Newborn Screening (NBS) in

MINNESOTA

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

Newborn screening (NBS) can save lives, but only if Minnesota 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] Minnesota is a leader in NBS and thanks to the Super Gav Act, the state screens for conditions that are included on the RUSP.[3] However, during the years between RUSP nomination and state adoption, the approximately 66,000 babies born in Minnesota 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?

Minnesotans can avoid a lengthy wait for MLD NBS.

When an intervention is available, we ask that Minnesota 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, approximately half 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 Quimby Family

Gavin Quimby – affectionately known as Super Gav after his love of superheroes – was diagnosed with MLD at age three and passed away at age five. Gavin and his family turned the fight against MLD into superhero level advocacy to ensure that children with rare diseases have the opportunity for NBS. Super Gav was not only a hometown hero in Winona, he inspired screening for children facing other rare diseases.

Gavin's parents, Nick and Shanna Quimby, along with Senator Jeremy Miller and Minnesota's NBS Committee passed a bill named The Super Gav Act to test newborns for several rare diseases - including another leukodystrophy called adrenoleukodystrophy or ALD. This NBS bill was made to detect and treat diseases successfully before symptoms present. Since the Super Gav Act came into effect in February 2017, seventy children have come back positive for ALD, giving them an opportunity medical guidance because of NBS.

However, more work still needs to be done as the condition that Gavin battled, MLD, is not included on Minnesota NBS panel.

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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://www.hrsa.gov/advisory-committees/heritable-disorders/rusp/previous-nominations.html

2 https://newbornscreening.hrsa.gov/newborn-screening-process

3 https://www.health.state.mn.us/people/newbornscreening/

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

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