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MPS II and SMA Newborn Screening in Missouri Go Live

Dear Practitioner, Laboratory Manager, or Newborn Nursery Manager:

The Missouri Department of Health and Senior Services' Newborn Screening (NBS) Program launched full population pilots in November 2018 and January 2019 to screen all newborns born in Missouri for MPS II and Spinal Muscular Atrophy (SMA), respectively. While screening results for these two disorders have not appeared on the routine NBS laboratory reports during this pilot/implementation period, all newborns were tested for these disorders, and any displaying high risk abnormal results were referred for further confirmatory testing. During this pilot/implementation phase, one infant with MPS II and three infants with SMA were successfully detected, confirmed, and provided life-saving intervention.

Effective October 1, 2019, the Missouri State Public Health Laboratory (MSPHL) will be going live with the routine reporting of these disorders on the standard NBS laboratory reports. MPS II results will be reported under the already existing Lysosomal Storage Disorder category while SMA will appear as an additional disorder category.

MPS II is a lysosomal storage disorder caused by a deficiency or absence of an enzyme called iduronate-2-sulfatase (IDS) and is a rare, progressive, and debilitating genetic disorder that interferes with the body's ability to break down and recycle a specific cellular substance. As this substance accumulates throughout the body's cells, signs of MPS II become more visible. MPS II affects approximately 1 in 100,000 - 170,000 total live births, and affects predominantly males. Symptoms may not present at birth, but frequently appear in early childhood and may include delayed development, enlarged internal organs, heart conditions, stunted growth, skeletal abnormalities, and joint problems. The range of symptoms and effect of the disease on individuals varies widely; however, MPS II is always progressive and life-limiting.

SMA is a progressive neurodegenerative disease that affects the motor nerve cells in the spinal cord which impedes the ability to sit up, walk, swallow, and in the most severe cases, breathe. It is the leading genetic cause of death for infants, affecting approximately 1 in 6,000 - 11,000 infants nationally. The U.S. Food and Drug Administration recently approved a promising new treatment for infants with SMA. Infants who are diagnosed before exhibiting symptoms and who receive treatment have achieved unprecedented milestones such as sitting, standing, and walking. Screening newborns for SMA is nationally recommended and was added to the Recommended Uniform Screening Panel by the Secretary of Health and Human Services in 2018.

All infants with high risk abnormal results for MPS II will continue to be referred for confirmatory testing at one of the four contracted genetic referral centers in Missouri. All infants with high risk abnormal results for SMA will continue to be referred to the SMA follow-up program manager, who will contact the primary care provider with more detailed information regarding recommended follow-up and confirmatory testing.

If you have questions or comments, please contact: Lori Swartz, NBS follow-up program manager, 573-751-6266, or by email at Lori.Swartz@health.mo.gov or Tracy Klug, Chief of Newborn Screening Laboratory, 573-751-2662, or by email at Tracy.Klug@health.mo.gov