



**Connecticut Department of Public Health**

**Testimony Presented Before the Public Health Committee**

**February 16, 2016**

**Commissioner Raul Pino, MD, MPH  
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**Raised Bill # 5132**

**An Act Concerning Newborn Screening for Pompe Disease**

The Department of Public Health (DPH) provides the following information in regards to Raised Bill # 5132. This bill would implement universal newborn screening for Pompe disease, bringing to sixty-eight the number of disorders in the newborn screening panel conducted by DPH's Public Health Laboratory (PHL).

Pompe disease is an inherited lysosomal storage disorder affecting about 1 in 28,000 people in the United States. It is caused by the buildup of a complex sugar called glycogen in the body's cells. This buildup in certain organs and tissues, especially muscles, impairs their ability to function normally. Researchers have described three forms of Pompe disease, differing in age of onset and severity.

The classic form of infantile-onset Pompe disease begins within a few months of birth. Affected infants experience muscle weakness, poor muscle tone, enlarged liver, heart defects, respiratory problems, and failure to thrive. If untreated, infantile-onset Pompe disease leads to fatal heart failure in the first year of life. The non-classic form of infantile-onset Pompe disease usually appears by age one, and is characterized by delayed motor skills, progressive muscle weakness, heart defects, and respiratory problems. Most children with non-classic infantile-onset Pompe disease live only into early childhood. Late-onset Pompe disease becomes apparent later in childhood, adolescence, or adulthood, and is typically milder, although progressive muscle weakness may lead to fatal respiratory failure. Treatment with enzyme replacement therapy is effective in preventing onset and advancement of symptoms, improving outcomes, and extending lifespans. In June 2013, the Discretionary Advisory Committee on Heritable Disorders in Newborns and Children, within the federal Department of Health and Human Services, conducted an evidence-based review of this disorder and advised this disorder be added to the Recommended Uniform Screening Panel.

There would be a significant cost to implement this screening, including one-time instrumentation and ongoing consumables/instrument maintenance contracts. As Governor Malloy stated in his 2016 State of the State Address, we must reset our expectations of what we can afford in light of the new economic reality facing Connecticut. Because an unbudgeted fiscal impact would result from implementing this new screening, the Department cannot support this proposal.

Thank you for your consideration of this information.

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