

# Health Advisory: Infantile Krabbe Result Requires Immediate Action

Minnesota Department of Health, Mon, Feb 26, 13:00 CST 2024

## Action Steps

**Local and tribal health department:** Please forward to birth centers, hospitals, primary care clinics and OBGYN clinics in your jurisdiction.

**Hospitals, clinics, and other facilities:** Please forward to family physicians, pediatricians, primary care clinicians, nursery staff, NICU physicians, pediatric neurologists, pediatric metabolic specialists, genetic counselors, laboratory staff, and emergency department staff.

**Health care providers:**

- Watch for MDH Newborn Screening Notifications from MDH for Early Infantile Onset Krabbe, a rare disease that requires a hematopoietic stem cell transplant in the first 30 days of life.
- Call M Health Fairview on-call metabolic specialists IMMEDIATELY for guidance on clinical follow-up at 612-672-7575.
- Contact the child's family IMMEDIATELY, as MDH does not contact them directly. Share the follow-up plan from M Health Fairview with them.
- Arrange for referrals and help the family coordinate follow-up. Most care will be provided via M Health Fairview's University of Minnesota hospital.
- Contact MDH's Newborn Screening genetic counselors with questions about any screening result at 651-201-3548.

## Background

Krabbe disease is a neurometabolic disorder caused by a deficiency in the GALC enzyme which leads to the deterioration of the myelin sheath and nerve damage. Initial symptoms of infantile Krabbe disease include irritability, sensitivity to loud noises, feeding difficulties, and muscle stiffness. If untreated, it can cause seizures, blindness, deafness, neurologic deterioration, and death within the first two years of life.

The MDH Newborn Screening program has just started testing for Krabbe disease. See the full [MDH Newborn Screening Panel and Timeline](https://www.health.state.mn.us/people/newbornscreening/program/newbornscreeningpanel.html) (<https://www.health.state.mn.us/people/newbornscreening/program/newbornscreeningpanel.html>) for a list of the over 60 conditions that is included in newborn screening.

There is no cure for Krabbe disease. Currently, the only treatment available for the early infantile onset form is hematopoietic stem cell transplantation (HSCT). **HSCT is most effective when performed before symptoms develop, ideally before 30 days of life.** HSCT has been shown to slow the progression of the disease, improve quality of life, and increase the lifespan. Supportive therapies and management like physical therapy and medications can also be beneficial for both early onset and later onset forms of the disease. Even when performed pre-symptomatically, treatment does not prevent all morbidity and mortality.

The M Health Fairview University of Minnesota hospital is the only center in the region who can perform stem cell transplants for children with infantile Krabbe disease. The major metabolic centers in the state

(Children's and Mayo) along with our North Dakota and South Dakota neighbors have elected to refer all consultations for infantile Krabbe disease to the University.

Due to the time-sensitive nature of treatment for infantile Krabbe disease, it's imperative that physicians who receive a Krabbe disease result from the MDH Newborn Screening program act on the information IMMEDIATELY. Things need to come together quickly for a newborn with infantile Krabbe disease to receive HSCT by day 30 of life. Families will need referrals to specialists that are only available at a select number of centers; additional diagnostic and genetic testing, as well as physical exams, will need to be performed; a transplant match must be found; preparation for transplant (typically myeloablative conditioning); etc. Additional complicating factors for care teams to consider including: issues surrounding insurance, appointment availability, parent resources and support (fiscal, transportation and lodging, ability to leave home and job for a significant period, ability of birthparent to travel so soon after giving birth, etc.).

## For More Information

- [Newborn Screening Program](https://www.health.state.mn.us/people/newbornscreening/index.html)  
(<https://www.health.state.mn.us/people/newbornscreening/index.html>)  
Additional information regarding the Minnesota Newborn Screening Program.
- [Blood Spot Screening Disorder Fact Sheets](https://www.health.state.mn.us/people/newbornscreening/materials/factsheets/bloodspotdisorders.html)  
(<https://www.health.state.mn.us/people/newbornscreening/materials/factsheets/bloodspotdisorders.html>)  
Fact sheets with disorder-specific information and next steps for both families and providers following an abnormal newborn screening result.

A copy of this HAN is available at: [MDH Health Alert Network](http://www.health.state.mn.us/han) (<http://www.health.state.mn.us/han>)  
The content of this message is intended for public health and health care personnel and response partners who have a need to know the information to perform their duties.