Iowa Registry for Congenital and Inherited Disorders Personnel

Director
Paul A. Romitti, Ph.D.

Clinical Director for Birth Defects
Kim Keppler-Noreuil, M.D.

Clinical Director for Neuromuscular Disorders
Katherine Mathews, M.D.

Clinical Directors for Stillbirths
Roger Williamson, M.D.
Kristi Borowski, M.D.

Program Manager
Kristine Hardin, M.S.

Clinical Editors
Sue Gorton, B.S.N., R.N., R.H.I.T.
Carrie Stephan, M.A., R.N.

Programming Staff
Bill Budelier, M.S.
Florence Foo, M.A.

Surveillance Field Staff
Carrie Fall, B.A.S., R.H.I.T.
Patricia Ganzer Holland
Jacquelyn Loesche, P.A.
Evelyn Redtree, M.S.
Patricia Steen, A.S., R.H.I.T.
Ann Vogt, R.N.

Internal Advisory Committee
James Torner, Ph.D., Chair
Trudy Burns, M.P.H., Ph.D.
Jane Borst, R.N., M.A.
Jeffrey Murray, M.D.
Roger Williamson, M.D., M.S.

Center for Congenital and Inherited Disorders, Iowa Department of Public Health
Kimberly Piper, R.N.C., B.S., C.P.H.
Dear members of the Legislature, health care professionals, and concerned citizens of Iowa:

We are happy to provide you with the 2009 report of the Iowa Registry for Congenital and Inherited Disorders (IRCID). The IRCID has conducted state-wide surveillance for birth defects since 1983, and state-wide surveillance for Duchenne/Becker muscular dystrophy and stillbirths, since 2002 and 2005, respectively. In 2009, the IRCID began surveillance for selected newborn screening disorders.

Nationally, the IRCID is the only active state-wide surveillance program for birth defects in the Midwest and serves as a model program for other states. IRCID staff members are active members of the National Birth Defects Prevention Network (NBDPN), an organization of individuals and programs that conduct birth defect monitoring, research, and prevention.

The IRCID is also a key partner with the Iowa Center of Excellence for Birth Defects Research and Prevention, which is a collaborative enterprise between the College of Public Health, Carver College of Medicine, and the College of Pharmacy at The University of Iowa. Its other partners include the Iowa Department of Public Health, the Iowa Chapter of the March of Dimes, and The University of Iowa Center for Health Effects of Environmental Contamination.

Additionally, the IRCID is a key partner with the Iowa site for the Muscular Dystrophy Surveillance, Tracking, and Research Network, which is a collaborative enterprise between the College of Public Health and Carver College of Medicine at The University of Iowa. Other partners include the Iowa Department of Public Health and the Muscular Dystrophy Association of Iowa.

Our strong state-wide program to monitor congenital and inherited disorders is a valuable resource for the state of Iowa. While taking care to preserve the privacy of families affected by these disorders, the IRCID provides important information to state policy makers and public health professionals. Not only does it provide irreplaceable information, it allows for important research on the causes, prevention, and treatment of such disorders. We are pleased to perform this important work on behalf of the citizens of Iowa.

Sincerely,

Paul A. Romitti, Ph.D.
Director and Associate Professor of Epidemiology
In the United States, the Centers for Disease Control and Prevention (CDC) recognize three types of surveillance systems; each is 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)

The IRCID conducts active surveillance to identify information about congenital and inherited disorders that occur in Iowa and to Iowa residents. The IRCID has collected information for over 46,000 children with various birth defects. This information has been used by health care providers and educators to provide treatment and support services. It is also used by researchers to study risk factors for birth defects and to evaluate treatments for birth defects. The IRCID also conducts surveillance for Duchenne/Becker muscular dystrophy and has identified nearly 100 children with this neuromuscular disease. In addition, the IRCID is collaborating with the Metropolitan Atlanta Congenital Defects Program to develop approaches to active surveillance for stillbirths.

### Surveillance for Birth Defects

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).

The IRCID has traditionally focused on structural birth defects, which typically involve a body part that is missing or malformed. Examples include heart defects, spina bifida, and cleft lip and palate. Starting with 2003 deliveries, the IRCID adopted the recommendations of the National Birth Defects Prevention Network (NBDPN) to focus on a core set of 45 defects (see Table 1). Prior to this change, the IRCID included many ‘minor’ conditions, so this change represents a reduction in the number of conditions that it monitors.
Table 1
Prevalence for birth defects in Iowa, 2002-2006 deliveries

<table>
<thead>
<tr>
<th>Condition</th>
<th>Total</th>
<th>Prevalence Rate †</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Brain/Spinal Cord</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Anencephalus</td>
<td>58</td>
<td>2.99</td>
</tr>
<tr>
<td>Encephalocele</td>
<td>16</td>
<td>0.83</td>
</tr>
<tr>
<td>Hydrocephalus without spina bifida</td>
<td>197</td>
<td>10.16</td>
</tr>
<tr>
<td>Microcephalus</td>
<td>193</td>
<td>9.95</td>
</tr>
<tr>
<td>Spina bifida without anencephalus</td>
<td>95</td>
<td>4.90</td>
</tr>
<tr>
<td><strong>Eye</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Aniridia</td>
<td>1</td>
<td>0.05</td>
</tr>
<tr>
<td>Anophthalmia/microphthalmia</td>
<td>56</td>
<td>2.89</td>
</tr>
<tr>
<td>Congenital cataract</td>
<td>47</td>
<td>2.42</td>
</tr>
<tr>
<td><strong>Ear</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Anotia/microtia</td>
<td>35</td>
<td>1.80</td>
</tr>
<tr>
<td><strong>Heart</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Aortic valve stenosis</td>
<td>75</td>
<td>3.87</td>
</tr>
<tr>
<td>Atrial septal defect</td>
<td>591</td>
<td>30.48</td>
</tr>
<tr>
<td>Coarctation of aorta</td>
<td>88</td>
<td>4.54</td>
</tr>
<tr>
<td>Common truncus</td>
<td>19</td>
<td>0.98</td>
</tr>
<tr>
<td>Ebstein's anomaly</td>
<td>17</td>
<td>0.88</td>
</tr>
<tr>
<td>Endocardial cushion defect</td>
<td>151</td>
<td>7.79</td>
</tr>
<tr>
<td>Hypoplastic left heart syndrome</td>
<td>41</td>
<td>2.11</td>
</tr>
<tr>
<td>Patent ductus arteriosus</td>
<td>545</td>
<td>28.10</td>
</tr>
<tr>
<td>Pulmonary valve atresia and stenosis</td>
<td>192</td>
<td>9.90</td>
</tr>
<tr>
<td>Tetralogy of Fallot</td>
<td>88</td>
<td>4.54</td>
</tr>
<tr>
<td>Transposition of great arteries</td>
<td>66</td>
<td>3.40</td>
</tr>
<tr>
<td>Tricuspid valve atresia and stenosis</td>
<td>23</td>
<td>1.19</td>
</tr>
<tr>
<td>Ventricular septal defect</td>
<td>977</td>
<td>50.38</td>
</tr>
</tbody>
</table>

† Prevalence per 10,000 live births.
Continued on next page...
Table 1 (continued from previous page)

<table>
<thead>
<tr>
<th>Condition</th>
<th>Total</th>
<th>Prevalence Rate †</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Oral/Facial</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Choanal atresia</td>
<td>37</td>
<td>1.91</td>
</tr>
<tr>
<td>Cleft lip with and without cleft palate</td>
<td>248</td>
<td>12.79</td>
</tr>
<tr>
<td>Cleft palate without cleft lip</td>
<td>155</td>
<td>7.99</td>
</tr>
<tr>
<td><strong>Digestive</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Biliary atresia</td>
<td>10</td>
<td>.52</td>
</tr>
<tr>
<td>Esophageal atresia / tracheoesophageal fistula</td>
<td>41</td>
<td>2.11</td>
</tr>
<tr>
<td>Hirschsprung's disease (congenital megacolon)</td>
<td>39</td>
<td>2.01</td>
</tr>
<tr>
<td>Pyloric stenosis</td>
<td>578</td>
<td>29.80</td>
</tr>
<tr>
<td>Rectal and large intestinal atresia/stenosis</td>
<td>95</td>
<td>4.90</td>
</tr>
<tr>
<td><strong>Genital/Urinary</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Bladder extrophy</td>
<td>11</td>
<td>0.57</td>
</tr>
<tr>
<td>Hypospadias and Epispadias</td>
<td>185</td>
<td><strong>18.58</strong></td>
</tr>
<tr>
<td>Obstructive genitourinary defect</td>
<td>528</td>
<td>27.23</td>
</tr>
<tr>
<td>Renal agenesis/hypoplasia</td>
<td>143</td>
<td>7.37</td>
</tr>
<tr>
<td><strong>Muscle/Skeletal</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Congenital hip dislocation</td>
<td>174</td>
<td>8.97</td>
</tr>
<tr>
<td>Diaphragmatic hernia</td>
<td>16</td>
<td>0.83</td>
</tr>
<tr>
<td>Gastrochisis</td>
<td>94</td>
<td>4.85</td>
</tr>
<tr>
<td>Omphalocele</td>
<td>53</td>
<td>2.73</td>
</tr>
<tr>
<td>Reduction deformity, lower limbs</td>
<td>43</td>
<td>2.22</td>
</tr>
<tr>
<td>Reduction deformity, upper limbs</td>
<td>89</td>
<td>4.59</td>
</tr>
<tr>
<td><strong>Syndromes</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Down syndrome (Trisomy 21)</td>
<td>329</td>
<td>16.96</td>
</tr>
<tr>
<td>Edwards syndrome (Trisomy 18)</td>
<td>54</td>
<td>2.78</td>
</tr>
<tr>
<td>Patau syndrome (Trisomy 13)</td>
<td>30</td>
<td>1.55</td>
</tr>
<tr>
<td><strong>Other</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Amniotic bands</td>
<td>18</td>
<td>0.93</td>
</tr>
<tr>
<td>Fetus or newborn affected by maternal alcohol use</td>
<td>8</td>
<td>0.41</td>
</tr>
</tbody>
</table>

† Prevalence rates per 10,000 live births.
* Includes epispadias and/or second or third degree hypospadias. Excludes hypospadias NOS and first degree hypospadias.
** Prevalence per 10,000 male live births.
Birth Defects Research

Approximately 1 in 33 newborns are affected by a major birth defect, making such conditions disturbingly common. These conditions come with personal and monetary costs, both for the families of these children and for society. Nearly 20% of all infant deaths are caused by birth defects. Hospitalizations associated with such conditions are longer than hospitalizations for other conditions. More than $8 billion is required to provide lifetime care for the children born with birth defects each year.

Because the causes of up to 70% of birth defects are unknown, research is a critical part of any strategy to prevent these conditions. For this reason, in 1996 the United States Congress directed the CDC to establish regional “centers of excellence” in birth defect research and prevention. Furthermore, interest in fostering collaboration among state birth defect programs led to the formation of the National Birth Defects Prevention Network in 1998.

National Birth Defects Prevention Network

The National Birth Defects Prevention Network (NBDPN) is a nationwide association of birth defect programs and individuals. The IRCID is an active member of the NBDPN and participates in many of its projects. For example, the NBDPN provides a set of guidelines to help birth defect registries around the country organize their work in a consistent manner. The NBDPN also provides educational materials to birth defect abstractors, as well as informational resources to promote Birth Defects Prevention Month each January. Another goal of the NBDPN is to encourage scientific collaboration among birth defect programs. The IRCID is currently participating in NBDPN projects for biliary atresia, neural tube defects, pyloric stenosis, and ventral wall defects.

2009 NBDPN Publications Using IRCID Data


Iowa Center of Excellence for Birth Defects Research and Prevention

The Iowa Center of Excellence for Birth Defects Research and Prevention was one of eight charter centers established by the CDC to study genetic and environmental (broadly defined) risk factors for birth defects. Iowa Center investigators participate in local (state-wide) projects as well as the National Birth Defects Prevention Study (NBDPS). The NBDPS is a population-based study that investigates genetic and environmental risk factors for over 30 major birth
defects. As a partner with the Iowa Center, the IRCID identifies children with NBDPS-eligible birth defects and secures permission from mothers and guardians to share information with researchers. 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 their pregnancies. Biologic samples are also collected from each family to study genetic factors that may contribute to these birth defects. Presently, over 35,000 interviews have been completed nationwide, and biologic samples have been collected from more than 19,333 families.

Over 200 research projects are currently underway nation-wide as part of the NBDPS. Some of them examine risk factors such as maternal nutrition. Others examine gene and environment interactions. Still others examine maternal behavior during pregnancy. For example, the Iowa Center recently led a project that examined the role of maternal caffeine consumption during pregnancy on the development of neural tube defects.

The research performed by Iowa investigators has the potential to positively affect the lives of Iowans. Current studies by Iowa investigators are focused on the relationships between birth defects and agricultural chemicals, cigarette smoking, alcohol consumption, medications, and compounds in drinking water.

2009 Iowa Center Publications Using IRCID Data
(Names listed in bold designate Iowa investigators)


2009 NBDPS Publications Using IRCID Data
(Names listed in bold designate Iowa investigators)


National Down Syndrome Project

The National Down Syndrome Project (NDSP) is a population-based study to investigate genetic and environmental risk factors for Down syndrome. This study is led by investigators at Emory University and is a combined effort of the Iowa Center and programs in five other states. Iowa women who gave birth to an infant affected with Down syndrome and women with an unaffected birth were interviewed about their health, diet, and lifestyle during pregnancy and biologic samples were collected from each family.

2009 NDSP Publications Using IRCID Data (Names listed in bold designate Iowa investigators)


Muscular Dystrophy Research

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. Approximately 1 in 3,500 boys have DBMD. Girls rarely have the disease, but they can be carriers of the gene mutation. Approximately one-third of boys with Duchenne muscular dystrophy did not inherit the disorder.

The Muscular Dystrophy Surveillance Tracking and Research Network

MD STARnet, the Muscular Dystrophy Surveillance, Tracking and Research Network, is a program currently active in six states. Its goal is to identify all people with childhood-onset Duchenne/Becker muscular dystrophies (DBMD). On behalf of the MD STARnet, the IRCID is undertaking surveillance of Iowans born since 1982 with DBMD. This surveillance consists of identification and ongoing medical chart review.

2009 MD STARnet Publications Using IRCID Data


Acknowledgements

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
- Carver College of Medicine
- College of Nursing
- College of Public Health
- Craniofacial Anomalies Research Center
- Governmental Relations Office
- Hygienic Laboratory
- Iowa Cancer Registry

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

**Registry surveillance activities are funded by:**
State of Iowa through a special appropriation to the Board of Regents
State of Iowa through a fee on issuance of birth certificates
Centers for Disease Control and Prevention

**Registry research activities are funded by:**
Centers for Disease Control and Prevention
National Institutes of Health

**Registry educational activities are funded by:**
Centers for Disease Control and Prevention
National Institutes of Health

Development and publication of this report was supported by funds appropriated by the Iowa General Assembly to the State Board of Regents.
The Iowa Registry for Congenital and Inherited Disorders is a collaborative program of the University of Iowa’s College of Public Health and the Iowa Department of Public Health.