WASHINGTON STATE BOARD OF HEALTH

**Date:** August 7, 2024

**To:** Washington State Board of Health Members

**From:** Kelly Oshiro, Board Member

**Subject:** Petition – Chapter 246-650 WAC, Newborn Screening, Request to add Wilson's Disease

## 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 July 26, 2024, the Washington State Board of Health (Board) received a rulemaking petition requesting to amend chapter 246-650 WAC to add Wilson's Disease as a condition for newborn screening. The petition states that early diagnosis of Wilson's Disease and pre-emptive treatment prior to symptom onset can prevent life-threatening complications.

Wilson's Disease is a rare and inherited metabolic disease that prevents the body from properly eliminating copper, causing it to accumulate in body tissues, especially the liver, brain and corneas of the eyes. Wilson's Disease is caused by two copies of an abnormal gene (*ATP7B*) that would normally help transport copper out of liver cells, allowing copper to be eliminated from body.[2,5] Excess copper then builds up in the liver and eventually in the blood and other organ systems.[3] Too much copper is toxic to the body's tissues and can lead to serious damage to the liver, nervous system, and other organ systems.[3]

Symptoms of Wilson's Disease vary widely and can occur anytime after age 3 through the age of 70.[1,3] People with Wilson's Disease may develop symptoms related to liver dysfunction initially, such as yellowing of skin and eyes, swelling, vomiting, and fatigue. Other people with Wilson's Disease may only develop brain-related symptoms such as behavioral changes, difficulty swallowing, muscle rigidity, difficulty with speech, and lack of coordination.[2,3]

Wilson's Disease is not being screened by any other state currently and has yet to be added to the Federal Recommended Uniform Screening Panel (RUSP). However, Washington's newborn screening lab has been working on a pilot project in partnership with Key Proteo in which 50,000 de-identified samples from residual newborn screening

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specimens have been tested to demonstrate the feasibility of a population-based screening test for Wilson's Disease.[4]

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 on the state's required newborn screening panel.

The Board has a process it follows when considering new conditions to include in the state's newborn screening panel. To determine which conditions to include in the panel, the Board may convene an advisory committee to evaluate candidate conditions using guiding principles and an established set of criteria. Before an advisory committee is convened, there should be sufficient scientific evidence available to apply the Board's criteria for inclusion. This may require a preliminary review.

I have invited Kelly Kramer, Board staff, and John Thompson, Director of the Department of Health's Newborn Screening Program, to provide an overview of the Board's process for adding a condition to the panel, the petition request, and a brief overview of Wilson's Disease

## Recommended Board Actions:

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

The Board declines the petition for rulemaking to add Wilson's Disease as a condition for newborn screening in Chapter 246-650 WAC, and directs staff to work with the Department of Health to perform a preliminary review of the condition for inclusion in WAC 246-650-020 and then report back to the Board so the Board can determine whether to establish a technical advisory committee to evaluate Wilson's Disease against the Board's criteria for adding conditions to the newborn screening rule.

OR

The Board declines the petition for rulemaking to add Wilson's Disease as a condition for newborn screening in Chapter 246-650 WAC, and directs staff to work with the Department of Health to move forward with convening a technical advisory committee to evaluate Wilson's Disease 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 accepts the petition for rulemaking to amend Chapter 246-650 WAC to add Wilson's Disease 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.

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Staff
Kelly Kramer

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PO Box 47990 • Olympia, WA 98504-7990
360-236-4110 • wsboh@sboh.wa.gov • sboh.wa.gov

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2. National Organization for Rare Disorders. Wilson Dease. Published March 2018. Accessed July 22, 2024. https://rarediseases.org/rare-diseases/wilson-disease
3. National Institutes of Health National Library of Medicine. Wilson Disease: StatPearls . Published August 2023. Accessed July 22, 2024. https://www.ncbi.nlm.nih.gov/books/NBK441990/
4. Key Proteo. Key Proteo Pilots Newborn Screening in Washington State. Published Mar 2022. Accessed July 22, 2024.
5. National Institutes of Health National Library of Medicine. Epidemiology, diagnosis, and treatment of Wilson's disease. Published November 2017. Accessed July 24, 2024. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5735277/