Newborn Screening (NBS) in

ARIZONA

Saving Children's Lives in Arizona through Timely, Equitable Access to Newborn Screening

Newborn screening (NBS) can save lives, but only if Arizona includes all devastating but addressable conditions on its state screening panel in a timely manner.

NBS is a simple blood test performed 24 to 48 hours after birth to screen for certain rare, genetic, hormone-related, and metabolic conditions that can cause serious injury or death but have an approved therapeutic option or viable intervention.[1] These conditions are not always visible or easily diagnosed.

The process for adding new conditions to federal and state screening panels can be lengthy. Through the Recommended Uniform Screening Panel (RUSP), the U.S. Secretary for Health and Human Services (HHS) puts forward a national recommendation for conditions that all newborns should be screened for at birth. However, each state has the authority to determine what conditions to include in their local NBS program.

On average, it takes 6 years for a condition to move through the federal nomination process and, if approved, be included on the RUSP.[2] Further, Arizona does not screen for all conditions that are included on the RUSP,[3] and during the years between RUSP nomination and state adoption, the nearly 77,000 babies born in Arizona each year miss the opportunity for screening, limiting their chances to receive an early diagnosis with a condition, and in some cases, access to potentially life-saving therapies.[4]

Why is timely screening important?

Left untreated, many of the conditions eligible for NBS can result in severe illness, disability, and often, premature death. Some of the conditions are neurodegenerative and rapidly progressing, so time is critical. If a child is not screened at birth, diagnosis is often delayed to the point where the child's condition may have rapidly progressed.

How Can You Help

When an intervention is available, we ask that Arizona add MLD to the screening panel through the authority of the NBS advisory committee or through legislation.

Timely NBS could ensure that every baby born in the state will be screened for this devastating condition – giving residents of the state an opportunity for a diagnosis and medical guidance.

What is MLD?

MLD is a very rare, fatal genetic disorder. The incidence of MLD is estimated to be 1 in 100,000 births in the U.S. or approximately 30 – 40 births per year.[5] In its most severe and common form, this rapidly neurodegenerative disease causes young children to lose the ability to walk, talk and interact with the world around them. Sadly, about 50% of children with the most severe form of MLD will die within 5 years of disease onset with palliative care often their only option.

Meet the Riley Family

Kendra and Dave's youngest daughter, Keira, was born in 2020, around the time that one of their other daughters, Olivia, started having trouble walking. Originally, it was thought that Olivia simply needed physical therapy. Then, she began having problems with vision and hypermobility. After getting an MRI, the first neurologist told them nothing was wrong.

Thankfully, the Rileys decided to get a second opinion. Three days later, the second neurologist informed them that he believed Olivia had MLD. Follow up testing confirmed his hunch.

Kendra and Dave were crushed. On top of that, testing for their youngest daughter, Keira, revealed she also had MLD. Processing this news and seeking care during a global pandemic was a uniquely challenging experience for the family. But the diagnosis Keira received as an infant enabled her to get specialized support she needed early on.

Despite the family's struggles, all three sisters continue to bring joy and light to their parents and love spending time together on Olivia's Make-a-Wish swing set. And Kendra and Dave now devote their time to supporting families like theirs and raising awareness of MLD and newborn screening.

1 in 100,000

Incidence of MLD

Arizona's Current Newborn Screening Panel

Arizona does not screen for all conditions that are included on the RUSP.

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State Resources

- CDC offers funding and assistance through the Newborn Screening Quality Assurance Program (NSQAP).
- Baby's First Test provides funding opportunities through grants.
- American Public Health Laboratories NewSTEPS program provides data, technical assistance, and training.

Citations

1. https://newbornscreening.hrsa.gov/newborn-screening-process
2. https://www.hrsa.gov/advisory-committees/heritable-disorders/rusp/previous nominations.html
3. https://www.azdhs.gov/preparedness/state-laboratory/newborn-screening/index.php
4. https://www.cdc.gov/nchs/fastats/state-and-territorial-data.htm
5. Bonkowsky JL et. al., Neurology 2010, Aug 23; 75(8), 718-725

**Based on average timelines for the four most recently added RUSP conditions*

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