Birth Defects Program Report

2024 Annual Report

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

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EXECUTIVE SUMMARY

Introduction

This 2024 annual report is prepared pursuant to 22 M.R.S. § 8945. Maine Center for Disease Control and Prevention Birth Defects Program (the *program*) is charged with identifying and researching birth defects in children and maintaining a central registry of cases of birth defects; the program is designed to be a central source of information to families and providers of the occurrence of birth defects in Maine. Health Resources and Services Administration, Maternal and Child Health Bureau block grant funds are used to support Maine's wide range of maternal and child health and children with special health needs activities across the State. This report summarizes the Program's activities and accomplishments for calendar year 2024 as well as planned activities for 2025 and it includes statistical information from the central registry. Data analyzed for birth defect occurrences and prevalence rates for annual reports covers a five-year span, 2016-2020 data is reflected in this 2024 report.

Background

The Maine Department of Health and Human Services is required to collect and report information on birth defects in Maine, pursuant to 22 M.R.S. §§ 8941-8945. The Department of Health and Human Services, Maine Center for Disease Control and Prevention - Birth Defects Program was established in 1999 and currently sits within the Division of Disease Prevention, 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.

Purpose

The program focuses on three established public health core functions of assessment, assurance, and policy development in conjunction with the requirements set out in statute. It aims 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 for surveillance purposes.
- Develop, expand, enhance and strengthen program impact.
- 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.

Related Activities

This 2024 annual report summarizes the current activities of the program, as well as the ongoing and upcoming activities for calendar year 2025.

Program Status

Over 2024, the program:

- Continued infrastructure building, including updating the database and evaluating abstractor case review processes to increase efficiency and accuracy.
- Maine CDC Maternal and Child Health (MCH) Program organizational structure was changed, and the Birth Defects Coordinator position was placed within the responsibilities of a new position, Newborn Health Coordinator. The Newborn Health Coordinator also coordinates newborn critical congenital heart disease (CCHD) screening surveillance and newborn bloodspot screening. In addition, a new epidemiologist was assigned to work with the program.
- Continued efforts with the program epidemiologist to refine data and assure quality.
- All babies with birth defects (confirmed or suspected) were referred to Early Intervention for ME and Maine CDC's Children and Youth with Special Health Care Needs Program.
- Met with contracted Cleft Lip and Palate Clinic Coordinator monthly to stay current with referrals and other information.
- New strategies were considered for birth defects surveillance procedures within the Maine Newborn Screening Portal to more accurately capture data.

Future Direction

Going forward, plans for the program include:

- Expanding support and services to pediatric specialty provider clinics who care for children with birth defects.
- Increasing collaboration with hospitals and communities to expand the reach of the program, including internal and external partners, and considering an advisory committee or work group as part of newborn health/screening and surveillance.
- Updating educational materials.
- Continuing to develop SMART Children's Health Screening Tracking System (CHSTS) for the State of Maine (SMART CHSTS Maine Newborn Screening Portal) to accurately capture birth defects data.
- Continuing to work with an epidemiologist to refine data and assure quality.
- Transition to new data capture processes and begin reporting program referral statistics.
- Meeting with the Maine Environmental Health Tracking Network.
- Continuing membership in the National Birth Defects Prevention Network.
- Attending National Birth Defects Prevention Network Conference in 2025.

Maine Birth Defects Program Full 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. A population-based birth defects surveillance and referral program that 1) uses multiple sources of data and 2) commits to early referral to an established network of services that are family-centered, culturally competent and community-based so that children achieve optimal health and develop to their full potential enables a surveillance program to accurately quantify morbidity and mortality, detect temporal trends, and assess the financial burden on families and State programs that birth defects may cause.[3] Many children who survive have a lifetime of major expenses from essential services such as specialty medical care, special education, rehabilitation, and developmental services.

Maine CDC Birth Defects Program (BDP) commits fully to participating 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 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 1999, chapter 344 established the program within the Maine Department of Health and Human Services. Program rules were formally adopted in April 2003 outlining reporting responsibilities and access to medical records, in accordance with 22 M.R.S. chapter 1687. Mandated reporting began May 2003.
- **September 2007** – Public Law 2007, chapter 450 amended statutory language specific to the registry and referrals to Child Development Services System.
- **May 2008** – Rules[3] were updated to include three additional reportable birth defects.

[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/
[3] Maine Birth Defects Program Rule, 10-144 CMR, c. 280; https://www.maine.gov/sos/sites/maine.gov.sos/files/content/assets/144c280.docx

- **April 2011** – Rules[4] 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[5] were amended to include additional birth defects recommended by the U.S. CDC and the National Birth Defects Prevention Network.

Stakeholder Engagement

The table below shows collaborators and partners to the program. National partners, the National Birth Defects Prevention Network and the Association for Maternal and Child Health Programs are also important to this work.

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

Program Description

Maine CDC Birth Defects Program (the *program* or Maine CDC BDP) began developing a birth defects surveillance system in 1999 with funding from the U.S. Centers for Disease Control and Prevention (CDC). The program was established within Maine CDC to identify newborns with birth defects, ensure they receive appropriate services, and to monitor birth defect trends.

As a surveillance unit, the program 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, those entities licensed under Title 22: Hospitals and Title 32: Licensed Professionals 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. Once a case is identified as a possible reportable birth defect, the case is assigned to an abstractor. The abstractor requests records from the hospital/location of birth and reviews those records to confirm the presence of a birth defect. Results of this abstraction process are entered into the CHSTS Nebulogic data base.

The program's current list of reportable birth defects aligns with the guidelines developed by the U.S. CDC National Birth Defects Prevention Network for the majority of core and recommended

[4] Maine Birth Defects Program Rule, 10-144 CMR, c. 280; https://www.maine.gov/sos/sites/maine.gov.sos/files/content/assets/144c280.docx
[5] Maine Birth Defects Program Rule, 10-144 CMR, c. 280; https://www.maine.gov/sos/sites/maine.gov.sos/files/content/assets/144c280.docx

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
- Birth defect is included on the program's list of reportable birth defects.

Potential cases are identified through weekly downloads of both the electronic birth and infant death certificates and medical records discharge data. Once a potential case is identified, abstraction is performed using a comprehensive electronic abstraction method. Information collected includes the nature and details of the birth defect, demographics, mother's health history, prenatal information, cytogenic and laboratory data, family history, and, when available, father's history, and mother's exposure to illegal drugs, medications, smoking, or alcohol. The data collected by the abstractors is entered into the birth defects surveillance section of the CHSTS tracking system.

Currently, the program reports on information gathered for 57 birth defects (see Appendix A for a complete list of reportable birth defects). These cases are confirmed usually within the first three months after birth. However, this timeframe can be longer depending on when the birth defect was reported and the ability of the abstractors to gather the necessary information from the birth hospital. There is also a lag time in verifying a birth defect because, by law, a birth defect can be diagnosed and reported within the first year of life.

Personnel and Funding Sources

Program staff include a .4 FTE coordinator and .15 FTE epidemiologist. The program currently contracts with partners to provide data registry abstraction services, as well as to facilitate the Cleft Lip and Palate clinical services.

Staff and contracted services are funded by state and federal funds allocated to promote and improve the health and well-being of the Maine's mothers, children, including children with special needs, and their families.

Goals, Activities and Achievements

The program gathers data about infants born each year with certain birth defects diagnosed within the first year of life. The statute requires the program to:

- Collect, analyze, and distribute information to identify the birth defects with regards to the following causes, risk factors and strategies for prevention and the provision of services,
- Establish a system for data collection that identifies prevalence and incidence rates by region and population group and identifies the morbidity and mortality rates resulting from birth defects,
- Conduct investigations to determine the nature, and extent of the disease or known or suspected causes of birth defects.
- Examples of Maine CDC BDP activity:

- Collection and abstraction of data on 57 birth defects. The Maine CDC BDP submits data bi-annually to the National Birth Defects Prevention Network and U.S. CDC.
- Collaboration with National Birth Defects Prevention Network on educational tools and promotion of prevention strategies.
- Collaboration with the Maine CDC Environmental Public Health Tracking Unit on relevant birth defects.

Cleft Lip and Palate

The program currently supports and collaborates with the Cleft Lip and Palate Clinic at MaineHealth Maine Medical Center. Until June 30, 2022, there were two active Cleft Lip and Palate clinics in Maine. Northern Light Health closed the Bangor clinic due to lack of specialists in the region. The program supported MaineHealth Maine Medical Center to increase their clinic's hours of operation to accommodate the additional patients. The Northern Light Cleft Clinic Coordinator continues to assist in the transition of patients.

Cleft lip and cleft palate are birth defects where the lip or roof of the mouth do not form properly during pregnancy. Together, these birth defects are commonly referred to as '*orofacial clefts*'. All families with a prenatal or postnatal diagnosis are offered a visit from a Public Health Nurse to provide support, answer questions, and connect families with the Cleft Lip and Palate clinic. Families may attend clinic until the child is 22 years of age, regardless of income. Families attending the clinic have access to multidisciplinary teams including a geneticist, genetic counselor, plastic surgeon, oral surgeon, prosthodontist, orthodontist, pediatric dentist, otolaryngologist (ears, nose, and throat) nurse practitioner, speech pathologist, social worker, and clinic coordinator. This team of specialists works collaboratively with families to develop the best treatment plan for each child's condition.

Children with a cleft lip and/or palate can find comprehensive care at Maine's Cleft Lip and Palate Clinic. In 2021, the clinic at MaineHealth Maine Medical Center became an American Cleft Palate-Craniofacial Association (ACPA) approved team. Clinic staff worked hard to maintain the necessary steps to remain ACPA- approved. The clinic staff also provides a full range of education and consultation services upon request in addition to monthly clinical services. Additionally, the Clinic Coordinator provides family and community educational opportunities on an annual basis. In 2022, 12 clinical sessions were offered serving over 194 families.

Maine CDC Birth Defects Tracking System

Tracking System

Maine CDC BDP began contracting with Nebulogic in 2019 to develop and implement a new comprehensive surveillance and tracking system. The Maine Newborn Screening Portal (MNSP) links birth defect data with multiple data sources that include birth and death certificates, hospital discharge data, and metabolic and newborn hearing screening data. 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, Maine CDC BDP can provide valuable public health data to State and national policy makers. In 2020, Maine CDC

BDP continued to work with Nebulogic staff to increase capacity on the Maine Newborn Screening Portal.

Statistical Reports

Maine CDC BDP submits data every other year for a specific span of years to the U.S. CDC, National Birth Defects Prevention Network. A birth defect meets criteria and is included in the Maine CDC BDP count if the birth defect is identified within the first year of life. This means the Maine CDC BDP may still be gathering data as late as December of the year after the birth of the baby. Consequently, time is needed to abstract the data necessary to confirm the birth defect. The next call for data will take place in March 2025, with a deadline in May 2025. The data included in this report (Appendix B) covers 2016 – 2020. The next report will cover 2018 – 2022.

Conclusion

The Maine Birth Defects Program provides an important resource to the state for tracking and identifying birth defects impacting children in Maine. We appreciate the opportunity to be a resource for families and clinicians and look forward to continuing to improve on our work.

Appendices

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
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 (2016-2020), Maine

Condition	Summary	Five Year Count	Prevalence per 10,000 Live Births and 95% Confidence Interval
Anencephalus	One in 8,370 live births	7	1.2 (0.5 - 2.5)
Anophthalmia/microphthalmia	One in 29,295 live births	2	0.3 (0 - 1.1)
Anotia/microtia	One in 8,370 live births	7	1.2 (0.5 - 2.5)
Aortic valve stenosis	One in 7,324 live births	8	1.4 (0.6 - 2.8)
Atrial septal defect	One in 514 live births	114	19.5 (15.8 - 23.7)
Atrioventricular septal defect	One in 3,662 live births	16	2.7 (1.5 - 4.4)
Biliary atresia	-----	0	-----
Bladder exstrophy	One in 19,530 live births	3	0.5 (0.1 - 1.5)
Choanal atresia	One in 9,765 live births	6	1 (0.4 - 2.2)
Cleft lip alone	One in 3,662 live births	16	2.7 (1.5 - 4.4)
Cleft lip with cleft palate	One in 1,674 live births	35	6 (4.2 - 8.3)
Cleft palate alone	One in 1,674 live births	35	6 (4.2 - 8.3)
Cloacal exstrophy	-----	0	-----
Clubfoot	One in 1,046 live births	56	9.6 (7.3 - 12.5)
Coarctation of the aorta	One in 3,084 live births	19	3.2 (1.9 - 5.0)
Common truncus (truncus arteriosus)	-----	0	-----
Congenital cataract	One in 19,530 live births	3	0.5 (0.1 - 1.5)
Congenital posterior urethral valves*	One in 29,727 live births	1	0.3 (0 - 1.7)
Craniosynostosis	One in 8,370 live births	7	1.2 (0.5 - 2.5)
Deletion 22q11.2	One in 29,295 live births	2	0.3 (0 - 1.1)
Diaphragmatic hernia	One in 14,647 live births	4	0.7 (0.2 - 1.8)
Double outlet right ventricle	One in 19,530 live births	3	0.5 (0.1 - 1.5)
Ebstein anomaly	-----	0	-----
Encephalocele	One in 29,295 live births	2	0.3 (0 - 1.1)
Esophageal atresia/tracheoesophageal	One in 8,370 live births	7	1.2 (0.5 - 2.5)
Gastroschisis	One in 3,906 live births	15	2.6 (1.5 - 4.3)
Holoprosencephaly	One in 19,530 live births	3	0.5 (0.1 - 1.5)
Hypoplastic left heart syndrome	One in 8,370 live births	7	1.2 (0.5 - 2.5)
Hypospadias*	One in 187 live births	159	53.5 (43.5 - 65.1)
Interrupted aortic arch	One in 14,647 live births	4	0.7 (0.2 - 1.8)
Limb deficiencies (reduction defects)	One in 5,326 live births	11	1.9 (0.9 - 3.4)
Omphalocele	One in 5,859 live births	10	1.7 (0.8 - 3.1)
Pulmonary valve atresia	-----	0	-----
Pulmonary valve atresia and stenosis	One in 5,859 live births	10	1.7 (0.8 - 3.1)
Rectal and large intestinal atresia/stenosis	One in 3,084 live births	19	3.2 (1.9 - 5.0)
Renal agenesis/hypoplasia	One in 2,253 live births	26	4.4 (2.9 - 6.4)
Single ventricle	One in 58,589 live births	1	0.2 (0 - 1.1)

Condition	Summary	Five Year Count	Prevalence per 10,000 Live Births and 95% Confidence Interval
Small intestinal atresia/stenosis	One in 29,295 live births	2	0.3 (0 - 1.1)
Spina bifida without anencephalus	One in 6,510 live births	9	1.5 (0.7 - 2.8)
Tetralogy of Fallot	One in 3,084 live births	19	3.2 (1.9 - 5)
Total anomalous pulmonary venous connection	-----	0	-----
Transposition of the great arteries	One in 6,510 live births	9	1.5 (0.7 - 2.8)
Tricuspid valve atresia	-----	0	-----
Tricuspid valve atresia and stenosis	-----	0	-----
Trisomy 13	One in 14,647 live births	4	0.7 (0.2 - 1.8)
Trisomy 18	One in 7,324 live births	8	1.4 (0.6 - 2.8)
Trisomy 21 (Down syndrome)	One in 945 live births	62	10.6 (8.1 - 13.6)
Turner syndrome*	One in 7,214 live births	4	1.4 (0.4 - 3.6)
Ventricular septal defect	One in 742 live births	79	13.5 (10.7 - 16.8)

Birth Data: Total live births include babies born to Maine residents in Maine.	
Total Live Births 2016-2020	*58,589*
Total Male Live Births 2016-2020	*29,727*
Total Female Live Births 2016-2020	*28,854*

Notes:
Count and rates are based on five years of available data for each condition during the reporting period 2016-2020.
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 and unknown gender live births.
Data Sources:
Birth Defects: Maine Birth Defects Registry, extract April 2023.
Births: Maine Data, Research and Vital Statistics (DRVS)

General comments: Case-finding is limited to babies born in Maine to Maine residents and to birth defects identified ir the first year of life and fetal deaths. A baby can be born with multiple conditions, adding up the number of defects will not yield the number of babies born with defects. In addition, this list represents only a portion of reportable birth defects collected and tracked in Maine.

National birth defects data for previous years can be found at the following link: Birth Defects Data Tables and Directory, Jan. 2022 - National Birth Defects Prevention Network (nbdpn.org)