WASHINGTON STATE BOARD OF HEALTH

Date: March 8, 2023

To: Washington State Board of Health Members

From: Kelly Oshiro, Board Chair

Subject: Petition – Chapter 246-650 WAC, Newborn Screening, Request to add Guanidinoacetate methyltransferase (GAMT) deficiency

Background and Summary:

The Administrative Procedures Act (RCW 34.05.330) allows any person to petition a state agency to request the adoption, amendment, or repeal of any rule. Upon receipt of a petition, the agency has sixty days to either (1) deny the petition in writing, stating the reasons and, as appropriate, offer other means for addressing the concerns raised by the petitioner, or (2) accept the petition and initiate rulemaking.

On February 24th, 2023, the Washington State Board of Health (Board) received a petition for rulemaking requesting an amendment to chapter 246-650 WAC to add Guanidinoacetate methyltransferase (GAMT) deficiency as a condition for newborn screening. The petitioner's request indicates they submitted the petition because GAMT deficiency was recently approved by the federal Advisory Committee on Heritable Disorders in Newborns and Children in June of 2022 for inclusion in the federal Recommended Uniform Screening Panel (RUSP).[1] The Secretary of Health and Human Services endorsed this decision on January 4th, 2023.[2]

GAMT deficiency is an inherited condition that prevents the body from properly producing creatine, an amino acid that helps organs store and use energy.[3,4] Without enough creatine, the body's organs do not get enough energy to support its vital functions, which can cause damage. This damage primarily affects the brain and muscles, as these organs need the most energy. Without early treatment, GAMT deficiency can cause serious cognitive impairments, and result in developmental delays for speech and mobility, intellectual disabilities, uncontrolled movements, muscle weakness, and seizures.

The Board has the authority under RCW 70.83.050 to adopt rules for screening Washington-born infants for hereditary conditions. WAC 246-650-010 defines the conditions, and WAC 246-650-020 lists the conditions for which all Washington-born newborns are to be screened.

Before an advisory committee is convened to review a candidate condition against the Board's five newborn screening requirements, a preliminary review (or qualifying assumption analysis) should be done to determine whether there is sufficient scientific evidence available to apply the criteria for inclusion. The Board uses a defined set of

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criteria to evaluate conditions for inclusion on the panel. These criteria are: available screening technology, diagnostic testing and treatment available, prevention potential and medical rationale, public health rationale, and cost-benefit/cost-effectiveness. If there is enough scientific evidence available to apply these criteria to a condition, the Board may then decide to convene a technical advisory committee (TAC) to inform its decision on whether a condition should be included in the newborn screening (NBS) panel.

I have invited Molly Dinardo, Board Staff, and John Thompson, Director of the Department of Health's Newborn Screening Program, to present preliminary information on GAMT deficiency and an overview of the Board's process and criteria for evaluating conditions for inclusion in the rule.

Recommended Board Actions:

The Board may wish to consider one of the following motions:

The Board declines the petition for rulemaking to add GAMT deficiency as a condition for newborn screening in Chapter 246-650 WAC but directs staff to work with the Department of Health to convene a technical advisory committee to evaluate GAMT deficiency using the Board's process and criteria to evaluate conditions for inclusion in WAC 246-650-020 and then make a recommendation to the Board.

OR

The Board determines that there is insufficient information available at this time to know whether GAMT deficiency meets the qualifying assumption for the Board's evaluation criteria for inclusion in the rule. The Board directs staff to update the information in two years and return to the Board for consideration at that time.

OR

The Board accepts the petition for rulemaking to amend Chapter 246-650 WAC to add Guanidinoacetate methyltransferase (GAMT) deficiency as a condition for newborn screening. The Board directs staff to notify the requestor of its decision and to file a CR-101, Preproposal of Inquiry, under its authority in RCW 70.83.050.

Staff

Molly Dinardo

1. Powell, Cynthia M. Advisory Committee on Heritable Disorders in Newborns and Children (ADHNC). ADHNC Chair Letter to Secretary of Health and Human Services for new ADHNC recommendation for Guanidinoacetate Methyltransferase Deficiency. June 7, 2022. Accessed February 22, 2023. https://www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/meetings/chair-letter-secretary-gamt.pdf
2. Becerra, Xavier. Secretary of Health and Human Services. Letter Response to ADHNC Recommendation Letter. January 4, 2023. Accessed February 22, 2023. https://www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/meetings/gamt-signed-response.pdf
3. National Institutes of Health| Guanidinoacetate methyltransferase deficiency | About the Disease | Genetic and Rare Diseases Information Center. Last Updated February 2023. Accessed February 22, 2023. https://rarediseases.info.nih.gov/diseases/2578/guanidinoacetate-methyltransferase-deficiency
4. Health Resources and Services Administration (HRSA) | Guanidinoacetate methyltransferase deficiency | Newborn Screening. Last Updated December 2022. Accessed February 22, 2023. https://newbornscreening.hrsa.gov/conditions/guanidinoacetate-methyltransferase-deficiency