



LEUKODYSTROPHY & NEWBORN SCREENING AWARENESS MONTH

MEET EMMETT KUIPERS



"We are grateful our son was not born in South Dakota. We are now equipped with the knowledge to help ensure Emmett has the best chance at life "

Todd and Heather Kuipers
Emmett's parents

Todd and Heather Kuipers adopted their son, Emmett, in early 2021. Because Emmett was born in Missouri - a state that includes Krabbe Disease on their Newborn Screening panel - Todd and Heather received a life-changing phone call from the adoption agency a few days later and were informed that additional testing needed to be done. They met with genetic counselors while the tests were being run and discovered that Emmett has other diagnoses as well, but shared that Emmett has juvenile or adult-onset Krabbe Disease.

After doing research on their own, the Kuipers were connected with Dr. Escolar and the Children's Hospital of Pittsburgh when Emmett was nine months old. While there, they did an MRI and other tests to gain more insight into Emmett's status related to Krabbe Disease. They were grateful to learn that there did not appear to be progression at the time.

Going forward, Emmett will continue to be monitored throughout childhood. If he becomes symptomatic at any point, Todd and Heather will begin the process of treatment to ensure that Emmett has the best chance at a full life.

Emmett is a happy child and loves jumping in his jumperoo and reading books with his parents. Because of his other diagnoses, his complex care team believes that he experiences cognitive delays. Todd and Heather are hopeful that this journey will settle down as he continues to grow and thrive and are grateful for the knowledge they have thanks to Newborn Screening.

NBS IN SOUTH DAKOTA

All babies born in the United States are screened for several conditions shortly after birth. Approximately 24-48 hours after a baby is held in the United States, a nurse pricks the heel to collect a small blood sample. Afterward, the nurse puts a series of blood drops onto a filter paper to create several "dried blood spots." Next, the Newborn Screening card is sent to the state laboratory for analysis.

South Dakota is not screening for Krabbe disease.

WHY SCREEN FOR KRABBE?

Krabbe disease is a severe neurodegenerative and rapidly progressing condition requiring immediate treatment for the most severe forms. The medical issues and symptoms of Krabbe disease are very significant and life-impacting. A delayed diagnosis, especially in the most severe forms, equates to palliative and supportive care as the only means of treatment until premature death.

WHAT IS KRABBE DISEASE?

Krabbe disease (pronounced krab A), is a rare genetic disorder, also known as globoid cell leukodystrophy. In the United States, Krabbe disease has been reported to affect approximately 1 in 100,000 individuals.¹ Infantile Krabbe disease is the most common and severe form causing infants to lose the ability to eat, extreme irritability, inability to sit up and grasp objects, blindness, and seizures. Sadly, infants die within the first 2-3 years of life in states that do not test for Krabbe disease.

We invite you to learn more at KrabbeConnect.org.



In 2020, there were²
10,960
live births in South Dakota



Number³
of conditions
screened for in South Dakota



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RESOURCES

- The Leukodystrophy Newborn Screening Action Network is dedicated to advancing newborn screening for leukodystrophies and lysosomal storage disorders, supporting newly-diagnosed families, and ensuring collaboration between all stakeholders. Learn more at <https://ldnbs.org/>.
- CDC offers funding and assistance through the Newborn Screening Quality Assurance Program (NSQAP). More information can be found at <https://www.cdc.gov/labstandards/nsqap.html>.
- Baby's First Test provides funding opportunities through grants. Learn more at <https://www.babysfirsttest.org/newborn-screening/funding-opportunities>.
- American Public Health Laboratories NewSTEPS program provides data, technical assistance, and training. Details at <https://www.newsteps.org/>.
- KrabbeConnect offers patient support services to help families navigate the burden of Krabbe disease. Learn more at <https://krabbeconnect.org/>.
- Hunter's Hope Foundation is a non-profit organization committed to giving hope through education, awareness, research, and family care for all leukodystrophies. Learn more at <https://www.huntershope.org/>.

CITATIONS

1. Wenger DA. Krabbe Disease. 2000 Jun 19 [Updated 2011 Mar 31]. In: Pagon RA, Adam MP, Ardinger HH, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2017.
2. "Fertility Rate: South Dakota, 2010-2020." March of Dimes | PeriStats, <https://www.marchofdimes.org/peristats/data?reg=29&top=2&stop=10&slev=4&obj=3&sreg=46&creg>. Accessed 31 July 2023.
3. "South Dakota | Baby's First Test | Newborn Screening | Baby Health." Babysfirsttest.org, 2015, <https://www.babysfirsttest.org/newborn-screening/states/south-dakota>. Accessed 31 July 2023.