STATE OF MICHIGAN
DEPARTMENT OF HEALTH AND HUMAN SERVICES
LANSING

RICK SNYDER
GOVERNOR

NICK LYON
DIRECTOR

September 2018

Newborn Screening Reporting 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 have been screened for the lysosomal storage disorders, **Pompe disease** and **mucopolysaccharidosis type I** (MPS I) since August 2017. Abnormal screening results for Pompe disease and MPS I have been reported separately from the rest of the newborn screening (NBS) results. Beginning October 2018, the screening results for Pompe Disease and MPS I will appear on the same report as the rest of the NBS results. A separate report for abnormal screening results for Pompe disease and MPS I will no longer be issued. Screening results for these two disorders will be in the row labeled "LSD" for lysosomal storage disorders (see below).

Disorder/Analyte(s)	Patient Screening Results	Expected Screening Results	Determination	Comments
Amino Acid Disorders	Within normal limits		Normal	
Fatty Acid Oxid. Disorders	Within normal limits		Normal	
Organic Acid Disorders	Within normal limits		Normal	
Endocrine Disorders	Within normal limits		Normal	
Enzyme Disorders	Within normal limits		Normal	
Hemoglobinopathy	Within normal limits		Normal	
Cystic Fibrosis	Within normal limits		Normal	
SCID	Within normal limits		Normal	
LSD	Within normal limits		Normal	

Second tier screening tests through Mayo Medical Laboratories have been implemented for both Pompe Disease and MPS I, which has reduced the number of infants with positive screens. Only infants who screen positive on the second tier test will be reported as positive. 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.

As a reminder, the follow-up coordinating centers for Pompe Disease and MPS I in Michigan are:

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

Sincerely,

Mary Kleyn

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