Iowa Registry for Congenital and Inherited Disorders

2024 Report

The Iowa Registry for Congenital and Inherited Disorders (IRCID) continues to be a national leader in surveillance of congenital and inherited disorders. IRCID conducts active surveillance to identify information about congenital and inherited disorders that occur in Iowa and to Iowa residents.

Since 1983, IRCID has collected information for over 64,000 children with various birth defects. This information is used by health care providers and educators to provide treatment and support services, and by researchers to study risk factors for birth defects and evaluate treatments for birth defects.

IRCID also conducts surveillance for muscular dystrophies – Duchenne, Becker, congenital, distal, Emery-Dreifuss, facioscapulohumeral, limb-girdle, myotonic, and oculopharyngeal. In addition, IRCID has collaborated with the Centers for Disease Control and Prevention (CDC) to develop approaches for active surveillance for stillbirths, newborn screening disorders, birth defects that may be related to Zika virus infection, delivery outcomes of pregnant women who tested positive for SARS-CoV-2, and congenital cytomegalovirus infection.

The surveillance and research efforts of 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, 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 (US), CDC recognizes three surveillance approaches, each rated differently for completeness of ascertainment of pregnancies with a birth defect.

- Vital Record Reporting: 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 Reporting: 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).

Approximately 1 in 33 newborns is affected by a major birth defect in the US. Major defects come with cph-webmaster@uiowa.edual and monetary costs for families of these children and for society. Nearly 20% of all infant deaths are caused by major defects. Hospitalizations associated with major defects are longer than those for other conditions and account for about $9 billion annually for infants.

IRCID has traditionally focused on structural birth defects, which involve a body part that is missing or malformed. Examples include heart defects, spina bifida, clubfoot, and cleft lip and palate. Since 2003, IRCID adopted the recommendations of the National Birth Defects Prevention Network (NBDPN) to focus largely on a core set of major birth defects (see Table 1).

Table 1. Prevalence (per 10,000 live births) for birth defects in Iowa, 2017-2021 deliveries

Birth Defect	Total	Prevalence
Brain/Spinal Cord		
Anencephalus	55	2.9
Encephalocele	22	1.2
Holoprosencephaly	47	2.5
Spina bifida without anencephalus	76	4.1
Eye		
Anophthalmia/microphthalmia	30	1.6
Congenital cataract	67	3.6
Ear		
Anotia/microtia	54	2.9
Heart		
Aortic valve stenosis	45	2.4
Atrial septal defect	387	20.7
Atrioventricular septal defect	105	5.6
Coarctation of aorta	128	6.9
Common truncus	13	0.7
Double outlet right ventricle	48	2.6
Ebstein anomaly	16	0.9
Hypoplastic left heart syndrome	48	2.6
Interrupted aortic arch	15	0.8
Pulmonary valve atresia and stenosis	173	9.3
Single ventricle	10	0.5
Tetralogy of Fallot	77	4.1
Total anomalous pulmonary venous return	24	1.3
Transposition of great arteries	50	2.7
Tricuspid valve atresia and stenosis	44	2.4
Ventricular septal defect	943	50.5
Oral/Facial		
Choanal atresia	10	0.5
Cleft lip only	71	3.8
Cleft lip with cleft palate	128	6.9
Cleft palate without cleft lip	139	7.4
Gastrointestinal		
Biliary atresia	5	0.3
Esophageal atresia/tracheoesophageal fistula	49	2.6
Hirschsprung's disease (congenital megacolon)	30	1.6
Pyloric stenosis	275	14.7
Rectal and large intestinal atresia/stenosis	61	3.3
Small intestinal atresia and stenosis	53	2.8
Genital/Urinary		
Bladder exstrophy	6	0.3
Cloacal exstrophy	0	0.0
Congenital posterior urethral valves[†]	21	2.2
Hypospadias[*,†]	571	59.9
Renal agenesis/hypoplasia	127	6.8

Table 1. (continued from previous page)

Birth Defect	Total	Prevalence
Muscle/Skeletal		
Clubfoot	341	18.3
Craniosynostosis	119	6.4
Diaphragmatic hernia	63	3.4
Gastroschisis	54	2.9
Limb deficiencies (reduction defects)	102	5.5
Omphalocele	52	2.8
Syndromes/Chromosomes		
Deletion 22q11.2	30	1.6
Down syndrome (Trisomy 21)	302	16.2
Edwards syndrome (Trisomy 18)	66	3.5
Patau syndrome (Trisomy 13)	30	1.6
Turner syndrome‡	39	4.3

*Includes first-, second-, and third-degree hypospadias.
†Prevalence per 10,000 male live births.
‡Prevalence per 10,000 female live births.

Birth Defect Research

Because the causes of up to 70% of major birth defects that occur are unknown, research is a critical part of any strategy to prevent these defects. In 1996 the US Congress directed CDC to establish regional centers in birth defect research and prevention. The Iowa Center for Birth Defects Research and Prevention (CBDRP) is one of eight centers initially established by CDC to study risk factors for major defects. Interest in fostering collaboration among state birth defect programs also led to the establishment of NBDPN in 1998.

National Birth Defects Prevention Network (NBDPN)

IRCID is an active member of NBDPN, a nationwide association of birth defect surveillance programs. NBDPN provides programs with guidelines to organize their work in a consistent manner and with educational materials and informational resources to promote Birth Defects Prevention Month each January. NBDPN also encourages scientific collaboration among surveillance programs.

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Iowa Center for Birth Defects Research and Prevention (CBDRP)

The Iowa CBDRP participated in the National Birth Defects Prevention Study (NBDPS) and currently participates in the Birth Defects Study To Evaluate Pregnancy exposureS (BD-STEPS). NBDPS investigated risk factors for over 30 major defects. IRCID identified children with NBDPS-eligible defects and secured permission from mothers to share information with researchers. Mothers with a pregnancy affected by a major defect and those with an unaffected pregnancy were interviewed about their health, diet, and lifestyle during pregnancy. Biological specimens were requested from families to study genetic factors. Nationwide, over 43,000 interviews were completed, and over 25,000 families provided specimens.

NBDPS and BD-STEPS projects conducted by the Iowa CBDRP have the potential to positively impact the lives of Iowans. These projects examined agricultural chemicals, cigarette smoking, alcohol consumption, diet, medications, and compounds in drinking water, along with genetic factors. Projects published in 2024 that used IRCID data are listed below. Bolded names refer to Iowa investigators.

Iowa NBDPS Project Spotlight

Neural tube defects (NTDs) are characterized by abnormal closure of the neural tube or abnormal formation of the brain and spinal cord from the neural tube during embryogenesis. Both genetic and non-genetic risk factors are thought to contribute to development of NTDs. Disinfection by-products (DBPs) are common contaminants formed during the water disinfection process when common disinfectants, such as chlorine, react with natural organic matter in water. Researchers analyzed NBDPS data to study whether maternal early pregnancy exposure to DBPs in drinking water was related to NTDs in offspring. Researchers observed that NTDs were not increased among mothers with higher exposure levels to DBPs compared to those with lower exposure levels.

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Surveillance for Muscular Dystrophy

Muscular dystrophies (MDs) are a group of genetic progressive muscle diseases affecting an estimated 33 per 100,000 individuals and are characterized by worsening muscle weakness. Historically, types of MDs were diagnosed by known changes in muscle and clinical presentation; presently, diagnosis is determined largely by genetic analysis. Ages at symptom onset of MDs can range from birth through late adulthood. In children, Duchenne is the most common childhood MD, followed by congenital MDs. In adults, myotonic dystrophy is the most common MD, followed by facioscapulohumeral MD.

Muscular Dystrophy Surveillance Tracking and Research Network (MD STARnet)

MD STAR*net* is a surveillance program currently active in seven states (Florida, Iowa, New York, North Carolina, South Carolina, Utah, Virginia) and funded by CDC. The goals of MD STAR*net* are to define and describe the MD population in the US, define and describe healthcare needs and outcomes for

individuals living with MD, and collect information to guide MD care, treatment, and policy. On behalf of MD STAR*net*, IRCID conducts surveillance of Iowans who have been diagnosed with one of eight MDs and meet residence, diagnostic, and treatment period criteria (Table 2). Our surveillance consists of identification and ongoing medical chart review to identify individuals with at least one eligible MD diagnostic code (International Classification of Disease [ICD], ICD-9, ICD-10). The table below summarizes the number of Iowa individuals identified for MD STAR*net,* followed by MD STARnet projects published in 2024. Bolded names refer to Iowa investigators.

Table 2. Number of individuals identified with a muscular dystrophy among Iowa residents

Phase of Surveillance/Muscular Dystrophy	Total
Phase I*	
Duchenne or Becker	140
Phase II†	
Becker	52
Congenital	24
Distal	5
Duchenne	105
Emery-Dreifuss	12
Facioscapulohumeral	81
Limb-Girdle	66
Myotonic	253
Oculopharyngeal	17
Phases III and IV	
Becker‡	30
Congenital^	35
Distal^	8
Duchenne‡	79
Emery-Dreifuss^	20
Facioscapulohumeral^	131
Limb-Girdle^	142
Myotonic^	419
Oculopharyngeal§	37

*Resident individual with MD diagnosis born on or after January 1, 1982 through December 31, 2011 who lived in Arizona, Colorado, Georgia, Hawaii, Iowa, or western New York.

†Resident individual with MD diagnosis and health encounter from January 1, 2007 through December 31, 2011 who lived in Arizona, Colorado, Iowa, or western New York.

‡Phase III: Resident individual with MD diagnosis born on or after January 1, 2000 and health encounter from January 1, 2000 through December 31, 2015 who lived in Colorado, Iowa, western New York, North Carolina, South Carolina, or Utah.
Phase IV: Resident individual with MD diagnosis born on or after January 1, 2000 and health encounter from January 1, 2000 through December 31, 2020 who lived in Florida, Iowa, western New York, North Carolina, South Carolina, Utah, or Virginia.

^Phase III: Resident individual with MD diagnosis since January 1, 2008 and health encounter from January 1, 2008 through December 31, 2016 who lived in Colorado, Iowa, western New York, North Carolina, South Carolina, or Utah.
Phase IV: Resident individual with MD diagnosis and health encounter from January 1, 2008 through December 31, 2020 who lived in Florida, Iowa, western New York, North Carolina, South Carolina, Utah, or Virginia.

§Phase III: Resident individual with MD diagnosis and health encounter from January 1, 2006 through December 31, 2016 who lived in Colorado, Iowa, western New York, North Carolina, South Carolina, or Utah.

Iowa MD STARnet Project Spotlight

Using MD STAR*net* data, researchers described respiratory testing and insufficiency among people with facioscapulohumeral muscular dystrophy (FSHD) diagnosed during 2008-2016. Frequencies and proportions for selected outpatient respiratory assessments and abnormal test results were calculated. Frequencies were examined by disease characteristics (FSHD type, ages of onset, non-ambulatory status, scoliosis, lordosis), obesity, and number of health encounters. Frequency of evaluations and respiratory insufficiency were higher among those with known risk factors and longer follow-up. Researchers observed low proportions of respiratory testing among all confirmed cases of FSHD, but relatively high proportions of mild respiratory insufficiency among those tested. The higher proportions of testing among people with conditions that increase risk of respiratory complications suggest targeted monitoring. Broad implementation of the FSHD guidelines recommending all individuals receive baseline respiratory evaluation at diagnosis could identify respiratory insufficiency as a complication of FSHD.

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Surveillance for Emerging Threats to Mothers and Babies Network (SET-NET)

SET-NET aims to understand effects of emerging and reemerging threats on pregnant women and their infants. To accomplish this, surveillance programs participating in SET-NET work to detect the effects of these threats by collecting data from pregnancy through childhood and use these data to inform clinical decision-making and public health action. IRCID continues to participate in national projects led by CDC.

Microcephaly and Other Birth Defects Related to Zika Virus Exposure

Congenital microcephaly (MC) is a serious birth defect characterized by an abnormally small head size in affected infants compared to infants of the same sex and gestational age. A dramatic increase in MC in infants in Brazil was linked to pregnant women infected with Zika virus. Zika virus exposure poses a serious risk to an unborn fetus; thus, more timely surveillance is needed for monitoring MC and other birth defects that may be related to Zika virus exposure among pregnant women. To conduct this surveillance, IRCID created a rapid response team comprised of experienced surveillance professionals.

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Outcomes Related to SARS-CoV-2 Infection among Pregnant Women

In 2021, IRCID joined the CDC SET-NET to study outcomes for pregnant women infected by the SARS-CoV-2 virus and their offspring. The initial focus of this work is to conduct statewide surveillance of birth outcomes among pregnant women with a laboratory-confirmed SARS-CoV-2 infection in 2020. To date, IRCID has identified more than 3,000 deliveries among pregnant women in Iowa with SARS-CoV-2 infection during pregnancy.

Outcomes Related to Cytomegalovirus Infection among Pregnant Women

In 2023, IRCID expanded its surveillance of emerging threats by studying outcomes related to cytomegalovirus infection (CMV) among pregnant women. To conduct this surveillance the Iowa Department of Health and Human Services issued a reporting order making CMV a reportable infection in Iowa beginning in September 2023. IRCID uses data on CMV infection to identify infants diagnosed with congenital CMV in the first weeks of life.

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