



LEUKODYSTROPHY & NEWBORN SCREENING AWARENESS MONTH

MEET BRYCE CLAUSEN



"It's devastating to know that a family attempted to add Krabbe disease to Indiana's NBS panel but were unsuccessful. If they had succeeded, Bryce would likely have been the first baby identified in Indiana.

Our son was born a mere twenty-five days after the proposed activation of screening. We knew we had to do something to change this."

Joel and Andrea Clausen
Bryce's parents

Joel and Andrea Clausen joyfully welcomed their second son, Bryce on January 25, 2018. Bryce spent fourteen days in the NICU primarily because his lungs had not fully developed. When he came home he was a happy baby, and although he was not the greatest eater, he was still gaining weight. Life proceeded as expected until Bryce was about six months old. Joel and Andrea began to notice that he was irritable, wasn't sleeping through the night anymore, and had lost some of his abilities. Pediatricians tried three different formulas to try to resolve the issues, but Bryce was starting to lose weight.

On November 1, 2018, when Bryce was nine months old, Joel and Andrea heard two words from their neurologist that changed their lives forever: Krabbe disease. They knew something must be wrong because they were called into the doctor's office for an after-hours appointment. The neurologist gave them the bad news as kindly as he could: "Your son has a terminal disease called Krabbe disease, there is no cure, and he will most likely pass away before his second birthday." Joel and Andrea were speechless as they attempted to process this news.

The Clausens made sure that Bryce had the most fulfilling life possible and took him on many adventures. Bryce loved seeing bright colors and listening to his brother talk to him. They said goodbye to Bryce just five months later, at fourteen months of age.

It is because of Bryce that Indiana now screens every baby for Krabbe disease at birth. Joel and Andrea worked to ensure that Indiana families have a different story than they did because they understand the impact of Newborn Screening for Krabbe disease: "Bryce's life would have been saved."

NBS IN INDIANA

All babies born 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.

Indiana began screening for Krabbe disease in July 2020, thanks to the Clausen family's advocacy efforts.

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 **78,616**² live births in Indiana



Number **58**³ of conditions screened for in Indiana



*All information in this fact sheet is based on data available before 7.31.2023



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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: Indiana, 2010-2020." March of Dimes | PeriStats, <https://www.marchofdimes.org/peristats/data?reg=42&top=2&stop=10&slev=4&obj=3&sreg=18&creg>. Accessed 31 July 2023.
3. "Indiana | Baby's First Test | Newborn Screening | Baby Health." Babysfirsttest.org, 2015, <https://www.babysfirsttest.org/newborn-screening/states/indiana>. Accessed 31 July 2023.