

EXECUTIVE SUMMARY

This summary reviews the information the federal advisory committee used when deciding whether to recommend adding Pompe Disease to the Recommended Uniform Screening Panel (RUSP) in 2013.

About the condition

Pompe Disease is a rare disease caused by a change in a single gene. Studies of patients with symptoms suggest that between 1 and 2.5 out of every 100,000 people have Pompe Disease. People with Pompe Disease do not have enough of the GAA enzyme that helps the body break down stored sugar. Babies with the disease appear normal. There are 2 types of Pompe Disease: infantile- and late-onset. The first type can cause muscle problems that begin in early infancy. Most children with Pompe Disease have the late-onset type. Problems from the disease can worsen quickly and cause death within the first year.

Treatment for Pompe Disease

There is no cure for Pompe Disease. Early diagnosis allows early treatment and improves outcomes for babies with Pompe Disease. Enzyme replacement therapy can stop Pompe Disease problems from getting worse.

Detecting Pompe Disease in newborns

Newborn screening for Pompe Disease can happen along with routine newborn screening for other conditions in the first few days of life. Newborn Pompe Disease screening measures GAA enzyme activity. This process uses the same dried blood spots already collected to screen for other disorders. Newborns with low GAA activity have higher risk for the disease. They need more testing to diagnose the condition.

Public health impact

Based on what is known about screening and the risk of being born with Pompe Disease, experts think that screening all newborns in the United States for Pompe Disease would find about 144 babies with the disease each year (about 3.6 out of every 100,000 children born). It would prevent up to 28 people with the disease from needing a breathing machine and up to 19 deaths due to the disease each year.

Committee decision

The Committee voted in 2013 to recommend adding Pompe Disease to the RUSP. As of 2015, the RUSP recommends that state newborn screening programs include Pompe Disease.