Date: July 21, 2022

From: South Dakota Department of Health Newborn Screening Program
State Hygienic Laboratory at the University of Iowa

RE: IMPLEMENTATION OF TESTING FOR A NEW DISORDER FOR NEWBORN SCREENING
Screening for Pompe disease will begin September 1, 2022

On July 19, 2022, the South Dakota Department of Health Newborn Screening Program completed the required administrative rule process to begin screening for Pompe disease.

Effective September 1, 2022, the South Dakota Department of Health Newborn Screening Program will begin screening for Pompe disease through the screening process of South Dakota’s contracted laboratory, the State Hygienic Laboratory at the University of Iowa.

On September 1, 2022, the State Hygienic Laboratory at the University of Iowa will begin charging birthing hospitals and providers for Pompe disease testing.

What is Pompe disease?
Pompe disease causes progressive muscle weakness and cardiomyopathy. Pompe disease is caused by pathological variation in the gene GAA, which codes for the enzyme “acid alpha-glucosidase” and which is needed for the normal processing of glycogen. This inability to process glycogen results in the accumulation of glycogen within cells and secondary cellular damage primarily affecting muscle tissues (both skeletal and cardiac). An individual must have two pathologic variations in the GAA gene in order to have Pompe disease (recessive inheritance). Early treatment of babies with infantile-onset Pompe disease is needed to avoid progressive, fatal cardiomyopathy. Without treatment, the infantile form of Pompe disease leads to death within the first two years of life. Later-onset forms of Pompe disease lead to physical disability due to muscle weakness and cardiomyopathy and increased mortality due to cardiomyopathy. Treatment for infantile-onset Pompe includes enzyme replacement therapy to correct the deficiency, as well as multi-disciplinary management of cardiopulmonary complications and functional problems. Treatment for late-onset Pompe disease also includes enzyme replacement therapy, but it is not given until the individual has symptoms.

Newborn screening for Pompe involves the measurement of acid-α-glucosidase (GAA) enzyme activity using Flow Injection Analyses-Tandem Mass Spectrometry (FIA-MS/MS). A screen positive result for Pompe is detected by a low or absent activity of the GAA enzyme, the biomarker for Pompe disease.

As a reminder, this is a screening test. A false negative or a false positive result must always be considered when screening. Therefore, clinical findings and status should be considered whenever interpreting laboratory results.