RARE READY

A GENETIC CONDITION COALITION

Expanding Newborn Screening: A Lifesaving Opportunity

Newborn screening is a public health program that involves collecting a few drops of blood from a newborn's heel to test for serious medical conditions that have effective, FDA-approved treatments. The number of conditions screened varies from state to state.

The Challenge

- More than **10,000 rare diseases** exist, affecting **1 in 10 Americans.**
- **80%** of rare diseases are genetic; approximately **70%** begin in childhood.

Why Expand Newborn Screening?

- Scientific breakthroughs have led to **treatments** for previously untreatable rare conditions.
- Delays in diagnosis have severe consequences. Nearly two-thirds of Americans living with a rare disease are children, and 30% of these children do not survive past their fifth birthday — **lives that could have been saved through screening.**
- **Early diagnosis** leads to timely interventions, improving quality of life, and survival rates.

Call to Action

- Support legislation to **expand state-specific newborn screening** programs.
- Ensure every child has the best possible start in life through **early detection and treatment.**

The Urgency to Act

- We are in an era of remarkable medical progress with advanced and even **curative treatments for genetic conditions.**
- The current state-specific newborn screening system has not kept pace with medical advancements, constrained by a slow and fragmented expansion process—screening for **fewer than 1% of rare diseases,** with many states **omitting conditions** that have FDA-approved treatments.
- On average, a rare disease diagnosis takes more than **six years** and nearly **17 medical encounters** after symptoms start, creating a significant economic burden—exceeding **$517,000 per patient** *(The Cost of Delayed Diagnosis in Rare Disease - EveryLife Foundation).*