

The State Genetics Plan for Iowa

A Plan of Action for the Iowa Department of Public Health

Iowa Department of Public Health

Administration

Stephen C. Gleason, D.O., Director

Julie McMahon, R.N., Director of Family and Community Health

Edward L. Schor, M.D., Medical Director, Family and Community Health

Birth Defects Institute

Jeannette Anderson, R.N., M.A., CPNP, State Coordinator for Genetic Services

Tonya Diehn, M.S., Community Health Consultant

Iowa Department of Public Health
Division of Family and Community Health

Birth Defects Institute

Lucas State Office Building

321 East 12th Street

Des Moines, IA 50319-0075

2001

Acknowledgements

The 2001 State Genetics Plan for Iowa is the result of the collaborative efforts of many individuals. The project would not have been possible without the help of the following workgroups and programs:

**Genetics Family Focus Group
Parent Educator Connection
Parent – to –Parent Initiative
Parent Training and Information Center of Iowa
The Birth Defects Advisory Committee
Division of Medical Genetics personnel, Department of Pediatrics, Univ. of Iowa
The Iowa Birth Defects Registry personnel
The University Hygienic Laboratory personnel
The Department of Obstetrics and Gynecology personnel, University of Iowa
The Data Assessment and Integration Task Force
The Collaborative Legislative Task Force
The Division of Family and Community Health personnel, Iowa Dept. of Public Health**

A special acknowledgement and warmest thanks to all of the individuals and families who so kindly shared their personal experience and expertise:

**Verabeth Bricker
Linda Brown
Renee Compiano
Paula Connelly
Danielle Garcia
Jane Eilers
Sharon Frerker**

**Laura Groenenboom
Michelle Hall
Lisa Headness
Jennifer Kokjohn
Cheryl Mack
Mike and Holly McCormick
Tracy Ollendick**

**Jim Olinger
Jule Reynolds
Marilee Pietz
Monique Stam
Kay Stroutman
David Taylor
Delores Wellman**

Additional information about this document and Genetic Services in Iowa can be obtained through:

**The State Coordinator for Genetic Services
Birth Defects Institute
Iowa Department of Public Health
Lucas State Office Building
321 E 12th Street
Des Moines, IA 50319-0075
Phone: 515-281-7584**

This project was supported by Project # 4 H46 MC 00156-01 from the Maternal and Child Health Bureau (Title V, Social Security Act), Health Resources and Services Administration, Department of Health and Human Services.

Introduction

Newborn metabolic and hemoglobinopathy screening programs exemplify how collaborative genetic and public health initiatives can benefit children and their families through early detection, appropriate treatment, and prevention of mental retardation, physical disability and death. The children diagnosed by newborn metabolic screening programs, however, account for only a small number of the individuals with genetic conditions and other special health needs seen in public health programs.

Advances in the understanding of the genetic basis of congenital anomalies, inherited disorders and chronically disabling diseases such as diabetes and cancer promise to offer solutions for the treatment and health care management of many more people. However, environmental factors, lifestyle and behavioral choices also impact the manifestation of disease and birth defects.

Genetic and public health collaboration is essential for understanding how gene interactions, environmental factors and lifestyle choices are involved in both health and in disease. Incorporating genetic knowledge within public health programs has the potential to optimize the identification and care of infants, children and adults with special health needs, as well as promote the health of the general population.

Iowa legislation mandates that the Iowa Department of Public Health develop and administer the state's policy with respect to the conduct of scientific investigations and research concerning the causes, mortality, methods of treatment, prevention and cure of birth defects in order to provide for the protection and promotion of the health of the state's citizens. The Birth Defects Institute was established within the department in 1976 to initiate and conduct these investigations. Mandated

activities also included the development of genetic and metabolic screening programs, designation of a central laboratory, provision of professional and public programs, the conducting and support of clinical counseling services and maintaining a central registry for report data. Currently, the Birth Defects Institute is made up of the Regional Genetics Consultation Service, the Neuromuscular and Related Genetic Disease Program, the Iowa Neonatal Metabolic Screening Program, the Expanded Maternal Serum Alpha-fetoprotein Screening Program, and the Iowa Birth Defects Registry.

An analysis of the genetic health care, laboratory and surveillance programs and their function and utilization within the Iowa Department of Public Health and throughout the state was performed in 2000. In addition, individuals, families, health care and human service professionals shared their personal experiences and described their genetic priorities and service needs. The State Genetics Plan for Iowa summarizes the findings of the analysis and outlines the priority issues identified during the evaluation. The document provides a plan of action for the Iowa Department of Public Health to facilitate further genetic and public health collaboration and to ensure quality, comprehensive genetic health care, education, diagnosis, treatment, and supportive services for all Iowans.

SUMMARY OF RECOMMENDATIONS

- Lead in the provision and promotion of meaningful and relevant public and professional genetic education and training with the input of the citizens, health professionals and educators of Iowa.
- Improve family, health care provider and community awareness of the genetic services and programs in Iowa and the prevention, health promotion, surveillance, education, testing and health care services they provide.
- Assure and enhance the availability and accessibility of quality and comprehensive genetic services for all Iowans.
- Administer additional assessment and quality assurance measures for the genetic and metabolic screening programs of the Birth Defects Institute.
- Evaluate and refine current birth defect registry legislation and policies.
- Inform families when their child has been placed on the Iowa Birth Defects Registry and provide them with information about early intervention and supportive services they may be eligible for.
- Facilitate the development of a statewide teratogen service.
- Provide the assurance and policy development necessary to establish an integrated information repository for the Iowa Department of Public Health Programs serving the health care and human service needs of Iowa's families.

Lead In The Provision And Promotion Of Meaningful And Relevant Public And Professional Genetic Education And Training With The Input Of The Citizens, Health Professionals And Educators Of Iowa.

Background

Genetic research and technology is rapidly advancing the understanding of the genetic basis of congenital anomalies, inherited disorders and chronically disabling diseases such as diabetes and cancer. While the genetic factors cannot be prevented, lifestyle changes, behavioral modification, and in some cases preventive treatment may affect the manifestation or severity of the disease. Advances in genetic knowledge potentially offer solutions for the treatment and health care management of many individuals. With this knowledge, comes the need and responsibility to provide the general public a fair assessment of what genetics can and can not do, including the benefits, risks, and limitations of genetic testing, diagnosis, and treatment and the possible consequences of genetic testing.

A fundamental responsibility of the Birth Defects Institute is to implement public educational programs to inform persons of the importance of genetic screening and to provide education and training of medical students, physicians, nurses, scientists and technicians in the causes, methods of treatment, prevention and cure of birth defects. The Regional Genetic Consultation Service and the Neuromuscular and Related Genetic Disease Program provide statewide genetics education to health care professionals and the general public to promote health and prevent disease.

Public health programs are most effective in informing, educating, and empowering people about genetic health issues when they address the concerns and needs of families impacted by a genetic condition and the community at large. Consumers, parents and health care and human service professionals alike need to have a basic knowledge of genetics to be full participants in the care of an individual with a genetic condition, chromosome abnormality, or birth defect.

Needs Assessment Tools

One-to-one meetings with individuals of Genetics Focus Group
Parent-to-Parent Initiative
Parent Training and Information Center of Iowa
Birth Defects Advisory Committee Survey
Genetic Service Provider Survey
Regional Genetic Consultation Service Needs Assessment
Neuromuscular and Related Genetic Disease Program Needs Assessment
Optimizing Genetic Services in a Social, Ethical, and Policy Context-Suggestions from Consumers and Providers in the New England Regional Genetics Group

Lead In The Provision And Promotion Of Meaningful And Relevant Public And Professional Genetic Education And Training With The Input Of The Citizens, Health Professionals And Educators Of Iowa.

Priority Issues

- Rapidly expanding genetics information and technology. The IDPH and the programs of the Birth Defects Institute have the responsibility to keep current of these issues and lead in the development, promotion and provision of genetic education programs.
- Engagement before education. Engage the community at large to see what knowledge is wanted, then provide education based on what is desired.
- Pertinent and simple information. Genetic health care providers should provide educational outreach to the community at large and practical help and education to families engaged in daily care. Public health genetics should ensure that individuals and families are provided information about the recent advances of genetics as well as education that will enable them to make informed decisions and maximize health.
- Genetic curriculum for health care and human service students and professionals. Students and professionals in the fields of medicine, psychology, dentistry, nursing, physician assistant, occupational therapy, physical therapy, special education and social work should have basic knowledge of genetic disorders, referral and information resources, how to identify consumers in need of genetic counseling, and the impact a genetic condition can have on an individual and families.

Recommended Strategies/ Goals

- Mobilize community partnerships at the state and local levels to identify genetic health problems and educational needs.
- Develop and provide educational inservices and presentations to inform health care professionals and the general public about the genetic contribution to both common and rare disorders and the benefits, risks, limitations and potential consequences of genetic testing.
- The Iowa Department of Public Health and genetic professionals at educational institutions should advocate for genetic curriculum coursework for all health care and human service students and continuing education for professionals.
- Utilize the National Coalition of Health Professional Education in Genetics' recommendations for structuring genetic curriculum.
- Utilize parents and the community in educational inservices and presentations.
- Utilize multiple mediums to provide updated information on genetics- written, oral, and videotaped materials developed at all educational levels.
- Update literature resources for genetic conditions and technology at the public libraries. Involve families, communities, and care providers to determine desired and needed resources.
- Implement a resource library, including video-lending services at the Iowa Department of Public Health and expand and update resources at the University of Iowa.
- Expand Birth Defects Institute website to include parent resource information, consumer issues, genetics newsletter and a genetic services directory for health professionals with information about clinical genetics services, consumer support, newborn screening, birth defect surveillance, prenatal genetics services, genetic laboratories and specialty clinics.
- Involve the community at large in education about what questions to ask health care provider, what information to access, what records to keep.
- Develop a central clearinghouse for health care professionals and the general public to obtain more information on genetic conditions.

Improve Family, Health Care Provider And Community Awareness Of The Genetic Services And Programs In Iowa And The Prevention, Health Promotion, Surveillance, Educational, Testing, And Health Care Services They Provide.

Background

Since its creation, the Birth Defects Institute, in partnership with the University of Iowa and health care providers throughout the state, has developed programs that have provided Iowans with state of the art genetics health care. The programs of the Birth Defects Institute address all steps of the life cycle: prenatal, neonatal, pediatric and adult. The Department of Obstetrics and Gynecology at the University of Iowa Hospitals and Clinics provides comprehensive care to women with abnormal screening results identified through the Expanded Maternal Serum Alpha-fetoprotein Program. The Hemoglobinopathy Screening and Comprehensive Care Program, the Metabolic Clinic and the Endocrine Clinic of the Department of Pediatrics at the University of Iowa Hospitals and Clinics provide comprehensive care to the individuals and families identified through the Iowa Neonatal Metabolic Screening Program. The Regional Genetic Consultation Service provides comprehensive genetic health care services and education to individuals and families throughout Iowa with statewide outreach clinics. Comprehensive care services include diagnostic evaluations and confirmatory testing, medical management, providing non-directive information to individuals and families, case management, individual and family support, education, consultation and referral. These services promote health and prevention of disease in children, adult and families with diagnosed or undiagnosed genetic conditions. The Neuromuscular and Related Genetic Disease Program provides healthcare services and education to individuals with diseases and their families at locations around the state. Neuromuscular disorders affect individuals of all ages, socioeconomic backgrounds and ethnicity. Neuromuscular services include, but are not limited to: diagnostic evaluation, case management of healthcare problems, patient education and genetic counseling, physical therapy, psychosocial support, community education and connection to research opportunities. Through the service programs of the Birth Defects Institute, patients and their families gain access to specialized healthcare, efficient and accurate diagnosis, decreased morbidity, improved quality of life and knowledgeable communities.

Families, communities, health professionals and public health agencies are often not aware of the Birth Defects Institute and the genetic services its programs and other programs within the state provide. Information about what genetic services are, how to access genetic services, the benefits of genetic services, genetic research and the health care, supportive, human service and educational programs available to individuals with genetic conditions is desired by individuals, families and professionals.

Needs Assessment Tools

One-to-one meetings with individuals of the Genetic Focus Group
Regional Genetic Consultation Service Needs Assessment
Neuromuscular and Related Genetic Disease Program Needs Assessment
Birth Defects Advisory Committee Survey
Genetic Service Providers Survey
Public Health Information System Needs Assessment

Improve Family, Health Care Provider And Community Awareness Of The Genetic Services And Programs In Iowa And The Prevention, Health Promotion, Surveillance, Educational, Testing, And Health Care Services They Provide.

Priority Issues

- Promotion of genetic services available in Iowa. The Iowa Department of Public Health should actively promote awareness of all genetic disorder-related programs within the state.
- Health care provider and service program outreach. Collaboration and involvement with community health care providers and health and human service programs will facilitate awareness of genetic services and identification of individuals in need of these services.
- Accessibility to health care and human service resource information. Comprehensive and widely accessible materials about the health and human services programs available to individuals with genetic conditions and other special health care needs will promote awareness of and utilization of genetic programs.

Recommended Strategy/Goals

- The Iowa Department of Public Health should release public service announcements and news articles to increase the awareness of genetic services in Iowa.
- Work with primary care providers, local health departments, area education agencies, Early ACCESS and Title V, X and XIX programs to provide training on such issues as when it is appropriate to refer consumers to genetics centers or specialists, and how to identify consumers in need of genetic counseling.
- Include information about genetic services in materials prepared for families of children with special health care and/or educational needs; in materials that adults and families receive related to prenatal, pediatric and adult care and in other relevant public health programmatic materials.
- Collaborate with AEA parent coordinators to increase awareness of genetic conditions associated with learning and developmental disabilities and the benefits of genetic evaluations.
- Collaborate with March of Dimes and other non-profit organizations to promote genetic education and service awareness.
- Provide newsletters that include update information about genetic services and activities within the state.
- Expand Birth Defects Institute web site to include parent resource information, consumer issues, a genetics newsletter and a genetic services directory for health professionals with information about clinical genetics services, consumer support, newborn screening, birth defects surveillance, prenatal genetics services, genetic laboratories and specialty clinics.
- Disseminate genetic services directory to health and human service providers, genetic support groups, and parent groups.
- Develop and widely disseminate a document for communities and families describing genetic services in Iowa as well as the other health care, human service, educational and supportive programs that work with individuals and families with special health care needs. Ensure that the document is responsive to the concerns and needs expressed by families and individuals with genetic conditions and other special health care needs.

Assure And Enhance The Availability And Accessibility Of Quality And Comprehensive Genetic Services For All Iowans.

Background

The Iowa Department of Public Health is responsible for the promotion of health and disease prevention and assuring the access of quality comprehensive care to all Iowans with or at risk for genetic conditions. Presently there are five programs with the Birth Defects Institute: the Iowa Neonatal Metabolic Screening Program, the Iowa Birth Defects Registry, the Expanded Maternal Serum Alpha-fetoprotein Screening Program, the Regional Genetic Consultation Service, and the Neuromuscular and Related Genetic Disease Program. While these programs address all steps of the life cycle, approximately 52 and 70 percent of the patients seen in the Neuromuscular and Related Genetic Disease Program and Regional Genetic Consultation Service respectively are under the age of 20.

Genetics is a rapidly developing discipline, which is providing us with the knowledge and potential to better treat genetic conditions, individualize medical treatment, prevent the occurrence of certain birth defects and decrease the burden of chronically disabling diseases such as heart disease, cancer and diabetes. Continuous advances are being made in the identification of genes that contribute to disease and the technology to screen for and diagnose these diseases. The need for genetic services is every growing, particularly for adult-onset and chronic disorders. It is critical that the public health community develops policies and programs that keep abreast of genetic advances and assure the availability of and access to quality genetic health care services.

Needs Assessment Tools

Birth Defects Advisory Committee Survey
Genetic Service Providers Survey
Regional Genetic Consultation Service Needs Assessment
Neuromuscular and Related Genetic Disease Program Needs Assessment
Birth Defects and Genetic Counseling Program Contract
University Hygienic Laboratory 28E Agreement
Muscular Dystrophy and Related Genetic Diseases Program Contract

Assure And Enhance The Availability And Accessibility of Quality And Comprehensive Genetic Services For All Iowans.

Priority Issues

- Expansion of Genetic Services within Iowa Department of Public Health and the State of Iowa. Advances in the understanding of the genetic basis of congenital anomalies, inherited disorders and chronically disabling diseases such as heart disease, diabetes and cancer promises to offer solutions for the treatment and health care management of many Iowans. Expansion of medical genetic knowledge and services within the public health infrastructure has the potential to optimize the identification and management of infants, children and adults with special health needs, as well as promote health preservation and disease prevention for the general population.
- Collaboration with private sector and public health programs. Genetic factors, environmental factors, lifestyle and behavioral choices impact the manifestation of disease and birth defects. Collaboration is essential for discernment of the environmental, lifestyle and gene interactions involved in both health and in disease.
- Statewide genetic service assessment capacity and capability. Most medical conditions have a genetic component. Comprehensive health care for individuals with genetic disorders and other special health care needs is provided by a variety of providers. Assessment of all health care providers within the state of Iowa is needed to most accurately determine service utilization by individuals with genetic conditions or genetic susceptibilities. Additionally, knowledge of the prevalence of genetic conditions and health risk factors among Iowans is essential for determining service needs and for program planning, provision and administration.
- Birth Defects Institute name. The name is limiting, potentially negative and says nothing about the prevention, promotion, and education the programs provide for Iowans of all ages.

Recommended Strategy/Goals

- Change the name of the Birth Defects Institute to reflect the comprehensive services the programs provide and to acknowledge that genetic services and issues span the human life cycle.
- Improve the ability to assess the utilization of genetic services provided by genetic service and non-genetic service providers.
- Development of health status indicators that will accurately measure the population of interest.
- Increased epidemiological support for evaluating the genetic services program and assessment of the genetics health care delivery system.
- Explore strategies that will lead to assessing the morbidity and mortality attributable to genetic conditions.
- Documentation of referral, case management, and coordination activities performed by NMP and RGCS.
- Monitor quality assurance of genetic service providers.
- Encourage funding for developing genetic infrastructure and services through state appropriations and grant writing.
- Develop a plan to promote inclusion of a statewide genetic screening component into aspects of well woman care, well child care and preventive health services for both children and adults.
- Expansion of genetic services to include those which focus on adult onset problems, cancer, and hemochromatosis for example.
- Increase identification, education and promotion of services to patients and families for disorders for which new diagnostic tests and/or treatment/surveillance modalities are available.
- Improve the awareness and support of genetic services, knowledge and technology in public health.
- Develop collaborations with Division of Health Promotion, Division of Environmental Health, Maternal and Child Health, Child Health Specialty Clinics and other primary care and specialty services.

Administer Additional Assessment And Quality Assurance Measures For Genetic And Metabolic Screening Programs Of The Birth Defects Institute.

Background

The Birth Defects Institute of the Iowa Department of Public Health is legislatively mandated to develop and administer genetic and metabolic screening programs to detect and prevent or treat birth defects statewide. The Iowa Neonatal Metabolic Screening Program and the Expanded Maternal Serum Alpha-fetoprotein (MSAFP) Screening Program within the Birth Defects Institute provide neonatal and prenatal screening laboratory, follow-up, consultative and educational services. Metabolic screening of all newborns is required in the state of Iowa and testing is performed at the designated central laboratory. Expanded MSAFP screening is voluntary; however, when a woman chooses to have screening, the testing is to be performed by the designated central laboratory. The University Hygienic Laboratory is the designated central screening laboratory for the State of Iowa. Three divisions of the Department of Pediatrics at the University of Iowa provide consultation services for the Iowa Neonatal Metabolic Screening Program; Medical Genetics, Endocrinology and Hematology and Oncology. The Department of Obstetrics and Gynecology of the University of Iowa provides consultation services for the Expanded MSAFP Screening Program. The State Coordinator for Genetic Services coordinates the activities of the Birth Defects Institute with respect to the neonatal metabolic and MSAFP screening programs.

Iowa Public Health Administrative Code outlines screening policies, the responsibilities of the attending physician, central laboratory and consulting physician, and screening test fee determination. A memorandum of understanding between the University of Iowa and the Iowa Department of Public Health further delineates program responsibilities and fiscal management. The Birth Defects Advisory Committee advises the Iowa Department of Public Health Director regarding adding new tests to the screening programs, fee increases, and the use of developmental funds in compliance with written policy and procedures. The committee represents the interests of the people of the State of Iowa and assists in the development of programs that assure availability and access to quality genetic services for all Iowans. The National Newborn Screening and Genetics Resource Center sponsored a site review of the Iowa Neonatal Metabolic Program at the request of the Iowa Department of Public Health in April 2000. The resulting consultation report addressed the Iowa Department of Public Health's administrative responsibilities and made recommendations for program improvement.

Needs Assessment Tools

Iowa Newborn Screening Program Consultation Reports
Birth Defects Advisory Committee Minutes
University Hygienic Laboratory 28E Agreement
Expanded Maternal Serum Alpha-fetoprotein Screening Program Reports
Neonatal Metabolic Screening Annual Reports
Genetic Service Provider Survey
Birth Defect Advisory Committee Survey
Data Integration and Assessment Task Force

Administer Additional Assessment And Quality Assurance Measures For The Genetic And Metabolic Screening Programs Of The Birth Defects Institute.

Priority Issues

- Accountability for the metabolic screening of Iowa's newborns. The Iowa Department of Public Health (IDPH) has the mandated responsibility for assuring that every newborn is screened unless the parent(s) sign a waiver refusing testing.
- Uniform short-term follow-up procedures for all disorders screened by the Iowa Neonatal Metabolic Screening Program. Short-term follow-up begins with the identification of abnormal or unsatisfactory screening results and ends when a satisfactory sample has been received and tested or when confirmatory results are received. IDPH is responsible for assuring that the follow-up is efficient and effective.
- Monitoring and tracking of newborns diagnosed through the Iowa Neonatal Metabolic Screening Program. The effectiveness of newborn screening diagnosis and subsequent treatment and care management can only be determined when the long term outcome and functionality of the patients diagnosed is known.
- Funding and administration of the metabolic formula program. Lifelong dietary treatment is advocated for some metabolic patients. The current program structure and funding is unable to meet the dietary management needs of all individuals with metabolic disorders.
- Expanded MSAFP testing at laboratories other than the University Hygienic Laboratory, the designated laboratory for the Birth Defects Institute. The Iowa Administrative Code 641-4 mandates that when a patient desires Expanded MSAFP testing, the sample is to be submitted to the designated laboratory.

Recommended Strategy/Goals

- The Iowa Department of Public Health should perform matching of the Iowa Neonatal Metabolic Screening Program clients to Vital Records data and follow-up on all unmatched infants.
- Utilize the newborn blood spot card identification number as an infant's universal identifier and require number in all databases collecting information about newborns: INMSP, Vital Records, Newborn Hearing Screening and the Immunization Registry.
- Develop mechanisms for assuring uniform short-term follow-up procedures for all conditions screened by the Iowa Neonatal Metabolic Screening Program (INMSP).
- Central follow-up coordinator at IDPH to act as a "clearinghouse" for all follow-up personnel, INMSP personnel, hospitals and community health care providers.
- Development of mechanisms to monitor and track newborns diagnosed through INMSP through adulthood.
- Document quality assurance practices of the clinical service programs providing comprehensive care for the patients diagnosed through INMSP.
- Develop methods to enhance data accessibility and data sharing among the programs of the Birth Defects Institute and with the Children with Special Health Care Needs program to improve continuity of patient care and service coordination.
- Audit of current PKU metabolic formula program and review of funding sources.
- IDPH and INMSP should provide ongoing and regular primary care provider and laboratory inservices about newborn screening policies, attendant responsibilities, and laboratory submission requirements.
- Evaluate reasons for MSAFP sample submission non-compliance.
- Development of measures to achieve Expanded MSAFP sample submission compliance.
- Refinement of the Expanded MSAFP testing policy stated in the Iowa Administrative Code 641-4.

Evaluate And Refine Current Birth Defect Registry Legislation And Policies.

Background

In 1976, the Iowa Department of Public Health (IDPH) was given the legislative power to develop and administer the state's policy in respect to how scientific investigations and research concerning the causes, prevention, treatment and cure of birth defects were conducted. The Birth Defects Institute (BDI) was established to initiate and conduct these investigations. Statutory authority is identified in the Code of Iowa Chapter 136A. The legislation was amended in 1983 to facilitate the compiling of statistical information for investigations of genetic disorders and birth defects by permitting IDPH to maintain a central registry to collect and store report data. Identifying information collected by the registry is confidential pursuant to Iowa Code 22.7, subsection 2. The Iowa Birth Defects Registry (IBDR) was established in 1984 to provide a method of data collection and surveillance within the state.

The Iowa Birth Defects Registry is based at the University of Iowa and operates an active surveillance system utilizing multiple sources of case ascertainment. Birth defects have been reportable conditions under the Iowa Administrative Code 641.1.2 since 1988. The State Health Registry of Iowa (Registry), which includes the Cancer Registry and the Iowa Birth Defects Registry, has the delegated responsibility of collecting the data through review of hospital, clinic, laboratory and physician office records. Birth and death certificates are used as a secondary source of information. An interagency agreement between the Iowa Department of Public Health and the University of Iowa assists in the maintenance of the Registry by transmitting birth and death records. The agreement permits the Registry to release birth and death records to researchers for scientific investigation and research projects which have been approved by the University of Iowa Institutional Review Board and for which research agreements with IDPH have been completed. The Vital Records Bureau Director determines who is issued an IDPH research agreement.

The Iowa Birth Defects Registry and Vital Records both collect information about birth defects. Vital Records collects data on a limited number of specific congenital anomalies observed within 24 hours of delivery and reported by the attending physician on the birth or fetal death certificate worksheet. The Iowa Birth Defects Registry collects data on all pregnancy outcomes and live births with one or more birth defects diagnosed within the first year of life born to Iowa residents. The reporting system describes birth defects among Iowa residents according to guidelines specified by the Centers for Disease Control. In 1998, 470 infants were identified by Vital Records to have birth defects. IBDR identified 1665 infants. 296 of the infants identified by Vital Records were confirmed to have reportable birth defects after medical record abstraction by the registry. Many programs utilize vital statistics information for program planning because it more accessible and more current than registry data. Birth defects are the leading cause of death in children less than one year of age. Improving the health of Iowa's children is a critical issue and public health programs need accurate and reliable data for planning outreach, intervention, preventive, and educational services.

Needs Assessment Tools

Iowa Code Chapter 136A
Iowa Administrative Code 641, Chapter 4
Maternal and Child Health State Plan
Healthy Iowans 2010

IDPH contracts, research agreements
Center of Health Statistics
Iowa Birth Defect Registry
US Standard Certificate Revisions Report

Evaluate And Refine Current Birth Defects Registry Legislation And Policies

Priority Issues

- Administrative Code rules and regulations. Iowa Administrative Code 641-Chapter 4 defines regulations and rules for implementing the Birth Defects Institute and its programs. There are no regulation and rules for the central registry.
- Administrative liability and accountability. The responsibilities of the Iowa Department of Public Health for the Iowa Birth Defects Registry are not well defined. An interagency agreement between IDPH and the University of Iowa is not current.
- Iowa Department of Public Health research agreements. No internal review process exists for the release of personally identifiable information for scientific investigations and research projects.
- Birth defects are a public health issue. At the state and community level, there is a demand for obtainable data that accurately describes the number of infants with birth defects and is relevant for planning health care and human service programs for children with special health needs.

Recommended Strategy/Goals

- Adoption of rules and regulations for the Iowa Birth Defects Registry in the Iowa Administrative Code.
- Review of Chapter 136A of the Code of Iowa by the Attorney General to determine the role of the Iowa Department of Public Health in the administration and sustainability of the Iowa Birth Defects Registry.
- Implementation of an internal review board at the Iowa Department of Public Health.
- Improved utilization, accessibility, and usefulness of birth defects surveillance data.
- Determine how public health and human service programs want aggregate birth defects surveillance data reported and by what mechanisms.
- Develop policies for utilization of Iowa Birth Defect Registry data by the Iowa Department of Public Health.
- Develop mechanisms for the Iowa Department of Public Health to assure timely collection and publication of birth defects surveillance data, including financial support for record abstraction or in-kind support for report compilation.

Inform Families When Their Child Has Been Placed On The Iowa Birth Defects Registry And Provide Them With Information About Early Intervention And Supportive Services They May be Eligible For.

Background

Birth defects are structural, genetic or chromosome abnormalities that can adversely affect a child's health and development. They are the leading cause of death in children less than one year of age and a principal cause of childhood morbidity. Birth defects are reportable conditions in Iowa. Physicians comply with this reporting obligation by cooperating with the surveillance activities of the Iowa Birth Defects Registry. Since the registration of individuals in the registry is a public health function legislatively mandated to the Iowa Department of Public Health, parental consent for registration is not required.

The Iowa Birth Defects Registry (IBDR) performs statewide surveillance to record the types, frequency and distribution of birth defects. This knowledge is necessary to investigate potential causes of birth defects, plan for appropriate care and services and assess the impact of intervention. An essential component of the registry is research to identify the genetic and environmental causes of specific birth defects. All IBDR research projects are submitted to the University of Iowa Human Subjects Review Board for review and approval. Family participation in research is critical to determining the potential causes of birth defects. Parents of children entered into the registry are only contacted to partake in research after the registry receives passive consent from the child's physician to do so. Participating families typically provide environmental exposure information via mailed questionnaires or telephone surveys. Some research projects ask for biological samples from the affected child and his or her family members.

While birth defects are a vital public health issue, the priorities for the public health and individual families of children with birth defects may be quite different. Discussion with individuals and parents of individuals with genetic conditions, chromosome abnormalities and birth defects identified four priority issues.

Needs Assessment Tools

Iowa Administrative Code
Iowa Birth Defects Registry Annual Report
One-to-one meetings with parents of Genetic Focus Group
Parent-to-Parent Initiative
Parent Educator Connection
KID Coalition
The Beach Center on Families and Disability
Optimizing Genetic Services in a Social, Ethical, and Policy Context – Suggestions from Consumers and Providers in the New England Regional Genetics Group

Inform Families When Their Child Has Been Placed On The Iowa Birth Defects Registry and Provide Them With Information About Early Intervention and Supportive Services They May Be Eligible For.

Priority Issues

- Parents are not informed that their child has been ascertained by the Iowa Birth Defects Registry. Birth defects research and prevention efforts require the involvement of parents with affected children, so they have the right to know about birth defect surveillance activities and the registration of their child.
- Communication of research practices and results. Many parents understand the importance of research especially when it could determine the cause of their child's condition and potentially benefit that child and their future children.
- Lack of awareness of available early intervention and supportive services. The parent(s) of a child with a major birth defect, genetic condition or chromosome abnormality are faced with significant challenges in the day-to-day activities many families take for granted. Their primary focus is to meet the daily nutritional, medical, communication, educational and recreational needs of their child. Parent to parent support increases a parent's capacity to meet these needs and to accept the challenges they face.
- Iowa Birth Defects Registry outreach. Early identification of a child with a birth defect coupled with early intervention services saves lives and improves long term chances of a meaningful life.

Recommended Strategy/Goals

- The health care professional diagnosing a child with a birth defect at birth should inform parents about the Iowa Birth Defects Registry and its purpose.
- Inform families by a letter from the Iowa Department of Public Health and the Iowa Birth Defects Registry when the child is entered into the registry.
- Develop an IBDR brochure with information about its purpose as well as information about early intervention and supportive services that the health care professional making a diagnosis could give to parents.
- Develop mechanisms to ensure that parents are not asked to participate in research without being informed about their child's registration first.
- Utilize newsletters to keep participating families informed of research results and any publications or reports generated from the studies.
- Resource information about early intervention and supportive services their child and the family may be eligible for should be provided when informing the family about the child's entry into the registry.
- Collaborate with parent groups, March of Dimes, Regional Genetic Consultation Service, Neuromuscular and Related Genetic Disease Program, Maternal and Child Health, Child Health Specialty Clinics and Early ACCESS to determine what resource information should be provided to families.
- Support existing parent to parent programs and family to family health information initiatives and utilize the programs to provide resources for parent support and information.
- The Iowa Birth Defects Registry should promote improved awareness and utilization of surveillance data by agencies providing early intervention services.
- Utilize collaboration to promote awareness and utilization of aggregate birth defects surveillance data for early intervention service planning and provision.

Facilitate The Development Of A Statewide Teratogen Service.

Background

Almost five pregnancies per day are affected by birth defects in the state of Iowa. The causes of many birth defects are unknown. Maternal use of illicit drugs, prescribed medications, alcohol, exposure to chemicals and radiation, and maternal illness and disease during an individual's prenatal development attribute to up to five percent of congenital anomalies. Environmental agents that cause birth defects are called teratogens. Teratogens are a cause of great concern to all in our society. Most birth defects caused by teratogenic exposures can be prevented. The prevention of these congenital anomalies is a public health issue.

State public health programs and their agencies should take the lead in supporting endeavors to decrease the number of preventable birth defects. Teratogen Information Systems have an important role in primary prevention. They are designed to work with the public/and or interact with health professionals and their patients and provide information about the risks that a particular environmental agent has for conception and prenatal development. A Teratogen Information System should be staffed by health professionals with training and/or experience in genetics, fetal development and teratology. The provision of educational programs and outreach about prevention preconceptionally, as well as prenatally, are an integral component. A Teratogen Information System should be in place in every state and its services should be available to all residents. A statewide Teratogen Information System is not in place in Iowa.

Needs Assessment Tools

Regional Genetic Consultation Service Needs Assessment
Birth Defects Registry Annual Report
Guidelines For Clinical Genetic Services For The Public's Health –Council of Regional Networks of Genetic Services
Guidelines For State Genetic Services Programs – Great Plains Genetics Service Network

Facilitate The Development Of A Statewide Teratogen Service

Priority Issues

- Health promotion and protection of our most vulnerable population—the unborn child. One such course of action is the development of a statewide teratogen information service, which would include a pregnancy hotline, educational programs for the public and health care professionals, preconception counseling and education.
- Prevention of birth defects caused by teratogenic exposures. Prevention of birth defects is facilitated by public awareness of potential risks and available resources for information.
- Capacity to develop a formal statewide Teratogen Information Service. A statewide clearinghouse for teratogen questions and referrals to appropriate services does not exist. Statewide consultation outreach, education, and activities do not occur.

Recommended Strategy/Goals

- Develop the standards, the format and expected outcomes of a statewide Teratogen Information Service.
- Collaborate with March of Dimes, Title V maternal health and family planning programs, the Division of Environmental Health, the Division of Health Promotion, Prevention, and Addictive Behaviors, the Iowa Birth Defects Registry, and the Regional Genetic Consultation Service, the UIHC genetics program, and community health care providers in this development.
- Partner with the state's perinatologists, private practitioners, the Regional Genetic Consultation Service and the University of Iowa's Genetic Clinic to improve teratogen services within the state.
- Explore financial support for a Statewide Information System including state appropriations and federal grants.
- Expand the capacity of Iowa Department of Public Health to provide a statewide Teratogen Information Service with a clearinghouse for teratogen information, referrals to appropriate services, and pregnancy outcome tracking.
- The Birth Defects Institute should develop and present education programs and activities aimed at promoting healthy behaviors and reducing the numbers of preventable birth defects.
- Referral to appropriate services near the patient's home should be made available (i.e. industrial hygienist, epidemiologist, high-resolution ultrasound, occupational health expert).
- Implement a toll-free telephone number for teratogen inquiries.
- Incorporate information regarding the teratogen service and teratogenic agents in public education for persons of reproductive age.

Provide The Assurance And Policy Development Necessary To Establish An Integrated Information Repository For The Iowa Department Of Public Health Programs Serving The Health Care And Human Service Needs Of Iowa's Families.

Background

The mission of the Iowa Department of Public Health is to promote and protect the health of Iowans. The essential functions of the department include monitoring health care status, identifying community health problems, linking people to needed personal health services, assuring the provision of health care, and evaluating the effectiveness, accessibility, and quality of individual and population-based health care programs. Efficient and accurate data collection, analysis, interpretation and dissemination are critical to address these public health care services. Data integration of the enabling, population-based and direct health care service programs within the department has the potential to greatly improve the understanding of health needs, barriers and service utilization of Iowa's mothers, children and families. Integration will improve the accessibility of data for performance measure reporting, health status indicator evaluation, identification of emerging issues and gaps in the provision of services, and in the assessment of strengths and weaknesses in service delivery.

A vision of the Iowa Department of Public Health (IDPH) is to have an integrated information repository containing the client-based data of all the IDPH programs serving the health care and human service needs of Iowa's children, women and families. As IDPH was developing this vision, the Federal Maternal and Child Health Bureau awarded the state of Iowa, a Genetics Planning Grant in the fall of 1999. The grant is entitled, "A Plan for the Development of a State Genetics Plan and Integrated Data Collection and Service System for Early Intervention." An integral component of the planning grant was the proposal for the development of an integrated child health information system.

The planning grant has served as a unique opportunity to bring together the data integration and data enhancement initiatives for women and children occurring at IDPH. Ten programs serving the health and human service needs of women, children and families at the Iowa Department of Public Health met in 1999 through 2000 to assess the feasibility of data integration and to examine the ethical, legal and confidentiality issues pertaining to data access and use. The Family and Community Health Division Director and representatives from the Family Services Bureau, Bureau of Information Management, Bureau of Childhood Immunization, Bureau of Vital Records, Iowa Neonatal Metabolic Screening Program, Iowa Birth Defects Registry, Childhood Lead Poisoning Prevention Program, Newborn Hearing Program, WIC, and the State Center for Health Statistics comprised the Data Assessment and Integration Task Force. A needs assessment of the current public health information system for programs collecting, maintaining and disseminating data on women, children and families was performed. The Data Assessment and Integration Task Force identified five priority issues that need to be resolved before an integrated information system addressing the needs of clients, health care and service providers and local, state and federal service programs can become a reality.

Needs Assessment Tools

Public Health Information System Needs Assessment
Data Assessment and Integration Task Force Report and Recommendations

Provide The Assurance And Policy Development Necessary To Establish An Integrated Information Repository For The Iowa Department Of Public Health Programs Serving The Health Care And Human Service Needs Of Iowa's Families.

Priority Issues

- Public Health Administrative Rules for data sharing and integration. Current rules do not permit data sharing between IDPH programs or development of a centralized information system.
- Duplication of data collected from the children, women and families the IDPH programs serve. Duplicative efforts are costly, time consuming and do not promote the health of children, women and families.
- Variability in how demographic variables are defined. Different demographic variable definitions impede data set linkages, data analysis and dissemination of good, quality aggregate data.
- Restricted accessibility to program data. Greater access at the local and state level is needed to provide, coordinate and manage healthcare and human services for clients without duplicative data collection and data entry. Improved accessibility would facilitate identification of clients for early intervention services.
- Independent integration initiatives within the health department. The multiple data systems evolving to meet the “ integration needs” of their individual programs do not service public health efforts effectively.
- Security of information system and data sharing. Confidentiality of personally identifiable information is of utmost importance.

Recommended Strategy/Goals

- Implement Public Health administrative rules for data sharing between the health care and human service programs that IDPH administers.
- Establish rules to permit the development of centralized information system resident in the Iowa Department of Public Health. Rules should include security measures for ensuring confidentiality.
- Establishment of core data elements for the health care and human service programs IDPH administers.
- Review process for any Iowa Department of Public Health data enhancement, integration, or data sharing project.
- Develop initiatives to improve data accessibility and utilization to meet the information needs of clients, health care providers, human service professionals, community clinics, and state and federal service programs.