

Massachusetts

Massachusetts Birth Defects Monitoring Program (MBDMP)

Purpose: Surveillance, Research, Public health program evaluation, Assist community health assessments

Partner: Hospitals, Environmental Agencies/Organizations, Advocacy Groups, Universities, Maternal and Child Health Programs, State Lab

Program status: Currently collecting data

Start year: 1997

Earliest year of available data: 1999

Organizational location: Department of Public Health (Bureau of Family Health and Nutrition)

Population covered annually: 69,000

Statewide: Yes

Current legislation or rule: Massachusetts General Laws, Chapter 111, Section 67E in 1963. In 2002 the Massachusetts legislature amended this statute, expanding the birth defects monitoring program. In 2009 regulations for a Congenital Anomalies Registry, 105 CMR 302.000, were promulgated.

Legislation year enacted: 1963 (amended 2002, regulations 2009)

Case Definition

Pregnancy outcome: Livebirths (All gestational ages and birth weights), Fetal deaths - stillbirths, spontaneous abortions, etc. (≥ 20 weeks gestation or ≥ 350 grams), Elective terminations (Unspecified non-live births (elective terminations at any gestational age, spontaneous losses < 20 weeks and < 350 grams))

Age: 1 year

Residence: In- and out-of-state births to state residents

Surveillance Methods

Case ascertainment: Active Case Finding

Vital records: Birth certificates, Death certificates, Matched birth/death file, Fetal birth certificate

Delivery hospitals: Disease index or discharge index, Postmortem/pathology logs, Specialty outpatient clinics

Pediatric & tertiary care hospitals: Disease index or discharge index, Postmortem/pathology logs, Specialty outpatient clinics

Other specialty facilities: Prenatal diagnostic facilities (ultrasound, etc.), Cytogenetic laboratories, Genetic counseling/clinical genetic facilities

Other sources: Accepting physician reports sent to us.

Case Ascertainment

Conditions warranting chart review in newborn period: Any chart with an ICD-9-CM code 740-759/ICD-10-CM code Q00-Q99, Any chart with a selected list of ICD-9-CM codes outside 740-759/ICD-10-CM codes outside Q00-Q99, Any chart with selected defects or medical conditions (i.e. abnormal facies, congenital heart disease), All stillborn infants, All prenatally diagnosed or suspected cases, Any birth certificate with a major birth defect box checked

Conditions warranting chart review beyond the newborn period: All infant deaths (excluding prematurity), Any infant with a codable defect

Coding: CDC coding system based on BPA

Data Collected

Infant/fetus: Identification information (name, address, date-of-birth, etc.), Demographic information (race/ethnicity, sex, etc.), Birth measurements (weight, gestation, Apgars, etc.), Tests and procedures, Infant complications, Birth defect diagnostic information

Mother: Identification information (name, address, date-of-birth, etc.), Demographic information (race/ethnicity, sex, etc.), Gravidity/parity, Illnesses/conditions, Prenatal care, Prenatal diagnostic information, Pregnancy/delivery complications, Family history

Father: Identification information (name, address, date-of-birth, etc.), Demographic information (race/ethnicity, sex, etc.), Family history

Data Collection Methods and Storage

Data collection: Printed abstract/report filled out by staff, Electronic file/report filled out by staff at facility (laptop, web-based, etc.)

Database collection and storage: Access

Data Analysis

Data analysis software: SAS, Access, Excel, Tableau

Quality assurance: Validity checks, Re-abstraction of cases, Double-checking of assigned codes, Comparison/verification between multiple data sources, Clinical review, Timeliness, Data/hospital audits as needed

Data use and analysis: Routine statistical monitoring, Public health program evaluation, Baseline rates, Rates by demographic and other variables, Monitoring outbreaks and cluster investigations, Time trends, Observed vs. expected analyses, Epidemiological studies (using only program data), Identification of potential cases for other epidemiologic studies, Grant proposals, Education/public awareness

System Integration

System links: Link to other state registries/databases, Link case finding data to final birth file, Link case finding data to final fetal death file

System integration: Link birth defects data to MDPH Pregnancy to Early Life Longitudinal (PELL) data system.

Funding

Funding source: 60% General state funds, 40% MCH funds

Other

Web site: www.mass.gov/dph/birthdefects

Surveillance reports on file: Annual or bi-annual reports since 1999

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DATA TABLES

Massachusetts
Birth Defects Counts and Prevalence 2014 - 2018 (Prevalence per 10,000 Live Births)

Defect	Maternal Race/Ethnicity					Total*	Notes
	White, Non-Hispanic	Black, Non-Hispanic	Hispanic	Asian or Pacific Islander, Non-Hispanic	American Alaska Native, Non-Hispanic		
Anencephalus	49 <i>2.3</i>	15 <i>4.2</i>	21 <i>3.2</i>	11 <i>3.4</i>	0 <i>0.0</i>	107 <i>3.0</i>	
Anophthalmia/microphthalmia	19 <i>0.9</i>	4 <i>1.1</i>	10 <i>1.5</i>	2 <i>0.6</i>	0 <i>0.0</i>	39 <i>1.1</i>	
Anotia/microtia	46 <i>2.2</i>	7 <i>2.0</i>	28 <i>4.2</i>	9 <i>2.8</i>	0 <i>0.0</i>	93 <i>2.6</i>	
Aortic valve stenosis	44 <i>2.1</i>	2 <i>0.6</i>	6 <i>0.9</i>	1 <i>0.3</i>	0 <i>0.0</i>	55 <i>1.6</i>	
Atrial septal defect	545 <i>25.9</i>	108 <i>30.4</i>	215 <i>32.6</i>	71 <i>22.0</i>	5 <i>41.5</i>	973 <i>27.4</i>	
Atrioventricular septal defect (Endocardial cushion defect)	124 <i>5.9</i>	49 <i>13.8</i>	74 <i>11.2</i>	15 <i>4.6</i>	1 <i>8.3</i>	279 <i>7.9</i>	
Biliary atresia	11 <i>0.5</i>	2 <i>0.6</i>	4 <i>0.6</i>	3 <i>0.9</i>	0 <i>0.0</i>	20 <i>0.6</i>	
Bladder exstrophy	6 <i>0.3</i>	1 <i>0.3</i>	2 <i>0.3</i>	0 <i>0.0</i>	0 <i>0.0</i>	9 <i>0.3</i>	
Choanal atresia	12 <i>0.6</i>	2 <i>0.6</i>	2 <i>0.3</i>	3 <i>0.9</i>	1 <i>8.3</i>	21 <i>0.6</i>	
Cleft lip alone	68 <i>3.2</i>	9 <i>2.5</i>	14 <i>2.1</i>	17 <i>5.3</i>	1 <i>8.3</i>	113 <i>3.2</i>	
Cleft lip with cleft palate	92 <i>4.4</i>	12 <i>3.4</i>	36 <i>5.5</i>	16 <i>5.0</i>	1 <i>8.3</i>	159 <i>4.5</i>	
Cleft palate alone	132 <i>6.3</i>	14 <i>3.9</i>	37 <i>5.6</i>	20 <i>6.2</i>	3 <i>24.9</i>	218 <i>6.1</i>	
Cloacal exstrophy	7 <i>0.3</i>	0 <i>0.0</i>	4 <i>0.6</i>	1 <i>0.3</i>	0 <i>0.0</i>	12 <i>0.3</i>	
Clubfoot	377 <i>17.9</i>	48 <i>13.5</i>	118 <i>17.9</i>	25 <i>7.7</i>	3 <i>24.9</i>	590 <i>16.6</i>	1
Coarctation of the aorta	102 <i>4.8</i>	9 <i>2.5</i>	33 <i>5.0</i>	13 <i>4.0</i>	1 <i>8.3</i>	161 <i>4.5</i>	
Common truncus (truncus arteriosus)	15 <i>0.7</i>	4 <i>1.1</i>	2 <i>0.3</i>	2 <i>0.6</i>	0 <i>0.0</i>	24 <i>0.7</i>	
Congenital cataract	69 <i>3.3</i>	11 <i>3.1</i>	24 <i>3.6</i>	6 <i>1.9</i>	0 <i>0.0</i>	112 <i>3.2</i>	
Congenital posterior urethral valves	23 <i>2.1</i>	11 <i>6.1</i>	12 <i>3.6</i>	2 <i>1.2</i>	0 <i>0.0</i>	49 <i>2.7</i>	2
Craniosynostosis	146 <i>6.9</i>	6 <i>1.7</i>	29 <i>4.4</i>	9 <i>2.8</i>	1 <i>8.3</i>	198 <i>5.6</i>	
Deletion 22q11.2	34 <i>1.6</i>	6 <i>1.7</i>	16 <i>2.4</i>	4 <i>1.2</i>	0 <i>0.0</i>	60 <i>1.7</i>	
Diaphragmatic hernia	71 <i>3.4</i>	11 <i>3.1</i>	19 <i>2.9</i>	9 <i>2.8</i>	1 <i>8.3</i>	114 <i>3.2</i>	
Double outlet right ventricle	41 <i>1.9</i>	8 <i>2.3</i>	13 <i>2.0</i>	5 <i>1.5</i>	0 <i>0.0</i>	69 <i>1.9</i>	
Ebstein anomaly	19 <i>0.9</i>	0 <i>0.0</i>	3 <i>0.5</i>	0 <i>0.0</i>	0 <i>0.0</i>	24 <i>0.7</i>	
Encephalocele	16 <i>0.8</i>	8 <i>2.3</i>	7 <i>1.1</i>	2 <i>0.6</i>	0 <i>0.0</i>	34 <i>1.0</i>	
Esophageal atresia/tracheoesophageal fistula	68 <i>3.2</i>	3 <i>0.8</i>	13 <i>2.0</i>	1 <i>0.3</i>	1 <i>8.3</i>	86 <i>2.4</i>	
Gastroschisis	58 <i>2.8</i>	8 <i>2.3</i>	32 <i>4.9</i>	7 <i>2.2</i>	2 <i>16.6</i>	110 <i>3.1</i>	
Holoprosencephaly	38 <i>1.8</i>	6 <i>1.7</i>	19 <i>2.9</i>	8 <i>2.5</i>	0 <i>0.0</i>	76 <i>2.1</i>	
Hypoplastic left heart syndrome	60 <i>2.8</i>	14 <i>3.9</i>	20 <i>3.0</i>	3 <i>0.9</i>	0 <i>0.0</i>	103 <i>2.9</i>	
Hypospadias	890 <i>82.1</i>	102 <i>56.7</i>	195 <i>58.3</i>	71 <i>43.0</i>	5 <i>80.0</i>	1,294 <i>71.3</i>	2
Interrupted aortic arch	6 <i>0.3</i>	2 <i>0.6</i>	2 <i>0.3</i>	0 <i>0.0</i>	0 <i>0.0</i>	10 <i>0.3</i>	

Massachusetts
Birth Defects Counts and Prevalence 2014 - 2018 (Prevalence per 10,000 Live Births)

Defect	Maternal Race/Ethnicity					Total*	Notes
	White, Non-Hispanic	Black, Non-Hispanic	Hispanic	Asian or Pacific Islander, Non-Hispanic	American Indian or Alaska Native, Non-Hispanic		
Limb deficiencies (reduction defects)	105 <i>5.0</i>	16 <i>4.5</i>	35 <i>5.3</i>	7 <i>2.2</i>	1 <i>8.3</i>	167 <i>4.7</i>	
Omphalocele	101 <i>4.8</i>	8 <i>2.3</i>	27 <i>4.1</i>	21 <i>6.5</i>	0 <i>0.0</i>	168 <i>4.7</i>	
Pulmonary valve atresia and stenosis	197 <i>9.3</i>	53 <i>14.9</i>	63 <i>9.6</i>	21 <i>6.5</i>	1 <i>8.3</i>	345 <i>9.7</i>	3
Pulmonary valve atresia	30 <i>1.4</i>	5 <i>1.4</i>	5 <i>0.8</i>	5 <i>1.5</i>	0 <i>0.0</i>	46 <i>1.3</i>	3
Rectal and large intestinal atresia/stenosis	97 <i>4.6</i>	7 <i>2.0</i>	24 <i>3.6</i>	8 <i>2.5</i>	1 <i>8.3</i>	142 <i>4.0</i>	
Renal agenesis/hypoplasia	195 <i>9.3</i>	31 <i>8.7</i>	51 <i>7.7</i>	17 <i>5.3</i>	2 <i>16.6</i>	309 <i>8.7</i>	
Single ventricle	9 <i>0.4</i>	3 <i>0.8</i>	6 <i>0.9</i>	2 <i>0.6</i>	0 <i>0.0</i>	23 <i>0.6</i>	
Small intestinal atresia/stenosis	49 <i>2.3</i>	9 <i>2.5</i>	18 <i>2.7</i>	11 <i>3.4</i>	1 <i>8.3</i>	92 <i>2.6</i>	
Spina bifida without anencephalus	91 <i>4.3</i>	10 <i>2.8</i>	44 <i>6.7</i>	7 <i>2.2</i>	0 <i>0.0</i>	165 <i>4.7</i>	
Tetralogy of Fallot	104 <i>4.9</i>	22 <i>6.2</i>	37 <i>5.6</i>	18 <i>5.6</i>	0 <i>0.0</i>	196 <i>5.5</i>	
Total anomalous pulmonary venous connection	11 <i>0.5</i>	2 <i>0.6</i>	5 <i>0.8</i>	9 <i>2.8</i>	0 <i>0.0</i>	28 <i>0.8</i>	
Transposition of the great arteries (TGA)	63 <i>3.0</i>	11 <i>3.1</i>	15 <i>2.3</i>	9 <i>2.8</i>	0 <i>0.0</i>	102 <i>2.9</i>	
Dextro-transposition of great arteries (d-TGA)	54 <i>2.6</i>	10 <i>2.8</i>	9 <i>1.4</i>	8 <i>2.5</i>	0 <i>0.0</i>	84 <i>2.4</i>	
Tricuspid valve atresia and stenosis	28 <i>1.3</i>	4 <i>1.1</i>	3 <i>0.5</i>	0 <i>0.0</i>	0 <i>0.0</i>	35 <i>1.0</i>	
Tricuspid valve atresia	19 <i>0.9</i>	4 <i>1.1</i>	2 <i>0.3</i>	0 <i>0.0</i>	0 <i>0.0</i>	25 <i>0.7</i>	
Trisomy 13	84 <i>4.0</i>	11 <i>3.1</i>	18 <i>2.7</i>	12 <i>3.7</i>	0 <i>0.0</i>	145 <i>4.1</i>	
Trisomy 18	185 <i>8.8</i>	31 <i>8.7</i>	47 <i>7.1</i>	31 <i>9.6</i>	1 <i>8.3</i>	325 <i>9.2</i>	
Trisomy 21 (Down syndrome)	628 <i>29.8</i>	99 <i>27.9</i>	152 <i>23.0</i>	67 <i>20.7</i>	8 <i>66.4</i>	1,031 <i>29.1</i>	
Turner syndrome	156 <i>15.2</i>	24 <i>13.7</i>	33 <i>10.2</i>	25 <i>15.9</i>	1 <i>17.2</i>	254 <i>14.7</i>	4
Ventricular septal defect	607 <i>28.8</i>	76 <i>21.4</i>	220 <i>33.4</i>	82 <i>25.4</i>	4 <i>33.2</i>	1,015 <i>28.6</i>	
Total live births	210,779	35,508	65,961	32,295	1,205	354,475	5
Male live births	108,397	17,987	33,453	16,527	625	181,425	
Female live births	102,379	17,521	32,508	15,768	580	173,047	

Massachusetts
Birth Defects Counts and Prevalence 2014 - 2018 (Prevalence per 10,000 Live Births)

Defect	Maternal Age (Years)		Total*	Notes
	Less than 35	35+		
Gastroschisis	103 <i>3.9</i>	7 <i>0.8</i>	110 <i>3.1</i>	
Trisomy 13	60 <i>2.2</i>	85 <i>9.7</i>	145 <i>4.1</i>	
Trisomy 18	113 <i>4.2</i>	212 <i>24.2</i>	325 <i>9.2</i>	
Trisomy 21 (Down syndrome)	353 <i>13.2</i>	678 <i>77.5</i>	1,031 <i>29.1</i>	
Total live births	266,975	87,490	354,475	5

Notes

1. Data for this condition is limited to those who require casting or other treatment if the case is live birth.
2. Data for this condition include male and unknown gender cases only. Prevalence is calculated per 10,000 male live births.
3. Data for this condition include cases of pulmonary valve atresia with a ventricular septal defect that were reviewed and determined not to be a variant of tetralogy of Fallot.
4. Data for this condition include female and unknown gender cases only. Prevalence is calculated per 10,000 female live births.
5. Data for total live births include unknown gender.

General comments

*Data for totals include unknown and/or other.

-Data for conditions exclude possible/probable cases.