Maine
Birth Defects Program
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by:
tment of Human Services
Bureau of Health
Division of Community & Family Health

Maine Birth Defects Program
Report to the Legislature-1999

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Background

The 119th Maine State Legislature enacted a law to establish the Maine Birth Defects Program within the Bureau of Health (H.P. 1322-L.D. 1905)(22 MRSA c. 1687). The purpose of this program is to identify and investigate birth defects in children. The program shall maintain a central registry of cases of birth defects, collect, analyze and distribute information and undertake necessary research to identify the following with regard to birth defects: cases, risk factors and strategies for prevention and the provision of services. The law requires the Department to report on activities and findings of the Maine Birth Defects Program to the Health & Human Services Committee of the Maine State Legislature.

The Bureau of Health received a cooperative agreement with the Centers for Disease Control and Prevention to develop and implement a state-based birth defects surveillance system and use the surveillance data to guide prevention and intervention programs. Maine was awarded $100,000 for each of three years February 1, 1999 to January 31, 2002.

This report outlines activities of year one of the program.

For more information on activities of the Maine Birth Defects Program, contact the Genetics Program at (207)287-5357 or 1-800-698-3624.

Problem Statement

Birth defects are the leading cause of infant mortality in the United States, accounting for one out of five infant deaths. It is estimated that between 3-5% of all live births are complicated by congenital anomalies.[i] Further, of the 100,000 to 150,000 infants born in the U.S. annually with a major birth defect, approximately 6000 die during the first 28 days of life and another 2,000 die before their first birthday.[ii] Though Maine has some information about the prevalence of certain birth defects in the state, the overall prevalence and types of defects are difficult to ascertain because we have not had a population-based surveillance system or registry. The data that is currently available through the Bureau of Health's Office of Data, Research and Vital Statistics (ODRVS) is based solely on vital records. It indicates the following:

- For the five year period, 1992-1996, 2% of live births were affected by congenital anomalies, which is approximately 250 babies per year. It is estimated that 400 -750 infants are identified with a serious birth defect by age one, each year;[iii] and
- For the five year period, 1992 - 1996, 24% of infant deaths were attributable to congenital anomalies and chromosomal abnormalities.[iv]

This data is derived from birth certificates, which alone, are not an adequate source of information. Birth certificates do not contain complete or accurate data and many conditions are not apparent at the time the birth certificate is completed.[v]

Though much is still to be known about the etiology and prevention of birth defects, it is known that folic acid consumption prior to conception can reduce the incidence of spina bifida (open spine) and other neural tube defects (defects of the brain and spina) (NTDs). The U. S. Public Health Service estimates indicate that up to 50% of all NTDs could be prevented if women of childbearing age consumed 0.4 mg of folic acid per day. [vi] Women who have had a pregnancy affected by a neural tube defect can reduce their risk of recurrence by up to 70% with increased consumption of folic acid preconception.[vii] Other studies suggest that folic acid may help avert other birth defects, such as cleft lip and palate and congenital heart defects, prematurity and low birth weight.[viii]

Without an ongoing population-based birth defects surveillance program that uses multiple sources of data, the full impact of birth defects on Maine children and their families is unknown. With Maine's birth rate of less than 14,000 live births per year, it will take several years of surveillance to obtain meaningful information. Without this data, it is not possible to accurately quantify morbidity and mortality or detect temporal trends. The need for prevention activities and services also cannot be thoroughly evaluated, or education and programs be appropriately targeted. In addition, clusters of specific birth defects may go undetected because surveillance and research efforts are underdeveloped, incomplete or not timely.

In November 1999, the Pew Environmental Health Commission released a report analyzing birth defect information from across the United States. They found that there are increases in certain birth defects that cannot be fully explained through existing data. Data is difficult to compare across states as surveillance systems vary in their methods, defects which are included, reporting and data sources and many states have no

surveillance systems. Maine is one of twenty states that either have no system or are just beginning to plan to implement a birth defects surveillance system. Each state was evaluated on seven elements of a system. Maine received a grade of "B" even though the program is not yet implemented. The report, "Healthy from the Start" and related reports can be viewed and downloaded from
http://pewenvirohealth.jhsph.edu\html\reports\menu.html>

Results or Benefits Expected

Through the activities of the Maine Birth Defects Program, we expect a significant increase in our knowledge of birth defects prevalence in Maine and our capability to serve affected families. With the implementation of a statewide monitoring system we will be able to quantify morbidity and mortality due to birth defects and to correlate our state data with national rates and trends. The registry will enhance our existing capability to identify all birth defect cases, including those diagnosed prenatally, throughout the state. With this information, we will be able to provide appropriate referrals and counseling to affected families, so that infants receive the care that they need and recurrent cases can be prevented. In addition, the data and information that is collected and analyzed through the Maine Birth Defects Program will subsequently influence program direction and collaboration among several Bureau of Health initiatives, including the Genetics Program, the Children with Special Health Needs Program, Public Health Nursing and the Women and Children's Preventive Health Services Program. An example of such an initiative would be implementing and evaluating prevention programs.

Activities and plans:

Workgroups provide input into program planning

There were three workgroups organized to provide input for program development for the Birth Defects Program.

Data workgroup:

This workgroup reviewed a list of recommended data items, as published in Public Health Surveillance, edited by Halperin and Baker,1992. Recommendations were made on which items would be included on the case reporting form and the medical record abstraction form. The group discussed examples of such forms from other state surveillance programs and made recommendations for the development of a form for Maine. The group briefly discussed the importance of including data from prenatally diagnosed cases, irrespective of outcome. Other data sources were briefly discussed. (Participants list is in Appendix A.)

Plan: Based on discussion, a revised list of data items will be developed which will include information on sources of such data. An abstraction form will be developed. The workgroup will review these and further discuss data sources, timeliness and accuracy and process for requesting data.

MBDP will use data from the hospital case reports, specialists and Genetics Centers as primary sources for case ascertainment. Other specialty clinics, birth certificates, other vital record data and hospital discharge data will be used as secondary sources. A medical record abstractor will review medical records of the mother and infant to collect clinical data, validate reported diagnosis and submit data to be entered into a database. Evaluation of timeliness, and accuracy will be an ongoing process.

Reporting workgroup:

This workgroup reviewed the legislation, cooperative agreement abstract, draft case reporting form, selected birth defects to be included, and proposed reporting requirement. Comments and recommendations were made regarding the case reporting form. A suggestion was made to consider including skeletal dysplasias and other chromosome anomalies. The group discussed religious objection and prenatally diagnosed cases. (Participants list is included in Appendix A.)

Plan: Several people were unable to attend this meeting. They will be contacted individually for input and another meeting may be scheduled. Input has been provided by perinatal nurse managers, other nurses, Dr. Hourihan, pediatric cardiologist, a medical records director and consumers.
Parental objection based on sincerely held religious beliefs is allowed under the law. This objection will be clearly stated in the medical record of mother and infant. No

information will be collected on these cases. Program evaluation will include the impact of such objections on population based surveillance.
Further consideration and discussion is needed on management of case reports on prenatally diagnosed cases. A recommendation to separate reporting of prenatal diagnosis from report of outcome of pregnancy has been made.

Resources workgroup:

This workgroup reviewed legislation and the cooperative agreement abstract. The case report form was discussed and suggestions were made. Timing of folic acid counseling and genetic counseling were discussed with differing opinions voiced. Participants discussed available resource directories and determined that there is no central location for complete and up-to-date information on resources in Maine. The program will consider an agreement to develop such a directory. Each participant was asked to consider elements of a home visit and other information that would be helpful for families to assure access to resources and services. Another meeting will be scheduled. (Participants list is included in Appendix A.)

Plan: Consider agreement to develop statewide resource directory. Review suggestions for home visit and information for families. The nurse making the home visit will be responsible to review or to present information on folic acid intake to reduce the risk of recurrence as appropriate. A plan is underway to provide an in-service to public health and community health nurses to assure that accurate and consistent information is provided to families.

Advisory Committee Convened

An Advisory Committee was convened which includes representatives from various professional medical organizations, physician specialties, service and early intervention programs, public health nursing, other Bureau of Health programs and consumers. The first meeting of this committee, held December 13, 1999, was successful and provided valuable input into the program, particularly regarding case reporting. (Advisory Committee members are listed in Appendix B.)

Rules and Regulations Drafted

Rules and regulations are being drafted that will outline the responsibilities of reporters and the program. The draft rules were discussed in an Advisory Committee meeting on December 13, 1999. The final draft of the rules will be announced in several newspapers statewide in February and comments will be solicited before adoption. As proposed, hospitals and providers who diagnose birth defects in infants and fetuses will be required to report the occurrence of a reportable birth defect to the MBDP by discharge, transfer to another facility or within 7 days of a diagnosis. Reporting cannot be required before the rules have been adopted.

Birth Defects to be included

To be included in the registry, the birth defect must occur in a live born, stillborn or prenatally diagnosed infant born in Maine whose mother is a Maine resident, of at least 20 weeks gestation (or earlier if prenatally diagnosed) and diagnosed before one year of age.

The cases to be reported initially will include major birth defects which meet the following criteria: a major defect in structure or function which is generally identified during the first few days of life, requires surgery and contributes to infant morbidity and mortality. These defects include the major neural tube defects (defects of the brain and open spine): spina bifida, anencephaly, and encephalocele, also gastroschesis and omphalocele (defects of the abdominal wall), cleft lip and/or palate, trisomy 13, 18 and 21 (chromosome anomalies). Major heart defects are being discussed with a pediatric cardiologist who will recommend defects to be included which also meet the criteria. Several other defects have been suggested, such as, skeletal abnormalities and are still being considered. The list of defects to be included will be expanded at a later time.

Use of Data

The surveillance system will be used to monitor the prevalence of birth defects in the state, identify specific cases for referral and follow-up services. All families of infants with birth defects will be offered a home visit by a nurse who will assist the family in identifying and accessing needed services. The nurses will provide or reinforce accurate and sensitive information on risk reduction for recurrence of NTD through the preconceptional use of folic acid. The Children with Special Health Needs Program will offer assistance with care coordination.

Data will be used to produce periodic reports to providers and other interested parties. The aggregate data will be available for research initiatives conducted both in Maine and by national epidemiological studies. Requests for use of identifiable data will be considered pending approval by the Bureau of Health Institutional Review Board.

Surveillance data will be used to monitor changes in occurrence of NTD that may result from food fortification and folic acid awareness campaigns. It will be used to target educational efforts. Data will be used to determine effectiveness of referral network. It will be used in consideration of outreach clinics for genetics and birth defects evaluation, diagnosis and treatment services.

Staffing

The Genetics Program director also serves as the coordinator for the MBDP. Multiple priorities have proved challenging. Collaboration with consultants, workgroups and providers has contributed to program progress. Technical assistance will be sought to orient and/or train new personnel in birth defect surveillance activities.

Some work has been accomplished through consultation with other division staff such as administrative support and computer programming.

One position is pending approval:

- Medical Record Abstractor: A job description and qualifications have been developed for a medical record abstractor (Comprehensive Health Planner I). A vacant position from another program will be upgraded and assigned to the MBDP pending budgetary approval.

Contractual agreements are developed for consultation in the following areas:

- Epidemiologist
- Dysmorphology consultant

Appendix A

Maine Birth Defects Program
Workgroups

Data workgroup:

Participants:
Don Lemieux, Director, Office of Data Research and Vital Statistics
Alice Rohman, Office of Data Research and Vital Statistics
Nancy Sonnenfeld, Epidimiologist, Division of Community & Family Health
Glen Palomaki, Director of Biometry, Foundation for Blood Research

Reporting workgroup:

Participants:
Dr. John Salvato, American Academy of Pediatrics, invited
Dr. Blum, Academy of Family Practitioners, invited
Kathleen Stuchiner, Maine Hospital Association, invited
Gordon Smith, Maine Medical Association, invited
Rhonda Spiro, MD, Foundation for Blood Research, attended
Laurent Beauregard, PhD, EMMC, Genetics Program and Administrator, invited
Beth Wilson, RNC, Perinatal Outreach/Perinatal Nurse Manager liason, attended
Dr. Ricka Wolman, MCH Medical Director, attended
Dr. David Whiteman, MMC, invited-input provided

Resources workgroup:

Participants:
Toni Wall, CSHN Director
Tammy Voisine, CSHN case manager
Margaret Squires, Maine Parent Federation, Co-director Special Parents Information Network (SPIN)
Judy Matthews, RN, Public Health Nurse
Kathy Philips, parent
Brenda Rodgerson, parent

Appendix B

Maine Birth Defects Program
Advisory Committee

Regular Members:

Dr. Stephen Amato, Pediatric Geneticist, EMMC
Dr. Laurent Beauregard, EMMC Genetics Program and Administrator, EMMC
Dr. Jacquelyn Blackstone, Perinatologist, MMC
Dr. Thomas Brewster, Pediatrician and Geneticist, Gorham
Jaci Holmes, Part C Coordinator, DOE
Jere Hoover, Executive Director, Maine Chapter March of Dimes
Donald Lemieux, Director, Office of Data Research and Vital Statistics
Kristina Lunner, Maine Medical Association
Judith Matthews, RN, Public Health Nursing
Ellie Mulcahy, RN, Director, Maine Genetics Program and Coordinator, MBDP
Glenn Palomaki, Director Biometry, Foundation for Blood Research (FBR)
Valerie Ricker, RN, Director Family Health Programs
Sharon Schulberger, Director, Program Services, Maine Chapter March of Dimes
Dr. Dan Sobel, Neonatologist, MMC, Maine Chapter American Academy of Pediatrics, Committee of Fetus and Newborn
Nancy Sonnenfeld, Epidemiologist, Division Community & Family Health
Dr. Rhonda Spiro, Medical Director, Southern Maine Genetics Service, FBR
Margaret Squires, Co-Director, Maine Parent Federation, Special Parents Information Network
Kathy Stuchiner, Maine Hospital Association
Toni Wall, Director, Children with Special Health Needs Program
Dr. David Whiteman, Geneticist/Dysmorphologist, MMC
Beth Wilson, RNC, Perinatal Outreach Program, MMC
Dr. Fredericka Wolman, MCH Medical Director, Division Community & Family Health
Paula Yoon, MPH, Centers for Disease Control

Ad Hoc Members:

Dr. Tim Boley, Perinatologist, EMMC
Dr. Mark Cooper, Obstetrician, Chair Maine Section Academy of Obstetricians and Gynecologists
Dr. Michael Curci, Pediatric Surgeon, MMC
Dr. Angela Gilladoga, Pediatric Cardiologist, EMMC
Dr. Kumar Kilesh, Neonatologist, EMMC
Dr. Thomas McGill, Pediatric Surgeon, EMMC
Dr. John Salvato, Pediatrician, Chair Maine Chapter American Academy of Pediatrics

Endnotes:

[i] Halperin, William and Baker, Edward L., eds. 1992. *Public Health Surveillance.* New York. Van Nostrand Reinhold.

[ii] Ibid.

[iii] Office of Data, Research and Vital Statistics, Bureau of Health, Maine Department of Human Services, 1998.

[iv] Ibid.

[v] Piper JM, Mitchel EF, et al. *Validation of 1989 Tennessee birth certificates using maternal and* newborn *hospital records.* Am J. Epidemiology 1993; 137:758-68.

[vi] Johnston, R.B. 1997. *Folic Acid: New Dimensions of an Old Friendship.* Advances in Pediatrics: 44:231-261.

[vii] ACOG Committee Opinion. 1993. *Folic Acid for the Prevention of Recurrent Neural Tube Defects.* Committee on Obstetrics: Maternal and Fetal Medicine. Number 120.

[viii] Johnston. 1997.

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This information is available in alternate formats upon request.