Iowa Registry for Congenital and Inherited Disorders

2025 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 healthcare 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 personal 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, 2018-2022 deliveries

Birth Defect	Total	Prevalence
Brain/Spinal Cord		
Anencephalus	51	2.8
Encephalocele	28	1.5
Holoprosencephaly	44	2.4
Spina bifida without anencephalus	76	4.1
Eye		
Anophthalmia/microphthalmia	31	1.7
Congenital cataract	59	3.2
Ear		
Anotia/microtia	43	2.3
Heart		
Aortic valve stenosis	42	2.3
Atrial septal defect	371	20.2
Atrioventricular septal defect	111	6.0
Coarctation of aorta	118	6.4
Common truncus	15	0.8
Double outlet right ventricle	47	2.6
Ebstein anomaly	13	0.7
Hypoplastic left heart syndrome	39	2.1
Interrupted aortic arch	13	0.7
Pulmonary valve atresia and stenosis	162	8.8
Single ventricle	9	0.5
Tetralogy of Fallot	84	4.6
Total anomalous pulmonary venous return	26	1.4
Transposition of great arteries	51	2.8
Tricuspid valve atresia and stenosis	45	2.4
Ventricular septal defect	892	48.4
Oral/Facial		
Choanal atresia	9	0.5
Cleft lip only	70	3.8
Cleft lip with cleft palate	111	6.0
Cleft palate without cleft lip	127	6.9
Gastrointestinal		
Biliary atresia	8	0.4
Esophageal atresia/tracheoesophageal fistula	49	2.7
Pyloric stenosis	262	14.2
Rectal and large intestinal atresia/stenosis	62	3.4
Small intestinal atresia and stenosis	46	2.5
Genital/Urinary		
Bladder exstrophy	4	0.2
Cloacal exstrophy	2	0.1
Congenital posterior urethral valves[†]	21	2.2
Hypospadias[*,†]	513	54.5
Renal agenesis/hypoplasia	109	5.9

Table 1. (continued from previous page)

Birth Defect	Total	Prevalence
Muscle/Skeletal		
Clubfoot	358	19.4
Craniosynostosis	111	6.0
Diaphragmatic hernia	68	3.7
Gastroschisis	55	3.0
Limb deficiencies (reduction defects)	95	5.2
Omphalocele	52	2.8
Syndromes/Chromosomes		
Deletion 22q11.2	42	2.3
Down syndrome (Trisomy 21)	301	16.3
Edwards syndrome (Trisomy 18)	65	3.5
Patau syndrome (Trisomy 13)	25	1.4
Turner syndrome‡	36	4.0

*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 birth defect centers. The Iowa Center for Birth Defects Research and Prevention (CBDRP) is one of the centers 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 a 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.

Iowa Center for Birth Defects Research and Prevention (CBDRP)

The Iowa CBDRP participated in the National Birth Defects Prevention Study (NBDPS) and the Birth Defects Study To Evaluate Pregnancy exposureS (BD-STEPS). IRCID identified children with NBDPS- and BD-STEPS-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.

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 2025 that used IRCID data are listed below. Bolded names refer to Iowa investigators.

Iowa CBDRP Project Spotlight

Influenza (flu) vaccination during pregnancy helps to reduce flu-related hospitalizations of pregnant women and of infants less than 6 months of age. However, current information on flu vaccination during pregnancy and the risk of birth defects is limited. Using BD-STEPS data, researchers studied whether use of inactivated flu vaccine in early pregnancy was related to having a baby with one of 14 birth defects. Among BD-STEPS participants who reported receiving the vaccine in early pregnancy, researchers did not find a relationship between vaccination and the birth defects, when compared with women who did not receive the vaccine in early pregnancy. This study supports use of inactivated flu vaccine during early pregnancy as recommended by the World Health Organization, United States Advisory Committee on Immunization Practices, and the American College of Obstetricians and Gynecologists.

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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 six (Florida, Iowa, New York, North Carolina, South Carolina, Utah) 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 STAR*net* projects published in 2025. 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

Table 2. (continued from previous page)

Phase of Surveillance/Muscular Dystrophy	Total
Phase V[€]	
Facioscapulohumeral	167

[*]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.

[€]Phase V: Resident individual with MD diagnosis and health encounter from January 1, 2015 through December 31, 2025 who lived in Colorado, Iowa, western New York, North Carolina, South Carolina, or Utah.

Iowa MD STAR*net* Project Spotlight

Using MD STAR*net* data, investigators described pain experiences and pain medications prescribed for individuals with one of several MDs, Duchenne and Becker, congenital, distal, Emery-Dreifuss, facioscapulohumeral, limb-girdle or myotonic. Percentages of individuals prescribed pain medications for at least 6 weeks were estimated. Researchers observed reports of pain varied from 13-53% depending upon MD type. Pain medications were more often prescribed for individuals aged 20 years and older (31-40%) than those aged less than 20 years (<15%). Among individuals prescribed pain medications, the first medication was typically a non-opioid. Additional medications prescribed included non-opioid and opioid medications. Pain medication is commonly prescribed for individuals with symptomatic MD with most prescribed only non-opioids. These data highlight pain management as a frequent component of MD care.

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

SET-NET aims to understand effects of emerging threats on pregnant women and their infants. To accomplish this, IRCID and other 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.

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.

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. Since September 2023, IRCID has identified over 50 infants diagnosed with congenital CMV.

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