



LEUKODYSTROPHY & NEWBORN SCREENING AWARENESS MONTH

MEET MAX BARNETT



"It shouldn't matter where you live in the United States: you should have the same Newborn Screening experience to give your child the best chance at a great life. My son might still be here had Arizona screened for Krabbe disease."

Allison Barnett, Max's mom

Max was born on February 6, 2014, to parents Allison and Josiah. He was healthy and such a happy baby, always smiling. At his six-month well-check appointment, the doctor noted that he was a little delayed with some milestones. Around eight months of age, they began to notice that Max was struggling to sit up straight, he was a little fussier, he stopped grabbing for things, and he wasn't rolling. At his nine-month well-check, the pediatrician told them to take him to the emergency room because his breathing had become a little more shallow and something was off. An MRI was performed and they were sent home with no real answers other than he had some vanishing white matter. The pediatrician arranged for Allison and Josiah to meet with a genetics doctor the following week where they did blood work to test for some rare disorders.

At the next appointment, the geneticist walked in with two neurologists, and with the worst possible news - Max had a leukodystrophy called Krabbe disease. The doctors told Allison and Josiah to take Max home and make him comfortable.

Max quickly lost his eyesight but he loved superheroes, music, and his older sister, Emma. He would always look in her direction when she was around. Allison and Josiah created a bucket list for Max and experienced many great things with him.

Newborn Screening would have given the Barnetts an opportunity to have Max treated and to have given him a chance at life.



In 2020, there were **76,947**² live births in Arizona



Number of conditions³ screened for in AZ



*All information in this fact sheet is based on data available before 9.22.2022

NBS IN ARIZONA

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, the heel is pricked by a nurse to collect a small sample of blood. 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.

Arizona does not currently screen for Krabbe disease despite grassroots efforts to have it included.

WHY SCREEN FOR KRABBE?

Krabbe disease is a severe neurodegenerative and rapidly progressing condition requiring immediate treatment for the most severe forms. Max was diagnosed with an early onset form of Krabbe disease at six months of age. This form, known as infantile Krabbe disease, requires treatment within the first 30-45 days of life. If Max was identified at birth with Krabbe disease, Max could have undergone a stem cell transplant. Due to his delayed diagnosis, palliative and supportive care were his only means of treatment.

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.

This project was supported by the following:





KRABBE DISEASE IN ARIZONA

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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: Ohio, 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=04>. Accessed 22 Sep. 2022.
3. "Minnesota | Baby's First Test | Newborn Screening | Baby Health." Babysfirsttest.org, 2015, <https://www.babysfirsttest.org/newborn-screening/states/arizona>. Accessed 22 Sept. 2022.