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**State of Wisconsin**  
Department of Health Services

**DIVISION OF PUBLIC HEALTH**

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Date: December 1, 2021

To: Hospital Administrators, Newborn Nursery Managers, Neonatal Intensive Care Units (NICU), Special Care Nurseries, Primary Care Providers, Wisconsin Academy of Family Physicians, Wisconsin Chapter of the American Academy of Pediatrics, Wisconsin Guild of Midwives, Wisconsin Hospital Association, and Wisconsin Medical Society

From: Paula Tran  
State Health Officer and Administrator  
Division of Public Health

James J. Schauer, PhD, PE, MBA  
Director  
Wisconsin State Laboratory of Hygiene  
School of Medicine and Public Health  
University of Wisconsin-Madison

Re: **Emergency Rule Issued to Add Pompe disease to Newborn Screening**

The Wisconsin Department of Health Services (DHS) has added, by emergency rule, screening for Pompe disease to the Wisconsin Newborn Screening Program's panel of conditions. This rule will go into effect once it is published in the Wisconsin State Journal, which is expected to occur on **January 10, 2022**.

Every infant born in Wisconsin is required to have Pompe disease screening within 24-48 hours after birth.

**Why is it important to screen for Pompe disease?**

Pompe disease is a very serious disorder that can be fatal or cause significant disability when diagnosis and/or treatment is delayed. When Pompe disease is detected early and proper treatment is started many babies remain relatively healthy.

**How is screening for Pompe disease done?**

As with all other required newborn screening conditions detected in the *blood spot*, Pompe disease is screened by laboratory tests. This condition will simply be added to the current newborn screen panel; no additional blood spot card is required.

**What happens if a newborn has a positive screen?**

If a newborn has a positive screen, s/he will need a definitive test for Pompe disease. When the positive screen is obtained, the Wisconsin State Laboratory of Hygiene, at which the screening tests are performed, will contact the newborn's primary care provider to let them know of the need for further testing. An appropriate specialist will also be informed so the baby's primary care provider will have necessary professional support. This will be true for babies born out of hospital as well.

### **How can I find out more information about Pompe disease?**

Additional information on Pompe disease can be found at

<http://ghr.nlm.nih.gov/condition/pompe-disease> and

[https://www.ninds.nih.gov/Disorders/All-Disorders/Pompe-Disease-Information-Page#:~:text=onset%20Pompe%20disease.,Pompe%20disease%20is%20a%20rare%20\(estimated%20at%201%20in%20every,alpha%2Dglucosidase%20\(GAA\)\)](https://www.ninds.nih.gov/Disorders/All-Disorders/Pompe-Disease-Information-Page#:~:text=onset%20Pompe%20disease.,Pompe%20disease%20is%20a%20rare%20(estimated%20at%201%20in%20every,alpha%2Dglucosidase%20(GAA)))

For questions for the Department of Health Services, please email Robert Steiner, MD, Newborn Screening Program Medical Consultant for the Bureau of Community Health Promotion, at [Robert.Steiner@dhs.wisconsin.gov](mailto:Robert.Steiner@dhs.wisconsin.gov).

For questions for the Newborn Screening Laboratory at the Wisconsin State Laboratory of Hygiene, you can reach Mei Baker, MD, Co-Director for the Newborn Screening Laboratory by email at [mei.baker@wisc.edu](mailto:mei.baker@wisc.edu), or by phone at (608) 890-1796.

