DEPARTMENT OF HEALTH AND HUMAN SERVICES

Advisory Committee on Heritable Disorders
in Newborns and Children
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November 2, 2017

Nicola Longo, M.D., Ph.D.
Professor and Chief
Medical Genetics/Pediatrics
University of Utah
295 Chipeta Way
Salt Lake City UT 84108

Marzia Pasquali, Ph.D., FACMG
Professor of Pathology
University of Utah School of Medicine
Medical Director, Biochemical Genetics and
Supplemental Newborn Screening

Dear Drs. Longo and Pasquali:

The Advisory Committee on Heritable Disorders in Newborns and Children (Committee) appreciates your updated nomination of Guanidinoacetate Methyltransferase (GAMT) Deficiency for inclusion on the Recommended Uniform Screening Panel (RUSP). As part of the formal review process, the Committee conducted a review of the updated materials provided for the nomination package. During the November 2016 Committee meeting results were presented for discussion. A copy of the presentation is enclosed.

The Committee recognizes GAMT as a medically serious condition which warrants consideration for inclusion on the RUSP. The Committee has determined that most of the criteria needed for moving the nomination of GAMT to evidence review have been met. GAMT results in serious neurodevelopmental sequellae for affected persons, there is a well-established case definition, the newborn screening test appears reasonable for the newborn screening system, and treatment is effective. However, at this time, the Committee has decided to not send the nomination forward for evidence review. The Committee's decision is based specifically on the fact that at this time, no cases have been identified prospectively through a newborn screening system. The Committee determined that the case presented in the revised nomination package did not meet the criterion of a prospectively identified case. The Committee criterion is that "the study should evaluate the newborn screening process

from collection through diagnosis and identify at least one screen positive newborn with confirmation of presence of the condition under consideration."

Since one state in the United States and several other countries are conducting prospective pilot studies on newborn screening for GAMT, the Committee has been following the results of these efforts and encourages you to resubmit the nomination when you become aware of the identification and diagnosis of at least one patient is made prospectively through a newborn screening system.

The Committee has also been following what is happening in the field. Of note, the Centers for Disease Control and Prevention has published a screening method for GAMT deficiency, has laboratory quality assurance materials, and is available to provide technical assistance for any state program that is interested in implementing newborn screening for GAMT deficiency.

The Committee also encourages the submission to the Committee of any updated and formalized treatment guidelines when available.

Please contact Dr. Catharine Riley (criley@hrsa.gov) when you have additional data to submit or if you have any questions or concerns.

Thank you for your nomination of GAMT for inclusion in the RUSP for state newborn screening programs. I look forward to hearing from you soon.

Sincerely yours,

/s/

Joseph A. Bocchini, Jr., M.D.
Chairperson

Enclosure:
ACHDNC Nomination and Prioritization Workgroup Presentation: GAMT

cc: Catharine Riley, PhD, MPH
Designated Federal Official
Health Resources and Services Administration