Maine Birth Defects Program

2025 Annual Report

1/1/2025 to 12/31/2025

Submitted by:
Maine Department of Health and Human Services
Maine Center for Disease Control and Prevention

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Executive Summary

Introduction

This 2025 annual report is prepared pursuant to 22 M.R.S. § 8945 and submitted by Maine Center for Disease Control and Prevention (Maine CDC). The Maine Birth Defects Program (the program) is charged with identifying cases of reportable birth defects in Maine and maintaining a central registry for monitoring children with identified birth defects and referrals to services, and reporting statistics annually. Data sets analyzed for birth defect occurrences and prevalence rates for annual reports cover a five-year span. This 2025 report reflects 2018-2023 data and statistical information from the central registry and summarizes program activities and accomplishments over the past calendar year and those planned activities for 2026.

Background

Maine's Department of Health and Human Services (the Department) is required to collect data to inform causes, risk factors, and strategies for improving prevalence of birth defects and access to services for children with a birth defect and their families. The Maine Birth Defects Program (Maine BDP) was established in 1999 and currently sits within Maine CDC, Maternal and Child Health Program. The program is charged with identifying and researching birth defects in children and maintaining a central registry of cases of birth defects in Maine.

Program Purpose

The Maine Birth Defects Program focuses on the three public health core functions of assessment, assurance, and policy development. These core functions are executed in conjunction with statutory requirements to:

- Provide an up-to-date birth defects registry.
- Assure data collection and sound data via epidemiological processes.
- Protect confidential information.
- Monitor additional birth defects recommended by the U.S. CDC and the National Birth Defects Prevention Network and determine which should be added to the Maine BDP.
- Develop, expand, enhance, and strengthen the program.
- Provide for primary prevention to decrease occurrence.
- Maintain components to educate populations about birth defects and systems.
- Refer those with birth defects to early intervention, Children and Youth with Special Health Care Needs and other support services.

Data and Statistical Reporting

By law, healthcare providers are required to report all cases involving a diagnosis of a reportable birth defect involving a fetus or infant born in the State of Maine to a Maine resident. The Maine BDP is authorized to access health care and other records for the purpose of investigating a reportable birth defect case. Information about potential cases identified through birth and infant death certificates and medical records discharge data is collected in the central registry which enables the program to produce State-level data for national reporting and to monitor birth defect trends. Maine's methodology for case identification combined passive case ascertainment with active case ascertainment. This means the Maine BDP may still be gathering data as late as December of the year after birth. Annually, the Maine BDP compiles information for this legislative report, and the Maine BDP also reports birth defects data every other year for a specific span of years to the U.S. CDC, National Birth Defects Prevention Network.

Related Program Activities

This 2025 annual report summarizes the current activities of the program, as well as the ongoing and upcoming activities for calendar year 2026. Activities from 2025 include:

- In2025A review of the hospital reporting of birth defects from discharge records identified lapses in reporting and following meetings with several hospitals, a new uniform data collection tool was implemented, and discharge reporting was re-started.
- An extensive review and evaluation of the abstractor case review process identified inefficiencies and, as a result, contracted services ended July 1, 2025.
- The Birth Defects Program Coordinator worked with the Epidemiologist to successfully prepare the data for submission to the National Birth Defects Prevention Network for the years 2018 to 2023.
- All 176 cases of reported birth effects (confirmed or suspected) were referred to Early Intervention for ME (EI4ME) and the Maine CDC's Children and Youth with Special Health Care Needs Program.
- Meetings with the Cleft Lip and Palate Clinic Coordinator were held monthly to stay current with referrals and other information.
- The Birth Defects Program Coordinator attended the National Birth Defects Prevention Network Conference in 2025.

Planned Activities

The program's goals for the future include:

- Expand support and services to pediatric specialty provider clinics who care for children with birth defects.
- Increase collaboration with hospitals and communities to expand the reach of the Program, including internal and external partners.
- Continue to update educational materials and make available information about current resources.
- Continue to work with an epidemiologist to refine data and assure quality and transition to new data capture processes.
- Convene with the Maine Environmental Health Tracking Network.
- Continue membership with the National Birth Defects Prevention Network.

Maine Birth Defects Program Annual Report

Background

A birth defect is defined as an abnormal condition that occurs before or at the time of birth. Birth defects include a wide range of abnormalities with varying levels of impact. Some birth defects are serious and can result in death, while others are less severe and can be treated with appropriate medical services. Birth defects may be caused by genetic factors, environmental, drug or medication exposures, while others remain unexplained. Birth defects can cause both mental and physical disabilities that affect children and their families for life.[1]

Birth defects affect about one in every 33 babies born in the United States each year. They are the leading cause of infant deaths, accounting for more than 20 percent of all infant deaths. Babies born with birth defects have a greater chance of illness and long-term disability than babies born without birth defects.[2] In order to prevent as many birth defects as possible and ensure the timely delivery of care, care coordination, and wrap-around services for families, it is essential to know what types of birth defects are occurring. With a surveillance program in place, it is possible to analyze birth defect data to detect temporal trends and assess impact for families and the care system.

The Maine Birth Defects Program commits fully to participate in epidemiological investigations as a means of informing public policy with respect to the equitable prevention activities and delivery of all components of care and treatment services.

Maine Law and Program Rules

Legislation supporting the Maine Birth Defects Program has and continues to define the program's purpose. Amendments made to the governing statute over time impacting the program's roles and responsibilities are highlighted below:

- **May 1999** - Public Law (P.L.1999 c. 344) established the Maine BDP within the Maine Department of Health and Human Services.
- **April 2003** - Program rules were formally adopted in April 2003 outlining reporting responsibilities and access to medical records. (22 M.R.S. c. 1687). Mandated reporting began May 2003, per Maine's birth defect program rule at 10-144 C.M.R. chapter 280.
- **May 2008** – Rules were updated to include three additional reportable birth defects.
- **April 2011** – Rules were amended to include the 45 birth defects recommended by the U. S. CDC and the National Birth Defects Prevention Network. Referral to the Part C Agency, Early Intervention for ME was also included.
- **December 2017** – Rules amended to include additional birth defects recommended by the U.S. CDC and the National Birth Defects Prevention Network.

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[1] Trust for America's Health "Birth defects and Developmental Disabilities: A Major Public Health Challenge"

[2] https://www.cdc.gov/ncbddd/birthdefects/index.html

[3] National Birth Defects Prevention Network http://www.nbdpn.org/

Partner Engagement

Key national-level partners include the National Birth Defects Prevention Network and the Association for Maternal and Child Health Programs (AMCHP). The Maine BDP also collaborates with a number of partners at the State and local levels, including:

- **Non-Governmental**
  - Parents and families
  - Hospitals and their staff
  - Primary care providers
  - Specialty physicians
  - Genetic counselors
  - Home Visiting
  - Family Voices
- **State of Maine**
  - Department of Education – Early Intervention for ME
  - Public Health Nursing
  - Environmental Public Health Tracking
  - Data, Research and Vital Statistics
  - Office of MaineCare Services
  - Pregnancy Risk Assessment Monitoring System

Maine CDC Birth Defects Program Summary

Maine CDC Birth Defects Program (the Program or Maine CDC BDP) was established within the Department of Health and Human services, Maine CDC - Maternal and Child Health (MCH) and is charged with tracking birth defects identified in children and maintaining a central registry for analyzing causes, risk factors, and program improvement strategies. Tracking cases of reported birth defects in the central registry enables the program to monitor birth defects trends and informs recommendations for improving access to services that monitor birth defect trends.

The Program began developing a birth defects surveillance system in 1999 with funding from the U.S. Centers for Disease Control and Prevention (U.S. CDC) to identify newborns with birth defects and monitor whether they receive appropriate services.

The Maine BDP’s current list of reportable birth defects is in alignment with the guidelines developed by the U.S. CDC National Birth Defects Prevention Network (NBDPN) for all of the core and recommended defects. The current list of Maine’s reportable birth defects may be found in Appendix A of this report.

For a case to be considered a reportable birth defect it must meet the following criteria:

- Infant was live born, stillborn, or prenatally diagnosed, with a gestational age of greater than 20 weeks or be a fetus less than 20 weeks gestation but with a prenatal diagnosis;
- The birth had to have occurred in Maine, and the mother is a Maine resident;
- Diagnosis was made before the infant reached 1 year of age; and
- The birth defect is included on the Maine BDP list of reportable birth defects.

Personnel and Funding Sources

State and federal funds are used to support MCH to promote and improve the health and well-being of the Maine’s children, including children with special needs, and their families. The Maine BDP consists of .4 FTE coordinator and .15 FTE Epidemiologist. The Maine BDP contracted with partners to provide data registry abstraction services until July 1, 2025. The program also supports the delivery of clinical services to infants and children with cleft lip and palate, a separate program focus area within MCH.

Program Operations

The Maine BDP contracted with Nebulogic in 2019 to develop and implement a new comprehensive surveillance and tracking system. The SMART Children's Health Screening Tracking System (CHSTS) links birth defect data with multiple data sources that include birth and death certificates; hospital discharge data; and newborn bloodspot, critical congenital heart defects (CCHD), and hearing screening data. The Maine Newborn Screening Portal (MNSP) maintains security/confidentiality of all records by assigning permission to access the system on an individual basis. By linking information from these existing data sources, the Maine BDP can provide valuable public health data to state and national policy makers. The Maine BDP continues to work with Nebulogic staff to increase capacity on the MNSP.

As a surveillance unit, the Maine BDP began passive case ascertainment with confirmation of cases by active case ascertainment on May 1, 2003. Passive case ascertainment with active case ascertainment is an approach whereby the surveillance program receives case reports of birth defects from a variety of data sources and then follows up with a review of the case. As required by statute, health care providers licensed under Title 22 and Title 32 are required to provide or make available health records and information relating to the occurrence of birth defects. Passive data sources include hospital case reports, prenatal assessments, birth and death certificates, and medical discharge records using diagnostic codes.

As part of the configuration of the surveillance system established for the Maine BDP, the Department contracted with an abstractor to perform case reviews to confirm the presence of a birth defect after the Maine BDP receives a provider's report. The abstractor requested records from the hospital/location of birth and reviewed those records to confirm the presence of a birth defect. Results of this abstraction process were entered into the CHSTS NebuLogic database. Following a recent evaluation of the case review process, due to the findings of delays and redundancy in and inefficiencies of the abstraction process, the abstraction service contract was not renewed beyond July 1, 2025. Maine BDP has continued to work with Department of Education - Child Development Services System directly on referrals to identify children eligible for services.

Referrals

Maine reports on over 50 birth defects and, according to rule and statute, the cases are entered into CHSTS for national reporting and the Maine BDP will refer to the Child Development Services System (CDS) children with a reportable birth defect who may be eligible for early intervention. (22 M.R.S. § 8943).

In the past, cases identified by the Maine BDP were confirmed through abstraction within the first three months after birth. However, due to a reduction in capacity with the provider used for abstractions, over the past several years, this timeframe lengthened significantly, with confirmation taking up to one year. Efforts to improve the data system are ongoing. MCH met with the Maine Part C Agency, Early Intervention for ME (EI4ME), and a decision was made to make referrals to CDS after passive ascertainment. At the time of this annual report, CDS reported 176 MCH referrals found eligible for referring to early intervention services. The Maine BDP coordinator continues to coordinate with DOE CDS regarding processing reported cases and referrals, without case confirmation. MCH is evaluating Maine's birth defect procedures to identify options that optimize both data quality and service timeliness.

Separate from Maine BDP referrals to CDS, reportable cases are also referred for care coordination through Maine's CYSHCN program. CYSHCN provides another level of MCH support to families to ensure children and youth with special health needs receive the appropriate care and services to achieve optimal health, development, and quality of life.

National Reporting

The Maine BDP submits data (counts of defects by defect, maternal age, maternal race and infant sex) for a specific span of years to the U.S. CDC, National Birth Defects Prevention Network. A birth defect is included if the birth defect is identified within the first year of life. This year, the NBDPN data request covered the years 2018 to 2023 and the data were submitted in September 2025. The official data summary for this report is not yet available. (See Appendix C for the last NBDPN report for Maine.) In addition, the NPDPN requested a state program "Performance Summary". (See Appendix B).

Improvement Strategies

Per 22 M.R.S. § 8945, the program looks for strategies to improve the operation of the program and the central registry. Recent work has included:

- A review of the hospital reporting of birth defects from discharge records revealed that several hospitals had stopped reporting.
    - Meetings with the hospitals were held, a new standardized reporting tool was created, and discharge reporting was re-initiated.
- An extensive review and evaluation were also made of the abstractor case review process as it was configured in the SMART, the software used for case management of the MCH screening and surveillance programs. As configured, the abstraction process was not currently meeting the data needs and efficient use of resources for the abstraction process to continue. The contract with the company doing the abstraction was discontinued on July 1, 2025.
    - To improve system efficiency and service timeliness regarding Maine's birth defects, MCH is reviewing current processes and methodologies, and, for future reports, ways of translating data into strategies to improve access to services and reduce birth defects rates and prevalence.

Conclusion

The Maine Birth Defects Program provides an important resource for tracking and identifying birth defects impacting children in Maine. The Maine BDP will continue to be a resource for families and clinicians and work with partners on strategies to improve the work of the Maine BDP and its registry.

Acknowledgements

The Maine Center for Disease Control and Prevention would like to extend our thanks the University of Southern Maine epidemiologists for their contributions and Maine's reporting facilities for their time and efforts to provide the case reports that are successful to this registry. Further, our thanks go to the Maine CDC staff, particularly from the Children and Youth with Special Health Needs program and the Office of Data, Research, and Vital Statistics for birth and death files, linkages, and support for reporting.

Appendix A

Reportable Birth Defects Included in Case Definition for 2023.

Birth Defect	**ICD-10-CM Codes**
Central Nervous System	
Anencephalous	Q00.0-Q00.1
Spina Bifida without anencephalous	Q05.0-Q05.9 Q07.01, Q07.03 w/o Q00.0-Q00.1
Encephalocele	Q01-Q01.9
Microcephalus	Q02
Holoprosencephaly	Q04.2
Eye	
Anophthalmia/microphthalmia	Q11.0-Q11.2
Congenital cataract	Q12.0
Ear	
Anotia/microtia	Q16.0, Q17.2
Cardiovascular	
Common truncus (truncus arteriosus or TA)	Q20.0
Double outlet right ventricle (DORV)	Q20.1
Interrupted aortic arch (IAA)	Q25.2, Q25.4 post 2016 25.21
Transposition of great arteries	Q20-Q20.9
Tetralogy of Fallot	Q21.3
Ventricular septal defect	Q21.0
Atrial septal defect	Q21.1
Atrioventricular septal defect (Endocardial cushion defect)	Q21.2
Pulmonary valve atresia and stenosis	Q22.0, 22.1
Tricuspid valve atresia and stenosis	Q22.4
Ebstein's anomaly	Q22.5
Aortic valve stenosis	Q23.0
Hypoplastic left heart syndrome	Q23.4
Coarctation of aorta	Q25.1
Total anomalous pulmonary venous connection (TAPVC)	Q26.2
Single Ventricle	Q20.4

Birth Defect	**ICD-10-CM Codes**
Orofacial	
Cleft palate without cleft lip	Q35.1 - Q35.9
Cleft lip with and without cleft palate	Q36.0 - 36.9, Q37.0 - Q37.9
Choanal atresia	Q30.0
Gastrointestinal	
Esophageal atresia/tracheoesophageal fistula	Q39.0 - 39.4
Rectal and large intestinal atresia/stenosis	Q42.0 - Q42.9
Biliary atresia	Q44.2 – Q44.3
Small intestinal atresia/stenosis	Q41.0 – Q41.9
Genitourinary	
Renal agenesis/hypoplasia	Q60 – Q60.6
Bladder exstrophy	Q64.10 – Q64.19
Hypospadias and Epispadias	Q51.0 - Q54.9 (excluding Q54.4)
Cloacal exstrophy	Q64.12
Congenital Posterior Urethral Valves	Q64.2
Musculoskeletal	
Reduction deformity, upper limbs	Q71.0-Q71.9, 73.0 – Q73.8
Reduction deformity, lower limbs	Q72.0- Q72.9
Gastroschisis	Q79.3
Omphalocele	Q79.2
Diaphragmatic hernia	Q79.0, Q79.1
Clubfoot	Q66.0, Q66.89
Craniosynostosis	Q75.0
Chromosomal	
Trisomy 13	Q91.4 – Q91.7
Down syndrome (Trisomy 21)	Q90.0 – Q90.9
Trisomy 18	Q91.0 – Q91.3
Deletion 22q11	Q93.81
Turner syndrome	Q96.0 – Q96.9

Appendix B.

Birth Defects Counts and Prevalence Rates (2018-2023)

Data Sources: Birth Defects: Maine Birth Defects Registry, extract August 2025.
Births: Maine CDC, Data, Research, and Vital Statistics (DRVS)
General comments: Case-finding is limited to babies born in Maine and to birth defects identified in the first year of life. A baby can be born with multiple conditions; adding up the number of defects will not yield the number of babies born with defects.
National birth defects data for previous years can be found at the following link:
https://nbdpn.org/birth-defects-data-tables-and-directory/

Condition	**Summary**	**Six Year Count**	**Prevalence per 10,000 Live Births and 95% Confidence Interval**
Anencephalus	One in 13,886 live births	5	0.7 (0.2 - 1.7)
Anophthalmia/microphthalmia	One in 69,431 live births	1	0.1 (0.0 - 0.8)
Anotia/microtia	One in 7,715 live births	9	1.3 (0.6 - 2.5)
Aortic valve stenosis	One in 13,886 live births	5	0.7 (0.2 - 1.7)
Atrial septal defect	One in 335 live births	207	29.8 (25.9 - 34.2)
Atrioventricular septal defect	One in 6,943 live births	10	1.4 (0.7 - 2.7)
Biliary atresia	One in 69,431 live births	1	0.1 (0.0 - 0.8)
Bladder exstrophy	One in 69,431 live births	1	0.1 (0.0 - 0.8)
Choanal atresia	One in 7,715 live births	9	1.3 (0.6 - 2.5)
Cleft lip alone	One in 3,654 live births	19	2.7 (1.7 - 4.3)
Cleft lip with cleft palate	One in 1,615 live births	43	6.2 (4.5 - 8.3)
Cleft palate alone	One in 1,877 live births	37	5.3 (3.8 - 7.4)
Cloacal exstrophy	-----	0	-----
Clubfoot	One in 694 live births	100	14.4 (11.7 - 17.5)
Coarctation of the aorta	One in 6,312 live births	11	1.6 (0.8 - 2.8)
Common truncus (truncus arteriosus)	-----	0	-----
Congenital cataract	One in 69,431 live births	1	0.1 (0.0 - 0.8)
Congenital posterior urethral valves*	One in 1,420 live births	25	7.0 (4.6 - 10.4)
Craniosynostosis	One in 4,084 live births	17	2.4 (1.4 - 3.9)
Deletion 22q11.2	One in 23,144 live births	3	0.4 (0.1 - 1.3)
Diaphragmatic hernia	One in 11,572 live births	6	0.9 (0.3 - 1.9)
Double outlet right ventricle	One in 34,716 live births	2	0.3 (0.0 - 1.0)
Ebstein anomaly	-----	0	-----
Encephalocele	-----	0	-----
Esophageal atresia/tracheoesophageal	One in 6,943 live births	10	1.4 (0.7 - 2.7)
Gastroschisis	One in 4,959 live births	14	2.0 (1.1 - 3.4)
Holoprosencephaly	One in 17,358 live births	4	0.6 (0.2 - 1.5)

Condition	**Summary**	**Six Year Count**	**Prevalence per 10,000 Live Births and 95% Confidence Interval**
Hypoplastic left heart syndrome	One in 17,358 live births	4	0.6 (0.2 - 1.5)
Hypospadias*	One in 197 live births	180	50.7 (43.6 - 58.7)
Interrupted aortic arch	One in 13,886 live births	5	0.7 (0.2 - 1.7)
Limb deficiencies (reduction defects)	One in 3,156 live births	22	3.2 (2.0 - 4.8)
Omphalocele	One in 7,715 live births	9	1.3 (0.6 - 2.5)
Pulmonary valve atresia	-----	0	-----
Pulmonary valve atresia and stenosis	One in 9,919 live births	7	1.0 (0.4 - 2.1)
Rectal and large intestinal atresia/stenosis	One in 6,943 live births	10	1.4 (0.7 - 2.7)
Renal agenesis/hypoplasia	One in 2,893 live births	24	3.5 (2.2 - 5.1)
Single ventricle	One in 69,431 live births	1	0.1 (0.0 - 0.8)
Small intestinal atresia/stenosis	One in 8,679 live births	8	1.2 (0.5 - 2.3)
Spina bifida without anencephalus	One in 5,786 live births	12	1.7 (0.9 - 3.0)
Tetralogy of Fallot	One in 4,084 live births	17	2.4 (1.4 - 3.9)
Total anomalous pulmonary venous connection	One in 23,144 live births	3	0.4 (0.1 - 1.3)
Transposition of the great arteries	-----	0	-----
Tricuspid valve atresia	-----	0	-----
Tricuspid valve atresia and stenosis	One in 69,431 live births	1	0.1 (0.0 - 0.8)
Trisomy 13	One in 17,358 live births	4	0.6 (0.2 - 1.5)
Trisomy 18	One in 11,572 live births	6	0.9 (0.3 - 1.9)
Trisomy 21 (Down syndrome)	One in 1,052 live births	66	9.5 (7.4 - 12.1)
Turner syndrome**	One in 4,840 live births	7	2.1 (0.8 - 4.3)
Ventricular septal defect	One in 352 live births	197	28.4 (24.6 - 32.6)

Birth Data: Total live births include babies born in Maine.	
Total Live Births 2018-2023	*69,431*
Total Male Live Births 2018-2023	*35,488*
Total Female Live Births 2018-2023	*33,880*

Notes:

Count and rates are based on six years of available data for each condition from 2018-2023.

Prevalence rates are calculated per 10,000 live births.

Ninety-five percent confidence intervals are provided for all rates.

*Congenital posterior urethral valves: prevalence per 10,000 male live births

*Hypospadias: prevalence per 10,000 male live births

**Turner Syndrome: prevalence per 10,000 female live births.