
Massachusetts Birth Defects Among Live and Stillbirths 2008-2009



Massachusetts Birth Defects Monitoring Program
Bureau of Family Health and Nutrition

Massachusetts Department of Public Health

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<http://www.mass.gov/dph/birthdefects>

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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.

<u>Section</u>	<u>Change or addition</u>
Chapter 1	This chapter contains a description of changes to the Massachusetts legislative regulations regarding case ascertainment and abstraction and the major ways in which ascertainment has been enhanced beginning with 2008 births.
Chapter 2	The methods section on <i>Data Collection</i> was updated to include enhancements that have resulted from the implementation of new legislative regulations.
Chapter 3	A figure depicting the trend in the overall prevalence of defects (Figure 1) was added. Table 5 was reconfigured/enhanced to reflect the counts of birth defects by <i>pattern of occurrence</i> rather than by cases with “one defect” compared to those with “two or more defects”.
Appendix	A table was added describing the distribution of folic acid use across a number of different demographic characteristics including race/ethnicity, maternal age, maternal education, household poverty level, and maternal nativity

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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 second leading cause of death in the first year of life, the third largest cause of death between the ages of one and fourteen, and in the top ten among 15-24 year olds in Massachusetts. Nationally, about 20% of all infant deaths—defined as the first year of life—result from a birth defect. In Massachusetts, 15.0% of all infant deaths were attributable to birth defects (MADPH 2012).

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 Centers for Disease Control and Prevention (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 cost for babies born with any of the 12 major structural birth defects was estimated to be \$133 million in 2007 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 and occupational limitations due to early death of or care for a child. The psychosocial costs cannot be calculated.

Over the past thirteen 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 such as phenylketonuria and congenital hypothyroidism are monitored separately by the state newborn screening program.

This report presents statewide data on the prevalence of birth defects among live births and stillbirths in Massachusetts during the years 2008 and 2009. The first

annual report presented Massachusetts birth defects data for the year 1999. Four subsequent reports compiled data from 2000-2001, 2002-2003, 2004-2005, and 2006-2007, respectively. Our ability to find and identify infants born with birth defects to Massachusetts residents has improved over time and is reflected in increasing prevalence rates year-on-year (Figure 1). The 2008-2009 data are presented in combined form since the numbers are relatively small for individual defects. As in previous years, given the BDMP's continued enhancement to birth defects ascertainment, interpretations of these data with respect to previous reports must be made with caution.

The data allow for some preliminary trend analyses and evaluation of the efficacy of public health 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 such as Early Intervention helps to ensure services for identified children and to provide population-based information to guide future program planning and prevention strategies.

Prevalence

The overall prevalence of birth defects among births to Massachusetts residents in 2008-2009 was 175.3 per 10,000 live births. Among the 151,935 live births and 798 still births to Massachusetts residents in 2008-2009, 2,609 (0.4%) live birth and 55 (6.9%) stillbirths had one or more birth defects. Though population-based, the decade of active, statewide surveillance conducted from 1999 through 2009 only provides baseline frequencies for birth defects. More years of data will establish the stability of these baseline frequencies. The ten most common defects were unchanged from the 2006-2007 report. Three of these ten 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 (2nd or 3rd degree), 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 21 selected defects. Massachusetts was one of 11 states with population-based monitoring programs to contribute birth defect data. These average prevalence rates cover deliveries from 2004-2006 (CDC 2010). When compared to other states' birth defects surveillance programs that utilize active case ascertainment, Massachusetts rates for 2008-2009 were significantly lower than the U.S. rates for approximately 40% of the defects and were about the same as the national estimates for the other 60%. 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.

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 2008-2009. Of infants born with birth defects, 46.6% were C-section deliveries, compared to 33.3% of non-birth defect births; 20.8% of birth defect cases were of low birthweight (<2500 grams; 5.5 lbs) as opposed to 7.5% of those without a birth defect; 20.2% of infants were premature (gestational age < 37 weeks) compared with 8.5% of those without a birth defect; and 5.1% of infants with a birth defect died before their first birthday, compared to 0.4% of those without a birth defect. While the number of infants with birth defects is relatively small, it is important to recognize the impact of these outcomes when diagnosing and treating a baby with a birth defect.

Sex

The birth defect case prevalence was 144.6 per 10,000 live births for females and 203.5 for males. While the prevalence of most types of birth defects did not significantly differ by sex of the infant/fetus, some did. The most common defects seen in males, in descending order of prevalence were hypospadias (2nd or 3rd degree), atrial septal defects (secundum and NOS), polydactyly/syndactyly, obstructive genitourinary defect, and clubfoot. The most common defects seen in females were atrial septal defects (secundum and NOS), ventricular septal defects (membranous and NOS), polydactyly/syndactyly, Down syndrome, and obstructive genitourinary defect.

Plurality

Examining birth defects by plurality is important since birth defects are more common among multiple births (twins, triplets, etc.), and the number of multiple births has increased in Massachusetts since 1994. Plurality information obtained from reviewing the medical record differed slightly from the plurality recorded on the birth and fetal death records. Because medical record abstraction may reveal early losses not recorded at birth and is therefore more accurate, plurality from the medical record abstraction is used in this report. When using the medical record, the birth defect case prevalence was 171.2 for singletons and 260.1 for multiple

births per 10,000 live births, so the risk of a baby who was born as part of a set of twins, triplets, etc. is significantly higher than a baby born alone.

Birth defects that occurred most often among multiple births (all of which occurred more often than in singleton births) were atrial septal defects (secundum and NOS), ventricular septal defects (membranous and NOS), hypospadias (2nd and 3rd degree), clubfoot, and obstructive genitourinary defect.

Analysis of Trends

The overall prevalence of birth defects in Massachusetts was 5.5% higher from 2008 to 2009 compared to 2006 to 2007 (overall rate, 175.3 and 166.1 per 10,000 live births, respectively). 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 change in individual birth defect rates. Trends in selected 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 is one defect that displayed an increase between 2000 and 2006, but it appears to have remained stable since that point. This trend parallels the national trend and may reflect the aggressive strategies for and improvements in surveillance.

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 178.1 for mothers younger than 20 years, 172.1 for those 20-24 years, 165.7 for those 25-29 years, 153.8 for those 30-34 years and 199.6 for those 35 years and older. Mothers younger than the age of 20 had the highest rate (18.6 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 only about half of babies with Down syndrome were born to women 35 years or older, the Down syndrome rate of 29.9 per 10,000 births for women 35 years and older was about four times that of mothers under the age of 35 years (7.6 per 10,000 live births). The pattern of higher Down syndrome rates among babies born to older women reflects the pattern of higher chromosomal defects in general among the oldest maternal age group.

In 2009, Massachusetts had the highest proportion of U.S. births conceived through assisted reproductive technology (ART) procedures among state residents (4.3% of MA births). Massachusetts also had the fourth-highest number of total ART births in the U.S. with 9,845. This high frequency of ART procedures may be due in part to more complete insurance coverage in Massachusetts (Sunderam et al. 2008). 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 overall prevalence of birth defects varied by maternal race and Hispanic ethnicity, but differences among groups were not statistically significant. The age-adjusted birth defect prevalence per 10,000 live births was 168.8 for non-Hispanic whites, 200.6 for non-Hispanic blacks, 139.1 for non-Hispanic Asians/Pacific Islanders and 182.8 for Hispanics. Due to small numbers, the rates for other races were not calculated individually. There were very few differences among the most commonly occurring defects according to race and ethnicity. All groups had atrial septal defects (secundum and NOS) and polydactyly/syndactyly amongst the most common defects. Hypospadias (2nd and 3rd degree) and obstructive genitourinary defect were in all but Hispanics' most commonly occurring defects. Though Asians had generally lower age-adjusted rates of chromosomal defects, the difference was not statistically significant when compared to the other racial/ethnic groups. This finding was different from the 2004-2005 report when the rate for white, non-Hispanics was about half the rate compared to black, non-Hispanics and Hispanics. Maternal birthplace (U.S. versus non-U.S.) may be a contributing factor in group differences, as women born in the U.S. had slightly higher birth defect rates than those born outside the U.S. 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 about the same as the rates of women born in the U.S. (200.1 and 196.9 per 10,000 live births, respectively).

Region

The birth defect rates among six Massachusetts regions in 2008-2009 were not statistically significantly different. The age-adjusted rates ranged from 163.3 per 10,000 live births in the Western region to 171.7 per 10,000 in the Boston region.

Severity

A severity scale was developed by the Center in collaboration with our partners at Boston University and the Massachusetts General Hospital. This scale was based on the usual outcome for a specific birth defect including its typical compatibility with survival, the need for immediate treatment, the need for long-term care and the amenability of the defect to correction. We developed an automated algorithm to classify the majority of cases into the categories of "severe", "serious", "moderate", and "mild" with a minority of cases requiring assignment by a clinician. 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 and lack of prenatal diagnosis reporting. 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). 18.4% 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. "Moderate" birth defects comprised about

70% of the total cases. All of these needed medical follow up and many may have required a number of surgeries and extensive treatment. "Mild" birth defects comprised a little over 9% of the affected infants. These defects may or may not have required corrective treatment.

Etiology and Pattern

The surveillance system in Massachusetts has allowed for the collection of relevant etiology (cause) information. Cases with known etiology accounted for about 18.4% of the birth defects in Massachusetts in 2008-2009. Of the 489 cases with known cause, "single gene" etiology accounted for 21.3%, "chromosomal" etiology accounted for 73.8% and "maternal-fetal factors" accounted for 2.7% of cases. The majority of birth defects cases in Massachusetts in 2008-2009 had an unknown etiology (81.6%).

Cases are also classified by the *pattern* of defects (i.e. whether one defect occurs with others). Of all 2,664 birth defect cases (2,609 live births and 55 stillbirths) 43.4% had a "solitary" (truly a single) defect pattern, 26.1% had "Major plus minors" (defined as having a major defect accompanied by one or more minor defects), 4.6% were a "sequence" (allowing for more than one major defect if the defects are related pathogenically), and 25.9% had "multiple major" defects. Cleft lip with and without cleft palate, hypospadias (2nd or 3rd degree), gastroschisis, and craniosynostosis appeared more often as a single defect rather than with other defects—all occurring more than 75% of the time as a "solitary" defect. Birth defects which appeared more often in conjunction with other defects included hydrocephaly, spina bifida, anotia/microtia, obstructive genitourinary defect, and all cardiovascular defects.

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.

Suggested Citation

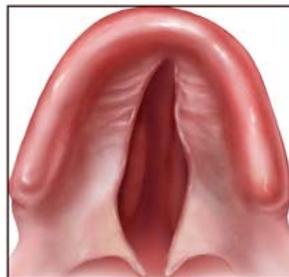
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Chapter 1

Introduction



Baby with cleft palate



Cleft palate

Courtesy of the Centers for Disease Control and Prevention

The Public Health Importance of Birth Defects

Each year in the United States, approximately 120,000 babies—1 in 33—are born with birth defects (MarchofDimes 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 thousand 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 can 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, 15% of all infant deaths were attributable to birth defects (MADPH 2012). The overall infant mortality rate for Massachusetts in 2009 was 4.9 per 1,000 births, and for the U.S. it was 6.3 per 1,000 births (Kochanek et al. (2011).

Causes of Birth Defects

The causes of most birth defects are poorly understood, but certain genetic and environmental factors have been associated with selected defects. These include prenatal environmental factors, such as infections (e.g., rubella), exposures to medications or other chemicals, drug or alcohol abuse, and nutritional deficiencies.

A single abnormal gene can cause certain birth defects. The gene may have an error in its code such as a missing piece or extra genetic material which can result in malformations. Other causes of birth defects may be multifactorial with genes and environmental factors both playing a role. For 70% of major birth defects, no known cause has been identified (CDC 2006). Chapter 6 presents data that most birth defect cases in Massachusetts have an 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 “periconceptual” period, the month before and three months after conception. For the developing pregnancy, “the environment” includes any exposure to the fetus as well as 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 “periconceptual” 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 in the US (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), administered 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 reported that taking folic acid can prevent birth defects. Also, 51.6% of the participating women ages 18-49 reported that they take folic acid on a daily basis (see table in Appendix).

An additional and potentially more relevant indicator of folic acid intake can be multivitamin use among women who recently had a live birth. The Massachusetts Pregnancy Risk Assessment Monitoring System (PRAMS) survey of resident women who had a live birth includes questions about prenatal multivitamin use. 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. Among respondents, 37.6% reported that they took multivitamins daily in the month prior to pregnancy. (MA PRAMS Report 2010; these data—stratified by a number of demographic categories—are presented in the 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). Additionally, the draft version of the new Healthy People 2020 adds an objective to increase the 1-year survival rates of children born with Down syndrome. 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);
- 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 eleven 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 medical 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, estimated the lifetime cost for families dealing with a baby with birth defects to be between \$75,000 and \$503,000 (CDC 1995). Their estimated lifetime cost for a baby born with spina bifida was \$385,896 in 2007.

Adjusting for inflation, the estimated combined lifetime cost in Massachusetts for babies born with 12 major structural birth defects was \$133 million in 2007 (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, which cannot be quantified.

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 extended the definition of 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.

Update on the Implementation of the Regulations

In the winter of 2009, Massachusetts enacted regulations (105 CMR 302) related to the Massachusetts Birth Defects Monitoring Program. Among its provisions, the regulations expanded the reporting requirements for birth defects cases identified at or after birth extended reporting to cases identified prenatally, and established an Advisory Committee.

Since the enactment of these regulations, the MBDP has been working on their implementation. Hospitals across the state were notified of the new requirements, and the expanded post-natal reporting was implemented at these sites with the utilization of a revised Confidential Reporting and Abstracting Form (CRAF). An advisory group of obstetrical and radiological clinicians was convened to develop the specifications related to prenatal reporting, and the MBDP visited Massachusetts tertiary hospitals to determine where and how birth defects are diagnosed prenatally. The implementation of the prenatal specifications began in January 2011. As such, the addition of prenatal report is not reflected in the 2008-2009 report. However, the enhancements to case ascertainment of additional reporting sites (emergency rooms, day surgeries, and outpatient centers) and standardized electronic reports is likely reflected, at least in part, in the increase in overall birth defect prevalence compared to the previous report for 2006-2007.

An advisory committee, comprised of patients, families, health care providers, researchers, and other interested parties, was also established. Advisory committee meetings have been held twice per year for a total of six times as of December 2012.

The 2008-2009 Surveillance Report

This report presents statewide data on the prevalence of birth defects in live births and stillbirths in Massachusetts during the years 2008 and 2009. 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, third, fourth, and fifth reports compiled data from 2000-2001, 2002-2003, 2004-2005, and 2006-2007 respectively. Our ability to find and identify infants born with birth defects to Massachusetts residents has improved over time. The approximate prevalence increases of 12% from 2000-

2001 to 2002-2003, and 9% from 2002-2003 to 2004-2005 are attributable to improved case ascertainment. The prevalence of all birth defects for this report (175.3/10,000 live births), is, similarly, slightly higher than the 2006-2007 report, increasing by 5.5%. Interpretations of these data must be made with caution as the continuing refinement and enhancement of our surveillance methods may have a continuing affect on the completeness of our case ascertainment and the subsequent estimation of defect prevalence.

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 (0.77 lbs).

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 2008 through 2009 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 (0.77 lbs).
- 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 population-based surveillance methods for statewide case ascertainment. Hospitals across the state submitted monthly discharge lists with birth defect diagnoses to the Center. Nursery and neonatal intensive care liaisons faxed or phoned in reports of birth defects. Abstractors reviewed medical charts for each potential case. Beginning with 2008 births, new regulations were implemented expanding the reporting sites to include outpatient centers, emergency rooms, day surgery, and hospital labs). Standardized electronic reporting from sites also began rolling out in 2008. Below is a list of selected enhancements made since the inception of the program:

- 2001 Massachusetts Eye & Ear Institute (specialty care) in Boston added
Women & Infants Hospital (maternity) and Hasbro Hospital (tertiary) in Rhode Island added to capture Southeastern Massachusetts out-of-state births and/or birth defect diagnosis
- 2002 Additional CNS diagnosis added
- 2005 Reviewed surveillance criteria that resulted in additions/clarifications resulting in increased ascertainment
- 2006 Additional GI diagnosis added
- 2007 Added DNA-based diagnosis and updated BPA/ICD-9 codes based on national surveillance guidelines
- 2009 New regulations promulgated affecting surveillance reporting: additional codes reported to capture existing reportable diagnoses, expanded site reporting (outpatient, ER, day surgery), and standardized electronic reporting from institutions.

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

The Center has developed extensive procedures to guarantee the confidentiality of the data and protect the privacy of families. These procedures uphold the Center's 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 race/ethnicity, and birth weight 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 2008-2009 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 rates in the tables is a range of possible values around the point estimate that has a 95% chance of including the actual 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 2008 through 2009. Due to the small numbers of birth defects and the fact that the BDMP data set is still relatively new, conclusions from these results—including inferences about multi-year trends—are not valid until a more extensive multi-year estimate establishes a stable, baseline rate.

2. Defects that are not diagnosed at birth and that do not need hospitalization may be underreported. Additionally, the diagnoses made after the first year of life and those made solely via prenatal testing could be missed.

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 at 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 losses (Forrester, Merz et al. 1998; Ethen and Canfield 2002).

6. Only diagnoses confirmed up to 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 may be detected by accident when a child is symptomatic (Holmes 1994). Many defects are also detected when diagnostic tests are performed to confirm a more severe, accompanying defect. Finally, many defects that result in developmental delays (e.g. fetal alcohol syndrome) may not be detected until those delays become evident as a child is gets 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 received special permission and 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 population demographics, living environment, and variances in surveillance/case ascertainment methods may contribute to differences in the prevalence of birth defects.

Glossary

A glossary of selected birth defect terms is included in the appendix 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 151,935 live births to Massachusetts residents in 2008-2009, 2,664 had one or more structural birth defects that were ascertained by Massachusetts BDMP. Among these, 55 stillbirths were identified with a birth defect. Overall, 1.8% of births in the state (175.3 per 10,000 live births) were identified as having at least one birth defect. The majority of defects occurred in the cardiovascular (occurring in 33.4% of defect cases) and musculoskeletal (occurring in 28.2%) categories. Figure 2 shows the percentage of reported birth defects by defect category. 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 2004-2006 (CDC 2010). Massachusetts rates for 2008-2009 were significantly lower than the US rates for approximately 40% of the defects and were about the same as the national estimates for the other 60% (see Table 2). Differences in rates may reflect variations in defect criteria between surveillance systems as well as regions. Also, in Massachusetts, birth defects are not reported when they are prenatally diagnosed and the pregnancy is electively terminated, which would tend to result in lower rates 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 per 10,000 live births. Massachusetts does not collect data on elective terminations and has a spina bifida prevalence of 2.17 per 10,000 live births for 2008-2009 which is over 60% lower than the national rate (see Table 2).

The CDC estimates that up to 50% of pregnancies with neural tube defects and up to 80% of pregnancies with anencephaly are electively terminated after prenatal diagnosis (Cragan and Khoury 2000). Substantial evidence from past studies has 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's 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 number 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 2008-2009 Massachusetts rates for anencephaly, spina bifida, trisomy 13, and trisomy 18 were significantly lower than the national estimates. The rate for Down syndrome was similar to the national estimate (Table 2). Using 2008-2009 rates, an estimated 50-72% of anencephaly cases are missed through exclusion of terminated cases. Upon adjusting the Massachusetts rate of anencephaly, two out of the three adjusted Massachusetts rates of anencephaly appear similar to the national estimate while the third, though lower, still is not significantly lower when assessing overlapping confidence intervals (see Appendix for description of confidence intervals). Likewise, with an estimated 29-46% of spina bifida cases missed through exclusion of terminated cases in surveillance, all three of the adjusted Massachusetts rates for spina bifida may also be similar to 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 do not appear to be statistically dissimilar 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 significantly exceeding the national estimate. Table 3 lists the adjusted rates. The adjusted rate of Down syndrome in Massachusetts that is higher than the national estimate possibly reflects the older average delivery age of pregnant women 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 prenatal diagnostic testing patterns and elective termination decision-making processes in the state. In recent years, coupled with the increasing numbers of pregnancies in older women, different diagnostic testing practices may have developed. Further evidence of these trends may be reflected in the Massachusetts Pregnancy Risk Assessment Monitoring System (PRAMS) survey data, collected from Massachusetts resident women who had a live born infant in 2009. Mothers of

singletons, 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. Of the survey respondents, 92.1% (N=1,388) replied that a healthcare provider had discussed the issue with them (MA PRAMS Report, 2012).

Figure 2 provides the overall prevalence of birth defects by interpregnancy interval (IPI), defined as the time period in completed months between the date of conception of one pregnancy and the date of delivery of the preceding pregnancy, among women whose preceding pregnancy resulted in a live birth. There were no clear trends in the overall prevalence of birth defects by IPI. However, this does not preclude the existence of trends for individual birth defects that may be obscured when looking at defects overall.

Defect Pattern

Table 5 shows the distribution of birth defects by *pattern*. Cases are classified by the pattern of defects (i.e. whether one defect occurs with others). Of all 2,664 birth defect cases (2,609 live births and 55 stillbirths), 43.4% had a “solitary” (truly a single) defect pattern, 26.1% had “Major plus minors” (defined as having a major defect accompanied by one or more minor defects), 4.6% were a “sequence” (allowing for more than one major defect if the defects are related pathogenically), and 25.9% had “multiple major” defects. Cleft lip with and without cleft palate, hypospadias (2nd or 3rd degree), gastroschisis, and craniosynostosis appeared more often as a single defect rather than with other defects—all occurring more than 75% of the time as a “solitary” defect. Birth defects which appeared more often in conjunction with other defects included hydrocephaly, spina bifida, anotia/microtia, obstructive genitourinary defect, and all cardiovascular defects.

Selected Pregnancy Outcomes

Figure 3 compares selected pregnancy outcome characteristics among infants born with birth defects to those born without birth defects in 2008-2009 by percentage. Of infants born with birth defects, 46.6% were delivered by Cesarean, compared to 33.3% of infants born with no birth defect births; 20.8% of infants with birth defects had low birthweight (<2,500 grams) as opposed to 7.5% of those without a birth defect; 20.2% of infants were premature (gestational age < 37 weeks), compared with 8.5% of those without a birth defect; 5.2% 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 144.6 for females and 203.5 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 (see Table 7). The most common defects seen in males, in order of prevalence beginning with the most prevalent defect, were hypospadias (2nd or 3rd degree), atrial septal defects (secundum and NOS), polydactyly/syndactyly, obstructive genitourinary defect, and clubfoot. The most common defects seen in females were atrial septal defects (secundum and NOS), ventricular septal defects (membranous and NOS), polydactyly/syndactyly, Down syndrome, and obstructive genitourinary defect. Selected birth defects by sex of infant are presented in Figure 4.

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 (1999-2004) 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 have contributed to any apparent increase in birth defect rates. This is evidenced by a steady increase in the overall prevalence of birth defects as seen in Figure 1. A comparison of selected cardiovascular birth defects rates from 2008-2009 to rates in previous 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.

Gastroschisis and hypospadias are some of the birth defects of interest due to increasing national prevalence trends. Surveillance in Massachusetts and elsewhere has suggested an increased risk of gastroschisis in pregnancies among very young women (under 20) up to 2006 with an apparent leveling of prevalence between 2006 and 2009, although the mechanisms for this are unknown. A recent study showed that although younger women had both a comparatively larger risk of gastroschisis and a greater prevalence of smoking, cigarette smoking had no effect on risk these younger. Smoking, however, increased the risk in older women, possibly due to the increased smoking history (Werler, Mitchell et al 2009). The rate of gastroschisis in Massachusetts increased 63.8% between 2000 and 2007 (Figure 5). This increasing trend paralleled increasing national rates of gastroschisis, although a factor in this increase may be better ascertainment of cases by the surveillance system in Massachusetts. This may have been due to aggressive strategies of case confirmation and improvements in surveillance, although over time, increases were observed in other locations around the U.S. and the world.

Table 1 Prevalence of Birth Defects, Massachusetts: 2008-2009

Defect ¹	Live birth Count	Stillbirth Count	Total Count	Rate per 10,000 Births	95% Confidence Interval
Total Cases	2609	55	2664	175.34	168.80-182.06
Central Nervous System: 243 cases					
Anencephaly	8	3	11	0.72	0.36-1.30
Encephalocele	3	0	3	0.20	0.04-0.58
Holoprosencephaly	9	1	10	0.66	0.32-1.21
Hydrocephaly w/o Spina Bifida	48	6	54	3.55	2.67-4.64
Microcephaly	25	0	25	1.65	1.06-2.43
Spina Bifida w/ and w/o Hydrocephaly	30	3	33	2.17	1.50-3.05
Spinal Cord	57	0	57	3.75	2.84-4.86
Other CNS ²	113	1	114	7.50	6.19-9.01
Eye: 90 cases					
Aniridia	1	0	1	0.07	0.00-0.37
Anophthalmia/Microphthalmia	22	0	22	1.45	0.91-2.19
Congenital Glaucoma, Congenital Cataract	46	0	46	3.03	2.22-4.04
Other Eye ²	47	0	47	3.09	2.27-4.11
Ear: 66 cases					
Anotia/Microtia	27	0	27	1.78	1.17-2.59
Other Ear ²	43	1	44	2.90	2.10-3.89
Cardiovascular: 890 cases					
Anomalous Pulmonary Venous Connection					
Total/Partial Anomalous Pulmonary Venous Connection	20	0	20	1.32	0.80-2.03
Atrioventricular Canal Defects					
ASD Primum	2	0	2	0.13	0.02-0.48
Common Atrium	10	0	10	0.66	0.32-1.21
Complete Atrioventricular Canal Defect	44	1	45	2.96	2.16-3.96
Endocardial Cushion (OS and NOS)	25	0	25	1.65	1.06-2.43
VSD, Canal Type	3	0	3	0.20	0.04-0.58
Conotruncal (Outlet) and Aortic Arch					
Double Outlet Right Ventricle	15	0	15	0.99	0.55-1.63
Interrupted Aortic Arch, Type B	5	0	5	0.33	0.11-0.77
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	60	0	60	3.95	3.01-5.08

Table 1 Prevalence of Birth Defects, Massachusetts: 2008-2009

Defect ¹	Live birth Count	Stillbirth Count	Total Count	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>					
Truncus	4	0	4	0.26	0.07-0.67
d-Transposition of the Great Arteries	39	1	40	2.63	1.88-3.58
<i>Ebstein Anomaly</i>					
Ebstein Anomaly	4	0	4	0.26	0.07-0.67
<i>Heterotaxy (Laterality Defects)</i>					
Heterotaxy	24	0	24	1.58	1.01-2.35
<i>Left-Sided Obstruction</i>					
Aortic Valve Stenosis	21	0	21	1.38	0.86-2.11
Coarctation of Aorta	65	1	66	4.34	3.36-5.53
Hypoplastic Left Heart Syndrome	18	2	20	1.32	0.80-2.03
Interrupted Aortic Arch (Type A and NOS)	1	0	1	0.07	0.00-0.37
<i>Patent Ductus Arteriosus</i>					
Patent Ductus Arteriosus	221	0	221	14.55	12.69-16.60
<i>Right-Sided Obstruction</i>					
Pulmonary Stenosis, Valvular	95	0	95	6.25	5.06-7.64
Pulmonary Valve Atresia w/intact septum	10	0	10	0.66	0.32-1.21
Pulmonary Valve Atresia with VSD	3	0	3	0.20	0.04-0.58
Tricuspid Valve Atresia	13	0	13	0.86	0.46-1.46
<i>Septal Defects</i>					
ASD (Secundum and NOS)	338	0	338	22.25	19.94-24.75
VSD (Membranous and NOS)	184	7	191	12.57	10.85-14.49
VSD, Conoventricular/Malalignment	27	2	29	1.91	1.28-2.74
<i>Single Ventricle and L-TGA</i>					
L-TGA	5	0	5	0.33	0.11-0.77
Single Ventricle	4	0	4	0.26	0.07-0.67
<i>Other Cardiovascular</i>					
Other Cardiovascular ²	305	6	311	20.47	18.26-22.87
<i>Respiratory: 53 cases</i>					
Choanal Atresia	10	0	10	0.66	0.32-1.21
Lung Anomalies	23	0	23	1.51	0.96-2.27
Other Respiratory ²	20	2	22	1.45	0.91-2.19

Table 1 Prevalence of Birth Defects, Massachusetts: 2008-2009

Defect ¹	Live birth Count	Stillbirth Count	Total Count	Rate per 10,000 Births	95% Confidence Interval
<i>Orofacial: 286 cases</i>					
Cleft Lip w/ and w/o Cleft Palate	121	2	123	8.10	6.73-9.66
Cleft Palate w/o Cleft Lip	86	1	87	5.73	4.59-7.06
Pierre Robin Sequence	35	0	35	2.30	1.60-3.20
Other Orofacial ²	79	2	81	5.33	4.23-6.63
<i>Gastrointestinal: 243 cases</i>					
Biliary Atresia	10	0	10	0.66	0.32-1.21
Esophageal Atresia/Tracheoesophageal Fistula	37	0	37	2.44	1.71-3.36
Hirschsprung Disease	35	0	35	2.30	1.60-3.20
Rectal and Large Intestinal Atresia/Stenosis	41	1	42	2.76	1.99-3.74
Small Intestinal Atresia	43	2	45	2.96	2.16-3.96
Other Gastrointestinal ²	99	3	102	6.71	5.47-8.15
<i>Genitourinary: 554 cases</i>					
Bladder Exstrophy	3	0	3	0.20	0.04-0.58
Hypospadias, 2nd or 3rd Degree ³	191	0	191	24.49	21.14-28.22
Obstructive Genitourinary Defect	226	2	228	15.01	13.12-17.09
Renal Agenesis/Hypoplasia	7	2	9	0.59	0.27-1.12
Other Genitourinary ²	255	2	257	16.92	14.91-19.11
<i>Musculoskeletal: 751 cases</i>					
Club Foot	199	7	206	13.56	11.77-15.54
Craniosynostosis	65	1	66	4.34	3.36-5.53
Diaphragmatic Hernia	37	0	37	2.44	1.71-3.36
Gastroschisis	48	2	50	3.29	2.44-4.34
Omphalocele	17	1	18	1.18	0.70-1.87
Polydactyly/Syndactyly	240	5	245	16.13	14.17-18.28
Reduction Deformity, Lower Limbs	17	2	19	1.25	0.75-1.95
Reduction Deformity, Upper Limbs	29	2	31	2.04	1.39-2.90
Skeletal Dysplasia	18	1	19	1.25	0.75-1.95
Other Musculoskeletal ²	174	5	179	11.78	10.12-13.64
<i>Chromosomal and other Syndromes: 445 cases</i>					
Klinefelter Syndrome	7	0	7	0.46	0.19-0.95
Trisomy 13	4	1	5	0.33	0.11-0.77

Table 1 Prevalence of Birth Defects, Massachusetts: 2008-2009

Defect ¹	Live birth Count	Stillbirth Count	Total Count	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>					
Trisomy 18	19	7	26	1.71	1.12-2.51
Trisomy 21 (Down Syndrome)	183	9	192	12.64	10.91-14.56
Turner Syndrome	12	3	15	0.99	0.55-1.63
Other Chromosomal Syndromes/Other Syndromes ²	204	3	207	13.62	11.83-15.61
<i>Other: 54 cases</i>					
Amniotic Bands	10	3	13	0.86	0.46-1.46
Skin Anomalies	27	0	27	1.78	1.17-2.59
Other, Specified ²	17	0	17	1.12	0.65-1.79

- 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 Rate may represent a heterogeneous group of defects.
- ³ Rate calculated using male live births.

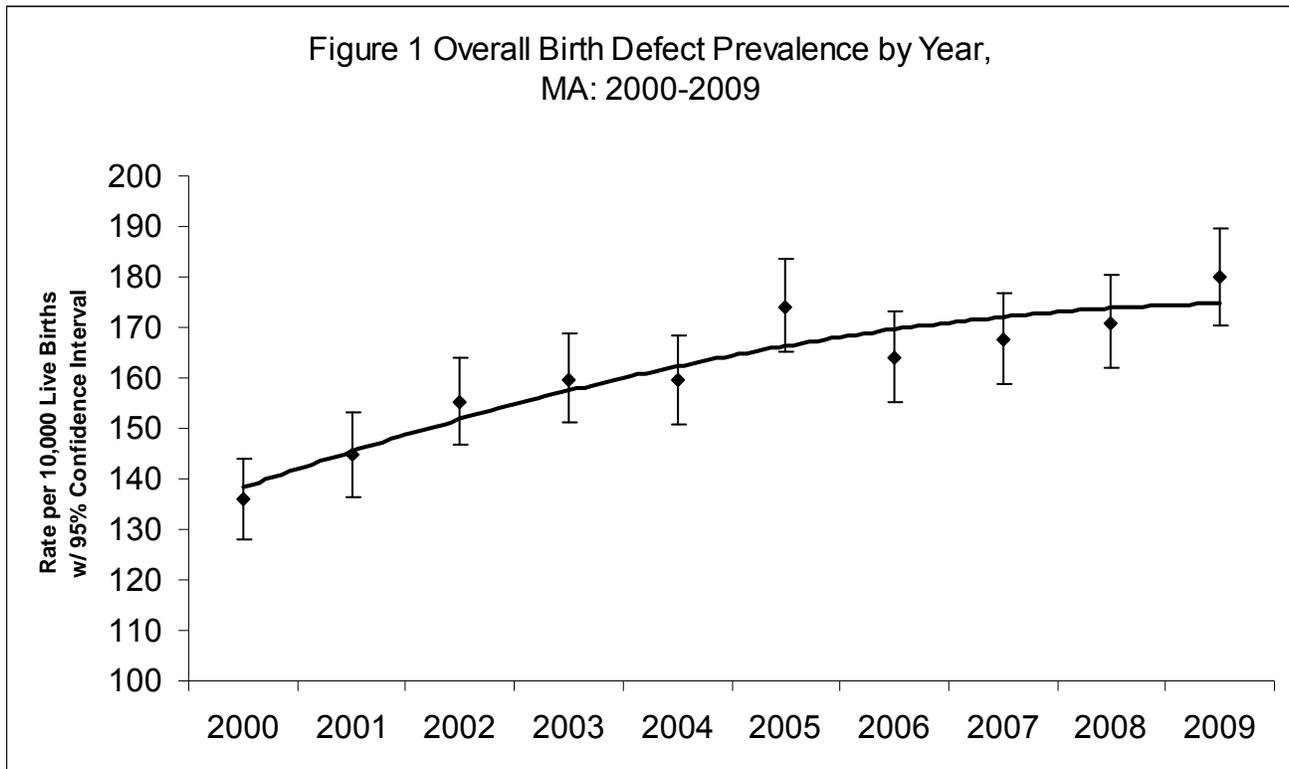


Table 2 Comparison of Selected Massachusetts 2008-2009 Birth Defect Rates to National Estimates

Defect	Count	Rate per 10,000 Births MA¹	95% Confidence Interval	Rate per 10,000 Births US²	95% Confidence Interval
Anencephaly	11	0.72	0.36 - 1.30	2.23	2.07 - 2.41
Spina bifida	33	2.17	1.50 - 3.05	3.72	3.52 - 3.94
Anophthalmia/microphthalmia	22	1.45	0.91 - 2.19	2.10	1.94 - 2.27
Truncus arteriosus (common truncus)	4	0.26	0.07 - 0.67	0.74	0.65 - 0.84
Transposition of the great arteries ³	45	2.96	2.16 - 3.96	3.04	2.85 - 3.24
Tetralogy of Fallot	60	3.95	3.01 - 5.08	4.05	3.83 - 4.28
Atrioventricular canal defect ⁴	85	5.59	4.47 - 6.92	4.70	4.45 - 4.96
Hypoplastic left heart syndrome	20	1.32	0.80 - 2.03	2.31	2.14 - 2.48
Cleft palate without cleft lip	87	5.73	4.59 - 7.06	6.45	6.17 - 6.74
Cleft lip with and without cleft palate	123	8.10	6.73 - 9.66	10.89	10.53 - 11.26
Esophageal atresia/tracheoesophageal fistula	37	2.44	1.71 - 3.36	2.12	1.96 - 2.29
Rectal and large intestinal atresia/stenosis	42	2.76	1.99 - 3.74	4.86	4.61 - 5.14
Reduction deformity, upper limbs	31	2.04	1.39 - 2.90	3.64	3.43 - 3.86
Reduction deformity, lower limbs	19	1.25	0.75 - 1.95	1.65	1.51 - 1.80
Gastroschisis	50	3.29	2.44 - 4.34	4.72	4.49 - 4.97
Omphalocele	18	1.18	0.70 - 1.87	1.92	1.77 - 2.08
Diaphragmatic hernia	37	2.44	1.71 - 3.36	2.60	2.42 - 2.79
Trisomy 21 (Down syndrome)	192	12.64	10.91 - 14.56	13.48	13.08 - 13.90
Trisomy 13	5	0.33	0.11 - 0.77	1.20	1.09 - 1.33
Trisomy 18	26	1.71	1.12 - 2.51	2.55	2.38 - 2.73

¹. MA rate is based on live births and stillbirths.

². Source: Updated National Birth Prevalence Estimates for Selected Birth Defects in the United States, 2004-2006. Birth Defects Research (Part A) **88**(12): 1008-1016. Only "active" surveillance system estimates are used and include systems that ascertain prenatally diagnosed and terminated pregnancies.

³. Includes d-TGA and L-TGA.

⁴. 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)

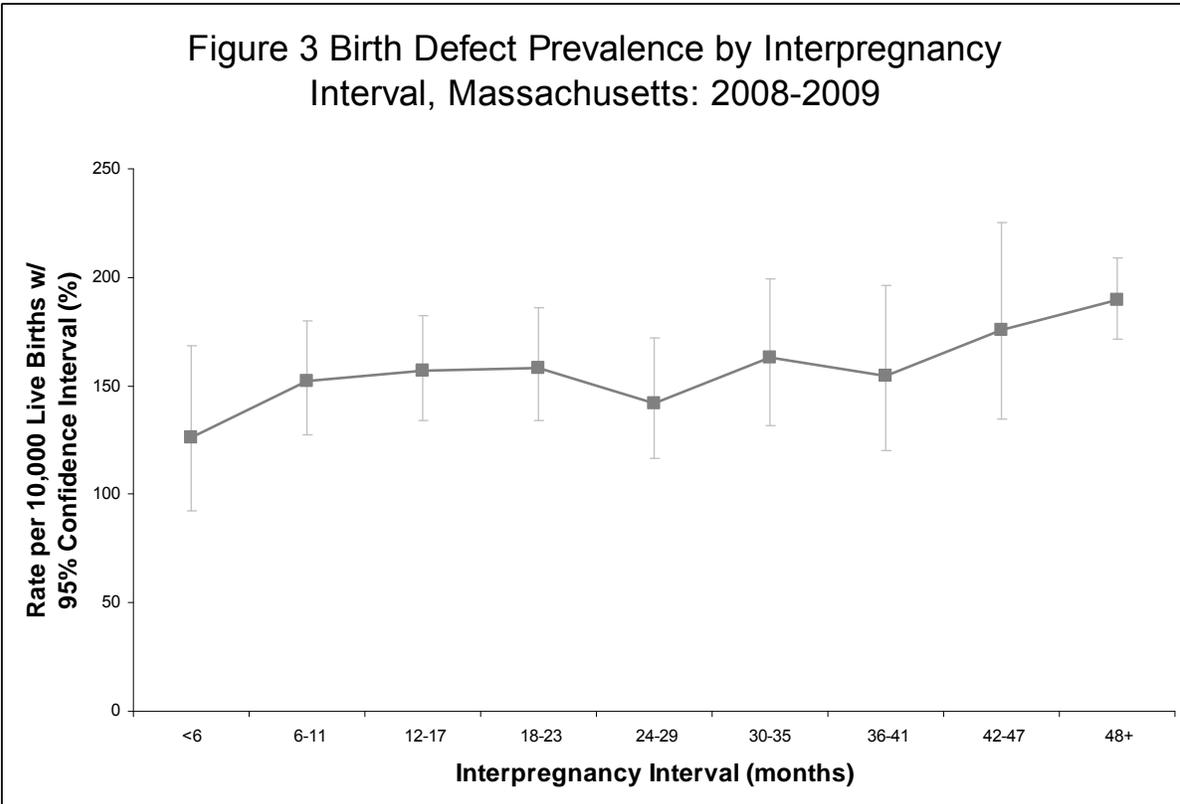
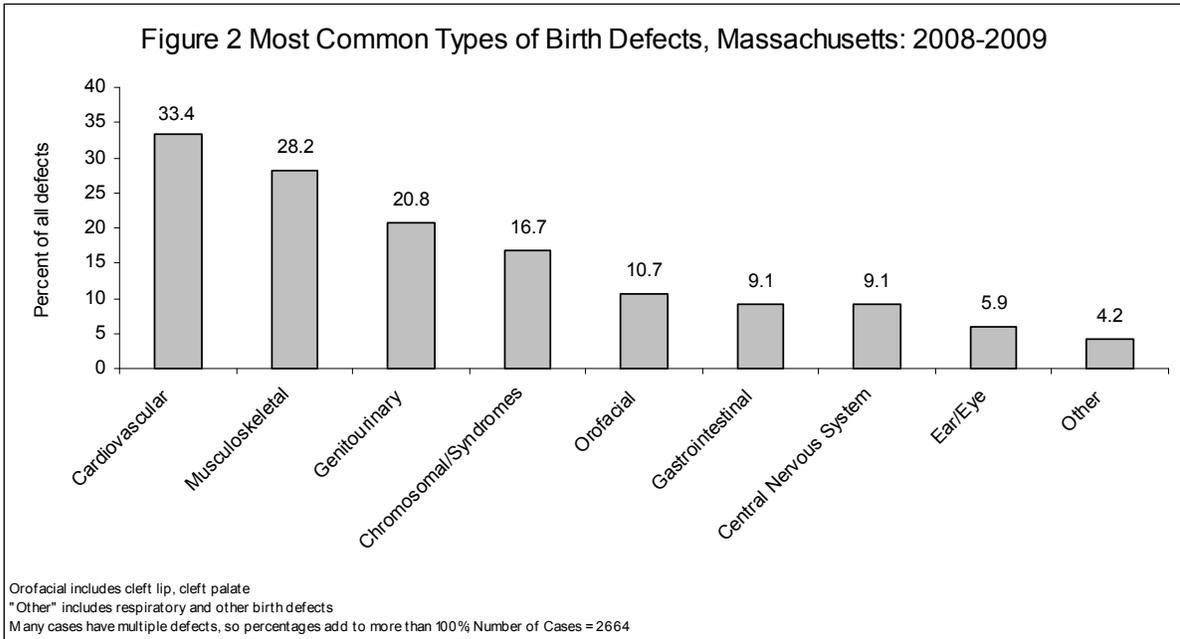
Defect	MA rate	95% Confidence Interval	Estimated Missing % ¹	Estimate Source ²	Adjusted MA rate ³	95% Confidence Interval	US rate ⁴	95% Confidence Interval
Anencephaly	0.72	0.36 – 1.30	66.7	Peller, et al.	2.17	1.50 - 3.05	2.23	2.07 – 2.41
			50.0	Cragan, et al.	1.45	0.91 - 2.19		
			72.0	Forrester, et al.	2.59	1.84 - 3.53		
Spina bifida	2.17	1.50 – 3.05	45.5	Peller, et al.	3.99	3.05 - 5.12	3.72	3.52 – 3.94
			29.0	Cragan, et al.	3.06	2.24 - 4.07		
			29.8	Forrester, et al.	3.09	2.27 - 4.11		
Trisomy 21 (Down syndrome)	12.64	10.91 – 14.56	35.2	Peller, et al.	19.50	17.34 - 21.85	13.48	13.08–13.90
			37.3	Forrester, et al.	20.15	17.96 - 22.54		
Trisomy 18	1.71	1.12 – 2.51	56.5	Peller, et al.	3.93	3.00 - 5.07	2.55	2.38 – 2.73
			49.0	Forrester, et al.	3.36	2.50 - 4.41		

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. Source: Updated National Birth Prevalence Estimates for Selected Birth Defects in the United States, 2004-2006. Birth Defects Research (Part A) 88(12): 1008-1016. Only "active" surveillance system estimates are used.



Interpregnancy Interval: the time period in completed months between the date of conception of one pregnancy and the date of delivery of the preceding pregnancy.
 Note: Prevalence estimates are among singleton births to multigravid women whose preceding pregnancy resulted in a live birth.

**Table 4 Most Common Defects among Live Births
and Stillbirths, Massachusetts: 2008 – 2009**

Defect¹	Category	Count	Rate per 10,000 Births	95% Confidence Interval
ASD (Secundum and NOS)	Cardiovascular	338	22.25	19.94-24.75
Polydactyly/Syndactyly	Musculoskeletal	245	16.13	14.17-18.28
Obstructive Genitourinary Defect	Genitourinary	228	15.01	13.12-17.09
Club Foot	Musculoskeletal	206	13.56	11.77-15.54
Trisomy 21 (Down Syndrome)	Chromosomal and other Syndromes	192	12.64	10.91-14.56
VSD (Membranous and NOS)	Cardiovascular	191	12.57	10.85-14.49
Hypospadias, 2nd or 3rd Degree	Genitourinary	191	24.49	21.14-28.22
Cleft Lip w/ and w/o Cleft Palate	Orofacial	123	8.10	6.73-9.66
Pulmonary Stenosis, Valvular	Cardiovascular	95	6.25	5.06-7.64
Cleft Palate w/o Cleft Lip	Orofacial	87	5.73	4.59-7.06

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

² Rate calculated using male live births.

Table 5 Counts of Birth Defects by Pattern among Live Births and Stillbirths, Massachusetts: 2008-2009

Defect ¹	Isolated Defects			Multiple Major Defects ³	Total Cases
	Solitary	Major + Minor(s) ²	Sequence		
<i>Central Nervous System</i>					
Anencephaly	9	1	0	1	11
Encephalocele	1	0	0	2	3
Holoprosencephaly	2	0	3	5	10
Hydrocephaly w/o Spina Bifida	18	11	1	24	54
Microcephaly	3	3	1	18	25
Spina Bifida w/ and w/o Hydrocephaly	1	1	24	7	33
Spinal Cord	5	11	15	26	57
Other CNS	23	34	10	47	114
<i>Eye</i>					
Aniridia	0	0	0	1	1
Anophthalmia/Microphthalmia	4	6	1	11	22
Congenital Glaucoma, Congenital Cataract	26	6	1	13	46
Other Eye	10	14	0	23	47
<i>Ear</i>					
Anotia/Microtia	5	9	2	11	27
Other Ear	3	9	0	32	44
<i>Cardiovascular</i>					
<i>Anomalous Pulmonary Venous Connection</i>					
Total/Partial Anomalous Pulmonary Venous Connection	2	13	3	2	20
<i>Atrioventricular Canal Defects</i>					
ASD Primum	0	2	0	0	2
Common Atrium	0	0	9	1	10
Complete Atrioventricular Canal Defect	1	5	6	33	45
Endocardial Cushion (OS and NOS)	0	8	3	14	25
VSD, Canal Type	0	1	0	2	3
<i>Conotruncal (Outlet) and Aortic Arch</i>					
Double Outlet Right Ventricle	0	9	4	2	15
Interrupted Aortic Arch, Type B	1	1	0	3	5
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	13	21	2	24	60
Truncus	1	2	0	1	4

Table 5 Counts of Birth Defects by Pattern among Live Births and Stillbirths, Massachusetts: 2008-2009

Defect ¹	Isolated Defects			Multiple Major Defects ³	Total Cases
	Solitary	Major + Minor(s) ²	Sequence		
<i>(cont'd)</i>					
d-Transposition of the Great Arteries	17	19	1	3	40
<i>Ebstein Anomaly</i>					
Ebstein Anomaly	1	3	0	0	4
Heterotaxy (Laterality Defects)					
Heterotaxy	1	0	20	3	24
<i>Left-Sided Obstruction</i>					
Aortic Valve Stenosis	2	13	2	3	20
Coarctation of Aorta	6	39	1	19	65
Hypoplastic Left Heart Syndrome	4	10	1	5	20
Interrupted Aortic Arch (Type A and NOS)	0	1	0	0	1
<i>Patent Ductus Arteriosus</i>					
Patent Ductus Arteriosus	15	80	4	122	221
<i>Right-Sided Obstruction</i>					
Pulmonary Stenosis, Valvular	30	50	3	12	95
Pulmonary Valve Atresia w/intact septum	3	5	1	1	10
Pulmonary Valve Atresia with VSD	0	3	0	0	3
Tricuspid Valve Atresia	1	9	0	3	13
<i>Septal Defects</i>					
ASD (Secundum and NOS)	54	141	6	137	338
VSD (Membranous and NOS)	21	96	2	73	192
VSD, Conoventricular/Malalignment	3	15	1	10	29
<i>Single Ventricle and L-TGA</i>					
L-TGA	0	4	1	0	5
Single Ventricle	1	3	0	0	4
<i>Other Cardiovascular</i>					
Other Cardiovascular	22	149	21	120	312
<i>Respiratory</i>					
Choanal Atresia	4	1	0	5	10
Lung Anomalies	14	3	1	5	23
Other Respiratory	3	2	2	15	22

Table 5 Counts of Birth Defects by Pattern among Live Births and Stillbirths, Massachusetts: 2008-2009

Defect ¹	Isolated Defects			Multiple Major Defects ³	Total Cases
	Solitary	Major + Minor(s) ²	Sequence		
<i>Orofacial</i>					
Cleft Lip w/ and w/o Cleft Palate	94	6	3	20	123
Cleft Palate w/o Cleft Lip	32	7	27	21	87
Pierre Robin Sequence	0	1	29	5	35
Other Orofacial	40	9	3	29	81
<i>Gastrointestinal</i>					
Biliary Atresia	5	1	1	3	10
Esophageal Atresia/Tracheoesophageal Fistula	10	1	0	26	37
Hirschsprung Disease	29	1	0	5	35
Rectal and Large Intestinal Atresia/Stenosis	14	3	1	24	42
Small Intestinal Atresia	16	10	1	18	45
Other Gastrointestinal	45	29	6	22	102
<i>Genitourinary</i>					
Bladder Exstrophy	2	1	0	0	3
Hypospadias, 2nd or 3rd Degree	154	12	0	25	191
Obstructive Genitourinary Defect	7	119	13	89	228
Renal Agenesis/Hypoplasia	0	1	7	1	9
Other Genitourinary	69	98	9	81	257
<i>Musculoskeletal</i>					
Club Foot	114	19	18	55	206
Craniosynostosis	50	0	0	16	66
Diaphragmatic Hernia	17	4	3	13	37
Gastroschisis	38	6	1	5	50
Omphalocele	6	4	0	8	18
Polydactyly/Syndactyly	110	68	7	60	245
Reduction Deformity, Lower Limbs	0	5	4	10	19
Reduction Deformity, Upper Limbs	9	4	6	12	31
Skeletal Dysplasia	0	0	0	19	19
Other Musculoskeletal	15	18	30	116	179
<i>Chromosomal and other Syndromes</i>					
Klinefelter Syndrome	0	0	0	7	7

Table 5 Counts of Birth Defects by Pattern among Live Births and Stillbirths, Massachusetts: 2008-2009

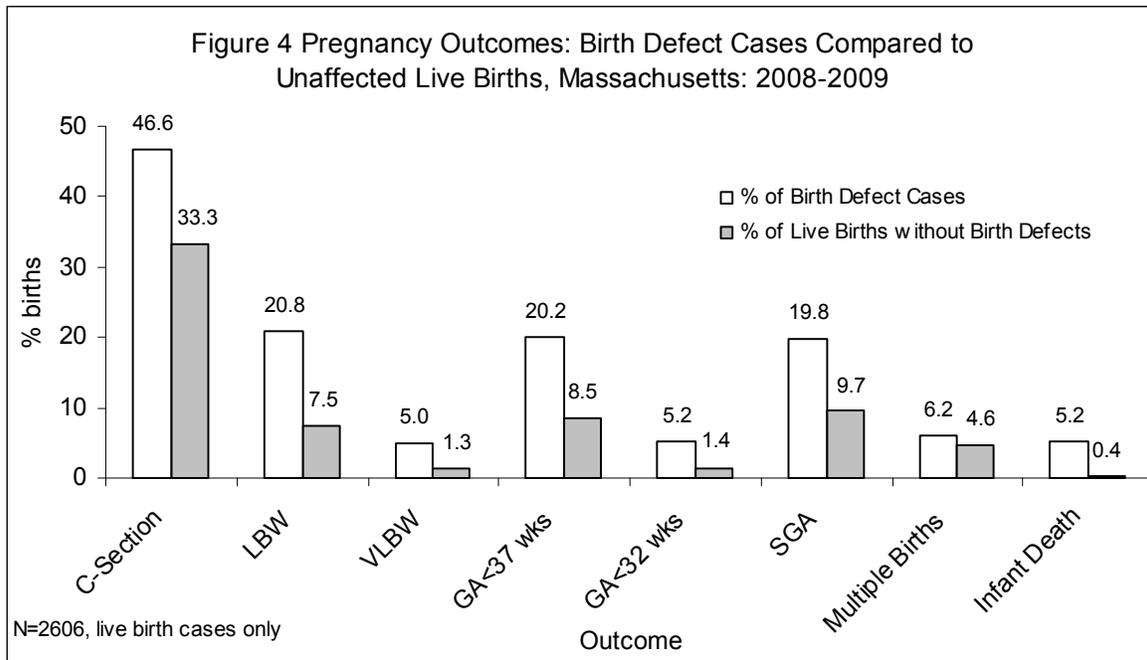
Defect ¹	Isolated Defects			Multiple Major Defects ³	Total Cases
	Solitary	Major + Minor(s) ²	Sequence		
<i>(cont'd)</i>					
Trisomy 13	0	0	0	5	5
Trisomy 18	0	0	0	26	26
Trisomy 21 (Down Syndrome)	0	0	0	192	192
Turner Syndrome	0	0	0	15	15
Other Chromosomal Syndromes/Other Syndromes	3	3	8	193	207
Other					
Amniotic Bands	0	0	11	2	13
Skin Anomalies	3	1	0	23	27
Other, Specified	2	2	7	6	17

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

² Major + Minor(s) includes Additive pattern.

³ Multiple major includes all recognized syndromes.

All pattern definitions can be found in the Appendix under *Technical Notes*



LBW: low birth weight; VLBW: very low birth weight; GA<37: gestational age less than 37 weeks; GA<32: gestational age less than 32 weeks; SGA: small-for-gestational age, defined as birth weight below the 10th percentile for gestational age on basis of a sex-specific US standard (Oken 2003)

Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2008-2009

Defect ¹	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
<i>Central Nervous System</i>				
Anencephaly	Male	9	1.15	0.53-2.19
	Female	2	0.27	0.03-0.98
Encephalocele	Male	0	0.00	0.00-0.47
	Female	3	0.41	0.08-1.19
Holoprosencephaly	Male	3	0.38	0.08-1.12
	Female	6	0.81	0.30-1.77
Hydrocephaly w/o Spina Bifida	Male	30	3.85	2.60-5.49
	Female	24	3.25	2.08-4.83
Microcephaly	Male	13	1.67	0.89-2.85
	Female	12	1.62	0.84-2.83
Spina Bifida w/ and w/o Hydrocephaly	Male	15	1.92	1.08-3.17
	Female	16	2.16	1.24-3.51
Spinal Cord	Male	32	4.10	2.81-5.79
	Female	25	3.38	2.19-4.99
Other CNS	Male	55	7.05	5.31-9.18
	Female	58	7.84	5.96-10.14
<i>Eye</i>				
Aniridia	Male	1	0.13	0.00-0.71
	Female	0	0.00	0.00-0.50
Anophthalmia/Microphthalmia	Male	10	1.28	0.61-2.36
	Female	12	1.62	0.84-2.83
Congenital Glaucoma, Congenital Cataract	Male	22	2.82	1.77-4.27
	Female	24	3.25	2.08-4.83
Other Eye	Male	22	2.82	1.77-4.27
	Female	25	3.38	2.19-4.99
<i>Ear</i>				
Anotia/Microtia	Male	14	1.80	0.98-3.01
	Female	13	1.76	0.94-3.01
Other Ear	Male	23	2.95	1.87-4.43
	Female	21	2.84	1.76-4.34

Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2008-2009

Defect ¹	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
<i>Cardiovascular</i>				
<i>Anomalous Pulmonary Venous Connection</i>				
Total/Partial Anomalous Pulmonary Venous Connection	Male	12	1.54	0.80-2.69
	Female	8	1.08	0.47-2.13
<i>Atrioventricular Canal Defects</i>				
ASD Primum	Male	1	0.13	0.00-0.71
	Female	1	0.14	0.00-0.75
Common Atrium	Male	2	0.26	0.03-0.93
	Female	8	1.08	0.47-2.13
Complete Atrioventricular Canal Defect	Male	19	2.44	1.47-3.80
	Female	26	3.52	2.30-5.15
Endocardial Cushion (OS and NOS)	Male	9	1.15	0.53-2.19
	Female	16	2.16	1.24-3.51
VSD, Canal Type	Male	3	0.38	0.08-1.12
	Female	0	0.00	0.00-0.50
<i>Conotruncal (Outlet) and Aortic Arch</i>				
Double Outlet Right Ventricle	Male	9	1.15	0.53-2.19
	Female	6	0.81	0.30-1.77
Interrupted Aortic Arch, Type B	Male	1	0.13	0.00-0.71
	Female	4	0.54	0.15-1.38
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	Male	39	5.00	3.56-6.84
	Female	21	2.84	1.76-4.34
Truncus	Male	1	0.13	0.00-0.71
	Female	3	0.41	0.08-1.19
d-Transposition of the Great Arteries	Male	26	3.33	2.18-4.89
	Female	14	1.89	1.03-3.18
<i>Ebstein Anomaly</i>				
Ebstein Anomaly	Male	2	0.26	0.03-0.93
	Female	2	0.27	0.03-0.98
<i>Heterotaxy (Laterality Defects)</i>				
Heterotaxy	Male	10	1.28	0.61-2.36
	Female	14	1.89	1.03-3.18

Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2008-2009

Defect ¹	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
<i>Left-Sided Obstruction</i>				
Aortic Valve Stenosis	Male	11	1.41	0.70-2.52
	Female	10	1.35	0.65-2.49
Coarctation of Aorta	Male	33	4.23	2.91-5.94
	Female	33	4.46	3.07-6.27
Hypoplastic Left Heart Syndrome	Male	10	1.28	0.61-2.36
	Female	10	1.35	0.65-2.49
Interrupted Aortic Arch (Type A and NOS)	Male	0	0.00	0.00-0.47
	Female	1	0.14	0.00-0.75
<i>Patent Ductus Arteriosus</i>				
Patent Ductus Arteriosus	Male	116	14.88	12.29-17.84
	Female	105	14.20	11.61-17.19
<i>Right-Sided Obstruction</i>				
Pulmonary Stenosis, Valvular	Male	43	5.51	3.99-7.43
	Female	52	7.03	5.25-9.22
Pulmonary Valve Atresia w/intact septum	Male	5	0.64	0.21-1.50
	Female	5	0.68	0.22-1.58
Pulmonary Valve Atresia with VSD	Male	2	0.26	0.03-0.93
	Female	1	0.14	0.00-0.75
Tricuspid Valve Atresia	Male	8	1.03	0.44-2.02
	Female	5	0.68	0.22-1.58
<i>Septal Defects</i>				
ASD (Secundum and NOS)	Male	162	20.77	17.70-24.23
	Female	176	23.80	20.41-27.59
VSD (Membranous and NOS)	Male	96	12.31	9.97-15.03
	Female	95	12.85	10.39-15.70
VSD, Conoventricular/Malalignment	Male	11	1.41	0.70-2.52
	Female	18	2.43	1.44-3.85
<i>Single Ventricle and L-TGA</i>				
L-TGA	Male	4	0.51	0.14-1.31
	Female	1	0.14	0.00-0.75

Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2008-2009

Defect ¹	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
Single Ventricle	Male	3	0.38	0.08-1.12
	Female	1	0.14	0.00-0.75
<i>Other Cardiovascular</i>				
Other Cardiovascular	Male	165	21.16	18.05-24.65
	Female	145	19.61	16.55-23.07
Lung Anomalies	Male	11	1.41	0.70-2.52
	Female	12	1.62	0.84-2.83
Other Respiratory	Male	9	1.15	0.53-2.19
	Female	13	1.76	0.94-3.01
<i>Orofacial</i>				
Cleft Lip w/ and w/o Cleft Palate	Male	82	10.52	8.36-13.05
	Female	41	5.54	3.98-7.52
Cleft Palate w/o Cleft Lip	Male	33	4.23	2.91-5.94
	Female	54	7.30	5.49-9.53
Pierre Robin Sequence	Male	13	1.67	0.89-2.85
	Female	22	2.97	1.86-4.50
Other Orofacial	Male	42	5.39	3.88-7.28
	Female	39	5.27	3.75-7.21
<i>Gastrointestinal</i>				
Biliary Atresia	Male	4	0.51	0.14-1.31
	Female	6	0.81	0.30-1.77
Esophageal Atresia/Tracheoesophageal Fistula	Male	18	2.31	1.37-3.65
	Female	19	2.57	1.55-4.01
Hirschsprung Disease	Male	28	3.59	2.39-5.19
	Female	7	0.95	0.38-1.95
Rectal and Large Intestinal Atresia/Stenosis	Male	26	3.33	2.18-4.89
	Female	16	2.16	1.24-3.51
Small Intestinal Atresia	Male	25	3.21	2.07-4.73
	Female	19	2.57	1.55-4.01
Other Gastrointestinal	Male	57	7.31	5.54-9.47
	Female	44	5.95	4.32-7.99

Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2008-2009

Defect ¹	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
<i>Genitourinary</i>				
Bladder Exstrophy	Male	3	0.38	0.08-1.12
	Female	0	0.00	0.00-0.50
Hypospadias, 2nd or 3rd Degree ³	Male	191	24.49	21.14-28.22
	Female	0	0.00	0.00-0.00
Obstructive Genitourinary Defect	Male	153	19.62	16.63-22.99
	Female	74	10.01	7.86-12.56
Renal Agenesis/Hypoplasia	Male	5	0.64	0.21-1.50
	Female	4	0.54	0.15-1.38
Other Genitourinary	Male	172	22.06	18.88-25.61
	Female	84	11.36	9.06-14.06
<i>Musculoskeletal</i>				
Club Foot	Male	130	16.67	13.93-19.80
	Female	74	10.01	7.86-12.56
Craniosynostosis	Male	43	5.51	3.99-7.43
	Female	23	3.11	1.97-4.67
Diaphragmatic Hernia	Male	21	2.69	1.67-4.12
	Female	15	2.03	1.14-3.35
Gastroschisis	Male	23	2.95	1.87-4.43
	Female	27	3.65	2.41-5.31
Omphalocele	Male	13	1.67	0.89-2.85
	Female	5	0.68	0.22-1.58
Polydactyly/Syndactyly	Male	154	19.75	16.75-23.13
	Female	91	12.31	9.91-15.11
Reduction Deformity, Lower Limbs	Male	11	1.41	0.70-2.52
	Female	8	1.08	0.47-2.13
Reduction Deformity, Upper Limbs	Male	16	2.05	1.17-3.33
	Female	15	2.03	1.14-3.35
Skeletal Dysplasia	Male	13	1.67	0.89-2.85
	Female	6	0.81	0.30-1.77
Other Musculoskeletal	Male	100	12.82	10.43-15.60
	Female	77	10.41	8.22-13.01

Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2008-2009

Defect ¹	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
<i>Chromosomal and other Syndromes</i>				
Klinefelter Syndrome	Male	7	0.90	0.36-1.85
	Female	0	0.00	0.00-0.50
Trisomy 13	Male	1	0.13	0.00-0.71
	Female	4	0.54	0.15-1.38
Trisomy 18	Male	9	1.15	0.53-2.19
	Female	16	2.16	1.24-3.51
Trisomy 21 (Down Syndrome)	Male	114	14.62	12.06-17.56
	Female	78	10.55	8.34-13.16
Turner Syndrome	Male	0	0.00	0.00-0.47
	Female	14	1.89	1.03-3.18
Other Chromosomal Syndromes/Other Syndromes	Male	101	12.95	10.55-15.74
	Female	105	14.20	11.61-17.19
<i>Other</i>				
Amniotic Bands	Male	6	0.77	0.28-1.67
	Female	7	0.95	0.38-1.95
Skin Anomalies	Male	11	1.41	0.70-2.52
	Female	16	2.16	1.24-3.51
Other, Specified	Male	7	0.90	0.36-1.85
	Female	10	1.35	0.65-2.49

¹ Cases can be included in more than one defect. Cases are counted once in the total for a defect category. Due to missing sex of infant counts may not match those in other tables.

² Rate may represent a heterogeneous group of defects.

³ Rate calculated using male live births.

**Table 7 Most Common Defects by Sex of Live Births
and Stillbirths, Massachusetts: 2008-2009**

Defect	Count	Rate per 10,000 Births	95% Confidence Interval
FEMALE			
ASD (Secundum and NOS)	176	23.80	20.41-27.59
VSD (Membranous and NOS)	95	12.85	10.39-15.70
Polydactyly/Syndactyly	91	12.31	9.91-15.11
Trisomy 21 (Down Syndrome)	78	10.55	8.34-13.16
Obstructive Genitourinary Defect	74	10.01	7.86-12.56
Club Foot	74	10.01	7.86-12.56
Cleft Palate w/o Cleft Lip	54	7.30	5.49-9.53
Pulmonary Stenosis, Valvular	52	7.03	5.25-9.22
Cleft Lip w/ and w/o Cleft Palate	41	5.54	3.98-7.52
Coarctation of Aorta	33	4.46	3.07-6.27
MALE			
Hypospadias, 2nd or 3rd Degree ²	191	24.49	21.14-28.22
ASD (Secundum and NOS)	162	20.77	17.70-24.23
Polydactyly/Syndactyly	154	19.75	16.75-23.13
Obstructive Genitourinary Defect	153	19.62	16.63-22.99
Club Foot	130	16.67	13.93-19.80
Trisomy 21 (Down Syndrome)	114	14.62	12.06-17.56
VSD (Membranous and NOS)	96	12.31	9.97-15.03
Cleft Lip w/ and w/o Cleft Palate	81	10.39	8.25-12.91
Pulmonary Stenosis, Valvular	43	5.51	3.99-7.43
Craniosynostosis	43	5.51	3.99-7.43

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

² Rate calculated using male live births.

Figure 5 Prevalence of Selected Birth Defects by Sex of Infant Among Live Births and Stillbirths, Massachusetts: 2008-2009

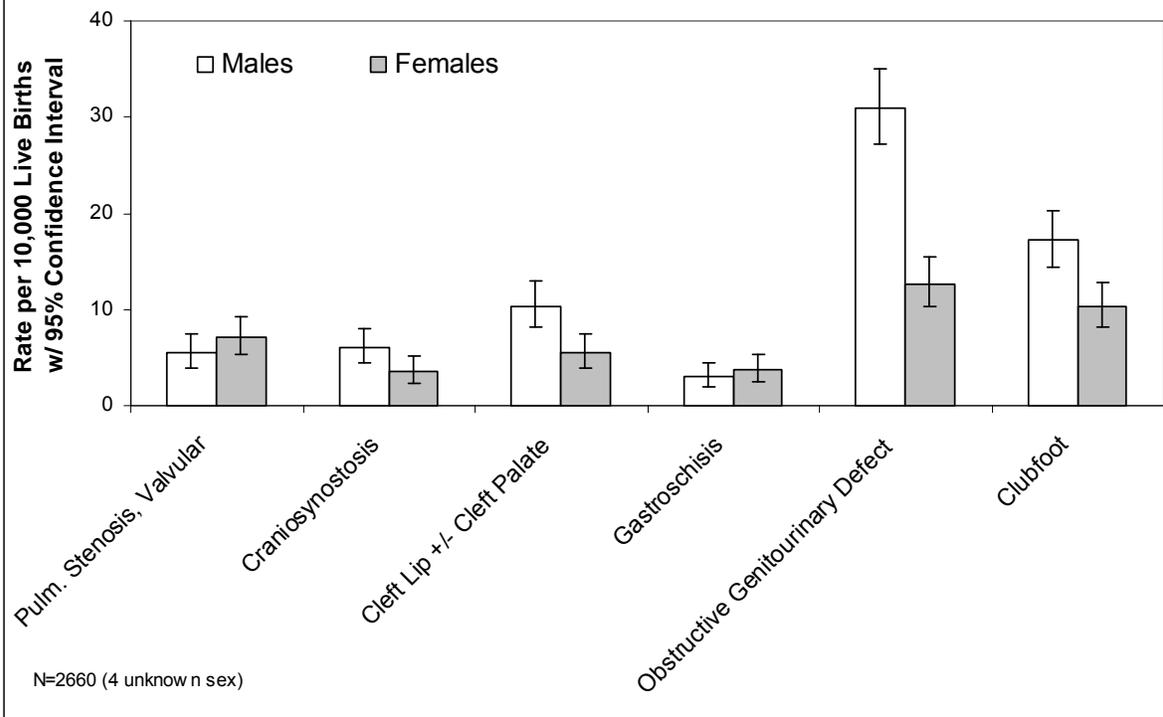
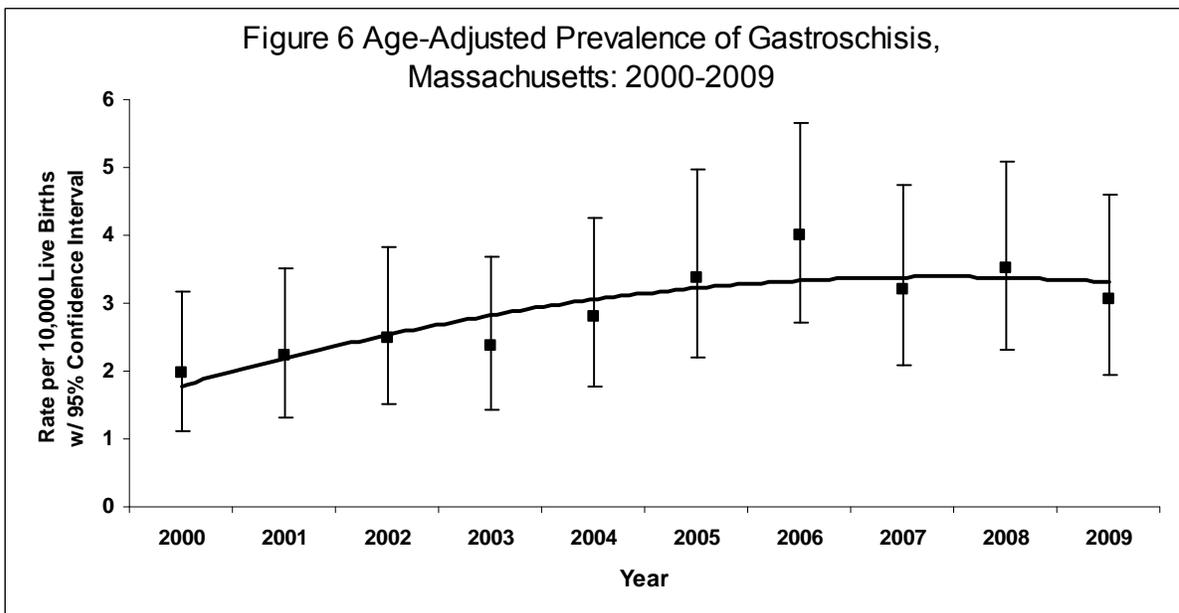


Figure 6 Age-Adjusted Prevalence of Gastroschisis, Massachusetts: 2000-2009



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. Plurality information obtained from reviewing the medical record differed slightly from the plurality recorded on the birth and fetal death records. Since medical record abstraction may reveal early losses not recorded at birth and is therefore more accurate, plurality from the medical record abstraction (rather than the birth certificate information) is used in this report. When using the medical record, the birth defect case prevalence was 171.2 for singletons and 260.1 for multiple births (more than one infant) per 10,000 live births. While multiple births comprised under 5% of all live births, they comprised almost 7% of birth defects cases among live births (see Figures 7a and 7b). Birth defects that occurred most often among multiple births (all of which occurred more often than in singleton births) were atrial septal defects (secundum and NOS), ventricular septal defects (membranous and NOS), hypospadias (2nd and 3rd degree), clubfoot, and obstructive genitourinary defect. Figure 8 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 rate of multiple births has been increasing in Massachusetts since 1994.

Maternal Age

The prevalence of birth defects varied by maternal age. For live births only, rates per 10,000 live births were 178.1 for mothers younger than 20 years, 172.1 for those 20-24 years, 165.7 for those 25-29 years, 153.8 for those 30-34 years and 199.6 for those 35 years and older.

As expected, there was a strong association between Down syndrome and advanced maternal age (see Figure 8). The Down syndrome rate of 28.4 per 10,000 live births for women 35 years and older was more than three times that of any other maternal age group. The pattern of higher Down syndrome rates among older women reflects the pattern of higher chromosomal defects in general among older women. Figure 9 shows the increase of chromosomal defects as maternal age increases; the rate of chromosomal defects among women in the 35+ age group is significantly higher than all other age groups. Additionally, as seen in Figure 10, the proportion of all birth defects that are chromosomal in nature was higher in the 35+ age group than in other age groups.

Figure 11 shows that younger mothers (aged 19 and under) had the highest rate of gastroschisis cases at 16.4 per 10,000 live births. 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 diaphragmatic hernia 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. Obstructive genitourinary defect, polydactyly/syndactyly, and atrial septal defect (secundum or NOS) were three of the most frequently occurring defects common to all maternal age groups, clubfoot was among the most common birth defects in all but the oldest 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 since the 1980s, and 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 now very similar (MADPH 2009).

The percentage of women giving birth in the state who are aged of 35 or over has doubled from 11.4% in 1989 to 22.3% in 2009. In addition, multiple births occurring in mothers aged 35 or over also doubled (3.4% to 7.5%) between 1989 and 2009 while multiple births to mothers under the age under 35 increased only slightly (2.3% to 3.9% of births to mothers under 35 years from 1989 to 2009, respectively) (MADPH 2009).

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 and 2004, a 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 fifth in the number of ART procedures performed in 2006 (8,305), after California, New York, Illinois and New Jersey. In 2006 Massachusetts had the highest ratio of the number of ART procedures among state residents at 1,291 per million residents. (Wright, Chang et al. 2008). 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 11 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 Plurality¹ of Live Births and Stillbirths, Massachusetts: 2008-2009

Defect ¹	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
<i>Central Nervous System</i>				
Anencephaly	Singleton	9	0.62	0.28-1.18
	Multiple	2	2.84	0.34-10.27
Encephalocele	Singleton	3	0.21	0.04-0.61
	Multiple	0	0.00	0.00-5.24
Holoprosencephaly	Singleton	10	0.69	0.33-1.27
	Multiple	0	0.00	0.00-5.24
Hydrocephaly w/o Spina Bifida	Singleton	49	3.38	2.50-4.47
	Multiple	5	7.11	2.31-16.58
Microcephaly	Singleton	24	1.66	1.06-2.46
	Multiple	1	1.42	0.04-7.92
Spina Bifida w/ and w/o Hydrocephaly	Singleton	29	2.00	1.34-2.87
	Multiple	4	5.68	1.55-14.55
Spinal Cord	Singleton	55	3.80	2.86-4.94
	Multiple	2	2.84	0.34-10.27
Other CNS ³	Singleton	107	7.38	6.05-8.92
	Multiple	7	9.95	4.00-20.50
<i>Eye</i>				
Aniridia	Singleton	1	0.07	0.00-0.38
	Multiple	0	0.00	0.00-5.24
Anophthalmia/Microphthalmia	Singleton	21	1.45	0.90-2.22
	Multiple	1	1.42	0.04-7.92
Congenital Glaucoma, Congenital Cataract	Singleton	44	3.04	2.21-4.08
	Multiple	2	2.84	0.34-10.27
Other Eye ³	Singleton	46	3.17	2.32-4.23
	Multiple	1	1.42	0.04-7.92
<i>Ear</i>				
Anotia/Microtia	Singleton	26	1.79	1.17-2.63
	Multiple	1	1.42	0.04-7.92
Other Ear ³	Singleton	43	2.97	2.15-4.00
	Multiple	1	1.42	0.04-7.92

Table 8 Prevalence of Birth Defects by Plurality¹ of Live Births and Stillbirths, Massachusetts: 2008-2009

Defect ¹	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
<i>Cardiovascular</i>				
<i>Anomalous Pulmonary Venous Connection</i>				
Total/Partial Anomalous Pulmonary Venous Connection	Singleton	20	1.38	0.84-2.13
	Multiple	0	0.00	0.00-5.24
<i>Atrioventricular Canal Defects</i>				
ASD Primum	Singleton	2	0.14	0.02-0.50
	Multiple	0	0.00	0.00-5.24
Common Atrium	Singleton	8	0.55	0.24-1.09
	Multiple	2	2.84	0.34-10.27
Complete Atrioventricular Canal Defect	Singleton	44	3.04	2.21-4.08
	Multiple	1	1.42	0.04-7.92
Endocardial Cushion (OS and NOS)	Singleton	24	1.66	1.06-2.46
	Multiple	1	1.42	0.04-7.92
VSD, Canal Type	Singleton	3	0.21	0.04-0.61
	Multiple	0	0.00	0.00-5.24
<i>Conotruncal (Outlet) and Aortic Arch</i>				
Double Outlet Right Ventricle	Singleton	15	1.04	0.58-1.71
	Multiple	0	0.00	0.00-5.24
Interrupted Aortic Arch, Type B	Singleton	5	0.35	0.11-0.81
	Multiple	0	0.00	0.00-5.24
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	Singleton	54	3.73	2.80-4.86
	Multiple	6	8.53	3.13-18.56
Truncus	Singleton	3	0.21	0.04-0.61
	Multiple	1	1.42	0.04-7.92
d-Transposition of the Great Arteries	Singleton	38	2.62	1.86-3.60
	Multiple	2	2.84	0.34-10.27
<i>Ebstein Anomaly</i>				
Ebstein Anomaly	Singleton	4	0.28	0.08-0.71
	Multiple	0	0.00	0.00-5.24
<i>Heterotaxy (Laterality Defects)</i>				
Heterotaxy	Singleton	21	1.45	0.90-2.22
	Multiple	3	4.26	0.88-12.46

Table 8 Prevalence of Birth Defects by Plurality¹ of Live Births and Stillbirths, Massachusetts: 2008-2009

Defect ¹	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
<i>Left-Sided Obstruction</i>				
Aortic Valve Stenosis	Singleton	20	1.38	0.84-2.13
	Multiple	1	1.42	0.04-7.92
Coarctation of Aorta	Singleton	59	4.07	3.10-5.25
	Multiple	7	9.95	4.00-20.50
Hypoplastic Left Heart Syndrome	Singleton	18	1.24	0.74-1.96
	Multiple	2	2.84	0.34-10.27
Interrupted Aortic Arch (Type A and NOS)	Singleton	1	0.07	0.00-0.38
	Multiple	0	0.00	0.00-5.24
<i>Patent Ductus Arteriosus</i>				
Patent Ductus Arteriosus	Singleton	216	14.91	12.99-17.03
	Multiple	5	7.11	2.31-16.58
<i>Right-Sided Obstruction</i>				
Pulmonary Stenosis, Valvular	Singleton	89	6.14	4.93-7.56
	Multiple	6	8.53	3.13-18.56
Pulmonary Valve Atresia w/intact septum	Singleton	10	0.69	0.33-1.27
	Multiple	0	0.00	0.00-5.24
Pulmonary Valve Atresia with VSD	Singleton	3	0.21	0.04-0.61
	Multiple	0	0.00	0.00-5.24
Tricuspid Valve Atresia	Singleton	13	0.90	0.48-1.53
	Multiple	0	0.00	0.00-5.24
<i>Septal Defects</i>				
ASD (Secundum and NOS)	Singleton	312	21.53	19.21-24.06
	Multiple	26	36.95	24.14-54.14
VSD (Membranous and NOS)	Singleton	172	11.87	10.16-13.78
	Multiple	19	27.00	16.26-42.16
VSD, Conoventricular/Malalignment	Singleton	24	1.66	1.06-2.46
	Multiple	5	7.11	2.31-16.58
<i>Single Ventricle and L-TGA</i>				
L-TGA	Singleton	5	0.35	0.11-0.81
	Multiple	0	0.00	0.00-5.24

Table 8 Prevalence of Birth Defects by Plurality¹ of Live Births and Stillbirths, Massachusetts: 2008-2009

Defect ¹	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
Single Ventricle	Singleton	4	0.28	0.08-0.71
	Multiple	0	0.00	0.00-5.24
<i>Other Cardiovascular</i>				
Other Cardiovascular ³	Singleton	292	20.15	17.91-22.60
	Multiple	19	27.00	16.26-42.16
<i>Respiratory</i>				
Choanal Atresia	Singleton	9	0.62	0.28-1.18
	Multiple	1	1.42	0.04-7.92
Lung Anomalies	Singleton	21	1.45	0.90-2.22
	Multiple	2	2.84	0.34-10.27
Other Respiratory ³	Singleton	21	1.45	0.90-2.22
	Multiple	1	1.42	0.04-7.92
<i>Orofacial</i>				
Cleft Lip w/ and w/o Cleft Palate	Singleton	117	8.07	6.68-9.68
	Multiple	6	8.53	3.13-18.56
Cleft Palate w/o Cleft Lip	Singleton	83	5.73	4.56-7.10
	Multiple	4	5.68	1.55-14.55
Pierre Robin Sequence	Singleton	34	2.35	1.63-3.28
	Multiple	1	1.42	0.04-7.92
Other Orofacial ³	Singleton	76	5.25	4.13-6.56
	Multiple	5	7.11	2.31-16.58
<i>Gastrointestinal</i>				
Biliary Atresia	Singleton	9	0.62	0.28-1.18
	Multiple	1	1.42	0.04-7.92
Esophageal Atresia/Tracheoesophageal Fistula	Singleton	34	2.35	1.63-3.28
	Multiple	3	4.26	0.88-12.46
Hirschsprung Disease	Singleton	35	2.42	1.68-3.36
	Multiple	0	0.00	0.00-5.24
Rectal and Large Intestinal Atresia/Stenosis	Singleton	42	2.90	2.09-3.92
	Multiple	0	0.00	0.00-5.24

Table 8 Prevalence of Birth Defects by Plurality¹ of Live Births and Stillbirths, Massachusetts: 2008-2009

Defect ¹	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
Small Intestinal Atresia	Singleton	42	2.90	2.09-3.92
	Multiple	3	4.26	0.88-12.46
Other Gastrointestinal ³	Singleton	94	6.49	5.24-7.94
	Multiple	8	11.37	4.91-22.40
<i>Genitourinary</i>				
Bladder Exstrophy	Singleton	3	0.21	0.04-0.61
	Multiple	0	0.00	0.00-5.24
Hypospadias, 2nd or 3rd Degree ⁴	Singleton	173	23.25	19.91-26.98
	Multiple	18	50.49	29.92-79.80
Obstructive Genitourinary Defect	Singleton	217	14.98	13.05-17.11
	Multiple	11	15.63	7.80-27.97
Renal Agenesis/Hypoplasia	Singleton	8	0.55	0.24-1.09
	Multiple	1	1.42	0.04-7.92
Other Genitourinary ³	Singleton	241	16.63	14.60-18.87
	Multiple	16	22.74	13.00-36.92
<i>Musculoskeletal</i>				
Club Foot	Singleton	193	13.32	11.51-15.34
	Multiple	13	18.47	9.84-31.59
Craniosynostosis	Singleton	63	4.35	3.34-5.56
	Multiple	3	4.26	0.88-12.46
Diaphragmatic Hernia	Singleton	32	2.21	1.51-3.12
	Multiple	5	7.11	2.31-16.58
Gastroschisis	Singleton	49	3.38	2.50-4.47
	Multiple	1	1.42	0.04-7.92
Omphalocele	Singleton	15	1.04	0.58-1.71
	Multiple	3	4.26	0.88-12.46
Polydactyly/Syndactyly	Singleton	236	16.29	14.28-18.50
	Multiple	9	12.79	5.85-24.28
Reduction Deformity, Lower Limbs	Singleton	18	1.24	0.74-1.96
	Multiple	1	1.42	0.04-7.92

Table 8 Prevalence of Birth Defects by Plurality¹ of Live Births and Stillbirths, Massachusetts: 2008-2009

Defect ¹	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
Reduction Deformity, Upper Limbs	Singleton	29	2.00	1.34-2.87
	Multiple	2	2.84	0.34-10.27
Skeletal Dysplasia	Singleton	19	1.31	0.79-2.05
	Multiple	0	0.00	0.00-5.24
Other Musculoskeletal ³	Singleton	158	10.90	9.27-12.74
	Multiple	21	29.84	18.47-45.62
<i>Chromosomal and other Syndromes</i>				
Klinefelter Syndrome	Singleton	6	0.41	0.15-0.90
	Multiple	1	1.42	0.04-7.92
Trisomy 13	Singleton	4	0.28	0.08-0.71
	Multiple	1	1.42	0.04-7.92
Trisomy 18	Singleton	24	1.66	1.06-2.46
	Multiple	2	2.84	0.34-10.27
Trisomy 21 (Down Syndrome)	Singleton	183	12.63	10.87-14.60
	Multiple	9	12.79	5.85-24.28
Turner Syndrome	Singleton	14	0.97	0.53-1.62
	Multiple	1	1.42	0.04-7.92
Other Chromosomal Syndromes/Other Syndromes ³	Singleton	196	13.53	11.70-15.56
	Multiple	11	15.63	7.80-27.97
<i>Other</i>				
Amniotic Bands	Singleton	10	0.69	0.33-1.27
	Multiple	3	4.26	0.88-12.46
Skin Anomalies	Singleton	24	1.66	1.06-2.46
	Multiple	3	4.26	0.88-12.46
Other, Specified ³	Singleton	16	1.10	0.63-1.79
	Multiple	1	1.42	0.04-7.92

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

² Cases can be included in more than one defect. Cases are counted once in the total for a defect category. Due to missing plurality of infant counts may not match those in other tables.

³ Rate may represent a heterogeneous group of defects.

⁴ Rate calculated using male live births.

Table 9 Most Common Defects by Plurality¹ of Live Births and Stillbirths, Massachusetts: 2008-2009

Defect ²	Count	Rate per 10,000 Births	95% Confidence Interval
MULTIPLE			
ASD (Secundum and NOS)	26	36.95	24.14-54.14
VSD (Membranous and NOS)	19	27.00	16.26-42.16
Hypospadias, 2nd or 3rd Degree ³	18	50.49	29.92-79.80
Club Foot	13	18.47	9.84-31.59
Obstructive Genitourinary Defect	11	15.63	7.80-27.97
Polydactyly/Syndactyly	9	12.79	5.85-24.28
Trisomy 21 (Down Syndrome)	9	12.79	5.85-24.28
Coarctation of Aorta	7	9.95	4.00-20.50
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	6	8.53	3.13-18.56
Pulmonary Stenosis, Valvular	6	8.53	3.13-18.56
SINGLETON			
ASD (Secundum and NOS)	312	21.53	19.21-24.06
Polydactyly/Syndactyly	236	16.29	14.28-18.50
Obstructive Genitourinary Defect	217	14.98	13.05-17.11
Club Foot	193	13.32	11.51-15.34
Trisomy 21 (Down Syndrome)	183	12.63	10.87-14.60
Hypospadias, 2nd or 3rd Degree ³	173	23.25	19.91-26.98
VSD (Membranous and NOS)	172	11.87	10.16-13.78
Cleft Lip w/ and w/o Cleft Palate	117	8.07	6.68-9.68
Pulmonary Stenosis, Valvular	89	6.14	4.93-7.56
Cleft Palate w/o Cleft Lip	83	5.73	4.56-7.10

- 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 case
- ³ Rate calculated using male live births.

Figure 7a Total Live Births, Massachusetts: 2008-2009

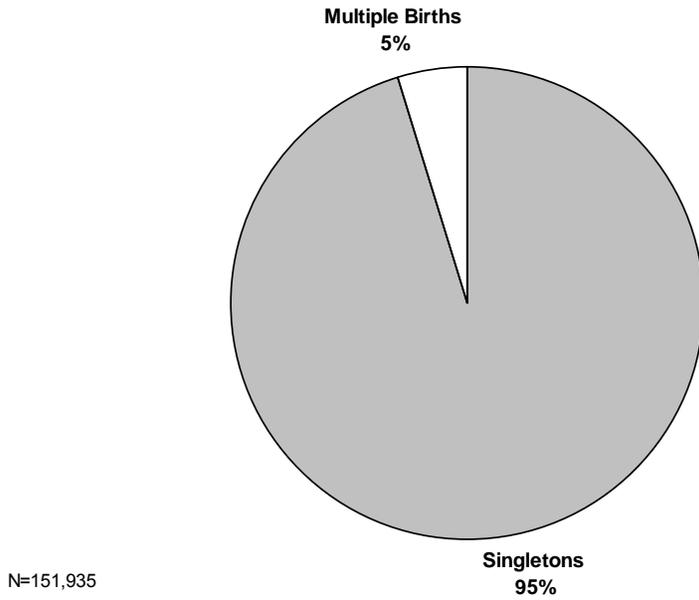


Figure 7b Birth Defect Cases, Massachusetts: 2008-2009

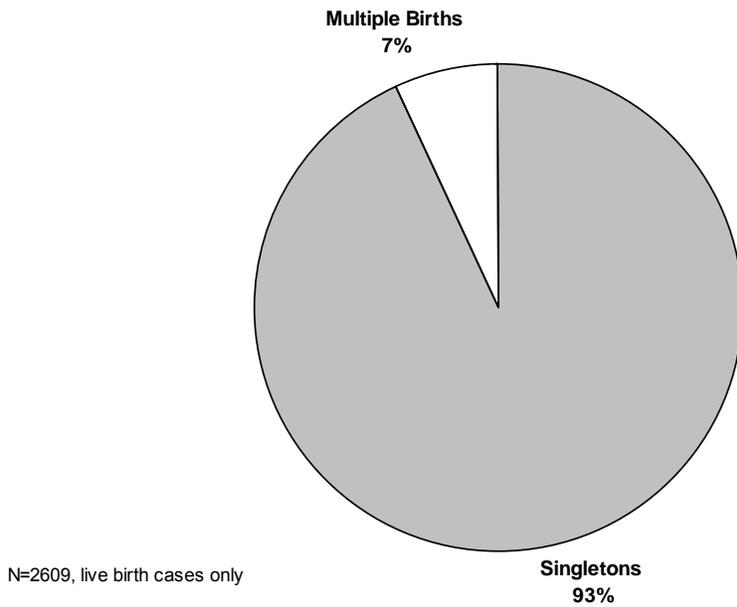
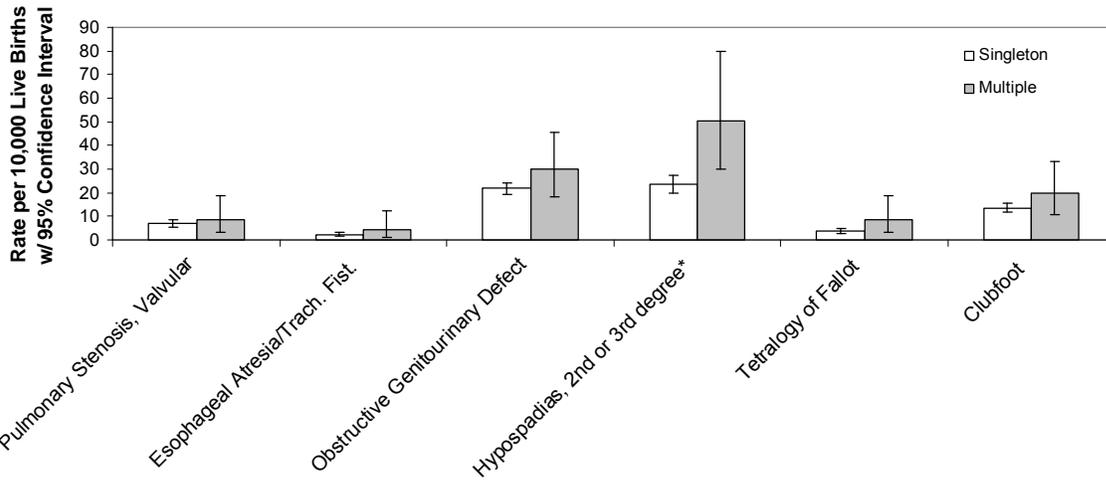


Figure 8 Prevalence of Selected Birth Defects by Plurality among Live Births and Stillbirths, Massachusetts: 2008-2009



N=2664

*prevalence estimate for hypospadias calculated using male births only for denominator

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2009

Defect ¹	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
<i>Central Nervous System</i>				
Anencephaly	<20	0	0.00	0.00-4.03
	20-24	4	1.63	0.44-4.18
	25-29	3	0.80	0.17-2.34
	30-34	1	0.22	0.01-1.20
	35+	0	0.00	0.00-1.07
Encephalocele	<20	0	0.00	0.00-4.03
	20-24	0	0.00	0.00-1.50
	25-29	2	0.53	0.06-1.93
	30-34	1	0.22	0.01-1.20
	35+	0	0.00	0.00-1.07
Holoprosencephaly	<20	0	0.00	0.00-4.03
	20-24	5	2.04	0.66-4.76
	25-29	2	0.53	0.06-1.93
	30-34	0	0.00	0.00-0.80
	35+	2	0.58	0.07-2.10
Hydrocephaly w/o Spina Bifida	<20	7	7.65	3.08-15.76
	20-24	13	5.30	2.82-9.07
	25-29	8	2.13	0.92-4.20
	30-34	9	1.94	0.89-3.69
	35+	11	3.19	1.59-5.71
Microcephaly	<20	2	2.19	0.26-7.89
	20-24	6	2.45	0.90-5.33
	25-29	6	1.60	0.59-3.48
	30-34	7	1.51	0.61-3.12
	35+	4	1.16	0.32-2.97
Spina Bifida w/ and w/o Hydrocephaly	<20	1	1.09	0.03-6.09
	20-24	5	2.04	0.66-4.76
	25-29	8	2.13	0.92-4.20
	30-34	8	1.73	0.75-3.40
	35+	7	2.03	0.82-4.18

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2009

Defect ¹	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
Spinal Cord	<20	2	2.19	0.26-7.89
	20-24	10	4.08	1.96-7.50
	25-29	11	2.93	1.46-5.25
	30-34	18	3.89	2.30-6.14
	35+	16	4.64	2.65-7.54
Other CNS	<20	16	17.48	9.99-28.39
	20-24	23	9.38	5.95-14.07
	25-29	20	5.34	3.26-8.24
	30-34	26	5.62	3.67-8.23
	35+	28	8.12	5.40-11.74
<i>Eye</i>				
Aniridia	<20	0	0.00	0.00-4.03
	20-24	0	0.00	0.00-1.50
	25-29	1	0.27	0.01-1.49
	30-34	0	0.00	0.00-0.80
	35+	0	0.00	0.00-1.07
Anophthalmia/Microphthalmia	<20	1	1.09	0.03-6.09
	20-24	3	1.22	0.25-3.58
	25-29	9	2.40	1.10-4.56
	30-34	4	0.86	0.24-2.21
	35+	5	1.45	0.47-3.38
Congenital Glaucoma, Congenital Cataract	<20	6	6.56	2.41-14.27
	20-24	11	4.49	2.24-8.03
	25-29	12	3.20	1.65-5.59
	30-34	9	1.94	0.89-3.69
	35+	8	2.32	1.00-4.57
Other Eye	<20	5	5.46	1.77-12.75
	20-24	12	4.89	2.53-8.55
	25-29	15	4.00	2.24-6.60
	30-34	7	1.51	0.61-3.12
	35+	8	2.32	1.00-4.57

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2009

Defect ¹	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
<i>Ear</i>				
Anotia/Microtia	<20	1	1.09	0.03-6.09
	20-24	4	1.63	0.44-4.18
	25-29	8	2.13	0.92-4.20
	30-34	7	1.51	0.61-3.12
	35+	7	2.03	0.82-4.18
Other Ear	<20	0	0.00	0.00-4.03
	20-24	10	4.08	1.96-7.50
	25-29	13	3.47	1.85-5.93
	30-34	11	2.38	1.19-4.25
	35+	9	2.61	1.19-4.96
<i>Cardiovascular</i>				
<i>Anomalous Pulmonary Venous Connection</i>				
Total/Partial Anomalous Pulmonary Venous Connection	<20	3	3.28	0.68-9.58
	20-24	1	0.41	0.01-2.27
	25-29	5	1.33	0.43-3.11
	30-34	3	0.65	0.13-1.89
	35+	8	2.32	1.00-4.57
<i>Atrioventricular Canal Defects</i>				
ASD Primum	<20	0	0.00	0.00-4.03
	20-24	0	0.00	0.00-1.50
	25-29	0	0.00	0.00-0.98
	30-34	1	0.22	0.01-1.20
	35+	1	0.29	0.01-1.62
Common Atrium	<20	0	0.00	0.00-4.03
	20-24	1	0.41	0.01-2.27
	25-29	4	1.07	0.29-2.73
	30-34	3	0.65	0.13-1.89
	35+	2	0.58	0.07-2.10

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2009

Defect ¹	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
Complete Atrioventricular Canal Defect	<20	1	1.09	0.03-6.09
	20-24	9	3.67	1.68-6.97
	25-29	9	2.40	1.10-4.56
	30-34	10	2.16	1.04-3.97
	35+	15	4.35	2.44-7.18
Endocardial Cushion (OS and NOS)	<20	0	0.00	0.00-4.03
	20-24	3	1.22	0.25-3.58
	25-29	3	0.80	0.17-2.34
	30-34	6	1.30	0.48-2.82
	35+	13	3.77	2.01-6.45
VSD, Canal Type	<20	0	0.00	0.00-4.03
	20-24	0	0.00	0.00-1.50
	25-29	0	0.00	0.00-0.98
	30-34	2	0.43	0.05-1.56
	35+	1	0.29	0.01-1.62
<i>Conotruncal (Outlet) and Aortic Arch</i>				
Double Outlet Right Ventricle	<20	0	0.00	0.00-4.03
	20-24	2	0.82	0.10-2.95
	25-29	4	1.07	0.29-2.73
	30-34	6	1.30	0.48-2.82
	35+	3	0.87	0.18-2.54
Interrupted Aortic Arch, Type B	<20	0	0.00	0.00-4.03
	20-24	1	0.41	0.01-2.27
	25-29	1	0.27	0.01-1.49
	30-34	2	0.43	0.05-1.56
	35+	1	0.29	0.01-1.62
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	<20	3	3.28	0.68-9.58
	20-24	16	6.52	3.73-10.60
	25-29	16	4.27	2.44-6.93
	30-34	11	2.38	1.19-4.25
	35+	14	4.06	2.22-6.81

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2009

Defect ¹	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
Truncus	<20	0	0.00	0.00-4.03
	20-24	1	0.41	0.01-2.27
	25-29	1	0.27	0.01-1.49
	30-34	1	0.22	0.01-1.20
	35+	1	0.29	0.01-1.62
d-Transposition of the Great Arteries	<20	4	4.37	1.19-11.19
	20-24	6	2.45	0.90-5.33
	25-29	9	2.40	1.10-4.56
	30-34	9	1.94	0.89-3.69
	35+	11	3.19	1.59-5.71
Ebstein Anomaly				
Ebstein Anomaly	<20	0	0.00	0.00-4.03
	20-24	0	0.00	0.00-1.50
	25-29	1	0.27	0.01-1.49
	30-34	2	0.43	0.05-1.56
	35+	1	0.29	0.01-1.62
Heterotaxy (Laterality Defects)				
Heterotaxy	<20	2	2.19	0.26-7.89
	20-24	6	2.45	0.90-5.33
	25-29	6	1.60	0.59-3.48
	30-34	7	1.51	0.61-3.12
	35+	3	0.87	0.18-2.54
Left-Sided Obstruction				
Aortic Valve Stenosis	<20	1	1.09	0.03-6.09
	20-24	2	0.82	0.10-2.95
	25-29	6	1.60	0.59-3.48
	30-34	4	0.86	0.24-2.21
	35+	8	2.32	1.00-4.57

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2009

Defect ¹	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
Coarctation of Aorta	<20	5	5.46	1.77-12.75
	20-24	9	3.67	1.68-6.97
	25-29	17	4.53	2.64-7.26
	30-34	17	3.67	2.14-5.88
	35+	17	4.93	2.87-7.90
Hypoplastic Left Heart Syndrome	<20	2	2.19	0.26-7.89
	20-24	3	1.22	0.25-3.58
	25-29	4	1.07	0.29-2.73
	30-34	2	0.43	0.05-1.56
	35+	7	2.03	0.82-4.18
Interrupted Aortic Arch (Type A and NOS)	<20	0	0.00	0.00-4.03
	20-24	0	0.00	0.00-1.50
	25-29	1	0.27	0.01-1.49
	30-34	0	0.00	0.00-0.80
	35+	0	0.00	0.00-1.07
<i>Patent Ductus Arteriosus</i>				
Patent Ductus Arteriosus	<20	5	5.46	1.77-12.75
	20-24	34	13.86	9.60-19.37
	25-29	54	14.40	10.82-18.79
	30-34	63	13.61	10.46-17.41
	35+	65	18.86	14.55-24.03
<i>Right-Sided Obstruction</i>				
Pulmonary Stenosis, Valvular	<20	5	5.46	1.77-12.75
	20-24	11	4.49	2.24-8.03
	25-29	21	5.60	3.47-8.56
	30-34	38	8.21	5.81-11.27
	35+	20	5.80	3.54-8.96

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2009

Defect ¹	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
Pulmonary Valve Atresia w/intact septum	<20	2	2.19	0.26-7.89
	20-24	1	0.41	0.01-2.27
	25-29	1	0.27	0.01-1.49
	30-34	3	0.65	0.13-1.89
	35+	3	0.87	0.18-2.54
Pulmonary Valve Atresia with VSD	<20	1	1.09	0.03-6.09
	20-24	1	0.41	0.01-2.27
	25-29	0	0.00	0.00-0.98
	30-34	0	0.00	0.00-0.80
	35+	1	0.29	0.01-1.62
Tricuspid Valve Atresia	<20	1	1.09	0.03-6.09
	20-24	2	0.82	0.10-2.95
	25-29	2	0.53	0.06-1.93
	30-34	5	1.08	0.35-2.52
	35+	3	0.87	0.18-2.54
Septal Defects				
ASD (Secundum and NOS)	<20	16	17.48	9.99-28.39
	20-24	43	17.53	12.69-23.62
	25-29	73	19.47	15.26-24.48
	30-34	101	21.82	17.77-26.51
	35+	105	30.46	24.91-36.87
VSD (Membranous and NOS)	<20	9	9.83	4.50-18.67
	20-24	27	11.01	7.26-16.02
	25-29	39	10.40	7.40-14.22
	30-34	49	10.58	7.83-13.99
	35+	60	17.41	13.28-22.40
VSD, Conoventricular/Malalignment	<20	3	3.28	0.68-9.58
	20-24	3	1.22	0.25-3.58
	25-29	6	1.60	0.59-3.48
	30-34	6	1.30	0.48-2.82
	35+	9	2.61	1.19-4.96

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2009

Defect ¹	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
Single Ventricle and L-TGA				
L-TGA	<20	0	0.00	0.00-4.03
	20-24	0	0.00	0.00-1.50
	25-29	3	0.80	0.17-2.34
	30-34	0	0.00	0.00-0.80
	35+	2	0.58	0.07-2.10
Single Ventricle	<20	1	1.09	0.03-6.09
	20-24	0	0.00	0.00-1.50
	25-29	1	0.27	0.01-1.49
	30-34	1	0.22	0.01-1.20
	35+	1	0.29	0.01-1.62
Other Cardiovascular				
Other Cardiovascular	<20	12	13.11	6.78-22.91
	20-24	44	17.94	13.04-24.09
	25-29	72	19.21	15.03-24.19
	30-34	89	19.22	15.44-23.66
	35+	88	25.53	20.47-31.45
Respiratory				
Choanal Atresia	<20	0	0.00	0.00-4.03
	20-24	2	0.82	0.10-2.95
	25-29	3	0.80	0.17-2.34
	30-34	2	0.43	0.05-1.56
	35+	3	0.87	0.18-2.54
Lung Anomalies	<20	0	0.00	0.00-4.03
	20-24	2	0.82	0.10-2.95
	25-29	8	2.13	0.92-4.20
	30-34	7	1.51	0.61-3.12
	35+	6	1.74	0.64-3.79

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2009

Defect ¹	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
Other Respiratory	<20	0	0.00	0.00-4.03
	20-24	5	2.04	0.66-4.76
	25-29	8	2.13	0.92-4.20
	30-34	4	0.86	0.24-2.21
	35+	3	0.87	0.18-2.54
<i>Orofacial</i>				
Cleft Lip w/ and w/o Cleft Palate	<20	11	12.02	6.00-21.51
	20-24	16	6.52	3.73-10.60
	25-29	34	9.07	6.28-12.67
	30-34	32	6.91	4.73-9.76
	35+	28	8.12	5.40-11.74
Cleft Palate w/o Cleft Lip	<20	3	3.28	0.68-9.58
	20-24	18	7.34	4.35-11.60
	25-29	25	6.67	4.32-9.84
	30-34	26	5.62	3.67-8.23
	35+	14	4.06	2.22-6.81
Pierre Robin Sequence	<20	2	2.19	0.26-7.89
	20-24	6	2.45	0.90-5.33
	25-29	14	3.73	2.04-6.27
	30-34	9	1.94	0.89-3.69
	35+	4	1.16	0.32-2.97
Other Orofacial	<20	2	2.19	0.26-7.89
	20-24	16	6.52	3.73-10.60
	25-29	19	5.07	3.05-7.91
	30-34	22	4.75	2.98-7.19
	35+	20	5.80	3.54-8.96

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2009

Defect ¹	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
<i>Gastrointestinal</i>				
Biliary Atresia	<20	3	3.28	0.68-9.58
	20-24	1	0.41	0.01-2.27
	25-29	1	0.27	0.01-1.49
	30-34	4	0.86	0.24-2.21
	35+	1	0.29	0.01-1.62
Esophageal Atresia/Tracheoesophageal Fistula	<20	0	0.00	0.00-4.03
	20-24	4	1.63	0.44-4.18
	25-29	11	2.93	1.46-5.25
	30-34	6	1.30	0.48-2.82
	35+	16	4.64	2.65-7.54
Hirschsprung Disease	<20	2	2.19	0.26-7.89
	20-24	5	2.04	0.66-4.76
	25-29	13	3.47	1.85-5.93
	30-34	10	2.16	1.04-3.97
	35+	5	1.45	0.47-3.38
Rectal and Large Intestinal Atresia/Stenosis	<20	2	2.19	0.26-7.89
	20-24	10	4.08	1.96-7.50
	25-29	11	2.93	1.46-5.25
	30-34	9	1.94	0.89-3.69
	35+	9	2.61	1.19-4.96
Small Intestinal Atresia	<20	4	4.37	1.19-11.19
	20-24	7	2.85	1.15-5.88
	25-29	10	2.67	1.28-4.91
	30-34	9	1.94	0.89-3.69
	35+	12	3.48	1.80-6.08
Other Gastrointestinal	<20	14	15.30	8.36-25.67
	20-24	15	6.12	3.42-10.09
	25-29	28	7.47	4.96-10.79
	30-34	18	3.89	2.30-6.14
	35+	23	6.67	4.23-10.01

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2009

Defect ¹	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
<i>Genitourinary</i>				
Bladder Exstrophy	<20	0	0.00	0.00-4.03
	20-24	0	0.00	0.00-1.50
	25-29	1	0.27	0.01-1.49
	30-34	2	0.43	0.05-1.56
	35+	0	0.00	0.00-1.07
Hypospadias, 2nd or 3rd Degree ²	<20	3	6.37	1.31-18.61
	20-24	24	19.18	12.29-28.54
	25-29	40	20.58	14.71-28.03
	30-34	77	32.37	25.55-40.46
	35+	47	26.81	19.70-35.65
Obstructive Genitourinary Defect	<20	19	20.76	12.50-32.42
	20-24	40	16.31	11.65-22.21
	25-29	51	13.60	10.13-17.89
	30-34	60	12.96	9.89-16.68
	35+	55	15.96	12.02-20.77
Renal Agenesis/Hypoplasia	<20	0	0.00	0.00-4.03
	20-24	1	0.41	0.01-2.27
	25-29	1	0.27	0.01-1.49
	30-34	4	0.86	0.24-2.21
	35+	1	0.29	0.01-1.62
Other Genitourinary	<20	8	8.74	3.77-17.23
	20-24	41	16.72	12.00-22.68
	25-29	62	16.54	12.68-21.20
	30-34	73	15.77	12.36-19.83
	35+	70	20.31	15.83-25.66
<i>Musculoskeletal</i>				
Club Foot	<20	15	16.39	9.17-27.04
	20-24	37	15.09	10.62-20.80
	25-29	53	14.14	10.59-18.49
	30-34	54	11.66	8.76-15.22
	35+	40	11.60	8.29-15.80

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2009

Defect ¹	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
Craniosynostosis	<20	0	0.00	0.00-4.03
	20-24	3	1.22	0.25-3.58
	25-29	17	4.53	2.64-7.26
	30-34	23	4.97	3.15-7.45
	35+	22	6.38	4.00-9.66
Diaphragmatic Hernia	<20	3	3.28	0.68-9.58
	20-24	8	3.26	1.41-6.43
	25-29	6	1.60	0.59-3.48
	30-34	10	2.16	1.04-3.97
	35+	9	2.61	1.19-4.96
Gastroschisis	<20	15	16.39	9.17-27.04
	20-24	19	7.75	4.66-12.10
	25-29	12	3.20	1.65-5.59
	30-34	2	0.43	0.05-1.56
	35+	0	0.00	0.00-1.07
Omphalocele	<20	2	2.19	0.26-7.89
	20-24	8	3.26	1.41-6.43
	25-29	1	0.27	0.01-1.49
	30-34	2	0.43	0.05-1.56
	35+	4	1.16	0.32-2.97
Polydactyly/Syndactyly	<20	22	24.04	15.07-36.40
	20-24	41	16.72	12.00-22.68
	25-29	62	16.54	12.68-21.20
	30-34	68	14.69	11.41-18.62
	35+	47	13.63	10.02-18.13
Reduction Deformity, Lower Limbs	<20	1	1.09	0.03-6.09
	20-24	5	2.04	0.66-4.76
	25-29	3	0.80	0.17-2.34
	30-34	6	1.30	0.48-2.82
	35+	2	0.58	0.07-2.10

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2009

Defect ¹	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
Reduction Deformity, Upper Limbs	<20	3	3.28	0.68-9.58
	20-24	8	3.26	1.41-6.43
	25-29	4	1.07	0.29-2.73
	30-34	10	2.16	1.04-3.97
	35+	4	1.16	0.32-2.97
Skeletal Dysplasia	<20	0	0.00	0.00-4.03
	20-24	6	2.45	0.90-5.33
	25-29	4	1.07	0.29-2.73
	30-34	5	1.08	0.35-2.52
	35+	3	0.87	0.18-2.54
Other Musculoskeletal	<20	11	12.02	6.00-21.51
	20-24	34	13.86	9.60-19.37
	25-29	45	12.00	8.76-16.06
	30-34	47	10.15	7.46-13.50
	35+	36	10.44	7.31-14.46
<i>Chromosomal and other Syndromes</i>				
Klinefelter Syndrome	<20	0	0.00	0.00-4.03
	20-24	0	0.00	0.00-1.50
	25-29	1	0.27	0.01-1.49
	30-34	1	0.22	0.01-1.20
	35+	5	1.45	0.47-3.38
Trisomy 13	<20	0	0.00	0.00-4.03
	20-24	2	0.82	0.10-2.95
	25-29	0	0.00	0.00-0.98
	30-34	1	0.22	0.01-1.20
	35+	1	0.29	0.01-1.62
Trisomy 18	<20	2	2.19	0.26-7.89
	20-24	3	1.22	0.25-3.58
	25-29	3	0.80	0.17-2.34
	30-34	2	0.43	0.05-1.56
	35+	9	2.61	1.19-4.96

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2009

Defect ¹	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
Trisomy 21 (Down Syndrome)	<20	6	6.56	2.41-14.27
	20-24	22	8.97	5.62-13.58
	25-29	24	6.40	4.10-9.53
	30-34	33	7.13	4.91-10.01
	35+	98	28.43	23.08-34.65
Turner Syndrome	<20	1	1.09	0.03-6.09
	20-24	4	1.63	0.44-4.18
	25-29	4	1.07	0.29-2.73
	30-34	0	0.00	0.00-0.80
	35+	3	0.87	0.18-2.54
Other Chromosomal Syndromes/Other Syndromes	<20	8	8.74	3.77-17.23
	20-24	24	9.79	6.27-14.56
	25-29	50	13.34	9.90-17.58
	30-34	58	12.53	9.51-16.20
	35+	64	18.57	14.30-23.71
<i>Other</i>				
Amniotic Bands	<20	2	2.19	0.26-7.89
	20-24	2	0.82	0.10-2.95
	25-29	1	0.27	0.01-1.49
	30-34	3	0.65	0.13-1.89
	35+	2	0.58	0.07-2.10
Skin Anomalies	<20	0	0.00	0.00-4.03
	20-24	6	2.45	0.90-5.33
	25-29	5	1.33	0.43-3.11
	30-34	9	1.94	0.89-3.69
	35+	7	2.03	0.82-4.18

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2009

Defect ¹	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
Other, Specified	<20	2	2.19	0.26-7.89
	20-24	2	0.82	0.10-2.95
	25-29	4	1.07	0.29-2.73
	30-34	5	1.08	0.35-2.52
	35+	4	1.16	0.32-2.97

¹ Cases can be included in 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. Due to missing age of mother counts may not match those in other tables.

² Rate calculated using male live births.

Figure 9 Rates of Down Syndrome by Maternal Age Group, Massachusetts: 2008-2009

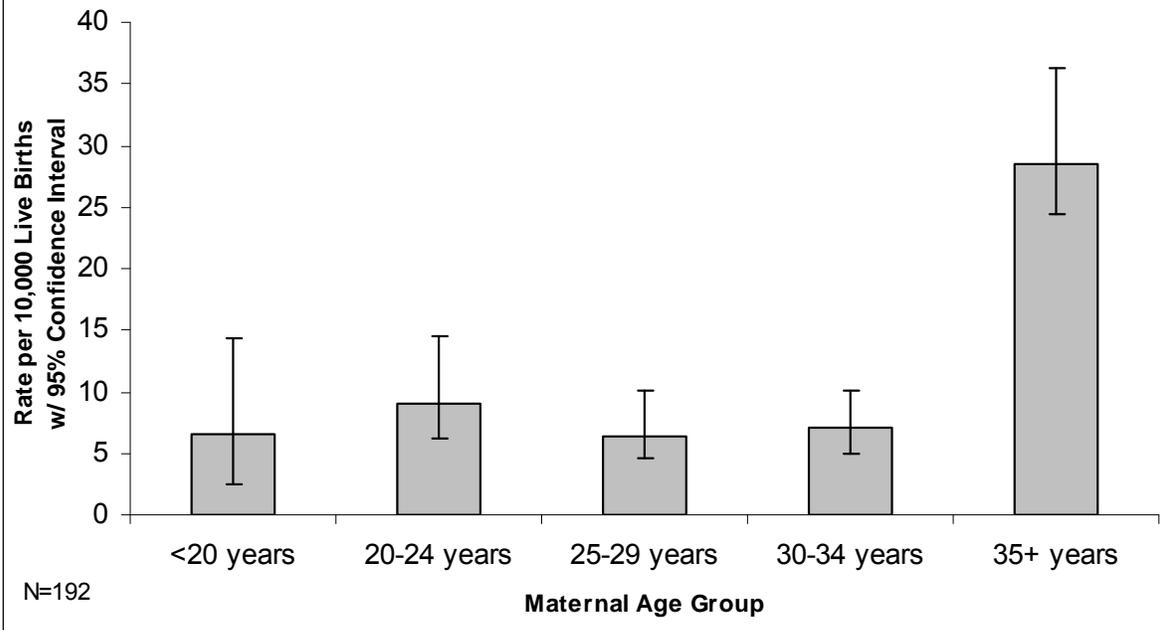


Figure 10 Chromosomal and All Other Defects by Maternal Age Group, Massachusetts: 2008-2009

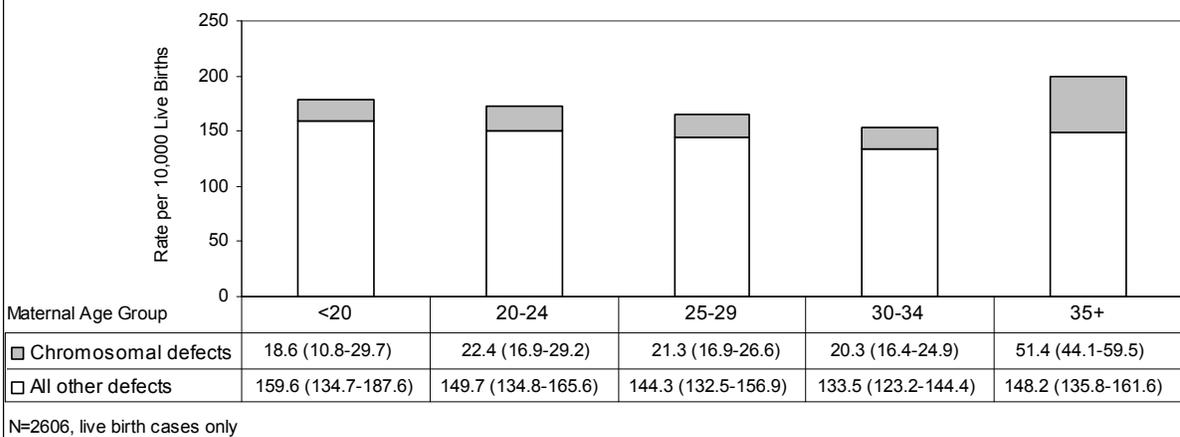


Figure 11 Rates of Gastroschisis by Maternal Age Group, Massachusetts: 2008-2009

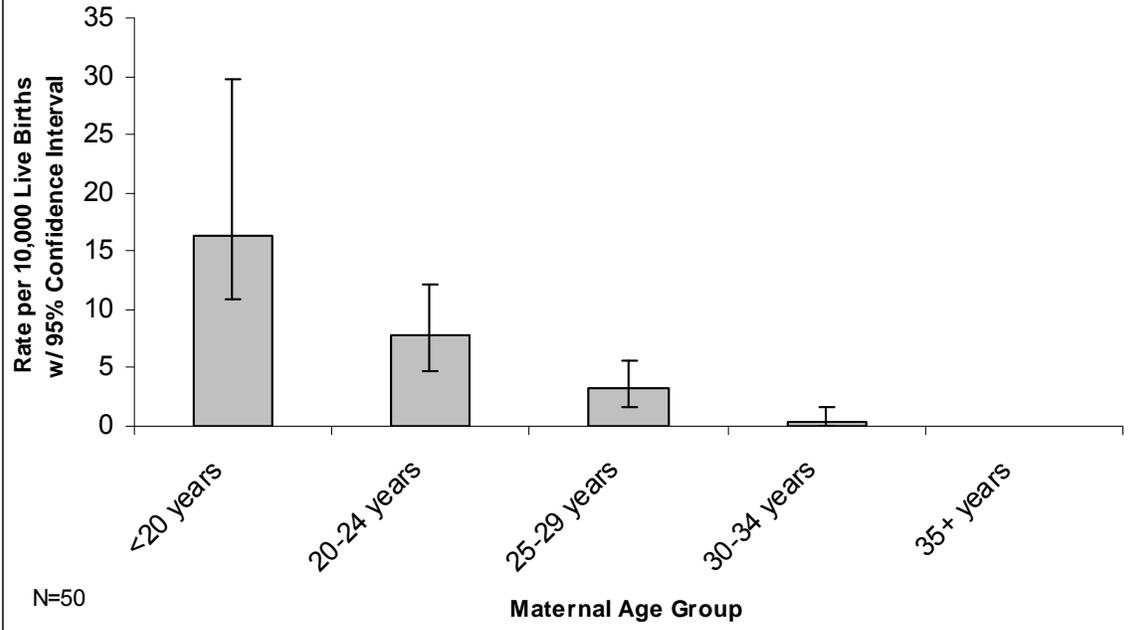


Figure 12 Births by ART Use and Plurality among Live Births, Massachusetts: 2002-2009

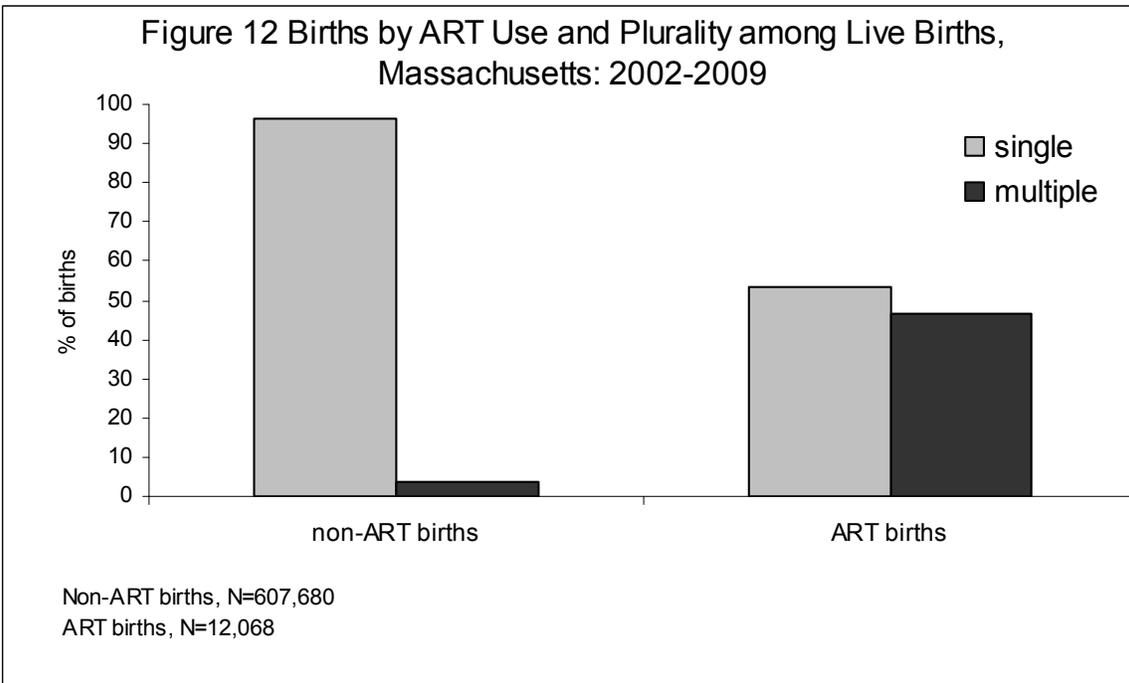


Table 11 Most Common Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2009

Age Group (yrs)	Defect ¹	Count	Rate per 10,000 Births	95% Confidence Interval
<20	Polydactyly/Syndactyly	22	24.04	15.07-36.40
	Obstructive Genitourinary Defect	19	20.76	12.50-32.42
	ASD (Secundum and NOS)	16	17.48	9.99-28.39
	Club Foot	15	16.39	9.17-27.04
	Gastroschisis	15	16.39	9.17-27.04
20-24	ASD (Secundum and NOS)	43	17.53	12.69-23.62
	Polydactyly/Syndactyly	41	16.72	12.00-22.68
	Obstructive Genitourinary Defect	40	16.31	11.65-22.21
	Club Foot	37	15.09	10.62-20.80
	VSD (Membranous and NOS)	27	11.01	7.26-16.02
25-29	ASD (Secundum and NOS)	73	19.47	15.26-24.48
	Polydactyly/Syndactyly	62	16.54	12.68-21.20
	Club Foot	53	14.14	10.59-18.49
	Obstructive Genitourinary Defect	51	13.60	10.13-17.89
	Hypospadias, 2nd or 3rd Degree ²	40	20.58	14.71-28.03
30-34	ASD (Secundum and NOS)	101	21.82	17.77-26.51
	Hypospadias, 2nd or 3rd Degree ²	77	25.55	25.55-40.46
	Polydactyly/Syndactyly	68	14.69	11.41-18.62
	Obstructive Genitourinary Defect	60	12.96	9.89-16.68
	Club Foot	54	11.66	8.76-15.22
35+	ASD (Secundum and NOS)	105	30.46	24.91-36.87
	Trisomy 21 (Down Syndrome)	98	28.43	23.08-34.65
	VSD (Membranous and NOS)	60	17.41	13.28-22.40
	Obstructive Genitourinary Defect	55	15.96	12.02-20.77
	Hypospadias, 2nd or 3rd Degree ²	47	26.81	19.70-35.65

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

² Rate calculated using male live births.

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 age-adjusted rate per 10,000 live births was 168.8 for non-Hispanic whites, 200.6 for non-Hispanic blacks, 139.1 for non-Hispanic Asians/Pacific Islanders and 182.8 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 most common defects by maternal race and Hispanic ethnicity. There were very few differences among the most commonly occurring defects in the different race/ethnicities. All groups had atrial septal defects (secundum and NOS) and polydactyly/syndactyly amongst the most common defects, and hypospadias (2nd and 3rd degree) and obstructive genitourinary defect was in all but one group’s most commonly occurring defects. Though non-Hispanic Asians had generally lower age-adjusted rates of chromosomal defects, all races/ethnicities were comparable and did not have statistically significant differences in the overall defect rates. This finding is different from the 2004-2005 report, where the non-Hispanic white group had about half the rate compared to non-Hispanic black and Hispanic. Maternal birthplace (U.S. versus non-U.S.) may be a contributing factor in group differences as women born in the U.S. had slightly higher rates than those born outside the U.S. 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 about the same as the rates of women born in the U.S (200.1 and 196.9 per 10,000 live births, respectively).

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 12 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.

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.

Trends in Maternal Race and Ethnicity

Figure 15 shows the age-adjusted birth defects rates between 2004 and 2009 in two-year intervals. The non-Hispanic black group saw a decrease in the overall prevalence of birth defects during the 2006-2007 period and a return to 2004-2005 levels in the most recent two-year-period. The other groups saw little change between 2004 and 2009.

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 age-adjusted birth defect rates by the six regions in 2008-2009 are shown in Figure 15. Although not statistically significantly different, the rates range from 163.3 per 10,000 in the Metro West region to 181.4 per 10,000 in the Boston region.

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008-2009

Defect	Maternal Race ¹	Count ²	Rate per 10,000 Births	95% Confidence Interval
<i>Total Cases</i>	White, Non-Hispanic	1723	168.8	160.80-186.8
	Black, Non-Hispanic	272	200.6	176.8-224.5
	Asian, Non-Hispanic	156	139.1	117.3-160.9
	Hispanic	395	182.8	164.8-200.8
<i>Central Nervous System</i>				
Anencephaly	White, Non-Hispanic	4	0.39	0.11-1.01
	Black, Non-Hispanic	1	0.74	0.02-4.10
	Asian, Non-Hispanic	1	0.84	0.02-4.68
	Hispanic	1	0.46	0.01-2.55
Encephalocele	White, Non-Hispanic	3	0.30	0.06-0.86
	Black, Non-Hispanic	0	0.00	0.00-2.71
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	0	0.00	0.00-1.69
Holoprosencephaly	White, Non-Hispanic	4	0.39	0.11-1.01
	Black, Non-Hispanic	1	0.74	0.02-4.10
	Asian, Non-Hispanic	1	0.84	0.02-4.68
	Hispanic	3	1.37	0.28-4.01
Hydrocephaly w/o Spina Bifida	White, Non-Hispanic	24	2.36	1.51-3.52
	Black, Non-Hispanic	9	6.62	3.03-12.57
	Asian, Non-Hispanic	1	0.84	0.02-4.68
	Hispanic	12	5.48	2.83-9.58
Microcephaly	White, Non-Hispanic	17	1.67	0.98-2.68
	Black, Non-Hispanic	4	2.94	0.80-7.53
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	3	1.37	0.28-4.01
Spina Bifida w/ and w/o Hydrocephaly	White, Non-Hispanic	18	1.77	1.05-2.80
	Black, Non-Hispanic	2	1.47	0.18-5.31
	Asian, Non-Hispanic	3	2.52	0.52-7.37
	Hispanic	4	1.83	0.50-4.68

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008-2009

Defect	Maternal Race ¹	Count ²	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
Spinal Cord	White, Non-Hispanic	37	3.64	2.57-5.02
	Black, Non-Hispanic	6	4.41	1.62-9.60
	Asian, Non-Hispanic	3	2.52	0.52-7.37
	Hispanic	9	4.11	1.88-7.81
Other CNS	White, Non-Hispanic	69	6.80	5.29-8.60
	Black, Non-Hispanic	20	14.71	8.98-22.72
	Asian, Non-Hispanic	5	4.20	1.36-9.81
	Hispanic	16	7.31	4.18-11.87
Eye				
Aniridia	White, Non-Hispanic	0	0.00	0.00-0.36
	Black, Non-Hispanic	0	0.00	0.00-2.71
	Asian, Non-Hispanic	1	0.84	0.02-4.68
	Hispanic	0	0.00	0.00-1.69
Anophthalmia/Microphthalmia	White, Non-Hispanic	12	1.18	0.61-2.06
	Black, Non-Hispanic	3	2.21	0.46-6.45
	Asian, Non-Hispanic	2	1.68	0.20-6.07
	Hispanic	5	2.29	0.74-5.33
Congenital Glaucoma, Congenital Cataract	White, Non-Hispanic	22	2.17	1.36-3.28
	Black, Non-Hispanic	8	5.88	2.54-11.59
	Asian, Non-Hispanic	3	2.52	0.52-7.37
	Hispanic	11	5.03	2.51-9.00
Other Eye	White, Non-Hispanic	31	3.05	2.07-4.33
	Black, Non-Hispanic	4	2.94	0.80-7.53
	Asian, Non-Hispanic	5	4.20	1.36-9.81
	Hispanic	7	3.20	1.29-6.59
Ear				
Anotia/Microtia	White, Non-Hispanic	15	1.48	0.83-2.44
	Black, Non-Hispanic	2	1.47	0.18-5.31
	Asian, Non-Hispanic	5	4.20	1.36-9.81
	Hispanic	5	2.29	0.74-5.33

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008-2009

Defect	Maternal Race ¹	Count ²	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
Other Ear	White, Non-Hispanic	30	2.96	1.99-4.22
	Black, Non-Hispanic	2	1.47	0.18-5.31
	Asian, Non-Hispanic	3	2.52	0.52-7.37
	Hispanic	6	2.74	1.01-5.97
<i>Cardiovascular</i>				
<i>Anomalous Pulmonary Venous Connection</i>				
Total/Partial Anomalous Pulmonary Venous Connection	White, Non-Hispanic	12	1.18	0.61-2.06
	Black, Non-Hispanic	2	1.47	0.18-5.31
	Asian, Non-Hispanic	3	2.52	0.52-7.37
	Hispanic	3	1.37	0.28-4.01
<i>Atrioventricular Canal Defects</i>				
ASD Primum	White, Non-Hispanic	2	0.20	0.02-0.71
	Black, Non-Hispanic	0	0.00	0.00-2.71
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	0	0.00	0.00-1.69
Common Atrium	White, Non-Hispanic	4	0.39	0.11-1.01
	Black, Non-Hispanic	2	1.47	0.18-5.31
	Asian, Non-Hispanic	1	0.84	0.02-4.68
	Hispanic	0	0.00	0.00-1.69
Complete Atrioventricular Canal Defect	White, Non-Hispanic	28	2.76	1.83-3.99
	Black, Non-Hispanic	7	5.15	2.07-10.61
	Asian, Non-Hispanic	3	2.52	0.52-7.37
	Hispanic	5	2.29	0.74-5.33
Endocardial Cushion (OS and NOS)	White, Non-Hispanic	18	1.77	1.05-2.80
	Black, Non-Hispanic	3	2.21	0.46-6.45
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	3	1.37	0.28-4.01
VSD, Canal Type	White, Non-Hispanic	2	0.20	0.02-0.71
	Black, Non-Hispanic	1	0.74	0.02-4.10
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	0	0.00	0.00-1.69

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008-2009

Defect	Maternal Race ¹	Count ²	Rate per 10,000 Births	95% Confidence Interval
<i>Conotruncal (Outlet) and Aortic Arch</i>				
Double Outlet Right Ventricle	White, Non-Hispanic	10	0.99	0.47-1.81
	Black, Non-Hispanic	1	0.74	0.02-4.10
	Asian, Non-Hispanic	1	0.84	0.02-4.68
	Hispanic	2	0.91	0.11-3.30
Interrupted Aortic Arch, Type B	White, Non-Hispanic	3	0.30	0.06-0.86
	Black, Non-Hispanic	2	1.47	0.18-5.31
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	0	0.00	0.00-1.69
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	White, Non-Hispanic	30	2.96	1.99-4.22
	Black, Non-Hispanic	8	5.88	2.54-11.59
	Asian, Non-Hispanic	4	3.36	0.92-8.61
	Hispanic	15	6.86	3.84-11.31
Truncus	White, Non-Hispanic	3	0.30	0.06-0.86
	Black, Non-Hispanic	0	0.00	0.00-2.71
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	0	0.00	0.00-1.69
d-Transposition of the Great Arteries	White, Non-Hispanic	32	3.15	2.16-4.45
	Black, Non-Hispanic	3	2.21	0.46-6.45
	Asian, Non-Hispanic	1	0.84	0.02-4.68
	Hispanic	3	1.37	0.28-4.01
<i>Ebstein Anomaly</i>				
Ebstein Anomaly	White, Non-Hispanic	2	0.20	0.02-0.71
	Black, Non-Hispanic	1	0.74	0.02-4.10
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	1	0.46	0.01-2.55
<i>Heterotaxy (Laterality Defects)</i>				
Heterotaxy	White, Non-Hispanic	14	1.38	0.75-2.31
	Black, Non-Hispanic	4	2.94	0.80-7.53
	Asian, Non-Hispanic	1	0.84	0.02-4.68
	Hispanic	2	0.91	0.11-3.30

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008-2009

Defect	Maternal Race ¹	Count ²	Rate per 10,000 Births	95% Confidence Interval
<i>Left-Sided Obstruction</i>				
Aortic Valve Stenosis	White, Non-Hispanic	16	1.58	0.90-2.56
	Black, Non-Hispanic	3	2.21	0.46-6.45
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	1	0.46	0.01-2.55
Coarctation of Aorta	White, Non-Hispanic	47	4.63	3.40-6.16
	Black, Non-Hispanic	3	2.21	0.46-6.45
	Asian, Non-Hispanic	1	0.84	0.02-4.68
	Hispanic	12	5.48	2.83-9.58
Hypoplastic Left Heart Syndrome	White, Non-Hispanic	13	1.28	0.68-2.19
	Black, Non-Hispanic	3	2.21	0.46-6.45
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	2	0.91	0.11-3.30
Interrupted Aortic Arch (Type A and NOS)	White, Non-Hispanic	0	0.00	0.00-0.36
	Black, Non-Hispanic	0	0.00	0.00-2.71
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	1	0.46	0.01-2.55
<i>Patent Ductus Arteriosus</i>				
Patent Ductus Arteriosus	White, Non-Hispanic	135	13.30	11.15-15.74
	Black, Non-Hispanic	28	20.59	13.68-29.76
	Asian, Non-Hispanic	13	10.93	5.82-18.69
	Hispanic	39	17.82	12.67-24.37
<i>Right-Sided Obstruction</i>				
Pulmonary Stenosis, Valvular	White, Non-Hispanic	59	5.81	4.42-7.50
	Black, Non-Hispanic	15	11.03	6.17-18.20
	Asian, Non-Hispanic	4	3.36	0.92-8.61
	Hispanic	17	7.77	4.53-12.44
Pulmonary Valve Atresia w/intact septum	White, Non-Hispanic	6	0.59	0.22-1.29
	Black, Non-Hispanic	1	0.74	0.02-4.10
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	2	0.91	0.11-3.30

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008-2009

Defect	Maternal Race ¹	Count ²	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
Pulmonary Valve Atresia with VSD	White, Non-Hispanic	0	0.00	0.00-0.36
	Black, Non-Hispanic	2	1.47	0.18-5.31
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	1	0.46	0.01-2.55
Tricuspid Valve Atresia	White, Non-Hispanic	9	0.89	0.41-1.68
	Black, Non-Hispanic	1	0.74	0.02-4.10
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	2	0.91	0.11-3.30
Septal Defects				
ASD (Secundum and NOS)	White, Non-Hispanic	214	21.08	18.35-24.10
	Black, Non-Hispanic	46	33.83	24.77-45.13
	Asian, Non-Hispanic	19	15.97	9.62-24.94
	Hispanic	52	23.76	17.75-31.16
VSD (Membranous and NOS)	White, Non-Hispanic	112	11.03	9.08-13.27
	Black, Non-Hispanic	20	14.71	8.98-22.72
	Asian, Non-Hispanic	16	13.45	7.69-21.84
	Hispanic	31	14.17	9.63-20.11
VSD, Conoventricular/Malalignment	White, Non-Hispanic	19	1.87	1.13-2.92
	Black, Non-Hispanic	4	2.94	0.80-7.53
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	4	1.83	0.50-4.68
Single Ventricle and L-TGA				
L-TGA	White, Non-Hispanic	4	0.39	0.11-1.01
	Black, Non-Hispanic	1	0.74	0.02-4.10
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	0	0.00	0.00-1.69
Single Ventricle	White, Non-Hispanic	4	0.39	0.11-1.01
	Black, Non-Hispanic	0	0.00	0.00-2.71
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	0	0.00	0.00-1.69

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008-2009

Defect	Maternal Race ¹	Count ²	Rate per 10,000 Births	95% Confidence Interval
<i>Other Cardiovascular</i>				
Other Cardiovascular	White, Non-Hispanic	185	18.22	15.69-21.05
	Black, Non-Hispanic	46	33.83	24.77-45.13
	Asian, Non-Hispanic	15	12.61	7.06-20.80
	Hispanic	49	22.39	16.57-29.61
<i>Respiratory</i>				
Choanal Atresia	White, Non-Hispanic	7	0.69	0.28-1.42
	Black, Non-Hispanic	1	0.74	0.02-4.10
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	1	0.46	0.01-2.55
Lung Anomalies	White, Non-Hispanic	13	1.28	0.68-2.19
	Black, Non-Hispanic	2	1.47	0.18-5.31
	Asian, Non-Hispanic	1	0.84	0.02-4.68
	Hispanic	6	2.74	1.01-5.97
Other Respiratory	White, Non-Hispanic	17	1.67	0.98-2.68
	Black, Non-Hispanic	0	0.00	0.00-2.71
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	3	1.37	0.28-4.01
<i>Orofacial</i>				
Cleft Lip w/ and w/o Cleft Palate	White, Non-Hispanic	83	8.18	6.51-10.14
	Black, Non-Hispanic	8	5.88	2.54-11.59
	Asian, Non-Hispanic	9	7.56	3.46-14.36
	Hispanic	20	9.14	5.58-14.12
Cleft Palate w/o Cleft Lip	White, Non-Hispanic	67	6.60	5.11-8.38
	Black, Non-Hispanic	3	2.21	0.46-6.45
	Asian, Non-Hispanic	2	1.68	0.20-6.07
	Hispanic	11	5.03	2.51-9.00
Pierre Robin Sequence	White, Non-Hispanic	29	2.86	1.91-4.10
	Black, Non-Hispanic	1	0.74	0.02-4.10
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	3	1.37	0.28-4.01

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008-2009

Defect	Maternal Race ¹	Count ²	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
Other Orofacial	White, Non-Hispanic	54	5.32	4.00-6.94
	Black, Non-Hispanic	10	7.35	3.53-13.53
	Asian, Non-Hispanic	3	2.52	0.52-7.37
	Hispanic	11	5.03	2.51-9.00
<i>Gastrointestinal</i>				
Biliary Atresia	White, Non-Hispanic	5	0.49	0.16-1.15
	Black, Non-Hispanic	1	0.74	0.02-4.10
	Asian, Non-Hispanic	3	2.52	0.52-7.37
	Hispanic	1	0.46	0.01-2.55
Esophageal Atresia/Tracheoesophageal Fistula	White, Non-Hispanic	30	2.96	1.99-4.22
	Black, Non-Hispanic	2	1.47	0.18-5.31
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	5	2.29	0.74-5.33
Hirschsprung Disease	White, Non-Hispanic	24	2.36	1.51-3.52
	Black, Non-Hispanic	0	0.00	0.00-2.71
	Asian, Non-Hispanic	4	3.36	0.92-8.61
	Hispanic	7	3.20	1.29-6.59
Rectal and Large Intestinal Atresia/Stenosis	White, Non-Hispanic	26	2.56	1.67-3.75
	Black, Non-Hispanic	5	3.68	1.19-8.58
	Asian, Non-Hispanic	4	3.36	0.92-8.61
	Hispanic	6	2.74	1.01-5.97
Small Intestinal Atresia	White, Non-Hispanic	28	2.76	1.83-3.99
	Black, Non-Hispanic	3	2.21	0.46-6.45
	Asian, Non-Hispanic	5	4.20	1.36-9.81
	Hispanic	6	2.74	1.01-5.97
Other Gastrointestinal	White, Non-Hispanic	59	5.81	4.42-7.50
	Black, Non-Hispanic	13	9.56	5.09-16.35
	Asian, Non-Hispanic	6	5.04	1.85-10.98
	Hispanic	18	8.23	4.88-13.00

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008-2009

Defect	Maternal Race ¹	Count ²	Rate per 10,000 Births	95% Confidence Interval
<i>Genitourinary</i>				
Bladder Exstrophy	White, Non-Hispanic	2	0.20	0.02-0.71
	Black, Non-Hispanic	0	0.00	0.00-2.71
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	1	0.46	0.01-2.55
Hypospadias, 2nd or 3rd Degree ⁴	White, Non-Hispanic	143	27.58	23.24-32.49
	Black, Non-Hispanic	20	28.69	17.52-44.30
	Asian, Non-Hispanic	10	16.17	7.76-29.74
	Hispanic	16	14.04	8.03-22.81
Obstructive Genitourinary Defect	White, Non-Hispanic	142	13.99	11.78-16.49
	Black, Non-Hispanic	19	13.97	8.41-21.82
	Asian, Non-Hispanic	19	15.97	9.62-24.94
	Hispanic	45	20.57	15.00-27.52
Renal Agenesis/Hypoplasia	White, Non-Hispanic	2	0.20	0.02-0.71
	Black, Non-Hispanic	3	2.21	0.46-6.45
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	1	0.46	0.01-2.55
Other Genitourinary	White, Non-Hispanic	172	16.94	14.51-19.67
	Black, Non-Hispanic	18	13.24	7.85-20.92
	Asian, Non-Hispanic	23	19.33	12.26-29.01
	Hispanic	37	16.91	11.91-23.31
<i>Musculoskeletal</i>				
Club Foot	White, Non-Hispanic	138	13.59	11.42-16.06
	Black, Non-Hispanic	19	13.97	8.41-21.82
	Asian, Non-Hispanic	8	6.72	2.90-13.25
	Hispanic	29	13.25	8.88-19.03
Craniosynostosis	White, Non-Hispanic	57	5.61	4.25-7.27
	Black, Non-Hispanic	1	0.74	0.02-4.10
	Asian, Non-Hispanic	1	0.84	0.02-4.68
	Hispanic	6	2.74	1.01-5.97

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008-2009

Defect	Maternal Race ¹	Count ²	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
Diaphragmatic Hernia	White, Non-Hispanic	20	1.97	1.20-3.04
	Black, Non-Hispanic	4	2.94	0.80-7.53
	Asian, Non-Hispanic	3	2.52	0.52-7.37
	Hispanic	6	2.74	1.01-5.97
Gastroschisis	White, Non-Hispanic	26	2.56	1.67-3.75
	Black, Non-Hispanic	8	5.88	2.54-11.59
	Asian, Non-Hispanic	2	1.68	0.20-6.07
	Hispanic	11	5.03	2.51-9.00
Omphalocele	White, Non-Hispanic	9	0.89	0.41-1.68
	Black, Non-Hispanic	4	2.94	0.80-7.53
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	4	1.83	0.50-4.68
Polydactyly/Syndactyly	White, Non-Hispanic	152	14.97	12.69-17.55
	Black, Non-Hispanic	32	23.53	16.10-33.22
	Asian, Non-Hispanic	14	11.77	6.43-19.74
	Hispanic	35	16.00	11.14-22.25
Reduction Deformity, Lower Limbs	White, Non-Hispanic	9	0.89	0.41-1.68
	Black, Non-Hispanic	2	1.47	0.18-5.31
	Asian, Non-Hispanic	1	0.84	0.02-4.68
	Hispanic	5	2.29	0.74-5.33
Reduction Deformity, Upper Limbs	White, Non-Hispanic	16	1.58	0.90-2.56
	Black, Non-Hispanic	3	2.21	0.46-6.45
	Asian, Non-Hispanic	2	1.68	0.20-6.07
	Hispanic	8	3.66	1.58-7.20
Skeletal Dysplasia	White, Non-Hispanic	10	0.99	0.47-1.81
	Black, Non-Hispanic	3	2.21	0.46-6.45
	Asian, Non-Hispanic	1	0.84	0.02-4.68
	Hispanic	3	1.37	0.28-4.01

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008-2009

Defect	Maternal Race ¹	Count ²	Rate per 10,000 Births	95% Confidence Interval
<i>(cont'd)</i>				
Other Musculoskeletal	White, Non-Hispanic	102	10.05	8.19-12.20
	Black, Non-Hispanic	28	20.59	13.68-29.76
	Asian, Non-Hispanic	9	7.56	3.46-14.36
	Hispanic	30	13.71	9.25-19.57
<i>Chromosomal and other Syndromes</i>				
Klinefelter Syndrome	White, Non-Hispanic	6	0.59	0.22-1.29
	Black, Non-Hispanic	0	0.00	0.00-2.71
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	1	0.46	0.01-2.55
Trisomy 13	White, Non-Hispanic	3	0.30	0.06-0.86
	Black, Non-Hispanic	1	0.74	0.02-4.10
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	0	0.00	0.00-1.69
Trisomy 18	White, Non-Hispanic	9	0.89	0.41-1.68
	Black, Non-Hispanic	3	2.21	0.46-6.45
	Asian, Non-Hispanic	1	0.84	0.02-4.68
	Hispanic	4	1.83	0.50-4.68
Trisomy 21 (Down Syndrome)	White, Non-Hispanic	110	10.84	8.91-13.06
	Black, Non-Hispanic	23	16.92	10.72-25.38
	Asian, Non-Hispanic	10	8.41	4.03-15.46
	Hispanic	37	16.91	11.91-23.31
Turner Syndrome	White, Non-Hispanic	8	0.79	0.34-1.55
	Black, Non-Hispanic	1	0.74	0.02-4.10
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	2	0.91	0.11-3.30
Other Chromosomal Syndromes/Other Syndromes	White, Non-Hispanic	145	14.28	12.05-16.81
	Black, Non-Hispanic	22	16.18	10.14-24.50
	Asian, Non-Hispanic	5	4.20	1.36-9.81
	Hispanic	31	14.17	9.63-20.11

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008-2009

Defect	Maternal Race ¹	Count ²	Rate per 10,000 Births	95% Confidence Interval
<i>Other</i>				
Amniotic Bands	White, Non-Hispanic	5	0.49	0.16-1.15
	Black, Non-Hispanic	3	2.21	0.46-6.45
	Asian, Non-Hispanic	0	0.00	0.00-3.10
	Hispanic	2	0.91	0.11-3.30
Skin Anomalies	White, Non-Hispanic	14	1.38	0.75-2.31
	Black, Non-Hispanic	5	3.68	1.19-8.58
	Asian, Non-Hispanic	3	2.52	0.52-7.37
	Hispanic	4	1.83	0.50-4.68
Other, Specified	White, Non-Hispanic	10	0.99	0.47-1.81
	Black, Non-Hispanic	3	2.21	0.46-6.45
	Asian, Non-Hispanic	1	0.84	0.02-4.68
	Hispanic	1	0.46	0.01-2.55

¹ Due to small numbers, races classified as "other" are not included.

² Cases can be included in 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. Due to missing race of mother counts may not match those in other tables.

³ Overall rates are standardized to the age distribution of Massachusetts

⁴ Rate calculated using male live births.

**Table 13 Most Common Defects by Maternal Race/Hispanic
Ethnicity for Live Births, Massachusetts: 2008-2009**

Race¹	Defect²	Count	Rate per 10,000 Births	95% Confidence Interval
White, Non-Hispanic	ASD (Secundum and NOS)	214	21.08	18.35-24.10
	Polydactyly/Syndactyly	152	14.97	12.69-17.55
	Hypospadias, 2nd or 3rd Degree ³	143	27.58	23.24-32.49
	Obstructive Genitourinary Defect	142	13.99	11.78-16.49
	Club Foot	138	13.59	11.42-16.06
Black, Non-Hispanic	ASD (Secundum and NOS)	46	33.83	24.77-45.13
	Polydactyly/Syndactyly	32	23.53	16.10-33.22
	Trisomy 21 (Down Syndrome)	23	16.92	10.72-25.38
	VSD (Membranous and NOS)	20	14.71	8.98-22.72
	Hypospadias, 2nd or 3rd Degree ³	20	17.52	17.52-44.30
Asian, Non-Hispanic	ASD (Secundum and NOS)	19	15.97	9.62-24.94
	Obstructive Genitourinary Defect	19	15.97	9.62-24.94
	VSD (Membranous and NOS)	16	13.45	7.69-21.84
	Polydactyly/Syndactyly	14	11.77	6.43-19.74
	Hypospadias, 2nd or 3rd Degree ³	10	16.17	7.76-29.74
Hispanic	ASD (Secundum and NOS)	52	23.76	17.75-31.16
	Obstructive Genitourinary Defect	45	20.57	15.00-27.52
	Trisomy 21 (Down Syndrome)	37	16.91	11.91-23.31
	Polydactyly/Syndactyly	35	16.00	11.14-22.25
	VSD (Membranous and NOS)	31	14.17	9.63-20.11

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.

³ Rate calculated using male live births.

Figure 13 Age-Adjusted Prevalence of Chromosomal and All Other Defects by Maternal Race / Hispanic Ethnicity, Massachusetts: 2008-2009

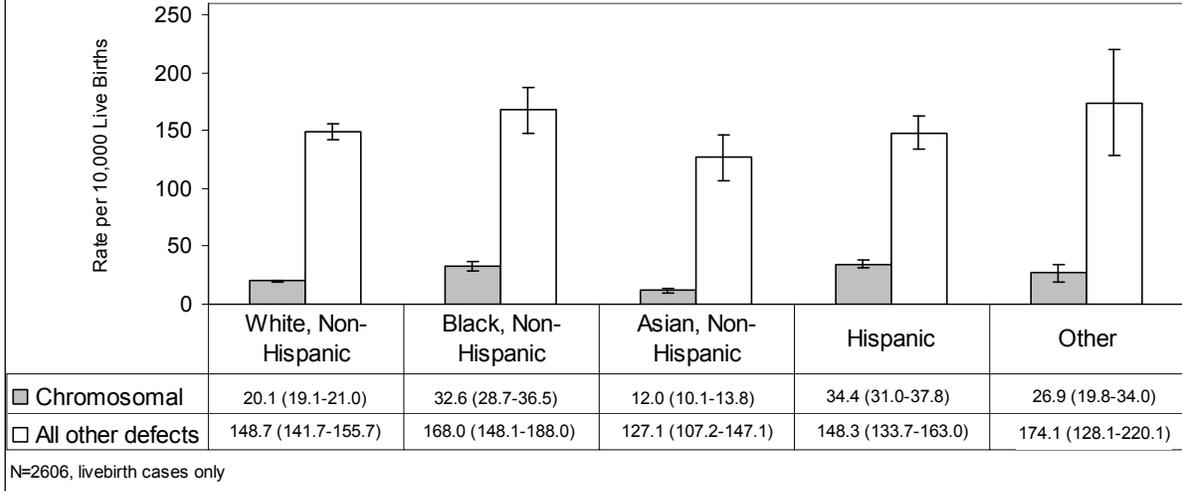


Figure 14 Prevalence of Birth Defects by Maternal Race / Hispanic Ethnicity and Birthplace, Massachusetts: 2008-2009

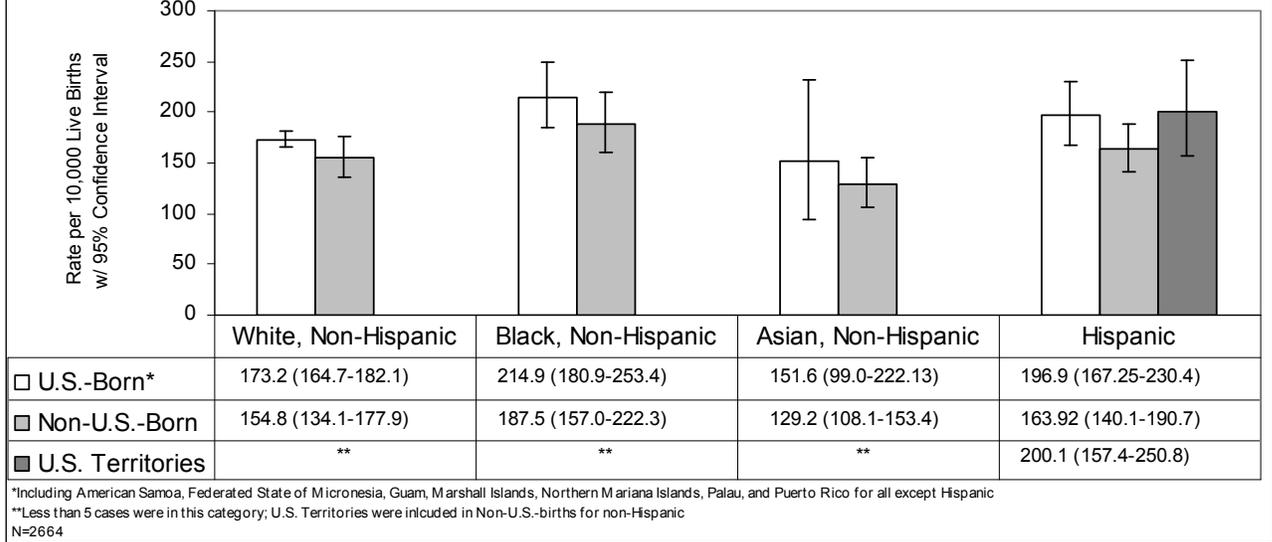


Figure 15 Age-Adjusted Prevalence of Birth Defects by Maternal Race / Hispanic Ethnicity, Massachusetts: 2004-2009

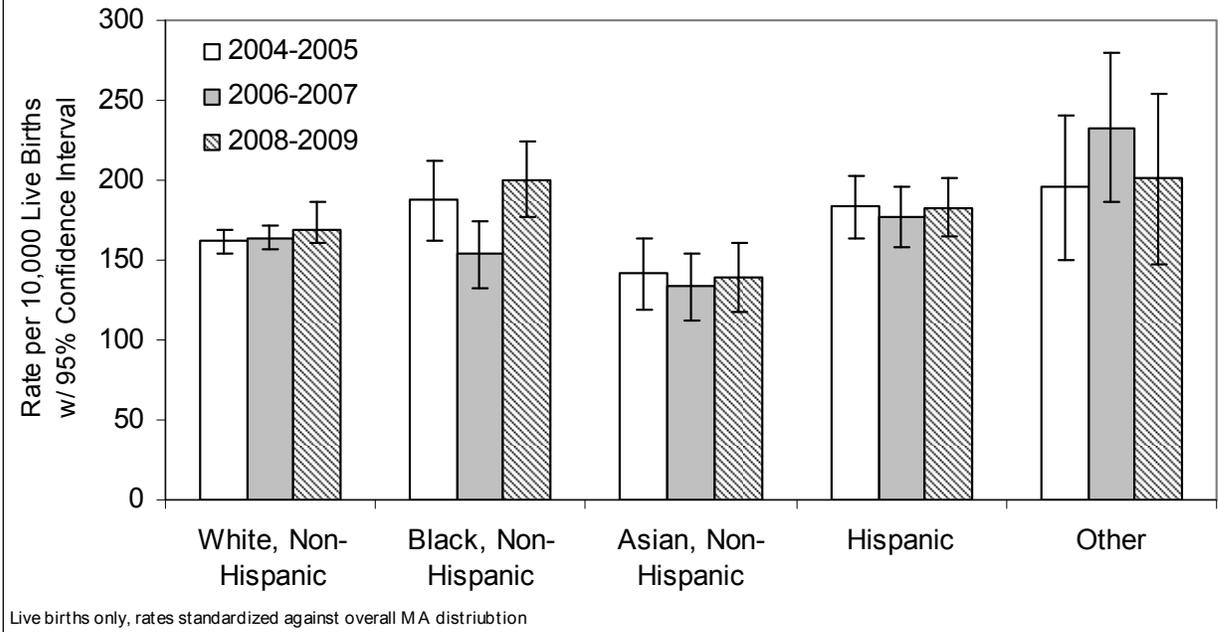
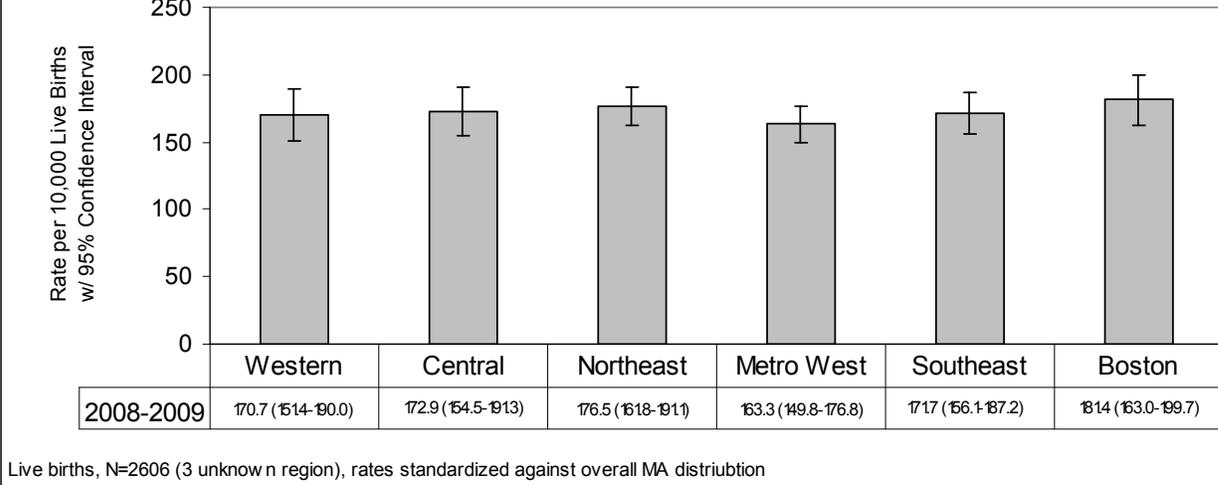


Figure 16 Age-Adjusted Prevalence of Birth Defect by Region, Massachusetts: 2008-2009



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. A severity scale was developed by the Center in collaboration with our partners at Boston University and the Massachusetts General Hospital. This scale was based on the usual outcome for a specific birth defect including its typical compatibility with survival, the need for immediate treatment, the need for long-term care and the amenability of the defect to correction.

We developed an automated algorithm to classify the majority of cases into the categories of “severe”, “serious”, “moderate”, and “mild” with a minority of cases requiring assignment by a clinician. “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 mostly 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.

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 and lack of prenatal diagnosis reporting. 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).

18.4% 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.

“Moderate” birth defects comprised about 70% of the total cases. All of these needed medical follow up and many may have required a number of surgeries and extensive treatment.

“Mild” birth defects comprised a little over 9% of the affected infants. These defects may or may not have required corrective treatment. 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) with 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 18.4% of the birth defects in Massachusetts in 2008-2009. Of the cases with known cause, “single gene” etiology accounted for 21.3%, “chromosomal” etiology accounted for 73.8% and “maternal-fetal factors” accounted for 2.7% of cases. The majority of birth defects cases in Massachusetts in 2008-2009 had an unknown etiology (81.6%).

As Figure 17 shows, single gene etiology accounted for 21.3% of the known etiology cases. Single gene defects include achondroplasia, Marfan syndrome (deletion 15q21.1), Smith-Lemli-Opitz syndrome and other examples of defects categorized as Mendelian syndrome. Chromosomal etiology accounted for almost 74% 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 and other factors accounted for 2.7% 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 “other factor” known etiology cases include 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 2008-2009 had an unknown etiology (2,175 of 2,664 live births and stillbirths). These were categorized by pattern and by description in Table 15.

Cases are also classified by the *pattern* of defects i.e. whether one defect occurs with others. Of all 2,664 birth defect cases (2,609 live births and 55 stillbirths) 43.4% had a “solitary” (truly a single) defect pattern, 26.1% had “Major plus minors” (defined as having a major defect accompanied by one or more minor defects), 4.6% were a “sequence” (allowing for more than one major defect if the defects are related pathogenically), and 25.9% had “multiple major” defects. Cleft lip with and without cleft palate, hypospadias (2nd or 3rd degree), gastroschisis, and craniosynostosis appeared more often as a single defect rather than with other defects—all occurring more than 75% of the time as a “solitary” defect. Birth defects which appeared more often in conjunction with other defects included Down syndrome (as well as the two other ascertained trisomies), obstructive genitourinary defect, and all cardiovascular defects.

See Figure 18 for the distribution of cases with unknown etiology among these patterns.

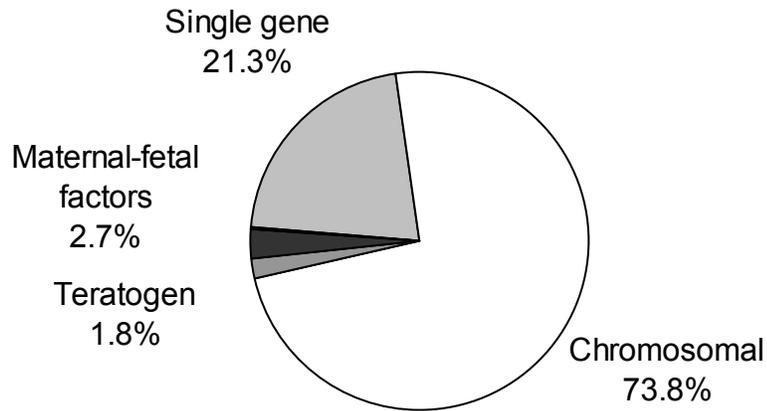
Table 14 Percentage of Birth Defect Cases by Severity Groups, Massachusetts: 2008-2009

SEVERITY CATEGORIES	PERCENTAGE OF BIRTH DEFECTS CASES
Severe, supportive measures, usually incompatible with life	2.8%
Serious, may be correctable, most have long-term needs	18.4%
Moderate, most correctable, many have long-term needs	69.5%
Mild, may be correctable, minimal long-term needs	9.2%

Table 15 Pattern and Etiology of Birth Defect Cases, Massachusetts: 2008-2009

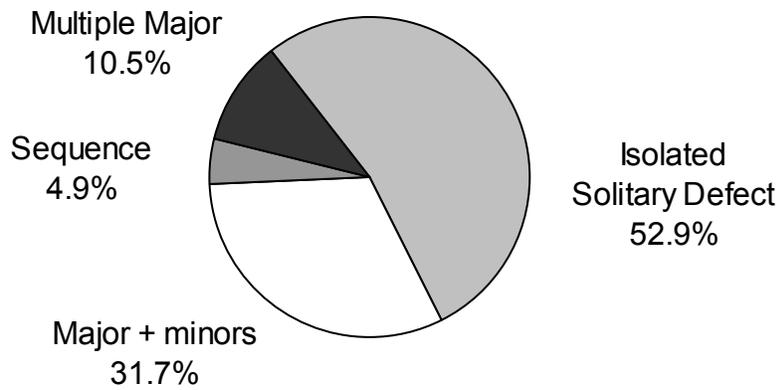
Pattern	Description	Cases	
		Known Etiology	Unknown Etiology
Isolated	Solitary defect	6	1,151
	Major and minors (different organ/body parts) or 2 or more defects (same organ/body part)	4	689
	Sequence: Common primary defect with consistent, related anomalies	15	107
Multiple Majors	2 or more major defects in different organs/body parts	462	228
Total		489 (18.4%)	2,175 (81.6%)

Figure 17 Birth Defect Cases with Known Etiology,
Massachusetts: 2008-2009



N=476
"other" contributes 0.4%

Figure 18 Patterns of Birth Defect Cases with Unknown Etiology,
Massachusetts: 2008-2009



N=2175

Appendices

Technical Notes

Definitions

2008-2009 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 Massachusetts EOHHS-Defined Regions and Age-Adjusted Overall Birth Defects Prevalence, 2008-2009

Folic Acid Use/Awareness Tables

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 would not substantially change the ratio.

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

Assignment of Race/Ethnicity

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

Calculation of 2009 Dollars

2009 dollars were calculated from the Bureau of Labor Statistics' CPI Inflation Calculator. The CPI inflation calculator uses the average Consumer Price Index for a given calendar year. This data represents changes in prices of all goods and services purchased for consumption by urban households. This index value has been calculated every year since 1913. For the current year, the latest monthly index value is used. http://www.bls.gov/data/inflation_calculator.htm

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.

A new automated algorithm was created for the 2004-2005 report 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 for 2008-2009 data was able to assign severity levels to about 96% of the cases, with the remaining 4% assigned manually by the Center Clinical Geneticist. The process that included the automated categorization system produced percentages of birth defects within each of the four severity categories in 2008-2009 that were similar to those attained in each of the categories in previous reports.

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.

Pattern assignment definitions

Solitary defect: Truly solitary defect

Major and Minor defects: More than one defect of the same organ or body part; Major plus minor defects in different organs or body parts

Sequence: Several defects in different organs/body parts that are related pathogenically

Multiple major defects: Multiple major defects in different organs and/or body parts including all defects that arise from a recognized syndrome.

Definitions

These definitions are derived from the Massachusetts Department of Public Health report titled Massachusetts Births, 2007 except where noted.

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.

Interpregnancy Interval (IPI): The time period in completed months between the date of conception of one pregnancy and the date of delivery of the preceding pregnancy.

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.

Small for gestational age: Term used to describe an infant whose birth weight is below the 10th percentile (i.e. below 90% of infants) for gestational age on basis of a sex-specific US standard (Oken 2003).

Stillbirth

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

2008 and 2009 Populations Used in Calculating Rates

Numbers of Live Births to MA Residents*				
		2008 N=76,969	2009 N=74,966	Total N=151,935
By Maternal Age	<20	4,623	4,528	9,151
	20-24	12,475	12,048	24,523
	25-29	19,019	18,469	37,488
	30-34	23,152	23,143	46,295
	35+	17,679	16,774	34,471
	Unknown**	3	4	7
By Infant's Sex	Male	39,447	38,534	77,981
	Female	37,521	36,431	73,952
	Unknown**	1	1	2
By Plurality	Singleton	73,475	71,423	144,898
	Multiple Birth	3,494	3,543	7,037
By Maternal Race/Ethnicity	White	51,760	49,759	101,519
	Black	6,652	6,945	13,597
	Hispanic	10,895	10,986	21,881
	Asian/PI	5,958	5,939	11,897
	American Indian**	145	122	267
	Other, non-Hisp**	1,417	1,036	2,453
	Unknown**	142	179	231
By EOHHS Region	Western	8,952	8,655	17,607
	Central	10,091	9,828	19,919
	Northeast	15,969	15,808	31,777
	Metro West	17,572	16,986	34,558
	Southeast	14,023	13,458	27,481
	Boston	10,362	10,231	20,593

* data runs for total year and two-year total in each demographic strata (i.e. age, sex, plurality, race/eth) are mutually exclusive.

** some variables not included (or categories are collapsed) for risk factor analysis due to low cell counts

Source: MassCHIP v3.0r326

Birth Defect Codes and Exclusions¹ by Defect Category		
Defect	ICD-9 / BPA²	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 palpebral 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 palpebral 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 Exclusions¹ by Defect Category (cont'd)		
Defect	ICD-9 / BPA²	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	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 Exclusions¹ 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 Exclusions¹ 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 Exclusions¹ 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 2008-2009

BPA Label	BPA Code	# of defects
Central Nervous System		
Anencephaly	740020	11
Meningomyelocele/myelomeningocele, Highest level, lumbar, Arnold Chiari malformation ± hydrocephalus, open lesion	741003	8
Meningomyelocele/myelomeningocele, Highest level unspecified, Arnold Chiari malformation ± hydrocephalus, open lesion	741009	1
Meningomyelocele/myelomeningocele, Highest level, lumbar, Arnold Chiari malformation ± hydrocephalus, closed lesion	741103	2
Lipomeningomyelocele, Highest level, sacral, Arnold Chiari malformation ± hydrocephalus, closed lesion	741144	2
Meningomyelocele/myelomeningocele, Highest level, thoracic, Hydrocephalus, other (aqueduct of Sylvius) or NOS, open lesion	741302	1
Meningomyelocele/myelomeningocele, Highest level, lumbar, Hydrocephalus, other (aqueduct of Sylvius) or NOS, open lesion	741303	2
Lipomeningomyelocele, Highest level, lumbar, Hydrocephalus, other (aqueduct of Sylvius) or NOS, closed lesion	741443	1
Meningomyelocele/myelomeningocele, Highest level, thoracic, No mentioned hydrocephalus, open lesion	741702	1
Meningomyelocele/myelomeningocele, Highest level, sacral, No mentioned hydrocephalus, open lesion	741704	1
Meningomyelocele/myelomeningocele, Highest level unspecified, No mentioned hydrocephalus, open lesion	741709	1
Unspecified spina bifida, Highest level, sacral, No mentioned hydrocephalus, open lesion	741794	1
Meningocele, Highest level, sacral, No mentioned hydrocephalus, closed lesion	741814	1
Lipomeningomyelocele, Highest level, lumbar, No mentioned hydrocephalus, closed lesion	741843	3
Lipomeningocele, Highest level, lumbar, No mentioned hydrocephalus, closed lesion	741853	2
Lipomeningocele, Highest level, sacral, No mentioned hydrocephalus, closed lesion	741854	1
Meningomyelocele/myelomeningocele, Highest level, lumbar, No mentioned hydrocephalus, unspecified open/closed lesion	741903	1
Meningomyelocele/myelomeningocele, Highest level, sacral, No mentioned hydrocephalus, unspecified open/closed lesion	741904	1
Meningocele, Highest level, sacral, No mentioned hydrocephalus, unspecified open/closed lesion	741914	1
Unspecified spina bifida, Highest level, lumbar, No mentioned hydrocephalus, unspecified open/closed lesion	741993	1
Unspecified spina bifida, Highest level unspecified, No mentioned hydrocephalus, unspecified open/closed lesion	741999	1
Encephalocele, Occipital	742000	1
Encephalocele, Frontal (including proencephalon)	742085	1
Encephalocele, NOS	742090	1
Microcephalus	742100	25
S Cerebrum anomalies	742200	2
Corpus callosum anomalies (don't code colpocephaly with ACC)	742210	69
Cerebellum anomalies	742230	7
Cerebellar Hypoplasia	742235	2
Agyria and lissencephaly	742240	4
Microgyria / polymicrogyria	742250	18
Holoprosencephaly, NOS	742260	2
Holoprosencephaly, Alobar	742265	2
Holoprosencephaly, Semilobar	742266	3
Holoprosencephaly, Lobar	742267	3
S Brain, reduction defect OS (8/02 Includes colpocephaly, pachygyria, schizencephaly) & absent septum pellucidum	742280	31
Hydrocephaly, Anomalies of Aqueduct of Sylvius	742300	9
Dandy-Walker Malformation	742310	10
Hydranencephaly	742320	7
Hydrocephaly, Other Specified	742380	13
Hydrocephaly, NOS	742390	16
Enlarged brain and head / enlarged head / enlarged brain / megalencephaly / macrocephaly	742400	17
Brain cysts: Porencephaly / porencephalic	742410	2
S Brain cysts: Cerebral / subependymal / periventricular	742420	2
Brain: Other specified anomalies / cortical atrophy / cranial nerve defects	742480	5
Spinal cord: Diastematomyelia	742520	1
Spinal cord: Other specified anomalies (Includes tethered cord) (and arachnoid cyst)	742580	57

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2008-2009

BPA Label	BPA Code	# of defects
Eye		
Microphthalmos, Left	743101	14
Microphthalmos, Right	743102	2
Microphthalmos, Bilateral	743104	6
Buphthalmos/Congenital Glaucoma, Laterality Unk	743200	1
Buphthalmos/Congenital Glaucoma, Left	743201	1
Buphthalmos/Congenital Glaucoma, Right	743202	1
Buphthalmos/Congenital Glaucoma, Bilateral	743204	7
Absence of lens/Congenital Aphakia, Right	743302	1
Cataract, NOS, Left	743321	9
Cataract, NOS, Right	743322	9
Cataract, NOS, Bilateral	743324	12
Coloboma of lens, Laterality Unk	743340	1
Cataract, anterior polar, Left	743351	2
Cataract, anterior polar, Right	743352	2
Cataract, other specified, Left	743361	2
Cataract, other specified, Bilateral	743364	2
S Cornea, other anomalies. Excludes: megalocornea (use 743.220)	743410	6
Absence of iris/Aniridia, Bilateral	743424	1
S Iris: Coloboma	743430	8
S Anterior segment: OS colobomas and anomalies (Use for Rieger SYNDROME, use 759800. For Reiger ANOMALY, use 743470-474)	743480	3
S Anterior segment: Unspecified colobomas and anomalies	743490	2
S Vitreous humor: Specified anomalies (includes PHPV)	743500	2
S Retina: Specified anomalies / congenital retinal aneurysm. Excludes:Stickler syndrome(use_759.860)	743510	3
S Optic disc: Specified anomalies / hypoplastic optic nerve / coloboma of the optic disc	743520	22
S Choroid: Coloboma	743535	5
S Eyelids: Coloboma	743636	1
S Tear ducts: # STENOSIS, stricture, or obstruction of lacrimal duct	743650	3
S Absence or stricture of auditory canal	744000	8
S Anomaly of middle ear / fusion of ossicles	744020	7
S Anomaly of inner ear / congenital anomaly of membranous labyrinth or organ of Corti	744030	9
S Ear: Unspecified anomalies with hearing impairment / congenital deafness, NOS	744090	5
S # Ear : ACCESSORY auricle / polyotia	744100	2
Microtia, Left	744211	10
Microtia, Right	744212	13
Microtia, Bilateral	744214	4
S Ear: Other misshapen ear / cleft / malformed /#POINTED / # ELFIN,pixie-like / # LOP / # CAULIFLOWER / # ABSENT or decreased cartilage -- a conditional exclusion if <36wks	744230	14
S Ear: Other specified anomalies (see also 744.230) / #DARWIN tubercle	744280	6
Cardiovascular		
Anomalous Pulmonary Venous Connection		
Total anomalous pulmonary venous return/connection/drainage	747420	9
Partial anomalous pulmonary venous return/connection/drainage	747430	11
Atrioventricular Canal Defects		
Atrial septal defect, primum type (ASD1)	745600	2
Common Atrium	745610	10
Complete atrioventricular canal (CAVC)	745630	45
Endocardial cushion defect, Other specified	745680	21
Ventricular septal defect, inflow type (subtricuspid, canal-type) (VSDavc)	745685	3
Endocardial cushion defect, NOS	745690	4
Conotruncal (Outlet) and Aortic Arch		

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2008-2009

BPA Label	BPA Code	# of defects
Truncus Arteriosus	745000	4
Dextro-transposition of great arteries (dTGA, dTGV) w/ intact ventricular septum	745100	22
Dextro-transposition of great arteries (dTGA, dTGV) w/ ventricular septal defect	745110	18
Double-outlet right ventricle (DORV) with normally related great arteries	745185	7
Double-outlet right ventricle (DORV) with transposed great arteries	745186	5
Double-outlet right ventricle (DORV), Other Specified	745188	1
Double-outlet right ventricle (DORV), NOS	745189	2
Tetralogy of Fallot	745200	41
Interrupted aortic arch, type B	747217	5
Pulmonary atresia with VSD (tetralogy of Fallot with pulmonary atresia)	747310	19
Ebstein Anomaly		
Ebstein Malformation or Anomaly	746200	4
Heterotaxy (Laterality Defects)		
Complete situs inversus w/ dextrocardia	759300	7
Situs inversus w/ levocardia	759310	1
Situs ambiguus, right; right isomerism	759350	2
Situs ambiguus, left; left isomerism	759360	2
Situs ambiguus, sidedness NOS	759380	13
Left-Sided Obstruction		
Aortic stenosis, valvar	746300	21
Hypoplastic left heart syndrome	746700	20
Coarctation of the aorta (COA), preductal (proximal)	747100	1
Coarctation of the aorta, juxtaductal	747120	10
Coarctation of the aorta, NOS	747190	55
Interrupted aortic arch, type A	747216	1
Other Cardiovascular		
Aortic septal defect / aortopulmonary window. Excludes: atrial septal defect(use 745.590)	745010	2
Pulmonary valve: Other specified anomalies. Excludes: infundibular PS (746.830)	746080	13
Aortic valve: bicuspid BAV / insufficiency or regurgitation / # MILD',MINIMAL', 'TRIVIAL',or 'PHYSIOLOGIC' ~	746400	1
Bicuspid Aortic Valve (BAV); new code for cases starting DOB 1/1/07	746470	60
Aortic valve: Other specified anomalies / aortic valve atresia. Excludes: supravalvular aortic stenosis(747.220)	746480	23
Mitral valve: Congenital mitral stenosis	746500	10
Mitral valve: Absence, atresia, or hypoplasia	746505	9
Mitral valve: insufficiency or regurgitation, congenital/# MILD', MINIMAL',TRIVIAL', or 'PHYSIOLOGIC' ~	746600	3
Situs: Dextrocardia without situs inversus / dextrocardia with situs solitus. Excludes: dextrocardia with situs inversus (use_759.300)	746800	5
Cor triatriatum	746820	2
Pulmonary infundibular (subvalvular) stenosis	746830	14
Heart: Other specified anomalies / ectopia cordis / mesocardia / conduction defects, NOS	746880	87
Hypoplastic left ventricle. Excludes: hypoplastic left heart syndrome (746.700)	746881	1
Hypoplastic right heart or right ventricle / Uhl's disease (parchment RV)	746882	7
Anomalies of coronary artery or sinus	746885	21
"Pulmonic" or pulmonary atresia, stenosis, or hypoplasia, NOS w/ no mention of whether valve or artery	746995	1
Aorta: Hypoplasia	747210	21
Supra-aortic stenosis / supravalvular aortic stenosis. Excludes: aortic stenosis, congenital(see 746.300)	747220	4
Aorta: Persistent right aortic arch	747230	35
Aorta: Vascular ring / double aortic arch / vascular ring compression of trachea	747250	8
Aorta: Congenital aneurysm / dilatation	747270	6
Aorta: Other specified anomalies	747280	1
S Pulmonary artery: stenosis. Use 746.995 if artery or valve is not specified	747320	15
S Pulmonary artery: other specified / pulmonary artery hypoplasia	747380	8
Persistent left superior vena cava	747410	42

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2008-2009

BPA Label	BPA Code	# of defects
Great veins: Other specified anomalies (includes IVC interruption, bilateral SVC)	747480	28
S Peripheral arteries: Other anomalies / aberrant subclavian artery	747640	27
Cerebral vessels: Other anomalies / vein of Galen	747810	1
S Circulatory system: Other specified anomalies. Excludes cong aneurysms: coronary ~ (746.880), peripheral ~ (747.640), pulmonary~ (747.330), retinal ~(743.510), ruptured cerebral arterioven	747880	7
Patent Ductus Arteriosus		
# PATENT ductus arteriosus (PDA). Always code if >=36 wks. and >=6 weeks of age. Always code if >=36 wks with a medical / surgical intervention such as indomethacinor surgical ligation. Otherwise, a	747000	221
Right-Sided Obstruction		
Pulmonary valve atresia/intact ventricular septum	746000	10
Pulmonic stenosis, valvar	746010	95
Pulmonary valve atresia with VSD (not TOF variant 747.310)	746030	3
Tricuspid atresia	746100	13
Septal Defects		
Ventricular septal defect, Perimembranous (type II, membranous) (VSDmem)	745485	180
Ventricular septal defect, Malalignment-type (type I, subarterial) (VSDmal)	745487	29
Ventricular septal defect, NOS	745490	11
Atrial septal defect, Secundum type (ASD2)	745510	289
Atrial septal defect, OS	745580	1
Atrial septal defect, NOS	745599	49
Single Ventricle and L-TGA		
L-TGA /Corrected transposition of great vessels / ventricular inversion. Excludes: dextrocardia (use 746800)	745120	5
Single ventricle, NOS	745300	1
Single ventricle, Double Inlet Left Ventricle	745310	2
Single ventricle, Double Inlet Right Ventricle	745320	1
Respiratory		
S Choanal stenosis (For NBDPS: choanal atresia = 748.010 etc)	748000	1
Choanal atresia, Left	748011	2
Choanal atresia, Right	748012	2
Choanal atresia, Bilateral	748014	6
Nose: Agenesis or underdevelopment	748100	5
Nose: Tubular / single nostril / proboscis (most cases with proboscis have holoprosencephaly)	748185	1
Larynx: Web, glottic	748205	1
Larynx: Web, NOS	748209	2
Congenital subglottic stenosis. Excludes: stenosis that the chart states was acquired or secondary to endotracheal (ET)intubation or ventilation.	748310	1
Other anomalies of trachea. Excludes: vascular ring compression of the trachea (use 747.250)	748330	3
S Other anomalies of bronchus	748350	1
Larynx: Cleft / laryngotracheoesophageal cleft / 1/04: use for laryngeal atresia/stenosis	748385	24
S Lung cysts: Single	748400	1
S Lung cysts: CCAM (cong cystic adenomatoid malf), Other specified	748480	13
S Lung agenesis or aplasia	748500	1
S # HYPOPLASIA of lung or pulmonary hypoplasia -- a conditional exclusion only in infants <36wks.	748510	2
S Lung: sequestration	748520	5
Lung: Bilobed right / right lung with left lung bronchial pattern	748625	2
S Lung: Other and unspecified anomalies	748690	2
S Respiratory system: Other specified anomalies / congenital lobar emphysema / lymphangiectasia of lung	748880	3
Orofacial		
Pierre Robin sequence (not a true "syndrome")	524080	35
S Branchial cleft, sinus, fistula, cyst, or pit	744400	35
S Other branchial cleft anomalies / dermal sinus of head	744480	12
S Face or neck: Other specified anomalies (6/03 eg. facial cleft)	744880	7

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2008-2009

BPA Label	BPA Code	# of defects
Nose: Fissured, notched, or cleft	748120	1
Nose: OS anomalies (For NBDPS: nasal pyriform aperture stenosis is here, not Cho Sten)/ small nose and nostril / absent nasal septum / # FLAT or WIDE nasal bridge/ #BEAKED nose	748180	25
Cleft hard palate, Bilateral	749010	7
Cleft hard palate, Central	749020	2
Cleft hard palate, NOS	749030	16
Cleft soft palate, Bilateral	749050	1
Cleft soft palate, Central	749060	1
Cleft soft palate, NOS	749070	45
Cleft palate, NOS	749090	15
Cleft lip, Unilateral, Left	749101	17
Cleft lip, Unilateral, Right	749102	15
Cleft lip, Bilateral	749110	2
S Cleft: Incomplete CL/ microform /pseudo / fused lip /healed lip	749190	5
Cleft lip, NOS	749195	4
Cleft lip and palate, Unilateral cleft lip, Left	749201	23
Cleft lip and palate, Unilateral cleft lip, Right	749202	22
Cleft lip and palate, Bilateral cleft lip	749210	30
Cleft lip and palate, Central cleft lip	749220	2
Cleft lip and palate, NOS	749290	3
Tongue: large / macroglossia	750120	2
Gastrointestinal		
Esophageal atresia without TE fistula	750300	4
Esophageal atresia with TE fistula	750310	29
Tracheoesophageal fistula without mention of esophageal atresia	750320	1
Tracheoesophageal fistula, "H" type	750325	3
Microgastria	750700	1
Persistent omphalomesenteric duct / persistent vitelline duct	751000	2
# MECKEL'S diverticulum	751010	16
Intestinal atresia/stenosis, Duodenum	751100	25
Intestinal atresia/stenosis, Jejunum	751110	15
Intestinal atresia/stenosis, Ileum	751120	9
Intestinal atresia/stenosis, Large Intestine, NOS	751200	2
Rectal atresia/stenosis without mention of fistula	751220	2
Anal atresia with fistula	751230	20
Anal atresia without mention of fistula	751240	18
Hirschsprung disease: Long-segment (aganglionosis beyond rectum)	751310	19
Hirschsprung disease: Short-segment (aganglionosis involving no more than the anal sphincter and the rectum)	751320	6
Hirschsprung disease, NOS	751330	10
Malrotation: cecum and/or colon	751400	1
Congenital adhesions or bands of omentum and peritoneum / Ladd's bands	751420	2
Malrotation: Other specified and unspecified	751490	52
Malrotation: small intestine alone	751495	2
Duplication of anus, appendix, cecum, or intestine / enterogenous cyst	751500	6
Microcolon	751520	8
Ectopic (displaced, anteriorly placed) anus	751530	9
Intestine: Other specified anomalies / # RECTAL fissures	751580	3
Biliary atresia, extrahepatic or NOS (use 751.670 for intrahepatic)	751650	10
Choledochal cysts	751660	3
Pancreas: Annular	751720	5
Genitourinary		
Gyne: S Ovaries, Other specified anomalies	752080	2
Gyne: S Ovaries, Multiple cysts	752085	5

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BPA Label	BPA Code	# of defects
Gyne: Uterus, other anomalies / bicornuate/ unicornis	752380	1
Gyne: Vulva Absence or anomaly / # FUSION / # HYPOPLASTIC labia majora -- conditional exclusion if <36 wks.	752440	1
Gyne: OS anomalies of cervix, vagina, or external female genitalia /# VAGINAL tags / # HYMENAL tags	752480	2
Hypospadias, Second Degree	752606	65
Hypospadias, Third Degree	752607	12
Hypospadias, Second Degree with Chordee	752626	75
Hypospadias, Third Degree with Chordee	752627	40
Indeterminate sex, NOS / ambiguous genitalia	752790	14
Testis and scrotum: Other anomalies / polyorchidism / bifid scrotum. Excludes: torsion of the testes or spermatic cord(608.200)	752820	13
Penis: Other anomalies / concealed penis / absent or hooded foreskin / #REDUNDANT foreskin (Redundant foreskin is never coded.)	752860	54
Penis: Small / hypoplastic / micropenis	752865	6
Genital organs: Other specified anomalies / microgenitalia / macrogenitalia	752880	1
Renal agenesis, bilateral	753000	7
Renal hypoplasia, bilateral	753005	2
S Kidney/renal: cyst, single	753100	8
Kidneys: Polycystic, infantile type (IPKD)	753110	3
Kidneys: Polycystic, adult type (APKD)	753120	1
S Kidneys: Multicystic renal dysplasia / multicystic kidney	753160	34
S Congenital hydronephrosis / pyelocaliectasis	753200	214
S Atresia, stricture, or stenosis of ureter / ureteropelvic junction obstruction or stenosis /ureterovesical junction obstruction or stenosis / hypoplastic ureter	753210	54
S Megaloureter, NOS / hydroureter	753220	37
S Other and unspecified obstructive defects of renal pelvis and ureter	753290	1
S Kidney: Double or triple, pelvis / pyelon duplex or triplex	753310	12
S Kidney: Lobulated, fused, or horseshoe / crossed fused ectopia	753320	7
S Kidney: Ectopic / pelvic	753330	7
S Kidney: Enlarged, hyperplastic, or giant	753340	1
S Ureter: Accessory / double ureter / duplex collecting system	753410	38
S Ureter: Ectopic	753420	9
S Ureter: Other specified anomalies / ureterocele	753480	25
S Ureter: Variations of vesicoureteral reflux	753485	91
Bladder exstrophy	753500	3
Urethra: Congenital posterior urethral valves or posterior urethral obstruction	753600	23
Other atresia, or stenosis of bladder neck	753610	2
Obstruction, atresia or stenosis of urinary meatus / meatal stenosis	753630	2
Other and unspecified atresia and stenosis of urethra and bladder neck	753690	3
# Urachus: PATENT	753700	3
Urachus: Cyst	753710	11
Absence of bladder or urethra	753800	4
Ectopic bladder	753810	1
Double urethra or urinary meatus	753840	1
Other specified anomalies of bladder and urethra	753880	3
Musculoskeletal		
Certain cong musculoskeletal anomalies face, face, jaw: Use for asymmetry of face	754000	6
Congenital postural scoliosis	754200	15
S Bowing, femur	754400	2
S Bowing, tibia and/or fibula	754410	2
S Dislocation of knee, congenital	754440	3
S Clubfoot: Talipes equinovarus	754500	93
S Clubfoot: Talipes calcaneovarus	754510	2
S Clubfoot: # METATARSUS varus or adductus	754520	12
S Clubfoot: Complex varus deformities	754530	2

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BPA Label	BPA Code	# of defects
S Unspecified varus deformities of feet	754590	1
S Talipes calcaneovalgus	754600	9
S Unspecified valgus deformities of foot	754690	1
S Clubfoot, NOS / talipes, NOS	754730	92
S Other specified deformities of ankle and / or toes / dorsiflexion of foot. Excludes: widely spaced first and second toes (use_755.600)	754780	3
S Other specified deformity of hands (see 755.500 for specified anomalies of fingers)	754880	1
S Polydactyly fingers / postaxial polydactyly, Type A	755005	60
S Polydactyly: Accessory thumbs (preaxial polydactyly)	755010	52
S Polydactyly: Accessory toes (postaxial)	755020	49
S Polydactyly: Accessory big toe (preaxial)	755030	11
S Polydactyly: Accessory digits hand, NOS (preaxial, postaxial not specified)	755095	4
S Polydactyly: Accessory digits foot, NOS (preaxial, postaxial not specified)	755096	11
S Syndactyly: Fused fingers	755100	25
S Syndactyly: Webbed fingers	755110	12
S Syndactyly: Fused toes	755120	25
S Syndactyly: Webbed toes / # WEBBING between the second and third toes.	755130	51
S Syndactyly: Unspecified (webbed vs. fused) thumb and / or fingers, NOS	755193	9
S Syndactyly: Unspecified toes	755194	4
S Syndactyly: Unspecified (webbed vs. fused) Toes	755199	6
Transverse deficiency or amputation of the arm, NOS, Bilateral	755204	1
Absence of the forearm and hand, Left	755241	3
Absence of the forearm and hand, Right	755242	1
Absence of hand or fingers, Left	755246	6
Absence of hand or fingers, Right	755247	4
Absence of hand or fingers, Bilateral	755249	3
Longitudinal deficiency of arm, NOS, Bilateral	755254	1
Split-Hand, Left	755256	1
Split-Hand, Right	755257	1
Split-Hand, Bilateral	755259	3
Thumb only missing or hypoplastic, Left	755261	2
Thumb only missing or hypoplastic, Right	755262	2
Thumb only missing or hypoplastic, Bilateral	755264	2
Radial aplasia/hypoplasia, Right	755267	3
Radial aplasia/hypoplasia, Bilateral	755269	1
Ulnar aplasia/hypoplasia, Right	755272	1
Ulnar aplasia/hypoplasia, Bilateral	755274	1
Transverse deficiency or amputation of the leg, NOS, Bilateral	755304	1
Absence of the lower leg and foot, Left	755341	1
Absence of foot or toes, Left	755346	2
Absence of foot or toes, Right	755347	4
Absence of foot or toes, Bilateral	755349	5
Split-Foot, Left	755356	1
Split-Foot, Bilateral	755359	3
Tibial aplasia/hypoplasia, Right	755367	1
Tibial aplasia/hypoplasia, Bilateral	755369	2
Fibular aplasia/hypoplasia, Left	755371	1
Fibular aplasia/hypoplasia, Right	755372	1
S Anomalies of fingers /camptodactyly/macro- /brachy-/clino-, triphalangeal thumb. Excludes:acrocephalosyndactyly(see756.050) /Apert synd(see756.055)	755500	38
S Radioulnar synostosis	755536	3
S Anomalies of elbow and upper arm	755540	3
S Upper limb: Hypoplasia / Fingers, hands, or arms: hypoplasia. Excludes: aplasia or absent upper limb (see 755.2)	755585	15
S Lower limb: other specified anomalies / hyperextended legs / shortening of legs	755680	2

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2008-2009

BPA Label	BPA Code	# of defects
S Lower limb: hypoplasia / Toes, feet, legs: hypoplasia. Excludes: aplasia of or absent lower limb (see 755.3)	755685	9
= Arthrogryposis multiplex congenita / distal arthrogryposis syndrome. Temporarily includes: one or more flexion contractures of individual joints	755800	14
Craniosynostosis, Sagittal	756005	38
Craniosynostosis, Metopic	756006	13
Craniosynostosis, Coronal, Left	756011	4
Craniosynostosis, Coronal, Right	756012	6
Craniosynostosis, Coronal, Bilateral	756014	5
Craniosynostosis, Lambdoidal, Left	756021	3
Craniosynostosis, Lambdoidal, Right	756022	2
Craniosynostosis, Lambdoidal, Bilateral	756024	2
Skull and face bone: Other specified anomalies / localized skull defects / mid-facial hypoplasia / prominent maxilla/hypotelorism / # FLATocciput / # PROMINENT occiput.	756080	6
Spine: Kyphosis / kyphoscoliosis	756120	10
Vertebrae, cervical: anomalies	756140	3
Vertebrae, cervical: hemivertebrae	756145	4
Vertebrae, thoracic: anomalies	756150	15
Vertebrae, thoracic: hemivertebrae	756155	16
Vertebrae, lumbar: anomalies	756160	8
Vertebrae, lumbar: hemivertebrae	756165	8
Vertebrae, sacrococcygeal: anomalies / agenesis of sacrum. Excludes: pilonidal sinus (see 685.100)	756170	22
Sacral agenesis	756175	1
Vertebrae: Other specified anomalies	756180	2
Vertebrae: Hemivertebrae, NOS	756185	1
Unspecified anomalies of spine	756190	1
S Ribs: Absence	756300	17
S Ribs: Fused	756320	10
S Ribs: Extra	756330	8
S Ribs: Other anomalies	756340	14
Sternum: Other anomalies / double ossification center in manubrium / bifid/ short	756380	2
= Achondroplasia	756430	4
= Thanatophoric dwarfism	756447	2
= Other specified chondrodystrophy. Excludes: Conradi's (use 756.575)	756480	3
= Osteogenesis imperfecta	756500	3
= Infantile cortical hyperostosis / Caffey syndrome	756530	1
= Conradi syndrome / chondrodysplasia punctata. Excludes: warfarin embryopathy	756575	2
= Other specified osteodystrophies	756580	2
= Unspecified osteodystrophies	756590	2
Diaphragmatic hernia, NOS (inc. absent/hemidiaphragm), Laterality Unk	756600	3
Diaphragmatic hernia, NOS (inc. absent/hemidiaphragm), Left	756601	23
Diaphragmatic hernia, NOS (inc. absent/hemidiaphragm), Right	756602	3
Diaphragmatic hernia, Bochdalek, Left	756611	6
Diaphragmatic hernia, Morgagni, Right	756617	1
Diaphragmatic hernia, Morgagni, Unilat, Side Unk	756618	1
S Diaphragm: Eventration	756620	8
Omphalocele	756700	18
Gastroschisis	756710	50
Prune belly syndrome	756720	2
Poland syndrome or anomaly	756800	4
S Other absent or hypoplastic muscle / absent pectoralis major. Excludes: prune belly syndrome (use 756720)	756810	1
= Amyotrophia congenita (= one specific type of arthrogryposis)	756840	1
Muscle, tendon, fascia and connective tissue: Other specified anomalies / myopathy, congenital NOS	756880	1

Chromosomal

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2008-2009

BPA Label	BPA Code	# of defects
DiGeorge S (10/02: Use for specific phenotype with chrome/FISH 22q, if available 758.370)	279110	4
Moebius syndrome (multiple cranial nerve palsies, esp. CN 7, limb, tongue abns)	352600	1
= Treacher-Collins syndrome / Mandibulofacial dysostosis	756045	2
= Other craniofacial syndromes / Hallermann-Streiff syndrome	756046	2
= Apert syndrome / Acrocephalosyndactyly types I or II	756055	1
= Goldenhar syndrome / oculoauriculovertebral dysplasia	756060	1
= Hemifacial microsomia	756065	7
= Ellis-van Creveld syndrome	756525	1
= Down syndrome: trisomy 21	758000	185
= Down syndrome: translocation 21, duplication 21q, Robertsonian translocation, isochromosome 21q	758020	4
= Down syndrome: mosaic	758040	3
= Trisomy 13 (archaic Patau syndrome): cytogenetics result in record	758100	4
= Trisomy 13: no cytogenetics in record	758190	1
= Trisomy 18 (archaic Edwards syndrome): cytogenetics result in record	758200	25
= Trisomy 18: translocation trisomy with duplication 18q	758220	1
= Deletion 21q, monosomy 21, or a G-group NOS (archaic)	758300	2
= Deletion 5p / Cri du chat syndrome	758310	5
= Deletion 17p or 18p / deletion of short arm chromosome 17 or 18	758350	3
= 22q11 deletion (Added 7/04: apply to 1/01. Also code phenotype if stated, eg. DGS 279.110)	758370	14
= Deletion: Autosome (not X or Y)(ie. #1-16, 4q,5q,19,20) / (From 8/02, used for 22q11, prior to the specific 22q code added 7/04)	758380	29
= Deletion: unspecified autosome	758390	1
= Trisomy 8	758500	2
= Trisomy: 6, 7, 9, 10, 11, 12 / Other trisomy C (archaic)	758510	3
= Trisomy, partial / 8/02 "partial trisomy" = "duplication". But, for "dup NOS" use 758930	758530	20
= 1/06 Clarified: "Other Trans" Incl Unbal AND Other Bal Translocations, OS. Excludes bal trans in normal (758.400)	758540	13
= Autosome OS: Other spec anomalies / marker / 8/02: Ring, derivative, mosaic, isochromosome, "additional" material / 3/03 inversions. 2/08 Never code "pericentric inv 9"	758580	16
= Turner phenotype: karyotype 45,X [XO] Note: The 7586xx code series that follows excludes pure gonadal dysgenesis(752.720)	758600	11
= Turner phenotype: variant karyotypes, eg. isochromosome, mosaic (eg X, XX,XY), partial X deletion, ring X chromosome. Excludes: Turner phenotype with normal karyotype	758610	4
= Klinefelter syndrome: 47, XXY	758700	5
= Klinefelter phenotype: other karyotype with additnl X chromosome, e.g., XXXY, XXY, XXXXY	758710	2
= Mosaic XO/XY, 45X/46XY. Excludes: with Turner phenotype(758.610)	758800	1
= XYY, male / 47,XYY / mosaic XYY male	758840	3
= XXX female / 47XXX / Triple X syndrome	758850	9
= Additional sex chromosomes, NOS	758860	1
= Sex chromosome: Other specified anomaly / fragile X	758880	5
= Unspecified chromosome: Deletion of chromosome(s), NOS	758920	1
Other specified DNA based diagnosis	758999	3
= Tuberous sclerosis / Bourneville's disease	759500	3
= Sturge-Weber syndrome/ Encephalocutaneous angiomatosis/	759610	2
= Malf. Syndromes/face: Aarskog /BOF /BOR /Fraser /FreemanSheldon / Kabuki / Miller-Dieker/ Noonan /Opitz G / oral-facial-digital/ Oto-palato-digital / Septo-optic dysplasia / Waardenburg / Williams	759800	19
= Malf. Syndromes/short stature: Smith-Lemli-Optiz /de Lange / Cockayne / Laurence-Moon-Biedl / Russell-Silver / Seckel	759820	5
= Malf. Syndromes/limbs: Baller-Gerold/ Carpenter / caudal regression /Fryns/ Holt-Oram / Klippel-Trenaunay-Webe/ LimbBodyWall /Roberts/ Rubinstein-Taybi / sirenomelia / thrombocytopenia-absent radius	759840	2
= Malf. Syndromes/other skeletal: Marfan / Stickler/ Beemer Langer	759860	8
= Malf. Syndromes/metabolic: Alagille /Alport / Beckwith-Wiedemann / Johansen-Blizzard/ Ieprechaunism / Lowe/ Menkes(kinky hair) /Prader-Willi/ Zellweger	759870	17
= Malf OS: VATER/VACTERL/Acardia/ Angelman/Bloom/CHARGE/hemihyper/Meckel-Gruber/Neu-Laxova/PentalogyCantrell/ Sotos/ TownesBrock/ WalkerWarburg/ Weaver / 10/02 VCFS,Shprintzen: code also chrome/FISH	759890	30

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BPA Label	BPA Code	# of defects
Other		
Amniotic band sequence	658800	13
= Collodion baby	757110	4
= Other and unspecified ichthyosis	757190	5
= X-linked ichthyosis	757196	3
Ichthyosiform erythroderma	757197	3
= Epidermolysis bullosa	757330	3
= Ectodermal dysplasia. Excludes: Ellis-van Creveld syndrome (756.525)	757340	3
= X-linked type ectodermal dysplasia (especially, HED Hypohidrotic Ectodermal Dysplasia)	757345	1
= Incontinentia pigmenti	757350	1
= Xeroderma pigmentosum	757360	1
Hair: Other specified anomalies	757480	3
Skin: Other specified anomalies / scalp defects. For specified anomalies of skin see 757.390. For specified anomalies of hair, see_757480. For specified anomalies of nails_757.580	757800	5
Spleen: Absence / asplenia	759000	2
Spleen: Accessory / 8/02 Use for polysplenia, though not exactly the same	759040	8
Spleen: Other specified anomalies	759080	1
Thyroglossal duct anomalies / thyroglossal cyst	759220	1
Anomalies of thymus / absent thymus / # THYMICHYPERTROPHY	759240	5

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

Birth Defects by Severity

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 Defect Terms¹

Achondroplasia: A genetic dysplasia of cartilage and long bones caused by mutations in the gene FGFR3. It results in disproportionate short stature with short limbs and relatively more normal trunk size. Persons affected with achondroplasia can have abnormalities of the foramen magnum potentially causing damage to the upper spinal cord. Spinal canal stenosis is a problem starting in late adolescence. There may also be lumbar lordosis, limited elbow extension and early arthritis. People with achondroplasia typically have normal intelligence.

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.

Birth Prevalence: (# of cases with birth defect A in an area and time period/ # of live births in that area and time period) x 10,000. *For more information see Technical Notes section.*

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).

Clubfoot: See *Talipes Equinovarus*.

Cluster: An apparently unusual concentration of a health condition in a particular area and time period.

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.

Confidence Interval (95%): The interval that contains the true prevalence (which we can only estimate) 95% of the time. *For more information see Technical Notes section.*

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 defects, 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.

Folate: B vitamin necessary for red blood cell production; folate deficiency can lead to anemia and, during embryogenesis, can affect the normal development of the fetus' neural tube; found in liver, green leafy vegetables, beans, beets, broccoli, cauliflower, citrus fruits, and sweet potatoes. *See folic acid.*

Folic Acid: One of the B vitamins especially important for a woman to take before conception to help prevent neural tube defects in a fetus; essential for DNA synthesis and therefore the growth and division of cells; obtained from fortified foods or from a multivitamin containing at least 4mg; also found in natural sources including liver, beans, and leafy green vegetables. While folate and folic acid are both forms of water-soluble B vitamins, folic acid refers to the synthetic vitamin used in supplements, whereas folate is the form found in foods.

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 proximal or middle segment(s) of a limb).

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

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.

Talipes equinovarus (Clubfoot): A development disorder of the foot and ankle that affects one (unilateral) or both (bilateral) feet. The foot is in an incorrect anatomical position, and is inclined inward, axially rotated outward, and points downward. Clubfoot is a complex disorder that is caused by genetic and environmental influences.

Tetralogy of Fallot (TOF): Congenital heart defect composed of ventricular septal defects, 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.

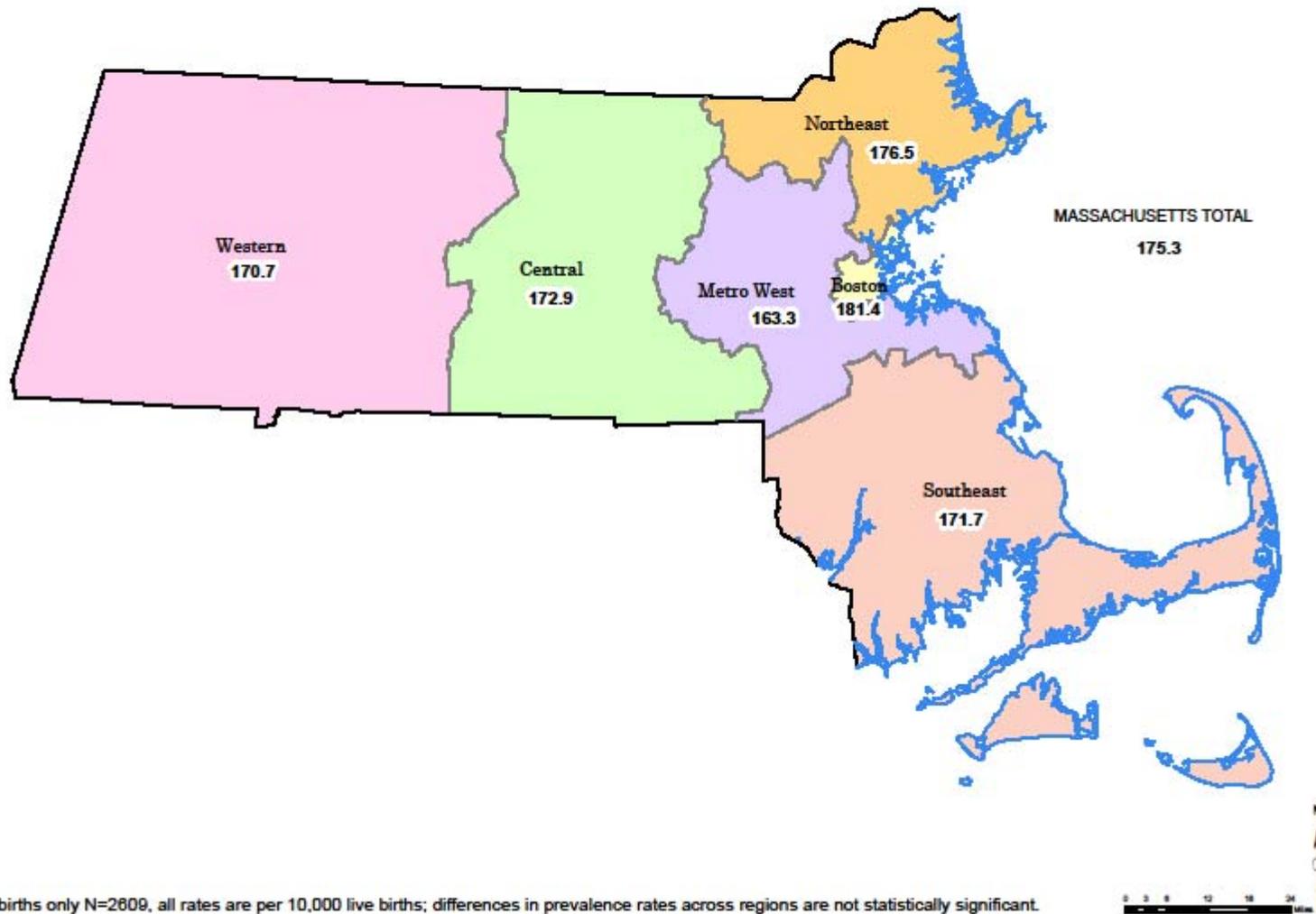
Trisomy 21: See **Down syndrome**.

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 Texas Birth Defects Epidemiology & Surveillance, Texas Department of State Health Services, <http://www.dshs.state.tx.us/birthdefects/glossary.shtm>. Modified 5/2/13, Accessed 3/14/13

Map of Massachusetts EOHHS-Designated Regions and Overall, Age-Adjusted Rate of Birth Defects, 2008-2009*



Frequency of Daily Multivitamin Use in the Month Prior to Pregnancy by Socio-demographic Characteristics, Massachusetts, 2009¹

Characteristic	Weighted n	Weighted %	95% CL	
Total	26921	37.6	34.3	- 41.1
Maternal race/ethnicity				
White, non-Hispanic	19958	41.7	36.8	- 46.6
Black, non-Hispanic	1830	28.2	23.4	- 33.6
Hispanic	2399	22.5	18.6	- 27.0
Asian, non-Hispanic	2397	42.1	36.4	- 48.0
Other, non-Hispanic	337	43.2	27.0	- 61.0
Maternal age (years)				
<20	630	15.8	10.0	- 24.1
20-29	8816	26.9	22.5	- 31.9
30-39	16342	50.5	45.3	- 55.7
40+	1133	46.3	28.7	- 64.8
Maternal education				
<High school	1523	24.1	17.0	- 32.9
High school diploma	4305	20.8	15.8	- 27.0
Some college	4746	34.1	27.3	- 41.7
College graduate	16347	53.4	48.0	- 58.7
Household poverty level				
≤100% FPL	3083	19.9	15.1	- 25.7
>100% FPL	23837	42.6	38.6	- 46.6
Maternal nativity				
Non-US-born	6038	31.7	27.5	- 36.1
US-born	20882	39.8	35.6	- 44.3

¹. Data obtained from resident women who had given live birth in Massachusetts in 2009. The survey was conducted by Massachusetts as part of the CDC Pregnancy Risk Assessment Monitoring System (PRAMS).

Folic Acid Awareness and Behavior in Women Ages 18-44,
Massachusetts 2000 and 2004¹

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. 2004 is the final year this question was included on the survey.

² Underlying sample size is less than 50 respondents (insufficient data).

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