Massachusetts Birth Defects

2015-2018

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

Massachusetts Department of Public Health

July 2022, updated March 29, 2023 and August 9, 2023

About Birth Defects

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

About the Massachusetts Center for Birth Defects Research and Prevention

The Massachusetts Birth Defects Monitoring Program has been collecting information on live births and stillbirths with birth defects since 1997. Beginning in 2012, we also began to collect data on other pregnancy losses with birth defects, which include terminations and miscarriages less than 20 weeks gestational age. Potential cases are identified through reports from delivery and specialty hospitals, prenatal diagnostic centers, vital records, and other sources.[2] As an active, population-based surveillance program, maternal and infant records for potential cases are reviewed to confirm the presence of birth defects.

Data collected through the program is used to inform public health policy and for program planning and prevention efforts. The program is constantly working to improve case-finding in order to provide the most complete data possible. As a result, comparisons to previous reports must be interpreted with caution. This brief report presents data on the prevalence of birth defects in Massachusetts for deliveries in the years 2015 through 2018.

Prevalence of Birth Defects in Massachusetts

From 2015 through 2018, we identified 10,289 cases with one or more birth defects (7,810 live births, 153 stillbirths, and 2,326 other pregnancy losses). The denominator used is 282,607 live births to Massachusetts residents during this same time period. This results in an overall prevalence of 364 per 10,000 live births in Massachusetts for 2015-2018.

Table 1 shows the case counts for specific types of birth defects overall and by birth outcome (live births, stillbirths, other pregnancy losses). In addition, the prevalence of each type of defect (case counts divided by number of live births) is presented with 95% confidence intervals.

Genitourinary, chromosomal, cardiovascular, and musculoskeletal defects are the most common in Massachusetts. Appendix 1 provides a list of the defects collected by the Massachusetts Birth Defects Monitoring Program and their associated codes. A glossary of birth defects can be found in Appendix 2.

References

1. CDC. About Birth Defects. https://www.cdc.gov/ncbddd/birthdefects/index.html.
2. Liberman RF, Getz KD, Lin AE, Higgins CA, Sekhavat S, Markenson GR, et al. Delayed diagnosis of critical congenital heart defects: trends and associated factors. *Pediatrics*. 2014;134(2):e373-381.10.1542/peds.2013-3949

For Additional Information on Massachusetts Birth Defects

For additional information on birth defects in Massachusetts, please see the Massachusetts Center for Birth Defects Research and Prevention website:
www.mass.gov/dph/birthdefects

Massachusetts Public Health Information Tool

A data portal for Massachusetts health data, including birth defects, may be found here:
https://www.mass.gov/orgs/population-health-information-tool

Massachusetts Environmental Public Health Tracking

Data on environmental, community and health indicators by community can be found here:
https://matracking.ehs.state.ma.us/

National Birth Defects Prevention Network

Data on major birth defects from US population-based Birth Defects Surveillance Programs, including Massachusetts, can be found here:
https://www.nbdpn.org/birth_defects_data_tables_and.php

General Birth Defects Information

Birth defects definitions and information from the Centers for Disease Control may be found here:
https://www.cdc.gov/ncbddd/birthdefects/facts.html

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>
> https://www.mass.gov/lists/massachusetts-birth-defects-surveillance-reports

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Table 1. Counts, Prevalence and 95% Confidence Intervals for Birth Defects, Massachusetts Birth Defects Monitoring Program, 2015-2018

Body system		Defect	Live Birth Count	Stillbirth Count	Other Loss Count	Total Count	Rate per 10,000 Births (95% Confidence Interval)
Central Nervous System		Anencephaly	10	10	71	91	3.2 (2.6-4.0)
		Spina Bifida with and without Hydrocephaly	65	0	62	127	4.5 (3.8-5.4)
		Hydrocephaly without Spina Bifida	102	8	73	183	6.5 (5.6-7.5)
		Encephalocele	5	1	16	22	0.8 (0.5-1.2)
		Microcephaly	134	0	5	139	4.9 (4.1-5.8)
		Holoprosencephaly	13	6	46	65	2.3 (1.8-2.9)
		Spinal Cord	130	0	5	135	4.8 (4.0-5.7)
		Other CNS	347	14	103	464	16.4 (15.0-18.0)
Eye		Anophthalmia/Microphthalmia	25	0	5	30	1.1 (0.7-1.5)
		Congenital Glaucoma, Congenital Cataract	103	0	0	103	3.6 (3.0-4.4)
		Aniridia	11	0	0	11	0.4 (0.2-0.7)
		Other Eye	112	0	3	115	4.1 (3.4-4.9)
Ear		Anotia/Microtia	66	1	2	69	2.4 (1.9-3.1)
		Other Ear	155	1	2	158	5.6 (4.8-6.5)
Cardiovascular	Anomalous Pulmonary Venous Connection	Partial	21	1	3	25	0.9 (0.6-1.3)
		Total	19	0	3	22	0.8 (0.5-1.2)
	Atrioventricular Canal Defects	Atrioventricular Septal Defect	139	13	26	178	6.3 (5.4-7.3)
	Conotruncal (Outlet) and Aortic Arch	Truncus	9	3	5	17	0.6 (0.4-1.0)
		dextro-Transposition of the Great Arteries	57	1	4	62	2.2 (1.7-2.8)
		Tetralogy of Fallot with or without Pulmonary Atresia	128	1	31	160	5.7 (4.8-6.6)
		Double Outlet Right Ventricle	31	2	20	53	1.9 (1.4-2.5)
		Interrupted Aortic Arch, Type B	4	0	0	4	0.1 (0.0-0.4)
	Ebstein Anomaly	Ebstein Anomaly	16	0	2	18	0.6 (0.4-1.0)

Table 1 cont.							
Body System		**Defect**	**Live Birth Count**	**Stillbirth Count**	**Other Loss Count**	**Total Count**	**Rate per 10,000 Births (95% Confidence Interval)**
Cardiovascular cont.	Left-Sided Obstruction	Aortic Valve Stenosis	43	1	2	46	1.6 (1.2-2.2)
		Hypoplastic Left Heart Syndrome	53	4	26	83	2.9 (2.3-2.6)
		Coarctation of Aorta	125	2	2	129	4.6 (3.8-5.4)
		Interrupted Aortic Arch (Type A and NOS)	4	0	2	6	0.2 (0.1-0.5)
		Aortic Arch Atresia without HLHS	3	0	1	4	0.1 (0.0-0.4)
	Right-Sided Obstruction	Tricuspid Valve Atresia	14	1	3	18	0.6 (0.4-1.0)
		Pulmonary Valve Atresia with VSD	9	2	2	13	0.5 (0.2-0.8)
		Pulmonary Valve Atresia with intact septum	11	1	10	22	0.8 (0.5-1.2)
		Pulmonary Valve Stenosis	241	1	5	247	8.7 (7.7-9.9)
	Septal Defects	VSD, Muscular, Membranous, Other	779	12	38	829	29.3 (27.4-31.4)
		ASD (Secundum and NOS)	792	2	4	798	28.2 (26.3-30.3)
		VSD, Conoventricular/ Malalignment	43	3	7	53	1.9 (1.4-2.5)
	Single Ventricle and L-TGA	Levo-Transposition of the Great Arteries	10	0	3	13	0.5 (0.2-0.8)
		Single Ventricle	10	0	11	21	0.7 (0.5-1.1)
	Other Cardiovascular	Other Cardiovascular	715	14	85	814	28.8 (26.9-30.9)
Respiratory		Choanal Atresia	15	0	0	15	0.5 (0.3-0.9)
		Lung Anomalies	78	5	10	93	3.3 (2.7-4.0)
		Other Respiratory	88	5	21	114	4.0 (3.3-4.9)

Table 1 cont.						
Body System	**Defect**	**Live Birth Count**	**Stillbirth Count**	**Other Loss Count**	**Total Count**	**Rate per 10,000 Births (95% Confidence Interval)**
Orofacial	Cleft Palate without Cleft Lip	160	1	8	169	6.0 (5.1-7.0)
	Cleft Lip with and without Cleft Palate	172	4	32	208	7.4 (6.4-8.4)
	Pierre Robin Sequence	41	0	1	42	1.5 (1.1-2.0)
	Other Orofacial	124	4	8	136	4.8 (4.0-5.7)
Gastrointestinal	Esophageal Atresia/Tracheoesophageal Fistula	66	1	0	67	2.4 (1.8-3.0)
	Rectal and Large Intestinal Atresia/Stenosis	108	3	7	118	4.2 (3.5-5.0)
	Hirschsprung Disease	50	0	0	50	1.8 (1.3-2.3)
	Biliary Atresia	17	0	0	17	0.6 (0.4-1.0)
	Small Intestinal Atresia	73	4	0	77	2.7 (2.2-3.4)
	Other Gastrointestinal	218	7	10	235	8.3 (7.3-9.5)
Genitourinary	Renal Agenesis/Hypoplasia	229	8	39	276	9.8 (8.7-11.0)
	Bladder Exstrophy	5	0	1	6	0.2 (0.1-0.5)
	Posterior Urethral Valve[a]	32	1	9	42	2.8 (2.0-3.9)
	Obstructive genitourinary-Other	1151	5	34	1190	42.1 (39.8-44.6)
	Hypospadias, 1st Degree or Other[a]	654	2	4	660	45.7 (42.3-49.3)
	Hypospadias, 2nd or 3rd Degree[a]	335	0	0	335	23.0 (20.6-25.7)
	Cloacal Exstrophy	11	0	0	11	0.4 (0.2-0.7)
	Other Genitourinary	1209	16	81	1306	46.2 (43.8-48.8)
Musculoskeletal	Reduction Deformity, Upper Limbs	62	6	23	91	3.2 (2.6-4.0)
	Reduction Deformity, Lower Limbs	25	3	17	45	1.6 (1.2-2.1)
	Gastroschisis	67	1	14	82	2.9 (2.3-3.6)
	Omphalocele	55	7	83	145	5.1 (4.3-6.0)
	Diaphragmatic Hernia	73	3	15	91	3.2 (2.6-4.0)

Table 1 cont.						
Body System	**Defect**	**Live Birth Count**	**Stillbirth Count**	**Other Loss Count**	**Total Count**	**Rate per 10,000 Births (95% Confidence Interval)**
Musculoskeletal cont.	Craniosynostosis	154	0	1	155	5.5 (4.7-6.4)
	Club Foot	433	24	80	537	19.0 (17.4-20.7)
	Polydactyly/Syndactyly	489	6	46	541	19.1 (17.6-20.8)
	Skeletal Dysplasia	51	3	27	81	2.9 (2.3-3.6)
	Other Musculoskeletal	344	21	95	460	16.3 (14.8-17.8)
Chromosomal and other Syndromes	Trisomy 13	15	3	101	119	4.2 (3.5-5.0)
	Trisomy 21 (Down Syndrome)	330	14	493	837	29.6 (27.7-31.7)
	Trisomy 18	25	22	218	265	9.4 (8.3-10.6)
	Turner Syndrome[b]	21	8	191	216	15.6 (13.6-17.9)
	Klinefelter Syndrome	50	0	21	71	2.5 (2.0-3.2)
	Deletion 22 q11.2	38	1	13	52	1.8 (1.4-2.4)
	Other Chromosomal Syndromes/Other Syndromes	496	21	790	1307	46.3 (43.8-48.8)
Other	Amniotic Bands	10	3	10	23	0.8 (0.5-1.2)
	Heterotaxy	36	2	7	45	1.6 (1.2-2.1)
	Skin Anomalies	43	1	0	44	1.6 (1.1-2.1)
	Other, Specified	49	9	18	76	2.7 (2.1-3.4)

Abbreviations: ASD-atrial septal defect; NOS-Not otherwise specified; VSD-ventricular septal defect.
Cases can be included in the count for more than one defect.
[a]Defect reported among Males only.
[b]Defect reported among Females only.

Appendix 1

List of Birth Defects and Codes Included in this Report				
Birth Defect	**ICD-9-CM Codes[1]**	**ICD-10-CM Codes[2]**	**Modified ICD-9-CM/BPA Codes[3]**	**Comments**
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-Q05.9 Q07.01, 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/ Microphthalmia	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	

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Birth Defect	**ICD-9-CM Codes[1]**	**ICD-10-CM Codes[2]**	**Modified ICD-9-CM/BPA Codes[3]**	**Comments**
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	Without Hypoplastic Left Heart Syndrome
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 (ASD) Primum, Common Atrium, Complete atrioventricular canal, Endocardial cushion defect, Ventricular Septal Defect (VSD), Canal type
ASD, Secundum, and Other	745.5	Q21.1, Q21.8, Q21.9	745.51, 745.58,745.59	
Coarctation of Aorta	747.10	Q25.1	747.10 – 747.19	
Dextro-Transposition of the Great Arteries	745.10	Q20.3, Q20.5, Q20.8	745.10, 745.11	Excludes 745.19
Double Outlet Right Ventricle	745.11	Q20.1, Q20.3, Q20.5, Q20.8	745.185, 745.186, 745.188, 745.189	
Ebstein Anomaly	746.2	Q22.5	746.20	
Hypoplastic Left Heart Syndrome	746.7	Q23.4	746.70	
Interrupted Aortic Arch	747.11	Q25.21, Q25.29, Q25.49	747.215 – 747.217	Includes Type A, Type B and Other
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 Atresia	746.00, 746.01	Q22.0, Q22.3	746.00, 746.03	With or without VSD

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Birth Defect	**ICD-9-CM Codes[1]**	**ICD-10-CM Codes[2]**	**Modified ICD-9-CM/BPA Codes[3]**	**Comments**
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 Atresia	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)	
VSD, Muscular, Membranous or Other	745.4	Q21.0, Q21.8, Q21.9	745.485, 745.486,745.49	
VSD, 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

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Orofacial				
Cleft Palate without Cleft Lip	749.0	Q35.1-Q35.9	749.00 – 749.07, 749.09	Excludes 749.08 (cleft uvula)
Cleft lip with/without Cleft Palate	749.1, 749.2	Q36.0-Q36.9 Q37.0-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	
Hypospadias[a]	752.61	Q54.0-Q54.9, excluding Q54.4	752.60, 752.62	In males only. Excludes 752.61 epispadias.
Posterior Urethral Valve[a]	753.6	Q64.2	753.60	Males only
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

List of Birth Defects and Codes Included in this Report				
Birth Defect	**ICD-9-CM Codes[1]**	**ICD-10-CM Codes[2]**	**Modified ICD-9-CM/BPA Codes[3]**	**Comments**
Renal Agenesis/ Hypoplasia	753.0	Q60.0-Q60.6	753.00- 753.01	
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
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	

List of Birth Defects and Codes Included in this Report				
Birth Defect	**ICD-9-CM Codes[1]**	**ICD-10-CM Codes[2]**	**Modified ICD-9-CM/BPA Codes[3]**	**Comments**
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 Syndrome[b]	758.6	Q96.0-Q96.9	758.60 – 758.69	In females only
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-	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),	Excludes Deletion 22 q11.2

List of Birth Defects and Codes Included in this Report				
Birth Defect	**ICD-9-CM Codes[1]**	**ICD-10-CM Codes[2]**	**Modified ICD-9-CM/BPA Codes[3]**	**Comments**
		Q98,9, Q99.0, Q99.8, Q99.9	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	

VSD: Ventricular Septal Defect; ASD: Atrial Septal Defect.
[a]Defect reported among Males only.
[b]Defect reported among Females only.

[1] International Classification of Diseases, 9th Revision.
[2] International Classification of Diseases, 10th Revision.
[3] Centers for Disease Control/Clinical Modification, British Pediatric Association.

NOTE: Some codes in the table above use shorthand with only 2 digits after the decimal point; for these the 3rd digit is implied and may include anything from 0 to 9.

Important changes to case ascertainment that may impact prevalence rates:
-Starting with deliveries in 1/1/2014, muscular ventricular septal defects (VSDs), unilateral renal agenesis, and hypospadias first degree and other began to be included in surveillance.
-Effective 7/1/2015, the surgical requirement was dropped for other genitourinary defects.

Appendix 2. Glossary of Terms Used in this Report

Agenesis The complete absence of part(s) of the body

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

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

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

Anotia A congenital absence of one or both ears.

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

Atresia Absence or closure of a normal opening.

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

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

Birth prevalence (# of cases with birth defect A in an area and time period ÷ #of live births in that area and period) X 10,000. See also Prevalence.

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

Cardiovascular See Heart Defects.

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

Central Nervous System Related to the brain or spinal cord.

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

Chromosomal Relating to chromosomes. Chromosomal defects involve abnormal structure or number of chromosomes, including partial or total absence of chromosomes or presence of extra chromosomes or parts of chromosomes. Examples include trisomy 13, Turner syndrome, and Down syndrome (trisomy 21).

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

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

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

Common truncus See Truncus arteriosus.

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

Congenital Existing at or dating from birth.

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

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

Down Syndrome (Trisomy 21) The chromosomal abnormality characterized by an extra copy of chromosome 21. In rare cases this syndrome is caused by translocation. Down syndrome can occur in mosaic, where there are some normal cells and some trisomy 21 cells. Many infants with Down syndrome also have congenital heart disease.

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

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

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

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

Fetal death See stillbirth.

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

Gastrointestinal Related to the digestive system. Includes defects of the stomach, esophagus, and liver. Examples include esophageal atresia,

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

Genital or Urinary (Genitourinary) Related to the genital or urinary organs.

Heart defects (Cardiovascular defects) Congenital heart defects affect the structure and function of a baby's heart and circulation system.

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

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

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

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

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

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

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

Limb defects See Reduction deformities.

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

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

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

Microtia A small or maldeveloped external ear and absent or closed off external auditory canal.

Musculoskeletal Related to the limbs or skeletal systems. Musculoskeletal defects may involve absence, abnormality or hypoplasia of limbs, like arms or legs, or structural abnormalities of limbs, muscles (such as diaphragmatic hernia), or abdominal wall (such as gastroschisis or omphalocele).

Neural Tube Defect A type of defect that occurs when the neural tube doesn't close properly in early pregnancy. Includes defects of the brain and spinal cord, like anencephaly, spina bifida and encephalocele.

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

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

Orofacial Related to the mouth or face. Examples include choanal atresia and clefts.

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

Posterior Urethral Valves Posterior urethral valves (PUV) are tissue folds in the posterior urethra. Congenital PUV is an abnormal membrane in males in the posterior urethra and is the most common cause of bladder outlet obstruction in male children.

Prevalence The number of birth defects observed during a period of time divided by the number of live births during the same time period. (# of cases with birth defect A in an area and time period ÷ #of live births in that area and period) X 10,000. See also Birth prevalence.

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

Reduction defects of the lower limbs The congenital absence of a portion of the lower limb (examples: a missing or shortened leg, missing toes.)

Reduction defects of the upper limbs The congenital absence of a portion of the upper limb (examples: a missing or shortened arm, missing fingers.)

Renal agenesis The failure of embryonic development of the kidney.

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

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

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

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

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

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

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

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

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

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

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

Trisomy 21 See Down Syndrome.

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

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

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

Definitions adapted from: Texas Department of State Health Services Glossary of Birth Defects Terms, last updated February 24, 2021: https://www.dshs.texas.gov/birthdefects/glossary.shtm

Additional references:
The Centers for Disease Control (CDC):
https://www.cdc.gov/ncbddd/index.html

The National Birth Defects Prevention Network (NBDPN):
https://www.nbdpn.org/docs/Appendix_3_1_BirthDefectsDescriptions_2017MAR24.pdf