Newborn Screening (NBS) in

ILLINOIS

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

Newborn screening (NBS) can save lives, but only if Illinois 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, in Illinois it takes an average of 2 years* for the state to begin screening for conditions once they are on the RUSP.[3] During these "gap" years between RUSP nomination and state adoption, the approximately 140,000 babies born in Illinois each year miss the opportunity for screening, limiting their chances to receive 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?

Illinoisans can avoid a lengthy wait for MLD NBS.

When an intervention is available, we ask that Illinois 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 progressing neurodegenerative disease causes young children to lose the ability to walk, talk and interact with the world around them. Sadly, 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 Axel's Family

"Newborn screening is very important to us. Since NBS cannot change our path at this time, we need to know we're making a difference so other families may not have to endure this type of heartache."

- Axel's mother, Lexi, on the future state of NBS

On October 15, 2017, Axel became a loving addition to Colby and Lexi's family of four. He was a happy healthy growing boy, completing all milestones the first year of life. However, after his 2nd birthday, Axel still struggled with walking. After multiple doctor appointments and various delayed testing, the Riordans still had no answers. It wasn't until an ER admission that Colby and Lexi were told the news that every parent fears. Testing revealed that Axel suffered from early infantile MLD. To say Colby and Lexi were devastated is an understatement.

They have witnessed Axel losing simple abilities such as sitting up and swallowing. They know with time the progressive disease will spread, but Axel's parents are so thankful for the support they have from their family and friends. It just so happens that a lot of their support team consists of real-life nurses, too. Also, Axel has the BEST big brother ever, Ryder, who loves him very much.

Axel's parents find themselves heading from Illinois to UPMC, which is out of state, for specialized medical care. Some days, Lexi feels very accomplished in terms of helping Axel, while other days are just completely exhausting. She knows they have a long, scary road ahead, but no matter what, they will continue to fight for "Ax man" because he deserves it, and nothing will hold him back!

In Illinois it takes an average of

2 years

for the state to begin screening for conditions once they are on the RUSP

During each "gap" year approximately

140,000

babies miss the opportunity for screening.

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

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://dph.illinois.gov/topics-services/life-stages-populations/newborn-screening
4 https://dph.illinois.gov/data-statistics/vital-statistics/birth-statistics.html
5 Bonkowsky JL et. al., Neurology 2010, Aug 23; 75(8), 718-725

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