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# Massachusetts Birth Defects

## 2013-2014

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Massachusetts Birth Defects Monitoring Program  
Massachusetts Center for Birth Defects Research and Prevention  
Bureau of Family Health and Nutrition

Massachusetts Department of Public Health

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This report can be found on our website: [www.mass.gov/dph/birthdefects](http://www.mass.gov/dph/birthdefects)

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## **EXECUTIVE SUMMARY**

One in 33 infants in the United States is born with a birth defect. Birth defects are defined as conditions that develop before birth affecting the structure of one or more parts of the body (1,2). Although birth defects are rare when compared to other adverse pregnancy outcomes like low birth weight or prematurity, birth defects are the leading cause of death in the first year of life. Nationally, about 20% of all infant deaths are attributable to birth defects. Birth defects may also result in mental and/or physical disability, may require costly medical care, and may cause economic, emotional, and social distress for families.

The causes of many birth defects are poorly understood. Certain genetic and environmental factors have been implicated in selected birth defects. These include prenatal environmental factors, such as infections, exposures to medications or other chemicals, drug or alcohol abuse, and nutritional deficiencies. Some birth defects can be caused by a single abnormal gene, while others arise due to a complex interplay between various genetic and environmental factors.

Studies have shown that the presence of adequate amounts of folic acid (vitamin B9) in the mother's system before conception and during the first trimester may help prevent birth defects of the brain and spinal cord known as neural tube defects (3). However, for more than 70% of all birth defects, no known cause has been identified (4). Researchers continue to investigate a wide variety of risk factors as possible causes.

The combined lifetime cost for infants born with 12 major structural birth defects in Massachusetts has been estimated at over 200 million dollars (5). Nationally, the lifetime cost of 17 common birth defects has been estimated to be over 9 billion in 2012 dollars (6).

This report presents data on the prevalence of birth defects in Massachusetts during the years 2013 and 2014. Two years of data are combined in this report, since the numbers of cases are often small for individual defects within a single year.

The Massachusetts Birth Defects Monitoring Program (BDMP) has data going back to 2000, allowing for some analysis of trends. Increasing rates over time reflect improved case ascertainment, the addition of new defects collected, and the collection of data on other pregnancy losses, which include terminations and early miscarriages. The program is constantly working to improve case-finding, in order to provide the most complete data possible to inform public health policy, planning and prevention efforts.

### **Prevalence of Birth Defects in Massachusetts**

Among 143,662 Massachusetts residents who delivered in 2013 or 2014, there are 3,894 cases (3,015 live births, 68 stillbirths, and 811 other pregnancy losses) with one or more structural birth defects. This results in an overall prevalence rate of 271 per 10,000 live births.

Among live births and stillbirths, cardiovascular defects are the most commonly occurring birth defects in Massachusetts, followed by musculoskeletal, genitourinary and chromosomal defects. When other pregnancy losses are included, then chromosomal birth defects become the most common. Specifically, trisomy 21 (Down syndrome) is the most common birth defect when all birth outcomes are included, followed by atrial septal defect (a cardiovascular defect).

## **Birth Defects in Massachusetts vs. the United States**

Massachusetts is one of 11 states with an active case ascertainment program and contributes data to published national prevalence estimates for selected birth defects. For most defects, Massachusetts rates in 2013-2014 are similar to the most recent published national rates from 2004-2006 (7). Massachusetts rates are significantly higher for trisomy 21, trisomy 13 and trisomy 18, even after age-adjustment, likely because of the inclusion of early miscarriages in Massachusetts. Massachusetts rates of omphalocele are also higher than national rates. Differences in surveillance system methodology, types of pregnancy outcomes included, and demographic variation may also contribute to the differences in rates for certain birth defects.

Until 2011, Massachusetts only collected information on live birth and stillbirth cases. Limiting the data to live births and stillbirths can result in undercounting of certain birth defects—especially those not compatible with life. Beginning in early 2011, Massachusetts began ascertaining prenatally-diagnosed birth defects in pregnancies that ended in other types of pregnancy losses. When Massachusetts data was limited to live births and stillbirths, rates of spina bifida and anencephaly were lower than national estimates, but in the current report, which includes these other pregnancy losses, the rates are similar.

## **Adverse Pregnancy Outcomes**

Live born infants with birth defects are more likely to have adverse outcomes such as low birth weight, prematurity, and small for gestational age (SGA) than those without birth defects. Infants with a birth defect are more than twice as likely to have low birth weight (less than 2500 grams) and to be born premature (before 37 weeks) compared to those without birth defects. Cesarean (C-section) deliveries are also more common among live born infants with birth defects than among unaffected infants.

## **Maternal Age**

The prevalence of birth defects varies by maternal age. The number of births to older mothers has been increasing over time in Massachusetts (8). Older mothers have a higher prevalence of birth defects compared to younger mothers. Overall birth defect rates were highest for mothers ages 35 years and older (350.7 per 10,000 live births).

There is a strong association between the rate of Down syndrome (trisomy 21) and advanced maternal age. In Massachusetts, the rate of Down syndrome in mothers ages 35 and older is 68 per 10,000 live births, over 5 times greater than the rate in mothers younger than 35.

Gastroschisis, a condition in which a child is born with the intestines protruding through a hole in the abdominal wall, occurs more often among younger mothers. In 2013-2014, mothers 20-24 years old had the highest rate of gastroschisis (14.3 per 10,000 live births), while the rate among mothers ages 35 and older was 0.6 per 10,000.

### **Assisted Reproductive Technology**

It is estimated that 1.5 percent of United States infants are conceived through the use of assisted reproductive technology (ART) (9). In 2013, Massachusetts had one of the highest rates of ART use in the nation. This may be partly due to high rates of insurance coverage for ART in Massachusetts and a higher proportion of older women of reproductive age trying to conceive. Infants conceived by ART have been shown to have an increased risk for certain birth defects compared to those conceived spontaneously (10).

### **Multiple Births**

Birth defects are significantly more common among multiple births (e.g., twins and triplets) than in singleton births. Approximately 4.3% of Massachusetts live births are multiple births (8). The birth defect prevalence rate in 2013-2014 is 265.6 per 10,000 live births for singletons and 385.2 per 10,000 live births for multiples.

### **Maternal Race/Ethnicity**

Birth defect rates may vary by maternal race and ethnicity. In 2013-2014 in Massachusetts, the overall age-adjusted prevalence rate of birth defects does not significantly differ by race/ethnicity, although as in previous years, Asians tend to have lower rates of birth defects.

### **Region**

The Massachusetts Executive Office of Health and Human Services divides the state into six regions, which are used for statistical, care coordination, and administrative purposes. In 2013-2014, the overall age-adjusted birth defect prevalence rate did not substantially differ by region, although the Metro West and Southeastern regions had the lowest rates. The end of medical record abstraction at Rhode Island hospitals may impact the Southeastern region rates.

### **Etiology and Pattern**

The surveillance system in Massachusetts collects information on etiology (cause), whenever available. The majority of birth defect cases in 2013-2014 had an unknown cause, which is consistent with the published research.

Pattern refers to whether a birth defect occurs with other defects. Approximately 34% of birth defects occurred with defects in other organs or as part of syndromes, while 66% occurred as isolated defects or were confined to the same organ, occurred with minor defects, or were part of a sequence of developmental events.

## **Analysis of Trends**

The overall prevalence of birth defects among live births and stillbirths in Massachusetts increased from 187.5 to 214.6 per 10,000 live births between 2011-2012 and 2013-2014. This increase is partly related to collecting more types of birth defects starting in 2014. Adding additional birth outcomes (other types of pregnancy losses) beginning in 2012 also contributed to an increase in birth defect prevalence rates.

This report includes selected trend analyses using current and previous years' data, with the understanding that there have been modifications to the surveillance system that may contribute to increases in birth defect rates, including expanded and improved case ascertainment, the addition of other pregnancy losses, and improved prenatal diagnosis.



# **CHAPTER 1: INTRODUCTION**

## **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 (1). Birth defects, or congenital anomalies, are serious abnormalities of body structure present before birth.

Although birth defects are rare when compared to other adverse pregnancy outcomes, they are the leading cause of infant death in the United States. Nationally, about 20% of all infant deaths result from birth defects (11), which is consistent with the number of infant deaths among cases found in the Massachusetts Birth Defects Monitoring Program surveillance system and with a recent report using Massachusetts death data from vital records (12).

Birth defects may cause significant physical or mental disability. There can be substantial costs to those affected and to their families, including direct costs of medical treatment, developmental services and special education, as well as indirect costs related to loss of work and productivity.

## **Causes of Birth Defects**

The causes of most birth defects are poorly understood, but certain genetic and environmental factors have been reported to be 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 birth defects, no known cause has been identified (4). Researchers are looking at a wide variety of environmental exposures and other possible risk factors as potential causes of birth defects.

## **Birth Defects and Folic Acid**

Studies have shown that the presence of adequate amounts of folic acid (vitamin B9) in the mother's system during the "periconceptional" period (one month before through three months after conception) may help prevent defects of the brain and spinal cord known as neural tube defects. 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 United States (13). However, some studies in certain populations suggest that not all cases of neural tube defects are preventable by increasing folic acid intake (14).

## **Healthy People 2020 Challenges**

The Healthy People 2020 objectives (15) include reducing rates of fetal and infant death, lowering the occurrence of neural tube defects, and reducing developmental disability. Birth defects surveillance is a critical component of the public health strategy to achieve these objectives. The birth defects surveillance program in Massachusetts allows the Department of Public Health to monitor the occurrence of birth defects in the state. This data makes it possible to identify changes in birth defect rates over time, to identify geographical areas with unusually high or low rates, and to allow for development of strategies for prevention and for providing services to affected families.

### **Birth Defects Surveillance in Massachusetts**

Since 1999, the Massachusetts Birth Defects Monitoring Program (BDMP) located within the Massachusetts Center for Birth Defects Research and Prevention has conducted statewide, population-based active surveillance of birth defects among Massachusetts residents. The Center collects data on birth defects and identifies related trends, searches for potential causative factors associated with birth defects, addresses community concerns about birth defects, provides information to families of children with birth defects, and collects information on related screening and prevention efforts.

The BDMP identifies cases with structural birth defects diagnosed through 1 year of age. The primary focus of the state surveillance system is the identification of major structural birth defects that occur with or without a chromosomal abnormality or other non-chromosomal malformation syndrome.

The program's active surveillance system uses multiple sources of ascertainment, including prenatal reports, delivery and specialty care hospitals and birthing centers. Vital records serve as an additional source of information, providing demographic and clinical information on cases and acting as an additional source of case-finding.

Potential birth defect cases, identified through these varied sources, are assigned to medical record abstractors who review maternal and infant medical records. All cases are coded according to the International Classification of Diseases, Ninth Revision, Clinical Modification, modified British Pediatric Association (ICD-9-CM/BPA) system. Complex cases and cases in which the infant died are reviewed by a clinical geneticist.

The birth defects included in Massachusetts surveillance are ICD-9-CM/BPA codes ranging from 740.0 to 759.9 and several other selected codes outside this range for defects such as DiGeorge syndrome, Pierre Robin sequence and amniotic bands. A list of the ICD-9-CM/BPA codes for defects presented in this report is provided in [Appendix 4](#).

### **Economic Impact on Massachusetts**

The estimated lifetime cost in Massachusetts for babies born with one of 12 major structural birth defects is 200 million in 2012 dollars (5). This includes 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 can also be social and emotional impacts, which are difficult to quantify.

## **Birth Defects Surveillance Regulations**

In 2009, Massachusetts promulgated regulations (105 CMR 302) related to the Massachusetts Birth Defects Monitoring Program, which expanded reporting requirements for birth defects cases identified at or after birth and extended reporting to cases identified prenatally.

## **The 2013-2014 Surveillance Report**

This report presents statewide data on the prevalence of birth defects in live births, stillbirths, and other pregnancy losses in Massachusetts during the years 2013-2014. Most of the data is presented with the years 2013 and 2014 combined, since the numbers are relatively small for individual defects in a single year.

In early 2011, Massachusetts began ascertaining prenatally-diagnosed birth defects in pregnancies that ended in pregnancy losses other than a live birth or stillbirth, with the first full year of data available in 2012. Unless otherwise noted, data presented includes all pregnancy outcomes.

In 2014, we expanded the defect codes collected in Surveillance to be consistent with national reporting guidelines. We discuss the impact of this expanded reporting in [Chapter 8: Trend Analysis](#).

## **CHAPTER 2: METHODS**

### **Case Definition**

This report presents data on selected birth defects among deliveries to Massachusetts residents occurring during the calendar years 2013 and 2014. Cases were included if they met the following criteria:

Live birth, or stillbirth (fetal death) with a gestational age of at least 20 weeks or with a weight of at least 350 grams, or other pregnancy loss (includes early fetal deaths <20 weeks and <350 grams, elective terminations).

The infant or fetus had a structural birth defect that met diagnostic criteria listed in [Appendix 4](#).

For live births, the diagnosis must have been confirmed during the first year of life.

### **Data Collection**

Hospitals across the state submit monthly reports with birth defect diagnoses to the BDMP. Abstractors review maternal and infant medical records to collect information for each potential case. Beginning with 2008 births, reporting sites were expanded to include outpatient centers, emergency rooms, day surgery clinics, and laboratories.

Each live born case in the BDMP is linked to a Registry of Vital Records and Statistics record of live birth. Each reportable fetal death case is linked to a fetal death certificate, when available. Demographic and clinical variables, including maternal age, race/ethnicity, gestational age, birth weight, method of delivery, plurality, and region of residence are obtained from the live birth or fetal death certificate. Infant sex is ascertained from birth defects surveillance data because it is usually considered to be more accurate. For stillbirths without a fetal death certificate and for other pregnancy losses, demographic and clinical information comes from surveillance data.

### **Changes in Massachusetts Birth Defects Surveillance Over time**

A number of changes have taken place in our surveillance program over the years, which impact trends over time in prevalence of birth defects. Recent surveillance changes include the use of an electronic case report form for abstraction and the discontinuation of abstraction at two Rhode Island hospitals in 2011, the collection of cases diagnosed prenatally that did not result in a live birth or stillbirth (other pregnancy losses) in 2012, and the addition of a number of several new diagnostic codes in 2014, consistent with national reporting guidelines.

The surveillance program changes are summarized in Table 2.1 below.

**Table 2.1. Major Changes to Birth Defects Surveillance,  
Massachusetts 2000-2014**

2001:	<ul style="list-style-type: none"> <li>-Added 2 tertiary Rhode Island hospitals near the Massachusetts border</li> <li>-Added a tertiary specialty referral hospital (Massachusetts Eye and Ear Infirmary)</li> </ul>
2002:	<ul style="list-style-type: none"> <li>-Expanded list of central nervous system defect codes collected</li> <li>-Expanded reporting sources to include physicians, outpatient clinics and genetic services</li> </ul>
2005:	<ul style="list-style-type: none"> <li>-Added birth defect diagnoses</li> <li>-Clarified rules around which diagnoses must be reported</li> </ul>
2006:	<ul style="list-style-type: none"> <li>-Expanded the list of gastrointestinal defect codes</li> </ul>
2007:	<ul style="list-style-type: none"> <li>-Added code for genetic diagnosis and modified coding to be consistent with other programs</li> </ul>
2009:	<ul style="list-style-type: none"> <li>-Standardized electronic case reporting</li> <li>-New Birth Defect Monitoring Program regulations promulgated: <ul style="list-style-type: none"> <li>-Included additional birth defect codes</li> <li>-Expanded reporting sites to include outpatient visits, emergency department, and day surgery</li> <li>-Allowed collection of additional birth outcomes and prenatal diagnostic reporting</li> </ul> </li> </ul>
2011:	<ul style="list-style-type: none"> <li>-Developed electronic case report form for abstraction</li> <li>-Discontinued abstraction at Rhode Island hospitals</li> <li>-State implementation of new birth certificate format, with additional categories for race and other changes</li> <li>-Began collecting information on other pregnancy losses, including early miscarriages and elective terminations—2012 is first full year</li> </ul>
2012:	<ul style="list-style-type: none"> <li>-First full year of including other pregnancy losses</li> </ul>
2014:	<ul style="list-style-type: none"> <li>-New birth defects diagnosis codes added, including hypospadias Grade 1 and muscular ventricular septal defects, to comply with national standards</li> </ul>

The discontinuation of data collection at Rhode Island hospitals, where some cases born to Massachusetts residents in the southeastern part of the state are delivered or receive treatment, is expected to reduce our case numbers, but the program will continue to ascertain cases with known birth outcomes that receive diagnosis or

treatment in Massachusetts. Based on 2008-2009 data, we estimate that fewer than 20 live birth cases per year would be affected by this change, and for many of these we would still be able to obtain the information through referral hospital or prenatal diagnostic information. The impact is expected to be greater for cases with other pregnancy losses.

The addition of other pregnancy losses to our surveillance adds a significant number of cases to our Surveillance program and brings our rates closer to national rates. The addition of additional diagnostic codes to our reporting adds a substantial number of cases. For example, the prevalence of overall birth defects in 2012 increased from approximately 195 per 10,000 for live births and stillbirths only, to 243 per 10,000 with other losses included (<https://www.mass.gov/lists/massachusetts-birth-defects-surveillance-reports#2011-2012->). The addition of more defects in 2014 also added a substantial number of cases.

## **Quality Control**

To ensure data quality, the BDMP performs regular data quality checks. Key demographic and clinical information on live births and fetal deaths is checked against vital records data. The BDMP system includes many built-in logic and range checks, as well as checks for missing information. In addition, each case receives clinical review, which provides an additional layer of checking, as well as ensuring that diagnostic information is as accurate as possible. Regular data reports are run to identify missing or unusual data values, and periodic re-abstraction and data entry checks are also performed.

## **Confidentiality**

The program has developed extensive procedures to safeguard the confidentiality of the data and to protect the privacy of families. These procedures uphold ethical and legal obligations to protect confidentiality and comply with the requirements of state and federal laws.

## **Data Analysis**

A birth defect may occur as a single event or in combination with other defects. If a case had more than one defect within the same defect category, only one of these defects was counted in the category total. If a 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. For live births and stillbirths in this report, maternal age, race/ethnicity, plurality and infant birth weight are drawn from Vital Records. In cases where a stillbirth lacks a fetal death certificate and for other pregnancy losses, demographic and clinical information comes from surveillance data. For all outcomes, infant sex is drawn from surveillance data because it is generally considered to be more accurate.

The occurrence of birth defects is reported as a prevalence rate. Prevalence rate is calculated as the number of birth defect cases per 10,000 live births delivered during

the same time period. Prevalence tables include the number of cases, the estimated prevalence rate per 10,000 live births and the 95% confidence interval (CI) for each rate. Prevalence rather than incidence rates are used because incidence (new cases) of birth defects would need to be based on the number of embryos conceived within a year. This cannot be fully measured because the total number of conceptions and the number of these conceptions resulting in a delivery with a birth defect are not known (16).

Information on counts used for denominators in rates can be found in [Appendix 3](#). The CI can be used to assess the magnitude and stability of a rate or ratio. The CI for rates presented in this report consists of a range of possible values around the point estimate that has a 95% chance of including the actual underlying rate of a birth defect. Wide CI reflects the large variation due to small numbers (see [Appendix 1: Technical Notes](#)).

## Limitations

1. Defects that are not diagnosed at birth and that do not require hospitalization may be underreported.
2. Misclassification of birth defects may occur as a result of surveillance system coding errors or incomplete diagnostic information.
3. Only diagnoses confirmed through 1 year of age are currently included in BDMP surveillance. This may lead to undercounting of defects that are difficult to detect by this time.
4. The discontinuation of case abstraction in Rhode Island in 2011 may lead to undercounting of cases that receive no care in Massachusetts, especially cases born to residents of the southeastern part of the state after 2010.
5. Patent ductus arteriosus (PDA) is not included in this report, because this defect is often minor and is normal for infants born prematurely.
6. Comparisons between Massachusetts data and national estimates should be interpreted with caution, as there are differences in surveillance system methodologies, types of pregnancy outcomes included, and demographic variations.
7. Trends over time should be interpreted carefully, with consideration of the changes that have taken place over time, such as the addition of new codes and additional pregnancy outcomes.

Additional report notes can be found in [Appendix 1: Technical Notes](#).

## Glossary

A glossary of selected terms used in this report is included in [Appendix 2](#).

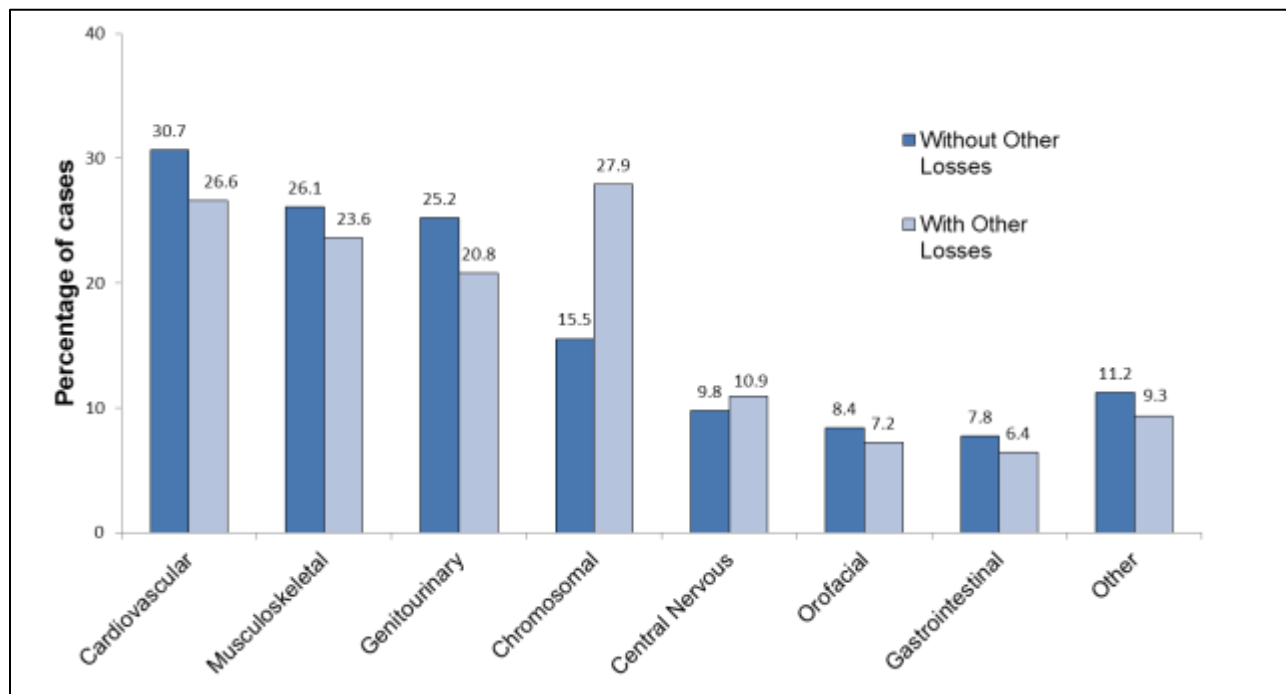
## CHAPTER 3: PREVALENCE OF BIRTH DEFECTS

### Overall Prevalence of Birth Defects

There were 143,662 live births to Massachusetts residents in 2013-2014, and our surveillance identified 3894 deliveries with at least one structural birth defect. This represents an overall Massachusetts birth defect rate of 271.0 (95% CI: 262.7-279.6) per 10,000 live births. Counts and rates for specific birth defects are shown in [Table T.1](#).

Figure 3.1. shows the percentage of reported birth defects by body system category with and without including other pregnancy losses. Among live birth and stillbirth cases, cardiovascular defects are the most common, followed by musculoskeletal, genitourinary and chromosomal defects. When we add the other pregnancy loss cases, then chromosomal defects become the most common. Because cases can have multiple defects, the same case may be included in more than one body system category.

**Figure 3.1. Birth Defects by Body System, with and without Other Pregnancy Losses, Massachusetts, 2013-2014**



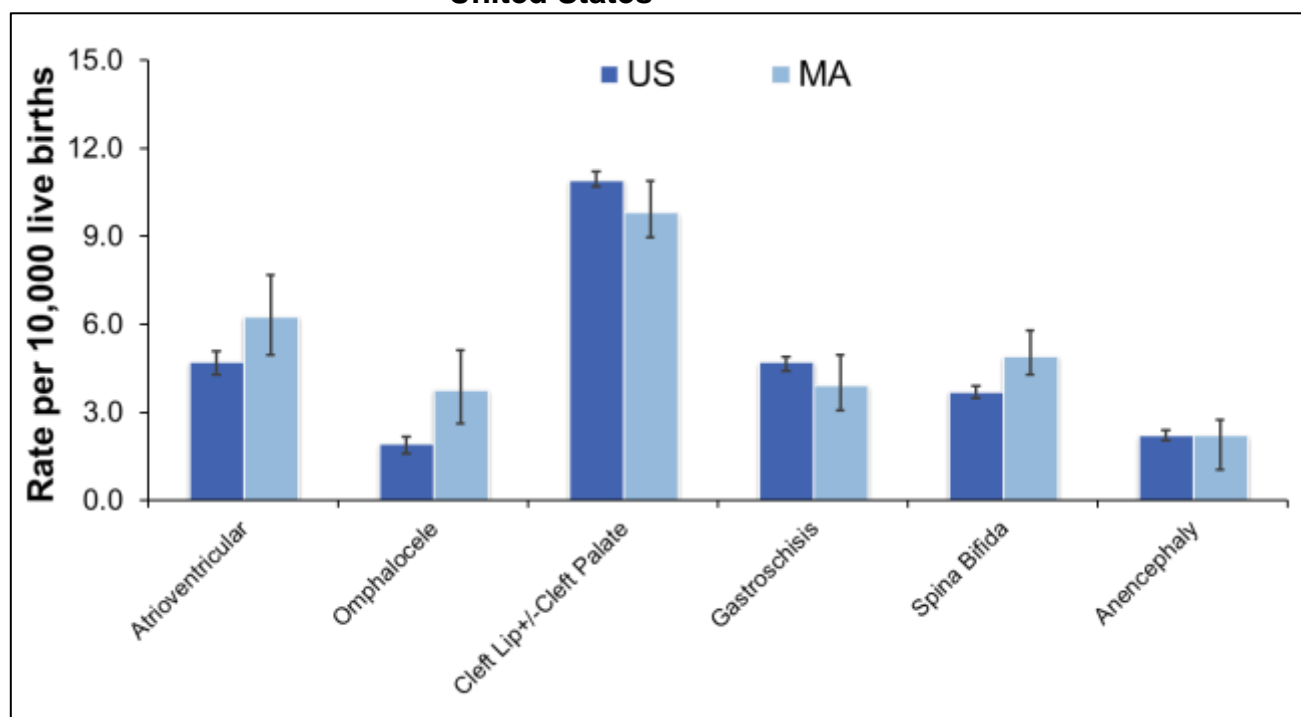
Because cases can have defects in more than one body system, percentages sum to more than 100.



## Birth Defects in Massachusetts vs. United States

Massachusetts is one of 11 states that has an active case ascertainment program and contributes birth defects data to published national prevalence estimates for selected birth defects. For many defects, Massachusetts rates are similar to national rates. However, Massachusetts rates for 2013-2014 are significantly higher for omphalocele, atrioventricular septal defects, and for trisomy 21 (Down syndrome), trisomy 13, and trisomy 18, even after age-adjustment (Figure 3.2a, Figure 3.2b, [Table T.2](#)). Differences in surveillance system methodology, types of pregnancy outcomes included, and demographic variation may account for the differences in rates for certain defects. Massachusetts rates in 2013-2014 included other pregnancy losses, including early fetal deaths <20 weeks, which are not included in national estimates.

**Figure 3.2a. Prevalence of Selected Birth Defects, Massachusetts vs. United States**

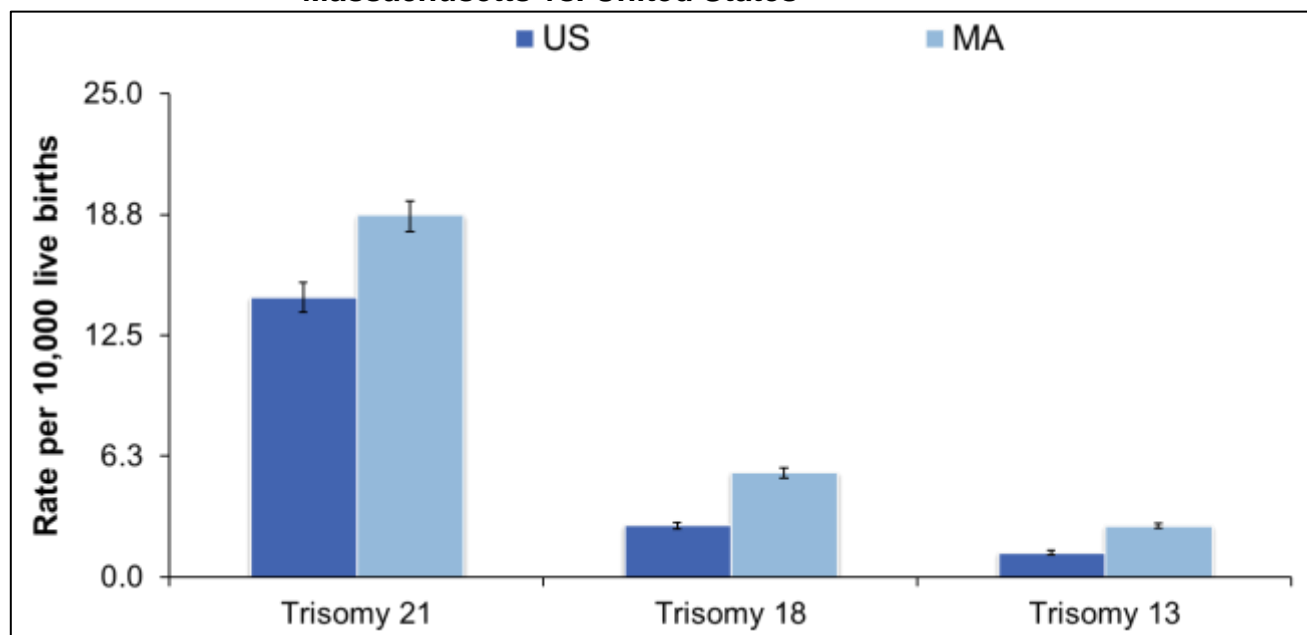


MA rates based on live births, stillbirths, and other pregnancy losses, N=3894.

US rates based on crude, pooled prevalence data from 11 active case-ascertainment programs, including Massachusetts (7). Nine of the 11 states contributing to the pooled estimates include elective terminations but not fetal deaths less than 20 weeks or less than 350 grams.

Error bars represent 95% confidence interval.

**Figure 3.2b. Age-Adjusted Prevalence of Selected Birth Defects,  
Massachusetts vs. United States**



MA rates based on live births, stillbirths, and other pregnancy losses, N=3894 and adjusted to maternal age distribution of US population 2006. National Vital Statistics Reports 2009 Volume 57, Number 7.

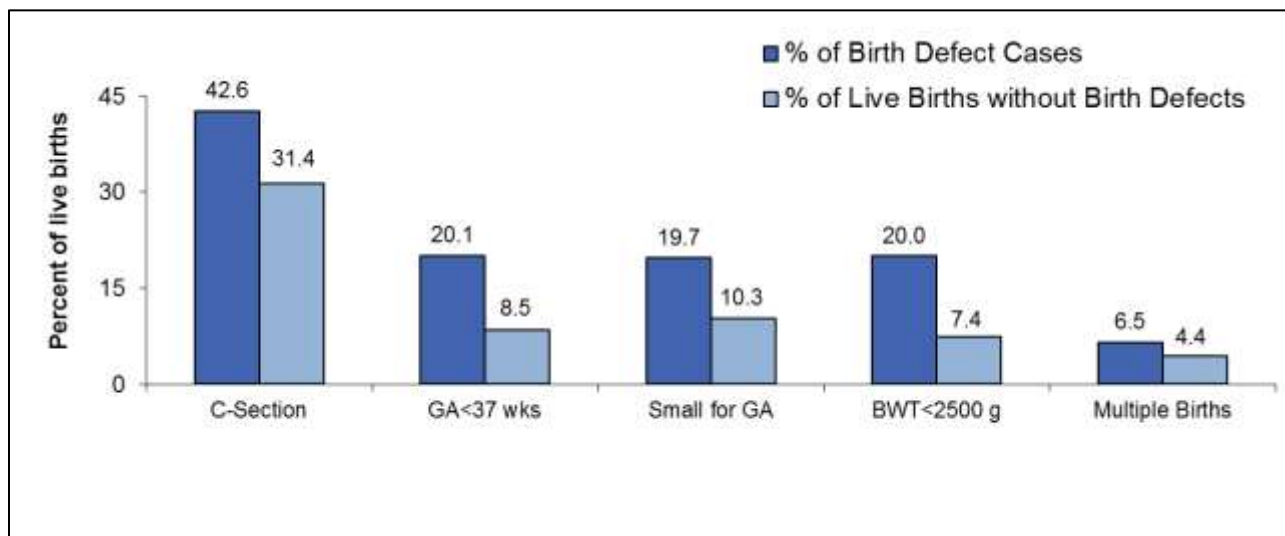
US rates based on crude, pooled prevalence data from 11 active case-ascertainment programs, including Massachusetts (7). Nine of the 11 states contributing to the pooled estimates include elective terminations but not fetal deaths less than 20 weeks or less than 350 grams.

Error bars represent 95% confidence interval.

## Selected Pregnancy Outcomes

Adverse pregnancy outcomes such as Cesarean section (C-section), low birth weight, prematurity, and small for gestational age (SGA) are more frequent among live born infants with birth defects than among unaffected infants. Infants with a birth defect are 2.7 times more likely to have low birth weight (less than 2500 grams) or very low birth weight (less than 1500 grams) and 2.4 times more likely to be born premature (before 37 weeks) compared to those without birth defects (Figure 3.3). Cesarean (C-section) deliveries are more common among live born infants with birth defects (43%) compared to unaffected infants (31%). In addition, infants with birth defects are more likely to die in their first year of life.

**Figure 3.3. Pregnancy Outcomes among Live Births with and without Birth Defects, Massachusetts: 2013-2014**



N=3015 live births with birth defects; N=140,647 live births without birth defects.

C-section: Cesarean section; GA: gestational age; BWT: birth weight

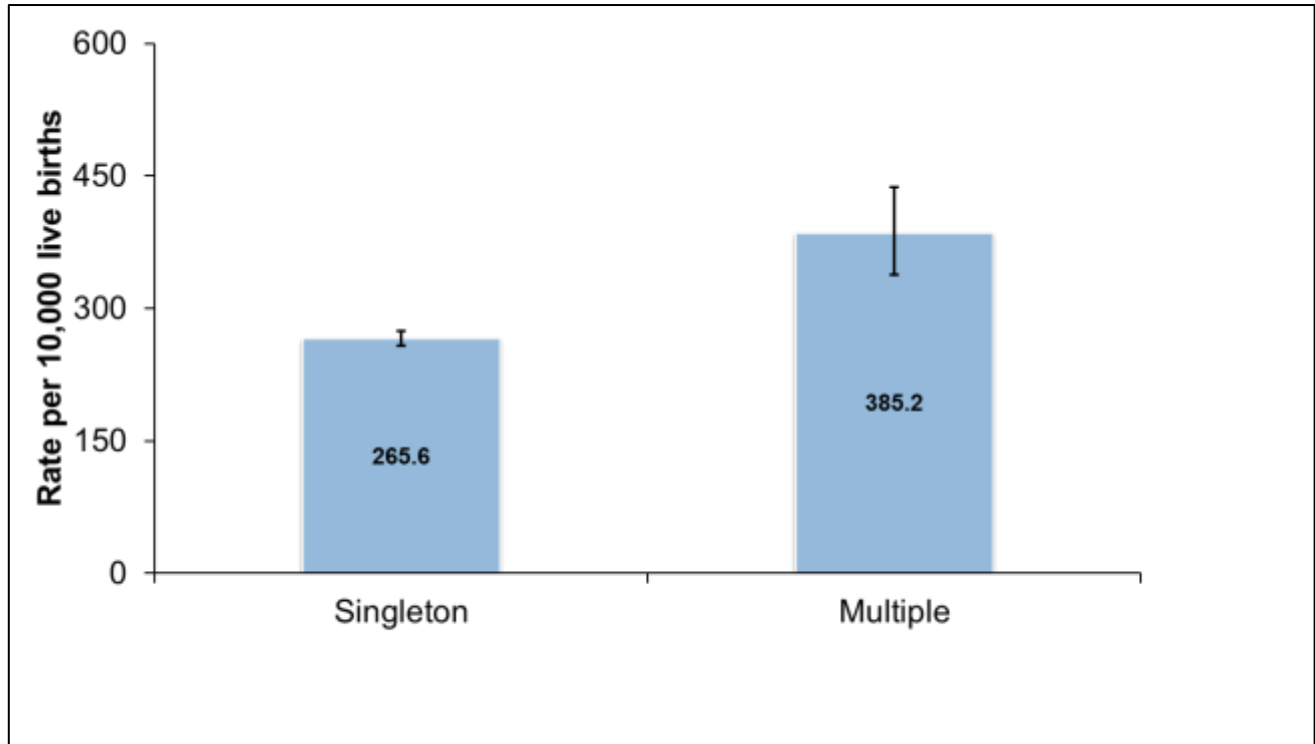
Small for GA calculation based on method described by Oken et al. in 2003, defined as birth weight below the 10th percentile for gestational age based on a sex-specific US standard (17).

## CHAPTER 4: PREVALENCE OF BIRTH DEFECTS BY SEX AND PLURALITY

Several birth defects are more common in males than in females, including club foot ([Table T.3](#)). Also, several obstructive genitourinary defects occur exclusively in males, leading to higher rates of these defects as well.

The overall birth defect rate in singletons is 265.6 (95% CI: 257.1-274.2) per 10,000 live births, while the rate in multiples is 385.2 (95% CI: 338.4-436.6) per 10,000 live births. While many individual defects do not differ significantly by plurality (Figure 4), the rates of septal defects and atrial and ventricular septal defects are significantly higher in multiples than in singletons ([Table T.4](#)).

**Figure 4. Prevalence of Birth Defects in Singleton and Multiple Births, Massachusetts: 2013-2014**



Live births, stillbirths and other pregnancy losses, N=3894.  
Error bars represent 95% confidence interval.

## CHAPTER 5: PREVALENCE OF BIRTH DEFECTS BY MATERNAL AGE

### Maternal Age

The prevalence of birth defects varies by maternal age (Table 5.1), with rates highest for mothers ages 35 years and older (350.7 per 10,000 live births) and second highest for mothers younger than 20 years of age (275.7 per 10,000 live births).

**Table 5.1. Overall Prevalence of Birth Defects by Maternal Age, Massachusetts: 2013-2014**

Maternal Age (years)	Cases	Rate	95% Confidence Interval
<20	143	275.7	232.4-324.8
20-24	455	232.6	212.0-254.8
25-29	832	234.7	219.2-251.0
30-34	1294	258.3	244.6-272.5
35+	1170	350.7	331.2-371.0

Live births, stillbirths, and other pregnancy losses. N=3894.  
Rate per 10,000 live births.

Monitoring birth defects by maternal age is important in part because the percentage of women giving birth in the state who are age 35 or older doubled from 11.4% in 1989 to 22.2% in 2011 (8), with more multiple births among mothers over age 35 than among younger mothers.

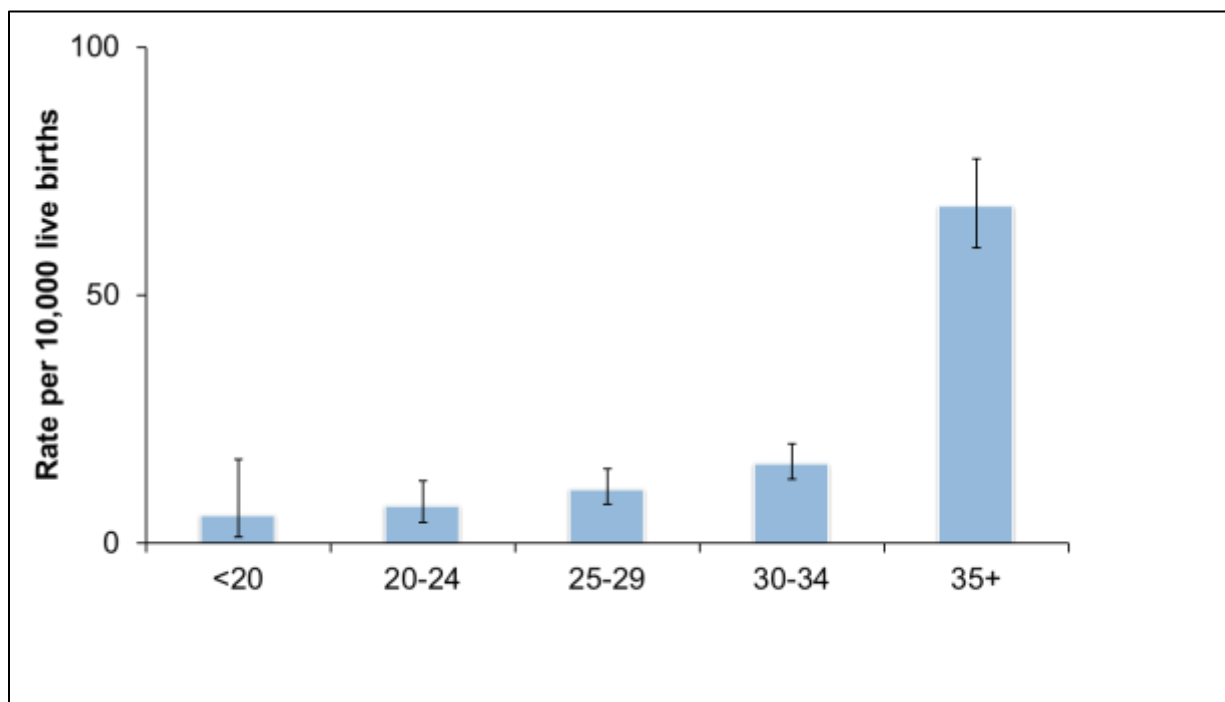
The use of assisted reproductive technology (ART) is a factor in the increased percentage of women ages 35 and older giving birth and the increased frequency of multiple births among Massachusetts mothers. Massachusetts has the highest ART rate in the nation, with 4.5% of live births conceived with ART (9).

The prevalence rates of specific birth defects by age group are shown in [Table T.5](#). Gastroschisis rates are highest in women under 25 years of age, while trisomy 21 (Down syndrome) rates are highest in women ages 35 and older.

## Down Syndrome

There is a strong association between Down syndrome and advanced maternal age (Figure 5.1). In Massachusetts, the rate of Down syndrome in mothers 35 and older is 68 per 10,000 live births, which is more than 5 times greater than the rate in mothers younger than 35. This reflects the general pattern of higher chromosomal defect rates among older women.

**Figure 5.1. Prevalence of Down Syndrome by Maternal Age, Massachusetts: 2013-2014**

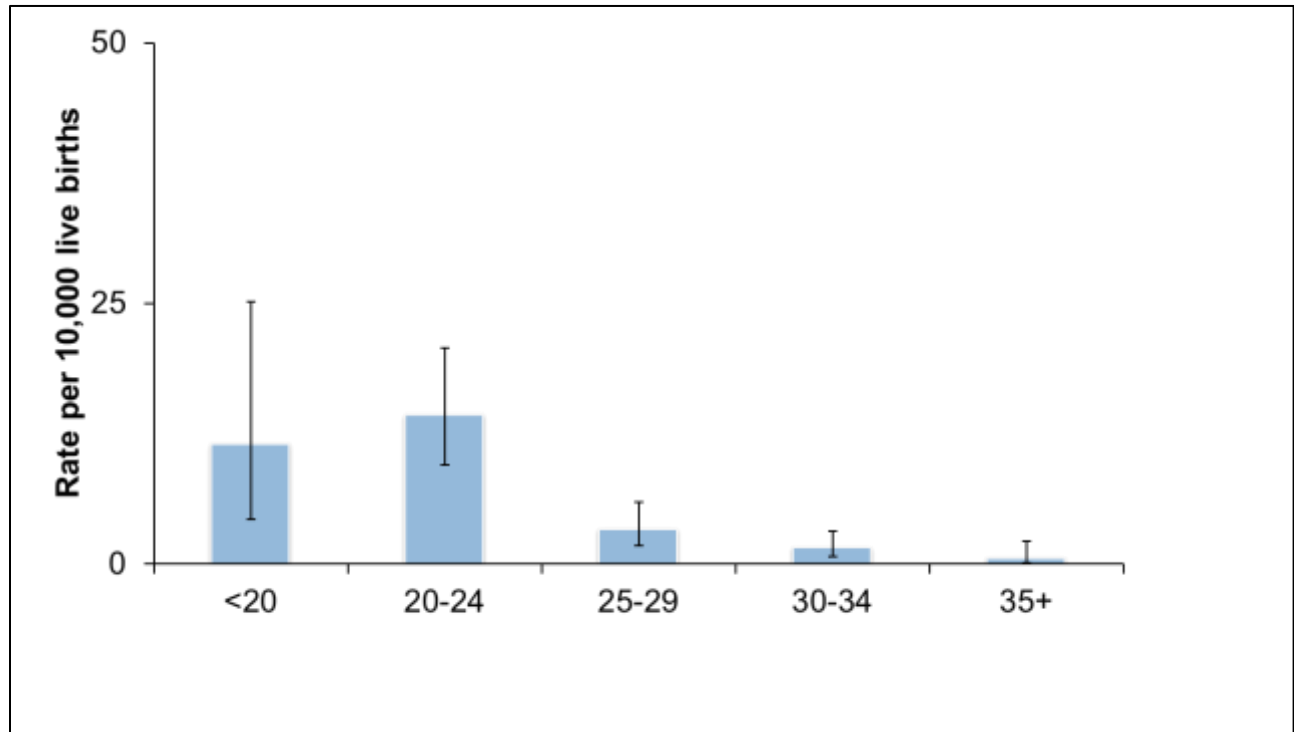


Live births, stillbirths, and other losses, N=365.  
Error bars represent 95% confidence interval.

## Gastroschisis

Mothers ages 20-24 had the highest rate of gastroschisis cases at 14.3 per 10,000 live births (Figure 5.2). The association between gastroschisis and younger maternal age has been shown in previous studies (18).

**Figure 5.2. Prevalence of Gastroschisis by Maternal Age, Massachusetts: 2013-2014**



Live birth, stillbirths, and other losses, N=56.  
Error bars represent 95% confidence interval.

## CHAPTER 6: PREVALENCE OF BIRTH DEFECTS BY MATERNAL RACE/ETHNICITY AND REGION

### Maternal Race/Ethnicity

In Massachusetts and nationally, birth defect rates vary by maternal race/ethnicity. Table 6.1 shows the variation in age-adjusted birth defect rates by racial/ethnic group in Massachusetts during the current reporting period.

**Table 6.1. Age-Adjusted Prevalence of Birth Defects by Maternal Race/Ethnicity, Massachusetts: 2013-2014**

Maternal Race	Cases	Age-Adjusted Rate <sup>1</sup>	95% Confidence Interval
White, Non-Hispanic	2,452	274.3	258.8-289.9
Black, Non-Hispanic	387	280.9	251.7-310.1
Asian, Non-Hispanic	277	220.0	146.1-293.9
Hispanic	121	248.4	231.1-265.7
Other, Non-Hispanic <sup>2</sup>	30	221.1	135.9-306.2

Live births, stillbirths, and other pregnancy losses. N=3847. Excludes 47 cases missing race information.

<sup>1</sup> Rate per 10,000 live births, adjusted to statewide maternal age distribution of the birthing population.

<sup>2</sup> Includes American Indian.

The age-adjusted overall birth defect rates did not differ significantly by race, although the rate in Asian mothers was slightly lower than the rate among other racial groups. Possible explanations for racial differences include genetic variation, diet differences, and varying access to prenatal screening and health care services. For example, data from the 2011 Massachusetts Pregnancy Risk Assessment Monitoring System shows racial/ethnic differences in the use of a multivitamin in the month prior to pregnancy (19). These results are shown in [Appendix 5. Table T.6.](#) shows the prevalence rates of the individual birth defects by maternal race/ethnicity. Trends over time in maternal race/ethnicity can be found in [Chapter 8.](#)



## Maternal Region of Residence

The Massachusetts Executive Office of Health and Human Services divides the state into six regions for statistical, care coordination and administrative purposes. The six regions are based on geographical groupings of cities and towns: Western, Central, Northeast, Metro West, Southeast, and Boston. A map of these regions is provided in [Appendix 6](#). The age-adjusted birth defect rates by the six regions in 2013-2014 are shown in Table 6.2. There were no significant differences in rates by region. We also observed no evidence of trends over time (Figure 6.2).

**Table 6.2. Age-Adjusted Prevalence of Birth Defects by Maternal Residence Region, Massachusetts: 2013-2014**

Region	Cases	Age-Adjusted Rate <sup>1</sup>	95% Confidence Interval
Western	419	265.4	240.6-290.1
Central	518	292.4	258.1-326.8
Northeast	864	284.7	264.9-304.4
Metro West	898	251.9	236.0-267.8
Southeast	620	252.4	233.8-271.2
Boston	575	283.3	253.9-312.7

Live births, stillbirths, and other pregnancy losses, N=3894.

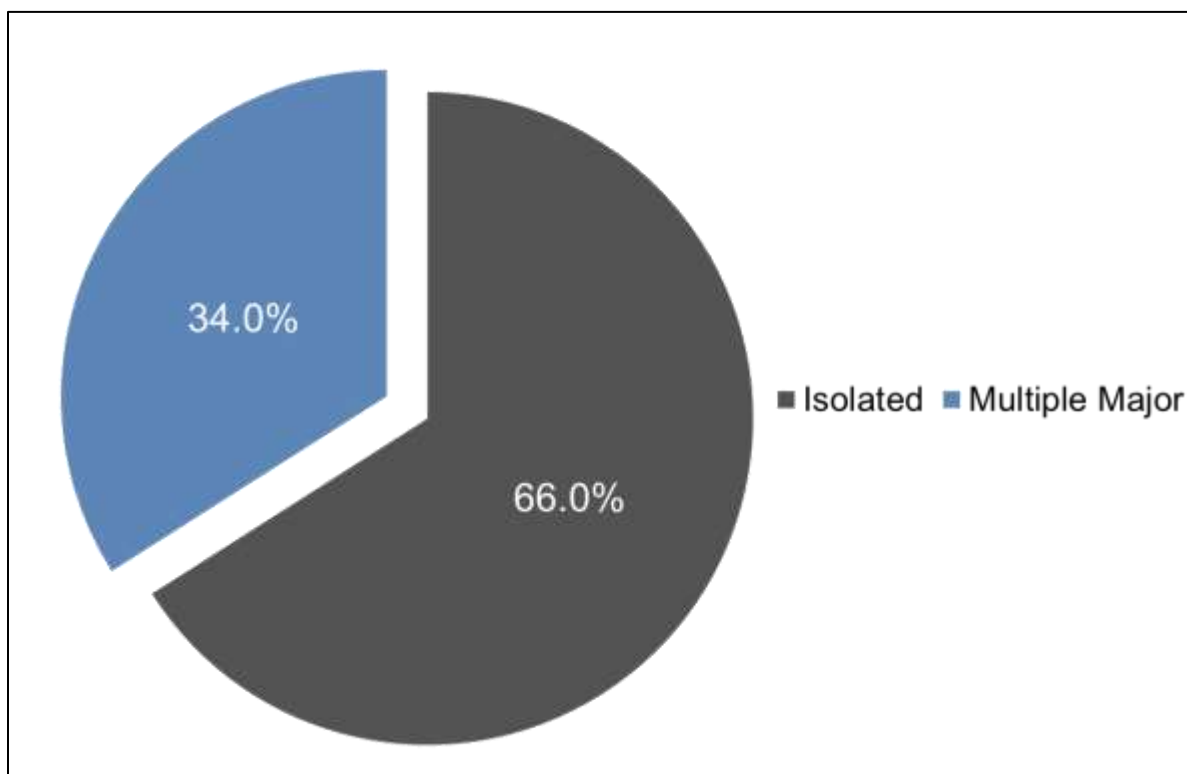
<sup>1</sup> Rate per 10,000 live births, adjusted to statewide maternal age distribution of the birthing population.

## CHAPTER 7: BIRTH DEFECTS BY PATTERN AND ETIOLOGY

### Pattern

Cases are classified based on their pattern (i.e., whether a defect occurs with others). Of the 3894 birth defect cases in 2013-2014, 34% had multiple major defects or syndromes, and 66% were isolated (Figure 7.1). Approximately 59% of the isolated cases had single defects, and nearly 34% had multiple defects within the same organ or a single major defect with one or more minor defects; the remaining 7% are part of a sequence of developmental events. Birth defects that tend to occur as solitary defects include hypospadias, gastroschisis, and craniosynostosis.

**Figure 7.1. Birth Defects by Pattern, Massachusetts: 2013-2014**



Live births, stillbirths and other pregnancy losses, N=3894.

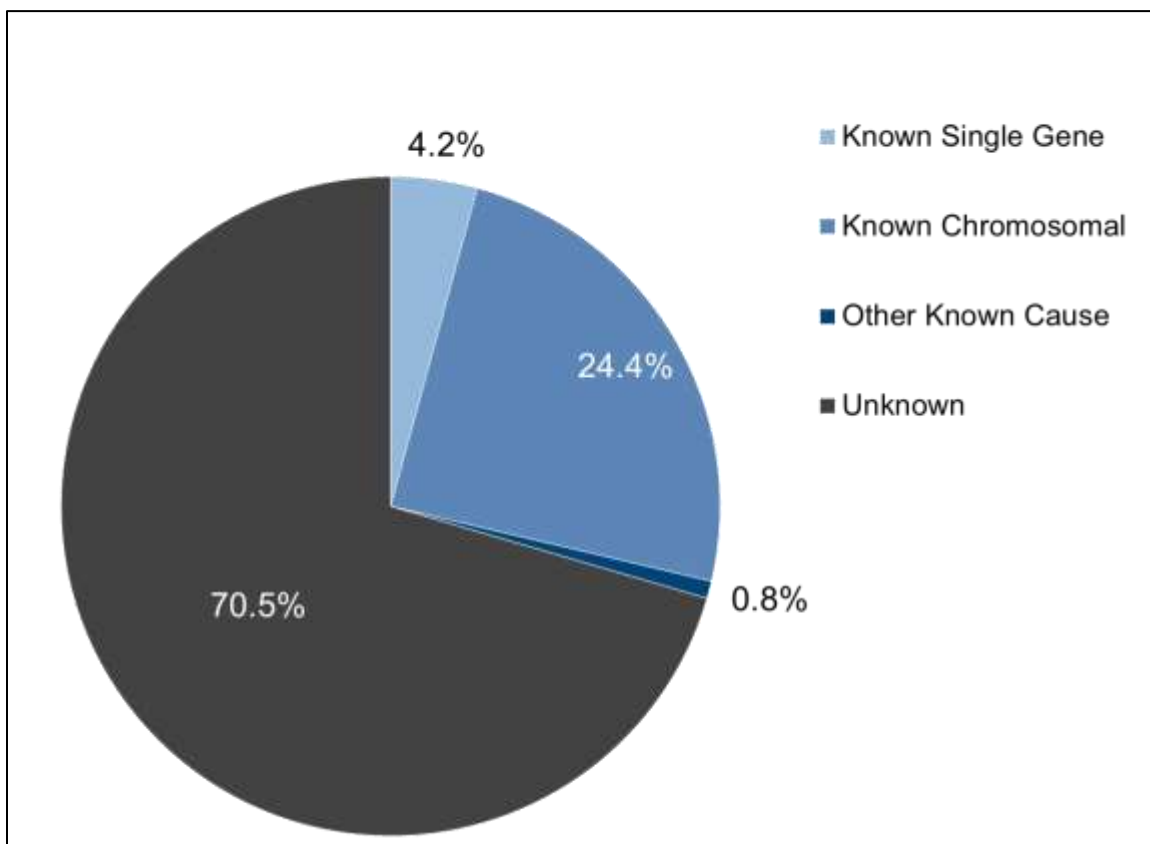
Isolated defects include cases with a single defect, those with multiple defects in the same organ, those with a single major defect and one or more minor defects, and those with defects that occur as part of a sequence of events.

## Etiology

The surveillance system in Massachusetts collects etiology or causal information for birth defects, when available. Cases are classified into etiology categories, with similar cases grouped using knowledge of pathogenesis and embryologic mechanisms. Etiology classification considers each case as a biologic entity rather than a collection of individual defects. The schema was developed based on general principles outlined in the literature (20,21,22).

The majority of birth defects cases in Massachusetts in 2013-2014 (70.5%) had an unknown cause (Figure 7.2). Most of the cases with known etiology were whole chromosome abnormalities (extra copy or missing copy of a chromosome), such as trisomy 13 and Turner syndrome. Other examples of defects with known etiologies include single gene defects, such as achondroplasia, Smith-Lemli-Opitz syndrome and other defects considered to be a Mendelian syndrome. Examples of known etiologies include teratogens (e.g., thalidomide), maternal conditions (e.g., diabetes), and conditions of the uterine environment (e.g., didelphys uterus).

**Figure 7.2. Birth Defects by Etiology, Massachusetts: 2013-2014**



Live births, stillbirths, and other pregnancy losses, N=3894. Percentages may not add to 100% due to rounding.

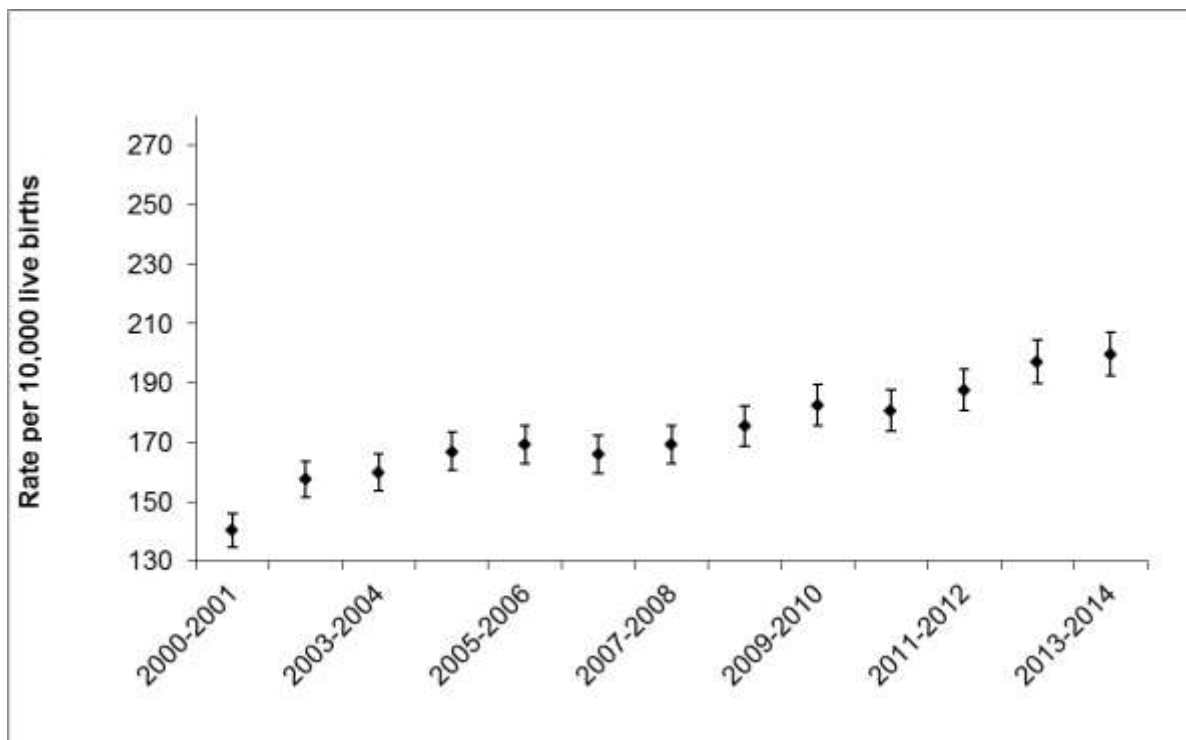
## CHAPTER 8: TREND ANALYSIS

The rate of birth defects in Massachusetts has increased steadily over time (Figure 8.1a). This likely reflects improvements in case ascertainment or confirmation as described above, changes in the distribution of demographic variables over time (e.g., more births to older mothers), changes in survival to diagnosis, and random variation, more than true increases in the overall rate of birth defects. The addition of new reporting sources and codes would also have impacted these trends.

The addition of other pregnancy losses in 2012 resulted in a large increase in birth defect prevalence rates (Figure 8.1b). The collection of additional defects beginning in 2014, including grade 1 hypospadias, resulted in further increases in prevalence rates.

**Figure 8.1a. Overall Prevalence of Birth Defects in Massachusetts,  
2-year Rolling Average  
Live Births and Stillbirths, 2000-2014**

*Excludes new defects added in 2014*

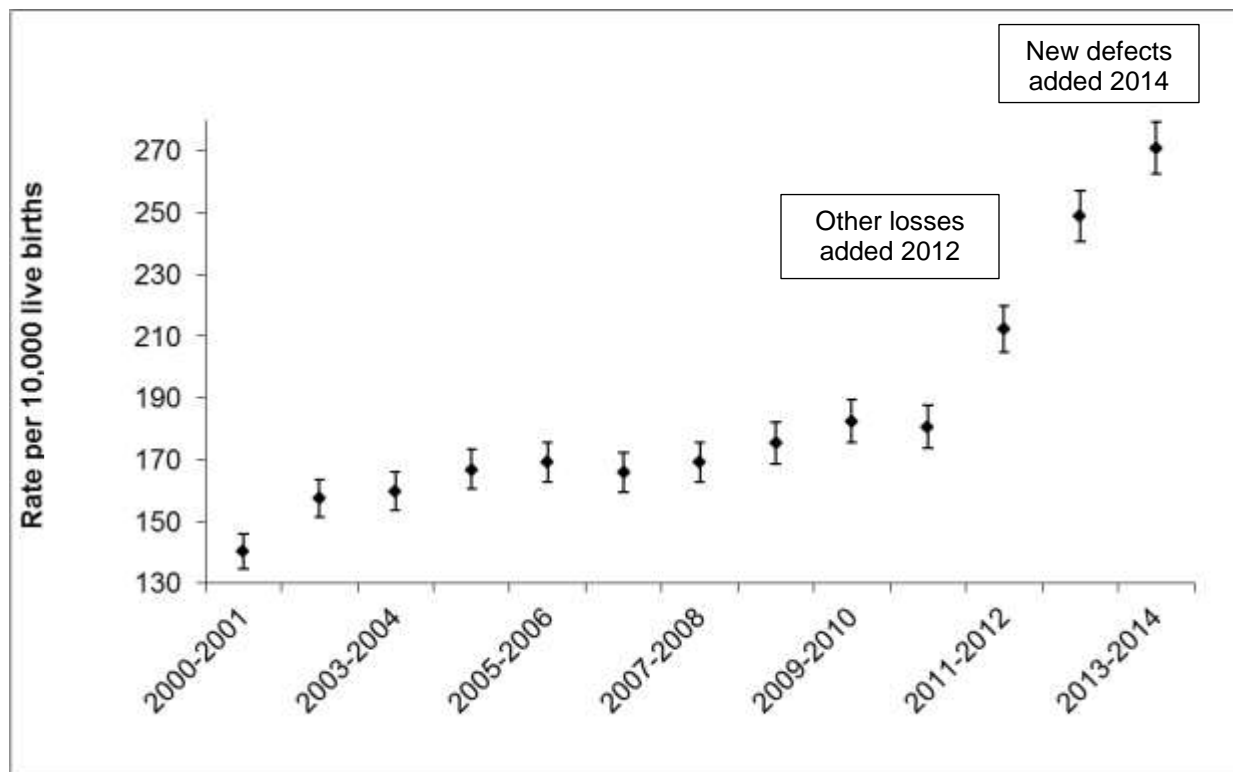


Note: Y axis begins at 130.

Error bars represent 95% confidence interval.

**Figure 8.1b. Overall Prevalence of Birth Defects in Massachusetts,  
2-year Rolling Average  
Live Births, Stillbirths, and Other Pregnancy Losses, 2000-2014**

*Includes new defects added in 2014*



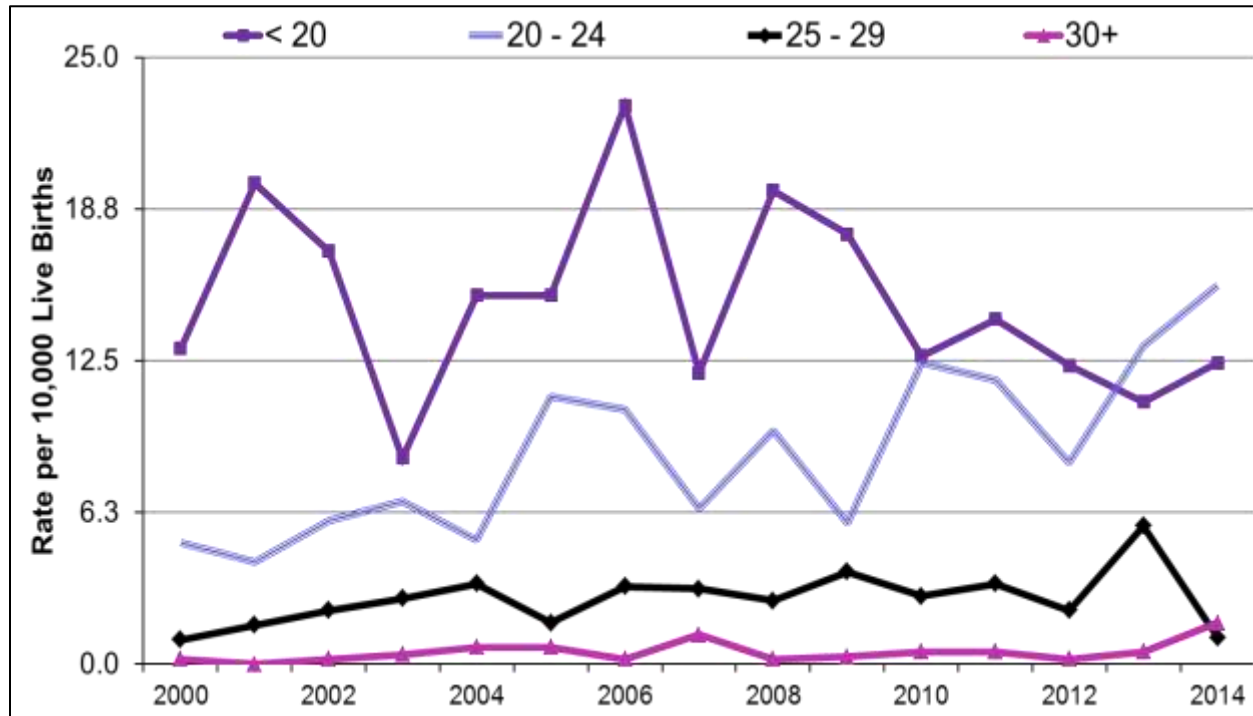
Note: Y axis begins at 130.

Error bars represent 95% confidence interval.

## Trends in Gastroschisis

Figure 8.2. presents gastroschisis rates by maternal age in Massachusetts over a 12-year time span. Rates of gastroschisis have been increasing over time among 20-24 year old mothers in Massachusetts. This is similar to trends seen nationally (23).

**Figure 8.2. Prevalence of Gastroschisis by Year and Maternal Age, Massachusetts: 2000-2014**

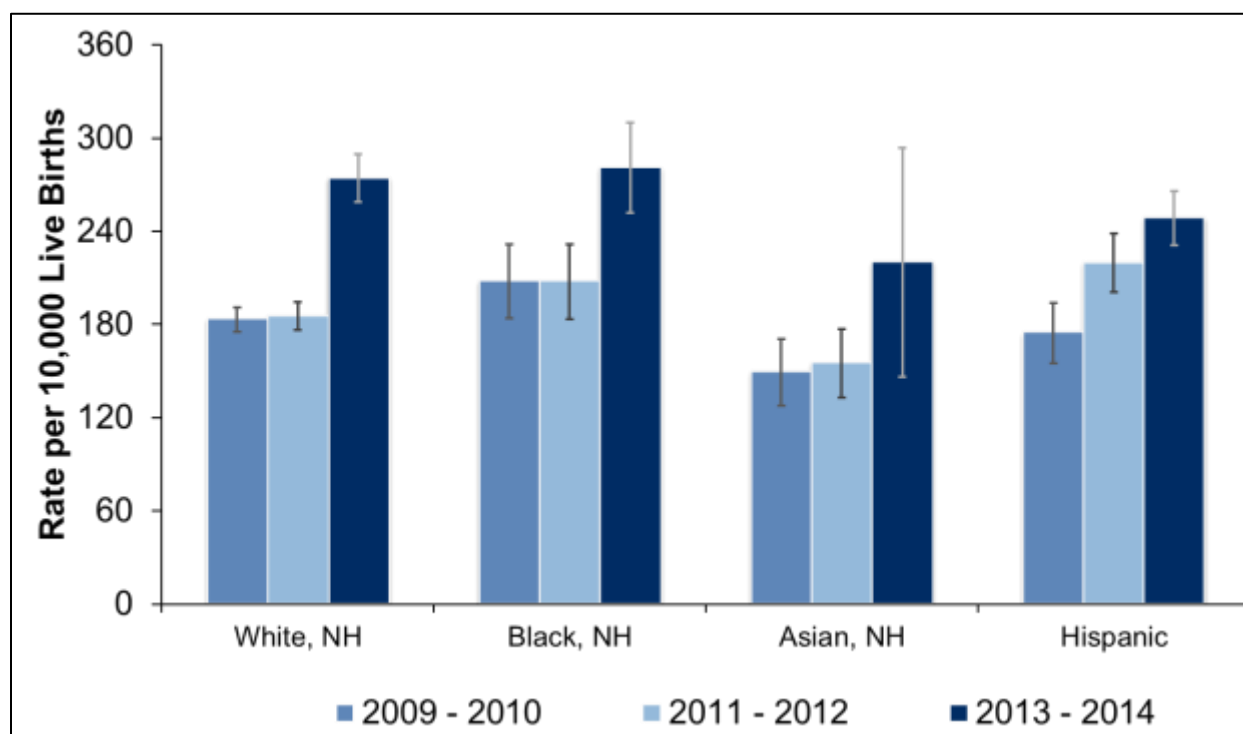


Includes live births and stillbirths.

## Trends in Maternal Race/Ethnicity

Figure 8.3. shows the age-adjusted birth defect rates by race/ethnicity between 2009 and 2014 in two-year intervals. Rates in 2013-2014 did not differ significantly by race/ethnicity. In 2009-2010 and 2011-2012, Asians had significantly lower rates of birth defects, but with the addition of other pregnancy losses in 2013-2014, this difference is no longer significant.

**Figure 8.3. Prevalence of Birth Defects by Maternal Race/Ethnicity, Massachusetts: 2009-2014**

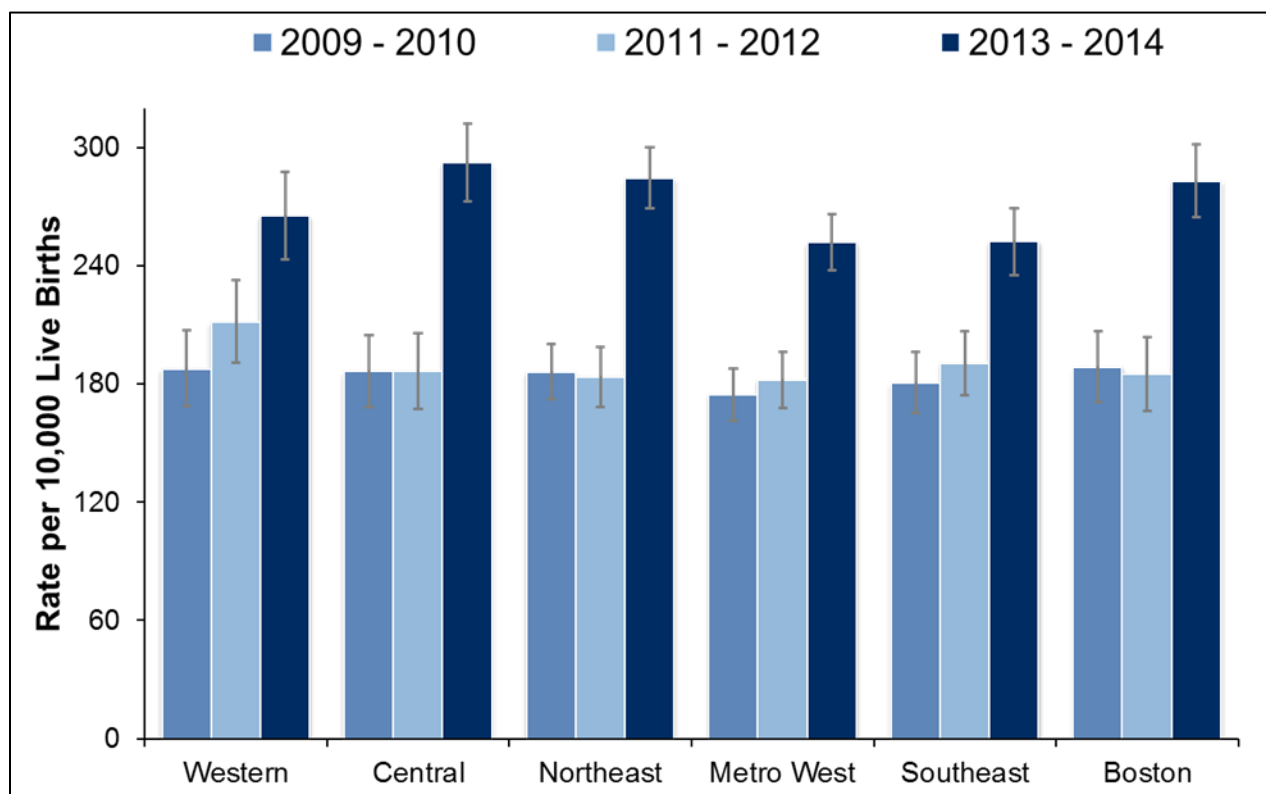


Includes live births, stillbirths, and other pregnancy losses (2013 on). NH=Non-Hispanic. Adjusted to statewide maternal age distribution of the Massachusetts birth population in each 2-year period. Error bars represent 95% confidence interval.

## Trends in Maternal Region of Residence

Figure 8.4 shows prevalence of birth defects by region of maternal residence over time. In 2013-2014 the overall age-adjusted birth defect prevalence was lowest in the Metro West and Southeastern regions and highest for the Central and Northeast regions. The end of abstraction of Rhode Island hospitals likely impacted the Southeastern region rates.

**Figure 8.4. Prevalence of Birth Defects by Maternal Residence Region, Massachusetts: 2009-2014**



Includes live births, stillbirths, and other pregnancy losses (2013 on). NH=Non-Hispanic.  
Adjusted to statewide maternal age distribution of the Massachusetts birth population in each 2-year period.  
Error bars represent 95% confidence interval.



## RESOURCES, SUGGESTED CITATION, CONTACT INFORMATION

### Resources

For additional information on birth defects:

[www.mass.gov/dph/birthdefects](http://www.mass.gov/dph/birthdefects)

### Suggested Citation

*Massachusetts Birth Defects 2013-2014*. Boston, MA: Center for Birth Defects Research and Prevention, Bureau of Family Health and Nutrition, Massachusetts Department of Public Health. September 2019.

<http://www.mass.gov/eohhs/gov/departments/dph/programs/family-health/birth-defect/monitoring/surveillance-reports.html>

Friendly URL: [www.mass.gov/dph/birthdefects](http://www.mass.gov/dph/birthdefects)

### Contact Information

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## TABLES

**Table T.1. Prevalence of Birth Defects, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Live Birth Count	Stillbirth Count	Other Pregnancy Loss Count	Total Count	Rate per 10,000 Live Births	95% Confidence Interval
<b>Total Cases with a Reportable Birth Defect</b>	3015	68	811	3894	271.0	262.7-279.6
<b>Central Nervous System</b>						
Anencephaly	1	1	29	31	2.16	1.47-3.06
Encephalocele	10	1	12	23	1.60	1.01-2.40
Holoprosencephaly	8	3	12	23	1.60	1.01-2.40
Hydrocephaly without Spina Bifida	59	4	16	79	5.50	4.35-6.85
Microcephaly	52	2	2	56	3.90	2.94-5.06
Spina Bifida with/without Hydrocephaly	32	4	34	70	4.87	3.80-6.16
Spinal Cord <sup>2</sup>	59	1	1	61	4.25	3.25-5.45
Other Central Nervous System <sup>2</sup>	146	4	35	185	12.88	11.09-14.87
<b>Eye</b>						
Aniridia	6		0	6	0.42	0.15-0.91
Anophthalmia/Microphthalmia	17		2	19	1.32	0.80-2.07
Congenital Glaucoma, Congenital Cataract	57		0	57	3.97	3.01-5.14
Other Eye <sup>2</sup>	67		0	67	4.66	3.61-5.92
<b>Ear</b>						
Anotia/Microtia	34	3	1	38	2.65	1.87-3.63
Other Ear <sup>2</sup>	83	1	1	85	5.92	4.73-7.32
<b>Cardiovascular</b>						
<i>Anomalous Pulmonary Venous Connection</i>						
Anomalous Pulmonary Venous Connection	27		1	28	1.95	1.30-2.82
<i>Atrioventricular Canal Defects</i>						
Atrial Septal Defect (ASD) Primum	1	1	1	3	0.21	0.04-0.61
Common Atrium	1		0	1	0.07	0.00-0.39
Complete Atrioventricular Canal Defect	43	7	14	64	4.45	3.43-5.69
Endocardial Cushion Defect	11	1	4	16	1.11	0.64-1.81
Ventricular Septal Defect (VSD), Canal Type	4	1	1	6	0.42	0.15-0.91

**Table T.1. Prevalence of Birth Defects, Massachusetts: 2013-2014**

<b>Defect<sup>1</sup></b>	<b>Live Birth Count</b>	<b>Stillbirth Count</b>	<b>Other Pregnancy Loss Count</b>	<b>Total Count</b>	<b>Rate per 10,000 Live Births</b>	<b>95% Confidence Interval</b>
<b><i>Conotruncal (Outlet) and Aortic Arch</i></b>						
Double Outlet Right Ventricle	23	3	7	33	2.30	1.58-3.23
Interrupted Aortic Arch, Type B	3		0	3	0.21	0.04-0.61
Tetralogy of Fallot	65	2	10	77	5.36	4.23-6.70
Truncus Arteriosus (Common Truncus)	7	1	1	9	0.63	0.29-1.19
Dextro-Transposition of the Great Arteries	37	1	3	41	2.85	2.05-3.87
<b><i>Ebstein Anomaly</i></b>						
Ebstein Anomaly	7	0	0	7	0.49	0.20-1.00
<b><i>Heterotaxy (Laterality Defects)</i></b>						
Heterotaxy	18	2	4	24	1.67	1.07-2.49
<b><i>Left-Sided Obstruction</i></b>						
Aortic Arch Atresia without Hypoplastic Left Heart Syndrome	0	1	0	1	0.07	0.00-0.39
Aortic Valve Stenosis	16	0	2	18	1.25	0.74-1.98
Coarctation of Aorta	68	1	0	69	4.80	3.74-6.08
Hypoplastic Left Heart Syndrome	21	3	15	39	2.71	1.93-3.71
Interrupted Aortic Arch, Type A or Not otherwise specified (NOS)	1	0	1	2	0.14	0.02-0.50
<b><i>Right-Sided Obstruction</i></b>						
Pulmonary Stenosis, Valvar	106	1	4	111	7.73	6.36-9.30
Pulmonary Valve Atresia with Intact Septum	7	1	1	9	0.63	0.29-1.19
Pulmonary Valve Atresia with VSD	7	0	3	10	0.70	0.33-1.28
Tricuspid Valve Atresia	10	0	1	11	0.77	0.38-1.37
<b><i>Septal Defects</i></b>						
ASD (Secundum and NOS)	327	1	6	334	23.25	20.82-25.88
VSD (Membranous and NOS)	189	6	16	211	14.69	12.77-16.81
VSD (Muscular)	160	1	1	162	11.28	9.61-13.15
VSD, Conoventricular/Malalignment	23	1	2	26	1.81	1.18-2.65
<b><i>Single Ventricle and Levo-Transposition</i></b>						
Levo-Transposition of the Great Arteries	7	0	0	7	0.49	0.20-1.00
Single Ventricle	4	0	2	6	0.42	0.15-0.91

**Table T.1. Prevalence of Birth Defects, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Live Birth Count	Stillbirth Count	Other Pregnancy Loss Count	Total Count	Rate per 10,000 Live Births	95% Confidence Interval
<b><i>Other Cardiovascular</i></b>						
Other Cardiovascular <sup>2,3</sup>	318	7	31	356	24.78	22.28-27.49
<b>Respiratory</b>						
Choanal Atresia	11	0	0	11	0.77	0.38-1.37
Lung Anomalies	34	1	1	36	2.51	1.76-3.47
Other Respiratory <sup>2</sup>	16	3	3	22	1.53	0.96-2.32
<b>Orofacial</b>						
Cleft Lip with/without Cleft Palate	124	1	15	140	9.75	8.20-11.50
Cleft Palate without Cleft Lip	85	2	4	91	6.33	5.10-7.78
Pierre Robin Sequence	25	0	0	25	1.74	1.13-2.57
Other Orofacial <sup>2</sup>	48	0	3	51	3.55	2.64-4.67
<b>Gastrointestinal</b>						
Biliary Atresia	4	0	0	4	0.28	0.08-0.71
Esophageal Atresia/Tracheoesophageal Fistula	40	0	1	41	2.85	2.05-3.87
Hirschsprung Disease	25	0	0	25	1.74	1.13-2.57
Rectal and Large Intestinal Atresia/Stenosis	53	0	5	58	4.04	3.07-5.22
Small Intestinal Atresia	42	0	1	43	2.99	2.17-4.03
Other Gastrointestinal <sup>2</sup>	102	4	5	111	7.73	6.36-9.30
<b>Genitourinary</b>						
Bladder Exstrophy	5	0	0	5	0.35	0.11-0.81
Cloacal Exstrophy	2	0	0	2	0.14	0.02-0.50
Hypospadias <sup>4</sup> , 1st Degree or NOS	228	0	1	229	31.16	27.26-35.47
Hypospadias <sup>4</sup> , 2nd or 3rd Degree	176	1	0	177	24.08	20.67-27.91
<b>Obstructive Genitourinary Defect</b>	201	2	12	215	14.97	13.03-17.11
Renal Agenesis/Hypoplasia	36	2	10	48	3.34	2.46-4.43
Other Genitourinary <sup>2</sup>	296	6	13	315	21.93	19.57-24.48
<b>Musculoskeletal</b>						
Club Foot	202	5	28	235	16.36	14.33-18.59
Craniosynostosis	88	0	1	89	6.20	4.98-7.62
Diaphragmatic Hernia	38	4	9	51	3.55	2.64-4.67
Gastroschisis	52	2	2	56	3.90	2.94-5.06

**Table T.1. Prevalence of Birth Defects, Massachusetts: 2013-2014**

<b>Defect<sup>1</sup></b>	<b>Live Birth Count</b>	<b>Stillbirth Count</b>	<b>Other Pregnancy Loss Count</b>	<b>Total Count</b>	<b>Rate per 10,000 Live Births</b>	<b>95% Confidence Interval</b>
Omphalocele	18	8	28	54	3.76	2.82-4.90
Polydactyly/Syndactyly	221	1	12	234	16.29	14.27-18.51
Reduction Deformity, Lower Limbs	19	1	8	28	1.95	1.30-2.82
Reduction Deformity, Upper Limbs	48	0	11	59	4.11	3.13-5.30
Skeletal Dysplasia	31	1	8	40	2.78	1.99-3.79
Other Musculoskeletal <sup>2</sup>	159	6	37	202	14.06	12.19-16.14
<b>Chromosomal and other Syndromes</b>						
Klinefelter Syndrome	9	1	3	13	0.90	0.48-1.55
Trisomy 13	4	1	45	50	3.48	2.58-4.59
Trisomy 18	15	13	82	110	7.66	6.29-9.23
Trisomy 21 (Down Syndrome)	165	15	185	365	25.41	22.87-28.15
Turner Syndrome <sup>5</sup>	7	3	68	78	10.83	8.53-13.56
Other Chromosomal Syndromes/Other Syndromes <sup>2</sup>	239	10	231	480	33.41	30.49-36.53
<b>Other</b>						
Amniotic Bands	8	0	4	12	0.84	0.43-1.46
Skin Anomalies <sup>2</sup>	21	0	1	22	1.53	0.96-2.32
Other, Specified <sup>2</sup>	20	1	3	24	1.67	1.07-2.49

NOS: Not Otherwise Specified; ASD: Atrial Septal Defect; VSD: Ventricular Septal Defect.

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

<sup>2</sup> Rate represents a heterogeneous group of defects.

<sup>3</sup> Excludes Patent Ductus Arteriosus.

<sup>4</sup> Rate calculated using male live births.

<sup>5</sup> Rate calculated using female live births.

<b>Table T.2. Prevalence of Selected Birth Defects, Massachusetts vs. United States</b>					
<b>Defect</b>	<b>Count MA</b>	<b>Rate MA<sup>1</sup></b>	<b>95% CI</b>	<b>Rate US<sup>2</sup></b>	<b>95% Confidence Interval</b>
Anencephaly	31	2.16	1.47-3.06	2.23	2.07-2.41
Spina Bifida without Anencephaly	70	4.87	3.80-6.16	3.72	3.52-3.94
Anophthalmia/Microphthalmia	19	1.32	0.80-2.07	2.10	1.94-2.27
Truncus Arteriosus (Common Truncus)	9	0.63	0.29-1.19	0.74	0.65-0.84
Transposition of the Great Arteries	48	3.34	2.46-4.43	3.04	2.85-3.24
Tetralogy of Fallot	77	5.36	4.23-6.70	4.05	3.83-4.28
Atrioventricular Septal Defect <sup>3</sup>	90	6.26	5.04-7.70	4.70	4.45-4.96
Hypoplastic Left Heart Syndrome	39	2.71	1.93-3.71	2.31	2.14-2.48
Cleft Palate without Cleft Lip	91	6.33	5.10-7.78	6.45	6.17-6.74
Cleft Lip with/without Cleft Palate	140	9.75	8.20-11.50	10.89	10.53-11.26
Esophageal Atresia/Tracheoesophageal Fistula	41	2.85	2.05-3.87	2.12	1.96-2.29
Rectal and Large Intestinal Atresia/Stenosis	58	4.04	3.07-5.22	4.86	4.61-5.14
Reduction Deformity, Lower Limbs	28	1.95	1.30-2.82	1.65	1.51-1.80
Reduction Deformity, Upper Limbs	59	4.11	3.13-5.30	3.64	3.43-3.86
Gastroschisis	56	3.90	2.94-5.06	4.72	4.49-4.97
Omphalocele	54	3.76	2.82-4.90	1.92	1.77-2.08
Diaphragmatic Hernia	51	3.55	2.64-4.67	2.60	2.42-2.79
Trisomy 21 (Down Syndrome) <sup>4</sup>	365	18.70	17.88-19.44	14.47	14.11-14.83
Trisomy 13 <sup>4</sup>	50	2.64	2.53-2.75	1.26	1.16-1.37
Trisomy 18 <sup>4</sup>	110	5.39	5.14-5.63	2.66	2.50-2.81

<sup>1</sup> Rate per 10,000 live births. Includes live births, stillbirths, and other pregnancy losses, 2013 and 2014.

<sup>2</sup> Rate per 10,000 live births. US rates based on crude, pooled prevalence data from 11 active case-ascertainment programs, including Massachusetts, from 2004-2006 (7). Nine of the other states contributing to the pooled estimates include elective terminations in addition to live births and stillbirths.

<sup>3</sup> Includes endocardial cushion defect, complete atrioventricular canal defect, atrial septal defect (ASD) primum, common atrium, and canal type ventricular septal defect (VSD).

<sup>4</sup> Adjusted for maternal age distribution of US population 2006.

**Table T.3. Prevalence of Birth Defects by Infant Sex, Live Births, Stillbirths and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Sex	Count	Rate per 10,000 Live Births	95% Confidence Interval
<b>Central Nervous System</b>				
Anencephaly	Male	17	2.31	1.35-3.70
	Female	6	0.86	0.31-1.86
Encephalocele	Male	10	1.36	0.65-2.50
	Female	10	1.43	0.68-2.62
Holoprosencephaly	Male	10	1.36	0.65-2.50
	Female	13	1.85	0.99-3.17
Hydrocephaly	Male	43	5.85	4.23-7.88
	Female	34	4.85	3.36-6.77
Microcephaly	Male	25	3.40	2.20-5.02
	Female	30	4.28	2.88-6.10
Spina Bifida with/without Hydrocephaly	Male	32	4.35	2.98-6.15
	Female	33	4.70	3.24-6.60
Spinal Cord <sup>2</sup>	Male	31	4.22	2.87-5.99
	Female	30	4.28	2.88-6.10
Other Central Nervous System <sup>2</sup>	Male	103	14.02	11.44-17.00
	Female	75	10.69	8.41-13.40
<b>Eye</b>				
Aniridia	Male	4	0.54	0.15-1.39
	Female	2	0.29	0.03-1.03
Anophthalmia/Microphthalmia	Male	8	1.09	0.47-2.14
	Female	11	1.57	0.78-2.80
Congenital Glaucoma, Congenital Cataract	Male	36	4.90	3.43-6.78
	Female	21	2.99	1.85-4.57
Other Eye <sup>2</sup>	Male	24	3.27	2.09-4.86
	Female	43	6.13	4.43-8.25
<b>Ear</b>				
Anotia/Microtia	Male	19	2.59	1.56-4.04
	Female	18	2.57	1.52-4.05
Other Ear <sup>2</sup>	Male	39	5.31	3.77-7.25
	Female	46	6.56	4.80-8.74

**Table T.3. Prevalence of Birth Defects by Infant Sex, Live Births, Stillbirths and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Sex	Count	Rate per 10,000 Live Births	95% Confidence Interval
<b>Cardiovascular</b>				
<i>Anomalous Pulmonary Venous Connection</i>				
Anomalous Pulmonary Venous Connection	Male	14	1.91	1.04-3.20
	Female	14	2.00	1.09-3.35
<i>Atrioventricular Canal Defects</i>				
Atrial Septal Defect (ASD) Primum	Male	2	0.27	0.03-0.98
	Female	1	0.14	0.00-0.79
Common Atrium	Male	1	0.14	0.00-0.76
	Female	0	0	-
Complete Atrioventricular Canal Defect	Male	33	4.49	3.09-6.31
	Female	30	4.28	2.88-6.10
Endocardial Cushion Defect	Male	5	0.68	0.22-1.59
	Female	10	1.43	0.68-2.62
Ventricular Septal Defect (VSD), Canal Type	Male	4	0.54	0.15-1.39
	Female	2	0.29	0.03-1.03
<i>Conotruncal (Outlet) and Aortic Arch</i>				
Double Outlet Right Ventricle	Male	22	2.99	1.88-4.53
	Female	10	1.43	0.68-2.62
Interrupted Aortic Arch, Type B	Male	1	0.14	0.00-0.76
	Female	2	0.29	0.03-1.03
Tetralogy of Fallot	Male	35	4.76	3.32-6.62
	Female	41	5.84	4.19-7.93
Truncus Arteriosus (Common Truncus)	Male	3	0.41	0.08-1.19
	Female	6	0.86	0.31-1.86
Dextro-Transposition of the Great Arteries	Male	28	3.81	2.53-5.51
	Female	13	1.85	0.99-3.17
<i>Ebstein Anomaly</i>				
Ebstein Anomaly	Male	2	0.27	0.03-0.98
	Female	5	0.71	0.23-1.66



**Table T.3. Prevalence of Birth Defects by Infant Sex, Live Births, Stillbirths and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Sex	Count	Rate per 10,000 Live Births	95% Confidence Interval
<b><i>Heterotaxy (Laterality Defects)</i></b>				
Heterotaxy	Male	13	1.77	0.94-3.02
	Female	10	1.43	0.68-2.62
<b><i>Left-Sided Obstruction</i></b>				
Aortic Arch Atresia without Hypoplastic Left Heart Syndrome	Male	1	0.14	0.00-0.76
	Female	0	-	-
Aortic Valve Stenosis	Male	15	2.04	1.14-3.37
	Female	3	0.43	0.09-1.25
Coarctation of Aorta	Male	37	5.03	3.54-6.94
	Female	32	4.56	3.12-6.44
Hypoplastic Left Heart Syndrome	Male	22	2.99	1.88-4.53
	Female	11	1.57	0.78-2.80
Interrupted Aortic Arch (Type A and NOS)	Male	1	0.14	0.00-0.76
	Female	1	0.14	0.00-0.79
<b><i>Right-Sided Obstruction</i></b>				
Pulmonary Stenosis, Valvular	Male	52	7.08	5.28-9.28
	Female	58	8.27	6.28-10.68
Pulmonary Valve Atresia with intact septum	Male	4	0.54	0.15-1.39
	Female	5	0.71	0.23-1.66
Pulmonary Valve Atresia with VSD	Male	6	0.82	0.30-1.78
	Female	3	0.43	0.09-1.25
Tricuspid Valve Atresia	Male	5	0.68	0.22-1.59
	Female	6	0.86	0.31-1.86
<b><i>Septal Defects</i></b>				
ASD (Secundum and NOS)	Male	168	22.86	19.53-26.59
	Female	165	23.51	20.06-27.39
VSD (Membranous and NOS)	Male	101	13.74	11.19-16.70
	Female	107	15.25	12.50-18.43
VSD (Muscular)	Male	70	9.53	7.43-12.03
	Female	91	12.97	10.44-15.92

**Table T.3. Prevalence of Birth Defects by Infant Sex, Live Births, Stillbirths and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Sex	Count	Rate per 10,000 Live Births	95% Confidence Interval
VSD, Conoventricular/Malalignment	Male	15	2.04	1.14-3.37
	Female	11	1.57	0.78-2.80
<b><i>Single Ventricle and Levo-Transposition</i></b>				
Levo-Transposition of the Great Arteries	Male	5	0.68	0.22-1.59
	Female	2	0.29	0.03-1.03
Single Ventricle	Male	4	0.54	0.15-1.39
	Female	1	0.14	0.00-0.79
<b><i>Other Cardiovascular</i></b>				
Other Cardiovascular <sup>2,3</sup>	Male	195	26.53	22.94-30.53
	Female	155	22.09	18.75-25.85
<b>Respiratory</b>				
Choanal Atresia	Male	4	0.54	0.15-1.39
	Female	7	1.00	0.40-2.06
Lung Anomalies	Male	19	2.59	1.56-4.04
	Female	17	2.42	1.41-3.88
Other Respiratory <sup>2</sup>	Male	17	2.31	1.35-3.70
	Female	4	0.57	0.16-1.46
<b>Orofacial</b>				
Cleft Lip with/without Cleft Palate	Male	84	11.43	9.12-14.15
	Female	53	7.55	5.66-9.88
Cleft Palate without Cleft Lip	Male	38	5.17	3.66-7.10
	Female	53	7.55	5.66-9.88
Pierre Robin Sequence	Male	14	1.91	1.04-3.20
	Female	11	1.57	0.78-2.80
Other Orofacial <sup>2</sup>	Male	23	3.13	1.98-4.70
	Female	27	3.85	2.54-5.60
<b>Gastrointestinal</b>				
Biliary Atresia	Male	1	0.14	0.00-0.76
	Female	3	0.43	0.09-1.25
Esophageal Atresia/Tracheoesophageal Fistula	Male	25	3.40	2.20-5.02
	Female	16	2.28	1.30-3.70

**Table T.3. Prevalence of Birth Defects by Infant Sex, Live Births, Stillbirths and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Sex	Count	Rate per 10,000 Live Births	95% Confidence Interval
Hirschsprung Disease	Male	17	2.31	1.35-3.70
	Female	8	1.14	0.49-2.25
Rectal and Large Intestinal Atresia/Stenosis	Male	27	3.67	2.42-5.35
	Female	31	4.42	3.00-6.27
Small Intestinal Atresia	Male	26	3.54	2.31-5.18
	Female	17	2.42	1.41-3.88
Other Gastrointestinal <sup>2</sup>	Male	58	7.89	5.99-10.20
	Female	53	7.55	5.66-9.88
<b>Genitourinary</b>				
Bladder Exstrophy	Male	2	0.27	0.03-0.98
	Female	3	0.43	0.09-1.25
Cloacal Exstrophy	Male	0	0	-
	Female	2	0.29	0.03-1.03
Hypospadias <sup>4</sup> , 1st Degree or NOS	Male	229	31.16	27.26-35.47
	Female	0	-	-
Hypospadias <sup>4</sup> , 2nd or 3rd Degree	Male	177	24.08	20.67-27.91
	Female	0	0	-
Obstructive Genitourinary Defect	Male	143	19.46	16.40-22.92
	Female	72	10.26	8.03-12.92
Renal Agenesis/Hypoplasia	Male	30	4.08	2.75-5.83
	Female	17	2.42	1.41-3.88
Other Genitourinary <sup>2</sup>	Male	197	26.81	23.19-30.82
	Female	113	16.10	13.27-19.36
<b>Musculoskeletal</b>				
Club Foot	Male	146	19.87	16.77-23.36
	Female	83	11.83	9.42-14.66
Craniosynostosis	Male	57	7.76	5.87-10.05
	Female	32	4.56	3.12-6.44
Diaphragmatic Hernia	Male	18	2.45	1.45-3.87
	Female	32	4.56	3.12-6.44

**Table T.3. Prevalence of Birth Defects by Infant Sex, Live Births, Stillbirths and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Sex	Count	Rate per 10,000 Live Births	95% Confidence Interval
Gastroschisis	Male	32	4.35	2.98-6.15
	Female	24	3.42	2.19-5.09
Omphalocele	Male	25	3.40	2.20-5.02
	Female	23	3.28	2.08-4.92
Polydactyly/Syndactyly	Male	137	18.64	15.65-22.04
	Female	96	13.68	11.08-16.71
Reduction Deformity, Lower Limbs	Male	20	2.72	1.66-4.20
	Female	6	0.86	0.31-1.86
Reduction Deformity, Upper Limbs	Male	30	4.08	2.75-5.83
	Female	28	3.99	2.65-5.77
Skeletal Dysplasia	Male	21	2.86	1.77-4.37
	Female	18	2.57	1.52-4.05
Other Musculoskeletal <sup>2</sup>	Male	101	13.74	11.19-16.70
	Female	93	13.25	10.70-16.24
<b>Chromosomal and other Syndromes</b>				
Klinefelter Syndrome	Male	13	1.77	0.94-3.02
	Female	0	-	-
Trisomy 13	Male	24	3.27	2.09-4.86
	Female	23	3.28	2.08-4.92
Trisomy 18	Male	55	7.48	5.64-9.74
	Female	46	6.56	4.80-8.74
Trisomy 21 (Down Syndrome)	Male	175	23.81	20.42-27.61
	Female	174	24.80	21.25-28.77
Turner Syndrome <sup>5</sup>	Male	1	0.14	0.00-0.76
	Female	76	10.83	8.53-13.56
Other Chromosomal Syndromes/Other Syndromes <sup>2</sup>	Male	235	31.98	28.02-36.34
	Female	238	33.92	29.74-38.51
<b>Other</b>				
Amniotic Bands	Male	3	0.41	0.08-1.19
	Female	7	1.00	0.40-2.06

**Table T.3. Prevalence of Birth Defects by Infant Sex, Live Births, Stillbirths and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Sex	Count	Rate per 10,000 Live Births	95% Confidence Interval
Skin Anomalies	Male	14	1.91	1.04-3.20
	Female	8	1.14	0.49-2.25
Other, Specified <sup>2</sup>	Male	16	2.18	1.24-3.54
	Female	8	1.14	0.49-2.25

NOS: Not Otherwise Specified. ASD: Atrial Septal Defect; VSD: Ventricular Septal Defect.

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

<sup>2</sup> Rate represents a heterogeneous group of defects.

<sup>3</sup> Excludes Patent Ductus Arteriosus.

<sup>4</sup> Rate calculated using male live births.

<sup>5</sup> Rate calculated using female live births.

<b>Table T.4. Prevalence of Birth Defects by Plurality, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014</b>				
<b>Defect<sup>1</sup></b>	<b>Plurality</b>	<b>Count</b>	<b>Rate per 10,000 Live Births</b>	<b>95% Confidence Interval</b>
<b>Central Nervous System</b>				
Anencephaly	Singleton	31	2.26	1.53-3.20
	Multiple	0	0	-
Encephalocele	Singleton	23	1.67	1.06-2.51
	Multiple	0	0	-
Holoprosencephaly	Singleton	23	1.67	1.06-2.51
	Multiple	0	0	-
Hydrocephaly without Spina Bifida	Singleton	77	5.61	4.43-7.01
	Multiple	2	3.16	0.38-11.40
Microcephaly	Singleton	55	4.01	3.02-5.21
	Multiple	1	1.58	0.04-8.79
Spina Bifida with/without Hydrocephaly	Singleton	69	5.02	3.91-6.36
	Multiple	1	1.58	0.04-8.79
Spinal Cord <sup>2</sup>	Singleton	57	4.15	3.14-5.38
	Multiple	4	6.31	1.72-16.17
Other Central Nervous System <sup>2</sup>	Singleton	179	13.03	11.19-15.09
	Multiple	6	9.47	3.48-20.61
<b>Eye</b>				
Aniridia	Singleton	6	0.44	0.16-0.95
	Multiple	0	0	-
Anophthalmia/Microphthalmia	Singleton	19	1.38	0.83-2.16
	Multiple	0	0	-
Congenital Glaucoma, Congenital Cataract	Singleton	57	4.15	3.14-5.38
	Multiple	0	0	-
Other Eye <sup>2</sup>	Singleton	66	4.81	3.72-6.11
	Multiple	1	1.58	0.04-8.79
<b>Ear</b>				
Anotia/Microtia	Singleton	37	2.69	1.90-3.71
	Multiple	1	1.58	0.04-8.79
Other Ear <sup>2</sup>	Singleton	82	5.97	4.75-7.41
	Multiple	3	4.74	0.98-13.84

<b>Table T.4. Prevalence of Birth Defects by Plurality, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014</b>				
<b>Defect<sup>1</sup></b>	<b>Plurality</b>	<b>Count</b>	<b>Rate per 10,000 Live Births</b>	<b>95% Confidence Interval</b>
<b>Cardiovascular</b>				
<i>Anomalous Pulmonary Venous Connection</i>				
Anomalous Pulmonary Venous Connection	Singleton	27	1.97	1.30-2.86
	Multiple	1	1.58	0.04-8.79
<i>Atrioventricular Canal Defects</i>				
Atrial Septal Defect (ASD) primum	Singleton	3	0.22	0.05-0.64
	Multiple	0	0	-
Common Atrium	Singleton	1	0.07	0.00-0.41
	Multiple	0	0	-
Complete Atrioventricular Canal Defect	Singleton	62	4.51	3.46-5.79
	Multiple	2	3.16	0.38-11.40
Endocardial Cushion Defect	Singleton	14	1.02	0.56-1.71
	Multiple	2	3.16	0.38-11.40
Ventricular Septal Defect (VSD), Canal Type	Singleton	6	0.44	0.16-0.95
	Multiple	0	0	-
<i>Conotruncal (Outlet) and Aortic Arch</i>				
Double Outlet Right Ventricle	Singleton	33	2.40	1.65-3.37
	Multiple	0	0	-
Interrupted Aortic Arch, Type B	Singleton	3	0.22	0.05-0.64
	Multiple	0	0	-
Tetralogy of Fallot	Singleton	73	5.32	4.17-6.68
	Multiple	4	6.31	1.72-16.17
Truncus Arteriosus (Common Truncus)	Singleton	7	0.51	0.20-1.05
	Multiple	2	3.16	0.38-11.40
Dextro-Transposition of the Great Arteries	Singleton	39	2.84	2.02-3.88
	Multiple	2	3.16	0.38-11.40
<i>Ebstein Anomaly</i>				
Ebstein Anomaly	Singleton	7	0.51	0.20-1.05
	Multiple	0	0	-
<i>Heterotaxy (Laterality Defects)</i>				
Heterotaxy	Singleton	23	1.67	1.06-2.51
	Multiple	1	1.58	0.04-8.79

<b>Table T.4. Prevalence of Birth Defects by Plurality, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014</b>				
<b>Defect<sup>1</sup></b>	<b>Plurality</b>	<b>Count</b>	<b>Rate per 10,000 Live Births</b>	<b>95% Confidence Interval</b>
<b><i>Left-Sided Obstruction</i></b>				
Aortic Valve Atresia	Singleton	1	0.07	0.00-0.41
	Multiple	0	0	-
Aortic Valve Stenosis	Singleton	17	1.24	0.72-1.98
	Multiple	1	1.58	0.04-8.79
Coarctation of Aorta	Singleton	61	4.44	3.40-5.71
	Multiple	8	12.63	5.45-24.88
Hypoplastic Left Heart Syndrome	Singleton	38	2.77	1.96-3.80
	Multiple	1	1.58	0.04-8.79
Interrupted Aortic Arch (Type A and NOS)	Singleton	2	0.15	0.02-0.53
	Multiple	0	0	-
<b><i>Right-Sided Obstruction</i></b>				
Pulmonary Stenosis, Valvar	Singleton	97	7.06	5.73-8.62
	Multiple	14	22.10	12.08-37.08
Pulmonary Valve Atresia with intact septum	Singleton	8	0.58	0.25-1.15
	Multiple	1	1.58	0.04-8.79
Pulmonary Valve Atresia with Ventricular Septal Defect	Singleton	9	0.66	0.30-1.24
	Multiple	1	1.58	0.04-8.79
Tricuspid Valve Atresia	Singleton	10	0.73	0.35-1.34
	Multiple	1	1.58	0.04-8.79
<b><i>Septal Defects</i></b>				
ASD (Secundum and NOS)	Singleton	307	22.36	19.93-25.00
	Multiple	27	42.62	28.09-62.01
VSD (Membranous and NOS)	Singleton	193	14.05	12.14-16.18
	Multiple	18	28.41	16.84-44.91
VSD, Muscular	Singleton	145	10.56	8.91-12.42
	Multiple	17	26.84	15.63-42.97
VSD, Conoventricular/Malalignment	Singleton	25	1.82	1.18-2.69
	Multiple	1	1.58	0.04-8.79
<b><i>Single Ventricle and L-TGA</i></b>				
Levo-Transposition of the Great Arteries	Singleton	6	0.44	0.16-0.95
	Multiple	1	1.58	0.04-8.79



<b>Table T.4. Prevalence of Birth Defects by Plurality, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014</b>				
<b>Defect<sup>1</sup></b>	<b>Plurality</b>	<b>Count</b>	<b>Rate per 10,000 Live Births</b>	<b>95% Confidence Interval</b>
Single Ventricle	Singleton	6	0.44	0.16-0.95
	Multiple	0	0	-
Other Cardiovascular <sup>2,3</sup>	Singleton	340	24.76	22.20-27.53
	Multiple	16	25.26	14.44-41.01
<b>Respiratory</b>				
Choanal Atresia	Singleton	11	0.80	0.40-1.43
	Multiple	0	0	-
Lung Anomalies	Singleton	35	2.55	1.78-3.54
	Multiple	1	1.58	0.04-8.79
Other Respiratory <sup>2</sup>	Singleton	21	1.53	0.95-2.34
	Multiple	1	1.58	0.04-8.79
<b>Orofacial</b>				
Cleft Lip with and without Cleft Palate	Singleton	133	9.68	8.11-11.48
	Multiple	7	11.05	4.44-22.77
Cleft Palate without Cleft Lip	Singleton	87	6.34	5.07-7.81
	Multiple	4	6.31	1.72-16.17
Pierre Robin Sequence	Singleton	23	1.67	1.06-2.51
	Multiple	2	3.16	0.38-11.40
Other Orofacial <sup>2</sup>	Singleton	46	3.35	2.45-4.47
	Multiple	5	7.89	2.56-18.42
<b>Gastrointestinal</b>				
Biliary Atresia	Singleton	4	0.29	0.08-0.75
	Multiple	0	0	-
Esophageal Atresia/Tracheoesophageal Fistula	Singleton	38	2.77	1.96-3.80
	Multiple	3	4.74	0.98-13.84
Hirschsprung Disease	Singleton	24	1.75	1.12-2.60
	Multiple	1	1.58	0.04-8.79
Rectal and Large Intestinal Atresia/Stenosis	Singleton	57	4.15	3.14-5.38
	Multiple	1	1.58	0.04-8.79
Small Intestinal Atresia	Singleton	39	2.84	2.02-3.88
	Multiple	4	6.31	1.72-16.17

<b>Table T.4. Prevalence of Birth Defects by Plurality, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014</b>				
<b>Defect<sup>1</sup></b>	<b>Plurality</b>	<b>Count</b>	<b>Rate per 10,000 Live Births</b>	<b>95% Confidence Interval</b>
Other Gastrointestinal <sup>2</sup>	Singleton	107	7.79	6.39-9.42
	Multiple	4	6.31	1.72-16.17
<b>Genitourinary</b>				
Bladder Exstrophy	Singleton	5	0.36	0.12-0.85
	Multiple	0	0	-
Cloacal Exstrophy	Singleton	2	0.15	0.02-0.53
	Multiple	0	0	-
Hypospadias <sup>4</sup> , 1 <sup>st</sup> Degree or NOS	Singleton	206	29.30	25.43-33.58
	Multiple	23	72.42	45.91-108.66
Hypospadias <sup>4</sup> , 2nd or 3rd Degree	Singleton	167	23.75	20.28-27.64
	Multiple	10	31.49	15.1-57.9
Obstructive Genitourinary Defect	Singleton	201	14.64	12.68-16.81
	Multiple	14	22.10	12.08-37.08
Renal Agenesis/Hypoplasia	Singleton	46	3.35	2.45-4.47
	Multiple	2	3.16	0.38-11.40
Other Genitourinary <sup>2</sup>	Singleton	300	21.85	19.44-24.46
	Multiple	15	23.68	13.25-39.05
<b>Musculoskeletal</b>				
Club Foot	Singleton	222	16.17	14.11-18.44
	Multiple	13	20.52	10.93-35.09
Craniosynostosis	Singleton	84	6.12	4.88-7.57
	Multiple	5	7.89	2.56-18.42
Diaphragmatic Hernia	Singleton	47	3.42	2.51-4.55
	Multiple	4	6.31	1.72-16.17
Gastroschisis	Singleton	54	3.93	2.95-5.13
	Multiple	2	3.16	0.38-11.40
Omphalocele	Singleton	50	3.64	2.70-4.80
	Multiple	4	6.31	1.72-16.17
Polydactyly/Syndactyly	Singleton	223	16.24	14.18-18.52
	Multiple	11	17.36	8.67-31.07
Reduction Deformity, Lower Limbs	Singleton	25	1.82	1.18-2.69
	Multiple	3	4.74	0.98-13.84

<b>Table T.4. Prevalence of Birth Defects by Plurality, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014</b>				
<b>Defect<sup>1</sup></b>	<b>Plurality</b>	<b>Count</b>	<b>Rate per 10,000 Live Births</b>	<b>95% Confidence Interval</b>
Reduction Deformity, Upper Limbs	Singleton	55	4.01	3.02-5.21
	Multiple	4	6.31	1.72-16.17
Skeletal Dysplasia	Singleton	39	2.84	2.02-3.88
	Multiple	1	1.58	0.04-8.79
Other Musculoskeletal <sup>2</sup>	Singleton	189	13.76	11.87-15.87
	Multiple	13	20.52	10.93-35.09
<b>Chromosomal/other Syndromes</b>				
Klinefelter Syndrome	Singleton	13	0.95	0.50-1.62
	Multiple	0	0	-
Trisomy 13	Singleton	50	3.64	2.70-4.80
	Multiple	0	0	-
Trisomy 18	Singleton	107	7.79	6.39-9.42
	Multiple	3	4.74	0.98-13.84
Trisomy 21 (Down Syndrome)	Singleton	357	26.00	23.37-28.83
	Multiple	8	12.63	5.45-24.88
Turner Syndrome <sup>5</sup>	Singleton	78	10.83	8.53-13.56
	Multiple	0	0	-
Other Chromosomal <sup>2</sup>	Singleton	468	34.08	31.07-37.31
	Multiple	12	18.94	9.79-33.09
<b>Other</b>				
Amniotic Bands	Singleton	11	0.80	0.40-1.43
	Multiple	1	1.58	0.04-8.79
Skin Anomalies	Singleton	22	1.60	1.00-2.43
	Multiple	0	0	-
Other, Specified <sup>2</sup>	Singleton	23	1.67	1.06-2.51
	Multiple	1	1.58	0.04-8.79

NOS: Not Otherwise Specified. ASD: Atrial Septal Defect; VSD: Ventricular Septal Defect.

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

<sup>2</sup> Rate represents a heterogeneous group of defects.

<sup>3</sup> Excludes Patent Ductus Arteriosus.

<sup>4</sup> Rate calculated using male live births.

<sup>5</sup> Rate calculated using female live births.

**Table T.5. Prevalence of Birth Defects by Maternal Age, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Age	Count	Rate per 10,000 Live Births	95% Confidence Interval
<b>Central Nervous System</b>				
Anencephaly	<20	2	3.86	0.47-13.93
	20-24	4	2.05	0.56-5.24
	25-29	12	3.38	1.75-5.91
	30-34	8	1.60	0.69-3.15
	35+	5	1.50	0.49-3.50
Encephalocele	<20	0	0	-
	20-24	6	3.07	1.13-6.68
	25-29	5	1.41	0.46-3.29
	30-34	6	1.20	0.44-2.61
	35+	6	1.80	0.66-3.91
Holoprosencephaly	<20	2	3.86	0.47-13.93
	20-24	4	2.05	0.56-5.24
	25-29	6	1.69	0.62-3.68
	30-34	6	1.20	0.44-2.61
	35+	5	1.50	0.49-3.50
Hydrocephaly without Spina Bifida	<20	5	9.64	3.13-22.50
	20-24	20	10.23	6.25-15.79
	25-29	15	4.23	2.37-6.98
	30-34	18	3.59	2.13-5.68
	35+	21	6.30	3.90-9.62
Microcephaly	<20	5	9.64	3.13-22.50
	20-24	10	5.11	2.45-9.40
	25-29	19	5.36	3.23-8.37
	30-34	12	2.39	1.24-4.18
	35+	10	3.00	1.44-5.51
Spina Bifida with/without Hydrocephaly	<20	3	5.78	1.19-16.90
	20-24	7	3.58	1.44-7.37
	25-29	12	3.38	1.75-5.91
	30-34	28	5.59	3.71-8.08
	35+	20	6.00	3.66-9.26

**Table T.5. Prevalence of Birth Defects by Maternal Age, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Age	Count	Rate per 10,000 Live Births	95% Confidence Interval
Spinal Cord <sup>2</sup>	<20	4	7.71	2.10-19.74
	20-24	8	4.09	1.77-8.06
	25-29	10	2.82	1.35-5.19
	30-34	28	5.59	3.71-8.08
	35+	11	3.30	1.65-5.90
Other Central Nervous System <sup>2</sup>	<20	11	21.21	10.59-37.94
	20-24	36	18.41	12.89-25.48
	25-29	43	12.13	8.78-16.34
	30-34	53	10.58	7.92-13.84
	35+	42	12.59	9.07-17.02
<b>Eye</b>				
Aniridia	<20	0	0	-
	20-24	0	0	-
	25-29	2	0.56	0.07-2.04
	30-34	4	0.80	0.22-2.04
	35+	0	0	-
Anophthalmia/Microphthalmia	<20	1	1.93	0.05-10.74
	20-24	5	2.56	0.83-5.97
	25-29	1	0.28	0.01-1.57
	30-34	4	0.80	0.22-2.04
	35+	8	2.40	1.04-4.73
Congenital Glaucoma, Congenital Cataract	<20	1	1.93	0.05-10.74
	20-24	12	6.14	3.17-10.72
	25-29	12	3.38	1.75-5.91
	30-34	21	4.19	2.59-6.41
	35+	11	3.30	1.65-5.90
Other Eye <sup>2</sup>	<20	2	3.86	0.47-13.93
	20-24	16	8.18	4.68-13.29
	25-29	10	2.82	1.35-5.19
	30-34	25	4.99	3.23-7.37
	35+	14	4.20	2.29-7.04

**Table T.5. Prevalence of Birth Defects by Maternal Age, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Age	Count	Rate per 10,000 Live Births	95% Confidence Interval
<b>Ear</b>				
Anotia/Microtia	<20	1	1.93	0.05-10.74
	20-24	7	3.58	1.44-7.37
	25-29	13	3.67	1.95-6.27
	30-34	12	2.39	1.24-4.18
	35+	5	1.50	0.49-3.50
Other Ear <sup>2</sup>	<20	0	0	-
	20-24	7	3.58	1.44-7.37
	25-29	22	6.21	3.89-9.40
	30-34	31	6.19	4.20-8.78
	35+	25	7.49	4.85-11.06
<b>Cardiovascular</b>				
<i>Anomalous Pulmonary Venous Connection</i>				
Anomalous Pulmonary Venous Connection	<20	0	0	-
	20-24	4	2.05	0.56-5.24
	25-29	5	1.41	0.46-3.29
	30-34	10	2.00	0.96-3.67
	35+	9	2.70	1.23-5.12
<i>Atrioventricular Canal Defects</i>				
Atrial Septal Defect (ASD) Primum	<20	0	0	-
	20-24	0	0	-
	25-29	1	0.28	0.01-1.57
	30-34	1	0.20	0.01-1.11
	35+	1	0.30	0.01-1.67
Common Atrium	<20	0	0	-
	20-24	0	0	-
	25-29	1	0.28	0.01-1.57
	30-34	0	0	-
	35+	0	0	-

**Table T.5. Prevalence of Birth Defects by Maternal Age, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Age	Count	Rate per 10,000 Live Births	95% Confidence Interval
Complete Atrioventricular Canal Defect	<20	1	1.93	0.05-10.74
	20-24	9	4.60	2.10-8.74
	25-29	13	3.67	1.95-6.27
	30-34	16	3.19	1.83-5.19
	35+	25	7.49	4.85-11.06
Endocardial Cushion Defect	<20	1	1.93	0.05-10.74
	20-24	0	0	-
	25-29	0	0	-
	30-34	8	1.60	0.69-3.15
	35+	7	2.10	0.84-4.32
Ventricular Septal Defect (VSD), Canal Type	<20	0	0	-
	20-24	0	0	-
	25-29	1	0.28	0.01-1.57
	30-34	2	0.40	0.05-1.44
	35+	3	0.90	0.19-2.63
<b><i>Conotruncal (Outlet) and Aortic Arch</i></b>				
Double Outlet Right Ventricle	<20	1	1.93	0.05-10.74
	20-24	7	3.58	1.44-7.37
	25-29	7	1.97	0.79-4.07
	30-34	11	2.20	1.10-3.93
	35+	7	2.10	0.84-4.32
Interrupted Aortic Arch, Type B	<20	1	1.93	0.05-10.74
	20-24	1	0.51	0.01-2.85
	25-29	0	0	-
	30-34	1	0.20	0.01-1.11
	35+	0	0	-
Tetralogy of Fallot	<20	2	3.86	0.47-13.93
	20-24	7	3.58	1.44-7.37
	25-29	17	4.80	2.79-7.68
	30-34	30	5.99	4.04-8.55
	35+	21	6.30	3.90-9.62

**Table T.5. Prevalence of Birth Defects by Maternal Age, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Age	Count	Rate per 10,000 Live Births	95% Confidence Interval
Truncus Arteriosus (Common Truncus)	<20	1	1.93	0.05-10.74
	20-24	2	1.02	0.12-3.69
	25-29	0	0	-
	30-34	4	0.80	0.22-2.04
	35+	2	0.60	0.07-2.17
Dextro-Transposition of the Great Arteries	<20	1	1.93	0.05-10.74
	20-24	6	3.07	1.13-6.68
	25-29	8	2.26	0.97-4.45
	30-34	16	3.19	1.83-5.19
	35+	10	3.00	1.44-5.51
<b><i>Ebstein Anomaly</i></b>				
Ebstein Anomaly	<20	0	0	-
	20-24	0	0	-
	25-29	2	0.56	0.07-2.04
	30-34	2	0.40	0.05-1.44
	35+	3	0.90	0.19-2.63
<b><i>Heterotaxy (Laterality Defects)</i></b>				
Heterotaxy	<20	1	1.93	0.05-10.74
	20-24	5	2.56	0.83-5.97
	25-29	5	1.41	0.46-3.29
	30-34	9	1.80	0.82-3.41
	35+	4	1.20	0.33-3.07
<b><i>Left-Sided Obstruction</i></b>				
Aortic Arch Atresia without Hypoplastic Left Heart	<20	0	0	-
	20-24	1	0.51	0.01-2.85
	25-29	0	0	-
	30-34	0	0	-
	35+	0	0	-



**Table T.5. Prevalence of Birth Defects by Maternal Age, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Age	Count	Rate per 10,000 Live Births	95% Confidence Interval
Aortic Valve Stenosis	<20	0	0	-
	20-24	2	1.02	0.12-3.69
	25-29	6	1.69	0.62-3.68
	30-34	6	1.20	0.44-2.61
	35+	4	1.20	0.33-3.07
Coarctation of Aorta	<20	3	5.78	1.19-16.90
	20-24	6	3.07	1.13-6.68
	25-29	13	3.67	1.95-6.27
	30-34	36	7.18	5.03-9.95
	35+	11	3.30	1.65-5.90
Hypoplastic Left Heart Syndrome	<20	0	0	-
	20-24	5	2.56	0.83-5.97
	25-29	8	2.26	0.97-4.45
	30-34	18	3.59	2.13-5.68
	35+	8	2.40	1.04-4.73
Interrupted Aortic Arch (Type A and NOS)	<20	0	0	-
	20-24	1	0.51	0.01-2.85
	25-29	0	0	-
	30-34	1	0.20	0.01-1.11
	35+	0	0	-
<b><i>Right-Sided Obstruction</i></b>				
Pulmonary Stenosis, Valvular	<20	5	9.64	3.13-22.50
	20-24	11	5.62	2.81-10.06
	25-29	26	7.33	4.79-10.75
	30-34	34	6.79	4.70-9.48
	35+	35	10.49	7.31-14.59
Pulmonary Valve Atresia with Intact septum	<20	1	1.93	0.05-10.74
	20-24	1	0.51	0.01-2.85
	25-29	2	0.56	0.07-2.04
	30-34	2	0.40	0.05-1.44
	35+	3	0.90	0.19-2.63

**Table T.5. Prevalence of Birth Defects by Maternal Age, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Age	Count	Rate per 10,000 Live Births	95% Confidence Interval
Pulmonary Valve Atresia with VSD	<20	1	1.93	0.05-10.74
	20-24	1	0.51	0.01-2.85
	25-29	2	0.56	0.07-2.04
	30-34	3	0.60	0.12-1.75
	35+	3	0.90	0.19-2.63
Tricuspid Valve Atresia	<20	0	0	-
	20-24	2	1.02	0.12-3.69
	25-29	1	0.28	0.01-1.57
	30-34	5	1.00	0.32-2.33
	35+	3	0.90	0.19-2.63
<b><i>Septal Defects</i></b>				
ASD (Secundum and NOS)	<20	14	26.99	14.76-45.29
	20-24	36	18.41	12.89-25.48
	25-29	63	17.77	13.66-22.74
	30-34	114	22.75	18.77-27.33
	35+	107	32.08	26.29-38.76
VSD (Membranous and NOS)	<20	14	26.99	14.76-45.29
	20-24	19	9.71	5.85-15.17
	25-29	51	14.39	10.71-18.92
	30-34	62	12.37	9.49-15.86
	35+	65	19.48	15.04-24.84
VSD (Muscular)	<20	4	7.71	2.10-19.74
	20-24	19	9.71	5.85-15.17
	25-29	31	8.74	5.94-12.41
	30-34	67	13.37	10.36-16.98
	35+	41	12.29	8.82-16.67
VSD, Conoventricular/Malalignment	<20	2	3.86	0.47-13.93
	20-24	3	1.53	0.32-4.48
	25-29	5	1.41	0.46-3.29
	30-34	7	1.40	0.56-2.88
	35+	9	2.70	1.23-5.12

**Table T.5. Prevalence of Birth Defects by Maternal Age, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Age	Count	Rate per 10,000 Live Births	95% Confidence Interval
<b><i>Single Ventricle and Levo-Transposition</i></b>				
Levo-Transposition of the Great Arteries	<20	0	0	-
	20-24	2	1.02	0.12-3.69
	25-29	0	0	-
	30-34	2	0.40	0.05-1.44
	35+	3	0.90	0.19-2.63
Single Ventricle	<20	0	0	-
	20-24	1	0.51	0.01-2.85
	25-29	3	0.85	0.17-2.47
	30-34	1	0.20	0.01-1.11
	35+	1	0.30	0.01-1.67
<b><i>Other Cardiovascular</i></b>				
Other Cardiovascular <sup>2,3</sup>	<20	16	30.85	17.63-50.09
	20-24	41	20.96	15.04-28.44
	25-29	77	21.72	17.14-27.15
	30-34	132	26.34	22.04-31.24
	35+	90	26.98	21.69-33.16
<b><i>Respiratory</i></b>				
Choanal Atresia	<20	0	0	-
	20-24	2	1.02	0.12-3.69
	25-29	2	0.56	0.07-2.04
	30-34	5	1.00	0.32-2.33
	35+	2	0.60	0.07-2.17
Lung Anomalies <sup>2</sup>	<20	1	1.93	0.05-10.74
	20-24	1	0.51	0.01-2.85
	25-29	10	2.82	1.35-5.19
	30-34	13	2.59	1.38-4.44
	35+	11	3.30	1.65-5.90

**Table T.5. Prevalence of Birth Defects by Maternal Age, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Age	Count	Rate per 10,000 Live Births	95% Confidence Interval
Other Respiratory	<20	1	1.93	0.05-10.74
	20-24	4	2.05	0.56-5.24
	25-29	3	0.85	0.17-2.47
	30-34	8	1.60	0.69-3.15
	35+	6	1.80	0.66-3.91
<b>Orofacial</b>				
Cleft Lip with/without Cleft Palate	<20	2	3.86	0.47-13.93
	20-24	14	7.16	3.91-12.01
	25-29	37	10.44	7.35-14.39
	30-34	47	9.38	6.89-12.47
	35+	40	11.99	8.57-16.33
Cleft Palate without Cleft Lip	<20	4	7.71	2.10-19.74
	20-24	9	4.60	2.10-8.74
	25-29	24	6.77	4.34-10.07
	30-34	29	5.79	3.88-8.31
	35+	25	7.49	4.85-11.06
Pierre Robin Sequence	<20	1	1.93	0.05-10.74
	20-24	4	2.05	0.56-5.24
	25-29	6	1.69	0.62-3.68
	30-34	10	2.00	0.96-3.67
	35+	4	1.20	0.33-3.07
Other Orofacial <sup>2</sup>	<20	0	0	-
	20-24	11	5.62	2.81-10.06
	25-29	7	1.97	0.79-4.07
	30-34	21	4.19	2.59-6.41
	35+	12	3.60	1.86-6.28

**Table T.5. Prevalence of Birth Defects by Maternal Age, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Age	Count	Rate per 10,000 Live Births	95% Confidence Interval
<b>Gastrointestinal</b>				
Biliary Atresia	<20	0	0	-
	20-24	0	0	-
	25-29	1	0.28	0.01-1.57
	30-34	3	0.60	0.12-1.75
	35+	0	0	-
Esophageal Atresia/Tracheoesophageal Fistula	<20	2	3.86	0.47-13.93
	20-24	5	2.56	0.83-5.97
	25-29	7	1.97	0.79-4.07
	30-34	17	3.39	1.98-5.43
	35+	10	3.00	1.44-5.51
Hirschsprung Disease	<20	0	0	-
	20-24	4	2.05	0.56-5.24
	25-29	5	1.41	0.46-3.29
	30-34	12	2.39	1.24-4.18
	35+	4	1.20	0.33-3.07
Rectal and Large Intestinal Atresia/Stenosis	<20	4	7.71	2.10-19.74
	20-24	13	6.65	3.54-11.37
	25-29	10	2.82	1.35-5.19
	30-34	20	3.99	2.44-6.16
	35+	11	3.30	1.65-5.90
Small Intestinal Atresia	<20	3	5.78	1.19-16.90
	20-24	3	1.53	0.32-4.48
	25-29	10	2.82	1.35-5.19
	30-34	15	2.99	1.68-4.94
	35+	12	3.60	1.86-6.28
Other Gastrointestinal <sup>2</sup>	<20	6	11.57	4.25-25.18
	20-24	15	7.67	4.29-12.65
	25-29	32	9.03	6.17-12.74
	30-34	31	6.19	4.20-8.78
	35+	27	8.09	5.33-11.78

**Table T.5. Prevalence of Birth Defects by Maternal Age, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Age	Count	Rate per 10,000 Live Births	95% Confidence Interval
<b>Genitourinary</b>				
Bladder Exstrophy	<20	0	0	-
	20-24	1	0.51	0.01-2.85
	25-29	2	0.56	0.07-2.04
	30-34	2	0.40	0.05-1.44
	35+	0	0	-
Cloacal Exstrophy	<20	0	0	-
	20-24	1	0.51	0.01-2.85
	25-29	0	0	-
	30-34	1	0.20	0.01-1.11
	35+	0	0	-
Hypospadias <sup>4</sup> , 1st Degree or NOS	<20	13	48.93	26.05-83.67
	20-24	27	26.85	17.7-39.07
	25-29	62	34.37	26.35-44.06
	30-34	81	31.78	25.24-39.51
	35+	46	26.66	19.52-35.56
Hypospadias <sup>4</sup> , 2nd or 3rd Degree	<20	5	18.82	6.11-43.92
	20-24	24	23.87	15.29-35.51
	25-29	40	22.17	15.84-30.19
	30-34	68	26.68	20.72-33.83
	35+	40	23.18	16.56-31.57
Obstructive Genitourinary Defect	<20	14	26.99	14.76-45.29
	20-24	26	13.29	8.68-19.48
	25-29	47	13.26	9.74-17.63
	30-34	81	16.17	12.84-20.09
	35+	47	14.09	10.35-18.74
Renal Agenesis/Hypoplasia	<20	2	3.86	0.47-13.93
	20-24	6	3.07	1.13-6.68
	25-29	17	4.80	2.79-7.68
	30-34	15	2.99	1.68-4.94
	35+	8	2.40	1.04-4.73

**Table T.5. Prevalence of Birth Defects by Maternal Age, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Age	Count	Rate per 10,000 Live Births	95% Confidence Interval
Other Genitourinary <sup>2</sup>	<20	16	30.85	17.63-50.09
	20-24	34	17.38	12.04-24.29
	25-29	62	17.49	13.41-22.42
	30-34	122	24.35	20.22-29.07
	35+	81	24.28	19.28-30.18
<b>Musculoskeletal</b>				
Club Foot	<20	7	13.50	5.43-27.81
	20-24	33	16.87	11.61-23.70
	25-29	56	15.80	11.93-20.51
	30-34	73	14.57	11.42-18.32
	35+	66	19.78	15.30-25.17
Craniosynostosis	<20	0	0	-
	20-24	7	3.58	1.44-7.37
	25-29	21	5.92	3.67-9.05
	30-34	35	6.99	4.87-9.71
	35+	26	7.79	5.09-11.42
Diaphragmatic Hernia	<20	1	1.93	0.05-10.74
	20-24	6	3.07	1.13-6.68
	25-29	20	5.64	3.45-8.71
	30-34	14	2.79	1.53-4.69
	35+	10	3.00	1.44-5.51
Gastroschisis	<20	6	11.57	4.25-25.18
	20-24	28	14.32	9.51-20.69
	25-29	12	3.38	1.75-5.91
	30-34	8	1.60	0.69-3.15
	35+	2	0.60	0.07-2.17
Omphalocele	<20	2	3.86	0.47-13.93
	20-24	5	2.56	0.83-5.97
	25-29	8	2.26	0.97-4.45
	30-34	20	3.99	2.44-6.16
	35+	19	5.70	3.43-8.89

**Table T.5. Prevalence of Birth Defects by Maternal Age, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Age	Count	Rate per 10,000 Live Births	95% Confidence Interval
Polydactyly/Syndactyly	<20	10	19.28	9.25-35.45
	20-24	41	20.96	15.04-28.44
	25-29	57	16.08	12.18-20.83
	30-34	74	14.77	11.60-18.54
	35+	52	15.59	11.64-20.44
Reduction Deformity, Lower Limbs	<20	1	1.93	0.05-10.74
	20-24	6	3.07	1.13-6.68
	25-29	9	2.54	1.16-4.82
	30-34	9	1.80	0.82-3.41
	35+	3	0.90	0.19-2.63
Reduction Deformity, Upper Limbs	<20	2	3.86	0.47-13.93
	20-24	15	7.67	4.29-12.65
	25-29	11	3.10	1.55-5.55
	30-34	17	3.39	1.98-5.43
	35+	14	4.20	2.29-7.04
Skeletal Dysplasia	<20	2	3.86	0.47-13.93
	20-24	3	1.53	0.32-4.48
	25-29	10	2.82	1.35-5.19
	30-34	13	2.59	1.38-4.44
	35+	12	3.60	1.86-6.28
Other Musculoskeletal <sup>2</sup>	<20	10	19.28	9.25-35.45
	20-24	26	13.29	8.68-19.48
	25-29	42	11.85	8.54-16.01
	30-34	76	15.17	11.95-18.99
	35+	48	14.39	10.61-19.08
<b>Chromosomal and other Syndromes</b>				
Klinefelter Syndrome	<20	0	0	-
	20-24	1	0.51	0.01-2.85
	25-29	2	0.56	0.07-2.04
	30-34	5	1.00	0.32-2.33
	35+	5	1.50	0.49-3.50



**Table T.5. Prevalence of Birth Defects by Maternal Age, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Age	Count	Rate per 10,000 Live Births	95% Confidence Interval
Trisomy 13	<20	1	1.93	0.05-10.74
	20-24	0	0	-
	25-29	10	2.82	1.35-5.19
	30-34	10	2.00	0.96-3.67
	35+	29	8.69	5.82-12.49
Trisomy 18	<20	1	1.93	0.05-10.74
	20-24	2	1.02	0.12-3.69
	25-29	11	3.10	1.55-5.55
	30-34	24	4.79	3.07-7.13
	35+	72	21.58	16.89-27.18
Trisomy 21 (Down Syndrome)	<20	3	5.78	1.19-16.90
	20-24	15	7.67	4.29-12.65
	25-29	39	11.00	7.82-15.04
	30-34	81	16.17	12.84-20.09
	35+	227	68.05	59.48-77.50
Turner Syndrome <sup>5</sup>	<20	3	11.86	2.45-34.65
	20-24	9	9.47	4.33-17.98
	25-29	18	10.34	6.13-16.34
	30-34	28	11.37	7.56-16.44
	35+	20	12.42	7.59-19.18
Other Chromosomal Syndromes/Other Syndromes <sup>2</sup>	<20	17	32.77	19.09-52.47
	20-24	43	21.99	15.91-29.61
	25-29	90	25.39	20.41-31.21
	30-34	159	31.73	26.99-37.07
	35+	171	51.26	43.87-59.55
<b>Other</b>				
Amniotic Bands	<20	1	1.93	0.05-10.74
	20-24	1	0.51	0.01-2.85
	25-29	4	1.13	0.31-2.89
	30-34	3	0.60	0.12-1.75
	35+	3	0.90	0.19-2.63

**Table T.5. Prevalence of Birth Defects by Maternal Age, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Age	Count	Rate per 10,000 Live Births	95% Confidence Interval
Skin Anomalies	<20	0	0	-
	20-24	2	1.02	0.12-3.69
	25-29	9	2.54	1.16-4.82
	30-34	7	1.40	0.56-2.88
	35+	4	1.20	0.33-3.07
Other, Specified <sup>2</sup>	<20	0	0	-
	20-24	5	2.56	0.83-5.97
	25-29	7	1.97	0.79-4.07
	30-34	9	1.80	0.82-3.41
	35+	3	0.90	0.19-2.63

NOS: Not Otherwise Specified. ASD: Atrial Septal Defect; VSD: Ventricular Septal Defect.

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

<sup>2</sup> Rate represents a heterogeneous group of defects.

<sup>3</sup> Excludes Patent Ductus Arteriosus.

<sup>4</sup> Rate calculated using male live births.

<sup>5</sup> Rate calculated using female live births.

**Table T.6. Prevalence of Birth Defects by Maternal Race, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Race	Count	Rate per 10,000 Live Births	95% Confidence Interval
<b>Central Nervous System</b>				
Anencephaly	White	20	2.25	1.38-3.48
	Black	2	1.42	0.17-5.13
	Asian	1	0.79	0.02-4.40
	Hispanic	4	1.60	0.44-4.10
Encephalocele	White	8	0.90	0.39-1.78
	Black	5	3.55	1.15-8.28
	Asian	5	3.95	1.28-9.22
	Hispanic	4	1.60	0.44-4.10
Holoprosencephaly	White	16	1.80	1.03-2.93
	Black	1	0.71	0.02-3.95
	Asian	2	1.58	0.19-5.71
	Hispanic	3	1.20	0.25-3.51
Hydrocephaly without Spina Bifida	White	38	4.28	3.03-5.88
	Black	12	8.51	4.40-14.87
	Asian	8	6.32	2.73-12.45
	Hispanic	18	7.20	4.27-11.38
Microcephaly	White	27	3.04	2.01-4.43
	Black	8	5.68	2.45-11.19
	Asian	4	3.16	0.86-8.09
	Hispanic	17	6.80	3.96-10.89
Spina Bifida with/without Hydrocephaly	White	48	5.41	3.99-7.17
	Black	3	2.13	0.44-6.22
	Asian	3	2.37	0.49-6.93
	Hispanic	12	4.80	2.48-8.39
Spinal Cord <sup>2</sup>	White	41	4.62	3.32-6.27
	Black	5	3.55	1.15-8.28
	Asian	5	3.95	1.28-9.22
	Hispanic	9	3.60	1.65-6.84

**Table T.6. Prevalence of Birth Defects by Maternal Race, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Race	Count	Rate per 10,000 Live Births	95% Confidence Interval
Other Central Nervous System <sup>2</sup>	White	109	12.29	10.09-14.82
	Black	24	17.03	10.91-25.34
	Asian	16	12.64	7.23-20.53
	Hispanic	33	13.21	9.09-18.55
<b>Eye</b>				
Aniridia	White	6	0.68	0.25-1.47
	Black	0	0	-
	Asian	0	0	-
	Hispanic	0	0	-
Anophthalmia/Microphthalmia	White	12	1.35	0.70-2.36
	Black	2	1.42	0.17-5.13
	Asian	2	1.58	0.19-5.71
	Hispanic	2	0.80	0.10-2.89
Congenital Glaucoma, Congenital Cataract	White	33	3.72	2.56-5.22
	Black	8	5.68	2.45-11.19
	Asian	3	2.37	0.49-6.93
	Hispanic	13	5.20	2.77-8.90
Other Eye <sup>2</sup>	White	46	5.19	3.80-6.92
	Black	4	2.84	0.77-7.27
	Asian	8	6.32	2.73-12.45
	Hispanic	8	3.20	1.38-6.31
<b>Ear</b>				
Anotia/Microtia	White	19	2.14	1.29-3.34
	Black	4	2.84	0.77-7.27
	Asian	3	2.37	0.49-6.93
	Hispanic	11	4.40	2.20-7.88
Other Ear <sup>2</sup>	White	55	6.20	4.67-8.07
	Black	6	4.26	1.56-9.27
	Asian	9	7.11	3.25-13.50
	Hispanic	13	5.20	2.77-8.90

**Table T.6. Prevalence of Birth Defects by Maternal Race, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Race	Count	Rate per 10,000 Live Births	95% Confidence Interval
<b>Cardiovascular</b>				
<i>Anomalous Pulmonary Venous Connection</i>				
Anomalous Pulmonary Venous Connection	White	16	1.80	1.03-2.93
	Black	4	2.84	0.77-7.27
	Asian	4	3.16	0.86-8.09
	Hispanic	3	1.20	0.25-3.51
<i>Atrioventricular Canal Defects</i>				
Atrial Septal Defect (ASD) Primum	White	2	0.23	0.03-0.81
	Black	0	0	-
	Asian	0	0	-
	Hispanic	1	0.40	0.01-2.23
Common Atrium	White	0	0	-
	Black	1	0.71	0.02-3.95
	Asian	0	0	-
	Hispanic	0	0	-
Complete Atrioventricular Canal Defect	White	34	3.83	2.65-5.36
	Black	9	6.39	2.92-12.12
	Asian	4	3.16	0.86-8.09
	Hispanic	15	6.00	3.36-9.90
Endocardial Cushion Defect	White	5	0.56	0.18-1.32
	Black	8	5.68	2.45-11.19
	Asian	1	0.79	0.02-4.40
	Hispanic	2	0.80	0.10-2.89
Ventricular Septal Defect (VSD), Canal Type	White	3	0.34	0.07-0.99
	Black	1	0.71	0.02-3.95
	Asian	0	0	-
	Hispanic	2	0.80	0.10-2.89
<i>Conotruncal (Outlet) and Aortic Arch</i>				
Double Outlet Right Ventricle	White	19	2.14	1.29-3.34
	Black	4	2.84	0.77-7.27
	Asian	2	1.58	0.19-5.71
	Hispanic	7	2.80	1.13-5.77

**Table T.6. Prevalence of Birth Defects by Maternal Race, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Race	Count	Rate per 10,000 Live Births	95% Confidence Interval
Interrupted Aortic Arch, Type B	White	1	0.11	0.00-0.63
	Black	1	0.71	0.02-3.95
	Asian	0	0	-
	Hispanic	1	0.40	0.01-2.23
Tetralogy of Fallot	White	48	5.41	3.99-7.17
	Black	5	3.55	1.15-8.28
	Asian	8	6.32	2.73-12.45
	Hispanic	14	5.60	3.06-9.40
Truncus Arteriosus (Common Truncus)	White	6	0.68	0.25-1.47
	Black	2	1.42	0.17-5.13
	Asian	0	0	-
	Hispanic	1	0.40	0.01-2.23
Dextro-Transposition of the Great Arteries	White	33	3.72	2.56-5.22
	Black	4	2.84	0.77-7.27
	Asian	1	0.79	0.02-4.40
	Hispanic	3	1.20	0.25-3.51
<b><i>Ebstein Anomaly</i></b>				
Ebstein Anomaly	White	6	0.68	0.25-1.47
	Black	0	0	-
	Asian	0	0	-
	Hispanic	0	0	-
<b><i>Heterotaxy (Laterality Defects)</i></b>				
Heterotaxy	White	12	1.35	0.70-2.36
	Black	4	2.84	0.77-7.27
	Asian	3	2.37	0.49-6.93
	Hispanic	5	2.00	0.65-4.67
<b><i>Left-Sided Obstruction</i></b>				
Aortic Arch Atresia without Hypoplastic Left Heart	White	0	0	-
	Black	0	0	-
	Asian	0	0	-
	Hispanic	1	0.40	0.01-2.23

**Table T.6. Prevalence of Birth Defects by Maternal Race, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Race	Count	Rate per 10,000 Live Births	95% Confidence Interval
Aortic Valve Stenosis	White	17	1.92	1.12-3.07
	Black	1	0.71	0.02-3.95
	Asian	0	0	-
	Hispanic	0	0	-
Coarctation of Aorta	White	48	5.41	3.99-7.17
	Black	8	5.68	2.45-11.19
	Asian	5	3.95	1.28-9.22
	Hispanic	8	3.20	1.38-6.31
Hypoplastic Left Heart Syndrome	White	21	2.37	1.47-3.62
	Black	5	3.55	1.15-8.28
	Asian	4	3.16	0.86-8.09
	Hispanic	6	2.40	0.88-5.23
Interrupted Aortic Arch (Type A and NOS)	White	1	0.11	0.00-0.63
	Black	1	0.71	0.02-3.95
	Asian	0	0	-
	Hispanic	0	0	-
<b><i>Right-Sided Obstruction</i></b>				
Pulmonary Stenosis, Valvar	White	66	7.44	5.75-9.47
	Black	16	11.35	6.49-18.44
	Asian	8	6.32	2.73-12.45
	Hispanic	21	8.40	5.20-12.85
Pulmonary Valve Atresia with intact septum	White	4	0.45	0.12-1.15
	Black	2	1.42	0.17-5.13
	Asian	2	1.58	0.19-5.71
	Hispanic	1	0.40	0.01-2.23
Pulmonary Valve Atresia with VSD	White	8	0.90	0.39-1.78
	Black	2	1.42	0.17-5.13
	Asian	0	0	-
	Hispanic	0	0	-

**Table T.6. Prevalence of Birth Defects by Maternal Race, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Race	Count	Rate per 10,000 Live Births	95% Confidence Interval
Tricuspid Valve Atresia	White	8	0.90	0.39-1.78
	Black	1	0.71	0.02-3.95
	Asian	0	0	-
	Hispanic	2	0.80	0.10-2.89
<i>Septal Defects</i>				
ASD (Secundum and NOS)	White	216	24.35	21.21-27.82
	Black	38	26.96	19.08-37.01
	Asian	22	17.38	10.89-26.32
	Hispanic	51	20.41	15.20-26.83
VSD (Membranous and NOS)	White	125	14.09	11.73-16.79
	Black	25	17.74	11.48-26.19
	Asian	16	12.64	7.23-20.53
	Hispanic	42	16.81	12.11-22.72
VSD (Muscular)	White	111	12.51	10.29-15.07
	Black	10	7.10	3.40-13.05
	Asian	10	7.90	3.79-14.53
	Hispanic	29	11.60	7.77-16.67
VSD, Conoventricular/Malalignment	White	10	1.13	0.54-2.07
	Black	7	4.97	2.00-10.23
	Asian	3	2.37	0.49-6.93
	Hispanic	5	2.00	0.65-4.67
<i>Single Ventricle and Levo-Transposition</i>				
Levo-Transposition of the Great Arteries	White	4	0.45	0.12-1.15
	Black	0	0	-
	Asian	0	0	-
	Hispanic	3	1.20	0.25-3.51
Single Ventricle	White	4	0.45	0.12-1.15
	Black	0	0	-
	Asian	1	0.79	0.02-4.40
	Hispanic	1	0.40	0.01-2.23



**Table T.6. Prevalence of Birth Defects by Maternal Race, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Race	Count	Rate per 10,000 Live Births	95% Confidence Interval
<b><i>Other Cardiovascular</i></b>				
Other Cardiovascular <sup>2,3</sup>	White	229	25.81	22.58-29.38
	Black	39	27.67	19.68-37.83
	Asian	24	18.96	12.15-28.21
	Hispanic	59	23.61	17.97-30.45
<b>Respiratory</b>				
Choanal Atresia	White	8	0.90	0.39-1.78
	Black	1	0.71	0.02-3.95
	Asian	1	0.79	0.02-4.40
	Hispanic	1	0.40	0.01-2.23
Lung Anomalies	White	18	2.03	1.20-3.21
	Black	5	3.55	1.15-8.28
	Asian	5	3.95	1.28-9.22
	Hispanic	8	3.20	1.38-6.31
Other Respiratory <sup>2</sup>	White	10	1.13	0.54-2.07
	Black	0	0	-
	Asian	2	1.58	0.19-5.71
	Hispanic	9	3.60	1.65-6.84
<b>Orofacial</b>				
Cleft Lip with/without Cleft Palate	White	99	11.16	9.07-13.59
	Black	8	5.68	2.45-11.19
	Asian	15	11.85	6.63-19.55
	Hispanic	18	7.20	4.27-11.38
Cleft Palate without Cleft Lip	White	59	6.65	5.06-8.58
	Black	5	3.55	1.15-8.28
	Asian	9	7.11	3.25-13.50
	Hispanic	16	6.40	3.66-10.40
Pierre Robin Sequence	White	17	1.92	1.12-3.07
	Black	2	1.42	0.17-5.13
	Asian	1	0.79	0.02-4.40
	Hispanic	4	1.60	0.44-4.10

**Table T.6. Prevalence of Birth Defects by Maternal Race, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Race	Count	Rate per 10,000 Live Births	95% Confidence Interval
Other Orofacial <sup>2</sup>	White	33	3.72	2.56-5.22
	Black	5	3.55	1.15-8.28
	Asian	5	3.95	1.28-9.22
	Hispanic	6	2.40	0.88-5.23
<b>Gastrointestinal</b>				
Biliary Atresia	White	1	0.11	0.00-0.63
	Black	0	0	-
	Asian	1	0.79	0.02-4.40
	Hispanic	2	0.80	0.10-2.89
Esophageal Atresia/Tracheoesophageal Fistula	White	34	3.83	2.65-5.36
	Black	3	2.13	0.44-6.22
	Asian	1	0.79	0.02-4.40
	Hispanic	3	1.20	0.25-3.51
Hirschsprung Disease	White	13	1.47	0.78-2.51
	Black	5	3.55	1.15-8.28
	Asian	2	1.58	0.19-5.71
	Hispanic	5	2.00	0.65-4.67
Rectal and Large Intestinal Atresia/Stenosis	White	33	3.72	2.56-5.22
	Black	7	4.97	2.00-10.23
	Asian	3	2.37	0.49-6.93
	Hispanic	14	5.60	3.06-9.40
Small Intestinal Atresia	White	26	2.93	1.91-4.29
	Black	4	2.84	0.77-7.27
	Asian	3	2.37	0.49-6.93
	Hispanic	9	3.60	1.65-6.84
Other Gastrointestinal <sup>2</sup>	White	68	7.67	5.95-9.72
	Black	14	9.93	5.43-16.67
	Asian	7	5.53	2.22-11.40
	Hispanic	19	7.60	4.58-11.87

**Table T.6. Prevalence of Birth Defects by Maternal Race, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Race	Count	Rate per 10,000 Live Births	95% Confidence Interval
<b>Genitourinary</b>				
Bladder Exstrophy	White	3	0.34	0.07-0.99
	Black	1	0.71	0.02-3.95
	Asian	0	0	-
	Hispanic	1	0.40	0.01-2.23
Cloacal Exstrophy	White	1	0.11	0.00-0.63
	Black	0	0	-
	Asian	0	0	-
	Hispanic	1	0.40	0.01-2.23
Hypospadias <sup>4</sup> , 1st Degree or NOS	White	170	37.44	32.02-43.51
	Black	10	13.86	6.65-25.49
	Asian	9	13.92	6.36-26.42
	Hispanic	38	29.75	21.05-40.84
Hypospadias <sup>4</sup> , 2nd or 3rd Degree	White	122	26.87	22.31-32.08
	Black	21	29.11	18.02-44.50
	Asian	7	10.83	4.35-22.31
	Hispanic	25	19.57	12.67-28.90
Obstructive Genitourinary Defect	White	133	14.99	12.55-17.77
	Black	20	14.19	8.67-21.92
	Asian	15	11.85	6.63-19.55
	Hispanic	42	16.81	12.11-22.72
Renal Agenesis/Hypoplasia	White	36	4.06	2.84-5.62
	Black	5	3.55	1.15-8.28
	Asian	1	0.79	0.02-4.40
	Hispanic	5	2.00	0.65-4.67
Other Genitourinary <sup>2</sup>	White	201	22.66	19.63-26.02
	Black	35	24.84	17.30-34.54
	Asian	28	22.12	14.70-31.97
	Hispanic	45	18.01	13.13-24.10

**Table T.6. Prevalence of Birth Defects by Maternal Race, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Race	Count	Rate per 10,000 Live Births	95% Confidence Interval
<b>Musculoskeletal</b>				
Club Foot	White	164	18.49	15.77-21.54
	Black	17	12.06	7.03-19.31
	Asian	11	8.69	4.34-15.55
	Hispanic	35	14.01	9.76-19.48
Craniosynostosis	White	72	8.12	6.35-10.22
	Black	3	2.13	0.44-6.22
	Asian	2	1.58	0.19-5.71
	Hispanic	11	4.40	2.20-7.88
Diaphragmatic Hernia	White	33	3.72	2.56-5.22
	Black	3	2.13	0.44-6.22
	Asian	6	4.74	1.74-10.32
	Hispanic	9	3.60	1.65-6.84
Gastroschisis	White	34	3.83	2.65-5.36
	Black	5	3.55	1.15-8.28
	Asian	2	1.58	0.19-5.71
	Hispanic	11	4.40	2.20-7.88
Omphalocele	White	33	3.72	2.56-5.22
	Black	2	1.42	0.17-5.13
	Asian	4	3.16	0.86-8.09
	Hispanic	11	4.40	2.20-7.88
Polydactyly/Syndactyly	White	116	13.08	10.81-15.68
	Black	46	32.64	23.90-43.54
	Asian	21	16.59	10.27-25.36
	Hispanic	49	19.61	14.51-25.92
Reduction Deformity, Lower Limbs	White	23	2.59	1.64-3.89
	Black	3	2.13	0.44-6.22
	Asian	2	1.58	0.19-5.71
	Hispanic	0	0	-

**Table T.6. Prevalence of Birth Defects by Maternal Race, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Race	Count	Rate per 10,000 Live Births	95% Confidence Interval
Reduction Deformity, Upper Limbs	White	35	3.95	2.75-5.49
	Black	8	5.68	2.45-11.19
	Asian	2	1.58	0.19-5.71
	Hispanic	13	5.20	2.77-8.90
Skeletal Dysplasia	White	28	3.16	2.10-4.56
	Black	5	3.55	1.15-8.28
	Asian	2	1.58	0.19-5.71
	Hispanic	3	1.20	0.25-3.51
Other Musculoskeletal <sup>2</sup>	White	118	13.30	11.01-15.93
	Black	30	21.29	14.36-30.39
	Asian	14	11.06	6.05-18.56
	Hispanic	35	14.01	9.76-19.48
<b>Chromosomal and other Syndromes</b>				
Klinefelter Syndrome	White	11	1.24	0.62-2.22
	Black	0	0	-
	Asian	0	0	-
	Hispanic	1	0.40	0.01-2.23
Trisomy 13	White	36	4.06	2.84-5.62
	Black	1	0.71	0.02-3.95
	Asian	3	2.37	0.49-6.93
	Hispanic	3	1.20	0.25-3.51
Trisomy 18	White	54	6.09	4.57-7.94
	Black	14	9.93	5.43-16.67
	Asian	14	11.06	6.05-18.56
	Hispanic	18	7.20	4.27-11.38
Trisomy 21 (Down Syndrome)	White	229	25.81	22.58-29.38
	Black	34	24.13	16.71-33.71
	Asian	22	17.38	10.89-26.32
	Hispanic	57	22.81	17.28-29.55

**Table T.6. Prevalence of Birth Defects by Maternal Race, Live Births, Stillbirths, and Other Pregnancy Losses, Massachusetts: 2013-2014**

Defect <sup>1</sup>	Maternal Race	Count	Rate per 10,000 Live Births	95% Confidence Interval
Turner Syndrome <sup>5</sup>	White	48	11.08	8.17-14.70
	Black	6	8.72	3.20-18.98
	Asian	5	8.08	2.62-18.85
	Hispanic	5	4.09	1.33-9.55
Other Chromosomal Syndromes/Other Syndromes <sup>2</sup>	White	303	34.16	30.42-38.22
	Black	35	24.84	17.30-34.54
	Asian	33	26.07	17.95-36.62
	Hispanic	65	26.01	20.07-33.15
<b>Other</b>				
Amniotic Bands	White	8	0.90	0.39-1.78
	Black	2	1.42	0.17-5.13
	Asian	0	0	-
	Hispanic	2	0.80	0.10-2.89
Skin Anomalies	White	15	1.69	0.95-2.79
	Black	4	2.84	0.77-7.27
	Asian	1	0.79	0.02-4.40
	Hispanic	2	0.80	0.10-2.89
Other, Specified <sup>2</sup>	White	13	1.47	0.78-2.51
	Black	4	2.84	0.77-7.27
	Asian	4	3.16	0.86-8.09
	Hispanic	3	1.20	0.25-3.51

Race/ethnic groups used: White, Non-Hispanic; Black, Non-Hispanic; Asian, Non-Hispanic; Hispanic.

Other, Non-Hispanic not presented due to small numbers.

NOS: Not Otherwise Specified. ASD: Atrial Septal Defect; VSD: Ventricular Septal Defect.

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

<sup>2</sup> Rate represents a heterogeneous group of defects.

<sup>3</sup> Excludes Patent Ductus Arteriosus.

<sup>4</sup> Rate calculated using male live births.

<sup>5</sup> Rate calculated using female live births.

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## APPENDICES

### Appendix 1: Technical Notes

#### Data Sources

Surveillance records were matched to records from the Registry of Vital Records and Statistics to obtain demographic and clinical information. For live births, birth certificate data were used as the source of information for maternal age, region of maternal residence (based on city), race/ethnicity, birth weight, plurality and gestational age (clinical estimate). All diagnostic information and infant sex were obtained from surveillance data. All live births were matched to a birth certificate. For fetal deaths that did not match to a fetal death certificate, surveillance data was used to obtain diagnostic and clinical information. Information on other pregnancy losses was obtained from surveillance.

#### Prevalence, Rates and Confidence Interval

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 live births is used as an approximation.

The rates provided in the tables and figures are estimations of the proportion of deliveries with birth defects overall and within subgroups. This rate is expressed as birth defect births per 10,000 live births and is calculated by the formula:

Number of Cases / total number live births x 10,000

Fetal deaths and other pregnancy losses 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 methods outlined in Sever, 2004 and the National Birth Defects Prevention Network “Guidelines for Conducting Birth Defects Surveillance”:  
[https://www.nbdpn.org/docs/Ch\\_8\\_Statistics6-04\\_2016DEC14.pdf](https://www.nbdpn.org/docs/Ch_8_Statistics6-04_2016DEC14.pdf).

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 a birth defect is identical in two subpopulations, the observed rates for the subpopulations may differ because of random variation. The width of the CI indicates the precision of the observed rate as an estimate of the underlying risk of having a birth defect, with a wider interval indicating less certainty about this estimate. The width of the CI reflects the size of the subpopulation and the number of cases of birth defects. Smaller subpopulations with fewer defects lead to wider CI. The 95% CI used in the report is calculated using the Poisson method, except for the CI for the age-adjusted rates, which are calculated using the standard method. If CI for two rates overlaps, this means that we cannot rule out random variation to explain any differences in the rates.



## **Assignment of Race/Ethnicity**

The Massachusetts Center for Birth Defects Research and Prevention follows the recommendation of the National Center for Health Statistics of classifying births according to the self-reported race/ethnicity of the mother. The 2003 version of the Certificate of Live Birth was implemented in Massachusetts in early 2011, and this new certificate allows for reporting of more than one racial and ethnic category. Race/ethnicity is a self-reported item and is subject to the usual limitations of this type of information.

## **Calculation of 2012 Dollars**

Calculation of cost of birth defects in 2012 dollars was made using the Bureau of Labor Statistics consumer price index (CPI) Inflation Calculator. The CPI inflation calculator uses the average Consumer Price Index for a given calendar year.

## **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.

## **Isolated vs. Multiple Major designation**

*Isolated cases:* cases that have only a single defect, those with multiple defects within the same organ, those with a single major defect with one or more minor defects, and those with defects that are part of a sequence of developmental events.

*Multiple major cases:* cases that have multiple major defects in different organs with or without being part of a recognized syndrome.

## Appendix 2: Glossary of Terms Used in this Report

**Agenesis, aplasia, or hypoplasia** The absence or incomplete development of an organ or body part.

**Anencephaly** Congenital absence of the skull, with cerebral hemispheres completely missing or reduced to small masses attached to the base of the skull. Anencephaly is not compatible with life.

**Anophthalmia** A developmental defect characterized by complete absence of the eyes, or by the presence of vestigial eyes.

**Anotia** A congenital absence of one or both ears.

**Aortic valve stenosis** A cardiac anomaly characterized by a narrowing or stricture of the aortic valve. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. This condition can be repaired surgically in some cases.

**Atresia** Absence or closure of a normal opening.

**Atrial septal defect (ASD)** A congenital cardiac malformation in which there are one or more openings in the atrial septum (muscular and fibrous wall between the right and left atria) allowing a mixing of oxygenated and unoxygenated blood. The openings vary in size and may resolve without treatment or may require surgical treatment.

**Biliary atresia** A congenital absence or underdevelopment of one or more of the ducts in the biliary tract. Correctable surgically.

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

**Birth prevalence** the number of birth defect cases at a given time, calculated as follows:

$$\frac{\text{The number of cases with birth defect A in an area and time period}}{\text{The number of live births in that area and time period}} \times 10,000$$

**Bladder exstrophy** Incomplete closure of the anterior wall of the bladder and the abdominal cavity. The abdominal wall and underlying organs do not fuse properly so that the bladder is exposed on the outside of the body.

**Cataract** An opacity (clouding) of the lens of the eye.

**Choanal atresia or stenosis** A congenital anomaly in which a bony or membranous formation blocks the passageway between the nose and the pharynx. This defect is usually repaired surgically after birth.

**Cleft lip** The congenital failure of the fetal components of the lip to fuse or join, forming a groove or fissure in the lip.

**Cleft palate** The congenital failure of the palate to fuse properly, forming a grooved depression or fissure in the roof of the mouth. This defect varies in degree of severity. The fissure can extend into the hard and soft palate and into the nasal cavities.

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

**Coarctation of the aorta** Localized narrowing of the aorta. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. This condition can vary from mild to severe.

**Confidence interval (CI) (95%)** The interval that contains the true prevalence (which we can only estimate) 95% of the time.

**Congenital** Existing at or dating from birth.

**Craniosynostosis** A premature closing of the cranial sutures before or soon after birth. This condition is occasionally associated with other skeletal defects. If no surgical correction is made, the growth of the skull is inhibited, and the head is deformed.

**Diaphragmatic hernia** A failure of the diaphragm to form completely, leaving a hole. Abdominal organs may protrude through the hole into the chest cavity and interfere with development of the heart and lungs.

**Down syndrome (Trisomy 21)** The chromosomal abnormality characterized by an extra copy of chromosome 21. In rare cases this syndrome is caused by translocation. Down syndrome can occur in mosaic (i.e., there is a population of normal cells and a population of trisomy 21 cells). Many infants have congenital heart disease.

**Ebstein anomaly** A congenital heart defect in which the tricuspid valve is displaced downward into the right ventricle causing abnormal patterns of cardiac circulation.

**Embryogenesis** The development and growth of an embryo, especially the period from the second through the eighth week after conception.

**Encephalocele** The protrusion of the brain substance through a defect in the skull.

**Endocardial cushion defect** A variety of septal defects (malformations of the walls separating the two atria and two ventricles of the heart) resulting from imperfect fusion of the endocardial cushions in the embryonic heart.

**Esophageal stenosis or atresia** A narrowing or incomplete formation of the esophagus. Usually a surgical emergency. Frequently associated with a tracheoesophageal fistula.

**Fetal death** See stillbirth.

**Fistula** An abnormal passage from an internal organ to the body surface or between two internal organs or structures.

**Folate** A 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.

**Folic acid** One of the B vitamins especially important for a woman to take before conception to help prevent neural tube defect. Folic acid refers to the synthetic vitamin used in supplements, whereas folate is the form found in foods.

**Gastroschisis** A congenital opening of the abdominal wall with protrusion of the intestines. This condition is surgically treated.

**Hernia** A protrusion of an organ or part through connective tissue or through a wall of the cavity in which it is normally enclosed.

**Hirschsprung disease** The congenital absence of autonomic ganglia (nerves controlling involuntary and reflexive movement) in the muscles of the colon. This results in immobility of the intestines and may cause obstruction or stretching of the intestines. This condition is repaired surgically in early childhood by the removal of the affected portion of the intestine.

**Holoprosencephaly** Failure of the brain to develop into two equal halves, so there is structural abnormality of the brain. There may be associated midline facial defects including cyclopia (fusion of the eye orbits into a single cavity containing one eye) in severe cases. About half the cases are probably due to a single gene defect (the HPE gene). Frequently occurs with Trisomy 13.

**Hydrocephalus** The abnormal accumulation of fluid within the spaces of the brain.

**Hypoplasia** A condition of arrested development in which an organ or body part remains below the normal size or in an immature state.

**Hypoplastic left heart syndrome** Atresia, or marked hypoplasia, of the aortic opening or valve, with hypoplasia of the ascending aorta and defective development of the left ventricle (with mitral valve atresia). This condition is usually fatal if not treated.

**Hypospadias** A congenital defect in males in which the urinary meatus (urinary outlet) is on the underside of the penis or on the perineum (area between the genitals and the anus). The condition may be surgically corrected if needed for cosmetic, urologic, or reproductive reasons.

**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.

**Limb defects** See Reduction deformities.

**Live Birth** Any infant who breathes or shows any other evidence of life at birth.

**Microcephaly** Congenital small size of the head relative to the height, with corresponding small brain size.

**Microphthalmia** The congenital abnormal smallness of one or both eyes. Can occur in the presence of other ocular defects.

**Microtia** A small or maldeveloped external ear and atretic or stenotic external auditory canal.

**Mosaic** In genetics, this refers to an individual organism that has two or more kinds of genetically different cell types. The degree of abnormality depends on the type of tissue containing affected cells. Individuals may vary from near normal to full manifestation of the genetic syndrome. Can occur in any chromosome abnormality syndrome.

**Neural tube defect** A defect resulting from failure of the neural tube to close in the first month of pregnancy. The major conditions include anencephaly, spina bifida, and encephalocele.

**Obstructive genitourinary defect** Stenosis or atresia of the urinary tract at any level. Severity of the defect depends largely upon the level of the obstruction. Urine accumulates behind the obstruction and damages the organs.

**Omphalocele** The protrusion of an organ into the umbilicus. The defect is usually closed surgically soon after birth. Contrast with Gastroschisis.

**Other Pregnancy Loss/Other Loss** Spontaneous pregnancy loss at less than 20 weeks gestation and weighing less than 350 grams OR elective termination.

**Patent ductus arteriosus (PDA)** A hole between the pulmonary artery and the aorta. This is normal in fetal life, but can cause problems after birth. The vast majority close spontaneously and cause no problems. Medical or surgical correction may be done. PDA in a premature infant is not considered a birth defect.

**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.

**Poisson regression** A type of statistical analysis based on the Poisson distribution used to compare rates of rare occurrences such as birth defects between different population groups, different areas, or different times.

**Prevalence** With respect to the prevalence of birth defects, see “*Birth prevalence*”.

**Pulmonary valve atresia or stenosis** A congenital heart condition characterized by absence or constriction of the pulmonary valve. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. This condition can vary from mild to severe. Mild forms are relatively well tolerated and require no intervention. More severe forms are surgically corrected.

**Reduction defects of the lower limbs** The congenital absence of a portion of the lower limb.

**Reduction defects of the upper limbs** The congenital absence of a portion of the upper limb.

**Renal agenesis** The failure of embryonic development of the kidney.

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

**Spina bifida** A neural tube defect resulting from failure of the spinal neural tube to close. The spinal cord and/or meninges may or may not protrude. This usually results in damage to the spinal cord with paralysis of the involved limbs. Includes myelomeningocele (involving both spinal cord and meninges) and meningocele (involving just the meninges).

**Stenosis** A narrowing or constriction of the diameter of a bodily passage or orifice.

**Stenosis or atresia of large intestine, rectum and anus** The absence, closure or constriction of the large intestine, rectum or anus. Can be surgically corrected or bypassed.

**Stenosis or atresia of the small intestine** A narrowing or incomplete formation of the small intestine obstructing movement of food through the digestive tract.

**Stillbirth (Fetal Death)** Death of a fetus delivered of at least 20 weeks gestation, or with a weight of at least 350 grams.

**Tetralogy of Fallot** A congenital cardiac anomaly consisting of four defects: ventricular septal defect, pulmonary valve stenosis or atresia, displacement of the aorta to the right, and hypertrophy of right ventricle. The condition is corrected surgically.

**Tracheoesophageal fistula** An abnormal passage between the esophagus and trachea. Corrected surgically. It is frequently associated with esophageal atresia.

**Translocation** The rearrangement of genetic material within the same chromosome or the transfer of a segment of one chromosome to another one. People with balanced translocations do not always manifest genetic syndromes, but may be carriers of genetic syndromes and can have children with unbalanced translocations. Can occur with any chromosomal anomaly syndrome.

**Transposition of the great vessels (Transposition of the great arteries/TGA)** A congenital malformation in which the aorta arises from the right ventricle and the pulmonary artery from the left ventricle (opposite of normal), so that the venous return from the peripheral circulation is recirculated without being oxygenated in the lungs. Can occur in Levo (L-) or Dextro (d-) form. Dextro usually requires immediate surgical correction.

**Tricuspid valve atresia or stenosis** A congenital cardiac condition characterized by the absence or constriction of the tricuspid valve. The opening between the right atrium and right ventricle is absent or restricted, and normal circulation is not possible. This condition is often associated with other cardiac defects. This condition is surgically corrected depending on the severity.

**Trisomy** A chromosomal abnormality characterized by one more than the normal number of chromosomes. Normally, cells contain two of each chromosome. In trisomy, cells contain three copies of a specific chromosome.

**Trisomy 13** The chromosomal abnormality caused by an extra chromosome 13. The syndrome can occur in mosaic so that there is a population of normal cells and a population of trisomy 13 cells. The syndrome is characterized by impaired midline facial development, cleft lip and palate, polydactyly and mental retardation. Most infants do not survive beyond 6 months of life.

**Trisomy 18** The chromosomal abnormality characterized by an extra copy of chromosome 18. Trisomy 18 can occur in mosaic. The syndrome is characterized by mental retardation, neonatal hepatitis, low-set ears, skull malformation and short digits. Cardiac and renal anomalies are also common. Survival for more than a few months is rare.

**Trisomy 21** See Down syndrome.

**Truncus arteriosus (Common truncus)** A congenital heart defect in which the common arterial trunk fails to divide into pulmonary artery and aorta. This is corrected surgically.

**Ventricle** One of the two lower chambers of the heart (plural ventricles). The right ventricle sends blood to the lungs, and the left ventricle passes oxygen-rich blood to the rest of the body.

**Ventricular septal defect (VSD)** A congenital cardiac malformation in which there are one or more openings in the ventricular septum (muscular and fibrous wall between the right and left ventricle or right and left lower chambers of the heart) allowing a mixing of oxygenated and deoxygenated blood. The openings vary in size and may resolve without treatment or require surgical treatment.

**Definitions adapted from:** Texas Department of State Health Services Glossary of Birth Defects Terms, April 2010.

### Appendix 3: Massachusetts 2013-2014 Live Birth Populations Used in Calculating Rates

Numbers of Live Births to Massachusetts Residents				
		2013 N=71,794	2014 N=71,868	Total N=143,662
By Maternal Age	<20	2768	2419	5187
	20-24	9947	9611	19,558
	25-29	17,622	17,829	35,451
	30-34	24,857	25,248	50,105
	35+	16,598	16,761	33,359
	Unknown	2	0	2
By Infant Sex	Male	36,600	36,890	73,490
	Female	35,194	34,978	70,172
	Unknown/Ambiguous	0	0	0
By Plurality	Singleton	68,526	68,801	137,327
	Multiple Birth	3268	3067	6335
By Maternal Race/Ethnicity	White	44,581	44,129	88,710
	Black	7022	7071	14,093
	Hispanic	12,320	12,670	24,990
	Asian/Pacific Islander	6231	6426	12,657
	American Indian/Other	760	624	1384
	Unknown	880	948	1828
By Region	Western	8119	8059	16,178
	Central	9032	9130	18,162
	Northeast	15,207	15,217	30,424
	Metro West	16,804	16,914	33,718
	Southeast	12,476	12,514	24,990
	Boston	10,156	10,034	143,662

Based on data from the Massachusetts Registry of Vital Records and Statistics. Division of Research and Epidemiology, Bureau of Health Information, Statistics, Research, and Evaluation, Massachusetts Department of Public Health “Massachusetts Births 2013” and “Massachusetts Births 2014” with the addition of updated vital records information (e.g. from late-filed birth certificates).

#### Appendix 4: ICD-9-CM/BPA Birth Defect Codes Used in this Report and Inclusions/Exclusions

Birth Defect	ICD-9-CM Codes <sup>1</sup>	Modified ICD-9-CM/BPA Codes <sup>2</sup>	Comments
<b>Central Nervous System</b>			
Anencephaly	740.0 –740.1	740.00 – 740.10, 740.20, 740.21, 740.29	
Encephalocele	742.0	742.00 – 742.09	
Holoprosencephaly	742.2	742.26	
Hydrocephaly without Spina Bifida	742.3	742.30-742.32, 742.38-742.39	
Microcephaly	742.1	742.10, 742.286	
Spina bifida with and without Hydrocephaly	741.0, 741.9	741.00 – 741.99	
Spinal Cord anomalies	348.0, 745.51, 742.53, 742.59	742.50, 742.51, 742.52, 742.53, 742.54, 742.58	
Other Central Nervous System	742.2, 742.4, 742.8, 742.9	742.20, 742.21, 742.23-742.25, 742.27-742.29, 742.40-742.42, 742.480, 742.485, 742.88, 742.90	
<b>Eye</b>			
Aniridia	743.45	743.420-743.424	
Anophthalmia/Microphthalmia	743.0, 743.1	743.00 – 743.10	
Congenital Glaucoma/Congenital Cataract	743.30 – 743.34	743.20, 743.32, 743.35, 743.36	
Other Eye	743.35, 743.41-44, 743.46-743.49, 743.51-743.59, 743.66	743.300-743.314, 743.340-743.344, 743.410, 743.430, 743.440, 743.460-743.474, 743.480-743.530, 743.535, 743.580, 743.590, 743.610, 743.620, 743.636, 743.650, 743.800	
<b>Ear</b>			
Anotia/Microtia	744.01, 744.23	744.01, 744.21	
Other Ear	744.02-744.09, 744.24, 744.3	744.00, 744.02-744.10, 744.23-744.25, 744.280, 744.300	



Birth Defect	ICD-9-CM Codes <sup>1</sup>	Modified ICD-9-CM/BPA Codes <sup>2</sup>	Comments
<b>Cardiovascular</b>			
Aortic Arch Atresia	747.22	747.200	Without Hypoplastic Left Heart Syndrome
Aortic Valve Stenosis	746.3	746.30	
Atrial Septal Defect (ASD), Primum	745.61	745.60	
ASD, Secundum, and Not otherwise specified (NOS)	745.5	745.51, 745.58, 745.59	
Coarctation of Aorta	747.10	747.10 – 747.19	
Common Atrium	745.69	745.61	
Complete Atrioventricular Canal Defect	745.69	745.62, 745.63	
Dextro-Transposition of the Great Arteries	745.10	745.10, 745.11	Excludes 745.19 (NOS)
Double Outlet Right Ventricle	745.11	745.185, 745.186, 745.188, 745.189	
Ebstein Anomaly	746.2	746.20	
Endocardial cushion defect	745.60, 745.69	745.68, 745.69	Includes other specified (OS) and NOS
Hypoplastic Left Heart Syndrome	746.7	746.70	
Interrupted Aortic Arch	747.11	747.215 – 747.217	Includes Type A, Type B and NOS
Levo-Transposition of the Great Arteries	745.10, 745.12	745.12	
Partial anomalous pulmonary venous connection	747.42	747.43	
Pulmonary Valve Atresia	746.01	746.00, 746.03	With intact ventricular septum (746.000), with VSD (746.030)
Pulmonary Stenosis, Valvular	746.02	746.01	
Single Ventricle	745.3	745.30-745.33, 745.38	
Tetralogy of Fallot	745.2	745.20, 747.31	
Total and partial anomalous pulmonary venous connection	747.41	747.42, 747.43	
Tricuspid Valve Atresia	746.1	746.10	Excludes tricuspid valve stenosis (746.106)
Truncus Arteriosus (Common Truncus)	745.0	745.00 (excluding 745.01)	
Ventricular Septal Defect (VSD), Canal Type	745.69	745.685	

Birth Defect	ICD-9-CM Codes <sup>1</sup>	Modified ICD-9-CM/BPA Codes <sup>2</sup>	Comments
VSD, Conoventricular/Malalignment	745.4	745.487	Excludes canal type and muscular (745.486).
VSD, Membranous or NOS	745.4	745.485, 745.49	Excludes canal type and muscular (745.486).
VSD, Muscular	745.4	745.486	
Other Cardiovascular	746.4, 746.8, 746.9, 747.2, 747.40, 747.49, 747.6, 747.8	745.010, 746.080, 746.090, 746.106, 746.400-746.505, 746.600, 746.800, 746.820, 746.830, 746.850, 746.880-746.882, 746.885, 746.900, 746.995, 747.210, 747.220, 747.230, 747.250, 747.270, 747.280, 747.300, 747.320, 747.380, 747.410, 747.480, 747.490, 747.620, 747.640, 747.650, 747.680, 747.800, 747.810, 747.880	
<b>Respiratory</b>			
Choanal Atresia	748.0	748.00, 748.01	
Lung Anomalies	748.4, 748.5	748.40, 748.41, 748.48, 748.50, 748.51, 748.52, 748.58, 748.88	
Other Respiratory	748.3, 748.6, 748.8	748.000, 748.100, 748.185, 748.205, 748.209, 748.310, 748.330-748.350, 748.380, 748.385, 748.390, 748.625, 748.690	Excludes laryngo-tracheomalacia
<b>Orofacial</b>			
Cleft Palate without Cleft Lip	749.0	749.00 – 749.09	Prior to 2014, excludes isolated submucous cleft palate
Cleft lip with/without Cleft Palate	749.1, 749.2	749.10 – 749.19, 749.20-749.29	
Pierre Robin Sequence	756.0	524.080	
Other Orofacial	744.8	744.400, 744.480, 744.880, 748.120, 748.180, 750.120, 750.130	
<b>Gastrointestinal</b>			
Biliary Atresia	751.61	751.65	
Esophageal Atresia/ Tracheoesophageal Fistula	750.3	750.30 – 750.35	
Hirschsprung Disease	751.3	751.30-751.34	
Rectal and Large Intestinal Atresia/Stenosis	751.2	751.20 – 751.24	

Birth Defect	ICD-9-CM Codes <sup>1</sup>	Modified ICD-9-CM/BPA Codes <sup>2</sup>	Comments
Small Intestinal Atresia	751.1	751.10 – 751.19	
Other Gastrointestinal	750.4,750.6, 750.7,750.8, 751.5, 751.7	750.380, 750.430, 750.480, 750.60,750.70, 750.80, 751.00, 751.010, 751.400-751.420, 751.490, 751.495, 751.50,751.52, 751.53, 751.54, 751.56, 751.58, 751.61-751.64, 751.66, 751.67, 751.70, 751.72, 751.74, 751.80	
<b>Genitourinary</b>			
Bladder Exstrophy	753.5	753.50	
Cloacal Exstrophy	751.5	751.555	
Hypospadias	752.61	752.60-752.62	In males only. Excludes 752.61 and 752.621 Prior to 2014, excludes 752.600, 752.605, 752.620,752.625
Obstructive Genitourinary Defect	753.2, 753.6	753.20 - 753.22,753.29, 753.60-753.69	Includes posterior urethral valve requires surgery or other defect
Renal Agenesis/Hypoplasia	753.0	753.00- 753.01	Prior to 2014 excludes unilateral renal agenesis
Other Genitourinary	752.0, 752.1, 752.3, 752.4, 752.8, 753.0- 753.8	752.00, 752.08, 752.085, 752.10,752.20, 752.30,752.32, 752.38, 752.40-752.44, 752.48, 752.70, 752.79-752.82, 752.85, 752.860, 752.865, 752.880, 752.901, 753.10-753.12, 753.13 753.16, 753.18, 753.31-753.34,753.38,753.40, 753.410, 753.420,753.480,753.485, 753.70, 753.710, 753.790-753.820, 753.84, 753.88	
<b>Musculoskeletal</b>			
Club Foot	754.51, 754.70	754.50, 754.51, 754.52, 754.53, 754.59, 754.60, 754.68, 754.69, 754.73 (excluding 754.735)	Requires casting or surgery for live births
Craniosynostosis	No specific code	756.00 – 756.03	
Diaphragmatic Hernia	756.6	756.600 - 756.605, 756.610 – 756.617, 756.618-756.619	
Gastroschisis	756.73	756.71	

Birth Defect	ICD-9-CM Codes <sup>1</sup>	Modified ICD-9-CM/BPA Codes <sup>2</sup>	Comments
Omphalocele	756.72	756.70	
Polydactyly/syndactyly	755.0, 755.1	755.005, 755.01-755.03, 755.095-755.096, 755.10-755.13, 755.19-755.199	Hands require bone or cartilage involvement. Excludes webbing of toes 2-3
Reduction Deformity, Lower limbs	755.3	755.30-755.39	
Reduction Deformity, Upper limbs	755.2	755.20-755.29	
Skeletal Dysplasia	755.55, 756.4, 756.5	755.555, 756.400, 756.41, 756.43, 756.447, 756.46, 756.480, 756.49, 756.50, 756.53, 756.54, 756.575, 756.58, 756.59	
Other Musculoskeletal	756.19, 756.3, 756.8	754.00, 754.20, 754.21, 754.22, 754.400, 754.410, 754.430, 754.440, 754.780, 754.820, 754.840, 754.880, 755.44-755.50, 755.530, 755.536, 755.54, 755.58, 755.585, 755.640, 755.650, 755.680, 755.685, 755.800, 756.080, 756.110, 756.120, 756.140, 756.145, 756.146, 756.150, 756.155, 756.156, 756.160, 756.165, 756.166, 756.170, 756.175, 756.180, 756.185, 756.190, 756.300, 756.310-756.350, 756.380, 756.620, 756.680, 756.690, 756.720, 756.790, 756.795, 756.80, 756.81, 756.84, 756.88	
<b>Chromosomal</b>			
Klinefelter Syndrome	758.7	758.70-758.71, 758.79	
Trisomy 13	758.1	758.10 – 758.19	
Trisomy 18	758.2	758.20 – 758.29	
Trisomy 21 (Down syndrome)	758.0	758.00 – 758.09	
Turner Syndrome	758.6	758.60 – 758.69	In females only

Birth Defect	ICD-9-CM Codes <sup>1</sup>	Modified ICD-9-CM/BPA Codes <sup>2</sup>	Comments
Other Chromosomal	various	279.110, 352.600, 756.040, 756.045 756.046, 756.050, 756.055-756.057, 756.060, 756.065, 756.525 756.550 - 756.570, 756.830, 756.850, 757.300 758.300 - 758.400, 758.50-758.54, 758.580, 758.585, 758.586, 758.590, 758.80 - 758.86, 758.88, 758.89, 758.90 - 759.93 758.990, 758.999, 759.340, 759.400-759.480, 759.500, 759.610, 759.620, 759.800, 759.820, 759.840, 759.860, 759.870, 759.890	
<b>Other</b>			
Amniotic Bands	No specific code	658.80	
Heterotaxy/Situs Inversus	759.3	759.30-759.33, 759.35-759.395	Displayed as part of the group of cardiovascular defects in tables
Skin Anomalies	757.1, 757.31, 757.39, 757.4, 757.8	757.34, 757.36, 757.48, 757.80, 757.35, 757.33, 757.11, 757.19, 757.195-757.197	
Other	various	255.20, 658.80, 759.00, 759.01, 759.04, 759.08, 759.21, 759.22, 759.24, 759.68, 759.70, 759.90	

NOS: Not Otherwise Specified; VSD: Ventricular Septal Defect; ASD: Atrial Septal Defect.

<sup>1</sup> International Classification of Diseases, 9<sup>th</sup> Revision.

<sup>2</sup> Centers for Disease Control/Clinical Modification, British Pediatric Association.

Note on codes: Some codes in the table above use shorthand with only 2 digits after the decimal point. If not specified, the third digit is implied and can be anything from 0-9.

## Appendix 5: Pre-Pregnancy Multivitamin Use, Massachusetts: 2011

### Prevalence of Multivitamin Use in the Month Prior to Pregnancy, Massachusetts: 2011

Frequency	%
Did not take a multivitamin at all	46.9
1-3 times per week	6.1
4-6 times per week	7.7
Daily	39.4

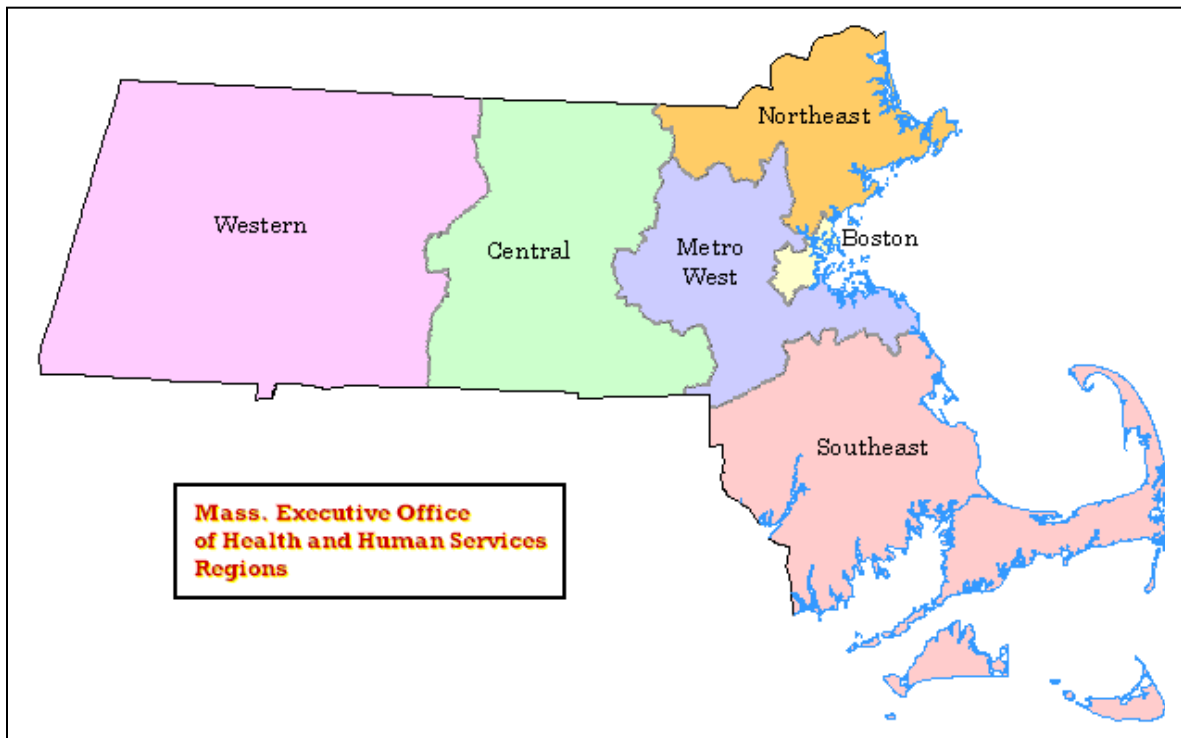
### Daily Multivitamin Use in the Month Prior to Pregnancy by Socio-demographic Characteristics, Massachusetts: 2011

Characteristic	Weighted n	Weighted %	95% Confidence Interval
<b>Total</b>	27520	39.4	36.2 - 42.7
<b>Maternal race/ethnicity</b>			
White, non-Hispanic	18917	43.9	39.2 - 48.7
Black, non-Hispanic	2019	31.4	24.7 - 39.0
Hispanic	3190	25.7	21.0 - 31.0
Asian, non-Hispanic	2202	44.6	35.1 - 54.5
Other, non-Hispanic	890	33.6	20.5 - 49.9
<b>Maternal age (years)</b>			
<20	530	14.7	6.7 - 29.2
20-29	7524	27.1	22.6 - 32.0
30-39	17910	50.2	45.5 - 54.9
40+	1557	55.4	39.9 - 70.0
<b>Maternal education</b>			
<High school	1432	19.9	13.7 - 28.1
High school diploma	4055	24.2	18.6 - 30.8
Some college	4692	30.6	24.5 - 37.4
College graduate	16759	56.6	51.4 - 61.6
<b>Household poverty level</b>			
≤100% FPL <sup>1</sup>	3892	22.2	17.5 - 27.7
>100% FPL	21909	47.5	43.3 - 51.8
<b>Maternal nativity</b>			
Non-US-born	7548	35.3	30.7 - 40.2
US-born	19840	41.0	36.8 - 45.3

<sup>1</sup>Federal Poverty Line

Source: Office of Data Translation, Bureau of Family Health and Nutrition, Massachusetts Department of Public Health (2015) Massachusetts Pregnancy Risk Assessment Monitoring System (PRAMS) 2011 Surveillance Report.

## Appendix 6: Map of Massachusetts Regions



## EVALUATION FORM

### Massachusetts Birth Defects 2013-2014

#### TO OUR READERS:

To better serve our users, we are enclosing this evaluation form. Please complete this questionnaire and Fax, email or mail using the contact information at the bottom of this page.

What tables and figures do you find MOST useful?
What tables and figures do you find LEAST useful?
Are there other tables and figures that you would like added to this publication? If yes, please describe.
Do you have other comments or suggestions?

Thank you.

Please return your comments to:  
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Bureau of Family Health & Nutrition  
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