



DIVISION OF FAMILY & COMMUNITY HEALTH

Child & Family Services | Disease Prevention & Health Promotion

Date: August 24, 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**
please note updated implementation date

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 leads 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.

- The newborn screening fee will increase from the current rate of \$91.00 to \$98.00 on **September 1, 2022**.



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- There will be no charge for repeat specimens when requested by the South Dakota Department of Health or the State Hygienic Laboratory.
- The CPT procedure and service code used for the Newborn Screening panel, including Pompe disease screening, is **S3620**. This is a universal code and is the only code to use per APP and HIPPA.

HCPCS Level II codes are part of the standard procedure code set under the Health Insurance Portability and Accountability Act of 1996.

Newborn metabolic screening panel, includes test kit, postage, and the laboratory tests specified by the state for inclusion in this panel (eg, galactose; hemoglobin, electrophoresis; hydroxyprogesterone, 17-D; phenylalanine [phenylketonuria (PKU)]; and thyroxine, total)

- **NOTE:** The appropriate blood collection code (36416). **DO NOT** bill for each test separately
- **Specimen collection will not change from current procedures - nothing different needs to be done for the collection of the newborn blood spot specimen.**
- Additional Pompe disease information is available upon request. If interested, please contact the South Dakota Department of Health Newborn Screening Program at 605-983-1389. Pompe disease information is also available from the US National Library of Medicine (<https://medlineplus.gov/genetics/condition/pompe-disease/>) and Baby's First Test (<https://babysfirsttest.org/newborn-screening/conditions/pompe>).

Questions regarding Pompe disease screening may be directed to the following individuals:

Questions for the South Dakota Newborn Screening Program:

Bernadette Boes, RN
Newborn Screening Program Coordinator
South Dakota Department of Health
605-983-1389
Bernadette.boes@state.sd.us

SHL Newborn Screening Laboratory Questions:

Kenneth Coursey
Clinical Lab Supervisor, SHL Newborn Screening Laboratory
515-725-1630
kenneth-coursey@uiowa.edu

SHL Newborn Screen Billing Questions:

John Negley
State Hygienic Laboratory
2490 Crosspark Road
Coralville, IA 52241
319-335-4500/Fax 319-335-4171
shl-receivable@uiowa.edu



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Screening Follow-up and Treatment Questions:

Sharina Tveit, RN
South Dakota Newborn Screening Program
Follow-up Coordinator
605-312-0976
Sharina.tveit@sanfordhealth.org

The South Dakota Department of Health and the State Hygienic Laboratory intend to work closely with you to implement screening for Pompe disease with the goal of saving the lives and improving outcomes for affected infants. Please feel free to contact us with any questions regarding Pompe disease screening.

Sincerely,

Beth Dokken
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