MASSACHUSETTS

BIRTH DEFECTS REPORT 2019-2021

Massachusetts Birth Defects Monitoring Program
Division for Surveillance, Research, and Promotion of Perinatal Health
Bureau of Family Health and Nutrition
Massachusetts Department of Public Health
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MASSACHUSETTS

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Table of Contents

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 Birth Defects Monitoring Program

The Massachusetts Birth Defects Monitoring Program has been collecting information on live births and stillbirths with birth defects born to Massachusetts residents since 1997. Beginning in 2012, we began to collect data on birth defects among pregnancies that ended in other types of losses, including terminations at any gestational age and miscarriages less than 20 weeks gestation. 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 are 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 2019, 2020, and 2021.

Prevalence of Birth Defects in Massachusetts

From 2019 through 2021, we identified 8,173 cases with one or more birth defects (5,962 live births, 109 stillbirths, and 2,102 other pregnancy losses). The denominator used is live births to Massachusetts residents during this same time period. This results in an overall prevalence of 399 per 10,000 live births in Massachusetts for 2019-2021.

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 birth defect (case counts divided by number of live births x 10,000) 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 birth defects collected by the Massachusetts Birth Defects Monitoring Program and their associated codes. A glossary of birth defects can be found in Appendix 2.

[1]CDC About Birth Defects. www.cdc.gov/ncbddd/birthdefects/index.html

[2]Fothergill A, Liberman RF, Nestoridi E, Mai CT, Yeung LF, Higgins C and Yazdy MM, 2024. Expanding the Massachusetts Birth Defects Monitoring Program to include additional pregnancy outcomes: Programmatic efforts and impacts on case ascertainment, 2012–2020. Birth Defects Research, 116(3), p.e2323

Additional Information on Birth Defects

For additional information on birth defects in Massachusetts, please see the Massachusetts Birth Defects Monitoring Program website:
mass.gov/dph/birthdefects

Massachusetts Public Health Information Tool
A data portal for Massachusetts health data, including birth defects.
mass.gov/orgs/population-health-information-tool

Massachusetts Environmental Public Health Tracking
Data on environmental, community, and health indicators by community.
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.
nbdpn.org/birth_defects_data_tables_and.php

General Birth Defects Information
Birth defects definitions and information from the Centers for Disease Control.
cdc.gov/ncbddd/birthdefects/facts.html

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Table 1. Counts, Overall Prevalence Rate, and 95% Confidence Intervals for Birth Defects, Massachusetts, 2019-2021

Body System	Birth Defect	Livebirth Count	Stillbirth Count	Other Loss Count	Total Count	Rate per 10,000 Live Births (95% Confidence Interval)
Central Nervous System	Anencephaly	2	4	57	63	3.1 (2.4-3.9)
	Encephalocele	13	0	14	27	1.3 (0.9-1.9)
	Holoprosencephaly	12	2	30	44	2.2 (1.6-2.9)
	Hydrocephaly without Spina Bifida	86	5	61	152	7.4 (6.3-8.7)
	Microcephaly	58	0	2	60	2.9 (2.2-3.8)
	Spina Bifida with and without Hydrocephaly	50	2	37	89	4.4 (3.5-5.4)
	Spinal Cord Anomalies	106	1	5	112	5.5 (4.5-6.6)
	Other Central Nervous System	266	8	105	379	18.5 (16.7-20.5)
Eye	Aniridia	5	0	0	5	0.2 (0.1-0.6)
	Anophthalmia/ Microphthalmia	29	1	8	38	1.9 (1.3-2.6)
	Congenital Glaucoma/ Congenital Cataract	73	0	0	73	3.6 (2.8-4.5)
	Other Eye	108	1	9	118	5.8 (4.8-6.9)
Ear	Anotia/Microtia	37	1	3	41	2.0 (1.4-2.7)
	Other Ear	90	0	5	95	4.6 (3.8-5.7)
Cardiovascular	Anomalous Pulmonary Venous Connection					
	Partial	11	1	0	12	0.6 (0.3-1.0)
	Total	23	0	1	24	1.2 (0.8-1.7)
	Conotruncal (Outlet) and Aortic Arch					
	Double Outlet Right Ventricle	22	3	9	34	1.7 (1.2-2.3)
	Interrupted Aortic Arch	11	0	1	12	0.6 (0.3-1.0)
	Tetralogy of Fallot with or without Pulmonary Atresia	66	5	19	90	4.4 (3.5-5.4)

Body System	Birth Defect	Livebirth Count	Stillbirth Count	Other Loss Count	Total Count	Rate per 10,000 Births (95% Confidence Interval)
Cardiovascular	Conotruncal (Outlet) and Aortic Arch					
	Truncus Arteriosus	7	1	3	11	0.5 (0.3-1.0)
	dextro-Transposition of the Great Arteries	48	0	3	51	2.5 (1.9-3.3)
	Ebstein Anomaly	10	1	1	12	0.6 (0.3-1.0)
	Left-Sided Obstruction					
	Aortic Arch Atresia	1	0	1	2	0.1 (0.0-0.4)
	Aortic Valve Stenosis	35	0	1	36	1.8 (1.2-2.4)
	Coarctation of the Aorta	97	0	1	98	4.8 (3.9-5.8)
	Hypoplastic Left Heart Syndrome	25	0	15	40	2.0 (1.4-2.7)
	Right-Sided Obstruction					
	Pulmonary Valve Stenosis	154	0	3	157	7.7 (6.5-9.0)
	Pulmonary Valve Atresia with/without VSD	19	1	7	27	1.3 (0.9-1.9)
	Tricuspid Valve Atresia	6	1	4	11	0.5 (0.3-1.0)
	Septal defects					
	ASD Secundum/ Other	495	2	3	500	24.4 (22.3-26.7)
	Atrioventricular Septal Defect[a]	98	9	27	134	6.6 (5.5-7.8)
	VSD Cono-ventricular/ Malalignment	42	2	2	46	2.3 (1.7-3.0)

Body System	Birth Defect	Livebirth Count	Stillbirth Count	Other Loss Count	Total Count	Rate per 10,000 Live Births (95% Confidence Interval)
Cardiovascular	VSD Muscular, Membranous, Other	598	4	24	626	30.6 (28.2-33.1)
	Single Ventricle	11	0	7	18	0.9 (0.5-1.4)
	levo-Transposition of the Great Arteries	13	0	3	16	0.8 (0.5-1.3)
	Other Cardiovascular	561	7	66	634	31.0 (28.6-33.5)
Respiratory	Choanal Atresia	23	0	0	23	1.1 (0.7-1.7)
	Lung Anomalies	88	2	9	99	4.8 (3.9-5.9)
	Other Respiratory	47	1	13	61	3.0 (2.3-3.8)
Orofacial	Cleft Lip with and without Cleft Palate	124	6	38	168	8.2 (7.0-9.6)
	Cleft Palate without Cleft Lip	116	1	9	126	6.2 (5.1-7.3)
	Pierre Robin Sequence	39	0	1	40	2.0 (1.4-2.7)
	Other Orofacial	98	1	10	109	5.3 (4.4-6.4)
Gastrointestinal	Biliary Atresia	16	0	0	16	0.8 (0.5-1.3)
	Esophageal Atresia/Tracheoe-sophageal Fistula	46	1	1	48	2.3 (1.7-3.1)
	Hirschsprung Disease	42	0	0	42	2.1 (1.5-2.8)
	Rectal and Large Intestinal Atresia/ Stenosis	69	2	3	74	3.6 (2.8-4.5)
	Small Intestinal Atresia	50	2	0	52	2.5 (1.9-3.3)
	Other Gastrointestinal	151	1	8	160	7.8 (6.7-9.1)

Body System	Birth Defect	Livebirth Count	Stillbirth Count	Other Loss Count	Total Count	Rate per 10,000 Live Births (95% Confidence Interval)
Genitourinary	Bladder Exstrophy	7	0	1	8	0.4 (0.2-0.8)
	Cloacal Exstrophy	4	0	1	5	0.2 (0.1-0.6)
	Hypospadias[b], 1st Degree or NOS	454	3	2	459	43.9 (40.0-48.1)
	Hypospadias[b], 2nd or 3rd Degree	233	0	1	234	22.4 (19.6-25.4)
	Obstructive Genitourinary-Not PUV	1024	3	22	1049	51.3 (48.2-54.4)
	Posterior Urethral Valve (PUV)[a]	19	0	8	27	2.6 (1.7-3.8)
	Renal Agenesis/ Hypoplasia	163	4	24	191	9.3 (8.1-10.8)
	Other Genitourinary	914	9	61	984	48.1 (45.1-51.2)
Musculoskeletal	Club Foot	320	8	44	372	18.2 (16.4-20.1)
	Craniosynostosis	118	0	1	119	5.8 (4.8-7.0)
	Diaphragmatic Hernia	58	1	9	68	3.3 (2.6-4.2)
	Gastroschisis	39	5	9	53	2.6 (1.9-3.4)
	Omphalocele	29	4	63	96	4.7 (3.8-5.7)
	Polydactyly/ Syndactyly	359	8	39	406	19.8 (18.0-21.9)
	Reduction Defect, Lower Limbs	27	3	13	43	2.1 (1.5-2.8)
	Reduction Defect, Upper Limbs	46	4	33	83	4.1 (3.2-5.0)
	Skeletal Dysplasia	33	3	19	55	2.7 (2.0-3.5)
	Other Musculoskeletal	244	8	75	327	16.0 (14.3-17.8)

Body System	Birth Defect	Livebirth Count	Stillbirth Count	Other Loss Count	Total Count	Rate per 10,000 Live Births (95% Confidence Interval)
Chromosomal and Other Syndromes	Deletion 22q11.2	33	0	13	46	2.3 (1.7-3.0)
	Klinefelter Syndrome	54	1	22	77	3.8 (3.0-4.7)
	Trisomy 13	9	3	97	109	5.3 (4.4-6.4)
	Trisomy 18	29	12	158	199	9.7 (8.4-11.2)
	Trisomy 21 (Down syndrome)	244	13	457	714	34.9 (32.4-37.5)
	Turner Syndrome[c]	33	7	172	212	21.2 (18.4-24.2)
	Other Chromosomal/ Syndromes	446	28	825	1299	63.5 (60.1-67.0)
Other	Amniotic Bands	9	2	7	18	0.9 (0.5-1.4)
	Heterotaxy/Situs Inversus	33	0	5	38	1.9 (1.3-2.6)
	Skin Anomalies	24	0	1	25	1.2 (0.8-1.8)
	Other	49	4	12	65	3.2 (2.5-4.1)

Abbreviations: ASD-atrial septal defect; NOS-Not otherwise specified; VSD-ventricular septal defect.
Note: Cases can be included in the count for more than one defect.

[a]Atrioventricular septal defect includes: ASD primum, Common atrium, Complete atrioventricular canal defect, Endocardial cushion defect, and VSD, Canal type
[b]In Males only
[c]In 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	
Holoprosen-cephaly	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	

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/ Other	745.5	Q21.1, Q21.8, Q21.9	745.51, 745.58,745.59	
Coarctation of the 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	

Birth Defect	ICD-9-CM Codes[1]	ICD-10-CM Codes[2]	Modified ICD-9-CM/ BPA Codes[3]	Comments
Cardiovascular				
Partial Anomalous Pulmonary Venous Connection	747.42	Q26.3, Q26.4	747.43	
Pulmonary Valve Atresia with/without VSD	746.00, 746.01	Q22.0, Q22.3	746.00, 746.03	With or without VSD
Pulmonary Valve Stenosis	746.02	Q22.1	746.01	
Single Ventricle	745.3	Q20.4, Q20.8	745.30-745.33,745.38	
Tetralogy of Fallot with or without pulmonary atresia	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, Conoventricular/ Malalignment	745.4	Q21.0, Q21.8, Q21.9	745.487	
VSD, Muscular, Membranous, Other	745.4	Q21.0, Q21.8, Q21.9	745.485, 745.486, 745.49	
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	

Birth Defect	ICD-9-CM Codes[1]	ICD-10-CM Codes[2]	Modified ICD-9-CM/ BPA Codes[3]	Comments
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 Lip with and 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)
Cleft Palate without Cleft Lip	749.0	Q35.1-Q35.9	749.00 – 749.07, 749.09	Excludes 749.08 (cleft uvula)
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, 750.150	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, 750.50-750.70, 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	

Birth Defect	ICD-9-CM Codes[1]	ICD-10-CM Codes[2]	Modified ICD-9-CM/BPA Codes[3]	Comments
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 (PUV)[a]	753.6	Q64.2	753.60	In males only
Other Obstructive Genitourinary Defect-Not PUV	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	
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, 752.72, 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	

Birth Defect	ICD-9-CM Codes[1]	ICD-10-CM Codes[2]	Modified ICD-9-CM/ BPA Codes[3]	Comments
Musculoskeletal				
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
Reduction Defects—Upper or Lower Limbs	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	

Birth Defect	ICD-9-CM Codes[1]	ICD-10-CM Codes[2]	Modified ICD-9-CM/ BPA Codes[3]	Comments
Chromosomal and Other Syndromes				
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/ Syndromes	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	

Abbreviations: 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]International Classification of Diseases, 9th Revision, 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 on 1/1/2014, muscular ventricular septal defects (VSDs), unilateral renal agenesis, and hypospadias first degree and NOS began to be included in surveillance.
- Effective 7/1/2015, the surgical requirement was dropped for obstructive genitourinary defects.

Appendix 2: Glossary of Terms Used in this Report

Agenesis	The complete absence of part(s) of the body (see also aplasia or hypoplasia).
Aplasia or hypoplasia	The absence or incomplete development of an organ or body part (see also agenesis).
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. It can be treated 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 misshapen.
Diaphragmatic herbia	A failure of the diaphragm to form completely, leaving a hole. Abdominal organs 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 cells. Many infants with Down syndrome also have congenital heart disease.
Ebstein anomaly	A congenital heart defect in which the triscupid 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 protusion of an organ or part through connective tissue or through a wall of the cavity in which it is normally enclosed.
Hisrchsprung 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 Defects.
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/ 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.)

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 form 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 intellectual disability. 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 intellectual disability, 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, accessed March 7, 2024: www.dshs.texas.gov/birthdefects/glossary.shtm

Additional references:
National Center on Birth Defects and Developmental Disabilities,(NCBDDD), The Centers for Disease Control (CDC): www.cdc.gov/ncbddd/index.html

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

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