Petition for Rulemaking:

Chapter 246-650 WAC, Request to Add Metachromatic Leukodystrophy (MLD)

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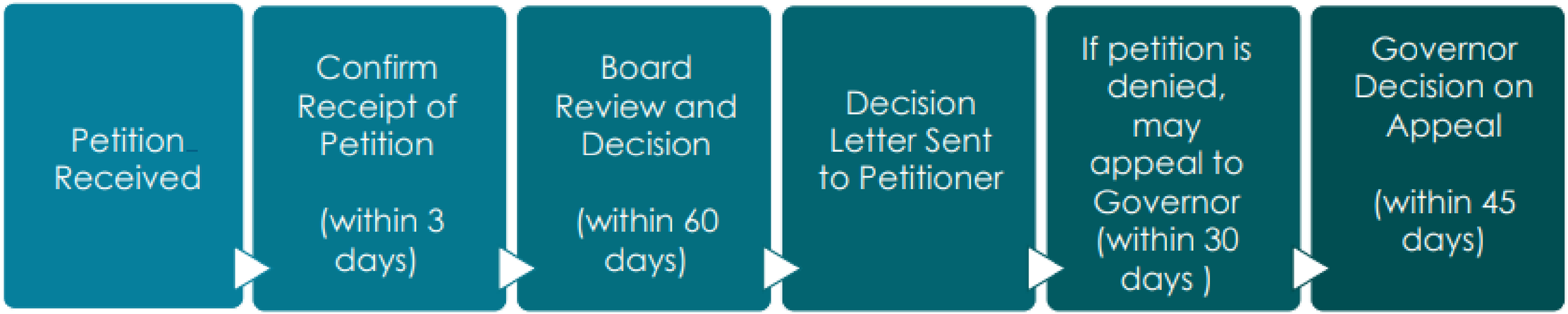
WASHINGTON STATE BOARD OF HEALTH

Background

Under the Administrative Procedures Act, RCW 34.05.330, any person may petition a state agency to adopt, repeal, or amend any rule within its authority.

Overview of the Board's Petition Process

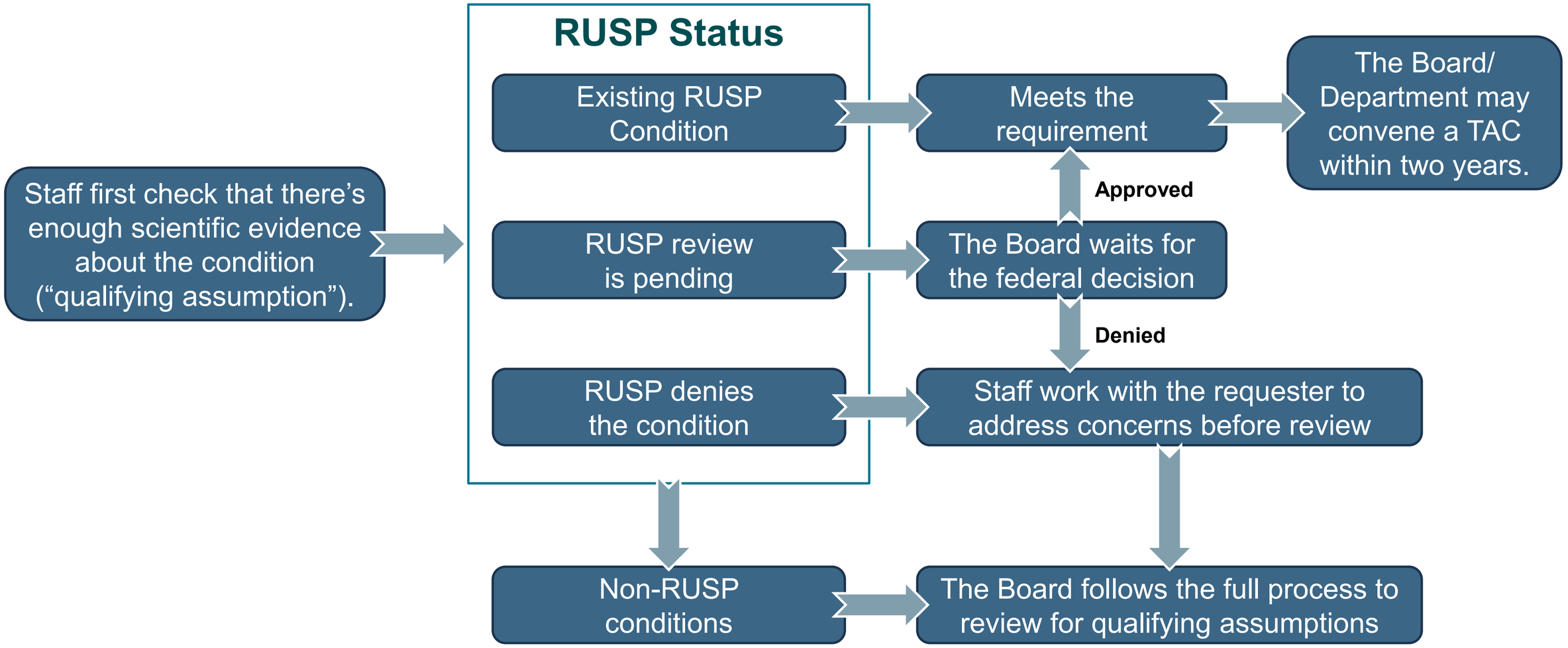


RCW 70.83.050 grants the Board authority to adopt rules for newborn screening in Washington State. These rules are within chapter 246-650 WAC.

Board Policy for Newborn Screening

Three guiding principles govern all aspects of evaluating candidate conditions for possible inclusion in Washington's newborn screening panel:

Qualifying Assumption



Petition for Rulemaking

- Received on October 6, 2025.
- Request to add MLD to Washington State's newborn screening panel (chapter 246-650 WAC).
- Petition highlights:
  - MLD screening can be done using existing lab equipment.
  - A reliable screening test is available.
  - Other states are implementing or planning screening for MLD.
  - Early detection is critical – treatment works best before symptoms appear.

Metachromatic Leukodystrophy (MLD)

What is it?

- Rare condition that prevents the body from breaking down and recycling fatty molecules
- Inherited and caused by changes in the ARSA gene
- Leads to nerve damage that worsens over time, affecting movement and thinking
- If untreated, it is often fatal within a few years of diagnosis

Types of MLD (based on when symptoms start):

- Late-infantile: Symptoms before 30 months of age (most common, 39-80% of cases)
- Early Juvenile: Symptoms between 30 months and 7 years (2nd most common)
- Late-juvenile and adult-onset (not the target of NBS and ineligible for gene therapy)

Impact of MLD:

- Affects approximately 1 in 100,000 newborns

Available Screening Technology

- **First tier**: Testing of sulfatides from dried bloodspots via liquid chromatography-tandem mass spectrometry (LC-MS/MS)
  - Technology already used in the WA NBS lab; can be tested with other conditions
- **Second tier**: Testing of ARSA enzyme activity in dried bloodspot (likely send out to contract lab)
- **Optional third tier**: ARSA gene sequencing using dried bloodspot to help confirm screening results and predict disease type (often sent out to a contract lab at the same time as the second tier)

- **Screening Accuracy**: Multi-step testing appears to have high performance (very few false positives or missed cases).
- **Current States Screening**: Four states (NY, MN, PA, IL) are working towards the implementation of MLD newborn screening.

Diagnostics and Available Treatment

Diagnostic Testing (After a Positive Screen):

- ARSA enzyme activity (whole blood)
- Sulfatide measurement (urine)
- DNA testing for changes in the ARSA gene

Treatment:

- Gene therapy Atidarsagene autotemcel (arsa-cel):
  - FDA-approved in 2024 for presymptomatic cases of late infantile MLD and presymptomatic/early symptomatic cases of early juvenile MLD
  - Five treatment centers offering gene therapy; nearest is Benioff Children's Hospital (San Francisco, CA)
- Later-onset MLD:
  - Stem cell transplant after symptoms appear to slow disease progression

Prevention Potential and Medical Rationale

- Results from MLD newborn screening can be ready within a few days to a few weeks, depending on how many tiers of testing are used and whether samples are sent to outside labs.
- Timing matters. Gene therapy works best when started early, ideally:
  - By 6 months of age for late-infantile MLD
  - By 12 months of age for early-juvenile MLD

Public Health Rationale

- Most babies with MLD do not have a family history:
  - Presymptomatic detection via universal newborn screening is the best way to identify affected babies early.
- Early detection is important:
  - Once symptoms appear and a clinical diagnosis is made, it’s often too late for gene therapy.
- Treatment works best before symptoms appear:
  - Gene therapy given early improves survival and helps preserve cognitive function.

Cost-Benefit and Public Health Infrastructure Readiness

- A 2024 health economic analysis in the UK found that adding MLD to newborn screening is cost-effective for the National Health Service (NHS) health system.
- Screening could improve quality of life, with an estimated 246 additional quality-adjusted life years (QALYs) in babies identified early.
- Costs are higher (mainly due to gene therapy), but the cost per QALY (£33,212) is considered acceptable.

Bean K, Jones SA, Chakrapani A, Vijay S, Wu T, Church H, Chanson C, Olaye A, Miller B, Jensen I, Pang F. Exploring the Cost-Effectiveness of Newborn Screening for Metachromatic Leukodystrophy (MLD) in the UK. Int J Neonatal Screen. 2024 Jun 26;10(3):45. doi: 10.3390/ijns10030045. PMID: 39051401; PMCID: PMC11270184.

MLD Federal Review

- MLD is currently under review by the Health Resources Services Administration (HRSA) for possible addition to the RUSP.
- Initial nomination submitted to Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) in 2024.
  - MLD was one of two conditions under review by ACHDNC before the committee was dissolved.
- The American Academy of Pediatrics published the evidence-based review of MLD in August 2025.
- HRSA held a public comment period on the report and accepted additional comments and evidence; closed on September 15, 2025.
- No updates have been announced yet on the outcome of the HRSA review.

For Board Member Discussion

- What questions do Board members have about the presentation?
- Would the Board consider accepting or denying this petition? Why or why not?
- Should Board Members direct staff to:
  - Monitor the outcome of the federal review of MLD, and report back to the Board later for a decision on forming a TAC?
  - Proceed with convening a TAC within two years.
  - Other?
- Discussion and justification for the Board's decision will be included in the formal response letter to the petitioner.

Citations:

1. U.S. National Institutes of Health, Genetic and Rare Diseases Information Center. Metachromatic leukodystrophy. September 2025. Accessed October 29, 2025. https://rarediseases.info.nih.gov/diseases/3230/metachromatic-leukodystrophy

2. Wendy K.K. Lam, Margie A. Ream, Scott D. Grosse, Jelili Ojodu, Elizabeth Jones, Hadley Stevens Smith, Anne Marie Comeau, Susan Tanksley, Alex Kemper; Evidence Regarding Metachromatic Leukodystrophy Newborn Screening. Pediatrics 2025; 10.1542/peds.2025-073188; https://doi.org/10.1542/peds.2025-073188

3. Cleveland Clinic. Metachromatic Leukodystrophy. Last reviewed February 6, 2023. Accessed October 29, 2025. https://my.clevelandclinic.org/health/diseases/6067-metachromatic-leukodystrophy

4. U.S. Department of Health and Human Services, Health Resources and Services Administration. Notice with request for comment: Consideration of adding metachromatic leukodystrophy to the Recommended Uniform Screening Panel (90 FR 39196). August 14, 2025. Accessed October 29, 2025. https://www.federalregister.gov/documents/2025/08/14/2025-15432/notice-with-request-for-comment-consideration-of-adding-metachromatic-leukodystrophy-to-the

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