Unique Approaches to Expanding Routine NBS Panels: State Case Studies

Newborn screening (NBS) programs are operated and funded by each state government. A federal process for recommending conditions suitable for NBS to states through the Recommended Uniform Screening Panel, or RUSP, is undergoing change. In April 2025, the committee responsible for evaluating evidence to support additions to the RUSP was dissolved. Despite this change, the RUSP remains the federal standard for testing panels, and 13 states have passed legislation that aligns testing with the RUSP.

With the operation of NBS programs remaining a state-based responsibility, states have flexibility in how they conduct their programs, including making decisions about which conditions to screen for or add to their screening panels. The manner in which a state's NBS program is conducted is unique to that state's capabilities. Some states have the facilities and expertise to conduct pilot studies before implementing population screening. Ten states have defined and made public their process for nominating new conditions. Twelve states use another state's laboratory for testing, and four (plus the District of Columbia) outsource testing to a commercial laboratory. Others rely on different advisory committee structures and legislative bodies to assess and advance proposals for new screening programs. See this state-by-state listing of key details.

Three case studies presented here illustrate the differences between states' approaches to NBS. Even if none of these cases relate to a state you're most interested in, there is value in understanding different approaches as a means to inform the questions you ask, the data you gather, and the proposals you draft and put forward.

Case Study: Washington State

In the state of Washington, new conditions are added to the newborn screening (NBS) panel through a formal, evidence-based, state-led process. The process is led by the State Board of Health (BOH), and supported by the Department of Health (DOH), which runs the state's NBS program. Under state law, the BOH sets rules for newborn screening, and the DOH carries out the requirements.

The BOH and DOH rely on newborn screening Technical Advisory Committees (TACs), composed of groups of experts from various fields, to review candidate conditions and recommend to the Board whether they should be added to the screening panel.

Washington's laboratory also processes the dried blood spots for Idaho and Hawaiiso this process affects available screening tests for those states as well.

Guiding Principles: According to Washington's published process, three principles guide the evaluation of new candidates for possible inclusion in its NBS panel:

- A decision to add a screening test should be driven by evidence. For example, test reliability and available treatments must have been scientifically evaluated, and those treatments must be shown to improve health outcomes for affected children.
- All children who screen positive should have reasonable access to diagnostic and treatment services.
- The benefits of screening for the disease or condition should outweigh the harm to families, children, and society.

The Petition and Initial Review: The most common way to nominate a condition for the NBS panel is by submitting a rulemaking petition, which is a formal request asking a state agency or board to change one of its rules. Anyone can submit a petition under state law (RCW 34.05.330). (Note: The state petition form is not specific to the DOH NBS program. The relevant rule that lists conditions the DOH screens all newborns for is chapter 246-650-020.)

When a state agency or board receives a petition, they have 60 days to respond in writing to the petitioner. They must either:

1. Accept the petition and begin rulemaking, or
2. Deny the petition, and explain why, including how the agency plans to address the concern, if appropriate.

For newborn screening, petitions to add a condition are always denied because the BOH follows certain steps before beginning the rulemaking process. First, the BOH must determine whether there is enough scientific evidence to apply its newborn screening criteria (called a qualifying assumption). These criteria are:

State law permits anyone to submit a Petition; the state has 60 days to make an initial response. Staff will evaluate the petition to determine whether it includes enough evidence to apply these specific criteria:

1. **Available Screening Technology**: Sensitive, specific, and timely tests are available that can be adapted to mass screening
2. **Diagnostic Testing and Treatment Available**: Accurate diagnostic tests, medical expertise, and effective treatment are available for evaluation and care of all infants identified with the condition.

3. **Prevention Potential and Medical Rationale**: The newborn identification of the condition allows for early diagnosis and intervention.
4. **Public Health Rationale**: The nature of the condition justifies population-based screening rather than risk-based screening or other approaches.
5. **Cost-benefit/Cost-effectiveness**: The outcomes outweigh the costs of screening. All outcomes, both positive and negative, need to be considered in the analysis.
6. **Public Health Infrastructure Readiness**: The NBS program's capacity to implement screening within a reasonable timeframe has been considered

Expert Review: If the BOH finds there is enough evidence to meet the qualifying assumption, they form a TAC. This committee includes a mix of experts, such as doctors, parents of children affected by rare diseases, insurance representatives, public health specialists, ethicists, and other representatives relevant to the condition.

Most of the work happens before the TAC meeting, as BOH and DOH staff prepare materials and arrange for subject-matter experts to present to the committee. During the meeting, the TAC evaluates the condition using six criteria, including a cost-benefit analysis. The committee then votes and makes a recommendation to the BOH. A favorable cost-benefit analysis is not required for a condition to be recommended.

BOH Decision: The TAC's recommendation, including the summary of information, committee votes and comments, is presented to the BOH by DOH and BOH staff. The BOH then decides whether to approve or deny adding the condition. The evidence is reviewed during one of the Board's public meetings. The public may always submit written comments or sign up for public comment for a Board meeting. A recent example of the TAC's recommendations to the BOH for Wilson Disease (dated August 2024), including a cost-benefit analysis, can be found here.

Before making a final decision, the BOH reviews and considers the TAC's recommendation along with all available evidence presented to the committee, which may include public input.

Additional Information

Legislative Activity: In recent years, the state legislature has directed the BOH to evaluate candidate conditions. Examples included branched chain ketoacid dehydrogenase kinase (BCKDK) deficiency and congenital cytomegalovirus (cCMV).

Relationship to the RUSP: Under the state's current newborn screening process, when a condition is added to the federal Recommended Uniform Screening Panel

(RUSP), it may automatically meet the qualifying assumption for review in Washington. In these cases, the state may form a TAC to review the condition within two years.
Note: After the termination of the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) in April 2025, the BOH changed the language from "will convene" to "may convene", reflecting the uncertainty around future federal reviews.

Funding and Implementation
When the BOH approves a new condition, the DOH must secure legislative approval to increase the newborn screening fee before the condition can be officially added and screening can begin. This fee pays for the new test, staff time, follow-up for positive results, and other administrative costs. Additionally, because the Washington State Healthcare Authority (HCA), which administers the state's Medicaid program, covers about 40% of births in the state HCA must also receive legislative approval to adjust Medicaid managed care rates to cover the higher fee. In some cases, DOH and HCA need to submit these requests multiple times before they are fully approved.

Takeaways From This State's Process

Washington's process for adding new conditions to its NBS panel highlights:

- A relationship between the RUSP and a state process that reflects the state's unique capabilities and governing process, while also leveraging the evidence-based method of review traditionally used to expand the RUSP
- A transparent, evidence-based process for assessing candidates for NBS, with a pathway for members of the public to present potential candidate conditions and related evidence.
- A cost-benefit-based approach to analyzing and supporting the costs of adding new conditions to NBS panels.

Additional case studies coming soon