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DIVISION OF PUBLIC HEALTH

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Date: September 13, 2019

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: Jeanne F. Ayers, R.N., M.P.H.
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 Carnitine Palmitoyltransferase 1A Deficiency and Spinal Muscular Atrophy to Newborn Screening**

The Wisconsin Department of Health Services (DHS) has added, by emergency rule, screening for Carnitine Palmitoyltransferase 1A deficiency (CPT1A) and Spinal Muscular Atrophy (SMA) 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 **October 15, 2019**.

Every infant born in a hospital is required to have CPT1A and SMA screening prior to discharge. Babies born out of hospital are also required to be screened.

Why is it important to screen for CPT1A and SMA?

CPT1A requires prompt diagnosis to prevent potential complications (such as hypoglycemia), especially during times of illness and other stress. The most severe types of SMA require early diagnosis and treatment to avoid significant neuromuscular (including respiratory) disability. Babies with undetected CPT1A or SMA are at risk for death or significant disability.

How are screening for CPT1A and SMA done?

As with all other required newborn screening conditions detected in the *blood spot*, CPT1A and SMA are screened by laboratory tests. These two conditions 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 CPT1A or SMA. 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 CPT1A and SMA?

Additional information on CPT1A can be found at the Newborn Screening Info CPT1 page.

<https://www.newbornscreening.info/Parents/fattyaciddisorders/CPT1.html>

Additional information about SMA can be found at Baby's First Test Spinal Muscular Atrophy page.

<https://www.babysfirsttest.org/newborn-screening/conditions/spinal-muscular-atrophy>

For questions for the Department of Health Services, please email Gary Kirk, MD, MPH, Chief Medical Officer for the Bureau of Community Health Promotion, at gary.kirk@dhs.wisconsin.gov or call him at (608)266-5818.

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 or by phone at (608)890-1796. You can also reach Patrice Held, PhD, Co-Director for the Newborn Screening Laboratory by email at patrice.held@slh.wisc.edu, or by phone at (608)265-5968.

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