# Massachusetts Birth Defects 2006-2007

Massachusetts Birth Defects Monitoring Program
Bureau of Family Health and Nutrition

Massachusetts Department of Public Health

October 2010

# Massachusetts Birth Defects 2006-2007

Deval L. Patrick, Governor
Timothy P. Murray, Lieutenant Governor
JudyAnn Bigby, MD, Secretary, Executive Office of Health and Human Services
John Auerbach, Commissioner, Massachusetts Department of Public Health
Ron Benham, Director, Bureau of Family Health and Nutrition
Marlene Anderka, Director, Massachusetts Center for Birth Defects Research and Prevention
Cathleen Higgins, Birth Defects Surveillance Coordinator
Christopher Borger, Birth Defects Epidemiologist

Massachusetts Department of Public Health
*http://www.mass.gov/dph/birthdefects*

October 2010

# Acknowledgements

This report was prepared by the staff of the Massachusetts Center for Birth Defects Research and Prevention (MCBDRP) including: Marlene Anderka, Elizabeth Bingay, Christopher Borger, Han Fang, Kelly Getz, Cathleen Higgins, Angela Lin, Rebecca Lovering, Gerlinde Munshi, and Samantha Parker.

Data in this report have been collected through the efforts of the field staff of the MCBDRP including: Roberta Aucoin, Daniel Sexton, Lori Tetrault, Marie-Noel Westgate and Susan Winship.

We would like to acknowledge the following individuals for their time and commitment to supporting our efforts in improving the MCBDRP.

Lewis Holmes, MD, MassGeneral Hospital for Children
Carol Louik, ScD, Slone Epidemiology Center, Boston University
Allen Mitchell, MD, Slone Epidemiology Center, Boston University
Martha Werler, ScD, Slone Epidemiology Center, Boston University

Ed Doherty, March of Dimes Massachusetts Chapter

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

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

Funding for surveillance of 2006-2007 birth defects in Massachusetts was provided in part by the Centers for Disease Control and Prevention via the Centers for Birth Defects Research and Prevention Grant # U50-CCU113247.

**For more information, contact:**

Massachusetts Department of Public Health
Bureau of Family Health and Nutrition
Massachusetts Center for Birth Defects Research and Prevention
Massachusetts Birth Defects Monitoring Program
250 Washington Street
Boston, MA 02108
(617) 624-5510

**This and other Department of Public Health publications and materials can be downloaded from the Internet at:**
***http://www.mass.gov/dph***

### *Note to Readers: Changes in this year's report*

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

| *Section* | *Change or addition* |
|---|---|
| *Chapter 1* | *This chapter contains a description of changes to the Massachusetts legislative regulations regarding case ascertainment and abstraction.* |
| *Chapter 3* | *A figure showing the overall prevalence of defects by interpregnancy inverval (IPI) (Figure 2) was added.* |
| | *Small for gestational age (SGA) was added as a pregnancy outcome in Figure 3.* |
| *Appendix* | *A list of birth defects classified by severity level was added.* |
| | *The map of Massachusetts regions has been enhanced to include age-adjusted regional prevalences.* |

# TABLE OF CONTENTS

# Executive Summary

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

The causes of birth defects are poorly understood. For 60-70% of major birth defects, no known cause has been identified. Researchers are looking at a wide variety of environmental exposures and risk factors as possible causes. Because most of the structural development of the fetus occurs during early pregnancy, studies usually focus on the "periconceptional" period, the month before and three months after conception. For the developing pregnancy, the environment includes any exposures to the fetus as well as any exposures to the mother.

Folic acid deficiency is related to certain birth defects. The awareness of folic acid intake to prevent birth defects may be increasing, although almost half of Massachusetts mothers (47.7%) with recent live births surveyed as part of the CDC Pregnancy Risk Assessment Monitoring System (PRAMS) did not take any multivitamins or prenatal vitamins during the month prior to pregnancy (MA PRAMS Report 2010).

The Massachusetts combined lifetime costs for babies born with any of 12 major structural birth defects was estimated at $129 million in 2007 dollars (see Technical Notes for inflation adjustment) (Harris and James 1997). These figures include direct costs of medical treatment, developmental services and special education, as well as indirect costs to society for lost wages due to early death or occupational limitations. The psychosocial costs cannot be calculated.

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

This report presents statewide data on the prevalence of birth defects among live births and stillbirths in Massachusetts during the years 2006 and 2007. The first annual report presented Massachusetts birth defects data for the year 1999. Three subsequent reports compiled data from 2000-2001, 2002-2003, and 2004-2005, respectively. Our ability to find and identify infants born with birth defects to Massachusetts residents has improved over time and is reflected in increased

prevalence rates. The 2006-2007 data are presented in combined form since the numbers are relatively small for individual defects. Although with the 2006-2007 report we appear to have established a preliminary baseline rate, given its comparability with previous reports, interpretations of these data with respect to the 2004-2005 report must still be made with caution.

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

Planning for children with special health care needs is essential to support affected infants and families. Coordination between the BDMP and maternal and child health programs helps to ensure services for identified children and to provide population-based information to guide future program planning and prevention strategies.

## Prevalence

The overall prevalence of birth defects among births to Massachusetts residents in 2006-2007 was 166.1 per 10,000 live births. Among the 155,604 live births to Massachusetts residents in 2006-2007, 2,528 had one or more birth defects. In addition, 57 stillbirths were identified as having a birth defect. Although population-based, active surveillance statewide from 1999 through 2007 provides baseline frequencies for birth defects, more years of data will establish the stability of these preliminary baseline frequencies. The ten most common defects were unchanged from the 2005-2006 report. Three of these ten most common defects were cardiovascular defects: atrial septal defects (secundum and NOS), ventricular septal defects (membranous and NOS), and pulmonary stenosis, valvular. Common non-cardiovascular defects included hypospadias ($2^{nd}$ or $3^{rd}$ degree*)*, clubfoot, polydactyly/syndactyly, Down syndrome, obstructive genitourinary defects, cleft lip with and without cleft palate and cleft palate without cleft lip.

The CDC published improved national prevalence estimates for 18 selected major defects. Massachusetts was one of 11 states with population-based monitoring programs to contribute birth defect data. These average prevalence rates cover deliveries from 1999-2001 (CDC 2006). Massachusetts rates for 2006-2007 were significantly lower than the U.S. rates for approximately one-third of the defects and were about the same as the national estimates for the other two-thirds. The lower rates for the other defects may reflect differences in surveillance system methodology and regional variation. Also, birth defects are not reported in Massachusetts when they are prenatally diagnosed and the pregnancy is electively terminated, which would tend to result in lower rates for Massachusetts for certain defects. Based on previous studies of defects in association with prenatal screening and subsequent elective termination (Forrester, Merz et al. 1998; Cragan and Khoury 2000; Peller, Westgate et al. 2004), adjusted Massachusetts

rates that include cases currently not included in surveillance become similar to or slightly higher than the average U.S. rates. In addition, spontaneous deliveries of stillbirths ≥ 20 weeks of gestation were reported by birthing hospitals, but limited information about the stillbirth is included in the maternal record. Thus, some birth defects are not well documented and cannot be confirmed for inclusion in state surveillance.

## Cases with One Defect vs. Two or More Defects

Of all 2,585 birth defect cases (2,528 live births and 57 stillbirths) 55.1% had one defect and 44.9% had more than one (defined as having more than one defect among defects that were included in this surveillance report). Anencephaly, cleft lip with and without cleft palate, biliary atresia and hypospadias (2nd or 3rd degree) appeared more often as a single defect rather than with other defects. Birth defects which appeared more often in conjunction with other defects included holoprosencephaly, anophthalmia/microphthalmia, cloacal exstrophy, omphalocele, obstructive genitourinary defect, reduction deformity of upper or lower limbs, trisomy 18, Down syndrome, and all cardiovascular defects.

## Selected Pregnancy Outcomes

We compared selected pregnancy outcomes (C-sections, birthweight, gestational age, multiple birth and infant death) among infants born with birth defects to those born without birth defects in 2004-2005. Of infants born with birth defects, 44.1% were C-section deliveries, compared to 33.2% of non-birth defect births; 21.4% of birth defect cases were of low birthweight (<2500 grams) as opposed to 7.7% of those without a birth defect; 18.3% of infants were defined as small for gestational age, compared with 9.8% of those without a birth defect; 5.3% of infants with a birth defect died before their 1st birthday, compared to 0.4% of those without a birth defect. While numbers of infants with birth defects are relatively small, it is important to recognize the impact of these outcomes when diagnosing and treating a baby with a birth defect.

## Sex

The birth defect case prevalence was 135.6 for females and 195.1 for males per 10,000 live births. While the prevalence of most types of birth defects did not substantially differ by sex of the infant/fetus, some conditions were associated with sex. The most common defects seen in males, in order of prevalence beginning with the most prevalent defect, were hypospadias (2nd or 3rd degree), obstructive genitourinary defect, polydactyly/syndactyly, atrial septal defects (secundum and NOS), Down syndrome, and clubfoot. The most common defects seen in females were atrial septal defects (secundum and NOS), polydactyly/syndactyly, Down syndrome, obstructive genitourinary defect, clubfoot, ventricular septal defects (membranous and NOS), and valvular pulmonary stenosis.

## Plurality

Examining birth defects by plurality is important since birth defects are more common among multiple births (more than one infant), and the number of multiple births has been on the rise in Massachusetts since 1994. Plurality information obtained from reviewing the medical record differed slightly from the plurality recorded on the birth and fetal death records. Because medical record abstraction may reveal early losses not recorded at birth and is therefore more accurate, plurality from the medical record abstraction is used in this report. When using the medical record, the birth defect case prevalence was 159.7 for singletons and 301.7 for multiple births per 10,000 live births. Birth defect case prevalence from vital records for singletons was 160.5, but for multiple births it was 284.5.

Birth defects that occurred most often among multiple births (all of which occurred more often than in singleton births) were obstructive genitourinary defect, atrial septal defects (secundum and NOS), hypospadias ($2^{nd}$ and $3^{rd}$ degree), polydactyly/syndactyly, clubfoot, ventricular septal defects (membranous and NOS), Down syndrome, cleft lip with and without cleft palate, tetralogy of Fallot, and valvular pulmonary stenosis.

## Analysis of Trends

Given that the overall prevalence of birth defects in Massachusetts described in the 2004-2005 and 2006-2007 reports remained stable (overall rate, 166.8 and 166.1, respectively), preliminary trend analysis using data from this report and previous reports may be performed with the understanding that better ascertainment of cases and increased use of diagnostic technologies may be factors in any apparent change in individual birth defect rates. Trends in selected cardiovascular, orofacial and musculoskeletal birth defects for which we have large numbers of cases during each report interval allow for preliminary baseline prevalence rates to be estimated. Gastroschisis is one defect that displayed an increase between 2000 and 2007. This increase parallels the national trend and may reflect the aggressive strategies for and improvements in surveillance.

## Maternal Age

Monitoring birth defects by maternal age is important since the number of births to older mothers has been increasing over time in Massachusetts. The prevalence of birth defects varied by maternal age group. For live births only, rates per 10,000 live births were 147.4 for mothers younger than 20 years, 153.9 for those 20-24 years, 152.2 for those 25-29 years, 157.4 for those 30-34 years and 190.0 for those 35 years and older. Mothers younger than the age of 20 had the highest rate (16.4 per 10,000) of gastroschisis. This association has been shown in previous studies (Fillingham and Rankin 2008; Vu, Nobuhara et al. 2008). As expected, there was a strong association between Down syndrome and advanced maternal age. Although 44% of babies with Down syndrome were born to women under 35, the Down syndrome rate of 30.1 per 10,000 births for women 35 years and older was about three times that of the next highest Down syndrome rate (10.4

per 10,000 births occurred in the 30-34 age group) and nearly 6 times the lowest age-group rate (5.1 per 10,000 births in mothers aged 20-24 years). The pattern of higher Down syndrome rates among babies born to older women reflects the pattern of higher chromosomal defects in general among older women.

In 2006, Massachusetts had the highest proportion of births conceived through assisted reproductive technology (ART) procedures among state residents (1,291 per million residents). In 2006, Massachusetts was one of the five states with the highest frequency of ART procedures performed (Sunderam S., Chang J. et al 2009). This high frequency of ART procedures may be due in part to more complete insurance coverage in Massachusetts, and may, in fact, be an underestimate (Zhang Z, Macaluso M et al. 2009). In the U.S., ART has also been associated with some birth defects such as septal heart defects and cleft lip with and without cleft palate (Reefhuis, Honein et al. 2009).

## Maternal Race / Hispanic Ethnicity

The prevalence of birth defects varied by maternal race and Hispanic ethnicity. The age-adjusted birth defect prevalence per 10,000 live births was 163.9 for Non-Hispanic Whites, 153.4 for Non-Hispanic Blacks, 133.5 for Non-Hispanic Asians/Pacific Islanders and 176.7 for Hispanics. Due to small numbers, the rates for other races were not calculated individually. The most common defects among Hispanics included obstructive genitourinary defect, polydactyly/syndactyly, clubfoot, atrial septal defects (secundum and NOS), and Down syndrome. Among Blacks, the most common defects included polydactyly/syndactyly, obstructive genitourinary defect, Down syndrome, atrial septal defects (secundum and NOS), and hypospadias ($2^{nd}$ and $3^{rd}$ degree). The most common defects among Whites included atrial septal defects (secundum and NOS), obstructive genitourinary defect, polydactyly/syndactyly, hypospadias ($2^{nd}$ and $3^{rd}$ degree), and Down syndrome. Among Asians, the most common defects included polydactyly/syndactyly, obstructive genitourinary defect, atrial septal defects (secundum and NOS), and cleft lip with and without cleft palate. Though Asians had generally lower age-adjusted rates of chromosomal defects, all races (Whites, Blacks, Asians, Hispanics and the combined “other”) were comparable. This finding is a different from the 2004-2005 report, where Whites had about half the rate compared to Blacks, Hispanics, and “other”. Maternal birthplace (U.S. versus non-U.S.) may be a contributing factor in group differences, as women born in the U.S. had slightly higher rates than those born outside the U.S. The prevalence of birth defects in children of Hispanic women born in the U.S. Territories (including Puerto Rico, U.S. Virgin Islands and Guam) was higher than the rates of women born both in and outside the U.S, but the overlapping confidence intervals indicate a lack of statistical significance in the difference.

## Region

The birth defect rates among six Massachusetts regions in 2006-2007 were not statistically significantly different. The rates ranged from 154.8 per 10,000 in Boston to 174.7 per 10,000 in Southeast Massachusetts.

## Severity

A severity scale was developed by the Center in collaboration with our partners at Boston University and the Massachusetts General Hospital. This scale was based on the usual outcome for a specific birth defect including its typical compatibility with survival, the need for immediate treatment, the need for long-term care and the amenability of the defect to correction. We developed a new automated algorithm to classify the cases into the categories of “severe”, “serious”, “moderate”, and “mild”. Nearly 3% of cases with birth defects were classified as “severe” and most of these cases did not survive. This percentage was an underestimate of severe cases due to limited data. For example, researchers at CDC estimated that up to 80% of anencephaly cases and 50% of any neural tube defect may be electively terminated after prenatal diagnosis (Cragan and Khoury 2000). Nineteen percent of cases were affected with a “serious” birth defect. Many of these cases needed intensive medical care and planning for continuing care and long-term disability. “Moderate” birth defects comprised about 70% of the total cases. All of these needed medical follow up and many may have required a number of surgeries and extensive treatment. “Mild” birth defects comprised nearly 9% of the affected infants. These defects may or may not have required corrective treatment.

## Etiology and Pattern

The surveillance system in Massachusetts allowed for the collection of relevant etiology (cause) information. Cases with known etiology accounted for about 18% of the birth defects in Massachusetts in 2006-2007. Of the cases with known cause, “single gene” etiology accounts for almost 19%, “chromosomal” etiology accounts for almost 77% and “maternal-fetal factors” accounted for about 5%. The majority of birth defects cases in Massachusetts in 2006-2007 had an unknown etiology (82%). Among the cases with unknown etiology, the “multiple major” (2 or more major defects in different organs/body parts) pattern comprised 10%, while the “isolated” pattern comprised the majority (about 90%). Within the “isolated” pattern, three subgroups further described the occurrence of birth defects cases. These “isolated” subgroups are solitary defect, major and minors (different organ/body parts) or 2 or more defects (same organ/body part), and sequence (common primary defect with consistent, related anomalies).

## Related Public Health Resources

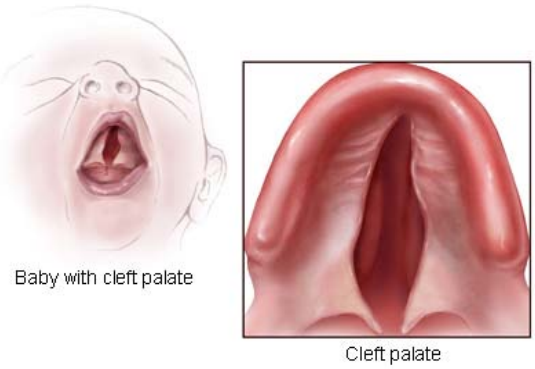
Two resources include: "Public Health Resources in Massachusetts" and a list of "Selected National Resources." Both may be found through the Massachusetts Department of Public Health website: www.mass.gov/dph/birthdefects.

## Suggested Citation

*Massachusetts Birth Defects 2006-2007*. Boston, MA: Center for Birth Defects Research and Prevention, Bureau of Family Health & Nutrition, Massachusetts Department of Public Health. October 2010.

# Chapter 1

# Introduction




*Courtesy of the Centers for Disease Control and Prevention*

## The Public Health Importance of Birth Defects

Each year in the United States, approximately 120,000 babies—1 in 33—are born with birth defects (March of Dimes 2010). Birth defects, sometimes called congenital anomalies, are abnormalities of structure, function or metabolism present before birth. These abnormalities may be fatal or may result in physical or mental disability. Several thousand defects have been identified. Some are life threatening while others are less severe.

Birth defects can lead to lifelong disability, require costly medical care and cause great distress in families. The economic, emotional and social impact on families may be catastrophic.

Although birth defects are rare when compared to other adverse birth outcomes, such as low birthweight or prematurity, they are the leading cause of death in the first year of life in the United States. Nationally, about 20% of all infant deaths result from a birth defect. In Massachusetts, 16.0% of all infant deaths were attributable to birth defects (MADPH 2007). The overall infant mortality rate for Massachusetts in 2008 was 4.9 per 1,000 births, and for the U.S. it was 6.7 per 1,000 births.

### Causes of Birth Defects

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

A single abnormal gene can cause certain birth defects. The gene may have an error in its code, a missing piece or extra genetic material, all of which can result in malformations. Other birth defects may be caused by a combination of factors such as genes interacting with environmental factors. For 70% of major birth defects, no known cause has been identified (CDC 2006). We show in Chapter 6 that about 82% of birth defect cases in Massachusetts have unknown etiology. These cases were further classified by birth defect patterns.

Researchers are looking at a wide variety of environmental exposures and risk factors as causes of birth defects. Because most of the structural development of the fetus occurs during early pregnancy, studies usually focus on the "periconceptional" period, the month before and three months after conception. For the developing pregnancy, "the environment" includes any exposure to the fetus as well as any exposure to the mother.

### Birth Defects and Folic Acid Awareness and Behavior

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

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

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

An additional and potentially more relevant indicator of folic acid intake can be multivitamin use among women who recently had a live birth. The Massachusetts Pregnancy Risk Assessment Monitoring System (PRAMS) survey of resident women who had a live birth includes questions about prenatal multivitamin use. Mothers of singletons, twins and triplets (higher order multiples were excluded) were asked about their weekly consumption of a multivitamin or prenatal vitamin the month prior to becoming pregnant. Among respondents, 36.1% reported that did not take multivitamins, 16.2% took 1-6 multivitamins per week, and 47.7% took multivitamins on a daily basis (MA PRAMS Report 2010) These data are presented in the Appendix).

**Healthy People 2010 Challenges**

Healthy People 2010 established the objectives of reducing the fetal and infant death rates by 40%, developmental disabilities rates by 50% and neural tube defect rates by 50% (DHHS 2000). Additionally, the draft version of the new Healthy People 2020 adds an objective to increase the 1-year survival rates of children born with Down syndrome. Birth defects surveillance is a critical component of the public health strategy to achieve these objectives. The active surveillance program in Massachusetts allows the Department of Public Health to monitor the extent and occurrence of birth defects within the Commonwealth. These data make it possible to identify:

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

## Birth Defects Surveillance in Massachusetts

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

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

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

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

### Economic Impact on Massachusetts

Estimating the economic impact of birth defects on the state of Massachusetts is challenging. The California Birth Defects Monitoring Program and the Metropolitan Atlanta Congenital Defects Program, using 1992 data, estimated the lifetime cost for families dealing with a baby with birth defects to be between $75,000 and $503,000 (CDC 1995). Their estimated lifetime cost for a baby born with spina bifida was $385,896 in 2007.

Adjusting for inflation, the estimated combined lifetime cost in Massachusetts for babies born with 12 major structural birth defects was $129 million in 2007 (see Technical Notes). These figures included direct costs of medical treatment, developmental services and special education, as well as indirect costs to society for lost wages due to early death or occupational limitations. There are also social and emotional impacts.

## Legislative Changes Regarding Birth Defects Surveillance

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

## Update on the Implementation of the Regulations

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

Since their enactment, the program has been working on the implementation of the regulations. Hospitals across the Commonwealth were notified of the new requirements, and the expanded post-natal reporting was implemented in these sites with the utilization of a revised Confidential Reporting and Abstracting Form (CRAF). An advisory group of obstetrical and radiological clinicians was convened to develop the specifications related to prenatal reporting, and visits were made to the Massachusetts tertiary hospitals to determine where and how birth defects are diagnosed prenatally. Implementation of the prenatal specifications will begin in November, 2010, and are expected to be fully implemented in all maternity hospitals by June, 2011. As such, these changes are not reflected in the current, 2006-2007 report. An advisory committee, comprised of patients, families, health care providers, researchers, and other interested parties, was also established. Two advisory committee meetings have been held in 2010, with one more planned for October 2010.

## The 2006-2007 Surveillance Report

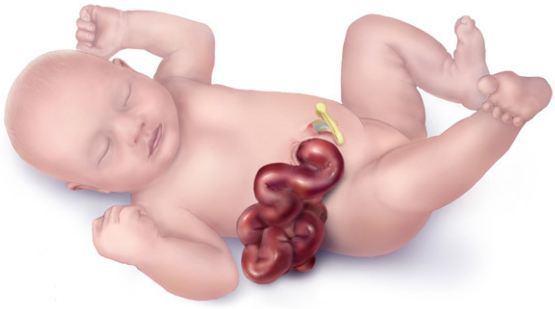
This report presents statewide data on the prevalence of birth defects in live births and stillbirths in Massachusetts during the years 2006 and 2007. The data are presented in combined form since the numbers are relatively small for individual defects. The first annual report presented Massachusetts data for birth defects for the year 1999. Our second, third, and fourth reports compiled data from 2000-2001, 2002-2003, and 2004-2005, respectively. Our ability to find and identify infants born with birth defects to Massachusetts residents has improved over time. The approximate prevalence increases of 12% from 2000-2001 to 2002-2003, and 9% from 2002-2003 to 2004-2005 are attributable to improved case ascertainment. The prevalence of all birth defects for this report is similar to the 2004-2005 report, decreasing by 0.4%. Interpretations of these data must be made with caution until a multi-year estimate establishes a stable, baseline rate.

With the introduction of the 2006-2007 report the Massachusetts Birth Defects Monitoring Program (BDMP) seems to have leveled-off in terms of defect prevalence estimates. The overall rate reported for birth defects in MA in 2006-2007 (166.1 per 10,000 live births) is virtually identical to the corresponding rate in the 2004-2005 report.

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

# Chapter 2

# Methods



Baby with gastroschisis

*Courtesy of the Centers for Disease Control and Prevention*

## Case Definition

This report summarizes data on selected birth defects present in births occurring during the calendar years 2006 through 2007 to Massachusetts residents. Cases met the following criteria:

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

## Data Collection

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

## Confidentiality

Great care was taken to protect the confidentiality of data. The Center has developed extensive procedures to guarantee the confidentiality of the data and protect the privacy of families. These procedures uphold the Center's ethical and legal obligations to safeguard confidentiality and fully comply with the strict requirements of state and federal laws.

## Data Analysis

A defect may have occurred as a single event or in combination with other defects. If the case had more than one defect within the same defect category, only one of these defects was counted in the category total. If the case had more than one defect in different defect categories, the case was listed in the total for each of these defect categories. Thus the counts in the defect categories presented in the prevalence tables represent the total number of defects and not the total number of cases with birth defects. Each case in the BDMP was linked to a Registry of Vital Records and Statistics record. In this report, maternal age and race/ethnicity are drawn from the birth certificate. Because birth certificate data are more accurate for these fields than fetal death reports, analyses of maternal age and race/ethnicity are limited to live births.

The occurrence of birth defects is reported as prevalence. Prevalence is calculated as the number of birth defect cases born during the period 2006-2007 per 10,000 live births born during the same period. Prevalence tables include the number of cases found, the estimated prevalence rate per 10,000 live births and the 95% confidence interval for that rate. The incidence (new cases) of birth defects (based upon the number of embryos conceived within a year) is not fully measured because both the total number of conceptions that occur and the number of these conceptions resulting in a defect are not known (Sever 2004).

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

## Data Limitations

1. Birth defect counts for this report are only for calendar years 2006 through 2007. Due to the small numbers of birth defects, conclusions from these results are not valid until a more extensive multi-year estimate establishes a stable, baseline rate.

2. Currently, the Massachusetts BDMP ascertains cases only at birthing hospitals, two non-birthing tertiary care centers, two non-birthing community hospitals and one specialty care hospital. Thus, defects that are not diagnosed at birth and that do not need hospitalization may be underreported (e.g., cardiac defects that are detected in an outpatient setting after the immediate newborn period).

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

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

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

6. Only diagnoses confirmed before one year of age are currently included. The frequency of diagnosed malformations can be higher among older children due to 'hidden' abnormalities such as kidney malformations or certain heart defects which are detected by accident, in conjunction or when a child is symptomatic (Holmes 1994). Many defects are also detected when diagnostic tests are performed to confirm a more severe, accompanying defect. Finally, many defects that result in developmental delays (e.g. fetal alcohol syndrome) may not be detected until those delays become evident when a child is much older.

7. In 2000, 1,318 births occurred to Massachusetts residents at out-of-state hospitals. Of these births, 68.9% occurred in Rhode Island (RI) hospitals. In order to capture data on infants with birth defects residing in the southeastern region of Massachusetts that were born or treated at RI hospitals, we began receiving hospital discharge lists and abstracting medical records on infants with birth defects at two RI hospitals. Deliveries and diagnoses that occurred in other out-of-state facilities are not included at this time.

8. There are limitations when comparing the Massachusetts BDMP data to data from other states and national estimates. Factors such as differences in population demographics, living environment , and surveillance/case ascertainment methods used by different monitoring programs may contribute to differences in the prevalence of birth defects.

## Glossary

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

# Chapter 3

# Prevalence of Birth Defects

Baby with anencephaly

*Courtesy of the Centers for Disease Control and Prevention*

## Overall Prevalence of Birth Defects

Table 1 shows the prevalence of defects for all births and for live births and stillbirths separately. Among the 155,604 live births to Massachusetts residents in 2006-2007, 2,528 had one or more structural birth defects that were ascertained by Massachusetts BDMP. In addition, 57 stillbirths were identified with a birth defect. Overall, 1.7% of births in the state (166.1 per 10,000 live births) were identified as having at least one birth defect. The majority of defects occurred in the cardiovascular (30.9%) and musculoskeletal (29.6%) categories. Figure 1 shows the percentage of reported birth defects by defect category. Cases can be included in more than one defect category.

The CDC published improved national prevalence estimates for 18 selected major defects. Massachusetts was one of 11 states with population-based monitoring programs to contribute birth defect data. These average prevalence rates cover deliveries from 1999-2001 (CDC 2006). Massachusetts rates for 2006-2007 were significantly lower than the US rates for approximately one-third of the defects and were about the same as the national estimates for the other two-thirds (see Table 2). Differences in rates may reflect variations in defect criteria between surveillance systems as well as regions. Also, in Massachusetts, birth defects are not reported when they are prenatally diagnosed and the pregnancy is electively terminated, which would tend to result in lower rates for certain defects. Spontaneous deliveries of stillbirths equal to or greater than 20 weeks of gestation were reported by birthing hospitals but limited information about the stillbirth is included in the maternal record. Thus, some birth defects are not well documented and are unable to be confirmed for inclusion in state surveillance.

Another data source to which we can compare Massachusetts rates is the neural tube defect (NTD) ascertainment project of the National Birth Defects Prevention Network at CDC. Massachusetts has submitted data quarterly since 1999. Using data from 1999-2000, researchers from CDC calculated prevalence rates for spina bifida and anencephaly, two serious birth defects that occur early in pregnancy (CDC 2004). Birth defect programs which included prenatally diagnosed cases of spina bifida that are subsequently electively terminated had a prevalence rate of 4.1. Massachusetts does not collect data on elective terminations and has a spina bifida prevalence of 1.5 per 10,000 for 2006-2007 which is over 60% lower than the national rate (see Table 2).

CDC estimates that up to 50% of pregnancies with neural tube defects and up to 80% of pregnancies with anencephaly may be electively terminated after prenatal diagnosis (Cragan and Khoury 2000). Substantial evidence from past studies have examined the effect of prenatal diagnoses and elective termination on the prevalence of various birth defects (Forrester, Merz et al. 1998). Researchers at Brigham and Women Hospital (BWH), where 11% of resident births occurred in 2004-2005, looked at past trends in elective termination in Massachusetts. For the two years 1994 and 1999, 40-80% of pregnancies prenatally diagnosed with either lethal or very severe defects were terminated (Peller, Westgate et al. 2004),

suggesting that epidemiologic studies of major malformations must include elective terminations to be complete.

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

The unadjusted 2006-2007 Massachusetts rates for anencephaly, spina bifida and trisomy 18 were lower than the national estimates. The rate for Down syndrome was similar to the national estimate (Table 2). Using 2006-2007 rates an estimated 50-72% of anencephaly cases are missed through exclusion of terminated cases. Upon adjusting the Massachusetts rate of anencephaly, one out of the three adjusted Massachusetts rates of anencephaly may be similar to the national estimate while the other two adjusted rates are lower than the national estimate. Likewise, with an estimated 29-46% of spina bifida cases missed through exclusion of terminated cases in surveillance, one of the three adjusted Massachusetts rates of spina bifida may also be similar to the national estimate while the other two are lower than the national estimate. In the case of trisomy 18, where 49-57% of cases may be missed due to elective termination, the two adjusted Massachusetts rates are similar to the national estimate. The estimated 35-37% of Down Syndrome cases missed due to elective termination based on two sources translates to adjusted Massachusetts rates exceeding the national estimate. Table 3 lists the adjusted rates. The adjusted rates of trisomy 18 and Down syndrome in Massachusetts are similar and higher, respectively, compared to national estimates, possibly reflecting the older population having children in Massachusetts.

The adjusted rates calculated here in Table 3 are based on previous studies examining data from various locations and dating as far back as 1974 through 1999. Therefore, the adjusted rates for these defects may be an underestimation, because the studies cited are not recent and may not reflect the current behaviors in prenatal diagnostic testing and elective termination decision-making processes in the state. In recent years, due to the increasing numbers of pregnancies in older women, different diagnostic testing behaviors may have developed. Further evidence of these trends may be reflected in the Massachusetts Pregnancy Risk Assessment Monitoring System (PRAMS) survey data, collected from Massachusetts resident women who had a live born infant in 2007 and 2008. Mothers of singletons, twins, and triplets (higher order multiples were excluded)

were asked whether they had discussed with healthcare providers the availability of tests to screen for birth defects or diseases during prenatal care visits. 93.2% of the respondents (N=2,592) replied that a healthcare provider had discussed the issue with them, and Black, non-Hispanics (88.8%) and Asian, non-Hispanics (86.4%) were significantly less likely than White, non-Hispanics (94.6%) to report that a healthcare provider had discussed the issue with them (MA PRAMS Report, 2010).

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

## Cases with One Defect vs. Two or More Defects

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

Among birth defect cases, 55.1% had one defect and 44.9% had more than one defect. Defects that appeared more often as a one defect rather than with other defects included anencephaly, cleft lip with and without cleft palate, biliary atresia, and hypospadias (2$^{nd}$ or 3$^{rd}$ degree). Birth defects that appeared more often in conjunction with other defects included holoprosencephaly, anophthalmia/microphthalmia, cloacal exstrophy, omphalocele, reduction deformity of lower limbs, trisomy 18, and all of the cardiovascular defects. Overall, in 2006-2007, a cardiovascular defect was almost 4 times more likely to occur with another defect.

## Selected Pregnancy Outcomes

Figure 3 compares selected pregnancy outcome characteristics among infants born with birth defects to those born without birth defects in 2006-2007 by percentage. Of infants born with birth defects, 44.1% were delivered by Cesarean, compared to 33.2% of infants born with no birth defect births; 21.4% of infants with birth defects had low birthweight (<2,500 grams) as opposed to 7.7% of those without a birth defect; 18.3% of infants were defined as small for gestational age, compared with 9.8% of those without a birth defect; 5.3% of infants with a birth defect died before their 1$^{st}$ birthday, compared to 0.4% of those without a birth defect. While numbers of infants with birth defects are relatively small, it is important to recognize the impact of these outcomes when diagnosing and treating a baby with a birth defect.

## Prevalence of Birth Defects by Sex

Table 6 presents the prevalence of birth defects by sex of the infant/fetus. The overall prevalence was 135.6 for females and 195.1 for males per 10,000 live births. While the prevalence of most types of birth defects did not differ by sex of the infant/fetus, some conditions were significantly associated with sex. Table 7 shows the most common birth defects for females and males. The most common defects seen in females were atrial septal defects (secundum and NOS), polydactyly/syndactyly, and Down syndrome. The most common defects seen in males were hypospadias (2nd or 3rd degree), obstructive genitourinary defects, and polydactyly/syndactyly. Selected birth defects by sex of infant are presented in Figure 4.

## Trend Analyses of Selected Birth Defects

The statewide data on the prevalence of birth defects in live births and stillbirths during multiple years from this surveillance report as well as three prior surveillance reports allow for some trend analysis. Data from the first few reports must be interpreted with caution since surveillance techniques were being established and executed. However, preliminary trend analysis with recent reports may be performed with the understanding that better ascertainment of cases and increased use of diagnostic technologies may have contributed to any apparent increase in birth defect rates. A comparison of selected cardiovascular birth defects rates from 2006-2007 to rates in the previous two reports suggests that most rates have remained steady or slightly increased due to better case confirmation. Similar trends are found in selected orofacial and musculoskeletal birth defects as well.

Defects such as gastroschisis and hypospadias are some of the defects of interest due to increasing national prevalence trends. Surveillance in Massachusetts and elsewhere has suggested an increased risk of gastroschisis in pregnancies among very young women (under 20), although the mechanisms for this are unknown. A recent study has shown that although younger women had comparatively larger risk of gastroschisis and greater prevalence of smoking, cigarette smoking had no effect on risk in women under 20, but smoking (possibly the duration of smoking) increased the risk in older women (Werler, Mitchel et al. manuscript). The rate of gastroschisis in Massachusetts has increased 63.8% between 2000 and 2007 (Figure 5). This increasing trend parallels the increasing national rates of gastroschisis, although a factor in this increase may be better ascertainment of cases by the surveillance system in Massachusetts. This may be due to aggressive strategies of case confirmation and improvements in surveillance, although over time, increases have been observed in other locations, around the U.S. and internationally as well.

# Table 1 Prevalence of Birth Defects, Massachusetts: 2006-2007

| Defect[1] | Count among live births | Count among stillbirths | Total Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|---|
| ***Total Cases*** | 2,528 | 57 | 2,585 | 166.1 | 159.8-172.6 |
| ***Central Nervous System: 223 cases*** | | | | | |
| Anencephaly | 6 | 2 | 8 | 0.51 | 0.22-1.01 |
| Encephalocele | 3 | 0 | 3 | 0.19 | 0.04-0.56 |
| Holoprosencephaly | 5 | 2 | 7 | 0.45 | 0.18-0.93 |
| Hydrocephaly w/o spina bifida | 33 | 5 | 38 | 2.44 | 1.73-3.35 |
| Microcephaly | 30 | 1 | 31 | 1.99 | 1.35-2.83 |
| Spina bifida w/ and w/o hydrocephaly | 21 | 2 | 23 | 1.48 | 0.94-2.22 |
| Tethered cord | 55 | 0 | 55 | 3.53 | 2.66-4.60 |
| Other CNS* | 110 | 3 | 113 | 7.26 | 5.98-8.73 |
| ***Eye: 84 cases*** | | | | | |
| Aniridia | 1 | 0 | 1 | 0.06 | 0.00-0.36 |
| Anophthalmia/microphthalmia | 16 | 0 | 16 | 1.03 | 0.59-1.67 |
| Congenital glaucoma, congenital cataract | 50 | 0 | 50 | 3.21 | 2.38-4.24 |
| Other eye* | 31 | 0 | 31 | 1.99 | 1.35-2.83 |
| ***Ear: 56 cases*** | | | | | |
| Anotia/microtia | 31 | 0 | 31 | 1.99 | 1.35-2.83 |
| Other ear* | 30 | 0 | 30 | 1.93 | 1.30-2.75 |
| ***Cardiovascular: 801 cases*** | | | | | |
| ***Anomalous Pulmonary Venous Connection*** | | | | | |
| Total/partial anomalous pulmonary venous connection | 24 | 0 | 24 | 1.54 | 0.99-2.29 |
| ***Atrioventricular Canal Defects*** | | | | | |
| ASD primum | 8 | 0 | 8 | 0.51 | 0.22-1.01 |
| Common atrium | 5 | 0 | 5 | 0.32 | 0.10-0.75 |
| Complete atrioventricular canal defect | 43 | 4 | 47 | 3.02 | 2.22-4.02 |
| Endocardial cushion (OS and NOS) | 6 | 1 | 7 | 0.45 | 0.18-0.93 |
| VSD, canal type | 4 | 1 | 5 | 0.32 | 0.10-0.75 |
| ***Conotruncal (Outlet) and Aortic Arch*** | | | | | |
| Double outlet right ventricle | 20 | 2 | 22 | 1.41 | 0.89-2.14 |
| d-Transposition of the great arteries | 32 | 2 | 34 | 2.19 | 1.51-3.05 |
| Interrupted aortic arch, type B | 8 | 0 | 8 | 0.51 | 0.22-1.01 |

# Table 1 Prevalence of Birth Defects, Massachusetts: 2006-2007

| Defect[1] | Count among live births | Count among stillbirths | Total Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|---|
| *(cont'd)* | | | | | |
| Tetralogy of Fallot w/ and w/o pulmonary atresia | 65 | 3 | 68 | 4.37 | 3.39-5.54 |
| Truncus | 6 | 0 | 6 | 0.39 | 0.14-0.84 |
| ***Ebstein Anomaly*** | | | | | |
| Ebstein anomaly | 7 | 0 | 7 | 0.45 | 0.18-0.93 |
| ***Heterotaxy (Laterality Defects)*** | | | | | |
| Heterotaxy | 15 | 2 | 17 | 1.09 | 0.64-1.75 |
| ***Left-Sided Obstruction*** | | | | | |
| Aortic valve stenosis | 29 | 0 | 29 | 1.86 | 1.25-2.68 |
| Coarctation of aorta | 62 | 0 | 62 | 3.98 | 3.05-5.11 |
| Hypoplastic left heart syndrome | 28 | 1 | 29 | 1.86 | 1.25-2.68 |
| Interrupted aortic arch (type A and NOS) | 5 | 0 | 5 | 0.32 | 0.10-0.75 |
| ***Patent Ductus Arteriosus*** | | | | | |
| Patent ductus arteriosus | 283 | 0 | 283 | 18.19 | 16.13-20.43 |
| ***Right-Sided Obstruction*** | | | | | |
| Pulmonary stenosis, valvular | 99 | 0 | 99 | 6.36 | 5.17-7.75 |
| Pulmonary valve atresia w/intact septum | 7 | 0 | 7 | 0.45 | 0.18-0.93 |
| Pulmonary valve atresia with VSD | 6 | 0 | 6 | 0.39 | 0.14-0.84 |
| Tricuspid valve atresia | 7 | 0 | 7 | 0.45 | 0.18-0.93 |
| ***Septal Defects*** | | | | | |
| ASD (secundum and NOS) | 252 | 2 | 254 | 16.32 | 14.38-18.46 |
| VSD (membranous and NOS) | 148 | 4 | 152 | 9.77 | 8.28-11.45 |
| VSD, conoventricular/malalignment | 19 | 0 | 19 | 1.22 | 0.74-1.91 |
| ***Single Ventricle and L-TGA*** | | | | | |
| L-TGA | 6 | 0 | 6 | 0.39 | 0.14-0.84 |
| Single ventricle | 12 | 0 | 12 | 0.77 | 0.40-1.35 |
| ***Other Cardiovascular*** | | | | | |
| Other cardiovascular* | 261 | 2 | 263 | 16.90 | 14.92-19.07 |
| ***Respiratory: 54 cases*** | | | | | |
| Choanal atresia | 14 | 0 | 14 | 0.90 | 0.49-1.51 |
| Lung anomalies | 21 | 1 | 22 | 1.41 | 0.89-2.14 |
| Other Respiratory* | 22 | 0 | 22 | 1.41 | 0.89-2.14 |

# Table 1 Prevalence of Birth Defects, Massachusetts: 2006-2007

| Defect[1] | Count among live births | Count among stillbirths | Total Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|---|
| *(cont'd)* | | | | | |
| ***Orofacial: 266 cases*** | | | | | |
| Cleft lip w/ and w/o cleft palate | 115 | 6 | 121 | 7.78 | 6.45-9.29 |
| Cleft palate w/o cleft lip | 73 | 2 | 75 | 4.82 | 3.79-6.04 |
| Pierre Robin sequence | 23 | 0 | 23 | 1.48 | 0.94-2.22 |
| Other orofacial* | 72 | 0 | 72 | 4.63 | 3.62-5.83 |
| ***Gastrointestinal: 250 cases*** | | | | | |
| Biliary atresia | 9 | 0 | 9 | 0.58 | 0.26-1.10 |
| Esophageal atresia/tracheoesophageal fistula | 37 | 0 | 37 | 2.38 | 1.67-3.28 |
| Hirschsprung disease | 24 | 0 | 24 | 1.54 | 0.99-2.29 |
| Rectal and large intestinal atresia/stenosis | 58 | 0 | 58 | 3.73 | 2.83-4.82 |
| Small intestinal atresia | 55 | 0 | 55 | 3.53 | 2.66-4.60 |
| Other gastrointestinal* | 102 | 2 | 104 | 6.68 | 5.46-8.10 |
| ***Genitourinary: 570 cases*** | | | | | |
| Bladder exstrophy | 1 | 0 | 1 | 0.06 | 0.00-0.36 |
| Cloacal exstrophy | 2 | 0 | 2 | 0.13 | 0.02-0.46 |
| Hypospadias, 2nd or 3rd degree | 181 | 0 | 181 | 11.63 | 10.00-13.46 |
| Obstructive genitourinary defect | 242 | 0 | 242 | 15.55 | 13.65-17.64 |
| Renal agenesis/hypoplasia | 2 | 2 | 4 | 0.26 | 0.07-0.66 |
| Other genitourinary* | 271 | 4 | 275 | 17.67 | 15.65-19.89 |
| ***Musculoskeletal: 766 cases*** | | | | | |
| Clubfoot | 181 | 7 | 188 | 12.08 | 10.42-13.94 |
| Craniosynostosis | 72 | 0 | 72 | 4.63 | 3.62-5.83 |
| Diaphragmatic hernia | 42 | 2 | 44 | 2.83 | 2.05-3.80 |
| Gastroschisis | 52 | 4 | 56 | 3.60 | 2.72-4.67 |
| Omphalocele | 19 | 2 | 21 | 1.35 | 0.84-2.06 |
| Polydactyly/syndactyly | 231 | 3 | 234 | 15.04 | 13.17-17.09 |
| Reduction deformity, lower limbs | 23 | 0 | 23 | 1.48 | 0.94-2.22 |
| Reduction deformity, upper limbs | 50 | 4 | 54 | 3.47 | 2.61-4.53 |
| Skeletal dysplasia | 17 | 1 | 18 | 1.16 | 0.69-1.83 |
| Other musculoskeletal* | 163 | 8 | 171 | 10.99 | 9.40-12.77 |
| | | | | | |

# Table 1 Prevalence of Birth Defects, Massachusetts: 2006-2007

| Defect[1] | Count among live births | Count among stillbirths | Total Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|---|
| *(cont'd)* | | | | | |
| ***Chromosomal and other Syndromes: 428 cases*** | | | | | |
| Klinefelter syndrome | 7 | 0 | 7 | 0.45 | 0.18-0.93 |
| Trisomy 13 | 12 | 4 | 16 | 1.03 | 0.59-1.67 |
| Trisomy 18 | 14 | 10 | 24 | 1.54 | 0.99-2.29 |
| Trisomy 21 (Down syndrome) | 193 | 4 | 197 | 12.66 | 10.95-14.56 |
| Turner syndrome | 10 | 5 | 15 | 0.96 | 0.54-1.59 |
| Other chromosomal syndromes/other syndromes* | 168 | 4 | 172 | 11.05 | 9.46-12.83 |
| ***Other: 59 cases*** | | | | | |
| Amniotic bands | 12 | 4 | 16 | 1.03 | 0.59-1.67 |
| Skin anomalies | 18 | 0 | 18 | 1.16 | 0.69-1.83 |
| Other, specified* | 24 | 1 | 25 | 1.61 | 1.04-2.37 |

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

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

# Table 2 Comparison of Selected Massachusetts 2006-2007 Birth Defect Rates to National Estimates

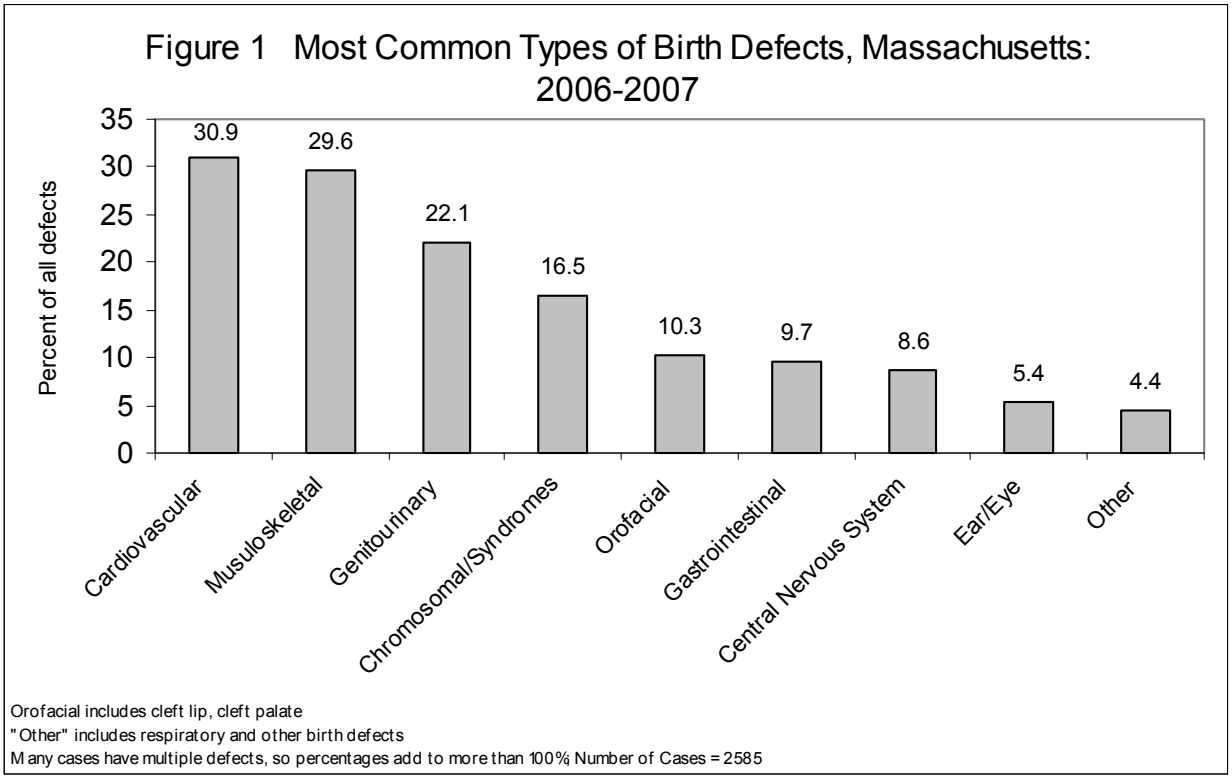
| Defect | Count | Rate per 10,000 Births MA[1] | 95% Confidence Interval | Rate per 10,000 Births US[2] | 95% Confidence Interval |
|---|---|---|---|---|---|
| Anencephaly[3] | 8 | 0.51 | 0.22-1.01 | 3.45 | 3.27 – 3.64 |
| Spina bifida[3] | 23 | 1.48 | 0.94-2.22 | 4.10 | 3.90 – 4.30 |
| Anophthalmia/microphthalmia | 16 | 1.03 | 0.59-1.67 | 2.08 | 1.90 – 2.27 |
| Truncus arteriosus (common truncus) | 6 | 0.39 | 0.14-0.84 | 0.82 | 0.71 – 0.93 |
| Transposition of the great arteries[4] | 40 | 2.57 | 1.84-3.50 | 4.73 | 4.47 – 5.00 |
| Tetralogy of Fallot | 68 | 4.37 | 3.39-5.54 | 3.92 | 3.67 – 4.17 |
| Endocardial cushion defect[5] | 66 | 4.24 | 3.28-5.40 | 4.35 | 4.10 – 4.62 |
| Hypoplastic left heart syndrome | 29 | 1.86 | 1.25-2.68 | 2.43 | 2.24 – 2.63 |
| Cleft palate without cleft lip | 75 | 4.82 | 3.79-6.04 | 6.39 | 6.08 – 6.71 |
| Cleft lip with and without cleft palate | 121 | 7.78 | 6.45-9.29 | 10.48 | 10.08 – 10.88 |
| Esophageal atresia/tracheoesophageal fistula | 37 | 2.38 | 1.67-3.28 | 2.37 | 2.18 – 2.56 |
| Rectal and large intestinal atresia/stenosis | 58 | 3.73 | 2.83-4.82 | 4.81 | 4.54 – 5.08 |
| Reduction deformity, upper limbs | 54 | 3.47 | 2.61-4.53 | 3.79 | 3.55 – 4.03 |
| Reduction deformity, lower limbs | 23 | 1.48 | 0.94-2.22 | 1.90 | 1.73 – 2.07 |
| Gastroschisis | 56 | 3.60 | 2.72-4.67 | 3.73 | 3.49 – 3.97 |
| Omphalocele | 21 | 1.35 | 0.84-2.06 | 2.09 | 1.91 – 2.27 |
| Diaphragmatic hernia | 44 | 2.83 | 2.05-3.80 | 2.94 | 2.73 – 3.15 |
| Trisomy 21 (Down syndrome) | 193 | 12.66 | 10.95-14.56 | 13.65 | 13.19 – 14.12 |
| Trisomy 13 | 16 | 1.03 | 0.59-1.67 | 1.33 | 1.18 – 1.47 |
| Trisomy 18 | 24 | 1.54 | 0.99-2.29 | 2.41 | 2.22 – 2.61 |

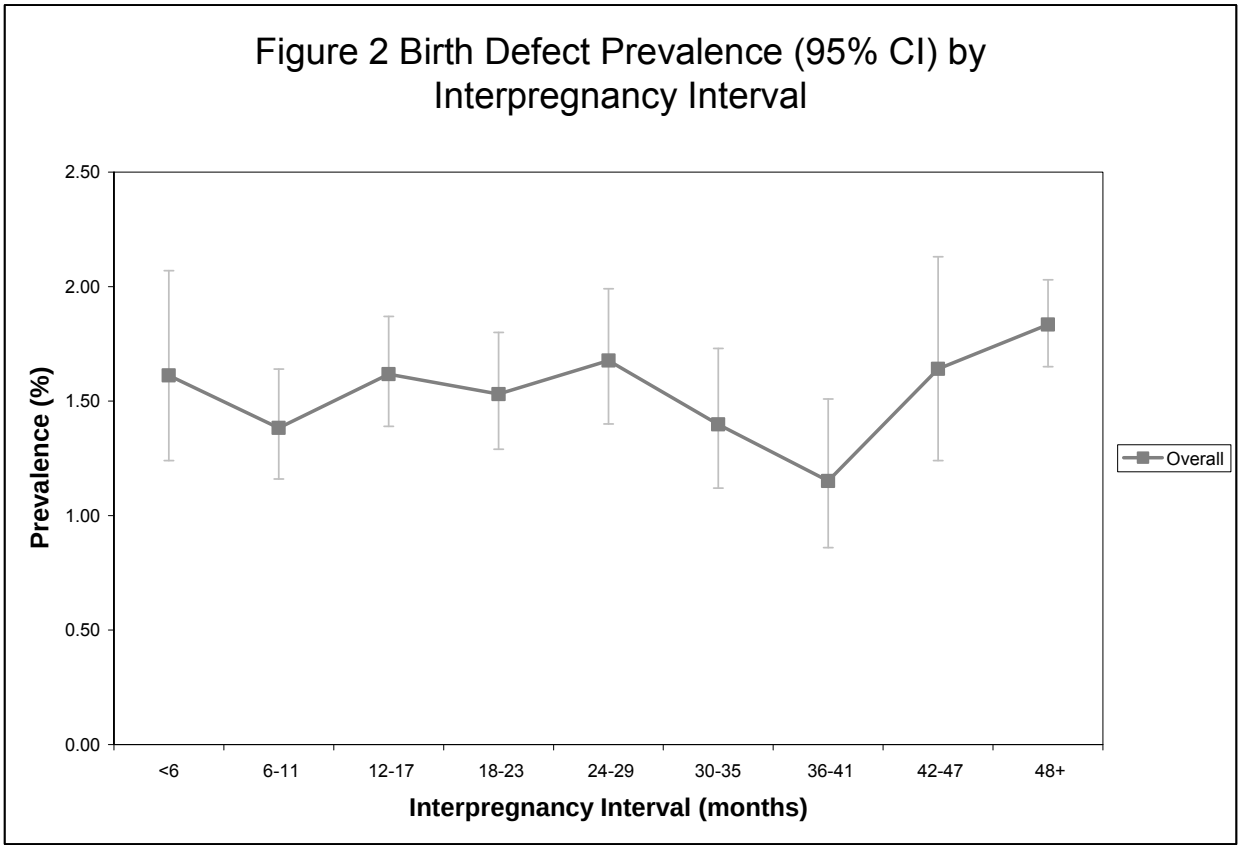
1. MA rate is based on live births and stillbirths.
2. All US rates except for anencephaly and spina bifida are from the CDC MMWR report presenting improved national prevalence estimates based on the average prevalence from 11 states including MA (CDC 2006). Rates were adjusted for race–specific distribution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

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

| Defect | MA rate | 95% Confidence Interval | Estimated Missing %[1] | Estimate Source[2] | Adjusted MA rate[3] | 95% Confidence Interval | US rate | 95% Confidence Interval |
|---|---|---|---|---|---|---|---|---|
| Anencephaly | 0.51 | 0.22 – 1.01 | 66.7 | Peller, et al. | 1.53 | 0.91 – 2.16 | 3.45[4] | 3.27 – 3.64 |
| | | | 50.0 | Cragan, et al. | 1.02 | 0.60 – 1.44 | | |
| | | | 72.0 | Forrester, et al. | 1.82 | 1.08 – 2.57 | | |
| Spina bifida | 1.48 | 0.94 – 2.22 | 45.5 | Peller, et al. | 2.72 | 2.12 – 3.31 | 4.10[4] | 3.90 – 4.30 |
| | | | 29.0 | Cragan, et al. | 2.08 | 1.63 – 2.54 | | |
| | | | 29.8 | Forrester, et al. | 2.11 | 1.65 – 2.57 | | |
| Trisomy 21 (Down syndrome) | 12.66 | 10.95 – 14.56 | 35.2 | Peller, et al. | 19.54 | 18.13 – 20.94 | 12.94[5] | 12.51–13.39 |
| | | | 37.3 | Forrester, et al. | 20.10 | 18.65 – 21.54 | | |
| Trisomy 18 | 1.54 | 0.99 – 2.29 | 56.5 | Peller, et al. | 3.54 | 2.59 – 4.49 | 2.29[5] | 2.11 – 2.48 |
| | | | 49.0 | Forrester, et al. | 3.02 | 2.21 – 3.83 | | |

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




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

## Table 4 Most Common Defects among Live Births and Stillbirths, Massachusetts: 2006 – 2007

| Defect[1] | Category | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| ASD (Secundum and NOS) | Cardiovascular | 254 | 16.32 | 14.38-18.46 |
| Obstructive Genitourinary Defect | Genitourinary | 242 | 15.55 | 13.65-17.64 |
| Polydactyly/Syndactyly | Musculoskeletal | 234 | 15.04 | 13.17-17.09 |
| Trisomy 21 (Down Syndrome) | Chromosomal and other Syndromes | 197 | 12.66 | 10.95-14.56 |
| Clubfoot | Musculoskeletal | 188 | 12.08 | 10.42-13.94 |
| Hypospadias, 2nd or 3rd Degree | Genitourinary | 181 | 11.63 | 10.00-13.46 |
| VSD (Membranous and NOS) | Cardiovascular | 152 | 9.77 | 8.28-11.45 |
| Cleft Lip w/ and w/o Cleft Palate | Orofacial | 121 | 7.78 | 6.45-9.29 |
| Pulmonary Stenosis, Valvular | Cardiovascular | 99 | 6.36 | 5.17-7.75 |
| Cleft Palate w/o Cleft Lip | Orofacial | 75 | 4.82 | 3.79-6.04 |

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

# Table 5 Cases with One Defect vs. Two or More Defects among Live Births and Stillbirths, Massachusetts: 2006-2007

| Defect[1] | Cases with one Defect | Cases with two or more defects[2] | Total Cases |
|---|---|---|---|
| ***Central Nervous System: 223 cases*** | | | |
| Anencephaly | 8 | 0 | 8 |
| Encephalocele | 1 | 2 | 3 |
| Holoprosencephaly | 0 | 7 | 7 |
| Hydrocephaly w/o spina bifida | 3 | 35 | 38 |
| Microcephaly | 9 | 22 | 31 |
| Spina bifida w/ and w/o hydrocephaly | 7 | 16 | 23 |
| Tethered cord | 17 | 38 | 55 |
| Other CNS | 40 | 73 | 113 |
| ***Eye: 84 cases*** | | | |
| Aniridia | 0 | 1 | 1 |
| Anophthalmia/microphthalmia | 1 | 15 | 16 |
| Congenital glaucoma, congenital cataract | 33 | 17 | 50 |
| Other eye | 7 | 24 | 31 |
| ***Ear: 56 cases*** | | | |
| Anotia/microtia | 14 | 17 | 31 |
| Other ear | 5 | 25 | 30 |
| ***Cardiovascular: 801 cases*** | | | |
| ***Anomalous Pulmonary Venous Connection*** | | | |
| Total/partial anomalous pulmonary venous connection | 3 | 21 | 24 |
| ***Atrioventricular Canal Defects*** | | | |
| ASD primum | 0 | 8 | 8 |
| Common atrium | 0 | 5 | 5 |
| Complete atrioventricular canal defect | 0 | 47 | 47 |
| Endocardial cushion (OS and NOS) | 0 | 7 | 7 |
| VSD, canal type | 0 | 5 | 5 |
| ***Conotruncal (Outlet) and Aortic Arch*** | | | |
| Double outlet right ventricle | 0 | 22 | 22 |
| d -Transposition of the great arteries | 12 | 22 | 34 |
| Interrupted aortic arch, type B | 0 | 8 | 8 |
| Tetralogy of Fallot w/ and w/o pulmonary atresia | 18 | 50 | 68 |
| Truncus | 0 | 6 | 6 |

# Table 5 Cases with One Defect vs. Two or More Defects among Live Births and Stillbirths, Massachusetts: 2006-2007

| Defect[1] | Cases with one Defect | Cases with two or more defects[2] | Total Cases |
|---|---|---|---|
| *(cont'd)* | | | |
| ***Ebstein Anomaly*** | | | |
| Ebstein anomaly | 2 | 5 | 7 |
| ***Heterotaxy (Laterality Defects)*** | | | |
| Heterotaxy | 1 | 16 | 17 |
| ***Left-Sided Obstruction*** | | | |
| Aortic valve stenosis | 2 | 27 | 29 |
| Coarctation of aorta | 8 | 54 | 62 |
| Hypoplastic left heart syndrome | 7 | 22 | 29 |
| Interrupted aortic arch (type A and NOS) | 0 | 5 | 5 |
| ***Patent Ductus Arteriosus*** | | | |
| Patent ductus arteriosus | 5 | 278 | 283 |
| ***Right-Sided Obstruction*** | | | |
| Pulmonary stenosis, valvular | 41 | 58 | 99 |
| Pulmonary valve atresia w/intact septum | 2 | 5 | 7 |
| Pulmonary valve atresia with VSD | 1 | 5 | 6 |
| Tricuspid valve atresia | 0 | 7 | 7 |
| ***Septal Defects*** | | | |
| ASD (Secundum and NOS) | 37 | 217 | 254 |
| VSD (Membranous and NOS) | 43 | 109 | 152 |
| VSD, conoventricular/malalignment | 1 | 18 | 19 |
| ***Other Cardiovascular*** | | | |
| Other cardiovascular | 26 | 237 | 263 |
| ***Single Ventricle and L-TGA*** | | | |
| L-TGA | 0 | 6 | 6 |
| Single ventricle | 0 | 12 | 12 |
| ***Respiratory: 54 cases*** | | | |
| Choanal atresia | 6 | 8 | 14 |
| Lung anomalies | 15 | 7 | 22 |
| Other respiratory | 7 | 15 | 22 |
| ***Orofacial: 266 cases*** | | | |
| Cleft lip w/ and w/o cleft palate | 99 | 22 | 121 |
| Cleft palate w/o cleft lip | 37 | 38 | 75 |

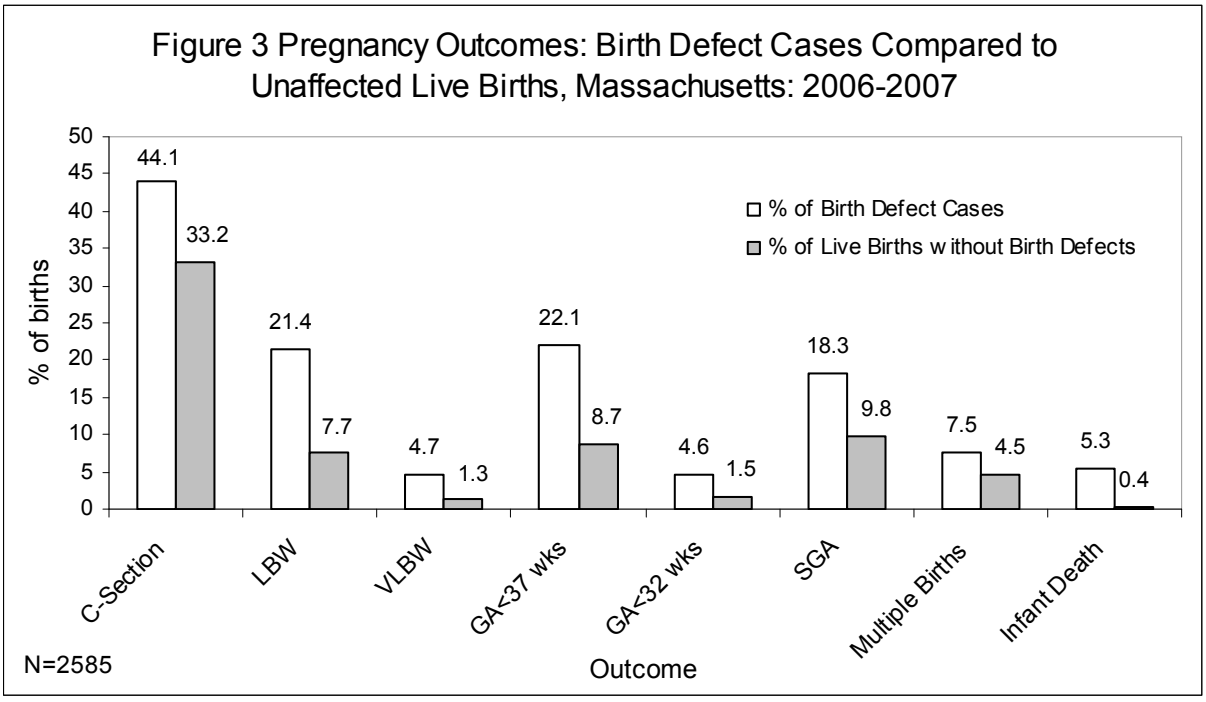
# Table 5 Cases with One Defect vs. Two or More Defects among Live Births and Stillbirths, Massachusetts: 2006-2007

| Defect[1] | Cases with one Defect | Cases with two or more defects[2] | Total Cases |
|---|---|---|---|
| *(cont'd)* | | | |
| Pierre Robin sequence | 0 | 23 | 23 |
| Other orofacial | 46 | 26 | 72 |
| ***Gastrointestinal: 250 cases*** | | | |
| Biliary atresia | 8 | 1 | 9 |
| Esophageal atresia/tracheoesophageal fistula | 8 | 29 | 37 |
| Hirschsprung disease | 20 | 4 | 24 |
| Rectal and large Intestinal atresia/stenosis | 18 | 40 | 58 |
| Small intestinal atresia | 22 | 33 | 55 |
| Other gastrointestinal | 38 | 66 | 104 |
| ***Genitourinary: 570 cases*** | | | |
| Bladder exstrophy | 1 | 0 | 1 |
| Cloacal exstrophy | 0 | 2 | 2 |
| Hypospadias, 2nd or 3rd degree | 134 | 47 | 181 |
| Obstructive genitourinary defect | 56 | 186 | 242 |
| Renal agenesis/hypoplasia | 1 | 3 | 4 |
| Other genitourinary | 94 | 181 | 275 |
| ***Musculoskeletal: 766 cases*** | | | |
| Clubfoot | 127 | 61 | 188 |
| Craniosynostosis | 63 | 9 | 72 |
| Diaphragmatic hernia | 20 | 24 | 44 |
| Gastroschisis | 42 | 14 | 56 |
| Omphalocele | 7 | 14 | 21 |
| Polydactyly/syndactyly | 164 | 70 | 234 |
| Reduction deformity, lower limbs | 3 | 20 | 23 |
| Reduction deformity, upper limbs | 20 | 34 | 54 |
| Skeletal dysplasia | 12 | 6 | 18 |
| Other musculoskeletal | 17 | 154 | 171 |
| ***Chromosomal and other Syndromes: 428 cases*** | | | |
| Klinefelter syndrome | 6 | 1 | 7 |
| Trisomy 13 | 1 | 15 | 16 |
| Trisomy 18 | 5 | 19 | 24 |

## Table 5 Cases with One Defect vs. Two or More Defects among Live Births and Stillbirths, Massachusetts: 2006-2007

| Defect[1] | Cases with one Defect | Cases with two or more defects[2] | Total Cases |
|---|---|---|---|
| *(cont'd)* | | | |
| Trisomy 21 (Down syndrome) | 69 | 127 | 196 |
| Turner syndrome | 9 | 6 | 15 |
| Other chromosomal syndromes/other syndromes | 31 | 141 | 172 |
| ***Other: 5 cases*** | | | |
| Amniotic bands | 2 | 14 | 16 |
| Skin anomalies | 12 | 6 | 18 |
| Other, specified | 2 | 23 | 25 |

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




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

# Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2006-2007

| Defect[1] | Sex | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| ***Central Nervous System*** | | | | |
| Anencephaly | Male | 4 | 0.50 | 0.14-1.29 |
| | Female | 4 | 0.53 | 0.14-1.35 |
| Encephalocele | Male | 1 | 0.13 | 0.00-0.7 |
| | Female | 2 | 0.26 | 0.03-0.95 |
| Holoprosencephaly | Male | 0 | 0.00 | 0.00-0.38 |
| | Female | 7 | 0.92 | 0.37-1.90 |
| Hydrocephaly w/o spina bifida | Male | 14 | 1.76 | 0.96-2.95 |
| | Female | 24 | 3.16 | 2.03-4.86 |
| Microcephaly | Male | 13 | 1.63 | 0.87-2.79 |
| | Female | 18 | 2.37 | 1.41-3.75 |
| Spina bifida w/ and w/o hydrocephaly | Male | 5 | 0.63 | 0.20-1.46 |
| | Female | 18 | 2.37 | 1.41-3.75 |
| Tethered cord | Male | 26 | 3.26 | 2.13-4.78 |
| | Female | 29 | 3.82 | 2.56-5.49 |
| Other CNS | Male | 61 | 7.65 | 5.85-9.83 |
| | Female | 52 | 6.85 | 5.12-8.98 |
| ***Eye*** | | | | |
| Aniridia | Male | 1 | 0.13 | 0.00-0.70 |
| | Female | 0 | 0.00 | 0.00-0.39 |
| Anophthalmia/microphthalmia | Male | 4 | 0.50 | 0.14-1.29 |
| | Female | 12 | 1.58 | 0.82-2.76 |
| Congenital glaucoma, congenital cataract | Male | 27 | 3.39 | 2.23-4.93 |
| | Female | 23 | 3.03 | 1.92-4.55 |
| Other eye | Male | 16 | 2.01 | 1.15-3.26 |
| | Female | 15 | 1.98 | 1.11-3.26 |
| ***Ear*** | | | | |
| Anotia/microtia | Male | 19 | 2.38 | 1.44-3.72 |
| | Female | 12 | 1.58 | 0.82-2.76 |
| Other ear | Male | 17 | 2.13 | 1.24-3.42 |
| | Female | 13 | 1.71 | 0.91-2.93 |

# Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2006-2007

| Defect[1] | Sex | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| ***Cardiovascular*** | | | | |
| ***Anomalous Pulmonary Venous Connection*** | | | | |
| Total/partial anomalous pulmonary venous connection | Male | 14 | 1.76 | 0.96-2.95 |
| | Female | 10 | 1.32 | 0.63-2.42 |
| ***Atrioventricular Canal Defects*** | | | | |
| ASD primum | Male | 5 | 0.63 | 0.20-1.46 |
| | Female | 3 | 0.40 | 0.08-1.16 |
| Common atrium | Male | 5 | 0.63 | 0.20-1.46 |
| | Female | 0 | 0.00 | 0.00-0.39 |
| Complete atrioventricular canal defect | Male | 18 | 2.26 | 1.34-3.57 |
| | Female | 29 | 3.82 | 2.56-5.49 |
| Endocardial cushion (OS and NOS) | Male | 2 | 0.25 | 0.03-0.91 |
| | Female | 5 | 0.66 | 0.21-1.54 |
| VSD, canal type | Male | 0 | 0.00 | 0.00-0.38 |
| | Female | 5 | 0.66 | 0.21-1.54 |
| ***Conotruncal (Outlet) and Aortic Arch*** | | | | |
| Double outlet right ventricle | Male | 12 | 1.51 | 0.78-2.63 |
| | Female | 10 | 1.32 | 0.63-2.42 |
| d-Transposition of the great arteries | Male | 21 | 2.64 | 1.63-4.03 |
| | Female | 13 | 1.71 | 0.91-2.93 |
| Interrupted aortic arch, type B | Male | 5 | 0.63 | 0.2-1.46 |
| | Female | 3 | 0.40 | 0.08-1.16 |
| Tetralogy of Fallot w/ and w/o pulmonary atresia | Male | 31 | 3.89 | 2.64-5.52 |
| | Female | 37 | 4.87 | 3.43-6.72 |
| Truncus | Male | 2 | 0.25 | 0.03-0.91 |
| | Female | 4 | 0.53 | 0.14-1.35 |
| ***Ebstein Anomaly*** | | | | |
| Ebstein anomaly | Male | 4 | 0.50 | 0.14-1.29 |
| | Female | 3 | 0.40 | 0.08-1.16 |
| ***Heterotaxy (Laterality Defects)*** | | | | |
| Heterotaxy | Male | 11 | 1.38 | 0.69-2.47 |
| | Female | 6 | 0.79 | 0.29-1.72 |

# Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2006-2007

| Defect[1] | Sex | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| ***Left-Sided Obstruction*** | | | | |
| Aortic valve stenosis | Male | 20 | 2.51 | 1.53-3.88 |
| | Female | 9 | 1.19 | 0.54-2.25 |
| Coarctation of aorta | Male | 38 | 4.77 | 3.37-6.54 |
| | Female | 24 | 3.16 | 2.03-4.70 |
| Hypoplastic left heart syndrome | Male | 14 | 1.76 | 0.96-2.95 |
| | Female | 15 | 1.98 | 1.11-3.26 |
| Interrupted aortic arch (type A and NOS) | Male | 2 | 0.25 | 0.03-0.91 |
| | Female | 3 | 0.40 | 0.08-1.16 |
| ***Patent Ductus Arteriosus*** | | | | |
| Patent ductus arteriosus | Male | 145 | 18.19 | 15.35-21.41 |
| | Female | 138 | 18.18 | 15.27-21.48 |
| ***Right-Sided Obstruction*** | | | | |
| Pulmonary stenosis, valvular | Male | 46 | 5.77 | 4.23-7.7 |
| | Female | 53 | 6.98 | 5.23-9.13 |
| Pulmonary valve atresia w/intact septum | Male | 5 | 0.63 | 0.2-1.46 |
| | Female | 2 | 0.26 | 0.03-0.95 |
| Pulmonary valve atresia with VSD | Male | 5 | 0.63 | 0.20-1.46 |
| | Female | 1 | 0.13 | 0.00-0.73 |
| Tricuspid valve atresia | Male | 0 | 0.00 | 0.00-0.38 |
| | Female | 7 | 0.92 | 0.37-2.14 |
| ***Septal Defects*** | | | | |
| ASD (secundum and NOS) | Male | 128 | 16.06 | 13.40-19.1 |
| | Female | 126 | 16.60 | 13.83-19.76 |
| VSD (membranous and NOS) | Male | 79 | 9.91 | 7.85-12.35 |
| | Female | 73 | 9.62 | 7.54-12.09 |
| VSD, conoventricular/malalignment | Male | 8 | 1.00 | 0.43-1.98 |
| | Female | 11 | 1.45 | 0.72-2.59 |
| ***Single Ventricle and L-TGA*** | | | | |
| L-TGA | Male | 4 | 0.50 | 0.14-1.29 |
| | Female | 2 | 0.26 | 0.03-0.95 |

# Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2006-2007

| Defect[1] | Sex | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| Single ventricle | Male | 5 | 0.63 | 0.20-1.46 |
| | Female | 7 | 0.92 | 0.37-1.90 |
| ***Other Cardiovascular*** | | | | |
| Other cardiovascular | Male | 147 | 18.45 | 15.58-21.68 |
| | Female | 116 | 15.28 | 12.63-18.33 |
| ***Respiratory*** | | | | |
| Choanal atresia | Male | 5 | 0.63 | 0.20-1.46 |
| | Female | 9 | 1.19 | 0.54-2.25 |
| Lung anomalies | Male | 9 | 1.13 | 0.52-2.14 |
| | Female | 13 | 1.71 | 0.91-2.93 |
| Other respiratory | Male | 10 | 1.25 | 0.60-2.31 |
| | Female | 12 | 1.58 | 0.82-2.76 |
| ***Orofacial*** | | | | |
| Cleft lip w/ and w/o cleft palate | Male | 69 | 8.66 | 6.74-10.96 |
| | Female | 52 | 6.85 | 5.12-8.98 |
| Cleft palate w/o cleft lip | Male | 40 | 5.02 | 3.59-6.83 |
| | Female | 35 | 4.61 | 3.21-6.41 |
| Pierre Robin sequence | Male | 14 | 1.76 | 0.96-2.95 |
| | Female | 9 | 1.19 | 0.54-2.25 |
| Other orofacial | Male | 41 | 5.14 | 3.69-6.98 |
| | Female | 31 | 4.08 | 2.77-5.80 |
| ***Gastrointestinal*** | | | | |
| Biliary atresia | Male | 4 | 0.50 | 0.14-1.29 |
| | Female | 5 | 0.66 | 0.21-1.54 |
| Esophageal atresia/tracheoesophageal fistula | Male | 22 | 2.76 | 1.73-4.18 |
| | Female | 15 | 1.98 | 1.11-3.26 |
| Hirschsprung disease | Male | 19 | 2.38 | 1.44-3.72 |
| | Female | 5 | 0.66 | 0.21-1.54 |
| Rectal and large intestinal atresia/stenosis | Male | 35 | 4.39 | 3.06-6.11 |
| | Female | 23 | 3.03 | 1.92-4.55 |

# Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2006-2007

| Defect[1] | Sex | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| Small intestinal atresia | Male | 25 | 3.14 | 2.03-4.63 |
| | Female | 30 | 3.95 | 2.67-5.64 |
| Other gastrointestinal | Male | 58 | 7.28 | 5.53-9.41 |
| | Female | 46 | 6.06 | 4.44-8.08 |
| ***Genitourinary*** | | | | |
| Bladder exstrophy | Male | 1 | 0.13 | 0-0.7 |
| | Female | 0 | 0.00 | 0.00-0.39 |
| Cloacal exstrophy | Male | 0 | 0.00 | 0.00-0.38 |
| | Female | 2 | 0.26 | 0.03-0.95 |
| Hypospadias, 2nd or 3rd degree | Male | 181 | 22.71 | 19.52-26.27 |
| | Female | 0 | 0.00 | 0.00-0.39 |
| Obstructive genitourinary defect | Male | 166 | 20.83 | 17.78-24.25 |
| | Female | 76 | 10.01 | 7.89-12.53 |
| Renal agenesis/hypoplasia | Male | 2 | 0.25 | 0.03-0.91 |
| | Female | 2 | 0.26 | 0.03-0.95 |
| Other genitourinary | Male | 205 | 25.72 | 22.32-29.50 |
| | Female | 69 | 9.09 | 7.07-11.50 |
| ***Musculoskeletal*** | | | | |
| Clubfoot | Male | 112 | 14.05 | 11.57-16.91 |
| | Female | 75 | 9.88 | 7.77-12.39 |
| Craniosynostosis | Male | 45 | 5.65 | 4.12-7.56 |
| | Female | 27 | 3.56 | 2.34-5.18 |
| Diaphragmatic hernia | Male | 33 | 4.14 | 2.85-5.82 |
| | Female | 11 | 1.45 | 0.72-2.59 |
| Gastroschisis | Male | 26 | 3.26 | 2.13-4.78 |
| | Female | 29 | 3.82 | 2.56-5.49 |
| Omphalocele | Male | 14 | 1.76 | 0.96-2.95 |
| | Female | 7 | 0.92 | 0.37-1.90 |
| Polydactyly/syndactyly | Male | 138 | 17.32 | 14.55-20.46 |
| | Female | 98 | 12.91 | 10.48-15.73 |

# Table 6 Prevalence of Birth Defects by Sex of Infant among Live Births and Stillbirths, Massachusetts: 2006-2007

| Defect[1] | Sex | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| Reduction deformity, lower limbs | Male | 10 | 1.25 | 0.6-2.31 |
| | Female | 13 | 1.71 | 0.91-2.93 |
| Reduction deformity, upper limbs | Male | 23 | 2.89 | 1.83-4.33 |
| | Female | 31 | 4.08 | 2.77-5.80 |
| Skeletal dysplasia | Male | 10 | 1.25 | 0.6-2.31 |
| | Female | 8 | 1.05 | 0.46-2.08 |
| Other musculoskeletal | Male | 94 | 11.79 | 9.53-14.43 |
| | Female | 77 | 10.14 | 8.01-12.68 |
| ***Chromosomal and other Syndromes*** | | | | |
| Klinefelter syndrome | Male | 7 | 0.88 | 0.35-1.81 |
| | Female | 0 | 0.00 | 0.00-0.39 |
| Trisomy 13 | Male | 6 | 0.75 | 0.28-1.64 |
| | Female | 10 | 1.32 | 0.63-2.42 |
| Trisomy 18 | Male | 5 | 0.63 | 0.20-1.46 |
| | Female | 19 | 2.50 | 1.51-3.91 |
| Trisomy 21 (Down syndrome) | Male | 117 | 14.68 | 12.14-17.59 |
| | Female | 80 | 10.54 | 8.36-13.12 |
| Turner syndrome | Male | 3 | 0.38 | 0.08-1.10 |
| | Female | 12 | 1.58 | 0.82-2.76 |
| Other chromosomal syndromes/other syndromes | Male | 89 | 11.17 | 8.97-13.74 |
| | Female | 84 | 11.07 | 8.83-13.70 |
| ***Other*** | | | | |
| Amniotic bands | Male | 3 | 0.38 | 0.08-1.10 |
| | Female | 12 | 1.58 | 0.82-2.76 |
| Skin anomalies | Male | 10 | 1.25 | 0.60-2.31 |
| | Female | 8 | 1.05 | 0.46-2.08 |
| Other, specified | Male | 14 | 1.76 | 0.96-2.95 |
| | Female | 11 | 1.45 | 0.72-2.59 |

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

# Table 7 Most Common Defects by Sex of Live Births and Stillbirths, Massachusetts: 2006-2007

| Defect[1] | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|
| **FEMALE** | | | |
| ASD (secundum and NOS) | 126 | 16.60 | 13.83-19.76 |
| Polydactyly/syndactyly | 98 | 12.91 | 10.48-15.73 |
| Trisomy 21 (Down syndrome) | 80 | 10.54 | 8.36-13.12 |
| Obstructive genitourinary defect | 76 | 10.01 | 7.89-12.53 |
| Clubfoot | 72 | 9.49 | 7.42-11.95 |
| VSD (membranous and NOS) | 73 | 9.62 | 7.54-12.09 |
| Pulmonary stenosis, valvular | 53 | 6.98 | 5.23-9.13 |
| Cleft lip w/ and w/o cleft palate | 48 | 6.32 | 4.66-8.38 |
| Tetralogy of Fallot w/ and w/o pulmonary atresia | 34 | 4.48 | 3.10-6.26 |
| Cleft palate w/o cleft lip | 34 | 4.48 | 3.10-6.26 |
| **MALE** | | | |
| Hypospadias, 2nd or 3rd degree | 181 | 22.71 | 19.52-26.27 |
| Obstructive genitourinary defect | 166 | 20.83 | 17.78-24.25 |
| Polydactyly/syndactyly | 133 | 16.69 | 13.97-19.78 |
| ASD (secundum and NOS) | 128 | 16.06 | 13.40-19.10 |
| Trisomy 21 (Down syndrome) | 113 | 14.18 | 11.69-17.05 |
| Clubfoot | 109 | 13.68 | 11.23-16.50 |
| VSD (membranous and NOS) | 79 | 9.91 | 7.85-12.35 |
| Cleft lip w/ and w/o cleft palate | 67 | 8.41 | 6.52-10.68 |
| Pulmonary stenosis, valvular | 46 | 5.77 | 4.23-7.70 |
| Craniosynostosis | 45 | 5.65 | 4.12-7.56 |

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

Figure 4 Prevalence of Selected Birth Defects by Sex of Infant Among Live Births and Still Births, Massachusetts: 2006-2007

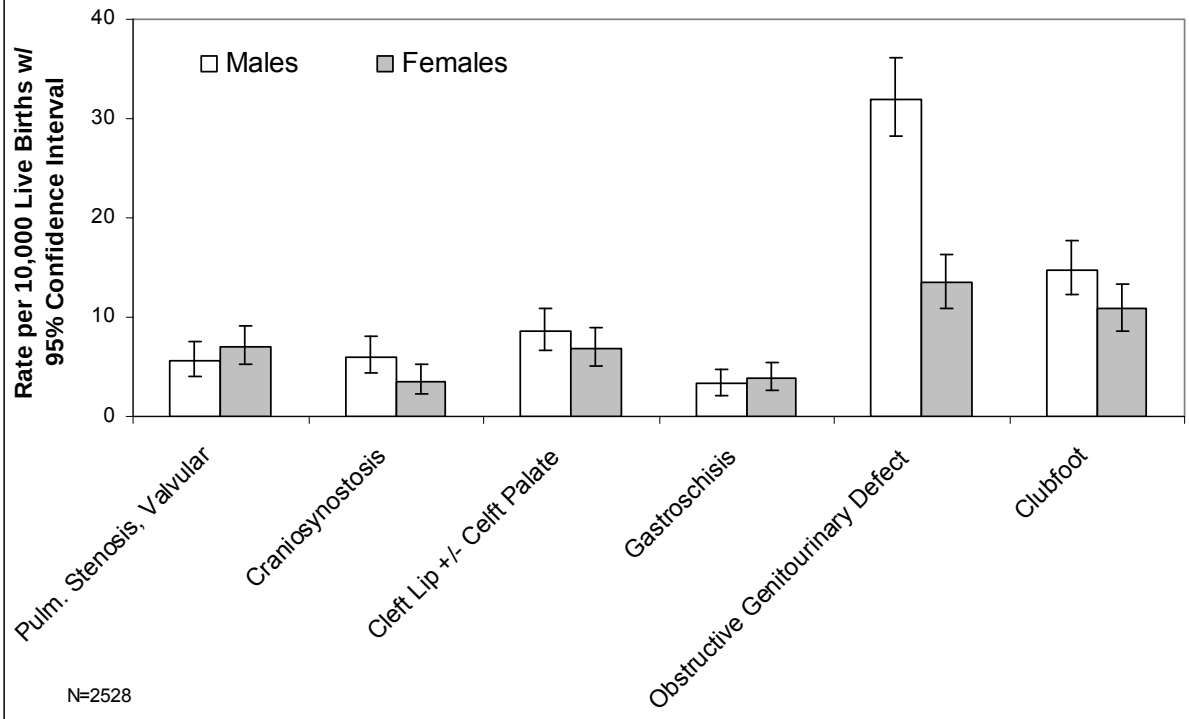
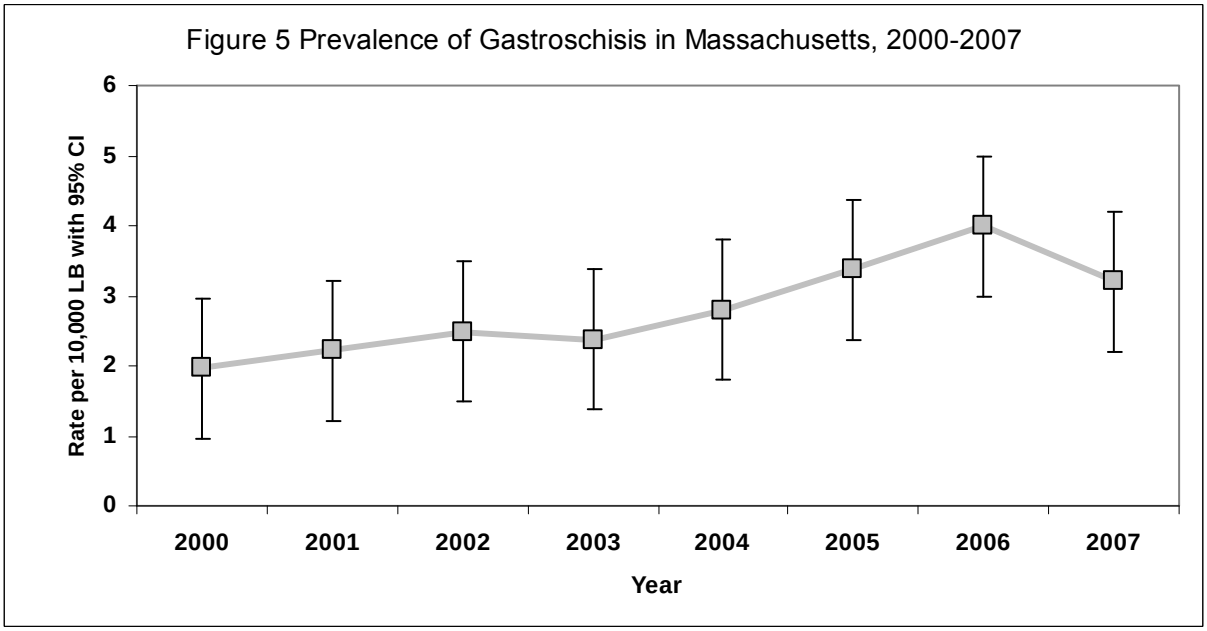



Figure 5 Prevalence of Gastroschisis in Massachusetts, 2000-2007



# Chapter 4

# Prevalence of Birth Defects by Plurality and Maternal Age

Baby with cleft lip

*Courtesy of the Centers for Disease Control and Prevention*

## Plurality

Table 8 shows the distribution of birth defects by plurality. Plurality information obtained from reviewing the medical record differed slightly from the plurality recorded on the birth and fetal death records. Because medical record abstraction may reveal early losses not recorded at birth and is therefore more accurate, plurality from the medical record abstraction is used in this report. When using the medical record, the birth defect case prevalence was 159.7 for singletons and 301.7 for multiple births (more than one infant) per 10,000 live births. Plurality from vital records for singletons was 160.5 but for multiple births was 284.5. While multiple births comprised 4.5% of all live births, they comprised 8.1% of birth defects cases among live births (see Figure 6). Birth defects that occurred most often in multiple births (all of which occurred more often than in singleton births) were obstructive genitourinary defect, atrial septal defects (secundum and NOS), hypospadias (2nd and 3rd degree), polydactyly/syndactyly, clubfoot, ventricular septal defects (membranous and NOS), Down syndrome, cleft lip with and without cleft palate, tetralogy of Fallot, and valvular pulmonary stenosis. Figure 7 presents rates for selected birth defects for singletons and multiples. Table 9 lists the most common defects among singletons and multiples. Examining birth defects by plurality is important since the rate of multiple births has been increasing in Massachusetts since 1994.

## Maternal Age

The prevalence of birth defects varied by maternal age. For live births only, rates per 10,000 live births were 147.4 for mothers younger than 20 years, 153.9 for those 20-24 years, 152.2 for those 25-29 years, 157.4 for those 30-34 years and 190.0 for those 35 years and older. Based on the calculation of population attributable fraction, 42.6% of Down syndrome cases in Massachusetts may be due to births in women greater than or equal to 35 years old. Table 10 shows the rates for birth defects by maternal age.

As expected, there was a strong association between Down syndrome and advanced maternal age (see Figure 8). The Down syndrome rate of 30.1 per 10,000 births for women 35 years and older was more than two times that of any other maternal age group. The pattern of higher Down syndrome rates among older women reflects the pattern of higher chromosomal defects in general among older women. Figure 9 shows the increase of chromosomal defects as maternal age increases; the rate of chromosomal defects among women in the 35+ age group is significantly higher than all other age groups. The proportion of all birth defects that are chromosomal defects was higher in the 35+ age group than in other age groups.

Figure 10 shows that younger mothers (aged 19 and under) had the highest rate (16.4) of gastroschisis cases. This association has been shown in previous studies (Forrester and Merz 1999). Mothers younger than 25 years of age had infants with higher rates of gastroschisis and diaphragmatic hernia than other age groups. Older mothers had higher rates for many defects including Down

syndrome and ventricular septal defects (membranous and NOS). While results for other defects also differed by age group, the small numbers from two years of surveillance were not sufficient for interpretation.

Table 11 displays the most common birth defects for live births by maternal age groups. Obstructive genitourinary defect and polydactyly/syndactyly were two of the five most frequently occurring defects common to all maternal age groups. Hypospadias (2nd or 3rd degree) and clubfoot were among the five most common birth defects in every age group, with the exception of maternal age greater than 35 years.

Monitoring birth defects by maternal age is important since the number of births to older mothers has been increasing over time in Massachusetts. Birth rates for women ages 30+ have increased steadily from 1980 to 2006. The number of births to women ages 30+ surpassed the number of births to women below age 30 in 1996. Recent data suggest that the numbers of births to women ages 30+ peaked in 2002 and has slightly decreased since, whereas the numbers of births to women ages below 30 reached a low point in 2004 and may be slightly increasing so that the numbers of births to women in the two age groups are converging (MADPH 2009). The number of births to women ages 35 in 2007 is almost 60% greater than the number in 1989, and likewise, the number of births to women under age 35 in 2006 is less than 25% of the number in 1989 (MassCHIP v3.0 r324).

The percentage of women giving birth in the state who are aged of 35 or over has doubled from 11.4% in 1989 to 22.7% in 2007. In addition, multiple births occurring in mothers aged 35 or over also doubled (3.4% to 6.8%) between 1989 and 2007 while multiple births to mothers under the age under 35 decreased slightly (18.3% to 12.6% from 1989 to 2007, respectively) (MassCHIP v3.0 r324).

A factor in both the increased percentage of women giving birth at an age of 35 or over and the disparity of multiple births among these older women giving birth may be the use of assisted reproductive technology (ART). Between 1997 to 2004, a steady 2% of Massachusetts births resulted from ART (CDC 2009) though this may be an underestimate (Zhang Z, Macaluso M et al). According to the CDC, Massachusetts ranked 5th highest in number of ART procedures performed in 2006 (8,305), after California, New York, Illinois and New Jersey. In 2006 Massachusetts had the highest ratio of the number of ART procedures among state residents at 1,291 per million residents. (Sunderam S., Chang J. et al 2009). Of the infants born in 2005 as a result of ART procedures, 43.5% were born in multiple birth deliveries (Wright, Chang et al. 2008). Figure 11 shows the disparity in percent of births that are single versus multiple from ART births and non-ART births in Massachusetts (CDC 2009). The vast majority of non-ART births are single deliveries, whereas almost half of ART births are multiple deliveries. ART poses many risks associated with multiple births that may lead to adverse maternal and infant outcomes such as low birthweight and preterm delivery. ART has also

been associated with some birth defects such as septal heart defects and cleft lip with or without cleft palate (Reefhuis, Honein et al. 2009).

# Table 8 Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2006-2007

| Defect[2] | Plurality | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| ***Central Nervous System*** | | | | |
| Anencephaly | Singleton | 8 | 0.54 | 0.23-1.06 |
| | Multiple | 0 | 0.00 | 0.00-5.30 |
| Encephalocele | Singleton | 3 | 0.20 | 0.04-0.59 |
| | Multiple | 0 | 0.00 | 0.00-5.30 |
| Holoprosencephaly | Singleton | 7 | 0.47 | 0.19-0.97 |
| | Multiple | 0 | 0.00 | 0.00-5.30 |
| Hydrocephaly w/o spina bifida | Singleton | 33 | 2.22 | 1.53-3.12 |
| | Multiple | 5 | 7.18 | 2.33-16.76 |
| Microcephaly | Singleton | 30 | 2.02 | 1.36-2.88 |
| | Multiple | 1 | 1.44 | 0.04-8.01 |
| Spina bifida w/ and w/o hydrocephaly | Singleton | 20 | 1.35 | 0.82-2.08 |
| | Multiple | 3 | 4.31 | 0.89-12.60 |
| Tethered cord | Singleton | 50 | 3.36 | 2.50-4.43 |
| | Multiple | 5 | 7.18 | 2.33-16.76 |
| Other CNS | Singleton | 103 | 6.93 | 5.66-8.40 |
| | Multiple | 10 | 14.37 | 6.89-26.42 |
| ***Eye*** | | | | |
| Aniridia | Singleton | 1 | 0.07 | 0.00-0.37 |
| | Multiple | 0 | 0.00 | 0.00-5.30 |
| Anophthalmia/microphthalmia | Singleton | 16 | 1.08 | 0.62-1.75 |
| | Multiple | 0 | 0.00 | 0.00-5.30 |
| Congenital glaucoma, congenital cataract | Singleton | 47 | 3.16 | 2.32-4.20 |
| | Multiple | 3 | 4.31 | 0.89-12.60 |
| Other eye | Singleton | 30 | 2.02 | 1.36-2.88 |
| | Multiple | 1 | 1.44 | 0.04-8.01 |
| ***Ear*** | | | | |
| Anotia/microtia | Singleton | 28 | 1.88 | 1.25-2.72 |
| | Multiple | 3 | 4.31 | 0.89-12.60 |
| Other ear | Singleton | 28 | 1.88 | 1.25-2.72 |
| | Multiple | 2 | 2.87 | 0.35-10.38 |

# Table 8 Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2006-2007

| Defect[2] | Plurality | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| ***Cardiovascular*** | | | | |
| ***Anomalous Pulmonary Venous Connection*** | | | | |
| Total/partial anomalous pulmonary venous connection | Singleton | 21 | 1.41 | 0.87-2.16 |
| | Multiple | 3 | 4.31 | 0.89-12.60 |
| ***Atrioventricular Canal Defects*** | | | | |
| ASD primum | Singleton | 7 | 0.47 | 0.19-0.97 |
| | Multiple | 1 | 1.44 | 0.04-8.01 |
| Common atrium | Singleton | 4 | 0.27 | 0.07-0.69 |
| | Multiple | 1 | 1.44 | 0.04-8.01 |
| Complete atrioventricular canal defect | Singleton | 45 | 3.03 | 2.21-4.05 |
| | Multiple | 2 | 2.87 | 0.35-10.38 |
| Endocardial cushion (OS and NOS) | Singleton | 5 | 0.34 | 0.11-0.78 |
| | Multiple | 2 | 2.87 | 0.35-10.38 |
| VSD, canal type | Singleton | 5 | 0.34 | 0.11-0.78 |
| | Multiple | 0 | 0.00 | 0.00-5.30 |
| ***Conotruncal (Outlet) and Aortic Arch*** | | | | |
| Double outlet right ventricle | Singleton | 20 | 1.35 | 0.82-2.08 |
| | Multiple | 2 | 2.87 | 0.35-10.38 |
| d-Transposition of the great arteries | Singleton | 33 | 2.22 | 1.53-3.12 |
| | Multiple | 1 | 1.44 | 0.04-8.01 |
| Interrupted aortic arch, type B | Singleton | 7 | 0.47 | 0.19-0.97 |
| | Multiple | 1 | 1.44 | 0.04-8.01 |
| Tetralogy of Fallot w/ and w/o pulmonary atresia | Singleton | 57 | 3.83 | 2.90-4.97 |
| | Multiple | 10 | 14.37 | 6.89-26.42 |
| Truncus | Singleton | 4 | 0.27 | 0.07-0.69 |
| | Multiple | 2 | 2.87 | 0.35-10.38 |
| ***Ebstein Anomaly*** | | | | |
| Ebstein anomaly | Singleton | 7 | 0.47 | 0.19-0.97 |
| | Multiple | 0 | 0.00 | 0.00-5.30 |
| ***Laterality Defects*** | | | | |
| Heterotaxy | Singleton | 17 | 1.14 | 0.67-1.83 |
| | Multiple | 0 | 0.00 | 0.00-5.30 |

# Table 8 Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2006-2007

| Defect[2] | Plurality | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| ***Left-Sided Obstruction*** | | | | |
| Aortic valve stenosis | Singleton | 26 | 1.75 | 1.14-2.56 |
| | Multiple | 3 | 4.31 | 0.89-12.60 |
| Coarctation of aorta | Singleton | 58 | 3.90 | 2.96-5.04 |
| | Multiple | 4 | 5.75 | 1.57-14.71 |
| Hypoplastic left heart syndrome | Singleton | 26 | 1.75 | 1.14-2.56 |
| | Multiple | 3 | 4.31 | 0.89-12.60 |
| Interrupted aortic arch (type A and NOS) | Singleton | 5 | 0.34 | 0.11-0.78 |
| | Multiple | 0 | 0.00 | 0.00-5.30 |
| ***Patent Ductus Arteriosus*** | | | | |
| Patent ductus arteriosus | Singleton | 267 | 17.96 | 15.87-20.25 |
| | Multiple | 16 | 22.99 | 13.14-37.33 |
| ***Right-Sided Obstruction*** | | | | |
| Pulmonary stenosis, valvular | Singleton | 90 | 6.05 | 4.87-7.44 |
| | Multiple | 9 | 12.93 | 5.91-24.55 |
| Pulmonary valve atresia w/intact septum | Singleton | 7 | 0.47 | 0.19-0.97 |
| | Multiple | 0 | 0.00 | 0.00-5.30 |
| Pulmonary valve atresia with VSD | Singleton | 6 | 0.40 | 0.15-0.88 |
| | Multiple | 0 | 0.00 | 0.00-5.30 |
| Tricuspid valve atresia | Singleton | 4 | 0.27 | 0.07-0.69 |
| | Multiple | 3 | 4.31 | 0.89-12.60 |
| ***Septal Defects*** | | | | |
| ASD (secundum and NOS) | Singleton | 229 | 15.41 | 13.48-17.54 |
| | Multiple | 25 | 35.92 | 23.25-53.02 |
| VSD (membranous and NOS) | Singleton | 138 | 9.28 | 7.80-10.97 |
| | Multiple | 14 | 20.11 | 11.00-33.75 |
| VSD, conoventricular/malalignment | Singleton | 19 | 1.28 | 0.77-2.00 |
| | Multiple | 0 | 0.00 | 0.00-5.30 |
| ***Single Ventricle and L-TGA*** | | | | |
| L-TGA | Singleton | 5 | 0.34 | 0.11-0.78 |
| | Multiple | 1 | 1.44 | 0.04-8.01 |

# Table 8 Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2006-2007

| Defect[2] | Plurality | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| Single ventricle | Singleton | 10 | 0.67 | 0.32-1.24 |
| | Multiple | 2 | 2.87 | 0.35-10.38 |
| ***Other Cardiovascular*** | | | | |
| Other cardiovascular | Singleton | 236 | 15.88 | 13.92-18.04 |
| | Multiple | 27 | 38.79 | 25.56-56.44 |
| ***Respiratory*** | | | | |
| Choanal atresia | Singleton | 13 | 0.87 | 0.47-1.50 |
| | Multiple | 1 | 1.44 | 0.04-8.01 |
| Lung anomalies | Singleton | 19 | 1.28 | 0.77-2.00 |
| | Multiple | 3 | 4.31 | 0.89-12.60 |
| Other respiratory | Singleton | 21 | 1.41 | 0.87-2.16 |
| | Multiple | 1 | 1.44 | 0.04-8.01 |
| ***Orofacial*** | | | | |
| Cleft lip w/ and w/o cleft palate | Singleton | 108 | 7.27 | 5.96-8.77 |
| | Multiple | 13 | 18.68 | 9.95-31.94 |
| Cleft palate w/o cleft lip | Singleton | 71 | 4.78 | 3.73-6.02 |
| | Multiple | 4 | 5.75 | 1.57-14.71 |
| Pierre Robin sequence | Singleton | 23 | 1.55 | 0.98-2.32 |
| | Multiple | 0 | 0.00 | 0.00-5.30 |
| Other orofacial | Singleton | 69 | 4.64 | 3.61-5.87 |
| | Multiple | 3 | 4.31 | 0.89-12.60 |
| ***Gastrointestinal*** | | | | |
| Biliary atresia | Singleton | 9 | 0.61 | 0.28-1.15 |
| | Multiple | 0 | 0.00 | 0.00-5.30 |
| Esophageal atresia/tracheoesophageal fistula | Singleton | 33 | 2.22 | 1.53-3.12 |
| | Multiple | 4 | 5.75 | 1.57-14.71 |
| Hirschsprung disease | Singleton | 24 | 1.61 | 1.03-2.40 |
| | Multiple | 0 | 0.00 | 0.00-5.30 |
| Rectal and large intestinal Atresia/stenosis | Singleton | 51 | 3.43 | 2.55-4.51 |
| | Multiple | 7 | 10.06 | 4.04-20.72 |
| Small intestinal atresia | Singleton | 51 | 3.43 | 2.55-4.51 |
| | Multiple | 4 | 5.75 | 1.57-14.71 |

# Table 8 Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2006-2007

| Defect[2] | Plurality | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| Other gastrointestinal | Singleton | 97 | 6.53 | 5.29-7.96 |
| | Multiple | 7 | 10.06 | 4.04-20.72 |
| ***Genitourinary*** | | | | |
| Bladder exstrophy | Singleton | 1 | 0.07 | 0.00-0.37 |
| | Multiple | 0 | 0.00 | 0.00-5.30 |
| Cloacal exstrophy | Singleton | 2 | 0.13 | 0.02-0.49 |
| | Multiple | 0 | 0.00 | 0.00-5.30 |
| Hypospadias, 2nd or 3rd degree | Singleton | 164 | 11.03 | 9.41-12.86 |
| | Multiple | 17 | 24.43 | 14.23-39.11 |
| Obstructive genitourinary defect | Singleton | 216 | 14.53 | 12.66-16.60 |
| | Multiple | 26 | 37.36 | 24.40-54.74 |
| Renal agenesis/hypoplasia | Singleton | 3 | 0.20 | 0.04-0.59 |
| | Multiple | 1 | 1.44 | 0.04-8.01 |
| Other genitourinary | Singleton | 256 | 17.22 | 15.18-19.47 |
| | Multiple | 19 | 27.30 | 16.44-42.63 |
| ***Musculoskeltal*** | | | | |
| Clubfoot | Singleton | 172 | 11.57 | 9.91-13.44 |
| | Multiple | 16 | 22.99 | 13.14-37.33 |
| Craniosynostosis | Singleton | 69 | 4.64 | 3.61-5.87 |
| | Multiple | 3 | 4.31 | 0.89-12.60 |
| Diaphragmatic hernia | Singleton | 43 | 2.89 | 2.09-3.90 |
| | Multiple | 1 | 1.44 | 0.04-8.01 |
| Gastroschisis | Singleton | 54 | 3.63 | 2.73-4.74 |
| | Multiple | 2 | 2.87 | 0.35-10.38 |
| Omphalocele | Singleton | 18 | 1.21 | 0.72-1.91 |
| | Multiple | 3 | 4.31 | 0.89-12.60 |
| Polydactyly/syndactyly | Singleton | 217 | 14.60 | 12.72-16.68 |
| | Multiple | 17 | 24.43 | 14.23-39.11 |
| Reduction deformity, lower limbs | Singleton | 21 | 1.41 | 0.87-2.16 |
| | Multiple | 2 | 2.87 | 0.35-10.38 |
| Reduction deformity, upper limbs | Singleton | 51 | 3.43 | 2.55-4.51 |
| | Multiple | 3 | 4.31 | 0.89-12.60 |

# Table 8 Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2006-2007

| Defect[2] | Plurality | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| Skeletal dysplasia | Singleton | 17 | 1.14 | 0.67-1.83 |
| | Multiple | 1 | 1.44 | 0.04-8.01 |
| Other musculoskeletal | Singleton | 148 | 9.96 | 8.42-11.70 |
| | Multiple | 23 | 33.05 | 20.95-49.59 |
| ***Chromosomal and other Syndromes*** | | | | |
| Klinefelter syndrome | Singleton | 5 | 0.34 | 0.11-0.78 |
| | Multiple | 2 | 2.87 | 0.35-10.38 |
| Trisomy 13 | Singleton | 14 | 0.94 | 0.51-1.58 |
| | Multiple | 2 | 2.87 | 0.35-10.38 |
| Trisomy 18 | Singleton | 23 | 1.55 | 0.98-2.32 |
| | Multiple | 1 | 1.44 | 0.04-8.01 |
| Trisomy 21 (Down syndrome) | Singleton | 183 | 12.31 | 10.59-14.23 |
| | Multiple | 14 | 20.11 | 11.00-33.75 |
| Turner syndrome | Singleton | 14 | 0.94 | 0.51-1.58 |
| | Multiple | 1 | 1.44 | 0.04-8.01 |
| Other chromosomal syndromes/other syndromes | Singleton | 156 | 10.49 | 8.91-12.28 |
| | Multiple | 16 | 22.99 | 13.14-37.33 |
| ***Other*** | | | | |
| Amniotic bands | Singleton | 14 | 0.94 | 0.51-1.58 |
| | Multiple | 2 | 2.87 | 0.35-10.38 |
| Skin anomalies | Singleton | 17 | 1.14 | 0.67-1.83 |
| | Multiple | 1 | 1.44 | 0.04-8.01 |
| Other, specified | Singleton | 23 | 1.55 | 0.98-2.32 |
| | Multiple | 2 | 2.87 | 0.35-10.38 |

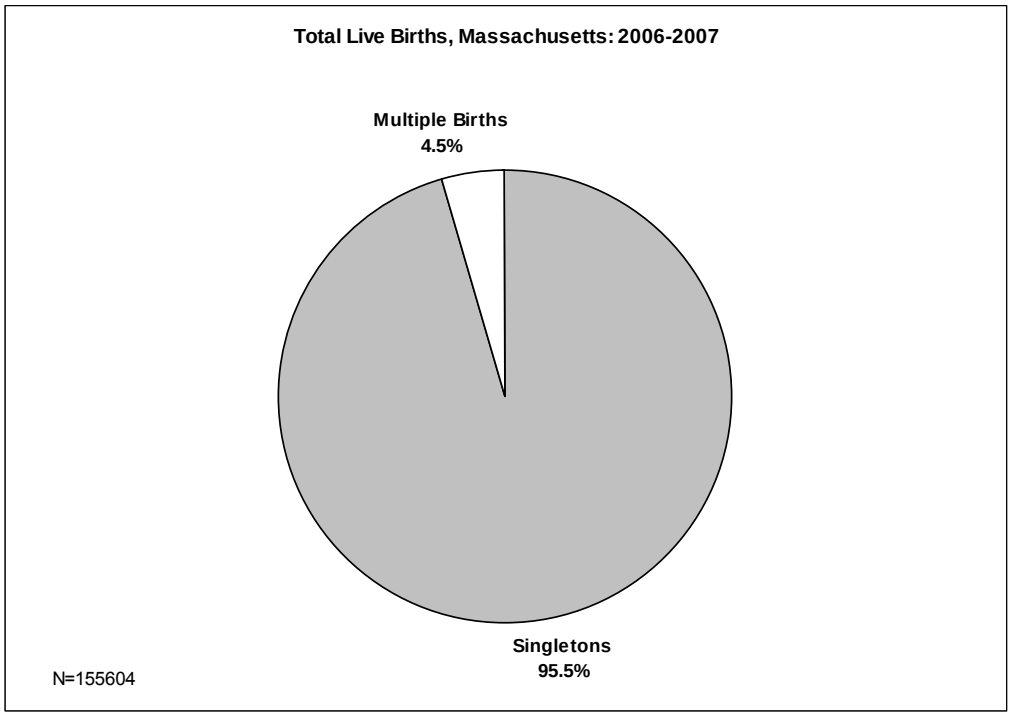
1. Plurality is the number of births to a woman from the same pregnancy. A singleton is the birth of one infant; multiple represents more than one infant.
2. Cases can be included in more than one defect. Cases are counted once in the total for a defect category. Due to missing plurality of infant counts may not match those in other tables.

# Table 9 Most Common Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 2006-2007

| Defect[2] | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|
| **MULTIPLE** | | | |
| Obstructive genitourinary defect | 26 | 37.36 | 24.40-54.74 |
| ASD (secundum and NOS) | 25 | 35.92 | 23.25-53.02 |
| Hypospadias, 2nd or 3rd degree | 17 | 24.43 | 14.23-39.11 |
| Polydactyly/syndactyly | 17 | 24.43 | 14.23-39.11 |
| Clubfoot | 16 | 22.99 | 13.14-37.33 |
| VSD (membranous and NOS) | 14 | 20.11 | 11.00-33.75 |
| Trisomy 21 (Down syndrome) | 14 | 20.11 | 11.00-33.75 |
| Cleft lip w/ and w/o cleft palate | 13 | 18.68 | 9.95-31.94 |
| Tetralogy of Fallot w/ and w/o pulmonary atresia | 10 | 14.37 | 6.89-26.42 |
| Pulmonary stenosis, valvular | 9 | 12.93 | 5.91-24.55 |
| **SINGLETON** | | | |
| ASD (secundum and NOS) | 229 | 15.41 | 13.48-17.54 |
| Polydactyly/syndactyly | 217 | 14.60 | 12.72-16.68 |
| Obstructive genitourinary defect | 216 | 14.53 | 12.66-16.60 |
| Trisomy 21 (Down syndrome) | 183 | 12.31 | 10.59-14.23 |
| Clubfoot | 172 | 11.57 | 9.91-13.44 |
| Hypospadias, 2nd or 3rd degree | 164 | 11.03 | 9.41-12.86 |
| VSD (membranous and NOS) | 138 | 9.28 | 7.80-10.97 |
| Cleft lip w/ and w/o cleft palate | 108 | 7.27 | 5.96-8.77 |
| Pulmonary stenosis, valvular | 90 | 6.05 | 4.87-7.44 |
| Cleft palate w/o cleft lip | 71 | 4.78 | 3.73-6.02 |

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

Figure 6 Plurality of All Live Births and Birth Defect Cases, Live Births Only, Massachusetts: 2006-2007



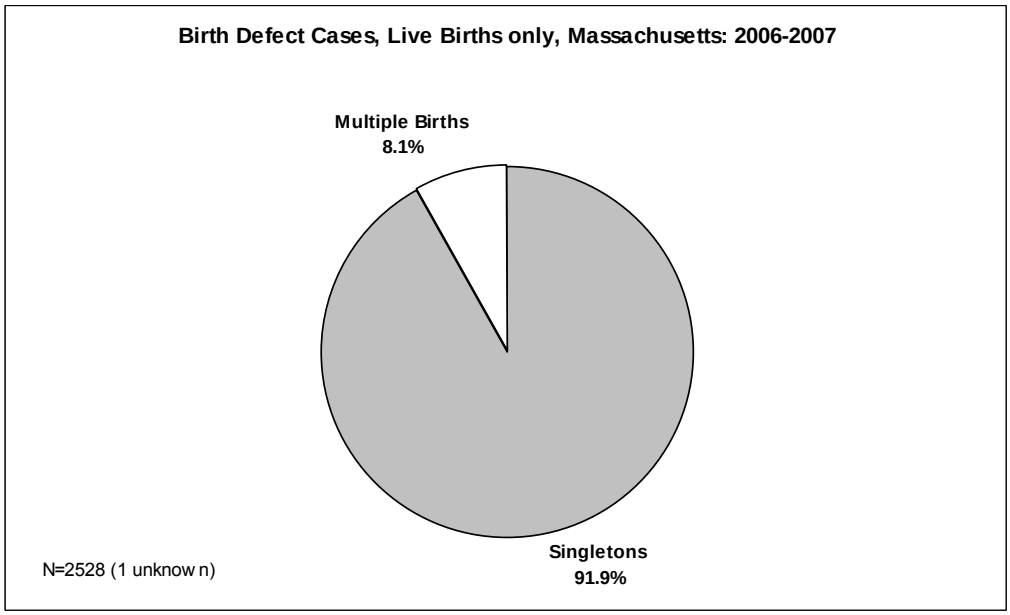
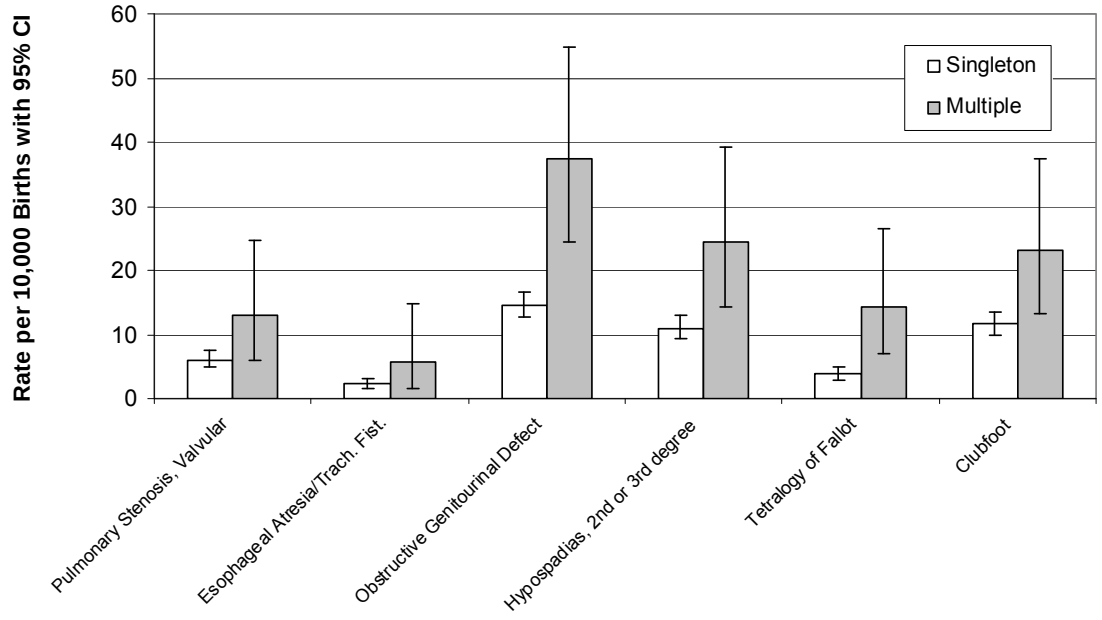



Figure 7 Prevalence of Selected Birth Defects by Plurality among Live Births and Stillbirths, Massachusetts: 2006-2007




N=2585

# Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2006-2007

| Defect[1] | Maternal Age | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| ***Central Nervous System*** | | | | |
| Anencephaly | <20 | 2 | 2.05 | 0.25-7.39 |
| | 20-24 | 0 | 0.00 | 0.00-1.47 |
| | 25-29 | 1 | 0.26 | 0.01-1.47 |
| | 30-34 | 2 | 0.43 | 0.05-1.54 |
| | 35+ | 1 | 0.28 | 0.01-1.55 |
| Encephalocele | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 2 | 0.80 | 0.10-2.89 |
| | 25-29 | 0 | 0.00 | 0.00-0.97 |
| | 30-34 | 0 | 0.00 | 0.00-0.79 |
| | 35+ | 1 | 0.28 | 0.01-1.55 |
| Holoprosencephaly | <20 | 1 | 1.02 | 0.03-5.70 |
| | 20-24 | 2 | 0.80 | 0.10-2.89 |
| | 25-29 | 1 | 0.26 | 0.01-1.47 |
| | 30-34 | 1 | 0.21 | 0.01-1.19 |
| | 35+ | 0 | 0.00 | 0.00-1.03 |
| Hydrocephaly w/o spina bifida | <20 | 2 | 2.05 | 0.25-7.39 |
| | 20-24 | 5 | 2.00 | 0.65-4.66 |
| | 25-29 | 6 | 1.58 | 0.58-3.44 |
| | 30-34 | 9 | 1.92 | 0.88-3.64 |
| | 35+ | 11 | 3.06 | 1.53-5.48 |
| Microcephaly | <20 | 1 | 1.02 | 0.03-5.70 |
| | 20-24 | 5 | 2.00 | 0.65-4.66 |
| | 25-29 | 8 | 2.11 | 0.91-4.16 |
| | 30-34 | 9 | 1.92 | 0.88-3.64 |
| | 35+ | 7 | 1.95 | 0.78-4.01 |
| Spina Bifida w/ and w/o hydrocephaly | <20 | 3 | 3.07 | 0.63-8.97 |
| | 20-24 | 1 | 0.40 | 0.01-2.23 |
| | 25-29 | 9 | 2.37 | 1.09-4.51 |
| | 30-34 | 5 | 1.07 | 0.35-2.49 |
| | 35+ | 3 | 0.83 | 0.17-2.44 |

# Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2006-2007

| Defect[1] | Maternal Age | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| Tethered cord | <20 | 6 | 6.14 | 2.25-13.37 |
| | 20-24 | 7 | 2.80 | 1.12-5.76 |
| | 25-29 | 15 | 3.96 | 2.21-6.53 |
| | 30-34 | 19 | 4.05 | 2.44-6.32 |
| | 35+ | 8 | 2.23 | 0.96-4.39 |
| Other CNS | <20 | 11 | 11.26 | 5.62-20.14 |
| | 20-24 | 20 | 7.99 | 4.88-12.35 |
| | 25-29 | 27 | 7.12 | 4.69-10.36 |
| | 30-34 | 23 | 4.90 | 3.11-7.35 |
| | 35+ | 29 | 8.07 | 5.40-11.59 |
| ***Eye*** | | | | |
| Aniridia | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 1 | 0.40 | 0.01-2.23 |
| | 25-29 | 0 | 0.00 | 0.00-0.97 |
| | 30-34 | 0 | 0.00 | 0.00-0.79 |
| | 35+ | 0 | 0.00 | 0.00-1.03 |
| Anophthalmia/microphthalmia | <20 | 2 | 2.05 | 0.25-7.39 |
| | 20-24 | 4 | 1.60 | 0.44-4.09 |
| | 25-29 | 4 | 1.05 | 0.29-2.70 |
| | 30-34 | 2 | 0.43 | 0.05-1.54 |
| | 35+ | 4 | 1.11 | 0.30-2.85 |
| Congenital glaucoma, congenital cataract | <20 | 3 | 3.07 | 0.63-8.97 |
| | 20-24 | 8 | 3.20 | 1.38-6.30 |
| | 25-29 | 10 | 2.64 | 1.26-4.85 |
| | 30-34 | 18 | 3.83 | 2.27-6.06 |
| | 35+ | 11 | 3.06 | 1.53-5.48 |
| Other eye | <20 | 3 | 3.07 | 0.63-8.97 |
| | 20-24 | 6 | 2.40 | 0.88-5.22 |
| | 25-29 | 4 | 1.05 | 0.29-2.70 |
| | 30-34 | 10 | 2.13 | 1.02-3.92 |
| | 35+ | 8 | 2.23 | 0.96-4.39 |

# Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2006-2007

| Defect[1] | Maternal Age | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| ***Ear*** | | | | |
| Anotia/microtia | <20 | 3 | 3.07 | 0.63-8.97 |
| | 20-24 | 3 | 1.20 | 0.25-3.50 |
| | 25-29 | 8 | 2.11 | 0.91-4.16 |
| | 30-34 | 11 | 2.34 | 1.17-4.19 |
| | 35+ | 6 | 1.67 | 0.61-3.63 |
| Other ear | <20 | 1 | 1.02 | 0.03-5.70 |
| | 20-24 | 8 | 3.20 | 1.38-6.30 |
| | 25-29 | 4 | 1.05 | 0.29-2.70 |
| | 30-34 | 10 | 2.13 | 1.02-3.92 |
| | 35+ | 7 | 1.95 | 0.78-4.01 |
| ***Cardiovascular*** | | | | |
| ***Anomalous Pulmonary Venous Connection*** | | | | |
| Total/Partial anomalous pulmonary venous connection | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 4 | 1.60 | 0.44-4.09 |
| | 25-29 | 5 | 1.32 | 0.43-3.08 |
| | 30-34 | 9 | 1.92 | 0.88-3.64 |
| | 35+ | 6 | 1.67 | 0.61-3.63 |
| ***Atrioventricular Canal Defects*** | | | | |
| ASD primum | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 1 | 0.40 | 0.01-2.23 |
| | 25-29 | 1 | 0.26 | 0.01-1.47 |
| | 30-34 | 2 | 0.43 | 0.05-1.54 |
| | 35+ | 4 | 1.11 | 0.30-2.85 |
| Common atrium | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 1 | 0.40 | 0.01-2.23 |
| | 25-29 | 1 | 0.26 | 0.01-1.47 |
| | 30-34 | 1 | 0.21 | 0.01-1.19 |
| | 35+ | 2 | 0.56 | 0.07-2.01 |

# Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2006-2007

| Defect[1] | Maternal Age | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| Complete atrioventricular canal defect | <20 | 2 | 2.05 | 0.25-7.39 |
| | 20-24 | 5 | 2.00 | 0.65-4.66 |
| | 25-29 | 9 | 2.37 | 1.09-4.51 |
| | 30-34 | 10 | 2.13 | 1.02-3.92 |
| | 35+ | 17 | 4.73 | 2.76-7.57 |
| Endocardial cushion (OS and NOS) | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 0 | 0.00 | 0.00-1.47 |
| | 25-29 | 0 | 0.00 | 0.00-0.97 |
| | 30-34 | 4 | 0.85 | 0.23-2.18 |
| | 35+ | 2 | 0.56 | 0.07-2.01 |
| VSD, canal type | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 1 | 0.40 | 0.01-2.23 |
| | 25-29 | 0 | 0.00 | 0.00-0.97 |
| | 30-34 | 0 | 0.00 | 0.00-0.79 |
| | 35+ | 3 | 0.83 | 0.17-2.44 |
| ***Conotruncal (Outlet) and Aortic Arch*** | | | | |
| Double outlet right ventricle | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 9 | 3.60 | 1.64-6.83 |
| | 25-29 | 4 | 1.05 | 0.29-2.70 |
| | 30-34 | 4 | 0.85 | 0.23-2.18 |
| | 35+ | 3 | 0.83 | 0.17-2.44 |
| d -Transposition of the great arteries | <20 | 1 | 1.02 | 0.03-5.70 |
| | 20-24 | 3 | 1.20 | 0.25-3.50 |
| | 25-29 | 12 | 3.16 | 1.64-5.53 |
| | 30-34 | 9 | 1.92 | 0.88-3.64 |
| | 35+ | 7 | 1.95 | 0.78-4.01 |
| Interrupted aortic arch, type B | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 1 | 0.40 | 0.01-2.23 |
| | 25-29 | 3 | 0.79 | 0.16-2.31 |
| | 30-34 | 3 | 0.64 | 0.13-1.87 |
| | 35+ | 1 | 0.28 | 0.01-1.55 |

# Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2006-2007

| Defect[1] | Maternal Age | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| Tetralogy of Fallot w/ and w/o pulmonary atresia | <20 | 1 | 1.02 | 0.03-5.70 |
| | 20-24 | 17 | 6.80 | 3.96-10.88 |
| | 25-29 | 12 | 3.16 | 1.64-5.53 |
| | 30-34 | 17 | 3.62 | 2.11-5.80 |
| | 35+ | 18 | 5.01 | 2.97-7.91 |
| Truncus | <20 | 1 | 1.02 | 0.03-5.70 |
| | 20-24 | 1 | 0.40 | 0.01-2.23 |
| | 25-29 | 2 | 0.53 | 0.06-1.91 |
| | 30-34 | 2 | 0.43 | 0.05-1.54 |
| | 35+ | 0 | 0.00 | 0.00-1.03 |
| ***Ebstein Anomaly*** | | | | |
| Ebstein anomaly | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 0 | 0.00 | 0.00-1.47 |
| | 25-29 | 1 | 0.26 | 0.01-1.47 |
| | 30-34 | 2 | 0.43 | 0.05-1.54 |
| | 35+ | 4 | 1.11 | 0.30-2.85 |
| ***Heterotaxy (Laterality Defects)*** | | | | |
| Heterotaxy | <20 | 1 | 1.02 | 0.03-5.70 |
| | 20-24 | 2 | 0.80 | 0.10-2.89 |
| | 25-29 | 5 | 1.32 | 0.43-3.08 |
| | 30-34 | 4 | 0.85 | 0.23-2.18 |
| | 35+ | 3 | 0.83 | 0.17-2.44 |
| ***Left-Sided Obstruction*** | | | | |
| Aortic valve stenosis | <20 | 3 | 3.07 | 0.63-8.97 |
| | 20-24 | 5 | 2.00 | 0.65-4.66 |
| | 25-29 | 7 | 1.85 | 0.74-3.80 |
| | 30-34 | 12 | 2.56 | 1.32-4.47 |
| | 35+ | 2 | 0.56 | 0.07-2.01 |
| Coarctation of aorta | <20 | 1 | 1.02 | 0.03-5.70 |
| | 20-24 | 6 | 2.40 | 0.88-5.22 |
| | 25-29 | 21 | 5.54 | 3.43-8.47 |
| | 30-34 | 18 | 3.83 | 2.27-6.06 |

# Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2006-2007

| Defect[1] | Maternal Age | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | 35+ | 16 | 4.45 | 2.54-7.23 |
| | | | | |
| Hypoplastic left heart syndrome | <20 | 1 | 1.02 | 0.03-5.70 |
| | 20-24 | 5 | 2.00 | 0.65-4.66 |
| | 25-29 | 10 | 2.64 | 1.26-4.85 |
| | 30-34 | 5 | 1.07 | 0.35-2.49 |
| | 35+ | 7 | 1.95 | 0.78-4.01 |
| Interrupted aortic arch (type A and NOS) | <20 | 2 | 2.05 | 0.25-7.39 |
| | 20-24 | 0 | 0.00 | 0.00-1.47 |
| | 25-29 | 0 | 0.00 | 0.00-0.97 |
| | 30-34 | 1 | 0.21 | 0.01-1.19 |
| | 35+ | 2 | 0.56 | 0.07-2.01 |
| ***Other Cardiovascular*** | | | | |
| Other cardiovascular | <20 | 10 | 10.23 | 4.91-18.82 |
| | 20-24 | 34 | 13.59 | 9.41-18.99 |
| | 25-29 | 75 | 19.78 | 15.56-24.80 |
| | 30-34 | 74 | 15.76 | 12.38-19.79 |
| | 35+ | 68 | 18.92 | 14.69-23.98 |
| ***Patent Ductus Arteriosus*** | | | | |
| Patent ductus arteriosus | <20 | 10 | 10.23 | 4.91-18.82 |
| | 20-24 | 39 | 15.59 | 11.09-21.31 |
| | 25-29 | 48 | 12.66 | 9.33-16.78 |
| | 30-34 | 87 | 18.53 | 14.84-22.86 |
| | 35+ | 99 | 27.54 | 22.39-33.53 |
| ***Right-Sided Obstruction*** | | | | |
| Pulmonary stenosis, valvular | <20 | 2 | 2.05 | 0.25-7.39 |
| | 20-24 | 12 | 4.80 | 2.48-8.38 |
| | 25-29 | 17 | 4.48 | 2.61-7.18 |
| | 30-34 | 30 | 6.39 | 4.31-9.12 |
| | 35+ | 38 | 10.57 | 7.48-14.51 |
| Pulmonary valve atresia w/intact septum | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 2 | 0.80 | 0.10-2.89 |
| | 25-29 | 1 | 0.26 | 0.01-1.47 |

# Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2006-2007

| Defect[1] | Maternal Age | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | 30-34 | 3 | 0.64 | 0.13-1.87 |
| | 35+ | 1 | 0.28 | 0.01-1.55 |
| Pulmonary valve atresia with VSD | <20 | 1 | 1.02 | 0.03-5.70 |
| | 20-24 | 1 | 0.40 | 0.01-2.23 |
| | 25-29 | 3 | 0.79 | 0.16-2.31 |
| | 30-34 | 1 | 0.21 | 0.01-1.19 |
| | 35+ | 0 | 0.00 | 0.00-1.03 |
| Tricuspid valve atresia | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 0 | 0.00 | 0.00-1.47 |
| | 25-29 | 1 | 0.26 | 0.01-1.47 |
| | 30-34 | 1 | 0.21 | 0.01-1.19 |
| | 35+ | 5 | 1.39 | 0.45-3.25 |
| ***Septal Defects*** | | | | |
| ASD (secundum and NOS) | <20 | 7 | 7.16 | 2.88-14.76 |
| | 20-24 | 34 | 13.59 | 9.41-18.99 |
| | 25-29 | 60 | 15.82 | 12.08-20.37 |
| | 30-34 | 64 | 13.63 | 10.50-17.41 |
| | 35+ | 87 | 24.21 | 19.39-29.86 |
| VSD (membranous and NOS) | <20 | 8 | 8.19 | 3.53-16.13 |
| | 20-24 | 16 | 6.40 | 3.66-10.39 |
| | 25-29 | 35 | 9.23 | 6.43-12.84 |
| | 30-34 | 37 | 7.88 | 5.55-10.86 |
| | 35+ | 52 | 14.47 | 10.81-18.97 |
| VSD, conoventricular/malalignment | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 3 | 1.20 | 0.25-3.50 |
| | 25-29 | 6 | 1.58 | 0.58-3.44 |
| | 30-34 | 6 | 1.28 | 0.47-2.78 |
| | 35+ | 4 | 1.11 | 0.30-2.85 |
| ***Single Ventricle and L-TGA*** | | | | |
| L-TGA | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 1 | 0.40 | 0.01-2.23 |
| | 25-29 | 1 | 0.26 | 0.01-1.47 |
| | 30-34 | 1 | 0.21 | 0.01-1.19 |

# Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2006-2007

| Defect[1] | Maternal Age | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | 35+ | 3 | 0.83 | 0.17-2.44 |
| Single ventricle | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 4 | 1.60 | 0.44-4.09 |
| | 25-29 | 2 | 0.53 | 0.06-1.91 |
| | 30-34 | 1 | 0.21 | 0.01-1.19 |
| | 35+ | 5 | 1.39 | 0.45-3.25 |
| ***Respiratory*** | | | | |
| Choanal atresia | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 5 | 2.00 | 0.65-4.66 |
| | 25-29 | 2 | 0.53 | 0.06-1.91 |
| | 30-34 | 3 | 0.64 | 0.13-1.87 |
| | 35+ | 4 | 1.11 | 0.30-2.85 |
| Lung anomalies | <20 | 3 | 3.07 | 0.63-8.97 |
| | 20-24 | 3 | 1.20 | 0.25-3.50 |
| | 25-29 | 3 | 0.79 | 0.16-2.31 |
| | 30-34 | 6 | 1.28 | 0.47-2.78 |
| | 35+ | 6 | 1.67 | 0.61-3.63 |
| Other respiratory | <20 | 3 | 3.07 | 0.63-8.97 |
| | 20-24 | 3 | 1.20 | 0.25-3.50 |
| | 25-29 | 2 | 0.53 | 0.06-1.91 |
| | 30-34 | 6 | 1.28 | 0.47-2.78 |
| | 35+ | 8 | 2.23 | 0.96-4.39 |
| ***Orofacial*** | | | | |
| Cleft lip w/ and w/o cleft palate | <20 | 6 | 6.14 | 2.25-13.37 |
| | 20-24 | 16 | 6.40 | 3.66-10.39 |
| | 25-29 | 33 | 8.70 | 5.99-12.22 |
| | 30-34 | 30 | 6.39 | 4.31-9.12 |
| | 35+ | 30 | 8.35 | 5.63-11.92 |
| Cleft palate w/o cleft lip | <20 | 5 | 5.12 | 1.66-11.94 |
| | 20-24 | 11 | 4.40 | 2.19-7.87 |
| | 25-29 | 15 | 3.96 | 2.21-6.53 |
| | 30-34 | 23 | 4.90 | 3.11-7.35 |
| | 35+ | 19 | 5.29 | 3.18-8.26 |

# Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2006-2007

| Defect[1] | Maternal Age | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| Pierre Robin sequence | <20 | 1 | 1.02 | 0.03-5.70 |
| | 20-24 | 3 | 1.20 | 0.25-3.50 |
| | 25-29 | 8 | 2.11 | 0.91-4.16 |
| | 30-34 | 4 | 0.85 | 0.23-2.18 |
| | 35+ | 7 | 1.95 | 0.78-4.01 |
| Other orofacial | <20 | 5 | 5.12 | 1.66-11.94 |
| | 20-24 | 11 | 4.40 | 2.19-7.87 |
| | 25-29 | 16 | 4.22 | 2.41-6.85 |
| | 30-34 | 18 | 3.83 | 2.27-6.06 |
| | 35+ | 22 | 6.12 | 3.84-9.27 |
| ***Gastrointestinal*** | | | | |
| Biliary atresia | <20 | 1 | 1.02 | 0.03-5.70 |
| | 20-24 | 4 | 1.60 | 0.44-4.09 |
| | 25-29 | 1 | 0.26 | 0.01-1.47 |
| | 30-34 | 1 | 0.21 | 0.01-1.19 |
| | 35+ | 2 | 0.56 | 0.07-2.01 |
| Esophageal atresia/tracheoesophageal fistula | <20 | 3 | 3.07 | 0.63-8.97 |
| | 20-24 | 6 | 2.40 | 0.88-5.22 |
| | 25-29 | 1 | 0.26 | 0.01-1.47 |
| | 30-34 | 9 | 1.92 | 0.88-3.64 |
| | 35+ | 18 | 5.01 | 2.97-7.91 |
| Hirschsprung disease | <20 | 2 | 2.05 | 0.25-7.39 |
| | 20-24 | 6 | 2.40 | 0.88-5.22 |
| | 25-29 | 6 | 1.58 | 0.58-3.44 |
| | 30-34 | 4 | 0.85 | 0.23-2.18 |
| | 35+ | 6 | 1.67 | 0.61-3.63 |
| Rectal and large intestinal atresia/stenosis | <20 | 6 | 6.14 | 2.25-13.37 |
| | 20-24 | 10 | 4.00 | 1.92-7.35 |
| | 25-29 | 15 | 3.96 | 2.21-6.53 |
| | 30-34 | 14 | 2.98 | 1.63-5.00 |
| | 35+ | 13 | 3.62 | 1.93-6.19 |

# Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2006-2007

| Defect[1] | Maternal Age | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| Small intestinal atresia | <20 | 3 | 3.07 | 0.63-8.97 |
| | 20-24 | 10 | 4.00 | 1.92-7.35 |
| | 25-29 | 10 | 2.64 | 1.26-4.85 |
| | 30-34 | 18 | 3.83 | 2.27-6.06 |
| | 35+ | 14 | 3.90 | 2.13-6.54 |
| Other gastrointestinal | <20 | 6 | 6.14 | 2.25-13.37 |
| | 20-24 | 18 | 7.19 | 4.26-11.37 |
| | 25-29 | 26 | 6.86 | 4.48-10.05 |
| | 30-34 | 33 | 7.03 | 4.84-9.87 |
| | 35+ | 19 | 5.29 | 3.18-8.26 |
| ***Genitourinary*** | | | | |
| Bladder exstrophy | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 0 | 0.00 | 0.00-1.47 |
| | 25-29 | 0 | 0.00 | 0.00-0.97 |
| | 30-34 | 1 | 0.21 | 0.01-1.19 |
| | 35+ | 0 | 0.00 | 0.00-1.03 |
| Cloacal exstrophy | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 0 | 0.00 | 0.00-1.47 |
| | 25-29 | 0 | 0.00 | 0.00-0.97 |
| | 30-34 | 2 | 0.43 | 0.05-1.54 |
| | 35+ | 0 | 0.00 | 0.00-1.03 |
| Hypospadias, 2nd or 3rd degree | <20 | 9 | 9.21 | 4.21-17.49 |
| | 20-24 | 23 | 9.19 | 5.83-13.79 |
| | 25-29 | 39 | 10.29 | 7.31-14.06 |
| | 30-34 | 58 | 12.35 | 9.38-15.97 |
| | 35+ | 52 | 14.47 | 10.81-18.97 |
| Obstructive genitourinary defect | <20 | 20 | 20.47 | 12.50-31.61 |
| | 20-24 | 30 | 11.99 | 8.09-17.12 |
| | 25-29 | 53 | 13.98 | 10.47-18.28 |
| | 30-34 | 80 | 17.04 | 13.51-21.21 |
| | 35+ | 59 | 16.42 | 12.50-21.17 |

# Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2006-2007

| Defect[1] | Maternal Age | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| Renal agenesis/hypoplasia | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 1 | 0.40 | 0.01-2.23 |
| | 25-29 | 1 | 0.26 | 0.01-1.47 |
| | 30-34 | 0 | 0.00 | 0.00-0.79 |
| | 35+ | 0 | 0.00 | 0.00-1.03 |
| Other genitourinary | <20 | 14 | 14.33 | 7.83-24.04 |
| | 20-24 | 44 | 17.59 | 12.78-23.61 |
| | 25-29 | 62 | 16.35 | 12.54-20.96 |
| | 30-34 | 82 | 17.47 | 13.89-21.68 |
| | 35+ | 69 | 19.20 | 14.94-24.30 |
| ***Musculoskeletal*** | | | | |
| Clubfoot | <20 | 10 | 10.23 | 4.91-18.82 |
| | 20-24 | 32 | 12.79 | 8.75-18.06 |
| | 25-29 | 45 | 11.87 | 8.66-15.88 |
| | 30-34 | 62 | 13.21 | 10.13-16.93 |
| | 35+ | 32 | 8.90 | 6.09-12.57 |
| Craniosynostosis | <20 | 1 | 1.02 | 0.03-5.70 |
| | 20-24 | 9 | 3.60 | 1.64-6.83 |
| | 25-29 | 17 | 4.48 | 2.61-7.18 |
| | 30-34 | 29 | 6.18 | 4.14-8.87 |
| | 35+ | 16 | 4.45 | 2.54-7.23 |
| Diaphragmatic hernia | <20 | 4 | 4.09 | 1.12-10.48 |
| | 20-24 | 8 | 3.20 | 1.38-6.30 |
| | 25-29 | 11 | 2.90 | 1.45-5.19 |
| | 30-34 | 12 | 2.56 | 1.32-4.47 |
| | 35+ | 7 | 1.95 | 0.78-4.01 |
| Gastroschisis | <20 | 16 | 16.37 | 9.36-26.59 |
| | 20-24 | 20 | 7.99 | 4.88-12.35 |
| | 25-29 | 11 | 2.90 | 1.45-5.19 |
| | 30-34 | 5 | 1.07 | 0.35-2.49 |
| | 35+ | 0 | 0.00 | 0.00-1.03 |

# Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2006-2007

| Defect[1] | Maternal Age | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| Omphalocele | <20 | 2 | 2.05 | 0.25-7.39 |
| | 20-24 | 3 | 1.20 | 0.25-3.50 |
| | 25-29 | 6 | 1.58 | 0.58-3.44 |
| | 30-34 | 4 | 0.85 | 0.23-2.18 |
| | 35+ | 4 | 1.11 | 0.30-2.85 |
| Polydactyly/syndactyly | <20 | 12 | 12.28 | 6.35-21.45 |
| | 20-24 | 36 | 14.39 | 10.08-19.92 |
| | 25-29 | 53 | 13.98 | 10.47-18.28 |
| | 30-34 | 77 | 16.40 | 12.94-20.50 |
| | 35+ | 53 | 14.75 | 11.05-19.29 |
| Reduction deformity, lower limbs | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 7 | 2.80 | 1.12-5.76 |
| | 25-29 | 9 | 2.37 | 1.09-4.51 |
| | 30-34 | 3 | 0.64 | 0.13-1.87 |
| | 35+ | 4 | 1.11 | 0.30-2.85 |
| Reduction deformity, upper limbs | <20 | 1 | 1.02 | 0.03-5.70 |
| | 20-24 | 13 | 5.20 | 2.77-8.89 |
| | 25-29 | 13 | 3.43 | 1.83-5.86 |
| | 30-34 | 14 | 2.98 | 1.63-5.00 |
| | 35+ | 9 | 2.50 | 1.15-4.75 |
| Skeletal dysplasia | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 3 | 1.20 | 0.25-3.50 |
| | 25-29 | 4 | 1.05 | 0.29-2.70 |
| | 30-34 | 5 | 1.07 | 0.35-2.49 |
| | 35+ | 5 | 1.39 | 0.45-3.25 |
| Other musculoskeletal | <20 | 16 | 16.37 | 9.36-26.59 |
| | 20-24 | 29 | 11.59 | 7.76-16.65 |
| | 25-29 | 34 | 8.97 | 6.21-12.53 |
| | 30-34 | 45 | 9.59 | 6.99-12.83 |
| | 35+ | 39 | 10.85 | 7.72-14.83 |

# Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2006-2007

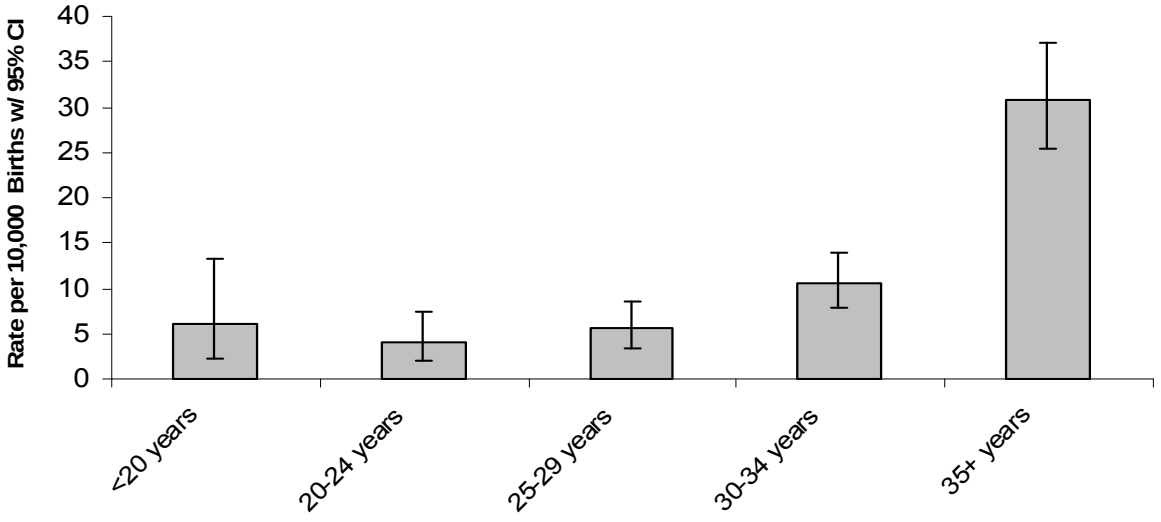
| Defect[1] | Maternal Age | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| ***Chromosomal and other Syndromes*** | | | | |
| Klinefelter syndrome | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 2 | 0.80 | 0.10-2.89 |
| | 25-29 | 2 | 0.53 | 0.06-1.91 |
| | 30-34 | 1 | 0.21 | 0.01-1.19 |
| | 35+ | 2 | 0.56 | 0.07-2.01 |
| Trisomy 13 | <20 | 1 | 1.02 | 0.03-5.70 |
| | 20-24 | 1 | 0.40 | 0.01-2.23 |
| | 25-29 | 3 | 0.79 | 0.16-2.31 |
| | 30-34 | 1 | 0.21 | 0.01-1.19 |
| | 35+ | 6 | 1.67 | 0.61-3.63 |
| Trisomy 18 | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 3 | 1.20 | 0.25-3.50 |
| | 25-29 | 3 | 0.79 | 0.16-2.31 |
| | 30-34 | 2 | 0.43 | 0.05-1.54 |
| | 35+ | 6 | 1.67 | 0.61-3.63 |
| Trisomy 21 (Down syndrome) | <20 | 5 | 5.12 | 1.66-11.94 |
| | 20-24 | 10 | 4.00 | 1.92-7.35 |
| | 25-29 | 21 | 5.54 | 3.43-8.47 |
| | 30-34 | 49 | 10.44 | 7.72-13.80 |
| | 35+ | 108 | 30.05 | 24.65-36.28 |
| Turner syndrome | <20 | 0 | 0.00 | 0.00-3.78 |
| | 20-24 | 1 | 0.40 | 0.01-2.23 |
| | 25-29 | 2 | 0.53 | 0.06-1.91 |
| | 30-34 | 3 | 0.64 | 0.13-1.87 |
| | 35+ | 4 | 1.11 | 0.30-2.85 |
| Other chromosomal syndromes/other syndromes | <20 | 7 | 7.16 | 2.88-14.76 |
| | 20-24 | 23 | 9.19 | 5.83-13.79 |
| | 25-29 | 45 | 11.87 | 8.66-15.88 |
| | 30-34 | 50 | 10.65 | 7.91-14.04 |
| | 35+ | 43 | 11.96 | 8.66-16.12 |

# Table 10 Prevalence of Birth Defects by Maternal Age Group for Live Births, Massachusetts: 2006-2007

| Defect[1] | Maternal Age | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| ***Other*** | | | | |
| Amniotic bands | <20 | 2 | 2.05 | 0.25-7.39 |
| | 20-24 | 3 | 1.20 | 0.25-3.50 |
| | 25-29 | 4 | 1.05 | 0.29-2.70 |
| | 30-34 | 2 | 0.43 | 0.05-1.54 |
| | 35+ | 1 | 0.28 | 0.01-1.55 |
| Skin anomalies | <20 | 3 | 3.07 | 0.63-8.97 |
| | 20-24 | 3 | 1.20 | 0.25-3.50 |
| | 25-29 | 8 | 2.11 | 0.91-4.16 |
| | 30-34 | 1 | 0.21 | 0.01-1.19 |
| | 35+ | 3 | 0.83 | 0.17-2.44 |
| Other, specified | <20 | 1 | 1.02 | 0.03-5.70 |
| | 20-24 | 3 | 1.20 | 0.25-3.50 |
| | 25-29 | 7 | 1.85 | 0.74-3.80 |
| | 30-34 | 7 | 1.49 | 0.60-3.07 |
| | 35+ | 6 | 1.67 | 0.61-3.63 |

1. Cases can be included in more than one defect. Cases are counted once in the total for a defect category. Because only live births are presented on this table, case totals are not listed. Due to missing age of mother counts may not match those in other tables.

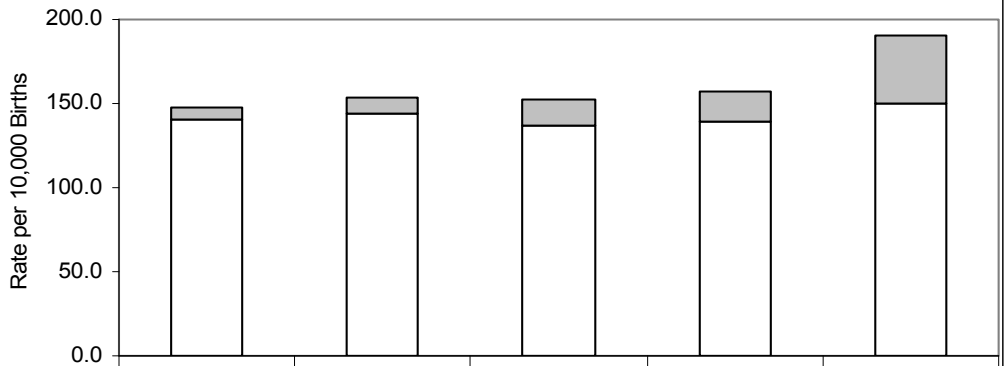
## Figure 8 Rates of Down syndrome among Maternal Age Groups, Massachusetts: 2006-2007




N=197

**Maternal Age Group**

## Figure 9 Chromosomal and All Other Defects by Maternal Age, Massachusetts: 2006-2007




| Maternal Age Group | <20 | 20-24 | 25-29 | 30-34 | 35+ |
|---|---|---|---|---|---|
| Chromosomal defects | 7.2 (2.9-14.8) | 10.4 (6.8-15.2) | 14.8 (11.2-19.2) | 18.3 (14.7-22.6) | 40.6 (34.3-47.8) |
| All other defects | 140.2 (117.7-165.8) | 143.5 (129.1-159.0) | 137.4 (125.9-149.6) | 139.1 (128.7-150.1) | 149.4 (137.1-162.5) |

N=2528, live births only

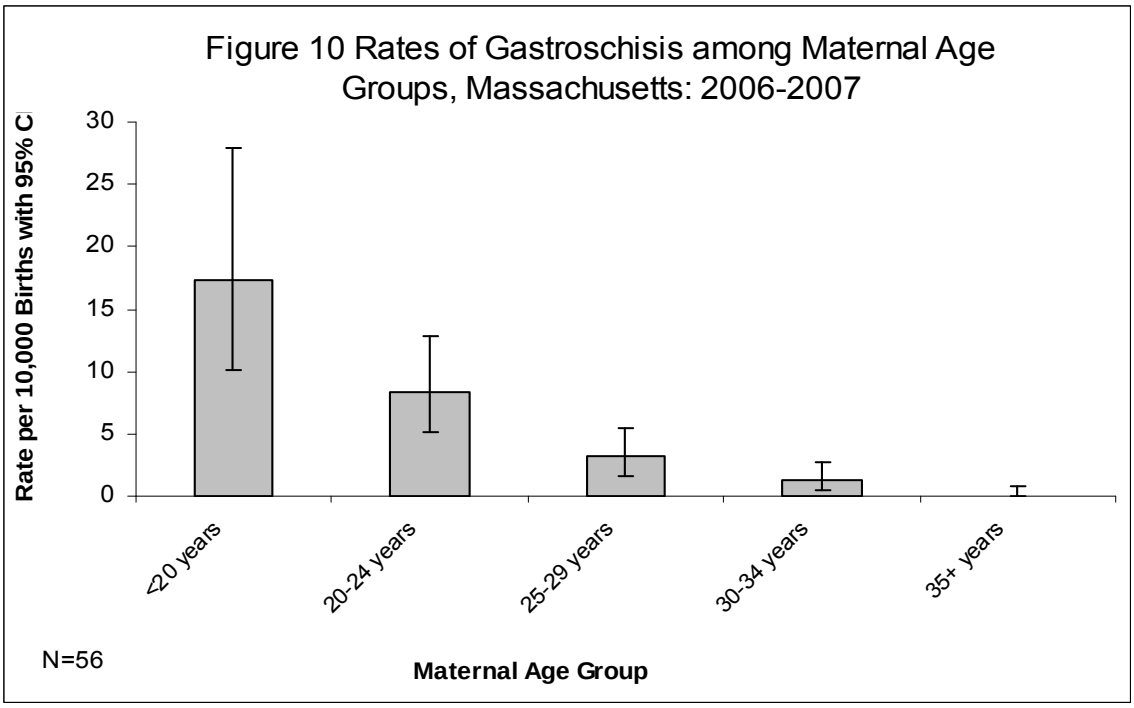


Figure 10 Rates of Gastroschisis among Maternal Age Groups, Massachusetts: 2006-2007
Rate per 10,000 Births with 95% C
30
25
20
15
10
5
0
<20 years
20-24 years
25-29 years
30-34 years
35+ years
N=56
Maternal Age Group

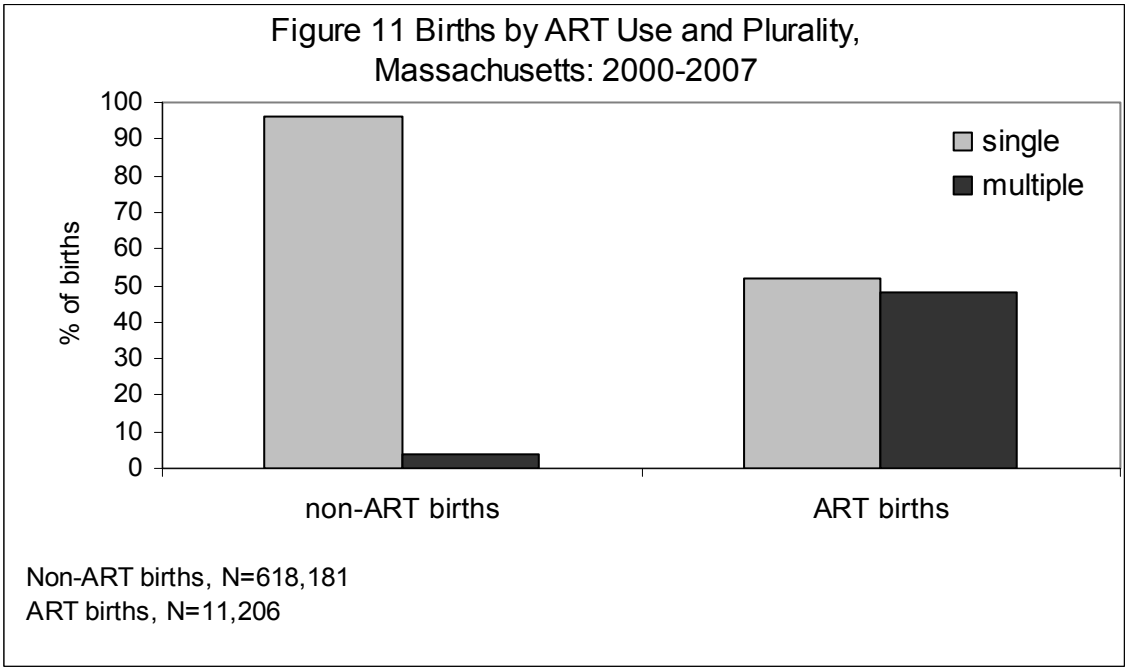


Figure 11 Births by ART Use and Plurality, Massachusetts: 2000-2007
% of births
100
90
80
70
60
50
40
30
20
10
0
single
multiple
non-ART births
ART births
Non-ART births, N=618,181
ART births, N=11,206

# Table 11 Most Common Defects by Maternal Age Group for Live Births, Massachusetts: 2006-2007

| Age Group (yrs) | Defect[1] | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| <20 | Obstructive genitourinary defect | 20 | 20.47 | 12.50-31.61 |
| | Gastroschisis | 16 | 16.37 | 9.36-26.59 |
| | Polydactyly/syndactyly | 12 | 12.28 | 6.35-21.45 |
| | Clubfoot | 10 | 10.23 | 4.91-18.82 |
| | Hypospadias, 2nd or 3rd degree | 9 | 9.21 | 4.21-17.49 |
| 20-24 | Polydactyly/ syndactyly | 36 | 14.39 | 10.08-19.92 |
| | ASD (secundum and NOS) | 34 | 13.59 | 9.41-18.99 |
| | Clubfoot | 32 | 12.79 | 8.75-18.06 |
| | Obstructive genitourinary defect | 30 | 11.99 | 8.09-17.12 |
| | Hypospadias, 2nd or 3rd degree | 23 | 9.19 | 5.83-13.79 |
| 25-29 | ASD (secundum and NOS) | 60 | 15.82 | 12.08-20.37 |
| | Obstructive genitourinary defect | 53 | 13.98 | 10.47-18.28 |
| | Polydactyly/syndactyly | 53 | 13.98 | 10.47-18.28 |
| | Clubfoot | 45 | 11.87 | 8.66-15.88 |
| | Hypospadias, 2nd or 3rd degree | 39 | 10.29 | 7.31-14.06 |
| 30-34 | Obstructive genitourinary defect | 80 | 17.04 | 13.51-21.21 |
| | Polydactyly/syndactyly | 77 | 16.40 | 12.94-20.50 |
| | ASD (secundum and NOS) | 64 | 13.63 | 10.50-17.41 |
| | Clubfoot | 62 | 13.21 | 10.13-16.93 |
| | Hypospadias, 2nd or 3rd degree | 58 | 12.35 | 9.38-15.97 |
| 35+ | Trisomy 21 (Down syndrome) | 108 | 30.05 | 24.65-36.28 |
| | ASD (secundum and NOS) | 87 | 24.21 | 19.39-29.86 |
| | Obstructive genitourinary defect | 59 | 16.42 | 12.50-21.17 |
| | VSD (membranous and NOS) | 52 | 14.47 | 10.81-18.97 |
| | Polydactyly/syndactyly | 53 | 14.75 | 11.05-19.29 |

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

# Chapter 5

# Prevalence of Birth Defects by Race / Ethnicity and Region

Baby with encephalocele

*Courtesy of the Centers for Disease Control and Prevention*

## Maternal Race / Hispanic Ethnicity

Table 12 shows the variation in prevalence of birth defects by maternal race and Hispanic ethnicity. The rate per 10,000 live births was 163.9 for non-Hispanic Whites, 153.4 for non-Hispanic Blacks, 133.5 for non-Hispanic Asians/Pacific Islanders and 176.7 for Hispanics. In some analyses, the rates for other races were not calculated due to small numbers or the rates for other races were combined as one "other" category.

Table 13 shows the more common defects by maternal race and Hispanic ethnicity. The most common defects in Hispanics included obstructive genitourinary defect, polydactyly/syndactyly, clubfoot, atrial septal defects (secundum and NOS), and Down syndrome. In Blacks, the most common defects included polydactyly/syndactyly, obstructive genitourinary defect, Down syndrome, atrial septal defects (secundum and NOS), and hypospadias ($2^{nd}$ and $3^{rd}$ degree). The most common defects in Whites included atrial septal defects (secundum and NOS), obstructive genitourinary defect, polydactyly/syndactyly, hypospadias ($2^{nd}$ and $3^{rd}$ degree), and Down syndrome. In Asians, the most common defects included polydactyly/syndactyly, obstructive genitourinary defect, atrial septal defects (secundum and NOS), and cleft lip with and without cleft palate.

To understand birth defect trends or patterns in maternal race and ethnicity, we explored differences among the groups for certain categories of birth defects. Figure 12 shows the rate of chromosomal defects and all other defects according to maternal race and ethnicity. These rates were age-adjusted because chromosomal defects as well as other defects may be related to maternal age and differences may exist between racial and ethnic groups.

Multiple factors likely contribute to differences in prevalence by racial and ethnic groups including genetic variation, diet and lifestyle, differential access or use of health care services including prenatal screening and diagnosis, or socioeconomic differences. Interestingly, maternal birthplace may also be a contributing factor (see Figure 13). Children born to women born in the U.S. had a slightly higher, though non-significant, rate of birth defects than children of women born outside the U.S. The rate of birth defects in children of Hispanic women born in the U.S. territories (including Puerto Rico, U.S. Virgin Islands and Guam) was non-significantly higher than the rates among women born both in and outside the U.S. More years of data and in-depth studies are needed to affirm the stability of these rates and to understand racial and ethnic patterns.

## Trends in Maternal Race and Ethnicity

Figure 14 shows the age-adjusted birth defects rates between 2004-2005 and 2006-2007. In Blacks, Asians and Hispanics, the rates decreased between the two time periods. Among whites the rates slightly increased. The birth defects rate in other races increased. These changes were not significant between the two time periods examined.

## Birth Defects by Massachusetts Region

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

The birth defect rates by the six regions in 2006-2007 are shown in Figure 15. Although not statistically significantly different, the rates range from 154.8 per 10,000 in Boston to 174.7 per 10,000 in Southeast Massachusetts.

# Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2006-2007

| Defect | Maternal Race[1] | Count[2] | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| ***Central Nervous System*** | | | | |
| Anencephaly | White, Non-Hispanic | 2 | 0.19 | 0.02-0.68 |
| | Black, Non-Hispanic | 1 | 0.77 | 0.02-4.31 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 2 | 0.93 | 0.11-3.35 |
| Encephalocele | White, Non-Hispanic | 1 | 0.09 | 0.00-0.53 |
| | Black, Non-Hispanic | 0 | 0.00 | 0.00-2.86 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 1 | 0.46 | 0.01-2.58 |
| Holoprosencephaly | White, Non-Hispanic | 2 | 0.19 | 0.02-0.68 |
| | Black, Non-Hispanic | 2 | 1.55 | 0.19-5.59 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 1 | 0.46 | 0.01-2.58 |
| Hydrocephaly w/o spina bifida | White, Non-Hispanic | 17 | 1.61 | 0.94-2.58 |
| | Black, Non-Hispanic | 9 | 6.97 | 3.19-13.23 |
| | Asian, Non-Hispanic | 1 | 0.89 | 0.02-4.96 |
| | Hispanic | 4 | 1.86 | 0.51-4.75 |
| Microcephaly | White, Non-Hispanic | 17 | 1.61 | 0.94-2.58 |
| | Black, Non-Hispanic | 5 | 3.87 | 1.26-9.04 |
| | Asian, Non-Hispanic | 1 | 0.89 | 0.02-4.96 |
| | Hispanic | 7 | 3.25 | 1.31-6.69 |
| Spina Bifida w/ and w/o hydrocephaly | White, Non-Hispanic | 14 | 1.33 | 0.72-2.22 |
| | Black, Non-Hispanic | 2 | 1.55 | 0.19-5.59 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 5 | 2.32 | 0.75-5.41 |
| Tethered cord | White, Non-Hispanic | 41 | 3.88 | 2.79-5.27 |
| | Black, Non-Hispanic | 2 | 1.55 | 0.19-5.59 |
| | Asian, Non-Hispanic | 3 | 2.67 | 0.55-7.81 |
| | Hispanic | 8 | 3.71 | 1.60-7.31 |
| Other CNS | White, Non-Hispanic | 61 | 5.78 | 4.42-7.42 |
| | Black, Non-Hispanic | 16 | 12.39 | 7.08-20.12 |
| | Asian, Non-Hispanic | 6 | 5.34 | 1.96-11.63 |
| | Hispanic | 25 | 11.60 | 7.51-17.12 |

# Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2006-2007

| Defect | Maternal Race[1] | Count[2] | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| ***Eye*** | | | | |
| Aniridia | White, Non-Hispanic | 0 | 0.00 | 0.00-0.35 |
| | Black, Non-Hispanic | 0 | 0.00 | 0.00-2.86 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 1 | 0.46 | 0.01-2.58 |
| Anophthalmia/microphthalmia | White, Non-Hispanic | 4 | 0.38 | 0.10-0.97 |
| | Black, Non-Hispanic | 8 | 6.19 | 2.67-12.21 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 4 | 1.86 | 0.51-4.75 |
| Congenital glaucoma, congenital cataract | White, Non-Hispanic | 39 | 3.69 | 2.63-5.05 |
| | Black, Non-Hispanic | 7 | 5.42 | 2.18-11.17 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 3 | 1.39 | 0.29-4.07 |
| Other eye | White, Non-Hispanic | 21 | 1.99 | 1.23-3.04 |
| | Black, Non-Hispanic | 4 | 3.10 | 0.84-7.93 |
| | Asian, Non-Hispanic | 2 | 1.78 | 0.22-6.44 |
| | Hispanic | 3 | 1.39 | 0.29-4.07 |
| ***Ear*** | | | | |
| Anotia/microtia | White, Non-Hispanic | 21 | 1.99 | 1.23-3.04 |
| | Black, Non-Hispanic | 1 | 0.77 | 0.02-4.31 |
| | Asian, Non-Hispanic | 3 | 2.67 | 0.55-7.81 |
| | Hispanic | 6 | 2.78 | 1.02-6.06 |
| Other ear | White, Non-Hispanic | 22 | 2.08 | 1.31-3.15 |
| | Black, Non-Hispanic | 2 | 1.55 | 0.19-5.59 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 5 | 2.32 | 0.75-5.41 |

# Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2006-2007

| Defect | Maternal Race[1] | Count[2] | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| ***Cardiovascular*** | | | | |
| ***Anomalous Pulmonary Venous Connection*** | | | | |
| Total/partial anomalous pulmonary venous connection | White, Non-Hispanic | 17 | 1.61 | 0.94-2.58 |
| | Black, Non-Hispanic | 2 | 1.55 | 0.19-5.59 |
| | Asian, Non-Hispanic | 2 | 1.78 | 0.22-6.44 |
| | Hispanic | 3 | 1.39 | 0.29-4.07 |
| ***Atrioventricular Canal Defects*** | | | | |
| ASD primum | White, Non-Hispanic | 6 | 0.57 | 0.21-1.24 |
| | Black, Non-Hispanic | 2 | 1.55 | 0.19-5.59 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 0 | 0.00 | 0.00-1.71 |
| Common atrium | White, Non-Hispanic | 3 | 0.28 | 0.06-0.83 |
| | Black, Non-Hispanic | 1 | 0.77 | 0.02-4.31 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 1 | 0.46 | 0.01-2.58 |
| Complete atrioventricular canal defect | White, Non-Hispanic | 33 | 3.13 | 2.15-4.39 |
| | Black, Non-Hispanic | 5 | 3.87 | 1.26-9.04 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 4 | 1.86 | 0.51-4.75 |
| Endocardial cushion (OS and NOS) | White, Non-Hispanic | 4 | 0.38 | 0.10-0.97 |
| | Black, Non-Hispanic | 1 | 0.77 | 0.02-4.31 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 1 | 0.46 | 0.01-2.58 |
| VSD, canal type | White, Non-Hispanic | 2 | 0.19 | 0.02-0.68 |
| | Black, Non-Hispanic | 1 | 0.77 | 0.02-4.31 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 0 | 0.00 | 0.00-1.71 |
| ***Conotruncal (Outlet) and Aortic Arch*** | | | | |
| Double outlet right ventricle | White, Non-Hispanic | 8 | 0.76 | 0.33-1.49 |
| | Black, Non-Hispanic | 4 | 3.10 | 0.84-7.93 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 5 | 2.32 | 0.75-5.41 |

# Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2006-2007

| Defect | Maternal Race[1] | Count[2] | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| d-Transposition of the great arteries | White, Non-Hispanic | 21 | 1.99 | 1.23-3.04 |
| | Black, Non-Hispanic | 1 | 0.77 | 0.02-4.31 |
| | Asian, Non-Hispanic | 1 | 0.89 | 0.02-4.96 |
| | Hispanic | 6 | 2.78 | 1.02-6.06 |
| Interrupted aortic arch, type B | White, Non-Hispanic | 5 | 0.47 | 0.15-1.11 |
| | Black, Non-Hispanic | 2 | 1.55 | 0.19-5.59 |
| | Asian, Non-Hispanic | 1 | 0.89 | 0.02-4.96 |
| | Hispanic | 0 | 0.00 | 0.00-1.71 |
| Tetralogy of Fallot w/ and w/o pulmonary atresia | White, Non-Hispanic | 41 | 3.88 | 2.79-5.27 |
| | Black, Non-Hispanic | 7 | 5.42 | 2.18-11.17 |
| | Asian, Non-Hispanic | 9 | 8.02 | 3.67-15.22 |
| | Hispanic | 5 | 2.32 | 0.75-5.41 |
| Truncus | White, Non-Hispanic | 5 | 0.47 | 0.15-1.11 |
| | Black, Non-Hispanic | 0 | 0.00 | 0.00-2.86 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 1 | 0.46 | 0.01-2.58 |
| ***Ebstein Anomaly*** | | | | |
| Ebstein anomaly | White, Non-Hispanic | 6 | 0.57 | 0.21-1.24 |
| | Black, Non-Hispanic | 0 | 0.00 | 0.00-2.86 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 1 | 0.46 | 0.01-2.58 |
| ***Heterotaxy (Laterality Defects)*** | | | | |
| Heterotaxy | White, Non-Hispanic | 6 | 0.57 | 0.21-1.24 |
| | Black, Non-Hispanic | 3 | 2.32 | 0.48-6.79 |
| | Asian, Non-Hispanic | 2 | 1.78 | 0.22-6.44 |
| | Hispanic | 3 | 1.39 | 0.29-4.07 |
| ***Left-Sided Obstruction*** | | | | |
| Aortic valve stenosis | White, Non-Hispanic | 19 | 1.80 | 1.08-2.81 |
| | Black, Non-Hispanic | 3 | 2.32 | 0.48-6.79 |
| | Asian, Non-Hispanic | 1 | 0.89 | 0.02-4.96 |
| | Hispanic | 5 | 2.32 | 0.75-5.41 |

# Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2006-2007

| Defect | Maternal Race[1] | Count[2] | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| Coarctation of aorta | White, Non-Hispanic | 49 | 4.64 | 3.43-6.13 |
| | Black, Non-Hispanic | 3 | 2.32 | 0.48-6.79 |
| | Asian, Non-Hispanic | 2 | 1.78 | 0.22-6.44 |
| | Hispanic | 5 | 2.32 | 0.75-5.41 |
| Hypoplastic left heart syndrome | White, Non-Hispanic | 20 | 1.89 | 1.16-2.93 |
| | Black, Non-Hispanic | 0 | 0.00 | 0.00-2.86 |
| | Asian, Non-Hispanic | 1 | 0.89 | 0.02-4.96 |
| | Hispanic | 5 | 2.32 | 0.75-5.41 |
| Interrupted aortic arch (type A and NOS) | White, Non-Hispanic | 4 | 0.38 | 0.10-0.97 |
| | Black, Non-Hispanic | 0 | 0.00 | 0.00-2.86 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 0 | 0.00 | 0.00-1.71 |
| ***Other Cardiovascular*** | | | | |
| Other cardiovascular | White, Non-Hispanic | 180 | 17.05 | 14.65-19.73 |
| | Black, Non-Hispanic | 23 | 17.81 | 11.29-26.72 |
| | Asian, Non-Hispanic | 12 | 10.69 | 5.52-18.67 |
| | Hispanic | 34 | 15.77 | 10.92-22.04 |
| ***Patent Ductus Arteriosus*** | | | | |
| Patent ductus arteriosus | White, Non-Hispanic | 193 | 18.28 | 15.79-21.05 |
| | Black, Non-Hispanic | 26 | 20.13 | 13.15-29.50 |
| | Asian, Non-Hispanic | 19 | 16.92 | 10.19-26.43 |
| | Hispanic | 32 | 14.84 | 10.15-20.96 |
| ***Right-Sided Obstruction*** | | | | |
| Pulmonary stenosis, valvular | White, Non-Hispanic | 63 | 5.97 | 4.58-7.63 |
| | Black, Non-Hispanic | 7 | 5.42 | 2.18-11.17 |
| | Asian, Non-Hispanic | 7 | 6.23 | 2.51-12.85 |
| | Hispanic | 18 | 8.35 | 4.95-13.20 |
| Pulmonary valve atresia w/intact septum | White, Non-Hispanic | 6 | 0.57 | 0.21-1.24 |
| | Black, Non-Hispanic | 0 | 0.00 | 0.00-2.86 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 1 | 0.46 | 0.01-2.58 |

# Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2006-2007

| Defect | Maternal Race[1] | Count[2] | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| Pulmonary valve atresia with VSD | White, Non-Hispanic | 3 | 0.28 | 0.06-0.83 |
| | Black, Non-Hispanic | 0 | 0.00 | 0.00-2.86 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 3 | 1.39 | 0.29-4.07 |
| Tricuspid valve atresia | White, Non-Hispanic | 7 | 0.66 | 0.27-1.37 |
| | Black, Non-Hispanic | 0 | 0.00 | 0.00-2.86 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 0 | 0.00 | 0.00-1.71 |
| ***Septal Defects*** | | | | |
| ASD (secundum and NOS) | White, Non-Hispanic | 181 | 17.14 | 14.73-19.83 |
| | Black, Non-Hispanic | 15 | 11.62 | 6.50-19.16 |
| | Asian, Non-Hispanic | 17 | 15.14 | 8.82-24.24 |
| | Hispanic | 26 | 12.06 | 7.88-17.67 |
| VSD (membranous and NOS) | White, Non-Hispanic | 96 | 9.09 | 7.36-11.10 |
| | Black, Non-Hispanic | 14 | 10.84 | 5.93-18.19 |
| | Asian, Non-Hispanic | 10 | 8.91 | 4.27-16.38 |
| | Hispanic | 22 | 10.21 | 6.40-15.45 |
| VSD, conoventricular/malalignment | White, Non-Hispanic | 10 | 0.95 | 0.45-1.74 |
| | Black, Non-Hispanic | 3 | 2.32 | 0.48-6.79 |
| | Asian, Non-Hispanic | 4 | 3.56 | 0.97-9.12 |
| | Hispanic | 2 | 0.93 | 0.11-3.35 |
| ***Single Ventricle and L-TGA*** | | | | |
| L-TGA | White, Non-Hispanic | 6 | 0.57 | 0.21-1.24 |
| | Black, Non-Hispanic | 0 | 0.00 | 0.00-2.86 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 0 | 0.00 | 0.00-1.71 |
| Single ventricle | White, Non-Hispanic | 10 | 0.95 | 0.45-1.74 |
| | Black, Non-Hispanic | 1 | 0.77 | 0.02-4.31 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 1 | 0.46 | 0.01-2.58 |

# Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2006-2007

| Defect | Maternal Race[1] | Count[2] | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| ***Respiratory*** | | | | |
| Choanal atresia | White, Non-Hispanic | 12 | 1.14 | 0.59-1.99 |
| | Black, Non-Hispanic | 0 | 0.00 | 0.00-2.86 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 2 | 0.93 | 0.11-3.35 |
| Lung anomalies | White, Non-Hispanic | 16 | 1.52 | 0.87-2.46 |
| | Black, Non-Hispanic | 2 | 1.55 | 0.19-5.59 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 3 | 1.39 | 0.29-4.07 |
| Other respiratory | White, Non-Hispanic | 15 | 1.42 | 0.80-2.34 |
| | Black, Non-Hispanic | 5 | 3.87 | 1.26-9.04 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 2 | 0.93 | 0.11-3.35 |
| ***Orofacial*** | | | | |
| Cleft lip w/ and w/o cleft palate | White, Non-Hispanic | 81 | 7.67 | 6.09-9.53 |
| | Black, Non-Hispanic | 7 | 5.42 | 2.18-11.17 |
| | Asian, Non-Hispanic | 13 | 11.58 | 6.17-19.80 |
| | Hispanic | 12 | 5.57 | 2.88-9.72 |
| Cleft palate w/o cleft lip | White, Non-Hispanic | 57 | 5.40 | 4.09-6.99 |
| | Black, Non-Hispanic | 0 | 0.00 | 0.00-2.86 |
| | Asian, Non-Hispanic | 2 | 1.78 | 0.22-6.44 |
| | Hispanic | 12 | 5.57 | 2.88-9.72 |
| Pierre Robin sequence | White, Non-Hispanic | 18 | 1.70 | 1.01-2.69 |
| | Black, Non-Hispanic | 0 | 0.00 | 0.00-2.86 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 5 | 2.32 | 0.75-5.41 |
| Other orofacial | White, Non-Hispanic | 52 | 4.92 | 3.68-6.46 |
| | Black, Non-Hispanic | 11 | 8.52 | 4.25-15.24 |
| | Asian, Non-Hispanic | 2 | 1.78 | 0.22-6.44 |
| | Hispanic | 6 | 2.78 | 1.02-6.06 |

# Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2006-2007

| Defect | Maternal Race[1] | Count[2] | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| ***Gastrointestinal*** | | | | |
| Biliary atresia | White, Non-Hispanic | 6 | 0.57 | 0.21-1.24 |
| | Black, Non-Hispanic | 0 | 0.00 | 0.00-2.86 |
| | Asian, Non-Hispanic | 1 | 0.89 | 0.02-4.96 |
| | Hispanic | 1 | 0.46 | 0.01-2.58 |
| Esophageal atresia/tracheoesophageal Fistula | White, Non-Hispanic | 25 | 2.37 | 1.53-3.49 |
| | Black, Non-Hispanic | 3 | 2.32 | 0.48-6.79 |
| | Asian, Non-Hispanic | 1 | 0.89 | 0.02-4.96 |
| | Hispanic | 6 | 2.78 | 1.02-6.06 |
| Hirschsprung disease | White, Non-Hispanic | 16 | 1.52 | 0.87-2.46 |
| | Black, Non-Hispanic | 1 | 0.77 | 0.02-4.31 |
| | Asian, Non-Hispanic | 2 | 1.78 | 0.22-6.44 |
| | Hispanic | 2 | 0.93 | 0.11-3.35 |
| Rectal and large intestinal atresia/stenosis | White, Non-Hispanic | 36 | 3.41 | 2.39-4.72 |
| | Black, Non-Hispanic | 1 | 0.77 | 0.02-4.31 |
| | Asian, Non-Hispanic | 4 | 3.56 | 0.97-9.12 |
| | Hispanic | 12 | 5.57 | 2.88-9.72 |
| Small intestinal atresia | White, Non-Hispanic | 41 | 3.88 | 2.79-5.27 |
| | Black, Non-Hispanic | 5 | 3.87 | 1.26-9.04 |
| | Asian, Non-Hispanic | 2 | 1.78 | 0.22-6.44 |
| | Hispanic | 6 | 2.78 | 1.02-6.06 |
| Other gastrointestinal | White, Non-Hispanic | 64 | 6.06 | 4.67-7.74 |
| | Black, Non-Hispanic | 8 | 6.19 | 2.67-12.21 |
| | Asian, Non-Hispanic | 3 | 2.67 | 0.55-7.81 |
| | Hispanic | 17 | 7.89 | 4.59-12.63 |

# Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2006-2007

| Defect | Maternal Race[1] | Count[2] | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| ***Genitourinary*** | | | | |
| Bladder exstrophy | White, Non-Hispanic | 1 | 0.09 | 0.00-0.53 |
| | Black, Non-Hispanic | 0 | 0.00 | 0.00-2.86 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 0 | 0.00 | 0.00-1.71 |
| Cloacal exstrophy | White, Non-Hispanic | 0 | 0.00 | 0.00-0.35 |
| | Black, Non-Hispanic | 0 | 0.00 | 0.00-2.86 |
| | Asian, Non-Hispanic | 1 | 0.89 | 0.02-4.96 |
| | Hispanic | 1 | 0.46 | 0.01-2.58 |
| Hypospadias, 2nd or 3rd degree | White, Non-Hispanic | 139 | 13.16 | 11.07-15.54 |
| | Black, Non-Hispanic | 15 | 11.62 | 6.50-19.16 |
| | Asian, Non-Hispanic | 8 | 7.13 | 3.08-14.04 |
| | Hispanic | 12 | 5.57 | 2.88-9.72 |
| Obstructive genitourinary defect | White, Non-Hispanic | 157 | 14.87 | 12.63-17.38 |
| | Black, Non-Hispanic | 18 | 13.94 | 8.26-22.03 |
| | Asian, Non-Hispanic | 18 | 16.03 | 9.50-25.34 |
| | Hispanic | 39 | 18.09 | 12.86-24.73 |
| Renal agenesis/hypoplasia | White, Non-Hispanic | 1 | 0.09 | 0.00-0.53 |
| | Black, Non-Hispanic | 1 | 0.77 | 0.02-4.31 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 0 | 0.00 | 0.00-1.71 |
| Other genitourinary | White, Non-Hispanic | 189 | 17.90 | 15.44-20.64 |
| | Black, Non-Hispanic | 17 | 13.16 | 7.67-21.08 |
| | Asian, Non-Hispanic | 27 | 24.05 | 15.85-34.99 |
| | Hispanic | 29 | 13.45 | 9.01-19.32 |

# Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2006-2007

| Defect | Maternal Race[1] | Count[2] | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| ***Musculoskeletal*** | | | | |
| Clubfoot | White, Non-Hispanic | 132 | 12.50 | 10.46-14.82 |
| | Black, Non-Hispanic | 7 | 5.42 | 2.18-11.17 |
| | Asian, Non-Hispanic | 8 | 7.13 | 3.08-14.04 |
| | Hispanic | 28 | 12.99 | 8.63-18.77 |
| Craniosynostosis | White, Non-Hispanic | 56 | 5.30 | 4.01-6.89 |
| | Black, Non-Hispanic | 2 | 1.55 | 0.19-5.59 |
| | Asian, Non-Hispanic | 1 | 0.89 | 0.02-4.96 |
| | Hispanic | 12 | 5.57 | 2.88-9.72 |
| Diaphragmatic hernia | White, Non-Hispanic | 30 | 2.84 | 1.92-4.06 |
| | Black, Non-Hispanic | 4 | 3.10 | 0.84-7.93 |
| | Asian, Non-Hispanic | 1 | 0.89 | 0.02-4.96 |
| | Hispanic | 2 | 0.93 | 0.11-3.35 |
| Gastroschisis | White, Non-Hispanic | 29 | 2.75 | 1.84-3.94 |
| | Black, Non-Hispanic | 3 | 2.32 | 0.48-6.79 |
| | Asian, Non-Hispanic | 2 | 1.78 | 0.22-6.44 |
| | Hispanic | 12 | 5.57 | 2.88-9.72 |
| Omphalocele | White, Non-Hispanic | 11 | 1.04 | 0.52-1.86 |
| | Black, Non-Hispanic | 2 | 1.55 | 0.19-5.59 |
| | Asian, Non-Hispanic | 1 | 0.89 | 0.02-4.96 |
| | Hispanic | 4 | 1.86 | 0.51-4.75 |
| Polydactyly/syndactyly | White, Non-Hispanic | 143 | 13.54 | 11.41-15.95 |
| | Black, Non-Hispanic | 27 | 20.91 | 13.78-30.42 |
| | Asian, Non-Hispanic | 20 | 17.81 | 10.88-27.51 |
| | Hispanic | 34 | 15.77 | 10.92-22.04 |
| Reduction deformity, lower limbs | White, Non-Hispanic | 11 | 1.04 | 0.52-1.86 |
| | Black, Non-Hispanic | 3 | 2.32 | 0.48-6.79 |
| | Asian, Non-Hispanic | 1 | 0.89 | 0.02-4.96 |
| | Hispanic | 7 | 3.25 | 1.31-6.69 |

# Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2006-2007

| Defect | Maternal Race[1] | Count[2] | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| Reduction deformity, upper limbs | White, Non-Hispanic | 32 | 3.03 | 2.07-4.28 |
| | Black, Non-Hispanic | 2 | 1.55 | 0.19-5.59 |
| | Asian, Non-Hispanic | 2 | 1.78 | 0.22-6.44 |
| | Hispanic | 12 | 5.57 | 2.88-9.72 |
| Skeletal dysplasia | White, Non-Hispanic | 12 | 1.14 | 0.59-1.99 |
| | Black, Non-Hispanic | 0 | 0.00 | 0.00-2.86 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 5 | 2.32 | 0.75-5.41 |
| Other musculoskeletal | White, Non-Hispanic | 110 | 10.42 | 8.56-12.56 |
| | Black, Non-Hispanic | 11 | 8.52 | 4.25-15.24 |
| | Asian, Non-Hispanic | 13 | 11.58 | 6.17-19.80 |
| | Hispanic | 26 | 12.06 | 7.88-17.67 |
| ***Chromosomal and other Syndromes*** | | | | |
| Klinefelter syndrome | White, Non-Hispanic | 6 | 0.57 | 0.21-1.24 |
| | Black, Non-Hispanic | 0 | 0.00 | 0.00-2.86 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 1 | 0.46 | 0.01-2.58 |
| Trisomy 13 | White, Non-Hispanic | 8 | 0.76 | 0.33-1.49 |
| | Black, Non-Hispanic | 1 | 0.77 | 0.02-4.31 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 3 | 1.39 | 0.29-4.07 |
| Trisomy 18 | White, Non-Hispanic | 5 | 0.47 | 0.15-1.11 |
| | Black, Non-Hispanic | 4 | 3.10 | 0.84-7.93 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 5 | 2.32 | 0.75-5.41 |
| Trisomy 21 (Down syndrome) | White, Non-Hispanic | 133 | 12.60 | 10.55-14.93 |
| | Black, Non-Hispanic | 17 | 13.16 | 7.67-21.08 |
| | Asian, Non-Hispanic | 10 | 8.91 | 4.27-16.38 |
| | Hispanic | 23 | 10.67 | 6.76-16.01 |

# Table 12 Prevalence of Birth Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2006-2007

| Defect | Maternal Race[1] | Count[2] | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| *(cont'd)* | | | | |
| Turner syndrome | White, Non-Hispanic | 7 | 0.66 | 0.27-1.37 |
| | Black, Non-Hispanic | 0 | 0.00 | 0.00-2.86 |
| | Asian, Non-Hispanic | 1 | 0.89 | 0.02-4.96 |
| | Hispanic | 1 | 0.46 | 0.01-2.58 |
| Other chromosomal syndromes/other syndromes | White, Non-Hispanic | 118 | 11.17 | 9.25-13.38 |
| | Black, Non-Hispanic | 14 | 10.84 | 5.93-18.19 |
| | Asian, Non-Hispanic | 9 | 8.02 | 3.67-15.22 |
| | Hispanic | 25 | 11.60 | 7.51-17.12 |
| ***Other*** | | | | |
| Amniotic bands | White, Non-Hispanic | 7 | 0.66 | 0.27-1.37 |
| | Black, Non-Hispanic | 0 | 0.00 | 0.00-2.86 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 5 | 2.32 | 0.75-5.41 |
| Skin anomalies | White, Non-Hispanic | 11 | 1.04 | 0.52-1.86 |
| | Black, Non-Hispanic | 5 | 3.87 | 1.26-9.04 |
| | Asian, Non-Hispanic | 0 | 0.00 | 0.00-3.29 |
| | Hispanic | 2 | 0.93 | 0.11-3.35 |
| Other, specified | White, Non-Hispanic | 15 | 1.42 | 0.80-2.34 |
| | Black, Non-Hispanic | 2 | 1.55 | 0.19-5.59 |
| | Asian, Non-Hispanic | 1 | 0.89 | 0.02-4.96 |
| | Hispanic | 3 | 1.39 | 0.29-4.07 |

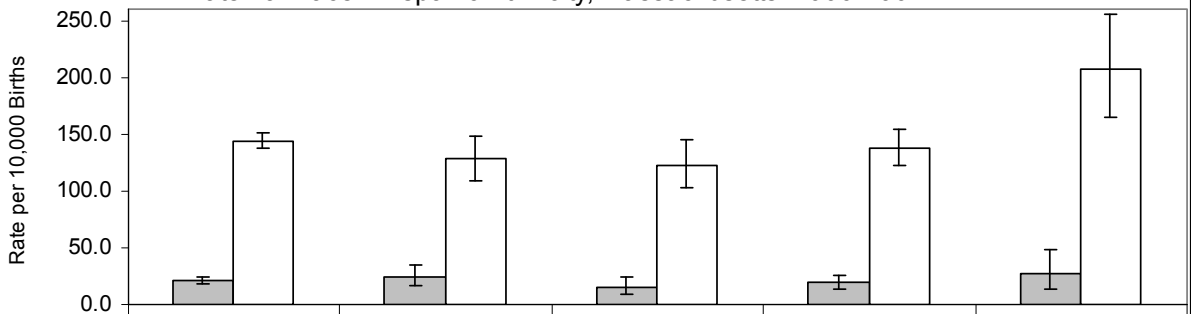
1. Due to small numbers, races classified as "other" are not included.
2. Cases can be included in more than one defect. Cases are counted once in the total for a defect category. Because only live births are presented on this table, case totals are not listed. Due to missing race of mother counts may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

# Table 13 Most Common Defects by Maternal Race/Hispanic Ethnicity for Live Births, Massachusetts: 2006-2007

| Race[1] | Defect[2] | Count | Rate per 10,000 Births | 95% Confidence Interval |
|---|---|---|---|---|
| **White, Non-Hispanic** | ASD (secundum and NOS) | 181 | 17.14 | 14.73-19.83 |
| | Obstructive genitourinary defect | 157 | 14.87 | 12.63-17.38 |
| | Polydactyly/syndactyly | 143 | 13.54 | 11.41-15.95 |
| | Hypospadias, 2nd or 3rd degree | 139 | 13.16 | 11.07-15.54 |
| | Trisomy 21 (Down syndrome) | 133 | 12.6 | 10.55-14.93 |
| **Black, Non-Hispanic** | Polydactyly/syndactyly | 27 | 20.91 | 13.78-30.42 |
| | Obstructive genitourinary defect | 18 | 13.94 | 8.26-22.03 |
| | Trisomy 21 (Down syndrome) | 17 | 13.16 | 7.67-21.08 |
| | ASD (secundum and NOS) | 15 | 11.62 | 6.50-19.16 |
| | Hypospadias, 2nd or 3rd degree | 15 | 11.62 | 6.50-19.16 |
| **Asian, Non-Hispanic** | Polydactyly/syndactyly | 20 | 17.81 | 10.88-27.51 |
| | Obstructive genitourinary defect | 18 | 16.03 | 9.50-25.34 |
| | ASD (secundum and NOS) | 17 | 15.14 | 8.82-24.24 |
| | Cleft lip w/ and w/o cleft palate | 13 | 11.58 | 6.17-19.80 |
| | VSD (membranous and NOS) | 10 | 8.91 | 4.27-16.38 |
| | Trisomy 21 (Down syndrome) | 10 | 8.91 | 4.27-16.38 |
| **Hispanic** | Obstructive genitourinary defect | 39 | 18.09 | 12.86-24.73 |
| | Polydactyly/syndactyly | 34 | 15.77 | 10.92-22.04 |
| | Clubfoot | 28 | 12.99 | 8.63-18.77 |
| | ASD (secundum and NOS) | 26 | 12.06 | 7.88-17.67 |
| | Trisomy 21 (Down syndrome) | 23 | 10.67 | 6.76-16.01 |

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

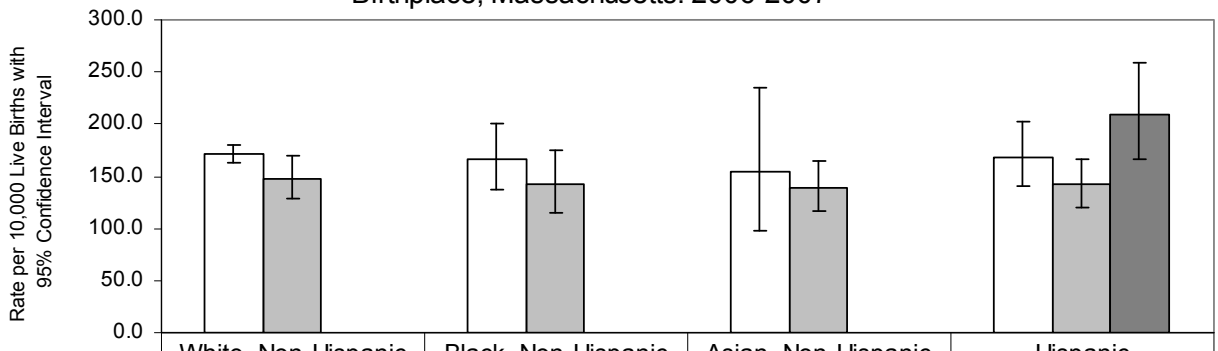
Figure 12 Age-Adjusted Prevalence of Chromosomal and All Other Defects by Maternal Race / Hispanic Ethnicity, Massachusetts: 2006-2007




| | White, Non-Hispanic | Black, Non-Hispanic | Asian, Non-Hispanic | Hispanic | Other |
|---|---|---|---|---|---|
| Chromosomal | 20.9 (18.3-23.9) | 24.0 (16.3-34.1) | 15.1 (8.8-24.2) | 19.5 (14.0-26.3) | 26.7 (13.3.-47.8) |
| All other defects | 144.0 (136.9-151.4) | 172.8 (109.0-148.8) | 122.9 (103.3-145.2) | 138.2 (123.0-154.9) | 206.4 (164.8-255.2) |

Livebirths only, N=2528

Figure 13 Prevalence of Birth Defects by Maternal Race / Hispanic Ethnicity and Birthplace, Massachusetts: 2006-2007




| | White, Non-Hispanic | Black, Non-Hispanic | Asian, Non-Hispanic | Hispanic |
|---|---|---|---|---|
| U.S.-Born* | 170.8 (162.6-179.4) | 166.6 (136.8-200.9) | 155.0 (97.2-234.7) | 168.8 (140.-201.8) |
| Non-U.S.-Born | 148.0 (128.5-169.6) | 142.9 (115.1-175.5) | 139.7 (117.3-165.1) | 141.7 (119.8-166.3) |
| U.S. Territories | ** | ** | ** | 208.6 (165.9-258.9) |

*Including American Samoa, Federated State of Micronesia, Guam, Marshall Islands, Northern Mariana Islands, Palau, and Puerto Rico for all except Hispanic
**Less than 5 cases were in this category; U.S. Territories were inlcuded in Non-U.S.-births for non-Hispanic
N=2585

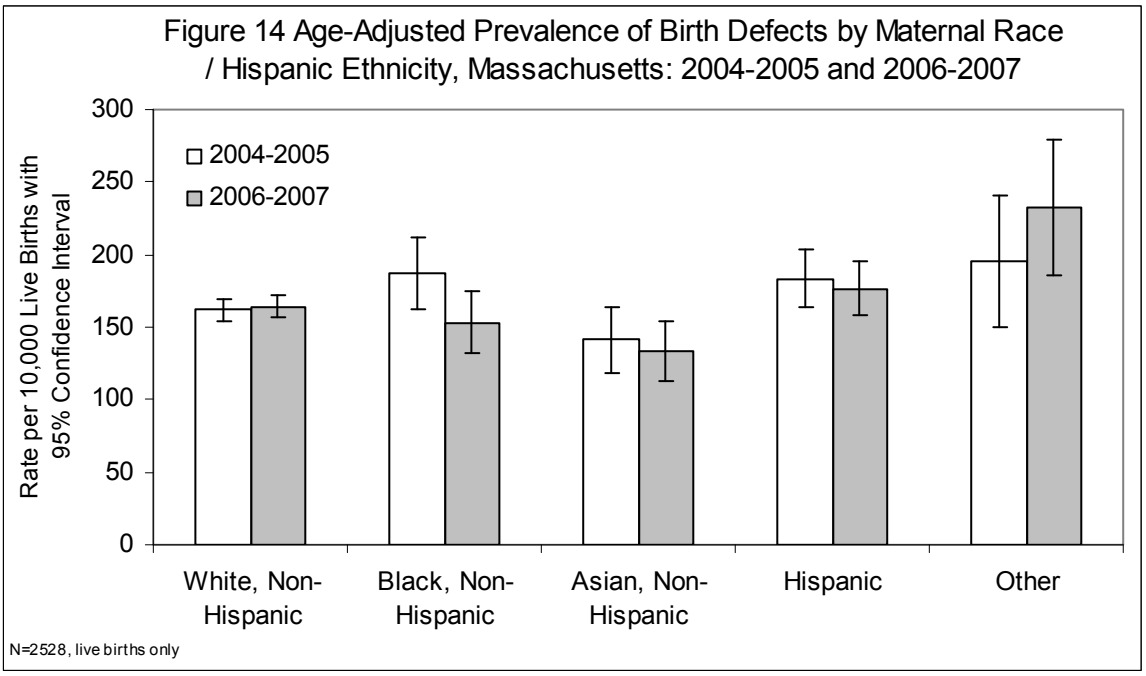


Figure 14 Age-Adjusted Prevalence of Birth Defects by Maternal Race / Hispanic Ethnicity, Massachusetts: 2004-2005 and 2006-2007
Rate per 10,000 Live Births with 95% Confidence Interval
300
250
200
150
100
50
0
2004-2005
2006-2007
White, Non-Hispanic
Black, Non-Hispanic
Asian, Non-Hispanic
Hispanic
Other
N=2528, live births only

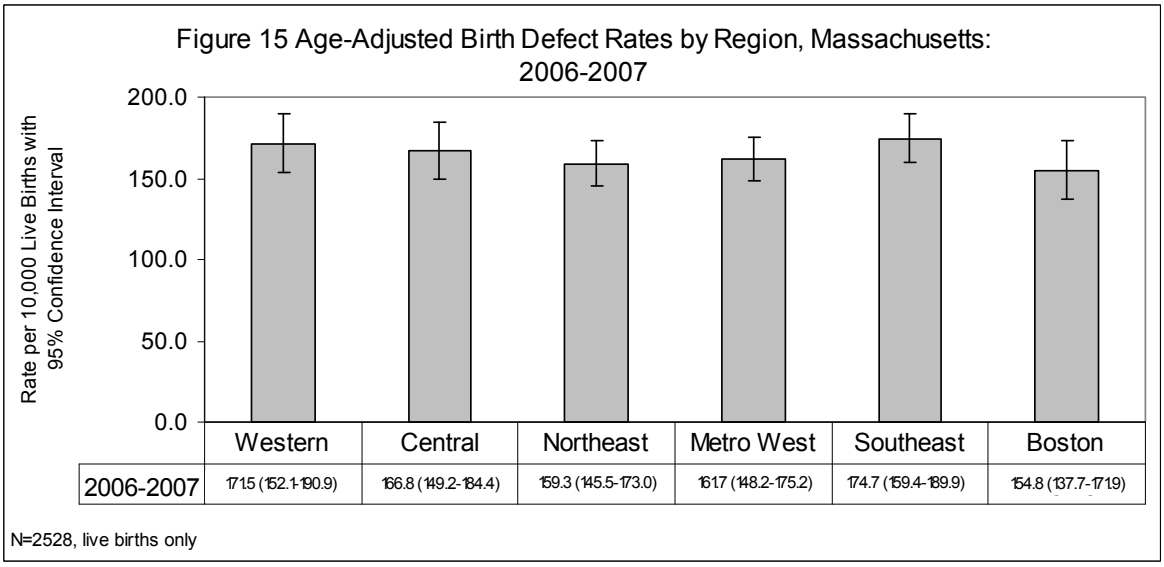
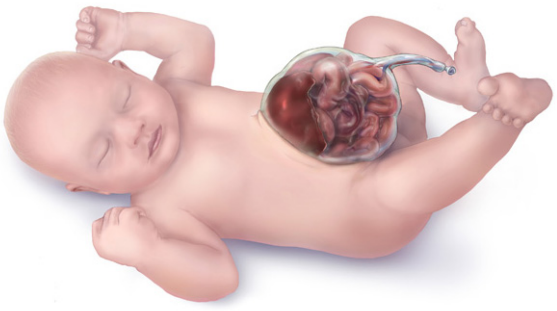


Figure 15 Age-Adjusted Birth Defect Rates by Region, Massachusetts: 2006-2007
Rate per 10,000 Live Births with 95% Confidence Interval
200.0
150.0
100.0
50.0
0.0
Western
Central
Northeast
Metro West
Southeast
Boston
2006-2007
171.5 (152.1-190.9)
166.8 (149.2-184.4)
159.3 (145.5-173.0)
161.7 (148.2-175.2)
174.7 (159.4-189.9)
154.8 (137.7-171.9)
N=2528, live births only

# Chapter 6

# Prevalence of Birth Defects by Severity, Etiology and Pattern



Baby with omphalocele

*Courtesy of the Centers for Disease Control and Prevention*

## Prevalence of Birth Defects by Severity

Birth defects cases were categorized by their level of severity. This scale was based on the usual outcome for a specific birth defect including its typical compatibility with survival, the need for immediate treatment, the need for long-term care and the amenability of the defect to correction.

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

Table 14 shows the percentage of birth defect cases by severity groups. Nearly three percent of cases had birth defects classified as “severe,” and most did not survive the neonatal period. This percentage was an underestimate of these most “severe” cases due to limitations of the data and because we were missing many “severe” defects including the estimated 80% of anencephaly cases and 50% of any neural tube defects that were electively terminated after prenatal diagnosis (Cragan and Khoury 2000).

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

“Moderate” birth defects comprised almost 70% of the total cases. All of these children needed medical follow-up, and many needed surgeries and extensive treatment.

“Mild” birth defects comprised nearly 9% of the cases. Within the classification of “mild severity,” there was variability. For example, children with microphthalmia (small eyes) could have mild reduction in the size of the globe or a more severe reduction resulting in visual loss or the need for intrusive ophthalmologic medical care. In contrast, infants with isolated dextrocardia (heart in the right side of the chest instead of the left) and no other heart defect have no clinical consequence.

## Prevalence of Birth Defects by Etiology and Pattern

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

Cases with known etiology accounted for about 18% of the birth defects (476 of 2,585 live births and stillbirths) in Massachusetts in 2006-2007. Etiology groups include “single gene,” “chromosomal,” “maternal-fetal factors” (including teratogens or uterine factors) and “other factors.”

As Figure 16 shows, single gene etiology accounted for 18.5% (88) of the known etiology cases. Single gene defects include achondroplasia, Marfan syndrome (deletion 15q21.1), Smith-Lemli-Opitz syndrome and other examples of defects categorized as Mendelian syndrome. Chromosomal etiology accounted for almost 76.7% (365) of the cases with known etiology. Cases with chromosomal etiology include trisomy 13, 18 and 21, Turner syndrome and other chromosomal duplications and deletions. Maternal-fetal and other factors accounted for about 4.6% (23) of all cases with known etiology. Maternal-fetal factors include teratogens such as maternal diabetes and uterine factors such as deformation or didelphy uterus. The “other factor” known etiology cases include conjoined twins. Cases with known etiology may also be classified by pattern, with the categories and definitions listed in Table 15. The majority of cases with known etiology fall within the “multiple majors” pattern including combinations of two or more major defects.

While the CDC estimates that about 70% of birth defects have unknown cause, the majority (82%) of birth defects cases in Massachusetts in 2006-2007 had unknown etiology (2,109 of 2,585 live births and stillbirths). These were categorized by pattern and by description in Table 15.

The “isolated” pattern is the largest category among the cases with unknown etiology, comprising about 90%. Within this pattern are three subgroups based on the number and type of defects found in a case. The largest subgroup of isolated cases have a truly solitary defect such as cleft palate, gastroschisis or anencephaly, comprising about 52% of total cases with unknown etiology. The next largest isolated pattern subgroup accounts for about 35% of the cases with unknown etiology. These cases have 2 or more defects in the same organ or body part, or they have an isolated major and some minor defects in different body parts. An example of this pattern is coarctation of the aorta and aortic stenosis (valvar). The third subgroup accounts for the remaining cases (a little over 4% of all unknown cases) within the isolated pattern and is defined by a sequence of a common primary defect with consistent related anomalies. An example of this pattern is spina bifida, clubfoot, hydrocephalus and hip dislocation. The “multiple majors” pattern comprises about 10% of cases with unknown etiology, and these cases include combinations of two or more major defects such as hydrocephalus and cleft palate and anotia. See Figure 17 for the distribution of cases with unknown etiology among these patterns.

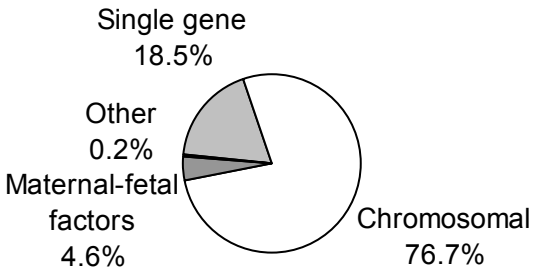
## Table 14 Percentage of Birth Defect Cases by Severity Groups, Massachusetts: 2006-2007

| SEVERITY CATEGORIES | PERCENTAGE OF BIRTH DEFECTS CASES |
|---|---|
| Severe, supportive measures, usually incompatible with life | 2.7% |
| Serious, may be correctable, most have long-term needs | 19.0% |
| Moderate, most correctable, many have long-term needs | 69.6% |
| Mild, may be correctable, minimal long-term needs | 8.7% |
| | |

## Table 15 Patterns and Etiology of Birth Defect Cases, Massachusetts: 2006-2007

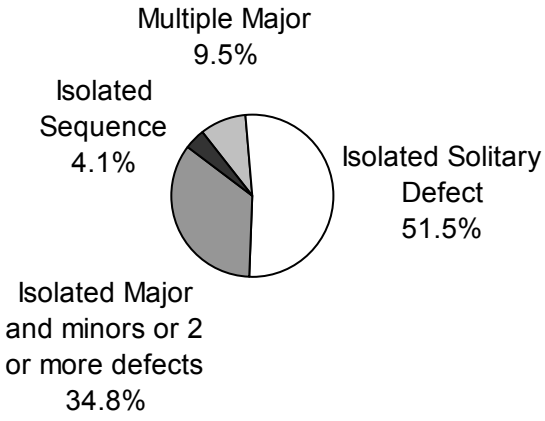
| Pattern | Description | Cases | |
|---|---|---|---|
| | | Known Etiology | Unknown Etiology |
| Isolated | Solitary defect | 4 | 1,087 |
| | Major and minors (different organ/body parts) or 2 or more defects (same organ/body part) | 7 | 734 |
| | Sequence: Common primary defect with consistent, related anomalies | 4 | 87 |
| Multiple Majors | 2 or more major defects in different organs/body parts | 461 | 201 |
| Total | | 476 (18.4%) | 2,109 (81.6%) |

Figure 16 Birth Defect Cases with Known Etiology, Massachusetts: 2006-2007




N=476

Figure 17 Patterns of Birth Defect Cases with Unknown Etiology, Massachusetts: 2006-2007




N=2109

# Appendices

Technical Notes

Definitions

2006-2007 Denominators Used in Calculating Rates

Birth Defects Codes and Exclusions by Defect Category

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

Birth Defects by Severity

Glossary of Selected Birth Defect Terms

Map of Massachusetts EOHHS-Defined Regions and Age-Adjusted Overall Birth Defects Prevalence, 2006-2007

References

## Technical Notes

### Data Sources

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

### Prevalence, Rates and Confidence Intervals

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

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

Cases/total number live births x 10,000

Fetal deaths are included in the numerator but not in the denominator, so the result is technically a ratio and not a rate. This method of calculating rates is consistent with the national "Guidelines for Conducting Birth Defects Surveillance." (National Birth Defects Prevention Network, June 2004) Because the number of fetal deaths is so small, the inclusion of fetal deaths in the denominator does not substantially change the ratio.

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

### Assignment of Race/Ethnicity

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

### Calculation of 2007 Dollars

2007 dollars were calculated from the Gross Domestic Product Deflator Inflation Index, an inflation calculator for adjusting costs from one year to another using the Gross Domestic Product (GDP) Deflator inflation index of 1.0319 representing the inflation from 2005 to 2007. (Based on this calculator, costs increased from 125 million to 129 million during this period.) This inflation calculator is based on the inflation rate during the US Government Fiscal Year, which begins on October 1 and ends on September 30. http://cost.jsc.nasa.gov/inflateGDP.html.

**Assignment of Severity**

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

A new automated algorithm was created for the 2004-2005 report based on modified rules that had been developed to determine severity of defects described in previous birth defects surveillance reports, and the program was validated using the data described in the reports. The automated process for 2006-2007 data was able to assign severity levels to about 96% of the cases, with the remaining 4% assigned manually by the Center Clinical Geneticist. The process that included the automated categorization system produced percentages of birth defects within each of the four severity categories in 2006-2007 that were similar to those attained in each of the categories in previous reports.

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

## Definitions

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

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

1 pound = 453.6 grams
1,000 grams = 2 pounds and 3 ounces

**Infant**
A child whose age is less than one year (365 days).

**Infant Death**
Death of a child whose age is less than one year.

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

**Live Birth**
Any infant who breathes or shows any other evidence of life (such as beating of the heart, pulsation of the umbilical cord, or definite movement of voluntary muscles) after separation from the mother's uterus, regardless of the duration of gestation.

**Neonatal**
Infant under 28 days of age.

**Neonatal Death**
Death of a child whose age is less than 28 days.

**Plurality**
The number of births to a woman produced in the same gestational period. A singleton is the birth of one infant; twins represent the births of two infants, etc.

**Resident Birth**
The birth of an infant whose mother reports that her usual place of residence is in Massachusetts. In Massachusetts, a resident is a person with a permanent address in one of the 351 cities or towns.

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

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

# 2006 and 2007 Populations Used in Calculating Rates

| Numbers of Live Births to MA Residents* | | | | |
|---|---|---|---|---|
| | | **2006 N=77670** | **2007 N=77934** | **Total N=155,604** |
| By Maternal Age | <20 | 4778 | 4993 | 9771 |
| | 20-24 | 12420 | 12598 | 25018 |
| | 25-29 | 18633 | 19283 | 37916 |
| | 30-34 | 23611 | 23334 | 46945 |
| | 35+ | 18218 | 17724 | 35942 |
| | Unknown | 10** | 2** | 12** |
| By Infant's Sex | Male | 39670 | 40026 | 79696 |
| | Female | 37999 | 37907 | 75906 |
| By Plurality | Singleton | 74146 | 74498 | 148644 |
| | Multiple Birth | 3524 | 3436 | 6960 |
| By Maternal Race/Ethnicity | White | 52975 | 52620 | 105595 |
| | Black | 6452 | 6462 | 12914 |
| | Hispanic | 10696 | 10861 | 21577 |
| | Asian/PI | 5469 | 5758 | 11227 |
| | *American Indian* | *151*** | *157*** | *308*** |
| | *Other, non-Hisp* | *1844*** | *1967*** | *3811*** |
| | *Unknown* | *83*** | *109*** | *192*** |

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

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

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

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

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

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

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

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

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

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

# ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2006-2007

| BPA label | BPA Code | # of Defects |
|---|---|---|
| **Central Nervous System** | | |
| Agyria and lissencephaly | 742240 | 6 |
| Anencephaly | 740020 | 8 |
| Arrhinencephaly | 742270 | 1 |
| Brain cysts: Porencephaly / porencephalic | 742410 | 1 |
| Brain: Other specified anomalies / cortical atrophy / cranial nerve defects | 742480 | 7 |
| Cerebellar Hypoplasia | 742235 | 5 |
| Cerebellum anomalies | 742230 | 6 |
| Corpus callosum anomalies (don't code colpocephaly with ACC) | 742210 | 58 |
| Dandy-Walker Malformation | 742310 | 19 |
| Encephalocele, Occipital | 742000 | 3 |
| Enlarged brain and head / enlarged head / enlarged brain / megalencephaly / macrocephaly | 742400 | 12 |
| Holoprosencephaly, Alobar | 742265 | 2 |
| Holoprosencephaly, Lobar | 742267 | 1 |
| Holoprosencephaly, NOS | 742260 | 1 |
| Holoprosencephaly, Semilobar | 742266 | 3 |
| Hydranencephaly | 742320 | 5 |
| Hydrocephaly, NOS | 742390 | 10 |
| Hydrocephaly, Anomalies of Aqueduct of Sylvius | 742300 | 7 |
| Hydrocephaly, Other Specified | 742380 | 5 |
| Lipomeningomyelocele, Highest level unspecified, No mentioned hydrocephalus, closed lesion | 741849 | 1 |
| Lipomeningomyelocele, Highest level, lumbar, No mentioned hydrocephalus, closed lesion | 741843 | 4 |
| Meningocele, Highest level unspecified, Hydrocephalus, other (aqueduct of Sylvius) or NOS, closed lesion | 741419 | 1 |
| Meningocele, Highest level, sacral, No mentioned hydrocephalus, closed lesion | 741814 | 1 |
| Meningocele, Highest level, sacral, No mentioned hydrocephalus, open lesion | 741714 | 1 |
| Meningocele, Highest level, thoracic, No mentioned hydrocephalus, closed lesion | 741812 | 1 |
| Meningomyelocele/myelomeningocele, Highest level unspecified, Hydrocephalus, other (aqueduct of Sylvius) or NOS, unspec. open/closed | 741509 | 1 |
| Meningomyelocele/myelomeningocele, Highest level, lumbar, Arnold Chiari malformation ± hydrocephalus, closed lesion | 741103 | 3 |
| Meningomyelocele/myelomeningocele, Highest level, lumbar, Arnold Chiari malformation ± hydrocephalus, open lesion | 741003 | 5 |
| Meningomyelocele/myelomeningocele, Highest level, lumbar, Arnold Chiari malformation ± hydrocephalus, unspec. open/closed lesion | 741203 | 1 |
| Meningomyelocele/myelomeningocele, Highest level, lumbar, Hydrocephalus, other (aqueduct of Sylvius) or NOS, unspec. open/closed | 741503 | 1 |
| Meningomyelocele/myelomeningocele, Highest level, sacral, No mentioned hydrocephalus, closed lesion | 741804 | 1 |
| Meningomyelocele/myelomeningocele, Highest level, sacral, No mentioned hydrocephalus, open lesion | 741704 | 1 |
| Microcephalus | 742100 | 31 |
| Microgyria / polymicrogyria | 742250 | 9 |
| Myelocele, Highest level, cervical, No mentioned hydrocephalus, closed lesion | 741821 | 1 |

# ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2006-2007

| BPA label (cont'd) | BPA Code | # of Defects |
|---|---|---|
| **Eye** | | |
| Absence of iris/Aniridia, Bilateral | 743424 | 1 |
| Absence of lens/Congenital Aphakia, Laterality Unk | 743300 | 1 |
| Anophthalmos, Bilateral | 743004 | 1 |
| Anophthalmos, Right | 743002 | 1 |
| Axenfeld Anomaly, Bilateral | 743469 | 1 |
| Axenfeld Anomaly, Left | 743466 | 1 |
| Buphthalmos/Congenital Glaucoma, Bilateral | 743204 | 9 |
| Buphthalmos/Congenital Glaucoma, Left | 743201 | 1 |
| Buphthalmos/Congenital Glaucoma, Right | 743202 | 2 |
| Cataract, anterior polar, Left | 743351 | 2 |
| Cataract, anterior polar, Right | 743352 | 1 |
| Cataract, NOS, Left | 743321 | 9 |
| Cataract, NOS, Bilateral | 743324 | 10 |
| Cataract, NOS, Right | 743322 | 13 |
| Cataract, other specified, Left | 743361 | 2 |
| Cataract, other specified, Bilateral | 743364 | 3 |
| Microphthalmos, Bilateral | 743104 | 9 |
| Microphthalmos, Laterality Unk | 743100 | 1 |
| Microphthalmos, Left | 743101 | 2 |
| Microphthalmos, Right | 743102 | 3 |
| Peters Anomaly, Bilateral | 743464 | 1 |
| Peters Anomaly, Right | 743462 | 1 |
| S Anterior segment: OS colobomas and anomalies (Use for Rieger SYNDROME, use 759800. For Reiger ANOMALY, use 743470-474) | 743480 | 2 |
| S Anterior segment: Unspecified colobomas and anomalies | 743490 | 1 |
| S Choroid: Coloboma | 743535 | 2 |
| S Cornea, other anomalies. Excludes: megalocornea (use 743.220) | 743410 | 2 |
| S Eyelid: Ectropion | 743610 | 1 |
| S Eyelids: Coloboma | 743636 | 2 |
| **Ear** | | |
| Anotia, Left | 744011 | 1 |
| Anotia, Right | 744012 | 1 |
| Microtia, Bilateral | 744214 | 5 |
| Microtia, Left | 744211 | 7 |
| Microtia, Right | 744212 | 19 |
| S # Ear : ACCESSORY auricle / polyotia | 744100 | 2 |
| S Absence or stricture of auditory canal | 744000 | 5 |
| S Anomaly of inner ear / congenital anomaly of membranous labyrinth or organ of Corti | 744030 | 4 |
| S Anomaly of middle ear / fusion of ossicles | 744020 | 3 |
| S Ear: Other specified anomalies (see also 744.230) / #DARWIN tubercle | 744280 | 6 |
| S Ear: Other misshapen ear / cleft / malformed /#POINTED / # ELFIN,pixie-like / # LOP / # CAULIFLOWER / # ABSENT or decreased cartilage -- a conditional exclusion if <36wks | 744230 | 10 |

# ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2006-2007

| BPA label (cont'd) | BPA Code | # of Defects |
|---|---|---|
| S Ear: Unspecified anomalies / congenital anomaly (any part) / anomaly,deformity,NOS | 744300 | 1 |
| **Cardiovascular** | | |
| **Anomalous Pulmonary Venous Connection** | | |
| Partial anomalous pulmonary venous return/connection/drainage | 747430 | 10 |
| Total anomalous pulmonary venous return/connection/drainage | 747420 | 14 |
| **Atrioventricular Canal Defects** | | |
| Atrial septal defect, primum type (ASD1) | 745600 | 8 |
| Common Atrium | 745610 | 5 |
| Complete atrioventricular canal (CAVC) | 745630 | 47 |
| Endocardial cushion defect, NOS | 745690 | 2 |
| Endocardial cushion defect, Other specified | 745680 | 5 |
| Ventricular septal defect, inflow type (subtricuspid, canal-type) (VSDavc) | 745685 | 5 |
| Atrial septal defect, primum type (ASD1) | 745600 | 8 |
| **Conotruncal (Outlet) and Aortic Arch** | | |
| Dextro-transposition of great arteries (dTGA, dTGV) w/ intact ventricular septum | 745100 | 21 |
| Dextro-transposition of great arteries (dTGA, dTGV) w/ ventricular septal defect | 745110 | 13 |
| Double-outlet right ventricle (DORV) with normally related great arteries | 745185 | 9 |
| Double-outlet right ventricle (DORV) with transposed great arteries | 745186 | 8 |
| Double-outlet right ventricle (DORV), NOS | 745189 | 4 |
| Double-outlet right ventricle (DORV), Other Specified | 745188 | 1 |
| Interrupted aortic arch, type B | 747217 | 8 |
| Pulmonary atresia with VSD (tetralogy of Fallot with pulmonary atresia) | 747310 | 10 |
| Tetralogy of Fallot | 745200 | 58 |
| Truncus Arteriosus | 745000 | 6 |
| **Ebstein Anomaly** | | |
| Ebstein Malformation or Anomaly | 746200 | 7 |
| **Heterotaxy (Laterality Defects)** | | |
| Complete situs inversus w/ dextrocardia | 759300 | 2 |
| Heterotaxy, NOS | 759395 | 3 |
| Situs ambiguus, left; left isomerism | 759360 | 3 |
| Situs ambiguus, sidedness NOS | 759380 | 7 |
| Situs ambiguus, sidedness unclear | 759370 | 1 |
| Situs inversus abdominis | 759330 | 2 |
| Complete situs inversus w/ dextrocardia | 759300 | 2 |
| Heterotaxy, NOS | 759395 | 3 |
| **Left – Sided Obstruction** | | |
| Aortic stenosis, valvar | 746300 | 30 |
| Coarctation of the aorta (COA), preductal (proximal) | 747100 | 2 |
| Coarctation of the aorta, juxtaductal | 747120 | 3 |
| Coarctation of the aorta, NOS | 747190 | 57 |
| Hypoplastic left heart syndrome | 746700 | 29 |
| Interrupted aortic arch, NOS | 747215 | 3 |
| Interrupted aortic arch, type A | 747216 | 2 |

# ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2006-2007

| BPA label (cont'd) | BPA Code | # of Defects |
|---|---|---|
| **Right – Sided Obstruction** | | |
| Pulmonary valve atresia with VSD (not TOF variant 747.310) | 746030 | 6 |
| Pulmonary valve atresia/intact ventricular septum | 746000 | 7 |
| Pulmonic stenosis, valvar | 746010 | 98 |
| Tricuspid atresia | 746100 | 7 |
| **Septal Defects** | | |
| Atrial septal defect, NOS | 745599 | 70 |
| Atrial septal defect, OS | 745580 | 3 |
| Atrial septal defect, Secundum type (ASD2) | 745510 | 184 |
| Ventricular septal defect, NOS | 745490 | 9 |
| Ventricular septal defect, Malalignment-type (type I, subarterial) (VSDmal) | 745487 | 19 |
| Ventricular septal defect, Perimembranous (type II, membranous) (VSDmem) | 745485 | 143 |
| **Single Ventricle and L – TGA** | | |
| L-TGA /Corrected transposition of great vessels / ventricular inversion. Excludes: dextrocardia (use 746800) | 745120 | 6 |
| Single ventricle, NOS | 745300 | 4 |
| Single ventricle, Double Inlet Left Ventricle | 745310 | 4 |
| Single ventricle, Double Inlet Right Ventricle | 745320 | 4 |
| **Other Cardiovascular** | | |
| "Pulmonic" or pulmonary atresia, stenosis, or hypoplasia, NOS w/ no mention of whether valve or artery | 746995 | 1 |
| Anomalies of coronary artery or sinus | 746885 | 13 |
| Aorta: Hypoplasia | 747210 | 7 |
| Aorta: Other specified anomalies | 747280 | 1 |
| Aorta: Persistent right aortic arch | 747230 | 28 |
| Aorta: Vascular ring / double aortic arch / vascular ring compression of trachea | 747250 | 10 |
| Aortic septal defect / aortopulmonary window. Excludes: atrial septal defect(use 745.590) | 745010 | 2 |
| Aortic valve: bicuspid BAV / insufficiency or regurgitation / # MILD',MINIMAL', 'TRIVIAL',or 'PHYSIOLOGIC' ~ | 746400 | 36 |
| Aortic valve: Other specified anomalies / aortic valve atresia. Excludes: supravalvular aortic stenosis(747.220) | 746480 | 20 |
| Aortic valve: Unspecified anomalies | 746490 | 1 |
| Bicuspid Aortic Valve (BAV); new code for cases starting DOB 1/1/07 | 746470 | 24 |
| Cerebral vessels: Other anomalies / vein of Galen | 747810 | 2 |
| Cor triatriatum | 746820 | 4 |
| Great veins: Other specified anomalies (includes IVC interruption, bilateral SVC) | 747480 | 16 |
| Heart: Other specified anomalies / ectopia cordis / mesocardia / conduction defects, NOS | 746880 | 53 |
| Hypoplastic left ventricle. Excludes: hypoplastic left heart syndrome (746.700) | 746881 | 4 |
| Hypoplastic right heart or right ventricle / Uhl's disease (parchment RV) | 746882 | 4 |
| Mitral valve: Absence, atresia, or hypoplasia | 746505 | 6 |
| Mitral valve: Congenital mitral stenosis | 746500 | 5 |
| Persistent left superior vena cava | 747410 | 35 |
| Pulmonary infundibular (subvalvular) stenosis | 746830 | 13 |
| Pulmonary valve: Other specified anomalies. Excludes: infundibular PS (746.830) | 746080 | 22 |
| S Circulatory system: Other specified anomalies. Excludes cong aneurysms: coronary ~ (746.880),peripheral ~ (747.640), pulmonary~ (747.330), retinal ~(743.510), ruptured cerebral arterioven | 747880 | 3 |
| S Peripheral arteries: Other anomalies / aberrant subclavian artery | 747640 | 31 |
| S Pulmonary artery: atresia, absence or agenesis. Use 746.995 if artery or valve is not specified | 747300 | 1 |
| S Pulmonary artery: other specified / pulmonary artery hypoplasia | 747380 | 6 |
| S Pulmonary artery: stenosis. Use 746.995 if artery or valve is not specified | 747320 | 9 |
| Situs: Dextrocardia without situs inversus / dextrocardia with situs solitus. Excludes: dextrocardia with situs inversus (use_759.300) | 746800 | 9 |
| Supra-aortic stenosis / supravalvular aortic stenosis. Excludes: aortic stenosis, congenital(see 746.300) | 747220 | 5 |

# ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2006-2007

| BPA label (cont'd) | BPA Code | # of Defects |
|---|---|---|
| Valves: Unspecified anomalies | 746900 | 1 |

## Respiratory

| | | |
|---|---|---|
| Choanal atresia, Bilateral | 748014 | 6 |
| Choanal atresia, Left | 748011 | 1 |
| Choanal atresia, Right | 748012 | 7 |
| Larynx: Cleft / laryngotracheoesophageal cleft / 1/04: use for laryngeal atresia/stenosis | 748385 | 36 |
| Other anomalies of trachea. Excludes: vascular ring compression of the trachea (use 747.250) | 748330 | 6 |
| S Choanal stenosis (For NBDPS: choanal atresia = 748.010 etc) | 748000 | 5 |
| S Lung agenesis or aplasia | 748500 | 2 |
| S Lung cysts: CCAM (cong cystic adenomatoid malf), Other specified | 748480 | 11 |
| S Lung cysts: Single | 748400 | 1 |
| S Lung: other specified dysplasia / fusion of lobes | 748580 | 2 |
| S Lung: sequestration | 748520 | 7 |
| S Other anomalies of bronchus | 748350 | 1 |
| S Stenosis of bronchus | 748340 | 1 |

## Orofacial

| | | |
|---|---|---|
| Cleft hard palate, Bilateral | 749010 | 1 |
| Cleft hard palate, Central | 749020 | 5 |
| Cleft hard palate, NOS | 749030 | 21 |
| Cleft hard palate, Unilateral, Left | 749001 | 1 |
| Cleft hard palate, Unilateral, Right | 749002 | 1 |
| Cleft lip and palate, Bilateral cleft lip | 749210 | 24 |
| Cleft lip and palate, Central cleft lip | 749220 | 1 |
| Cleft lip and palate, NOS | 749290 | 3 |
| Cleft lip and palate, Unilateral cleft lip, Left | 749201 | 23 |
| Cleft lip and palate, Unilateral cleft lip, Right | 749202 | 13 |
| Cleft lip, Bilateral | 749110 | 5 |
| Cleft lip, Central | 749120 | 1 |
| Cleft lip, NOS | 749195 | 1 |
| Cleft lip, Unilateral, Left | 749101 | 31 |
| Cleft lip, Unilateral, Right | 749102 | 14 |
| Cleft palate, NOS | 749090 | 12 |
| Cleft soft palate, Central | 749060 | 3 |
| Cleft soft palate, NOS | 749070 | 30 |
| Cleft soft palate, Unilateral, Right | 749042 | 1 |
| Nose: Fissured, notched, or cleft | 748120 | 1 |
| Nose: OS anomalies (For NBDPS:nasal pyriform aperture stenosis is here, not Cho Sten)/ small nose and nostril / absent nasal septum / # FLAT or WIDE nasal bridge/ #BEAKED nose | 748180 | 16 |
| Pierre Robin sequence (not a true "syndrome") | 524080 | 23 |
| S Branchial cleft, sinus, fistula, cyst, or pit | 744400 | 26 |
| S Cleft: Incomplete CL/ microform /pseudo / fused lip /healed lip | 749190 | 5 |

# ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2006-2007

| BPA label (cont'd) | BPA Code | # of Defects |
|---|---|---|
| S Face or neck: Other specified anomalies (6/03 eg. facial cleft) | 744880 | 7 |

## Gastrointestinal

| | | |
|---|---|---|
| Anal atresia with fistula | 751230 | 23 |
| Anal atresia without mention of fistula | 751240 | 15 |
| Annular pancreas | 751720 | 4 |
| Biliary atresia, extrahepatic or NOS (use 751670 for intrahepatic) | 751650 | 5 |
| Bronchoesophageal fistula with or without mention of esophageal atresia | 750330 | 1 |
| Congenital hiatal hernia / cardiac displacement through esophageal hiatus / partial thoracic stomach. Excludes: congenital diaphragmatic hernia (756.610) | 750600 | 1 |
| Duplication of anus, appendix, cecum, or intestine / enterogenous cyst | 751500 | 6 |
| Ectopic (displaced, anteriorly placed) anus | 751530 | 17 |
| Esophageal atresia with TE fistula | 750310 | 34 |
| Esophageal atresia without TE fistula | 750300 | 4 |
| Hirschsprung disease, NOS | 751330 | 12 |
| Hirschsprung disease: Long – segment (aganglionosis beyond rectum) | 751310 | 17 |
| Hirschsprung disease: Short – segment (aganglionosis involving no more than the anal sphincter and the rectum) | 751320 | 6 |
| Intestinal atresia/stenosis, duodenum | 751100 | 17 |
| Intestinal atresia/stenosis, ileum | 751120 | 8 |
| Intestinal atresia/stenosis, jejunum | 751110 | 22 |
| Intestinal atresia/stenosis, large intestine, NOS | 751200 | 3 |
| Malrotation: cecum and/or colon | 751400 | 1 |
| Malrotation: Other specified and unspecified | 751490 | 48 |
| Malrotation: small intestine alone | 751495 | 3 |
| Meckel's diverticulum | 751010 | 10 |
| Microcolon | 751520 | 1 |
| Other specified anomalies of intestine / rectal fissures | 751580 | 2 |
| Other specified anomalies of upper alimentary tract | 750800 | 1 |
| Persistent omphalomesenteric duct / persistent vitelline duct | 751000 | 4 |
| Rectal atresia/stenosis without mention of fistula | 751220 | 6 |
| Tracheoesophageal fistula without mention of esophageal atresia | 750320 | 1 |

## Genitourinary

| | | |
|---|---|---|
| # Urachus: PATENT | 753700 | 4 |
| Bladder exstrophy | 753500 | 1 |
| Cloacal exstrophy | 751550 | 2 |
| Double urethra or urinary meatus | 753840 | 3 |
| Genital organs: Other specified anomalies / microgenitalia / macrogenitalia | 752880 | 1 |
| Gyne: OS anomalies of cervix, vagina, or external female genitalia /# VAGINAL tags / # HYMENAL tags | 752480 | 2 |
| Gyne: S Ovaries, Multiple cysts | 752085 | 3 |

# ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2006-2007

| BPA label (cont'd) | BPA Code | # of Defects |
|---|---|---|
| Gyne: S Ovaries, Other specified anomalies | 752080 | 1 |
| Gyne: Uterus, other anomalies / bicornuate/ unicornis | 752380 | 2 |
| Gyne: Vagina, absence or atresia complete or partial | 752410 | 1 |
| Hypospadias, Second Degree | 752606 | 71 |
| Hypospadias, Second Degree with Chordee | 752626 | 58 |
| Hypospadias, Third Degree | 752607 | 13 |
| Hypospadias, Third Degree with Chordee | 752627 | 39 |
| Indeterminate sex, NOS / ambiguous genitalia | 752790 | 21 |
| Kidneys: Polycystic, adult type (APKD) | 753120 | 1 |
| Kidneys: Polycystic, infantile type (IPKD) | 753110 | 2 |
| Other and unspecified atresia and stenosis of urethra and bladder neck | 753690 | 2 |
| Other atresia, or stenosis of bladder neck | 753610 | 1 |
| Other specified anomalies of bladder and urethra | 753880 | 5 |
| Penis: Other anomalies / concealed penis / absent or hooded foreskin / #REDUNDANT foreskin (Redundant foreskin is never coded.) | 752860 | 84 |
| Penis: Small / hypoplastic / micropenis | 752865 | 14 |
| Renal agenesis, bilateral | 753000 | 1 |
| Renal agenesis, left + renal hypoplasia, right | 753006 | 2 |
| Renal hypoplasia, bilateral | 753005 | 1 |
| S Absence of testis / monorchidism, NOS | 752800 | 1 |
| S Atresia, stricture, or stenosis of ureter / ureteropelvic junction obstruction or stenosis /ureterovesical junction obstruction or stenosis / hypoplastic ureter | 753210 | 58 |
| S Congenital hydronephrosis / pyelocaliectasis | 753200 | 225 |
| S Kidney/renal: cyst, single | 753100 | 9 |
| S Kidney: Double or triple, pelvis / pyelon duplex or triplex | 753310 | 20 |
| S Kidney: Ectopic / pelvic | 753330 | 5 |
| S Megaloureter, NOS / hydroureter | 753220 | 48 |
| S Ureter: Accessory / double ureter / duplex collecting system | 753410 | 30 |
| S Ureter: Ectopic | 753420 | 11 |
| S Ureter: Other specified anomalies / ureterocele | 753480 | 30 |
| S Ureter: Variations of vesicoureteral reflux | 753485 | 80 |
| S Kidney: Lobulated, fused, or horseshoe / crossed fused ectopia | 753320 | 7 |
| S Kidneys: Multicystic renal dysplasia / multicystic kidney | 753160 | 29 |
| Testis and scrotum: Other anomalies / polyorchidism / bifid scrotum. Excludes: torsion of the testes or spermatic cord(608.200) | 752820 | 11 |
| True hermaphroditism / ovotestis | 752700 | 3 |

## Musculoskeletal

| | | |
|---|---|---|
| = Achondroplasia | 756430 | 5 |
| = Amyotrophia congenita (= one specific type of arthrogryposis) | 756840 | 2 |
| = Arthrogryposis multiplex congenita / distal arthrogryposis syndrome. Temporarily includes: one or more flexion contractures of individual joints | 755800 | 18 |
| = Cleidocranial dysostosis | 755555 | 1 |
| = Osteogenesis imperfecta | 756500 | 6 |
| = Other specified osteodystrophies | 756580 | 3 |

# ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2006-2007

| BPA label (cont'd) | BPA Code | # of Defects |
|---|---|---|
| = Spondyloepiphyseal dysplasia | 756460 | 1 |
| = Thanatophoric dwarfism | 756447 | 2 |
| Absence of foot or toes, Bilateral | 755349 | 5 |
| Absence of foot or toes, Left | 755346 | 4 |
| Absence of foot or toes, Right | 755347 | 6 |
| Absence of hand or fingers, Bilateral | 755249 | 3 |
| Absence of hand or fingers, Left | 755246 | 18 |
| Absence of hand or fingers, Right | 755247 | 11 |
| Absence of the forearm and hand, Bilateral | 755244 | 1 |
| Absence of the forearm and hand, Left | 755241 | 1 |
| Absence of the forearm and hand, Right | 755242 | 1 |
| Absence of the lower leg and foot, Left | 755341 | 1 |
| Absence of thigh only (lower leg and foot present), Right | 755332 | 1 |
| Congenital postural scoliosis | 754200 | 20 |
| Craniosynostosis, Coronal, Bilateral | 756014 | 6 |
| Craniosynostosis, Coronal, Laterality Unk | 756010 | 1 |
| Craniosynostosis, Coronal, Left | 756011 | 6 |
| Craniosynostosis, Coronal, Right | 756012 | 3 |
| Craniosynostosis, Lambdoidal, Left | 756021 | 2 |
| Craniosynostosis, Lambdoidal, Right | 756022 | 1 |
| Craniosynostosis, Metopic | 756006 | 14 |
| Craniosynostosis, Sagittal | 756005 | 42 |
| Diaghragmatic hernia, Morgagni, Bilateral | 756619 | 1 |
| Diaphragmatic hernia, Bochdalek, Left | 756611 | 11 |
| Diaphragmatic hernia, Bochdalek, Right | 756612 | 2 |
| Diaphragmatic hernia, Morgagni, Left | 756616 | 1 |
| Diaphragmatic hernia, Morgagni, Right | 756617 | 4 |
| Diaphragmatic hernia, NOS (inc. absent/hemidiaphragm), Laterality Unk | 756600 | 1 |
| Diaphragmatic hernia, NOS (inc. absent/hemidiaphragm), Left | 756601 | 21 |
| Diaphragmatic hernia, NOS (inc. absent/hemidiaphragm), Right | 756602 | 3 |
| Fibular aplasia/hypoplasia, Left | 755371 | 1 |
| Fibular aplasia/hypoplasia, Right | 755372 | 1 |
| Gastroschisis | 756710 | 56 |
| Longitudinal deficiency of arm, NOS, Bilateral | 755254 | 1 |
| Omphalocele | 756700 | 21 |
| Prune belly syndrome | 756720 | 2 |
| Radial aplasia/hypoplasia, Bilateral | 755269 | 4 |
| Radial aplasia/hypoplasia, Left | 755266 | 4 |
| Radial aplasia/hypoplasia, Right | 755267 | 3 |
| S Anomalies of elbow and upper arm | 755540 | 1 |
| S Anomalies of fingers /camptodactyly/macro- /brachy-/clino-, triphalangeal thumb. Excludes:acrocephalosyndactyly(see756.050) /Apert synd(see756.055) | 755500 | 46 |
| S Bowing, tibia and/or fibula | 754410 | 3 |
| S Clubfoot, NOS / talipes, NOS | 754730 | 92 |
| S Clubfoot: # METATARSUS varus or adductus | 754520 | 16 |
| S Clubfoot: Complex varus deformities | 754530 | 3 |

# ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2006-2007

| BPA label (cont'd) | BPA Code | # of Defects |
|---|---|---|
| S Clubfoot: Talipes calcaneovarus | 754510 | 1 |
| S Clubfoot: Talipes equinovarus | 754500 | 72 |
| S Diaphragm: Eventration | 756620 | 2 |
| S Diaphragm: Other specified anomalies | 756680 | 2 |
| S Genu recurvatum | 754430 | 3 |
| S Lower limb: hypoplasia / Toes, feet, legs: hypoplasia. Excludes: aplasia of or absent lower limb (see 755.3) | 755685 | 4 |
| S Other specified deformities of ankle and / or toes / dorsiflexion of foot. Excludes: widely spaced first and second toes (use_755.600) | 754780 | 3 |
| S Other specified valgus deformities of foot | 754680 | 2 |
| S Polydactyly fingers / postaxial polydactyly, Type A | 755005 | 63 |
| S Polydactyly: Accessory big toe (preaxial) | 755030 | 13 |
| S Polydactyly: Accessory digits foot, NOS (preaxial, postaxial not specified) | 755096 | 9 |
| S Polydactyly: Accessory digits hand, NOS (preaxial, postaxial not specified) | 755095 | 9 |
| S Polydactyly: Accessory thumbs (preaxial polydactyly) | 755010 | 50 |
| S Polydactyly: Accessory toes (postaxial) | 755020 | 53 |
| S Ribs: Absence | 756300 | 20 |
| S Ribs: Extra | 756330 | 13 |
| S Ribs: Fused | 756320 | 8 |
| S Ribs: Other anomalies | 756340 | 4 |
| S Syndactyly: Fused fingers | 755100 | 10 |
| S Syndactyly: Fused toes | 755120 | 33 |
| S Syndactyly: Unspecified (see below for specified site) | 755190 | 3 |
| S Syndactyly: Unspecified (webbed vs. fused) thumb and / or fingers, NOS | 755193 | 10 |
| S Syndactyly: Unspecified (webbed vs. fused) Toes | 755199 | 1 |
| S Syndactyly: Unspecified toes | 755194 | 3 |
| S Syndactyly: Webbed fingers | 755110 | 20 |
| S Syndactyly: Webbed toes / # WEBBING between the second and third toes. | 755130 | 37 |
| S Talipes calcaneovalgus | 754600 | 12 |
| S Unspecified reduction defect of lower limb | 755390 | 2 |
| S Unspecified valgus deformities of foot | 754690 | 2 |
| S Unspecified varus deformities of feet | 754590 | 1 |
| S Upper leg: anomalies / anteversion of femur | 755650 | 1 |
| S Upper limb: Hypoplasia / Fingers, hands, or arms: hypoplasia. Excludes: aplasia or absent upper limb (see 755.2) | 755585 | 9 |
| S Upper limb: Other specified anomalies / hyperextensibility of upper limb / shortening of upper limb | 755580 | 3 |
| Sacral agenesis | 756175 | 3 |
| Skull and face bone: Other specified anomalies / localized skull defects / mid-facial hypoplasia / prominent maxilla/hypotelorism / # FLATocciput / # PROMINENT occiput. | 756080 | 14 |
| Spine: Kyphosis / kyphoscoliosis | 756120 | 9 |
| Split-Foot, Bilateral | 755359 | 1 |
| Split-Foot, Right | 755357 | 2 |
| Split-Hand, Bilateral | 755259 | 3 |
| Split-Hand, Left | 755256 | 3 |
| Split-Hand, Right | 755257 | 3 |
| Sternum: Other anomalies / double ossification center in manubrium / bifid/ short | 756380 | 1 |
| Thumb only missing or hypoplastic, Bilateral | 755264 | 2 |
| Thumb only missing or hypoplastic, Left | 755261 | 2 |
| Thumb only missing or hypoplastic, Right | 755262 | 1 |

# ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2006-2007

| BPA label (cont'd) | BPA Code | # of Defects |
|---|---|---|
| Tibial aplasia/hypoplasia, Right | 755367 | 1 |
| Ulnar aplasia/hypoplasia, Bilateral | 755274 | 2 |
| Ulnar aplasia/hypoplasia, Right | 755272 | 2 |
| Unspecified anomalies of spine | 756190 | 1 |
| Vertebrae, cervical: anomalies | 756140 | 2 |

## Chromosomal and Other Syndromes

| | | |
|---|---|---|
| = 1/06 Clarified: "Other Trans" Incl Unbal AND Other Bal Translocations, OS. Excludes bal trans in normal (758.400) | 758540 | 7 |
| = 22q11 deletion (Added 7/04: apply to 1/01. Also code phenotype if stated, eg. DGS 279.110) | 758370 | 21 |
| = Autosome OS: Other spec anomalies / marker / 8/02: Ring, derivative, mosaic, isochrome, "additional" material / 3/03 inversions. 2/08 Never code "pericentric inv 9" | 758580 | 15 |
| = Crouzon disease | 756040 | 1 |
| = Deletion 17p or 18p / deletion of short arm chromosome 17 or 18 | 758350 | 1 |
| = Deletion 4p / Wolff-Hirschorn syndrome | 758320 | 5 |
| = Deletion 5p / Cri du chat syndrome | 758310 | 2 |
| = Deletion: Autosome (not X or Y)(ie. #1-16, 4q,5q,19,20) / (From 8/02, used for 22q11, prior to the specific 22q code added 7/04) | 758380 | 9 |
| = Deletion: unspecified autosome | 758390 | 1 |
| = Down syndrome: diagnosed clinically, but no karyotype report in medical record | 758090 | 1 |
| = Down syndrome: mosaic | 758040 | 1 |
| = Down syndrome: translocation 21, duplication 21q, Robertsonian translocation, isochrome 21q | 758020 | 3 |
| = Down syndrome: trisomy 21 | 758000 | 192 |
| = Goldenhar syndrome / oculoauriculovertebral dysplasia | 756060 | 4 |
| = Hemifacial microsomia | 756065 | 6 |
| = Klinefelter syndrome: 47, XXY | 758700 | 7 |
| = Mosaic XO/XY, 45X/46XY. Excludes: with Turner phenotype(758.610) | 758800 | 3 |
| = Other craniofacial syndromes / Hallermann-Streiff syndrome | 756046 | 3 |
| = Other specified acrocephalosyndactylies | 756057 | 2 |
| = Sex chromosome : Unspecified anomaly | 758890 | 1 |
| = Sex chromosome: Other specified anomaly / fragile X | 758880 | 5 |
| = Treacher-Collins syndrome / Mandibulofacial dysostosis | 756045 | 2 |
| = Triploidy | 758586 | 3 |
| = Trisomy 13 (archaic Patau syndrome): cytogenetics result in record | 758100 | 16 |
| = Trisomy 18 (archaic Edwards syndrome): cytogenetics result in record | 758200 | 24 |
| = Trisomy 8 | 758500 | 1 |
| = Trisomy, partial / 8/02 "partial trisomy" = "duplication". But, for "dup NOS" use 758930 | 758530 | 10 |
| = Trisomy: Other total trisomy syndromes / trisomy 22 / trisomy, NOS | 758520 | 3 |
| = Tuberous sclerosis / Bourneville's disease | 759500 | 6 |
| = Turner phenotype: karyotype 45,X [XO] Note: The 7586xx code series that follows excludes pure gonadal dysgenesis(752.720) | 758600 | 9 |
| = Turner phenotype: variant karyotypes, eg. isochromosome, mosaic (eg X, XX,XY), partial X deletion, ring X chromosome. Excludes: Turner phenotype with normal karyotype | 758610 | 6 |
| = Unspecified chromosome: Unspecified anomaly of chromosome(s) | 758990 | 1 |
| = XXX female / 47XXX / Triple X syndrome | 758850 | 8 |

# ICD9/BPA Codes with Counts - Live Births and Stillbirths, Massachusetts 2006-2007

| BPA label (cont'd) | BPA Code | # of Defects |
|---|---|---|
| = XYY, male / 47,XYY / mosaic XYY male | 758840 | 1 |
| = Malf OS: VATER/VACTERL/Acardia/ Angelman/Bloom/CHARGE/hemihyper/Meckel-Gruber/Neu-Laxova/PentalogyCantrell/ Sotos/ TownesBrock/ WalkerWarburg/ Weaver / 10/02 VCFS,Shprintzen: code also chrome/FISH | 759890 | 22 |
| = Malf. Syndromes/face: Aarskog /BOF /BOR /Fraser /FreemanSheldon / Kabuki / Miller-Dieker/ Noonan /Opitz G / oral-facial-digita/ Oto-palato-digital / Septo-optic dysplasia / Waardenburg / Williams | 759800 | 18 |
| = Malf. Syndromes/limbs: Baller-Gerold/ Carpenter / caudal regression /Fryns/ Holt-Oram / Klippel-Trenaunay-Webe/ LimbBodyWall /Roberts/ Rubinstein-Taybi / sirenomelia / thrombocytopenia-absent radius | 759840 | 6 |
| = Malf. Syndromes/metabolic: Alagille /Alport / Beckwith-Wiedemann / Johansen-Blizzard/ leprechaunism / Lowe/ Menkes(kinky hair) /Prader-Willi/ Zellweger | 759870 | 11 |
| = Malf. Syndromes/other skeletal: Marfan / Stickler/ Beemer Langer | 759860 | 4 |
| = Malf. Syndromes/short stature: Smith-Lemli-Optiz /de Lange / Cockayne / Laurence-Moon-Biedl / Russell-Silver / Seckel | 759820 | 3 |
| DiGeorge S (10/02: Use for specific phenotype with chrome/FISH 22q, if available 758.370) | 279110 | 10 |
| = 1/06 Clarified: "Other Trans" Incl Unbal AND Other Bal Translocations, OS. Excludes bal trans in normal (758.400) | 758540 | 7 |
| = 22q11 deletion (Added 7/04: apply to 1/01. Also code phenotype if stated, eg. DGS 279.110) | 758370 | 21 |
| = Autosome OS: Other spec anomalies / marker / 8/02: Ring, derivative, mosaic, isochrome, "additional" material / 3/03 inversions. 2/08 Never code "pericentric inv 9" | 758580 | 15 |

## Other

| | | |
|---|---|---|
| = Collodion baby | 757110 | 2 |
| = Ectodermal dysplasia. Excludes: Ellis-van Creveld syndrome (756.525) | 757340 | 2 |
| = Epidermolysis bullosa | 757330 | 2 |
| = X-linked ichthyosis | 757196 | 5 |
| Adrenogenital syndrome / adrenal hyperplasia | 255200 | 2 |
| Amniotic band sequence | 658800 | 16 |
| Anomalies of thymus / absent thymus / # THYMICHYPERTROPHY | 759240 | 3 |
| Ichthyosiform erythroderma | 757197 | 1 |
| Multiple congenital anomalies (In MA, ="MCA NOS", not "MCA no specific dx") / anomaly, multiple, NOS / deformity, multiple, NOS | 759700 | 3 |
| Skin: Other specified anomalies / scalp defects. For specified anomalies of skin see 757.390. For specified anomalies of hair, see_757480. For specified anomalies of nails_757.580 | 757800 | 7 |
| Spleen: Absence / asplenia | 759000 | 6 |
| Spleen: Accessory / 8/02 Use for polysplenia, though not exactly the same | 759040 | 8 |
| Spleen: Hypoplasia | 759010 | 1 |

# Birth Defects by Severity

**Severe, supportive measures, usually incompatible with life**

Anencephaly

Bilateral renal agenesis

Trisomy 13

Trisomy 18

Severe identifiable syndrome or condition, not elsewhere classified

Severe isolated defects, not elsewhere classified

Multiple severe defects, (Severe MCA,NEC)

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

Achondroplasia

Aniridia

Anophthalmia

Arthrogryposis

Biliary atresia

Bladder exstrophy

Cloacal exstrophy

CHD, multiple mod – severe, not elsewhere classified

Double outlet right ventricle

Encephalocele

Heterotaxy with CHD

Holoprosencephaly

Hypoplastic left heart syndrome

Limb reductions, mod – severe

Osteogenesis imperfecta

Sacral agenesis, caudal regression, sirenomelia

Single ventricle

Spina bifida

Amniotic band complex

Down syndrome

Mod serious syndrome/condition, not elsewhere classified

Mod serious defect, not elsewhere classified

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

**Moderate, most correctable, many have long – term needs**

Aortic valve stenosis

Atrial septal defect

Atrioventricular canal AVC / AVSD / ECD

Choanal atresia

Cleft lip/ palate

Coarctation

Cataract, glaucoma

Clubfoot

Coloboma

CHD, Mult mild – mod not, listed elsewhere

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

Craniosynostosis

Dandy – Walker malformation

Diaphragmatic hernia

Esophageal atresia/ TEF

Ebstein anomaly

Gastroschisis

Genitourinary, obstructive

Hirschsprung disease

Penis, buried, hidden

Hydrocephalus

Hypospadias, 2nd or 3rd degree

Intestinal atresia: duod, jejunal, ileal

Imperforate anus/rectal atresia and stenosis

Interrupted aortic arch

Klinefelter syndrome

Limb reductions, mild – mod

Malrotation

Microcephaly

Microtia

Omphalocele

Pulm sequestration/ CCAM

Pulmonary atresia/stenosis

Tethered cord

Tetralogy of Fallot

Total /partial anom. pulm venous return

Transposition great arteries

Tricuspid atresia/stenosis

Turner syndrome

Ventricular septal defect

Moderate syndrome/condition, not elsewhere classified

Moderate defect, not elsewhere classified

Moderate multiple severe defects, (Moderate MCA,NEC)

DiGeorge/ VCF/ 22q11 del spectrum

Goldenhar/FAVS/ OAVD

**Mild, may be correctable, minimal long – term needs**

Bicuspid aortic valve

Meckel's diverticulum

Microphthalmia

CHD, OS, asymptomatic

Patent ductus arteriosus

Polydactyly, accessory thumbs, syndactyly

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

Mild defect, not listed above

## Glossary of Selected Birth Defects Terms[1]

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

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

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

**Anencephaly:** Congenital absence of the skull and brain.

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

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

**Anotia:** Congenital absence of the ear.

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

**Arthrogryposis:** Multiple congenital contractures of various joints.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

**Hypertrophy:** Overgrowth due to enlargement of existing cells.

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

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

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

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

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

**Meninges:** Membranes that cover the brain and spinal cord.

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

**Microphthalmia:** Congenital smallness of the eye globe.

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

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

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

**NOS:** Not Otherwise Specified

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

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

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

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

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

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

**Renal agenesis:** Congenital absence of the kidney.

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

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

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

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

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

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

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

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

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

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

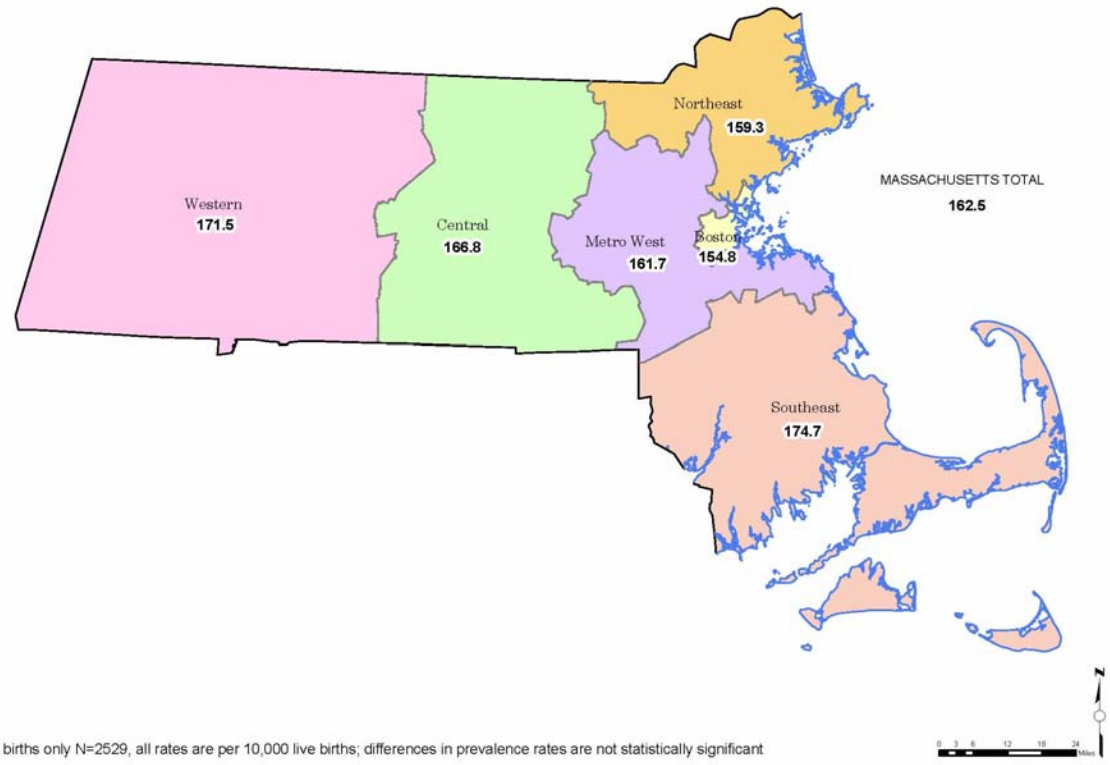
**Trisomy 21:** See **Down syndrome**.

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

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

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

## Map of Massachusetts EOHHS-Designated Regions and Overall, Age-Adjusted Rate of Birth Defects, 2006-2007*




*live births only N=2529, all rates are per 10,000 live births; differences in prevalence rates are not statistically significant

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

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

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

## Frequency of Multivitamin Use during Month Prior to Pregnancy, Massachusetts, 2007/2008[1]

| Frequency of multivitamin or prenatal vitamin consumption | % of respondents (N=2973) |
|---|---|
| None | 47.7 |
| 1 – 3 times per week | 9.1 |
| 4 – 6 times per week | 7.1 |
| Every day of the week | 36.1 |

## References


Cary, J., M. Feldkamp, et al. (2005). "An etiologic classification of congenital malformations, including orofacial clefts, in Utah." Proceedings of the Greenwood Genetic Center **24**: 69.

CDC (1995). "Economic costs of birth defects and cerebral palsy-United States, 1992." MMWR Morb Mortal Wkly Rep **44**(37): 694-9.

CDC (1998). "Trends in infant mortality attributable to birth defects-United States, 1980-1995." MMWR Morb Mortal Wkly Rep **47**(37): 773-8.

CDC (2004). "Spina bifida and anencephaly before and after folic acid mandate-United States, 1995-1996 and 1999-2000." MMWR Morb Mortal Wkly Rep **53**(17): 362-5.

CDC (2006). "Improved national prevalence estimates for 18 selected major birth defects-United States, 1999-2001." MMWR Morb Mortal Wkly Rep **54**(51): 1301-5.

CDC (2009). "Assisted reproductive technology and trends in low birthweight-Massachusetts, 1997-2004." MMWR Morb Mortal Wkly Rep **58**(3): 49-52.

Cragan, J. D. and M. J. Khoury (2000). "Effect of prenatal diagnosis on epidemiologic studies of birth defects." Epidemiology **11**(6): 695-9.

DHHS (2000). Healthy People 2010: Understanding and Improving Health. U. S. D. o. H. a. H. Services. Washington, DC, U.S. Government Printing Office.

Ethen, M. K. and M. A. Canfield (2002). "Impact of including elective pregnancy terminations before 20 weeks gestation on birth defect rates." Teratology **66 Suppl 1**: S32-5.

Fillingham, A. and J. Rankin (2008). "Prevalence, prenatal diagnosis and survival of gastroschisis." Prenatal Diagnosis **28**(13): 1232-1237.

Forrester, M. B. and R. D. Merz (1999). "Epidemiology of abdominal wall defects, Hawaii, 1986-1997." Teratology **60**(3): 117-23.

Forrester, M. B., R. D. Merz, et al. (1998). "Impact of prenatal diagnosis and elective termination on the prevalence of selected birth defects in Hawaii." Am J Epidemiol **148**(12): 1206-11.

Harris, J. A. and L. James (1997). "State-by-state cost of birth defects-1992." Teratology **56**(1-2): 11-6.

Heseker, H. B., J. B. Mason, et al. (2008). "Not all cases of neural-tube defect can be prevented by increasing the intake of folic acid." Br J Nutr: 1-8.

Holmes, L. (1994). Congenital Malformations. Principles and Practices of Pediatrics. Philadelphia, J.B. Lippincott Co.**:** 277-279.

Lu, E. (2010). Bureau of Family Health and Nutrition, Massachusetts Department of Public Health**:** personal communication.

MADPH (2007). Massachusetts Births 2005. Boston, Division of Research and Epidemiology, Center for Health Information, Statistics, Research, and Evaluation, Massachusetts Department of Public Health.

MADPH (2007). Massachusetts Deaths 2004. C. f. H. I. Division of Research and Epidemiology, Statistics, Research, and Evaluation, Massachusetts Department of Public Health. Boston.

MADPH (2008). Massachusetts Births 2006. Boston, Division of Research and Epidemiology, Bureau of Health Information, Statistics, Research, and Evaluation, Massachusetts Department of Public Health.

MADPH (2009). Massachusetts Births 2007. Boston, Division of Research and Epidemiology, Bureau of Health Information, Statistics, Research, and Evaluation, Massachusetts Department of Public Health.

MarchofDimes (2006). Birth Defects. March of Dimes Birth Defects Foundation.

Mills, J. L. and C. Signore (2004). "Neural tube defect rates before and after food fortification with folic acid." Birth Defects Res A Clin Mol Teratol **70**(11): 844-5.

Mosley, B. S., M. A. Cleves, et al. (2009). "Neural tube defects and maternal folate intake among pregnancies conceived after folic acid fortification in the United States." Am J Epidemiol **169**(1): 9-17.

Oken E., K.P. Kleinman, et al. (2003). "A nearly continuous measure of birth weight for gestational age using a United States national reference." BMC Pediatr. 3 (6). (Open Access: http://www.biomedcentral.com/1471-2431/3/6)

Peller, A. J., M. N. Westgate, et al. (2004). "Trends in congenital malformations, 1974-1999: effect of prenatal diagnosis and elective termination." Obstet Gynecol **104**(5 Pt 1): 957-64.

Rasmussen, S. A., R. S. Olney, et al. (2003). "Guidelines for case classification for the National Birth Defects Prevention Study." Birth Defects Res A Clin Mol Teratol **67**(3): 193-201.

Reefhuis, J., M. A. Honein, et al. (2009). "Assisted reproductive technology and major structural birth defects in the United States." Hum Reprod **24**(2): 360-6.
Sever, L., Ed. (2004). Guidelines for Conducting Birth Defects Surveillance. Atlanta, National Birth Defects Prevention Network, Inc.
Vu, L. T., K. K. Nobuhara, et al. (2008). "Increasing Prevalence of Gastroschisis: Population-based Study in California." The Journal of Pediatrics **152**(6): 807-811.
Werler, M., A. Mitchel, et al. (manuscript). Is there epidemiological evidence to support vascular disruption as a pathogenesis of gastroschisis? Boston.
Sunderam S., Chang J. et al (2009). "Assisted reproductive technology surveillance—United States, 2006." MMWR Surveill Summ 58(SS05): 1-25.
Zhang Z, Macaluso M, Cohen BB, Schieve LA, Nannini, A, Chen, M, et al. Accuracy of assisted reproductive technology information of the Massachusetts birth certificate, 1997–2000. *Fertil Steril* 2009: In Press.

Massachusetts Birth Defects 2006-2007 Evaluation Form

**TO OUR READERS:**
To better serve our users, we are enclosing this evaluation form. Please complete this questionnaire and return it to the address at the bottom of the 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 them in detail. |
| Do you have other comments or suggestions? |

Thank you.

Please return your comments to:

Cathleen A. Higgins, Surveillance Coordinator

Center for Birth Defects Research and Prevention
Bureau of Family Health & Nutrition
Massachusetts Department of Public Health
250 Washington Street, 5th floor, Boston, MA 02108