Massachusetts Birth Defects Among Live and Stillbirths 2008-2010

Massachusetts Birth Defects Monitoring Program
Bureau of Family Health and Nutrition

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Note to Readers: Changes in this year's report

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

Section	Change or addition
Chapter 3	The figure for the overall trend in birth defect prevalence added in the last report was modified to represent a 2-year rolling.
	A chart depicting a ten-year trend in the prevalence of hypospadias was added.
Chapter 5	The previous bar-chart comparing two-year, age-adjusted overall birth defect prevalence rates among race/ethnicities was replaced by a line-graph showing the 2-year rolling averages for the overall, age-adjusted birth defect prevalence rates. This does a better job showing trends over time.

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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 among the top ten causes of death among 15-24 year olds in Massachusetts. Nationally, about 20% of all infant deaths—defined as death within 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, from the month before through the 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. Although there is increased awareness of the importance of folic acid in preventing birth defects, almost half of Massachusetts mothers in Massachusetts 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 $141.8 million in 2010 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, 2009, and 2010 and adds one year of new data to the previously published 2008-2009 report. Due to a change to the birth certificate in 2011 the BDMP decided to produce a report that includes three years of available data prior to this changeover. 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 2008-2010 data are presented in combined form since the numbers are relatively small for individual defects. As in previous years, given the BDMP's continued improvements in 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 newborn screening. 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. Preliminary trend analysis using data from this report and previous reports may be performed with the understanding that better ascertainment of cases and increased use of diagnostic technologies may be factors in any apparent increase in individual and overall 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

Understanding frequencies and trends of various birth defects allows for better planning of services for families of affected infants and children who may have special health care needs. Coordination between the BDMP and Maternal & Child Health Programs such as Early Intervention helps to ensure services for these children and their families. Identifying trends in birth defects over time can also help to guide future program planning and prevention strategies.

Prevalence

Figure 1 shows the trend in birth defect prevalence over the ten-year period ending in 2010. The overall prevalence of birth defects among live births to Massachusetts residents in 2008-2010 was 178.5 per 10,000 live births. Among the 224,770 live births and 1,135 stillbirths to Massachusetts residents in 2008-2010, 3,931 (1.7%) live births and 81 (7.1 %) stillbirths had one or more birth defects. While Table 1 shows the prevalence of all defects tracked by the BDMP, table 4 shows those that are the most common between 2008 and 2010. The ten most common defects were unchanged from the previous two reports. Three of these ten defects were cardiovascular defects: atrial septal defects, ventricular septal defects, 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 publishes periodic national prevalence estimates for 21 selected defects. In the last review, 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 2008-2010 were significantly lower than national rates for approximately 45% of the defects and were about the same as the national estimates for the other 55%. The lower rates for certain defects may reflect differences in surveillance system methodology and regional variation. Also, Massachusetts does not report prenatally diagnosed birth defects if the pregnancy is electively terminated, and this 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 rate estimates that include such cases became similar to or slightly higher than the average U.S. rates. In addition, since information on stillbirths with a birth defect is sometimes limited, these cases cannot always be included in surveillance, leading to lower estimated prevalence rates.

Selected Pregnancy Outcomes

We compared selected pregnancy outcomes, including Cesarean-sections (C-sections), birth weight, gestational age, multiple birth and infant death among infants born with birth defects to those born without birth defects in 2008-2010. Of infants born with a birth defect, 46.7% were C-section deliveries, compared to 33.4% of unaffected births; 21.3% of birth defect cases were of low birth weight (<2500 grams; 5.5 lbs) as opposed to 7.5% of those without a birth defect; 20.5% of infants with defects were premature (gestational age < 37 weeks) compared with 8.5% of those without a birth defect; and 4.8% 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 adverse outcomes (Figure 4).

Sex

The birth defect case prevalence was 146.8 per 10,000 live births for females and 207.3 per 10,000 live births for males (Table 6). While the prevalence of most types of birth defects did not significantly differ by sex of the infant/fetus, some did. As seen in Table 7, the most common defects seen in males, in descending order of prevalence were hypospadias (2nd or 3rd degree), atrial septal defects, obstructive genitourinary defect, polydactyly/syndactyly, and clubfoot. The most common defects seen in females were atrial septal defects, ventricular septal defects, 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 since the number of multiple births has been increasing 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 late losses not recorded at birth and is therefore thought to be more accurate, plurality from the BDMP's medical record abstraction is used in this report. The birth defect case prevalence was 173.8 per 10,000 live births for singletons and 275.0 per 10,000 live births for multiple births, so the risk of a baby who was born as part of a set of twins, triplets, etc. is considerably higher than a baby born alone (Table 8).

The birth defects that occurred most often among multiple births (all of which occurred more often than in singleton births) were atrial septal defects, ventricular septal defects, hypospadias (2nd and 3rd degree), clubfoot, and obstructive genitourinary defect (Table 9).

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; table 10 shows overall and defect-specific prevalence rates across age groups for the 2008-2010 period. For live births only, rates per 10,000 live births were 178.7 for mothers younger than 20 years, 177.0 for those 20-24 years, 167.1 for those 25-29 years, 157.7 for those 30-34 years and 203.5 for those 35 years and older. Mothers younger than the age of 20 had the highest rate (14.5 per 10,000) of gastroschisis among live births. 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 live born babies with Down syndrome were born to women 35 years or older, the Down syndrome rate of 29.5 per 10,000 live births for women 35 years and older was about four times that of mothers under the age of 35 years (7.3 per 10,000 live births). The pattern of higher Down syndrome rates among babies born to older women reflects the general pattern of greater frequency of chromosomal defects among the oldest maternal age group. One defect that appears to very age-specific is gastroschisis. This defect displayed an increase between 2000 and 2006 but 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 (Figure 10).

Maternal Race / Hispanic Ethnicity

The overall prevalence of birth defects varied by maternal race and Hispanic ethnicity. The age-adjusted birth defect prevalence per 10,000 live births was 174.2 for non-Hispanic whites, 203.5 for non-Hispanic blacks, 143.4 for non-

Hispanic Asians/Pacific Islanders and 180.6 for Hispanics (Table 12). 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), polydactyly/syndactyly, and obstructive genitourinary defect amongst the most common defects. Down syndrome was among the most commonly occurring defects in all but non-Hispanic whites. Asians had lower age-adjusted rates of chromosomal defects and had significantly lower rates than any of the other racial/ethnic groups and, though the Hispanic rate was not significantly different from non-Hispanic-white or –black rates, the non-Hispanic black defect prevalence rate was significantly higher than that for the non-Hispanic whites. Figure 15 shows the trends in birth defect prevalence rates from 2000 to the most current data year. Asian women born in the U.S. had slightly higher, though not statistically significantly higher, a birth defect rate than those born outside the U.S (160.7 vs. 134.2 per 10,000 live births) Given that a much higher percentage of Asian women in Massachusetts (85.2%) were born outside the U.S. compared to other race/ethnicities, maternal birthplace may be a contributing factor in group differences—particular among Asian women. Conversely, though over 50% of non-Hispanic black women were born outside the U.S. the birth defect prevalence rates did not differ nearly as much as Asian women (205.3 vs. 203.2 per 10,000 live births respectively for those born inside and outside the U.S.)

Region

The birth defect rates among six Massachusetts regions in 200 8-2010 were not statistically significantly different. The age-adjusted rates ranged from 165.53 per 10,000 live births in the Metro West region to 182.3 per 10,000 in the Boston region.

Assisted Reproductive Technology (ART)

In the last review of ART use in the U.S. in 2012, Massachusetts had the highest proportion of U.S. births conceived through assisted reproductive technology (ART) procedures among state residents (4.3% of MA births compared to 1.4% of births nationally). 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. 2012). 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).

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 allows for the collection of relevant etiology (cause) information. Cases with known etiology accounted for about 18.5% of the birth defects in Massachusetts in 2008-2010. Of the 743 cases with known cause, "single gene" etiology accounted for 21.5%, "chromosomal" etiology accounted for 72.8% and "maternal-fetal factors" accounted for 3.4% of cases. The vast majority of birth defects cases in Massachusetts in 2008-2010 (81.4% or 3,266 or 4,012 cases) had an unknown etiology.

Cases are also classified by the *pattern* of defects (i.e. whether one defect occurs with others). Of all 4,012 birth defect cases (3,931 live births and 81 stillbirths) 43.5% had a "solitary" (truly a single) defect pattern, 26.4% had "Major plus minors" (defined as having a major defect accompanied by one or more minor defects), 4.3% were a "sequence" (allowing for more than one major defect if the defects are related pathogenically), and 25.6% had "multiple major" defects. Cleft lip with and without cleft palate, hypospadias (2nd or 3rd degree), gastroschisis, and craniosynostosis appeared overwhelmingly as single defects rather than with other defects—all occurring more that 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.

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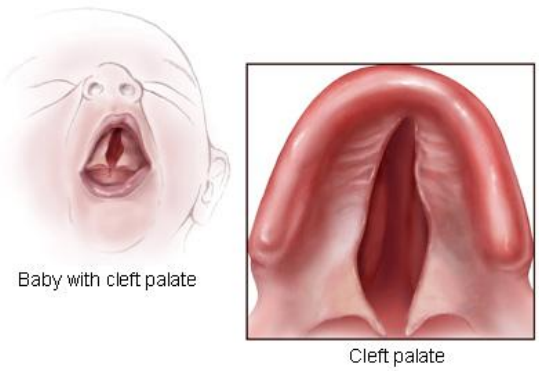
Chapter 1

Introduction



Courtesy of the Centers for Disease Control and Prevention

The Public Health Importance of Birth Defects

Each year in the United States, approximately 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 "periconceptional" period, the month before and three months after conception. For the developing pregnancy, "the environment" includes any exposure to the fetus as well as the mother.

Birth Defects and Folic Acid Awareness and Behavior

Studies have shown that the presence of adequate amounts of folic acid (vitamin B9) in the mother's system during the "periconceptional" period may help prevent defects of the brain and spinal cord known as neural tube defects. Mandatory fortification of cereal grains with folic acid has resulted in a 26% reduction in the

number of babies born with these neural tube defects 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 2020 Challenges

Healthy People 2020 has established the objectives of reducing the infant death related to any birth defect and specifically to congenital heart defects by 10%, spina bifida and anencephaly rates by 10%, and increase the 1-year survival rates of children born with Down syndrome by 10% (DHHS 2020). 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.

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 ($439,059 in 2013 dollars).

Adjusting for inflation, the estimated combined lifetime cost in Massachusetts for babies born with 12 major structural birth defects was $151,322,760 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.

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.

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 throu gh age three. It also extend ed 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 s uch 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.

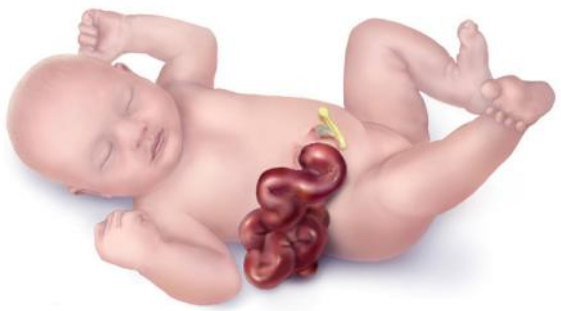
The 2008-2010 Surveillance Report
This report presents statewide data on the prevalence of birth defects in live births and stillbirths in Massachusetts duri ng the years 2008, 2009, and 2010. The data are presented in combined form since the numbers are relatively small for individual defects. The first annual report presented Massachusetts data for birth defects for the year 1999. Since that time we have produced reports every two years that reflect the nature of birth defects for a corresponding two -year period. Our ability to find and identify infants born with birth defects to Massachusetts

residents has improved over time. The prevalence increases from the 2000-2001 report through this current version are at least partially attributable to continually improved case ascertainment. Because there were major changes to the Massachusetts birth certificate in 2011 that are delaying its release, the decision was made to produce this interim, 3-year report rather than wait to produce a two-year report as has been the practice. Once the new certificate has been released we will generate a regular two-year report for 2010-2011. As a result, the prevalence of all birth defects for this report (178.5 per 10,000 live births) is very similar to that the previous 2008-2009 report (175.3 per 10,000 live births). Interpretations of the report 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 2010 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 2008-2010 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 2010. Due to the small numbers of birth defects relative to the number of births and the fact that the BDMP data ascertainment is constantly being refined, conclusions from these results—including inferences about multi-year trends—are not valid until a extensive multi-year estimates establish 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.

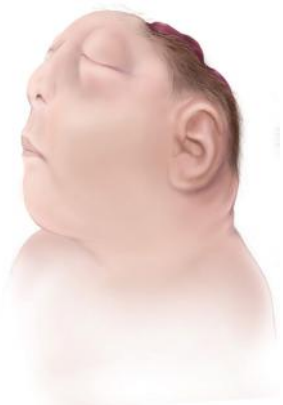
8. There are limitations when comparing the Massachusetts BDMP data to data from other states and national estimates. Factors such as differences in population demographics, living environment, and variances in surveillance/case ascertainment methods may contribute to differences in the prevalence of 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 for the report period 2008-2010 while Figure 1 shows the overall trend in birth defects from 2000 to 2010. Among births to Massachusetts residents in 2008-2010, 4,012 had one or more structural birth defects that were ascertained by Massachusetts BDMP. Among these, 81 stillbirths were identified with a birth defect. Overall, 1.8% of births in the state (178.5 per 10,000 live births) were identified as having at least one birth defect. The majority of defects occurred in the cardiovascular (occurring in 33.2% of defect cases) and musculoskeletal (occurring in 28.5%) 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 2008-2010 were significantly lower than the US rates for approximately 45% of the defects and were about the same as the national estimates for the other 55% (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.

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 2008-2010 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 2008-2010 rates, an estimated 50-72% of anencephaly cases are missed through exclusion of terminated cases. Upon adjusting the Massachusetts rate of for these four defects and assessing overlapping confidence intervals (see Appendix for description of confidence intervals), Massachusetts rates of anencephaly appear significantly lower than the national estimate for all three estimates while Trisomy 21 appears significantly higher than the national estimate. Both spina bifida and Trisomy 18 rates, when adjusted, do not appear to be significantly dissimilar from the U.S. estimate. Table 3 lists the adjusted rates. The age-adjusted rate of Down syndrome in Massachusetts that is higher than the national estimate possibly reflects the higher birth rate for mothers over the age of 35 compared to the national average (CDC 2012). The lower anencephaly rate may the result of a very small number of actual cases having a disproportionate effect. Between 2008 and 2010 Massachusetts averaged just over 74,000 births and fewer than five cases per year.

Figure 3 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* (as does Table 15). Cases are classified by the pattern of defects (i.e. whether one defect occurs with others). Of all 4,012 birth defect cases (3,931 live births and 81 stillbirths), 43.6% had a “solitary” (truly a single) defect pattern, 26.5% had “Major plus minors” (defined as having a major defect accompanied by one or more minor defects), 4.3% were a “sequence” (allowing for more than one major defect if the defects are related pathogenically), and 25.6% 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 4 compares selected pregnancy outcome characteristics among infants born with birth defects to those born without birth defects in 2008-2010 by percentage. Of infants born with birth defects, 46.7% were delivered by Cesarean, compared to 33.4% of infants born with no birth defect births; 21.3% of infants with birth defects had low birthweight (<2,500 grams) as opposed to 7.5% of those without a birth defect; 20.5% of infants were premature (gestational age < 37 weeks), compared with 8.5% of those without a birth defect; 4.8% of infants with a birth defect died before their 1st birthday, compared to 0.4% of those without a birth defect. While numbers of infants with birth defects are relatively small, it is important to recognize the impact of these outcomes when diagnosing and treating a baby with a birth defect.

Prevalence of Birth Defects by Sex

Table 6 presents the prevalence of birth defects by sex of the infant/fetus. The overall prevalence was 146.8 for females and 207.3 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), obstructive genitourinary defect, polydactyly/syndactyly, 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 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. 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 2008-2010 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 2010, 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 in women under 20, but smoking (possibly the duration of smoking) increased the risk in older women (Werler, Mitchell et al 2009). The rate of gastroschisis in Massachusetts 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. As mentioned above, the rate of gastroschisis since 2007 seems to remain unchanged. Alternatively, the rate of hypospadias—the most common defect among male births and one that overwhelmingly appears as a solitary defect—appears to have seen an increase in the last few years. This increase may be a result of new reporting originating from outpatient facilities (see Chapter 2: Methods). Before this additional source for ascertaining defects was implemented, all hypospadias reports were the result of the identification of a surgical procedure only. Given that this increase in a relatively new finding, it will be important to focus on whether this is simply a result of better ascertainment or a genuine increase in the rates of this defect.

Table 1 Prevalence of Birth Defects, Massachusetts: 2008 -2010

Defect[1]	Live birth Count	Stillbirth Count	Total Count	Rate per 10,000 Births	95% Confidence Interval
Total Cases	3931	81	4012	178.49	173.06-184.05
Central Nervous System					
Anencephaly	9	4	13	0.58	0.31-0.99
Encephalocele	5	0	5	0.22	0.07-0.52
Holoprosencephaly	14	1	15	0.67	0.37-1.10
Hydrocephaly w/o Spina Bifida	71	8	79	3.51	2.78-4.38
Microcephaly	35	0	35	1.56	1.08-2.17
Spina Bifida w/ and w/o Hydrocephaly	51	3	54	2.40	1.80-3.13
Spinal Cord	87	0	87	3.87	3.10-4.77
Other CNS[2]	165	3	168	7.47	6.39-8.69
Eye					
Aniridia	3	0	3	0.13	0.03-0.39
Anophthalmia/Microphthalmia	32	2	34	1.51	1.05-2.11
Congenital Glaucoma, Congenital Cataract	67	0	67	2.98	2.31-3.79
Other Eye[2]	69	0	69	3.07	2.39-3.89
Ear					
Anotia/Microtia	39	0	39	1.74	1.23-2.37
Other Ear[2]	73	1	74	3.29	2.59-4.13
Cardiovascular					
Anomalous Pulmonary Venous Connection					
Total/Partial Anomalous Pulm Venous Connection	38	0	38	1.69	1.20-2.32
Atrioventricular Canal Defects					
ASD Primum	5	0	5	0.22	0.07-0.52
Common Atrium	12	0	12	0.53	0.28-0.93
Complete Atrioventricular Canal Defect	65	2	67	2.98	2.31-3.79
Endocardial Cushion (OS and NOS)	35	0	35	1.56	1.08-2.17
VSD, Canal Type	7	0	7	0.31	0.13-0.64
Conotruncal (Outlet) and Aortic Arch					
Double Outlet Right Ventricle	24	0	24	1.07	0.68-1.59
Interrupted Aortic Arch, Type B	8	0	8	0.36	0.15-0.70
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	89	1	90	4.00	3.22-4.92
Truncus	6	0	6	0.27	0.10-0.58

Table 1 Prevalence of Birth Defects, Massachusetts: 2008 -2010

Defect[1]	Live birth Count	Stillbirth Count	Total Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)					
d-Transposition of the Great Arteries	54	1	55	2.45	1.84-3.19
Ebstein Anomaly					
Ebstein Anomaly	8	0	8	0.36	0.15-0.70
Heterotaxy (Laterality Defects)					
Heterotaxy	36	0	36	1.60	1.12-2.22
Left-Sided Obstruction					
Aortic Valve Stenosis	31	0	31	1.38	0.94-1.96
Coarctation of Aorta	96	1	97	4.32	3.50-5.26
Hypoplastic Left Heart Syndrome	29	2	31	1.38	0.94-1.96
Interrupted Aortic Arch (Type A and NOS)	1	0	1	0.04	0.00-0.25
Patent Ductus Arteriosus					
Patent Ductus Arteriosus	307	0	307	13.66	12.17-15.27
Right-Sided Obstruction					
Pulmonary Stenosis, Valvular	156	0	156	6.94	5.89-8.12
Pulmonary Valve Atresia w/intact septum	13	1	14	0.62	0.34-1.05
Pulmonary Valve Atresia with VSD	6	0	6	0.27	0.10-0.58
Tricuspid Valve Atresia	17	0	17	0.76	0.44-1.21
Septal Defects					
ASD (Secundum and NOS)	495	0	495	22.02	20.13-24.05
VSD (Membranous and NOS)	263	11	274	12.19	10.79-13.72
VSD, Conoventricular/Malalignment	38	2	40	1.78	1.27-2.42
Single Ventricle and L-TGA					
L-TGA	7	0	7	0.31	0.13-0.64
Single Ventricle	9	0	9	0.40	0.18-0.76
Other Cardiovascular					
Other Cardiovascular[2]	473	11	484	21.53	19.66-23.54
Respiratory					
Choanal Atresia	16	0	16	0.71	0.41-1.16
Lung Anomalies	43	0	43	1.91	1.38-2.58
Other Respiratory[2]	26	3	29	1.29	0.86-1.85
Orofacial					
Cleft Lip w/ and w/o Cleft Palate	178	4	182	8.10	6.96-9.36

Table 1 Prevalence of Birth Defects, Massachusetts: 2008 -2010

Defect[1]	Live birth Count	Stillbirth Count	Total Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)					
Cleft Palate w/o Cleft Lip	122	2	124	5.52	4.59-6.58
Pierre Robin Sequence	43	0	43	1.91	1.38-2.58
Other Orofacial[2]	115	2	117	5.21	4.30-6.24
Gastrointestinal					
Biliary Atresia	14	0	14	0.62	0.34-1.05
Esophageal Atresia/Tracheoesophageal Fistula	51	0	51	2.27	1.69-2.98
Hirschsprung Disease	52	0	52	2.31	1.73-3.03
Rectal and Large Intestinal Atresia/Stenosis	68	1	69	3.07	2.39-3.89
Small Intestinal Atresia	64	2	66	2.94	2.27-3.74
Other Gastrointestinal	160	3	163	7.25	6.18-8.45
Genitourinary					
Bladder Exstrophy	4	0	4	0.18	0.05-0.46
Cloacal Exstrophy	2	0	2	0.09	0.01-0.32
Hypospadias, 2nd or 3rd Degree[3]	309	0	309	13.75	12.26-15.37
Obstructive Genitourinary Defect	344	4	348	15.48	13.90-17.20
Renal Agenesis/Hypoplasia	8	3	11	0.49	0.24-0.88
Other Genitourinary[2]	395	7	402	17.88	16.18-19.72
Musculoskeletal					
Club Foot	316	10	326	14.50	12.97-16.17
Craniosynostosis	109	1	110	4.89	4.02-5.90
Diaphragmatic Hernia	55	1	56	2.49	1.88-3.24
Gastroschisis	73	3	76	3.38	2.66-4.23
Omphalocele	24	2	26	1.16	0.76-1.69
Polydactyly/Syndactyly	353	6	359	15.97	14.36-17.71
Reduction Deformity, Lower Limbs	27	2	29	1.29	0.86-1.85
Reduction Deformity, Upper Limbs	53	3	56	2.49	1.88-3.24
Skeletal Dysplasia	29	1	30	1.33	0.90-1.91
Other Musculoskeletal[2]	254	7	261	11.61	10.25-13.11
Chromosomal and other Syndromes					
Klinefelter Syndrome	8	0	8	0.36	0.15-0.70
Trisomy 13	6	4	10	0.44	0.21-0.82
Trisomy 18	27	11	38	1.69	1.20-2.32

Table 1 Prevalence of Birth Defects, Massachusetts: 2008 -2010

Defect[1]	Live birth Count	Stillbirth Count	Total Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)					
Trisomy 21 (Down Syndrome)	277	12	289	12.86	11.42-14.43
Turner Syndrome	15	7	22	0.98	0.61-1.48
Other Chromosomal Syndromes/Other Syndromes [2]	306	4	310	13.79	12.30-15.41
Other					
Amniotic Bands	17	4	21	0.93	0.58-1.43
Skin Anomalies	38	0	38	1.69	1.20-2.32
Other, Specified[2]	29	0	29	1.29	0.86-1.85

[1.] Cases can be included in the count for more than one defect. Cases are counted once in the tota l for a defect category.
[2] Rate may represent a heterogeneous group of defects.
[3] Rate calculated using male live births.

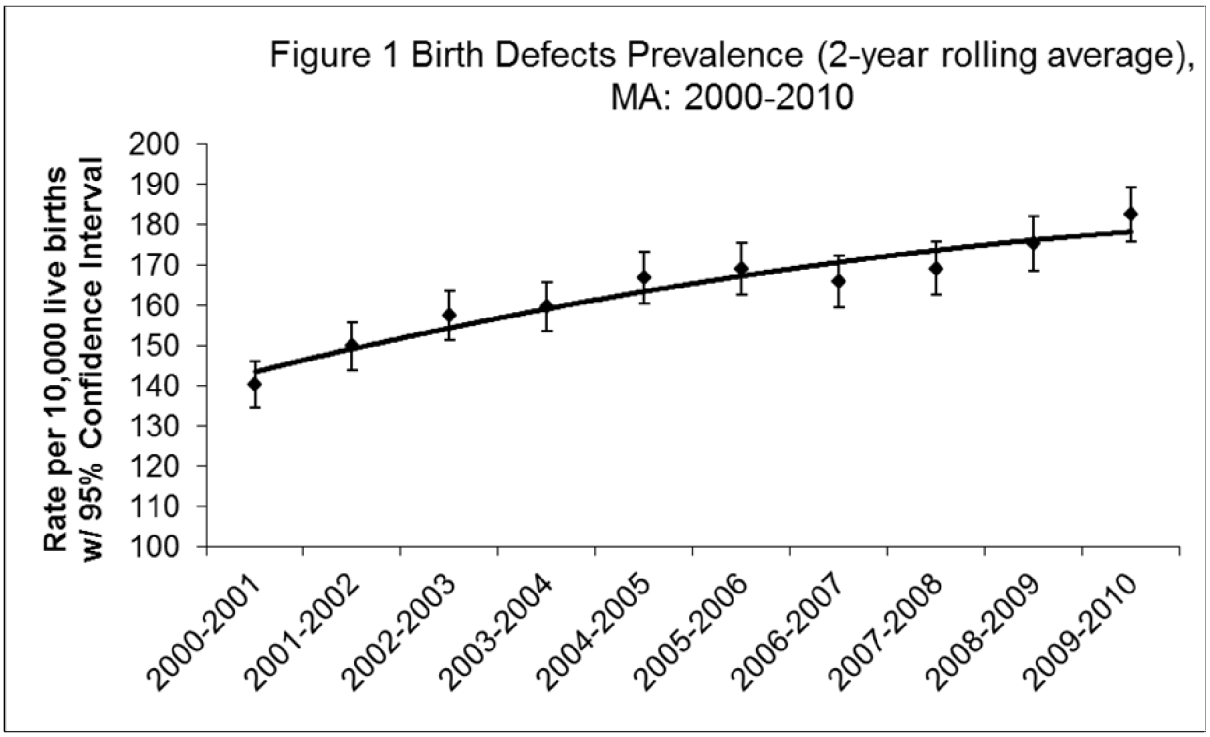


Table 2 Comparison of Selected Massachusetts Birth Defect Rates to National Estimates, 2008-2010

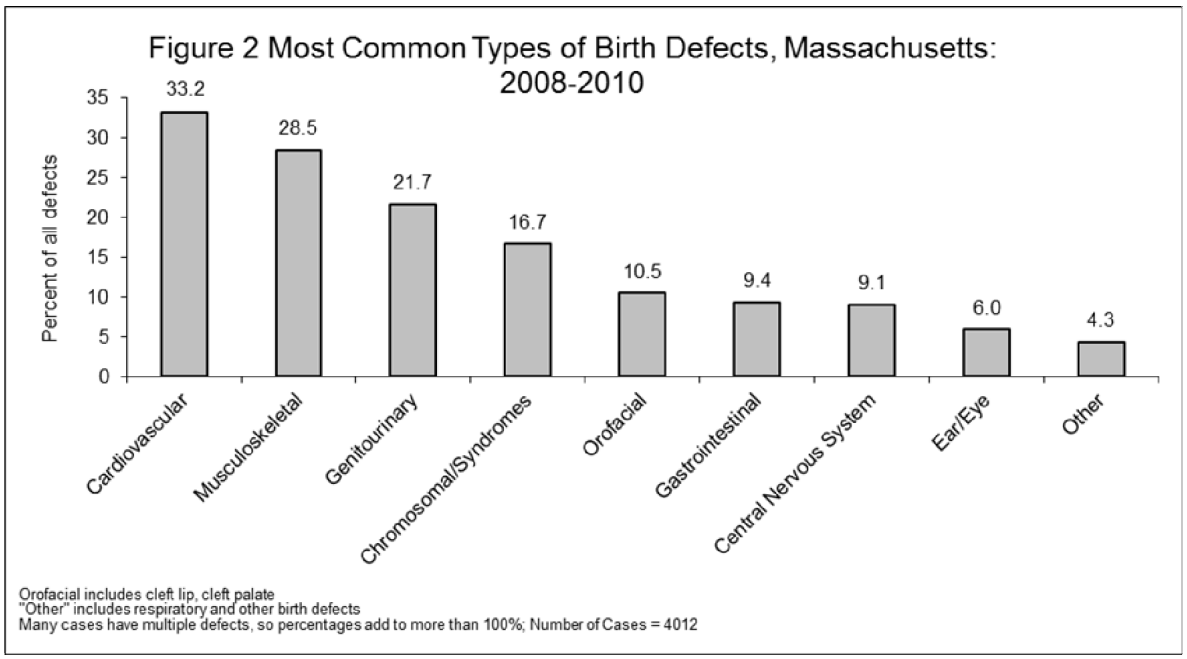
Defect	Count	Rate per 10,000 Births MA[1]	95% Confidence Interval	Rate per 10,000 Births US[2]	95% Confidence Interval
Anencephaly	13	0.58	0.31-0.99	2.23	2.07 - 2.41
Spina bifida	54	2.40	1.80-3.13	3.72	3.52 - 3.94
Anophthalmia/microphthalmia	34	1.51	1.05-2.11	2.10	1.94 - 2.27
Truncus arteriosus (common truncus)	6	0.27	0.10-0.58	0.74	0.65 - 0.84
Transposition of the great arteries[3]	62	2.76	2.11-3.54	3.04	2.85 - 3.24
Tetralogy of Fallot	90	4.00	3.22-4.92	4.05	3.83 - 4.28
Atrioventricular canal defect [4]	126	5.61	4.67-6.67	4.70	4.45 - 4.96
Hypoplastic left heart syndrome	31	1.38	0.94-1.96	2.31	2.14 - 2.48
Cleft palate without cleft lip	124	5.52	4.59-6.58	6.45	6.17 – 6.74
Cleft lip with and without cleft palate	182	8.10	6.96-9.36	10.89	10.53 - 11.26
Esophageal atresia/tracheoesophageal fistula	51	2.27	1.69-2.98	2.12	1.96 - 2.29
Rectal and large intestinal atresia/stenosis	69	3.07	2.39-3.89	4.86	4.61 - 5.14
Reduction deformity, upper limbs	56	2.49	1.88-3.24	3.64	3.43 - 3.86
Reduction deformity, lower limbs	29	1.29	0.86-1.85	1.65	1.51 - 1.80
Gastroschisis	76	3.38	2.66-4.23	4.72	4.49 - 4.97
Omphalocele	26	1.16	0.76-1.69	1.92	1.77 - 2.08
Diaphragmatic hernia	56	2.49	1.88-3.24	2.60	2.42 – 2.79
Trisomy 21 (Down syndrome)	289	12.86	11.42-14.43	13.48	13.08 - 13.90
Trisomy 13	10	0.44	0.21-0.82	1.20	1.09 - 1.33
Trisomy 18	38	1.69	1.20-2.32	2.55	2.38 - 2.73

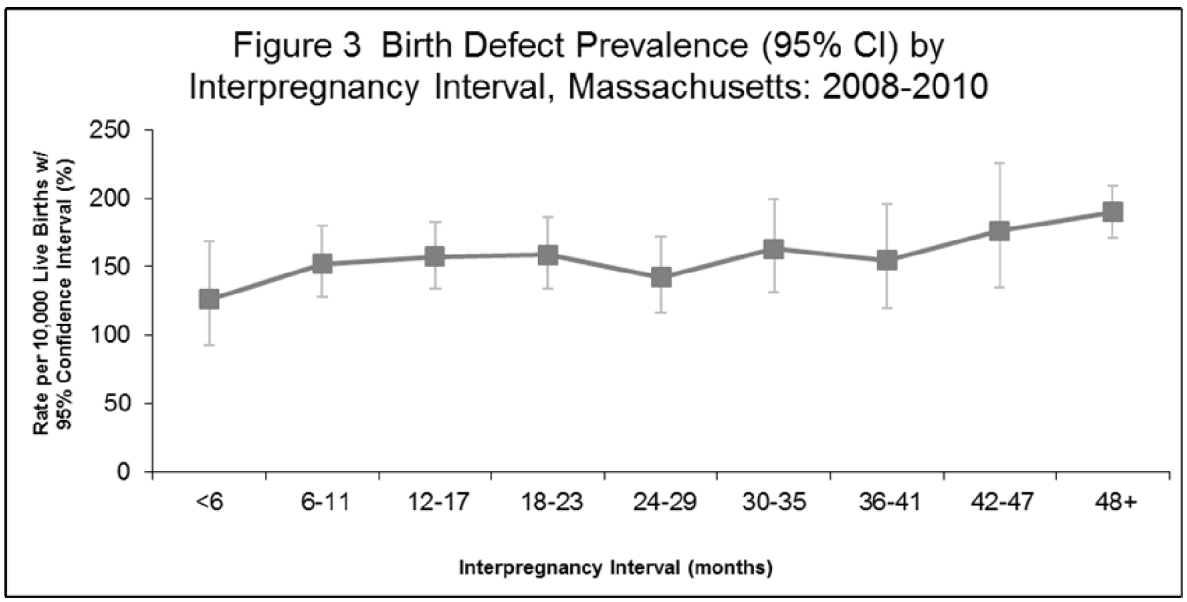
1. MA rate is based on live births and stillbirths.
2. Source: Updated National Birth Prevalence Estimates for Selected Birth Defects in the United States, 2004-2006. Birth Defects Research (Part A) **88**(12): 1008-1016. Only "active" surveillance system estimates are used and include systems that ascertain 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	0.58	0.31-0.99	66.7	Peller, et al.	0.98	0.61 - 1.48	2.23	2.07 – 2.41
			50.0	Cragan, et al.	0.89	0.54 - 1.37		
			72.0	Forrester, et al.	0.98	0.61 - 1.48		
Spina bifida	2.40	1.80-3.13	45.5	Peller, et al.	4.00	3.22 - 4.92	3.72	3.52 – 3.94
			29.0	Cragan, et al.	3.60	2.86 - 4.48		
			29.8	Forrester, et al.	4.14	3.34 - 5.07		
Trisomy 21 (Down syndrome)	12.86	11.42-14.43	35.2	Peller, et al.	21.44	19.57 - 23.44	13.48	13.08–13.90
			37.3	Forrester, et al.	22.11	20.21 - 24.14		
Trisomy 18	1.69	1.20-2.32	56.5	Peller, et al.	2.80	2.15 - 3.59	2.55	2.38 – 2.73
			49.0	Forrester, et al.	2.89	2.23 - 3.69		

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 p receding pregnancy.
Note: Prevalence estimates are among singleton births to multigravid women whose preceding pre gnancy resulted in a live birth.

Table 4 Most Common Defects among Live Births and Stillbirths, Massachusetts: 2008 – 2010

Defect	Category	Count	Rate per 10,000 Births	95% Confidence Interval
ASD (Secundum and NOS)	Cardiovascular	495	22.02	20.13-24.05
Polydactyly/Syndactyly	Musculoskeletal	359	15.97	14.36-17.71
Obstructive Genitourinary Defect	Genitourinary	348	15.48	13.90-17.20
Club Foot	Musculoskeletal	326	14.50	12.97-16.17
Hypospadias, 2nd or 3rd Degree	Genitourinary	309	13.75	12.26-15.37
Trisomy 21 (Down Syndrome)	Chromosomal and other Syndromes	289	12.86	11.42-14.43
VSD (Membranous and NOS)	Cardiovascular	274	12.19	10.79-13.72
Cleft Lip w/ and w/o Cleft Palate	Orofacial	182	8.10	6.96-9.36
Pulmonary Stenosis, Valvular	Cardiovascular	156	6.94	5.89-8.12
Cleft Palate w/o Cleft Lip	Orofacial	124	5.52	4.59-6.58

Hypospadias rate calculated using only male live births

Table 5 Counts of Birth Defects by Pattern among Live Births and Stillbirths, Massachusetts: 2008-2010

Defect[1]	Isolated Defects			Multiple Major Defects[3]	Total Cases
	Solitary	Major + Minor(s)[2]	Sequence		
Total Cases	1746	1061	174	1028	4009
Central Nervous System					
Anencephaly	10	2	0	1	13
Encephalocele	1	2	0	2	5
Holoprosencephaly	2	0	4	9	15
Hydrocephaly w/o Spina Bifida	26	14	2	37	79
Microcephaly	4	5	1	25	35
Spina Bifida w/ and w/o Hydrocephaly	1	3	42	8	54
Spinal Cord	10	15	29	33	87
Other CNS	31	49	15	73	168
Eye					
Aniridia	0	0	0	3	3
Anophthalmia/Microphthalmia	4	8	1	21	34
Congenital Glaucoma, Congenital Cataract	39	9	1	18	67
Other Eye	12	19	1	37	69
Ear					
Anotia/Microtia	7	12	2	18	39
Other Ear	14	13	0	47	74
Cardiovascular					
Anomalous Pulmonary Venous Connection					
Total/Partial Anomalous Pulm Venous Conn	4	19	9	6	38
Atrioventricular Canal Defects					
ASD Primum	0	2	1	2	5
Common Atrium	0	0	10	2	12
Complete Atrioventricular Canal Defect	1	6	9	51	67
Endocardial Cushion (OS and NOS)	0	9	4	22	35
VSD, Canal Type	1	2	0	4	7
Conotruncal (Outlet) and Aortic Arch					
Double Outlet Right Ventricle	0	11	5	8	24
Interrupted Aortic Arch, Type B	1	2	0	5	8
Tetralogy of Fallot w/ and w/o Pulm Atresia	17	34	2	37	90

Table 5 Counts of Birth Defects by Pattern among Live Births and Stillbirths, Massachusetts: 2008-2010

Defect[1]	Isolated Defects			Multiple Major Defects[3]	Total Cases
	Solitary	Major + Minor(s)[2]	Sequence		
(cont'd)					
Truncus	1	2	0	3	6
d-Transposition of the Great Arteries	21	29	2	3	55
Ebstein Anomaly					
Ebstein Anomaly	3	4	0	1	8
Heterotaxy (Laterality Defects)					
Heterotaxy	1	0	31	4	36
Left-Sided Obstruction					
Aortic Valve Stenosis	4	19	2	6	31
Coarctation of Aorta	10	57	2	28	97
Hypoplastic Left Heart Syndrome	5	17	1	8	31
Interrupted Aortic Arch (Type A and NOS)	0	1	0	0	1
Patent Ductus Arteriosus					
Patent Ductus Arteriosus	22	104	4	177	307
Right-Sided Obstruction					
Pulmonary Stenosis, Valvular	50	78	4	24	156
Pulmonary Valve Atresia w/intact septum	4	7	1	2	14
Pulmonary Valve Atresia with VSD	0	3	3	0	6
Tricuspid Valve Atresia	1	12	1	3	17
Other Cardiovascular					
Other Cardiovascular	29	228	32	195	484
Septal Defects					
ASD (Secundum and NOS)	85	203	12	195	495
VSD (Membranous and NOS)	37	134	2	101	274
VSD, Conoventricular/Malalignment	6	19	1	14	40
Single Ventricle and L-TGA					
L-TGA	0	5	1	1	7
Single Ventricle	1	5	3	0	9
Respiratory					
Choanal Atresia	6	2	0	8	16
Lung Anomalies	32	4	1	6	43
Other Respiratory	6	2	4	17	29

Table 5 Counts of Birth Defects by Pattern among Live Births and Stillbirths, Massachusetts: 2008-2010

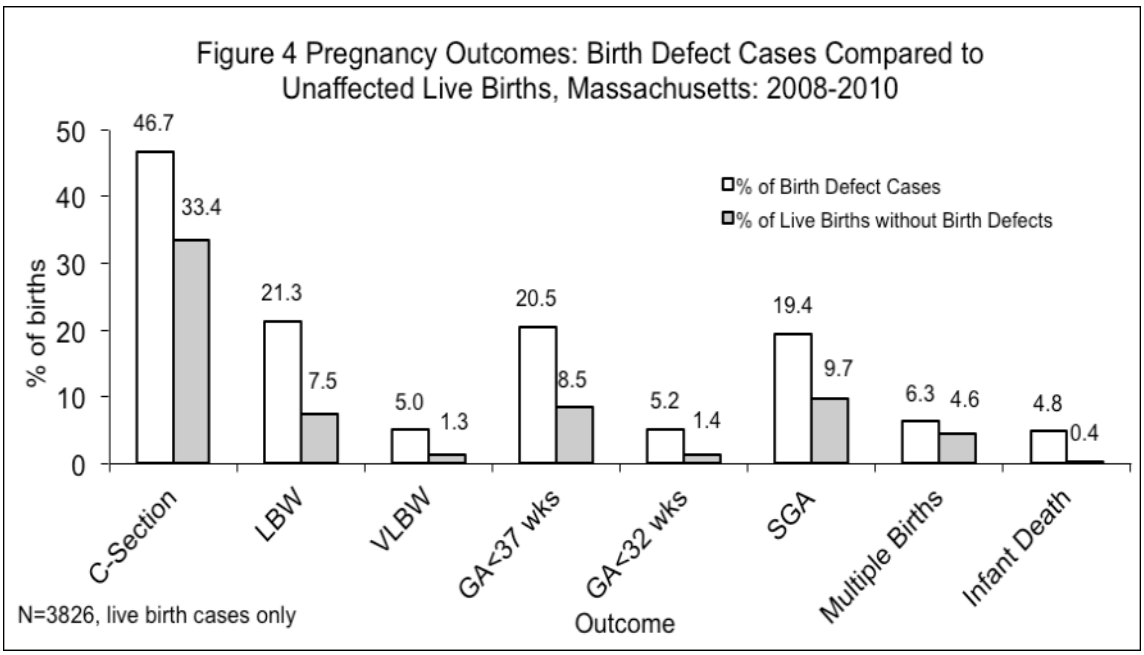
Defect[1]	Isolated Defects			Multiple Major Defects[3]	Total Cases
	Solitary	Major + Minor(s)[2]	Sequence		
Orofacial					
Cleft Lip w/ and w/o Cleft Palate	130	13	4	35	182
Cleft Palate w/o Cleft Lip	51	18	28	27	124
Pierre Robin Sequence	4	3	29	7	43
Other Orofacial	57	12	4	44	117
Gastrointestinal					
Biliary Atresia	8	1	2	3	14
Esophageal Atresia/Tracheoesophageal Fistula	13	4	0	34	51
Hirschsprung Disease	39	4	0	9	52
Rectal and Large Intestinal Atresia/Stenosis	24	7	1	37	69
Small Intestinal Atresia	22	19	1	24	66
Other Gastrointestinal	60	45	17	41	163
Genitourinary					
Bladder Exstrophy	3	1	0	0	4
Cloacal Exstrophy	0	0	1	1	2
Hypospadias, 2nd or 3rd Degree	246	24	1	38	309
Obstructive Genitourinary Defect	11	178	17	142	348
Renal Agenesis/Hypoplasia	1	1	8	1	11
Other Genitourinary	99	148	14	141	402
Musculoskeletal					
Club Foot	174	37	28	87	326
Craniosynostosis	81	6	0	23	110
Diaphragmatic Hernia	24	5	3	24	56
Gastroschisis	60	9	1	6	76
Omphalocele	8	4	0	14	26
Polydactyly/Syndactyly	159	105	9	86	359
Reduction Deformity, Lower Limbs	3	9	4	13	29
Reduction Deformity, Upper Limbs	14	10	11	21	56
Skeletal Dysplasia	0	0	0	30	30
Other Musculoskeletal	22	29	41	169	261

Table 5 Counts of Birth Defects by Pattern among Live Births and Stillbirths, Massachusetts: 2008-2010

Defect[1]	Isolated Defects			Multiple Major Defects[3]	Total Cases
	Solitary	Major + Minor(s)[2]	Sequence		
Chromosomal and other Syndromes					
Klinefelter Syndrome	0	0	0	8	8
Trisomy 13	0	0	0	10	10
Trisomy 18	0	0	0	38	38
Trisomy 21 (Down Syndrome)	0	0	0	289	289
Turner Syndrome	0	0	0	22	22
Other Chrom Syndromes/Other Syndromes	4	8	8	290	310
Other					
Amniotic Bands	0	1	17	3	21
Skin Anomalies	6	1	1	30	38
Other, Specified	2	6	14	7	29

1. Cases can be included in more than one defect. Cases are counted once in the total for a defect category. (3 unk pattern)
2. Major + Minor(s) includes Additive pattern.
3. Multiple major includes all recognized syndromes .
4. Counts may reflect a heterogeneous group of defects.

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



LBW: low birth weight (<2500gm); VLBW: very low birth weight (<1500gm); 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: 2008-2010

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
Total Cases	Male	2690	207.3	199.2-215.7
	Female	1607	146.8	139.7-154.1
Central Nervous System				
Anencephaly	Male	10	0.87	0.42-1.60
	Female	1	0.09	0.00-0.51
Encephalocele	Male	0	0.00	0.00-0.32
	Female	5	0.46	0.15-1.07
Holoprosencephaly	Male	6	0.52	0.19-1.13
	Female	8	0.73	0.32-1.44
Hydrocephaly w/o Spina Bifida	Male	40	3.47	2.48-4.73
	Female	38	3.47	2.46-4.76
Microcephaly	Male	17	1.47	0.86-2.36
	Female	18	1.64	0.97-2.60
Spina Bifida w/ and w/o Hydrocephaly	Male	28	2.43	1.61-3.51
	Female	24	2.19	1.40-3.26
Spinal Cord	Male	49	4.25	3.14-5.62
	Female	38	3.47	2.46-4.76
Other CNS[2]	Male	81	7.03	5.58-8.73
	Female	86	7.85	6.28-9.70
Eye				
Aniridia	Male	2	0.17	0.02-0.63
	Female	1	0.09	0.00-0.51
Anophthalmia/Microphthalmia	Male	17	1.47	0.86-2.36
	Female	17	1.55	0.90-2.49
Congenital Glaucoma, Congenital Cataract	Male	34	2.95	2.04-4.12
	Female	33	3.01	2.07-4.23
Other Eye[2]	Male	34	2.95	2.04-4.12
	Female	35	3.20	2.23-4.45
Ear				
Anotia/Microtia	Male	24	2.08	1.33-3.10
	Female	15	1.37	0.77-2.26

Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2008-2010

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Other Ear[2]	Male	42	3.64	2.63-4.93
	Female	32	2.92	2.00-4.13
Cardiovascular				
Anomalous Pulmonary Venous Connection				
Total/Partial Anomalous Pulmonary Venous Connection	Male	23	2.00	1.26-2.99
	Female	15	1.37	0.77-2.26
Atrioventricular Canal Defects				
ASD Primum	Male	3	0.26	0.05-0.76
	Female	2	0.18	0.02-0.66
Common Atrium	Male	2	0.17	0.02-0.63
	Female	10	0.91	0.44-1.68
Complete Atrioventricular Canal Defect	Male	31	2.69	1.83-3.82
	Female	35	3.20	2.23-4.45
Endocardial Cushion (OS and NOS)	Male	13	1.13	0.60-1.93
	Female	22	2.01	1.26-3.04
VSD, Canal Type	Male	5	0.43	0.14-1.01
	Female	2	0.18	0.02-0.66
Conotruncal (Outlet) and Aortic Arch				
Double Outlet Right Ventricle	Male	15	1.30	0.73-2.15
	Female	9	0.82	0.38-1.56
Interrupted Aortic Arch, Type B	Male	2	0.17	0.02-0.63
	Female	6	0.55	0.20-1.19
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	Male	56	4.86	3.67-6.31
	Female	34	3.11	2.15-4.34
Truncus	Male	2	0.17	0.02-0.63
	Female	4	0.37	0.10-0.94
d-Transposition of the Great Arteries	Male	37	3.21	2.26-4.42
	Female	18	1.64	0.97-2.60
Ebstein Anomaly				
Ebstein Anomaly	Male	4	0.35	0.09-0.89
	Female	4	0.37	0.10-0.94

Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2008-2010

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
Heterotaxy (Laterality Defects)				
Heterotaxy	Male	19	1.65	0.99-2.57
	Female	17	1.55	0.90-2.49
Left-Sided Obstruction				
Aortic Valve Stenosis	Male	18	1.56	0.93-2.47
	Female	13	1.19	0.63-2.03
Coarctation of Aorta	Male	51	4.42	3.29-5.82
	Female	46	4.20	3.08-5.60
Hypoplastic Left Heart Syndrome	Male	19	1.65	0.99-2.57
	Female	12	1.10	0.57-1.91
Interrupted Aortic Arch (Type A and NOS)	Male	0	0.00	0.00-0.32
	Female	1	0.09	0.00-0.51
Patent Ductus Arteriosus				
Patent Ductus Arteriosus	Male	157	13.62	11.57-15.93
	Female	150	13.70	11.59-16.07
Right-Sided Obstruction				
Pulmonary Stenosis, Valvular	Male	70	6.07	4.73-7.67
	Female	86	7.85	6.28-9.70
Pulmonary Valve Atresia w/intact septum	Male	7	0.61	0.24-1.25
	Female	6	0.55	0.20-1.19
Pulmonary Valve Atresia with VSD	Male	5	0.43	0.14-1.01
	Female	1	0.09	0.00-0.51
Tricuspid Valve Atresia	Male	11	0.95	0.48-1.71
	Female	6	0.55	0.20-1.19
Septal Defects				
ASD (Secundum and NOS)	Male	231	20.04	17.54-22.80
	Female	264	24.11	21.29-27.20
VSD (Membranous and NOS)	Male	132	11.45	9.58-13.58
	Female	142	12.97	10.92-15.29
VSD, Conoventricular/Malalignment	Male	16	1.39	0.79-2.25
	Female	22	2.01	1.26-3.04

Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2008-2010

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
Single Ventricle and L-TGA				
L-TGA	Male	5	0.43	0.14-1.01
	Female	2	0.18	0.02-0.66
Single Ventricle	Male	7	0.61	0.24-1.25
	Female	2	0.18	0.02-0.66
Other Cardiovascular				
Other Cardiovascular[2]	Male	258	22.38	19.73-25.29
	Female	222	20.27	17.69-23.12
Respiratory				
Choanal Atresia	Male	4	0.35	0.09-0.89
	Female	12	1.10	0.57-1.91
Lung Anomalies	Male	23	2.00	1.26-2.99
	Female	20	1.83	1.12-2.82
Other Respiratory[2]	Male	11	0.95	0.48-1.71
	Female	17	1.55	0.90-2.49
Orofacial				
Cleft Lip w/ and w/o Cleft Palate	Male	120	10.41	8.63-12.45
	Female	61	5.57	4.26-7.16
Cleft Palate w/o Cleft Lip	Male	41	3.56	2.55-4.83
	Female	82	7.49	5.96-9.30
Pierre Robin Sequence	Male	17	1.47	0.86-2.36
	Female	26	2.37	1.55-3.48
Other Orofacial[2]	Male	64	5.55	4.28-7.09
	Female	53	4.84	3.63-6.33
Gastrointestinal				
Biliary Atresia	Male	6	0.52	0.19-1.13
	Female	8	0.73	0.32-1.44
Esophageal Atresia/Tracheoesophageal Fistula	Male	26	2.26	1.47-3.30
	Female	25	2.28	1.48-3.37
Hirschsprung Disease	Male	39	3.38	2.41-4.63
	Female	13	1.19	0.63-2.03

Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2008-2010

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Rectal and Large Intestinal Atresia/Stenosis	Male	39	3.38	2.41-4.63
	Female	30	2.74	1.85-3.91
Small Intestinal Atresia	Male	36	3.12	2.19-4.32
	Female	28	2.56	1.70-3.70
Other Gastrointestinal[2]	Male	94	8.15	6.59-9.98
	Female	69	6.30	4.90-7.97
Genitourinary				
Bladder Exstrophy	Male	3	0.26	0.05-0.76
	Female	1	0.09	0.00-0.51
Cloacal Exstrophy	Male	0	0.00	0.00-0.32
	Female	2	0.18	0.02-0.66
Hypospadias, 2nd or 3rd Degree	Male	309	26.81	23.90-29.96
	Female	0	0.00	0.00-0.34
Obstructive Genitourinary Defect	Male	230	19.95	17.46-22.71
	Female	118	10.78	8.92-12.91
Renal Agenesis/Hypoplasia	Male	7	0.61	0.24-1.25
	Female	4	0.37	0.10-0.94
Other Genitourinary[2]	Male	260	22.56	19.90-25.47
	Female	141	12.88	10.84-15.19
Musculoskeletal				
Club Foot	Male	213	18.48	16.08-21.13
	Female	110	10.05	8.26-12.11
Craniosynostosis	Male	71	6.16	4.81-7.77
	Female	39	3.56	2.53-4.87
Diaphragmatic Hernia	Male	36	3.12	2.19-4.32
	Female	18	1.64	0.97-2.60
Gastroschisis	Male	33	2.86	1.97-4.02
	Female	43	3.93	2.84-5.29
Omphalocele	Male	20	1.74	1.06-2.68
	Female	6	0.55	0.20-1.19

Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2008-2010

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Polydactyly/Syndactyly	Male	221	19.17	16.73-21.87
	Female	138	12.60	10.59-14.89
Reduction Deformity, Lower Limbs	Male	17	1.47	0.86-2.36
	Female	11	1.00	0.50-1.80
Reduction Deformity, Upper Limbs	Male	28	2.43	1.61-3.51
	Female	27	2.47	1.62-3.59
Skeletal Dysplasia	Male	16	1.39	0.79-2.25
	Female	14	1.28	0.70-2.15
Other Musculoskeletal[2]	Male	144	12.49	10.54-14.71
	Female	115	10.50	8.67-12.61
Chromosomal and other Syndromes				
Klinefelter Syndrome	Male	8	0.69	0.30-1.37
	Female	0	0.00	0.00-0.34
Trisomy 13	Male	3	0.26	0.05-0.76
	Female	6	0.55	0.20-1.19
Trisomy 18	Male	17	1.47	0.86-2.36
	Female	19	1.74	1.04-2.71
Trisomy 21 (Down Syndrome)	Male	163	14.14	12.05-16.49
	Female	120	10.96	9.09-13.10
Turner Syndrome	Male	0	0.00	0.00-0.32
	Female	18	1.64	0.97-2.60
Other Chromosomal Syndromes/Other Syndromes[2]	Male	153	13.27	11.25-15.55
	Female	155	14.16	12.01-16.57

Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2008-2010

Defect[1]	Sex	Count	Rate per 10,000 Births	95% Confidence Interval
Other				
Amniotic Bands	Male	9	0.78	0.36-1.48
	Female	10	0.91	0.44-1.68
Skin Anomalies	Male	18	1.56	0.93-2.47
	Female	20	1.83	1.12-2.82
Other, Specified[2]	Male	15	1.30	0.73-2.15
	Female	14	1.28	0.70-2.15

[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: 2008-2010

Defect	Count	Rate per 10,000 Births	95% Confidence Interval
FEMALE			
ASD (Secundum and NOS)	264	24.11	21.29-27.20
VSD (Membranous and NOS)	142	12.97	10.92-15.29
Polydactyly/Syndactyly	138	12.60	10.59-14.89
Trisomy 21 (Down Syndrome)	120	10.96	9.09-13.10
Obstructive Genitourinary Defect	118	10.78	8.92-12.91
Club Foot	110	10.05	8.26-12.11
Pulmonary Stenosis, Valvular	86	7.85	6.28-9.70
Cleft Palate w/o Cleft Lip	82	7.49	5.96-9.30
Cleft Lip w/ and w/o Cleft Palate	61	5.57	4.26-7.16
Coarctation of Aorta	46	4.20	3.08-5.60
MALE			
Hypospadias, 2nd or 3rd Degree	309	26.81	23.90-29.96
ASD (Secundum and NOS)	231	20.04	17.54-22.80
Obstructive Genitourinary Defect	230	19.95	17.46-22.71
Polydactyly/Syndactyly	221	19.17	16.73-21.87
Club Foot	213	18.48	16.08-21.13
Trisomy 21 (Down Syndrome)	163	14.14	12.05-16.49
VSD (Membranous and NOS)	132	11.45	9.58-13.58
Cleft Lip w/ and w/o Cleft Palate	120	10.41	8.63-12.45
Craniosynostosis	71	6.16	4.81-7.77
Pulmonary Stenosis, Valvular	70	6.07	4.73-7.67

Hypospadias rate calculated using male live births.

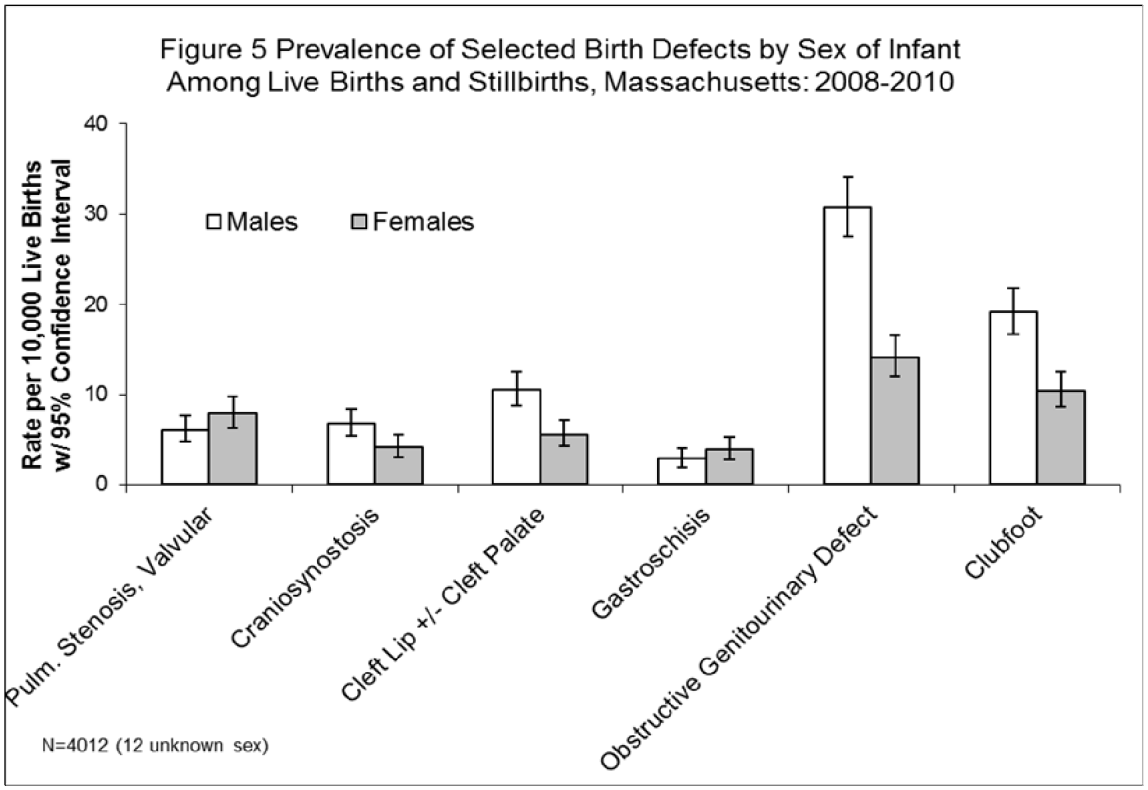


Figure 5 Prevalence of Selected Birth Defects by Sex of Infant
Among Live Births and Stillbirths, Massachusetts: 2008-2010
Rate per 10,000 Live Births
w/ 95% Confidence Interval
40
30
20
10
0
Males
Females
Pulm. Stenosis, Valvular
Craniosynostosis
Cleft Lip +/- Cleft Palate
Gastroschisis
Obstructive Genitourinary Defect
Clubfoot
N=4012 (12 unknown sex)

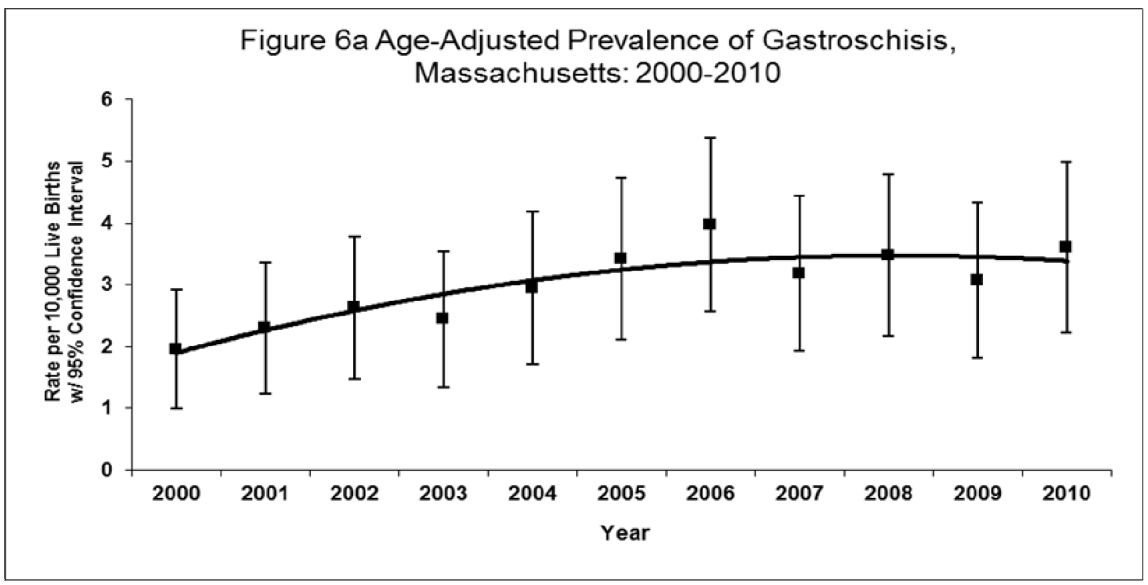


Figure 6a Age-Adjusted Prevalence of Gastroschisis,
Massachusetts: 2000-2010
Rate per 10,000 Live Births
w/ 95% Confidence Interval
6
5
4
3
2
1
0
2000
2001
2002
2003
2004
2005
2006
2007
2008
2009
2010
Year

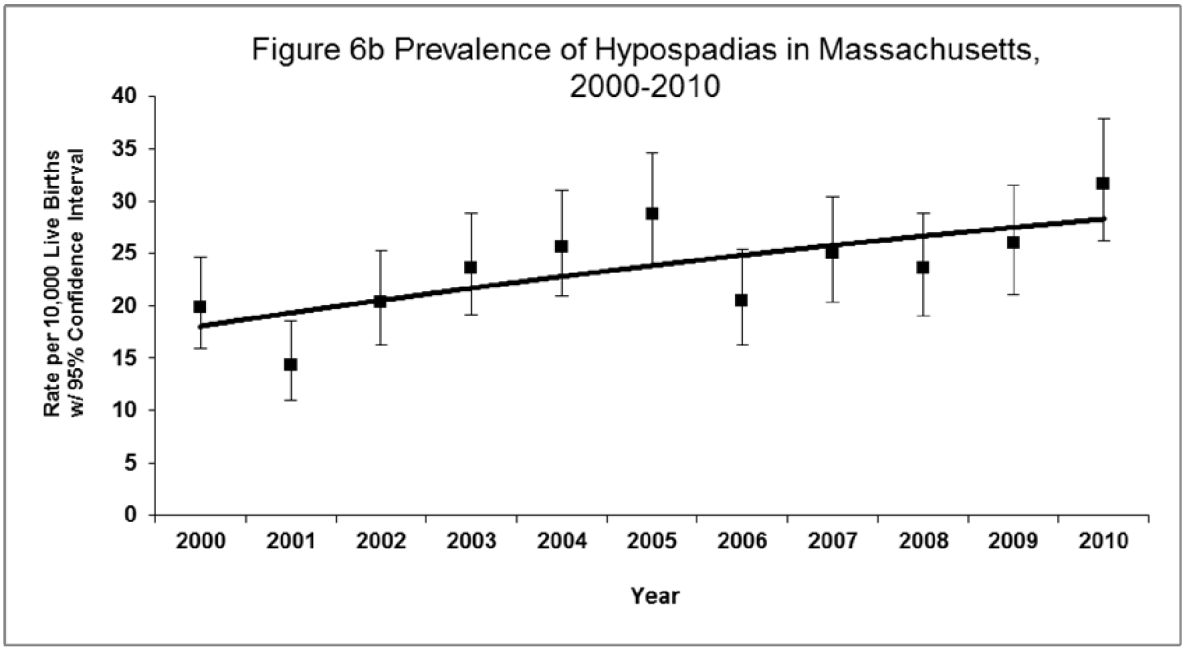


Figure 6b Prevalence of Hypospadias in Massachusetts, 2000-2010
Rate per 10,000 Live Births w/ 95% Confidence Interval
40
35
30
25
20
15
10
5
0
2000
2001
2002
2003
2004
2005
2006
2007
2008
2009
2010
Year

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 173.83 for singletons and 275.00 for multiple births (more than one infant) per 10,000 live births. While multiple births comprised under 5% of all live births, they comprised over 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. Defect rates among live born infants per 10,000 live births were 178.67 for mothers younger than 20 years, 177.00 for those 20-24 years, 167.11 for those 25-29 years, 157.66 for those 30-34 years and 203.52 for those 35 years and older.

As expected, there was a strong association between Down syndrome and advanced maternal age (see Figure 9). The Down syndrome rate of 31.1 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 Down syndrome rate as maternal age increases. 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 and primarily accounts for most of the difference between that age group and the others.

Figure 11 shows that younger mothers (aged 19 and under) had the highest rate of gastroschisis cases at 14.5 per 10,000 live births. This association has been shown in previous studies (Forrester and Merz 1999). The youngest mothers had infants with higher rates of gastroschisis and reduction deformities of the upper limb than other age groups. Older mothers had higher rates for other 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 three 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. The percentage of live births delivered to women ages 30+ has increased steadily since the 1980s to the point where the percent of total births delivered to women 30 years and older surpassed the percent of births to women below age 30 in 1996. More recent data suggest that the percent of births to women ages 30+ peaked around 2004 and has decreased only slightly since. There has been a marked change in the age distribution of Massachusetts women giving birth since 1980. Approximately 25% of women giving birth in 1980 were 30 years and older compared with 54% in 2010. Additionally, the proportion of multiple births in mothers over 35 years was twice that of mothers less than 35 years of age in 2010 (MADPH 2013).

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, 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 12 shows the disparity in percent of births that are single versus multiple from ART births and non-ART births in Massachusetts (CDC 2009). The vast majority of non-ART births are single deliveries, whereas almost half of ART births are multiple deliveries. ART poses many risks associated with multiple births that may lead to adverse maternal and infant outcomes such as low birthweight and preterm delivery. ART has also been associated with some birth defects such as septal heart defects and cleft lip with or without cleft palate (Reefhuis, Honein et al. 2009).

Table 8 Prevalence of Birth Defects by Plurality [1] of Live Births and Stillbirths, Massachusetts: 2008 -2010

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
Total Cases	Singleton	3727	173.8	168.3-179.5
	Multiple	285	275.0	244.0-308.8
Central Nervous System				
Anencephaly	Singleton	11	0.51	0.26-0.92
	Multiple	2	1.93	0.23-6.97
Encephalocele	Singleton	5	0.23	0.08-0.54
	Multiple	0	0.00	0.00-3.56
Holoprosencephaly	Singleton	14	0.65	0.36-1.10
	Multiple	1	0.96	0.02-5.38
Hydrocephaly w/o Spina Bifida	Singleton	73	3.40	2.67-4.28
	Multiple	6	5.79	2.12-12.60
Microcephaly	Singleton	34	1.59	1.10-2.22
	Multiple	1	0.96	0.02-5.38
Spina Bifida w/ and w/o Hydrocephaly	Singleton	48	2.24	1.65-2.97
	Multiple	6	5.79	2.12-12.60
Spinal Cord	Singleton	85	3.96	3.17-4.90
	Multiple	2	1.93	0.23-6.97
Other CNS[3]	Singleton	158	7.37	6.26-8.61
	Multiple	10	9.65	4.63-17.74
Eye				
Aniridia	Singleton	3	0.14	0.03-0.41
	Multiple	0	0.00	0.00-3.56
Anophthalmia/Microphthalmia	Singleton	33	1.54	1.06-2.16
	Multiple	1	0.96	0.02-5.38
Congenital Glaucoma, Congenital Cataract	Singleton	65	3.03	2.34-3.86
	Multiple	2	1.93	0.23-6.97
Other Eye[3]	Singleton	67	3.12	2.42-3.97
	Multiple	2	1.93	0.23-6.97
Ear				
Anotia/Microtia	Singleton	38	1.77	1.25-2.43
	Multiple	1	0.96	0.02-5.38

Table 8 Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2008-2010

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Other Ear[3]	Singleton	73	3.40	2.67-4.28
	Multiple	1	0.96	0.02-5.38
Cardiovascular				
Anomalous Pulmonary Venous Connection				
Total/Partial Anomalous Pulmonary Venous Connection	Singleton	38	1.77	1.25-2.43
	Multiple	0	0.00	0.00-3.56
Atrioventricular Canal Defects				
ASD Primum	Singleton	5	0.23	0.08-0.54
	Multiple	0	0.00	0.00-3.56
Common Atrium	Singleton	10	0.47	0.22-0.86
	Multiple	2	1.93	0.23-6.97
Complete Atrioventricular Canal Defect	Singleton	66	3.08	2.38-3.92
	Multiple	1	0.96	0.02-5.38
Endocardial Cushion (OS and NOS)	Singleton	33	1.54	1.06-2.16
	Multiple	2	1.93	0.23-6.97
VSD, Canal Type	Singleton	7	0.33	0.13-0.67
	Multiple	0	0.00	0.00-3.56
Conotruncal (Outlet) and Aortic Arch				
Double Outlet Right Ventricle	Singleton	23	1.07	0.68-1.61
	Multiple	1	0.96	0.02-5.38
Interrupted Aortic Arch, Type B	Singleton	8	0.37	0.16-0.74
	Multiple	0	0.00	0.00-3.56
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	Singleton	81	3.78	3.00-4.70
	Multiple	9	8.68	3.97-16.48
Truncus	Singleton	5	0.23	0.08-0.54
	Multiple	1	0.96	0.02-5.38
d-Transposition of the Great Arteries	Singleton	53	2.47	1.85-3.23
	Multiple	2	1.93	0.23-6.97
Ebstein Anomaly				
Ebstein Anomaly	Singleton	8	0.37	0.16-0.74
	Multiple	0	0.00	0.00-3.56

Table 8 Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2008-2010

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
Heterotaxy (Laterality Defects)				
Heterotaxy	Singleton	33	1.54	1.06-2.16
	Multiple	3	2.89	0.60-8.46
Left-Sided Obstruction				
Aortic Valve Stenosis	Singleton	29	1.35	0.91-1.94
	Multiple	2	1.93	0.23-6.97
Coarctation of Aorta	Singleton	89	4.15	3.33-5.11
	Multiple	8	7.72	3.33-15.21
Hypoplastic Left Heart Syndrome	Singleton	28	1.31	0.87-1.89
	Multiple	3	2.89	0.60-8.46
Interrupted Aortic Arch (Type A and NOS)	Singleton	1	0.05	0.00-0.26
	Multiple	0	0.00	0.00-3.56
Patent Ductus Arteriosus				
Patent Ductus Arteriosus	Singleton	296	13.81	12.28-15.47
	Multiple	11	10.61	5.30-18.99
Right-Sided Obstruction				
Pulmonary Stenosis, Valvular	Singleton	143	6.67	5.62-7.86
	Multiple	13	12.54	6.68-21.45
Pulmonary Valve Atresia w/intact septum	Singleton	14	0.65	0.36-1.10
	Multiple	0	0.00	0.00-3.56
Pulmonary Valve Atresia with VSD	Singleton	6	0.28	0.10-0.61
	Multiple	0	0.00	0.00-3.56
Tricuspid Valve Atresia	Singleton	17	0.79	0.46-1.27
	Multiple	0	0.00	0.00-3.56
Septal Defects				
ASD (Secundum and NOS)	Singleton	454	21.17	19.27-23.21
	Multiple	41	39.56	28.39-53.67
VSD (Membranous and NOS)	Singleton	246	11.47	10.08-13.00
	Multiple	28	27.02	17.95-39.05
VSD, Conoventricular/Malalignment	Singleton	34	1.59	1.10-2.22
	Multiple	6	5.79	2.12-12.60

Table 8 Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2008-2010

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
Single Ventricle and L-TGA				
L-TGA	Singleton	7	0.33	0.13-0.67
	Multiple	0	0.00	0.00-3.56
Single Ventricle	Singleton	9	0.42	0.19-0.80
	Multiple	0	0.00	0.00-3.56
Other Cardiovascular				
Other Cardiovascular[3]	Singleton	454	21.17	19.27-23.21
	Multiple	30	28.95	19.53-41.32
Respiratory				
Choanal Atresia	Singleton	13	0.61	0.32-1.04
	Multiple	3	2.89	0.60-8.46
Lung Anomalies	Singleton	40	1.87	1.33-2.54
	Multiple	3	2.89	0.60-8.46
Other Respiratory[3]	Singleton	27	1.26	0.83-1.83
	Multiple	2	1.93	0.23-6.97
Orofacial				
Cleft Lip w/ and w/o Cleft Palate	Singleton	168	7.84	6.70-9.11
	Multiple	14	13.51	7.39-22.66
Cleft Palate w/o Cleft Lip	Singleton	119	5.55	4.60-6.64
	Multiple	5	4.82	1.57-11.26
Pierre Robin Sequence	Singleton	42	1.96	1.41-2.65
	Multiple	1	0.96	0.02-5.38
Other Orofacial[3]	Singleton	108	5.04	4.13-6.08
	Multiple	9	8.68	3.97-16.48
Gastrointestinal				
Biliary Atresia	Singleton	13	0.61	0.32-1.04
	Multiple	1	0.96	0.02-5.38
Esophageal Atresia/Tracheoesophageal Fistula	Singleton	47	2.19	1.61-2.92
	Multiple	4	3.86	1.05-9.88
Hirschsprung Disease	Singleton	49	2.29	1.69-3.02
	Multiple	3	2.89	0.60-8.46

Table 8 Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2008-2010

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Rectal and Large Intestinal Atresia/Stenosis	Singleton	67	3.12	2.42-3.97
	Multiple	2	1.93	0.23-6.97
Small Intestinal Atresia	Singleton	63	2.94	2.26-3.76
	Multiple	3	2.89	0.60-8.46
Other Gastrointestinal[3]	Singleton	151	7.04	5.96-8.26
	Multiple	12	11.58	5.98-20.23
Genitourinary				
Bladder Exstrophy	Singleton	4	0.19	0.05-0.48
	Multiple	0	0.00	0.00-3.56
Cloacal Exstrophy	Singleton	2	0.09	0.01-0.34
	Multiple	0	0.00	0.00-3.56
Hypospadias, 2nd or 3rd Degree[4]	Singleton	282	25.73	11.66-14.78
	Multiple	27	51.31	17.17-37.90
Obstructive Genitourinary Defect	Singleton	332	15.48	13.86-17.24
	Multiple	16	15.44	8.82-25.07
Renal Agenesis/Hypoplasia	Singleton	10	0.47	0.22-0.86
	Multiple	1	0.96	0.02-5.38
Other Genitourinary[3]	Singleton	378	17.63	15.90-19.50
	Multiple	24	23.16	14.84-34.46
Musculoskeletal				
Club Foot	Singleton	308	14.37	12.81-16.06
	Multiple	18	17.37	10.29-27.45
Craniosynostosis	Singleton	104	4.85	3.96-5.88
	Multiple	6	5.79	2.12-12.60
Diaphragmatic Hernia	Singleton	48	2.24	1.65-2.97
	Multiple	8	7.72	3.33-15.21
Gastroschisis	Singleton	73	3.40	2.67-4.28
	Multiple	3	2.89	0.60-8.46
Omphalocele	Singleton	22	1.03	0.64-1.55
	Multiple	4	3.86	1.05-9.88

Table 8 Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2008-2010

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Polydactyly/Syndactyly	Singleton	343	16.00	14.35-17.78
	Multiple	16	15.44	8.82-25.07
Reduction Deformity, Lower Limbs	Singleton	27	1.26	0.83-1.83
	Multiple	2	1.93	0.23-6.97
Reduction Deformity, Upper Limbs	Singleton	54	2.52	1.89-3.29
	Multiple	2	1.93	0.23-6.97
Skeletal Dysplasia	Singleton	28	1.31	0.87-1.89
	Multiple	2	1.93	0.23-6.97
Other Musculoskeletal[3]	Singleton	233	10.87	9.52-12.36
	Multiple	28	27.02	17.95-39.05
Chromosomal and other Syndromes				
Klinefelter Syndrome	Singleton	7	0.33	0.13-0.67
	Multiple	1	0.96	0.02-5.38
Trisomy 13	Singleton	9	0.42	0.19-0.80
	Multiple	1	0.96	0.02-5.38
Trisomy 18	Singleton	35	1.63	1.14-2.27
	Multiple	3	2.89	0.60-8.46
Trisomy 21 (Down Syndrome)	Singleton	273	12.73	11.27-14.34
	Multiple	16	15.44	8.82-25.07
Turner Syndrome	Singleton	21	0.98	0.61-1.50
	Multiple	1	0.96	0.02-5.38
Other Chromosomal Syndromes/Other Syndromes[3]	Singleton	288	13.43	11.93-15.08
	Multiple	22	21.23	13.30-32.14

Table 8 Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2008-2010

Defect[2]	Plurality	Count	Rate per 10,000 Births	95% Confidence Interval
Other				
Amniotic Bands	Singleton	16	0.75	0.43-1.21
	Multiple	5	4.82	1.57-11.26
Skin Anomalies	Singleton	34	1.59	1.10-2.22
	Multiple	4	3.86	1.05-9.88
Other, Specified[3]	Singleton	27	1.26	0.83-1.83
	Multiple	2	1.93	0.23-6.97

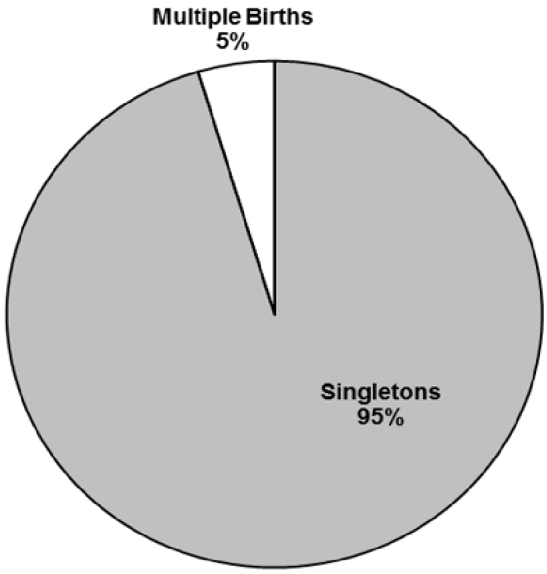
1. Plurality is the number of births to a woman from the same pregnancy. A singleton is the birth of one infant; multiple represents more than one infant.
2. Cases can be included in more than one defect. Due to missing plurality of infants, counts may not match those in other tables.
3. Rate may represent a heterogeneous group of defects.
4. Rate calculated using male live births.

Table 9 Most Common Defects by Plurality [1] of Live Births and Stillbirths, Massachusetts: 2008-2010

Defect[2]	Count	Rate per 10,000 Births	95% Confidence Interval
MULTIPLE			
ASD (Secundum and NOS)	41	39.56	28.39-53.67
VSD (Membranous and NOS)	28	27.02	17.95-39.05
Hypospadias, 2nd or 3rd Degree [3]	27	26.05	17.17-37.90
Club Foot	18	17.37	10.29-27.45
Obstructive Genitourinary Defect	16	15.44	8.82-25.07
Polydactyly/Syndactyly	16	15.44	8.82-25.07
Trisomy 21 (Down Syndrome)	16	15.44	8.82-25.07
Cleft Lip w/ and w/o Cleft Palate	14	13.51	7.39-22.66
Pulmonary Stenosis, Valvular	13	12.54	6.68-21.45
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	9	8.68	3.97-16.48
SINGLETON			
ASD (Secundum and NOS)	454	21.17	19.27-23.21
Polydactyly/Syndactyly	343	16.00	14.35-17.78
Obstructive Genitourinary Defect	332	15.48	13.86-17.24
Club Foot	308	14.37	12.81-16.06
Hypospadias, 2nd or 3rd Degree [3]	282	13.15	11.66-14.78
Trisomy 21 (Down Syndrome)	273	12.73	11.27-14.34
VSD (Membranous and NOS)	246	11.47	10.08-13.00
Cleft Lip w/ and w/o Cleft Palate	168	7.84	6.70-9.11
Pulmonary Stenosis, Valvular	143	6.67	5.62-7.86
Cleft Palate w/o Cleft Lip	119	5.55	4.60-6.64

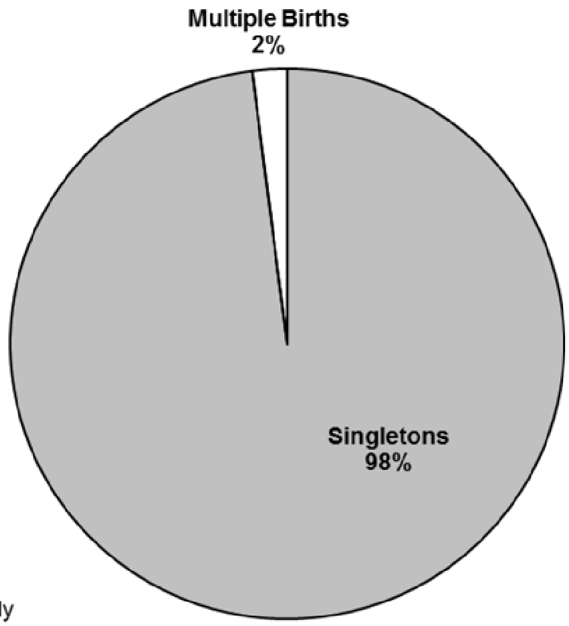
1. Plurality is the number of births to a woman from the same pregnancy. A singleton is the birth of one infant; multiple represents more than one infant.
2. Rate calculated using male live births.

Figure 7a Plurality of Live Births, Massachusetts: 2008-2010



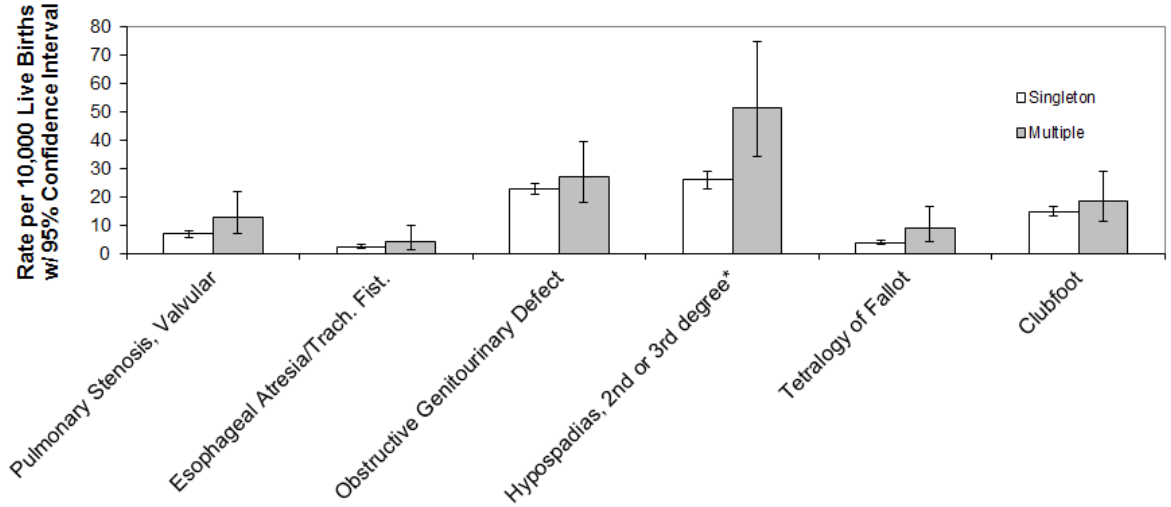
N=224,770

Figure 7b Plurality of Birth Defects Cases, Massachusetts: 2008-2010



N=2609, live birth cases only

Figure 8 Prevalence of Selected Birth Defects by Plurality among Live Births and Stillbirths, Massachusetts: 2008-2010



N=4012
*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: 2008-2010

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Total Cases	<20	234	178.7	156.5-203.1
	20-24	634	177.0	163.6-191.2
	25-29	928	167.1	156.6-178.1
	30-34	1095	157.7	148.5-167.2
	35+	1035	203.5	191.4-216.2
Central Nervous System				
Anencephaly	<20	1	0.76	0.02-4.25
	20-24	4	1.12	0.30-2.86
	25-29	3	0.54	0.11-1.58
	30-34	1	0.14	0.00-0.80
	35+	0	0.00	0.00-0.73
Encephalocele	<20	0	0.00	0.00-2.82
	20-24	1	0.28	0.01-1.56
	25-29	2	0.36	0.04-1.30
	30-34	1	0.14	0.00-0.80
	35+	1	0.20	0.00-1.10
Holoprosencephaly	<20	0	0.00	0.00-2.82
	20-24	5	1.40	0.45-3.26
	25-29	4	0.72	0.20-1.84
	30-34	3	0.43	0.09-1.26
	35+	2	0.39	0.05-1.42
Hydrocephaly w/o Spina Bifida	<20	10	7.64	3.66-14.04
	20-24	18	5.02	2.98-7.94
	25-29	13	2.34	1.25-4.00
	30-34	15	2.16	1.21-3.56
	35+	15	2.95	1.65-4.86
Microcephaly	<20	3	2.29	0.47-6.69
	20-24	8	2.23	0.96-4.40
	25-29	11	1.98	0.99-3.54
	30-34	8	1.15	0.50-2.27
	35+	5	0.98	0.32-2.29

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2010

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Spina Bifida w/ and w/o Hydrocephaly	<20	3	2.29	0.47-6.69
	20-24	8	2.23	0.96-4.40
	25-29	16	2.88	1.65-4.68
	30-34	12	1.73	0.89-3.02
	35+	11	2.16	1.08-3.87
Spinal Cord	<20	2	1.53	0.18-5.52
	20-24	10	2.79	1.34-5.13
	25-29	24	4.32	2.77-6.43
	30-34	30	4.32	2.91-6.17
	35+	21	4.13	2.56-6.31
Other CNS[2]	<20	21	16.03	9.93-24.51
	20-24	34	9.49	6.57-13.26
	25-29	37	6.66	4.69-9.18
	30-34	36	5.18	3.63-7.18
	35+	37	7.28	5.12-10.03
Eye				
Aniridia	<20	0	0.00	0.00-2.82
	20-24	0	0.00	0.00-1.03
	25-29	2	0.36	0.04-1.30
	30-34	0	0.00	0.00-0.53
	35+	1	0.20	0.00-1.10
Anophthalmia/Microphthalmia	<20	1	0.76	0.02-4.25
	20-24	3	0.84	0.17-2.45
	25-29	11	1.98	0.99-3.54
	30-34	8	1.15	0.50-2.27
	35+	9	1.77	0.81-3.36
Congenital Glaucoma, Congenital Cataract	<20	7	5.34	2.15-11.01
	20-24	16	4.47	2.55-7.25
	25-29	14	2.52	1.38-4.23
	30-34	16	2.30	1.32-3.74
	35+	14	2.75	1.51-4.62

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2010

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Other Eye[2]	<20	9	6.87	3.14-13.04
	20-24	16	4.47	2.55-7.25
	25-29	21	3.78	2.34-5.78
	30-34	12	1.73	0.89-3.02
	35+	11	2.16	1.08-3.87
Ear				
Anotia/Microtia	<20	2	1.53	0.18-5.52
	20-24	8	2.23	0.96-4.40
	25-29	13	2.34	1.25-4.00
	30-34	8	1.15	0.50-2.27
	35+	8	1.57	0.68-3.10
Other Ear[2]	<20	1	0.76	0.02-4.25
	20-24	17	4.75	2.76-7.60
	25-29	20	3.60	2.20-5.56
	30-34	18	2.59	1.54-4.10
	35+	17	3.34	1.95-5.35
Cardiovascular				
Anomalous Pulmonary Venous Connection				
Total/Partial Anomalous Pulmonary Venous Connection	<20	3	2.29	0.47-6.69
	20-24	3	0.84	0.17-2.45
	25-29	8	1.44	0.62-2.84
	30-34	13	1.87	1.00-3.20
	35+	11	2.16	1.08-3.87
Atrioventricular Canal Defects				
ASD Primum	<20	0	0.00	0.00-2.82
	20-24	0	0.00	0.00-1.03
	25-29	2	0.36	0.04-1.30
	30-34	1	0.14	0.00-0.80
	35+	2	0.39	0.05-1.42

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2010

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Common Atrium	<20	0	0.00	0.00-2.82
	20-24	1	0.28	0.01-1.56
	25-29	5	0.90	0.29-2.10
	30-34	4	0.58	0.16-1.47
	35+	2	0.39	0.05-1.42
Complete Atrioventricular Canal Defect	<20	2	1.53	0.18-5.52
	20-24	11	3.07	1.53-5.49
	25-29	12	2.16	1.12-3.77
	30-34	20	2.88	1.76-4.45
	35+	20	3.93	2.40-6.07
Endocardial Cushion (OS and NOS)	<20	1	0.76	0.02-4.25
	20-24	7	1.95	0.79-4.03
	25-29	4	0.72	0.20-1.84
	30-34	7	1.01	0.41-2.08
	35+	16	3.15	1.80-5.11
VSD, Canal Type	<20	0	0.00	0.00-2.82
	20-24	1	0.28	0.01-1.56
	25-29	0	0.00	0.00-0.66
	30-34	2	0.29	0.03-1.04
	35+	4	0.79	0.21-2.01
Conotruncal (Outlet) and Aortic Arch				
Double Outlet Right Ventricle	<20	0	0.00	0.00-2.82
	20-24	2	0.56	0.07-2.02
	25-29	7	1.26	0.51-2.60
	30-34	10	1.44	0.69-2.65
	35+	5	0.98	0.32-2.29
Interrupted Aortic Arch, Type B	<20	0	0.00	0.00-2.82
	20-24	1	0.28	0.01-1.56
	25-29	3	0.54	0.11-1.58
	30-34	3	0.43	0.09-1.26
	35+	1	0.20	0.00-1.10

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2010

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	<20	3	2.29	0.47-6.69
	20-24	20	5.58	3.41-8.62
	25-29	20	3.60	2.20-5.56
	30-34	23	3.31	2.10-4.97
	35+	23	4.52	2.87-6.79
Truncus	<20	0	0.00	0.00-2.82
	20-24	1	0.28	0.01-1.56
	25-29	2	0.36	0.04-1.30
	30-34	1	0.14	0.00-0.80
	35+	2	0.39	0.05-1.42
d-Transposition of the Great Arteries	<20	5	3.82	1.24-8.91
	20-24	9	2.51	1.15-4.77
	25-29	10	1.80	0.86-3.31
	30-34	14	2.02	1.10-3.38
	35+	16	3.15	1.80-5.11
Ebstein Anomaly				
Ebstein Anomaly	<20	0	0.00	0.00-2.82
	20-24	1	0.28	0.01-1.56
	25-29	2	0.36	0.04-1.30
	30-34	3	0.43	0.09-1.26
	35+	2	0.39	0.05-1.42
Heterotaxy (Laterality Defects)				
Heterotaxy	<20	3	2.29	0.47-6.69
	20-24	7	1.95	0.79-4.03
	25-29	9	1.62	0.74-3.08
	30-34	12	1.73	0.89-3.02
	35+	5	0.98	0.32-2.29

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2010

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Left-Sided Obstruction				
Aortic Valve Stenosis	<20	1	0.76	0.02-4.25
	20-24	1	0.28	0.01-1.56
	25-29	8	1.44	0.62-2.84
	30-34	9	1.30	0.59-2.46
	35+	12	2.36	1.22-4.12
Coarctation of Aorta	<20	6	4.58	1.68-9.97
	20-24	13	3.63	1.93-6.21
	25-29	24	4.32	2.77-6.43
	30-34	28	4.03	2.68-5.83
	35+	25	4.92	3.18-7.26
Hypoplastic Left Heart Syndrome	<20	4	3.05	0.83-7.82
	20-24	4	1.12	0.30-2.86
	25-29	6	1.08	0.40-2.35
	30-34	5	0.72	0.23-1.68
	35+	10	1.97	0.94-3.62
Interrupted Aortic Arch (Type A and NOS)	<20	0	0.00	0.00-2.82
	20-24	0	0.00	0.00-1.03
	25-29	1	0.18	0.00-1.00
	30-34	0	0.00	0.00-0.53
	35+	0	0.00	0.00-0.73
Patent Ductus Arteriosus				
Patent Ductus Arteriosus	<20	11	8.40	4.19-15.03
	20-24	41	11.45	8.21-15.53
	25-29	69	12.43	9.67-15.73
	30-34	83	11.95	9.52-14.81
	35+	103	20.25	16.53-24.56
Right-Sided Obstruction				
Pulmonary Stenosis, Valvular	<20	8	6.11	2.64-12.04
	20-24	23	6.42	4.07-9.63
	25-29	35	6.30	4.39-8.77
	30-34	55	7.92	5.97-10.31
	35+	35	6.88	4.79-9.57

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2010

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Pulmonary Valve Atresia w/intact septum	<20	2	1.53	0.18-5.52
	20-24	1	0.28	0.01-1.56
	25-29	2	0.36	0.04-1.30
	30-34	4	0.58	0.16-1.47
	35+	4	0.79	0.21-2.01
Pulmonary Valve Atresia with VSD	<20	1	0.76	0.02-4.25
	20-24	2	0.56	0.07-2.02
	25-29	0	0.00	0.00-0.66
	30-34	2	0.29	0.03-1.04
	35+	1	0.20	0.00-1.10
Tricuspid Valve Atresia	<20	1	0.76	0.02-4.25
	20-24	3	0.84	0.17-2.45
	25-29	2	0.36	0.04-1.30
	30-34	8	1.15	0.50-2.27
	35+	3	0.59	0.12-1.72
Septal Defects				
ASD (Secundum and NOS)	<20	22	16.80	10.53-25.43
	20-24	69	19.26	14.99-24.38
	25-29	106	19.09	15.63-23.09
	30-34	144	20.73	17.49-24.41
	35+	154	30.28	25.69-35.46
VSD (Membranous and NOS)	<20	17	12.98	7.56-20.78
	20-24	40	11.17	7.98-15.21
	25-29	56	10.08	7.62-13.10
	30-34	71	10.22	7.98-12.89
	35+	79	15.53	12.30-19.36
VSD, Conoventricular/Malalignment	<20	3	2.29	0.47-6.69
	20-24	4	1.12	0.30-2.86
	25-29	9	1.62	0.74-3.08
	30-34	9	1.30	0.59-2.46
	35+	13	2.56	1.36-4.37

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2010

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Single Ventricle and L-TGA				
L-TGA	<20	0	0.00	0.00-2.82
	20-24	0	0.00	0.00-1.03
	25-29	4	0.72	0.20-1.84
	30-34	1	0.14	0.00-0.80
	35+	2	0.39	0.05-1.42
Single Ventricle	<20	1	0.76	0.02-4.25
	20-24	1	0.28	0.01-1.56
	25-29	2	0.36	0.04-1.30
	30-34	3	0.43	0.09-1.26
	35+	2	0.39	0.05-1.42
Other Cardiovascular				
Other Cardiovascular[2]	<20	20	15.27	9.33-23.58
	20-24	65	18.15	14.00-23.13
	25-29	108	19.45	15.95-23.48
	30-34	138	19.87	16.69-23.47
	35+	142	27.92	23.52-32.91
Respiratory				
Lung Anomalies	<20	1	0.76	0.02-4.25
	20-24	3	0.84	0.17-2.45
	25-29	14	2.52	1.38-4.23
	30-34	15	2.16	1.21-3.56
	35+	10	1.97	0.94-3.62
Other Respiratory[2]	<20	0	0.00	0.00-2.82
	20-24	5	1.40	0.45-3.26
	25-29	11	1.98	0.99-3.54
	30-34	7	1.01	0.41-2.08
	35+	3	0.59	0.12-1.72

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2010

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
Orofacial				
Cleft Lip w/ and w/o Cleft Palate	<20	14	10.69	5.84-17.94
	20-24	29	8.10	5.42-11.63
	25-29	49	8.82	6.53-11.67
	30-34	49	7.06	5.22-9.33
	35+	37	7.28	5.12-10.03
Cleft Palate w/o Cleft Lip	<20	5	3.82	1.24-8.91
	20-24	22	6.14	3.85-9.30
	25-29	33	5.94	4.09-8.35
	30-34	36	5.18	3.63-7.18
	35+	26	5.11	3.34-7.49
Pierre Robin Sequence	<20	2	1.53	0.18-5.52
	20-24	8	2.23	0.96-4.40
	25-29	16	2.88	1.65-4.68
	30-34	11	1.58	0.79-2.83
	35+	6	1.18	0.43-2.57
Other Orofacial[2]	<20	3	2.29	0.47-6.69
	20-24	21	5.86	3.63-8.96
	25-29	28	5.04	3.35-7.29
	30-34	33	4.75	3.27-6.67
	35+	30	5.90	3.98-8.42
Gastrointestinal				
Biliary Atresia	<20	3	2.29	0.47-6.69
	20-24	1	0.28	0.01-1.56
	25-29	3	0.54	0.11-1.58
	30-34	5	0.72	0.23-1.68
	35+	2	0.39	0.05-1.42
Esophageal Atresia/Tracheoesophageal Fistula	<20	1	0.76	0.02-4.25
	20-24	6	1.67	0.61-3.65
	25-29	17	3.06	1.78-4.90
	30-34	8	1.15	0.50-2.27
	35+	19	3.74	2.25-5.83

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2010

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Hirschsprung Disease	<20	2	1.53	0.18-5.52
	20-24	10	2.79	1.34-5.13
	25-29	17	3.06	1.78-4.90
	30-34	18	2.59	1.54-4.10
	35+	5	0.98	0.32-2.29
Rectal and Large Intestinal Atresia/Stenosis	<20	2	1.53	0.18-5.52
	20-24	16	4.47	2.55-7.25
	25-29	16	2.88	1.65-4.68
	30-34	16	2.30	1.32-3.74
	35+	18	3.54	2.10-5.59
Small Intestinal Atresia	<20	5	3.82	1.24-8.91
	20-24	8	2.23	0.96-4.40
	25-29	16	2.88	1.65-4.68
	30-34	16	2.30	1.32-3.74
	35+	19	3.74	2.25-5.83
Other Gastrointestinal[2]	<20	16	12.22	6.98-19.84
	20-24	25	6.98	4.52-10.30
	25-29	42	7.56	5.45-10.22
	30-34	36	5.18	3.63-7.18
	35+	41	8.06	5.79-10.94
Genitourinary				
Bladder Exstrophy	<20	0	0.00	0.00-2.82
	20-24	1	0.28	0.01-1.56
	25-29	1	0.18	0.00-1.00
	30-34	2	0.29	0.03-1.04
	35+	0	0.00	0.00-0.73
Cloacal Exstrophy	<20	0	0.00	0.00-2.82
	20-24	0	0.00	0.00-1.03
	25-29	1	0.18	0.00-1.00
	30-34	1	0.14	0.00-0.80
	35+	0	0.00	0.00-0.73

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2010

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Hypospadias, 2nd or 3rd Degree[3]	<20	10	7.64	3.66-14.04
	20-24	40	11.17	7.98-15.21
	25-29	73	13.15	10.30-16.53
	30-34	107	15.41	12.63-18.62
	35+	79	15.53	12.30-19.36
Obstructive Genitourinary Defect	<20	30	22.91	15.45-32.70
	20-24	61	17.03	13.03-21.87
	25-29	77	13.87	10.94-17.33
	30-34	98	14.11	11.46-17.20
	35+	78	15.34	12.12-19.14
Renal Agenesis/Hypoplasia	<20	0	0.00	0.00-2.82
	20-24	2	0.56	0.07-2.02
	25-29	1	0.18	0.00-1.00
	30-34	4	0.58	0.16-1.47
	35+	1	0.20	0.00-1.10
Other Genitourinary[2]	<20	16	12.22	6.98-19.84
	20-24	61	17.03	13.03-21.87
	25-29	94	16.93	13.68-20.71
	30-34	112	16.13	13.28-19.40
	35+	112	22.02	18.13-26.50
Musculoskeletal				
Club Foot	<20	24	18.32	11.74-27.27
	20-24	68	18.98	14.74-24.07
	25-29	71	12.79	9.99-16.13
	30-34	92	13.25	10.68-16.25
	35+	61	11.99	9.17-15.41
Craniosynostosis	<20	2	1.53	0.18-5.52
	20-24	6	1.67	0.61-3.65
	25-29	26	4.68	3.06-6.86
	30-34	43	6.19	4.48-8.34
	35+	32	6.29	4.30-8.88

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2010

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Diaphragmatic Hernia	<20	5	3.82	1.24-8.91
	20-24	9	2.51	1.15-4.77
	25-29	10	1.80	0.86-3.31
	30-34	15	2.16	1.21-3.56
	35+	15	2.95	1.65-4.86
Gastroschisis	<20	19	14.51	8.73-22.65
	20-24	33	9.21	6.34-12.94
	25-29	17	3.06	1.78-4.90
	30-34	3	0.43	0.09-1.26
	35+	1	0.20	0.00-1.10
Omphalocele	<20	3	2.29	0.47-6.69
	20-24	8	2.23	0.96-4.40
	25-29	2	0.36	0.04-1.30
	30-34	4	0.58	0.16-1.47
	35+	7	1.38	0.55-2.84
Polydactyly/Syndactyly	<20	25	19.09	12.35-28.18
	20-24	61	17.03	13.03-21.87
	25-29	95	17.11	13.84-20.91
	30-34	101	14.54	11.84-17.67
	35+	71	13.96	10.90-17.61
Reduction Deformity, Lower Limbs	<20	2	1.53	0.18-5.52
	20-24	7	1.95	0.79-4.03
	25-29	4	0.72	0.20-1.84
	30-34	9	1.30	0.59-2.46
	35+	5	0.98	0.32-2.29
Reduction Deformity, Upper Limbs	<20	9	6.87	3.14-13.04
	20-24	13	3.63	1.93-6.21
	25-29	7	1.26	0.51-2.60
	30-34	17	2.45	1.43-3.92
	35+	7	1.38	0.55-2.84

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2010

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Skeletal Dysplasia	<20	0	0.00	0.00-2.82
	20-24	11	3.07	1.53-5.49
	25-29	7	1.26	0.51-2.60
	30-34	6	0.86	0.32-1.88
	35+	5	0.98	0.32-2.29
Other Musculoskeletal[2]	<20	21	16.03	9.93-24.51
	20-24	52	14.52	10.84-19.04
	25-29	57	10.26	7.77-13.30
	30-34	70	10.08	7.86-12.73
	35+	54	10.62	7.98-13.85
Chromosomal and other Syndromes				
Klinefelter Syndrome	<20	0	0.00	0.00-2.82
	20-24	0	0.00	0.00-1.03
	25-29	1	0.18	0.00-1.00
	30-34	1	0.14	0.00-0.80
	35+	6	1.18	0.43-2.57
Trisomy 13	<20	0	0.00	0.00-2.82
	20-24	2	0.56	0.07-2.02
	25-29	1	0.18	0.00-1.00
	30-34	1	0.14	0.00-0.80
	35+	2	0.39	0.05-1.42
Trisomy 18	<20	2	1.53	0.18-5.52
	20-24	3	0.84	0.17-2.45
	25-29	3	0.54	0.11-1.58
	30-34	5	0.72	0.23-1.68
	35+	14	2.75	1.51-4.62
Trisomy 21 (Down Syndrome)	<20	10	7.64	3.66-14.04
	20-24	30	8.37	5.65-11.96
	25-29	32	5.76	3.94-8.14
	30-34	55	7.92	5.97-10.31
	35+	150	29.50	24.96-34.61

Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2010

Defect[1]	Maternal Age	Count	Rate per 10,000 Births	95% Confidence Interval
(cont'd)				
Turner Syndrome	<20	3	2.29	0.47-6.69
	20-24	4	1.12	0.30-2.86
	25-29	4	0.72	0.20-1.84
	30-34	0	0.00	0.00-0.53
	35+	4	0.79	0.21-2.01
Other Chromosomal Syndromes/Other Syndromes[2]	<20	15	11.45	6.41-18.89
	20-24	37	10.33	7.27-14.24
	25-29	69	12.43	9.67-15.73
	30-34	91	13.10	10.55-16.09
	35+	94	18.48	14.94-22.62
Other				
Amniotic Bands	<20	3	2.29	0.47-6.69
	20-24	4	1.12	0.30-2.86
	25-29	1	0.18	0.00-1.00
	30-34	4	0.58	0.16-1.47
	35+	5	0.98	0.32-2.29
Skin Anomalies	<20	2	1.53	0.18-5.52
	20-24	8	2.23	0.96-4.40
	25-29	6	1.08	0.40-2.35
	30-34	13	1.87	1.00-3.20
	35+	9	1.77	0.81-3.36
Other, Specified[2]	<20	2	1.53	0.18-5.52
	20-24	3	0.84	0.17-2.45
	25-29	8	1.44	0.62-2.84
	30-34	8	1.15	0.50-2.27
	35+	8	1.57	0.68-3.10

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

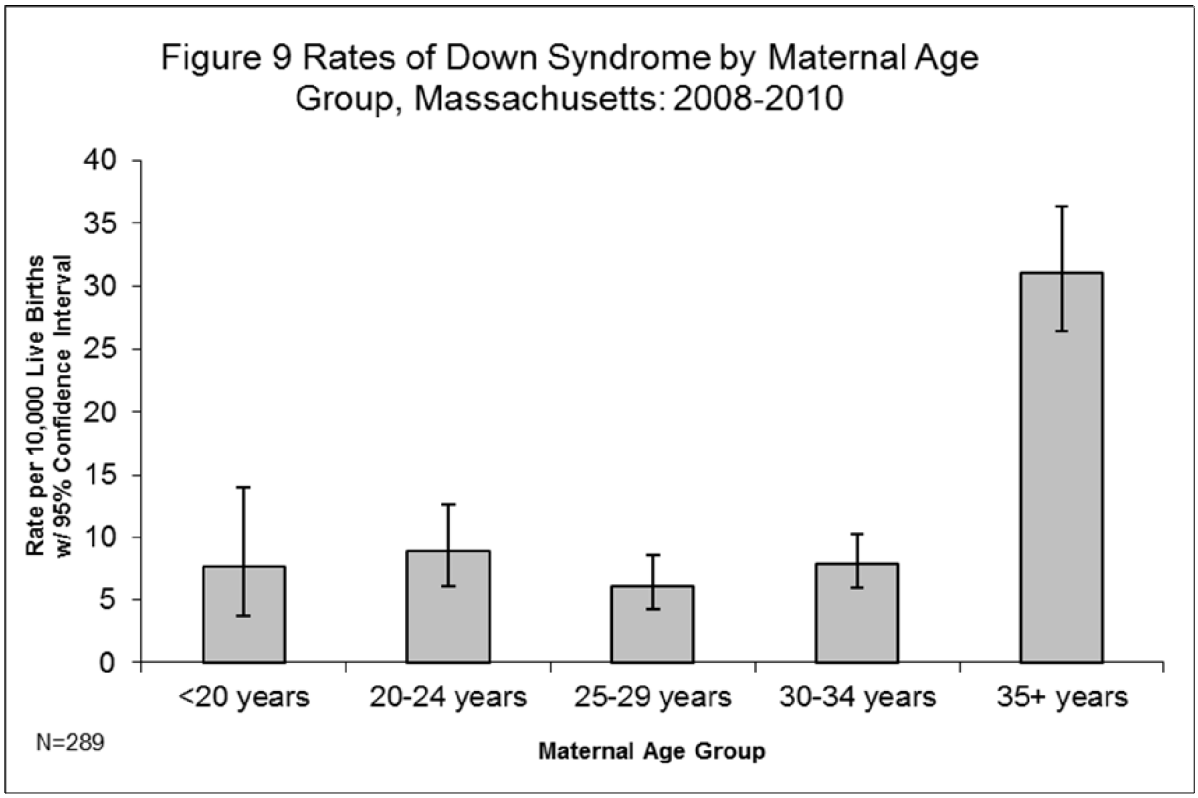


Figure 9 Rates of Down Syndrome by Maternal Age Group, Massachusetts: 2008-2010
Rate per 10,000 Live Births w/ 95% Confidence Interval
40
35
30
25
20
15
10
5
0
<20 years
20-24 years
25-29 years
30-34 years
35+ years
N=289
Maternal Age Group

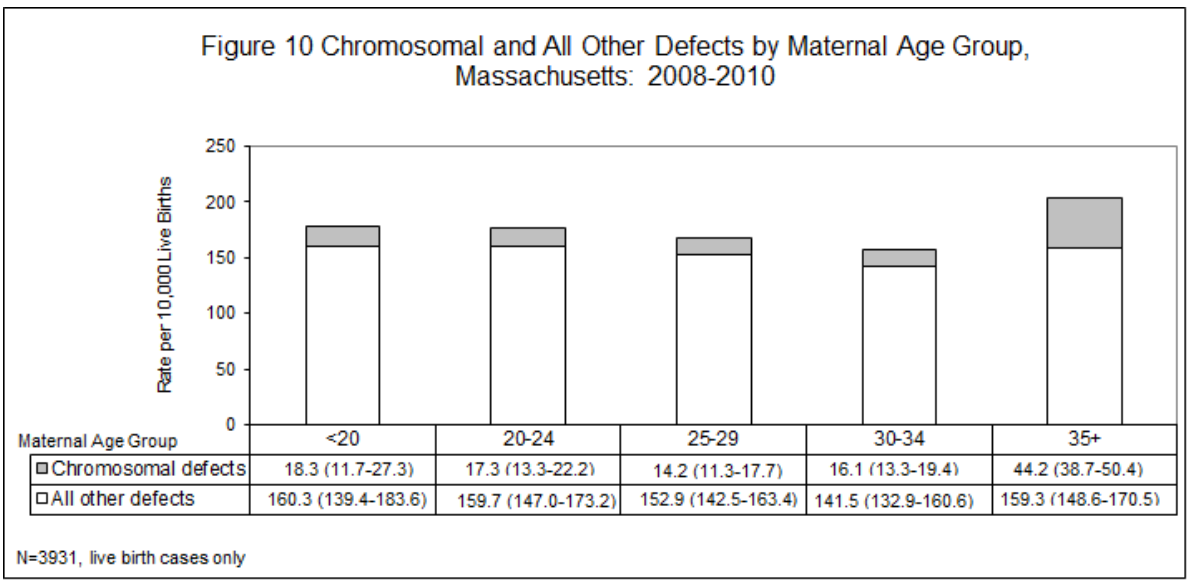


Figure 10 Chromosomal and All Other Defects by Maternal Age Group, Massachusetts: 2008-2010
Rate per 10,000 Live Births
250
200
150
100
50
0
Maternal Age Group
<20
20-24
25-29
30-34
35+
Chromosomal defects
18.3 (11.7-27.3)
17.3 (13.3-22.2)
14.2 (11.3-17.7)
16.1 (13.3-19.4)
44.2 (38.7-50.4)
All other defects
160.3 (139.4-183.6)
159.7 (147.0-173.2)
152.9 (142.5-163.4)
141.5 (132.9-160.6)
159.3 (148.6-170.5)
N=3931, live birth cases only

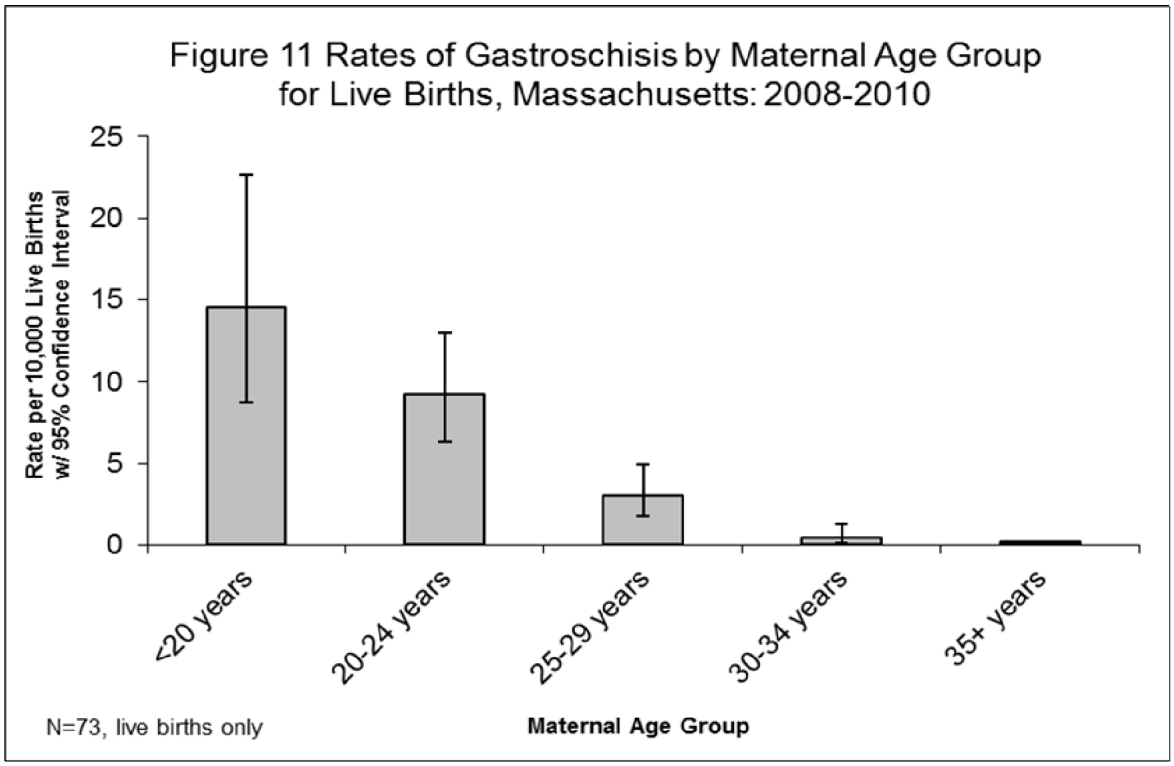


Figure 11 Rates of Gastroschisis by Maternal Age Group for Live Births, Massachusetts: 2008-2010
Rate per 10,000 Live Births w/ 95% Confidence Interval
25
20
15
10
5
0
<20 years
20-24 years
25-29 years
30-34 years
35+ years
N=73, live births only
Maternal Age Group

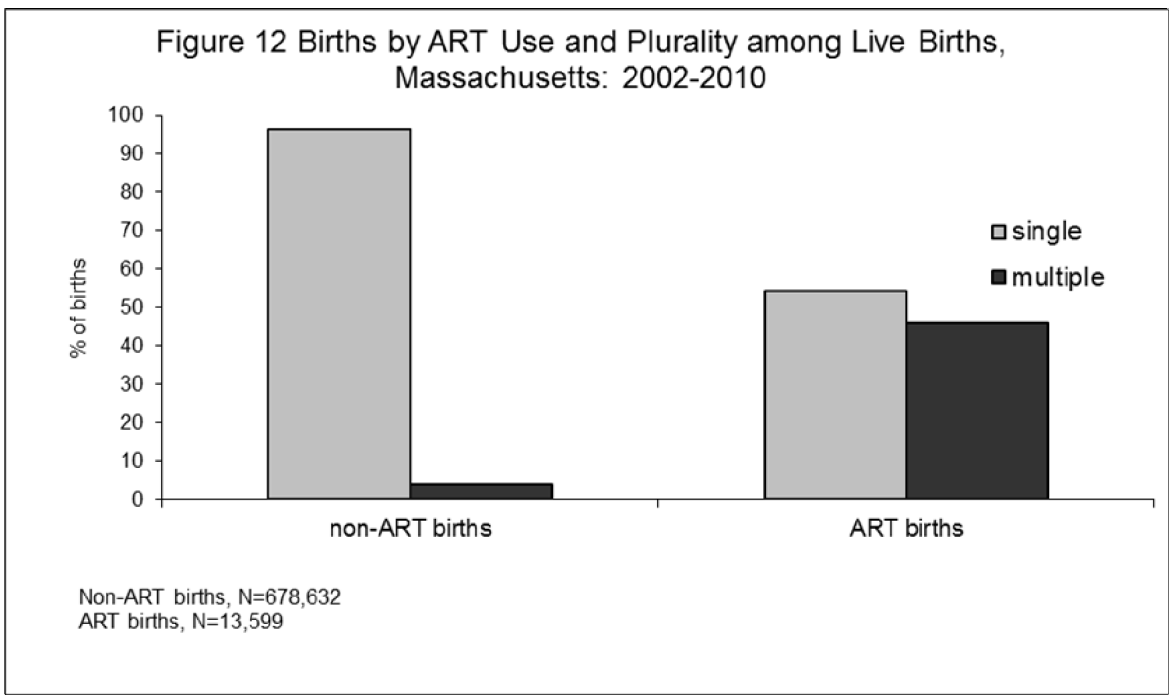


Figure 12 Births by ART Use and Plurality among Live Births, Massachusetts: 2002-2010
% of births
100
90
80
70
60
50
40
30
20
10
0
single
multiple
non-ART births
ART births
Non-ART births, N=678,632
ART births, N=13,599

Table 11 Most Common Defects by Maternal Age Group for Live Births, Massachusetts: 2008-2010

Maternal Age	Defect	Count	Rate per 10,000 Births	95% Confidence Interval
<20	Obstructive Genitourinary Defect	30	22.91	15.45-32.70
	Polydactyly/Syndactyly	25	19.09	12.35-28.18
	Club Foot	24	18.32	11.74-27.27
	ASD (Secundum and NOS)	22	16.80	10.53-25.43
	Gastroschisis	19	14.51	8.73-22.65
20-24	ASD (Secundum and NOS)	69	19.26	14.99-24.38
	Club Foot	68	18.98	14.74-24.07
	Obstructive Genitourinary Defect	61	17.03	13.03-21.87
	Polydactyly/Syndactyly	61	17.03	13.03-21.87
	VSD (Membranous and NOS)	40	11.17	7.98-15.21
25-29	ASD (Secundum and NOS)	106	19.09	15.63-23.09
	Polydactyly/Syndactyly	95	17.11	13.84-20.91
	Obstructive Genitourinary Defect	77	13.87	10.94-17.33
	Hypospadias, 2nd or 3rd Degree[1]	73	13.15	10.30-16.53
	Club Foot	71	12.79	9.99-16.13
30-34	ASD (Secundum and NOS)	144	20.73	17.49-24.41
	Hypospadias, 2nd or 3rd Degree[1]	107	15.41	12.63-18.62
	Polydactyly/Syndactyly	101	14.54	11.84-17.67
	Obstructive Genitourinary Defect	98	14.11	11.46-17.20
	Club Foot	92	13.25	10.68-16.25
35+	ASD (Secundum and NOS)	154	30.28	25.69-35.46
	Trisomy 21 (Down Syndrome)	150	29.50	24.96-34.61
	VSD (Membranous and NOS)	79	15.53	12.30-19.36
	Hypospadias, 2nd or 3rd Degree[1]	79	15.53	12.30-19.36
	Obstructive Genitourinary Defect	78	15.34	12.12-19.14

[1]Hypospadias 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 174.2 for non-Hispanic whites, 203.5 for non-Hispanic blacks, 143.4 for non-Hispanic Asians/Pacific Islanders and 180.6 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), polydactyly/syndactyly, and obstructive genitourinary defect amongst the most common defects. Down syndrome among the most common in all groups except white, non-Hispanic while hypospadias (2nd and 3rd degree) was among the common only in white, non-Hispanic and black, non-Hispanic. Non-Hispanic Asians had a significantly lower age-adjusted rate of chromosomal defects that was significantly lower than the non-Hispanic white rate. The non-Hispanic black and Hispanic rates were significantly different from one another, but both were significantly greater than the non-Hispanic white rate for chromosomal defects. This finding is similar to the 2008-2009 report but different from the 2006-2007 report, where the non-Hispanic black and Hispanic rates of chromosomal defects were not significantly different from non-Hispanic white rate. Maternal birthplace (U.S. versus non-U.S.) may be a contributing factor in racial/ethnic differences as women born in the U.S. had slightly—though not statistically significantly—higher rates than those born outside the U.S for most groups especially among non-Hispanic Asians more than 85% of whom were born outside the U.S. The non-Hispanic black rate of birth defects for non-U.S. residents was the only one of the four major racial/ethnic group that had the same as the birth rate for U.S. residents and non-residents. 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 (182.9.1 and 188.7 per 10,000 live births, respectively).

To better understand birth defect patterns in maternal race and ethnicity, we explored differences among the groups for certain categories of birth defects. Figure 13 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.

Figure 15 shows the rolling 2-year, age-adjusted birth defect prevalence rates between 2000 and 2010. The non-Hispanic black group, which saw a drop in prevalence rates starting in 2004-2005, has seen the greatest increase in prevalence rate since 2006-2007 while Hispanic rates, which had been increasing, saw rates dropping from 2007-2008 to the current two-year average. The other groups, non-Hispanic Asian and non-Hispanic white, saw only modest increases in prevalence rates since 2006-2007.

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 2008-2010 are shown in Figure 16. Although the regional differences in birth defect prevalence rate among the regions are not statistically significant from one another, the rates range from 165.5 per 10,000 live births in the Metro West region to 182.3 per 10,000 in the Boston region.

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008 -2010

Defect	Maternal Race[1]	Count[2]	Rate per 10,000 Births[3]	95% Confidence Interval
Total Cases	White, Non-Hispanic	2618	174.22	167.5-180.9
	Black, Non-Hispanic	415	203.47	183.9-223.0
	Asian, Non-Hispanic	243	143.37	125.3-161.4
	Hispanic	568	180.64	165.8-195.5
Central Nervous System				
Anencephaly	White, Non-Hispanic	4	0.27	0.07-0.68
	Black, Non-Hispanic	1	0.49	0.01-2.73
	Asian, Non-Hispanic	1	0.56	0.01-3.15
	Hispanic	2	0.62	0.07-2.23
Encephalocele	White, Non-Hispanic	3	0.20	0.04-0.58
	Black, Non-Hispanic	0	0.00	0.00-1.81
	Asian, Non-Hispanic	0	0.00	0.00-2.08
	Hispanic	2	0.62	0.07-2.23
Holoprosencephaly	White, Non-Hispanic	7	0.47	0.19-0.96
	Black, Non-Hispanic	1	0.49	0.01-2.73
	Asian, Non-Hispanic	1	0.56	0.01-3.15
	Hispanic	4	1.23	0.34-3.15
Hydrocephaly w/o Spina Bifida	White, Non-Hispanic	38	2.53	1.79-3.48
	Black, Non-Hispanic	13	6.38	3.39-10.90
	Asian, Non-Hispanic	1	0.56	0.01-3.15
	Hispanic	16	4.93	2.82-8.00
Microcephaly	White, Non-Hispanic	24	1.60	1.03-2.38
	Black, Non-Hispanic	4	1.96	0.53-5.02
	Asian, Non-Hispanic	1	0.56	0.01-3.15
	Hispanic	5	1.54	0.50-3.59
Spina Bifida w/ and w/o Hydrocephaly	White, Non-Hispanic	32	2.13	1.46-3.01
	Black, Non-Hispanic	5	2.45	0.80-5.72
	Asian, Non-Hispanic	3	1.69	0.35-4.95
	Hispanic	8	2.46	1.06-4.85
Spinal Cord	White, Non-Hispanic	59	3.93	2.99-5.07
	Black, Non-Hispanic	8	3.92	1.69-7.73
	Asian, Non-Hispanic	6	3.39	1.24-7.37
	Hispanic	12	3.70	1.91-6.46

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008 -2010

Defect	Maternal Race[1]	Count[2]	Rate per 10,000 Births[3]	95% Confidence Interval
(cont'd)				
Other CNS[5]	White, Non-Hispanic	97	6.47	5.24-7.89
	Black, Non-Hispanic	28	13.73	9.12-19.85
	Asian, Non-Hispanic	8	4.52	1.95-8.90
	Hispanic	29	8.93	5.98-12.83
Eye				
Aniridia	White, Non-Hispanic	2	0.13	0.02-0.48
	Black, Non-Hispanic	0	0.00	0.00-1.81
	Asian, Non-Hispanic	1	0.56	0.01-3.15
	Hispanic	0	0.00	0.00-1.14
Anophthalmia/Microphthalmia	White, Non-Hispanic	20	1.33	0.81-2.06
	Black, Non-Hispanic	3	1.47	0.30-4.30
	Asian, Non-Hispanic	2	1.13	0.14-4.08
	Hispanic	7	2.16	0.87-4.44
Congenital Glaucoma, Congenital Cataract	White, Non-Hispanic	35	2.33	1.63-3.25
	Black, Non-Hispanic	11	5.39	2.69-9.65
	Asian, Non-Hispanic	3	1.69	0.35-4.95
	Hispanic	16	4.93	2.82-8.00
Other Eye[5]	White, Non-Hispanic	45	3.00	2.19-4.01
	Black, Non-Hispanic	5	2.45	0.80-5.72
	Asian, Non-Hispanic	5	2.82	0.92-6.59
	Hispanic	14	4.31	2.36-7.23
Ear				
Anotia/Microtia	White, Non-Hispanic	20	1.33	0.81-2.06
	Black, Non-Hispanic	2	0.98	0.12-3.54
	Asian, Non-Hispanic	7	3.95	1.59-8.14
	Hispanic	9	2.77	1.27-5.26
Other Ear[5]	White, Non-Hispanic	50	3.33	2.47-4.40
	Black, Non-Hispanic	3	1.47	0.30-4.30
	Asian, Non-Hispanic	9	5.08	2.32-9.64
	Hispanic	9	2.77	1.27-5.26

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008 -2010

Defect	Maternal Race[1]	Count[2]	Rate per 10,000 Births[3]	95% Confidence Interval
Cardiovascular				
Anomalous Pulmonary Venous Connection				
Total/Partial Anomalous Pulmonary Venous Connection	White, Non-Hispanic	23	1.53	0.97-2.30
	Black, Non-Hispanic	2	0.98	0.12-3.54
	Asian, Non-Hispanic	6	3.39	1.24-7.37
	Hispanic	6	1.85	0.68-4.02
Atrioventricular Canal Defects				
ASD Primum	White, Non-Hispanic	4	0.27	0.07-0.68
	Black, Non-Hispanic	0	0.00	0.00-1.81
	Asian, Non-Hispanic	1	0.56	0.01-3.15
	Hispanic	0	0.00	0.00-1.14
Common Atrium	White, Non-Hispanic	6	0.40	0.15-0.87
	Black, Non-Hispanic	2	0.98	0.12-3.54
	Asian, Non-Hispanic	1	0.56	0.01-3.15
	Hispanic	0	0.00	0.00-1.14
Complete Atrioventricular Canal Defect	White, Non-Hispanic	42	2.80	2.02-3.79
	Black, Non-Hispanic	10	4.90	2.35-9.02
	Asian, Non-Hispanic	4	2.26	0.62-5.78
	Hispanic	7	2.16	0.87-4.44
Endocardial Cushion (OS and NOS)	White, Non-Hispanic	27	1.80	1.19-2.62
	Black, Non-Hispanic	3	1.47	0.30-4.30
	Asian, Non-Hispanic	1	0.56	0.01-3.15
	Hispanic	3	0.92	0.19-2.70
VSD, Canal Type	White, Non-Hispanic	4	0.27	0.07-0.68
	Black, Non-Hispanic	2	0.98	0.12-3.54
	Asian, Non-Hispanic	0	0.00	0.00-2.08
	Hispanic	1	0.31	0.01-1.72
Conotruncal (Outlet) and Aortic Arch				
Double Outlet Right Ventricle	White, Non-Hispanic	19	1.27	0.76-1.98
	Black, Non-Hispanic	1	0.49	0.01-2.73
	Asian, Non-Hispanic	1	0.56	0.01-3.15
	Hispanic	2	0.62	0.07-2.23

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008 -2010

Defect	Maternal Race[1]	Count[2]	Rate per 10,000 Births[3]	95% Confidence Interval
(cont'd)				
Interrupted Aortic Arch, Type B	White, Non-Hispanic	5	0.33	0.11-0.78
	Black, Non-Hispanic	2	0.98	0.12-3.54
	Asian, Non-Hispanic	0	0.00	0.00-2.08
	Hispanic	1	0.31	0.01-1.72
Tetralogy of Fallot w/ and w/o Pulmonary Atresia	White, Non-Hispanic	48	3.20	2.36-4.24
	Black, Non-Hispanic	11	5.39	2.69-9.65
	Asian, Non-Hispanic	7	3.95	1.59-8.14
	Hispanic	19	5.85	3.52-9.14
Truncus	White, Non-Hispanic	5	0.33	0.11-0.78
	Black, Non-Hispanic	0	0.00	0.00-1.81
	Asian, Non-Hispanic	0	0.00	0.00-2.08
	Hispanic	0	0.00	0.00-1.14
d-Transposition of the Great Arteries	White, Non-Hispanic	42	2.80	2.02-3.79
	Black, Non-Hispanic	4	1.96	0.53-5.02
	Asian, Non-Hispanic	3	1.69	0.35-4.95
	Hispanic	5	1.54	0.50-3.59
Ebstein Anomaly				
Ebstein Anomaly	White, Non-Hispanic	5	0.33	0.11-0.78
	Black, Non-Hispanic	1	0.49	0.01-2.73
	Asian, Non-Hispanic	0	0.00	0.00-2.08
	Hispanic	2	0.62	0.07-2.23
Heterotaxy (Laterality Defects)				
Heterotaxy	White, Non-Hispanic	22	1.47	0.92-2.22
	Black, Non-Hispanic	4	1.96	0.53-5.02
	Asian, Non-Hispanic	3	1.69	0.35-4.95
	Hispanic	4	1.23	0.34-3.15
Left-Sided Obstruction				
Aortic Valve Stenosis	White, Non-Hispanic	25	1.67	1.08-2.46
	Black, Non-Hispanic	4	1.96	0.53-5.02
	Asian, Non-Hispanic	0	0.00	0.00-2.08
	Hispanic	2	0.62	0.07-2.23

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008 -2010

Defect	Maternal Race[1]	Count[2]	Rate per 10,000 Births[3]	95% Confidence Interval
(cont'd)				
Coarctation of Aorta	White, Non-Hispanic	69	4.60	3.58-5.82
	Black, Non-Hispanic	8	3.92	1.69-7.73
	Asian, Non-Hispanic	2	1.13	0.14-4.08
	Hispanic	16	4.93	2.82-8.00
Hypoplastic Left Heart Syndrome	White, Non-Hispanic	20	1.33	0.81-2.06
	Black, Non-Hispanic	5	2.45	0.80-5.72
	Asian, Non-Hispanic	0	0.00	0.00-2.08
	Hispanic	4	1.23	0.34-3.15
Interrupted Aortic Arch (Type A and NOS)	White, Non-Hispanic	0	0.00	0.00-0.25
	Black, Non-Hispanic	0	0.00	0.00-1.81
	Asian, Non-Hispanic	0	0.00	0.00-2.08
	Hispanic	1	0.31	0.01-1.72
Patent Ductus Arteriosus				
Patent Ductus Arteriosus	White, Non-Hispanic	197	13.13	11.36-15.10
	Black, Non-Hispanic	34	16.67	11.55-23.30
	Asian, Non-Hispanic	22	12.42	7.78-18.80
	Hispanic	48	14.78	10.90-19.60
Right-Sided Obstruction				
Pulmonary Stenosis, Valvular	White, Non-Hispanic	95	6.33	5.12-7.74
	Black, Non-Hispanic	28	13.73	9.12-19.85
	Asian, Non-Hispanic	8	4.52	1.95-8.90
	Hispanic	23	7.08	4.49-10.63
Pulmonary Valve Atresia w/intact septum	White, Non-Hispanic	8	0.53	0.23-1.05
	Black, Non-Hispanic	2	0.98	0.12-3.54
	Asian, Non-Hispanic	0	0.00	0.00-2.08
	Hispanic	2	0.62	0.07-2.23
Pulmonary Valve Atresia with VSD	White, Non-Hispanic	2	0.13	0.02-0.48
	Black, Non-Hispanic	2	0.98	0.12-3.54
	Asian, Non-Hispanic	1	0.56	0.01-3.15
	Hispanic	1	0.31	0.01-1.72

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008 -2010

Defect	Maternal Race[1]	Count[2]	Rate per 10,000 Births[3]	95% Confidence Interval
(cont'd)				
Tricuspid Valve Atresia	White, Non-Hispanic	11	0.73	0.37-1.31
	Black, Non-Hispanic	2	0.98	0.12-3.54
	Asian, Non-Hispanic	1	0.56	0.01-3.15
	Hispanic	2	0.62	0.07-2.23
Septal Defects				
ASD (Secundum and NOS)	White, Non-Hispanic	316	21.07	18.81-23.52
	Black, Non-Hispanic	65	31.88	24.60-40.63
	Asian, Non-Hispanic	34	19.19	13.29-26.82
	Hispanic	72	22.17	17.35-27.93
VSD (Membranous and NOS)	White, Non-Hispanic	164	10.93	9.32-12.74
	Black, Non-Hispanic	26	12.75	8.33-18.68
	Asian, Non-Hispanic	25	14.11	9.13-20.83
	Hispanic	42	12.94	9.32-17.48
VSD, Conoventricular/Malalignment	White, Non-Hispanic	29	1.93	1.29-2.78
	Black, Non-Hispanic	4	1.96	0.53-5.02
	Asian, Non-Hispanic	0	0.00	0.00-2.08
	Hispanic	5	1.54	0.50-3.59
Single Ventricle and L-TGA				
L-TGA	White, Non-Hispanic	6	0.40	0.15-0.87
	Black, Non-Hispanic	1	0.49	0.01-2.73
	Asian, Non-Hispanic	0	0.00	0.00-2.08
	Hispanic	0	0.00	0.00-1.14
Single Ventricle	White, Non-Hispanic	6	0.40	0.15-0.87
	Black, Non-Hispanic	1	0.49	0.01-2.73
	Asian, Non-Hispanic	2	1.13	0.14-4.08
	Hispanic	0	0.00	0.00-1.14
Other Cardiovascular				
Other Cardiovascular[5]	White, Non-Hispanic	305	20.34	18.12-22.75
	Black, Non-Hispanic	59	28.93	22.03-37.32
	Asian, Non-Hispanic	27	15.24	10.04-22.18
	Hispanic	69	21.25	16.53-26.89

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008 -2010

Defect	Maternal Race[1]	Count[2]	Rate per 10,000 Births[3]	95% Confidence Interval
Respiratory				
Lung Anomalies	White, Non-Hispanic	22	1.47	0.92-2.22
	Black, Non-Hispanic	4	1.96	0.53-5.02
	Asian, Non-Hispanic	1	0.56	0.01-3.15
	Hispanic	15	4.62	2.59-7.62
Other Respiratory[5]	White, Non-Hispanic	22	1.47	0.92-2.22
	Black, Non-Hispanic	0	0.00	0.00-1.81
	Asian, Non-Hispanic	1	0.56	0.01-3.15
	Hispanic	3	0.92	0.19-2.70
Orofacial				
Cleft Lip w/ and w/o Cleft Palate	White, Non-Hispanic	128	8.53	7.12-10.15
	Black, Non-Hispanic	11	5.39	2.69-9.65
	Asian, Non-Hispanic	12	6.77	3.50-11.83
	Hispanic	25	7.70	4.98-11.37
Cleft Palate w/o Cleft Lip	White, Non-Hispanic	94	6.27	5.06-7.67
	Black, Non-Hispanic	4	1.96	0.53-5.02
	Asian, Non-Hispanic	5	2.82	0.92-6.59
	Hispanic	16	4.93	2.82-8.00
Pierre Robin Sequence	White, Non-Hispanic	33	2.20	1.51-3.09
	Black, Non-Hispanic	1	0.49	0.01-2.73
	Asian, Non-Hispanic	1	0.56	0.01-3.15
	Hispanic	6	1.85	0.68-4.02
Other Orofacial[5]	White, Non-Hispanic	80	5.33	4.23-6.64
	Black, Non-Hispanic	14	6.87	3.75-11.52
	Asian, Non-Hispanic	7	3.95	1.59-8.14
	Hispanic	13	4.00	2.13-6.85
Gastrointestinal				
Biliary Atresia	White, Non-Hispanic	6	0.40	0.15-0.87
	Black, Non-Hispanic	1	0.49	0.01-2.73
	Asian, Non-Hispanic	4	2.26	0.62-5.78
	Hispanic	3	0.92	0.19-2.70

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008 -2010

Defect	Maternal Race[1]	Count[2]	Rate per 10,000 Births[3]	95% Confidence Interval
(cont'd)				
Esophageal Atresia/Tracheoesophageal Fistula	White, Non-Hispanic	42	2.80	2.02-3.79
	Black, Non-Hispanic	3	1.47	0.30-4.30
	Asian, Non-Hispanic	1	0.56	0.01-3.15
	Hispanic	5	1.54	0.50-3.59
Hirschsprung Disease	White, Non-Hispanic	32	2.13	1.46-3.01
	Black, Non-Hispanic	2	0.98	0.12-3.54
	Asian, Non-Hispanic	7	3.95	1.59-8.14
	Hispanic	11	3.39	1.69-6.06
Rectal and Large Intestinal Atresia/Stenosis	White, Non-Hispanic	46	3.07	2.25-4.09
	Black, Non-Hispanic	8	3.92	1.69-7.73
	Asian, Non-Hispanic	5	2.82	0.92-6.59
	Hispanic	8	2.46	1.06-4.85
Small Intestinal Atresia	White, Non-Hispanic	40	2.67	1.91-3.63
	Black, Non-Hispanic	3	1.47	0.30-4.30
	Asian, Non-Hispanic	9	5.08	2.32-9.64
	Hispanic	11	3.39	1.69-6.06
Other Gastrointestinal[5]	White, Non-Hispanic	99	6.60	5.36-8.04
	Black, Non-Hispanic	21	10.30	6.38-15.74
	Asian, Non-Hispanic	13	7.34	3.91-12.55
	Hispanic	24	7.39	4.74-11.00
Genitourinary				
Bladder Exstrophy	White, Non-Hispanic	3	0.20	0.04-0.58
	Black, Non-Hispanic	0	0.00	0.00-1.81
	Asian, Non-Hispanic	0	0.00	0.00-2.08
	Hispanic	1	0.31	0.01-1.72
Cloacal Exstrophy	White, Non-Hispanic	1	0.07	0.00-0.37
	Black, Non-Hispanic	1	0.49	0.01-2.73
	Asian, Non-Hispanic	0	0.00	0.00-2.08
	Hispanic	0	0.00	0.00-1.14

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008 -2010

Defect	Maternal Race[1]	Count[2]	Rate per 10,000 Births[3]	95% Confidence Interval
(cont'd)				
Hypospadias, 2nd or 3rd Degree	White, Non-Hispanic	228	15.20	13.29-17.31
	Black, Non-Hispanic	36	17.65	12.37-24.44
	Asian, Non-Hispanic	15	8.47	4.74-13.97
	Hispanic	26	8.01	5.23-11.73
Obstructive Genitourinary Defect	White, Non-Hispanic	223	14.87	12.98-16.95
	Black, Non-Hispanic	31	15.20	10.33-21.58
	Asian, Non-Hispanic	25	14.11	9.13-20.83
	Hispanic	61	18.79	14.37-24.13
Renal Agenesis/Hypoplasia	White, Non-Hispanic	3	0.20	0.04-0.58
	Black, Non-Hispanic	3	1.47	0.30-4.30
	Asian, Non-Hispanic	0	0.00	0.00-2.08
	Hispanic	1	0.31	0.01-1.72
Other Genitourinary[5]	White, Non-Hispanic	272	18.14	16.04-20.42
	Black, Non-Hispanic	34	16.67	11.55-23.30
	Asian, Non-Hispanic	29	16.37	10.96-23.51
	Hispanic	53	16.32	12.23-21.35
Musculoskeletal				
Club Foot	White, Non-Hispanic	224	14.93	13.04-17.02
	Black, Non-Hispanic	30	14.71	9.93-21.00
	Asian, Non-Hispanic	13	7.34	3.91-12.55
	Hispanic	41	12.63	9.06-17.13
Craniosynostosis	White, Non-Hispanic	91	6.07	4.88-7.45
	Black, Non-Hispanic	4	1.96	0.53-5.02
	Asian, Non-Hispanic	3	1.69	0.35-4.95
	Hispanic	9	2.77	1.27-5.26
Diaphragmatic Hernia	White, Non-Hispanic	34	2.27	1.57-3.17
	Black, Non-Hispanic	5	2.45	0.80-5.72
	Asian, Non-Hispanic	3	1.69	0.35-4.95
	Hispanic	8	2.46	1.06-4.85

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008 -2010

Defect	Maternal Race[1]	Count[2]	Rate per 10,000 Births[3]	95% Confidence Interval
(cont'd)				
Gastroschisis	White, Non-Hispanic	39	2.60	1.85-3.55
	Black, Non-Hispanic	12	5.88	3.04-10.28
	Asian, Non-Hispanic	2	1.13	0.14-4.08
	Hispanic	17	5.24	3.05-8.38
Omphalocele	White, Non-Hispanic	14	0.93	0.51-1.57
	Black, Non-Hispanic	6	2.94	1.08-6.40
	Asian, Non-Hispanic	0	0.00	0.00-2.08
	Hispanic	4	1.23	0.34-3.15
Polydactyly/Syndactyly	White, Non-Hispanic	217	14.47	12.61-16.53
	Black, Non-Hispanic	48	23.54	17.36-31.21
	Asian, Non-Hispanic	28	15.81	10.50-22.85
	Hispanic	52	16.02	11.96-21.00
Reduction Deformity, Lower Limbs	White, Non-Hispanic	16	1.07	0.61-1.73
	Black, Non-Hispanic	2	0.98	0.12-3.54
	Asian, Non-Hispanic	1	0.56	0.01-3.15
	Hispanic	8	2.46	1.06-4.85
Reduction Deformity, Upper Limbs	White, Non-Hispanic	37	2.47	1.74-3.40
	Black, Non-Hispanic	3	1.47	0.30-4.30
	Asian, Non-Hispanic	3	1.69	0.35-4.95
	Hispanic	10	3.08	1.48-5.66
Skeletal Dysplasia	White, Non-Hispanic	17	1.13	0.66-1.81
	Black, Non-Hispanic	5	2.45	0.80-5.72
	Asian, Non-Hispanic	1	0.56	0.01-3.15
	Hispanic	5	1.54	0.50-3.59
Other Musculoskeletal[5]	White, Non-Hispanic	160	10.67	9.08-12.45
	Black, Non-Hispanic	33	16.18	11.14-22.73
	Asian, Non-Hispanic	14	7.90	4.32-13.26
	Hispanic	41	12.63	9.06-17.13

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008 -2010

Defect	Maternal Race[1]	Count[2]	Rate per 10,000 Births[3]	95% Confidence Interval
Chromosomal and other Syndromes				
Klinefelter Syndrome	White, Non-Hispanic	7	0.47	0.19-0.96
	Black, Non-Hispanic	0	0.00	0.00-1.81
	Asian, Non-Hispanic	0	0.00	0.00-2.08
	Hispanic	1	0.31	0.01-1.72
Trisomy 13	White, Non-Hispanic	5	0.33	0.11-0.78
	Black, Non-Hispanic	1	0.49	0.01-2.73
	Asian, Non-Hispanic	0	0.00	0.00-2.08
	Hispanic	0	0.00	0.00-1.14
Trisomy 18	White, Non-Hispanic	13	0.87	0.46-1.48
	Black, Non-Hispanic	4	1.96	0.53-5.02
	Asian, Non-Hispanic	1	0.56	0.01-3.15
	Hispanic	7	2.16	0.87-4.44
Trisomy 21 (Down Syndrome)	White, Non-Hispanic	172	11.47	9.82-13.32
	Black, Non-Hispanic	34	16.67	11.55-23.30
	Asian, Non-Hispanic	18	10.16	6.02-16.06
	Hispanic	49	15.09	11.16-19.95
Turner Syndrome	White, Non-Hispanic	9	0.60	0.27-1.14
	Black, Non-Hispanic	1	0.49	0.01-2.73
	Asian, Non-Hispanic	0	0.00	0.00-2.08
	Hispanic	3	0.92	0.19-2.70
Other Chromosomal Syndromes/Other Syndromes[5]	White, Non-Hispanic	218	14.53	12.67-16.60
	Black, Non-Hispanic	27	13.24	8.73-19.27
	Asian, Non-Hispanic	15	8.47	4.74-13.97
	Hispanic	44	13.55	9.85-18.19
Other				
Amniotic Bands	White, Non-Hispanic	11	0.73	0.37-1.31
	Black, Non-Hispanic	4	1.96	0.53-5.02
	Asian, Non-Hispanic	0	0.00	0.00-2.08
	Hispanic	2	0.62	0.07-2.23

Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008 -2010

Defect	Maternal Race[1]	Count[2]	Rate per 10,000 Births[3]	95% Confidence Interval
(cont'd)				
Skin Anomalies	White, Non-Hispanic	20	1.33	0.81-2.06
	Black, Non-Hispanic	8	3.92	1.69-7.73
	Asian, Non-Hispanic	3	1.69	0.35-4.95
	Hispanic	6	1.85	0.68-4.02
Other, Specified[5]	White, Non-Hispanic	19	1.27	0.76-1.98
	Black, Non-Hispanic	4	1.96	0.53-5.02
	Asian, Non-Hispanic	2	1.13	0.14-4.08
	Hispanic	2	0.62	0.07-2.23

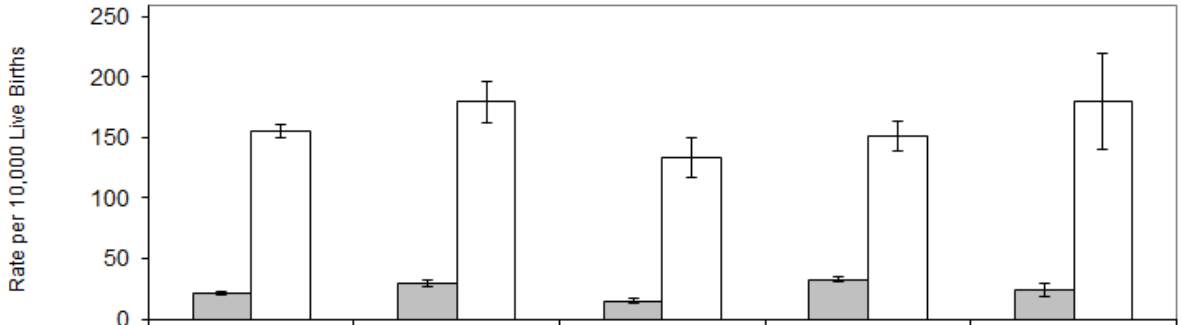
[1.] Due to small numbers, races classified as "other" are not included.
[2.] Cases can be included in more than one defect. Cases are counted once in the total for a defect category. Because only live births are presented on this table, case totals are not listed. Due to missing race of mother counts 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.
[5] Rate may represent a heterogeneous group of defects.

Table 13 Most Common Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2008-2010

Maternal Race[1]	Defect	Count	Rate per 10,000 Births	95% Confidence Interval
White, Non-Hispanic	ASD (Secundum and NOS)	316	21.07	18.81-23.52
	Hypospadias, 2nd or 3rd Degree[2]	228	15.20	13.29-17.31
	Club Foot	224	14.93	13.04-17.02
	Obstructive Genitourinary Defect	223	14.87	12.98-16.95
	Polydactyly/Syndactyly	217	14.47	12.61-16.53
Black, Non-Hispanic	ASD (Secundum and NOS)	65	31.88	24.60-40.63
	Polydactyly/Syndactyly	48	23.54	17.36-31.21
	Hypospadias, 2nd or 3rd Degree[2]	36	17.65	12.37-24.44
	Trisomy 21 (Down Syndrome)	34	16.67	11.55-23.30
	Obstructive Genitourinary Defect	31	15.20	10.33-21.58
Asian, Non-Hispanic	ASD (Secundum and NOS)	34	19.19	13.29-26.82
	Polydactyly/Syndactyly	28	15.81	10.50-22.85
	VSD (Membranous and NOS)	25	14.11	9.13-20.83
	Obstructive Genitourinary Defect	25	14.11	9.13-20.83
	Trisomy 21 (Down Syndrome)	18	10.16	6.02-16.06
Hispanic	ASD (Secundum and NOS)	72	22.17	17.35-27.93
	Obstructive Genitourinary Defect	61	18.79	14.37-24.13
	Polydactyly/Syndactyly	52	16.02	11.96-21.00
	Trisomy 21 (Down Syndrome)	49	15.09	11.16-19.95
	VSD (Membranous and NOS)	42	12.94	9.32-17.48

1. Due to small numbers, races classified as “other” are not included.
2. Rate calculated using male live births.

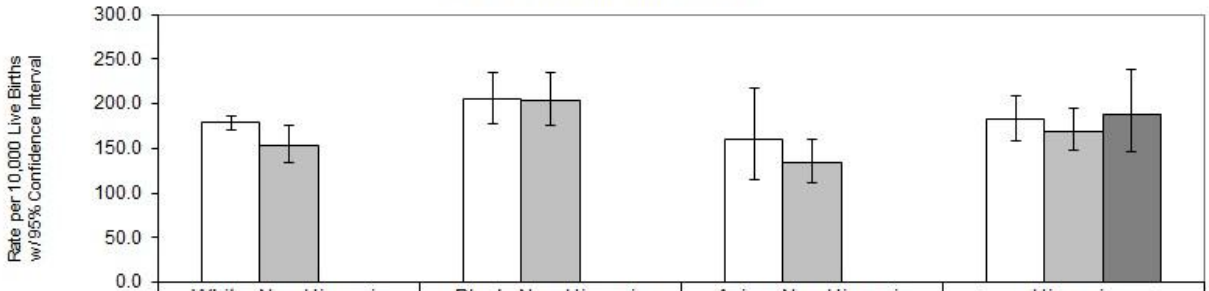
Figure 13 Age-Adjusted Prevalence of Chromosomal and All Other Defects by Maternal Race / Hispanic Ethnicity, Massachusetts: 2008-2010



	White, Non-Hispanic	Black, Non-Hispanic	Asian, Non-Hispanic	Hispanic	Other
Chromosomal	21.2 (20.4-22.0)	29.0 (26.2-31.7)	15.0 (13.1-16.9)	32.8 (30.1-35.4)	24.6 (19.1-30.0)
All other defects	155.3 (149.4-161.2)	179.8 (162.7-196.9)	133.8 (117.3-150.4)	151.7 (139.5-164.0)	179.6 (139.8-219.5)

Live births only, N = 3919 (12 unknown race)

Figure 14 Prevalence of Birth Defects by Maternal Race / Hispanic Ethnicity and Birthplace, Massachusetts: 2008-2010



	White, Non-Hispanic	Black, Non-Hispanic	Asian, Non-Hispanic	Hispanic
U.S.-Born*	178.5 (171.4-185.9)	205.3 (177.8-235.8)	160.7 (115.8-217.2)	182.9 (159.3-209.0)
Non-U.S.-Born	154.1 (136.8-173.1)	203.2(177.1-232.0)	134.2 (116.2-154.1)	169.7 (149.6-191.6)
U.S. Territories	**	**	**	188.7 (154.0-228.9)

*Including American Samoa, Federated State of Micronesia, Guam, Marshall Islands, Northern Mariana Islands, Palau, and Puerto Rico for all except Hispanic
**Too few cases in this category to calculae prevalence; U.S. Territories were inlcuded in Non-U.S.-births for non-Hispanic
N=4012

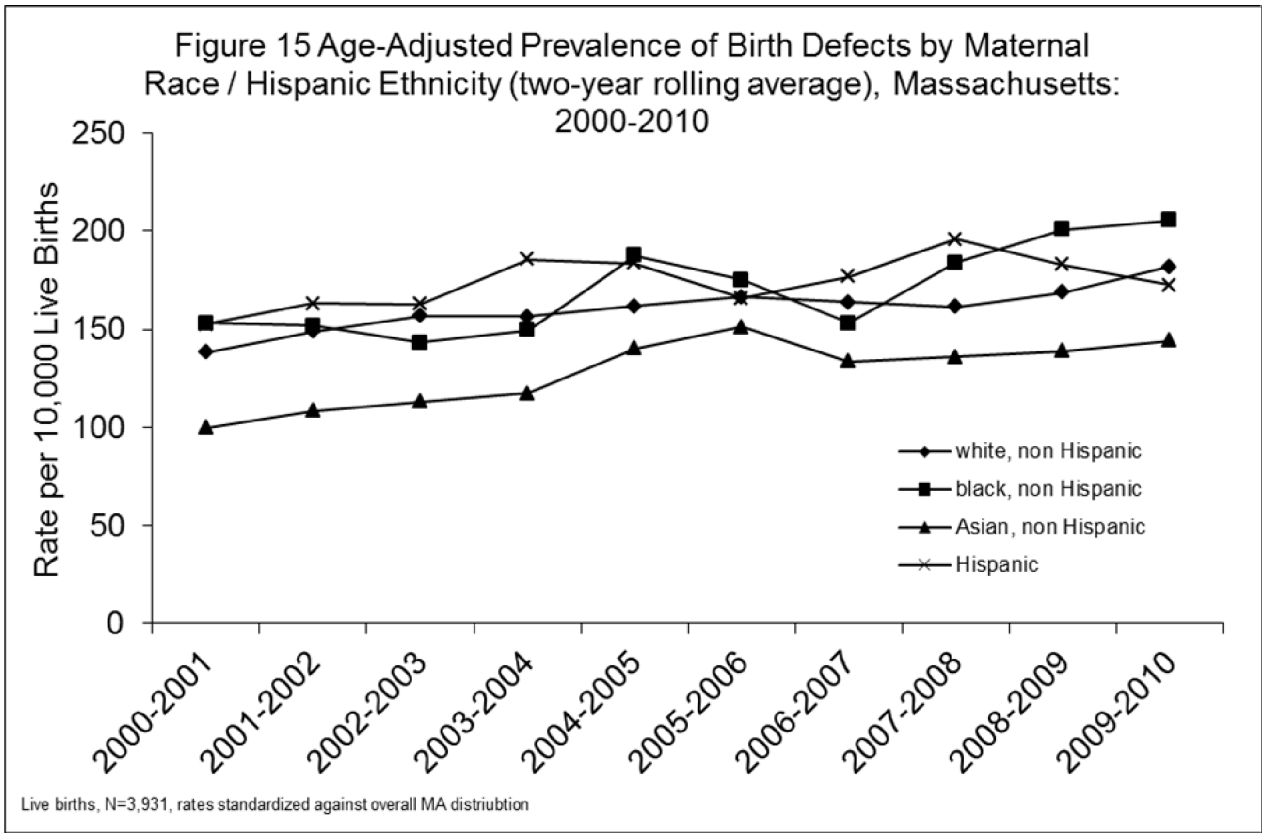


Figure 15 Age-Adjusted Prevalence of Birth Defects by Maternal Race / Hispanic Ethnicity (two-year rolling average), Massachusetts: 2000-2010
Rate per 10,000 Live Births
250
200
150
100
50
0
2000-2001
2001-2002
2002-2003
2003-2004
2004-2005
2005-2006
2006-2007
2007-2008
2008-2009
2009-2010
white, non Hispanic
black, non Hispanic
Asian, non Hispanic
Hispanic
Live births, N=3,931, rates standardized against overall MA distriubtion

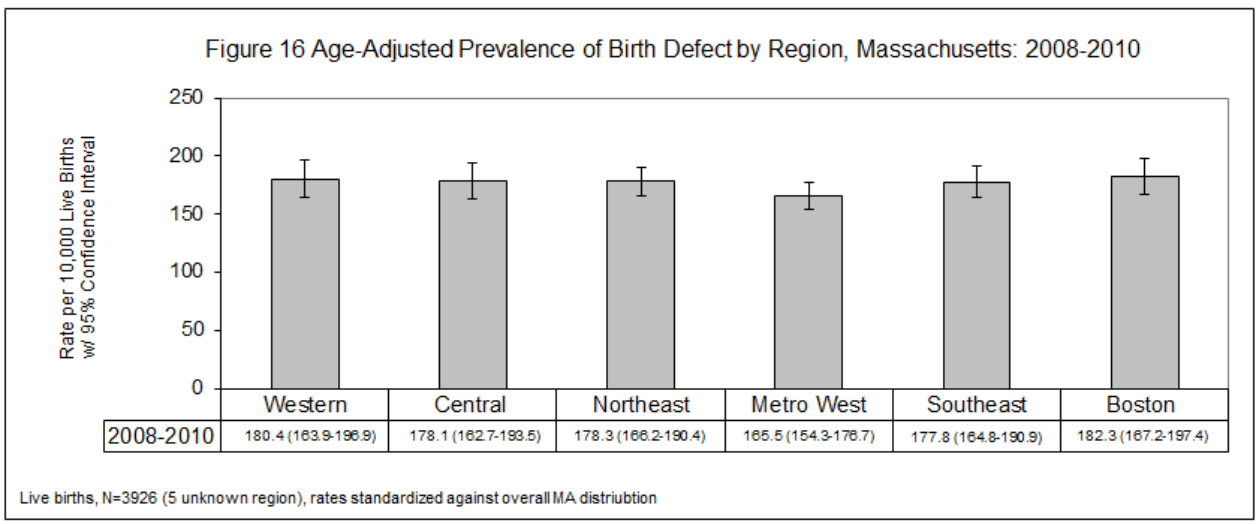
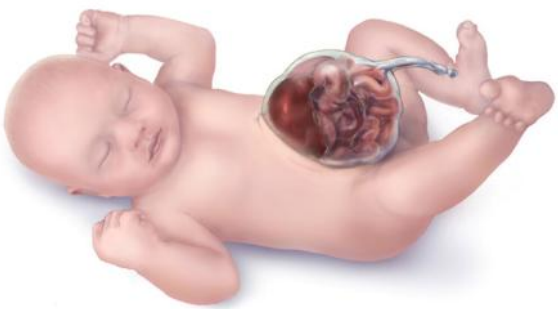


Figure 16 Age-Adjusted Prevalence of Birth Defect by Region, Massachusetts: 2008-2010
Rate per 10,000 Live Births w/ 95% Confidence Interval
250
200
150
100
50
0
Western
Central
Northeast
Metro West
Southeast
Boston
2008-2010
180.4 (163.9-196.9)
178.1 (162.7-193.5)
178.3 (166.2-190.4)
165.5 (154.3-176.7)
177.8 (164.8-190.9)
182.3 (167.2-197.4)
Live births, N=3926 (5 unknown region), rates standardized against overall MA distriubtion

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).

CDC estimates that about 30% of birth defectsnationwide have a known cause (or etiology). In Massachusetts, cases with known etiology accounted for only 18.5% of the birth defects in 2008-2010. Of the cases with known cause, "single gene" etiology accounted for 21.6%, "chromosomal" etiology accounted for 72.8% and "maternal-fetal factors" accounted for 3.4% of cases. The vast majority of birth defects cases in Massachusetts in 2008-2010 (81.4%, or 3,266 of 4,012 total cases had an unknown cause.

As Figure 17 shows, single gene etiology accounted for 21.5% 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 73% 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 3.4% 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 which contribute less that 1% of cases and are not shown in Figure 17 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.

Cases are also classified by the *pattern* of defects i.e. whether one defect occurs with others. Of all 4,012 birth defect cases (3,931 live births and 81 stillbirths) 43.5% had a "solitary" (truly a single) defect pattern, 26.4% had "Major plus minors" (defined as having a major defect accompanied by one or more minor defects), 4.3% were a "sequence" (allowing for more than one major defect if the defects are related pathogenically), and 25.6% had "multiple major" defects. As shown in Table 15, among only those 743 cases that had a known cause, the vast majority (93%, or 695 of 743 cases) were defined as having multiple major defects. This finding is in line with the majority of defects with a known etiology being chromosomal in natural and having diagnostic testing to determine a specific cause. 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 anopthalmia/micropthalmia, complete atrioventricular canal defect (CAVC), esophageal atresia, 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: 2008-2010

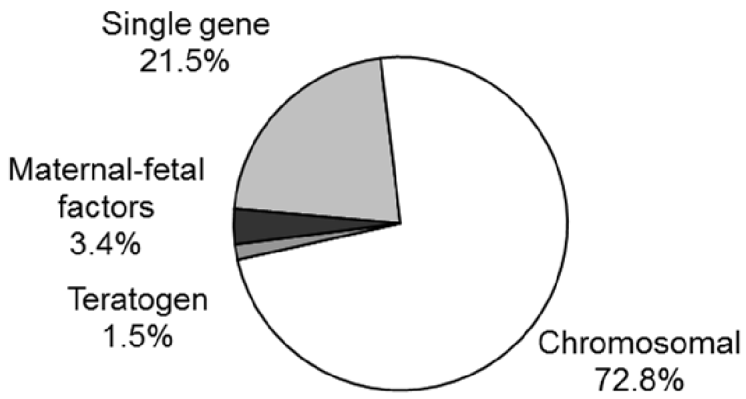
Severity Categories	Percentage of Birth Defects Cases
Severe, supportive measures, usually incompatible with life	2.8%
Serious, may be correctable, most have long-term needs	18.4%
Moderate, most correctable, many have long-term needs	69.5%
Mild, may be correctable, minimal long-term needs	9.2%

Table 15 Pattern and Etiology of Birth Defect Cases, Massachusetts: 2008-2010

Pattern	Description	Cases	
		Known Etiology	Unknown Etiology
Isolated	Solitary defect	8	1,738
	Major and minors (different organ/body parts) or 2 or more defects (same organ/body part)	16	1,045
	Sequence: Common primary defect with consistent, related anomalies	24	150
Multiple Majors	2 or more major defects in different organs/body parts	695	333
Total		743 (18.5%)	3,266 (81.4%)

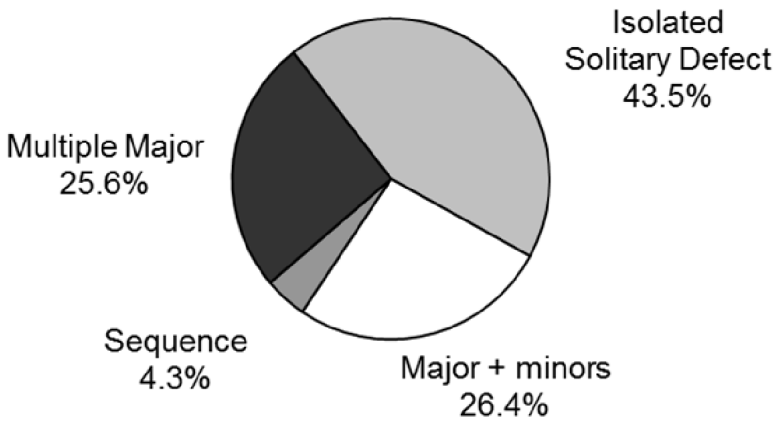
2 cases had an unknown pattern

Figure 17 Causes of Birth Defect Cases with Known Etiology, Massachusetts: 2008-2010



N=743 (18.5% of birth defect cases)
"other" contributes less than 1% and is not pictured the graph

Figure 18 Pattern of Birth Defect Cases with Unknown Etiology, Massachusetts: 2008-2010



N=4012

Appendices

Technical Notes

Definitions

2008-2010 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, 2008-2010

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 2010 Dollars
2010 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.

An 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. As codes have been added to the BDMP's surveillance system, those codes have been added to the severity assignment algorithm. The automated process for 2008-2010 data was able to assign severity levels to the majority of the cases, with the remaining assigned manually by the Center's Clinical Geneticist. The process that included the automated categorization system produced percentages of birth defects within each of the four severity categories in 2008-2010 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.

2008, 2009, and 2010 Populations Used in Calculating Rates

Numbers of Live Births to MA Residents*					
		2008 N=76,969	**2009 N=74,966**	**2010 N=72,835**	**Total N=224,770**
By Maternal Age	<20	4,623	4,528	3,946	13,097
	20-24	12,475	12,048	11,298	35,821
	25-29	19,019	18,469	18,043	55,531
	30-34	23,152	23,143	23,158	69,453
	35+	17,679	16,774	16,385	50,838
	Unknown**	3	4	5	12
By Infant's Sex	Male	39,447	38,534	37,287	115,268
	Female	37,521	36,431	35,546	109,498
	Unknown**	1	1	2	4
By Plurality	Singleton	73,475	71,423	69,508	214,406
	Multiple Birth	3,494	3,543	3,327	10,364
By Maternal Race/Ethnicity	White	51,760	49,759	48,466	149,985
	Black	6,652	6,945	6,794	20,391
	Hispanic	10,895	10,986	10,588	32,469
	Asian/PI	5,958	5,939	5,817	17,714
	American Indian**	145	122	105	372
	Other, non-Hisp**	1,417	1,036	978	3,431
	Unknown**	142	179	87	408
By EOHHS Region	Western	8,952	8,655	8,485	26,092
	Central	10,091	9,828	9,347	29,266
	Northeast	15,969	15,808	15,252	47,029
	Metro West	17,572	16,986	16,669	51,227
	Southeast	14,023	13,458	12,995	40,476
	Boston	10,362	10,231	10,087	30,680

* 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 v3.0r32 7

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

Birth Defect Codes and Exclusions[1] by Defect Category (cont'd)		
Defect	**ICD-9 / BPA[2]**	**NOTES**
Cardiovascular		
Anomalous Pulmonary Venous Connection		
Total/partial anomalous pulmonary venous connection	747.420, 747.430	
Atrioventricular Canal Defects		
ASD primum	745.600	
Common atrium	745.610	
Complete atrioventricular canal defect	745.620, 745.630	
Endocardial cushion defect (OS and NOS)	745.680, 745.690	
VSD, canal type	745.685	
Conotruncal (Outlet) and Aortic Arch		
Double outlet right ventricle	745.185-745.189	
d-Transposition of the great arteries	745.100, 745.110	
Interrupted aortic arch, type B	747.217	
Tetralogy of Fallot w/ and w/o pulmonary atresia	745.200, 747.310	
Truncus	745.000	
Ebstein Anomaly		
Ebstein anomaly	746.200	
Laterality Defects		
Heterotaxy, situs inversus	759.300-759.395	
Left-Sided Obstruction		
Aortic valve stenosis	746.300	
Coarctation of aorta	747.100-747.190	
Hypoplastic left heart syndrome	746.700, 747.200	
Interrupted aortic arch (type A and NOS)	747.215, 747.216	

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

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

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

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

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

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

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2008-2010

BPA Label	BPA Code	# of defects
Central Nervous System		
Anencephaly	740020	13
Meningomyelocele/myelomeningocele, Highest level, thoracic, Arnold Chiari malformation ± hydrocephalus, open lesion	741002	1
Meningomyelocele/myelomeningocele, Highest level, lumbar, Arnold Chiari malformation ± hydrocephalus, ope n lesion	741003	15
Meningomyelocele/myelomeningocele, Highest level, sacral, Arnold Chiari malformation ± hydrocephalus, open lesion	741004	1
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	5
Meningocele, Highest level, lumbar, Arnold Chiari malformation ± hydrocephalus, closed lesion	741113	1
Lipomeningomyelocele, Highest level, sacral, Arnold Chiari malformation ± hydrocephalus, closed lesion	741144	2
Meningomyelocele/myelomeningocele, Highest level, lumbar, Arnold Chiari malformation ± hydrocephalus, unspec. open/closed lesion	741203	1
Meningomyelocele/myelomeningocele, Highest level, sacral, Arnold Chiari malformation ± hydrocephalus, unspec. open/closed lesion	741204	1
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
Meningomyelocele/myelomeningocele, Highest level, sacral, Hydrocephalus, other (aqueduct of Sylvius) or NOS, clos ed lesion	741404	1
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, lumbar, No mentioned hydrocephalus, open lesion	741703	1
Meningomyelocele/myelomeningocele, Highest level, sacral, No mentioned hydrocephalus, open lesion	741704	1
Meningomyelocele/myelomeningocele, Highes t 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, lumbar, No mentioned hydrocephalus, closed lesion	741813	1
Meningocele, Highest level, sacral, No mentioned hydrocephalus, closed lesion	741814	1
Lipomeningomyelocele, Highest level, lumbar, No mentioned hydrocephalus, closed lesion	741843	5
Lipomeningocele, Highest level, lumbar, No mentioned hydrocephal us, closed lesion	741853	2
Lipomeningocele, Highest level, sacral, No mentioned hydrocephalus, closed lesion	741854	2
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	2
Encephalocele, Other Specified (including midline)	742080	1
Encephalocele, Frontal (including proencephalon)	742085	1
Encephalocele, Parietal	742086	2
Encephalocele, NOS	742090	1
Microcephalus	742100	35
S Cerebrum anomalies	742200	3
Corpus callosum anomalies (don't code colpocephaly with ACC)	742210	101
Cerebellum anomalies	742230	11
Cerebellar Hypoplasia	742235	7
Agyria and lissencephaly	742240	5
Microgyria / polymicrogyria	742250	23
Holoprosencephaly, NOS	742260	5
Holoprosencephaly, Alobar	742265	2
Holoprosencephaly, Semilobar	742266	3
Holoprosencephaly, Lobar	742267	5
S Brain, reduction defect OS (8/02 Includes colpocephaly, pachygyria, schizencephaly) & absent septum pellucidum	742280	49
Hydrocephaly, Anomalies of Aqueduct of S ylvius	742300	15
Dandy-Walker Malformation	742310	20
Hydranencephaly	742320	8
Hydrocephaly, Other Specified	742380	16
Hydrocephaly, NOS	742390	23

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2008-2010

BPA Label	BPA Code	# of defects
Enlarged brain and head / enlarged head / enlarged brain / megalencephaly / macrocephaly	742400	22
Brain cysts: Porencephaly / porencephalic	742410	2
S Brain cysts: Cerebral / subependymal / periventricular	742420	5
Brain: Other specified anomalies / cortical atrophy / cranial nerve defects	742480	7
Spinal cord: Diastematomyelia	742520	1
Spinal cord: Hydromyelia / hydrorachis	742540	2
Spinal cord: Other specified anomalies (Includes tethered cord) (and arachnoid cyst)	742580	85
Eye		
Anophthalmos, Right	743002	1
Microphthalmos, Laterality Unk	743100	1
Microphthalmos, Left	743101	15
Microphthalmos, Right	743102	5
Microphthalmos, Unilateral, Side Unk	743103	1
Microphthalmos, Bilateral	743104	12
Buphthalmos/Congenital Glaucoma, Laterality Unk	743200	1
Buphthalmos/Congenital Glaucoma, Left	743201	2
Buphthalmos/Congenital Glaucoma, Right	743202	1
Buphthalmos/Congenital Glaucoma, Bilateral	743204	9
Absence of lens/Congenital Aphakia, Laterality Unk	743300	1
Absence of lens/Congenital Aphakia, Right	743302	1
Cataract, NOS, Left	743321	12
Cataract, NOS, Right	743322	17
Cataract, NOS, Bilateral	743324	16
Coloboma of lens, Laterality Unk	743340	1
Cataract, anterior polar, Left	743351	3
Cataract, anterior polar, Right	743352	2
Cataract, other specified, Left	743361	2
Cataract, other specified, Right	743362	1
Cataract, other specified, Bilateral	743364	3
S Cornea, other anomalies. Excludes: megalocornea (use 743.220)	743410	9
Absence of iris/Aniridia, Laterality Unk	743420	1
Absence of iris/Aniridia, Bilateral	743424	2
S Iris: Coloboma	743430	11
S Iris,other anomalies: polycoria/ectopic pupil (For Peter's SYNDROME, use 759800. For Peter's ANOMALY,use 743460 - 469).Excludes:Brushfield spots	743440	1
Peters Anomaly, Left	743461	1
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	3
S Vitreous humor: Specified anomalies (includes PHPV)	743500	4
S Retina: Specified anomalies / congenital retinal aneurysm. Excludes:Stickler syndrome(use_759.860)	743510	6
S Optic disc: Specified anomalies / hypoplastic optic nerve / coloboma of the optic disc	743520	34
S Choroid: Coloboma	743535	6
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	13
S Anomaly of middle ear / fusion of ossicles	744020	13
S Anomaly of inner ear / congenital anomaly of membranous labyrinth or organ of Corti	744030	16
S Ear: Unspecified anomalies with hearing impairment / congenital deafness, NOS	744090	6
S # Ear : ACCESSORY auricle / polyotia	744100	10
Microtia, Laterality Unk	744210	1
Microtia, Left	744211	14
Microtia, Right	744212	17
Microtia, Bilateral	744214	7

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2008-2010

BPA Label	BPA Code	# of defects
S Ear: Other misshapen ear / cleft / malformed /#POINTED / # ELFIN,pixie -like / # LOP / # CAULIFLOWER / # ABSENT or decreased cartilage -- a conditional exclusion if <36wks	744230	17
S Ear: Other specified anomalies (see also 744.2 30) / #DARWIN tubercle	744280	10
Cardiovascular		
Anomalous Pulmonary Venous Connection		
Total anomalous pulmonary venous return/connection/drainage	747420	20
Partial anomalous pulmonary venous return/connection/drainage	747430	18
Atrioventricular Canal Defects		
Atrial septal defect, primum type (ASD1)	745600	5
Common Atrium	745610	12
Complete atrioventricular canal (CAVC)	745630	67
Endocardial cushion defect, Other specified	745680	30
Ventricular septal defect, inflow type (subtricuspi d, canal-type) (VSDavc)	745685	7
Endocardial cushion defect, NOS	745690	5
Conotruncal (Outlet) and Aortic Arch		
Truncus Arteriosus	745000	6
Dextro-transposition of great arteries (dTGA, dTGV) w/ intact ventricular septum	745100	29
Dextro-transposition of great arteries (dTGA, dTGV) w/ ventricular septal defect	745110	26
Double-outlet right ventricle (DORV) with normally related great arteries	745185	9
Double-outlet right ventricle (DORV) with transposed great arteries	745186	8
Double-outlet right ventricle (DORV), Other Specified	745188	4
Double-outlet right ventricle (DORV), NOS	745189	3
Tetralogy of Fallot	745200	65
Interrupted aortic arch, type B	747217	8
Pulmonary atresia with VSD (tetralogy of Fallot with pulmonary atresia)	747310	25
Ebstein Anomaly		
Ebstein Malformation or Anomaly	746200	8
Heterotaxy (Laterality Defects)		
Complete situs inversus w/ dextrocardia	759300	10
Situs inversus w/ levocardia	759310	1
Situs inversus abdominis	759330	1
Situs ambiguus, right; right isomerism	759350	3
Situs ambiguus, left; left isomerism	759360	3
Situs ambiguus, sidedness NOS	759380	18
Heterotaxy, NOS	759395	1
Left-Sided Obstruction		
Aortic stenosis, valvar	746300	31
Hypoplastic left heart syndrome	746700	30
Coarctation of the aorta (COA), preductal (proximal)	747100	1
Coarctation of the aorta (COA), postductal (distal)	747110	1
Coarctation of the aorta, juxtaductal	747120	12
Coarctation of the aorta, NOS	747190	83
Aorta: Atresia / absence	747200	1
Interrupted aortic arch, type A	747216	1
Other Cardiovascular		
Aortic septal defect / aortopulmonary window. Excludes: atrial septal defect(use 745.590)	745010	3
Pulmonary valve: Other specified anomalies. Excludes: infundibular PS (746.830)	746080	31
Tricuspid stenosis or hypoplasia *added 10/12*	746106	3
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	97
Aortic valve: Other specified anomalies / aortic valve atresia. Excludes: supravalvular aortic stenosis(747.220)	746480	29
Aortic valve: Unspecified anomalies	746490	1
Mitral valve: Congenital mitral stenosis	746500	15

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BPA Label	BPA Code	# of defects
Mitral valve: Absence, atresia, or hypoplasia	746505	12
Mitral valve: insufficiency or regurgitation, congenital/# MILD', MINIMAL','TRIVIAL', or 'PHYSIOLOGIC' ~	746600	3
Situs: Dextrocardia without situs inversus / dextrocardia with situs solitus. Excludes: dextrocar dia with situs inversus (use_759.300)	746800	7
Cor triatriatum	746820	3
Pulmonary infundibular (subvalvular) stenosis	746830	21
Heart: Other specified anomalies / ectopia cordis / mesocardia / conduction defects, NOS	746880	126
Hypoplastic left ventricle. Excludes: hypoplastic left heart syndrome (746.700)	746881	7
Hypoplastic right heart or right ventricle / Uhl's disease (parchment RV)	746882	8
Anomalies of coronary artery or sinus	746885	27
Valves: Unspecified anomalies	746900	1
"Pulmonic" or pulmonary atresia, stenosis, or hypoplasia, NOS w/ no mention of whether valve or artery	746995	2
Aorta: Hypoplasia	747210	33
Supra-aortic stenosis / supravalvular aortic stenosis. Excludes: aortic stenosis, congenital(see 746.300)	747220	5
Aorta: Persistent right aortic arch	747230	54
Aorta: Vascular ring / double aortic arch / vascular ring compression of trachea	747250	12
Aorta: Congenital aneurysm / dilatation	747270	7
Aorta: Other specified anomalies	747280	1
S Pulmonary artery: stenosis. Use 746.995 if artery or valve is not specified	747320	19
S Pulmonary artery: other specified / pulmonary artery hypoplasia	747380	12
Persistent left superior vena cava	747410	62
Great veins: Other specified anomalies (includes IVC interr uption, bilateral SVC)	747480	43
S Peripheral arteries: Other anomalies / aberrant subclavian artery	747640	44
S Peripheral vascular system:Other anomalies / primary pulmonary artery hypertension	747680	1
Arteriovenous malformation or aneurysm of brain	747800	1
Cerebral vessels: Other anomalies / vein of Galen	747810	2
S Circulatory system: Other specified anomalies. Excludes cong aneurysms: coronary ~ (746.880),peripheral ~ (747.640), pulmonary~ (747.330), retinal ~(743.510), ruptured cereb ral arterioven	747880	12
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	307
Right-Sided Obstruction		
Pulmonary valve atresia/intact ventricular septum	746000	14
Pulmonic stenosis, valvar	746010	156
Pulmonary valve atresia with VSD (not TOF variant 747.310)	746030	6
Tricuspid atresia	746100	17
Septal Defects		
Ventricular septal defect, Perimembranous (type II, membranous) (VSDmem)	745485	257
Ventricular septal defect, Malalignment-type (type I, subarterial) (VSDmal)	745487	40
Ventricular septal defect, NOS	745490	17
Atrial septal defect, Secundum type (ASD2)	745510	422
Atrial septal defect, OS	745580	3
Atrial septal defect, NOS	745599	70
Single Ventricle and L-TGA		
L-TGA /Corrected transposition of great vessels / ventricular inversion. Excludes: dextrocardia (use 746800)	745120	7
Single ventricle, NOS	745300	4
Single ventricle, Double Inlet Left Ventricle	745310	3
Single ventricle, Double Inlet Right Ventricle	745320	2
Respiratory		
S Choanal stenosis (For NBDPS: choanal atresia = 748.010 etc)	748000	2
Choanal atresia, Left	748011	2
Choanal atresia, Right	748012	6
Choanal atresia, Bilateral	748014	8
Nose: Agenesis or underdevelopment	748100	6

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BPA Label	BPA Code	# of defects
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	5
S Stenosis of bronchus	748340	1
S Other anomalies of bronchus	748350	1
Larynx: Cleft / laryngotracheoesophageal cleft / 1/04: use for laryngeal atresia/stenosis	748385	12
S Lung cysts: Single	748400	1
S Lung cysts: CCAM (cong cystic adenomatoid malf), Other specified	748480	24
S Lung agenesis or aplasia	748500	1
S # HYPOPLASIA of lung or pulmonary hypoplasia -- a conditional exclusion only in infants <36wks.	748510	3
S Lung: sequestration	748520	13
Lung: Bilobed right / right lung with left lung bronchial pattern	748625	3
S Lung: Other and unspecified anomalies	748690	4
S Respiratory system: Other specified anomalies / congenital lobar emphysema / lymphangiectasia of lung	748880	4
Orofacial		
Pierre Robin sequence (not a true "syndrome")	524080	43
S Branchial cleft, sinus, fistula, cyst, or pit	744400	47
S Other branchial cleft anomalies / dermal sinus of head	744480	21
S Face or neck: Other specified anomalies (6/03 eg. facial cleft)	744880	8
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	34
Cleft hard palate, Bilateral	749010	8
Cleft hard palate, Central	749020	5
Cleft hard palate, NOS	749030	23
Cleft soft palate, Bilateral	749050	2
Cleft soft palate, Central	749060	4
Cleft soft palate, NOS	749070	64
Cleft palate, NOS	749090	18
Cleft lip, Unilateral, Left	749101	29
Cleft lip, Unilateral, Right	749102	19
Cleft lip, Bilateral	749110	4
Cleft lip, Central	749120	1
S Cleft: Incomplete CL/ microform /pseudo / fused lip /healed lip	749190	7
Cleft lip, NOS	749195	4
Cleft lip and palate, Unilateral cleft lip, Left	749201	37
Cleft lip and palate, Unilateral cleft lip, Right	749202	30
Cleft lip and palate, Bilateral cleft lip	749210	41
Cleft lip and palate, Central cleft lip	749220	2
Cleft lip and palate, NOS	749290	8
Tongue: large / macroglossia	750120	7
Orofacial		
Pierre Robin sequence (not a true "syndrome")	524080	43
S Branchial cleft, sinus, fistula, cyst, or pit	744400	47
S Other branchial cleft anomalies / dermal sinus of head	744480	21
S Face or neck: Other specified anomalies (6/03 eg. facial cleft)	744880	8
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	34
Cleft hard palate, Bilateral	749010	8
Cleft hard palate, Central	749020	5

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BPA Label	BPA Code	# of defects
Cleft hard palate, NOS	749030	23
Cleft soft palate, Bilateral	749050	2
Cleft soft palate, Central	749060	4
Cleft soft palate, NOS	749070	64
Cleft palate, NOS	749090	18
Cleft lip, Unilateral, Left	749101	29
Cleft lip, Unilateral, Right	749102	19
Cleft lip, Bilateral	749110	4
Cleft lip, Central	749120	1
S Cleft: Incomplete CL/ microform /pseudo / fused lip /healed lip	749190	7
Cleft lip, NOS	749195	4
Cleft lip and palate, Unilateral cleft lip, Left	749201	37
Cleft lip and palate, Unilateral cleft lip, Right	749202	30
Cleft lip and palate, Bilateral cleft lip	749210	41
Cleft lip and palate, Central cleft lip	749220	2
Cleft lip and palate, NOS	749290	8
Tongue: large / macroglossia	750120	7
Gastrointestial		
Esophageal atresia without TE fistula	750300	6
Esophageal atresia with TE fistula	750310	39
Tracheoesophageal fistula without mention of esophageal atresi a	750320	2
Tracheoesophageal fistula, "H" type	750325	4
Microgastria	750700	1
Persistent omphalomesenteric duct / persistent vitelline duct	751000	3
# MECKEL'S diverticulum	751010	19
Intestinal atresia/stenosis, Duodenum	751100	38
Intestinal atresia/stenosis, Jejunum	751110	20
Intestinal atresia/stenosis, Ileum	751120	12
Intestinal atresia/stenosis, Large Intestine, NOS	751200	2
Rectal atresia/stenosis without mention of fistula	751220	5
Anal atresia with fistula	751230	38
Anal atresia without mention of fistula	751240	24
Hirschsprung disease: Long-segment (aganglionosis beyond rectum)	751310	29
Hirschsprung disease: Short-segment(aganglionosis involving no more than the anal sphincter and the rectum)	751320	9
Hirschsprung disease, NOS	751330	14
Malrotation: cecum and/or colon	751400	1
Anomalies of mesentery	751410	1
Congenital adhesions or bands of omentum and peritoneum / Ladd's bands	751420	2
Malrotation: Other specified and unspecified	751490	79
Malrotation: small intestine alone	751495	2
Duplication of anus, appendix, cecum, or intestine / enterogenous cyst	751500	12
Microcolon	751520	12
Ectopic (displaced, anteriorly placed) anus	751530	14
Congenital anal fistula	751540	2
Duodenal web	751560	1
Intestine: Other specified anomalies / # RECTAL fissures	751580	6
Cystic or fibrocystic disease of liver	751610	3
Other anomalies of liver / # HEPATOMEGALY / # HEPATOSPLENOMEGALY (also use code 759.020). Excludes: Budd -Chiari syndrome (453.000)	751620	9
Agenesis or hypoplasia of gallbladder	751630	1
Other anomalies of gallbladder / duplication of gall bladder	751640	1
Biliary atresia, extrahepatic or NOS (use 751.670 for intrahepatic)	751650	14
Choledochal cysts	751660	3
Other anomalies of hepatic or bile ducts	751670	3

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BPA Label	BPA Code	# of defects
Pancreas: Annular	751720	8
Pancreatic cyst. Excludes: fibrocystic disease of pancreas (277.000)	751740	1
Genitourinary		
Cloacal exstrophy	751550	2
Gyne: S Ovaries absence or agenesis	752000	3
Gyne: S Ovaries, Other specified anomalies	752080	4
Gyne: S Ovaries, Multiple cysts	752085	7
Gyne: S Fallopian tube or broad ligament, absent	752100	1
Gyne: Uterus, other anomalies / bicornuate/ unicornis	752380	2
Gyne: Vagina, absence or atresia complete or partial	752410	1
Gyne: Hymen # IMPERFORATE	752430	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	7
Hypospadias, Second Degree	752606	111
Hypospadias, Third Degree	752607	15
Hypospadias, Second Degree with Chordee	752626	125
Hypospadias, Third Degree with Chordee	752627	59
Indeterminate sex, NOS / ambiguous genitalia	752790	20
Testis and scrotum: Other anomalies / polyorchidism / bifid scrotum. Excludes: torsion of the testes or spermatic cord(608.200)	752820	25
Penis: Other anomalies / concealed penis / absent or hooded foreskin / #REDUNDANT foreskin (Redundant foreskin is never coded.)	752860	82
Penis: Small / hypoplastic / micropenis	752865	15
Genital organs: Other specified anomalies / microgenitalia / macrogenitalia	752880	2
Renal agenesis, bilateral	753000	9
Renal hypoplasia, bilateral	753005	2
S Kidney/renal: absence, agenesis, dysplasia, or hypoplasia, NOS	753009	11
S Kidney/renal: cyst, single	753100	12
Kidneys: Polycystic, infantile type (IPKD)	753110	11
Kidneys: Polycystic, adult type (APKD)	753120	1
S Kidneys: Multicystic renal dysplasia / multicystic kidney	753160	55
Kidney: Other specified disease / cystic NOS	753180	2
S Congenital hydronephrosis / pyelocaliectasis	753200	327
S Atresia, stricture, or stenosis of ureter / ureteropelvic junction obstruction or stenosis /ureterovesical junction obstruction or stenosis / hypoplastic ureter	753210	80
S Megaloureter, NOS / hydroureter	753220	58
S Other and unspecified obstructive defects of renal pelvis and ureter	753290	1
S Kidney: Double or triple, pelvis / pyelon duplex or triplex	753310	19
S Kidney: Lobulated, fused, or horseshoe / crossed fused ectopia	753320	9
S Kidney: Ectopic / pelvic	753330	10
S Kidney: Enlarged, hyperplastic, or giant	753340	1
S Kidney: Other specified anomalies	753380	3
S Ureter: Accessory / double ureter / duplex collecting system	753410	50
S Ureter: Ectopic	753420	13
S Ureter: Other specified anomalies / ureterocele	753480	35
S Ureter: Variations of vesicoureteral reflux	753485	119
Bladder exstrophy	753500	4
Urethra: Congenital posterior urethral valves or posterior urethral obstruction	753600	33
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	6
# Urachus: PATENT	753700	5
Urachus: Cyst	753710	14
Urachus: Other and unspecified anomaly	753790	1
Absence of bladder or urethra	753800	5

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BPA Label	BPA Code	# of defects
Ectopic bladder	753810	1
Congenital diverticulum or hernia of bladder	753820	2
Double urethra or urinary meatus	753840	3
Other specified anomalies of bladder and urethra	753880	5
Musculoskeletal		
Certain cong musculoskeletal anomalies face, face, jaw: Use for asymmetry of face	754000	7
Congenital postural scoliosis	754200	21
S Bowing, femur	754400	2
S Bowing, tibia and/or fibula	754410	4
S Genu recurvatum	754430	1
S Dislocation of knee, congenital	754440	4
S Clubfoot: Talipes equinovarus	754500	138
S Clubfoot: Talipes calcaneovarus	754510	3
S Clubfoot: # METATARSUS varus or adductus	754520	27
S Clubfoot: Complex varus deformities	754530	4
S Unspecified varus deformities of feet	754590	2
S Talipes calcaneovalgus	754600	14
S Other specified valgus deformities of foot	754680	1
S Unspecified valgus deformities of foot	754690	1
S Clubfoot, NOS / talipes, NOS	754730	145
S Other specified deformities of ankle and / or toes / dorsiflexion of foot. Excludes: widely spaced first and second toes (use_755.600)	754780	5
S Other specified deformity of hands (see 755.500 for specified anomalies of fingers)	754880	1
S Polydactyly fingers / postaxial polydactyly, Type A	755005	88
S Polydactyly: Accessory thumbs (preaxial polydactyly)	755010	81
S Polydactyly: Accessory toes (postaxial)	755020	78
S Polydactyly: Accessory big toe (preaxial)	755030	16
S Polydactyly: Accessory digits hand, NOS (preaxial, postaxial not specified)	755095	7
S Polydactyly: Accessory digits foot, NOS (preaxial, postaxial not specified)	755096	17
S Syndactyly: Fused fingers	755100	37
S Syndactyly: Webbed fingers	755110	22
S Syndactyly: Fused toes	755120	36
S Syndactyly: Webbed toes / # WEBBING between the second and third toes.	755130	68
S Syndactyly: Unspecified (webbed vs. fused) thumb and / o r fingers, NOS	755193	13
S Syndactyly: Unspecified toes	755194	7
S Syndactyly: Unspecified (webbed vs. fused) Toes	755199	9
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	2
Absence of hand or fingers, Laterality Unk	755245	1
Absence of hand or fingers, Left	755246	11
Absence of hand or fingers, Right	755247	9
Absence of hand or fingers, Bilateral	755249	5
Longitudinal deficiency of arm, NOS, Bilateral	755254	1
Split-Hand, Left	755256	3
Split-Hand, Right	755257	3
Split-Hand, Bilateral	755259	3
Thumb only missing or hypoplastic, Left	755261	3
Thumb only missing or hypoplastic, Right	755262	5
Thumb only missing or hypoplastic, Bilateral	755264	2
Radial aplasia/hypoplasia, Left	755266	2
Radial aplasia/hypoplasia, Right	755267	5
Radial aplasia/hypoplasia, Bilateral	755269	1
Ulnar aplasia/hypoplasia, Right	755272	1

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BPA Label	BPA Code	# of defects
Ulnar aplasia/hypoplasia, Bilateral	755274	1
Transverse deficiency or amputation of the leg, NOS, Bilateral	755304	1
Absence of thigh only (lower leg and foot present), Left	755331	1
Absence of the lower leg and foot, Left	755341	1
Absence of foot or toes, Left	755346	3
Absence of foot or toes, Right	755347	7
Absence of foot or toes, Bilateral	755349	6
Split-Foot, Left	755356	1
Split-Foot, Right	755357	2
Split-Foot, Bilateral	755359	4
Tibial aplasia/hypoplasia, Right	755367	2
Tibial aplasia/hypoplasia, Bilateral	755369	2
Fibular aplasia/hypoplasia, Left	755371	1
Fibular aplasia/hypoplasia, Right	755372	2
S Anomalies of fingers /camptodactyly/macro - /brachy-/clino-, triphalangeal thumb. Excludes:acrocephalosyndactyly(see756.050) /Apert synd(see756.055)	755500	50
S Radioulnar synostosis	755536	3
S Anomalies of elbow and upper arm	755540	4
S Upper limb: Other specified anomalies / hyperextensibility of upper limb / shortening of upper limb	755580	2
S Upper limb: Hypoplasia / Fingers, hands, or arms: hypoplasia. Excludes: aplasia or absent upper limb (see 755.2)	755585	25
S Knee: anomalies / hyperextended knee	755640	1
S Upper leg: anomalies / anteversion of femur	755650	1
S Lower limb: other specified anomalies / hyperextended legs / shortening of legs	755680	4
S Lower limb: hypoplasia / Toes, feet, legs: hypoplasia. Excludes: aplasia of or absent lower limb (see 755.3)	755685	14
= Arthrogryposis multiplex congenita / distal arthrogryposis syndrome. Temporarily includes: one or more flexion contrac tures of individual joints	755800	21
Craniosynostosis, Sagittal	756005	70
Craniosynostosis, Metopic	756006	18
Craniosynostosis, Coronal, Left	756011	4
Craniosynostosis, Coronal, Right	756012	13
Craniosynostosis, Coronal, Bilateral	756014	8
Craniosynostosis, Lambdoidal, Left	756021	3
Craniosynostosis, Lambdoidal, Right	756022	4
Craniosynostosis, Lambdoidal, Bilateral	756024	3
Skull and face bone: Other specified anomalies / localized skull defects / mid -facial hypoplasia / prominent maxilla/hypotelorism / # FLATocciput / # PROMINENT occiput.	756080	9
Klippel-Feil syndrome / Wildervanck syndrome	756110	1
Spine: Kyphosis / kyphoscoliosis	756120	16
Vertebrae, cervical: anomalies	756140	8
Vertebrae, cervical: hemivertebrae	756145	5
Vertebrae, thoracic: anomalies	756150	20
Vertebrae, thoracic: hemivertebrae	756155	21
Vertebrae, lumbar: anomalies	756160	10
Vertebrae, lumbar: hemivertebrae	756165	9
Vertebrae, sacrococcygeal: anomalies / agenesis of sacrum. Excludes: pilonidal sin us (see 685.100)	756170	27
Sacral agenesis	756175	3
Vertebrae: Other specified anomalies	756180	4
Vertebrae: Hemivertebrae, NOS	756185	2
Unspecified anomalies of spine	756190	1
S Ribs: Absence	756300	24
S Ribs: Fused	756320	14
S Ribs: Extra	756330	15
S Ribs: Other anomalies	756340	17
Sternum: Other anomalies / double ossification center in manubrium / bifid/ short	756380	3

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BPA Label	BPA Code	# of defects
= Achondroplasia	756430	9
= Thanatophoric dwarfism	756447	4
= Other specified chondrodystrophy. Excludes: Conradi's (use 756.575)	756480	5
= Osteogenesis imperfecta	756500	4
= Infantile cortical hyperostosis / Caffey syndrome	756530	2
= 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	5
Diaphragmatic hernia, NOS (inc. absent/hemidiaphragm), Left	756601	29
Diaphragmatic hernia, NOS (inc. absent/hemidiaphragm), Right	756602	4
Diaphragmatic hernia, Bochdalek, Left	756611	13
Diaphragmatic hernia, Morgagni, Right	756617	3
Diaphragmatic hernia, Morgagni, Unilat, Side Unk	756618	1
Diaghragmatic hernia, Morgagni, Bilateral	756619	1
S Diaphragm: Eventration	756620	10
Omphalocele	756700	26
Gastroschisis	756710	76
Prune belly syndrome	756720	2
Poland syndrome or anomaly	756800	7
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		
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	4
= 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
= Nail-patella syndrome	756830	1
= Specified syndromes, not elsewhere classified, involving skin anomalies	757300	1
= Down syndrome: trisomy 21	758000	277
= Down syndrome: translocation 21, duplication 21q, Robertsonian translocation, isochrome 21q	758020	6
= Down syndrome: mosaic	758040	5
= Down syndrome: diagnosed clinically, but no karyotype report in medical record	758090	1
= Trisomy 13 (archaic Patau syndrome): cytogenetics result in record	758100	9
= Trisomy 13: no cytogenetics in record	758190	1
= Trisomy 18 (archaic Edwards syndrome): cytogenetics result in record	758200	37
= Trisomy 18: translocation trisomy with duplication 18q	758220	1
= Deletion 21q, monosomy 21, or a G-group NOS (archaic)	758300	3
= Deletion 5p / Cri du chat syndrome	758310	5
= Deletion 13q / deletion of long arm of 13	758330	2
= 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	23
= 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	40
= Deletion: unspecified autosome	758390	1
= Trisomy 8	758500	3
= Trisomy: 6, 7, 9, 10, 11, 12 / Other trisomy C (archaic)	758510	5
= Trisomy, partial / 8/02 "partial trisomy" = "duplication". But, for "dup NOS" use 758930	758530	33
= 1/06 Clarified: "Other Trans" Incl Unbal AND Other Bal Translocations, OS. Excludes bal trans in normal (758.400)	758540	21

ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2008-2010

BPA Label	BPA Code	# of defects
= Autosome OS: Other spec anomalies / marker / 8/02: Ring, derivative, mosaic, is ochrome, "additional" material / 3/03 inversions. 2/08 Never code "pericentric inv 9"	758580	25
= Triploidy	758586	1
= Turner phenotype: karyotype 45,X [XO] Note: The 7586xx code series that follows excludes pure gonadal dysgenesis(752.720)	758600	17
= Turner phenotype: variant karyotypes, eg. isochromosome, mosaic (eg X, XX,XY), partial X deletion, ring X chromosome. Excludes: Turner phenotype with normal karyotype	758610	5
= Klinefelter syndrome: 47, XXY	758700	6
= Klinefelter phenotype: other karyotype with additnl X chrome, e.g., XXXY, XXYY, XXXXY	758710	2
= Mosaic XO/XY, 45X/46XY. Excludes: with Turner phenotype(758.610)	758800	1
= Mosaic including XXXXY, 49XXXXY. Excludes: with Klinefelter phenotype (use 758.710)	758830	1
= XYY, male / 47,XYY / mosaic XYY male	758840	4
= XXX female / 47XXX / Triple X syndrome	758850	14
= Additional sex chromosomes, NOS	758860	1
= Sex chromosome: Other specified anomaly / fragile X	758880	6
= Unspecified chromosome: Deletion of c hromosome(s), NOS	758920	1
Other specified DNA based diagnosis	758999	4
= Tuberous sclerosis / Bourneville's disease	759500	7
= Sturge-Weber syndrome/ Encephalocutaneous angiomatosis/	759610	3
= Malf. Syndromes/face: Aarskog /BOF /BOR /Fraser /Fr eemanSheldon / Kabuki / Miller-Dieker/ Noonan /Opitz G / oral-facial-digita/ Oto-palato-digital / Septo-optic dysplasia / Waardenburg / Williams	759800	28
= Malf. Syndromes/short stature: Smith-Lemli-Optiz /de Lange / Cockayne / Laurence-Moon-Biedl / Russell-Silver / Seckel	759820	6
= Malf. Syndromes/limbs: Baller-Gerold/ Carpenter / caudal regression /Fryns/ Holt-Oram / Klippel-Trenaunay-Webe/ LimbBodyWall /Roberts/ Rubinstein-Taybi / sirenomelia / thrombocytopenia-absent radius	759840	4
= Malf. Syndromes/other skeletal: Marfan / Stickler/ Beemer Langer	759860	8
= Malf. Syndromes/metabolic: Alagille /Alport / Beckwith-Wiedemann / Johansen-Blizzard/ leprechaunism / Lowe/ Menkes(kinky hair) /Prader-Willi/ Zellweger	759870	29
= 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	45
DiGeorge S (10/02: Use for specific phenotype with chrome/FISH 22q, i f available 758.370)	279110	5
Other		
Amniotic band sequence	658800	21
= Collodion baby	757110	5
= Other and unspecified ichthyosis	757190	7
= X-linked ichthyosis	757196	5
Ichthyosiform erythroderma	757197	5
= 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	4
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	9
Spleen: Absence / asplenia	759000	6
Spleen: Accessory / 8/02 Use for polysplenia, though not exactly the same	759040	11
Spleen: Other specified anomalies	759080	3
Thyroglossal duct anomalies / thyroglossal cyst	759220	1
Anomalies of thymus / absent thymus / # T HYMICHYPERTROPHY	759240	7
Multiple congenital anomalies (In MA, ="MCA NOS", not "MCA no specific dx") / anomaly, multiple, NOS / deformity, multiple, NOS	759700	1

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

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

EOHHS
Executive Office of Health and Human Services for the Massachusetts Department of Public Health

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 beatin g 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 10^{th} 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.

Glossary of Selected Birth Defect Terms[1]

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.

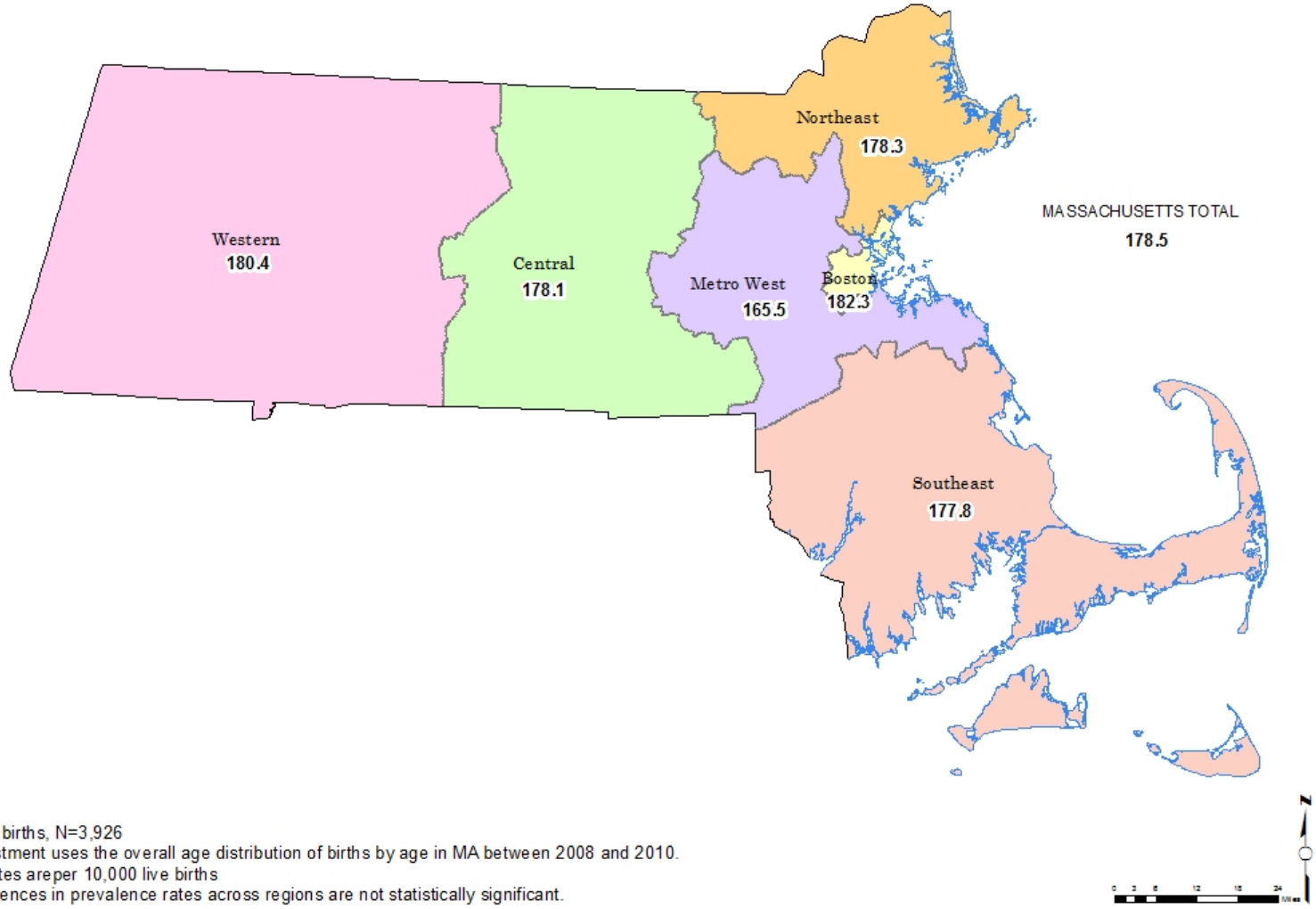
Trisomy 21: See **Down syndrome**.

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, 2008-2010*



*Live births, N=3,926
Adjustment uses the overall age distribution of births by age in MA between 2008 and 2010.
All rates areper 10,000 live births
Differences in prevalence rates across regions are not statistically significant.

Frequency of Daily Multivitamin Use in the Month Prior to Pregnancy by Socio-demographic Characteristics, Massachusetts, 2009[1]

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 2004 [1]

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

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