



LEUKODYSTROPHY & NEWBORN SCREENING AWARENESS MONTH

MEET LILY SMITH



"Newborn Screening saves lives, but only if we're screening."

Until Maryland is screening for Krabbe disease, our story will continue to be the standard for babies born here, and that is not acceptable."

**Kathleen Smith,
Lily's Mom**

Lily Smith was born in 2011 to parents Kathleen and Ben and completed their family. When she was about 5 ½ months old, Kathleen noticed that Lily was no longer holding her head up or reaching for her toys. She sought advice from the pediatrician, who noted that Lily had regressed in some other milestones as well. When the pitch of Lily's cry changed, Kathleen was advised to take her to the Emergency Room at Children's National Medical Center for testing. When they were brought back to talk to the doctors, Kathleen showed them videos of what Lily had been able to do only a week before and they immediately ordered a CT scan. The CT scan showed white matter on the brain and the doctor advised them to admit Lily for an MRI the following morning. The results of the MRI confirmed that Lily had Krabbe disease; the doctor explained that she would most likely not live until her second birthday and encouraged them to contact hospice and take lots of pictures.

Kathleen and Ben did not want to think about losing their child but figured at this point that they should learn as much as they could from Lily and her Krabbe journey. They found Dr. Escolar in Pittsburgh and learned that she was conducting research on Krabbe disease. Dr. Escolar wanted to see them quickly so they drove to Pittsburgh. Lily went through a great deal of testing and evaluation over the course of several days; they were informed that Lily was eligible to receive a stem cell transplant, which would help preserve her sight and hearing as well as slow the progression of the disease drastically.

The decision needed to be made quickly, and they decided that treatment was the best decision for their family. Kathleen left her two older children at home and spent one hundred days with Lily in Pittsburgh. The transplant was not easy and Lily had many complications along the way, including many days on a ventilator in the ICU, and having a VP shunt placed for hydrocephalus.

Despite the challenge of the journey, Lily just celebrated her 11th birthday in September and is an amazing young lady. She uses her communication device to express her wants and needs and loves traveling, camping, reading, crafts, watching television shows, and playing games. She amazes and inspires her family daily with her strength and her big smile.



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

NBS IN MARYLAND

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 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.

Maryland 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 were ²
68,554
live births in Maryland



Number ³
of conditions screened
for in Maryland



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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: Maryland, 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=34>. Accessed 13 August 2023.
3. "Maryland | Baby's First Test | Newborn Screening | Baby Health." Babysfirsttest.org, 2015, <https://www.babysfirsttest.org/newborn-screening/states/Maryland>. Accessed 13 August 2023.