Dear members of the Legislature, health care professionals, and concerned citizens of Iowa:

IT IS WITH PLEASURE that I provide you with the 2006 report of the Iowa Registry for Congenital and Inherited Disorders. For over two decades, the Registry has worked to advance the health of Iowa children by monitoring the entire state for various congenital and inherited disorders. By providing data for research studies and educational programs, the Registry works for the prevention and treatment of these disorders.

For the second year in a row, the Registry has expanded its mission. This year, we are expanding our activities to include surveillance for stillborn children. Each year there are over 200 stillborn children in the state of Iowa. Despite the emotional toll that accompanies a deceased child, there has been relatively little study on what causes fetal death. Our new activities will allow us to come up with a more accurate estimate of the prevalence of stillborn deliveries, and we will also be able to provide a comprehensive description of these cases. These efforts will hopefully allow researchers to learn more about fetal death. We will also be working with the Iowa Department of Public Health to learn what parents of stillborn children need to get through their difficult times.

The Registry continues its mission to work with researchers as they work to find the causes of various congenital and inherited disorders. For example, Registry data have been used to investigate risk factors for cleft lip and palate and the effect of folic acid fortification on spina bifida. Registry information is also used to determine whether the families of children with muscular dystrophy are getting the resources they need. In addition, we respond to requests from Iowa citizens who have questions about birth defects in their communities. All of this is done with the utmost diligence in maintaining the privacy of the families whose information is contained in the Registry.

Since 1983, the Registry has conducted its efforts on behalf of Iowa families. We are pleased to carry this tradition of service and research into 2006.

Sincerely,

Paul A. Romitti, Ph.D.
Director
Assistant Professor of Epidemiology
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 in Iowa and 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 Registry 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 Registry has collected information for over 42,000 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 also performs surveillance for Duchenne/Becker muscular dystrophy and has identified 68 children with that neuromuscular disease.

RESEARCH
Data collected by the Registry 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 Registry 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 Registry 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 Registry 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 1998 through 2002, a record review of 9,080 infants identified by the Registry’s active surveillance system showed that vital records would have identified only 1,644, 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.

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 12 states and 1 territory that have birth defect registries that meet the criteria for an active, state-wide surveillance system. As shown on the map below, the Iowa Registry for Congenital and Inherited Disorders is the only active surveillance system in the Midwest.
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.

**BIRTH DEFECT RATES IN IOWA, 1998 - 2002**

<table>
<thead>
<tr>
<th>Type of Birth Defect</th>
<th>Rate per 1,000 Live Births</th>
</tr>
</thead>
<tbody>
<tr>
<td>Central Nervous System Brain/Spinal Cord</td>
<td>4.4</td>
</tr>
<tr>
<td>Congenital Infection</td>
<td>0.3</td>
</tr>
<tr>
<td>Congenital Tumors</td>
<td>3.3</td>
</tr>
<tr>
<td>Gastrointestinal (Digestive)</td>
<td>5.7</td>
</tr>
<tr>
<td>Ear</td>
<td>4.9</td>
</tr>
<tr>
<td>Eye</td>
<td>3.7</td>
</tr>
<tr>
<td>Genitourinary (Genital/Urinary)</td>
<td>10.4</td>
</tr>
<tr>
<td>Cardiovascular (Heart)</td>
<td>12.9</td>
</tr>
<tr>
<td>Muscle/Skeletal</td>
<td>14.0</td>
</tr>
<tr>
<td>Oral/Facial</td>
<td>6.0</td>
</tr>
<tr>
<td>Respiratory</td>
<td>1.1</td>
</tr>
<tr>
<td>Skin</td>
<td>4.0</td>
</tr>
<tr>
<td>Syndromes</td>
<td>4.1</td>
</tr>
<tr>
<td>Other*</td>
<td>1.4</td>
</tr>
</tbody>
</table>

*Includes blood, hormonal, and metabolic conditions

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 1998 - 2002. 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 years of age) in such counties.
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 1998 - 2002. For all counties combined, the average rate of occurrence of birth defects for this time period was 48.2 per 1,000 live births.
STILLBIRTH SURVEILLANCE

During the 80th General Assembly of Iowa (legislative session 2004), legislation was introduced that would require the Iowa Department of Public Health to establish a work group to make recommendations for a stillbirth evaluation protocol, and to redesign the Iowa Registry for Congenital and Inherited Disorders to include surveillance of stillbirths. The intent of this legislation is for the Iowa Department of Public Health to devise strategies for the prevention of stillbirths and other congenital or inherited disorders which cause the death and disability of newborns in Iowa.

State Representative Janet Petersen and her husband, Brian Pattinson, had a daughter, Grace Pattinson, who was stillborn in the summer of 2003. Grace’s umbilical cord was compressed, and this restricted the blood flow to her from the placenta. As Representative Petersen and her husband soon found out, there are other babies that are stillborn due to similar circumstances. Through church and social circles, Rep. Petersen was connected with other women who had recently had stillborn children. As they shared their stories, Rep. Petersen, Tiffan Yamen, and Jan Caruthers became determined to work to prevent stillbirths from occurring.

These three women worked with the March of Dimes, the Iowa Department of Public Health, and a network of other families of stillborn infants to draft a bill that Rep. Petersen would introduce in the House. This bill, House File 2362, passed the House and Senate and was signed by Governor Vilsack in May of 2004. Rep. Petersen was also able to secure $26,000 in state funding to support this legislation.

The Iowa Department of Public Health established a work group of experts on stillbirths, newborn health, maternal health, family health, pathology, grief counseling, ultrasounds and radiology, and parents of stillborn infants. The work group met in September of 2004, and has established an evaluation protocol for birthing hospitals to use when caring for stillborn infants and their families. This protocol provides assessment guidelines, recommendations for testing and counseling, and resources for staff and family support.

Representative Petersen, Tiffan Yamen, and Jan Caruthers traveled to Washington D.C. to speak with U.S. Senator Tom Harkin about federal funding for stillbirth evaluation and prevention efforts. Senator Harkin was able to secure an appropriation to the Centers for Disease Control and Prevention earmarked for pilot projects for stillbirth surveillance in Iowa and metropolitan Atlanta, Georgia for federal fiscal year 2006. The Iowa Registry and the Iowa Department of Public Health have already begun program development and implementation of the stillbirth evaluation protocol.

Representative Janet Petersen, Tiffan Yamen, and Jan Caruthers have truly honored their children: Grace Pattinson, Madeline Yamen, and Jayden Caruthers; along with all of the other parents of stillborn infants, by establishing this legislation to support stillbirth prevention programs in Iowa.
MDSTARnet, the Muscular Dystrophy Surveillance Tracking and Research Network, is a program currently active in four states (with a fifth state starting soon). Its goal is to identify all people with childhood-onset Duchenne and Becker muscular dystrophies (DBMD). On behalf of MDSTARnet, the Iowa Registry for Congenital and Inherited Disorders is undertaking surveillance of Iowans born since 1982 with DBMD. This surveillance will consist of identification and ongoing chart review.

The Iowa Registry recently completed its first year of gathering data for this project. Thus far, we have identified 68 cases of DBMD in Iowa. The map to the right illustrates the number of cases in the western, central, and eastern sections of the state. Metropolitan areas tend to have higher numbers of affected people largely because there are more people in those areas. Central Iowa includes several metropolitan communities, making it the most populous section of the state. It is also the section with the most cases of DBMD. If the differences in population are taken into account, there does not appear to be a higher rate of DBMD in metropolitan areas compared to rural areas.

A summary of the current ages of Iowans with DBMD (as of late 2005) is presented in the graph to the right. Fewer people have been identified who are age five years and under as compared to other age categories. This finding is expected as DBMD is often not diagnosed until the child reaches school-age. The middle three age categories are nearly even, demonstrating a steady prevalence of DBMD in the state. Data in the upper age category (21 years to 25 years) is incomplete because MDSTARnet only identifies patients born since 1982. Therefore, all Iowans portrayed in this graph are less than 24 years of age.

As part of MDSTARnet, families of people with DBMD will be asked to take part in interviews with researchers. The purpose of the interview is to gather information related to the disease that might not be found in the medical record. The interview is in the final stages of development, and families will be recruited to participate in early 2006. Iowa families are also being invited to participate in an additional interview designed to identify specific problems, barriers to overcoming those problems, and resources that are particularly helpful.
The Iowa Registry for Congenital and Inherited Disorders provides data for several other 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 Network**  
The National Birth Defects Prevention Network (NBDPN) 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 NBDPN.

**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 over 30 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 defects and women with an unaffected pregnancy are interviewed about their health, diet and lifestyle during pregnancy. 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 Foundation for Facial Reconstruction**  
*Funding Agency: Centers for Disease Control and Prevention*  
The Registry is working with the National Foundation for Facial Reconstruction (NFFR) to investigate quality of care for people with craniofacial anomalies. People in Iowa, Arkansas, and western New York state with craniosynostosis, anotia, microtia, and cleft lip and palate are being asked to participate in this study. Medical records are reviewed to evaluate standard of care, and participants are interviewed about their experience with the medical delivery system.
The Iowa Registry contributes data to a variety of research projects. The following scientific publications have relied in part on Iowa Registry data.


State birth defect surveillance programs like the Iowa Registry provide information that is essential for the proper allocation of public health resources. In addition to state-level data, it is also important to determine nationwide rates for major defects. This information allows for the comparison of rates between individual states and nationwide totals. Nationwide rates are also essential for states that do not have a birth defect monitoring program.

Each year, the National Birth Defects Prevention Network (NBDPN) collects and publishes state prevalence data from birth defect registries across the United States. Recently, the Iowa Registry was one of eleven active surveillance programs that participated in an NBDPN-sponsored study to determine nationwide prevalence rates for 18 major birth defects. These data were published in the January, 2006, issue of the *Morbidity and Mortality Weekly Report*.

Among the 18 conditions examined by this article, the condition with the highest prevalence was orofacial clefts, which was estimated to affect almost 6,800 children annually in the United States. Almost 5,500 children were estimated to be affected with Down syndrome. The least frequently occurring condition that was included in this study was common truncus, a heart defect that affects approximately 330 children each year.
REGISTRY SPOTLIGHT

SUE GORTON

Each week, Registry representatives send dozens of abstracts from the field to the central office at The University of Iowa. The abstracts contain highly technical medical information, and each of them must be individually reviewed to ensure quality control. This takes extensive medical knowledge and an eye for detail. The Registry is very fortunate to have Ms. Sue Gorton filling this role as its clinical editor.

When Ms. Gorton joined the Iowa Registry in 2001, she brought a unique set of skills with her. She had just earned her certification as a Registered Health Information Technician. Prior to that, Ms. Gorton worked for many years as a nurse in a neo-natal intensive care unit. The combination of her understanding of health information systems with her extensive medical knowledge has proven to be quite valuable to the Registry.

After working for the Registry as a field representative for two years, Ms. Gorton moved to her current position in the central office. As clinical editor, she uses her expertise to make sure that all diagnostic information is correct and properly documented and that any new information fits in well with existing Registry records. By making sure that the entire record tells a coherent story, she makes the information useful to those who use it for research and public health initiatives.

“It is very rewarding to know that the information we gather will be used for a good purpose,” says Ms. Gorton, “the best part of my job is knowing that our work is used to help prevent birth defects.”
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.

**IOWA REGISTRY FOR CONGENITAL AND INHERITED DISORDERS**
http://www.public-health.uiowa.edu/rcid

**IOWA**
The University of Iowa and University of Iowa Health Care
http://www.public-health.uiowa.edu/
http://www.medicine.uiowa.edu/
http://www.uihealthcare.com/depts/childrenshospitalofiowa/
http://www.uihealthcare.com/depts/med/pediatrics/
Iowa Department of Public Health
http://www.idph.state.ia.us
IDPH Center for Congenital and Inherited Disorders
http://www.idph.state.ia.us/genetics/
Autism Society of IA
http://www.autismia.org
Arc of Iowa (Persons with Mental Retardation)
http://www.thearc.org
Children’s Miracle Network: Iowa
http://www.uihealthcare.com/depts/childrensmiraclenetwork/
Iowa Compass (information and referral services)
http://www.medicine.uiowa.edu/iowacompass
Iowa Department of Education, Special Education
http://www.state.ia.us/educate
Healthy Families Information and Referral Services
1-800-369-BABY
Virtual Hospital
http://www.vh.org

**UNITED STATES**
Alliance of Genetic Support Groups
http://www.geneticalliance.org
American Academy of Pediatrics
http://www.aap.org
Birth Defect Research for Children, Inc.
http://www.birthdefects.org/
Birth Defects Prevention Legislation Committee
http://www.birthdefectsprevention.org

Child Statistics
http://www.childstats.gov
GeneClinics
http://www.geneclinics.org
HuGE Net
http://www.cdc.gov/genetics/hugenet
KidNeeds.com (information and resource center)
http://www.kidneeds.com
March of Dimes
http://www.modimes.org
MD STARnet
http://www.cdc.gov/ncbddd/duchenne/mdstarnet.htm
Muscular Dystrophy Association of America
http://www.mdausa.org/
National Association for the Education of Young Children
http://www.naeyc.org
National Birth Defects Prevention Network
http://www.nbdpn.org/
National Information Center for Children and Youth with Disabilities
http://www.nichcy.org
National Institutes of Health
http://www.nih.gov
National Organization on Fetal Alcohol Syndrome
http://www.nofas.org
Parent Project for Muscular Dystrophy
http://www.parentprojectmd.org
National Society of Genetic Counselors
http://www.nsgc.org
Organization of Teratology Information Services
http://www.otispregnancy.org/
Phenylketonuria Organization
http://www.pkunews.org
Spina Bifida
http://www.spbaa.org
Teratology Society
http://www.teratology.org
Family Village: Information Center sponsored in part by U. of Wisconsin
http://www.familyvillage.wisc.edu/index.htm (click on library)

**INTERNATIONAL**
The Birth Defects Foundation
http://www.bdfcharity.co.uk
International Clearinghouse for Birth Defects Monitoring Systems
http://www.icbd.org
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 Center for Congenital and Inherited Disorders Advisory Committee

Iowa Regional Genetic Consultation Service

Iowa Board of Regents
March of Dimes Birth Defects Foundation
KID Coalition
ASK Resource Center

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National Institutes of Health

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Centers for Disease Control and Prevention
National Institutes of Health

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