

NEWBORN SCREENING

RECOMMENDED UNIFORM SCREENING PANEL (RUSP) ALIGNMENT LEGISLATION

For over 50 years, every newborn in the US has been afforded the chance to be screened for a range of debilitating and deadly diseases. Yet, as new technology allows for screening of more diseases, states have lagged in implementing these tests. As a result, children and their families miss the opportunity to receive optimal benefit from early treatment and intervention.

What is the Recommended Uniform Screening Panel (RUSP)?

The RUSP is a list of disorders recommended for states to screen as part of their universal newborn screening programs by the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) and approved by the Secretary of Health and Human Services.

New conditions are nominated to the RUSP by patient organizations and are added through science-driven, evidence-based review processes.

The RUSP currently includes 38 conditions, that without early detection and intervention, can cause disabilities, developmental delays, serious illness, or even death.

States decide individually how and when they will add new conditions to their newborn screening panels. While states generally follow the lead of the RUSP, the time it takes for states to begin screening for diseases newly added to the RUSP varies.

What is the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC)*?*

Composed of experts from key newborn screening stakeholder communities, the ACHDNC provides federal newborn screening recommendations to the Secretary of Health and Human Services.

Committee members recommend disorders to the RUSP based on evidence that supports the potential net benefit of screening, the ability of states to screen for the disorder, and the availability of effective treatments.

What is RUSP Alignment Legislation?

1. Requires that states consider screening newborn babies for any disorder on the RUSP.
2. Implements a timeline for states to begin screening for new disorders added to the RUSP.
3. Ensures resources are available to facilitate the addition of new disorders.

Why is RUSP Alignment important?

It currently takes an average of 5-6 years for states to add a disease to their state panel. [1]

Early detection allows for the diseases to be managed successfully and at a lower long-term cost- not only saving lives, but also saving state funds.

Over 12,000 infants are identified each year with one of the disorders included in the RUSP .[2]

Provides funding and resources to newborn screening programs to shore up delays between the availability of new treatments and implementation of screening.

[1] *https://www.mdpi.com/2409-515X/6/2/35*
[2] *https://www.cdc.gov/mmwr/volumes/69/wr/mm6936a6.htm*

NEWBORN SCREENING IN ACTION

Christina was seven months pregnant in the summer of 2016, when the first case of mosquito-borne Zika virus was contracted within the US- just blocks away from her Miami home. She and her husband Scott decided to be cautious and relocate temporarily to Chicago, her childhood home. In September, they had a beautiful baby girl named Ada.

A Chance Diagnosis

They were relieved to have escaped Zika, but when Ada was 10-days old they learned she had screened positive for a rare disease, called Pompe. A google search revealed that some infants with the disease do not live past their first birthday. Christina and Scott were terrified.

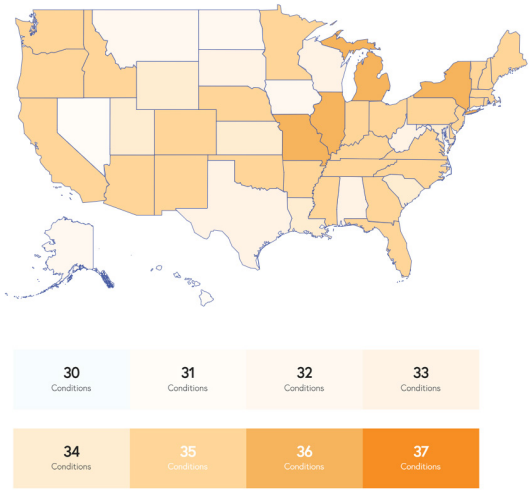
Follow-up testing confirmed the diagnosis and Ada was able to benefit from more advanced genomic testing and early intervention.

A Tale of Two States

If Ada had been born in Miami, her hometown, she would not have been screened for Pompe. Christina feels lucky that Ada was born in another state, where she was able to get an early diagnosis. More than four years later, Ada is a healthy toddler, learning new words every day.

Christina with baby Ada

Newborn screening programs vary widely by state, and many states are not testing for all diseases on the federal Recommended Uniform Screening Panel, leading to disparate health outcomes. We can do better.



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The EveryLife Foundation for Rare Diseases is a 501(c)(3) nonprofit, nonpartisan organization dedicated to empowering the rare disease patient community to advocate for impactful, science-driven legislation and policy that advances the equitable development of and access to lifesaving diagnoses, treatments and cures.