2014 Report

The Iowa Registry for Congenital and Inherited Disorders (IRCID) continues to be a national leader in surveillance of congenital and inherited disorders and serves as a model program for other states. 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 55,000 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 140 children with this neuromuscular disease. In addition, the IRCID has collaborated with the Metropolitan Atlanta Congenital Defects Program to develop approaches to active surveillance for stillbirths and also newborn screening disorders.

The surveillance and research efforts of the IRCID and its partners provide 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. We are pleased to perform this important work on behalf of the citizens of Iowa.

Surveillance for Birth Defects

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 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. Since 2003, the IRCID adopted the recommendations of the National Birth Defects Prevention Network (NBDPN) to focus largely on a core set of birth 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, 2007-2011 deliveries

Condition	Total	Prevalence Estimate*
Brain/Spinal Cord		
Anencephalus	63	3.2
Encephalocele	20	1.0
Hydrocephalus without spina bifida	251	12.7
Microcephalus	223	11.3
Spina bifida without anencephalus	84	4.3
Eye		
Aniridia	2	0.1
Anophthalmia/microphthalmia	43	2.2
Congenital cataract	54	2.7
Ear		
Anotia/microtia	48	2.4
Heart		
Aortic valve stenosis	59	3.0
Atrial septal defect	610	30.9
Atrioventricular septal defect	125	6.3
Coarctation of aorta	116	5.9
Common truncus	7	0.4
Ebstein's anomaly	21	1.1
Hypoplastic left heart syndrome	55	2.8
Patent ductus arteriosus	523	26.5
Pulmonary valve atresia	24	1.2
Pulmonary valve atresia and stenosis	249	12.6
Tetralogy of Fallot	77	3.9
Total anomalous pulmonary venous return	19	1.0
Transposition of great arteries	71	3.6
dextro-Transposition of great arteries	33	1.7
Tricuspid valve atresia	13	0.7
Tricuspid valve atresia and stenosis	57	2.9
Ventricular septal defect	1115	56.5

* Prevalence per 10,000 live births.
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Table 1 (continued from previous page)

Condition	Total	Prevalence Estimate*
Oral/Facial		
Choanal atresia	30	1.5
Cleft lip with and without cleft palate	200	10.1
Cleft palate without cleft lip	134	6.8
Digestive		
Biliary atresia	9	0.5
Esophageal atresia / tracheoesophageal fistula	51	2.6
Hirschsprung's disease (congenital megacolon)	34	1.7
Pyloric stenosis	444	22.5
Rectal and large intestinal atresia/stenosis	96	4.9
Genital/Urinary		
Bladder exstrophy	3	0.2
Hypospadias and Epispadias	259	25.7
Obstructive genitourinary defect	612	31.0
Renal agenesis/hypoplasia	121	6.1
Muscle/Skeletal		
Congenital hip dislocation	76	3.8
Diaphragmatic hernia	55	2.8
Gastroschisis	121	6.1
Omphalocele	49	2.5
Reduction deformity, lower limbs	40	2.0
Reduction deformity, upper limbs	99	5.0
Syndromes		
Down syndrome (Trisomy 21)	264	13.4
Edwards syndrome (Trisomy 18)	67	3.4
Patau syndrome (Trisomy 13)	33	1.7
Other		
Amniotic bands	25	1.3
Fetus or newborn affected by maternal alcohol use	6	0.3

* 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 is 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. Further, 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, gastroschisis, and pyloric stenosis.

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 participated in local (state-wide) projects as well as the National Birth Defects Prevention Study (NBDPS). The NBDPS was a population-based study that investigated genetic and environmental risk factors for over 30 major birth defects. As a partner with the Iowa Center, the IRCID identified children with NBDPS-eligible birth defects and secured permission from mothers to share information with researchers. Women with a pregnancy affected by one or more of the defects and women with an unaffected pregnancy were interviewed about their health, diet, and lifestyle during their pregnancies. Biological specimens were also requested from each family to study genetic factors that may contribute to these birth defects. Over 43,000 interviews have been completed nationwide, and biological specimens have been collected from more than 25,000 families.

Over 200 research projects are currently underway nation-wide as part of the NBDPS. Some of these projects examine risk factors, such as maternal nutrition. Others examine gene and environment interaction effects. Still others examine maternal behavior during pregnancy.

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.

2014 Iowa Center Publications Using IRCID Data
(Names listed in bold designate Iowa investigators)

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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 STAR*net*, 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/Becker muscular dystrophies (DBMD). On behalf of the MD STAR*net*, the IRCID is conducting surveillance of Iowans born since 1982 with DBMD. This surveillance consists of identification and ongoing medical chart review.

2014 MD STAR*net* Publications Using IRCID Data
(Names listed in bold designate Iowa investigators)

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Iowa Registry for Congenital and Inherited Disorders Personnel

Director
Paul A Romitti, PhD

Clinical Directors for Birth Defects
Thomas Scholz, MD
Oleg Shchelochkov, MD

Clinical Director for Neuromuscular Disorders
Katherine Mathews, MD

Clinical Directors for Stillbirths
Patricia Kirby, MBBCh, M Med, MRCPath, FRCPath

Clinical Editors
Sue Gorton, BSN, RN, RHIT
Kim Martin, RNC

Data Manager
Florence Foo, MA

Field Staff Supervisor
Carrie Fall, BAS, RHIT

Surveillance Field Staff
Leanna Darnold, BA, MT, CIC
Patricia Ganzer Holland
Ann Vogt, RN

Center for Congenital and Inherited Disorders, Iowa Department of Public Health
Kimberly Piper, RNC, BS, CPH

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National Institutes of Health

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Contact Information

Iowa Registry for Congenital and Inherited Disorders
UI Research Park
100 BVC, Room W260-A
Iowa City, IA 52242-5000
Phone: 319-335-4107
Fax: 319-335-4030
E-mail: ircid@uiowa.edu
Website: http://www.public-health.uiowa.edu/ircid