Newborn Screening (NBS) in

CALIFORNIA

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

Newborn screening (NBS) can save lives, but only if California 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 California it takes an average of 1.75 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 372,000 babies born in California 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?

Californians can avoid a lengthy wait for MLD NBS.

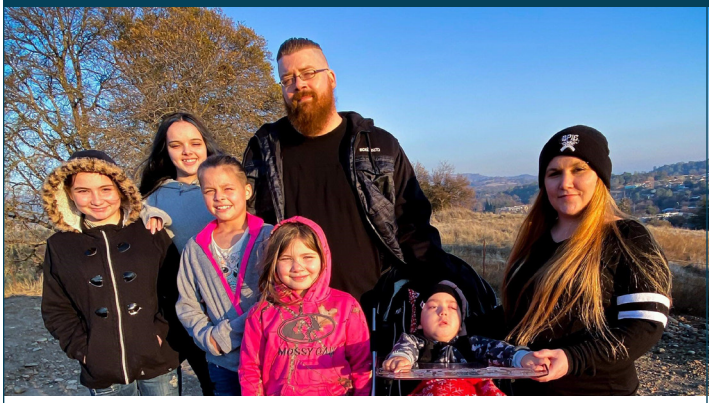
When an intervention is available, we ask that California add MLD to the screening panel through the authority of the NBS advisory committee or through legislation.

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 Killian's Family



Killian Viktorius Christie is a sweet 3-year-old boy from San Andreas, CA. He is the youngest sibling of 4 other sisters who do not have MLD.

Killian's parents became concerned about him when he was unable to balance while walking on his own. Eventually, he gave up on trying to walk. Doctors in the area were puzzled. Killian was eventually referred to a neurologist who ordered a genetic test, and Killian was diagnosed with MLD.

Watching Killian's frustration with losing his abilities was heartbreaking. Killian was aware that he had skills that were slipping away. Now, he has settled into his new life and lack of mobility and still smiles on some days. Killian is a constant humbling reminder that if he can smile through his tragedy, then we can also make something positive out of every day.

Animals love Killian. The Christie's have birds, cats, and dogs who follow him everywhere. Killian also loves hitting the road with his parents and sisters for the stimulation of the vibration from the road, the sound of music, and the vibrant colors of the scenery. His other favorite pastimes include cuddling in bed with Mom and Dad. Altogether, Killian's story is one of strength. MLD children are incredibly strong in spirit and they inevitably strengthen the people involved in their short, meaningful lives.

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

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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/previousnominations.html

3 https://www.cdph.ca.gov/Programs/CFH/DGDS/Pages/nbs/NBS-Disorders-Detectable.aspx

4 https://mn.gov/admin/demography/data-by-topic/births-fertility/

5 Bonkowsky JL et. al., Neurology 2010, Aug 23; 75(8), 718-725

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