WE ARE PLEASED TO PROVIDE YOU WITH THE 2005 REPORT of the Iowa Registry for Congenital and Inherited Disorders. Formerly called the Birth Defects Registry, the Registry was recently renamed to more accurately reflect its expanded programmatic and research activities. The University of Iowa supports this new focus and looks forward to working with the Iowa Department of Public Health to continue their long-standing partnership to promote the health and well-being of young Iowans.

In the past, the Registry has served as a model for other registries around the nation. In its new role, we believe that the Registry will continue to provide leadership at the national, state, and community levels to respond to emerging health issues in young populations. Already Registry staff have undertaken several important new initiatives. They have recently developed a partnership with the Muscular Dystrophy Surveillance Tracking and Research Network (MDSTAR.net) to track Duchenne/Becker Muscular Dystrophy in Iowa. In addition, they have also expanded the Registry’s outreach activities by contacting families to notify them when their children have been added to the Registry. As a result, the families have the opportunity to be connected to other state services and resources.

Data collected by Registry staff are utilized in a variety of research and educational activities both here on The University of Iowa campus and in collaboration with other research institutions across the nation. These data also provide the basis for program planning, health policies, and prevention efforts in support of the welfare of Iowa’s infants and children. Selected current topics being studied include: neural tube defects, cleft lip and palate, Down syndrome, and muscular dystrophy.

In the past, a strong statewide monitoring system has been a critical tool for birth defects prevention. We are confident that the Registry’s expertise will be invaluable to the study of other childhood disorders that can be prevented through further research and education.

Sincerely,

David J. Skorton, M.D.
President

Michael J. Hogan, Ph.D.
Provost
F. Wendell Miller
Professor of History

James A. Merchant, M.D., Dr.P.H.
Professor and Dean
College of Public Health
Dear members of the Legislature, health care professionals, and concerned citizens of Iowa:

I am proud to announce the release of the 2005 report of the Iowa Registry for Congenital and Inherited Disorders, formerly called the Iowa Birth Defects Registry. For many years, the Registry has been an important part of the state’s effort to promote the health of Iowa’s children. It provides data to researchers working to identify risk factors for various childhood disorders, and it also participates in outreach activities to educate and inform our citizens.

It is good news, therefore, that the Iowa Registry for Congenital and Inherited Disorders is expanding its surveillance and outreach activities. During the past year the Registry, in collaboration with the Iowa Department of Public Health, established a parent notification system to let parents know when their children’s information has been added to the Registry. This provides an opportunity to connect parents and families to state services when appropriate. This initiative was noted by the Trust for America’s Health, which rated the Registry with an “A” grade for its work in birth defect surveillance.

The Iowa Registry has also expanded its activities to include surveillance of patients with Duchenne/Becker Muscular Dystrophy. This attention to a developmental disorder represents a significant expansion for the Registry, which has traditionally focused on birth defects that are observed within the first year of life.

The Iowa legislature recently passed legislation that will provide additional funding for Registry activities. This is essential if the Registry is to continue surveillance and outreach in all 99 counties. The past year has been a year of growth for the Iowa Registry for Congenital and Inherited Disorders, and its new name is designed to reflect this expanded focus and capability.

Very truly yours,

[Signature]

Mary Mincer Hansen, R.N., Ph.D.
Director
Iowa Department of Public Health
A birth defect is a reportable condition in Iowa. The Iowa Birth Defects Registry was established in 1983 through the joint efforts of The University of Iowa, The Iowa Department of Public Health and the Iowa Department of Human Services.

**LOCATION**
The Iowa Registry for Congenital and Inherited Disorders is located within the College of Public Health at The University of Iowa. The Registry conducts active surveillance to identify information about congenital and inherited disorders that occur to Iowa residents. Active surveillance entails the use of field staff who collect information by reviewing medical records in hospitals and clinics in Iowa and in neighboring states that serve Iowa residents.

**MISSION**
The mission of the Iowa Registry for Congenital and Inherited Disorders is to: 1) maintain statewide surveillance for collecting information on selected congenital and inherited disorders in Iowa; 2) monitor annual trends in occurrence and mortality of these disorders; 3) provide data for research studies and educational activities for the prevention and treatment of these disorders.

**SURVEILLANCE**
The Iowa Registry for Congenital and Inherited Disorders has collected information for over 40,000 children with various birth defects. This information has been used by health care providers and educators to provide treatment and support services, and by researchers to study risk factors for birth defects and to evaluate treatments for birth defects. The Iowa Registry has recently started surveillance for Duchenne/Becker muscular dystrophy and has identified almost 50 children with that neuromuscular disease.

**RESEARCH**
Data collected by the Iowa Registry for Congenital and Inherited Disorders have been used in several research projects. Many of these projects include mailed or telephone surveys of women who have experienced a pregnancy affected by a birth defect, and, for comparison, women who have not experienced a pregnancy affected by a birth defect. Examples of birth defects studied by the Registry include Down syndrome, heart defects, neural tube defects, and cleft lip and palate.

**EDUCATION**
The Iowa Registry for Congenital and Inherited Disorders also participates in educational programs designed to help prevent the occurrence and recurrence of birth defects. Registry faculty and staff annually present lectures around the state and promote community awareness to students, families, health care workers, and multiple agencies. Awareness training is a cornerstone of our educational program.

**CONFIDENTIALITY**
Information collected by the Iowa Registry for Congenital and Inherited Disorders is kept confidential using computer security measures and locked files and offices. All staff members are required to sign a pledge to maintain the confidentiality of all information collected. These individuals are also reminded that their pledge remains in effect after the conclusion of their employment. Confidentiality is rigorously maintained so that the rights and welfare of the patients and families are not compromised.

**RECOGNITION**
The Iowa Registry for Congenital and Inherited Disorders has received national recognition for its role in birth defect surveillance, research, and education. In 1996, the Iowa Registry was one of only eight registries to receive an award from the Centers for Disease Control and Prevention to establish a “Center for Excellence in the Research and Prevention of Birth Defects.” In 1999 and again in 2002, the Iowa Registry received an “A” rating from the PEW Environmental Commission and the Trust for America’s Health, respectively, for its work in birth defect surveillance. In 2003, the Trust recognized the Iowa Registry’s continued improvement.
Early on, birth defect surveillance programs were established in response to:

- Clusters of birth defects, such as the high number of skeletal defects associated with a mother’s use of the morning sickness medication, thalidomide, and
- Concerns over environmental pollutants, such as the impact of contamination from toxic waste sites on the occurrence of birth defects.

More recently, birth defect surveillance programs have been established to permit health officials to:

- Implement and evaluate intervention programs, and
- Estimate the need for special health care services.

Often, birth defect surveillance programs, such as the Iowa Registry for Congenital and Inherited Disorders, attempt to provide a comprehensive monitoring program through surveillance, research and education efforts.

**BIRTH DEFECT SURVEILLANCE-UNITED STATES**

The Centers for Disease Control and Prevention (CDC) recognizes three types of birth defect surveillance systems, each rated differently for completeness of patient ascertainment:

- **Vital Records:** Use of birth and fetal death certificates provided by the state’s Department of Health (Rating: Poor)
- **Passive Reporting:** Use of medical reports submitted by staff from hospitals, clinics, or other facilities (Rating: Fair to Good)
- **Active System:** Use of trained personnel who systematically review records in hospitals, clinics, or other facilities (Rating: Excellent)

For the years 1997 through 2001, a record review of 9,091 infants identified by the Iowa Registry for Congenital and Inherited Disorders’ active surveillance system showed that vital records would have identified only 1,641, or 18%, of these infants. Therefore, for these years, if the Registry relied only on vital records for birth defect surveillance, on average, almost 4 out of every 5 infants diagnosed with a reportable birth defect would not have been identified.

**IOWA BIRTH DEFECTS ASCERTAINMENT, 1997-2001**

Although active surveillance systems may provide the most comprehensive surveillance for birth defects, they are also the most costly to maintain. In the United States, there are only 11 states and 1 territory that have birth defect registries that meet the criteria for an active surveillance system. As shown on the map, the Iowa Registry for Congenital and Inherited Disorders is the only active surveillance system in the Midwest.

**ACTIVE BIRTH DEFECTS SURVEILLANCE-UNITED STATES**
The term “defect” refers to abnormal development related to body structure, body function and metabolism, or an error in body chemistry.

Typically a defect is present at birth (congenital), but a recognizable defect may be diagnosed during pregnancy (prenatal) or following birth (postnatal). Birth defects can have many causes including genetic mutations, chromosomal abnormalities, environmental exposures, nutrition, and/or random events in the prenatal period. Many defects are thought to result from a combination of these factors. Examples of two major categories of birth defects include structural and metabolic defects. Metabolic defects often involve the inability of cells to produce a protein in the correct amount to regulate the chemistry of the body. Structural defects typically involve a body part that is missing or malformed. Examples include heart defects, spina bifida, and cleft lip and palate. Other terms often used for these types of defects include anomalies, malformations, and deformities. The Iowa Registry monitors for these and many other structural defects.

<table>
<thead>
<tr>
<th>Type of Birth Defect</th>
<th>Rate per 1,000 Live Births</th>
</tr>
</thead>
<tbody>
<tr>
<td>Brain/Spinal Cord</td>
<td>4.4</td>
</tr>
<tr>
<td>Congenital Infection</td>
<td>0.4</td>
</tr>
<tr>
<td>Congenital Tumors</td>
<td>3.2</td>
</tr>
<tr>
<td>Digestive</td>
<td>5.5</td>
</tr>
<tr>
<td>Ear</td>
<td>5.1</td>
</tr>
<tr>
<td>Eye</td>
<td>3.8</td>
</tr>
<tr>
<td>Genital/Urinary</td>
<td>10.4</td>
</tr>
<tr>
<td>Heart</td>
<td>13.0</td>
</tr>
<tr>
<td>Muscle/Skeletal</td>
<td>14.0</td>
</tr>
<tr>
<td>Oral/Facial</td>
<td>5.9</td>
</tr>
<tr>
<td>Respiratory</td>
<td>1.1</td>
</tr>
<tr>
<td>Skin</td>
<td>4.1</td>
</tr>
<tr>
<td>Syndromes</td>
<td>4.2</td>
</tr>
<tr>
<td>Other*</td>
<td>1.3</td>
</tr>
</tbody>
</table>

*Includes blood, hormonal, and metabolic conditions
NUMBER OF DELIVERIES AFFECTED BY BIRTH DEFECTS IN IOWA, 1997-2001

This map shows the number of deliveries in Iowa affected by one or more reportable birth defects by mother's county of residence for the period 1997-2001. When examining this map, note that numbers in some counties may be higher due to 1) greater population in certain counties and 2) greater numbers of reproductive-aged individuals (15-44) in such counties.

RATE OF DELIVERIES AFFECTED BY BIRTH DEFECTS IN IOWA, 1997-2001

This map shows the rate of deliveries in Iowa affected by one or more reportable birth defects by mother's county of residence for the period 1997-2001. For all counties combined, the average rate of occurrence of birth defects for this time period was 48.5/1,000 live births.
Muscular dystrophy refers to a group of genetic diseases that cause progressive muscle weakness. The most common form of muscular dystrophy affecting children is Duchenne/Becker muscular dystrophy (DBMD). Duchenne muscular dystrophy is the name that historically refers to the most severe form of this disorder. DBMD usually presents with weakness in early childhood. Weakness is progressive and children lose the ability to walk in late childhood. In the severe form, death occurs in young adulthood.

DBMD is caused by mutations in the dystrophin gene on the X chromosome. Girls rarely have the disease, but they can be carriers of the gene mutation. Approximately 1 in 3,500 boys have DBMD. Approximately one-third of boys with Duchenne muscular dystrophy did not inherit the disorder, but have a genetic mutation that is new in their family.

On behalf of MDSTARnet, the Iowa Registry for Congenital and Inherited Disorders is identifying and following the course of children with this disease. This long-term follow-up represents a major change for the Iowa Registry, which has primarily concentrated on providing comprehensive descriptions of anomalies that manifest within the first year of life. Through the information collected, we expect to be able to answer the following questions:

- How often does DBMD occur?
- Is it equally common in different racial and ethnic groups?
- What are the early signs and symptoms of DBMD?
- Do factors such as the type of care received or the type of gene changes affect the severity or course of DBMD?
- What medical and social services are families receiving?
- Do different populations receive different care?

In addition to the Iowa Registry, surveillance programs in Arizona, Colorado, and New York are also participating in this effort, which is being coordinated by The Centers for Disease Control and Prevention. The Muscular Dystrophy Association has endorsed this project and is working closely with each participating state.
The March of Dimes advocates for policies and programs that work to help Iowa families have healthier babies. In Iowa, volunteers represent a grassroots voice for mothers and babies. The Iowa State Chapter of the March of Dimes has an active advocacy network, key contacts, and committed volunteer leaders who work together to achieve success!

In the fall of 2002, the March of Dimes Iowa State Chapter staff and volunteers worked with key public policy/program advisors from the Iowa Medical Society, the Iowa Hospital Association, and the Iowa Department of Public Health on an appropriations bill for funding of active surveillance for the Iowa Registry for Congenital and Inherited Disorders. The decision was made to support a $5.00 increase in birth certificate fees from July 1, 2003 to July 1, 2005 and then another $5.00 increase after July 1, 2005 for a total of $10.00 to help fund active surveillance for the Registry. This would take birth certificate fees from $10 to $15 and then to $20 after July 1, 2005.

With the leadership of Rep. Pat Murphy (D) and Rep. Paul Wilderdyke (R) the bill made its way through the House and Senate. Throughout the session, March of Dimes relied on key individuals, Paul Romitti, Director of the Registry; Tonya Diehn, State Coordinator for Genetic Services, Department of Public Health; Lynh Patterson, Legislative Liaison, Iowa Department of Public Health, and Mark Braun, Director of State Relations, The University of Iowa to provide technical information and assistance. The Chapter also called on their advocacy network, key contacts and volunteers to voice support for this legislation. On May 1, 2003, Governor Vilsack signed the bill into law.

Unfortunately, there were no appropriations attached to this bill, thereby making it only a policy bill. The March of Dimes worked closely with both Democrat and Republican leadership to draft an appropriations bill for the 2004 legislative session. An appropriations bill was introduced and did pass through the Senate and House. Governor Vilsack signed the bill on March 18, 2004. The projected funding for the Iowa Registry for Congenital and Inherited Disorders active surveillance by 2005 is $250,000.

Governor Tom Vilsack signs the legislation that will help fund the activities of the Iowa Registry for Congenital and Inherited Disorders. Behind Governor Vilsack (from left to right) are Suzanne Heckenlaible from the March of Dimes, Joyce Kohl from the March of Dimes, Denise Ramsey from the Iowa Department of Public Health, and Paul Romitti, Director of the Registry.
RESEARCH

The Iowa Registry for Congenital and Inherited Disorders provides data for several research projects. A brief description of some of these projects is presented below:

**IOWA CHILD HEALTH STUDY**  
*Funding Agency: National Institutes of Health*  
This is a population-based study to investigate genetic and environmental risk factors for cleft lip and/or cleft palate (CL/P). Iowa women with a pregnancy affected by a CL/P and women with an unaffected pregnancy are asked to report information on family history, medical history, nutrition, lifestyle, and occupation to study environmental factors that may contribute to the development of CL/P. Biologic samples are collected from families to study genetic factors that may contribute to CL/P.

**NATIONAL BIRTH DEFECTS PREVENTION STUDY**  
*Funding Agency: Centers for Disease Control and Prevention*  
This is a population-based study to investigate genetic and environmental risk factors for 31 major birth defects. This study is a combined effort of the Iowa Registry and registries in seven other states. Women with a pregnancy affected by one or more of the 31 defects and women with an unaffected pregnancy are interviewed about their health, diet and lifestyle during their pregnancies. Biologic samples are also collected from each family to study genetic factors that may contribute to these birth defects.

**NATIONAL DOWN SYNDROME PROJECT**  
*Funding Agency: National Institutes of Health*  
This is a population-based study to investigate genetic and environmental risk factors for Down syndrome. This study is a combined effort of the Iowa Registry and registries in five other states. Iowa women who give birth to an infant affected with Down syndrome and women with an unaffected birth are interviewed about their health, diet and lifestyle during pregnancy. Biologic samples are collected from each family to better understand genetic factors that may contribute to Down syndrome.

**NATIONAL BIRTH DEFECTS PREVENTION NETWORK**  
The National Birth Defects Prevention Network is a nationwide association of birth defect registries, and one of its goals is to encourage scientific collaboration. Current projects are focused on such problems as preterm births, gastroschisis, and neural tube defects. The Iowa Registry for Congenital and Inherited Disorders is an active member of the National Birth Defects Prevention Network.


In 2000, the Iowa Department of Public Health (IDPH) conducted an assessment of its genetic health care, laboratory and surveillance programs to determine how they functioned and were used throughout the state. As part of this review, individuals, families, health care and human service professionals shared their personal experiences and described their priorities and needs for services.

This effort culminated in 2001 with the development of the State Genetics Plan for Iowa. It was determined that parents are very interested in the state’s surveillance for childhood disorders. Parents wanted to learn more about the Registry’s outreach activities, opportunities to participate in research, and early intervention resources.

In response to this need, the Iowa Administrative Code was revised to direct the Iowa Registry for Congenital and Inherited Disorders to inform parents when their child’s name and information is listed in the Registry. In addition, the Administrative Code directs the Registry to provide the parents with information about services for their child or family, such as early intervention services.

TAMMY O’HOLLEARN, Community Health Consultant with IDPH, was hired to facilitate the development of a parent notification system. Initially, a survey was mailed to physicians, and nurse practitioners throughout the state to assess their knowledge of the Registry and its activities, as well as to solicit input into the development of the parent notification system. A state-wide work group was then formed to develop the notification system. Participants in the work group included parents, pediatricians, social workers, service providers, attorneys and staff from the following agencies/programs: Area Education Agency, Early ACCESS, Child Health Specialty Clinics, Department of Public Health, Department of Human Services, Parent Training and Information Center, Parent Educator Connection, Registry staff and genetic service professionals. These individuals developed materials and provided invaluable input into the process that should occur when families are notified that their child’s name was placed in the Registry.

The Iowa Registry for Congenital and Inherited Disorders started parent notification for children born in Iowa on or after January 1, 2003. Parents receive a notification letter along with information about the Registry and a brochure outlining state resources that may be relevant to their situation. A postage-paid reply envelope is also included so that the family can have the opportunity to correct the Registry’s information. This provides a quality assurance check on the Registry’s records.

A copy of the notification letter is also sent to the family’s doctor who is encouraged to speak with the parents.

The parent notification letter process creates an additional opportunity for families seeking information to be connected to community resources. Heather, one of the parents who received the letter, stated “This was so important to me, personally. As a new parent it was really the first chance I had to learn about available resources to help my family and connect with other parents. It made such a difference.”
MR. BILL BUDELIER

There is an enormous amount of information stored within the files of the Iowa Registry for Congenital and Inherited Disorders. Over 40,000 cases are listed in the Registry, with about 2,000 more added each year. Managing all this information takes considerable effort and skill, and the Registry has been fortunate to have Bill Budelier working as a programmer to keep it organized and accessible.

In the 15 years Mr. Budelier has worked for the Registry, he has seen numerous changes in the way it works. Many records used to be indexed on note cards rather than by computer, and intensive data processing was done on a mainframe computer. The introduction of desktop PCs has led to a number of changes in information management. Keeping up with these changes is one of the more interesting and challenging parts of his job.

One of Mr. Budelier’s main responsibilities is to work closely with investigators to make sure they get the data they need for their research. He has also used his statistical skills as a co-author on various scientific publications. Being part of a research community is particularly rewarding for him, and he enjoys being part of the healthcare field as well. “I feel that research in the causes and prevention of birth defects is important,” says Mr. Budelier, “we are helping children and their families with the work we are doing here.”

DR. KATHERINE MATHEWS

The Iowa Registry for Congenital and Inherited Disorders has expanded its statewide surveillance to include childhood onset Duchenne/Becker Muscular Dystrophy (DBMD). This is a result of its participation in MDSTARnet (Muscular Dystrophy Surveillance and Tracking Network). Dr. Katherine Mathews has played an important role in this expansion of the Registry’s mission. As a co-principal investigator for MDSTARnet with Dr. Paul Romitti, she has been closely involved in the development of the surveillance protocol.

Dr. Mathews is optimistic about MDSTARnet and its potential to help people with DBMD. “This project allows us to examine all aspects of the disease across populations. We will learn about variability in DBMD and the impact of different treatments. We also have the opportunity to explore the effect of DBMD on family members as well as patients,” says Dr. Mathews. In addition to the standard MDSTARnet protocol, she is also coordinating a related project that assesses the needs of families of those who have DBMD.

Dr. Mathews completed residencies and fellowships in pediatrics, neurology, child neurology and genetics. Her research interests began with facioscapulohumeral dystrophy and have extended to all forms of muscular dystrophy. She is the Director of Child Neurology at the University of Iowa Hospitals and Clinics and has been the Director of the Iowa Neuromuscular Program for 8 years.
The Iowa Registry for Congenital and Inherited Disorders encourages readers to review these sites and discuss these topics in further detail with their medical providers. The Iowa Registry is not responsible for the quality of information provided at external sites.
We gratefully acknowledge the assistance of the following collaborating Iowa agencies and organizations:

THE UNIVERSITY OF IOWA
- Members of the internal advisory committee for the Iowa Registry for Congenital and Inherited Disorders
- Center for Health Effects of Environmental Contamination
- College of Liberal Arts
- College of Medicine
- College of Nursing
- College of Public Health
- Craniofacial Anomalies Research Center
- Iowa Cancer Registry
- University Hygienic Laboratory
- UI Governmental Relations Office

Iowa Department of Public Health and the members of the advisory committee for the Center for Congenital and Inherited Disorders

Iowa Regional Genetic Consultation Service

March of Dimes Birth Defects Foundation

KID Coalition

ASK Resource Center

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