Minutes for the Newborn Screening Technical Advisory Committee
January 14, 2025
Hybrid Meeting
ASL (or CART) and Spanish interpretation available
Seattle Airport Marriott Hotel
3201 S 176 St
Seattle, WA 98188
Virtual meeting: ZOOM Webinar

Technical Advisory Committee Members present:

In-Room Participants:

Kelly Oshiro, JD, Board Vice Chair and TAC Co-Chair
Nirupama (Nini) Shridhar, MPH, PhD, TAC Co-Chair
Heather Hinton, MultiCare Yakima Memorial
Eric Leung, Washington Chapter of the American Academy of Pediatrics (WCAAP)
Stephen Kutz, State Board of Health Member
Byron Raynz, Parent Advocate
Roberta (Bobbie) Salveson, Mary Bridge Children's Hospital Biochemical Genetics
Christina Lam, Seattle Children's Hospital Biochemical Genetics
Molly Parker, Provider and Chief Marketing Officer (CMO) of Population Health, Jefferson Healthcare
Michelle Whitlow, Parent Advocate, Lewis County Autism Coalition

Online Participants:

Joon-Ho Yu, Department of Epidemiology, University of Washington Bioethics, Treuman Katz Center for Pediatric Bioethics and Palliative Care
Kristine Alexander, Regence Health Plans
Lisa McGill Vargas, Sacred Heart Medical Center Neonatology Intensive Care Unit (NICU)
Taylor Kaminski, Global Perinatal Services
Priyanka Raut, Yakima Valley Farmworkers Clinic
Krystal Plonski, Naturopath, Seattle Children's Hospital, and Washington Association of Naturopathic Physicians (WANP)
Joan Chappel, Washington Healthcare Authority (HCA)
Sunpreet Bhangoo, Washington Healthcare Authority (HCA)
Peggy Harris, Parent/Child Advocate, Save Babies Through Screening Foundation

State Board of Health (Board) staff present:

Michelle Davis, Executive Director
Kelly Kramer, Newborn Screening Project Policy Advisor
Molly Dinardo, Policy Advisor
Melanie Hisaw, Executive Assistant
Crystal Ogle, Administrative Assistant
Michelle Larson, Communications Manager
Anna Burns, Communications Consultant

Guests and Participants:

Allegra Calder, Facilitator
John Thompson, Department of Health
Megan McCrillis, Department of Health
Samantha Fuller, Department of Health

Anna Hidle, Department of Health
Philip J. White, Associate Professor of Medicine, Division of Endocrinology, Metabolism & Nutrition, Duke University
Beth Ogata, University of Washington Genetic Medicine
Robert Steiner, Wisconsin Newborn Screening Program
Julie Thiel, Wisconsin Newborn Screening Program
Tami Horzewski, Wisconsin Newborn Screening Program

1. **WELCOME & INTRODUCTIONS**

Allegra Calder, Facilitator, and Kelly Kramer, Board staff, provided introductory remarks and overviews of the language interpretation channels and Zoom meeting functions.

Facilitator Calder then invited TAC members to introduce themselves and share their hopes for 2025.

2. **OCTOBER TAC RECAP, NOVEMBER BOARD UPDATES, ADDITIONAL CONSIDERATIONS FOR PROCESS RECOMMENDATION.**

Kelly K. reviewed the agenda with TAC members and then summarized the recommendations and outcomes of the October TAC meeting. Following this, Kelly K. highlighted the key discussions from the November Board meeting and presented additional Board recommendations. These recommendations were included in the meeting materials for members (see presentation on file).

Eric Leung, Committee Member, provided feedback on the updated process and criteria document on page 17 of the meeting materials. Member Leung suggested that staff clarify the definition of the “qualifying assumption.” Additionally, Member Leung recommended removing the specific reference to "five newborn screening criteria" and instead using the more general term "criteria."

Kelly Oshiro, Board Vice Chair and TAC Co-Chair, and Nini Shridhar, TAC Co-Chair, then outlined the purpose and objectives of the meeting, which were to evaluate branched chain ketoacid dehydrogenase kinase (BCKDK) deficiency and review the Board’s newborn screening criteria for assessing candidate conditions.

3. **OVERVIEW OF BRANCHED CHAIN KETOACID DEHYDROGENASE KINASE (BCKDK) DEFICIENCY**

Kelly K. provided an overview of BCKDK deficiency (see presentation on file) and noted that committee members would hear more about BCKDK from two subject matter experts later in the meeting.

Byron Raynz, Committee Member, asked staff for more information on Senate Bill 6234, which directed the Board to review BCKDK deficiency.

Kelly K. explained that Senator Wilson introduced the bill after researching autism spectrum disorder and discovering the connection with BCKDK deficiency. Senator Wilson proposed adding the condition to the newborn screening panel to address the prevalence of autism in Washington State. Kelly K. clarified that, to their knowledge, Senator Wilson did not know anyone personally with the condition; the bill stemmed from the Senator's personal interest.

4. FAMILY PERSPECTIVE

Kelly K. introduced Michelle Whitlow from the Lewis County Autism Coalition and Committee Member. Kelly K. explained that the Board and the Department of Health (Department) always try to include individuals with lived experiences in technical advisory committee reviews, particularly those with children diagnosed with the condition under review. Since BCKDK deficiency is extremely rare and no cases have been identified in the U.S., Board staff were unable to involve someone with direct experience of the condition. However, Kelly K. noted they were able to engage Michelle to provide a perspective from the autism spectrum disorder (ASD) community. While BCKDK deficiency is not directly linked to ASD, it is associated with epilepsy and certain forms of ASD. Michelle was invited to share insights on the broader connection between ASD and branched-chain amino acid disorders.

Member Whitlow, shared written testimony (see page 81 of the meeting materials) expressing concerns about including BCKDK deficiency in the Washington newborn screening panel due to its low prevalence. Member Whitlow highlighted that adding the condition could divert resources from existing conditions and raised questions about the cost-benefit ratio. Member Whitlow then called for further research and data collection to understand treatment efficacy and long-term outcomes, noting it would be difficult to add the condition without sufficient data.

As an advocate for the ASD community, Member Whitlow emphasized the principle of "nothing about us without us." While acknowledging a potential link between BCKDK deficiency and ASD, Member Whitlow noted that the broader ASD community has not been meaningfully engaged, particularly in the U.S. Member Whitlow stressed the need for an inclusive review process that carefully weighs the costs and benefits for both individuals with BCKDKD and the broader community. Member Whitlow concluded by expressing openness to learning more and ensuring future discussions include those directly impacted.

5. BCKDK DEFICIENCY: NATURAL HISTORY, DIAGNOSTIC TESTING, AND TREATMENT

Kelly K. introduced Dr. Phillip White, Associate Professor of Medicine within the Division of Endocrinology, Metabolism, and Nutrition from Duke University and Beth Ogata, a registered dietitian from the University of Washington Medical Genetics clinic to present information about BCKDK deficiency.

Dr. White presented the natural history of BCKDK deficiency, providing background on branched-chain amino acids (BCAAs) and their essential roles in the body. Dr. White noted that BCKDK deficiency, first described in 2012 by Novarino, is a very rare condition, with only 22 cases identified in the largest study to date. No report or study on the condition to date has provided a complete natural history of the condition. Dr. White shared that branched chain amino acids (BCAA) can be measured through a dried-blood spot, and high BCAA levels are currently used to identify Maple Syrup Urine Disease, and a similar test could likely be used to identify BCKDK deficiency (see presentation on file).

Christina Lam, Committee Member, asked about screening and whether any studies had been done on the sensitivity and specificity of the test.

Dr. White explained that BCKDK deficiency has not been widely screened for, and with only a few publications reporting on the condition, there is insufficient data to estimate the exact sensitivity and specificity of the screening test. Despite the lack of data, the measurement technologies for BCAA are very robust, and the variation in measurements is small.

Steve Kutz, Committee Member, inquired about treatments and dietary supplements for BCAA disorders and asked whether untreated or late-diagnosed BCKDK deficiency causes irreversible harm.

Dr. White explained that amino acids are derived from dietary proteins, and BCAA-enriched supplements are inexpensive and widely available, often marketed for muscle growth, strength, energy, and fat loss. Dr. White also noted that the few studies available indicate that early identification of BCKDK deficiency can prevent the onset of symptoms. However, those who received treatment later showed some improvement, but things like developmental disabilities caused by the condition did not reverse.

Member Kutz asked if any studies have used DNA testing to identify BCKDK deficiency in people with ASD retrospectively.

Dr. White mentioned a small study from Brazil that aimed to address this question but noted that the authors did not have expertise related to ASD.

Eric Leung, Committee Member, asked Dr. White if the rarity and newness of BCKDK deficiency means that its true prevalence is still unknown.

Dr. White confirmed this was accurate, but looking back at neonatal blood spot data could give a sense of the prevalence.

Member Leung thanked Dr. White and noted that the lack of prevalence data makes it difficult for the committee to assess the utility of universal newborn screening. Member Leung then asked Dr. White if they had worked with the Department of Health in North Carolina to look back at neonatal blood spot data.

Dr. White explained that while their expertise is in studying the biochemical pathways of BCAAs, from a cost-benefit perspective, it could be worth considering that neonatal blood spot testing already measures branched-chain amino acid levels. However, no established threshold exists to flag low values for potential follow-up, which would require additional consideration.

Lisa McGill Vargas, Committee Member, asked about the costs of screening for BCKDK deficiency, given that Maple Syrup Urine Disease is already screened for in Washington, inquiring if adding BCKDK deficiency would incur additional costs or require more staff. Member McGill Vargas also noted that in the NICU, premature babies on Total Parental Nutrition (TPN) often yield false positives for Maple Syrup

Urine Disease. TPN formulas in the U.S. are not tailored for preterm infants, and significantly affect their amino acid levels.

Dr. White responded that the costs would likely come from follow-up care, not the testing itself. Dr. White also noted that TPN might mask BCAA-related deficiencies in NICU newborns due to the supplementation provided.

Michelle Whitlow, Committee Member, inquired about the connection between microcephaly and BCKDK deficiency, and whether metabolic testing is typically conducted as part of standard protocol when the condition is identified in a newborn or young child.

Dr. White responded that, while they are not a pediatrician, existing literature suggests that microcephaly is progressive and commonly appears postnatally. It is not present at birth, and although head circumference is normal at birth, head growth becomes abnormal.

Member Leung then briefly shared information about microcephaly screening from a pediatrician's perspective.

Joon-Ho Yu, Committee Member, asked about a study Dr. White referenced, specifically the calculation methods used, and inquired how Dr. White interprets these calculations regarding the threshold issue.

Dr. White briefly shared their perspective on the study calculations and methods and the presentation of the data.

Beth shared insights on treatment from their role in the biochemical genetics program at the University of Washington, where they treat patients with conditions like BCKDK deficiency. Beth noted that current treatment approaches are based on four small studies, which offer clinician suggestions but are not formal treatment recommendations (see presentation on file). Beth explained that, although no patients with BCKDK deficiency have been seen in Washington State or the U.S., the standard care would be to refer patients to a biochemical genetics clinic. There, the patient would meet with a genetic counselor, nutritionist, and possibly a social worker for resources and support. The diagnosis would be confirmed, and a personalized treatment plan would be developed with the family.

Beth outlined a hypothetical treatment plan and concluded by discussing newborn screening treatment considerations, noting that while infrastructure and supports are available for managing conditions like BCKDK deficiency, challenges such as access to treatment, treatment burden, fatigue, and false positives must be carefully considered.

Priyanka Raut, Committee Member, inquired about the resources available to support community health workers or nurses involved in pregnancy care. Member Raut also asked how families could be better supported, seeking clarification on existing resources and potential opportunities for further development to direct these efforts effectively.

Beth responded that existing resources, such as clinics specializing in metabolic disorders and tertiary care centers, are available. Their team has worked closely with public health nurses and departments, particularly for patients in the Seattle area. However, Beth acknowledged a general shortage of these resources across the state.

Member Kutz asked Beth to elaborate on treatment fatigue.

Beth explained that treatment for BCKDK deficiency is life-long, and in one study, several patients discontinued treatment due to lack of reimbursement and other challenges.

Member Kutz followed up, asking if any adverse side effects of treatment might outweigh the benefits.

Beth responded that, hopefully, this would not be the case, which is why their clinic conducts long-term monitoring to ensure patients' health remains balanced.

Member McGill Vargas, inquired about the wait times for metabolic clinics and the availability of virtual support for children outside the Tacoma and Seattle areas.

Beth responded that the wait times for some services at Seattle Children's or Mary Bridge Hospital can be lengthy, but reserved spots for children identified through newborn screening help expedite their access to care. Beth mentioned that Bobbie Salveson from Mary Bridge would share more about access and equity considerations in the following presentation.

Member Lam asked Beth about the effectiveness of treatments for BCKDK deficiency.

Beth responded that, in their opinion, the data is not yet sufficient to support clear conclusions about treatment effectiveness. Beth emphasized the challenge of reconciling individualized treatment plans in the clinic with broader population recommendations, noting the need for more data to guide treatment decisions.

6. ACCESS AND EQUITY CONSIDERATIONS FOR BCKDK DEFICIENCY

Roberta "Bobbie" Salveson, Committee Member, shared perspective on access and equity for BCKDK deficiency. Member Salveson noted that initial diagnosis and follow-up are usually prompt, with patients at Mary Bridge seen within a week of receiving screening results. Member Salveson highlighted ongoing challenges with genetic and newborn screening, particularly the need for better education for parents and families, as many are unaware that their newborns have undergone screening. Member Salveson also emphasized the lack of cultural and linguistic services to support families navigating care.

Member Salveson added that while diagnostic services are generally covered, genetic testing can be expensive, ranging from $600 to $1,200 for the lab fee alone, not including additional costs. Insurance sometimes covers this, but coverage can be delayed. Treatment for infants and children, such as medical foods and formula, is usually covered by insurance, but this support stops once the child turns 18. Transportation access can also be a challenge, especially when tests or labs are

needed for follow-up care. While telemedicine visits are an option, logistical barriers remain. Member Salveson additionally pointed out the lack of geneticists specializing in adult care, adding to families' challenges. Member Salveson further mentioned that when there is only one center in the state capable of treating certain rare conditions, it significantly impacts the center's workload.

Member Salveson concluded by highlighting that their clinic typically doesn't receive autism referrals until children are at least three years old, and families can experience wait times of up to a year. By then, the clinic often cannot provide early intervention. Member Salveson stated that early treatment for BCKDK deficiency could potentially prevent an autism diagnosis, but navigating false positives remains a challenge due to the lack of data available on the condition.

Molly Parker, Committee Member, shared perspective as a primary care provider, noting two possible pathways for identifying BCKDK deficiency: through screening or during the natural progression of care, such as noticing microcephaly and motor delays. Member Parker inquired whether the earliest a primary care provider might recognize the condition would be around eight months, which was an optimistic estimate. Member Parker also highlighted barriers to timely diagnosis in general medicine, pointing out that a baby with microcephaly might take four to five months to be referred to a neurologist, leading to a potential diagnosis delay of up to one and a half to two years. Member Parker agreed that more data is needed to support a screening program for BCKDK deficiency.

Eric Leung, Committee Member, noted that the research on BCKDK deficiency did not specify how the eight-month-old with the condition was identified. However, the study included 21 patients across 13 families, half of which already had affected members. This suggests that having a family history helps with earlier identification of the condition. Member Leung concluded by sharing that it would be hard to diagnose the condition clinically without a known family history.

Michelle Whitlow, Committee Member, asked if any current newborn screenings, such as those for phenylketonuria (PKU), might also detect conditions like BCKDK deficiency. Specifically, they wondered if existing screenings could identify BCKDK deficiency or related metabolic conditions. Megan McCrillis, Department staff, mentioned that they would be covering this in the following presentation.

7. AVAILABLE SCREENING TECHNOLOGY

Megan McCrillis, Department staff presented the available screening technology for BCKDK deficiency, noting that no other states or countries are currently screening for the condition, and no prospective screening studies exist. Megan explained that while a screening test is available and Washington State already has the equipment for other conditions, thresholds for detecting low amino acid levels do not exist. This lack of established thresholds means that normal and abnormal ranges for amino acids remain undetermined, requiring further study to establish a baseline.

Megan also highlighted the potential for false positives due to this uncertainty. Additionally, while a diagnostic test exists, it is unavailable in-house at the Washington Public Health Laboratory (PHL) NBS program. It would need to be sent to an external

diagnostic lab for testing. Regarding sensitivity and specificity, Megan emphasized that no real-time data is available to determine these metrics due to the absence of prospective screening studies. Therefore, sensitivity and specificity remain unknown (see presentation on file).

Steve Kutz, Committee Member, inquired more about the methodology currently used by the newborn screening lab and whether it could be applied to screening for BCKDK deficiency.

Megan clarified that while the lab tests BCAA levels when screening for Maple Syrup Urine Disease, it only checks for elevated levels of BCAAs. There is no established threshold for what would be considered abnormal for low levels of BCAAs. Further research and pilot studies would be necessary to determine what constitutes a normal range for a typical newborn and what those ranges might look like for a baby with BCKDK deficiency. Megan concluded that while the technology and screening methodology are available, no one has yet conducted the necessary work to establish the low-end threshold for BCKDK deficiency.

John Thompson, Department staff, confirmed that while a diagnostic test for BCKDK deficiency is available, the Washington Newborn Screening Lab does not have it. Diagnostic tests are sent to a reference laboratory like Seattle Children's Hospital. John reiterated that there is no established normal range for low BCAAs, as prospective studies have not been conducted. John added that Megan collaborated with the Department's epidemiologist, running a week's worth of specimens through the Collaborative Laboratory Integrated Reports (CLIR) tool. While four specimens were flagged as low, it is unclear what this means, as no follow-up screenings have been conducted to determine if these results would normalize. The concept of screening for low BCAAs is still in the exploratory stage, with no prior studies to guide the process.

Joon-Ho Yu, Committee Member, inquired about the potential for false positive amino acid results due to nutritional supplements like total parenteral nutrition.

Megan explained that false positives due to interfering substances are common in NICU infants. The newborn screening follow-up team frequently monitors these cases, following up around 30 days of life or upon discharge. Megan added that once the infants are off supplemental nutrition, their values typically normalize, and that is when the team does the follow-up newborn screen to identify whether the results could have been false positives.

BREAK

8. COST-BENEFIT ANALYSIS

Megan McCrillis and Anna Hidle, Department staff, opened the discussion by revisiting the criteria for newborn screening panel inclusion, focusing on cost-benefit and cost-effectiveness.

Megan presented a cost-benefit model using a decision tree in Excel to compare the current outcomes for babies born with BCKDK in Washington State versus the potential outcomes if screening for the condition were implemented. Data for this model was

drawn from primary literature, states currently screening or piloting studies, and expert opinion. Megan emphasized the limited literature on the subject, noting that no states are currently screening for BCKDK or conducting studies on it, which made it challenging to run a model.

Anna H. discussed the challenges in economic analysis and cost-benefit modeling for conditions like BCKDK. Given the limited data, assigning costs to screening and treatment is particularly difficult and possibly risky. This lack of data introduces bias and makes the model's results misleading because it's based on unknown and weak published data.

Megan presented the cost-benefit model, comparing the status quo (no screening) with the potential scenario of screening for BCKDK. Megan discussed the variability in true positives, false positives, and false negatives and summarized the estimated costs and benefits. Due to limited data, no estimated cost was provided, though speculative figures could be shared upon request. Megan concluded by emphasizing that the quality of the cost-benefit analysis depends on the available data. While a benefit-cost ratio was unavailable, the model is built, and missing assumptions can be adjusted as more data becomes available (see presentation on file).

Michelle Whitlow, Committee Member, inquired whether Washington could conduct a pilot study of BCKDK deficiency screening, given the current lack of data on the condition.

John Thompson, Department staff, responded that when the TAC and Board recommend adding a new condition to the newborn screening panel, their team typically conducts a pilot study before screening begins. However, this is done with existing dried blood spot specimens. John confirmed that a prospective pilot study could be done, but it would require informed consent, and that no prospective pilot screening programs have been conducted since the 1980s, though some research studies have been done since then.

Steve Kutz, Committee Member, inquired if children with autism spectrum disorder are screened for BCKDK deficiency.

Member Whitlow responded no and clarified that while ongoing research is examining the incidence of genetic conditions, including metabolic disorders, and their association with autism spectrum disorder, this data is not yet available.

Eric Leung, Committee Member, clarified that in the studies reviewed, autism spectrum disorder was not the most common diagnosis among patients identified with BCKDK deficiency. Instead, developmental delays—such as motor, language, and behavioral delays—were more prevalent, with autism being just one subset.

Peggy Harris, Committee Member, shared the personal experience of having a child diagnosed with a rare condition and emphasized that if a condition can be feasibly screened for and there is a viable treatment, no matter how rare, it should be considered for screening.

Heather Hinton, Committee Member, shared concerns about the emotional impact of false positives on families, noting that ambiguous results often create significant worry. Member Hinton highlighted that, due to the limited data on BCKDK deficiency, there would be little information to provide for families if a positive result were to occur. Member Hinton also noted the lack of available resources or support groups for this condition, further complicating the situation from a genetic counseling perspective. Member Hinton emphasized the emotional toll such ambiguity can have on families.

Facilitator Calder reminded committee members that a vote would soon be required to decide whether BCKDK deficiency should be added to the newborn screening panel.

9. VOTE – EVALUATE BCKDKD WITH NEWBORN SCREENING CRITERIA

Kelly Kramer, Board staff, provided a brief recap of the Board's five criteria and gave instructions for voting.

Byron Raynz, Committee Member inquired before voting began whether BCKDK deficiency had ever been reviewed for the Recommended Uniform Screening Panel (RUSP) or if there had been any national efforts to review the condition. Member Raynz also asked if the staff had received any comments or testimonies from parents or families directly impacted by BCKDK deficiency.

Kelly K. responded that, to their knowledge, BCKDK deficiency has not been reviewed for newborn screening at the federal or state level. Additionally, no testimonies were received from families, though staff had contacted the bill sponsor for more information.

Facilitator Calder provided an update on the agenda, informing committee members that some items would be rearranged after lunch due to timing restraints and to accommodate guest speakers' schedules.

TAC members then participated in two anonymous online votes via Microsoft Forms. The first vote asked whether BCKDK deficiency meets the Board's five criteria for potential inclusion in the Washington Newborn Screening panel. The second vote was to recommend to the Board whether BCKDK should be added to the list of conditions for mandatory screening of all Washington-born newborns.

LUNCH

10. WA FIVE CRITERIA REVIEW AND DISCUSSION (Moved up from agenda item 13)

Facilitator Calder welcomed the committee members back and noted that the BCKDK deficiency votes would be reviewed after the committee hears from the Wisconsin Newborn Screening Program during the next agenda item.

Kelly K. reminded committee members that the agenda's next few sections would focus on reviewing the Board's five newborn screening criteria. To provide a broader perspective, staff invited Dr. Robert Steiner and his team from Wisconsin to present the criteria they use for newborn screening.

Robert Steiner, Julie Thiel, and Tami Horzewski from the Wisconsin Newborn Screening Program introduced themselves and thanked the committee for the opportunity to

present. Robert explained that 10 years ago, a task force developed Wisconsin's nine newborn screening criteria. Robert then outlined these nine criteria used to evaluate candidate conditions and mentioned there is some overlap with Washington's criteria (see materials on file).

Kelly K. asked Robert if their program had identified any criteria that other states might be missing in evaluating conditions, or if there were any wish list items they would like to add to their criteria.

Robert shared that their program typically doesn't receive feedback about missing criteria. Robert noted that the most contentious criterion is usually number nine, the cost issue, as it requires many assumptions and economic expertise, which is not always readily available. This often leads to significant discussion. Robert then asked the staff to add any additional comments.

Tami responded that criteria seven is often not applicable, and there have been some recent changes to criteria six to expand it.

Robert added that criteria seven is often not applicable, as it concerns the need for a new sample collection system, which is typically unnecessary (most tests use dried blood spot cards). However, it was relevant for hearing and congenital heart tests due to different mechanisms. Regarding Criteria 6, Robert shared that recent changes include adding test characteristics (specificity, sensitivity, positive predictive value) and the requirement for convincing medical evidence through experience, natural history, or literature.

Christina Lam, Committee Member, inquired whether there is a specific definition of "serious health risks" as mentioned in criteria one.

Robert acknowledged that this criterion is subjective and depends on the judgment of reviewers. However, most conditions reviewed pose serious health risks, such as death or disability, if untreated. Robert added that in childhood, this is particularly critical, as disorders with both early and late-onset forms present challenges, and states are struggling with how and whether to screen for both forms of the condition. Robert mentioned Krabbe Disease as an example of this.

Heather Hinton, Committee Member, noted that access to specialty services is limited in their rural Washington practice. Member Hinton asked if the team could expand on the wording of criterion four to clarify "reasonable availability."

Robert noted that Wisconsin faces similar rural access challenges. Robert mentioned that telehealth and satellite clinics have improved specialty care for babies in rural areas. Robert also highlighted a recent discussion on Krabbe Disease, where stem cell transplantation is limited to a few specialized centers. This led to a workgroup being formed to assess in-state treatment options and the possibility of out-of-state referrals. Criterion four has been particularly relevant in this context.

Facilitator Calder asked the presenters for feedback on Washington's criteria and if anything stood out to them as unclear or missing from their review.

Robert noted that Washington's criteria are clear and succinct. For their program, they often struggle to cover all nine criteria within a reasonable timeframe, especially with the faster pace of disorder nominations. Robert suggested not simply increasing the number of criteria but considering the addition of a few or incorporating relevant wording into existing criteria.

Eric Leung, Committee Member, asked if Robert's team could share more about the history of Wisconsin's criteria.

Tami shared that over 10 years ago, a workgroup reviewed Wisconsin's original six criteria, examined relevant research and literature, including the Wilson and Younger criteria, and conducted a vote to create the nine criteria. This work was led by one of their ethicists.

Member Lam inquired whether Wisconsin has defined effective interventions in terms of outcomes and treatment, noting that sometimes treatments can lead to other diseases.

Robert responded that no specifics have been established to guide the committee, though this topic typically sparks robust discussion. Robert cited Krabbe Disease as an example. Robert expressed concern about trying to more precisely define effective treatment, as it may be too challenging to have a standardized definition.

Steve Kutz, Committee Member, asked about the funding for new conditions Wisconsin adds to their newborn screening panel and how they prioritize adding new conditions.

Robert explained that, like Washington, Wisconsin's funding decisions must go through the Legislature. The program undergoes a rulemaking process to propose fee increases for blood spot cards, but it is uncertain whether the Legislature will approve the request. New conditions are prioritized on a first-come, first-served basis.

Molly Parker, Committee Member, inquired about Wisconsin's follow-up process for newborns with false positives.

Robert shared that efforts are made to minimize false positives. Specialists are committed to accommodating families and providers involved in false positive cases. Physicians and genetic counselors work to conduct confirmatory testing quickly, deliver results promptly, and support families in understanding that the baby is not affected. However, some subcommittee members expressed concerns during candidate condition reviews about the potential for 'vulnerable child syndrome' and other issues stemming from false positives. Robert concluded that false positives remain a challenging issue.

Facilitator Calder and Kelly K. thanked the presenters and mentioned that the committee had received an email with Wisconsin's nine criteria (also included in the meeting materials).

11. RESULTS AND DISCUSSION

Staff presented the BCKDK deficiency voting results, starting with the second vote on the committee's overall recommendation. 15 out of 16 members voted against recommending BCKDK deficiency for inclusion on the newborn screening panel, with most comments focusing on the lack of current data for the condition

Steve Kutz, Committee Member, raised a question about conditions reviewed with insufficient information, asking if the committee waits until the condition is brought to the committee again, if there is a mechanism to review it after a set number of years, or if the committee waits until it's added to the RUSP.

Eric Leung, Committee Member, noted that in the past, the committee has recommended reevaluating conditions after a certain period. Member Leung explained that trying to align with the RUSP allows for restarting the conversation if the condition is added, but families can also advocate for reconsideration at any time.

Facilitator Calder added that this conversation relates to Member Raynz's question about how the condition was brought to the committee, noting that it followed an unusual path, which is part of why the discussion on this condition has been challenging.

Nini Shridhar, TAC Co-Chair, commented on whether Washington should establish specific criteria for ultra-rare conditions, suggesting the need to evaluate how such conditions fit within the current criteria and whether specific data requirements should be met before adding them.

Facilitator Calder said that Co-Chair Shridhar's comments would be a good placeholder for the committee's criteria discussion shortly.

Heather Hinton, Committee Member, commented on how current genetic testing practices and technology may affect the identification of BCKDK deficiency prevalence. Member Hinton noted that if increased DNA testing leads to more identified cases in the future, the committee may want to review the condition

Staff then presented the results of the first vote, which evaluated BCKDK deficiency against the Board's five criteria. Staff asked committee members if they had additional questions or comments.

Member Kutz noted that during the vote, they considered the uncertainty of whether screening would prevent adverse health outcomes and asked about other conditions on the Washington panel with similar concerns.

Bobbie Salveson, Committee Member, discussed conditions with variable presentation and onset, such as X-ALD and Pompe disease, where treatment is limited to symptom monitoring and then management once symptoms appear. Member Salveson also highlighted the potential adverse effects of false-positive newborn screens but mentioned that families often seek a diagnosis for clarity and family planning, even without available treatment.

Member Leung asked if there is a condition on the screening panel with no negative outcomes.

John Thompson, Department staff noted 3MCC as the closest example, which is on the RUSP but not the panel. It's a benign biochemical trait but shares markers with harmful HMG deficiency, which is screened for.

Facilitator Calder confirmed with the group that the recommendation to the State Board of Health is to not recommend BCKDK deficiency. Facilitator Calder added that information and comments will be shared with the full board for review and will also be publicly available.

Kelly K. provided an overview of the next steps for presenting the TAC's recommendations to the Board.

Joon-Ho Yu, Committee Member, clarified that the comments section on the ballot is only prompted if 'unsure' is selected and requested staff ensure this is clear when presenting comments and votes to the Board.

Byron Raynz, Committee Member, questioned whether the recommendation to the Board should state "does not recommend *at this time*," to allow for future evaluation if needed.

Member Leung proposed that if the condition is revisited, it should be under the assumption that new data is available.

12. INTRODUCTION TO CRITERIA REVIEW

Kelly Kramer, Board staff ,introduced the criteria review (see presentation on file). Kelly K. reminded TAC members that recommendations for the criteria will not be reviewed or approved until the March 12 Board meeting.

13. CROSSWALK: RECOMMENDED UNIFORM SCREENING PANEL AND OTHER STATES' CRITERIA FOR CONDITION REVIEW

Megan McCrillis, Department staff, presented additional information from the Washington newborn screening program and other states to inform the committee's criteria review (see presentation on file). Megan explained that states not aligned with the RUSP use variations of the Wilson and Junger screening criteria, developed in the 1960s for all types of screening programs. These criteria also form the basis for Washington's newborn screening guidelines. Megan highlighted the strengths of Washington's current criteria and provided options for potential changes the committee could consider.

Steve Kutz, Committee Member, inquired about the costs associated with treating false positives and ruling them out.

John Thompson, Department staff, explained that their team's cost-benefit analysis includes costs related to false positives, based on data from programs already screening for these conditions. John noted that, for example, Pompe disease had a very

low predictive value, which was factored into the analysis. John also mentioned the advantages and disadvantages of being the first state to screen for a condition.

Eric Leung, Committee Member, and Member Kutz discussed the high false positive rates for cystic fibrosis screening. Member Leung clarified that the rate is skewed due to the preemie population in NICUs in Washington.

John added that if data from 2020 or 2021 had been used, the predictive value would have been higher. John noted a trade-off between better sensitivity and higher false positive rates. With the addition of cystic fibrosis DNA testing as a second-tier test, more cases with cystic fibrosis-causing variants were identified, requiring additional follow-up. While DNA testing is a valuable tool, it also has its drawbacks.

John provided additional context on the crosswalk Megan presented, noting that Pennsylvania was the only state with relevant criteria available for review.
Much of the crosswalk was based on Pennsylvania's criteria, with input from John and Megan incorporating ideas from the TAC's October discussion.

Byron Raynz, Committee Member, inquired whether the criteria considerations presented by Megan were recommendations from the newborn screening program or just general considerations.

Megan explained that the considerations were meant as ideas for discussion, based on efforts to identify more specific benchmarks. Megan noted that while many states were expected to have specific benchmark language, only Pennsylvania provided examples of this in their criteria.

Member Raynz expressed concerns about voting without input from subject matter experts (SMEs) and felt unsure about the potential impacts of changes to criteria. Member Raynz questioned if there were options to abstain or delegate votes for this important discussion.

John clarified that members could abstain and emphasized that if the discussion took a concerning direction, he would step in to offer support.

Member Kutz shared concerns about the costs of identifying rare conditions, referencing Pennsylvania's cost-based criteria, and emphasized the challenge of balancing resource allocation with the impact on families of rare conditions.

Christina Lam, Committee Member, noted that shifting responsibility away from future TACs could have long-term repercussions, aligning with Member Raynz's concerns.

Kelly Kramer, Board staff, acknowledged that today's task was vague and reiterated the Board's interest in modernizing criteria, emphasizing that the Board has the final say. Kelly K. also pointed out that the TAC could recommend that no changes need to be made to the criteria.

Nini Shridhar, TAC Co-Chair, reflected on the October meeting's discussion, where it was decided that criteria should be reviewed and potentially adjusted.

Kelly Oshiro, TAC Co-Chair, highlighted the increase in the number of conditions presented to the Board, suggesting a re-evaluation of the criteria to manage this influx. Co-Chair Oshiro emphasized the importance of ensuring newborns thrive and public health supports them.

Joon-Ho Yu, Committee Member, emphasized the need for transparency in the process and cautioned that, with rapidly advancing science and medical trends, criteria may need to adapt to account for emerging conditions, especially in the face of uncertainty and genetic testing advancements.

Krytsal Plonski, Committee Member, questioned whether the committee should consider aligning with RUSP criteria and suggested exploring the disclosure of criteria for RUSP conditions.

Member Kutz asked if conditions not on the RUSP had been reviewed in the past. Staff responded that yes, non RUSP conditions have been reviewed and recommended in the past.

Member Leung commented on the need for flexibility in the criteria due to the increasing volume of conditions and rapid advances in testing technology, using BCKDKD as an example. Member Leung suggested potentially postponing the cCMV review until the criteria are updated.

Co-Chair Shridhar agreed that it would be beneficial to use the same for the cCMV review, that was used for BCKDK deficiency for consistency, as it has already been discussed once.

John acknowledged Co-Chair Shridhar's point about using consistent criteria moving forward, but agreed with Member Leung's suggestion of postponing the cCMV review until the criteria are updated.

Facilitator Calder said the committee would take a break, and staff would huddle offline to discuss next steps.

BREAK

14. WA FIVE CRITERIA REVIEW AND DISCUSSION CONTINUED

Facilitator Calder shared that staff discussed options for the upcoming cCMV meeting in February. They considered whether to proceed with the current criteria or allow for further discussion. Changes could be addressed by email or before the cCMV meeting.

Kelly Kramer, Board staff, suggested the group could either provide concrete suggestions or leave the discussion open-ended.

Facilitator Calder clarified that the discussion would focus on ideas that are not problematic, not formal recommendations.

Steve Kutz, Committee Member, noted concerns about adding conditions and emphasized the need for criteria focused on identifying conditions in newborns, not later in life.

John Thompson, Department staff, responded that criteria three addresses this issue.

Eric Leung, Committee Member, felt the current criteria already cover identification in the newborn period.

Nini Shridhar, TAC Co-Chair, asked if natural history should be considered in the criteria.

Molly Dinardo, Board staff, referred to examples from Megan's presentation, noting the distinction between conditions with infantile onset detected through screening but not clinically in the first 24-48 hours.

Member Kutz explained the importance of newborn screening, as it provides a captive audience in the hospital to detect conditions early.

Kelly K. proposed a vote on whether to keep the criteria as is or make changes.

Lisa McGill Vargas, Committee Member, requested the current list of criteria be posted.

Kelly Oshiro, TAC Co-Chair, suggested a yes/no vote first, followed by further discussion about if changes are proposed.

Molly Parker, Committee Member, asked whether the purpose of the review was to modernize the language.

John Thompson, Department staff, shared historical context, noting that when the criteria were first developed in 2001, only minor changes were made in 2015. John suggested voting based on the current state or maintaining some ambiguity.

Member Kutz raised concerns that rejecting changes could lead to legislative pressure for further review.

Member Leung agreed with Member Kutz, noting that any changes would still need to meet the qualifying assumption that sufficient evidence supports the change.

John clarified that the Board's process for non-RUSP conditions requires meeting the qualifying assumption, which involves a preliminary review before a TAC.

Byron Raynz, Committee Member, asked if BCKDK met that review.

John explained that BCKDK did not go through the typical process because it was directed legislatively.

Staff created a voting ballot to have the committee vote on whether the team should review and update the current criteria, or leave the criteria as is.

15. VOTE – CRITERIA REVIEW

TAC members voted on whether to update the current newborn screening criteria. Seven members voted that the criteria should be changed, six voted no, they thought the criteria were already robust enough, and four voted unsure.

Eric Leung, Committee Member, found the current criteria robust but appreciated Wisconsin's criteria number 8 for its specific layout. Suggested considering rare conditions as testing becomes more available.

Kelly Oshiro, TAC Co-Chair, felt criteria 1 and 2 could benefit from more specificity, particularly with bullet points. Suggested adding Wisconsin's number 8 as a bullet under number 2 to improve clarity without overcomplicating the headlines.

Steve Kutz, Committee Member, advocated for moving the three guiding principles higher in the list, as they are crucial for accessibility and should be part of the vote.

Joon-Ho Yu, Committee Member, agreed to update criteria, especially number 4, which could use further consideration. Appreciated the bullet points for 3 and 5 and suggested further clarification of evidence-based principles across the criteria.

Krystal Plonski, Committee Member, agreed with the flexibility of the current format but suggested further consideration of the public health system's capacity to handle additional screening tests, as well as addressing concerns with criteria 4.

Nini Shridhar, TAC Co-Chair, voted yes, specifically for updates to criteria 1 and clarification of criteria 4.

Facilitator Calder recapped the committee's discussion so far.

Byron Raynz, Committee Member, expressed hesitation, as Wisconsin's criteria includes more prescriptive language. Washington's current criteria allow more flexibility to weigh evidence, and adding more prescriptive language could be a concern.

Member Yu appreciated the transparency of the RUSP decision matrix, which is less prescriptive but helps clarify the review process. Also noted the increasing types of evidence being considered, which offers a broader perspective.

Facilitator Calder asked staff if the next meeting would focus on reviewing criteria or specifically on cCMV.

Kelly K. acknowledged the need for further team discussions on whether to focus on cCMV or the criteria review.

16. DISCUSSION AND NEXT STEPS

Facilitator Calder noted the desire to revisit suggestions and that the project team will discuss timing further and connect with TAC members. The team would be in touch with committee members to discuss next steps.

ADJOURNMENT

Kelly Oshiro and Nini Shridhar, TAC Co-Chairs, adjourned the meeting at 2:30 p.m.

WASHINGTON STATE BOARD OF HEALTH

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Kelly Oshiro, TAC Co-Chair, and Nini Shridhar, TAC Co-Chair