DEPARTMENT OF HEALTH AND HUMAN SERVICES

Advisory Committee on Heritable Disorders
in Newborns and Children
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www.hrsa.gov/heritabledisorderscommittee

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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 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 nomination package. During the May 2016 Committee meeting results were presented for discussion. A copy of the presentation is enclosed.

The Committee recognizes GAMT as a medically important disorder that deserves serious consideration. GAMT has a well-established case definition, the performance characteristics of the proposed screening test appear to allow its incorporation into the newborn screening system, and treatment is noninvasive. However, at this time, the Committee has decided that the evidence available on the ability of the screening test to detect an affected person has not met our criteria. Therefore, the Committee decided to not send the nomination forward for evidence review. The Committee's decision is based primarily on the determination that the analytic validity of the screening test has not yet been determined, in part because no cases have been identified prospectively through newborn screening to date.

Since one state in the United States and several other countries are conducting prospective pilot studies on newborn screening for GAMT, the Committee encourages you to resubmit the nomination when identification of at least one patient has been made prospectively through newborn screening. The Committee will expedite its review to determine whether the new data provides sufficient support to request a formal review of the scientific evidence by the Committee's condition review workgroup.

Please contact Ann Ferrero (aferrero@hrsa.gov) when you are prepared to submit any additional data 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,

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

Enclosure:
ACHDNC Nomination and Prioritization Workgroup Presentation: GAMT

cc: Debi Sarkar, M.P.H.
Designated Federal Official
Health Resources and Services Administration