Congenital & Inherited Disorders

Division of Health Promotion & Chronic Disease Prevention
Phone: 1-800-383-3826
http://idph.iowa.gov/genetics

Protecting & Improving the Health of Iowans

IDPH Iowa Department of Public Health

Brody's Story

As a school psychologist, Kelsey works daily with children who have educational challenges. She and her husband did everything possible to give their son Brody a healthy start. One week after his birth, they were shocked to learn that Brody was diagnosed with profound biotinidase deficiency. This genetic condition can cause developmental delays, hearing and vision loss, coma, and even death. Fortunately, Brody's condition was caught in time. "We are humbled and grateful," Kelsey says. "With continuous treatment, Brody now has the chance to live a quality life full of joy, health and opportunity."

Did you know?

Each year, an average of 1,850 Iowa children are born with a congenital or inherited disorder, and approximately 200 babies are stillborn. Three of every 1,000 newborns or 120 babies in Iowa are diagnosed with hearing loss each year and another 2 to 3 per 1,000 children will develop hearing loss after birth. Childhood hearing loss is the most common birth defect. Most babies born with hearing loss are born to parents with normal hearing.

The Center for Congenital and Inherited Disorders (CCID) programs serve all phases of the life cycle: prenatal, neonatal, pediatric, and adult.

Why is the Center for Congenital and Inherited Disorders important to protecting and improving the health of Iowans?

- Screening programs for the early detection of inherited or congenital disorders help assure earlier interventions to eliminate or reduce disability and provide family support.
- Early detection and treatment can prevent mental retardation and even death in children born with an inherited or congenital disorder.
- Children born with a hearing loss who are identified early and given appropriate intervention before 6 months of age demonstrated significantly better speech and reading comprehension than children identified after 6 months of age (Yoshinaga-Itano, et al., 1998).
- By the time a child with hearing loss graduates from high school, more than $400,000 per child can be saved in special education costs if the child is identified early and given appropriate educational, medical, and audiological services (White & Maxon, 1995).

Which Iowa Public Health Goals are we working to achieve?

Strengthen the health infrastructure

Promote healthy living

What do we do?

CCID administers 8 programs that promote and improve access to comprehensive genetic health care services, laboratory services, early hearing detection and intervention, and surveillance. CCID assures statewide education is provided and develops policies and programs that assure the availability of and access to quality genetic health care, newborn screening, and laboratory services.

- Early Hearing Detection and Intervention (EHDI) program – provides universal newborn hearing screening, short-term follow up, and referrals to early intervention and family support services.
- Regional Genetics Consultation Services – regional clinics provide statewide medical consultation and counseling to people with a diagnosed genetic disorder.
- Neuromuscular & Related Disorders – provides medical consultation and counseling to those with a diagnosed neuromuscular disorder, such as muscular dystrophy.
- Iowa Newborn Screening Program (INSP) – conducts newborn testing and follow-up for metabolic disorders and cystic fibrosis. Testing is done for Iowa, North Dakota, and South Dakota. INMSP also provides metabolic formula and medical foods for people diagnosed with PKU and other inherited metabolism disorders that require medically necessary foods.
- Iowa Registry for Congenital and Inherited Disorders (IRCID) – conducts surveillance for congenital and inherited disorders and stillbirth on children born in Iowa.
- Stillbirth Surveillance Program – supports stillbirth surveillance activities of the Iowa Registry for Congenital and Inherited Disorders. Promotes stillbirth's awareness initiatives.
- Family Health History Initiative – provides resources to explore and compile family health history to determine the risk of inheriting disease. Provides resources for lifestyle/behavior changes and screening tests based on the results of the family health history.
- Maternal Prenatal Screening Program – conducts prenatal testing to screen for congenital/inherited disorders of the fetus.

How do we measure our progress?

❶ **Percent of newborns whose screening specimens are received by the State Hygienic Laboratory within ~~72~~ 60 hours of birth.** Data Source: INSP/SHL database. Data are available annually.

How are we doing? In 2016, 95% of Iowa newborn screening specimens were received by the State Hygienic Laboratory within 60 hours of birth (target – 95%).

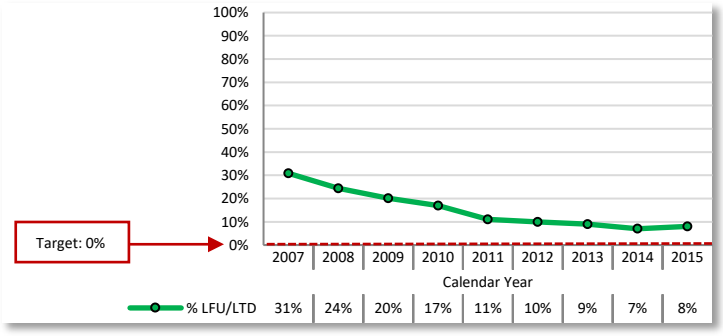
❷ **Percent of children, who do not have a parent-signed refusal, that are screened for disorders tested through the Iowa newborn screening panel.** Data Source: INSP/SHL database. Data are available annually.

How are we doing? Nearly all, 99%, Iowa newborns were screened using the Iowa newborn screening panel (target – 100%). There were 40 NBS refusals signed in CY2016 (number has increased due to improved documentation of refusals). One-hundred percent of the newborns whose parents did not refuse the screening received the blood spot screening.

❸ **Percent of infants screened at birth for hearing loss.**
Data Source: IDPH/EHDI database. Data are available annually. 2015 data is preliminary.
*Not eligible children data removed (families who refused screening and deceased).

How are we doing? Nearly all Iowa newborns (99%) are screened for hearing loss. Those not eligible for screening included infant deaths and parent refusals. There were 280 families who refused the hearing screen at birth; 231 (83%) were home birth families. An additional 21 children were considered lost at birth, 100% of those were home birth families who did not respond to repeated hearing screening requests. The number of refusals slightly decreased while the numbers of children lost at birth is consistent with 2014 data. Education and outreach continues to make a difference.

❹ **Percent of infants lost to follow up or documentation (LFU/LTD) among all infants who did not pass their initial birth hearing screen.**



Data Source: IDPH/EHDI database. Data are available annually. 2016 data are not yet available as some children may still be receiving follow up. 2015 data is preliminary.

How are we doing? The number of infants that do not return for a hearing re-screen has decreased steadily since 2007 indicating a greater percentage of children are receiving recommended follow up.

What can Iowans do to help?

1. Go to http://idph.iowa.gov/genetics to learn about CCID programs, and http://idph.iowa.gov/ehdi to learn more about EHDI programs.
2. Support and promote newborn screenings by having your children screened, and encouraging others to do the same.
3. Conduct your own family health history and talk to your health care provider about the results.
4. Talk to your legislators about funding for newborn screening and genetic programs.
5. Contact the EHDI advisory committee (http://idph.iowa.gov/ehdi/committee) with questions or issues.
6. Contact the Congenital and Inherited Disorders Advisory Committee (http://idph.iowa.gov/genetics/public/advisory-committee) with questions or issues

Health care professionals can

1. Teach patients about the benefits of newborn screening.
2. Provide information to pregnant women about monitoring fetal activity.
3. Help patients gather their family health history and discuss the results with them.
4. Learn more about science-based genetic research.

Policymakers can

1. Learn about science-based genetic research and genetic programs.
2. Provide funding for public health-based genetic programs, including public health surveillance.

Expenditures

General fund, federal funds, private grants*, & retained fees*: K07-0705/0709/0765; 0830-0830; 0153-0570/0722/0724. **EHDI**: general fund & federal funds: K05-0611; 0153-0544/0682

	State Fiscal Year 2015 Actual	State Fiscal Year 2016 Actual	State Fiscal Year 2017 Estimate
State funds	$1,037,327	$1,130,890	$1,162,083
Federal funds	$693,985	$504,023	$363,782
Other funds*	$224,869	$301,330	$244,969
Total funds	**$1,956,182**	**$1,936,243**	**$1,770,834**
FTEs	3.78	4.19	3.95

Note: Funding information is intended to provide an overview of funding related to the program area. It does not include all federal and state requirements and/or restrictions for the use of funds. Contact the program area for more detailed budget information.

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