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# Memo

**To:** Birthing Hospitals/Centers, Physician Offices & Health Departments

**From:** Scott J. Zimmerman, DrPH, MPH, HCLD (ABB)   
Director, NC State Laboratory of Public Health

**CC:** Jennifer Taylor, PhD

**Date:** August 5, 2016

**Re:** Newborn Screening Pilot for Mucopolysaccharidosis Type I (MPS-1)

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The North Carolina State Laboratory of Public Health (NCSLPH) in collaboration with RTI International will be conducting a pilot study to screen for mucopolysaccharidosis type 1 (MPS-1). MPS-1 is a lysosomal storage disorder that is caused by a missing enzyme,  $\alpha$ -L-iduronidase (IDUA). Infants with the severe form (previously referred to as Hurler syndrome) appear normal at birth but often have frequent respiratory infections, coarse features, skeletal changes, progressive developmental delay, progressive intellectual disability, and hearing loss. If left untreated, the severe form of MPS-1 usually leads to death in childhood.

The Health Resources and Services Administration's Advisory Committee on Heritable Disorders in Newborns and Children and the U.S. Secretary of Health and Human Services have recommended and approved this condition for inclusion in the Recommended Uniform Screening Panel (RUSP). The National Institutes of Health (NIH), Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD) awarded a contract to RTI in collaboration with the NCSLPH to rapidly conduct a pilot study on the implementation of a high throughput newborn screening test. The purpose of this pilot is to optimize testing conditions for identifying babies at greatest risk for MPS-1 and provide medical consultation to ensure appropriate care is provided to babies at greatest risk.





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The pilot study at the NCSLPH will use the dried blood spot specimen collected from the initial newborn heelstick specimen. In the majority of cases, no additional specimen will be required for MPS-1 risk assessment. The pilot study will be statewide and will evaluate 80,000 infant specimens. Results obtained during the pilot study will not appear on the newborn screening report. However, health care providers will be contacted if pilot study results identify an elevated risk for MPS-1 or if an additional specimen is needed to further evaluate the risk for MPS-1. Results obtained from babies that suggest an elevated risk for MPS-1 will be faxed to medical follow-up personnel, who will provide MPS-1 consultation services to the health care provider. Testing will begin on August 15, 2016 beginning with specimens received on August 11, 2016.

Please contact Dr. Jennifer Taylor at (919) 799-7834 or visit the MPS-1 pilot study website for more information: <http://ncnewbornscreeningpilots.org/MPS-1/>.

Thank you for your attention to this important announcement.

