WASHINGTON STATE BOARD OF HEALTH

Date: March 12, 2025

To: Washington State Board of Health Members

From: Kelly Oshiro, Board Member

Subject: Recommendations of the Branch-Chain Ketoacid Dehydrogenase Kinase Deficiency Newborn Screening Technical Advisory Committee

Background and Summary:

The Washington State Board of Health (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 on the state's required newborn screening panel.

During the 2023-2024 legislative session, Senate Bill 6234 passed, which directed the Board to conduct a review of branch-chain ketoacid dehydrogenase kinase (BCKDK) deficiency for Washington's mandatory newborn screening panel. BCKDK deficiency is a rare inherited genetic disorder that leads to a deficiency of branched-chain amino acids. It is caused by changes in the BCKDK gene, which produces the BCKDK enzyme. The BCKDK enzyme regulates the metabolism of branched-chain amino acids. Mutations with the BCKDK enzyme cause an overactive breakdown of branched-chain amino acids[1]. Without enough amino acids, proteins can't form properly, which impairs neurodevelopmental growth and development[1,2]. Signs and symptoms can vary but may include autism spectrum disorder, language impairment, seizures, and microcephaly[2]. There are 21 cases of BCKDK deficiency identified worldwide, with no cases yet identified in the United States[2].

On January 14, 2025, a technical advisory committee (TAC) convened to consider this condition against the Board's five newborn screening criteria. During the committee meeting, TAC Members heard presentations on the natural history of the condition, diagnostic testing and treatment, available screening technology, and cost-benefit analysis for adding this condition to the state's screening panel. The TAC then voted on individual criteria for BCKDK deficiency as well as an overall recommendation to the Board.

I have invited Megan McCrillis, Policy Analyst for the Department of Health's Newborn Screening Program, and Kelly Kramer, Policy Advisor to the Board, to present information from the BCKDK deficiency TAC for Board Member consideration.

Recommended Board Actions:

The Board may wish to consider and amend, if necessary, the following motions:

The Board directs staff to file a CR-101 to initiate rulemaking for chapter 246-650 WAC to consider adding branch-chain ketoacid dehydrogenase kinase (BCKDK) deficiency to the Washington state newborn screening panel.

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OR

The Board determines that branch-chain ketoacid dehydrogenase kinase (BCKDK) deficiency should not be considered for addition to the newborn screening panel at this time.

Staff

Kelly Kramer, Policy Advisor

1. Novarino, G., et al. Mutations in BCKD-kinase lead to a potentially treatable form of autism with epilepsy. Science 338: 394-397, 2012. [PubMed: 22956686]
2. Tangeraas, T., et al. BCKDK deficiency: a treatable neurodevelopmental disease amenable to newborn screening. Brain 146: 3003-3013, 2023. [PubMed: 36729635]