MLS Laboratory Update:

FEBRUARY 24, 2025

Purpose of this Message:

To inform MLS Laboratories that screening for Duchenne muscular dystrophy (DMD) and guanidinoacetate methyltransferase (GAMT) deficiency has begun.

Action Item:

Read the notification and share with other laboratorians and clinical partners at your site.

Laboratory Specific Information:

- Effective Monday, February 24, 2025, samples received by our laboratory are being tested for Duchenne muscular dystrophy (DMD) and guanidinoacetate methyltransferase (GAMT) deficiency.
- No change in specimen requirements or cost.
- Changes have been made to the newborn screening report. Duchenne muscular dystrophy will have it's own line in the grid. GAMT deficiency will be included in the Amino Acid Profile already present in the report.

Background:

The Minnesota Newborn Screening Program is pleased to announce that we are screening for Duchenne muscular dystrophy (DMD) and guanidinoacetate methyltransferase (GAMT) deficiency. The addition of DMD and GAMT deficiency will not add to the cost of newborn screening specimen cards at this time. Changes have been made to the newborn screening report. Duchenne muscular dystrophy will have it's own line. GAMT deficiency will included in the Amino Acid Profile already present in the report. Please reference the MLS alert that was sent on October 22, 2024 (https://www.health.state.mn.us/diseases/idlab/mls/labalerts/241022nbsprofileupdate.pdf) for additional information. An updated Newborn Screening report is available as well: Minnesota Department of Health Newborn Screening Final Report Example (https://www.health.state.mn.us/diseases/idlab/mls/labalerts/241022nbssamplereport.pdf).

Duchene muscular dystrophy (DMD) is caused by a mutation causing muscle breakdown. Muscle weakness is first noticed in legs and arms, eventually leading to loss of mobility and weakness of internal muscles needed for breathing. Symptoms of DMD, if not identified in the newborn period, can go unrecognized for years. On average, DMD is not diagnosed until the

age of 5, at which point muscle may already be severely damaged. Based on the experience of other states and Minnesota's birth rate, we expect about 300 infants per year to require additional testing. We expect about 6-10 of these infants to be diagnosed with DMD.

Please note, to reduce the number of unnecessary diagnostic test and referrals for DMD, if the initial newborn screen shows an elevated level of CK-MM (an analyte used to measure skeletal muscle damage), our genetic counselors will be contacting providers to order a second newborn screen to be **collected AFTER 2 weeks of age** to allow time for elevations in CK-MM to normalize after the birthing process.

Based on what we've heard from other states, **you will be seeing an increase in repeat specimens requested from us and will likely also see an increase in your outpatient lab orders**.

Screening for DMD among newborns allows families and medical specialists to tailor care, such as physical therapy, based on approved treatments available for children at certain ages. Disease-modifying treatments during the newborn period are available for 30% of affected newborns. Treatments are not curative, but symptoms and muscle breakdown are less severe.

Guanidinoacetate methyltransferase (GAMT) deficiency is a lifelong metabolic disorder causing a toxic buildup that results in serious brain and muscle problems if left untreated. GAMT deficiency symptoms typically begin before the age of 1 and as late as 3 years. These symptoms include late sitting, walking, speaking, and growth. If untreated, most children develop learning and behavioral challenges. Based on Minnesota's birthrate, it is expected that 0-1 children each year will benefit from GAMT deficiency being included in the newborn screen.

Additional Information:

As a reminder, S3620 is a valid healthcare common procedure coding system (HCPCS) code for newborn metabolic screening panel, it includes test kit, postage, and the laboratory tests specified by the state for inclusion in this panel. For repeat screens, report S3620 with the appropriate modifier for repeat services (-76, -77). This HCPCS code is provided as a resource to healthcare professionals. The final billing-related decisions must be made by the healthcare provider.

Please share this letter and its enclosure with the appropriate personnel within your hospital system. We hope you find this information helpful in your preparation, and we thank you for your continued support of newborn screening.

Questions: Please contact: Carrie Wolf, our Newborn Screening Program manager at carrie.wolf@state.mn.us or 651-201-5458.

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PLEASE FORWARD THIS TO ALL APPROPRIATE PERSONNEL WITHIN YOUR INSTITUTION AND HEALTH SYSTEM

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