Petition for Rulemaking: Newborn Screening

Chapter 246-650 WAC

Wilson's Disease

Kelly Kramer, Policy Advisor - August 7, 2024

WASHINGTON STATE BOARD OF HEALTH

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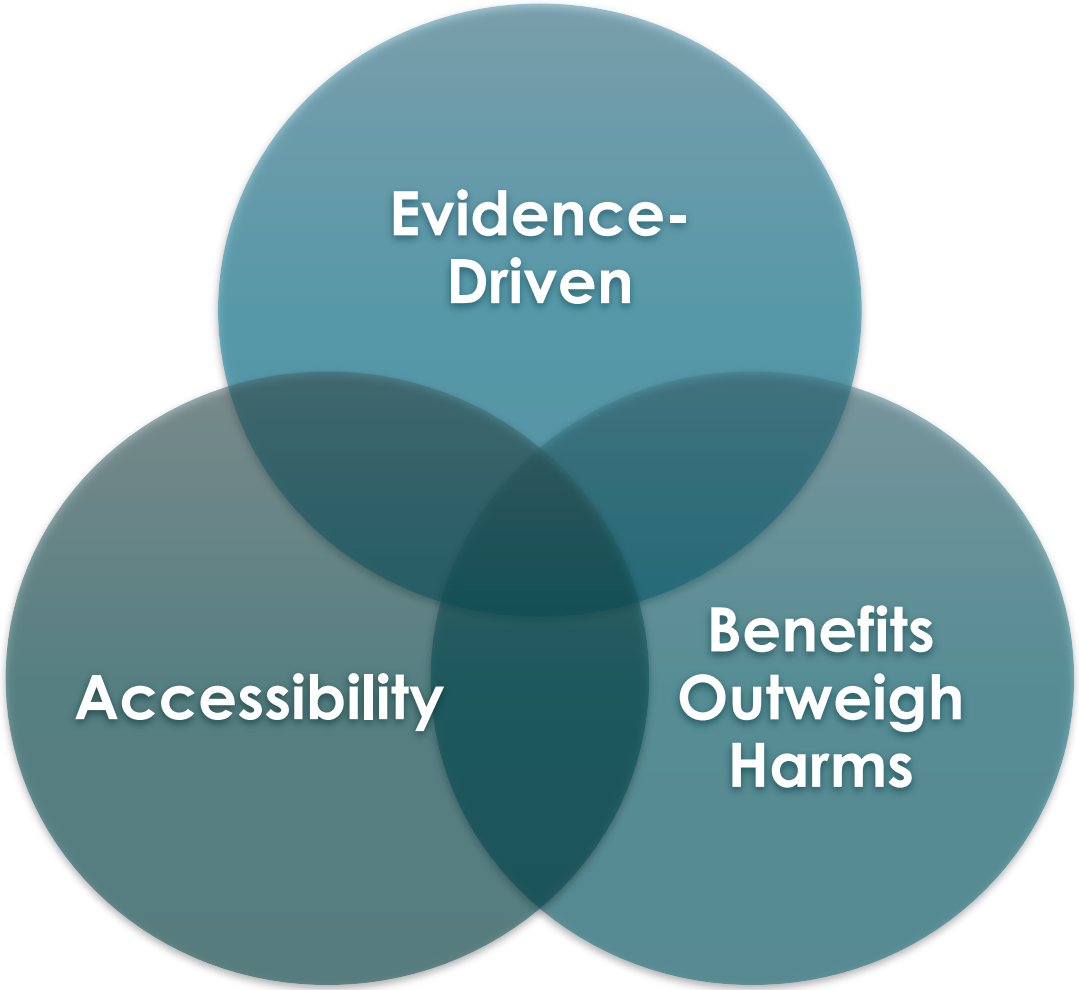
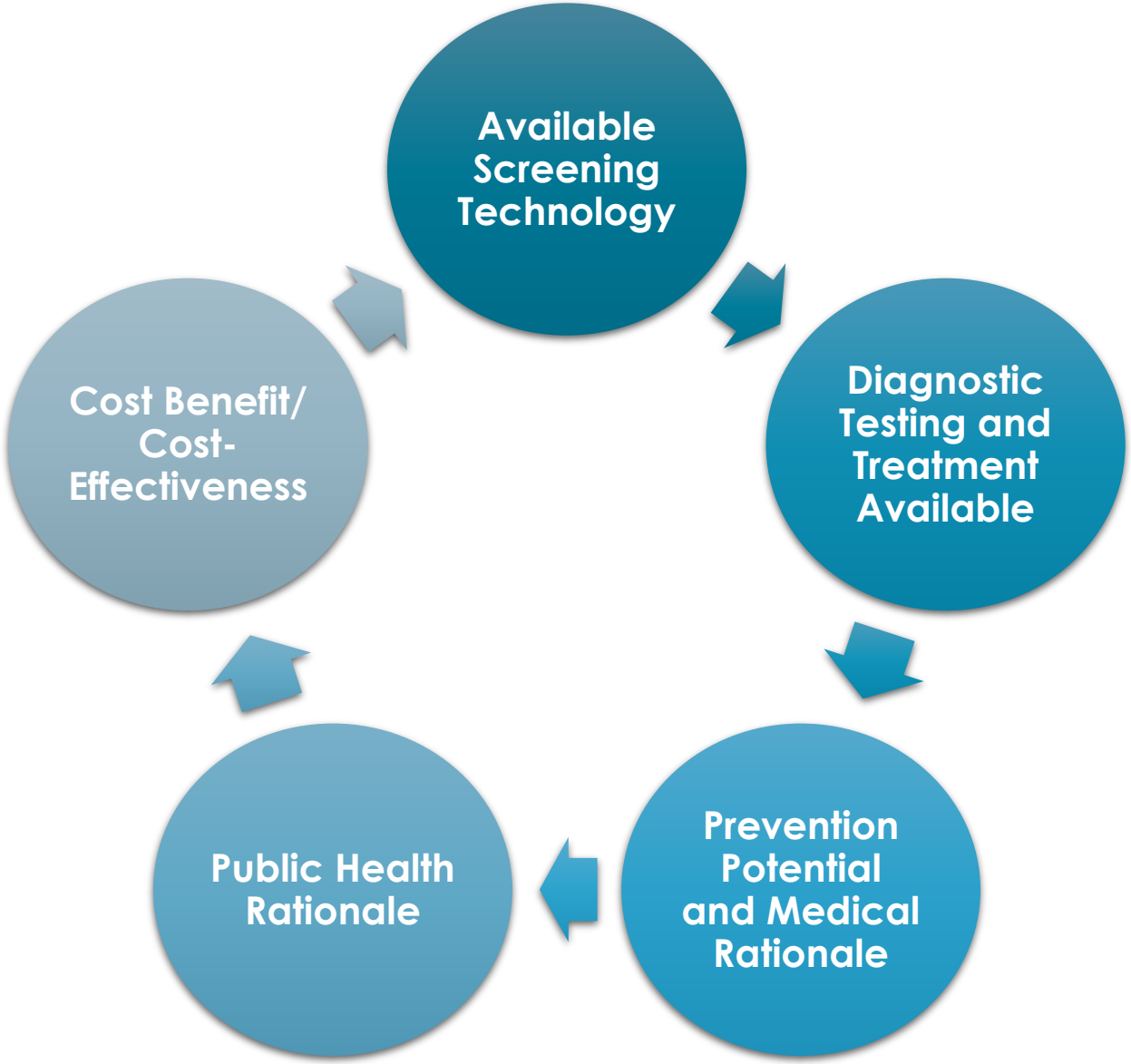
Board Policy for Newborn Screening

Three guiding principles govern all aspects of the evaluation of a candidate condition for possible inclusion in Washington's Newborn Screening panel:



Five Newborn Screening Criteria



Petition for Rulemaking

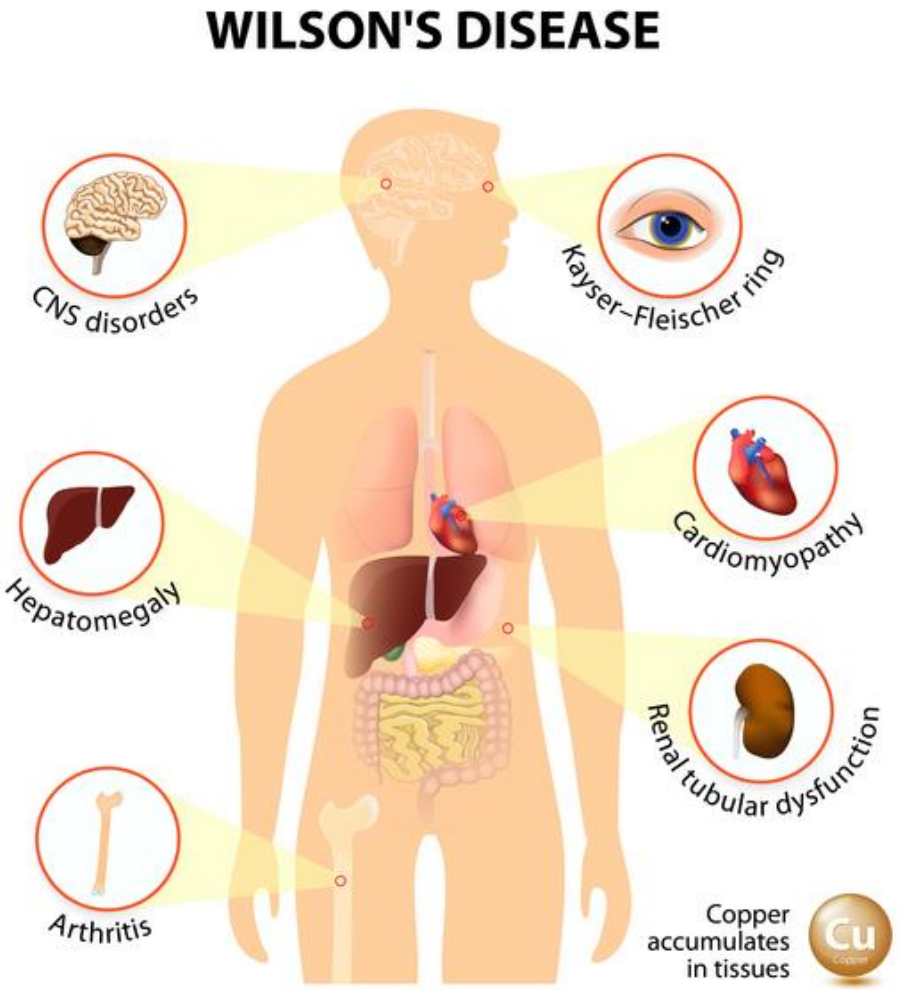
- On July 26, 2024, the Board received a petition request to amend chapter 246-650 WAC to add Wilson's Disease as a mandatory condition on the state's newborn screening panel.

- Early identification of an individual affected with Wilson's Disease would allow for early treatment and prevent tissue damage to the liver or nervous system.

What is Wilson's Disease

- Rare, inherited metabolic disorder
  - Autosomal recessive inheritance pattern
- Prevents body from eliminating excess copper
  - Genetic mutation prevents properly expelling copper
  - Copper builds up in tissues
  - Too much copper is toxic to body
- Symptoms
  - Can occur from age 3-70
  - Jaundice, fatigue, loss of appetite, swelling, difficulty with speech
  - Significant nervous system impairment
  - Death

[Source: NIH, National Library of Medicine; National Organization for Rare Disorders, Mayo Clinic]



Source: Medlineplus.gov/genetics/condition/Wilson-disease

Screening, Diagnostics, Treatment

- Available Screening Technology
  - Proteomic mass spectrometry
  - Key Proteo developed newborn screening kit, piloted at Washington Newborn Screening program

- Diagnostic Testing
  - Low ceruloplasmin in blood
  - High copper in urine
  - Sometimes, liver biopsy and/or brain imaging
  - Molecular testing

- Treatment
  - Copper chelation therapy
  - Liver transplant

[Source: Key Proteo, NIH, National Library of Medicine; National Organization for Rare Disorders]

Prevention Potential and Medical Rationale

- Literature recommends diagnosing Wilson's Disease as early as possible.
- Early connection to treatment prevents permanent neurological damage and liver disease.
  - Treatment is lifelong.
- Available treatments only resolve some complications related to Wilson's Disease.
- Damage to liver and brain is irreversible.

[Source: NIH, National Library of Medicine; GeneReviews; Mayo Clinic]

Public Health Rationale

- Autosomal recessive inheritance pattern
  - Prevalence: 1:32,400
  - Approximately 1 in 90 carry Wilson's Disease gene
  - If both parents are carriers, there's a 1 in 4 chance their child will have Wilson's Disease
  - If parents have a child with Wilson's Disease, they still have a 1 in 4 chance of having another child with Wilson's Disease, and chances stay the same for future children

- Wilson's Disease can impact all people equally
  - No differences based on sex, race, or ethnicity

[Source: National Organization for Rare Disorders]

Considerations

- No state in the US is screening for Wilson's Disease.
- Wilson's Disease is not on the Recommended Uniform Screening Panel (RUSP).
- The Washington Newborn Screening program is running a pilot project for Wilson's Disease screening.
- The petitioner and Washington Newborn Screening program have been working for over 15 years to develop newborn screening tests for Wilson's Disease.

For Board Member Discussion

- Would the Board consider accepting or denying this petition? Why or why not?
- Do Board Members want to direct staff to conduct a preliminary review of the condition and return to the Board at an upcoming meeting? Or proceed to a technical advisory committee?
- Discussion and justification for the Board's decision will be included in the Board's determination letter to the petitioner.

WASHINGTON STATE BOARD OF HEALTH

THANK YOU

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