STATE OF MICHIGAN
DEPARTMENT OF HEALTH AND HUMAN SERVICES
LANSING

RICK SNYDER
GOVERNOR

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DIRECTOR

October 2017

Newborn Screening for Lysosomal Storage Disorders
Pompe Disease and Mucopolysaccharidosis type I

Dear Practitioner(s):

Following recommendations by the Michigan Newborn Screening Quality Assurance Advisory Committee and approval by the Legislature, all Michigan newborns are now also screened for the lysosomal storage disorders, **Pompe disease** and **mucopolysaccharidosis type I** (MPS I or Hurler Syndrome). Until full integration with our laboratory software is completed in 2018, screening results for Pompe disease and MPS I will not appear on the laboratory report. Any abnormal results will be reported separately.

People with these conditions lack a specific enzyme function. The result is a build-up of large molecules in the lysosomes, which damages body tissues and organs. Babies with Pompe disease may be born with heart disease. They develop progressive muscle weakness affecting their ability to walk and breathe. Without enzyme replacement therapy, babies with infantile Pompe disease rarely live beyond their first year or two. We expect 2-3 babies with severe Pompe disease to be born in Michigan each year. MPS I causes progressive cognitive impairment and progressive problems with bones and joints. Without early treatment, life expectancy for a child with severe MPS I is 3 to 4 years. MPS I affects about 1 in 100,000 babies. With early detection by newborn screening, affected babies can be referred to a specialty center in the first weeks of life for treatment initiation.

The Michigan Department of Health and Human Services (MDHHS) Newborn Screening Laboratory uses enzyme activity assays to screen for these disorders. Acid alpha-glucosidase (GAA) activity is measured to test for Pompe disease and iduronidase (IDUA) activity for MPS I. Both screens are run using the dried filter paper blood spots collected as part of the current newborn screening process.

The NBS Program requests a repeat newborn screen for infants with borderline positive or inconclusive results for these disorders. For infants with strong positive results, the NBS Program notifies the primary care provider as well as one of our follow-up coordinating centers. Follow-up coordinating center staff have expertise in diagnosing and caring for patients with lysosomal storage disorders and will contact primary care providers to instruct them on the next steps after a strong positive screen result is received. To ensure the best possible outcome, it is very important to complete the follow-up steps as quickly as possible.

The lysosomal storage follow-up coordinating centers in Michigan are:

- Pediatric Genetics
  University of Michigan (Ann Arbor)
  Phone: (734) 764-0579
- Lysosomal Storage Disease Clinic
  Children's Hospital of Michigan (Detroit)
  Phone: (313) 832-9330

MDHHS and follow-up coordinating center staff will provide prompt communication and are available to answer questions. Additionally, more resources for providers and families are available on our website, www.michigan.gov/newbornscreening. Please do not hesitate to contact us at 517-335-4181 or newbornscreening@michigan.gov.

Sincerely,

Mary Kleyn

Mary Kleyn, MSc
Manager, Newborn Screening Follow-Up Program