WASHINGTON STATE BOARD OF HEALTH

Date: October 9, 2023

To: Washington State Board of Health Members

From: Kelly Oshiro, Board Vice Chair

Subject: Briefing – Recommendations of the GAMT and ARG1-D Newborn Screening Technical Advisory Committee

Background and Summary:

The Washington State Board of Health (Board) has the authority under RCW 70.83.050 to define and 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. To determine which conditions to include in the newborn screening panel, the Board convenes a multi-disciplinary technical advisory committee (TAC) to evaluate candidate conditions using guiding principles and an established set of criteria.

Earlier this year, the Board received petition requests to review Guanidinoacetate methyltransferase (GAMT) Deficiency and Arginase 1 Deficiency (ARG1-D) for possible inclusion in the state's newborn screening panel. After a preliminary review of these conditions at the Board's March and April 2023 meetings, Board Members directed Board staff to work with the Department of Health (Department) to convene a TAC for further evaluation.

GAMT Deficiency is a rare inherited metabolic condition that prevents the body from properly producing creatine.[1] Without enough creatine, organs like the brain and muscles cannot get enough energy, leading to neurological issues.[1,2] Children with this condition may begin showing symptoms from 3 months to age 3. Without early treatment, GAMT Deficiency can cause serious cognitive impairments and result in developmental delays, impaired speech and mobility, uncontrolled movements, muscle weakness, and seizures. However, when treatment is initiated early, existing literature shows that GAMT appears to be highly treatable.[3] Currently, the states of Michigan, New York, and Utah, as well as British Columbia, Ontario, and Victoria, Australia, screen for GAMT Deficiency. Approximately 130 individuals have been diagnosed with the condition worldwide.

ARG1-D is a rare inherited metabolic condition that causes the amino acid arginine (a building block of proteins) and ammonia to accumulate in the blood.[4,5] When ammonia levels become too high, it has toxic effects and can cause serious damage to the nervous system and other parts of the body. Signs of the condition can occur from

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about age 1 to 3, and symptoms include seizures, muscle tightness or stiffness, difficulty eating, vomiting, and trouble breathing.[6] Existing literature shows that individuals diagnosed later in life have an increased risk of disability and death, but early identification can allow babies to begin treatment early and lower arginine levels. While ARG1-D is only partially treatable, even a partial reduction in arginine has meaningful impacts on disease outcomes. Over 30 states currently screen for ARG1-D, and less than 260 individuals have been diagnosed with the condition worldwide.

The TAC Met on September 8th, 2023, to consider both conditions against the Board's five newborn screening criteria. During the committee meeting, TAC Members heard presentations on the natural history of the conditions, diagnostic testing and treatment, available screening technology, and cost-benefit analyses for adding these conditions to the state's screening panel. The TAC then voted on individual criteria for each condition as well as overall recommendations to the Board.

I have invited John Thompson, Director of the Department of Health's Newborn Screening Program, and Molly Dinardo, Policy Advisor to the Board, to present information from the GAMT and ARG1-D TAC meeting. First, they will present information and TAC recommendations for GAMT Deficiency, followed by ARG1-D, for Board Member consideration.

Recommended Board Actions:

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

GAMT Deficiency

The Board directs staff to file a CR-101 to initiate rulemaking for chapter 246-650 WAC to consider adding Guanidinoacetate methyltransferase (GAMT) Deficiency to the Washington State newborn screening panel.

OR

The Board determines that Guanidinoacetate methyltransferase (GAMT) should not be considered for addition to the newborn screening panel at this time and, further, that GAMT be reevaluated in two years as a candidate for mandatory newborn screening in Washington State.

ARG1-D

The Board directs staff to file a CR-101 to initiate rulemaking for chapter 246-650 WAC to consider adding Arginase 1 Deficiency (ARG1-D) to the Washington State newborn screening panel.

OR

The Board determines that Arginase 1 Deficiency (ARG1-D) should not be considered for addition to the newborn screening panel at this time and, further, that ARG1-D be

reevaluated in two years as a candidate for mandatory newborn screening in Washington State.

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