Maine Birth Defects Program

2010 Annual Report

Department of Health and Human Services
Maine Center for Disease Control and Prevention
Division of Family Health
Children with Special Health Needs

Summary

Effective September,1999, a Maine law went into effect (22 MRS §§8941-§8945) and authorized the Department of Health and Human Services to collect information on birth defects in Maine. The Maine Birth Defects Program was established within the Department of Health and Human Services, Maine Center for Disease Control and Prevention, Division of Family Health, Children with Special Health Needs Program. The Maine Birth Defects Program (MBDP) gathers data about infants born each year with certain birth defects diagnosed within the first year of life. The statute requires that the "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,

◊ Contact families to provide information about available resources and services,

◊ Conduct investigations to determine the nature and extent of the disease or known or suspected causes of birth defects.

MBDP is committed to fulfilling our mission that all infants with confirmed 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 primary goals of the Maine Birth Defects Program are to:

- Monitor incidence trends of birth defects to detect emerging health concerns and identify affected populations
  - ◊ Develop timely baseline birth defects rates
  - ◊ Monitor trends and relationships to environmental factors
- Ensure appropriate services are provided to affected families
  - ◊ Refer children to services and resources
  - ◊ Evaluate services utilization
- Prevent birth defects through targeted education
  - ◊ Provide data for services planning
  - ◊ Provide basis for prevention strategies
  - ◊ Evaluate efficacy of preventive strategies

- Educate health care providers and the public regarding birth defects
    - ◊ Inform the public about public health importance
    - ◊ Inform parents about resources and services
    - ◊ Provide data for studies

The statute did not include financial support for the activities of the Maine Birth Defects Program; therefore, MBDP is funded by the Maternal and Child Health Block Grant.

Introduction

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 quantify morbidity and mortality accurately, detect temporal trends, and assess the financial burden that birth defects may cause. Many children with birth defects who survive have a lifetime of major expenses. In addition, specialty medical care, special education, rehabilitation and developmental services are essential.

The Maine Birth Defects Program (MBDP) 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 MBDP is a comprehensive surveillance program that benefits the citizens 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 MBDP to participate fully in epidemiological investigations as a means of informing public policy, to develop prevention strategies in order to

[1] Trust for America's Health "Birth defects and Developmental Disabilities: A Major Public Health Challenge"
[2] http://www.cdc.gov/node.do/id0900f38000dffe

reduce birth defects, and to assess for timely referrals and follow-up care to reduce mortality and morbidity among children identified with birth defects.

Maine Birth Defects Program: A Progress Report

The Maine Birth Defects Program began passive case ascertainment on May 1, 2003. Passive case ascertainment is an approach whereby the surveillance program receives case reports of birth defects from a variety of data sources. As required by statute those entities licensed as hospitals under Title 22 and professionals under 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, birth and death certificates and medical discharge records using ICD-9 codes.

The Maine Birth Defects Program recently updated the 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 MBDP receives medical discharge data electronically from 27 of the 30 birth hospitals. MBDP is currently working with the 3 remaining hospitals to submit data electronically within the first six months of 2011. MBDP sends each hospital a monthly reminder to submit data.

In order for a case to be considered by the MBDP it must meet the following criteria:

- ◊ Infant was born alive, stillborn or prenatally diagnosed, with a gestational age of greater than 20 weeks,
- ◊ Fetuses less than 20 weeks gestation but with a prenatal diagnosis,
- ◊ The birth must occur in Maine and the mother must be a Maine resident,
- ◊ The diagnosis was made before the infant reached 1 year of age, and
- ◊ The birth defect is included in the MBDP list of reportable birth defects.

Potential cases are identified through weekly downloads of both the electronic birth and infant death certificates and medical record 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 and alcohol use.

The data collected by the abstractors is reviewed and entered in the birth defects surveillance and tracking system, called ChildLINK. ChildLINK was built to link existing state information systems with data obtained from hospitals, health care providers and others mandated to report. Once the birth defect is confirmed the family is notified by letter of the services available to them and how the Children with Special Health Needs Program can assist in locating those services.

Reportable Birth Defects

As of December 31, 2010, 679 reportable birth defects were confirmed; this includes children born in 2003 – 2007. Below is a table that reflects this information.

Selected birth defects counts and birth prevalence, Maine, 2003-2007

Birth Defect	5-year count	Average annual count	Average annual birth prevalence*
Cardiovascular			
Coarctation of aorta	27	5.4	4.0
Double outlet right ventricle	9	1.8	1.3
Hypoplastic left heart syndrome	20	4.0	2.9
Interrupted aortic arch	3	0.6	0.4
Pulmonary atresia with intact ventricular septum	2	0.4	0.3
Single ventricle	1	0.2	0.1
Tetralogy of Fallot	23	4.6	3.4
Transposition of great vessels	26	5.2	3.8
Tricuspid atresia	1	0.2	0.1
Truncus arteriosus	3	0.6	0.4
Central nervous system			
Anencephalus	5	1.0	0.7
Encephalocele	4	0.8	0.6
Spina bifida without anencephalus	19	3.8	2.8
Chromosomal			
Down syndrome	70	14.0	10.2
Musculoskeletal			
Gastroschisis	34	6.8	5.0
Omphalocele	15	3.0	2.2
Reduction deformity, lower limb	NA	NA	NA
Reduction deformity, upper limb	NA	NA	NA
Orofacial			
Cleft lip with and without cleft palate	62	12.4	9.1
Cleft palate without cleft lip	58	11.6	8.5
Genitourinary			
Hypospadias	NA	NA	NA
* Per 10,000 occurrent Maine births to Maine residents NA: Not available; surveillance for this condition began with 2008 births			
Notes: 1. Counts and prevalence are based on live births (excludes stillbirths and fetal deaths). 2. Table is based on ChildLINK data as of 3/22/2010.			

Currently, the Birth Defects Program reports on and gathers further information on 22 birth defects. These cases are confirmed usually within the first three months after birth, but this time frame can be longer depending on when the birth defect is reported to the Birth Defects Program and the ability of the abstractors to get the necessary information from the birth hospital. There is also a lag time in data of about one and one-half years because a reportable birth defect can be diagnosed within a

year of birth. Therefore, the program must wait at least one year to make sure a diagnosis is made. Data can become available to the Birth Defects Program at any time if the diagnosis is made within the year.

Accomplished Activities for CY2010 (January 1 – December 31, 2010)

1. The Maine Birth Defects Program has worked with the birth hospitals in Maine to help them send in their discharge data electronically. There are three birth hospitals left that need assistance to transition to electronic reporting.

2. The Maine Birth Defects Program began to contact families with a baby with a confirmed birth defect by letter to offer support, information regarding birth defects and available services.

3. The Maine Birth Defects Program continued to provide ongoing educational materials regarding the prevention of birth defects to parents, health providers and other interested parties.

4. The Maine Birth Defects Program worked with Environmental Public Health Tracking Unit (EPHTU) to present data on 12 birth defects that it is required to report on through their CDC Federal Grant.

https://tracking.publichealth.maine.gov/ephtnApp.portal;jsessionid=NkhbmWfrKM3Vv0KJTnJqJHjQk1lv10TGSss0C62XsVms7WWlv7p0!-849874375!-478816134?_nfpb=true&portlet_14_3_actionOverride=/ephtn/PreProcessQuickFacts&_windowLabel=portlet_14_3&portlet_14_3content_area=BIRTH_DEFECTS

5. The Maine Birth Defects Program continued to work collaboratively with the other New England states through the New England Birth Defects Consortium on issues pertinent to the region including data sharing and prevention activities as well as participating in the annual meeting.

6. The program coordinator met quarterly with the two abstractors who work for the Maine Birth Defects Program to continue to develop skills and share information pertinent to birth defects and abstraction.

Planned Activities for CY2011 (January 1 – December 31, 2011)

1. The Maine Birth Defects Program will continue to work collaboratively with the other New England states in providing prevention activities. Currently the Maine Birth Defects Program is working with WIC in the distribution of multivitamins, as are the other New England states' birth defects programs.

2. The Maine Birth Defects Program will work with New Hampshire's Birth Conditions Program collaboratively in a research project looking at arsenic in relationship to birth defects in the two

states. Maine is one of three New England states (in addition to New Hampshire and Massachusetts) that has naturally occurring arsenic in ground water.

3. The Maine Birth Defects Program will work with the remaining 3 birth hospitals to have them send in the discharge data electronically.

4. The Maine Birth Defects Program will finish updating the Birth Defects Rules to include the 45 birth defects recommended by the National Birth Defects Prevention Network and CDC as part of the reportable birth defects list in Maine. Currently MBDP reports on 22 defects.

5. The Maine Birth Defects Program will continue to provide ongoing educational materials regarding the prevention of birth defects to parents, health providers and other interested parties.

6. The Maine Birth Defects Program will begin follow-up phone calls with families that were contacted by letter once their children were found to have confirmed reportable birth defects to determine if their children's needs are being met.

7. The Maine Birth Defects Program will explore with available hospitals the possibility of accessing birth records remotely through internet access to do abstraction work.

8. The Maine Birth Defects Program will continue to work with Environmental Public Health Tracking Unit to provide the necessary data.

9. The Maine Birth Defects Program will continue to participate in the National Birth Defects Prevention Network by attending the annual meeting as well as participating in the New England Birth Defects Consortium.

10. The Maine Birth Defects Coordinator will continue regular meetings with the abstractors to increase their skill level and to share ongoing information about birth defects.

11. The Maine Birth Defects Program will design a web page to present information on all MBDP reportable birth defects.

12. The Maine Birth Defects Program will continue to refer babies with confirmed birth defects to the Department of Education, Child Development Services (Part C).

Appendix A: Listing of Current Reportable Birth Defects

Cardiovascular

- ◊ **Coarctation of the Aorta -** Narrowing of the descending aorta, which may obstruct blood flow from the heart to the rest of the body.
- ◊ **Double Outlet Right Ventricle -** Double outlet right ventricle (DORV) is a congenital heart disease in which the aorta rises from the right ventricle (the chamber of the heart that pumps blood to the lungs), instead of from the left ventricle (the normal pumping chamber to the body).
- ◊ **Hypoplastic Left Heart Syndrome (HLHS) -** A condition in which the structures on the left side of the heart and the aorta are extremely small. Classically, this condition includes hypoplasia of the left ventricle, atresia or severe hypoplasia of the mitral and aortic valves, and hypoplasia and coarctation of the aorta.
- ◊ **Interrupted Aortic Arch –** Interrupted aortic arch (IAA) is a relatively rare genetic disorder that usually occurs in association with a nonrestrictive ventricular septal defect (VSD) and ductus arteriosus or, less commonly, with a large aortopulmonary window or truncus arteriosus
- ◊ **Pulmonary Valve Atresia -** Pulmonary valve atresia – Lack of patency, or failure of formation altogether, of the pulmonary valve, resulting in obstruction of blood flow from the right ventricle to the pulmonary artery.
- ◊ **Single Ventricle –** Single ventricle defects is an umbrella term used to describe several very different complex congenital (present at birth) heart defects that share the same problem: the heart has only one functional ventricle.
- ◊ **Tetralogy of Fallot -** The simultaneous presence of a ventricular septal defect (VSD), pulmonic stenosis, a malpositioned aorta that overrides the ventricular septum, and right ventricular hypertrophy
- ◊ **Transposition of the Great Arteries (TGA) -** Transposition of the aorta and the pulmonary artery such that the aorta arises from the right ventricle (instead of the left) and the pulmonary artery arises from the left ventricle (instead of the right).
- ◊ **Tricuspid Valve Atresia and Stenosis -** Tricuspid valve atresia – lack of patency, or failure of formation altogether, of the tricuspid valve, resulting in obstruction of blood flow from the right atrium to the right ventricle.
- ◊ **Common Truncus (Truncus Arteriosus or TA) -** Failure of separation of the aorta and the pulmonary artery, resulting in a single common arterial trunk carrying blood from the heart to both the body and lungs.

◊ **Tricuspid Valve Stenosis** – Obstruction or narrowing of the tricuspid valve, which may impair blood flow from the right atrium to the right ventricle.

Central Nervous System

◊ **Anencephalus** - Partial or complete absence of the brain and skull.

◊ **Encephalocele -** Herniation of brain tissue and/or meninges through a defect in the skull. The hernia sac is usually covered by skin.

◊ **Spina Bifida without Anencephalus** - Incomplete closure of the vertebral spine (usually posteriorly) through which spinal cord tissue and/or the membranes covering the spine (meninges) herniate.

Chromosomal

◊ **Down Syndrome (Trisomy 21) -** The presence of three copies of all or a large part of chromosome 21.

Musculoskeletal

◊ **Gastroschisis -** A congenital opening or fissure in the anterior abdominal wall lateral to the umbilicus through which the small intestine, part of the large intestine, and occasionally the liver and spleen, may herniate.

◊ **Omphalocele -** A defect in the anterior abdominal wall in which the umbilical ring is widened, allowing herniation of abdominal organs, including the small intestine, part of the large intestine, and occasionally the liver and spleen, into the umbilical cord. The herniating organs are covered by a nearly transparent membranous sac.

◊ **Reduction Deformity, Upper Limbs -** Complete or partial absence of the upper arm (humerus), lower arm (radius and/or ulna), wrist (carpals), hand (metacarpals), or fingers (phalanges).

◊ **Reduction Deformity, Lower Limbs -** Complete or partial absence of the upper leg (femur), lower leg (tibia and/or fibula), ankle (tarsals), foot (metatarsals), or toes (phalanges).

Orofacial

◊ **Cleft Palate without Cleft Lip -** An opening in the roof of the mouth resulting from incomplete fusion of the shelves of the palate. The opening may involve the hard palate only, the soft palate only, or both.

◊ **Cleft Lip with and without Cleft Palate -** A defect in the upper lip resulting from incomplete fusion of the parts of the lip.

Genitourinary

- ◊ **Hypospadias -** Hypospadias – Displacement of the opening of the urethra (urethral meatus) ventrally and proximally (underneath and closer to the body) in relation to the tip of the glans of the penis.