

2020 Annual Report



Maine CDC Birth Defects Program

January 1, 2020 – December 31, 2020

Submitted to Joint Standing Committee on Health and Human Services



Acknowledgements

We would like to thank everyone from Children with Special Health Needs (CSHN) staff, USM Epidemiologist and Vital Records for birth and death files, linkages, and support for reporting. We thank all of Maine's reporting facilities for their time and efforts to provide the case reports that are essential to the success of this registry. We thank the members of the Advisory Committee for the partnership, advice and direction guiding and sustaining the program.

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EXECUTIVE SUMMARY

Background

In May 1999, the Department of Health and Human Services was authorized to collect information on birth defects in Maine by statute (Title 22: Health and Welfare: Subtitle: Facilities for Children and Adults; Chapter 1687; Birth Defects Program; §8941-§13375). The Maine CDC Birth Defects Program was established within the Department of Health and Human Services, Maine Center for Disease Control and Prevention (Maine CDC).

Purpose

The Maine Birth Defects Program focuses on the three public health core functions of assessment, assurance, and policy development in conjunction with the requirements set out in statute: provide

an up-to-date birth defects registry that facilitates the identification of risk factors, assures epidemiology, protects confidentiality, determines reportable birth defects through an advisory committee, provides for primary prevention to decrease occurrence, maintains components to educate populations about birth defects and systems, and refers those with birth defects to early intervention and other support services.

Activities

This 2020 annual report summarizes the current activities of the Maine CDC Birth Defects Program (BDP), the ongoing and upcoming activities for calendar year 2020 and shows selected birth defects counts and birth prevalence for the years 2013 - 2017. In 2020, the Maine CDC BDP:

- Completed and submitted the Annual Report of selected birth defects to the U.S. Centers for Disease Control and Prevention. The Annual Report to the National Birth Defects Prevention Network will not be required until 2021.
- Continued infrastructure building, including new data base, abstractor services and processes, and advisory group to increase efficiency and accuracy.
- Worked with epidemiologists to refine data and assure quality.
- Continued to make referrals for babies with a confirmed birth defect to the Maine Department of Education/Child Development Services (CDS) – Individuals with Disabilities Education Act (IDEA) Part C Agency.
- Met with the Maine CDC Environmental Public Health Tracking Unit to discuss updates to the Environmental Tracking Portal related to birth defects.
- Provided education to contracted abstractors to develop skills on new data portal.
- Provided educational materials about the prevention of birth defects to parents, health providers, and other interested parties.



- Participated in the virtual annual National Birth Defects Prevention Network Meeting in March 2020.
- Participated in Cleft Lip and Palate Conference at Maine Medical Center.
- Continued to work with the birth hospitals to support them sending critical congenital heart defects data electronically.
- Met with Cleft Lip and Palate Clinic Coordinators monthly to stay current with referrals and other information, as well as offer support during Covid 19 pandemic.
- Met with Maine Perinatal Nurse Managers to discuss Maine Birth Defects Program.

Future Direction

- Continue to update Birth Defects Program Manual to reflect current scope of responsibilities.
- Expand, enhance and strengthen Advisory Committee.
- Increase collaboration with hospitals and community to expand reach of Birth Defects Program.
- Continue to develop Maine Newborn Screening Portal.
- Continue to work with hospitals around screening and reporting Critical Congenital Heart Disease.
- Continue to work with epidemiologist to refine data and assure quality.
- Increase collaboration with internal and external partners.

For more information on the Maine CDC Birth Defects Program:

Contact Anna Cyr, Maternal and Child Health Coordinator, anna.cyr@maine.gov or 207-287-8424
<http://www.maine.gov/dhhs/mecdc/population-health/mch/cshn/birth-defects/index.html>

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% 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 birth defects, it is essential to know what types of birth defects are occurring. A population-based birth defects surveillance program that uses multiple sources of data allows 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.

The Maine CDC Birth Defects Program (Maine CDC BDP) is committed to fulfilling its mission that all infants with birth defects are identified early and referred to an established network of services, in order to achieve optimal health and develop to their fullest potential. The Maine CDC BDP is a comprehensive surveillance program that benefits the residents of Maine through the early identification of infants who have birth defects. Early identification ensures timely and appropriate access to systems of care that are family-centered, culturally competent and community-based. It is the intention of the Maine CDC BDP to participate fully in epidemiological investigations as a means of informing public policy, to develop prevention strategies in order to reduce birth defects and to assess for timely referrals and follow-up care to reduce mortality and morbidity among children identified with birth defects.

Legislation and Rules

Legislation supporting the Maine CDC BDP has and continues to define the purpose of the program. The statutes defining these roles and responsibilities are listed below.

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/>

May 1999 - Public Law (P.L.) 1322, 22 M.R.S.A. c. 1687, established the Maine CDC BDP within the Maine Department of Health and Human Services. Program rules were formally adopted April 2003 outlining reporting responsibilities and access to medical records. Mandated reporting began May 2003.

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, Child Developmental Services (CDS) 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.

Stakeholder Engagement

The following is a brief listing of organizations that have a strong association with the Maine CDC Birth Defects Program. The March of Dimes and the National Birth Defects Prevention Network are also key national-level partners.

<ul style="list-style-type: none"> • Consumers <ul style="list-style-type: none"> ○ Parents and families 	<ul style="list-style-type: none"> • Early Intervention Agencies <ul style="list-style-type: none"> ○ Department of Education – Child Development Services
<ul style="list-style-type: none"> • External groups involved with screening, follow-up and diagnosis <ul style="list-style-type: none"> ○ Hospitals and their staff ○ Nurses ○ Primary care providers ○ Specialist physicians ○ Genetic counselors 	<ul style="list-style-type: none"> • Other DHHS Programs <ul style="list-style-type: none"> ○ Maine CDC Newborn Hearing Screening Program ○ Maine CDC Newborn Bloodspot Screening Program ○ Maine Public Health Nursing ○ Maine CDC Environmental Public Health Tracking ○ Maine CDC Data, Research and Vital Statistics ○ Office of MaineCare Services ○ Pregnancy Risk Assessment Monitoring System

Maine CDC Birth Defects Advisory Committee

The Advisory Committee meets at least one time annually and more frequently, if needed. The Committee consists of interested parties including health professionals, outside agencies, and other state, community and private sector agencies, as well as the Maine CDC Children with Special Health Needs staff. The group provides consultation to the Maine CDC BDP on development, implementation, and evaluation of program policies, procedures, and activities.

Maine CDC Birth Defects Program Summary

Program Description

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

As a surveillance unit, the Maine CDC Birth Defects 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, 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 visits the hospital to review records to confirm the presence of a birth defect.

The Maine CDC Birth Defects Program is aligned with the most current listing of reportable birth defects to reflect the birth defects surveillance guidelines developed by the CDC National Birth Defects Prevention Network. The current listing of reportable birth defects may be found in Appendix A.

The Maine CDC BDP receives medical discharge data electronically from all birthing hospitals currently. Maine CDC BDP sends all hospitals a monthly electronic reminder to submit data.

For a case to be considered by the Maine Birth Defects Program, 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 in the Maine CDC BDP 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 reviewed and entered in the birth defects surveillance and tracking system. The system houses data obtained from hospitals, health care providers, and others mandated to report birth defects.

Currently, the Maine CDC Birth Defects Program reports on and gathers information on 57 birth defects (see Appendix A for a complete listing 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

The Maine CDC Birth Defects Program consists of 1 FTE coordinator and .12 FTE Epidemiologist. The Maine CDC BDP currently contracts with partners to provide data registry abstraction services, as well as, to facilitate the Cleft Lip and Palate clinical services.

The Birth Defects program and contracted services are funded by state and federal funds allocated to promoting and improving the health and well-being of the Maine's mothers, children, including children with special needs, and their families.

Goals, Activities and Achievements

The Maine CDC Birth Defects Program gathers data about infants born each year with certain birth defects diagnosed within the first year of life. The statute requires that the Maine Birth Defects Program:

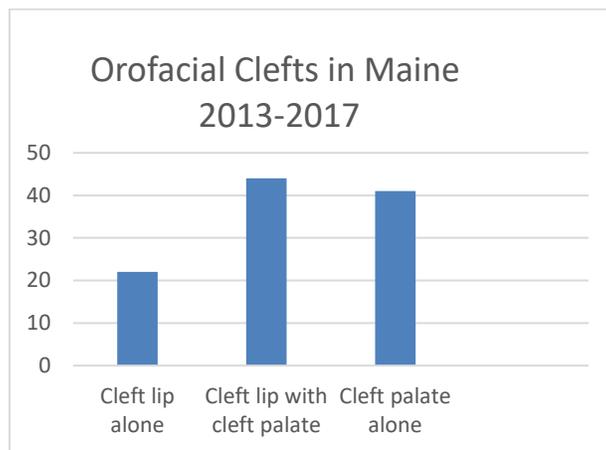
- Collect, analyze, and distribute information to identify the birth defects with regards to the following: causes, risk factors, and strategies for prevention and the provision of services,
- Establish a system for data collection that identifies prevalence and incidence rates by region and population 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 45 birth defects. The Maine CDC BDP submits data annually to the National Birth Defects Prevention Network and U.S. CDC.
 - Collaboration with the Maine CDC Environmental Public Health Tracking Unit to show 12 of the 45 birth defects on the Environmental Health Tracking Portal.
 - The baby identified with a confirmed birth defect is referred to the Department of Education Part C Agency (CDS) for follow-up for early intervention services.

Cleft Lip and Palate

The Maine Birth Defects program currently supports and collaborates with Cleft Lip and Palate clinics at MaineHealth Maine Medical Center and Northern Light Eastern Maine Medical Center. 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 clinics. Families may attend clinic until the child is 22 years of age regardless of income. Families attending clinics 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 locations. This year, each site began working toward becoming a team approved by the American Cleft Palate-Craniofacial Association. Clinic sites also provide a full range of education and consultation services upon request in addition to monthly clinical services. Additionally, clinical sites are working to provide family and community educational opportunities on an annual basis. In 2020, 23 clinical sessions were offered serving over 100 families, including telehealth clinics to support families during the coronavirus pandemic.



LEFT LIP AND CLEFT PALATE ARE BIRTH DEFECTS THAT OCCUR WHEN A BABY'S LIP OR MOUTH DO NOT FORM PROPERLY DURING PREGNANCY. TOGETHER, THESE BIRTH DEFECTS COMMONLY ARE CALLED "OROFACIAL CLEFTS".



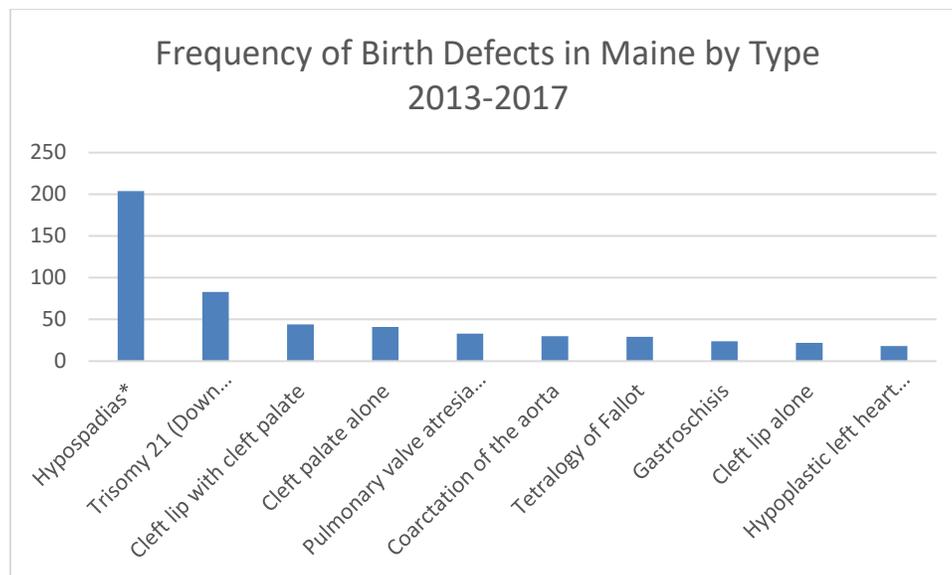
Maine CDC Birth Defects Tracking System

Tracking System

Maine CDC BDP began collaborating 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, the Birth Defects Program 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

The Maine CDC Birth Defects Program submits data annually for a specific span of years to the U.S. Centers for Disease Control and Prevention, National Birth Defects Prevention Network. The most recent data submitted to the U.S. CDC was 2013-2017, which will be available for publication in December 2020. The 24-month lag time in U.S. CDC data is based on the definition of a birth defect. 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 that 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.



Appendices

Appendix A

Reportable Birth Defects Included in Case Definition for 2019

Birth Defect	ICD-10-CM Codes
Central Nervous System	
Anencephalus	Q00.0-Q00.1
Spina Bifida without anencephalus	Q05.0-Q05.9 Q07.01, Q07.03 w/o Q00.0-Q00.1
Hydrocephalus without Spina Bifida	Q03-Q03.9
Encephalocele	Q01-Q01.9
Microcephalus	Q02
Holoprosencephaly	Q04.2
Eye	
Anophthalmia/microphthalmia	Q11.0-Q11.2
Congenital cataract	Q12.0
Aniridia	Q13.1
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
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
Patent ductus arteriosus	Q25.0
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
Pyloric stenosis	Q40.0
Hirschsprung's disease (congenital megacolon)	Q43.1
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
Obstructive genitourinary defect	Q62 – 62.39, Q64.2
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
Congenital hip dislocation	Q65 – Q65.5
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
Other	
Fetal alcohol syndrome	Q86.0
Amniotic bands	No code

Appendix B Birth Defects Counts and Prevalence Rates (2013-2017), Maine

Condition	Summary	Five Year Count	Prevalence per 10,000 Live Births and 95% Confidence Interval
Anencephalus	One in 3,711 live births	17	2.7 (1.6-4.3)
Anophthalmia/microphthalmia	One in 21,028 live births	3	0.5 (0.1-1.4)
Anotia/microtia	One in 5,257 live births	12	1.9 (1.0-3.3)
Aortic valve stenosis	One in 9,012 live births	7	1.1 (0.4-2.3)
Atrial septal defect	One in 387 live births	163	25.8 (22.0-30.1)
Atrioventricular septal defect (Endocardial cushion defect)	One in 3,004 live births	21	3.3 (2.1-5.1)
Biliary atresia	-----	0	0.0 (0.0-0.0)
Bladder exstrophy	One in 21,028 live births	3	0.5 (0.1-1.4)
Cleft lip alone	One in 2,867 live births	22	3.5 (2.2-5.3)
Cleft lip with cleft palate	One in 1,434 live births	44	7.0 (5.1-9.4)
Cleft palate alone	One in 1,539 live births	41	6.5 (4.7-8.8)
Coarctation of the aorta	One in 2,109 live births	30	4.8 (3.2-6.8)
Common truncus (truncus arteriosus)	---	0	---
Congenital cataract	One in 15,771 live births	4	0.6 (0.2-1.6)
Diaphragmatic hernia	One in 21,028 live births	3	0.5 (0.1-1.4)
Ebstein anomaly	One in 63,084 live births	1	0.2 (0.0-0.9)
Encephalocele	One in 7,886 live births	8	1.3 (0.5-2.5)
Esophageal atresia/tracheoesophageal fistula	One in 3,711 live births	17	2.7 (1.6-4.3)
Gastroschisis	One in 2,629 live births	24	3.8 (2.4-5.7)
Hypoplastic left heart syndrome	One in 3,505 live births	18	2.9 (1.7-4.5)
Hypospadias*	One in 159 live births	204	63.0 (54.6-72.2)
Limb deficiencies (reduction defects)	One in 4,506 live births	14	2.2 (1.2-3.7)
Omphalocele	One in 5,257 live births	12	1.9 (1.0-3.3)
Pulmonary valve atresia	One in 9,012 live births	7	0.0 (0.0-0.0)
Pulmonary valve atresia and stenosis	One in 1,912 live births	33	5.2 (3.6-7.3)
Rectal and large intestinal atresia/stenosis	One in 2,103 live births	30	4.8 (3.2-6.8)
Renal agenesis/hypoplasia	One in 1,502 live births	42	6.7 (4.8-9.0)
Spina bifida without anencephalus	One in 2,426 live births	26	4.1 (2.7-6.0)
Tetralogy of Fallot	One in 2,175 live births	29	4.6 (3.1-6.6)
continued			

Birth Defects Counts and Prevalence Rates (2013-2017), Maine

Condition	Summary	Five Year Count	Prevalence per 10,000 Live Births and 95% Confidence Interval
Transposition of the great arteries (TGA)	One in 3,943 live births	16	2.5 (1.4-4.1)
Tricuspid valve atresia and stenosis	One in 21,028 live births	3	0.5 (0.1-1.4)
Trisomy 13	One in 15,771 live births	4	0.6 (0.2-1.6)
Trisomy 18	One in 3,711 live births	17	2.7 (1.6-4.3)
Trisomy 21 (Down syndrome)	One in 760 live births	83	13.2 (10.5-16.3)
Ventricular septal defect	One in 535 live births	118	18.7 (15.5-22.4)

Birth Data

Total Live Births 2013-2017

63,084

Total Male Live Births 2013-2017

32,403

Notes

Count and rates are based on five years of available data for each condition during the reporting period 2013-2017.

*Hypospadias: prevalence per 10,000 male live births; total male live births are provided for Hypospadias rates.

Total live births and total male live births include babies born to Maine residents in and out of state.

Data Sources:

Birth Defects: Maine Birth Defects Registry, extract August 2019.

Births: United States Department of Health and Human Services (US DHHS), Centers for Disease Control and Prevention (CDC), National Center for Health Statistics (NCHS), Division of Vital Statistics, Natality public-use data 2007-2017, CDC WONDER Online Database August 2020. Accessed at <http://wonder.cdc.gov/natality-current.html>. Due to birth datafile limitations, total births include babies born to Maine residents in and out of state.

General comments: Case-finding is limited to babies born in Maine to Maine residents and to birth defects identified in 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:

http://www.nbdpn.org/annual_reports.php



**For more information contact
Maine Birth Defects Program**

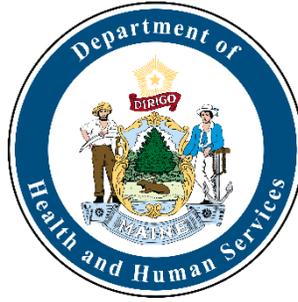
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