Congenital and Inherited Disorders Advisory Committee

Member Manual

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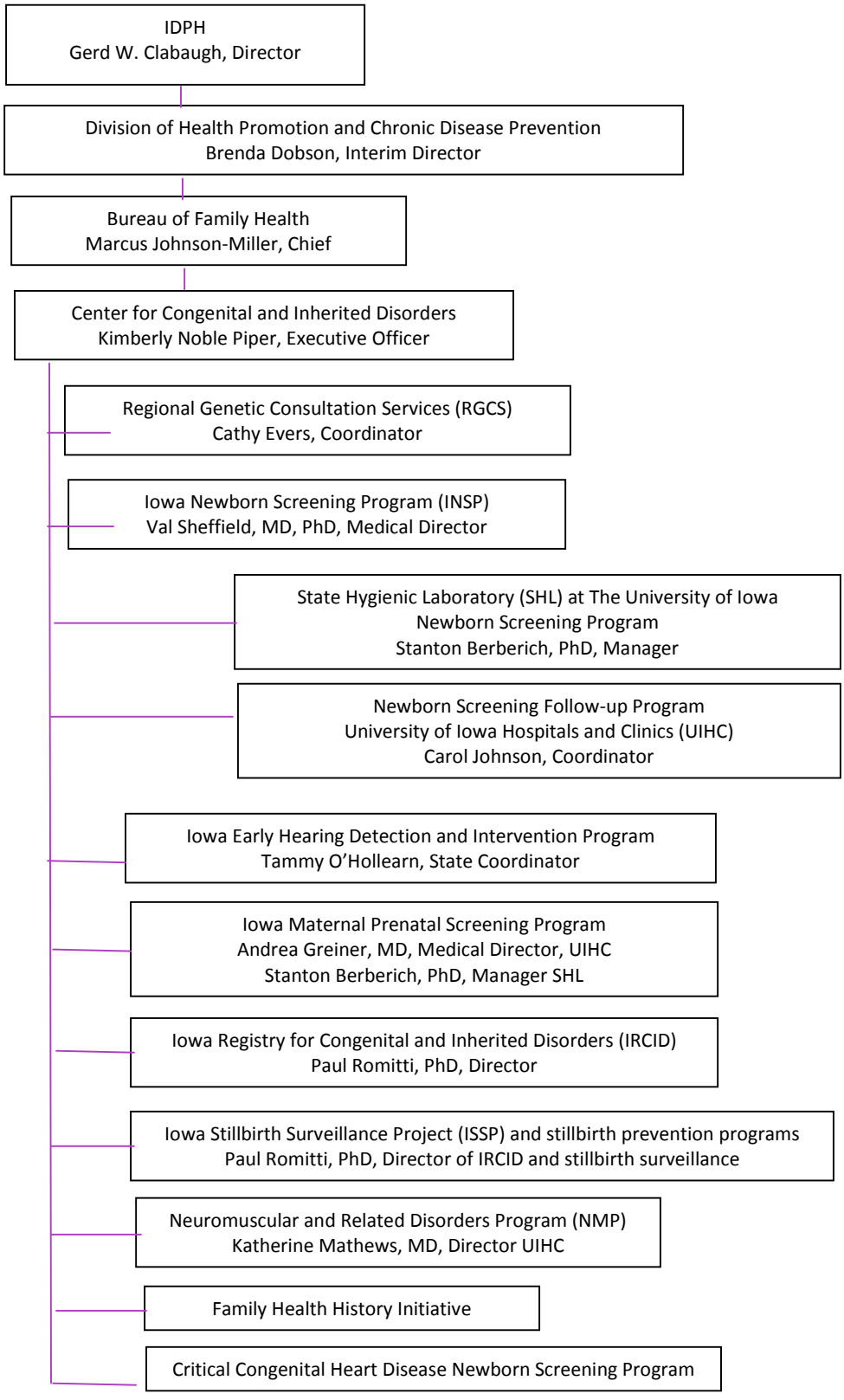
Iowa legislation mandates that the Iowa Department of Public Health develop and administer the state's policy with respect to the conduct of scientific investigations and research concerning the causes, prevention, treatment, and cure of birth defects. The Center for Congenital and Inherited Disorders (formerly called the Birth Defects Institute) was established within the department in 1976 to initiate, conduct, and supervise genetic investigations and research in order to provide for the protection and promotion of the health of Iowans. Since its creation, the Center for Congenital and Inherited Disorders, in partnership with the University of Iowa and health care providers throughout the state, have developed programs that have provided Iowa with state of the art genetics health care. The programs of the Center for Congenital and Inherited Disorders address all steps of the life cycle: preconception, prenatal, neonatal, pediatric and adult.

Established within the Center for Congenital and Inherited Disorders is the Congenital and Inherited Disorders Advisory Committee (CIDAC). The Center for Congenital and Inherited Disorders Advisory Committee represents the interests of the people of Iowa and assists in the development of programs that ensure the availability of and access to quality genetic and genomic health care services by all residents. The committee advises the director of the department of public health regarding issues related to genetics and hereditary and congenital disorders and makes recommendations about the design and implementation of the center’s programs.

The Executive Officer of CCID is the administrator of CCID programs on behalf of IDPH. The Executive Officer is also the designated State Genetics Coordinator (SGC).

CCID Organizational Chart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Neuromuscular and Related Disorders Program (NMP)
Katherine Mathews, MD, Director UIHC
Family Health History Initiative
Critical Congenital Heart Disease Newborn Screening Program

CCID Programming

There are nine programs within the Center for Congenital and Inherited Disorders which include the:

- Regional Genetic Consultation Service (RGCS)
- Iowa Newborn Screening Program (INSP)
- Iowa Early Hearing Detection and Intervention (EHDI) program
- Iowa Newborn Screening for Critical Congenital Heart Disease program
- Iowa Maternal Prenatal Screening Program (IMPSP)
- Iowa Registry for Congenital and Inherited Disorders (IRCID)
- Iowa Stillbirth Surveillance Project (ISSP) and stillbirth prevention programs
- Neuromuscular and Related Genetic Disorders Program (NMP)
- Family Health History Initiative

CCID also monitors and provides information about other population-based genetic services in the state, nationally, and internationally.

Regional Genetics Consultation Services

The University of Iowa Hospitals and Clinics Regional Genetics Consultation Service outreach clinics provide comprehensive, family centered, genetic health care to individuals and families throughout Iowa with statewide outreach clinics. This service has been in existence since 1976. The primary purpose of the RGCS is to provide this structure as an integral component of the state's health-care system. The secondary purpose is to assure the provision of statewide genetics education to promote health and prevent disease.

What services are provided?

- Diagnostic evaluations
- Confirmatory testing
- Medical management
- Provision of non-directive counseling to individuals and families
- Case management
- Individual and family support
- Education
- Consultation
- Referral

These services are provided to over 600 clients and 1,500 family members at 60 clinics in eight communities annually.

What is the cost for the service?

Some insurance companies reimburse for genetic consultation. Third party payers will be billed if a family has coverage. Laboratory procedures, if needed, will be billed separately.

Where are the clinics located?

Ames, Bettendorf, Des Moines, Dubuque, Fort Dodge, Iowa City, Mason City and Waterloo.

Iowa Newborn Screening Program

Newborn screening, also called newborn dried blood spot screening or heel stick screening, is a blood test that screens for certain treatable inherited disorders performed shortly after a baby's birth. Currently, Iowa law requires all newborns and infants born in the state of Iowa will be screened for the following:

- Biotinidase
- Congenital adrenal hyperplasia
- Congenital hypothyroidism
- Cystic fibrosis
- Expanded Panel Disorders
- Galactosemia
- Hemoglobinopathies
- SCID

The Iowa Newborn Screening Program (INSP) identifies babies who may have one of these disorders and alerts the baby's health care provider to the need for further testing and special care. With early diagnosis and treatment, complications from these serious disorders can usually be prevented.

How is the baby screened?

Birthing providers collect a small sample of blood from the baby (usually by doing a "heel stick"), and put the drops of blood on a special filter paper. The paper is then dried and sent to the state designated central testing laboratory - the State Hygienic Laboratory at the University of Iowa (SHL). Iowa enlists a courier service to pick up the filter paper collection forms and deliver to the SHL on the same day. The SHL screens the DBS specimen for over 50 disorders that can cause serious health problems like illness, physical disability, mental retardation, or even death if left untreated.

How are the results communicated?

Parents are usually **not** notified if the screen results are in a normal range. The health care provider will be informed when the screening results are completed. Generally, parents are notified only if rescreening is needed.

If rescreening is required, it should be done as soon as possible. Rescreening does not mean there is anything wrong with the baby. It

simply means that another sample must be obtained so that the complete set of screening tests can be performed. Some reasons for rescreening may include:

- Unsatisfactory specimens: There was something wrong with the sample and it needs to be recollected.
- Early collection: The sample was drawn before the baby was 24 hours old.
- Transfused: The baby received a blood transfusion before the sample was collected. Blood transfusions can lead to inaccurate screening results.
- Abnormal test result: An abnormal test result means there "may" be a disorder present. The doctor will work with the newborn screening program staff to determine if further evaluation is needed.

Many of these disorders cause irreversible damage, even death, in a short period of time, so immediate action is necessary.

Can parents refuse the screening?

Parents may refuse screening. If they refuse, they must inform the health care provider and accept the legal responsibility for the consequences of this decision, and sign a newborn screening refusal form. The health care provider or birthing hospital will notify the newborn screening program that the parents have waived the screen.

What is the cost of the screening?

The cost of newborn screening is established in Iowa law. The current fee is $122, and includes expenses for equipment, supplies and staff of the laboratory, a courier service that delivers the samples to the laboratory for same-day testing, and professional follow-up staff that include nurses and physician specialists; all to address over 50 heritable conditions. This fee is usually covered by in the hospital bill (included in the global newborn care fee) and will most likely be covered by the insurance company. There is no charge for a rescreen sample if one needs to be collected.

Storage and Use of Residual Newborn Screening Dried Blood Specimens

After the screening tests are done, there is often a very small amount of dried blood spot (DBS) left on the cards. Iowa law authorizes the SHL to store these residual, or leftover, DBS for a period of five years in a locked secure, facility at the SHL newborn screening lab in Ankeny, Iowa. The residual DBS specimens are stored for the first year at sub-zero temperatures, and then the remaining four years at room temperature in a locked, secure storage area. The specimens are destroyed after this time. All identifying information is stored separately from the residual DBS in an access protected data system. A unique identification number is used to link the identifying information to the DBS form if necessary.

Iowa stores leftover DBS for several reasons. To make sure the SHL maintains the highest quality of operations, samples of the residual

specimens are used for quality control testing. Quality control testing is a process to monitor the quality of testing and the accuracy of results. The baby's name and date of birth are not revealed when doing quality control testing. The SHL wants to be sure that the testing done one month gives the same result as the testing done the next month.

When testing begins for a new disease or when an improved testing method starts in the lab, professional staff needs to make sure the new test is running properly. This practice needs to be done on the same type of specimens that the testing will be done on, so all practice testing must be done on dried blood spots from newborns. The babies' names and dates of birth are not revealed when the dried blood spots are used for this testing.

In cases where a child later develops health problems, a parent or healthcare provider might request a repeat or other health-related testing on the residual specimen.

In some cases, results or samples are requested by the family or the baby's healthcare team:

- The baby's results can be repeated if needed without getting another blood sample from the baby
- The baby's sample is available for other health-related testing
- The baby's sample is available to help identify a missing or deceased child.
- Storage provides a record that the IDPH completed the screening.

Dried blood spots can help Iowa babies in other ways unrelated to newborn screening. If an infant dies without a cause of death being confirmed, and treatment prior to death (for example, blood transfusion) precludes evaluation, the infant's dried blood spots may be requested. For the purpose of closure in explaining the cause of the infant's death and providing risk information to the family for future pregnancies, a physician or genetic counselor may request a newborn specimen for diagnostic testing.

Throughout the world, dried blood spots are used to examine public health issues like childhood cancer, type I diabetes, lead poisoning, cytomegalovirus infection, and HIV/AIDS and other disorders affecting children.

Public health studies and research may be done only if the researchers follow these guidelines:

- The project must be done to help develop a new newborn screening test or to better understand diseases for the benefit of the general public.
- The baby's name and any identifying details about the baby are removed before the sample is provided.
- The project must be approved by the Institutional Review Board (IRB) where the researcher works to make sure it meets high ethical standards and to ensure that the privacy and safety of the babies is fully protected.

- The project must be approved by the Congenital and Inherited Disorders Advisory Committee to make sure it meets high ethical standards and to ensure that the privacy and safety of the babies is fully protected.
- The project must be approved by the Research and Ethics Review Committee (RERC) at the Iowa Department of Public Health to make sure all privacy and confidentiality laws are being followed.

Any researchers requesting residual DBS specimens with identifying information must go through the above channels, and in addition must first obtain informed, written consent from the individual, parent or guardian before the SHL will release any specimens.

Iowa Early Hearing Detection and Intervention (EHDI) Program

Iowa's Early Hearing Detection and Intervention Program works to ensure that all newborns and toddlers with hearing loss are identified as early as possible and provided with timely and appropriate audiological, educational, medical intervention and family support. The EHDI program is also dedicated to providing unbiased support to families of children who are deaf or hard of hearing.

Based on national statistics, we would expect approximately 120 Iowa children (out of approximately 40,000 births per year) to be born with a permanent hearing loss. Studies show that children who have a hearing loss can have delays in speech, language, cognitive development (thinking) and social and emotional development. If identification of hearing loss does not happen until after six months of age, a child's language skills at age three will be about half those of a child with normal hearing.

Given the serious ramifications of late identification of hearing loss, it is important to perform newborn hearing screening and as soon as possible make diagnostic referrals for infants who do not pass the screening. Because of this, Iowa legislature passed a law (Iowa Code section 135.131), which requires universal hearing screening of all newborns and infants in Iowa. The law further provides that any birthing hospital, birth center, physician, any facility including Area Education Agencies (AEAs), audiologists and other health care professional are legally required to report to the Iowa Department of Public Health (IDPH) the results of a hearing screen, re-screen, or diagnostic assessment for any child under three years of age.

Iowa’s Early Hearing Detection and Intervention Program Goal/Services:

- To develop and sustain a comprehensive coordinated system of care for Early Hearing Detection and intervention in Iowa.
- Provide technical assistance to birthing hospitals, Area Education Agencies, and private practice audiologists relative to the hearing screening program and their responsibility under the law.

- Statewide implementation of a Web-based surveillance system to assure all newborns are screened for hearing loss and receive follow up services, as needed.
- Facilitate data integration linkages with related screening, tracking, and surveillance programs to minimize infants “lost to follow-up”.
- Meet the National EHDI Goal of 1-3-6:

  "1" - All infants are screened for hearing loss before 1 month of age, preferably before hospital discharge.

  "3" - All infants who do not pass the screening will have a diagnostic audiologic evaluation before 3 months of age.

  "6" - All infants identified with a hearing loss receive appropriate early intervention services before 6 months of age.

- Review data to identify children with potential for hearing loss to ensure those children receive appropriate, timely early intervention services.
- Collaborate with Individuals with Disabilities Education Act, Part C (Early ACCESS) to strengthen early intervention services for children who are deaf or hard-of-hearing.
- Ensure families with children zero to three who are deaf, hard-of-hearing, or at risk of late-onset hearing loss will be linked to a medical home and receive family-to-family support.

Every Iowa newborn receives a hearing screen prior to hospital discharge. Results are reported to the EHDI data system. Any missed screen or abnormal screen results are monitored by the hearing screening follow-up staff at IDPH. EHDI follow-up staff work with the hospitals, health care providers, Iowa’s early intervention program – “Iowa Early Access,” and private audiologists and ENT providers to ensure the newborn receives timely screening, appropriate referrals, and early intervention. EHDI follow-up staff make approximately 300 calls per month to guarantee the baby and family receive comprehensive services.

Iowa Maternal Prenatal Screening Program (IMPSP)

The Iowa Maternal Prenatal Screen is a test available to all women during pregnancy. This screening test is designed to identify women with an increased risk to have a baby with Down syndrome, Trisomy 18, or an open neural tube defect. The screen may also identify women with an increased risk to have a baby with other kinds of birth defects or women at risk to develop a problem later in pregnancy. The Iowa MPSP is the first in a series of tests that may be offered. It cannot directly diagnose birth defects and chromosome abnormalities, but can help decide when other tests such as an ultrasound and

amniocentesis may be of value. The Iowa Integrated Screen is performed in two stages. Stage One involves an ultrasound and a blood sample and is done in the first trimester of pregnancy. Stage Two involves a second blood sample drawn between 15-20 weeks of pregnancy.

A screen-positive result does not necessarily mean that your baby has Down syndrome, Trisomy 18 or an open neural tube defect, only that there is an increased chance for one of these problems.

A screen-negative result provides reassurance but does not guarantee that your baby is healthy, only that there is a decreased chance for Down syndrome, Trisomy 18 and open neural tube defects.

If the Iowa Integrated Screen is screen-positive, the patient is offered an ultrasound and may be offered a diagnostic test such as an amniocentesis to find out if the developing baby has a chromosome abnormality or birth defect.

If the patient is too far along for Stage One of the IMPS, a result may still be calculated using Stage Two only (the Quad Screen).

If the patient misses Stage Two of the IMPS, a result may still be calculated using the information from Stage One, provided that the nuchal translucency (NT) measurement was done (the First Trimester Screen). Stage One alone cannot screen for open neural tube defects.

Iowa's integrated Maternal Prenatal Screening is a more effective screen for Down syndrome and Trisomy 18 than either Stage One or Stage Two alone.

State law requires that all maternal prenatal screening testing be conducted by the designated central testing laboratory - the State Hygienic Laboratory.

Unless the person being tested specifically prohibits such use in writing, the specimen and information obtained during the testing process may be used for program evaluation or research by the Iowa Department of Public Health or Department-approved scientific researchers to improve the health of mothers and children. An example of such research would be finding a blood test or tests which may predict the birth of a premature or growth retarded baby. Such studies are published without identifying the person or persons from whom these results were obtained. There are no known risks from being in these studies, and the person being tested will not benefit personally from participating in this research. However, others may benefit in the future from what we learn as results of these studies. The State Hygienic Laboratory stores residual maternal serum specimens at its laboratory in a locked, secure storage area.

Iowa Registry for Congenital and Inherited Disorders

The Registry for Congenital and Inherited Disorders is the reporting system to identify and monitor birth defects in the state of Iowa. Birth defects are reportable conditions in Iowa and the records of these birth defects are abstracted and maintained pursuant to 641 Iowa Administrative Code, Chapter 4.7(136A). The registry (formerly called the Birth Defects Registry) was established in 1983 through the joint efforts of the University of Iowa, the Iowa Department of Public Health, and the Iowa Department of Human Services. The program is located within the College of Public Health at the University of Iowa.

The mission of the Iowa Registry for Congenital and Inherited Disorders is to:

- maintain statewide surveillance for collecting information on birth defect occurrence;
- monitor annual trends in birth defect occurrence and mortality;
- conduct research to identify genetic and environmental risk factors for birth defects; and,
- promote educational activities for the prevention of birth defects.

The registry uses multiple sources to identify information about infants born with birth defects. Trained personnel review medical records at all Iowa hospitals and specialty clinics. Information such as diagnosis, prenatal exposures, prenatal testing, and family history is collected to help monitor trends and investigate causes of birth defects. Infants born in Iowa on or after January 1, 1983, who have specific birth defects diagnosed at birth or within the first year of life are included on the registry. Stillborn infants with specific birth defects are also registered. The registry only includes infants of mothers who were Iowa residents at the time of their birth.

Confidentiality is rigorously maintained so the rights and welfare of individuals included in the registry are not jeopardized.

Parents of children born after January 1, 2003, who are identified by IRCID, receive a notification letter and resource information within six months of being identified by the Registry. The child's primary care doctor also receives a copy of the notification letter.

The registry provides valuable information to the IDPH about the health status of children in Iowa. This enables the IDPH to plan for future services of those with special health or education needs.

The information provided by the Registry has the potential to help identify contributing factors of birth defects and allow for prevention education. The Registry may also connect families to local resources, which in turn may benefit your family by connecting you with other families that have similar circumstances or resources in your community.

The registry conducts research projects to study genetic and environmental risk factors for birth defects. Examples of birth defects currently studied by the registry include Down syndrome, congenital heart defects, neural tube defects, and cleft lip and palate. The University of Iowa Human Subjects Committee oversees all research projects. Family participation in research is critical to determine the potential causes of birth defects. Participating families typically provide environmental exposure information via mailed questionnaires or telephone surveys. Some research projects ask for biological samples from the affected child and his or her family members.

Iowa Stillbirth Surveillance Project and Stillbirth Prevention programs

In 2003, legislation was passed directing the Iowa Department of Public Health to establish a work group to develop a standardized stillbirth evaluation protocol and make recommendations for the surveillance of stillbirths occurring in Iowa. CCID received a cooperative agreement from the Centers for Disease Control and Prevention (CDC) to expand the existing Iowa Registry for Congenital and Inherited Disorders to include surveillance for fetal deaths (a.k.a. stillbirths). Information collected is analyzed for trends in fetal deaths occurrences, with the hope that prevention initiatives can be developed.

IRCID has conducted active surveillance to examine all Iowa stillbirths from 2000 through current day. IRCID now has over 2000 stillbirth cases in the registry. Each case identified is reviewed by a clinical reviewer to make sure it meets the case definition for stillbirth and to clarify possible causes and contributing factors to the stillbirth. Occasionally, information provided on the fetal death certificate provided to vital records is inaccurate. In such cases, IRCID does not have the authority to amend the fetal death certificate, but will correct the records in the registry. IRCID also does a match of fetal death certificates, stillbirths identified through active surveillance, and fetal death evaluation protocols submitted to the IDPH. For most years reported, there is not a one-to-one-to-one match of the records.

Neuromuscular and Related Genetic Disorders Program

The Neuromuscular and Related Genetics Disease Program provides comprehensive health services for individuals and families with a variety of neuromuscular and related disorders including Duchenne muscular dystrophy, Becker muscular dystrophy, limb-girdle muscular dystrophy, congenital muscular dystrophy, spinal muscular atrophy, myasthenia gravis, charcot-marie-tooth, and Friedreich's

ataxia. Additionally, the program gives educational presentations for families, healthcare providers, educators and others throughout Iowa.

What services are provided?

- Diagnostic evaluation
- Management of neuromuscular healthcare concerns
- Patient and family education
- Supportive patient and family services
- Physical therapy evaluation and recommendations
- Access to research opportunities

The Muscular Dystrophy Association approves service provision by the Neuromuscular and Related Genetic Disease Program.

Where are the clinics located?

Davenport, Des Moines, Dubuque, Iowa City, Mason City, Sioux City and Waterloo.

How do I make a referral?

Referrals are accepted from physicians, nurses, social workers, teachers, family members, and others. To arrange an appointment for neuromuscular services or inquire about educational presentations, call 319-353-6200.

- Katherine Mathews, MD
  Neuromuscular Program Director
  Division of Child Neurology
  Department of Pediatrics
  200 Hawkins Drive
  Iowa City, Iowa 52242
  319-356-7726 (ph)
  319-384-8818 (fax)
- Christina Trout, RN, MSN
  Advanced Practice Nurse
  Neuromuscular Program Department of Pediatrics
  200 Hawkins Drive
  Iowa City, Iowa 52242
  319-353-6200 (ph)
  319-384-8818 or 319-356-4855 (fax)

What is the cost for the services?

Insurance companies usually reimburse for medical consultation, nursing services, physical therapy, and respiratory therapy recommendations. Third party payers will be billed if coverage is available. Additionally, the Muscular Dystrophy Association offers financial assistance to families who qualify.

Iowa Neuromuscular and Related Disease Program Needs Assessment Report for the State of Iowa, June 2013; View the report

Family Health History Initiative

What is family history? Family history can be defined as a family's combination of shared genes, environment, behaviors and culture. Why should you determine your family's health history? A family health history can:

- Help you learn about diseases or conditions that you may be at an increased risk for developing;
- Point out behaviors (smoking, inactivity) or environmental exposures (secondhand smoke, farm chemicals) that may be increasing your risk for disease and that should be changed; and
- Help you take advantage of appropriate screening tests (genetic testing, mammography, colonoscopy, blood pressure checks) that may detect problems early.

The U. S. Surgeon General's Family Health History Initiative has developed a family health history tool that can help you collect information about your family health history. U.S. Surgeon General's Family Health History Initiative. This website offers two different types of tools - one in a downloadable electronic form that you can put on your own computer and update as needed, or hard copy forms in English or Spanish that you may print off to use.

The Genetic Alliance also offers family health history tools on their website that are customizable for different communities and cultures.

Iowa Newborn Screening for Critical Congenital Heart Disease

Congenital heart disease occurs in 9 of every 1,000 live births. About 25percent of these babies will have critical congenital heart disease (CCHD) whereby surgery or transcatheter intervention is required in the first year of life. In the United States, almost all types of congenital heart defects can be surgically repaired or palliated, and survival rates continue to improve. Early recognition and timely intervention can improve outcomes for these patients. The physical findings consistent with congenital heart disease, such as heart murmurs, tachypnea, or overt cyanosis may not be evident before the newborn is discharged from the hospital. Pulse-oximetry monitoring, a noninvasive method to determine oxygen saturation and identify hypoxemia has been proposed as one strategy for early detection of CCHD.

In August 2009, the American Academy of Pediatrics (AAP) and the American Heart Association (AHA) reviewed the available evidence and published a statement regarding the use of pulse oximetry to detect critical congenital heart disease in newborns. They concluded that it is a viable strategy to improve early detection of CCHD.

In September 2010, the US Health and Human Services (HHS) Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) recommended that critical congenital cyanotic heart disease be added to the uniform newborn screening panel. Their goal was to identify early in life those newborns with structural heart defects usually associated with hypoxia that could result in significant morbidity or death with closing of the ductus arteriosus or other physiologic changes in the newborn period. An expert technical panel recommended 7 specific lesions as the primary targets for screening: hypoplastic left heart syndrome (HLHS); pulmonary atresia; tetralogy of fallot; total anomalous pulmonary venous return; transposition of the great arteries; tricuspid atresia; and truncus arteriosus.

In September 2011, Department of Health and Human Services Secretary Kathleen Sebelius approved the addition of screening for critical congenital heart disease to each state's newborn screening panel. In 2012, Iowa passed legislation stating that all newborns and infants born in Iowa shall receive newborn screening for CCHD.

Screening for CCHD is accomplished by using pulse oximetry to estimate levels of arterial oxygen saturation in the newborn's hand and foot. Pulse oximetry is a noninvasive method of estimating the arterial oxygen saturation and pulse rate (PR) from pulsatile absorption signals derived from a sensor placed on the skin.

If a newborn has a positive (abnormal) screening result, the health care provider is notified and an electrocardiogram is done. The baby may be transferred to a neonatal intensive care unit for monitoring and treatment.

Hospitals and providers are not required to report CCHD screening results to the state at this time. When the CCID has implemented its new Iowa Newborn Screening Information System (INSIS), anticipated in 2016, reporting will be required.

Iowa Code and Administrative Code

Iowa Code 2014, Chapter 136A (8, 3)

CHAPTER 136A CENTER FOR CONGENITAL AND INHERITED DISORDERS

Referred to in §135.11

136A.1 Purpose.

To reduce and avoid adverse health conditions of inhabitants of the state, the Iowa department of public health shall initiate, conduct, and supervise screening and health care programs in order to detect and predict congenital or inherited disorders. The department shall assist in the translation and integration of

genetic and genomic advances into public health services to improve health outcomes throughout the life span of the inhabitants of the state.

136A.2 Definitions.

As used in this chapter, unless the context otherwise requires:

1. *"Attending health care provider"* means a licensed physician, nurse practitioner, certified nurse midwife, or physician assistant.
2. *"Congenital disorder"* means an abnormality existing prior to or at birth, including a stillbirth, that adversely affects the health and development of a fetus, newborn, child, or adult, including a structural malformation or a genetic, chromosomal, inherited, or biochemical disorder.
3. *"Department"* means the Iowa department of public health.
4. *"Disorder"* means a congenital or inherited disorder.
5. *"Genetics"* means the study of inheritance and how genes contribute to health conditions and the potential for disease.
6. *"Genomics"* means the functions and interactions of all human genes and their variation within human populations, including their interaction with environmental factors, and their contribution to health.
7. *"Inherited disorder"* means a condition caused by an abnormal change in a gene or genes passed from a parent or parents to their child. Onset of the disorder may be prior to or at birth, during childhood, or in adulthood.
8. *"Stillbirth"* means an unintended fetal death occurring after a gestation period of twenty completed weeks, or an unintended fetal death of a fetus with a weight of three hundred fifty or more grams.

136A.3 Establishment of center for congenital and inherited disorders — duties.

A center for congenital and inherited disorders is established within the department. The center shall do all of the following:

1. Initiate, conduct, and supervise statewide screening programs for congenital and inherited disorders amenable to population screening.
2. Initiate, conduct, and supervise statewide health care programs to aid in the early detection, treatment, prevention, education, and provision of supportive care related to congenital and inherited disorders.
3. Develop specifications for and designate a central laboratory in which tests conducted pursuant to the screening programs provided for in subsection 1 will be performed.
4. Gather, evaluate, and maintain information related to causes, severity, prevention, and methods of treatment for congenital and inherited disorders in conjunction with a central registry, screening programs, genetic health care programs, and ongoing scientific investigations and surveys.
5. Perform surveillance and monitoring of congenital and inherited disorders to determine the occurrence and trends of the disorders, to conduct thorough and complete epidemiological surveys, to assist in the planning for and provision of services to children with congenital and inherited disorders and their families, and to identify environmental and genetic risk factors for congenital and inherited disorders.
6. Provide information related to severity, causes, prevention, and methods of treatment for congenital and inherited disorders to the public, medical and scientific communities, and health science disciplines.
7. Implement public education programs, continuing education programs for health practitioners, and education programs for trainees of the health science disciplines related to genetics, congenital disorders, and inheritable disorders.
8. Participate in policy development to assure the appropriate use and confidentiality of genetic information and technologies to improve health and prevent disease.
9. Collaborate with state and local health agencies and other public and private organizations to provide education, intervention, and treatment for congenital and inherited disorders and to integrate genetics and genomics advances into public health activities and policies.

136A.4 Genetic health services.

The center may initiate, conduct, and supervise genetic health services for the inhabitants of the state, including the provision of regional genetic consultation clinics, comprehensive neuromuscular health care outreach clinics, and other outreach services and clinics as established by rule.

136A.5 Newborn metabolic screening.

1. All newborns born in this state shall be screened for congenital and inherited disorders in accordance with rules adopted by the department.
2. An attending health care provider shall ensure that every newborn under the provider's care is screened for congenital and inherited disorders in accordance with rules adopted by the department.
3. This section does not apply if a parent objects to the screening. If a parent objects to the screening of a newborn, the attending health care provider shall document the refusal in the newborn's medical record and shall obtain a written refusal from the parent and report the refusal to the department as provided by rule of the department.

136A.5A Newborn critical congenital heart disease screening.

1. Each newborn born in this state shall receive a critical congenital heart disease screening by pulse oximetry or other means as determined by rule, in conjunction with the metabolic screening required pursuant to section 136A.5.
2. An attending health care provider shall ensure that every newborn under the provider's care receives the critical congenital heart disease screening.
3. This section does not apply if a parent objects to the screening. If a parent objects to the screening of a newborn, the attending health care provider shall document the refusal in the newborn's medical record and shall obtain a written refusal from the parent and report the refusal to the department.

4. Notwithstanding any provision to the contrary, the results of each newborn's critical congenital heart disease screening shall only be reported in a manner consistent with the reporting of the results of metabolic screenings pursuant to section 136A.5 if funding is available for implementation of the reporting requirement.
5. This section shall be administered in accordance with rules adopted pursuant to section 136A.8.

136A.6 Central registry.

The center for congenital and inherited disorders shall maintain a central registry, or shall establish an agreement with a designated contractor to maintain a central registry, to compile, evaluate, retain, and disseminate information on the occurrence, prevalence, causes, treatment, and prevention of congenital disorders. Congenital disorders shall be considered reportable conditions in accordance with rules adopted by the department and shall be abstracted and maintained by the registry.

136A.7 Confidentiality.

The center for congenital and inherited disorders and the department shall maintain the confidentiality of any identifying information collected, used, or maintained pursuant to this chapter in accordance with section 22.7, subsection 2.

136A.8 Rules.

The center for congenital and inherited disorders, with assistance provided by the Iowa department of public health, shall adopt rules pursuant to chapter 17A to administer this chapter.

136A.9 Cooperation of other agencies.

All state, district, county, and city health or welfare agencies shall cooperate and participate in the administration of this chapter.

2004 Acts, ch 1031, §10, 12

CHAPTER 4 CENTER FOR CONGENITAL AND INHERITED DISORDERS
641—4.1(136A) Program overview. The center for congenital and inherited disorders within the department of public health provides administrative oversight to the following: Iowa newborn screening program, Iowa maternal prenatal screening program, regional genetic consultation service, neuromuscular and related genetic disease program, Iowa registry for congenital and inherited disorders, and Iowa early hearing detection and intervention program.
4.1(1) *Advisory committee.* The center for congenital and inherited disorders advisory committee represents the interests of the people of Iowa and assists in the development of programs that ensure the availability of and access to quality genetic and genomic health care services by all residents. The committee advises the director of the department of public health regarding issues related to genetics and hereditary and congenital disorders and makes recommendations about the design and implementation of the center's programs.
4.1(2) *Genetics coordinator.* The state genetics coordinator assigned within the department provides administrative oversight to the center for congenital and inherited disorders program within Iowa.
4.1(3) *Title V.* The center for congenital and inherited disorders has an association with the state Title V maternal child health program to promote comprehensive services for women, infants and children.
641—4.2(136A) Definitions. For the purposes of this chapter, the following definitions shall apply:
"Anonymized specimen" means a specimen that cannot be traced back to or linked with the particular individual from whom the specimen was obtained.
"Attending health care provider" means the licensed physician, nurse practitioner, certified midwife or physician assistant providing care to an infant at birth.
"Birthing facility" means a private or public facility licensed pursuant to Iowa Code chapter 135B that has a licensed obstetric unit or is licensed to provide obstetric services.
"Center" means the center for congenital and inherited disorders within the Iowa department of public health.
"Central laboratory" means the state hygienic laboratory (SHL), which is designated as the screening laboratory to perform testing and reporting for the Iowa newborn screening and Iowa maternal prenatal screening programs.
"Central registry" means the Iowa registry for congenital and inherited disorders (IRCID).
"Committee" means the congenital and inherited disorders advisory committee (CIDAC).
"Consulting physician" means a physician designated by the center for congenital and inherited disorders to interpret screen results and provide consultation to a licensed health care provider.
"Critical congenital heart disease" or *"CCHD"* means the presence of one or more specific heart lesions: hypoplastic left heart syndrome, pulmonary atresia, tetralogy of Fallot, total anomalous pulmonary venous return, transposition of the great arteries, tricuspid atresia, and truncus arteriosus.
"Department" means the Iowa department of public health.
"Director" means the director of the Iowa department of public health.
"Discharge" means a release of an infant from a hospital or birth center.
"Early ACCESS" means Iowa's Individuals with Disabilities Education Act (IDEA), Part C, program for infants and toddlers. Early ACCESS is a statewide, comprehensive, interagency system of integrated early intervention services that supports eligible children and their families as defined in 281—Chapter 120.
"Early hearing detection and intervention program" means Iowa's newborn hearing screening and follow-up program which ensures that all newborns and toddlers with hearing loss are identified as early as possible and provided with timely and appropriate audiological, educational and medical intervention and family support.
"Follow-up program" means the services provided to follow up on an abnormal screening result.
"Guardian" means a person who is not the parent of a minor child, but who has legal authority to make decisions regarding life or program issues for the child.

"Health care provider" means a licensed physician, nurse practitioner, certified nurse midwife, registered nurse, or physician assistant providing medical care to an individual.
"Iowa maternal prenatal screening program" or *"IMPSP"* means a program that provides a screening test designed to identify women with an increased risk of having a baby with a congenital or inherited disorder or women at risk of developing a problem later in pregnancy.
"Newborn critical congenital heart disease (CCHD) screening" means the screening of newborns for seven targeted heart conditions (hypoplastic left heart syndrome, pulmonary atresia, tetralogy of
Fallot, total anomalous pulmonary venous return, transposition of the great arteries, tricuspid atresia, and truncus arteriosus) using pulse oximetry or other means to detect blood oxygen saturation levels.
"Primary health care provider" means a licensed physician, physician assistant, nurse practitioner, or certified nurse midwife providing ongoing primary medical care to a patient.
"Receiving facility" means the facility receiving an infant from a birthing facility.
"Residual maternal prenatal serum screening specimen" means the portion of the specimen that may be left over after all necessary activities of the Iowa maternal prenatal screening program are completed.
"Residual newborn screening specimen" means the portion of the dried blood spot specimen that may be left over after all activities necessary for the Iowa newborn screening program are completed.
"Specialty genetics provider" means a geneticist, genetic nurse, or genetic counselor.
"State hygienic laboratory" or *"SHL"* means the designated central testing laboratory.
"Transferring facility" means the birthing facility that transfers the infant to another facility.
641—4.3(136A) Iowa newborn screening program (INSP). This program provides comprehensive newborn screening services for hereditary and congenital disorders for the state.
4.3(1) *Newborn screening policy.*
a. All newborns and infants born in the state of Iowa shall be screened for all congenital and inherited disorders specified by the center and approved by the state board of health.
b. As new disorders are recognized and new technologies and tests become available, the center shall follow protocols developed by the department in regard to the addition of disorders to or the deletion of disorders from the screening panel. The state board of health shall provide final approval for the addition of disorders to or the deletion of disorders from the screening panel.
c. The center may monitor individuals identified as having a genetic or metabolic disorder for the purpose of conducting public health surveillance or intervention and for determining whether early detection, treatment, and counseling lead to the amelioration or avoidance of the adverse outcomes of the disorder. Birthing facilities and health care providers shall provide patient data and records to the center upon request to facilitate the monitoring. Any identifying information provided to the center shall remain confidential pursuant to Iowa Code section 22.7(2).
d. For purposes of newborn screening, the department shall collect newborn screening specimens and data, test the specimens for disorders on the universal screening panel, conduct follow-up on abnormal screening results, conduct quality improvement and quality assurance activities, and store specimens for a time period determined by policies established by the CIDAC and the department.
4.3(2) *Newborn blood spot screening procedure for facilities and providers.*
a. Educating parent or guardian. Before a specimen from an infant is obtained, a parent or guardian shall be informed of the type of specimen, how it is obtained, the nature of the disorders for which the infant is being screened, the consequences of treatment and nontreatment, and the retention, use and disposition of residual specimens.
b. Refusal of screening. Should a parent or guardian refuse the screening, said refusal shall be documented in the infant's medical record, and the parent or guardian shall sign the refusal of screening form. The birthing facility or attending health care provider shall submit the signed refusal of screening form to the central laboratory within six days of the refusal. The birthing facility or attending health care provider may submit refusal forms via the courier service established for the transportation of newborn screening specimen collection forms.

c. Collection of specimens. A filter paper blood specimen shall be collected from the infant between 24 to 48 hours after the infant's birth. A specimen shall not be collected from an infant less than 24 hours after birth except as follows:
(1) A blood specimen must be collected before any initial transfusion, even if the infant is less than
24 hours old.
(2) A blood specimen must be collected before the infant leaves the hospital, whether by discharge or by transfer to another hospital, even if the infant is less than 24 hours old.
d. Submission of specimens. All specimens shall be delivered via courier service or, if courier service is not available, forwarded by first-class mail or other appropriate means within 24 hours after collection to the SHL.
e. Informed consent for the release of residual specimens for research use. The department shall establish policies and procedures, including an informed consent for release of specimens for research, to allow a parent or guardian the ability to provide informed consent prior to the release of the newborn's residual newborn screening specimen for research purposes. The parent or guardian, birthing facility or attending health care provider shall submit the informed consent form to the central laboratory pursuant to established policy and procedure. The informed consent procedure shall apply to all specimens collected on or after January 1, 2016. For specimens collected prior to January 1, 2016, a parent or guardian may send a letter stating that the newborn's specimen is not to be released for research purposes. This letter shall include the parent's or guardian's name, the newborn's name at birth, and the newborn's date of birth. The letter of notice shall be sent to the State Hygienic Laboratory at Newborn Screening Program,
State Hygienic Laboratory, 2220 S. Ankeny Blvd., Ankeny, Iowa 50023-9093.
4.3(3) *Primary health care provider responsibility.*
a. The health care provider shall ensure that infants under the provider's care are screened.
b. Procedures for specimen collection for newborn blood spot screening shall be followed in accordance with 4.3(2).
c. A physician or other health care professional who undertakes primary pediatric care of an infant delivered in Iowa shall arrange for the newborn screening if a newborn screening result is not in the infant's medical record.
4.3(4) *Birthing facility.* The birthing facility shall ensure that all infants receive newborn screening.
a. Designee. Each birthing facility shall designate an employee to be responsible for the newborn screening program in that institution.
b. Procedures for specimen collection for newborn screening shall be followed in accordance with
4.3(2).
c. Transfer. The following shall apply if an infant is transferred:
(1) If an infant is transferred within the hospital for acute care, the newborn nursery shall notify the acute care unit of the status of the newborn screening. The acute care unit shall then be responsible for the status of the newborn screening prior to discharge of the infant.
(2) If the infant is transferred to another facility within the state, the facility shall notify the receiving facility of the status of the newborn screening. The receiving facility shall then be responsible for completion of the newborn screening prior to discharge of the infant.
d. Discharge. Each birthing facility shall collect a newborn screening specimen on every infant prior to discharge, including under the following circumstances:
(1) The infant is discharged or transferred to another facility before the infant is 24 hours old.
(2) The infant is born with a condition that is incompatible with life.
(3) The infant has received a transfusion.
e. Notification. The birthing facility shall report the newborn screening results to the health care provider who has undertaken ongoing primary pediatric care of the infant.
4.3(5) *SHL responsibility.* The SHL shall:
a. Contract with a courier service to provide transportation and delivery of newborn screening specimens.
b. Contact all birthing facilities to inform them of the courier schedule.
c. Process specimens within 24 hours of receipt.
d. Notify the submitting health care provider, birthing facility, or drawing laboratory of an unacceptable specimen and the need for another specimen.

e. Report a presumptive positive screen result within 24 hours to the consulting physician or the physician's designee.
f. Distribute specimen collection forms, specimen collection procedures, refusal of newborn screening forms, and other materials to drawing laboratories, birthing facilities, and health care providers.
g. Report normal and abnormal screening results to the submitting facility or provider.
h. Submit a written annual report of the previous calendar year to the center by July 1of each year.
This report shall include:
(1) Number of infants screened,
(2) Number of repeat screens,
(3) Number of presumptive positive results by disorder,
(4) Number of rejected specimens,
(5) Number of waivers,
(6) Results of quality assurance testing including any updates to the INSP quality assurance policies, and
(7) Screening and educational activity details.
i. In collaboration with the program consulting physicians, submit a proposed budget and narrative justification for the upcoming state fiscal year by January 31 of each year.
j. Act as fiscal agent for program expenditures encompassing the analytical, technical, administrative, educational, and follow-up costs for the screening program.
k. Submit a fiscal expenditures report to the center within 90 days after the end of the state fiscal year.
4.3(6) *Follow-up program responsibility.* Follow-up programs shall be available for all individuals identified by the newborn screening as having an abnormal screen result.
a. The follow-up activities shall include care coordination, consultation, recommendations for treatment when indicated, case management, education and quality assurance.
b. The follow-up programs shall submit a written annual report of the previous calendar year by
July 1 of each year. The report shall include:
(1) The number of presumptive positive results and confirmed positive results by disorder,
(2) Number of confirmed cases receiving follow-up,
(3) A written summary of educational and follow-up activities.
c. In collaboration with the SHL, the follow-up programs shall submit a proposed budget and narrative justification for the upcoming fiscal year to the center by January 31 of each year.
d. The follow-up programs shall submit a fiscal expenditures report to the center within 90 days of the end of the state fiscal year.
4.3(7) *Sharing of information and confidentiality.* Reports, records, and other information collected by or provided to the Iowa newborn screening program relating to an infant's newborn screening results and follow-up information are confidential records pursuant to Iowa Code sections 22.7 and 136A.7.
INSP data may be retained indefinitely.
a. Personnel of the program shall maintain the confidentiality of all information and records used in the review and analysis of newborn screening and follow-up, including information that is confidential under Iowa Code chapter 22 or any other provisions of state law.
b. The program shall not release confidential information except to the following persons and entities, under the following conditions:
(1) The parent or guardian of an infant or child or the adult individual for whom the report is made.
(2) A primary health care provider, birthing facility, or submitting laboratory.
(3) A representative of a state or federal agency, to the extent that the information is necessary to perform a legally authorized function of that agency or the department. The state or federal agency will be subject to confidentiality regulations which are the same as or more stringent than those in the state of Iowa.
(4) A researcher, upon documentation of parental consent obtained by the researcher, and only to the extent that the information is necessary to perform research authorized by the department.
4.3(8) *Retention, use and disposition of residual newborn screening specimens.*

a. A newborn screening specimen collection form consists of a filter paper containing the dried blood spots (DBS) specimen and the attached requisition that contains information about the infant and birthing facility or drawing laboratory. The DBS specimen can be separated from the information contained in the requisition form. The INSP is the custodian of the specimens and related data for purposes of newborn screening, quality improvement and quality assurance activities.
(1) The residual DBS specimen shall be held for five years in a locked area at the SHL.
(2) The residual DBS specimen shall be stored for the first year at −70 degrees C.
(3) After one year, the residual DBS specimen shall be archived for four additional years at room temperature.
(4) The residual DBS specimen shall be incinerated after completion of the retention period.
b. The program shall not release a residual newborn screening specimen except to the following persons and entities:
(1) The parent or guardian of the infant or the individual adult upon whom the screening was performed.
(2) A health care provider acting on behalf of the patient.
(3) A medical examiner authorized to conduct an autopsy on a child or an investigation into the death of a child.
(4) A researcher for research purposes, under the terms and conditions provided in this rule.
(5) The newborn screening program, for operations as provided in this rule.
c. Research. A residual newborn screening specimen may be released for research purposes only if written consent has been received from a parent or guardian of the child, or the individual adult upon whom the screening was performed, and each of the following conditions is satisfied:
(1) Investigators shall submit proposals to use residual newborn screening specimens to the center.
Any intended use of the requested specimens as part of the research study must be clearly delineated in the proposal.
(2) Before research can commence, proposals shall be approved by the researcher's institutional review board, the congenital and inherited disorders advisory committee, and the department.
(3) Research on anonymized or identifiable residual newborn screening specimens shall be allowed only in instances where research would further: newborn screening activities; the health of an infant or child for whom no other specimens are available or readily attainable; general medical knowledge for existing public health surveillance activities; public health purposes; or medical knowledge to advance the public health.
d. Newborn screening program operations. Residual newborn screening specimens may be used for activities, testing, and procedures directly related to the operation of the newborn screening program, including confirmatory testing, laboratory quality control assurance and improvement, calibration of equipment, evaluation and improvement of the accuracy of newborn screening tests, and validation of equipment and screening methods, and the use of linked specimens in feasibility studies approved by the Congenital and Inherited Disorders Advisory Committee for the purpose of incorporating new tests or evaluating new test methodologies.
e. Prohibited uses. A residual newborn screening specimen shall not be released to any person or entity for commercial purposes or law enforcement purposes or to establish a database for forensic identification.
4.3(9) *Newborn screening for critical congenital heart disease.* All newborns and infants born in
Iowa shall receive newborn screening for CCHD, by pulse oximetry or other means in accordance with subparagraph 4.3(9)*"b"*(3). The purpose of newborn screening for CCHD is to identify newborns with structural heart defects usually associated with hypoxia in the newborn period which could have significant morbidity or mortality early in life with the closing of the ductus arteriosus or other physiological changes early in life.
a. Newborn CCHD screening procedure for providers and facilities.
(1) Educating parent or guardian. Before newborn screening for CCHD on an infant is conducted, a parent or guardian shall be informed of the type of screening, how it is performed, the nature of the disorders for which the infant is being screened, and the follow-up procedure for an abnormal screen result.

(2) Refusal. Should a parent or guardian refuse the screening, said refusal shall be documented in the infant's medical record, and the parent or guardian shall sign the refusal of screening form. The birthing facility or attending health care provider shall submit the signed refusal form to the central laboratory within six days of the refusal. The birthing facility or attending health care provider may submit refusal forms via the courier service established for the transportation of newborn screening specimen collection forms.
b. Newborn CCHD screening for newborns in low-risk or intermediate nurseries or out-of-hospital births.
(1) Screening should not begin until the newborn is at least 24 hours of age, or as late as possible if earlier discharge is planned, and should be completed on the second day of life.
(2) Screening shall be conducted using pulse oximeters or other means in accordance with subparagraph 4.3(9)*"b"*(3). Pulse oximeters shall:
1. Be motion tolerant;
2. Report functional oxygen saturation;
3. Be validated in low-perfusion conditions;
4. Be cleared by the Food and Drug Administration (FDA) for use on newborns; and
5. Have a 2 percent root-mean-square accuracy.
Disposable or reusable probes may be used. Reusable probes must be appropriately cleaned between uses according to manufacturer's instructions.
(3) Newborn CCHD screening shall be conducted by pulse oximetry or other means in accordance with the most recently published guidelines, algorithms, and protocols as outlined by the American
Academy of Pediatrics, the American College of Cardiology Foundation and the American Heart
Association, or subsequent guidance by the organizations listed in this subparagraph. Materials are available on the CCID Web page at
http://idph.state.ia.us/genetics/newborn_screening.asp.
c. Newborn CCHD screening for high-risk newborns in neonatal intensive care settings (NICU). Until such time that an evidence-based protocol for CCHD screening in infants discharged from the NICU is available, the attending health care provider shall conduct a comprehensive examination of the newborn to screen the infant for CCHD prior to discharge.
d. Primary health care provider responsibility. The health care provider shall ensure that infants under the provider's care are screened.
e. Reporting results of newborn CCHD screening. At such time as the CCHD reporting system is implemented, results of newborn CCHD screening shall be reported in a manner consistent with other newborn screening (formerly referenced as metabolic screening) reporting.
4.3(10) *INSP and IMPSP fees.*
a. The department shall annually review and determine the fee to be charged for all activities associated with the INSP and the IMPSP. The review and fee determination shall be completed at least one month prior to the beginning of the fiscal year. The newborn screening fee is $122.
b. The department shall include as part of the INSP fee an amount determined by the committee and department to fund the provision of special medical formula and foods for eligible individuals with
inherited diseases of amino acids and organic acids who are identified through the programs.
c. Funds collected through newborn screening fees shall be used for newborn screening program activities only.
d. Funds collected through maternal prenatal screening fees shall be used for maternal prenatal screening activities only.
e. In order to support newborn and maternal prenatal screening activities, the department shall authorize the expenditure and exchange of newborn screening and maternal prenatal screening funds between the SHL (as designated fiscal agent) and the department.
f. Upon department approval of proposed budgets, a portion of INSP and IMPSP fees shall be distributed to the department to support the percent of effort of the executive officer of the center for congenital and inherited disorders (CCID).
4.3(11) *Special medical formula and foods program.*

a. A special medical formula and foods program for individuals with inherited diseases of amino acids and organic acids who are identified through the Iowa newborn screening program is provided by the University of Iowa.
b. Payments received from clients based on third-party payment, sliding fee scales and donations shall be used to support the administration of and the purchase of special medical formula and foods.
c. The funding allocation from the Iowa newborn screening program fee will be used as the funder of last resort after all other available funding options have been pursued by the special medical formula and foods program.
d. Provisions of special medical formula and foods through this funding allocation shall be available to an individual only after the individual has shown that all benefits from third-party payers including, but not limited to, health insurers, health maintenance organizations, Medicare, Medicaid,
WIC and other government assistance programs have been exhausted. In addition, a full fee and a sliding fee scale shall be established and used for those persons able to pay all or part of the cost. Income and resources shall be considered in the application of the sliding fee scale. Individuals whose income is at or above 185 percent of the federal poverty level shall be charged a fee for the provision of special medical formula and foods. Placement of individuals on the sliding fee scale shall be determined and reviewed at least annually.
e. The SHL shall act as the fiscal agent.
f. The University of Iowa Hospitals and Clinics under the control of the state board of regents shall not receive indirect costs from state funds appropriated for this program.
641—4.4(136A) Iowa maternal prenatal screening program (IMPSP). This program provides comprehensive maternal prenatal screening services for the state.
4.4(1) *Maternal screening policy.* It shall be the policy of the state of Iowa that all pregnant women are offered maternal prenatal screening. The Iowa maternal prenatal screening program provides a risk assessment for open neural tube defects, ventral wall defects, Down syndrome, and Trisomy 18.
a. If a patient desires this screening test, her health care provider shall direct that a specimen be drawn and submitted to the SHL.
b. As new technologies and tests become available, the center shall follow protocols developed by the department with regard to the addition of disorders to or the deletion of disorders from the screening program.
4.4(2) *Maternal screening procedure.*
a. Collection of specimens. A serum or clotted blood specimen shall be collected from the patient within the appropriate gestational range indicated by the requested screen.
b. Processing of specimens. The SHL shall test specimens within three working days of receipt.
c. Reporting of abnormal results. Abnormal screen results shall be reported within 24 hours to the consulting physician or the physician's designee who shall then notify the primary health care provider.
On the next working day, this initial report shall be followed by a written report to the primary health care provider.
4.4(3) *Consulting physician responsibility.* A consulting physician shall be designated by the center in collaboration with the SHL to provide interpretation of screen results and consultation to the submitting health care provider. This physician shall provide consultation for abnormal screen results, assist with questions about management of identified cases, provide education and assist with quality assurance measures. The screening program, with assistance from the consulting physician, shall:
a. In collaboration with the SHL, submit a proposed budget and narrative justification for the upcoming fiscal year to the center by January 31 of each year, and
b. Submit a written annual report of the previous calendar year to the center by July 1 of each year.
The report shall include:
(1) Number of women screened,
(2) Number of repeat screens,
(3) Number of abnormal results by disorder,
(4) Number of rejected specimens,
(5) Results of quality assurance testing, and

(6) Screening and educational activity details.
4.4(4) *SHL responsibility.* The SHL shall:
a. Contract with a courier service to provide transportation and delivery of maternal prenatal serum specimens.
b. Contact all entities submitting specimens to inform them of the courier's schedule.
c. Test specimens within three working days of receipt.
d. Distribute specimen collection kits and other materials to health care provider offices and drawing facilities as required.
e. Inform the submitting health care provider or drawing facility of an unacceptable specimen and request another specimen.
f. Provide educational materials concerning specimen collection procedures.
g. Have available for review a written quality assurance program covering all aspects of its screening activity.
h. Act as a fiscal agent for program charges encompassing the analytical, technical, administrative, educational and follow-up costs for the screening program.
4.4(5) *IMPSP fee determination.* The department shall annually review and determine the fee to be charged for all activities associated with the IMPSP. The review and determination of the fee shall be completed at least one month prior to the beginning of the fiscal year.
4.4(6) *Sharing of information and confidentiality.* Reports, records, and other information collected by or provided to the IMPSP relating to a patient's maternal prenatal screening results and follow-up information are confidential records pursuant to Iowa Code section 22.7.
a. Personnel of the program shall maintain the confidentiality of all information and records used in the review and analysis of maternal serum screening and follow-up, including information that is confidential under Iowa Code chapter 22 or any other provisions of state law.
b. The program shall not release confidential information except to the following persons and entities, under the following conditions:
(1) The patient for whom the report is made.
(2) A primary health care provider or submitting laboratory.
(3) A representative of a state or federal agency, to the extent that the information is necessary to perform a legally authorized function of that agency or the department. The state or federal agency will be subject to confidentiality regulations which are the same as or more stringent than those in the state of Iowa.
(4) A researcher, upon documentation of patient consent obtained by the researcher, and only to the extent that the information is necessary to perform research authorized by the department and the state board of health.
4.4(7) *Retention, use and disposition of residual maternal prenatal screening specimens.*
a. A maternal serum screening specimen collection consists of laboratory tubes with maternal serum and associated information about the patient, health care provider, or drawing laboratory.
(1) The residual serum specimens shall be held for a specified period of time in a locked area at the
SHL in accordance with SHL policy and procedures.
(2) Reserved.
b. Research use.
(1) Investigators shall submit proposals to use residual serum specimens to the center. Any intent to utilize information associated with the requested specimens as part of the research study must be clearly delineated in the proposal.
(2) Before research can commence, proposals shall be approved by the researcher's institutional review board, the congenital and inherited disorders advisory committee, and the department.
(3) Personally identifiable residual specimens or records shall not be disclosed without documentation of informed patient consent obtained by the researcher.
(4) Research on anonymized or identifiable residual specimens shall be allowed in instances where research would further maternal prenatal screening activities or general medical knowledge for existing public health surveillance activities.
641—4.5(136A) Regional genetic consultation service (RGCS). This program provides comprehensive genetic and genomic services statewide through outreach clinics.

4.5(1) *Provision of comprehensive genetic and genomic services.* The department shall contract with the division of medical genetics within the department of pediatrics at the University of Iowa to provide genetic and genomic health care and education outreach services for individuals and families within Iowa. The contractor shall provide annual reports to the department as specified in the contract.
4.5(2) *Clinical services.* The services provided may include, but are not limited to: diagnostic evaluations, confirmatory testing, consultation by board-certified geneticists, genetic counseling, medical case management, and referral to appropriate agencies.
4.5(3) The University of Iowa Hospitals and Clinics under the control of the state board of regents shall not receive indirect costs from state funds appropriated for this program.
641—4.6(136A) Neuromuscular and other related genetic disease program (NMP). This program provides comprehensive services statewide for individuals and families with neuromuscular disorders through outreach clinics and statewide, active surveillance for selected neuromuscular disorders.
4.6(1) Provision of comprehensive services. The department shall contract with the department of pediatrics at the University of Iowa to provide neuromuscular health care, case management and education outreach services for individuals and families within Iowa. The contractor shall provide annual reports to the department as specified in the contract.
4.6(2) Clinical services. The services provided may include, but are not limited to: diagnostic evaluations, confirmatory testing, physical therapy, consultation by board-certified neurologists, genetic counseling, medical case management, supportive services and referral to appropriate agencies.
4.6(3) The University of Iowa Hospitals and Clinics under the control of the state board of regents shall not receive indirect costs from state funds appropriated for this program.
641—4.7(136A) Iowa registry for congenital and inherited disorders (IRCID). This program provides active statewide surveillance for congenital and inherited disorders. These disorders may include birth defects, neuromuscular disorders, metabolic disorders, and all stillbirths. The program also may conduct active statewide surveillance of live births without a reportable congenital or inherited disorder to serve as controls for epidemiological surveys. Surveillance activities for specific congenital and inherited disorders will be conducted for the period of time that adequate financial support is available.
4.7(1) *Definitions.*
a. Birth defects shall be defined as any major structural abnormality or metabolic disorder that may adversely affect a child's health and development. The abnormality or disorder must be diagnosed or its signs and symptoms must be recognized within the first two years of life.
b. Neuromuscular disorders shall be defined as Duchenne, Becker, congenital, distal, Emery-Dreifuss, fascioscapulohumeral, limb-girdle, myotonic, and oculopharyngeal muscular dystrophies.
c. Rescinded IAB 4/3/13, effective 5/8/13.
d. Stillbirths shall be defined as an unintended fetal death occurring after a gestational period of
20 completed weeks or an unintended fetal death of a fetus with a weight of 350 or more grams. Stillbirth is synonymous with fetal death.
e. A reportable congenital or inherited disorder occurring in a miscarriage or pregnancy may be included in the IRCID.
4.7(2) *Surveillance policy.*
a. Congenital disorders, including birth defects, occurring in Iowa are reportable conditions, and records of these disorders shall be abstracted pursuant to 641—1.3(139A) and maintained in the
IRCID. Congenital disorders surveillance shall be performed in order to determine the occurrence and trends of such disorders, to determine co-occurring conditions and treatments through annual follow-up abstraction, to conduct thorough and complete epidemiological surveys to identify environmental and genetic risk factors for congenital disorders, to contribute to prevention strategies, and to assist in the planning for and provision of services to children with congenital disorders and their families.
b. Records for neuromuscular disorders shall be abstracted pursuant to 641—1.3(139A) and maintained in the IRCID. Neuromuscular disorders surveillance for individuals of all ages shall be performed in order to determine the occurrence and trends of the selected

neuromuscular disorders, to determine co-occurring conditions and treatments through annual follow-up abstraction, to conduct thorough and complete epidemiological surveys through annual long-term follow-up, and to assist in the planning for and provision of services to individuals with selected neuromuscular disorders and their families.
c. Rescinded IAB 4/3/13, effective 5/8/13.
d. Stillbirths occurring in Iowa are reportable conditions, and records of these stillbirths shall be abstracted pursuant to 641—1.3(139A) and maintained in the IRCID. Stillbirth surveillance shall be performed in order to determine the occurrence and trends of stillbirths, to conduct thorough and complete epidemiological surveys to identify environmental and genetic risk factors for stillbirths, and to assist in the planning for and provision of services to prevent stillbirths.
4.7(3) *IRCID activities.*
a. The center shall establish an agreement with the University of Iowa to implement the activities of the IRCID.
b. The IRCID shall use the birth defects, neuromuscular disorders, metabolic disorders, and stillbirth coding schemes developed by the Centers for Disease Control and Prevention (CDC).
c. The IRCID staff shall review hospital records, clinical charts, physician's records, vital records, prenatal records, and fetal death evaluation protocols pursuant to 641—1.3(139A), information from the INSP, RGCS, NMP, and the IMPSP, and any other information that the IRCID deems necessary and appropriate for congenital and inherited disorders surveillance.
4.7(4) *Department responsibility.*
a. When a live infant's medical records are ascertained by the IRCID, the department or its designee shall inform the parent or legal guardian by letter that this information has been collected and provide the parent or guardian with information about services for which the child and family may be eligible.
b. The center and the IRCID shall annually release aggregate medical and epidemiological information to medical personnel and appropriate state and local agencies for the planning and monitoring of services for children with congenital or inherited disorders and their families.
4.7(5) *Confidentiality and disclosure of information.* Reports, records, and other information collected by or provided to the IRCID relating to a person known to have or suspected of having a congenital or inherited disorder are confidential records pursuant to Iowa Code sections 22.7 and
136A.7.
a. Personnel of the IRCID and the department shall maintain the confidentiality of all information and records used in the review and analysis of congenital or inherited disorders, including information which is confidential under Iowa Code chapter 22 or any other provisions of state law.
b. IRCID staff are authorized pursuant to 641—1.3(139A) to gather all information relevant to the review and analysis of congenital or inherited disorders. This information may include, but is not limited to, hospital records, physician's records, clinical charts, vital records, prenatal records, fetal death evaluation protocols, information from the INSP, RGCS, NMP, and the IMPSP, and any other information that the IRCID deems necessary and appropriate for congenital and inherited disorders surveillance.
IRCID staff are permitted to review hospital records, clinical charts, physician's records, vital records, and prenatal records, information from the INSP, RGCS, NMP, and IMPSP and any other information that the IRCID deems necessary and appropriate for live births without a reportable congenital or inherited disorder to serve as controls for epidemiological surveys.
c. No individual or organization providing information to the IRCID in accordance with this rule shall be deemed or held liable for divulging confidential information.
4.7(6) *Access to information in the IRCID.* The IRCID and the department shall not release confidential information except to the following, under the following conditions:
a. The parent or guardian of an infant or child for whom the report is made and who can demonstrate that the parent or guardian has received the notification letter.
b. An Early ACCESS service coordinator or an agency under contract with the department to administer the children with special health care needs program, upon receipt of written consent from the parent or guardian of the infant or child.

c. A local health care provider, upon receipt of written consent from the parent or guardian of the infant or child.
d. A representative of a federal agency, to the extent that the information is necessary to perform a legally authorized function of that agency or the department. The information provided shall not include the personal identifiers of an infant or child with a reportable congenital or inherited disorder.
e. Researchers, in accordance with the following:
(1) All proposals for research using the IRCID data to be conducted by persons other than program staff shall first be submitted to and accepted by the researcher's institutional review board. Proposals shall then be reviewed and approved by the department and the IRCID's internal advisory committee before research can commence.
(2) The IRCID shall submit to the IRCID's internal advisory committee for approval a protocol describing any research conducted by the IRCID in which the IRCID deems it necessary to contact case subjects and controls.
f. A representative of a state agency, to the extent that the information is necessary to perform a legally authorized function of that agency or the department. The state agency will be subject to confidentiality regulations that are the same as or more stringent than those in the state of Iowa.
641—4.8(135) Iowa's early hearing detection and intervention program. The goal of universal hearing screening of all newborns and infants in Iowa is the early detection of hearing loss to allow children and their families the earliest possible opportunity to obtain appropriate early intervention services. All newborns and infants born in Iowa, except those born with a condition that is incompatible with life, shall be screened for hearing loss. Early hearing detection and intervention programming and services will be provided pursuant to 641—Chapter 3.
641—4.9 and **4.10** Reserved.
CENTER FOR CONGENITAL AND INHERITED DISORDERS ADVISORY COMMITTEE (CIDAC)
641—4.11(136A) Purpose. CIDAC represents the interests of the people of Iowa and assists in the development of programs that ensure the availability of and access to quality genetic and genomic health care services by all residents. The committee advises the director regarding issues related to genetics and hereditary and congenital disorders.
641—4.12(136A) Duties of the committee. CIDAC shall perform the following duties:
4.12(1) Make recommendations about the design and implementation of the center's programs, including but not limited to:
a. The Iowa newborn screening program;
b. The regional genetics consultation service;
c. The maternal prenatal screening program;
d. The neuromuscular and related genetic disorders program; and
e. The Iowa registry for congenital and inherited disorders.
4.12(2) Support the development of special projects and conferences regarding genetic and genomic health care services and issues.
4.12(3) Advocate for quality genetic and genomic health care services for all residents in the state of Iowa.
641—4.13(136A) Membership. Membership will be comprised of representatives of professional
groups, agencies, legislators, parents, consumers, and professional health care providers.
4.13(1) CIDAC shall be comprised of regular, ex officio, and honorary members.
a. Potential regular members are considered from interest groups, consumer organizations, and genetic and genomic health care service providers. Two parent representatives and two consumer representatives shall serve as regular members of CIDAC.
b. The number of regular members shall not be fewer than 15 or more than 25.
c. No more than 30 percent of regular members shall be representatives of or employed by programs that are contractors of the center for congenital and inherited disorders in the Iowa department of public health.
d. Honorary members will be comprised of two legislators, one state senator and one state representative, and others deemed appropriate by the director.
e. Ex officio members are nominated by virtue of their positions held and the organizations they represent and are appointed by the director. These members provide expert information and consultation to CIDAC.

4.13(2) Every effort will be made to have gender balance and broad geographic representation on the advisory committee.
4.13(3) The director will appoint regular and honorary committee members for three fiscal years.
Reappointment of regular and honorary members shall be at the discretion of the director.
641—4.14(136A) Meetings.
4.14(1) Meetings of the committee will be held as necessary and at the call of the director or the chairperson. There shall be a minimum of four meetings per year.
4.14(2) All meetings are open to the public in accordance with the open meetings law, Iowa Code chapter 21.
4.14(3) A majority of the total number of regular members (50 percent plus one member) shall constitute a quorum. There must be a quorum of the regular members in attendance at a meeting for action to be taken.
4.14(4) Action can be taken by a vote of the regular members. Ex officio and honorary members are not eligible to vote.
4.14(5) Regular members who represent programs that are contractors of the center for congenital and inherited disorders in the department are expected to refrain from imposing undue influence on regular members and to recuse themselves from voting on issues which directly affect the operation of their programs.
4.14(6) Meeting attendance.
a. Attendance at a meeting is defined as presence at the meeting site in person, through the Iowa communications network (ICN), through webinar or web meeting, or via telephone.
b. Attendance by the regular member or the regular member's designee shall be expected at all meetings.
(1) A designee of similar standing must be able to reasonably fulfill the member's role on the committee in discussions.
(2) Designees are not eligible to vote.
c. Regular members, not designees, must attend at least two meetings per fiscal year to remain in good standing.
d. A regular member who misses more than three meetings in a fiscal year shall be deemed to have submitted a resignation.

[**ARC 0664C**, IAB 4/3/13, effective 5/8/13]
These rules are intended to implement Iowa Code chapter 136A.
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IAB 12/10/14, effective 1/14/15]

Membership

CIDAC Membership

1. American College of Obstetricians and Gynecologists, Iowa Chapter
2. Iowa Osteopathic Medical Association
3. American Academy of Pediatrics, Iowa Chapter
4. Academy of Family Physicians, Iowa Chapter
5. Nursing organization representative: e.g./ INA, IANP
6. Iowa Senator
7. Iowa Representative
8. Medical Geneticist, Division of Medical Genetics, U of I
9. Iowa Department of Education
10. Iowa Child Health Specialty Clinics
11. College of Public Health
12. National Association of Social Workers, Iowa Chapter
13. Iowa Insurance Commission
14. Ethics/Bioethics representative
15. Parent
16. Parent
17. Consumer/member at large
18. Consumer/member at large
19. Representative from community-based organizations, such as the March of Dimes, the ARC, Sickle Cell Society
20. Representative from Iowa Registry for Congenital and Inherited Disorders
21. Representative from University Hygienic Laboratories
22. Representative from Maternal Screening Programs

CIDAC Roster 2015

CONGENITAL AND INHERITED DISORDERS ADVISORY COMMITTEE (CIDAC)

Iowa Department of Public Health

Appointed by Director, Iowa Department of Public Health

Iowa Administrative Code, Chapter 4, Sections 641-4.1-4.7

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Policies and Procedures

CCID Policy #001 - Retention, Storage, and Use of Specimens Residual to Those Collected for Newborn Screening Services

CCID Policy #2 - Release of Data Policy and Procedure

CCID Policy #3 - Determining Conditions on the Iowa Newborn Screening Panel

CCID Policy #4 - Use of Iowa Newborn Screening Program and Iowa Maternal Prenatal Screening Developmental Funds