



# LEUKODYSTROPHY & NEWBORN SCREENING AWARENESS MONTH

## MEET WILLOW PIETZYK



**"We advocate for Nebraska to add Krabbe disease to its Newborn Screening panel so that other families don't have to spend months looking for answers and have the option to pursue treatment."**

**Taysha Pietzyk,**  
Willow's Mom

Willow Mae was born on December 29, 2020, the firstborn child of Taysha and Logan. They were in love with their baby girl and enjoyed watching her grow and learn. Around the time Willow was four months old, they began to notice that she was struggling to eat. After about a week, they went to the pediatrician and were sent home with acid reflux medicine. The medicine seemed to work for the next week, but then the issues restarted. They went back to the pediatrician and learned that Willow was losing weight, and they observed that she had lost head control. Taysha and Logan's concerns were brushed off, so they decided to pursue answers on their own.

She was admitted to Children's Hospital that day and had multiple blood draws, spinal taps, and MRIs done. The doctors there realized that something was abnormal with her brain. They were sent home with an NG tube and no answers.

On June 1, 2021, they had a follow-up appointment with Neurology that left them with no answers and complete heartache. The neurologist told them, "Some babies will just fade away and we have no answers. Willow will just fade away so go home and make her feel comfortable for the time she has left." No diagnosis was given.

With the encouragement of their family, Logan and Taysha decided to keep fighting for answers and for their daughter's life. They found a new pediatrician willing to fight alongside them - a three-month endeavor - and then received a referral to see Dr. Lulla at Boys Town National Research Hospital. On July 16th, Dr. Lulla immediately noticed that Willow was having a seizure, confirming Taysha and Logan's fears. In addition, he was not willing to accept that this was a "medical mystery" and it was because of him that genetic testing was performed.

On August 3rd, they finally received the diagnosis of Krabbe disease. The answer for which they had fought so hard brought relief because they finally knew what was causing her suffering; yet, with that relief came devastating grief. Equipped with this new knowledge they did their best to keep her comfortable, something they hadn't been able to do for her up to this point. They did their best to live every day to the fullest and did normal family things like going swimming, visiting the zoo, and going out to dinner. They chose joy in the midst of sadness.

Willow passed away on September 1, 2021, at eight months old.

## NBS IN NEBRASKA

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.

Nebraska 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.<sup>1</sup> 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](https://KrabbeConnect.org).



In 2020, there were <sup>2</sup>  
**24,291**  
live births in Nebraska



Number <sup>3</sup>  
of conditions screened  
for in Nebraska



## LEUKODYSTROPHY & NEWBORN SCREENING AWARENESS MONTH

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