Iowa Registry for Congenital and Inherited Disorders

2007 Research Report
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.

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Kristi Borowski, M.D.

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

I am happy to provide you with the 2007 research report of the Iowa Registry for Congenital and Inherited Disorders (IRCID). The IRCID performs state-wide surveillance for birth defects, stillbirths, and Duchenne/Becker muscular dystrophy. While taking care to preserve the privacy of families affected by these conditions, the Registry provides important information to state policy makers and public health professionals.

The IRCID also works closely with researchers who are focused on identifying the causes for many birth defects, and it is this work that is the focus of this year’s report. The IRCID is a key partner with the Iowa Center of Excellence for Birth Defects Research and Prevention, which is a collaborative enterprise between the Colleges of Medicine and Public Health 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.

There are nine such centers across the country, and each of them, including the Iowa Center, participates in the National Birth Defects Prevention Study (NBDPS). The NBDPS is the largest population-based study ever conducted in the United States to examine the causes of birth defects. The IRCID identifies children that are eligible for this study and secures permission from their parents before any research is performed with their information. Those who choose to join the study participate in research on a wide variety of conditions and risk factors.

The IRCID is also an active member of the National Birth Defects Prevention Network (NBDPN), an organization of individuals and programs that are concerned with birth defect monitoring, research, and prevention. The IRCID often participates in collaborative research projects sponsored by the Network. Recent studies have included a project that used state data to estimate the nation-wide prevalence for selected birth 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, fertility treatments, and compounds in drinking water. An area of focus is the interaction of genetic and environmental factors that might affect risk for cleft lip and palate.

A strong state-wide birth defect monitoring program is a valuable resource for the state of Iowa. Not only does it provide irreplaceable information for state leaders, it allows for important research on the causes and prevention of birth defects. 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 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 44,000 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 performs surveillance for Duchenne/Becker muscular dystrophy and has identified 78 children with that 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). Examples of two major categories of birth defects are 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.

The IRCID has traditionally focused on structural defects rather than metabolic conditions. (The Iowa Neonatal Metabolic Screening Program does an excellent job monitoring for those conditions.) 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, 2000-2004 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>61</td>
<td>3.21</td>
</tr>
<tr>
<td>Encephalocele</td>
<td>16</td>
<td>0.84</td>
</tr>
<tr>
<td>Hydrocephalus without Spina Bifida</td>
<td>202</td>
<td>10.64</td>
</tr>
<tr>
<td>Microcephalus</td>
<td>172</td>
<td>9.06</td>
</tr>
<tr>
<td>Spina bifida without anencephalus</td>
<td>103</td>
<td>5.42</td>
</tr>
<tr>
<td><strong>Eye</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Aniridia</td>
<td>5</td>
<td>0.26</td>
</tr>
<tr>
<td>Anophthalmia/ microphthalmia</td>
<td>50</td>
<td>2.63</td>
</tr>
<tr>
<td>Congenital cataract</td>
<td>43</td>
<td>2.26</td>
</tr>
<tr>
<td><strong>Ear</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Anotia/microtia</td>
<td>66</td>
<td>3.48</td>
</tr>
<tr>
<td><strong>Heart</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Aortic valve stenosis</td>
<td>74</td>
<td>3.90</td>
</tr>
<tr>
<td>Atrial septal defect</td>
<td>533</td>
<td>28.06</td>
</tr>
<tr>
<td>Coarctation of aorta</td>
<td>104</td>
<td>5.48</td>
</tr>
<tr>
<td>Common truncus</td>
<td>23</td>
<td>1.21</td>
</tr>
<tr>
<td>Ebstein's anomaly</td>
<td>23</td>
<td>1.21</td>
</tr>
<tr>
<td>Endocardial cushion defect</td>
<td>121</td>
<td>6.37</td>
</tr>
<tr>
<td>Hypoplastic left heart syndrome</td>
<td>53</td>
<td>2.79</td>
</tr>
<tr>
<td>Patent ductus arteriosus</td>
<td>536</td>
<td>28.22</td>
</tr>
<tr>
<td>Pulmonary valve atresia and stenosis</td>
<td>210</td>
<td>11.06</td>
</tr>
<tr>
<td>Tetralogy of Fallot</td>
<td>64</td>
<td>3.37</td>
</tr>
<tr>
<td>Transposition of great arteries</td>
<td>106</td>
<td>5.58</td>
</tr>
<tr>
<td>Tricuspid valve atresia and stenosis</td>
<td>20</td>
<td>1.05</td>
</tr>
<tr>
<td>Ventricular septal defect</td>
<td>868</td>
<td>45.70</td>
</tr>
</tbody>
</table>

† Prevalence per 10,000 live births.
Continued on next page...
<table>
<thead>
<tr>
<th>Condition</th>
<th>Total</th>
<th>Prevalence Rate&lt;sup&gt;†&lt;/sup&gt;</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Oral/Facial</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Choanal atresia</td>
<td>44</td>
<td>2.32</td>
</tr>
<tr>
<td>Cleft lip with and without cleft palate</td>
<td>241</td>
<td>12.69</td>
</tr>
<tr>
<td>Cleft palate without cleft lip</td>
<td>165</td>
<td>8.69</td>
</tr>
<tr>
<td><strong>Digestive</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Biliary atresia</td>
<td>13</td>
<td>0.68</td>
</tr>
<tr>
<td>Esophageal atresia/ tracheoesophageal fistula</td>
<td>52</td>
<td>2.74</td>
</tr>
<tr>
<td>Hirschsprung's disease (congenital megacolon)</td>
<td>46</td>
<td>2.42</td>
</tr>
<tr>
<td>Pyloric stenosis</td>
<td>627</td>
<td>33.01</td>
</tr>
<tr>
<td>Rectal and large intestinal atresia/stenosis</td>
<td>93</td>
<td>4.90</td>
</tr>
<tr>
<td><strong>Genital/Urinary</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Bladder extrophy</td>
<td>9</td>
<td>0.47</td>
</tr>
<tr>
<td>Hypospadias and Epispadias &lt;sup&gt;*&lt;/sup&gt;</td>
<td>134</td>
<td>7.06</td>
</tr>
<tr>
<td>Obstructive genitourinary defect</td>
<td>527</td>
<td>27.75</td>
</tr>
<tr>
<td>Renal agenesis/hypoplasia</td>
<td>140</td>
<td>7.37</td>
</tr>
<tr>
<td><strong>Muscle/Skeletal</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Congential hip dislocation</td>
<td>161</td>
<td>8.48</td>
</tr>
<tr>
<td>Diaphragmatic hernia</td>
<td>24</td>
<td>1.26</td>
</tr>
<tr>
<td>Gastrochisis</td>
<td>99</td>
<td>5.21</td>
</tr>
<tr>
<td>Omphalocele</td>
<td>49</td>
<td>2.58</td>
</tr>
<tr>
<td>Reduction deformity, lower limbs</td>
<td>54</td>
<td>2.84</td>
</tr>
<tr>
<td>Reduction deformity, upper limbs</td>
<td>106</td>
<td>5.58</td>
</tr>
<tr>
<td><strong>Syndromes</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Down syndrome (Trisomy 21)</td>
<td>301</td>
<td>15.85</td>
</tr>
<tr>
<td>Edwards syndrome (Trisomy 18)</td>
<td>51</td>
<td>2.69</td>
</tr>
<tr>
<td>Patau syndrome (Trisomy 13)</td>
<td>22</td>
<td>1.16</td>
</tr>
<tr>
<td><strong>Other</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Amniotic bands</td>
<td>29</td>
<td>1.53</td>
</tr>
<tr>
<td>Fetus or newborn affected by maternal alcohol use</td>
<td>6</td>
<td>0.32</td>
</tr>
</tbody>
</table>

<sup>†</sup> Prevalence per 10,000 live births.

<sup>*</sup> Includes epispadias and/or second or third degree hypospadias. Excludes hypospadias NOS and first degree hypospadias.
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 registries 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. Recent projects have been focused on such issues as preterm births, ventral wall defects, and neural tube defects. It has also provided national prevalence estimates for selected, common birth defects.

2006-2007 NBDPN Publications Using IRCID Data
(Names listed in bold designate Iowa investigators)


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 and also 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 cases and secures permission from parents 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. At this point, over 25,000 interviews have been completed nationwide, and biologic samples have been collected from more than 13,000 families.

Over 200 research projects are currently underway nation-wide as part of 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 with other centers examining the role of maternal alcohol consumption during pregnancy on the development of cleft lip and palate. The investigators found that the type of alcohol consumed affected the probability that a child would be born with a cleft. Distilled spirits was associated with the greatest risk, followed by wine, and then beer.

2006-2007 Iowa Center Publications Using IRCID Data
(Names listed in bold designate Iowa investigators)


**Warrington A, Vieira AR, Christensen K, Orioli IM, Castilla EE, Romitti PA, Murray JC.**

**2006-2007 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 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.

2006-2007 NDSP Publication Using IRCID Data
(Names listed in bold designate Iowa investigators)

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.

The Muscular Dystrophy Surveillance Tracking and Research Network

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

2006-2007 MDSTARnet Publication Using IRCID Data
(Names listed in bold designate Iowa investigators)

Acknowledgements

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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

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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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Contact Information

Iowa Registry for Congenital and Inherited Disorders
The University of Iowa
M107 Oakdale Hall
Iowa City, IA 52242-5000

Phone: 319-335-4107
Fax: 319-335-4030

E-mail: ircid@uiowa.edu

WWW: http://www.public-health.uiowa.edu/ircid

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.