	8.35	5.63 – 11.92
	30 – 34	58	11.61	8.82 – 15.01
	35+	47	12.80	9.40 – 17.02
Other				
Amniotic bands	<20	2	2.17	0.26 – 7.85
	20 – 24	4	1.70	0.46 – 4.36
	25 – 29	2	0.56	0.07 – 2.01
	30 – 34	3	0.60	0.12 – 1.76
	35+	3	0.82	0.17 – 2.39
Skin anomalies	<20	0	0.00	0.00 – 4.01
	20 – 24	1	0.43	0.01 – 2.37
	25 – 29	3	0.83	0.17 – 2.44
	30 – 34	3	0.60	0.12 – 1.76
	35+	1	0.27	0.01 – 1.52
Other, specified	<20	1	1.09	0.03 – 6.06
	20 – 24	5	2.13	0.69 – 4.97
	25 – 29	5	1.39	0.45 – 3.25
	30 – 34	10	2.00	0.96 – 3.68
	35+	6	1.63	0.60 – 3.56

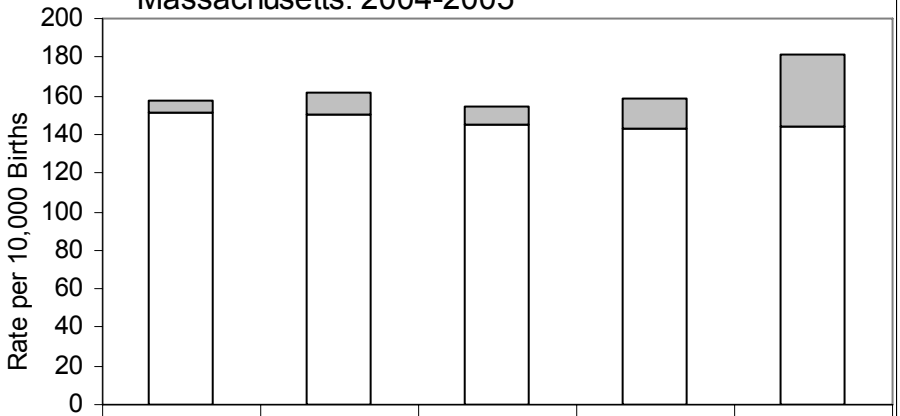
1. Cases can be included in the count for more than one defect. Cases are counted once in the total for a defect category.

Figure 7 Rates of Down Syndrome among Maternal Age Groups, Massachusetts: 2004-2005



N=181, live births only

Figure 8 Chromosomal and All Other Defects by Maternal Age, Massachusetts: 2004-2005



Maternal Age Group	<20	20-24	25-29	30-34	35+
Chromosomal defects	6.5 (2.4-14.2)	11.1 (7.2-16.2)	9.7 (6.8-13.5)	15.2 (12.0-19.1)	37.0 (31.1-43.8)
All other defects	151.1 (127.0-178.4)	150.4 (135.2-166.8)	145.0 (132.9-157.9)	143.2 (132.9-154.0)	144.1 (132.1-156.8)

N=2536, live births only

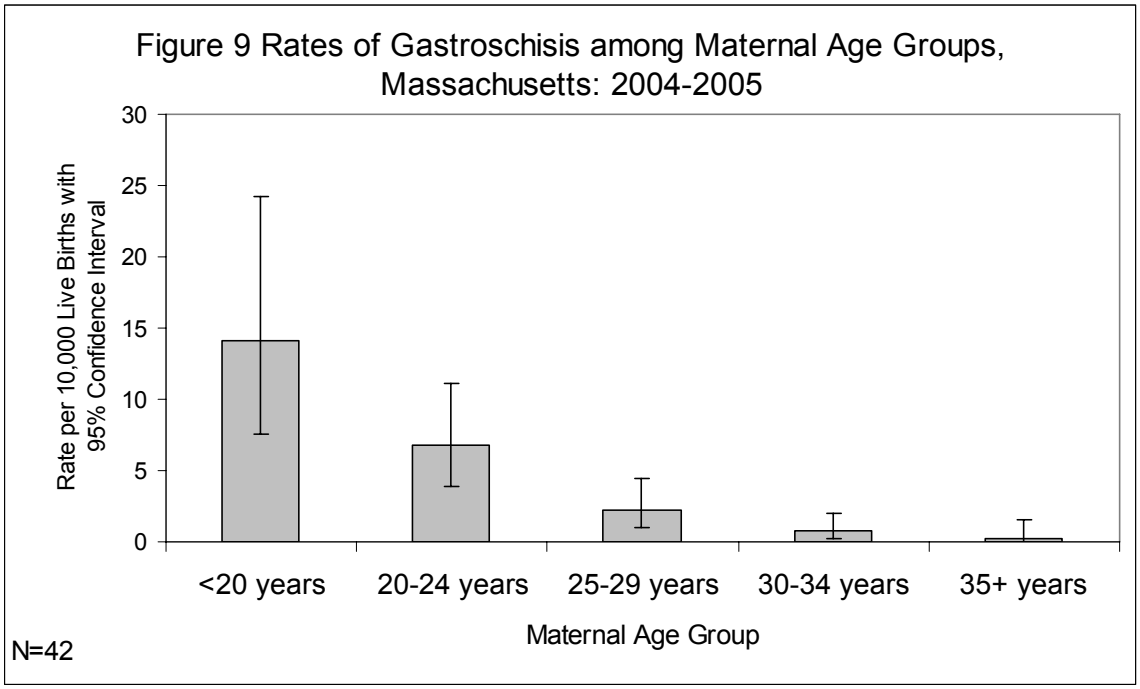


Figure 9 Rates of Gastroschisis among Maternal Age Groups, Massachusetts: 2004-2005
Rate per 10,000 Live Births with 95% Confidence Interval
30
25
20
15
10
5
0
<20 years
20-24 years
25-29 years
30-34 years
35+ years
Maternal Age Group
N=42

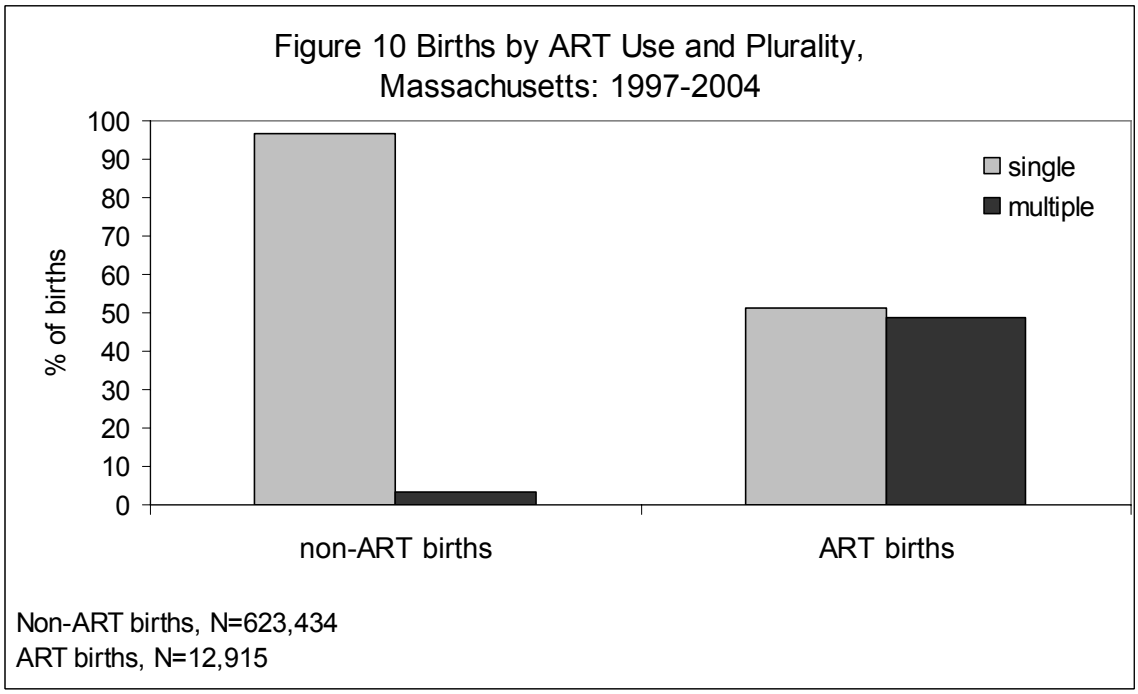


Figure 10 Births by ART Use and Plurality, Massachusetts: 1997-2004
% of births
100
90
80
70
60
50
40
30
20
10
0
single
multiple
non-ART births
ART births
Non-ART births, N=623,434
ART births, N=12,915

Table 11 Most Common Defects by Maternal Age Group for Live Births, Massachusetts: 2004-2005

Age Group (yrs)	Defect[1]	Count	Rate per 10,000 Births	95% Confidence Interval
<20	ASD (secundum and NOS)	14	15.22	8.32 – 25.53
	Gastroschisis	13	14.13	7.52 – 24.17
	Polydactyly/syndactyly	12	13.04	6.74 – 22.79
	Clubfoot	9	9.78	4.47 – 18.57
	Tetralogy of Fallot w/ and w/o pulmonary atresia	7	7.61	3.06 – 15.68
20 – 24	ASD (secundum and NOS)	41	17.47	12.53 – 23.69
	Clubfoot	37	15.76	11.10 – 21.73
	Hypospadias, 2nd or 3rd degree	35	14.91	10.39 – 20.74
	Obstructive genitourinary defect	29	12.35	8.27 – 17.74
	Polydactyly/syndactyly	24	10.22	6.55 – 15.21
25 – 29	ASD (secundum and NOS)	59	16.42	12.50 – 21.18
	Hypospadias, 2nd or 3rd degree	42	11.69	8.42 – 15.80
	Polydactyly/syndactyly	41	11.41	8.19 – 15.48
	Clubfoot	41	11.41	8.19 – 15.48
	Obstructive genitourinary defect	36	10.02	7.02 – 13.87
30 – 34	ASD (secundum and NOS)	88	17.62	14.13 – 21.71
	Hypospadias, 2nd or 3rd degree	75	15.02	11.81 – 18.82
	Polydactyly/syndactyly	65	13.01	10.04 – 16.59
	Clubfoot	56	11.21	8.47 – 14.56
	Obstructive genitourinary defect	52	10.41	7.78 – 13.65
35+	Trisomy 21 (Down syndrome)	99	26.96	21.91 – 32.82
	ASD (secundum and NOS)	82	22.33	17.76 – 27.72
	Hypospadias, 2nd or 3rd degree	52	14.16	10.58 – 18.57
	Obstructive genitourinary defect	47	12.80	9.40 – 17.02
	VSD (membranous and NOS)	44	11.98	8.71 – 16.09

1. Excludes patent ductus arteriosus (PDA) due to the high number of cases and the mild severity of the majority of these cases.

Chapter 5

Prevalence of Birth Defects by Race / Ethnicity and Region

Baby with encephalocele

Courtesy of the Centers for Disease Control and Prevention

Maternal Race / Hispanic Ethnicity

Table 12 shows the variation in prevalence of birth defects by maternal race and Hispanic ethnicity. The rate per 10,000 live births was 161.9 (154.5-169.6) for Non-Hispanic Whites, 180.5 (157.4-206.1) for Non-Hispanic Blacks, 142.0 (120.3-166.4) for Non-Hispanic Asians/Pacific Islanders and 166.7 (149.3-185.5) for Hispanics. In some analyses, the rates for other races were not calculated due to small numbers or the rates for other races were combined as one "other" category.

Table 13 shows the more common defects by maternal race and Hispanic ethnicity. The most common defects in Hispanics included Down syndrome, septal defects, clubfoot, cleft lip with and without cleft palate and obstructive genitourinary defects. In Blacks, the most common defects included atrial septal defects (secundum and NOS), Down syndrome, polydactyly/syndactyly, clubfoot, hypospadias and obstructive genitourinary defects. The most common defects in Whites included atrial septal defects, hypospadias, clubfoot, obstructive genitourinary defects, polydactyly/syndactyly and Down syndrome. In Asians, the most common defects included polydactyly/syndactyly, septal defects, obstructive genitourinary defects, Down syndrome and cleft palate.

To understand birth defect trends or patterns in maternal race and ethnicity, we explored differences among the groups for certain categories of birth defects. Figure 11 shows the rate of chromosomal defects and all other defects according to maternal race and ethnicity. These rates were age-adjusted because chromosomal defects as well as other defects may be related to maternal age and differences may exist between racial and ethnic groups. The rates of chromosomal defects in Blacks, Hispanics and "Other" race/ethnicity were about two times the rate in Whites and almost three times the rate in Asians.

Multiple factors likely contribute to differences in prevalence by racial and ethnic groups including genetic variation, diet and lifestyle, differential access or use of health care services including prenatal screening and diagnosis, or socioeconomic differences. Interestingly, maternal birthplace may also be a contributing factor (see Figure 12). Children born to White and Black women born in the U.S. had a slightly higher, though non-significant, rate of birth defects than children of White and Black women born outside the U.S. In contrast, children born to Asian women born in the U.S. had a lower, though non-significant, rate than children of Asian women born outside the U.S. The birth defect rates in children of Hispanic women born in the U.S. were slightly lower, though non-significant, than rates of Hispanic women born outside the U.S. The rate of birth defects in children of Hispanic women born in the U.S. territories (including Puerto Rico, U.S. Virgin Islands and Guam) was non-significantly higher than the rates among women born both in and outside the U.S. More years of data and in-depth studies are needed to affirm the stability of these rates and to understand racial and ethnic patterns.

Trends in Maternal Race and Ethnicity

Figure 13 shows the age-adjusted birth defects rates between 2002-2003 and 2004-2005. In Whites, Blacks, Asians and Hispanics, the rates increased, with the highest increase among Black women and the lowest increase among White women. The birth defects rate in other races decreased slightly. These changes, potentially due to more aggressive and comprehensive case surveillance strategies, were not significant between the two time periods examined.

Birth Defects by Massachusetts Region

The Massachusetts Commonwealth's Executive Office of Health and Human Services delineates regions for use by the Department of Public Health for statistical, care coordination and administrative purposes. The six regions are based on geographical groupings of cities and towns: Western, Central, Northeast, Metro West, Boston and Southeast. A map of these regions is provided in the Appendix section of this report.

The birth defect rates by the six regions in 2004-2005 are shown in Figure 14. Although not statistically significantly different, the rates range from 148.7 per 10,000 in Western Massachusetts to 172.9 per 10,000 in Southeast Massachusetts.

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
Central Nervous System				
Anencephaly	White, Non-Hispanic	7	0.64	0.26 – 1.33
	Black, Non-Hispanic	0	0.00	0.00 – 3.04
	Asian, Non-Hispanic	0	0.00	0.00 – 3.45
	Hispanic	1	0.50	0.01 – 2.81
Encephalocele	White, Non-Hispanic	3	0.28	0.06 – 0.81
	Black, Non-Hispanic	0	0.00	0.00 – 3.04
	Asian, Non-Hispanic	0	0.00	0.00 – 3.45
	Hispanic	0	0.00	0.00 – 1.86
Holoprosencephaly	White, Non-Hispanic	4	0.37	0.10 – 0.94
	Black, Non-Hispanic	0	0.00	0.00 – 3.04
	Asian, Non-Hispanic	0	0.00	0.00 – 3.45
	Hispanic	2	1.01	0.12 – 3.64
Hydrocephaly w/o spina bifida	White, Non-Hispanic	25	2.30	1.49 – 3.39
	Black, Non-Hispanic	3	2.47	0.51 – 7.23
	Asian, Non-Hispanic	3	2.80	0.58 – 8.19
	Hispanic	11	5.54	2.77 – 9.91
Microcephaly	White, Non-Hispanic	19	1.75	1.05 – 2.73
	Black, Non-Hispanic	3	2.47	0.51 – 7.23
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
	Hispanic	7	3.52	1.42 – 7.26
Spina bifida w/ and w/o hydrocephaly	White, Non-Hispanic	19	1.75	1.05 – 2.73
	Black, Non-Hispanic	1	0.82	0.02 – 4.59
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
	Hispanic	2	1.01	0.12 – 3.64
Spinal cord	White, Non-Hispanic	30	2.76	1.86 – 3.94
	Black, Non-Hispanic	4	3.30	0.90 – 8.44
	Asian, Non-Hispanic	3	2.80	0.58 – 8.19
	Hispanic	6	3.02	1.11 – 6.58
Other CNS	White, Non-Hispanic	67	6.16	4.77 – 7.82
	Black, Non-Hispanic	9	7.42	3.39 – 14.08
	Asian, Non-Hispanic	9	8.41	3.84 – 15.96
	Hispanic	13	6.55	3.49 – 11.19

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Eye				
Anophthalmia/microphthalmia	White, Non-Hispanic	5	0.46	0.15 – 1.07
	Black, Non-Hispanic	2	1.65	0.20 – 5.96
	Asian, Non-Hispanic	0	0.00	0.00 – 3.45
	Hispanic	0	0.00	0.00 – 1.86
Congenital glaucoma, congenital cataract	White, Non-Hispanic	29	2.67	1.79 – 3.83
	Black, Non-Hispanic	6	4.95	1.82 – 10.77
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
	Hispanic	7	3.52	1.42 – 7.26
Other eye	White, Non-Hispanic	25	2.30	1.49 – 3.39
	Black, Non-Hispanic	5	4.12	1.34 – 9.62
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
	Hispanic	1	0.50	0.01 – 2.81
Ear				
Anotia/microtia	White, Non-Hispanic	13	1.19	0.64 – 2.04
	Black, Non-Hispanic	1	0.82	0.02 – 4.59
	Asian, Non-Hispanic	3	2.80	0.58 – 8.19
	Hispanic	7	3.52	1.42 – 7.26
Other ear	White, Non-Hispanic	15	1.38	0.77 – 2.27
	Black, Non-Hispanic	2	1.65	0.20 – 5.96
	Asian, Non-Hispanic	4	3.74	1.02 – 9.57
	Hispanic	1	0.50	0.01 – 2.81
Cardiovascular				
Anomalous Pulmonary Venous Connection				
Total/partial anomalous pulmonary venous connection	White, Non-Hispanic	22	2.02	1.27 – 3.06
	Black, Non-Hispanic	3	2.47	0.51 – 7.23
	Asian, Non-Hispanic	2	1.87	0.23 – 6.75
	Hispanic	4	2.01	0.55 – 5.16
Conotruncal (Outlet) and Aortic Arch				
ASD primum	White, Non-Hispanic	7	0.64	0.26 – 1.33
	Black, Non-Hispanic	1	0.82	0.02 – 4.59
	Asian, Non-Hispanic	0	0.00	0.00 – 3.45

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Common atrium	Hispanic	1	0.50	0.01 – 2.81
	White, Non-Hispanic	2	0.18	0.02 – 0.66
	Black, Non-Hispanic	2	1.65	0.20 – 5.96
	Asian, Non-Hispanic	0	0.00	0.00 – 3.45
	Hispanic	1	0.50	0.01 – 2.81
Complete atrioventricular canal defect	White, Non-Hispanic	35	3.22	2.24 – 4.47
	Black, Non-Hispanic	10	8.24	3.95 – 15.16
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
	Hispanic	9	4.53	2.07 – 8.6
Endocardial cushion defect (OS and NOS)	White, Non-Hispanic	9	0.83	0.38 – 1.57
	Black, Non-Hispanic	0	0.00	0.00 – 3.04
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
	Hispanic	2	1.01	0.12 – 3.64
VSD, canal type	White, Non-Hispanic	6	0.55	0.20 – 1.20
	Black, Non-Hispanic	0	0.00	0.00 – 3.04
	Asian, Non-Hispanic	0	0.00	0.00 – 3.45
	Hispanic	4	2.01	0.55 – 5.16
Conotruncal (Outlet) and Aortic Arch				
Double outlet right ventricle	White, Non-Hispanic	19	1.75	1.05 – 2.73
	Black, Non-Hispanic	2	1.65	0.2 – 5.96
	Asian, Non-Hispanic	2	1.87	0.23 – 6.75
	Hispanic	5	2.52	0.82 – 5.88
d – Transposition of the great arteries	White, Non-Hispanic	24	2.21	1.41 – 3.28
	Black, Non-Hispanic	3	2.47	0.51 – 7.23
	Asian, Non-Hispanic	2	1.87	0.23 – 6.75
	Hispanic	6	3.02	1.11 – 6.58
Interrupted aortic arch, type B	White, Non-Hispanic	1	0.09	0.00 – 0.51
	Black, Non-Hispanic	0	0.00	0.00 – 3.04
	Asian, Non-Hispanic	1	0.93	0.02 – 5.2
	Hispanic	0	0.00	0.00 – 1.86

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Tetralogy of Fallot w/ and w/o pulmonary atresia	White, Non-Hispanic	49	4.50	3.33 – 5.95
	Black, Non-Hispanic	4	3.3	0.90 – 8.44
	Asian, Non-Hispanic	4	3.74	1.02 – 9.57
Truncus arteriosus	Hispanic	12	6.04	3.12 – 10.56
	White, Non-Hispanic	3	0.28	0.06 – 0.81
	Black, Non-Hispanic	0	0.00	0.00 – 3.04
	Asian, Non-Hispanic	0	0.00	0.00 – 3.45
	Hispanic	0	0.00	0.00 – 1.86
Ebstein Anomaly				
Ebstein anomaly	White, Non-Hispanic	6	0.55	0.20 – 1.20
	Black, Non-Hispanic	1	0.82	0.02 – 4.59
	Asian, Non-Hispanic	0	0.00	0.00 – 3.45
	Hispanic	1	0.50	0.01 – 2.81
Laterality Defects				
Heterotaxy	White, Non-Hispanic	14	1.29	0.70 – 2.16
	Black, Non-Hispanic	9	7.42	3.39 – 14.08
	Asian, Non-Hispanic	3	2.80	0.58 – 8.19
	Hispanic	2	1.01	0.12 – 3.64
Left – Sided Obstruction				
Aortic valve stenosis	White, Non-Hispanic	19	1.75	1.05 – 2.73
	Black, Non-Hispanic	2	1.65	0.20 – 5.96
	Asian, Non-Hispanic	0	0.00	0.00 – 3.45
	Hispanic	2	1.01	0.12 – 3.64
Coarctation of aorta	White, Non-Hispanic	59	5.42	4.13—7.00
	Black, Non-Hispanic	7	5.77	2.32 – 11.89
	Asian, Non-Hispanic	4	3.74	1.02 – 9.57
	Hispanic	4	2.01	0.55 – 5.16
Hypoplastic left heart syndrome	White, Non-Hispanic	14	1.29	0.70 – 2.16
	Black, Non-Hispanic	2	1.65	0.20 – 5.96
	Asian, Non-Hispanic	1	0.93	0.02 – 5.2
	Hispanic	3	1.51	0.31 – 4.41

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Interrupted aortic arch (type A and NOS)	White, Non-Hispanic	1	0.09	0.00 – 0.51
	Black, Non-Hispanic	1	0.82	0.02 – 4.59
	Asian, Non-Hispanic	0	0.00	0.00 – 3.45
	Hispanic	0	0.00	0.00 – 1.86
Patent Ductus Arteriosus				
Patent ductus arteriosus	White, Non-Hispanic	191	17.56	15.15 – 20.23
	Black, Non-Hispanic	50	41.22	30.59 – 54.34
	Asian, Non-Hispanic	17	15.88	9.25 – 25.43
	Hispanic	34	17.12	11.86 – 23.92
Right – Sided Obstruction				
Pulmonary stenosis, valvular	White, Non-Hispanic	62	5.70	4.37 – 7.31
	Black, Non-Hispanic	10	8.24	3.95 – 15.16
	Asian, Non-Hispanic	5	4.67	1.52 – 10.90
	Hispanic	12	6.04	3.12 – 10.56
Pulmonary valve atresia w/intact septum	White, Non-Hispanic	7	0.64	0.26 – 1.33
	Black, Non-Hispanic	2	1.65	0.20 – 5.96
	Asian, Non-Hispanic	0	0.00	0.00 – 3.45
	Hispanic	1	0.50	0.01 – 2.81
Pulmonary valve atresia with VSD	White, Non-Hispanic	6	0.55	0.20 – 1.20
	Black, Non-Hispanic	0	0.00	0.00 – 3.04
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
	Hispanic	0	0.00	0.00 – 1.86
Tricuspid valve atresia	White, Non-Hispanic	2	0.18	0.02 – 0.66
	Black, Non-Hispanic	0	0.00	0.00 – 3.04
	Asian, Non-Hispanic	0	0.00	0.00 – 3.45
	Hispanic	0	0.00	0.00 – 1.86
Septal Defects				
ASD (secundum and NOS)	White, Non-Hispanic	194	17.83	15.41 – 20.53
	Black, Non-Hispanic	41	33.80	24.26 – 45.85
	Asian, Non-Hispanic	16	14.95	8.54 – 24.27
	Hispanic	30	15.11	10.19 – 21.57

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
VSD (membranous and NOS)	White, Non-Hispanic	105	9.65	7.89 – 11.68
	Black, Non-Hispanic	11	9.07	4.53 – 16.23
	Asian, Non-Hispanic	11	10.28	5.13 – 18.39
	Hispanic	20	10.07	6.15 – 15.55
VSD, conoventricular/malalignment	White, Non-Hispanic	12	1.10	0.57 – 1.93
	Black, Non-Hispanic	3	2.47	0.51 – 7.23
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
	Hispanic	2	1.01	0.12 – 3.64
Single Ventricle and L – TGA				
L – TGA	White, Non-Hispanic	5	0.46	0.15 – 1.07
	Black, Non-Hispanic	2	1.65	0.20 – 5.96
	Asian, Non-Hispanic	0	0.00	0.00 – 3.45
	Hispanic	1	0.50	0.01 – 2.81
Single ventricle	White, Non-Hispanic	5	0.46	0.15 – 1.07
	Black, Non-Hispanic	3	2.47	0.51 – 7.23
	Asian, Non-Hispanic	0	0.00	0.00 – 3.45
	Hispanic	1	0.50	0.01 – 2.81
Other Cardiovascular				
Other cardiovascular	White, Non-Hispanic	207	19.03	16.52 – 21.8
	Black, Non-Hispanic	31	25.56	17.36 – 36.28
	Asian, Non-Hispanic	17	15.88	9.25 – 25.43
	Hispanic	34	17.12	11.86 – 23.92
Respiratory				
Choanal atresia	White, Non-Hispanic	9	0.83	0.38 – 1.57
	Black, Non-Hispanic	0	0.00	0.00 – 3.04
	Asian, Non-Hispanic	0	0.00	0.00 – 3.45
	Hispanic	0	0.00	0.00 – 1.86
Lung anomalies	White, Non-Hispanic	27	2.48	1.64 – 3.61
	Black, Non-Hispanic	4	3.30	0.90 – 8.44
	Asian, Non-Hispanic	2	1.87	0.23 – 6.75
	Hispanic	5	2.52	0.82 – 5.88

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Other respiratory	White, Non-Hispanic	18	1.65	0.98 – 2.61
	Black, Non-Hispanic	1	0.82	0.02 – 4.59
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
	Hispanic	4	2.01	0.55 – 5.16
Orofacial				
Cleft lip w/ and w/o cleft palate	White, Non-Hispanic	85	7.81	6.24 – 9.66
	Black, Non-Hispanic	5	4.12	1.34 – 9.62
	Asian, Non-Hispanic	6	5.60	2.06 – 12.2
	Hispanic	20	10.07	6.15 – 15.55
Cleft palate w/o cleft lip	White, Non-Hispanic	65	5.97	4.61 – 7.62
	Black, Non-Hispanic	9	7.42	3.39 – 14.08
	Asian, Non-Hispanic	6	5.60	2.06 – 12.20
	Hispanic	12	6.04	3.12 – 10.56
Pierre Robin sequence	White, Non-Hispanic	29	2.67	1.79 – 3.83
	Black, Non-Hispanic	2	1.65	0.20 – 5.96
	Asian, Non-Hispanic	2	1.87	0.23 – 6.75
	Hispanic	1	0.50	0.01 – 2.81
Other orofacial	White, Non-Hispanic	46	4.23	3.10 – 5.64
	Black, Non-Hispanic	6	4.95	1.82 – 10.77
	Asian, Non-Hispanic	5	4.67	1.52 – 10.90
	Hispanic	11	5.54	2.77 – 9.91
Gastrointestinal				
Biliary atresia	White, Non-Hispanic	3	0.28	0.06 – 0.81
	Black, Non-Hispanic	0	0.00	0.00 – 3.04
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
	Hispanic	1	0.50	0.01 – 2.81
Esophageal atresia/tracheoesophageal fistula	White, Non-Hispanic	29	2.67	1.79 – 3.83
	Black, Non-Hispanic	2	1.65	0.20 – 5.96
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
	Hispanic	4	2.01	0.55 – 5.16

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Hirschsprung disease	White, Non-Hispanic	25	2.30	1.49 – 3.39
	Black, Non-Hispanic	3	2.47	0.51 – 7.23
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
	Hispanic	6	3.02	1.11 – 6.58
Rectal and large intestinal atresia/stenosis	White, Non-Hispanic	30	2.76	1.86 – 3.94
	Black, Non-Hispanic	2	1.65	0.20 – 5.96
	Asian, Non-Hispanic	5	4.67	1.52 – 10.90
	Hispanic	7	3.52	1.42 – 7.26
Small intestinal atresia	White, Non-Hispanic	23	2.11	1.34 – 3.17
	Black, Non-Hispanic	8	6.60	2.85—13.00
	Asian, Non-Hispanic	2	1.87	0.23 – 6.75
	Hispanic	8	4.03	1.74 – 7.94
Other gastrointestinal	White, Non-Hispanic	61	5.61	4.29 – 7.20
	Black, Non-Hispanic	8	6.60	2.85—13.00
	Asian, Non-Hispanic	4	3.74	1.02 – 9.57
	Hispanic	11	5.54	2.77 – 9.91
Genitourinary				
Bladder exstrophy	White, Non-Hispanic	3	0.28	0.06 – 0.81
	Black, Non-Hispanic	1	0.82	0.02 – 4.59
	Asian, Non-Hispanic	0	0.00	0.00 – 3.45
	Hispanic	0	0.00	0.00 – 1.86
Cloacal exstrophy	White, Non-Hispanic	4	0.37	0.10 – 0.94
	Black, Non-Hispanic	0	0.00	0.00 – 3.04
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
	Hispanic	0	0.00	0.00 – 1.86
Hypospadias, 2nd or 3rd degree	White, Non-Hispanic	156	14.34	12.18 – 16.77
	Black, Non-Hispanic	15	12.37	6.92 – 20.4
	Asian, Non-Hispanic	17	15.88	9.25 – 25.43
	Hispanic	20	10.07	6.15 – 15.55

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Obstructive genitourinary defect	White, Non-Hispanic	121	11.12	9.23 – 13.29
	Black, Non-Hispanic	12	9.89	5.11 – 17.28
	Asian, Non-Hispanic	10	9.34	4.48 – 17.18
	Hispanic	20	10.07	6.15 – 15.55
Renal agenesis/hypoplasia	White, Non-Hispanic	2	0.18	0.02 – 0.66
	Black, Non-Hispanic	0	0.00	0.00 – 3.04
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
	Hispanic	1	0.50	0.01 – 2.81
Other genitourinary	White, Non-Hispanic	166	15.26	13.03 – 17.76
	Black, Non-Hispanic	13	10.72	5.71 – 18.33
	Asian, Non-Hispanic	15	14.01	7.84 – 23.11
	Hispanic	38	19.13	13.54 – 26.26
Musculoskeletal				
Clubfoot	White, Non-Hispanic	128	11.77	9.82 – 13.99
	Black, Non-Hispanic	17	14.01	8.16 – 22.44
	Asian, Non-Hispanic	5	4.67	1.52 – 10.90
	Hispanic	25	12.59	8.15 – 18.58
Craniosynostosis	White, Non-Hispanic	58	5.33	4.05 – 6.89
	Black, Non-Hispanic	2	1.65	0.20 – 5.96
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
	Hispanic	7	3.52	1.42 – 7.26
Diaphragmatic hernia	White, Non-Hispanic	28	2.57	1.71 – 3.72
	Black, Non-Hispanic	3	2.47	0.51 – 7.23
	Asian, Non-Hispanic	4	3.74	1.02 – 9.57
	Hispanic	7	3.52	1.42 – 7.26
Gastroschisis	White, Non-Hispanic	32	2.94	2.01 – 4.15
	Black, Non-Hispanic	2	1.65	0.20 – 5.96
	Asian, Non-Hispanic	0	0.00	0.00 – 3.45
	Hispanic	4	2.01	0.55 – 5.16

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Omphalocele	White, Non-Hispanic	8	0.74	0.32 – 1.45
	Black, Non-Hispanic	1	0.82	0.02 – 4.59
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
	Hispanic	1	0.50	0.01 – 2.81
Polydactyly/syndactyly	White, Non-Hispanic	120	11.03	9.15 – 13.19
	Black, Non-Hispanic	20	16.49	10.07 – 25.46
	Asian, Non-Hispanic	19	17.75	10.69 – 27.72
	Hispanic	16	8.06	4.61 – 13.08
Reduction deformity, lower limbs	White, Non-Hispanic	15	1.38	0.77 – 2.27
	Black, Non-Hispanic	3	2.47	0.51 – 7.23
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
Reduction deformity, upper limbs	Hispanic	1	0.50	0.01 – 2.81
	White, Non-Hispanic	34	3.13	2.16 – 4.37
	Black, Non-Hispanic	3	2.47	0.51 – 7.23
	Asian, Non-Hispanic	3	2.80	0.58 – 8.19
	Hispanic	6	3.02	1.11 – 6.58
Skeletal dysplasia	White, Non-Hispanic	16	1.47	0.84 – 2.39
	Black, Non-Hispanic	0	0.00	0.00 – 3.04
	Asian, Non-Hispanic	3	2.80	0.58 – 8.19
	Hispanic	4	2.01	0.55 – 5.16
Other musculoskeletal	White, Non-Hispanic	119	10.94	9.06 – 13.09
	Black, Non-Hispanic	7	5.77	2.32 – 11.89
	Asian, Non-Hispanic	8	7.47	3.23 – 14.73
	Hispanic	24	12.09	7.74 – 17.98
Chromosomal and other Syndromes				
Klinefelter syndrome	White, Non-Hispanic	7	0.64	0.26 – 1.33
	Black, Non-Hispanic	0	0.00	0.00 – 3.04
	Asian, Non-Hispanic	0	0.00	0.00 – 3.45
	Hispanic	1	0.50	0.01 – 2.81

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Trisomy 13	White, Non-Hispanic	7	0.64	0.26 – 1.33
	Black, Non-Hispanic	2	1.65	0.20 – 5.96
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
	Hispanic	0	0.00	0.00 – 1.86
Trisomy 18	White, Non-Hispanic	7	0.64	0.26 – 1.33
	Black, Non-Hispanic	2	1.65	0.20 – 5.96
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
	Hispanic	2	1.01	0.12 – 3.64
Trisomy 21 (Down syndrome)	White, Non-Hispanic	114	10.48	8.64 – 12.59
	Black, Non-Hispanic	20	16.49	10.07 – 25.46
	Asian, Non-Hispanic	10	9.34	4.48 – 17.18
	Hispanic	31	15.61	10.61 – 22.16
Turner syndrome	White, Non-Hispanic	10	0.92	0.44 – 1.69
	Black, Non-Hispanic	1	0.82	0.02 – 4.59
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
	Hispanic	1	0.50	0.01 – 2.81
Other chromosomal syndromes/other syndromes	White, Non-Hispanic	110	10.11	8.31 – 12.19
	Black, Non-Hispanic	19	15.66	9.43 – 24.46
	Asian, Non-Hispanic	10	9.34	4.48 – 17.18
	Hispanic	23	11.58	7.34 – 17.38
Other				
Amniotic bands	White, Non-Hispanic	7	0.64	0.26 – 1.33
	Black, Non-Hispanic	2	1.65	0.20 – 5.96
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
	Hispanic	2	1.01	0.12 – 3.64
Skin anomalies	White, Non-Hispanic	6	0.55	0.20 – 1.20
	Black, Non-Hispanic	1	0.82	0.02 – 4.59
	Asian, Non-Hispanic	1	0.93	0.02 – 5.20
	Hispanic	0	0.00	0.00 – 1.86

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2004-2005

Defect[1]	Maternal Race[2]	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Other, specified	White, Non-Hispanic	15	1.38	0.77 – 2.27
	Black, Non-Hispanic	8	6.60	2.85—13.00
	Asian, Non-Hispanic	2	1.87	0.23 – 6.75
	Hispanic	2	1.01	0.12 – 3.64

[1.] Cases can be included in the count for more than one defect. Cases are counted once in the total for a defect category. Because only live births are presented on this table, case totals are not listed.
[2.] Due to small numbers, races classified as "other" are not included.

Table 13 Most Common Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2004-2005

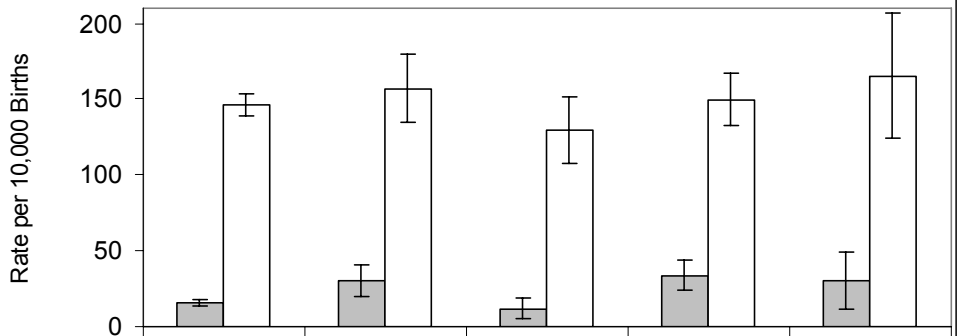
Race[1]	Defect[2]	Count	Rate per 10,000 Births	95% Confidence Interval
White, Non-Hispanic	ASD (secundum and NOS)	194	17.83	15.41 – 20.53
	Hypospadias, 2nd or 3rd degree	156	14.34	12.18 – 16.77
	Clubfoot	128	11.77	9.82 – 13.99
	Obstructive genitourinary defect	121	11.12	9.23 – 13.29
	Polydactyly/syndactyly	120	11.03	9.15 – 13.19
	Trisomy 21 (Down syndrome)	114	10.48	8.64 – 12.59
	VSD (membranous and NOS)	105	9.65	7.89 – 11.68
	Cleft lip w/ and w/o cleft palate	85	7.81	6.24 – 9.66
	Cleft palate w/o cleft lip	65	5.97	4.61 – 7.62
	Pulmonary stenosis, valvular	62	5.70	4.37 – 7.31
Black, Non-Hispanic	ASD (secundum and NOS)	41	33.80	24.26 – 45.85
	Trisomy 21 (Down syndrome)	20	16.49	10.07 – 25.46
	Polydactyly/syndactyly	20	16.49	10.07 – 25.46
	Clubfoot	17	14.01	8.16 – 22.44
	Hypospadias, 2nd or 3rd degree	15	12.37	6.92 – 20.40
	Obstructive genitourinary defect	12	9.89	5.11 – 17.28
	VSD (membranous and NOS)	11	9.07	4.53 – 16.23
	Complete atrioventricular canal defect	10	8.24	3.95 – 15.16
	Pulmonary stenosis, valvular	10	8.24	3.95 – 15.16
	Cleft palate w/o cleft pip	9	7.42	3.39 – 14.08
Asian, Non-Hispanic	Polydactyly/syndactyly	19	17.75	10.69 – 27.72
	Hypospadias, 2nd or 3rd degree	17	15.88	9.25 – 25.43
	ASD (secundum and NOS)	16	14.95	8.54 – 24.27
	VSD (membranous and NOS)	11	10.28	5.13 – 18.39
	Obstructive genitourinary defect	10	9.34	4.48 – 17.18
	Trisomy 21 (Down syndrome)	10	9.34	4.48 – 17.18
	Cleft palate w/o cleft lip	6	5.60	2.06 – 12.20
	Cleft lip w/ and w/o cleft palate	6	5.60	2.06 – 12.20
	Rectal and large intestinal atresia/stenosis	5	4.67	1.52 – 10.90
	Clubfoot	5	4.67	1.52 – 10.90

Table 13 Most Common Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2004-2005

Race[1]	Defect[2]	Count	Rate per 10,000 Births	95% Confidence Interval
(*cont'd*)				
Hispanic	Trisomy 21 (Down syndrome)	31	15.61	10.61 – 22.16
	ASD (secundum and NOS)	30	15.11	10.19 – 21.57
	Clubfoot	25	12.59	8.15 – 18.58
	VSD (membranous and NOS)	20	10.07	6.15 – 15.55
	Cleft lip w/ and w/o cleft palate	20	10.07	6.15 – 15.55
	Obstructive genitourinary defect	20	10.07	6.15 – 15.55
	Hypospadias, 2nd or 3rd degree	20	10.07	6.15 – 15.55
	Polydactyly/syndactyly	16	8.06	4.61 – 13.08
	Tetralogy of Fallot w/ and w/o pulmonary atresia	12	6.04	3.12 – 10.56
	Cleft palate w/o cleft lip	12	6.04	3.12 – 10.56

1. Due to small numbers, races classified as "other" are not included.
2. Excludes patent ductus arteriosus (PDA) due to the high number of cases and the mild severity of the majority of these cases.

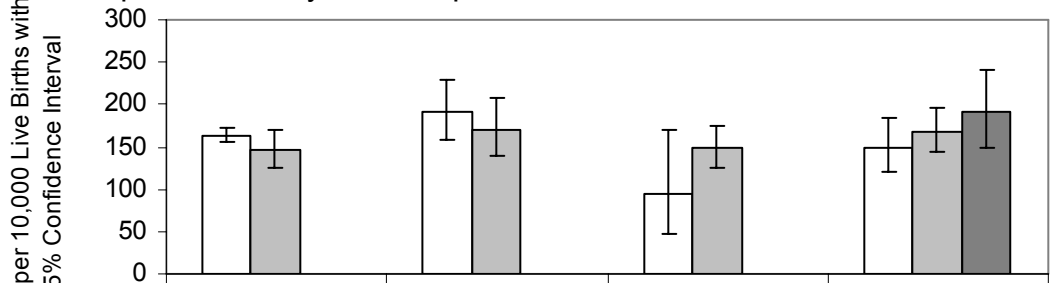
Figure 11 Age-Adjusted Prevalence of Chromosomal and All Other Defects by Maternal Race / Hispanic Ethnicity, Massachusetts: 2004-2005



	White, Non-Hispanic	Black, Non-Hispanic	Asian, Non-Hispanic	Hispanic	Other
Chromosomal defects	15.5 (13.2-17.7)	30.5 (20.1-40.9)	11.9 (5.4-18.4)	33.8 (23.6-44.0)	30.1 (11.4-48.8)
All other defects	146.4 (139.1-153.6)	157.0 (134.4-179.5)	129.5 (107.9-151)	149.6 (132.4-166.9)	165.4 (124.2-206.5)

Livebirths only, N=2535 (1 unknown race/ethnicity)

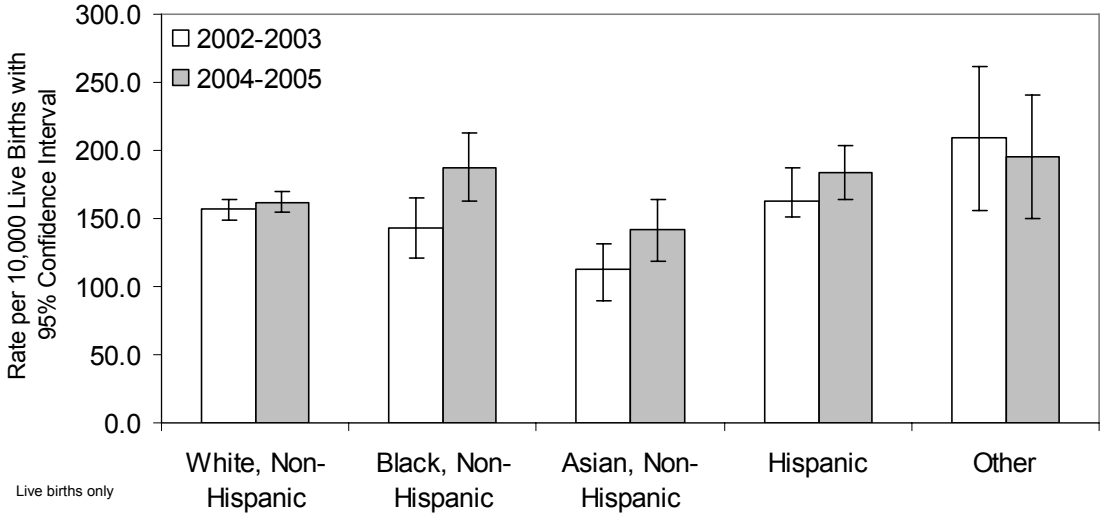
Figure 12 Prevalence of Birth Defects by Maternal Race / Hispanic Ethnicity and Birthplace, Massachusetts: 2004-2005



	White, Non-Hispanic	Black, Non-Hispanic	Asian, Non-Hispanic	Hispanic
U.S.-Born*	163.9 (156.0-172.2)	191.2 (158.7-229.7)	95.0 (47.4-169.8)	149.6 (140.9-192.2)
Non-U.S.-Born	146.4 (126.0-169.4)	170.6 (139.1-207.1)	147.7 (124.3-174.2)	168.3 (143.6-196.0)
U.S. Territories	**	**	**	190.7 (149.2-240.1)

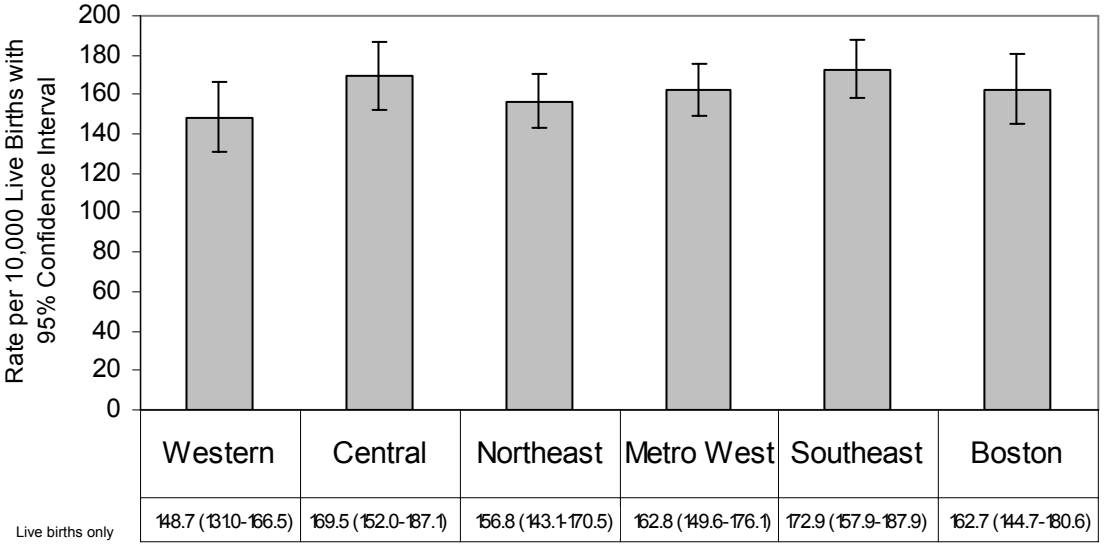
*Including Puerto Rico, U.S. Virgin Islands, and Guam for all except Hispanic
**Less than 5 cases were in this category
N=2590

Figure 13 Age-Adjusted Prevalence of Birth Defects by Maternal Race / Hispanic Ethnicity, Massachusetts: 2002-2003 and 2004-2005



Live births only

Figure 14 Age-Adjusted Prevalence of Birth Defect Rates by Region, Massachusetts: 2004-2005

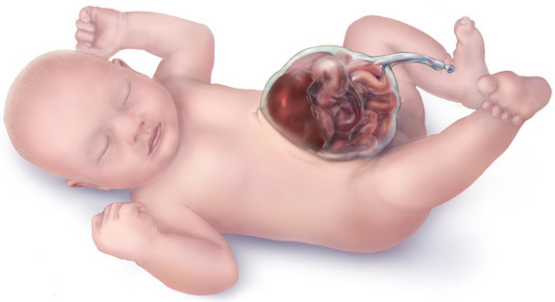


Western	Central	Northeast	Metro West	Southeast	Boston
148.7 (131.0-166.5)	169.5 (152.0-187.1)	156.8 (143.1-170.5)	162.8 (149.6-176.1)	172.9 (157.9-187.9)	162.7 (144.7-180.6)

Live births only

Chapter 6

Prevalence of Birth Defects by Severity, Etiology and Pattern



Baby with omphalocele

Courtesy of the Centers for Disease Control and Prevention

Prevalence of Birth Defects by Severity

Birth defects cases were categorized by their level of severity. 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.

These levels of severity included four categories: “severe” was defined as requiring supportive measures, usually incompatible with life; “serious” was defined as correctable, most having long-term needs; “moderate” was defined as most amenable to correction, many having long-term needs; and “mild” was defined as amenable to correction, with minimal long-term needs. The rules for designing the new automated algorithm are in the Technical Notes, and a list of selected defects within each severity category is in the Appendix.

Table 14 shows the percentage of birth defect 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 and Khoury 2000).

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

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

“Mild” birth defects comprised nearly 7% of the cases. Within the classification of “mild severity,” there was variability. For example, children with microphthalmia (small eyes) could have mild reduction in the size of the globe or a more severe reduction resulting in visual loss or the need for intrusive ophthalmologic medical care. In contrast, infants with isolated dextrocardia (heart in the right side of the chest instead of the left) and no other heart defect have no clinical consequence.

Prevalence of Birth Defects by Etiology and Pattern

To enhance the existing active birth defects surveillance program, a method was developed to classify cases by etiology and pattern. The surveillance system in Massachusetts allowed for the collection of relevant etiology information. Categories with sufficient detail were created, allowing similar cases to be grouped using knowledge of pathogenesis and embryologic mechanisms. The case classification defined a case as a biologic entity rather than a collection of individual defects. The schema was based upon general principles outlined in the literature (Rasmussen, Olney et al. 2003; Cary, Feldkamp et al. 2005).

Cases with known etiology accounted for about 18% of the birth defects (472 of 2,590 live births and stillbirths) in Massachusetts in 2004-2005. Etiology groups include “single gene,” “chromosomal,” “maternal-fetal factors” (including teratogens or uterine factors) and “other factors.”

As Figure 15 shows, single gene etiology accounted for almost 24% (112) of the known etiology cases. Single gene defects include achondroplasia, Marfan (Del 15q21.1), Smith-Lemli-Opitz syndrome and other examples of defects categorized as Mendelian syndrome. Chromosomal etiology accounted for almost 67% (320) of the cases with known etiology. Cases with chromosomal etiology include trisomy 13, 18 and 21, Turner syndrome and other chromosomal duplications and deletions. Maternal-fetal factors accounted for about 8% (37) of all cases with known etiology. Maternal-fetal factors include teratogens such as maternal diabetes and uterine factors such as deformation or didelphy uterus. The remaining known etiology cases included conjoined twins. Cases with known etiology may also be classified by pattern, with the categories and definitions listed in Table 15. The majority of cases with known etiology fall within the “multiple majors” pattern including combinations of two or more major defects.

While the CDC estimates that about 70% of birth defects have unknown cause, the majority (82%) of birth defects cases in Massachusetts in 2004-2005 had unknown etiology (2,118 of 2,590 live births and stillbirths). These were categorized by pattern and by description in Table 15.

The “isolated” pattern is the largest category among the cases with unknown etiology, comprising about 89%. Within this pattern are three subgroups based on the number and type of defects found in a case. The largest subgroup of isolated cases have a truly solitary defect such as cleft palate, gastroschisis or anencephaly, comprising about 54% of total cases with unknown etiology. The next largest isolated pattern subgroup accounts for about 30% of the cases with unknown etiology. These cases have 2 or more defects in the same organ or body part, or they have an isolated major and some minor defects in different body parts. An example of this pattern is coarctation of the aorta and aortic stenosis (valvar). The third subgroup accounts for the remaining cases within the isolated pattern and is defined by a sequence of a common primary defect with consistent related anomalies. An example of this pattern is spina bifida, clubfoot, hydrocephalus and hip dislocation. The “multiple majors” pattern comprises about 11% of cases with unknown etiology, and these cases include combinations of two or more major defects such as hydrocephalus and cleft palate and anotia. See Figure 16 for the distribution of cases with unknown etiology among these patterns.

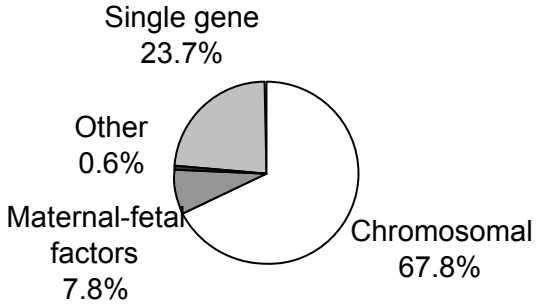
Table 14 Percentage of Birth Defect Cases by Severity Groups, Massachusetts: 2004-2005

SEVERITY CATEGORIES	PERCENTAGE OF BIRTH DEFECTS CASES
Severe, supportive measures, usually incompatible with life	2.7%
Serious, may be correctable, most have long-term needs	17.3%
Moderate, most correctable, many have long-term needs	73.0%
Mild, may be correctable, minimal long-term needs	7.0%

Table 15 Patterns and Etiology of Birth Defect Cases, Massachusetts: 2004-2005

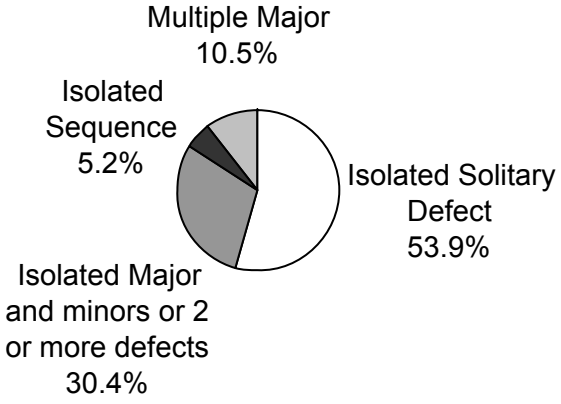
Pattern	Description	Cases	
		Known Etiology	Unknown Etiology
Isolated	Solitary defect	13	1,142
	Major and minors (different organ/body parts) or 2 or more defects (same organ/body part)	10	643
	Sequence: Common primary defect with consistent, related anomalies	17	110
Multiple Majors	2 or more major defects in different organs/body parts	432	223
Total		472 (18.2%)	2,118 (81.8%)

Figure 15 Birth Defect Cases with Known Etiology, Massachusetts: 2004-2005



N=472

Figure 16 Patterns of Birth Defect Cases with Unknown Etiology, Massachusetts: 2004-2005



N=2,118

Appendices

Technical Notes

Definitions

2004 – 2005 Denominators Used in Calculating Rates

Birth Defects Codes and Exclusions by Defect Category

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

Birth Defects by Severity

Glossary of Selected Birth Defect Terms

Map of MA Executive Office of Health and Human Services (EOHHS) Regions

References

Technical Notes

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

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

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

Cases/total number live births x 10,000

Fetal deaths are included in the numerator but not in the denominator, so the result is technically a ratio and 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. A difference is statistically significant when the confidence intervals do not overlap.

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

Calculation of 2005 Dollars
2005 dollars were calculated from the Gross Domestic Product Deflator Inflation Index, an inflation calculator for adjusting costs from one year to another using the Gross Domestic Product (GDP) Deflator inflation index of 1.0343 representing the inflation from 2001 to 2003. (Based on this calculator, costs increased from 118 million to 122 million during this period.) This inflation calculator is based on the inflation rate during the US Government Fiscal Year, which begins on October 1 and ends on September 30. http://cost.jsc.nasa.gov/inflateGDP.html.

Assignment of 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.

For this report, a new automated algorithm was created based on modified rules that had been developed to determine severity of defects described in previous birth defects surveillance reports, and the program was validated using the data described in the reports. The automated process was able to assign severity levels to about 95% of the cases, with the remaining 5% assigned manually by the Center Clinical Geneticist. The process that included the new automated categorization system produced percentages of birth defects within each of the four severity categories in 2004-2005 that were similar to those attained each of the categories in 2002-2003.

Some of the rules for assigning severity level are briefly described here. First, each defect labeled by an ICD9/BPA code was assigned a severity score or range of severity scores based on the defining characteristics of the defect. Each infant/fetus case was usually assigned a severity score based on the most severe defect it displayed. An exception was when the infant/fetus had 3 or more mild defects and was categorized as a moderate case. Cases with infant death when a lethal anomaly was not present were reviewed by the Center Clinical Geneticist and manually assigned a severity level. Cases with one or more defects that ranged in a single severity category may have required further review and manual assignment of severity level. Cases with a syndrome plus defect(s) were listed by the severity of the syndrome only. Syndromes were defined as a group of malformations that occurred together frequently enough to be recognized collectively as a distinct abnormal condition. The remainder of complex cases such as multiple major cases and syndromes required manual review by the Clinical Geneticist.

Definitions

These definitions are derived from the Massachusetts Department of Public Health report titled Massachusetts Births, 2005.

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
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
The birth of a fetus at greater than or equal to 20 weeks gestation, or with a weight of at least 350 grams.

2004 and 2005 Populations Used in Calculating Rates

		Numbers of Live Births to MA Residents		
		2004	**2005**	**Total**
Overall		78,460	76,824	155,284
By Maternal Age Group (yrs)	<20	4,601	4,598	9,199
	20 – 24	11,670	11,805	23,475
	25 – 29	17,914	18,026	35,940
	30 – 34	25,804	24,140	49,944
	35+	18,471	18,249	36,720
By Infant's Sex	Male	40,222	39,233	79,455
	Female	38,238	37,590	75,828
By Plurality	Singleton	74,677	73,258	147,935
	Multiple Birth	3,783	3,565	7,348
By Maternal Race/Ethnicity	White, NH	55,322	53,469	108,791
	Black, NH	6,053	6,077	12,130
	Hispanic	9,798	10,061	19,859
	Asian, NH	5,454	5,251	10,705

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

Birth Defect Codes and Exclusions[1] by Defect Category (cont'd)		
Defect	ICD-9 / BPA[2]	NOTES
Cardiovascular		
Anomalous Pulmonary Venous Connection		
Total/partial anomalous pulmonary venous connection	747.420, 747.430	
Atrioventricular Canal Defects		
ASD primum	745.600	
Common atrium	745.610	
Complete atrioventricular canal defect	745.620, 745.630	
Endocardial cushion defect (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 arteriosus	745.000	
Ebstein Anomaly		
Ebstein anomaly	746.200	
Laterality Defects		
Heterotaxy, situs inversus	759.300-759.395	
Left-Sided Obstruction		
Aortic valve stenosis	746.300	
Coarctation of aorta	747.100-747.190	
Hypoplastic left heart syndrome	746.700, 747.200	
Interrupted aortic arch (type A and NOS)	747.215, 747.216	

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

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

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

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

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

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

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2004-2005

BPA label	BPA Code	# of Defects
Central Nervous System		
Agyria and lissencephaly	742240	4
Anencephaly	740020	12
Brain cysts: Cerebral / subependymal / periventricular	742420	3
Brain cysts: Porencephaly / porencephalic	742410	3
Brain, reduction defect OS (8/02 Includes colpocephaly, pachygyria, schizencephaly)	742280	21
Brain: Other specified anomalies / cortical atrophy / cranial nerve defects	742480	7
Cerebellar hypoplasia	742235	5
Cerebellum anomalies	742230	7
Cerebrum anomalies	742200	4
Corpus callosum anomalies	742210	58
Dandy – Walker malformation	742310	10
Encephalocele, frontal (including proencephalon)	742085	1
Encephalocele, occipital	742000	2
Enlarged brain and head / enlarged head / enlarged brain / megalencephaly / macrocephaly	742400	12
Holoprosencephaly, lobar	742267	2
Holoprosencephaly, NOS	742260	5
Hydranencephaly	742320	4
Hydrocephaly, NOS	742390	20
Hydrocephaly, anomalies of aqueduct of Sylvius	742300	11
Hydrocephaly, other specified	742380	5
Lipomeningocele, highest level unspecified, no mentioned hydrocephalus, closed lesion	741859	1
Lipomeningocele, highest level, thoracic, no mentioned hydrocephalus, closed lesion	741852	1
Lipomeningomyelocele, highest level, lumbar, no mentioned hydrocephalus, closed	741843	5
Meningocele, highest level, lumbar, Arnold Chiari malformation ± hydrocephalus, open lesion	741013	1
Meningocele, highest level, sacral, no mentioned hydrocephalus, closed lesion	741814	2
Meningomyelocele/myelomeningocele, highest level, lumbar, Arnold Chiari malformation ± hydrocephalus, closed	741103	1
Meningomyelocele/myelomeningocele, highest level, lumbar, Arnold Chiari malformation ± hydrocephalus, open	741003	6
Meningomyelocele/myelomeningocele, highest level, lumbar, Hydrocephalus, other (aqueduct of Sylvius) or NOS, open	741303	2
Meningomyelocele/myelomeningocele, highest level, sacral, Arnold Chiari malformation ± hydrocephalus, open lesion	741004	3
Meningomyelocele/myelomeningocele, highest level, sacral, hydrocephalus, other (aqueduct of Sylvius) or NOS, open lesion	741304	1
Meningomyelocele/myelomeningocele, highest level, sacral, no mentioned hydrocephalus, open	741704	2
Microcephalus	742100	31
Microgyria / polymicrogyria	742250	9
Other specified spina bifida, highest level, lumbar, hydrocephalus, other (aqueduct of Sylvius) or NOS, unspec. open/closed	741583	1
Spinal cord: Other specified anomalies (Includes tethered cord)	742580	45
Eye		

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2004-2005

BPA label (cont'd)	BPA Code	# of Defects
Anophthalmos, bilateral	743004	1
Anterior segment: OS colobomas and anomalies	743480	2
Axenfeld Anomaly, right	743467	1
Buphthalmos/congenital glaucoma, bilateral	743204	5
Buphthalmos/congenital glaucoma, left	743201	6
Buphthalmos/ congenital glaucoma, right	743202	4
Cataract, anterior polar, bilateral	743354	2
Cataract, anterior polar, right	743352	1
Cataract, NOS, left	743321	7
Cataract, NOS, bilateral	743324	13
Cataract, NOS, right	743322	3
Cataract, other specified, left	743361	2
Cataract, other specified, bilateral	743364	3
Cataract, other specified, right	743362	2
Choroid: Coloboma	743535	4
Cornea, other anomalies. Excludes: megalocornea (use 743220)	743410	2
Eyelids: Coloboma	743636	3
Iris: Coloboma	743430	8
Microphthalmos, bilateral	743104	4
Microphthalmos, left	743101	2
Microphthalmos, right	743102	1
Optic disc: Specified anomalies / hypoplastic optic nerve / coloboma of the optic disc	743520	9
Peters anomaly, bilateral	743464	1
Peters anomaly, left	743461	2
Peters anomaly, right	743462	1
Retina: Specified anomalies / congenital retinal aneurysm.	743510	4
Riegers Anomaly, bilateral	743474	1
Vitreous humor: Specified anomalies (includes PHPV)	743500	3

Ear

Anotia, bilateral	744014	1
Anotia, right	744012	1
Microtia, Bilateral	744214	5
Microtia, left	744211	7
Microtia, right	744212	14
S # Ear : accessory auricle / polyotia	744100	2
S absence or anomaly of eustachian tube	744250	1
S absence or stricture of auditory canal	744000	2
S anomaly of inner ear / congenital anomaly of membranous labyrinth or organ of Corti	744030	3
S anomaly of middle ear / fusion of ossicles	744020	1
S ear: Other specified anomalies (see also 744.230) / #DARWIN tubercle	744280	3
S ear: Other misshapen ear / cleft / malformed /#POINTED / # ELFIN,pixie – like / # LOP / # CAULIFLOWER / # ABSENT or decreased cartilage – a conditional exclusion if <36wks	744230	16

Cardiovascular

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2004-2005

BPA label (cont'd)	BPA	# of
Anomalous Pulmonary Venous Connection		
Partial anomalous pulmonary venous return/connection/drainage	747430	9
Total anomalous pulmonary venous return/connection/drainage	747420	22
Atrioventricular Canal Defects		
Atrial septal defect, primum type (ASD1)	745600	9
Common atrium	745610	5
Complete atrioventricular canal (CAVC)	745630	58
Complete atrioventricular canal (CAVC) with ventricular septal defect	745620	1
Endocardial cushion defect, NOS	745690	3
Endocardial cushion defect, other specified	745680	10
Ventricular septal defect, inflow type (subtricuspid, canal – type) (VSDavc)	745685	11
Conotruncal (Outlet) and Aortic Arch		
Dextro – transposition of great arteries (dTGA, dTGV) w/ intact ventricular septum	745100	24
Dextro – transposition of great arteries (dTGA, dTGV) w/ ventricular septal defect	745110	12
Double – outlet right ventricle (DORV) with normally related great arteries	745185	7
Double – outlet right ventricle (DORV) with transposed great arteries	745186	9
Double – outlet right ventricle (DORV), NOS	745189	7
Double – outlet right ventricle (DORV), other specified	745188	6
Interrupted aortic arch, type B	747217	2
Pulmonary atresia with VSD (tetralogy of Fallot with pulmonary atresia)	747310	12
Tetralogy of Fallot	745200	58
Truncus arteriosus	745000	3
Ebstein Anomaly		
Ebstein malformation or anomaly	746200	8
Heterotaxy (Laterality Defects)		
Complete situs inversus w/ dextrocardia	759300	7
Heterotaxy, NOS	759395	5
Situs ambiguous, left; left isomerism	759360	4
Situs ambiguous, right; right isomerism	759350	2
Situs ambiguous, sidedness NOS	759380	4
Situs ambiguous, sidedness unclear	759370	2
Situs inversus abdominis	759330	3
Situs inversus w/ levocardia	759310	3
Left – Sided Obstruction		
Aortic stenosis, valvar	746300	23
Coarctation of the aorta (COA), postductal (distal)	747110	3
Coarctation of the aorta (COA), preductal (proximal)	747100	1
Coarctation of the aorta, juxtaductal	747120	10
Coarctation of the aorta, NOS	747190	62
Hypoplastic left heart syndrome	746700	21
Interrupted aortic arch, type A	747216	2
Patent Ductus Arteriosus		
Patent ductus arteriosus (PDA)	747000	303
Right – Sided Obstruction		
Pulmonary valve atresia with VSD (not TOF variant 747310)	746030	8
Pulmonary valve atresia/intact ventricular septum	746000	12
Pulmonic stenosis, valvar	746010	92
Tricuspid atresia	746100	7

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2004-2005

BPA label (cont'd)	BPA Code	# of Defects
Septal Defects		
Atrial septal defect, NOS	745599	83
Atrial septal defect, OS	745580	4
Atrial septal defect, secundum type (ASD2)	745510	198
Ventricular septal defect, NOS	745490	15
Ventricular septal defect, malalignment – type (type I, subarterial) (VSDmal)	745487	18
Ventricular septal defect, perimembranous (type II, membranous) (VSDmem)	745485	144
Single Ventricle and L – TGA		
L – TGA/corrected transposition of great vessels / ventricular inversion	745120	8
Single ventricle, NOS	745300	1
Single ventricle, double inlet left ventricle	745310	7
Single ventricle, other specified (e.g., absent right or left AV connection)	745380	1
Other Cardiovascular		
"Pulmonic" or pulmonary atresia, stenosis, or hypoplasia, NOS w/ no mention of whether valve of artery	746995	3
Anomalies of coronary artery or sinus	746885	11
Aorta: Hypoplasia	747210	11
Aorta: Other specified anomalies	747280	2
Aorta: Persistent right aortic arch	747230	42
Aorta: Vascular ring / double aortic arch / vascular ring compression of trachea	747250	11
Aortic septal defect / aortopulmonary window	745010	2
Aortic valve: bicuspid BAV	746400	63
Aortic valve: Other specified anomalies / aortic valve atresia	746480	21
Aortic valve: Unspecified anomalies	746490	1
Cerebral vessels: Other anomalies / vein of Galen	747810	3
Circulatory system: Other specified anomalies	747880	1
Cor triatriatum	746820	3
Great veins: Other specified anomalies (includes IVC interruption, bilateral SVC)	747480	24
Heart: Other specified anomalies / ectopia cordis / mesocardia / conduction defects, NOS	746880	62
Hypoplastic left ventricle. Excludes: hypoplastic left heart syndrome (746700)	746881	7
Hypoplastic right heart or right ventricle / Uhl's disease (parchment RV)	746882	2
Mitral valve: Absence, atresia, or hypoplasia	746505	9
Mitral valve: Congenital mitral stenosis	746500	13
Mitral valve: insufficiency or regurgitation, congenital	746600	2
Peripheral arteries: Other anomalies / aberrant subclavian artery	747640	33
Persistent left superior vena cava	747410	38
Pulmonary artery: atresia, absence or agenesis. Use 746995 if artery or valve is not specified	747300	1
Pulmonary artery: other specified / pulmonary artery hypoplasia	747380	14
Pulmonary artery: stenosis. Use 746.995 if artery or valve is not specified	747320	12
Pulmonary infundibular (subvalvular) stenosis	746830	8
Pulmonary valve: Other specified anomalies. Excludes: infundibular PS (746830)	746080	20
Pulmonary valve: Unspecified anomaly	746090	1
Situs: Dextrocardia without situs inversus / dextrocardia with situs solitus	746800	8
Supra – aortic stenosis / supravalvular aortic stenosis. Excludes: aortic stenosis, congenital (see 746300)	747220	3
Respiratory		
Agenesis or aplasia of lung	748500	3
Choanal atresia, Bilateral	748014	3

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2004-2005

BPA label (cont'd)	BPA Code	# of Defects
Choanal atresia, Left	748011	3
Choanal atresia, Right	748012	4
Choanal stenosis	748000	11
Hypoplasia of lung or pulmonary hypoplasia	748510	2
Larynx: Cleft / laryngotracheoesophageal cleft	748385	8
Lung cysts: CCAM (cong cystic adenomatoid malf), OS	748480	26
Other anomalies of trachea	748330	4
Other specified respiratory system anomalies / congenital lobar emphysema / lymphangiectasia of lung	748880	1
S lung cysts: Single	748400	1
S lung: Other and unspecified anomalies	748690	3
Sequestration of lung	748520	16

Orofacial

Branchial cleft, sinus, fistula, cyst, or pit	744400	29
Cleft hard palate, bilateral	749010	5
Cleft hard palate, central	749020	16
Cleft hard palate, NOS	749030	15
Cleft lip and palate, bilateral cleft lip	749210	22
Cleft lip and palate, central cleft lip	749220	1
Cleft lip and palate, NOS	749290	2
Cleft lip and palate, unilateral cleft lip, left	749201	28
Cleft lip and palate, unilateral cleft lip, right	749202	20
Cleft lip and palate, unilateral cleft lip, side unk	749203	2
Cleft lip, bilateral	749110	5
Cleft lip, unilateral, left	749101	27
Cleft lip, unilateral, right	749102	12
Cleft palate, NOS	749090	9
Cleft soft palate, bilateral	749050	1
Cleft soft palate, central	749060	9
Cleft soft palate, NOS	749070	38
Cleft soft palate, unilateral, side unk	749043	1
Cleft: Incomplete CL/ microform /pseudo / fused lip /healed lip	749190	3
Face or neck: Other specified anomalies (6/03 eg. facial cleft)	744880	12
Nose: OS anomalies: small nose and nostril / absent nasal septum / flat or wide nasal bridge/ beaked nose	748180	21
Other branchial cleft anomalies / dermal sinus of head	744480	6
Pierre Robin sequence	524080	34
Tongue: Dislocation or displacement / glossoptosis	750130	5
Tongue: large / macroglossia	750120	1

Gastrointestinal

Anal atresia with fistula	751230	23
Anal atresia without mention of fistula	751240	15
Annular pancreas	751720	4
Biliary atresia, extrahepatic or NOS (use 751670 for intrahepatic)	751650	5

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2004-2005

BPA label (cont'd)	BPA Code	# of Defects
Bronchoesophageal fistula with or without mention of esophageal atresia	750330	1
Congenital hiatal hernia / cardiac displacement through esophageal hiatus / partial thoracic stomach. Excludes: congenital diaphragmatic hernia (756.610)	750600	1
Duplication of anus, appendix, cecum, or intestine / enterogenous cyst	751500	6
Ectopic (displaced, anteriorly placed) anus	751530	17
Esophageal atresia with TE fistula	750310	34
Esophageal atresia without TE fistula	750300	4
Hirschsprung disease, NOS	751330	12
Hirschsprung disease: Long – segment (aganglionosis beyond rectum)	751310	17
Hirschsprung disease: Short – segment (aganglionosis involving no more than the anal sphincter and the rectum)	751320	6
Intestinal atresia/stenosis, duodenum	751100	17
Intestinal atresia/stenosis, ileum	751120	8
Intestinal atresia/stenosis, jejunum	751110	22
Intestinal atresia/stenosis, large intestine, NOS	751200	3
Malrotation: cecum and/or colon	751400	1
Malrotation: Other specified and unspecified	751490	48
Malrotation: small intestine alone	751495	3
Meckel's diverticulum	751010	10
Microcolon	751520	1
Other specified anomalies of intestine / rectal fissures	751580	2
Other specified anomalies of upper alimentary tract	750800	1
Persistent omphalomesenteric duct / persistent vitelline duct	751000	4
Rectal atresia/stenosis without mention of fistula	751220	6
Tracheoesophageal fistula without mention of esophageal atresia	750320	1

Genitourinary

Atresia, stricture, or stenosis of ureter / ureteropelvic junction obstruction or stenosis /ureterovesical junction obstruction or stenosis / hypoplastic ureter	753210	50
Bladder exstrophy	753500	4
Cloacal exstrophy	751550	5
Congenital hydronephrosis / pyelocaliectasis	753200	147
Double urethra or urinary meatus	753840	1
Genital organs: Other specified anomalies / microgenitalia / macrogenitalia	752880	1
Gyne: Ovaries, multiple cysts	752085	2
Gyne: Hymen imperforate	752430	1
Gyne: OS anomalies of cervix, vagina, or external female genitalia / Vaginal tags / Hymenal tags	752480	3
Gyne: S Ovaries, Other specified anomalies	752080	1
Gyne: Uterus fistulae connecting with digestive or urinary tract/ uterointestinal fistula / ureterovesical fistula	752320	1
Gyne: Uterus, other anomalies / bicornuate/ unicornis	752380	2
Hypospadias, second degree	752606	99
Hypospadias, second degree with chordee	752626	78
Hypospadias, third degree	752607	13
Hypospadias, third degree with chordee	752627	22
Indeterminate sex, NOS / ambiguous genitalia	752790	15
Kidney: Double or triple, pelvis / pyelon duplex or triplex	753310	10
Kidney: Ectopic / pelvic	753330	7
Kidney: Lobulated, fused, or horseshoe / crossed fused ectopia	753320	14

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2004-2005

BPA label (cont'd)	BPA Code	# of Defects
Kidneys: Multicystic renal dysplasia / multicystic kidney	753160	22
Kidneys: Polycystic, infantile type (IPKD)	753110	4
Megaloureter, NOS / hydroureter	753220	22
Obstruction, atresia or stenosis of anterior urethra	753620	1
Other specified anomalies of bladder and urethra	753880	4
Penis: Other anomalies / concealed penis / absent or hooded foreskin	752860	75
Penis: Small / hypoplastic / micropenis	752865	11
Renal agenesis, bilateral	753000	5
Renal agenesis, right + renal hypoplasia, left	753007	1
Renal hypoplasia, bilateral	753005	1
S absence of testis / monorchidism, NOS	752800	3
S kidney: Other specified anomalies	753380	3
Testis and scrotum: Other anomalies / polyorchidism / bifid scrotum	752820	13
Urachus: Cyst	753710	6
Urachus: Patent	753700	7
Ureter: Accessory / double ureter / duplex collecting system	753410	21
Ureter: Ectopic	753420	5
Ureter: Other specified anomalies / ureterocele	753480	18
Ureter: Variations of vesicoureteral reflux	753485	48
Urethra: Congenital posterior urethral valves or posterior urethral obstruction	753600	23

Musculoskeletal

= Amyotrophia congenita (= one specific type of arthrogryposis)	756840	1
= Unspecified chondrodystrophy. Excludes: lipochondrodystrophy (use 277.510)	756490	1
Absence of foot or toes, bilateral	755349	1
Absence of foot or toes, left	755346	9
Absence of foot or toes, right	755347	2
Absence of hand or fingers, left	755246	20
Absence of hand or fingers, right	755247	6
Absence of lower leg only (thigh and foot present), right	755337	1
Absence of the forearm and hand, Left	755241	4
Absence of the forearm and hand, right	755242	1
Absence of the lower leg and foot, right	755342	2
Achondroplasia	756430	7
Anomalies of fingers / camptodactyly/ macro – / brachy – / clino – , triphalangeal thumb	755500	50
Arthrogryposis multiplex congenita / distal arthrogryposis syndrome. Includes: one or more flexion contractures of individual joints	755800	24
Cleidocranial dysostosis	755555	1
Clubfoot, NOS / talipes, NOS	754730	81
Clubfoot: Metatarsus varus or adductus	754520	14
Clubfoot: Talipes calcaneovarus	754510	1
Clubfoot: Talipes equinovarus	754500	80
Congenital postural scoliosis	754200	22
Craniosynostosis, coronal, bilateral	756014	4
Craniosynostosis, coronal, laterality unk	756010	2
Craniosynostosis, Coronal, Left	756011	9

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2004-2005

BPA label (cont'd)	BPA	# of
Craniosynostosis, coronal, right	756012	7
Craniosynostosis, coronal, unilateral, side unk	756013	1
Craniosynostosis, lambdoidal, left	756021	1
Craniosynostosis, metopic	756006	11
Craniosynostosis, sagittal	756005	36
Diaphragmatic hernia, Bochdalek, left	756611	4
Diaphragmatic hernia, esophageal	756605	1
Diaphragmatic hernia, Morgagni, laterality unk	756615	2
Diaphragmatic hernia, morgagni, left	756616	2
Diaphragmatic hernia, NOS (inc. absent/hemidiaphragm), laterality unk	756600	1
Diaphragmatic hernia, NOS (inc. absent/hemidiaphragm), left	756601	21
Diaphragmatic hernia, NOS (inc. absent/hemidiaphragm), right	756602	14
Diaphragmatic hernia, NOS (inc. absent/hemidiaphragm), unilat, side unk	756603	1
Eventration of diaphragm	756620	4
Fibular aplasia/hypoplasia, right	755372	1
Gastroschisis	756710	48
Limb deficiencies, lower limb, not elsewhere classifiable, bilateral	755384	1
Limb deficiencies, upper limb, not elsewhere classifiable, bilateral	755284	1
Longitudinal deficiency of arm, NOS, bilateral	755254	1
Longitudinal deficiency of leg, NOS, left	755351	1
Lower limb: hypoplasia / toes, feet, legs: hypoplasia. Excludes: aplasia of or absent lower limb	755685	12
Lower limb: other specified anomalies / hyperextended legs / shortening of legs	755680	5
Omphalocele	756700	14
Osteogenesis imperfecta	756500	5
Other absent or hypoplastic muscle / absent pectoralis major. Excludes: prune belly syndrome (use 756720)	756810	1
Other specified chondrodystrophy. Excludes: Conradi's (use 756575)	756480	6
Other specified deformity of hands (see 755.500 for specified anomalies of fingers)	754880	2
Other specified osteodystrophies	756580	3
Poland syndrome or anomaly	756800	2
Polydactyly fingers / postaxial polydactyly, type A	755005	39
Polydactyly: Accessory big toe (preaxial)	755030	10
Polydactyly: Accessory digits foot, NOS (preaxial, postaxial not specified)	755096	2
Polydactyly: Accessory thumbs (preaxial polydactyly)	755010	45
Polydactyly: Accessory toes (postaxial)	755020	39
Prune belly syndrome	756720	2
Radial aplasia/hypoplasia, bilateral	755269	3
Radial aplasia/hypoplasia, left	755266	5
Ribs: Absence	756300	10
Ribs: Extra	756330	15
Ribs: Fused	756320	12
Ribs: Other anomalies	756340	5
S anomalies of forearm, NOS	755530	2
S clubfoot: Complex varus deformities	754530	1
S congenital deformities of foot, NOS	754735	2
S knee: anomalies / hyperextended knee	755640	1
S unspecified varus deformities of feet	754590	1
Sacral agenesis	756175	3

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2004-2005

BPA label (cont'd)	BPA Code	# of Defects
Skull and face bone: Other specified anomalies / localized skull defects / mid – facial hypoplasia / prominent maxilla/hypotelorism / flat occiput / prominent occiput	756080	7
Spine: Kyphosis / kyphoscoliosis	756120	8
Split – foot, bilateral	755359	2
Split – hand, left	755256	1
Sternum: Other anomalies / double ossification center in manubrium / bifid/ short	756380	3
Syndactyly: Fused fingers	755100	26
Syndactyly: Fused toes	755120	37
Syndactyly: Unspecified (see below for specified site)	755190	4
Syndactyly: Unspecified (webbed vs. fused) thumb and / or fingers, NOS	755193	12
Syndactyly: Unspecified (webbed vs. fused), digits not known	755199	1
Syndactyly: Unspecified toes	755194	4
Syndactyly: Webbed fingers	755110	11
Syndactyly: Webbed toes / webbing between the second and third toes	755130	25
Talipes calcaneovalgus	754600	11
Thanatophoric dwarfism	756447	1
Thumb only missing or hypoplastic, bilateral	755264	2
Thumb only missing or hypoplastic, Left	755261	7
Thumb only missing or hypoplastic, right	755262	2
Tibial aplasia/hypoplasia, right	755367	1
Total absence of the leg, left	755306	1
Ulnar aplasia/hypoplasia, right	755272	1
Upper limb: Hypoplasia / fingers, hands, or arms: hypoplasia. Excludes: aplasia or absent upper limb (see 7552)	755585	20
Upper limb: Other specified anomalies / hyperextensibility of upper limb / shortening of upper limb	755580	6
Vertebrae, cervical: anomalies	756140	5
Vertebrae, cervical: hemivertebrae	756145	1
Vertebrae, lumbar: anomalies	756160	7
Vertebrae, lumbar: hemivertebrae	756165	11
Vertebrae, sacrococcygeal: anomalies / agenesis of sacrum	756170	7
Vertebrae, thoracic: anomalies	756150	16
Vertebrae, thoracic: hemivertebrae	756155	22
Vertebrae: Other specified anomalies	756180	8

Chromosomal and Other Syndromes

= 22q11 deletion (Added 7/04: apply to 1/01. Also code phenotype if stated, eg. DGS 279.110)	758370	11
= Additional sex chromosomes, NOS	758860	1
= Deletion: unspecified autosome	758390	1
= Down syndrome: translocation trisomy 21 – duplication 21q	758020	1
= Klinefelter phenotype: other karyotype with additnl X chrome, e.g., XXXY, XXYY, XXXXY	758710	1
= Sturge – Weber syndrome/ encephalocutaneous angiomatosis/	759610	5
= Trisomy 8	758500	1
= Turner phenotype: unspecified karyotype / Bonneville – Ullrich syndrome	758690	1
Apert syndrome / Acrocephalosyndactyly types I or II	756055	3
Autosome NOS: Other spec anomalies / marker / 8/02: Ring, derivative, mosaic, isochrome, "additional" material / 3/03 inversions	758580	13
Conjoined twins: Other specified	759480	2
Deletion 17p or 18p / deletion of short arm chromosome 17 or 18	758350	1

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2004-2005

BPA label (cont'd)	BPA Code	# of Defects
Deletion 17q or 18q / deletion of the long arm of chromosome 17 or 18	758340	2
Deletion 4p / Wolff – Hirschorn syndrome	758320	1
Deletion 5p / cri du chat syndrome	758310	3
Deletion: Autosome (not X or Y)(ie. #1 – 16, 4q,5q,19,20) / 8/02 Include microdeletion / 10/02: use for 22q11 (also code phenotypes DGS and VCFS, if available)	758380	9
DiGeorge syndrome: 10/02: Code specific phenotype + chrome/FISH 22, if available	279110	11
Down syndrome: mosaic	758040	5
Down syndrome: translocation trisomy – duplication of a G group chromosome, NOS	758030	2
Down syndrome: trisomy 21	758000	176
Goldenhar syndrome / oculoauriculovertebral dysplasia	756060	7
Hemifacial microsomia	756065	16
Klinefelter syndrome: 47, XXY	758700	8
Malf OS: VATER/VACTERL/acardia/ angelman/bloom/CHARGE/hemihyper/Meckel – Gruber/Neu – Laxova/PentalogyCantrell/ Sotos/ TownesBrock/ WalkerWarburg/ Weaver / 10/02 VCFS,Shprintzen	759890	19
Malf. Syndromes/face: Aarskog /BOF /BOR /Fraser /FreemanSheldon / Kabuki / Miller – Dieker/ Noonan /Opitz G / oral – facial – digita/ Oto – palato – digital / Septo – optic dysplasia / Waardenburg / Williams	759800	22
Malf. Syndromes/limbs: Baller – Gerold/ Carpenter / caudal regression /Fryns/ Holt – Oram / Klippel – Trenaunay – Webe/ LimbBodyWall /Roberts/ Rubinstein – Taybi / sirenomelia / thrombocytopenia – absent radius	759840	5
Malf. Syndromes/metabolic: Alagille /Alport / Beckwith – Wiedemann / Johansen – Blizzard/ leprechaunism / Lowe/ Menkes(kinky hair) /Prader – Willi/ Zellweger	759870	17
Malf. Syndromes/other skeletal: Marfan / Stickler/ Beemer Langer	759860	5
Malf. Syndromes/short stature: Smith – Lemli – Optiz /de Lange / Cockayne / Laurence – Moon – Biedl / Russell – Silver / Seckel	759820	6
Mosaic XO/XX. Excludes: with Turner phenotype (758610)	758810	1
Mosaic XO/XY, 45X/46XY. Excludes: with Turner phenotype (758610)	758800	3
Other craniofacial syndromes / Hallermann – Streiff syndrome	756046	2
Other specified acrocephalosyndactylies	756057	4
Sex chromosome: Other specified anomaly / fragile X	758880	3
Treacher – Collins syndrome / Mandibulofacial dysostosis	756045	1
Trisomy 13: Patau syndrome	758100	11
Trisomy 18: Edwards syndrome	758200	22
Trisomy, partial / 8/02 "partial trisomy" = "duplication". But, for "dup NOS" use 758930	758530	6
Tuberous sclerosis / Bourneville's disease	759500	9
Turner syndrome: karyotype 45,X [XO] Note: The 7586xx code series that follows excludes pure gonadal dysgenesis(752720)	758600	5
Turner syndrome: variant karyotypes, eg. isochromosome, mosaic (eg X, XX,XY), partial X deletion, ring X chromosome. Excludes: Turner phenotype with normal karyotype	758610	10
Unbalanced translocations, OS. Excludes: bal trans in normal (75.400)	758540	4
XXX female / 47XXX / Triple X syndrome	758850	3
XYY, male / 47,XYY / mosaic XYY male	758840	1

Other

= Collodion baby	757110	1
= X – linked ichthyosis	757196	1
Adrenogenital syndrome / adrenal hyperplasia	255200	1
Amniotic band sequence	658800	15
Anomalies of thymus / absent thymus / thymichypertrophy	759240	6
Epidermolysis bullosa	757330	1
Hamartomas: Other specified	759680	3
Ichthyosiform erythroderma	757197	2

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2004-2005

BPA label (cont'd)	BPA Code	# of Defects
Multiple congenital anomalies (In MA, ="MCA NOS", not "MCA no specific dx") / anomaly, multiple, NOS / deformity, multiple, NOS	759700	1
Other and unspecified ichthyosis	757190	1
Skin: Other specified anomalies / scalp defects. For specified anomalies of skin see 757390. For specified anomalies of hair, see 757480. For specified anomalies of nails 757580	757800	3
Spleen: Absence / asplenia	759000	6
Spleen: Accessory / 8/02 Use for polysplenia, though not exactly the same	759040	10

Birth Defects by Severity

Severe, supportive measures, usually incompatible with life

Anencephaly

Bilateral renal agenesis

Trisomy 13

Trisomy 18

Severe identifiable syndrome or condition, not elsewhere classified

Severe isolated defects, not elsewhere classified

Multiple severe defects, (Severe MCA,NEC)

Serious, may be correctable, most have long – term needs

Achondroplasia

Aniridia

Anophthalmia

Arthrogryposis

Biliary atresia

Bladder exstrophy

Cloacal exstrophy

CHD, multiple mod – severe, not elsewhere classified

Double outlet right ventricle

Encephalocele

Heterotaxy with CHD

Holoprosencephaly

Hypoplastic left heart syndrome

Limb reductions, mod – severe

Osteogenesis imperfecta

Sacral agenesis, caudal regression, sirenomelia

Single ventricle

Spina bifida

Amniotic band complex

Down syndrome

Mod serious syndrome/condition, not elsewhere classified

Mod serious defect, not elsewhere classified

Mod – severe multiple defects, (Mod – severe MCA,NEC)

Moderate, most correctable, many have long – term needs

Aortic valve stenosis

Atrial septal defect

Atrioventricular canal AVC / AVSD / ECD

Choanal atresia

Cleft lip/ palate

Coarctation

Cataract, glaucoma

Clubfoot

Coloboma

CHD, Mult mild – mod not, listed elsewhere

Moderate, most correctable, many have long – term needs (cont'd)

Craniosynostosis

Dandy – Walker malformation

Diaphragmatic hernia

Esophageal atresia/ TEF

Ebstein anomaly

Gastroschisis

Genitourinary, obstructive

Hirschsprung disease

Penis, buried, hidden

Hydrocephalus

Hypospadias, 2nd or 3rd degree

Intestinal atresia: duod, jejunal, ileal

Imperforate anus/rectal atresia and stenosis

Interrupted aortic arch

Klinefelter syndrome

Limb reductions, mild – mod

Malrotation

Microcephaly

Microtia

Omphalocele

Pulm sequestration/ CCAM

Pulmonary atresia/stenosis

Tethered cord

Tetralogy of Fallot

Total /partial anom. pulm venous return

Transposition great arteries

Tricuspid atresia/stenosis

Turner syndrome

Ventricular septal defect

Moderate syndrome/condition, not elsewhere classified

Moderate defect, not elsewhere classified

Moderate multiple severe defects, (Moderate MCA,NEC)

DiGeorge/ VCF/ 22q11 del spectrum

Goldenhar/FAVS/ OAVD

Mild, may be correctable, minimal long – term needs

Bicuspid aortic valve

Meckel's diverticulum

Microphthalmia

CHD, OS, asymptomatic

Patent ductus arteriosus

Polydactyly, accessory thumbs, syndactyly

Heterotaxy without CHD, Situs inversus totalis without CHD, Situs inversus abdominis, isolated dextrocardia

Mild defect, not listed above

Glossary of Selected Birth Defects Terms[1]

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

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

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

Anencephaly: Congenital absence of the skull and brain.

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

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

Anotia: Congenital absence of the ear.

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

Arthrogryposis: Multiple congenital contractures of various joints.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Hypertrophy: Overgrowth due to enlargement of existing cells.

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

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

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

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

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

Meninges: Membranes that cover the brain and spinal cord.

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

Microphthalmia: Congenital smallness of the eye globe.

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

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

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

NOS: Not Otherwise Specified

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

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

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

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

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

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

Renal agenesis: Congenital absence of the kidney.

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

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

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

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

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

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

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

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

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

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

Trisomy 21: See **Down syndrome**.

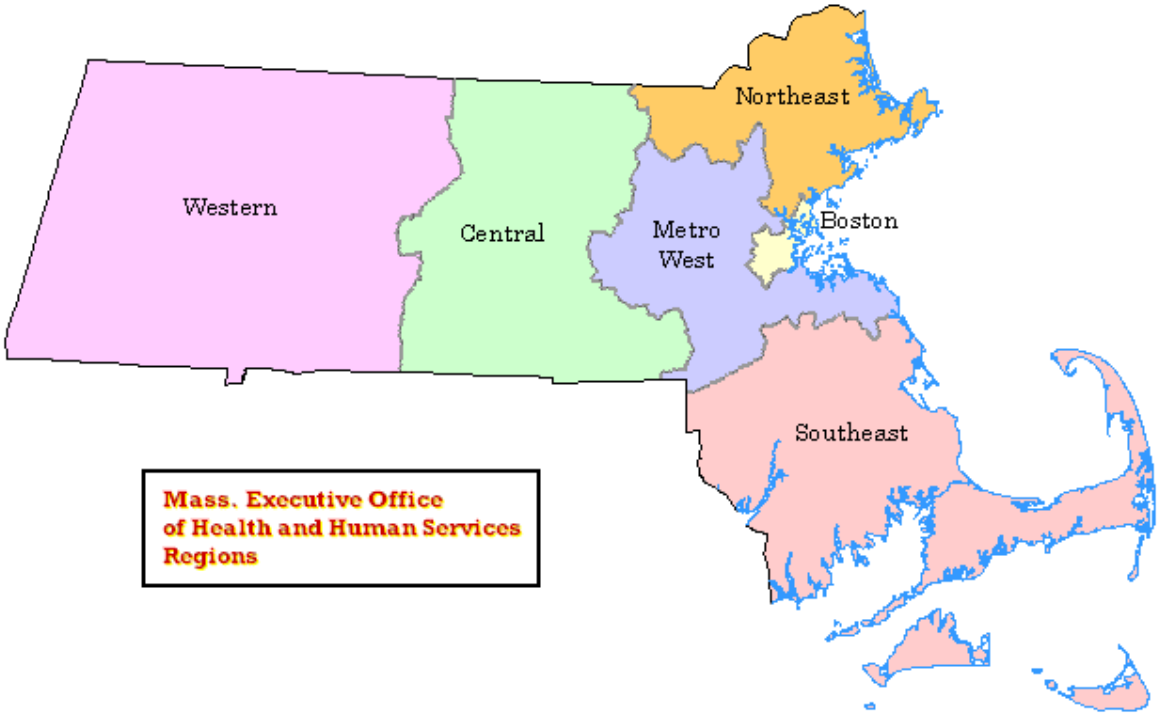
Truncus arteriosus: Congenital heart defect characterized by a single great arterial trunk, instead of a separate aorta and pulmonary artery.

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

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

Map of the Six Regions Designated by the Massachusetts EOHHS[1]

1. Obtained from the MA Executive Office of Health & Human Services (EOHHS) website (www.mass.gov/mgis/reg_eohhs.htm)



Folic Acid Awareness and Behavior in Women Ages 18-44, Massachusetts 2000 and 2004[1]

Survey Year, Age Group	Recognized that taking folic acid can prevent birth defects (%)	Take folic acid daily (%)
2000		
18 – 24	35.8	35.0
25 – 29	57.6	40.7
30 – 34	64.9	46.9
35 – 39	61.6	45.3
40 – 44	60.3	45.7
2004		
18 – 24	NA[2]	NA[2]
25 – 29	NA[2]	53.8
30 – 34	75.3	57.1
35 – 39	78.6	53.6
40 – 44	63.6	55.1

1. Data obtained from women surveyed by the Behavioral Risk Factor Surveillance System (BRFSS), maintained by the Massachusetts Department of Public Health, Bureau of Health Statistics, Research and Evaluation. The difference in the data may be associated with demographical factors such as age, gender and race/ethnicity of the respondent groups.
2. Underlying sample size is less than 50 respondents (insufficient data).

Frequency of Multivitamin Use during Month Prior to Pregnancy, Massachusetts Jan – Mar 2005[1]

Frequency of multivitamin or prenatal vitamin consumption	% of respondents (N=348)
None	42.5
1 – 3 times per week	12.4
4 – 6 times per week	9.1
Every day of the week	35.8
Don't know/blank	0.2

1. Data obtained from resident women 18 years or older who had given live birth between January to March 2005. The survey was conducted by Massachusetts for a pilot study as part of the CDC Pregnancy Risk Assessment Monitoring System (PRAMS).

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Birth Defects Monitoring Program
Bureau of Family Health & Nutrition
Massachusetts Department of Public Health
250 Washington Street, 5th floor, Boston, MA 02108