



LEUKODYSTROPHY & NEWBORN SCREENING AWARENESS MONTH

MEET WILLIAM BRANCH



'We are grateful for William and try to not think about the "what ifs" for him - only the here and now; however, we do know that William's life would be very different had he been able to receive treatment earlier.'

Robert and Abby Branch,
William's Parents

Robert and Abbey Branch welcomed William into their lives in 2014. When he was about three months old, William began to show signs of colic and wasn't eating as much as he should. Their previously happy, engaged child was angry, uncomfortable, and had a cry that had almost turned into a scream. After being misdiagnosed with reflux and then milk protein intolerance, his symptoms worsened and they noticed his limbs were stiff and rigid. They took William to a third pediatrician who took everything into consideration and recommended that William have an MRI to see if there were any abnormalities.

Robert and Abbey were unprepared for the words they were about to hear - the doctor told them that William likely only had only a year to live, that he would become deaf and blind, and that he would stop smiling.

Eager to help, one of Abbey's friends found Dr. Maria Escolar's information; three days later, the family flew to Pittsburgh and learned that he was indeed eligible for a transplant because he hadn't progressed too far and was healthy enough for the procedure.

His transplant was successful, but because he wasn't diagnosed at birth he experiences epilepsy and is immobile. That doesn't stop William from going to school and communicating through an eye-gaze device. He loves watching ballet and telling jokes, but he doesn't love math and makes that clear through his expressions. His smile is biggest when Abbey sings and dances and when Robert reads him stories. William is incredibly happy and loves to be the center of attention.

NBS IN IOWA

All babies in the United States are screened for several conditions shortly after birth. Approximately 24-48 hours after a baby is born 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. Each state determines what conditions are included on its panel.

Iowa does not currently screen 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 were ²
36,114
live births in Iowa



Number ³
of conditions
screened for in Iowa



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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: Iowa, 2010-2020." March of Dimes | PeriStats, <https://www.marchofdimes.org/peristats/data?reg=99&top=2&stop=1&lev=1&slev=4&obj=1&sreg=19&creg>. Accessed 31 July 2023.
3. "Iowa | Baby's First Test | Newborn Screening | Baby Health." Babysfirsttest.org, 2015, <https://www.babysfirsttest.org/newborn-screening/states/new-jersey>. Accessed 31 July 2023.