Massachusetts Birth Defects

2011-2012

Massachusetts Birth Defects Monitoring Program
Bureau of Family Health and Nutrition

Massachusetts Department of Public Health

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EXECUTIVE SUMMARY

One of every 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 birth 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 among live births and stillbirths in Massachusetts during the years 2011 and 2012, as well as selected 2012 data that includes other types of pregnancy losses. Two years of live birth and stillbirth data are combined in this report, since the numbers of cases are often small for individual defects within a single year.

The data allow for some trend analyses. Increasing rates over time may reflect improved case ascertainment. The Birth Defects Monitoring 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 Massachusetts residents who delivered in 2011 or 2012, there are 2731 cases (2663 live births and 68 stillbirths) with one or more structural birth defects. This results in a prevalence rate of 187.5 birth defects per 10,000 live births.

Cardiovascular defects are the most commonly occurring birth defects in Massachusetts, followed by musculoskeletal, genitourinary and chromosomal defects.

Of the ten most common specific birth defects, three are cardiovascular—atrial septal defect, ventricular septal defect, and pulmonary valve stenosis. The most common non-cardiovascular defects are polydactyly/syndactyly, club foot, obstructive genitourinary defect, trisomy 21 (Down syndrome), and hypospadias.

Birth Defects in Massachusetts vs. the US

Massachusetts is one of 11 states with active case ascertainment programs and contributes data to published national prevalence estimates for selected birth defects. For most defects, Massachusetts rates in 2011-2012 are similar to the most recent national rates from 2004-2006 (7). However, Massachusetts rates are significantly lower than US rates for anencephaly, spina bifida, cleft lip, and gastroschisis. Massachusetts rates are significantly higher for atrioventricular canal defects and for esophageal atresia/tracheoesophageal fistula. Differences in surveillance system methodology, types of pregnancy outcomes included, and demographic variation may account for 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. A preliminary assessment of the impact of adding these other losses in 2012, the first full year of data available, shows that Massachusetts rates for spina bifida and anencephaly are similar to national rates when these other pregnancy outcomes are included.

Adverse Pregnancy Outcomes

Adverse outcomes such as low birth weight (LBW), prematurity, and small for gestational age (SGA) are more frequent among live births with birth defects than among unaffected infants. Infants with a birth defect are 3 times more likely to have low birth weight (less than 2500 grams) and 2.7 times more likely 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. In addition, infants with birth defects are roughly 15 times more likely to die in their first year of life.

Infant Sex

The prevalence of birth defects in Massachusetts in 2011-2012 is 216.9 per 10,000 male live births and 156.4 per 10,000 female live births. Males are significantly more likely than females to have obstructive genitourinary defects, club foot, and cleft lip.

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 (220.7 per 10,000 live births) compared to younger mothers.

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 33.7 per 10,000 live births, over 4 times greater than the rate in mothers younger than 35.

Gastroschisis, a condition in which a child is born with the intestines—and sometimes other organs—protruding through a hole in the abdominal wall, occurs more often among younger mothers. In 2011-2012, mothers less than 20 years old have the highest rate of gastroschisis (13.3 per 10,000 live births) among all age groups.

Assisted Reproductive Technology

It is estimated that 1.5 percent of US infants are conceived through the use of assisted reproductive technology (ART) (9). In 2011, Massachusetts had the highest rate 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 more common among multiple births (e.g. twins and triplets) than in singleton births. About 4.5% of Massachusetts live births are multiple births (8). The birth defect prevalence rate in 2011-2012 is 181.8 per 10,000 live births for singletons and 311.6 per 10,000 live births for multiples.

Maternal Race/Ethnicity

In Massachusetts and nationally, birth defect rates vary by maternal race and ethnicity. In 2011-2012 in Massachusetts, the overall age-adjusted prevalence rate of birth defects among Hispanic mothers is significantly higher than for white and Asian mothers, and the rate for black mothers is significantly higher than that for Asian mothers. Possible explanations for racial/ethnic differences include genetic variation, diet and lifestyle differences, and varying access to prenatal screening and health care services.

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 2011-2012 the overall age-adjusted birth defect prevalence rate was higher in the Western region than in the other regions, but this difference was not statistically significant.

Severity

A severity scale for birth defects was developed in collaboration with Boston University and Massachusetts General Hospital, based on the usual outcome for a specific birth defect, including the need for immediate treatment, the need for long-term care, and the amenability of the defect to correction. “Mild” defects generally require minimal long term care. “Moderate” defects tend to be correctable, although many affected infants have long term care needs. “Serious” defects may often be correctable, but affected infants typically have long term care needs. “Severe” defects are usually incompatible with life.

Approximately 8% of defects are classified as mild, 65% are classified as moderate, and roughly 23% of 2011-2012 cases are considered serious. Severe defects make up 3% of the live birth and stillbirth cases in 2011-2012.

Etiology and Pattern

The surveillance system in Massachusetts collects information on etiology (cause), whenever available. Most cases of birth defects have no known cause.

Pattern refers to whether a birth defect occurs with other defects. Approximately 24% of birth defects occur with defects in other organs or as part of syndromes, while 76% occur as isolated defects or are confined to the same organ, occur with minor defects, or are part of a sequence of developmental events.

Analysis of Trends

The overall prevalence of birth defects in Massachusetts increased slightly but not significantly in 2011-2012 compared with 2009-2010. The overall birth defect rate among live births and stillbirths in 2011-2012 was 187.5 per 10,000 live births compared with 181.5 per 10,000 live births in 2009-2010. This report includes selected trend analyses using current and previous years’ data, with the understanding that better case ascertainment and improved prenatal diagnosis may contribute to changes in birth defect rates.

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 structure present before birth.

Although birth defects are rare when compared to other adverse birth 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 US (13). However, some studies in certain populations suggest that not all cases of neural tube defects are preventable by increasing folate 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 Birth Defects Monitoring Program (BDMP) has conducted statewide, population-based active surveillance of birth defects among Massachusetts residents. 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 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 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 2011-2012 Surveillance Report

This report presents statewide data on the prevalence of birth defects in live births and stillbirths in Massachusetts during the years 2011 and 2012, as well as selected 2012 data that includes birth defects among other types of pregnancy losses. Most of the data is presented with the years 2011 and 2012 combined, since the numbers are relatively small for individual defects in a single year. Unless otherwise indicated, the case counts and prevalence rates in this report are based on live births and stillbirths.

In early 2011, Massachusetts began ascertaining prenatally-diagnosed birth defects in pregnancies that ended in pregnancy losses other than a live birth or stillbirth. Some 2012 data on these other losses is included in this report, although these data should be interpreted with caution, since only one year of data from a start-up year is included.

The Massachusetts Center for Birth Defects Research and Prevention

The Massachusetts Center for Birth Defects Research and Prevention 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.

CHAPTER 2: METHODS

Case Definition

This report presents data on selected birth defects among deliveries to Massachusetts residents occurring during the calendar years 2011 and 2012. 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 type of pregnancy loss (early miscarriage or elective termination—2012 data for selected analyses only)

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 outcomes, demographic and clinical information comes from surveillance data.

Recent changes to the program include:

Early 2011: Began collecting cases diagnosed prenatally that did not result in a live birth or stillbirth (other pregnancy losses)
Late 2011: Began use of electronic case report form for abstraction
Early 2012: Discontinued abstraction at two Rhode Island tertiary hospitals

The discontinuation of data collection at Rhode Island hospitals, where some cases born to Massachusetts residents in the southeastern part of the state deliver or receive treatment, is expected to slightly reduce our case numbers, but the program will continue to ascertain those cases that receive any diagnosis or treatment in Massachusetts. Based on 2008-2009 data, we estimate that fewer than 20 cases per year would be affected by this change, and for many of these we would still be able to obtain the information by contacting a physician.

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. In this report, maternal age, race/ethnicity, plurality and infant birth weight are drawn from Vital Records, except in rare cases where a stillbirth lacks a fetal death certificate, in which case surveillance data is used instead. Infant sex is drawn from surveillance data because it is generally considered to be more accurate.

The occurrence of birth defects is reported as prevalence. Prevalence is calculated as the number of birth defect cases delivered during the period 2011-2012 per 10,000 live births delivered during the same time period. Prevalence tables include the number of cases found, the estimated prevalence rate per 10,000 live births and the 95% confidence intervals for each rate. The incidence (new cases) of birth defects (based upon the number of embryos conceived within a year) cannot be fully measured because the total number of conceptions and the number of these conceptions resulting in a birth defect are not known (16).

The confidence interval (CI) can be used to assess the magnitude and stability of a rate or ratio. The CI for rates presented in this report consist of a range of possible values around the point estimate that has a 95% chance of including the actual underlying risk of an infant being born with a birth defect. Wide confidence intervals reflect the large variation due to small numbers (see Appendix 1: Technical Notes).

Changes to the Certificate of Live Birth

In early 2011, Massachusetts implemented the 2003 revision of the US Standard Certificate of Live Birth, which includes more racial and ethnic categories and which allows for the selection of multiple racial/ethnic categories. The number of live births classified as Hispanic has increased in recent years (8), perhaps partly as a result of this change.

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. Limiting the data to live births and stillbirths may result in undercounting of certain birth defects—especially those incompatible with life. For 2012, selected birth defect counts and rates are also presented with other pregnancy losses included.

4. Only diagnoses confirmed before 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.

5. 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. Based on 2008-2009 data, we estimate that fewer than 20 cases per year would be affected by this change, and for many of these we would still be able to obtain the information by contacting a physician.

6. Patent ductus arteriosus (PDA) is not included in this report, because this defect is often minor and is normal for infants born prematurely.

7. 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.

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.

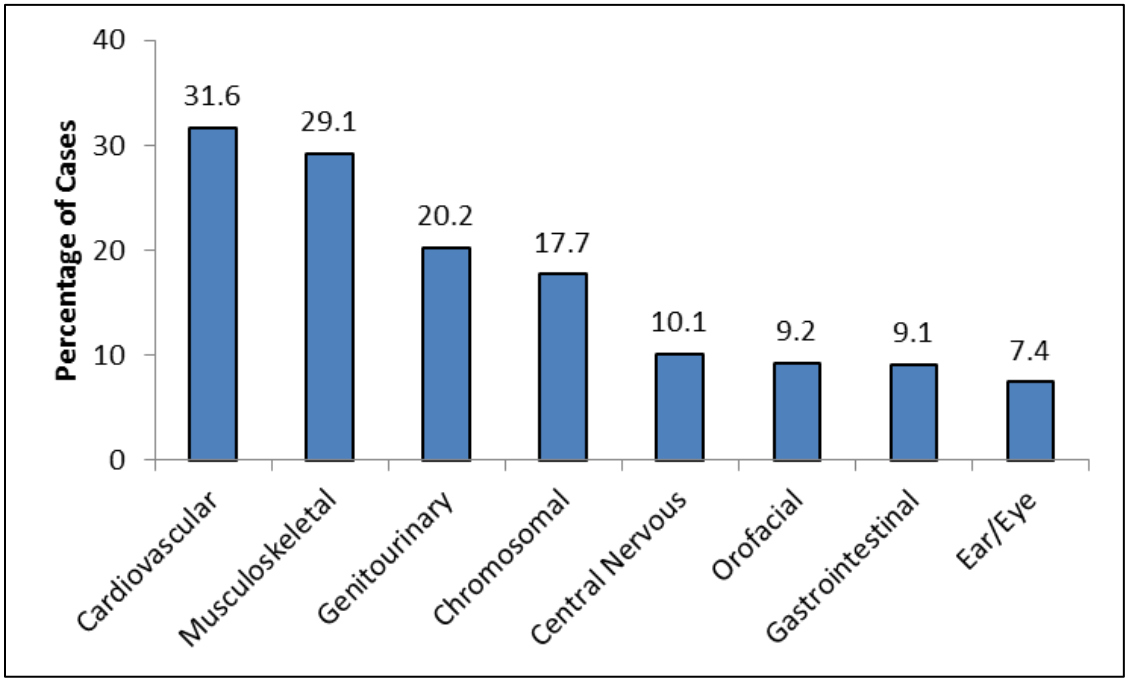
Chapter 3: Prevalence of Birth Defects

Overall Prevalence of Birth Defects

Among the 145,631 live births to Massachusetts residents in 2011-2012, 2663 live births and 68 stillbirths had at least one structural birth defect. This represents an overall Massachusetts birth defect rate of 187.5 (95% CI: 180.6-194.6) per 10,000 live births. Counts and rates for specific birth defects are shown in Table 1. The most common defects among live births and stillbirths are atrial septal defects, polydactyly/syndactyly, club foot, obstructive genitourinary defects, and trisomy 21 (Down syndrome).

Figure 1 shows the percentage of reported birth defects by defect category. Cardiovascular defects are the most commonly occurring birth defects in Massachusetts, followed by musculoskeletal, genitourinary and chromosomal defects. Because cases can have multiple defects, the same case may be included in more than one body system category.

Figure 1. Birth Defects by Body System, Massachusetts: 2011-2012



Live births and stillbirths, N=3809 defects among 2731 cases.
Figure shows the percentage of cases with defects in each body system.

Table 1. Prevalence of Birth Defects, Massachusetts: 2011-2012					
Defect[1]	**Live birth Count**	**Stillbirth Count**	**Total Count**	**Rate per 10,000 Births**	**95% Confidence Interval**
Total	***2663***	***68***	***2731***	***187.5***	***180.6-194.6***
Central Nervous System					
Anencephaly	6	2	8	0.55	0.24-1.08
Encephalocele	6	0	6	0.41	0.15-0.90
Holoprosencephaly	8	2	10	0.69	0.33-1.26
Hydrocephaly without Spina Bifida	47	3	50	3.43	2.55-4.53
Microcephaly	28	0	28	1.92	1.28-2.78
Spina Bifida with and without Hydrocephaly	24	4	28	1.92	1.28-2.78
Spinal Cord[2]	72	0	72	4.94	3.87-6.23
Eye					
Aniridia	4	0	4	0.27	0.07-0.70
Anophthalmia/Microphthalmia	14	0	14	0.96	0.53-1.61
Congenital Glaucoma, Congenital Cataract	45	0	45	3.09	2.25-4.13
Ear					
Anotia/Microtia	36	0	36	2.47	1.73-3.42
Cardiovascular[3]					
Anomalous Pulmonary Venous Connection					
Total/Partial Anomalous Pulmonary Venous Connection	21	0	21	1.44	0.89-2.20
Atrioventricular Canal Defects					
Atrial Septal Defect (ASD) Primum	1	0	1	0.07	0.00-0.38
Common Atrium	5	0	5	0.34	0.11-0.80
Complete Atrioventricular Canal Defect	59	2	61	4.19	3.20-5.38
Endocardial Cushion Defect, Other specified (OS) and Not otherwise specified (NOS)	18	2	20	1.37	0.84-2.12
Ventricular Septal Defect (VSD), Canal Type	8	1	9	0.62	0.28-1.17
Conotruncal (Outlet) and Aortic Arch					
Double Outlet Right Ventricle	14	2	16	1.10	0.63-1.78
Tetralogy of Fallot with and without Pulmonary Atresia	49	1	50	3.43	2.55-4.53
Truncus	5	0	5	0.34	0.11-0.80
dextro-Transposition of the Great Arteries	34	0	34	2.33	1.62-3.26
Ebstein Anomaly					
Ebstein Anomaly	5	0	5	0.34	0.11-0.80

Table 1. Prevalence of Birth Defects, Massachusetts: 2011-2012					
Defect[1]	**Live birth Count**	**Stillbirth Count**	**Total Count**	**Rate per 10,000 Births**	**95% Confidence Interval**
Heterotaxy (Laterality Defects)					
Heterotaxy	7	1	8	0.55	0.24-1.08
Left-Sided Obstruction					
Aortic Valve Stenosis	16	1	17	1.17	0.68-1.87
Aortic Arch Atresia	1	0	1	0.07	0.00-0.38
Coarctation of Aorta	68	0	68	4.67	3.63-5.92
Hypoplastic Left Heart Syndrome	23	1	24	1.65	1.06-2.45
Interrupted Aortic Arch (Type A and NOS)	4	0	4	0.27	0.07-0.70
Right-Sided Obstruction					
Pulmonary Stenosis, Valvular	119	0	119	8.17	6.77-9.78
Pulmonary Valve Atresia with intact septum	6	0	6	0.41	0.15-0.90
Pulmonary Valve Atresia with Ventricular Septal Defect	1	0	1	0.07	0.00-0.38
Tricuspid Valve Atresia	3	0	3	0.21	0.04-0.60
Septal Defects					
ASD (Secundum and NOS)	330	0	330	22.66	20.28-25.24
VSD (Membranous and NOS)	169	9	178	12.22	10.49-14.16
VSD (Conoventricular/Malalignment)	22	1	23	1.58	1.00-2.37
Single Ventricle and L-TGA					
levo-Transposition of the Great Arteries	5	1	6	0.41	0.15-0.90
Single Ventricle	3	0	3	0.21	0.04-0.60
Respiratory					
Choanal Atresia	12	0	12	0.82	0.43-1.44
Lung Anomalies[2]	37	2	39	2.68	1.90-3.66
Orofacial					
Cleft Lip with/without Cleft Palate	93	3	96	6.59	5.34-8.05
Cleft Palate without Cleft Lip	85	0	85	5.84	4.66-7.22
Pierre Robin Sequence	36	0	36	2.47	1.73-3.42
Gastrointestinal					
Biliary Atresia	14	0	14	0.96	0.53-1.61
Esophageal Atresia/Tracheoesophageal Fistula	49	0	49	3.36	2.49-4.45
Hirschsprung Disease	23	0	23	1.58	1.00-2.37
Rectal and Large Intestinal Atresia/Stenosis	53	2	55	3.78	2.85-4.92
Small Intestinal Atresia	29	4	33	2.27	1.56-3.18

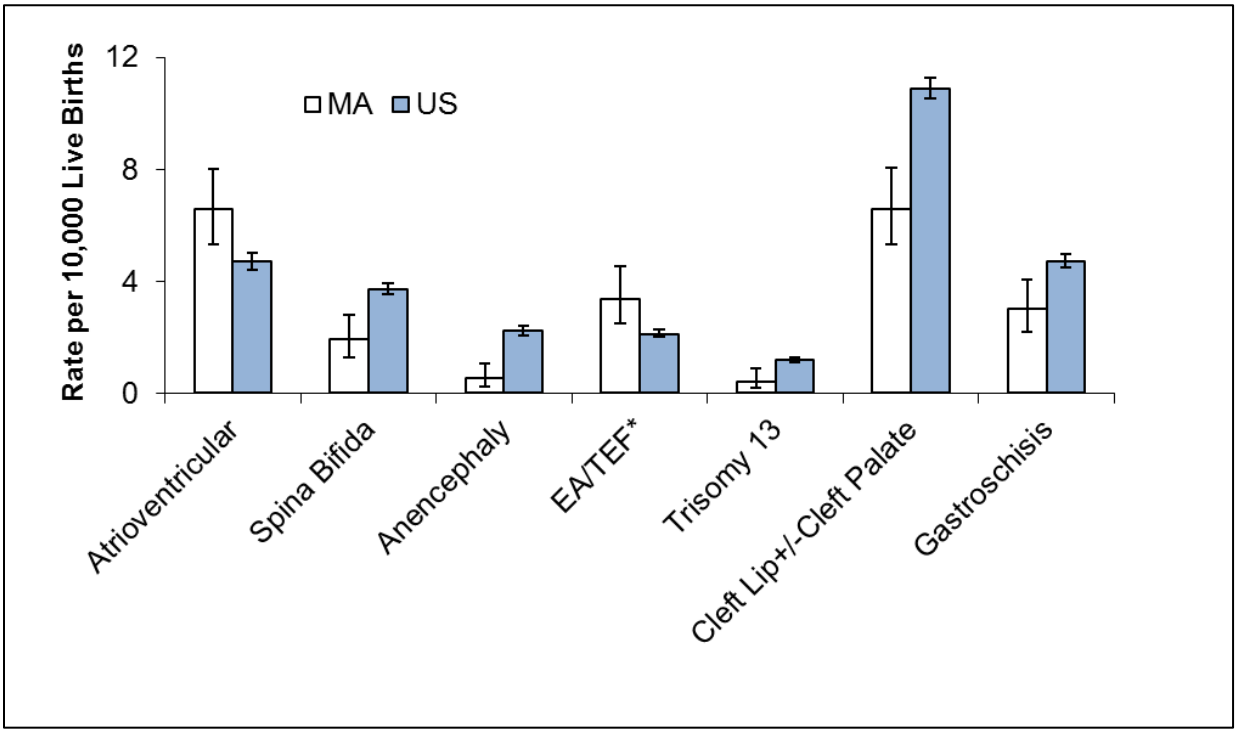
Table 1. Prevalence of Birth Defects, Massachusetts: 2011-2012					
Defect[1]	**Live birth Count**	**Stillbirth Count**	**Total Count**	**Rate per 10,000 Births**	**95% Confidence Interval**
Genitourinary					
Bladder Exstrophy	3	0	3	0.21	0.04-0.60
Cloacal Exstrophy	5	2	7	0.48	0.19-0.99
Hypospadias, 2nd or 3rd Degree[4]	163	0	163	21.85	18.62-25.47
Obstructive Genitourinary Defect	211	2	213	14.63	12.73-16.73
Renal Agenesis/Hypoplasia[5]	5	0	5	0.34	0.11-0.80
Musculoskeletal					
Club Foot	210	7	217	14.90	12.98-17.02
Craniosynostosis	67	0	67	4.60	3.57-5.84
Diaphragmatic Hernia	32	2	34	2.33	1.62-3.26
Gastroschisis	42	2	44	3.02	2.20-4.06
Omphalocele	25	3	28	1.92	1.28-2.78
Polydactyly/Syndactyly	241	3	244	16.75	14.72-18.99
Reduction Deformity, Lower Limbs	21	2	23	1.58	1.00-2.37
Reduction Deformity, Upper Limbs	43	0	43	2.95	2.14-3.98
Skeletal Dysplasia	26	4	30	2.06	1.39-2.94
Chromosomal and other Syndromes					
Klinefelter Syndrome	7	0	7	0.48	0.19-0.99
Trisomy 13	4	2	6	0.41	0.15-0.90
Trisomy 18	15	21	36	2.47	1.73-3.42
Trisomy 21 (Down Syndrome)	182	12	194	13.32	11.51-15.33
Turner Syndrome [6]	9	1	10	1.34	0.64-2.47
Other Chromosomal Syndromes/Other Syndromes[2]	231	6	237	16.27	14.27-18.48
Other					
Amniotic Bands	18	1	19	1.30	0.79-2.04
Skin Anomalies[2]	24	0	24	1.65	1.06-2.45

[1] Cases can be included in the count for more than one defect. Cases are counted once in the total for a defect category.
[2] Rate represents a heterogeneous group of defects.
[3] Excludes Patent Ductus Arteriosus.
[4] Rate calculated using male live births.
[5] Bilateral only.
[6] Rate calculated using female live births.

Birth Defects in Massachusetts vs. United States

Massachusetts is one of 11 states with active case ascertainment programs 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 2011-2012 are significantly lower than the most recent US rates (7) for anencephaly, spina bifida, cleft lip +/- palate, and gastroschisis. Massachusetts rates are significantly higher for atrioventricular septal defects and for esophageal atresia/tracheoesophageal fistula (Figure 2, Table 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 2011-12 are based on live births and stillbirths, whereas most other programs that contribute to these rates also collect information on elective terminations. This could result in undercounting of certain defects in Massachusetts, especially those not compatible with life. In Chapter 8, we look at the impact of including additional pregnancy outcomes.

Figure 2. Prevalence of Selected Birth Defects, Massachusetts vs. United States



*Includes tracheoesophageal fistula.
MA rates based on live births and stillbirths, N=2731.
US rates based on crude, pooled prevalence data from 11 active case-ascertainment programs, including Massachusetts (7). Nine of the other states contributing to the pooled estimates include elective terminations in addition to live births and stillbirths.
Error bars represent 95% confidence intervals.

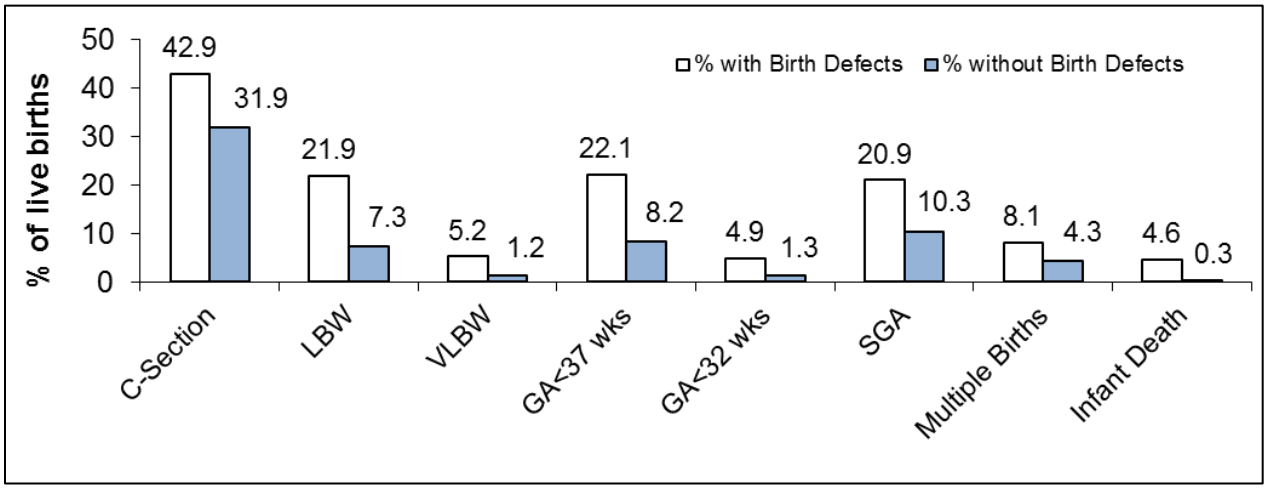
Table 2. Prevalence of Selected Birth Defects, Massachusetts vs. United States					
Defect	**Count MA**	**Rate MA[1]**	**95% CI**	**Rate US[2]**	**95% CI**
Anencephaly	8	.55	0.24-1.08	2.23	2.07-2.41
Spina bifida without anencephaly	28	1.92	1.28-2.78	3.72	3.52-3.94
Anophthalmia/Microphthalmia	14	0.96	0.53-1.61	2.10	1.94-2.27
Truncus arteriosus (common truncus)	5	0.34	0.11-0.80	0.74	0.65-0.84
Transposition of the Great Arteries	40	2.75	1.96-3.74	3.04	2.85-3.24
Tetralogy of Fallot	50	3.43	2.55-4.53	4.05	3.83-4.28
Atrioventricular septal defect[3]	96	6.59	5.34-8.05	4.70	4.45-4.96
Hypoplastic left heart syndrome	24	1.65	1.06-2.45	2.31	2.14-2.48
Cleft Palate without Cleft Lip	85	5.84	4.66-7.22	6.45	6.17-6.74
Cleft Lip with and without Cleft Palate	96	6.59	5.34-8.05	10.89	10.53-11.26
Esophageal Atresia/Tracheoesophageal Fistula	49	3.36	2.49-4.45	2.12	1.96-2.29
Rectal and Large Intestinal Atresia/Stenosis	55	3.78	2.85-4.92	4.86	4.61-5.14
Reduction Deformity, Lower Limbs	23	1.58	1.00-2.37	1.65	1.51-1.80
Reduction Deformity, Upper Limbs	45	2.95	2.14-3.98	3.64	3.43-3.86
Gastroschisis	44	3.02	2.20-4.06	4.72	4.49-4.97
Omphalocele	28	1.92	1.28-2.78	1.92	1.77-2.08
Diaphragmatic Hernia	34	2.33	1.62-3.26	2.60	2.42-2.79
Trisomy 21 (Down Syndrome)	194	13.32	11.51-15.33	13.48	13.08-13.90
Trisomy 13	6	0.41	0.15-0.90	1.20	1.09-1.33
Trisomy 18	36	2.47	1.73-3.42	2.55	2.38-2.73

[1] Rate per 10,000 live births. Includes live births and stillbirths, 2011 and 2012.
[2] 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.
[3] Includes endocardial cushion defect, complete atrioventricular canal defect, atrial septal defect (ASD) primum, common atrium, and canal type ventricular septal defect (VSD).

Selected Pregnancy Outcomes

Adverse pregnancy outcomes such as Cesarean section (C-section), low birth weight (LBW), prematurity, and small for gestational age (SGA) are more frequent among infants born with birth defects than among unaffected infants. Infants with a birth defect are 3 times more likely to have low birth weight (less than 2500 grams) or very low birth weight (less than 1500 grams) and 2.7 times more likely to be born premature (before 37 weeks) compared to those without birth defects (Figure 3). Cesarean (C-section) deliveries are more common among live born infants with birth defects (43%) compared to unaffected infants (32%). In addition, infants with birth defects are about 15 times more likely to die in their first year of life.

Figure 3. Pregnancy Outcomes among Live Births with and without Birth Defects, Massachusetts: 2011-2012

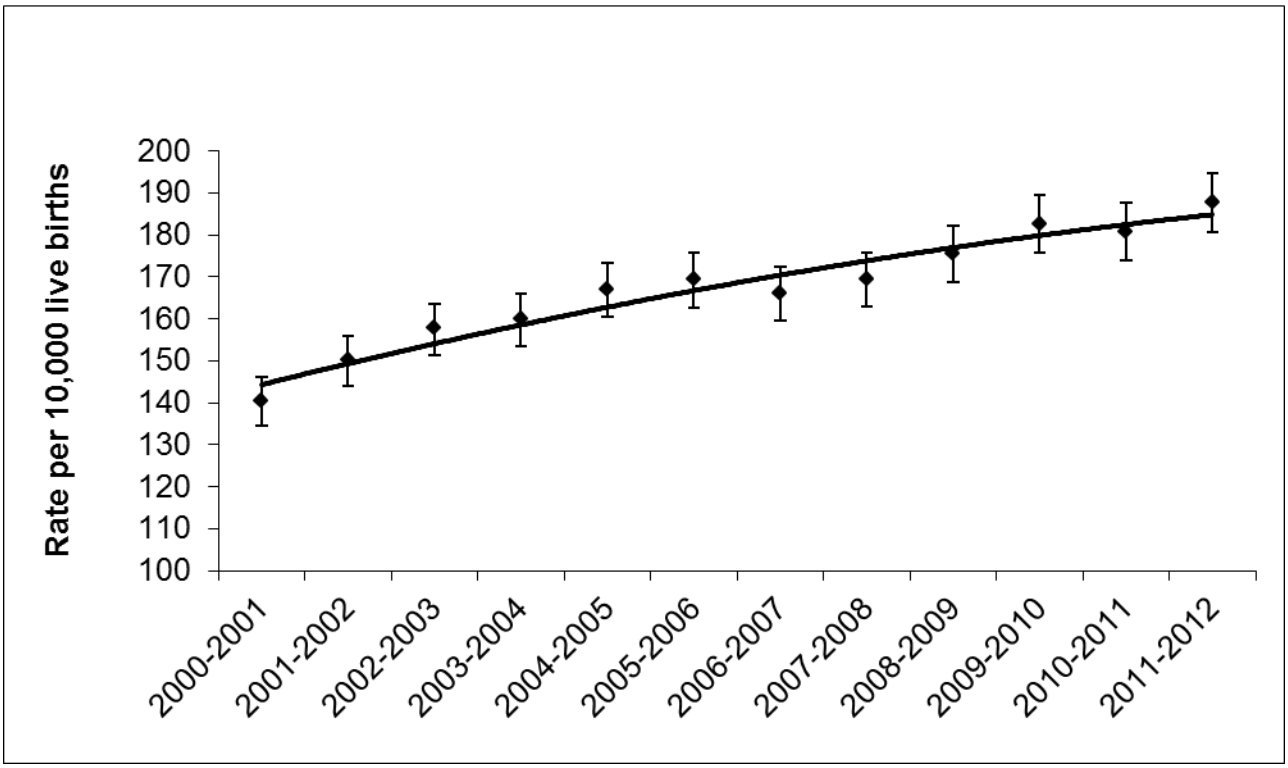


N=2663 live births with birth defects; N=145,631 live births without birth defects.
C-section: Cesarean section; LBW: low birth weight; VLBW: very low birth weight; GA: gestational age; SGA: small-for-gestational age.
SGA calculation based on method described by Oken et al. in 2003, defined as birth weight below the 10th percentile for gestational age based on a sex-specific US standard (17).

Trend Analysis

The rate of birth defects in Massachusetts has increased steadily over time (Figure 4). This may reflect improvements in case ascertainment or confirmation, changes in the distribution of demographic variables over time (e.g. more births to older mothers), changes in survival to diagnosis, random variation, or true increases in the overall rate of birth defects.

Figure 4. Overall Prevalence of Birth Defects by Year, Massachusetts: 2000-2012, 2-year rolling average



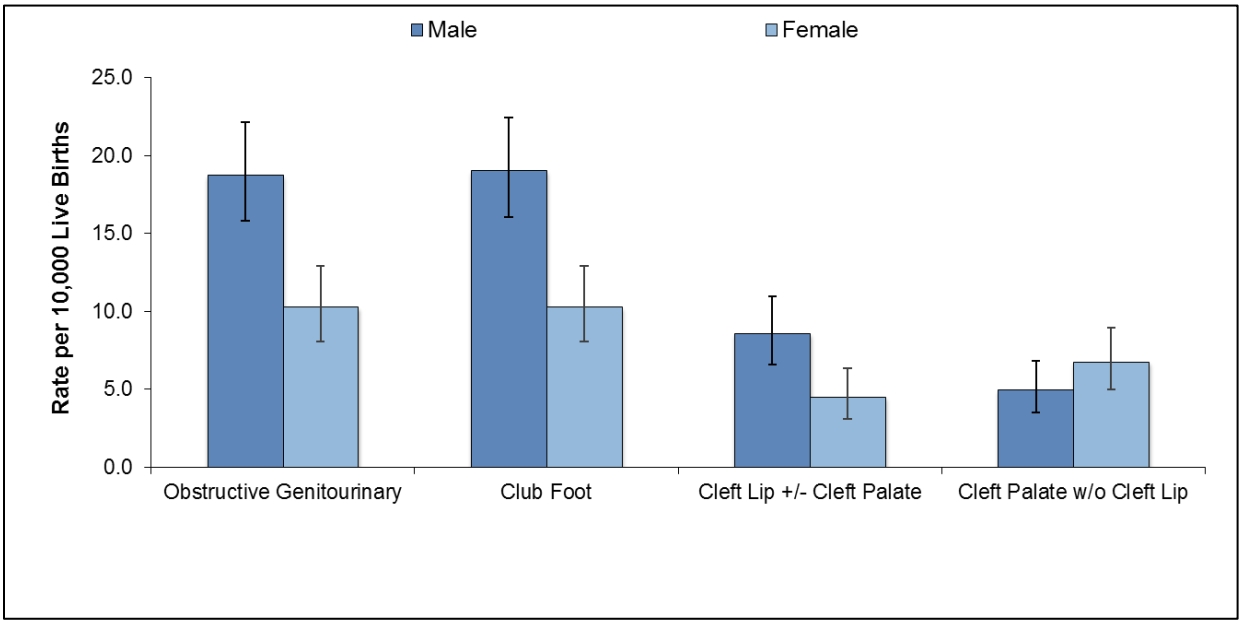
Live births and stillbirths.
Note: Y axis begins at 100.
Error bars represent 95% confidence intervals.

Chapter 4: Prevalence of Birth Defects by Infant Sex and Plurality

Infant Sex

Among males, the prevalence of birth defects in Massachusetts in 2011-2012 is 216.9 (95% CI: 206.6-227.6) per 10,000 live births, and among females the prevalence is 156.4 (95% CI: 147.4-165.5) per 10,000 live births. Males are significantly more likely to have obstructive genitourinary defects, club foot, and cleft lip (Figure 5, Table 3).

Figure 5. Prevalence of Selected Birth Defects by Infant Sex, Massachusetts: 2011-2012



Live births and stillbirths, N=2731.
Error bars represent 95% confidence intervals.

Table 3. Prevalence of Birth Defects by Infant Sex, Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Sex**	**Count**	**Rate[1]**	**95% CI**
Central Nervous System				
Anencephaly	Male	7	0.99	0.40-2.03
	Female	1	0.13	0.00-0.75
Encephalocele	Male	3	0.42	0.09-1.23
	Female	3	0.40	0.08-1.18
Holoprosencephaly	Male	5	0.70	0.23-1.64
	Female	5	0.67	0.22-1.56
Hydrocephaly without Spina Bifida	Male	26	3.66	2.39-5.36
	Female	24	3.22	2.06-4.79
Microcephaly	Male	14	1.97	1.08-3.31
	Female	14	1.88	1.03-3.15
Spina Bifida with and without Hydrocephaly	Male	16	2.25	1.29-3.66
	Female	12	1.61	0.83-2.81
Spinal Cord	Male	41	5.77	4.14-7.83
	Female	31	4.16	2.82-5.90
Eye				
Aniridia	Male	1	0.14	0.00-0.78
	Female	3	0.40	0.08-1.18
Anophthalmia/Microphthalmia	Male	5	0.70	0.23-1.64
	Female	9	1.21	0.55-2.29
Congenital Glaucoma, Congenital Cataract	Male	25	3.52	2.28-5.20
	Female	20	2.68	1.64-4.14
Ear				
Anotia/Microtia	Male	20	2.82	1.72-4.35
	Female	16	2.14	1.23-3.48
Cardiovascular				
Anomalous Pulmonary Venous Connection				
Total Anomalous Pulmonary Venous Connection	Male	11	1.55	0.77-2.77
	Female	10	1.34	0.64-2.47
Atrioventricular Canal Defects				
Atrial Septal Defect (ASD) Primum	Male	1	0.14	0.00-0.78
	Female	0	-	-

Table 3. Prevalence of Birth Defects by Infant Sex, Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Sex**	**Count**	**Rate[1]**	**95% CI**
Common Atrium	Male	1	0.14	0.00-0.78
	Female	4	0.54	0.15-1.37
Complete Atrioventricular Canal Defect	Male	23	3.24	2.05-4.86
	Female	38	5.09	3.60-6.99
Endocardial Cushion Defect, Other specified (OS) and Not otherwise specified (NOS)	Male	8	1.13	0.49-2.22
	Female	12	1.61	0.83-2.81
Ventricular Septal Defect (VSD), Canal Type	Male	7	0.99	0.40-2.03
	Female	2	0.27	0.03-0.97
Conotruncal (Outlet) and Aortic Arch				
Double Outlet Right Ventricle	Male	11	1.55	0.77-2.77
	Female	5	0.67	0.22-1.56
Tetralogy of Fallot with and without Pulmonary Atresia	Male	28	3.94	2.62-5.70
	Female	22	2.95	1.85-4.47
Truncus	Male	0	-	-
	Female	5	0.67	0.22-1.56
dextro-Transposition of the Great Arteries	Male	24	3.38	2.16-5.03
	Female	10	1.34	0.64-2.47
Ebstein Anomaly				
Ebstein Anomaly	Male	3	0.42	0.09-1.23
	Female	2	0.27	0.03-0.97
Heterotaxy (Laterality Defects)				
Heterotaxy	Male	3	0.42	0.09-1.23
	Female	5	0.67	0.22-1.56
Left-Sided Obstruction				
Aortic Valve Stenosis	Male	9	1.27	0.58-2.41
	Female	8	1.07	0.46-2.11
Aortic Arch Atresia	Male	1	0.14	0.00-0.78
	Female	0	-	-
Coarctation of Aorta	Male	40	5.63	4.02-7.67
	Female	28	3.75	2.49-5.42
Hypoplastic Left Heart Syndrome	Male	14	1.97	1.08-3.31
	Female	10	1.34	0.64-2.47

Table 3. Prevalence of Birth Defects by Infant Sex, Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Sex**	**Count**	**Rate[1]**	**95% CI**
Interrupted Aortic Arch (Type A and NOS)	Male	2	0.28	0.03-1.02
	Female	2	0.27	0.03-0.97
Right-Sided Obstruction				
Pulmonary Stenosis, Valvular	Male	51	7.18	5.35-9.44
	Female	68	9.12	7.08-11.56
Pulmonary Valve Atresia with intact septum	Male	5	0.70	0.23-1.64
	Female	1	0.13	0.00-0.75
Pulmonary Valve Atresia with VSD	Male	0	-	-
	Female	1	0.13	0.00-0.75
Tricuspid Valve Atresia	Male	3	0.42	0.09-1.23
	Female	0	-	-
Septal Defects				
ASD (Secundum and NOS)	Male	156	21.96	18.65-25.69
	Female	174	23.33	19.99-27.06
VSD (Membranous and NOS)	Male	89	12.53	10.06-15.42
	Female	89	11.93	9.58-14.68
VSD, Conoventricular/Malalignment	Male	13	1.83	0.97-3.13
	Female	10	1.34	0.64-2.47
Single Ventricle and L-TGA				
levo-Transposition of the Great Arteries	Male	4	0.56	0.15-1.44
	Female	2	0.27	0.03-0.97
Single Ventricle	Male	2	0.28	0.03-1.02
	Female	1	0.13	0.00-0.75
Respiratory				
Choanal Atresia	Male	4	0.56	0.15-1.44
	Female	8	1.07	0.46-2.11
Lung Anomalies	Male	27	3.80	2.51-5.53
	Female	12	1.61	0.83-2.81
Orofacial				
Cleft Lip with and without Cleft Palate	Male	64	9.01	6.94-11.51
	Female	32	4.29	2.93-6.06
Cleft Palate without Cleft Lip	Male	37	5.21	3.67-7.18
	Female	48	6.43	4.74-8.53

Table 3. Prevalence of Birth Defects by Infant Sex, Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Sex**	**Count**	**Rate[1]**	**95% CI**
Pierre Robin Sequence	Male	17	2.39	1.39-3.83
	Female	19	2.55	1.53-3.98
Biliary Atresia	Male	2	0.28	0.03-1.02
	Female	12	1.61	0.83-2.81
Esophageal Atresia/Tracheoesophageal Fistula	Male	31	4.36	2.97-6.19
	Female	18	2.41	1.43-3.81
Hirschsprung Disease	Male	19	2.67	1.61-4.18
	Female	4	0.54	0.15-1.37
Rectal and Large Intestinal Atresia/Stenosis	Male	22	3.10	1.94-4.69
	Female	33	4.42	3.05-6.21
Small Intestinal Atresia	Male	18	2.53	1.50-4.01
	Female	15	2.01	1.13-3.32
Genitourinary				
Bladder Exstrophy	Male	3	0.42	0.09-1.23
	Female	0	-	-
Cloacal Exstrophy	Male	0	-	-
	Female	7	0.94	0.38-1.93
Hypospadias, 2nd or 3rd Degree[2]	Male	163	22.95	19.56-26.75
	Female	0	-	-
Obstructive Genitourinary Defect	Male	140	19.71	16.58-23.26
	Female	73	9.79	7.67-12.30
Renal Agenesis/Hypoplasia	Male	5	0.70	0.23-1.64
	Female	0	-	-
Musculoskeletal				
Club Foot	Male	142	19.99	16.84-23.56
	Female	73	9.79	7.67-12.30
Craniosynostosis	Male	44	6.19	4.50-8.32
	Female	23	3.08	1.95-4.63
Diaphragmatic Hernia	Male	15	2.11	1.18-3.48
	Female	19	2.55	1.53-3.98
Gastroschisis	Male	25	3.52	2.28-5.20
	Female	18	2.41	1.43-3.81

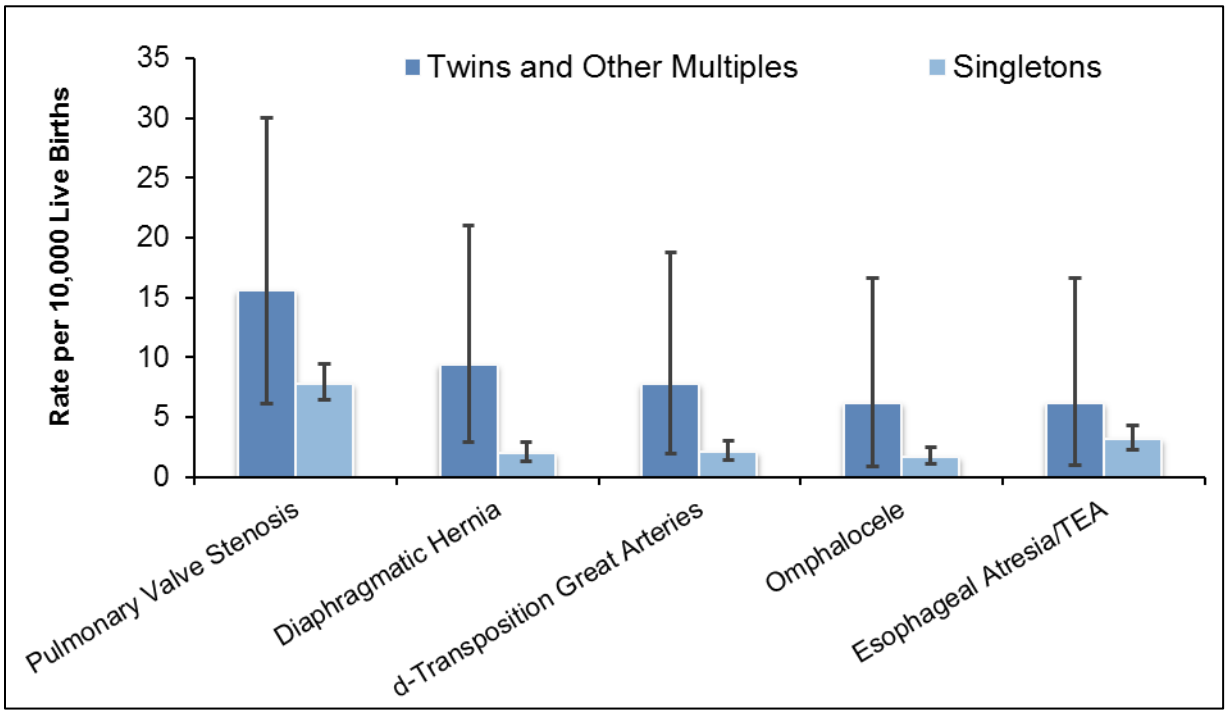
Table 3. Prevalence of Birth Defects by Infant Sex, Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Sex**	**Count**	**Rate[1]**	**95% CI**
Omphalocele	Male	20	2.82	1.72-4.35
	Female	8	1.07	0.46-2.11
Polydactyly/Syndactyly	Male	144	20.27	17.10-23.87
	Female	99	13.27	10.79-16.16
Reduction Deformity, Lower Limbs	Male	8	1.13	0.49-2.22
	Female	15	2.01	1.13-3.32
Reduction Deformity, Upper Limbs	Male	27	3.80	2.51-5.53
	Female	16	2.14	1.23-3.48
Skeletal Dysplasia	Male	12	1.69	0.87-2.95
	Female	18	2.41	1.43-3.81
Chromosomal and other Syndromes				
Klinefelter Syndrome	Male	7	0.99	0.40-2.03
	Female	0	-	-
Trisomy 13	Male	2	0.28	0.03-1.02
	Female	4	0.54	0.15-1.37
Trisomy 18	Male	13	1.83	0.97-3.13
	Female	23	3.08	1.95-4.63
Trisomy 21 (Down Syndrome)	Male	106	14.92	12.22-18.05
	Female	88	11.80	9.46-14.53
Turner Syndrome [3]	Male	0	-	-
	Female	10	1.34	0.64-2.47
Other				
Amniotic Bands	Male	5	0.70	0.23-1.64
	Female	13	1.74	0.93-2.98
Skin Anomalies	Male	19	2.67	1.61-4.18
	Female	5	0.67	0.22-1.56

[1] Rate per 10,000 live births.
[2] In males.
[3] In females.

Plurality

The overall birth defect rate in singletons is 181.8 (95% CI: 174.9-189.0) per 10,000 live births, while the rate in multiples is 311.6 (95% CI: 269.9-357.9) per 10,000 live births. While many individual defects do not differ significantly by plurality (Figure 6), the rates of septal defects, hypospadias, and diaphragmatic hernia are significantly higher in multiples than in singletons (Table 4).

Figure 6. Prevalence of Selected Birth Defects by Single and Multiple Births, Massachusetts: 2011-2012



Live births and stillbirths, N=2731.
TEA=tracheoesophageal fistula.
Error bars represent 95% confidence intervals.

Table 4. Prevalence of Birth Defects by Plurality, Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Plurality**	**Count**	**Rate[1]**	**95% CI**
Central Nervous System				
Anencephaly	Singleton	7	0.50	0.20-1.04
	Multiple	1	1.56	0.04-8.68
Encephalocele	Singleton	6	0.43	0.16-0.94
	Multiple	0	-	-
Holoprosencephaly	Singleton	10	0.72	0.34-1.32
	Multiple	0	-	-
Hydrocephaly without Spina Bifida	Singleton	45	3.23	2.36-4.33
	Multiple	5	7.79	2.53-18.18
Microcephaly	Singleton	27	1.94	1.28-2.82
	Multiple	1	1.56	0.04-8.68
Spina Bifida with and without Hydrocephaly	Singleton	28	2.01	1.34-2.91
	Multiple	0	-	-
Spinal Cord	Singleton	65	4.67	3.60-5.95
	Multiple	7	10.91	4.39-22.47
Eye				
Aniridia	Singleton	4	0.29	0.08-0.74
	Multiple	0	-	-
Anophthalmia/Microphthalmia	Singleton	13	0.93	0.50-1.60
	Multiple	1	1.56	0.04-8.68
Congenital Glaucoma, Congenital Cataract	Singleton	44	3.16	2.30-4.24
	Multiple	1	1.56	0.04-8.68
Ear				
Anotia/Microtia	Singleton	35	2.51	1.75-3.50
	Multiple	1	1.56	0.04-8.68
Cardiovascular				
Anomalous Pulmonary Venous Connection				
Total Anomalous Pulmonary Venous Connection	Singleton	20	1.44	0.88-2.22
	Multiple	1	1.56	0.04-8.68
Atrioventricular Canal Defects				
Atrial Septal Defect (ASD) primum	Singleton	1	0.07	0.00-0.40
	Multiple	0	-	-

Table 4. Prevalence of Birth Defects by Plurality, Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Plurality**	**Count**	**Rate[1]**	**95% CI**
Common Atrium	Singleton	5	0.36	0.12-0.84
	Multiple	0	-	-
Complete Atrioventricular Canal Defect	Singleton	57	4.09	3.10-5.30
	Multiple	4	6.23	1.70-15.96
Endocardial Cushion Defect, Other specified (OS) and Not otherwise specified (NOS)	Singleton	18	1.29	0.77-2.04
	Multiple	2	3.12	0.38-11.26
Ventricular Septal Defect (VSD), Canal Type	Singleton	9	0.65	0.30-1.23
	Multiple	0	-	-
Conotruncal (Outlet) and Aortic Arch				
Double Outlet Right Ventricle	Singleton	16	1.15	0.66-1.87
	Multiple	0	-	-
Tetralogy of Fallot with/without Pulmonary Atresia	Singleton	46	3.30	2.42-4.41
	Multiple	4	6.23	1.70-15.96
Truncus	Singleton	3	0.22	0.04-0.63
	Multiple	2	3.12	0.38-11.26
dextro-Transposition of the Great Arteries	Singleton	29	2.08	1.40-2.99
	Multiple	5	7.79	2.53-18.18
Ebstein Anomaly				
Ebstein Anomaly	Singleton	5	0.36	0.12-0.84
	Multiple	0	-	-
Heterotaxy (Laterality Defects)				
Heterotaxy	Singleton	8	0.57	0.25-1.13
	Multiple	0	-	-
Left-Sided Obstruction				
Aortic Valve Stenosis	Singleton	15	1.08	0.60-1.78
	Multiple	2	3.12	0.38-11.26
Aortic arch atresia	Singleton	1	0.07	0.00-0.40
	Multiple	0	-	-
Coarctation of Aorta	Singleton	67	4.81	3.73-6.11
	Multiple	1	1.56	0.04-8.68
Hypoplastic Left Heart Syndrome	Singleton	23	1.65	1.05-2.48
	Multiple	1	1.56	0.04-8.68

Table 4. Prevalence of Birth Defects by Plurality, Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Plurality**	**Count**	**Rate[1]**	**95% CI**
Interrupted Aortic Arch (Type A and NOS)	Singleton	3	0.22	0.04-0.63
	Multiple	1	1.56	0.04-8.68
Right-Sided Obstruction				
Pulmonary Stenosis, Valvular	Singleton	109	7.83	6.43-9.45
	Multiple	10	15.58	7.47-28.65
Pulmonary Valve Atresia with intact septum	Singleton	6	0.43	0.16-0.94
	Multiple	0	-	-
Pulmonary Valve Atresia with Ventricular Septal Defect	Singleton	1	0.07	0.00-0.40
	Multiple	0	-	-
Tricuspid Valve Atresia	Singleton	3	0.22	0.04-0.63
	Multiple	0	-	-
Septal Defects				
ASD (Secundum and NOS)	Singleton	295	21.19	18.84-23.75
	Multiple	35	54.53	37.99-75.84
VSD (Membranous and NOS)	Singleton	161	11.57	9.85-13.50
	Multiple	17	26.49	15.43-42.41
VSD, Conoventricular/Malalignment	Singleton	21	1.51	0.93-2.31
	Multiple	2	3.12	0.38-11.26
Single Ventricle and L-TGA				
levo-Transposition of the Great Arteries	Singleton	6	0.43	0.16-0.94
	Multiple	0	-	-
Single Ventricle	Singleton	3	0.22	0.04-0.63
	Multiple	0	-	-
Respiratory				
Choanal Atresia	Singleton	9	0.65	0.30-1.23
	Multiple	3	4.67	0.96-13.66
Lung Anomalies	Singleton	37	2.66	1.87-3.66
	Multiple	2	3.12	0.38-11.26
Orofacial				
Cleft Lip with and without Cleft Palate	Singleton	93	6.68	5.39-8.18
	Multiple	3	4.67	0.96-13.66

Table 4. Prevalence of Birth Defects by Plurality, Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Plurality**	**Count**	**Rate[1]**	**95% CI**
Cleft Palate without Cleft Lip	Singleton	81	5.82	4.62-7.23
	Multiple	4	6.23	1.70-15.96
Pierre Robin Sequence	Singleton	34	2.44	1.69-3.41
	Multiple	2	3.12	0.38-11.26
Gastrointestinal				
Biliary Atresia	Singleton	13	0.93	0.50-1.60
	Multiple	1	1.56	0.04-8.68
Esophageal Atresia/Tracheoesophageal Fistula	Singleton	45	3.23	2.36-4.33
	Multiple	4	6.23	1.70-15.96
Hirschsprung Disease	Singleton	22	1.58	0.99-2.39
	Multiple	1	1.56	0.04-8.68
Rectal and Large Intestinal Atresia/Stenosis	Singleton	54	3.88	2.91-5.06
	Multiple	1	1.56	0.04-8.68
Small Intestinal Atresia	Singleton	31	2.23	1.51-3.16
	Multiple	2	3.12	0.38-11.26
Genitourinary				
Bladder Exstrophy	Singleton	3	0.22	0.04-0.63
	Multiple	0	-	-
Cloacal Exstrophy	Singleton	7	0.50	0.20-1.04
	Multiple	0	-	-
Hypospadias, 2nd or 3rd Degree[1]	Singleton	142	19.90	16.76-23.45
	Multiple	21	64.98	40.22-99.32
Obstructive Genitourinary Defect	Singleton	202	14.51	12.58-16.66
	Multiple	11	17.14	8.56-30.67
Renal Agenesis/Hypoplasia	Singleton	4	0.29	0.08-0.74
	Multiple	1	1.56	0.04-8.68
Musculoskeletal				
Club Foot	Singleton	201	14.44	12.51-16.58
	Multiple	16	24.93	14.25-40.48
Craniosynostosis	Singleton	64	4.60	3.54-5.87
	Multiple	3	4.67	0.96-13.66

Table 4. Prevalence of Birth Defects by Plurality, Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Plurality**	**Count**	**Rate[1]**	**95% CI**
Diaphragmatic Hernia	Singleton	28	2.01	1.34-2.91
	Multiple	6	9.35	3.43-20.35
Gastroschisis	Singleton	43	3.09	2.24-4.16
	Multiple	1	1.56	0.04-8.68
Omphalocele	Singleton	24	1.72	1.10-2.57
	Multiple	4	6.23	1.70-15.96
Polydactyly/Syndactyly	Singleton	233	16.74	14.66-19.03
	Multiple	11	17.14	8.56-30.67
Reduction Deformity, Lower Limbs	Singleton	22	1.58	0.99-2.39
	Multiple	1	1.56	0.04-8.68
Reduction Deformity, Upper Limbs	Singleton	42	3.02	2.17-4.08
	Multiple	1	1.56	0.04-8.68
Skeletal Dysplasia	Singleton	25	1.80	1.16-2.65
	Multiple	5	7.79	2.53-18.18
Chromosomal/other Syndromes				
Klinefelter Syndrome	Singleton	7	0.50	0.20-1.04
	Multiple	0	-	-
Trisomy 13	Singleton	5	0.36	0.12-0.84
	Multiple	1	1.56	0.04-8.68
Trisomy 18	Singleton	33	2.37	1.63-3.33
	Multiple	3	4.67	0.96-13.66
Trisomy 21 (Down Syndrome)	Singleton	181	13.00	11.18-15.04
	Multiple	13	20.26	10.79-34.64
Turner Syndrome[3]	Singleton	10	1.34	0.64-2.47
	Multiple	0	-	-
Other				
Amniotic Bands	Singleton	17	1.22	0.71-1.96
	Multiple	2	3.12	0.38-11.26
Skin Anomalies	Singleton	24	1.72	1.10-2.57
	Multiple	0	-	-

[1] Rates per 10,000 live births.
[2] Rate calculated using male live births.
[3] Rate calculated using female live births.

Chapter 5: Prevalence of Birth Defects by Maternal Age

Maternal Age

The prevalence of birth defects varies by maternal age (Table 5), with rates highest for mothers ages 35 years and older (220.7 per 10,000 live births) and second highest for mothers younger than 20 years of age (190.7 per 10,000 live births).

Table 5. Overall Prevalence of Birth Defects by Maternal Age, Massachusetts: 2011-2012

Maternal Age (years)	Cases	Rate	95% CI
<20	129	190.7	159.2-226.6
20-24	385	174.9	158.0-193.1
25-29	643	177.7	164.3-191.8
30-34	854	177.8	166.2-190.0
35+	720	220.7	205.0-237.2

Live births and stillbirths, N=2731.
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 has 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 6. 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.

Table 6. Prevalence of Birth Defects by Maternal Age Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Maternal Age**	**Count**	**Rate[1]**	**95% CI**
Central Nervous System				
Anencephaly	<20	0	-	-
	20-24	1	0.45	0.01-2.53
	25-29	2	0.55	0.07-2.00
	30-34	1	0.21	0.01-1.16
	35+	4	1.23	0.33-3.14
Encephalocele	<20	0	-	-
	20-24	5	2.27	0.74-5.30
	25-29	1	0.28	0.01-1.54
	30-34	0	-	-
	35+	0	-	-
Holoprosencephaly	<20	1	1.48	0.04-8.24
	20-24	2	0.91	0.11-3.28
	25-29	1	0.28	0.01-1.54
	30-34	3	0.62	0.13-1.83
	35+	3	0.92	0.19-2.69
Hydrocephaly without Spina Bifida	<20	3	4.44	0.91-12.96
	20-24	11	5.00	2.49-8.94
	25-29	12	3.32	1.71-5.79
	30-34	15	3.12	1.75-5.15
	35+	9	2.76	1.26-5.24
Microcephaly	<20	1	1.48	0.04-8.24
	20-24	2	0.91	0.11-3.28
	25-29	8	2.21	0.95-4.36
	30-34	8	1.67	0.72-3.28
	35+	9	2.76	1.26-5.24
Spina Bifida with and without Hydrocephaly	<20	2	2.96	0.36-10.68
	20-24	5	2.27	0.74-5.30
	25-29	6	1.66	0.61-3.61
	30-34	7	1.46	0.59-3.00
	35+	8	2.45	1.06-4.83

Table 6. Prevalence of Birth Defects by Maternal Age Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Maternal Age**	**Count**	**Rate[1]**	**95% CI**
Spinal Cord	<20	0	-	-
	20-24	9	4.09	1.87-7.76
	25-29	19	5.25	3.16-8.20
	30-34	24	5.00	3.20-7.43
	35+	20	6.13	3.74-9.47
Eye				
Aniridia	<20	0	-	-
	20-24	1	0.45	0.01-2.53
	25-29	0	-	-
	30-34	2	0.42	0.05-1.50
	35+	1	0.31	0.01-1.71
Anophthalmia/Microphthalmia	<20	3	4.44	0.91-12.96
	20-24	1	0.45	0.01-2.53
	25-29	1	0.28	0.01-1.54
	30-34	6	1.25	0.46-2.72
	35+	3	0.92	0.19-2.69
Congenital Glaucoma, Congenital Cataract	<20	2	2.96	0.36-10.68
	20-24	6	2.73	1.00-5.93
	25-29	12	3.32	1.71-5.79
	30-34	14	2.91	1.59-4.89
	35+	11	3.37	1.68-6.03
Ear				
Anotia/Microtia	<20	2	2.96	0.36-10.68
	20-24	4	1.82	0.50-4.65
	25-29	7	1.93	0.78-3.99
	30-34	14	2.91	1.59-4.89
	35+	9	2.76	1.26-5.24

Table 6. Prevalence of Birth Defects by Maternal Age *Live Births and Stillbirths, Massachusetts: 2011-2012*				
Defect	**Maternal Age**	**Count**	**Rate[1]**	**95% CI**
Cardiovascular				
Anomalous Pulmonary Venous Connection				
Total Anomalous Pulmonary Venous Connection	<20	0	-	-
	20-24	2	0.91	0.11-3.28
	25-29	6	1.66	0.61-3.61
	30-34	6	1.25	0.46-2.72
	35+	7	2.15	0.86-4.42
Atrioventricular Canal Defects				
Atrial Septal Defect (ASD) primum	<20	0	-	-
	20-24	0	-	-
	25-29	0	-	-
	30-34	1	0.21	0.01-1.16
	35+	0	-	-
Common Atrium	<20	0	-	-
	20-24	1	0.45	0.01-2.53
	25-29	1	0.28	0.01-1.54
	30-34	1	0.21	0.01-1.16
	35+	2	0.61	0.07-2.21
Complete Atrioventricular Canal Defect	<20	5	7.39	2.40-17.25
	20-24	5	2.27	0.74-5.30
	25-29	10	2.76	1.33-5.08
	30-34	13	2.71	1.44-4.63
	35+	28	8.58	5.70-12.41
Endocardial Cushion Defect, Other specified (OS) and Not otherwise specified (NOS)	<20	1	1.48	0.04-8.24
	20-24	6	2.73	1.00-5.93
	25-29	1	0.28	0.01-1.54
	30-34	5	1.04	0.34-2.43
	35+	7	2.15	0.86-4.42

Table 6. Prevalence of Birth Defects by Maternal Age Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Maternal Age**	**Count**	**Rate[1]**	**95% CI**
Ventricular Septal Defect (VSD), canal type	<20	0	-	-
	20-24	0	-	-
	25-29	2	0.55	0.07-2.00
	30-34	4	0.83	0.23-2.13
	35+	3	0.92	0.19-2.69
Conotruncal (Outlet) and Aortic Arch				
Double Outlet Right Ventricle	<20	0	-	-
	20-24	4	1.82	0.50-4.65
	25-29	3	0.83	0.17-2.42
	30-34	5	1.04	0.34-2.43
	35+	4	1.23	0.33-3.14
Tetralogy of Fallot with and without Pulmonary Atresia	<20	4	5.91	1.61-15.14
	20-24	11	5.00	2.49-8.94
	25-29	13	3.59	1.91-6.14
	30-34	15	3.12	1.75-5.15
	35+	7	2.15	0.86-4.42
Truncus	<20	0	-	-
	20-24	1	0.45	0.01-2.53
	25-29	2	0.55	0.07-2.00
	30-34	0	-	-
	35+	2	0.61	0.07-2.21
dextro-Transposition of the Great Arteries	<20	1	1.48	0.04-8.24
	20-24	2	0.91	0.11-3.28
	25-29	5	1.38	0.45-3.22
	30-34	10	2.08	1.00-3.83
	35+	16	4.90	2.80-7.96
Ebstein Anomaly				
Ebstein Anomaly	<20	0	-	-
	20-24	1	0.45	0.01-2.53
	25-29	2	0.55	0.07-2.00
	30-34	0	-	-
	35+	2	0.61	0.07-2.21

Table 6. Prevalence of Birth Defects by Maternal Age Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Maternal Age**	**Count**	**Rate[1]**	**95% CI**
Heterotaxy (Laterality Defects)				
Heterotaxy	<20	1	1.48	0.04-8.24
	20-24	1	0.45	0.01-2.53
	25-29	2	0.55	0.07-2.00
	30-34	2	0.42	0.05-1.50
	35+	2	0.61	0.07-2.21
Left-Sided Obstruction				
Aortic Valve Stenosis	<20	2	2.96	0.36-10.68
	20-24	2	0.91	0.11-3.28
	25-29	3	0.83	0.17-2.42
	30-34	5	1.04	0.34-2.43
	35+	5	1.53	0.50-3.58
Aortic arch atresia	<20	0	-	-
	20-24	0	-	-
	25-29	0	-	-
	30-34	1	0.21	0.01-1.16
	35+	0	-	-
Coarctation of Aorta	<20	3	4.44	0.91-12.96
	20-24	11	5.00	2.49-8.94
	25-29	12	3.32	1.71-5.79
	30-34	27	5.62	3.70-8.18
	35+	15	4.60	2.57-7.58
Hypoplastic Left Heart Syndrome	<20	1	1.48	0.04-8.24
	20-24	5	2.27	0.74-5.30
	25-29	6	1.66	0.61-3.61
	30-34	6	1.25	0.46-2.72
	35+	6	1.84	0.67-4.00
Interrupted Aortic Arch (Type A and NOS)	<20	0	-	-
	20-24	0	-	-
	25-29	3	0.83	0.17-2.42
	30-34	1	0.21	0.01-1.16
	35+	0	-	-

Table 6. Prevalence of Birth Defects by Maternal Age Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Maternal Age**	**Count**	**Rate[1]**	**95% CI**
Right-Sided Obstruction				
Pulmonary Stenosis, Valvular	<20	4	5.91	1.61-15.14
	20-24	15	6.81	3.81-11.24
	25-29	33	9.12	6.28-12.81
	30-34	39	8.12	5.77-11.10
	35+	28	8.58	5.70-12.41
Pulmonary Valve Atresia with intact septum	<20	0	-	-
	20-24	2	0.91	0.11-3.28
	25-29	1	0.28	0.01-1.54
	30-34	1	0.21	0.01-1.16
	35+	2	0.61	0.07-2.21
Pulmonary Valve Atresia with VSD	<20	1	1.48	0.04-8.24
	20-24	0	-	-
	25-29	0	-	-
	30-34	0	-	-
	35+	0	-	-
Tricuspid Valve Atresia	<20	0	-	-
	20-24	0	-	-
	25-29	0	-	-
	30-34	2	0.42	0.05-1.50
	35+	1	0.31	0.01-1.71
Septal Defects				
ASD (Secundum and NOS)	<20	10	14.78	7.09-27.19
	20-24	47	21.35	15.69-28.39
	25-29	74	20.45	16.06-25.67
	30-34	102	21.23	17.31-25.78
	35+	97	29.73	24.11-36.27
VSD (Membranous and NOS)	<20	6	8.87	3.26-19.31
	20-24	25	11.36	7.35-16.76
	25-29	38	10.50	7.43-14.41
	30-34	59	12.28	9.35-15.84
	35+	50	15.33	11.38-20.21

Table 6. Prevalence of Birth Defects by Maternal Age Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Maternal Age**	**Count**	**Rate[1]**	**95% CI**
VSD, Conoventricular/Malalignment	<20	0	-	-
	20-24	1	0.45	0.01-2.53
	25-29	6	1.66	0.61-3.61
	30-34	10	2.08	1.00-3.83
	35+	6	1.84	0.67-4.00
Single Ventricle and Levo-Transposition of the Great Arteries				
levo-Transposition of the Great Arteries	<20	1	1.48	0.04-8.24
	20-24	0	-	-
	25-29	0	-	-
	30-34	2	0.42	0.05-1.50
	35+	3	0.92	0.19-2.69
Single Ventricle	<20	0	-	-
	20-24	0	-	-
	25-29	0	-	-
	30-34	3	0.62	0.13-1.83
	35+	0	-	-
Respiratory				
Choanal Atresia	<20	0	-	-
	20-24	1	0.45	0.01-2.53
	25-29	2	0.55	0.07-2.00
	30-34	3	0.62	0.13-1.83
	35+	6	1.84	0.67-4.00
Lung Anomalies	<20	3	4.44	0.91-12.96
	20-24	5	2.27	0.74-5.30
	25-29	12	3.32	1.71-5.79
	30-34	12	2.50	1.29-4.36
	35+	7	2.15	0.86-4.42

Table 6. Prevalence of Birth Defects by Maternal Age Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Maternal Age**	**Count**	**Rate[1]**	**95% CI**
Orofacial				
Cleft Lip with and without Cleft Palate	<20	8	11.83	5.11-23.30
	20-24	14	6.36	3.48-10.67
	25-29	27	7.46	4.92-10.86
	30-34	31	6.45	4.38-9.16
	35+	16	4.90	2.80-7.96
Cleft Palate without Cleft Lip	<20	3	4.44	0.91-12.96
	20-24	13	5.91	3.14-10.10
	25-29	22	6.08	3.81-9.21
	30-34	26	5.41	3.54-7.93
	35+	21	6.44	3.98-9.84
Pierre Robin Sequence	<20	3	4.44	0.91-12.96
	20-24	3	1.36	0.28-3.98
	25-29	9	2.49	1.14-4.72
	30-34	9	1.87	0.86-3.56
	35+	12	3.68	1.90-6.43
Gastrointestinal				
Biliary Atresia	<20	1	1.48	0.04-8.24
	20-24	0	-	-
	25-29	3	0.83	0.17-2.42
	30-34	2	0.42	0.05-1.50
	35+	8	2.45	1.06-4.83
Esophageal Atresia/Tracheoesophageal Fistula	<20	0	-	-
	20-24	7	3.18	1.28-6.55
	25-29	9	2.49	1.14-4.72
	30-34	15	3.12	1.75-5.15
	35+	18	5.52	3.27-8.72
Hirschsprung Disease	<20	0	-	-
	20-24	3	1.36	0.28-3.98
	25-29	4	1.11	0.30-2.83
	30-34	11	2.29	1.14-4.10
	35+	5	1.53	0.50-3.58

Table 6. Prevalence of Birth Defects by Maternal Age Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Maternal Age**	**Count**	**Rate[1]**	**95% CI**
Rectal and Large Intestinal Atresia/Stenosis	<20	3	4.44	0.91-12.96
	20-24	8	3.63	1.57-7.16
	25-29	12	3.32	1.71-5.79
	30-34	15	3.12	1.75-5.15
	35+	17	5.21	3.04-8.34
Small Intestinal Atresia	<20	0	-	-
	20-24	3	1.36	0.28-3.98
	25-29	10	2.76	1.33-5.08
	30-34	10	2.08	1.00-3.83
	35+	10	3.07	1.47-5.64
Genitourinary				
Bladder Exstrophy	<20	0	-	-
	20-24	0	-	-
	25-29	2	0.55	0.07-2.00
	30-34	1	0.21	0.01-1.16
	35+	0	-	-
Cloacal Exstrophy	<20	1	1.48	0.04-8.24
	20-24	0	-	-
	25-29	1	0.28	0.01-1.54
	30-34	4	0.83	0.23-2.13
	35+	1	0.31	0.01-1.71
Hypospadias, 2nd or 3rd Degree[2]	<20	7	20.21	8.13-41.65
	20-24	16	14.26	8.15-23.16
	25-29	38	20.47	14.48-28.09
	30-34	59	23.95	18.23-30.89
	35+	43	25.73	18.62-34.66
Obstructive Genitourinary Defect	<20	11	16.26	8.12-29.10
	20-24	28	12.72	8.45-18.38
	25-29	42	11.61	8.37-15.69
	30-34	73	15.20	11.91-19.11
	35+	59	18.09	13.77-23.33

Table 6. Prevalence of Birth Defects by Maternal Age Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Maternal Age**	**Count**	**Rate[1]**	**95% CI**
Renal Agenesis/Hypoplasia	<20	1	1.48	0.04-8.24
	20-24	1	0.45	0.01-2.53
	25-29	1	0.28	0.01-1.54
	30-34	2	0.42	0.05-1.50
	35+	0	-	-
Musculoskeletal				
Club Foot	<20	14	20.70	11.32-34.73
	20-24	35	15.90	11.07-22.11
	25-29	51	14.09	10.49-18.53
	30-34	60	12.49	9.53-16.08
	35+	57	17.47	13.23-22.64
Craniosynostosis	<20	1	1.48	0.04-8.24
	20-24	5	2.27	0.74-5.30
	25-29	14	3.87	2.12-6.49
	30-34	32	6.66	4.56-9.40
	35+	15	4.60	2.57-7.58
Diaphragmatic Hernia	<20	1	1.48	0.04-8.24
	20-24	5	2.27	0.74-5.30
	25-29	5	1.38	0.45-3.22
	30-34	14	2.91	1.59-4.89
	35+	9	2.76	1.26-5.24
Gastroschisis	<20	9	13.31	6.08-25.26
	20-24	22	9.99	6.26-15.13
	25-29	10	2.76	1.33-5.08
	30-34	0	-	-
	35+	3	0.92	0.19-2.69
Omphalocele	<20	1	1.48	0.04-8.24
	20-24	2	0.91	0.11-3.28
	25-29	4	1.11	0.30-2.83
	30-34	7	1.46	0.59-3.00
	35+	14	4.29	2.35-7.20

Table 6. Prevalence of Birth Defects by Maternal Age Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Maternal Age**	**Count**	**Rate[1]**	**95% CI**
Polydactyly/Syndactyly	<20	17	25.13	14.64-40.24
	20-24	39	17.72	12.60-24.22
	25-29	66	18.24	14.11-23.21
	30-34	66	13.74	10.63-17.48
	35+	56	17.17	12.97-22.29
Reduction Deformity, Lower Limbs	<20	3	4.44	0.91-12.96
	20-24	3	1.36	0.28-3.98
	25-29	9	2.49	1.14-4.72
	30-34	5	1.04	0.34-2.43
	35+	3	0.92	0.19-2.69
Reduction Deformity, Upper Limbs	<20	3	4.44	0.91-12.96
	20-24	6	2.73	1.00-5.93
	25-29	10	2.76	1.33-5.08
	30-34	16	3.33	1.90-5.41
	35+	8	2.45	1.06-4.83
Skeletal Dysplasia	<20	2	2.96	0.36-10.68
	20-24	4	1.82	0.50-4.65
	25-29	8	2.21	0.95-4.36
	30-34	10	2.08	1.00-3.83
	35+	6	1.84	0.67-4.00
Chromosomal and other Syndromes				
Klinefelter Syndrome	<20	0	-	-
	20-24	1	0.45	0.01-2.53
	25-29	0	-	-
	30-34	2	0.42	0.05-1.50
	35+	4	1.23	0.33-3.14
Trisomy 13	<20	0	-	-
	20-24	1	0.45	0.01-2.53
	25-29	4	1.11	0.30-2.83
	30-34	0	-	-
	35+	1	0.31	0.01-1.71

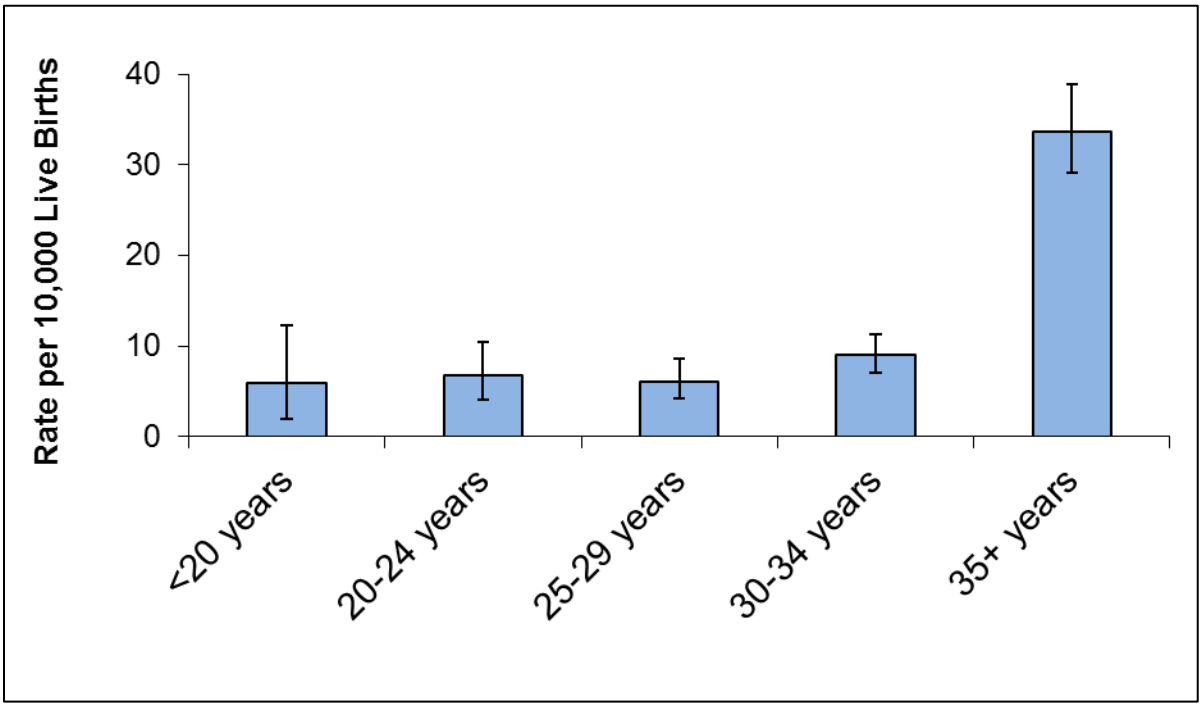
Table 6. Prevalence of Birth Defects by Maternal Age Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Maternal Age**	**Count**	**Rate[1]**	**95% CI**
Trisomy 18	<20	1	1.48	0.04-8.24
	20-24	3	1.36	0.28-3.98
	25-29	2	0.55	0.07-2.00
	30-34	5	1.04	0.34-2.43
	35+	25	7.66	4.96-11.31
Trisomy 21 (Down Syndrome)	<20	4	5.91	1.61-15.14
	20-24	15	6.81	3.81-11.24
	25-29	22	6.08	3.81-9.21
	30-34	43	8.95	6.48-12.06
	35+	110	33.72	27.71-40.64
Turner Syndrome[3]	<20	0	-	-
	20-24	5	4.63	1.50-10.81
	25-29	1	0.57	0.01-3.16
	30-34	2	0.85	0.10-3.09
	35+	2	1.26	0.15-4.54
Other				
Amniotic Bands	<20	2	2.96	0.36-10.68
	20-24	1	0.45	0.01-2.53
	25-29	7	1.93	0.78-3.99
	30-34	5	1.04	0.34-2.43
	35+	4	1.23	0.33-3.14
Skin Anomalies	<20	2	2.96	0.36-10.68
	20-24	2	0.91	0.11-3.28
	25-29	7	1.93	0.78-3.99
	30-34	6	1.25	0.46-2.72
	35+	7	2.15	0.86-4.42

[1] Rate per 10,000 live births.
[2] Rate calculated using male live births.
[3] Rate calculated using female live births.

Down Syndrome

There is a strong association between Down syndrome and advanced maternal age (Figure 7). In Massachusetts, the rate of Down syndrome in mothers 35 and older is 33.7 per 10,000 live births, which is more than 4 times greater than the rate in mothers younger than 35. This reflects the pattern of higher chromosomal defect rates in general among older women.

Figure 7. Prevalence of Down Syndrome by Maternal Age, Massachusetts: 2011-2012

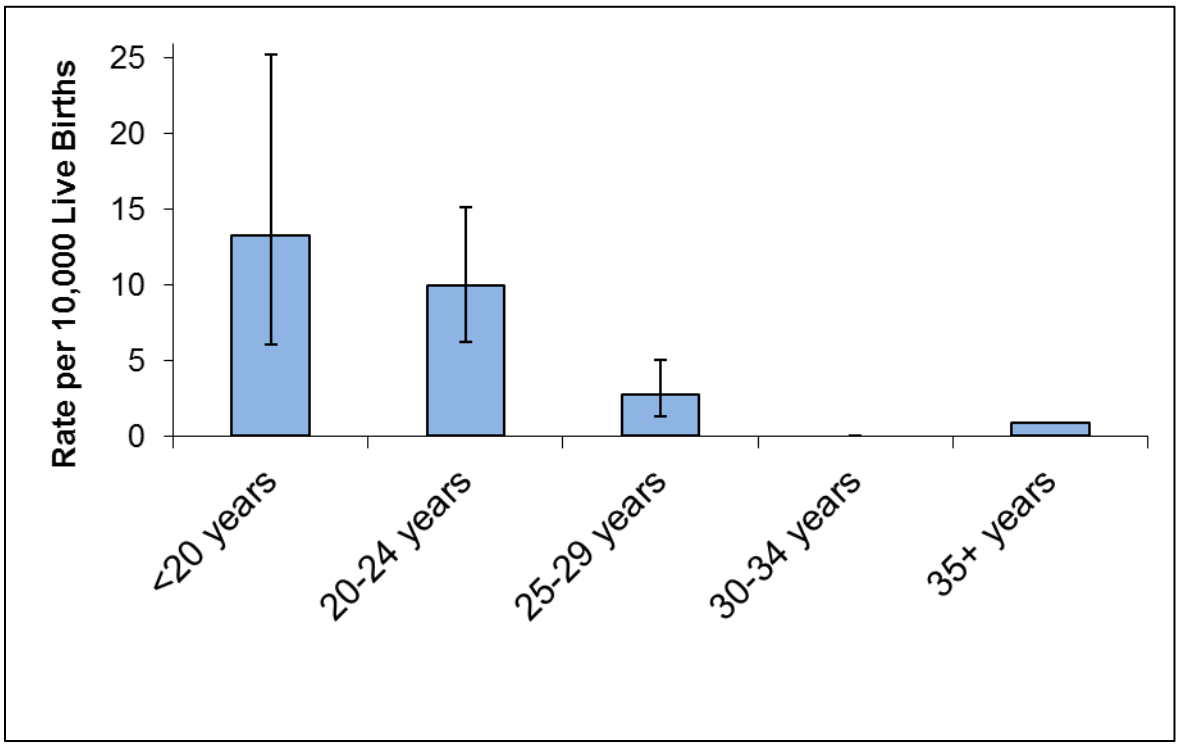


Live births and stillbirths, N=194.
Error bars represent 95% confidence intervals.

Gastroschisis

Mothers ages 19 and under had the highest rate of gastroschisis cases at 13.3 per 10,000 live births (Figure 8). The association between gastroschisis and younger maternal age has been shown in previous studies (18).

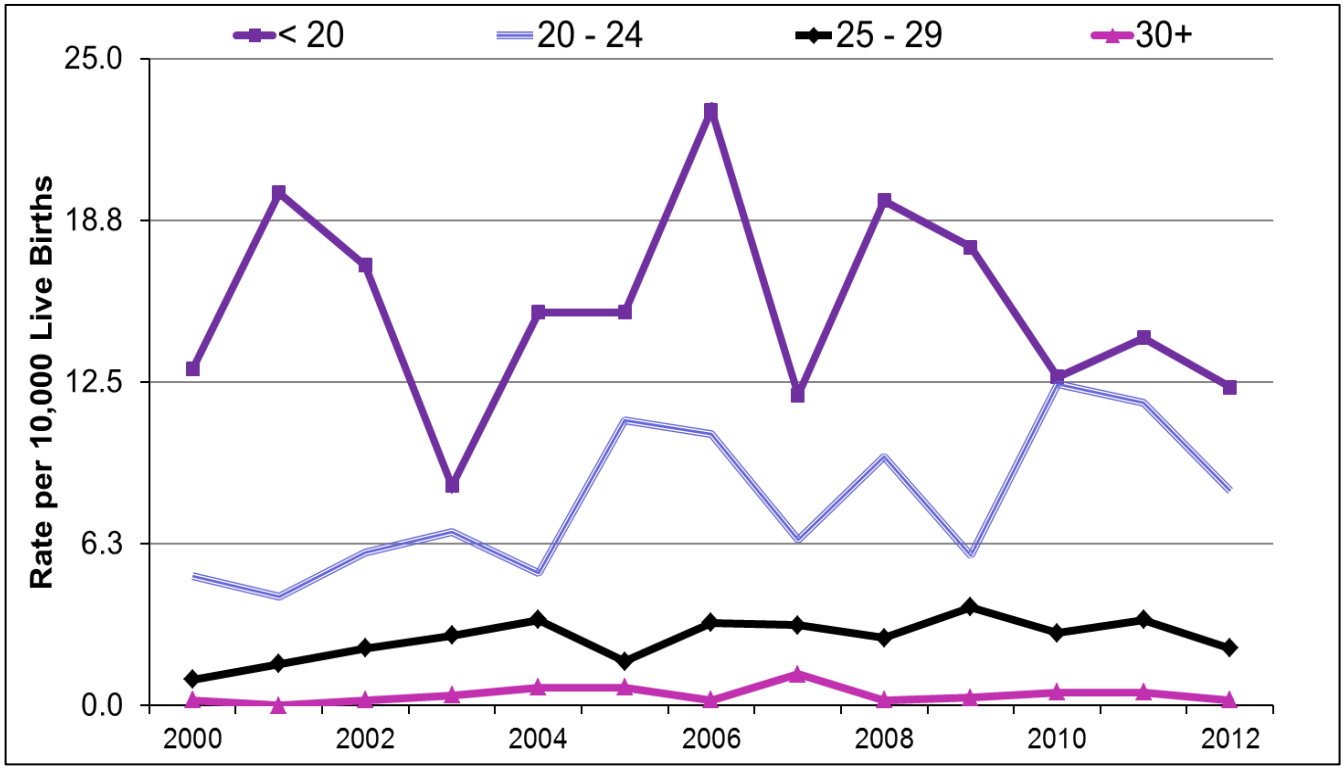
Figure 8. Prevalence of Gastroschisis by Maternal Age, Massachusetts: 2011-2012



Live births and stillbirths, N=44.
Error bars represent 95% confidence intervals.

Figure 9 presents gastroschisis rates by maternal age in Massachusetts over a 12-year time span.

Figure 9. Prevalence of Gastroschisis by Year and Maternal Age, Massachusetts: 2000-2012



Live births and stillbirths.

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 7 shows the variation in age-adjusted birth defect rates by racial/ethnic group in Massachusetts during the current reporting period.

Table 7. Age-Adjusted Prevalence of Birth Defects by Maternal Race/Ethnicity, Massachusetts: 2011-2012

Maternal Race	Cases	Age-Adjusted Rate[1]	95% CI
White, Non-Hispanic	1690	185.5	176.6-194.3
Black, Non-Hispanic	285	207.8	183.7-232.0
Asian, Non-Hispanic	190	154.8	132.8-176.8
Hispanic	529	219.8	201.1-238.6
Other, Non-Hispanic[2]	30	213.6	137.2-290.0

Live births and stillbirths, N=2724.
[1] Rate per 10,000 live births, adjusted to statewide maternal age distribution of the birthing population.
[2] Includes American Indian.

The age-adjusted birth defect rate in offspring of Hispanic mothers is significantly higher than that of white and Asian mothers. In addition, the age-adjusted birth defect rate among those born to black mothers is significantly higher than the rate among those born to Asian mothers. Possible explanations include genetic variation, diet and lifestyle differences, and varying access to prenatal screening and health care services. For example, data from the 2011 Massachusetts Pregnancy Risk Assessment Monitoring System shows that Hispanic women are less likely to use a multivitamin in the month prior to pregnancy (19). These results are shown in Appendix 7. Table 8 shows the prevalence rates of the individual birth defects by maternal race/ethnicity.

Table 8. Prevalence of Birth Defects by Maternal Race/Ethnicity, Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Maternal Race/Ethnicity[1]**	**Count**	**Rate[2]**	**95% CI**
Central Nervous System				
Anencephaly	White	4	0.44	0.12-1.13
	Black	0	-	-
	Asian	0	-	-
	Hispanic	4	1.55	0.42-3.96
Encephalocele	White	2	0.22	0.03-0.79
	Black	0	-	-
	Asian	0	-	-
	Hispanic	4	1.55	0.42-3.96
Holoprosencephaly	White	4	0.44	0.12-1.13
	Black	3	2.16	0.45-6.31
	Asian	0	-	-
	Hispanic	3	1.16	0.24-3.39
Hydrocephaly without Spina Bifida	White	28	3.08	2.04-4.45
	Black	3	2.16	0.45-6.31
	Asian	5	3.98	1.29-9.30
	Hispanic	13	5.03	2.68-8.59
Microcephaly	White	19	2.09	1.26-3.26
	Black	2	1.44	0.17-5.20
	Asian	3	2.39	0.49-6.98
	Hispanic	4	1.55	0.42-3.96
Spina with/without Hydrocephaly	White	18	1.98	1.17-3.13
	Black	2	1.44	0.17-5.20
	Asian	0	-	-
	Hispanic	8	3.09	1.34-6.09
Spinal Cord	White	46	5.05	3.70-6.74
	Black	4	2.88	0.78-7.37
	Asian	5	3.98	1.29-9.30
	Hispanic	14	5.41	2.96-9.08
Eye				
Aniridia	White	3	0.33	0.07-0.96
	Black	1	0.72	0.02-4.01
	Asian	0	-	-
	Hispanic	0	-	-

Table 8. Prevalence of Birth Defects by Maternal Race/Ethnicity, Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Maternal Race/Ethnicity[1]**	**Count**	**Rate[2]**	**95% CI**
Anophthalmia/Microphthalmia	White	7	0.77	0.31-1.58
	Black	2	1.44	0.17-5.20
	Asian	0	-	-
	Hispanic	5	1.93	0.63-4.51
Congenital Glaucoma/Congenital Cataract	White	28	3.08	2.04-4.45
	Black	3	2.16	0.45-6.31
	Asian	3	2.39	0.49-6.98
	Hispanic	11	4.25	2.12-7.61
Ear				
Anotia/Microtia	White	23	2.53	1.60-3.79
	Black	2	1.44	0.17-5.20
	Asian	4	3.19	0.87-8.16
	Hispanic	7	2.71	1.09-5.58
Cardiovascular				
Anomalous Pulmonary Venous Connection				
Total Anomalous Pulmonary Venous Connection	White	6	0.66	0.24-1.44
	Black	5	3.60	1.17-8.40
	Asian	3	2.39	0.49-6.98
	Hispanic	7	2.71	1.09-5.58
Atrioventricular Canal Defects				
Atrial Septal Defect (ASD) Primum	White	1	0.11	0.00-0.61
	Black	0	-	-
	Asian	0	-	-
	Hispanic	0	-	-
Common Atrium	White	1	0.11	0.00-0.61
	Black	2	1.44	0.17-5.20
	Asian	0	-	-
	Hispanic	2	0.77	0.09-2.79
Complete Atrioventricular Canal Defect	White	34	3.74	2.59-5.22
	Black	9	6.48	2.96-12.30
	Asian	4	3.19	0.87-8.16
	Hispanic	14	5.41	2.96-9.08

Table 8. Prevalence of Birth Defects by Maternal Race/Ethnicity, Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Maternal Race/Ethnicity[1]**	**Count**	**Rate[2]**	**95% CI**
Endocardial Cushion Defect, Other specified (OS) and Not otherwise specified (NOS)	White	9	0.99	0.45-1.88
	Black	3	2.16	0.45-6.31
	Asian	1	0.80	0.02-4.44
	Hispanic	7	2.71	1.09-5.58
Ventricular Septal Defect (VSD), Canal Type	White	5	0.55	0.18-1.28
	Black	0	-	-
	Asian	0	-	-
	Hispanic	4	1.55	0.42-3.96
Conotruncal (Outlet) and Aortic Arch				
Double Outlet Right Ventricle	White	8	0.88	0.38-1.73
	Black	2	1.44	0.17-5.20
	Asian	2	1.59	0.19-5.76
	Hispanic	4	1.55	0.42-3.96
Interrupted Aortic Arch, Type B	.	.	.	.-.
Tetralogy of Fallot with/without Pulmonary Atresia	White	35	3.85	2.68-5.35
	Black	4	2.88	0.78-7.37
	Asian	2	1.59	0.19-5.76
	Hispanic	8	3.09	1.34-6.09
Truncus	White	2	0.22	0.03-0.79
	Black	1	0.72	0.02-4.01
	Asian	0	-	-
	Hispanic	2	0.77	0.09-2.79
dextro-Transposition of the Great Arteries	White	17	1.87	1.09-2.99
	Black	4	2.88	0.78-7.37
	Asian	4	3.19	0.87-8.16
	Hispanic	9	3.48	1.59-6.61
Ebstein Anomaly				
Ebstein Anomaly	White	3	0.33	0.07-0.96
	Black	0	-	-
	Asian	0	-	-
	Hispanic	2	0.77	0.09-2.79

Table 8. Prevalence of Birth Defects by Maternal Race/Ethnicity, Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Maternal Race/Ethnicity[1]**	**Count**	**Rate[2]**	**95% CI**
Heterotaxy (Laterality Defects)				
Heterotaxy	White	4	0.44	0.12-1.13
	Black	1	0.72	0.02-4.01
	Asian	0	-	-
	Hispanic	3	1.16	0.24-3.39
Left-Sided Obstruction				
Aortic Valve Stenosis	White	11	1.21	0.60-2.16
	Black	0	-	-
	Asian	2	1.59	0.19-5.76
	Hispanic	4	1.55	0.42-3.96
Aortic arch Atresia	White	0	-	-
	Black	0	-	-
	Asian	1	0.80	0.02-4.44
	Hispanic	0	-	-
Coarctation of Aorta	White	47	5.16	3.79-6.87
	Black	8	5.76	2.49-11.35
	Asian	2	1.59	0.19-5.76
	Hispanic	11	4.25	2.12-7.61
Hypoplastic Left Heart Syndrome	White	14	1.54	0.84-2.58
	Black	1	0.72	0.02-4.01
	Asian	2	1.59	0.19-5.76
	Hispanic	5	1.93	0.63-4.51
Interrupted Aortic Arch (Type A and NOS)	White	4	0.44	0.12-1.13
	Black	0	-	-
	Asian	0	-	-
	Hispanic	0	-	-
Right-Sided Obstruction				
Pulmonary Stenosis, Valvular	White	70	7.69	6.00-9.72
	Black	23	16.56	10.50-24.84
	Asian	6	4.78	1.75-10.40
	Hispanic	17	6.57	3.83-10.52

Table 8. Prevalence of Birth Defects by Maternal Race/Ethnicity, Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Maternal Race/Ethnicity[1]**	**Count**	**Rate[2]**	**95% CI**
Pulmonary Valve Atresia with intact septum	White	5	0.55	0.18-1.28
	Black	0	-	-
	Asian	0	-	-
	Hispanic	1	0.39	0.01-2.15
Pulmonary Valve Atresia with VSD	White	0	0.00	0.00-0.41
	Black	1	0.72	0.02-4.01
	Asian	0	-	-
	Hispanic	0	-	-
Tricuspid Valve Atresia	White	2	0.22	0.03-0.79
	Black	0	-	-
	Asian	0	-	-
	Hispanic	1	0.39	0.01-2.15
Septal Defects				
ASD (Secundum and NOS)	White	201	22.09	19.14-25.36
	Black	37	26.64	18.75-36.71
	Asian	26	20.71	13.53-30.35
	Hispanic	63	24.36	18.72-31.16
VSD (Membranous and NOS)	White	103	11.32	9.24-13.73
	Black	17	12.24	7.13-19.59
	Asian	19	15.14	9.11-23.64
	Hispanic	39	15.08	10.72-20.61
VSD, Conoventricular/Malalignment	White	9	0.99	0.45-1.88
	Black	3	2.16	0.45-6.31
	Asian	5	3.98	1.29-9.30
	Hispanic	6	2.32	0.85-5.05
Single Ventricle and levo-Transposition of the Great Arteries				
levo-Transposition of the Great Arteries	White	6	0.66	0.24-1.44
	Black	0	-	-
	Asian	0	-	-
	Hispanic	0	-	-

Table 8. Prevalence of Birth Defects by Maternal Race/Ethnicity, Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Maternal Race/Ethnicity[1]**	**Count**	**Rate[2]**	**95% CI**
Single Ventricle	White	2	0.22	0.03-0.79
	Black	1	0.72	0.02-4.01
	Asian	0	-	-
	Hispanic	0	-	-
Respiratory				
Choanal Atresia	White	10	1.10	0.53-2.02
	Black	0	-	-
	Asian	0	-	-
	Hispanic	2	0.77	0.09-2.79
Lung Anomalies	White	19	2.09	1.26-3.26
	Black	5	3.60	1.17-8.40
	Asian	3	2.39	0.49-6.98
	Hispanic	11	4.25	2.12-7.61
Orofacial				
Cleft Lip with and without Cleft Palate	White	58	6.37	4.84-8.24
	Black	7	5.04	2.03-10.38
	Asian	7	5.58	2.24-11.49
	Hispanic	22	8.51	5.33-12.88
Cleft Palate without Cleft Lip	White	49	5.38	3.98-7.12
	Black	14	10.08	5.51-16.91
	Asian	7	5.58	2.24-11.49
	Hispanic	14	5.41	2.96-9.08
Pierre Robin Sequence	White	22	2.42	1.51-3.66
	Black	3	2.16	0.45-6.31
	Asian	4	3.19	0.87-8.16
	Hispanic	7	2.71	1.09-5.58
Gastrointestinal				
Biliary Atresia	White	6	0.66	0.24-1.44
	Black	2	1.44	0.17-5.20
	Asian	3	2.39	0.49-6.98
	Hispanic	3	1.16	0.24-3.39

Table 8. Prevalence of Birth Defects by Maternal Race/Ethnicity, Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Maternal Race/Ethnicity[1]**	**Count**	**Rate[2]**	**95% CI**
Esophageal Atresia/Tracheoesophageal Fistula	White	33	3.63	2.50-5.09
	Black	4	2.88	0.78-7.37
	Asian	0	-	-
	Hispanic	12	4.64	2.40-8.10
Hirschsprung Disease	White	12	1.32	0.68-2.30
	Black	3	2.16	0.45-6.31
	Asian	5	3.98	1.29-9.30
	Hispanic	3	1.16	0.24-3.39
Rectal and Large Intestinal Atresia/Stenosis	White	38	4.18	2.95-5.73
	Black	2	1.44	0.17-5.20
	Asian	5	3.98	1.29-9.30
	Hispanic	8	3.09	1.34-6.09
Small Intestinal Atresia	White	21	2.31	1.43-3.53
	Black	4	2.88	0.78-7.37
	Asian	1	0.80	0.02-4.44
	Hispanic	7	2.71	1.09-5.58
Bladder Exstrophy	White	3	0.33	0.07-0.96
	Black	0	-	-
	Asian	0	-	-
	Hispanic	0	-	-
Cloacal Exstrophy	White	5	0.55	0.18-1.28
	Black	0	-	-
	Asian	0	-	-
	Hispanic	2	0.77	0.09-2.79
Hypospadias, 2nd or 3rd Degree[3]	White	117	25.11	20.76-30.09
	Black	17	23.83	7.13-19.59
	Asian	9	13.96	6.38-26.50
	Hispanic	14	10.57	5.78-17.74
Obstructive Genitourinary Defect	White	131	14.39	12.04-17.08
	Black	15	10.80	6.04-17.81
	Asian	13	10.36	5.51-17.71
	Hispanic	51	19.72	14.68-25.93

Table 8. Prevalence of Birth Defects by Maternal Race/Ethnicity, Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Maternal Race/Ethnicity[1]**	**Count**	**Rate[2]**	**95% CI**
Renal Agenesis/Hypoplasia	White	4	0.44	0.12-1.13
	Black	0	-	-
	Asian	0	-	-
	Hispanic	1	0.39	0.01-2.15
Musculoskeletal				
Club Foot	White	145	15.93	13.45-18.75
	Black	17	12.24	7.13-19.59
	Asian	11	8.76	4.37-15.68
	Hispanic	40	15.46	11.05-21.06
Craniosynostosis	White	47	5.16	3.79-6.87
	Black	2	1.44	0.17-5.20
	Asian	4	3.19	0.87-8.16
	Hispanic	12	4.64	2.40-8.10
Diaphragmatic Hernia	White	24	2.64	1.69-3.92
	Black	1	0.72	0.02-4.01
	Asian	2	1.59	0.19-5.76
	Hispanic	7	2.71	1.09-5.58
Gastroschisis	White	25	2.75	1.78-4.06
	Black	2	1.44	0.17-5.20
	Asian	4	3.19	0.87-8.16
	Hispanic	12	4.64	2.40-8.10
Omphalocele	White	20	2.20	1.34-3.39
	Black	0	-	-
	Asian	2	1.59	0.19-5.76
	Hispanic	6	2.32	0.85-5.05
Polydactyly/Syndactyly	White	127	13.96	11.63-16.60
	Black	47	33.83	24.86-44.99
	Asian	17	13.54	7.89-21.68
	Hispanic	51	19.72	14.68-25.93
Reduction Deformity, Lower Limbs	White	16	1.76	1.00-2.86
	Black	5	3.60	1.17-8.40
	Asian	1	0.80	0.02-4.44
	Hispanic	0	-	-

Table 8. Prevalence of Birth Defects by Maternal Race/Ethnicity, Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Maternal Race/Ethnicity[1]**	**Count**	**Rate[2]**	**95% CI**
Reduction Deformity, Upper Limbs	White	35	3.85	2.68-5.35
	Black	1	0.72	0.02-4.01
	Asian	1	0.80	0.02-4.44
	Hispanic	6	2.32	0.85-5.05
Skeletal Dysplasia	White	21	2.31	1.43-3.53
	Black	2	1.44	0.17-5.20
	Asian	1	0.80	0.02-4.44
	Hispanic	6	2.32	0.85-5.05
Chromosomal and other Syndromes				
Klinefelter Syndrome	White	3	0.33	0.07-0.96
	Black	0	-	-
	Asian	2	1.59	0.19-5.76
	Hispanic	2	0.77	0.09-2.79
Trisomy 13	White	4	0.44	0.12-1.13
	Black	1	0.72	0.02-4.01
	Asian	0	-	-
	Hispanic	1	0.39	0.01-2.15
Trisomy 18	White	20	2.20	1.34-3.39
	Black	5	3.60	1.17-8.40
	Asian	3	2.39	0.49-6.98
	Hispanic	8	3.09	1.34-6.09
Trisomy 21 (Down Syndrome)	White	118	12.97	10.73-15.53
	Black	19	13.68	8.23-21.36
	Asian	9	7.17	3.28-13.61
	Hispanic	47	18.17	13.35-24.16
Turner Syndrome[4]	White	8	1.80	0.78-3.55
	Black	0	-	-
	Asian	1	1.64	0.04-9.12
	Hispanic	0	-	-

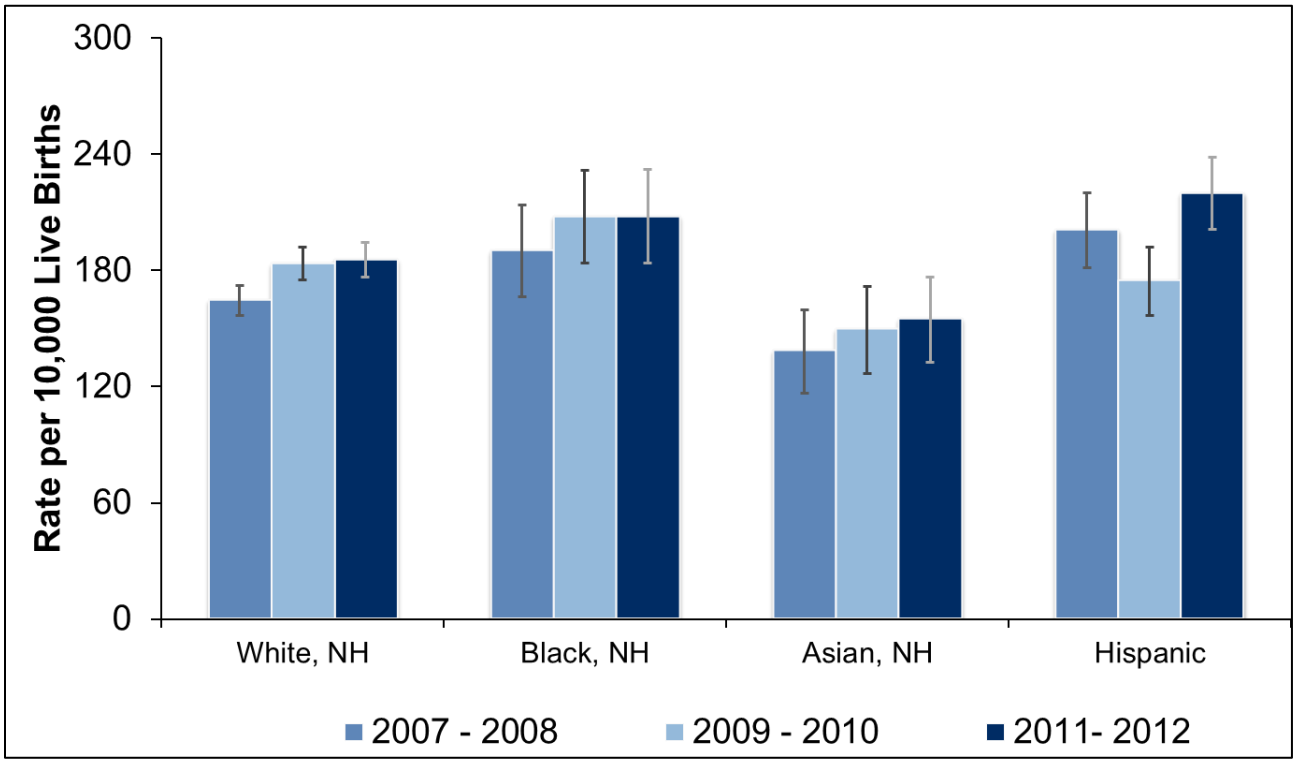
Table 8. Prevalence of Birth Defects by Maternal Race/Ethnicity, Live Births and Stillbirths, Massachusetts: 2011-2012				
Defect	**Maternal Race/Ethnicity[1]**	**Count**	**Rate[2]**	**95% CI**
Other				
Amniotic Bands	White	14	1.54	0.84-2.58
	Black	1	0.72	0.02-4.01
	Asian	0	-	-
	Hispanic	4	1.55	0.42-3.96
Skin Anomalies	White	13	1.43	0.76-2.44
	Black	2	1.44	0.17-5.20
	Asian	3	2.39	0.49-6.98
	Hispanic	5	1.93	0.63-4.51

[1] Race/ethnic groups used: White, Non-Hispanic; Black, Non-Hispanic; Asian, Non-Hispanic; Hispanic. Other, Non-Hispanic not presented due to small numbers.
[2] Rate per 10,000 live births.
[3] Rate calculated using male live births.
[4] Rate calculated using female live births.

Trends in Maternal Race/Ethnicity

Figure 10 shows the age-adjusted birth defect rates by race/ethnicity between 2007 and 2012 in two-year intervals. Hispanic rates for 2011-2012 are significantly higher than 2009-2010 rates, but are similar to 2007-2008 rates.

Figure 10. Prevalence of Birth Defects by Maternal Race/Ethnicity, Massachusetts: 2007-2012



Live births and stillbirths. NH=Non-Hispanic.
Adjusted to statewide maternal age distribution of the birthing population in each 2-year period.
Error bars represent 95% confidence intervals.

Maternal Region of Residence

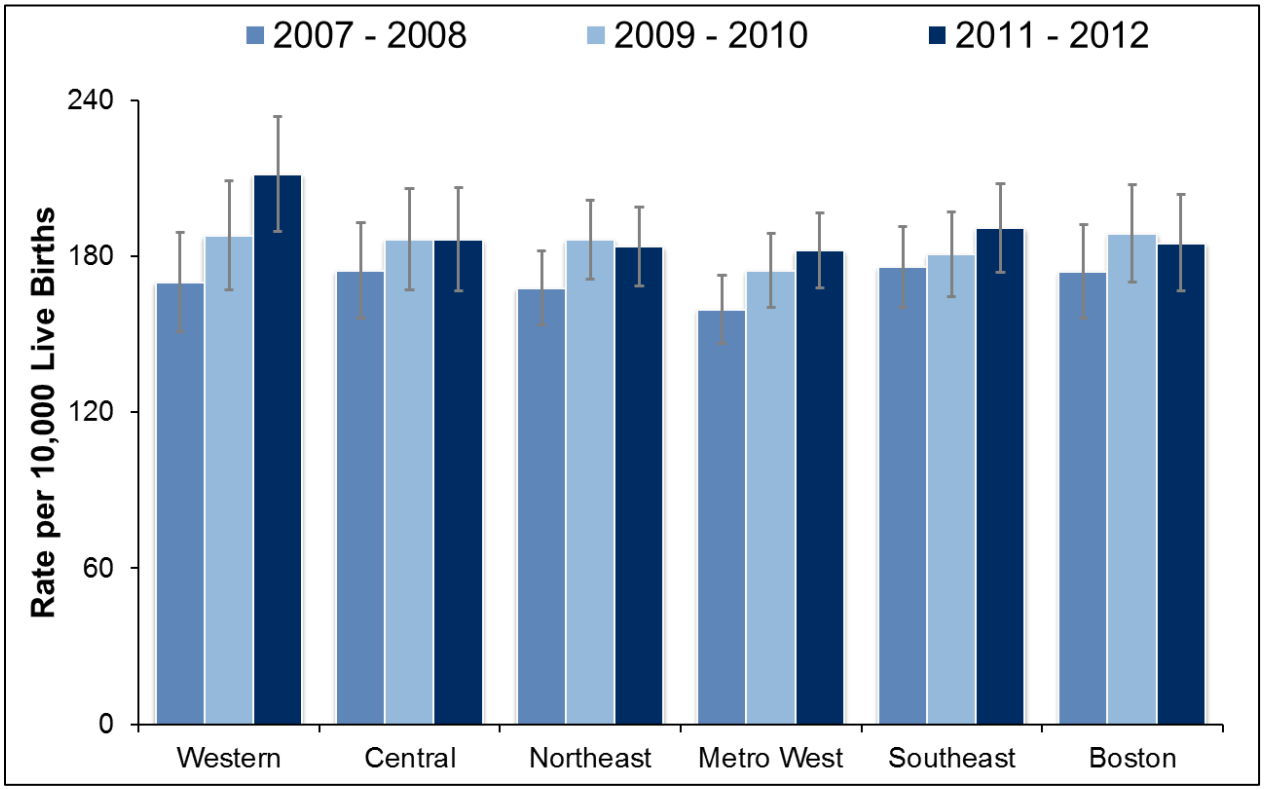
The Massachusetts Executive Office of Health and Human Services divides the state into six regions for statistical, care coordination and administrative purposes. The six regions are based on geographical groupings of cities and towns: Western, Central, Northeast, Metro West, Southeast, and Boston. A map of these regions is provided in Appendix 8. The age-adjusted birth defect rates by the six regions in 2011-2012 are shown in Table 9. Although not significantly different, the rates range from 182.2 per 10,000 in the Metro West region to 211.6 per 10,000 in the Western region. Rates in the Western region have increased over time (Figure 10), but the 2011-2012 rates are not significantly different from 2009-2010 rates. Regional differences may result from variation in the distribution of maternal and other characteristics across the regions.

Table 9. Age-Adjusted Prevalence of Birth Defects by Maternal Residence Region, Massachusetts: 2011-2012

Region	Cases	Age-Adjusted Rate[1]	95% CI
Western	348	211.6	189.4-233.8
Central	341	186.6	166.8-206.4
Northeast	558	183.7	168.5-199.0
Metro West	620	182.2	167.9-196.6
Southeast	482	190.8	173.8-207.9
Boston	382	185.1	166.5-203.6

Live births and stillbirths, N=2731.
[1] Rate per 10,000 live births, adjusted to statewide maternal age distribution of the birthing population.

Figure 11. Prevalence of Birth Defects by Maternal Residence Region, Massachusetts: 2007-2012



Live births and stillbirths.
Adjusted to statewide maternal age distribution of the birthing population in each 2-year period.
Error bars represent 95% confidence intervals.

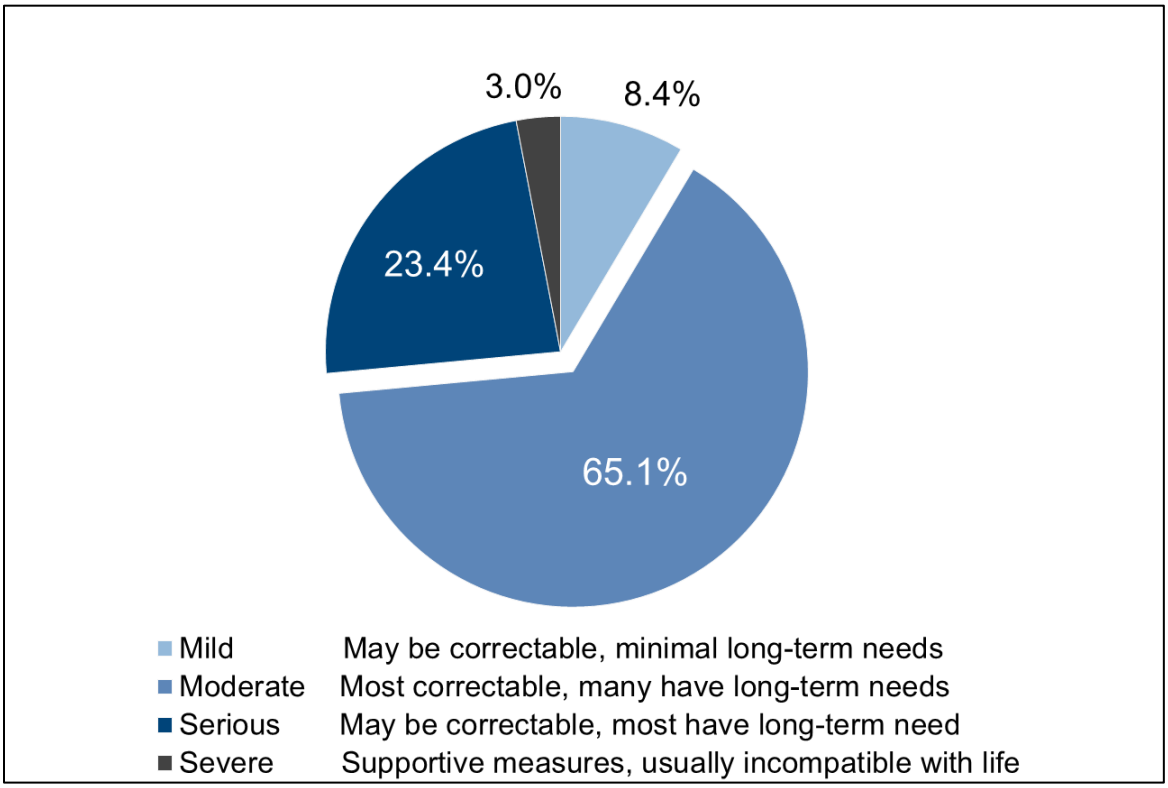
Chapter 7: Birth Defects by Severity, Pattern, and Etiology

Severity

Cases with birth defects ascertained in our surveillance system are categorized by their level of severity. A severity scale was developed by the BDMP in collaboration with Boston University and Massachusetts General Hospital. This scale is based on the usual outcome for a specific birth defect, including its typical compatibility with survival, the need for immediate treatment, the need for long-term care and the amenability of the defect to correction.

Birth defect cases were classified as “severe”, “serious”, “moderate”, or “mild”. Each case is assigned a severity level based on the most severe defect. Examples of birth defects by severity category are shown in Appendix 6. Figure 12 shows birth defects classified by severity.

Figure 12. Birth Defects by Severity, Massachusetts: 2011-2012



Live births and stillbirths, N=2731.
Percentages may not add to 100% due to rounding.

Approximately 8% of cases had defects that are classified as “mild”. These defects may or may not have required corrective treatment. Most defects were classified as moderate, comprising roughly 65% of the total cases. Cases with these types of defects tend to require medical follow up and sometimes require surgery or other treatment. Approximately 23% of

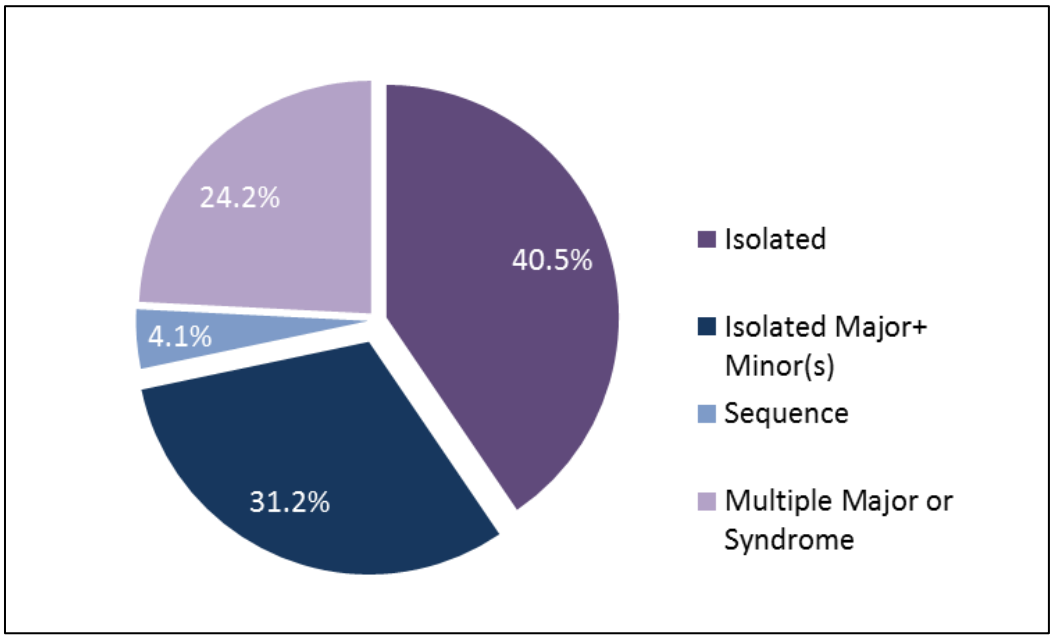
cases were classified as having a “serious” birth defect. Many of these cases need intensive medical care initially, as well as continuing care, and many will have long-term disability.

In 2011-2012, 3% of cases were classified as severe, which is usually incompatible with life. Within the severity categories, there can be considerable variability, particularly with defects classified as moderate.

Pattern

Cases are also classified based on their pattern (i.e. whether a defect occurs with others). Of the 2731 birth defect cases in 2011-2012, 24.2% had multiple major defects or syndromes (Figure 13). Approximately 40% of these cases had single defects, and 31% of these cases had multiple defects within the same organ or a single major defect with one or more minor defects. The remaining 4.1% of defects are part of a sequence of developmental events. Defects that tend to occur as solitary defects include hypospadias, gastroschisis, and craniosynostosis.

Figure 13. Birth Defects by Pattern, Massachusetts: 2011-2012



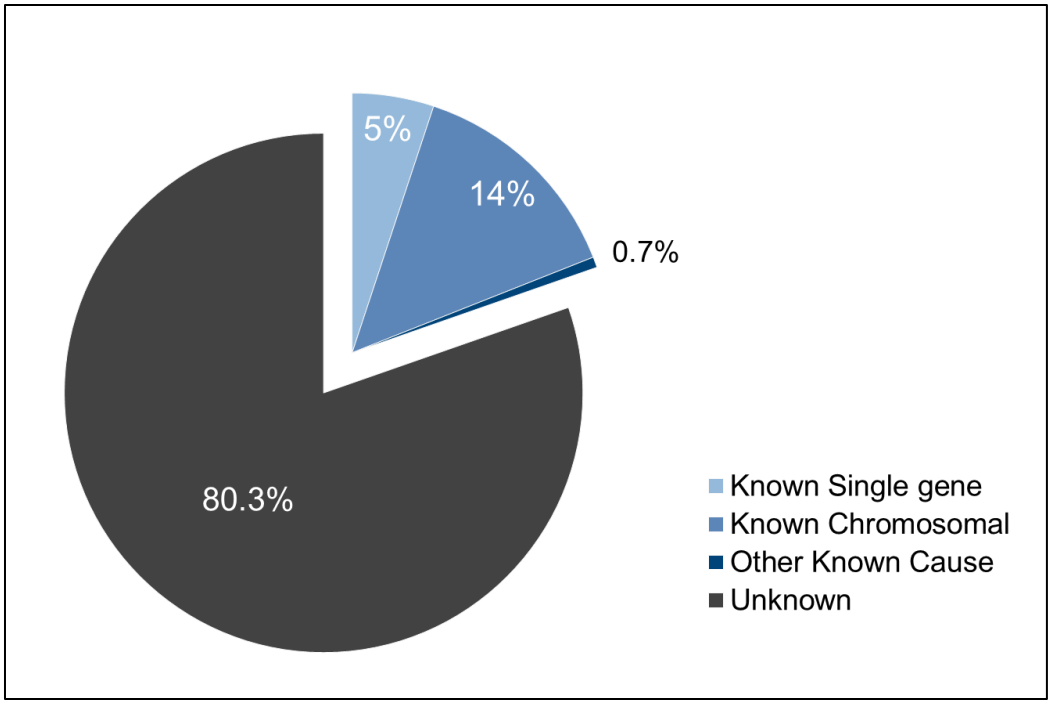
Live births and stillbirths, N=2731.
Percentages may not add to 100% due to rounding.

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 2011-2012 (80.3%) had an unknown etiology (Figure 14). Most of the cases with known etiology were whole chromosome abnormalities (extra copy or missing copy of a chromosome), such us trisomy 13 and Turner syndrome.

Figure 14. Birth Defects by Etiology, Massachusetts: 2011-2012



Live births and stillbirths, N=2731.
Percentages may not add to 100% due to rounding.

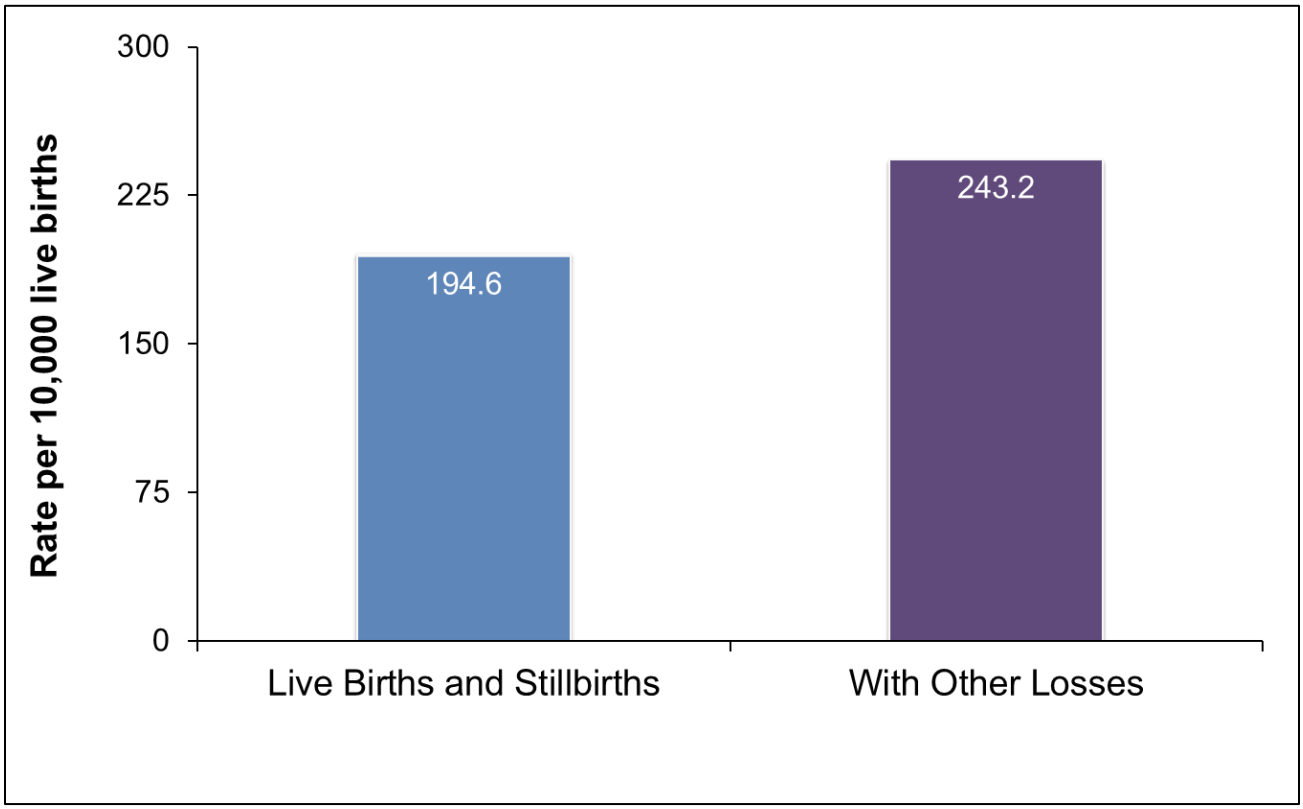
Other examples of defects with known etiologies include single gene defects, such as achondroplasia, Marfan syndrome (deletion 15q21.1), Smith-Lemli-Opitz syndrome and other defects considered to be a Mendelian syndrome. Examples of known etiologies include teratogens such as thalidomide, maternal conditions, such as diabetes, and conditions of the uterine environment, such as didelphys uterus.

Chapter 8: Other Pregnancy Losses—2012 data

In early 2011, the Massachusetts Birth Defects Monitoring Program began collecting information on prenatally diagnosed cases with an outcome other than live birth or stillbirth ("other pregnancy losses"). These include spontaneous early losses <20 weeks gestation and elective terminations. The first complete year of data that includes other pregnancy losses is 2012. The data on other pregnancy losses should be interpreted with caution, as only one year of data from a start-up period is included.

Including other pregnancy losses adds 361 additional cases and increases the overall birth defects prevalence rate in 2012 from 194.6 (95% CI: 184.7-204.9) to 243.2 (95% CI: 232.1-254.7) per 10,000 live births (Figure 15), which is closer to the 1 in 33 (3%) birth defects rate reported nationally (1).

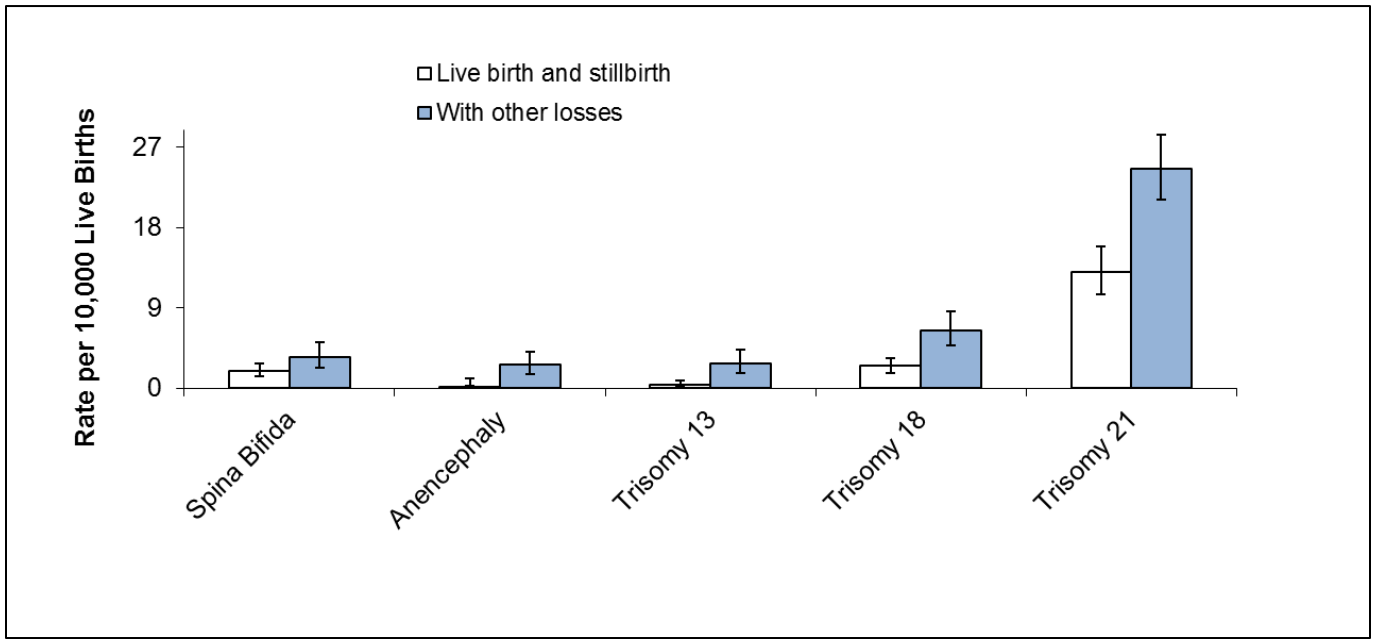
Figure 15. Prevalence of Birth Defects with and without Other Pregnancy Losses, Massachusetts: 2012



N=2731 live births and stillbirths; N=3092 live births, stillbirths and other pregnancy losses.

Figure 16 shows the rates of selected birth defects in Massachusetts in 2012 among live births and stillbirths alone compared to rates that include other pregnancy losses.

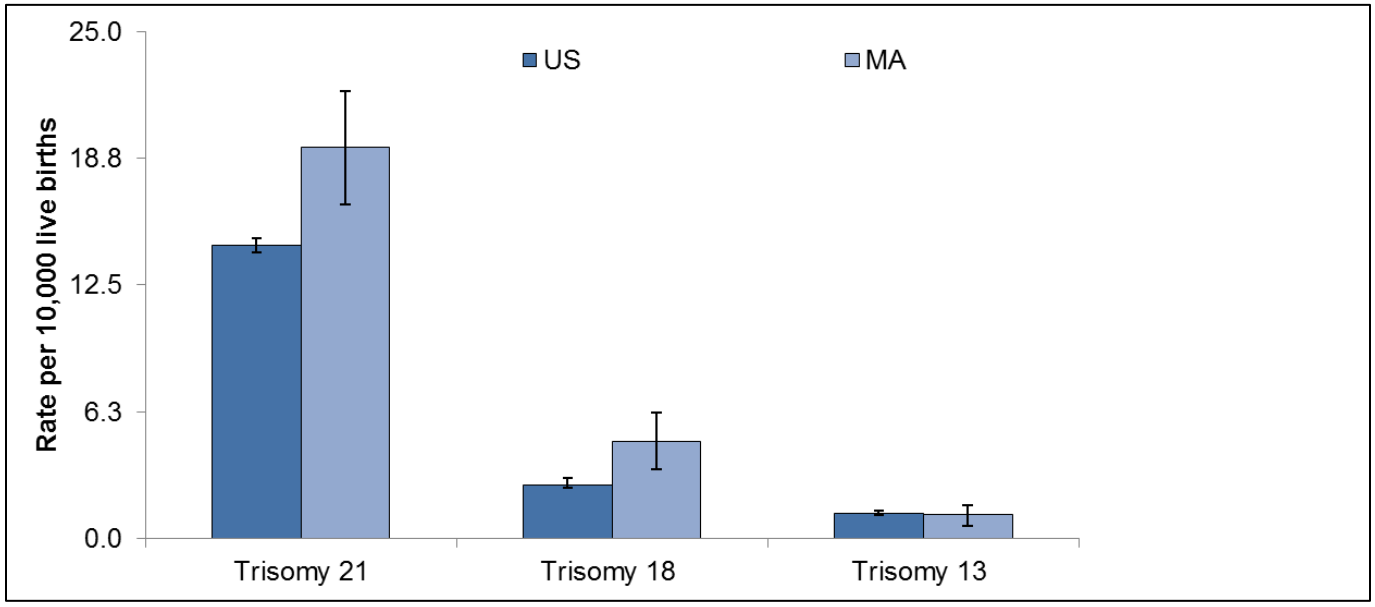
Figure 16. Prevalence of Selected Defects with and without Other Pregnancy Losses, Massachusetts: 2012



Error bars represent 95% confidence intervals.

The rates of trisomy 13, 18, 21, spina bifida and anencephaly are significantly higher when other losses are included, bringing these rates much closer to national estimates and bringing rates of Down Syndrome and trisomy 18 higher than national estimates, possibly due to the inclusion of early spontaneous losses, which most states do not include (Figure 17).

Figure 17. Prevalence of Selected Defects including Other Pregnancy Losses, Massachusetts[1] 2012 Compared to US[2] Rates



[1]Massachusetts rates include live births, stillbirths, and other pregnancy losses. Rates are standardized to US age distribution for 2006.

[2]US age-adjusted rates from Parker et al. based on crude, pooled prevalence data from 11 active case-ascertainment programs in 2004-2006, including Massachusetts (7). Nine of the other states contributing to the pooled estimates include elective terminations in addition to live births and stillbirths.

Error bars represent 95% confidence intervals.

Resources, Suggested Citation, Contact Information

Resources

For additional information on birth defects:

www.mass.gov/dph/birthdefects

Suggested Citation

Massachusetts Birth Defects 2011-2012. Boston, MA: Center for Birth Defects Research and Prevention, Bureau of Family Health and Nutrition, Massachusetts Department of Public Health. February 2016.

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

Friendly URL: www.mass.gov/dph/birthdefects

Contact Information

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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 Intervals

Prevalence is defined as the number of individuals with a disease or condition over a specified period of time divided by the number of individuals at risk during the same period. The numerator is the number of cases of birth defects. Since the preferred denominator is all pregnancies and since the number of pregnancies cannot be determined, the number of total 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 are included in the numerator but not in the denominator, so the result is technically a ratio and not a rate. This method of calculating rates is consistent with the national “Guidelines for Conducting Birth Defects Surveillance” (National Birth Defects Prevention Network, June 2004). Because the number of fetal deaths is so small, the inclusion of fetal deaths in the denominator would not substantially change the ratio.

The confidence interval (CI) is a method of assessing the magnitude and stability of a rate or ratio. The CI represents a range of values that has a 95% probability of including the true rate or ratio. Observed rates are subject to statistical variation. Thus, even if the underlying risk of a birth defect is identical in two subpopulations, the observed rates for the subpopulations may differ because of random variation. The width of the confidence interval 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 interval reflects the size of the subpopulation and the number of cases of birth defects. Smaller subpopulations with fewer defects lead to wider confidence intervals. The 95% confidence intervals used in the report are calculated using the Poisson method, except for the CIs for the age-adjusted rates, which are calculated using the standard method. If confidence intervals for two rates overlap, this means that we cannot rule out random variation to explain any differences in the rates.

Assignment of Race/Ethnicity

The Center follows the recommendation of the National Center for Health Statistics of classifying births according to the self-reported race/ethnicity of the mother. The 2003 version of the Certificate of Live Birth was implemented in Massachusetts in early 2011 (http://www.cdc.gov/nchs/vital_certs_rev.htm), 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.

Assignment of Severity

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

Some of the rules for assigning severity level are briefly described here. First, each defect labeled by an ICD9/BPA code was assigned a severity score or range of severity scores based on the defining characteristics of the defect. Each case was assigned a severity score based on the most severe defect that occurred within that case. Cases with infant death when a lethal anomaly was not present were reviewed by the Center Clinical Geneticist and manually assigned a severity level. Cases with a syndrome plus an additional defect(s) were categorized according to the severity of the syndrome.

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:

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

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.

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 350g 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 10th 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 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 2011-2012 Live Birth Populations Used in Calculating Rates

Numbers of Live Births to Massachusetts Residents				
		2011 N=73,174	**2012** N=72,457	**Total** N=145,631
By Maternal Age	<20	3511	3253	6764
	20-24	11,145	10,869	22,014
	25-29	18,241	17,947	36,188
	30-34	24,017	24,020	48,037
	35+	16,256	16,367	32,623
	Unknown	4	1	5
By Infant Sex	Male	37,512	37,086	74,598
	Female	35,660	35,369	71,029
	Unknown/Ambiguous	2	2	4
By Plurality	Singleton	69,931	69,274	139,213
	Multiple Birth	3235	3183	6418
By Maternal Race/Ethnicity	White	45,974	45,032	91,006
	Black	6999	6892	13,891
	Hispanic	12,777	13,088	25,865
	Asian/Pacific Islander	6023	6530	12,553
	American Indian	265	253	518
	Other	523	395	918
	Unknown	613	267	880
By Region	Western	8567	8397	16,964
	Central	9297	9254	18,551
	Northeast	15,336	15,192	30,528
	Metro West	16,889	16,698	33,587
	Southeast	12,708	12,668	25,376
	Boston	10,377	10,247	20,624

Based on data from 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 (2014) “Massachusetts Births 2011-2012”.

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

Birth Defect	ICD-9-CM Codes[1]	Modified ICD-9-CM/BPA Codes[2]	Comments
Central Nervous System			
Anencephaly	740.0 – 740.1	740.00 – 740.10	
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	Incl. if 2 standard deviations below the mean, adjusted for sex, gestational age and length
Spina bifida with and without Hydrocephaly	741.0, 741.9 740.0 - 740.1	741.00 – 741.99 740.00 – 740.10	
Spinal Cord anomalies	348.0, 745.51, 742.53, 742.59	742.52,742.54,742.58	
Eye			
Aniridia	743.45	743.42-743.424	
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	
Ear			
Anotia/Microtia	744.01, 744.23	744.01, 744.21	
Cardiovascular			
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 – 745.59	Includes ASD Secundum, Other and not otherwise specified (NOS)
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	

Birth Defect	ICD-9-CM Codes[1]	Modified ICD-9-CM/BPA Codes[2]	Comments
Dextro-Transposition of the Great Arteries	745.10	745.10, 745.11	Excludes 745.19 (NOS)
Double Outlet Right Ventricle	745.11	745.185, 186, 188, 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	745.3	
Tetralogy of Fallot with/without pulmonary valve atresia	745.2	745.20 – 745.21, 747.31	747.31 is Pulmonary artery atresia with septal defect
Total anomalous pulmonary venous connection	747.41	747.42	
Tricuspid Valve Atresia	746.1	746.10	Excludes tricuspid valve stenosis (746.106)
Truncus	745.0	745.00 (excluding 745.01)	
Ventricular Septal Defect (VSD), Canal Type	745.69	745.685	
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).
Respiratory			
Choanal Atresia	748.0	748.00	
Lung Anomalies	748.4, 748.5	748.40, 748.41, 748.48, 748.50, 748.51, 748.52, 748.58, 748.88	
Orofacial			
Cleft Palate without Cleft Lip	749.0	749.00 – 749.09	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	

Birth Defect	ICD-9-CM Codes[1]	Modified ICD-9-CM/BPA Codes[2]	Comments
Gastrointestinal			
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	
Small Intestinal Atresia	751.1	751.10 – 751.19	
Genitourinary			
Bladder Exstrophy	753.5	753.50	
Cloacal Exstrophy	751.5	751.555	
Hypospadias, 2nd or 3rd degree	752.61	752.60 – 752.62 (excluding 752.61 and 752.621)	In males only
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	Bilateral only
Musculoskeletal			
Club Foot	754.51, 754.70	754.50, 754.73 (excluding 754.735)	Requires casting or surgery
Craniosynostosis	No specific code	756.00 – 756.03	
Diaphragmatic Hernia	756.6	756.60-756.605, 756.61 – 756.617, 756.618-756.619	
Gastroschisis	756.73	756.71	
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	755.4 codes not used
Reduction Deformity, Upper limbs	755.2	755.20 – 755.29	755.4 codes not used
Skeletal Dysplasia	755.55, 756.4, 756.5	755.555, 756.41, 756.43, 756.447, 756.46, 756.480, 756.49, 756.50, 756.53, 756.54, 756.575, 756.58, 756.59	

Birth Defect	ICD-9-CM Codes[1]	Modified ICD-9-CM/BPA Codes[2]	Comments
Chromosomal			
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
Other			
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	

[1] International Classification of Diseases, 9th Revision.
[2] Centers for Disease Control/Clinical Modification, British Pediatric Association.

Appendix 5: Complete List of ICD-9-CM/BPA Birth Defect Codes and Counts Massachusetts, Live Births and Stillbirths: 2011-2012

ICD-9-CM/BPA Code Label	Code	Count
Cardiovascular		
Anomalous Pulmonary Venous Connection		
Partial anomalous pulmonary venous return/connection/drainage	747430	9
Total anomalous pulmonary venous return/connection/drainage	747420	12
Atrioventricular Canal Defects		
Atrial septal defect (ASD), primum type	745600	1
Common Atrium	745610	5
Complete atrioventricular canal	745630	60
Complete atrioventricular canal with ventricular septal defect	745620	1
Endocardial cushion defect, Not otherwise specified (NOS)	745690	3
Endocardial cushion defect, Other specified (OS)	745680	17
Ventricular septal defect (VSD), inflow type (subtricuspid, canal-type)	745685	9
Conotruncal and Aortic Arch		
Dextro-transposition of great arteries with intact ventricular septum	745100	17
Dextro-transposition of great arteries with VSD	745110	17
Double-outlet right ventricle with normally related great arteries	745185	8
Double-outlet right ventricle with transposed great arteries	745186	5
Double-outlet right ventricle, NOS	745189	2
Double-outlet right ventricle, OS	745188	1
Pulmonary atresia with VSD (Tetralogy of Fallot with pulmonary atresia)	747310	7
Tetralogy of Fallot	745200	43
Truncus Arteriosus	745000	5
Ebstein Anomaly		
Ebstein Malformation or Anomaly	746200	5
Heterotaxy (Laterality Defects)		
Situs ambiguus, left; left isomerism	759360	1
Situs ambiguus, sidedness NOS	759380	4
Situs ambiguus, sidedness unclear	759370	2
Situs inversus abdominis	759330	1
Left-Sided Obstruction		
Aorta: Atresia/absence	747200	1
Aortic stenosis, valvar	746300	17
Coarctation of the aorta, postductal (distal)	747110	1
Coarctation of the aorta, preductal (proximal)	747100	1
Coarctation of the aorta, juxtaductal	747120	10
Coarctation of the aorta, NOS	747190	56
Hypoplastic left heart syndrome	746700	24
Interrupted aortic arch, NOS	747215	1
Interrupted aortic arch, type A	747216	3

ICD-9-CM/BPA Code Label	Code	Count
Right-Sided Obstruction		
Pulmonary valve atresia with VSD (not 747.310)	746030	1
Pulmonary valve atresia/intact ventricular septum	746000	6
Pulmonic stenosis, valvar	746010	119
Tricuspid atresia	746100	3
Septal Defects		
Atrial septal defect, NOS	745599	27
Atrial septal defect, OS	745580	1
Atrial septal defect (ASD), Secundum type	745510	302
Ventricular septal defect (VSD), NOS	745490	10
VSD, Malalignment-type (type I, subarterial)	745487	23
VSD, Perimembranous (type II, membranous)	745485	168
Single Ventricle and L-TGA		
Levo-Transposition of the great arteries/great vessels/ventricular inversion Excludes: dextrocardia (use 746.800)	745120	6
Single ventricle, NOS	745300	2
Single ventricle, Double Inlet Left Ventricle	745310	1
Other Cardiovascular		
Anomalies of coronary artery or sinus	746885	17
Aorta: Hypoplasia	747210	4
Aorta: Congenital aneurysm/dilatation	747270	2
Aorta: Other specified anomalies	747280	1
Aorta: Persistent right aortic arch	747230	16
Aorta: Vascular ring/double aortic arch/vascular ring compression of trachea	747250	5
Aortic septal defect/aorto pulmonary window. Excludes ASD (use 745.590)	745010	7
Aortic valve: bicuspid/insufficiency or regurgitation Excludes ‘mild', ‘minimal', 'trivial', or 'physiologic'	746400	1
Aortic valve: Other specified anomalies/aortic valve atresia. Excludes: supravalvular aortic stenosis (747.220)	746480	18
Arteriovenous malformation, peripheral Excludes: pulmonary (747.340), cerebral (747.800), and retinal (743.510) arteriovenous malformations	747620	1
Bicuspid Aortic Valve	746470	74
Cerebral vessels: Other anomalies/vein of Galen	747810	1
Circulatory system: Other specified anomalies. Excludes aneurysms: coronary, peripheral, pulmonary, retinal, ruptured cerebral	747880	2
Cor triatriatum	746820	2
Great veins: Other specified anomalies	747480	6
Heart: Other specified anomalies/ectopia cordis/mesocardia/conduction defects, NOS	746880	37
Mitral valve: Absence, atresia, or hypoplasia	746505	1
Mitral valve: Congenital mitral stenosis	746500	5
Pericardium: Anomalies	746850	1
Peripheral arteries: Other anomalies/aberrant subclavian artery	747640	13
Peripheral vascular system: Other anomalies/primary pulmonary artery hypertension	747680	2
Persistent left superior vena cava	747410	14
Pulmonary artery: other specified/pulmonary artery hypoplasia	747380	4
Pulmonary artery: stenosis. Use 746.995 if artery or valve is not specified	747320	12

ICD-9-CM/BPA Code Label	Code	Count
Pulmonary infundibular (subvalvular) stenosis	746830	5
Pulmonary valve: Other specified anomalies. Excludes: infundibular stenosis (746.830)	746080	31
Situs: Dextrocardia without situs inversus/dextrocardia with situs solitus	746800	5
Tricuspid stenosis or hypoplasia	746106	2
Central Nervous System		
Agyria and lissencephaly	742240	3
Anencephaly	740020	8
Brain cysts: Cerebral/subependymal/periventricular	742420	3
Brain, reduction defect OS (Includes colpocephaly, pachygyria, schizencephaly) and absent septum pellucidum	742280	12
Brain: Other specified anomalies cortical atrophy/cranial nerve defects	742480	4
Cerebellar Hypoplasia	742235	5
Cerebellum anomalies	742230	4
Cerebrum anomalies	742200	3
Corpus callosum anomalies	742210	77
Dandy-Walker Malformation	742310	12
Encephalocele, NOS	742090	1
Encephalocele, Occipital	742000	3
Encephalocele, Parietal	742086	2
Enlarged brain and head/enlarged head/enlarged brain/megalencephaly/macrocephaly	742400	20
Holoprosencephaly, Alobar	742265	2
Holoprosencephaly, Lobar	742267	3
Holoprosencephaly, NOS	742260	2
Holoprosencephaly, Semilobar	742266	3
Hydranencephaly	742320	2
Hydrocephaly, NOS	742390	15
Hydrocephaly, Anomalies of Aqueduct of Sylvius	742300	18
Hydrocephaly, Other Specified	742380	3
Lipomeningocele, Highest level, lumbar, No mentioned hydrocephalus, closed lesion	741853	1
Lipomeningomyelocele, Highest level, lumbar, No mentioned hydrocephalus, closed lesion	741843	3
Meningocele, Highest level unspecified, No mentioned hydrocephalus, unspecified open/closed lesion	741919	1
Meningocele, Highest level, lumbar, No mentioned hydrocephalus, unspecified open/closed lesion	741913	1
Meningocele, Highest level, sacral, No mentioned hydrocephalus, closed lesion	741814	1
Meningocele, Highest level, thoracic, No mentioned hydrocephalus, closed lesion	741812	1
Meningomyelocele/myelomeningocele, Highest level, lumbar, Arnold Chiari malformation ± hydrocephalus, closed lesion	741103	1
Meningomyelocele/myelomeningocele, Highest level, lumbar, Arnold Chiari malformation ± hydrocephalus, open lesion	741003	14
Meningomyelocele/myelomeningocele, Highest level, lumbar, No hydrocephalus, unspecified open/closed lesion	741903	1
Meningomyelocele/myelomeningocele, Highest level, sacral, Arnold Chiari malformation ± hydrocephalus, open lesion	741004	1
Meningomyelocele/myelomeningocele, Highest level, sacral, No hydrocephalus, open lesion	741704	1
Microcephalus	742100	28
Microgyria/polymicrogyria	742250	11

ICD-9-CM/BPA Code Label	Code	Count
Myelocele, Highest level, lumbar, Arnold Chiari malformation ± hydrocephalus, open lesion	741023	1
Myelocystocele, Highest level, lumbar, No mentioned hydrocephalus, closed lesion	741833	1
Spinal cord: Other specified anomalies (Includes tethered cord and arachnoid cyst)	742580	72
Chromosomal		
22q11 deletion	758370	9
Apert syndrome/Acrocephalosyndactyly types I or II	756055	2
Autosome Other: Other specified anomalies/marker/Ring, derivative, mosaic, isochrome, "additional" material, inversions Excludes "pericentric inversion 9"	758580	14
Deletion 17p or 18p/deletion of short arm chromosome 17 or 18	758350	3
Deletion 17q or 18q/deletion of the long arm of chromosome 17 or 18	758340	4
Deletion 21q, monosomy 21, or a G-group NOS (archaic)	758300	1
Deletion 5p/Cri du chat syndrome	758310	3
Deletion: Autosome (not X or Y) (1-16, 4q,5q,19,20)	758380	32
Down syndrome: diagnosed clinically, but no karyotype in record	758090	1
Down syndrome: mosaic	758040	2
Down syndrome: translocation 21, duplication 21q, Robertsonian translocation, isochrome 21q	758020	7
Down syndrome: trisomy 21	758000	184
Ehlers-Danlos syndrome	756850	1
Ellis-van Creveld syndrome	756525	1
Goldenhar syndrome/oculoauriculovertebral dysplasia	756060	4
Hemifacial microsomia	756065	10
Klinefelter syndrome, NOS	758790	1
Klinefelter syndrome: 47, XXY	758700	6
Malformation syndrome OS: VATER/VACTERL/Acardia/Angelman/Bloom/CHARGE/hemihyper/Meckel-Gruber/Neu-Laxova/PentalogyCantrell/Sotos/TownesBrock/Walker Warburg/Weaver/VCFS/Shprintzen	759890	29
Malformation syndromes-face: Aarskog/BOF/BOR/Fraser/FreemanSheldon/Kabuki/Miller-Dieker/Noonan/Opitz G oral-facial-digita/Oto-palato-digital/Septo-optic dysplasia/Waardenburg/Williams	759800	20
Malformation syndromes-limbs: Baller-Gerold/Carpenter/caudal regression/Fryns/Holt-Oram/Klippel-Trenaunay-Webe/Limb Body Wall/Roberts/Rubinstein-Taybi/sirenomelia/thrombocytopenia-absent radius	759840	5
Malformation syndromes-metabolic: Alagille/Alport/Beckwith-Wiedemann/Johansen-Blizzard/leprechaunism/Lowe/Menkes (kinky hair)/Prader-Willi/Zellweger	759870	20
Malformation syndromes-other skeletal: Marfan/Stickler/Beemer Langer	759860	4
Malformation syndromes-short stature: Smith-Lemli-Optiz/de Lange/Cockayne/Laurence-Moon-Biedl/Russell-Silver/Seckel	759820	2
Moebius syndrome (multiple cranial nerve palsies)	352600	1
Mosaic XO/XY, 45X/46XY Excludes: Turner phenotype (758.610)	758800	1
Mosaic XY/XXY, 46XY/47XXY. Excludes: Klinefelter phenotype (758.710)	758820	1
Other craniofacial syndromes/Hallermann-Streiff syndrome	756046	2
Other specified acrocephalosyndactylies	756057	1
Other specified DNA based diagnosis	758999	4
Other Translocations Including Unbalanced and Other Balanced Translocations	758540	8
Sex chromosome: Other specified anomaly/fragile X	758880	3
Sturge-Weber syndrome/Encephalocutaneous angiomatosis	759610	3
Treacher-Collins syndrome/Mandibulofacial dysostosis	756045	1
		87

ICD-9-CM/BPA Code Label	Code	Count
Trisomy 13 (archaic Patau syndrome): cytogenetics result in record	758100	5
Trisomy 13: translocation trisomy with duplication 13q	758120	1
Trisomy 18 (archaic Edwards syndrome): cytogenetics result in record	758200	36
Trisomy 8	758500	1
Trisomy, partial	758530	24
Trisomy: Other total trisomy syndromes/trisomy 22/trisomy, NOS	758520	1
Tuberous sclerosis/Bourneville's disease	759500	5
Turner phenotype: karyotype 45,X [XO] Note: The 7586xx code series that follows excludes pure gonadal dysgenesis(752.720)	758600	5
Turner phenotype: variant karyotypes, e.g. Isochromosome, mosaic (eg X, XX,XY), partial X deletion, ring X chromosome. Excludes: Turner phenotype with normal karyotype	758610	5
XXX female/47XXX/Triple X syndrome	758850	7
XYY, male/47,XYY/mosaic XYY male	758840	2
DiGeorge syndrome	279110	8
Ear		
Absence or stricture of auditory canal	744000	12
Anomaly of inner ear/congenital anomaly of membranous labyrinth or organ of Corti	744030	11
Anomaly of middle ear/fusion of ossicles	744020	5
Anotia, Right	744012	1
Ear: Accessory auricle/polyotia	744100	39
Ear: Other specified anomalies (see also 744.230)/Darwin tubercle	744280	4
Ear: Unspecified anomalies with hearing impairment/congenital deafness, NOS	744090	2
Ear: Other misshapen ear/cleft/malformed/pointed/elfin, pixie-like/lop/cauliflower/absent or decreased cartilage -- a conditional exclusion if <36wks	744230	9
Ear: Unspecified anomalies/congenital anomaly (any part)/anomaly, deformity, NOS	744300	1
Microtia, Bilateral	744214	5
Microtia, Left	744211	10
Microtia, Right	744212	20
Eye		
Absence of iris/Aniridia, Bilateral	743424	4
Anophthalmos, Bilateral	743004	1
Anterior segment: OS colobomas and anomalies (Use for Rieger Syndrome, use 759.800. For Reiger anomaly, use 743.470-474)	743480	1
Anterior segment: Unspecified colobomas and anomalies	743490	4
Buphthalmos/Congenital Glaucoma, Bilateral	743204	6
Buphthalmos/Congenital Glaucoma, Left	743201	1
Buphthalmos/Congenital Glaucoma, Right	743202	6
Cataract, anterior polar, Bilateral	743354	2
Cataract, anterior polar, Right	743352	1
Cataract, NOS, Left	743321	7
Cataract, NOS, Bilateral	743324	13
Cataract, NOS, Right	743322	8
Cataract, other specified, Left	743361	1
Choroid: Coloboma	743535	1
Cornea, other anomalies. Excludes: megalocornea (use 743.220)	743410	3

ICD-9-CM/BPA Code Label	Code	Count
Eye: Other specified anomalies/exophthalmos/epicanthal folds/downward slant/Brushfield spots	743800	9
Eyelid: Entropion	743620	1
Eyelids: Coloboma	743636	2
Iris,other anomalies: polycoria/ectopic pupil (For Peters syndrome, use 759.800. For Peters Anomaly, use 743.460-469) Excludes: Brushfield spots	743440	1
Iris: Coloboma	743430	5
Microphthalmos, Bilateral	743104	9
Microphthalmos, Left	743101	2
Microphthalmos, Right	743102	2
Optic disc: Specified anomalies/hypoplastic optic nerve/coloboma of the optic disc	743520	15
Peters Anomaly, Left	743461	1
Peters Anomaly, Right	743462	1
Retina: Specified anomalies/congenital retinal aneurysm. Excludes: Stickler syndrome (use 759.860)	743510	3
Tear ducts: Stenosis, stricture, or obstruction of lacrimal duct	743650	3
Vitreous humor: Specified anomalies (includes persistent hyperplastic primary vitreous)	743500	4
Gastrointestinal		
Agenesis or hypoplasia of gallbladder	751630	1
Anal atresia with fistula	751230	28
Anal atresia without mention of fistula	751240	18
Anomalies of mesentery	751410	1
Biliary atresia, extrahepatic or NOS (use 751.670 for intrahepatic)	751650	14
Choledochal cysts	751660	5
Congenital adhesions or bands of omentum and peritoneum/Ladd's bands	751420	2
Congenital anal fistula	751540	1
Cystic or fibrocystic disease of liver	751610	1
Duodenal web	751560	2
Duplication of anus, appendix, cecum, or intestine/enterogenous cyst	751500	10
Ectopic (displaced, anteriorly placed) anus	751530	15
Esophageal atresia with TE fistula	750310	39
Esophageal atresia without TE fistula	750300	6
Hirschsprung disease, NOS	751330	1
Hirschsprung disease: Long-segment (aganglionosis beyond rectum)	751310	14
Intestinal atresia/stenosis, Duodenum	751100	21
Intestinal atresia/stenosis, Ileum	751120	8
Intestinal atresia/stenosis, Jejunum	751110	3
Intestinal atresia/stenosis, Large Intestine, NOS	751200	3
Intestinal atresia/stenosis, Small Intestine, NOS	751190	1
Intestine: Other specified anomalies/rectal fissures	751580	2
Larynx, trachea, and bronchus: Unspecified anomalies	748390	1
Malrotation: Other specified and unspecified	751490	43
Malrotation: Small intestine alone	751495	1
Meckel's diverticulum	751010	6
Microcolon	751520	2

ICD-9-CM/BPA Code Label	Code	Count
Other anomalies of gallbladder/duplication of gall bladder	751640	1
Other specified anomalies of esophagus	750480	1
Other tracheoesophageal anomalies	750380	1
Pancreas: Annular	751720	8
Persistent omphalomesenteric duct/persistent vitelline duct	751000	5
Tracheoesophageal fistula without mention of esophageal atresia	750320	1
Tracheoesophageal fistula, "H" type	750325	3
Rectal atresia/stenosis with fistula	751210	1
Rectal atresia/stenosis without mention of fistula	751220	5
Genitourinary		
Absence of bladder or urethra	753800	1
Absence of testis/monorchidism, NOS	752800	2
Aplasia or hypoplasia of testis and scrotum/hypoplastic scrotum due to cryptorchidism	752810	2
Atresia, stricture, or stenosis of ureter/ureteropelvic junction obstruction or stenosis/ureterovesical junction obstruction or stenosis/hypoplastic ureter	753210	1
Bladder exstrophy	753500	3
Cloacal exstrophy	751550	7
Congenital diverticulum or hernia of bladder	753820	3
Congenital hydronephrosis/pyelocaliectasis	753200	206
Double urethra or urinary meatus	753840	1
Gynecological: OS anomalies of cervix, vagina, or external female genitalia	752480	2
Gynecological: Ovaries absence or agenesis	752000	1
Gynecological: Ovaries, Multiple cysts	752085	3
Gynecological: Ovaries, Other specified anomalies	752080	2
Gynecological: Uterus absence or agenesis	752300	1
Gynecological: Uterus doubling/doubling of uterus (any degree) or associated with doubling of cervix and vagina	752200	3
Gynecological: Uterus, other anomalies/bicornuate/unicornis	752380	2
Gynecological: Vulva Absence or anomaly	752440	1
Hypospadias, Second Degree	752606	54
Hypospadias, Second Degree with chordee	752626	68
Hypospadias, Third Degree	752607	10
Hypospadias, Third Degree with chordee	752627	31
Indeterminate sex, NOS/ambiguous genitalia	752790	16
Kidney/renal: absence, agenesis, dysplasia, or hypoplasia, NOS	753009	24
Kidney/renal: cyst, single	753100	5
Kidney: Double or triple, pelvis/pyelon duplex or triplex	753310	11
Kidney: Ectopic/pelvic	753330	6
Kidney: Lobulated, fused, or horseshoe/crossed fused ectopia	753320	9
Kidney: Other specified anomalies	753380	1
Kidney: Other specified disease/cystic NOS	753180	2
Kidneys: Multicystic renal dysplasia/multicystic kidney	753160	23
Kidneys: Polycystic, adult type	753120	1
Kidneys: Polycystic, infantile type	753110	10
Megaloureter, NOS/hydroureter	753220	4
		90

ICD-9-CM/BPA Code Label	Code	Count
Obstruction, atresia or stenosis of urinary meatus/meatal stenosis	753630	1
Penis: Other anomalies/concealed penis/absent or hooded foreskin	752860	60
Penis: Small/hypoplastic/micropenis	752865	7
Renal agenesis, bilateral	753000	4
Renal agenesis, right + renal hypoplasia, left	753007	1
Testis and scrotum: Other anomalies/polyorchidism/bifid scrotum. Excludes: torsion of the testes or spermatic cord	752820	21
True hermaphroditism/ovotestis	752700	1
Urachus: Cyst	753710	6
Urachus: Other and unspecified anomaly	753790	3
Urachus: Patent	753700	2
Ureter: Accessory/double ureter/duplex collecting system	753410	28
Ureter: Ectopic	753420	1
Ureter: Other specified anomalies/ureterocele	753480	8
Ureter: Variations of vesicoureteral reflux	753485	41
Urethra: Congenital posterior urethral valves or posterior urethral obstruction	753600	1
Musculoskeletal		
Absence of foot or toes, Bilateral	755349	4
Absence of foot or toes, Left	755346	6
Absence of foot or toes, Right	755347	3
Absence of hand or fingers, Bilateral	755249	1
Absence of hand or fingers, Left	755246	12
Absence of hand or fingers, Right	755247	5
Absence of the forearm and hand, Left	755241	3
Absence of the lower leg and foot, Left	755341	2
Achondroplasia	756430	12
Anomalies of elbow and upper arm	755540	1
Anomalies of fingers/camptodactyly/macro-/brachy-/clino-, triphalangeal thumb. Excludes: acrocephalosyndactyly (use756.050)/Apert syndrome (use 756.055)	755500	32
Anomalies of forearm, NOS	755530	1
Arthrogryposis multiplex congenita/distal arthrogryposis syndrome. Includes: one or more flexion contractures of individual joints	755800	4
Bowing, tibia and/or fibula	754410	1
Certain musculoskeletal anomalies face, face, jaw: Use for asymmetry of face	754000	8
Club foot, NOS/talipes, NOS	754730	99
Club foot: Metatarsus varus or adductus	754520	19
Club foot: Complex varus deformities	754530	2
Club foot: Talipes calcaneovarus	754510	2
Club foot: Talipes equinovarus	754500	81
Congenital deformities of foot, NOS	754735	1
Congenital postural scoliosis	754200	21
Craniosynostosis, Coronal, Bilateral	756014	5
Craniosynostosis, Coronal, Left	756011	5
Craniosynostosis, Coronal, Right	756012	9
Craniosynostosis, Lambdoidal, Left	756021	3

ICD-9-CM/BPA Code Label	Code	Count
Craniosynostosis, Metopic	756006	10
Craniosynostosis, Sagittal	756005	32
Craniosynostosis, Unspecified Type, Laterality Unknown	756000	3
Diaghragmatic hernia, Morgagni, Bilateral	756619	1
Diaphragm: Eventration	756620	3
Diaphragmatic hernia, Bochdalek, Left	756611	3
Diaphragmatic hernia, Morgagni, Left	756616	2
Diaphragmatic hernia, Morgagni, Right	756617	3
Diaphragmatic hernia, NOS (includes absent/hemidiaphragm), Laterality Unknown	756600	1
Diaphragmatic hernia, NOS (includes absent/hemidiaphragm), Left	756601	21
Diaphragmatic hernia, NOS (includes absent/hemidiaphragm), Right	756602	3
Dislocation of knee, congenital	754440	1
Fibular aplasia/hypoplasia, Left	755371	1
Fibular aplasia/hypoplasia, Right	755372	1
Gastroschisis	756710	44
Genu recurvatum	754430	5
Hallux or great toe only missing or hypoplastic, Left	755361	1
Hallux or great toe only missing or hypoplastic, Right	755362	1
Infantile cortical hyperostosis/Caffey syndrome	756530	1
Longitudinal deficiency of arm, NOS, Right	755252	1
Lower limb: hypoplasia/Toes, feet, legs: hypoplasia. Excludes: aplasia of or absent lower limb (see 755.3)	755685	7
Lower limb: other specified anomalies/hyperextended legs/shortening of legs	755680	1
Omphalocele	756700	28
Osteogenesis imperfecta	756500	8
Osteopetrosis/Albers-Schonberg syndrome/marble bones	756540	1
Other absent or hypoplastic muscle/absent pectoralis major. Excludes: prune belly syndrome (use 756.720)	756810	1
Other and unspecified anomalies of abdominal wall	756790	3
Other specified chondrodystrophy. Excludes: Conradi's (use 756.575)	756480	2
Other specified deformities of ankle and/or toes/dorsiflexion of foot. Excludes: widely spaced first and second toes	754780	11
Other specified osteodystrophies	756580	5
Other specified valgus deformities of foot	754680	1
Poland syndrome or anomaly	756800	1
Polydactyly fingers/postaxial polydactyly, Type A	755005	64
Polydactyly: Accessory big toe (preaxial)	755030	8
Polydactyly: Accessory digits foot, NOS (preaxial, postaxial not specified)	755096	6
Polydactyly: Accessory digits hand, NOS (preaxial, postaxial not specified)	755095	5
Polydactyly: Accessory thumbs (preaxial polydactyly)	755010	49
Polydactyly: Accessory toes (postaxial)	755020	38
Prune belly syndrome	756720	1
Radial aplasia/hypoplasia, Right	755267	1
Ribs: Absence	756300	5
Ribs: Extra	756330	3

ICD-9-CM/BPA Code Label	Code	Count
Ribs: Other anomalies	756340	1
Sacral agenesis	756175	1
Skull and face bone: Other specified anomalies/localized skull defects/mid-facial hypoplasia/prominent maxilla/hypotelorism/flat occiput/prominent occiput	756080	4
Spine: Kyphosis/kyphoscoliosis	756120	3
Split-Foot, Bilateral	755359	1
Split-Hand, Bilateral	755259	4
Split-Hand, Left	755256	4
Syndactyly: Fused fingers	755100	13
Syndactyly: Fused toes	755120	6
Syndactyly: Unspecified (webbed vs. fused) thumb and/or fingers, NOS	755193	2
Syndactyly: Unspecified (webbed vs. fused) Toes	755199	3
Syndactyly: Unspecified toes	755194	1
Syndactyly: Unspecified, laterality not specified	755196	2
Syndactyly: Webbed fingers	755110	15
Syndactyly: Webbed toes. Exclude webbing between toes 2-3	755130	32
Talipes calcaneovalgus	754600	9
Thanatophoric dwarfism	756447	1
Thumb only missing or hypoplastic, Bilateral	755264	2
Thumb only missing or hypoplastic, Left	755261	2
Thumb only missing or hypoplastic, Right	755262	8
Tibial aplasia/hypoplasia, Right	755367	2
Unspecified reduction defect of lower limb	755390	1
Unspecified varus deformities of feet	754590	3
Upper limb: Hypoplasia/Fingers, hands, or arms: hypoplasia. Excludes: aplasia or absent upper limb (use 755.2)	755585	12
Vertebrae, cervical: anomalies	756140	2
Vertebrae, lumbar: anomalies	756160	3
Vertebrae, lumbar: hemivertebrae	756165	4
Vertebrae, sacrococcygeal: anomalies/agenesis of sacrum. Excludes: pilonidal sinus (use 685.100)	756170	13
Vertebrae, thoracic: anomalies	756150	10
Vertebrae, thoracic: hemivertebrae	756155	12
Vertebrae: Hemivertebrae, NOS	756185	1
Vertebrae: Other specified anomalies	756180	1
Orofacial		
Branchial cleft, sinus, fistula, cyst, or pit	744400	23
Cleft hard palate, Bilateral	749010	9
Cleft hard palate, Central	749020	4
Cleft hard palate, NOS	749030	16
Cleft lip and palate, Bilateral cleft lip	749210	17
Cleft lip and palate, Central cleft lip	749220	2
Cleft lip and palate, NOS	749290	3
Cleft lip and palate, Unilateral cleft lip, Left	749201	18
Cleft lip and palate, Unilateral cleft lip, Right	749202	12

ICD-9-CM/BPA Code Label	Code	Count
Cleft lip, Bilateral	749110	6
Cleft lip, NOS	749195	1
Cleft lip, Unilateral, Left	749101	23
Cleft lip, Unilateral, Right	749102	10
Cleft lip, Unilateral, Side Unknown	749103	1
Cleft palate, NOS	749090	13
Cleft soft palate, Central	749060	2
Cleft soft palate, NOS	749070	41
Cleft: Incomplete Cleft lip/microform/pseudo/fused lip/healed lip	749190	3
Face or neck: Other specified anomalies (e.g. Facial cleft)	744880	9
Nose: OS anomalies (small nose and nostril/absent nasal septum)	748180	21
Other branchial cleft anomalies dermal sinus of head	744480	22
Pierre Robin sequence	524080	36
Tongue: Dislocation or displacement/glossoptosis	750130	1
Tongue: large/macroglossia	750120	1
Respiratory		
Hypoplasia of lung or pulmonary hypoplasia -- a conditional in infants <36wks.	748510	3
Choanal atresia, Bilateral	748014	5
Choanal atresia, Left	748011	2
Choanal atresia, Right	748012	5
Choanal stenosis	748000	10
Larynx: Cleft/laryngotracheoesophageal cleft --use for laryngeal atresia/stenosis	748385	8
Lung agenesis or aplasia	748500	2
Lung cysts: Congenital cystic adenomatoid malformation, OS	748480	14
Lung cysts: Single	748400	2
Lung: Bilobed right/right lung with left lung bronchial pattern	748625	1
Lung: other specified dysplasia/fusion of lobes	748580	1
Lung: sequestration	748520	15
Nose: Agenesis or underdevelopment	748100	1
Other anomalies of trachea. Excludes: vascular ring compression of the trachea (use 747.250)	748330	1
Respiratory system: Other specified anomalies/congenital lobar emphysema/lymphangiectasia of lung	748880	2
Other		
Adrenogenital syndrome/adrenal hyperplasia	255200	6
Amniotic band sequence	658800	19
Anomalies of thymus/absent thymus	759240	4
Anomalies of thyroid gland	759210	1
Collodion baby	757110	3
Epidermolysis bullosa	757330	4
Hair: Other specified anomalies	757480	1
Hamartomas: Other specified	759680	2
Ichthyosiform erythroderma	757197	3
Multiple congenital anomalies (anomaly or deformity, multiple, NOS)	759700	4
Other and unspecified ichthyosis	757190	4
		94

ICD-9-CM/BPA Code Label	Code	Count
Skin: Other specified anomalies/scalp defects. For specified anomalies of skin use 757.390 For specified anomalies of hair, use 757.480. For specified anomalies of nails use 757.580	757800	7
Spleen: Absence/asplenia	759000	6
Spleen: Accessory. Includes polysplenia	759040	2
Thyroglossal duct anomalies/thyroglossal cyst	759220	2
X-linked ichthyosis	757196	2

Note: Some of the defects included in this table would not be reported if they occurred alone but are reported here because they occur along with a reportable defect from the list in Appendix 4.

NOS: Not otherwise specified; OS: Other, specified

Appendix 6: Selected Birth Defects by Severity

Examples of Severe Birth Defects (usually incompatible with life)

Anencephaly

Bilateral renal agenesis

Trisomy 13

Trisomy 18

Examples of Serious Birth Defects (may be correctable, most have long-term needs)

Anophthalmia

Biliary atresia

Bladder exstrophy

Cloacal exstrophy

Encephalocele

Hypoplastic left heart syndrome

Spina bifida

Trisomy 21/Down syndrome

Examples of Moderate Birth Defects (most are correctable, many have long-term needs)

Choanal atresia

Cleft lip/palate

Diaphragmatic hernia

Esophageal atresia/tracheoesophageal fistula

Gastroschisis

Hirschsprung disease

Intestinal atresia

Imperforate anus/rectal atresia and stenosis

Microtia

Omphalocele

Tetralogy of Fallot

Examples of Mild Birth Defects (may be correctable, minimal long-term needs)

Microphthalmia

Polydactyly/syndactyly

Appendix 7: Pre-Pregnancy Multivitamin Use Massachusetts: 2011

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

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

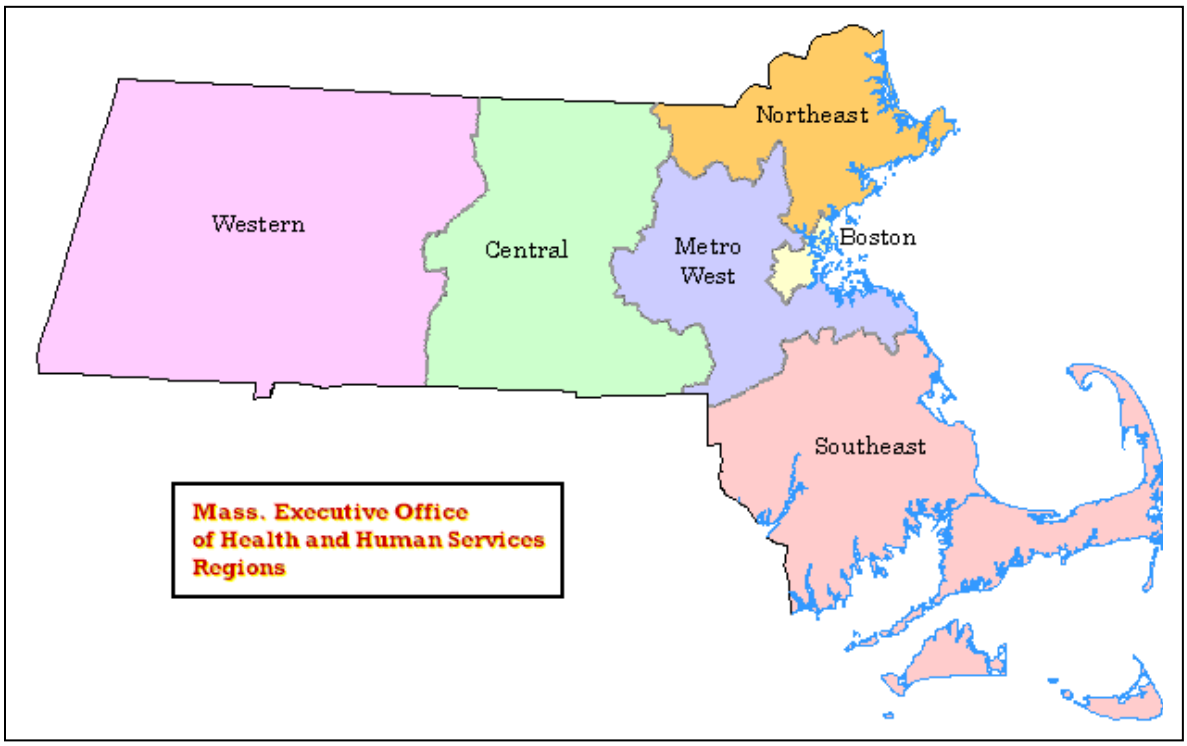
Daily Multivitamin Use in the Month Prior to Pregnancy by Socio-demographic Characteristics, Massachusetts: 2011

Characteristic	Weighted n	Weighted %	95% Confidence Interval
Total	27520	39.4	36.2 - 42.7
Maternal race/ethnicity			
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
Maternal age (years)			
<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
Maternal education			
<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
Household poverty level			
≤100% FPL[1]	3892	22.2	17.5 - 27.7
>100% FPL	21909	47.5	43.3 - 51.8
Maternal nativity			
Non-US-born	7548	35.3	30.7 - 40.2
US-born	19840	41.0	36.8 - 45.3

[1]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 8: Map of Massachusetts Regions



Massachusetts Birth Defects 2011-2012 Evaluation Form

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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