

Implementation of Testing for MPSI & Pompe Newborn Screening

April 19, 2023

Dear Newborn Screening Partners:

The Iowa Department of Health and Human Services (IHHS) is committed to promoting the health of newborns born in Iowa. Early detection of treatable hereditary and congenital disorders allows for the prevention or reduction of symptoms and, ultimately, saves babies' lives. Currently, newborns in Iowa are screened for 30 core disorders (full list can be found on the IDPH website: <https://idph.iowa.gov/newborn-screening>). The IHHS has authorized the Iowa Newborn Screening Program (INSP) to begin routine screening for Mucopolysaccharidosis type I (MPSI) and Glycogen storage disease type 2 (Pompe) on specimens received 05/15/2023. The addition of MPSI and Pompe requires no changes in the current collection and transportation of specimens.

MPSI is an autosomal recessively inherited lysosomal storage condition that results in the inability of a body to produce an enzyme called lysosomal alpha-L-iduronidase. Lack of this enzyme leads to build up of long chain sugar molecules called glycosaminoglycans. MPSI can be classified as a severe or attenuated case based on the age of onset, and severity of symptoms varies. Severe cases are characterized by deposits of glycosaminoglycans leading to skeletal abnormalities, cognitive impairment, heart disease, respiratory problems, enlarged liver (hepatomegaly) and spleen, characteristic facies, and reduced life expectancy. Attenuated forms of MPSI show a variability in onset and progression of disease and degree of cognitive impairment.

Pompe disease is an autosomal recessively inherited metabolic disorder that results in a deficiency in the enzyme acid alpha-glucosidase (acid maltase), needed to break down glycogen. Glycogen accumulation in lysosomes limits the cell's ability to break down waste products and impairs cell's functions, especially muscle cells. Onset of symptoms varies and is classified into two categories: infantile-onset, and late-onset. Infantile onset is the most common form and is characterized by muscle weakness (myopathy), poor muscle tone (hypotonia), an enlarged liver (hepatomegaly), cardiomyopathy, failure to thrive, breathing problems, and if left untreated, heart failure and death.

The decision to provide routine reporting is based on recommendations made by medical consultants to the Center for Congenital and Inherited Disorders after full evaluation of data collected during a pilot study conducted by INSP to identify Iowa infants at risk for MPSI & Pompe. The official addition of MPSI and Pompe on 05/15/23 requires no changes in the current collection and transportation of specimens, and the newborn screening fee will increase at the beginning of the fiscal year on **July 1, 2023**. Please refer to letter dated "03-15-2023" and titled "INBSP_Fee_Increase_notification" for more information about the newborn screening fee increase.

Public Health

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The State Hygienic Laboratory will test for enzyme activity of acid α -glucosidase (GAA), and α -L-Iduronidase (IDUA) using tandem mass spectrometry. Second tier testing will be performed for all specimens found to be presumptive positive on the first-tier screen. Newborn screening follow-up program staff will follow up on all out-of-range results with the patient's primary care provider, or hospital the newborn is currently receiving care, with results, recommendations, and education. Medical consultants will be available to assist with all abnormal assay results.

A reminder, this is a screening test. A false negative or a false positive result must always be considered when screening. Therefore, clinical findings and status should be considered whenever interpreting laboratory results.

Questions regarding MPSI and Pompe screening may be directed to the following persons

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