Expanding the Massachusetts Birth Defects Monitoring Program to Include Additional Pregnancy Outcomes: Programmatic Efforts and Impacts on Case Ascertainment 2012–2020

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The findings and conclusions in this report are those of the authors and do not necessarily represent the official position of the Centers for Disease Control and Prevention.

**Supplementary Tables**

* **Supplemental table S1.** Birth defects included in the Massachusetts Birth Defects Monitoring Program and year added to the program.
* **Supplemental table S2.** Counts and prevalence of birth defects per 10,000 live births ascertained by the Massachusetts Birth Defects Monitoring Program, overall and by maternal characteristics, 2012–2020.
* **Supplemental table S3.** Counts and prevalence of select birth defects per 10,000 live births ascertained by the Massachusetts Birth Defects Monitoring Program, overall and select birth defects types, 2012–2020.

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| **Supplemental Table S1.** Birth defects included in the Massachusetts Birth Defects Monitoring Program and year added to the programa |
| **Birth Defect** | **ICD-9-CM Codesb** | **ICD-10-CM** **Codesc** | **Modified ICD-9-CM/BPA** **Codesd** | **Comments** | **Codes changed in 2014 or later** |
| **Central Nervous System** |  |  |  |  |  |
| Anencephaly  | 740.0-740.1 | Q00.0-Q00.1 | 740.00, 740.10 |  |  |
| Encephalocele | 742.0 | Q01.0-Q01.9 | 742.00-742.09 |  |  |
| Holoprosencephaly | 742.2 | Q04.2 | 742.26 |  |  |
| Hydrocephaly without Spina Bifida | 742.3  | Q03.0, Q03.1, Q03.8, Q03.9 | 742.30-742.32, 742.38-742.39 |  |  |
| Microcephaly | 742.1 | Q02 | 742.10, 742.286 |  |  |
| Spina bifida with and without Hydrocephaly  | 741.0, 741.9 | Q05.0-Q05.9Q07.01,Q07.03 | 741.00-741.99 |  |  |
| Spinal Cord anomalies | 348.0, 745.51, 742.53, 742.59 | Q06.0-Q06.4, Q06.8  | 742.50, 742.51,742.52,742.53,742.54,742.58 | Includes arachnoid cyst |  |
| Other Central Nervous System | 742.2, 742.4, 742.8,742.9 | Q00.2, Q04.0-Q04.9, Q06.9, Q07.00, Q07.02, Q07.8, Q07.9, G90.1 | 740.20,740.21, 740.29,742.20, 742.21, 742.23-742.25, 742.27-742.29, 742.40-742.42, 742.480, 742.485, 742.88, 742.90 |  |  |
| **Eye** |  |  |  |  |  |
| Aniridia | 743.45 | Q13.1 | 743.420-743.424 |  |  |
| Anophthalmia/Micropthalmia | 743.0, 743.1 | Q11.0-Q11.2 | 743.00-743.10 |  |  |
| Congenital Glaucoma/ Congenital Cataract | 365.14, 743.20-743.22, 743.30 – 743.34 | Q12.0, Q15 | 743.20, 743.25, 743.26, 743.32, 743.35, 743.36  |  |  |
| Other Eye | 743.35-39, 743.41-44, 743.46-743.49, 743.51-743.59, 743.66, 743.8  | Q10.7, Q12.1-Q12.9, Q13.0, Q13.2, Q13.3-Q13.5, Q13.81, Q13.89, Q13.9, Q14.0-Q14.9, Q15.0, Q15.8  | 743.300-743.314, 743.340-743.344, 743.410, 743.430, 743.440, 743.460-743.474, 743.480-743.530, 743.535, 743.580, 743.590,743.610, 743.620, 743.636, 743.650, 743.800 |  |  |
| **Ear** |  |  |  |  |  |
| Anotia/Microtia | 744.01, 744.23 | Q16.0, Q16.1, Q17.2 | 744.01, 744.21 |  |  |
| Other Ear | 744.00, 744.02-744.09, 744.24, 744.29, 744.3 | Q16.2-Q16.9, Q17.3-Q17.9 | 744.00,744.02-744.10, 744.23-744.25, 744.280,744.300 |  |  |
| **Cardiovascular** |  |  |  |  |  |
| Aortic Arch Atresia | 747.22 | Q25.21, Q25.29, Q25.3, Q25.41, Q25.42, Q25.9 | 747.200 |  |  |
| Aortic Valve Stenosis | 746.3 | Q23.0, Q23.8, Q23.9 | 746.30 |  |  |
| Atrioventricular Septal Defect | 745.60, 745.61,745.66, 745.69 | Q21.2, Q21.0 | 745.60,745.61, 745.62, 745.63, 745.68, 745.69, 745.685 | Includes atrial septal defect primum, common atrium, complete atrioventricular canal, endocardial cushion defect, ventricular septal defect, canal type  |  |
| Atrial Septal Defect, Secundum, and Other | 745.5 | Q21.1, Q21.8, Q21.9 | 745.51, 745.58,745.59 |  |  |
| Coarctation of Aortad | 747.10 | Q25.1 | 747.10-747.19 |  |  |
| Dextro-Transposition of the Great Arteries d  | 745.10 | Q20.3, Q20.5, Q20.8 | 745.10, 745.11 | Excludes 745.19  |  |
| Double Outlet Right Ventricled | 745.11 | Q20.1, Q20.3, Q20.5, Q20.8 | 745.185, 745.186, 745.188, 745.189 |  |  |
| Ebstein Anomalyd | 746.2 | Q22.5 | 746.20 |  |  |
| Hypoplastic Left Heart Syndromed  | 746.7 | Q23.4 | 746.70 |  |  |
| Interrupted Aortic Archd | 747.11 | Q25.21, Q25.29, Q25.49 | 747.215-747.217 |  |  |
| Levo-Transposition of the Great Arteries | 745.12 | Q20.5, Q20.8 | 745.12  |  |  |
| Partial anomalous pulmonary venous connection | 747.42 | Q26.3, Q26.4 | 747.43 |  |  |
| Pulmonary Valve Atresiad | 746.00, 746.01 | Q22.0, Q22.3 | 746.00, 746.03 | With or without ventricular septal defect  |  |
| Pulmonary Stenosis, Valvular | 746.02 | Q22.1 | 746.01 |  |  |
| Single Ventricle | 745.3 | Q20.4, Q20.8 | 745.30-745.33, 745.38 |  |  |
| Tetralogy of Fallot | 745.2 | Q21.3, Q21.8 | 745.20, 747.31 |  |  |
| Total anomalous pulmonary venous connection | 747.41 | Q26.2, Q26.4 | 747.42 |  |  |
| Tricuspid Valve Atresiad | 746.1 | Q22.4, Q22.6, Q22.8, Q22.9 | 746.10 | Excludes tricuspid valve stenosis (746.106) |  |
| Truncus Arteriosus (Common Truncus) | 745.0 | Q20.0, Q21.4 | 745.00 (excluding 745.01) |  |  |
| Ventricular Septal Defect, Muscular, Membranous or Other | 745.4 | Q21.0, Q21.8, Q21.9 | 745.485, 745.486,745.49 |  | Prior to 2014, excluded Muscular VSD |
| Ventricular Septal Defect, Conoventricular or Malalignment | 745.4 | Q21.0, Q21.8, Q21.9 | 745.487 |  |  |
| Other Cardiovascular | 745.8, 746.09, 746.4, 746.5, 746.8, 746.9, 747.2, 747.32, 747.40, 747.49, 747.6, 747.8 | Q20.8, Q20.9, Q21.8, Q21.9, Q22.2, Q22.3, Q23.2, Q23.8, Q23.9, Q24.0- Q24.3, Q24.5, Q24.8, Q24.9, Q25.40, Q25.43-Q25.49, Q25.9, Q25.6, Q25.79, Q25.9, Q26.0, Q26.1, Q26.6, Q26.8, Q26.9, Q27.1-Q27.9, Q28.2-Q28.9 | 745.010, 746.080, 746.090, 746.106,746.400-746.505, 746.600, 746.800, 746.820, 746.830, 746.850, 746.880-746.882, 746.885, 746.900, 746.995, 747.210, 747.220, 747.230, 747.250, 747.270, 747.280, 747.300, 747.320, 747.380, 747.410, 747.480, 747.490, 747.620, 747.640, 747.650, 747.680, 747.800, 747.810, 747.880 |  |  |
| **Respiratory** |  |  |  |  |  |
| Choanal Atresia | 748.0 | Q30.0 | 748.01 |  |  |
| Lung Anomalies | 748.4, 748.5 | Q33.0, Q33.2, Q33.3, Q33.6, Q33.8 | 748.40, 748.41, 748.48, 748.50, 748.51, 748.52, 748.58 |  |  |
| Other Respiratory | 748.3,748.6, 748.8 | Q31.1-Q31.9, Q32.0-Q32.9, Q33.1, Q33.4, Q33.5, Q33.8, Q33.9, Q34.0-Q34.8 | 748.000, 748.100, 748.185, 748.205, 748.209, 748.310, 748.330-748.350, 748.380, 748.385, 748.390, 748.625, 748.690, 748.88 | Excludes laryngo-tracheomalacia |  |
| **Orofacial** |  |  |  |  |  |
| Cleft Palate without Cleft Lip | 749.0 | Q35.1-Q35.9 | 749.00 – 749.07, 749.09 | Excludes 749.08 (cleft uvula) | Prior to 2014, excludes isolated submucous cleft palate |
| Cleft lip with/without Cleft Palate | 749.1, 749.2 | Q36.0-Q36.9Q37.0-Q37.9 | 749.10-749.19, 749.20-749.29 | Excludes 749.191 (fused lip) |  |
| Pierre Robin Sequence | 756.0 | Q87.0, QQ87.08 | 524.08 |  |  |
| Other Orofacial | 744.4,744.8 | Q18.0-Q18.2 | 744.400,744.480,744.880, 748.120, 748.180,750.140 | Includes tongue fissure |  |
| **Gastrointestinal** |  |  |  |  |  |
| Biliary Atresia | 751.61 | Q44.2, Q44.3 | 751.65 |  |  |
| Esophageal Atresia/Tracheoesophageal Fistula | 750.3 | Q39.0-Q39.4, Q39.8 | 750.30-750.35 |  |  |
| Hirschsprung Disease | 751.3 | Q43.1, Q43.2 | 751.30-751.34 |  |  |
| Rectal and Large Intestinal Atresia/Stenosis | 751.2 | Q42.0-Q42.9 | 751.20-751.24 |  |  |
| Small Intestinal Atresia | 751.1 | Q41.0-Q41.9 | 751.10-751.19 |  |  |
| Other Gastrointestinal | 750.4,750.6,750.7,750.8, 751.0, 751.4,751.5, 751.62, 751.69, 751.7, 751.9 | Q39.5, Q39.6, Q39.8, Q39.9, Q40.1, Q40.2, Q40.8, Q43.0, Q43.3, Q43.4-Q43.9, Q44.0-Q44.7, Q45.0-Q45.3, | 750.380, 750.430, 750.480, 750.60, 750.70, 750.80, 751.00, 751.010, 751.400-751.420, 751.490, 751.495, 751.50,751.52, 751.53, 751.54, 751.56, 751.58, 751.61-751.64, 751.66, 751.67, 751.70, 751.72, 751.74, 751.80 |  |  |
| **Genitourinary** |  |  |  |  |  |
| Bladder Exstrophy | 753.5 | Q64.10-Q64.12,Q64.19 | 753.50 |  |  |
| Cloacal Exstrophy | 751.5 | Q64.12 | 751.55 |  |  |
| Hypospadiasf | 752.61 | Q54.0-Q54.9 (excluding Q54.4) | 752.60, 752.62 | Excludes 752.61 epispadias.  | Prior to 2014, excludes 752.600, 752.605, 752.620, 752.625 |
| Posterior Urethral Valvef | 753.6 | Q64.2 | 753.60 |  |  |
| Other Obstructive Genitourinary Defect | 753.2, 753.6 | Q62.0, Q62.10, Q62.11, Q62.12, Q62.2, Q62.31, Q62.32, Q62.39 | 753.20-753.22,753.29, 753.61-753.69 |  | For deliveries on or after 7/1/15, surgery not required |
| Renal Agenesis/Hypoplasia  | 753.0 | Q60.0-Q60.6 | 753.00-753.01 |  | Prior to 2014 excludes unilateral renal agenesis |
| Other Genitourinary | 752.0, 752.1, 752.2, 752.3, 752.4, 752.7, 753.0-753.8 | Q50.01, Q50.02, Q50.1, Q50.2, Q50.31, Q50.32, Q50.39, Q50.4-Q50.6, Q51.0, Q51.10, Q51.11, Q51.20-Q51.22, Q51.28, Q51.3, Q51.4, Q51.6, Q51.810, Q51.811, Q51.818, Q51.820, Q51.828, Q51.9, Q52.0, Q52.10- Q52.4, Q52.70, Q52.79, Q52.8, Q52.9, Q56.0-Q56.4 | 752.00, 752.08, 752.085, 752.10, 752.20, 752.30, 752.32, 752.38, 752.40-752.44, 752.48, 752.70, 752.79-752.82, 752.85, 752.860, 752.865, 752.880, 752.901, 753.10-753.12, 753.13 753.16, 753.18, 753.31-753.34,753.38,753.40, 753.410, 753.420,753.480,753.485, 753.70, 753.710, 753.790-753.820, 753.84, 753.88 |  |  |
| **Musculoskeletal** |  |  |  |  |  |
| Club Foot | 754.51, 754.70 | Q66.0, Q66.89, Q66.90, Q66.91 | 754.50, 754.51, 754.52, 754.53, 754.59, 754.60, 754.68, 754.69, 754.73 (excluding 754.735) | Requires casting or surgery for live births, other postnatal confirmation (e.g., autopsy) for nonlive births |  |
| Craniosynostosis | 756.0 | Q75.0 | 756.00-756.02 |  |  |
| Diaphragmatic Hernia | 756.6 | Q79.0, Q79.1 | 756.600-756.605, 756.610-756.617, 756.618-756.619 |  |  |
| Gastroschisis | 756.73  | Q79.3 | 756.71 |  |  |
| Omphalocele | 756.72 | Q79.2 | 756.70 |  |  |
| Polydactyly/syndactyly | 755.0, 755.1 | Q69.0-Q69.9, Q70.0-Q70.9 | 755.005, 755.01-755.03, 755.095-755.096, 755.10-755.13, 755.19 | Hands require bone or cartilage involvement. Excludes webbing of toes 2-3 |  |
| Limb Reduction Defects | 755.2, 755.3, 755.4 | Q71.0-Q71.9, Q72.0-Q72.9, Q73.0-Q73.9 | 755.20-755.29, 755.30-755.39, 755.40-755.49 |  |  |
| Skeletal Dysplasia | 755.55, 756.4, 756.5 | Q87.0, Q77.0-Q77,9, Q78.0-Q78.9 | 755.555, 756.400, 756.41, 756.43, 756.447, 756.46, 756.480, 756.49, 756.50, 756.53, 756.54, 756.575, 756.58, 756.59 |  |  |
| Other Musculoskeletal | 754.52, 752.53, 754.59, 755.50-755.54, 755.56, 755.58, 756.11-756.17, 756.19,756.3,756.8 | Q66.211-Q66.9, Q67.5, Q74.0, Q71.60-Q71.63, Q76.1- Q76.3, Q76.411-Q76.429, Q76.49, Q76.6-Q76.9, Q79.0, Q79.1, Q79.4, Q79.51, Q79.59 | 754.00,754.20, 754.21, 754.22, 754.400, 754.410, 754.430, 754.440, 754.780, 754.820, 754.840, 754.880, 755.44-755.50, 755.530, 755.536,755.54, 755.58, 755.585, 755.640, 755.650, 755.680, 755.685, 755.800, 756.080, 756.110, 756.120, 756.140, 756.145, 756.146, 756.150, 756.155, 756.156, 756.160, 756.165, 756.166, 756.170, 756.175, 756.180, 756.185, 756.190, 756.300, 756.310-756.350, 756.380, 756.620, 756.680, 756.690, 756.720, 756.790, 756.795, 756.80, 756.81, 756.84, 756.88 |  |  |
| **Chromosomal** |  |  |  |  |  |
| Deletion 22 q11.2 | 758.32 | Q93.81, D82.1 | 279.110, 758.37 |  |  |
| Klinefelter Syndrome | 758.7 | Q98.0, Q98.1, Q98.4 | 758.70-758.71, 758.79 |  |  |
| Trisomy 13  | 758.1 | Q91.4-Q91.7 | 758.10-758.19 |  |  |
| Trisomy 18 | 758.2 | Q91.0-Q91.3 | 758.20-758.29 |  |  |
| Trisomy 21 (Down syndrome) | 758.0 | Q90.0-Q90.9 | 758.00-758.09 |  |  |
| Turner Syndromeg | 758.6 | Q96.0-Q96.9 | 758.60-758.69 |  |  |
| Other Chromosomal | 756.83, 758.31-758.6 (except 758.32), 758.81-758.9 | Q79.60-Q79.69, Q93.0-Q93.9 (except Q93.81), Q95.0, Q95.1-Q95.9, Q96.0-Q96.9, Q97.0-Q97.9, Q98.5-Q98,9, Q99.0, Q99.8, Q99.9 | 352.600, 756.040, 756.045, 756.046, 756.050, 756.055-756.057, 756.060, 756.065, 756.525, 756.550-756.570, 756.830, 756.850, 757.300, 758.300-758.400 (except 758.32, 758.37),758.50-758.54, 758.580, 758.585, 758.586, 758.590, 758.80-758.86, 758.88, 758.89, 758.90 - 759.93, 758.990, 758.999, 759.340, 759.400-759.490, 759.500, 759.610, 759.620, 759.800-759.890 |  |  |
| **Other** |  |  |  |  |  |
| Amniotic Bands | No specific code |  | 658.80 |  |  |
| Heterotaxy/Situs Inversus | 759.3 | Q89.3, Q20.6 | 759.30-759.33, 759.35-759.395 |  |  |
| Skin Anomalies | 757.1, 757.31, 757.39 | Q80.0-Q80.9, Q81.0-Q81.9 Q82.1, Q82.4 | 757.34, 757.36, 757.48, 757.80, 757.35, 757.33, 757.11, 757.19, 757.195-757.197 |  |  |
| Other | 759.0-759.2, others | Q89.1, Q89.2, Q89.7, others | 255.20, 759.00, 759.01, 759.04, 759.05, 759.08, 759.11, 759.18, 759.21, 759.22, 759.24, 759.70, 759.90 |  |  |
| aAdapted with permission from Massachusetts Birth Defects 2013-2014 and Massachusetts Birth Defects 2015-2018.bInternational Classification of Diseases, 9th edition, Clinical Modification.cInternational Classification of Diseases, 10th edition, Clinical Modification.dCritical Congenital Heart Defect (CCHD)eInternational Classification of Diseases, 9th edition, Clinical Modification/modified British Pediatric Association (ICD-9-CM/BPA) fDefect reported among males onlygDefect reported among females only |

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| **Supplemental Table S2**. Counts and prevalence of birth defects per 10,000 live births ascertained by the Massachusetts Birth Defects Monitoring Program, overall and by maternal characteristics 2012–2020.  |
|  |  | 2012–2014(Births: 216,119) | 2015–2017(Births: 213,509) | 2018–2020(Births: 204,657) | 2012–2020(Births: 634,285) |
|  | Pregnancy outcomes includeda | Count | Prevalence (95% CI) | Count | Prevalence (95% CI) | Count | Prevalence (95% CI) | Count | Prevalence (95% CI) |
| **Overall** | LB, SB, OPL | 5569 | 257.7 (251.0, 264.5) | 7533 | 352.8 (345.0, 360.7) | 8075 | 394.6 (386.2, 403.1) | 21177 | 333.9 (329.4, 338.4) |
|  | LB, SB | 4396 | 203.4 (197.5, 209.5) | 5920 | 277.3 (270.4, 284.3) | 6013 | 293.8 (286.5, 301.2) | 16329 | 257.4 (253.5, 261.4) |
|  |  |  |  |  |  |  |  |  |  |
| **Maternal Characteristics** |  |  |  |  |  |  |  |  |  |
| **Age**, yearsb |  |  |  |  |  |  |  |  |  |
|  <20 | LB, SB, OPL | 217 | 257.1 (224.0, 293.7) | 190 | 319.5 (275.7, 368.3) | 158 | 344.6 (293.0, 402.7) | 565 | 297.8 (274.1, 323.0) |
|  | LB, SB | 197 | 233.4 (202.0, 268.4) | 169 | 284.2 (243.0, 330.4) | 143 | 311.9 (262.9, 367.4) | 509 | 268.3 (245.8, 292.3) |
|  20 to 24 | LB, SB, OPL | 666 | 218.9 (202.7, 236.0) | 787 | 306.9 (286.1, 328.7) | 719 | 333.4 (309.8, 358.2) | 2172 | 279.8 (268.3, 291.6) |
|  | LB, SB | 597 | 196.2 (180.9, 212.4) | 712 | 277.7 (257.9, 298.5) | 630 | 292.1 (270.1, 315.5) | 1939 | 249.8 (238.9, 261.0) |
|  25 to 29 | LB, SB, OPL | 1219 | 228.3 (215.8, 241.3) | 1629 | 311.6 (296.9, 326.9) | 1530 | 331.4 (315.2, 348.1) | 4378 | 288.3 (280.0, 296.9) |
|  | LB, SB | 1016 | 190.3 (178.9, 202.2) | 1395 | 266.9 (253.2, 281.1) | 1268 | 274.6 (259.9, 289.9) | 3679 | 242.3 (234.6, 250.2) |
|  30 to 34 | LB, SB, OPL | 1790 | 241.5 (230.6, 252.8) | 2505 | 324.9 (312.5, 337.6) | 2792 | 363.6 (350.5, 377.1) | 7087 | 310.8 (303.7, 318.0) |
|  | LB, SB | 1466 | 197.8 (187.9, 208.1) | 2003 | 259.8 (248.7, 271.2) | 2130 | 277.4 (265.9, 289.2) | 5599 | 245.6 (239.2, 252.0) |
|  ≥35 | LB, SB, OPL | 1677 | 337.3 (321.6, 353.5) | 2416 | 459.9 (442.2, 478.2) | 2873 | 517.3 (499.0, 536.0) | 6966 | 441.4 (431.4, 451.7) |
|  | LB, SB | 1120 | 225.2 (212.4, 238.7) | 1635 | 311.2 (296.6, 326.4) | 1840 | 331.3 (316.6, 346.5) | 4595 | 291.2 (283.0, 299.6) |
| **Race/Ethnicity** |  |  |  |  |  |  |  |  |  |
|  NH White | LB, SB, OPL | 3476 | 259.9 (251.4, 268.6) | 4573 | 361.0 (350.8, 371.4) | 4803 | 411.4 (400.0, 422.9) | 12852 | 340.7 (335.0, 346.6) |
|  | LB, SB | 2736 | 204.6 (197.1, 212.3) | 3567 | 281.5 (272.5, 290.8) | 3438 | 294.4 (284.8, 304.3) | 9741 | 258.3 (253.2, 263.4) |
|  NH Black | LB, SB, OPL | 548 | 261.1 (240.0, 283.6) | 678 | 318.4 (295.2, 342.9) | 794 | 375.0 (349.8, 401.5) | 2020 | 318.4 (304.8, 332.3) |
|  | LB, SB | 488 | 232.6 (212.6, 253.9) | 566 | 265.8 (244.6, 288.3) | 665 | 314.1 (291.0, 338.5) | 1719 | 270.9 (258.4, 283.8) |
|  NH Asian/PI | LB, SB, OPL | 400 | 208.5 (188.7, 229.7) | 626 | 321.7 (297.4, 347.5) | 655 | 339.7 (314.6, 366.3) | 1681 | 290.2 (276.7, 304.2) |
|  | LB, SB | 308 | 160.5 (143.2, 179.3) | 441 | 226.6 (206.2, 248.5) | 459 | 238.1 (217.0, 260.6) | 1208 | 208.6 (197.1, 220.5) |
|  NH American Indian | LB, SB, OPL | 12 | 164.2 (84.8, 286.8) | 31 | 416.7 (283.1, 591.4) | 29 | 363.9 (243.7, 522.6) | 72 | 316.9 (248.0, 399.1) |
|  | LB, SB | 12 | 164.2 (84.8, 286.8) | 20 | 268.8 (164.2, 415.2) | 26 | 326.2 (213.1, 478.0) | 58 | 255.3 (193.9, 330.0) |
|  Other NH | LB, SB, OPL | 41 | 201.8 (144.8, 273.7) | 96 | 472.0 (382.3, 576.4) | 116 | 498.9 (412.3, 598.4) | 253 | 395.9 (348.6, 447.8) |
|  | LB, SB | 39 | 191.9 (136.5, 262.4) | 62 | 304.8 (233.7, 390.8) | 78 | 335.5 (265.2, 418.7) | 179 | 280.1 (240.6, 324.3) |
|  Hispanic | LB, SB, OPL | 907 | 238.2 (223.1, 254.0) | 1311 | 331.5 (314.1, 349.7) | 1481 | 354.5 (337.0, 372.7) | 3699 | 309.8 (300.1, 319.8) |
|  | LB, SB | 797 | 209.3 (195.2, 224.2) | 1155 | 292.1 (275.7, 309.2) | 1273 | 304.8 (288.5, 321.7) | 3225 | 270.1 (261.0, 279.5) |
|  Unknown/Missing | LB, SB, OPL | 197 | -- | 249 | -- | 226 | -- | 672 | -- |
|  | LB, SB | 28 | -- | 129 | -- | 100 | -- | 257 | -- |
| aLive Births (LB), Stillbirths (SB) and other pregnancy losses (OPLs; miscarriage (<20 weeks gestational age (GA)) or elective terminations (any GA)bN = 9 individuals are missing maternal age. Maternal age refers to biological mother, so in cases where donor eggs or embryos are used, we set maternal age to missingAbbreviations: Gestational age, GA; Live births, LB; Non-Hispanic, NH; Other pregnancy losses, OPLs; Stillbirths, SB |

|  |
| --- |
| **Supplemental Table S3**. Counts and prevalence of select birth defects per 10,000 live births ascertained by the Massachusetts Birth Defects Monitoring Program, overall and by select birth defects types, 2012–2020.  |
|  |  | 2012–2014(Births: 216,119) | 2015–2017(Births: 213,509) | 2018–2020(Births: 204,657) | 2012–2020(Births: 634,285) |
|  | Pregnancy outcomes includedb | Count | Prevalence (95% CI) | Count | Prevalence (95% CI) | Count | Prevalence (95% CI) | Count | Prevalence (95% CI) |
| **Overall** a | LB, SB, OPL | 5569 | 257.7 (251.0, 264.5) | 7533 | 352.8 (345.0, 360.7) | 8075 | 394.6 (386.2, 403.1) | 21177 | 333.9 (329.4, 338.4) |
|  | LB, SB | 4396 | 203.4 (197.5, 209.5) | 5920 | 277.3 (270.4, 284.3) | 6013 | 293.8 (286.5, 301.2) | 16329 | 257.4 (253.5, 261.4) |
|  |  |  |  |  |  |  |  |  |  |
| **Birth Defects Typesc** |  |  |  |  |  |  |  |  |  |
| **Cardiovascular** | LB, SB, OPL | 1496 | 69.2 (65.8, 72.8) | 1774 | 83.1 (79.3, 87.0) | 1809 | 88.4 (84.9, 93.0) | 5079 | 80.1 (78.2, 82.7) |
|  | LB, SB | 1373 | 63.5 (60.2, 67.0) | 1622 | 76.0 (72.8, 80.2) | 1656 | 80.9 (77.6, 85.4) | 4651 | 73.3 (71.2, 75.9) |
| Critical Congenital Heart Defectsd | LB, SB, OPL | 424 | 19.6 (17.8, 21.6) | 408 | 19.1 (17.3, 21.1) | 411 | 20.1 (18.2, 22.1) | 1243 | 19.6 (18.5, 20.7) |
|  | LB, SB | 368 | 17.0 (15.3, 18.9) | 336 | 15.7 (14.1, 17.5) | 333 | 16.3 (14.6, 18.1) | 1037 | 16.3 (15.4, 17.4) |
|  Hypoplastic Left Heart Syndrome | LB, SB, OPL | 53 | 2.5 (1.8, 3.2) | 61 | 2.9 (2.2, 3.7) | 50 | 2.4 (1.8, 3.2) | 164 | 2.6 (2.2, 3.0) |
|  | LB, SB | 35 | 1.6 (1.1, 2.3) | 45 | 2.1 (1.5, 2.8) | 28 | 1.4 (0.9, 2.0) | 108 | 1.7 (1.4, 2.1) |
|  Dextro-Transposition of the Great Arteries | LB, SB, OPL | 63 | 2.9 (2.2, 3.7) | 45 | 2.1 (1.5, 2.8) | 50 | 2.4 (1.8, 3.2) | 158 | 2.5 (2.1, 2.9) |
|  | LB, SB | 57 | 2.6 (2.0, 3.4) | 43 | 2.0 (1.5, 2.7) | 46 | 2.3 (1.7, 3.0) | 146 | 2.3 (1.9, 2.7) |
|  Single Ventricle | LB, SB, OPL | 8 | 0.4 (0.2, 0.7) | 19 | 0.9 (0.5, 1.4) | 12 | 0.6 (0.3, 1.0) | 39 | 0.6 (0.4, 0.8) |
|  | LB, SB | 6 | 0.3 (0.1, 0.6) | 9 | 0.4 (0.2, 0.8) | 9 | 0.4 (0.2, 0.8) | 24 | 0.4 (0.2, 0.6) |
|  Tetralogy of Fallot  | LB, SB, OPL | 113 | 5.2 (4.3, 6.3) | 106 | 5.0 (4.1, 6.0) | 114 | 5.6 (4.6, 6.7) | 333 | 5.3 (4.7, 5.9) |
|  | LB, SB | 98 | 4.5 (3.7, 5.5) | 88 | 4.1 (3.3, 5.1) | 89 | 4.4 (3.5, 5.4) | 275 | 4.3 (3.8, 4.9) |
|  |  |  |  |  |  |  |  |  |  |
| **Central Nervous System** | LB, SB, OPL | 613 | 28.4 (26.2, 30.7) | 753 | 35.3 (32.8, 37.9) | 752 | 36.7 (34.2, 39.5) | 2118 | 33.4 (32.0, 34.8) |
|  | LB, SB | 430 | 19.9 (18.1, 21.9) | 519 | 24.3 (22.3, 26.5) | 486 | 23.8 (21.7, 26.0) | 1435 | 22.6 (21.5, 23.8) |
| Neural Tube Defectse | LB, SB, OPL | 165 | 7.6 (6.5, 8.9) | 169 | 7.9 (6.8, 9.2) | 193 | 9.4 (8.1, 10.9) | 527 | 8.3 (7.6, 9.0) |
|  | LB, SB | 59 | 2.7 (2.1, 3.5) | 67 | 3.1 (2.4, 4.0) | 74 | 3.6 (2.8, 4.5) | 200 | 3.2 (2.7, 3.6) |
|  Anencephaly | LB, SB, OPL | 48 | 2.2 (1.6, 2.9) | 60 | 2.8 (2.1, 3.6) | 74 | 3.6 (2.8, 4.5) | 182 | 2.9 (2.5, 3.3) |
|  | LB, SB | 2 | 0.1 (0.0, 0.3) | 14 | 0.7 (0.4, 1.1) | 9 | 0.4 (0.2, 0.8) | 25 | 0.4 (0.3, 0.6) |
|  Spina Bifida | LB, SB, OPL | 89 | 4.1 (3.3, 5.1) | 100 | 4.7 (3.8, 5.7) | 87 | 4.3 (3.4, 5.2) | 276 | 4.4 (3.9, 4.9) |
|  | LB, SB | 45 | 2.1 (1.5, 2.8) | 52 | 2.4 (1.8, 3.2) | 49 | 2.4 (1.8, 3.2) | 146 | 2.3 (1.9, 2.7) |
|  Encephalocele | LB, SB, OPL | 34 | 1.6 (1.1, 2.2) | 11 | 0.5 (0.3, 0.9) | 35 | 1.7 (1.2, 2.4) | 80 | 1.3 (1.0, 1.6) |
|  | LB, SB | 14 | 0.7 (0.4, 1.1) | 2 | 0.1 (0,0.3) | 16 | 0.8 (0.5, 1.3) | 32 | 0.5 (0.4, 0.7) |
|  |  |  |  |  |  |  |  |  |  |
| **Chromosomal** | LB, SB, OPL | 1538 | 71.2 (67.7, 74.8) | 1996 | 93.5 (89.5, 97.7) | 2519 | 123.1 (118.4, 128.0) | 6053 | 95.4 (93.1, 97.9) |
|  | LB, SB | 666 | 30.8 (28.5, 33.2) | 763 | 35.7 (33.3, 38.4) | 866 | 42.3 (39.6, 45.2) | 2295 | 36.2 (34.7, 37.7) |
| Trisomy 13 | LB, SB, OPL | 68 | 3.2 (2.4, 4.0) | 85 | 4.0 (3.2, 4.9) | 107 | 5.2 (4.3, 6.3) | 260 | 4.1 (3.6, 4.6) |
|  | LB, SB | 6 | 0.3 (0.1, 0.6) | 15 | 0.7 (0.4, 1.2) | 11 | 0.5 (0.3, 1.0) | 32 | 0.5 (0.3, 0.7) |
| Trisomy 18 | LB, SB, OPL | 133 | 6.2 (5.2, 7.3) | 186 | 8.7 (7.5, 10.1) | 201 | 9.8 (8.5, 11.3) | 520 | 8.2 (7.5, 8.9) |
|  | LB, SB | 22 | 1.0 (0.6, 1.5) | 33 | 1.6 (1.1, 2.2) | 42 | 2.1 (1.5, 2.8) | 97 | 1.5 (1.2, 1.9) |
| Trisomy 21  | LB, SB, OPL | 523 | 24.2 (22.2, 26.4) | 613 | 28.7 (26.5, 31.1) | 695 | 34.0 (31.5, 36.6) | 1831 | 28.9 (27.6, 30.2) |
|  | LB, SB | 254 | 11.8 (10.4, 13.3) | 264 | 12.4 (10.9, 14.0) | 261 | 12.8 (11.3, 14.4) | 779 | 12.3 (11.4, 13.2) |
| **Orofacial** | LB, SB, OPL | 410 | 19.0 (17.2, 20.9) | 386 | 18.1 (16.3, 20.0) | 377 | 18.4 (16.6, 20.4) | 1173 | 18.5 (17.5, 19.6) |
|  | LB, SB | 371 | 17.2 (15.5, 19.0) | 351 | 16.4 (14.8, 18.3) | 333 | 16.3 (14.6, 18.1) | 1055 | 16.6 (15.6, 17.7) |
| Cleft Lip w/ and w/o Cleft Palate | LB, SB, OPL | 191 | 8.8 (7.6, 10.2) | 154 | 7.2 (6.1, 8.5) | 155 | 7.6 (6.4, 8.9) | 500 | 7.9 (7.2, 8.6) |
|  | LB, SB | 164 | 7.6 (6.5, 8.8) | 130 | 6.1 (5.1, 7.2) | 127 | 6.2 (5.2, 7.4) | 421 | 6.6 (6.0, 7.3) |
| Cleft Palate w/o Cleft Lip | LB, SB, OPL | 130 | 6.0 (5.0, 7.1) | 131 | 6.1 (5.1, 7.3) | 116 | 5.7 (4.7, 6.8) | 377 | 5.9 (5.4, 6.6) |
|  | LB, SB | 125 | 5.8 (4.8, 6.9) | 125 | 5.9 (4.9, 7.0) | 110 | 5.4 (4.4, 6.5) | 360 | 5.7 (5.1, 6.3) |
| **Ear** | LB, SB, OPL | 159 | 7.4 (6.3, 8.6) | 131 | 6.1 (5.1, 7.3) | 111 | 5.4 (4.5, 6.5) | 401 | 6.3 (5.7, 7.0) |
|  | LB, SB | 157 | 7.3 (6.2, 8.5) | 127 | 6.0 (5.0, 7.1) | 107 | 5.2 (4.3, 6.3) | 391 | 6.2 (5.6, 6.8) |
| **Eye** | LB, SB, OPL | 185 | 8.6 (7.4, 9.9) | 172 | 8.1 (6.9, 9.4) | 167 | 8.2 (7.0, 9.5) | 524 | 8.3 (7.6, 9.0) |
|  | LB, SB | 181 | 8.4 (7.2, 9.7) | 166 | 7.8 (6.6, 9.1) | 153 | 7.5 (6.3, 8.8) | 500 | 7.9 (7.2, 8.6) |
| **Gastrointestinal** | LB, SB, OPL | 376 | 17.4 (15.7, 19.3) | 384 | 18.0 (16.2, 19.9) | 349 | 17.1 (15.3, 18.9) | 1109 | 17.5 (16.5, 18.5) |
|  | LB, SB | 360 | 16.7 (15.0, 18.5) | 374 | 17.5 (15.8, 19.4) | 337 | 16.5 (14.8, 18.3) | 1071 | 16.9 (15.9, 17.9) |
| Esophageal Atresia/Tracheoesophageal Fistula | LB, SB, OPL | 62 | 2.9 (2.2, 3.7) | 47 | 2.2 (1.6, 2.9) | 55 | 2.7 (2.0, 3.5) | 164 | 2.6 (2.2, 3.0) |
|  | LB, SB | 61 | 2.8 (2.2, 3.6) | 47 | 2.2 (1.6, 2.9) | 54 | 2.6 (2.0, 3.4) | 162 | 2.6 (2.2, 3.0) |
| **Genitourinary** | LB, SB, OPL | 1104 | 51.1 (48.1, 54.2) | 2279 | 106.7 (102.4, 111.2) | 2361 | 115.4 (110.8, 120.1) | 5744 | 90.6 (88.2, 92.9) |
|  | LB, SB | 1052 | 48.7 (45.8, 51.7) | 2187 | 102.4 (98.2, 106.8) | 2259 | 110.4 (105.9, 115.0) | 5498 | 86.7 (84.4, 89.0) |
| Bilateral Renal Agenesis | LB, SB, OPL | 14 | 0.7 (0.4, 1.1) | 13 | 0.6 (0.3, 1.0) | 23 | 1.1 (0.7, 1.7) | 50 | 0.8 (0.6, 1.0) |
|  | LB, SB | 3 | 0.1 (0.0, 0.4) | 6 | 0.3 (0.1, 0.6) | 11 | 0.5 (0.3, 1.0) | 20 | 0.3 (0.2, 0.5) |
| **Musculoskeletal** | LB, SB, OPL | 1361 | 63.0 (59.7, 66.4) | 1464 | 68.6 (65.1, 72.2) | 1406 | 68.7 (65.2, 72.4) | 4231 | 66.7 (64.7, 68.7) |
|  | LB, SB | 1178 | 54.5 (51.5, 57.7) | 1233 | 57.8 (54.6, 61.1) | 1169 | 57.1 (53.9, 60.5) | 3580 | 56.4 (54.6, 58.3) |
| Diaphragmatic Hernia | LB, SB, OPL | 63 | 2.9 (2.2, 3.7) | 70 | 3.3 (2.6, 4.1) | 65 | 3.2 (2.5, 4.1) | 198 | 3.1 (2.7, 3.6) |
|  | LB, SB | 52 | 2.4 (1.8, 3.2) | 59 | 2.8 (2.1, 3.6) | 54 | 2.6 (2.0, 3.4) | 165 | 2.6 (2.2, 3.0) |
| Gastroschisis | LB, SB, OPL | 73 | 3.4 (2.7, 4.3) | 61 | 2.9 (2.2, 3.7) | 54 | 2.6 (2.0, 3.4) | 188 | 3.0 (2.6, 3.4) |
|  | LB, SB | 69 | 3.2 (2.5, 4.0) | 52 | 2.4 (1.8, 3.2) | 42 | 2.1 (1.5, 2.8) | 163 | 2.6 (2.2, 3.0) |
| Omphalocele | LB, SB, OPL | 75 | 3.5 (2.7, 4.4) | 102 | 4.8 (3.9, 5.8) | 99 | 4.8 (3.9, 5.9) | 276 | 4.4 (3.9, 4.9) |
|  | LB, SB | 28 | 1.3 (0.9, 1.9) | 46 | 2.2 (1.6, 2.9) | 38 | 1.9 (1.3, 2.6) | 112 | 1.8 (1.5, 2.1) |
| Limb Reduction | LB, SB, OPL | 124 | 5.7 (4.8, 6.8) | 93 | 4.4 (3.5, 5.3) | 112 | 5.5 (4.5, 6.6) | 329 | 5.2 (4.6, 5.8) |
|  | LB, SB | 96 | 4.4 (3.6, 5.4) | 64 | 3.0 (2.3, 3.8) | 76 | 3.7 (2.9, 4.7) | 236 | 3.7 (3.3, 4.2) |
| **Respiratory** | LB, SB, OPL | 100 | 4.6 (3.8, 5.6) | 139 | 6.5 (5.5, 7.7) | 177 | 8.7 (7.4, 10.0) | 416 | 6.6 (5.9, 7.2) |
|  | LB, SB | 90 | 4.2 (3.4, 5.1) | 117 | 5.5 (4.5, 6.6) | 155 | 7.6 (6.4, 8.9) | 362 | 5.7 (5.1, 6.3) |
| aIndividuals with multiple birth defects are included once in the overall counts as unique cases, while stratified rows represent each birth defect separately. Individuals with birth defects that could not be classified beyond “other” are included in the overall case-counts, but not in stratified birth defects counts.bLive Births (LB), Stillbirths (SB) and other pregnancy losses (OPLs; miscarriage (<20 weeks gestational age (GA)) or elective terminations (any GA)cAll birth defects codes included in each overall category are listed in Supplemental Table S1. Specific birth defects listed here within each category are not exhaustive of all birth defects for that category. Birth defects reported here are modeled after the list of birth defects routinely collected from state programs through the National Birth Defects Prevention Network due to their public health importance and ability to be identified at birth. dCritical congenital heart defects includes: Coarctation of Aorta, Double Outlet Right Ventricle, Ebstein Anomaly, Hypoplastic Left Heart Syndrome, Interrupted Aortic Arch, Pulmonary Valve Atresia (with and without Ventricular Septal Defect), Single Ventricle, Dextro-Transposition of the Great Arteries, Tetralogy of Fallot with and without Pulmonary Atresia, Total Anomalous Pulmonary Venous Connection, Tricuspid Valve Atresia, and Truncus Arteriosus (Common Truncus).eNeural tube defects includes: Anencephaly, Encephalocele, and Spina Bifida with and without hydrocephalusAbbreviations: Gestational age, GA; Live births, LB; Non-Hispanic, NH; Oher pregnancy losses, OPLs; Stillbirths, SB. |