Fact Sheet

march of dimes
North Carolina

All Newborns Should Be Screened for Pompe Disease

What is Pompe Disease?

Pompe disease is a rare, inherited disorder that causes progressive muscle weakness. It is a part of a group of disorders referred to as Lysosomal Storage Disorders (LSDs) and is classified as either infantile- or late-onset disease. Pompe disease is caused by mutations in the gene that make an enzyme (GAA) that breaks down glycogen (sugars). If not treated, the muscles weaken as glycogen accumulates, affecting feeding, weight gain, movement, breathing, and in infants, heart contraction. This can lead to death early in childhood, with many children not surviving past the first year of life. Pompe disease occurs in about 1 in 40,000 in the United States.

The U.S. Secretary of Health and Human Services added Pompe disease to the federal Recommended Uniform Screening Panel (RUSP) in 2015.

Why Screen for Pompe Disease?

Pompe disease can be detected using the traditional newborn dried bloodspot. Research has shown that early treatment and management can lead to better outcomes for those diagnosed with infantile-onset Pompe disease. Infants with Pompe disease can appear healthy at first, but without early intervention, many will die in early childhood. Treatments such as Enzyme Replacement Therapy (ERT) can lead to longer life and less invasive disease management, but must be administered before severe symptoms occur to be effective.

All Newborns Should Be Screened for Pompe Disease

The March of Dimes supports screening all newborns for conditions placed on the RUSP by the US. Secretary of Health and Human Services. The RUSP includes conditions for which there is a documented medical benefit to the affected infant from early detection and treatment; there is a reliable screening test for the disorder; and early detection can be made from newborn bloodspots or other means. Infants with Pompe disease have options for treatment. Early detection for Pompe disease through newborn screening will allow affected infants to receive life-saving interventions promptly.

The Ask: Will you support funding to add Pompe, MPS1 and X-ALD to NC's Newborn Screening Panel?

Key Points

- Pompe disease is a rare, heritable disorder that causes muscle weakness. Although cases can occur late in life, those with onset in infancy tend to be fatal in early childhood.
- Without treatment, Pompe disease causes muscle weakness that affects feeding, weight gain, movement, breathing, and heart contraction in infants. This leads to premature mortality.
- Evidence suggests that the earlier a newborn can receive treatment (e.g., within the first three months of life), the higher the success rate.
- Pompe disease can be identified using the traditional newborn dried bloodspot.
- Screening has been estimated to cost $1-2 per infant, in addition to laboratory start-up costs.
- Treatment with Enzyme Replacement Therapy can cost up to $100,000 annually, but delayed therapy may result in irreversible muscle damage.
- Addition to the newborn screening panel endorsed by Child Fatality Task Force and The NC Newborn Screening Advisory Committee.

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