Newborn Screening (NBS) in

NORTH CAROLINA

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

Newborn screening (NBS) can save lives, but only if North Carolina 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] North Carolina passed legislation in 2021 that implements a three-year timeline for federal RUSP alignment, which is an important step forward in supporting children and families with rare diseases.[3] However, during the years between RUSP nomination and state adoption, the more than 119,000 babies born in North Carolina 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?

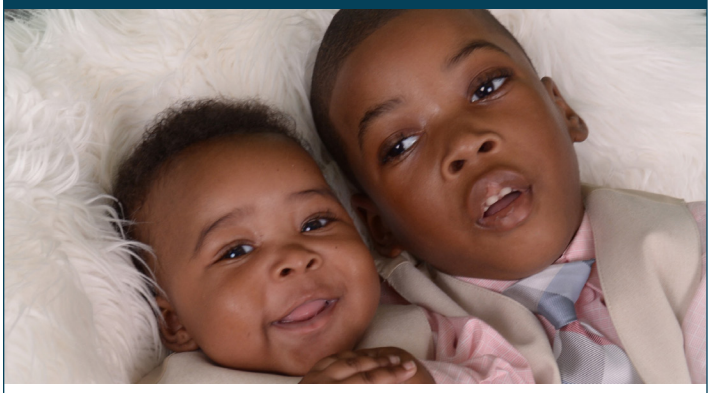
When an intervention is available, we ask that North Carolina add MLD to the screening panel through the authority of the NBS advisory committee or through legislation.

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, 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 James Family



Two years ago, Anna James learned that her son Emmanuel had a rare neurodegenerative condition called metachromatic leukodystrophy (MLD). Since MLD is genetic, her youngest child, Samuel, also needed to be tested. The results came back and confirmed the worst. At just one month old, Samuel, who looked like a perfectly healthy baby boy, was diagnosed with the same fatal condition as his older brother.

Emmanuel's condition has progressed very rapidly, and he has lost the ability to walk, talk, eat and more. He has been in the hospital for almost a year and is now on a respirator.

His younger brother, Sammy, received a bone marrow transplant when he was four months old to try and slow the progression of his MLD. Sammy spent his first year in the hospital and has to return frequently for medical treatments. For Anna, this means weekly trips to the hospital, a nearly three-hour roundtrip drive, while also trying to find childcare for her two older children.

MLD has put tremendous strain on the James family between long hospital stays, frequent trips across the state, and the financial burden of caring for two medically complex children. Anna is grateful for little moments of love between the two brothers and their siblings, especially they visit Emmanuel in the hospital.

Every year, between the time a condition is nominated to RUSP and added to the state panel,

>119,000

babies born in North Carolina miss the opportunity for screening.

North Carolina's Current Newborn Screening Panel

North Carolina recently passed legislation that implements a three-year timeline for federal RUSP alignment.

Newborn Screening (NBS) in

NORTH CAROLINA

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.ncleg.gov/BillLookUp/2021/H736

4 https://www.cdc.gov/nchs/data/vsrr/vsrr020.pdf

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

These materials were supported by Orchard Therapeutics.