STATE OF MICHIGAN
DEPARTMENT OF HEALTH AND HUMAN SERVICES
LANSING

GRETCHEN WHITMER
GOVERNOR

ELIZABETH HERTEL
DIRECTOR

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Newborn Screening for guanidinoacetate methyltransferase (GAMT) deficiency

Dear Practitioner(s):

Following recommendations by the Michigan Newborn Screening (NBS) Quality Assurance Advisory Committee and approval by the Legislature required by law (MCL 333.5431), all Michigan newborns will be screened for **guanidinoacetate methyltransferase (GAMT) deficiency** beginning September 19, 2022.

GAMT deficiency is an inborn error of metabolism that affects creatine synthesis. When the guanidinoacetate methyltransferase enzyme is damaged, creatine cannot be synthesized and guanidinoacetate (GAA) accumulates. This creates a shortage of creatine in the body and excessive amounts of GAA which is a neurotoxin. Creatine is an essential metabolite for regenerating energy, particularly in the brain. When the body does not get enough creatine and has extra GAA, it can cause developmental delay, speech problems, movement disorders, seizures and behavior issues such as autism and hyperactivity. Lack of early treatment can lead to lifelong cognitive impairments which can be severe. Treatment for GAMT deficiency consists of correcting the creatine deficiency and reducing GAA levels in the body and brain through dietary and medical interventions. Treatment is most effective if started early in life before symptoms arise. Michigan will be the third state in the United States to screen for GAMT deficiency.

The newborn will be screened for GAMT deficiency using the dried filter paper blood spots collected as part of the current newborn screening process. Analytes for GAMT deficiency include guanidinoacetate (GUAC) and the ratio of GUAC/creatine (CRE). The Michigan Department of Health and Human Services (MDHHS) Newborn Screening Laboratory will use a tandem mass spectrometry method to measure these analytes.

The NBS Program will request a repeat newborn screen for infants with borderline positive or inconclusive results for this disorder. For infants with strong positive results, the NBS Program will notify the primary care provider, as well as the follow-up coordinating center. Follow-up coordinating center staff will contact primary care providers to instruct them on the next steps after a strong positive screen result is received. The baby will be referred to a medical management center with expertise in diagnosing and caring for patients with GAMT deficiency. To ensure the best possible outcome, it is very important to complete the follow-up steps in a timely manner.

The GAMT deficiency follow-up coordinating center in Michigan is:

- 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