Washington State Department of HEALTH

WASHINGTON STATE BOARD OF HEALTH

Newborn Screening Technical Advisory Committee (TAC)

NOTICE OF PUBLIC MEETING

Tuesday, February 11, 2025
9:00 a.m. – 4:00 p.m.

Note: This is a hybrid meeting held via Zoom and in-person at the Washington State Department of Health, Town Center 1, 101 Israel Rd. S.E. Tumwater, WA 98501. Meeting rooms: 163 & 164. Meeting access and instructions are provided below. Language interpretation available.

Newborn Screening Technical Advisory Committee (TAC) Agenda

Review of the Criteria for Adding a Condition to the Mandatory Newborn Screening Panel and the Review of the Condition Congenital Cytomegalovirus (cCMV)

Time	Agenda Item	Speaker
9:00 a.m.	1. Welcome and Agenda	Kelly Oshiro, TAC Co-Chair, State Board of Health Nirupama Nini Shridhar, TAC Co-Chair, Department of Health Kelly Kramer, State Board of Health Allegra Calder, BERK Consulting
9:15 a.m.	2. January TAC Recap	Kelly Kramer, State Board of Health
9:30 a.m.	3. WA Criteria Review and Discussion	Kelly Kramer, State Board of Health Kelly Oshiro, TAC Co-Chair, State Board of Health Nirupama Nini Shridhar, TAC Co-Chair, Department of Health Allegra Calder, BERK Consulting
10:25 a.m.	Break	

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Time	Agenda Item	Speaker
10:35 a.m.	4. Washington Criteria Review and Discussion Continued	Kelly Kramer, State Board of Health Kelly Oshiro, TAC Co-Chair, State Board of Health Nirupama Nini Shridhar, TAC Co-Chair, Department of Health Allegra Calder, BERK Consulting
11:30 a.m.	5. Vote - Criteria review	Kelly Oshiro, TAC Co-Chair, State Board of Health Nirupama Nini Shridhar, TAC Co-Chair, Department of Health Allegra Calder, BERK Consulting
11:45 a.m.	Lunch	
12:15 p.m.	6. Discussion and Next Steps	Kelly Oshiro, TAC Co-Chair, State Board of Health Nirupama Nini Shridhar, TAC Co-Chair, Department of Health Allegra Calder, BERK Consulting
12:30 p.m.	7. Overview Congenital Cytomegalovirus (cCMV)	Kelly Kramer, State Board of Health
12:40 p.m.	8. Parent Perspective	Tawny Hooley Cathleen Ackley
1:00 p.m.	9. cCMV: Natural History, Diagnostic Testing, and Treatment	Dr. Ann Melvin, Seattle Children's Hospital, Infectious Disease

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Time	Agenda Item	Speaker
1:45 p.m.	10. Available Screening Technology	Megan McCrillis, Department of Health
2:15 PM	11. Overview: Early Hearing Detection, Diagnosis, and Intervention (EHDDI) Program	Julie Walker, Department of Health
2:35 p.m.	Break	
2:50 p.m.	12. Available Audiological Resources and Access	Michele Greenwood, Providence Spokane Ear Nose & Throat
3:20 p.m.	13. Discussion and Next Steps	Kelly Oshiro, TAC Co-Chair, State Board of Health Nirupama Nini Shridhar, TAC Co-Chair, Department of Health Allegra Calder, BERK Consulting
4:00 p.m.	Adjournment	
	Note: voting for cCMV will occur on the March 26, 2025, meeting, after the cost-benefit analysis presentation.	

Zoom Meeting Information:

Please click the link below to join the webinar:
https://us02web.zoom.us/j/83392553825?pwd=BkJVgIWHbVGK1r7vhYyHHpOtUQlxPY.1

You can also dial-in using your phone for listen-only mode:
Call in: +1 (253) 215-8782 (not toll-free)
International numbers available: https://us02web.zoom.us/u/kHSKfpCUv
Webinar ID: 833 9255 3825
Passcode: 281973

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Important Meeting Information to Know:

- This meeting is open to the public. The public can observe the meeting online.
- The Technical Advisory Committee will not take formal action or receive public comment. If you have comments or materials you would like to share with the full Board, please send them to wsboh@sboh.wa.gov.
- Times are estimates only. We reserve the right to alter the order of the agenda.
- Every effort will be made to provide Spanish interpretation, and American Sign Language (ASL). Should you need confirmation of these services, please email wsboh@sboh.wa.gov in advance of the meeting date.
- If you would like meeting materials in an alternate format or a different language, or if you are a person living with a disability and need reasonable modification, please contact the State Board of Health at (360) 236-4110 or by email wsboh@sboh.wa.gov. Please make your request as soon as possible to help us meet your needs. Some requests may take longer than two weeks to fulfill. TTY users can dial 711.

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AVISO DE REUNIÓN PÚBLICA

Martes, 11 de febrero de 2025
de 9:00 a. m. a 4:00 p. m.

Nota: Esta es una reunión híbrida que se realiza por Zoom y de forma presencial en el Departamento de Salud del Estado de Washington, Town Center 1, 101 Israel Rd. S.E. Tumwater, WA 98501. Salas de reunión: 163 y 164. A continuación, le proporcionamos el acceso a la reunión y las instrucciones. Hay servicios de interpretación a otros idiomas disponibles.

TAC (por su sigla en inglés, Comité de Asesoramiento Técnico) de la evaluación del recién nacido

Revisión del citomegalovirus congénito (CMVc) y de los criterios para incluirlo en el panel obligatorio de evaluación del recién nacido

Hora	Punto del orden del día	Orador/a
9:00 a. m.	1. Bienvenida y orden del día	Kelly Oshiro, copresidenta del TAC, Mesa Directiva de Salud del Estado Nirupama Nini Shridhar, copresidenta del TAC, Departamento de Salud Kelly Kramer, Mesa Directiva de Salud del Estado Allegra Calder, BERK Consulting
9:15 a. m.	2. Resumen del TAC del mes de enero	Kelly Kramer, Mesa Directiva de Salud del Estado
9:30 a. m.	3. Revisión y debate de los criterios de WA	Kelly Kramer, Mesa Directiva de Salud del Estado Kelly Oshiro, copresidenta del TAC, Mesa Directiva de Salud del Estado Nirupama Nini Shridhar, copresidenta del TAC, Departamento de Salud Allegra Calder, BERK Consulting
10:25 a. m.	Receso	

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Hora	Punto del orden del día	Orador/a
10:35 a. m.	4. Revisión y debate de los criterios de Washington (continuación)	Kelly Kramer, Mesa Directiva de Salud del Estado Kelly Oshiro, copresidenta del TAC, Mesa Directiva de Salud del Estado Nirupama Nini Shridhar, copresidenta del TAC, Departamento de Salud Allegra Calder, BERK Consulting
11:30 a. m.	5. Voto - Revisión de criterios	Kelly Oshiro, copresidenta del TAC, Mesa Directiva de Salud del Estado Nirupama Nini Shridhar, copresidenta del TAC, Departamento de Salud Allegra Calder, BERK Consulting
11:45 a. m.	Almuerzo	
12:15 p. m.	6. Debate y próximos pasos	Kelly Oshiro, copresidenta del TAC, Mesa Directiva de Salud del Estado Nirupama Nini Shridhar, copresidenta del TAC, Departamento de Salud Allegra Calder, BERK Consulting
12:30 p. m.	7. Resumen del citomegalovirus congénito (CMVc)	Kelly Kramer, Mesa Directiva de Salud del Estado
12:40 p. m.	8. Perspectiva del padre o madre	Tawny Hooley Cathleen Ackley
1:00 p. m.	9. cCMV: historia natural, pruebas de diagnóstico y tratamiento	Dr. Ann Melvin, Seattle Children's Hospital, Enfermedades Infecciosas

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Hora	Punto del orden del día	Orador/a
1:45 p. m.	10. Tecnología de detección disponible	Megan McCrillis, Departamento de Salud
2:15 p. m.	11. Resumen: Programa EHDDI (por su sigla en inglés, Programa de Detección, Diagnóstico e Intervención Temprana de la Audición)	Julie Walker, Departamento de Salud
2:35 p. m.	Receso	
2:50 p. m.	12. Recursos audiológicos disponibles y acceso	Michele Greenwood, Providence Spokane Ear Nose & Throat
3:20 p. m.	13. Debate y próximos pasos	Kelly Oshiro, copresidenta del TAC, Mesa Directiva de Salud del Estado Nirupama Nini Shridhar, copresidenta del TAC, Departamento de Salud Allegra Calder, BERK Consulting
4:00 p. m.	Levantamiento de la sesión	

Nota: La votación sobre el cCMV se llevará a cabo en la reunión del 26 de marzo de 2025, después de la presentación del análisis del costo-beneficio.

Información sobre la reunión por Zoom:

Para unirse al seminario web, haga clic en el siguiente enlace:
https://us02web.zoom.us/j/83392553825?pwd=BkJVgIWHbVGK1r7vhYyHHpOtUQlxPY.1

También puede participar por teléfono, mediante la modalidad de solo escucha:
Llamada: +1 (253) 215-8782 (no es un número gratuito)
Números internacionales disponibles: https://us02web.zoom.us/u/kHSKfpCUv
Id. del seminario web: 833 9255 3825
Contraseña: 281973

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Información importante de la reunión que debe saber:

- Esta reunión es pública. El público puede participar como oyente de la reunión.
- El Comité de Asesoramiento Técnico no tomará medidas formales ni recibirá comentarios del público. Si tiene algún comentario o material que desee compartir con toda la Mesa Directiva, envíelos a wsboh@sboh.wa.gov.
- Los horarios son estimativos. Nos reservamos el derecho de modificar el orden de los puntos que se tratarán en la reunión.
- Se hará todo lo posible para proporcionar interpretación en español y ASL (por su sigla en inglés, lenguaje de señas americano). Si necesita la confirmación de estos servicios, envíe un correo electrónico a wsboh@sboh.wa.gov antes de la fecha de la reunión.
- Si desea acceder a los materiales de la reunión en un formato alternativo o en otro idioma, o si tiene una discapacidad y necesita una modificación razonable, comuníquese con la Mesa Directiva de Salud llamando al (360) 236-4110 o enviando un correo electrónico a wsboh@sboh.wa.gov. Le pedimos que presente su solicitud lo antes posible para ayudarnos a satisfacer sus necesidades. Es posible que algunas solicitudes tarden más de dos semanas en atenderse.
  Los usuarios de TTY pueden marcar el número 711.

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Newborn Screening Technical Advisory Committee (TAC)

NBS TAC Membership

MEMBER	ALTERNATE	REPRESENTING
Kelly Oshiro, JD Board Co-Chair Assistant Attorney General		Washington State Board of Health (Board)
Nirupama (Nini) Shridhar, MPH, PhD Department Co-Chair State Genetics Coordinator		Department of Health (Department)
Joan Chappel, RN, MSN Nursing Consultant Advisor/Supervisor	**Sunpreet Bhangoo, RN** Occupational Nurse Consultant	Washington Health Care Authority (HCA)
Byron Raynz Parent Advocate		Parent/Child Advocacy
Emily Shelkowitz, MD Pediatrics, Medical Genetics	**Christina Lam, MD** Medical Director, Biochemical Genetics	Pediatric Specialty Care, Seattle Children's Hospital Biochemical Genetics
Eric Leung, MD Neonatologist		Neonatology and Washington Chapter of the American Academy of Pediatrics (WCAAP)
Heather Hinton, MS Certified Genetic Counselor		Genetic Counseling, MultiCare Yakima Memorial
Joon-Ho Yu, MPH, PhD Pediatrics/Public Health Bioethicist		Bioethics, Department of Epidemiology, University of Washington Bioethics, Treuman Katz Center for Pediatric Bioethics and Palliative Care
Kristine Alexander Senior Medical Policy Research Analyst		Private Insurers, Regence Health Plans
Krystal Plonski, ND, LAc, EAMP, FABNP Naturopathic Pediatrics and Acupuncturist		Naturopaths, Seattle Children's Hospital, and Washington Association of Naturopathic Physicians (WANP)

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Newborn Screening Technical Advisory Committee (TAC)

NBS TAC Membership

MEMBER	ALTERNATE	REPRESENTING
Lisa McGill Vargas, MD Neonatologist	**Rucha Shukla, MD** Neonatologist	Pediatrics, Neonatal-Perinatal Medicine, Sacred Heart Medical Center Neonatology Intensive Care Unit (NICU)
Peggy Harris Public Health and Children's Health Advocate		Parent/Child Advocacy, Save Babies Through Screening Foundation
Priyanka Raut, DNP, MHS, RN Senior Director of Nursing		Pediatrics, Yakima Valley Farmworkers Clinic
Roberta (Bobbie) Salveson, ARNP, PhD Pediatric Nurse Practitioner, Medical Genetics		Pediatric Specialty Care, Mary Bridge Children's Hospital Biochemical Genetics
Taylor Kaminski, Community Doula		Perinatal and Postpartum Care, Global Perinatal Services
María Sigüenza Executive Director		State Commissions, Commission on Hispanic Affairs
Molly Parker, MD, MPH Family Medicine Physician		Provider, Population Health, Jefferson Healthcare
Cathleen Ackley Parent Advocate		Parent/Child Advocacy
Steve Kutz, BSN, MPH Chair, Washington State American Indian Health Commission		State Commissions, American Indian Health Commission
Tawny Hooley Parent Advocate		Parent/Child Advocacy

NBS TAC Staff Support

Kelly Kramer, Board Newborn Screening Policy Advisor
John Thompson, Department Director of Newborn Screening
Megan McCrillis, Department Newborn Screening Policy Advisor
Molly Dinardo, Board Policy Advisor

Crystal Ogle, Board Administrative Assistant
Michelle Larson, Board Communications Manager
Anna Burns, Board Communications Consultant

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Newborn Screening Technical Advisory Committee (TAC)

Newborn Screening Technical Advisory Committee (TAC) Charter

Start Date: October 28, 2024 **End Date:** June 30, 2025 (tentative)
Members: See TAC Membership Addendum A

OBJECTIVE

Serve as an expert advisory committee on newborn screening for the Washington State Board of Health (Board). Review and recommend possible updates to the Board's current newborn screening process and criteria. Additionally, evaluate several candidate conditions for potential inclusion in the Washington State mandatory newborn screening panel and provide recommendations to the Board.

BACKGROUND

The Board establishes the rules for newborn screening in Washington, including deciding which conditions all newborns must be tested for at birth. To make these decisions, the Board assembles a multidisciplinary Technical Advisory Committee (TAC) comprised of family representatives and representatives from healthcare, social services, advocacy organizations, public health, and more. Using available evidence, the TAC then assesses candidate conditions using guiding principles and five newborn screening criteria to determine which conditions should be added to the panel.

KEY ACTIVITIES

This TAC is being convened to complete the following key activities:

- Review the Board's current newborn screening candidate condition review process and criteria and identify opportunities for improvement.
- Determine whether branched-chain ketoacid dehydrogenase kinase (BCKDK) deficiency meets the Board's criteria for newborn screening panel inclusion and provide a recommendation to the Board. This is a requirement of Senate Bill 6234 (Chapter 105, Laws of 2024).
- Determine whether congenital cytomegalovirus (cCMV) meets the Board's criteria for newborn screening and provide a recommendation to the Board. This is a requirement of Senate Bill 5829 (Chapter 96, Laws of 2024).
- Review other possible candidate conditions recently brought in front of the Board between 2024 and 2025.

TAC TIMELINES (Tentative)

- Meeting 1, Process and Criteria Review – Monday, October 28, 2024
- Meeting 2, BCKDK Deficiency Review – January 2025
- Meeting 3, Criteria Intro to cCMV – February 2025
- Meeting 4, Cost-Benefit Analysis of cCMV – March 2025

COMMITTEE NORMS AND EXPECTATIONS

- Be here now and stay purpose-oriented
- Listen for understanding; seek clarification and resist assumptions
- Appreciate the strength of diverse cultures and perspectives
- Engage respectfully; see with new eyes and hear with new ears
- Move up into a speaking role; move into a listening role
- Stay on topic and mind the time
- Assume positive intent; acknowledge and repair harms
- Try to avoid speaking with someone else is speaking
- Commit to using inclusive language in committee discussions and if possible, try to avoid using idioms or slang terms
- State your name each time you begin talking, and speak at a moderate pace to ensure language interpreters can appropriately translate what is being said
- Use acronyms where possible after introducing technical terms or proper nouns and encourage other committee members to do the same.

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Newborn Screening Technical Advisory Committee (TAC)

Newborn Screening Technical Advisory Committee (TAC) Charter

DECISION MAKING

- Proposed voting methods: This committee will use anonymous voting via Microsoft Forms and open discussion of results to inform committee decisions and recommendations.
- Proposed Primary or Alternative Member voting: Both primary and alternative TAC Members may attend these meetings, however, if both are in attendance the primary TAC member will be responsible for speaking and voting during the meeting. The alternative member only speaks and votes when the primary is not in attendance.

INFORMATION SHARING

The Newborn Screening TAC planning team will:

- Email and post meeting materials at least 48 hours before the scheduled meeting.
- Email updates and notices to TAC members and designated alternatives.
- Post information on the Newborn Screening Criteria Review Project webpage.

RESOURCES/REFERENCE MATERIALS

- Chapter 246-650 WAC – Newborn Screening.
- Washington State Board of Health Process to Evaluate Conditions for Inclusion in the Required Newborn Screening Panel.
- Washington Department of Health Newborn Screening Webpage

GUIDANCE FOR SPEAKING WITH LANGUAGE INTERPRETATION

The Washington State Board of Health (Board) offers American Sign Language and Spanish interpretation during our regular public meetings. We do this as a part of our work towards increasing language access.

We ask all speakers at Board meetings to follow this guidance to create an accessible meeting environment. If you have any questions or need guidance for presenting, please contact Board staff for support.

WHAT TO EXPECT DURING A BOARD MEETING

- You will receive a simplified version of this document at your seat on the day of the Board meeting.
- Board staff or interpreters may give you cues to slow down your pace. The cues may include:
  - Raising a paddle sign to signal you to slow down.
  - Making a brief verbal interruption asking you to slow down.

TIPS FOR SPEAKING AND PRESENTING DURING THE MEETING

We ask that you help us mitigate the need for interruptions by speaking at a comfortable pace. Our ASL and Spanish interpreters cannot deliver your message accurately if you speak too quickly.

- Take a breath after each sentence to give the interpreter time to deliver your message.
- If you are reading from a script, please be aware that you may read faster than you speak.
- To help the interpreters and audience identify you, state your name each time you begin talking.
- Wait until someone else finishes speaking before you speak. Interpreters can only choose one person to interpret at a time.
- Pause after introducing technical terms, proper nouns, dates, numbers, or figures to allow for interpretation.

TIPS FOR TECHNICAL TERMS

- We recommend including a pause after introducing technical terms, proper nouns, dates, numbers, or figures.
  - Example: *"This briefing will discuss rulemaking around newborn screening for Ornithine Transcarbamylase Deficiency (OTCD) [pause for interpretation, wait for cue from interpreter to continue], Chapter 246-650 WAC [pause for interpretation, wait for cue from interpreter to continue]."*
- After you introduce technical terms or proper nouns use their acronyms for the remainder of the introduction.
  - Example: *"For the remainder of this discussion, I will refer to this condition as OTCD."*
- If you are using visual materials (e.g., tables), incorporate descriptive language of the visual material.
  - Example: *"This is a table showing XXXX. And now, we'll look at this part of the table…"*

Newborn Screening Criteria Updates and Congenital Cytomegalovirus (cCMV) Review

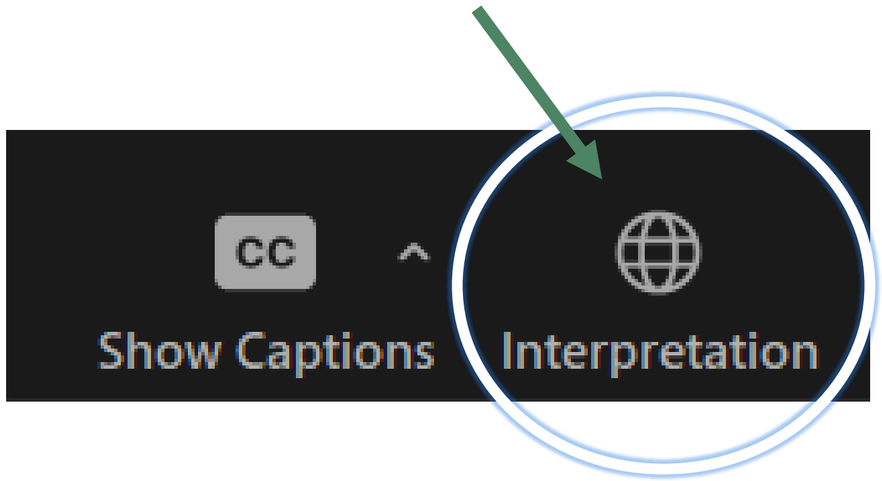
Newborn Screening Technical Advisory Committee (TAC)

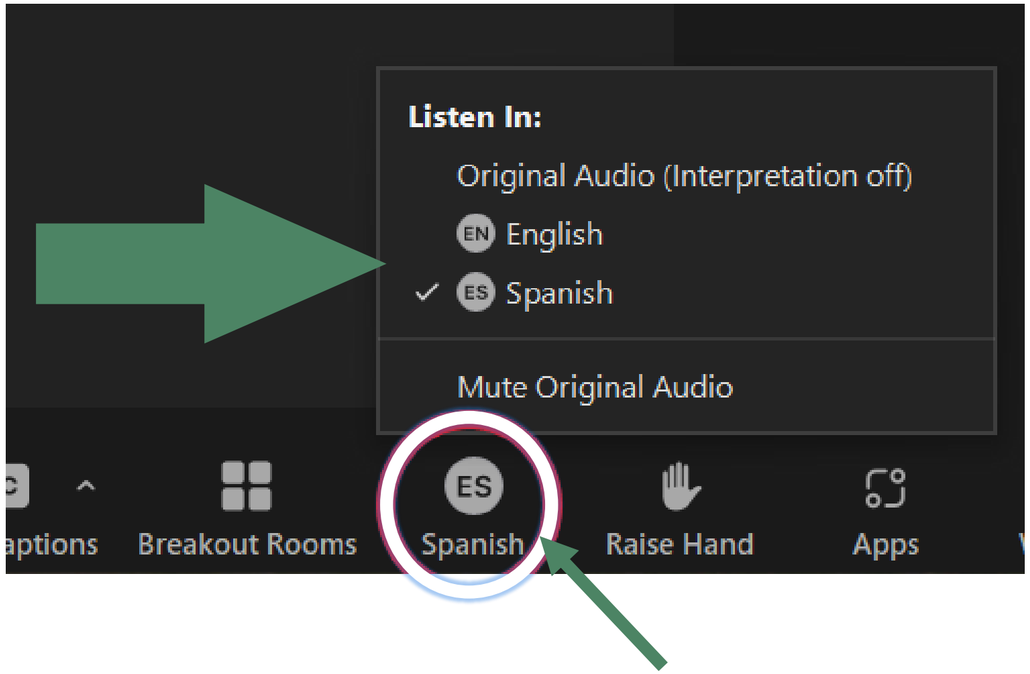
February 11, 2025

WASHINGTON STATE BOARD OF HEALTH

Canales de Idioma de Zoom
Zoom Language Channels

Canales de idioma
Language channels





Elige un idioma
Choose a language

Zoom Webinar Functions

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View participants, change your name

Closed Captioning/Live Transcription

Turn webcam on/off

Raise hand feature

Leave meeting

Note: Depending on your role, you may not have access to all functions identified on this slide.

Introductions

Agenda

- Meeting Introduction and Overview
- Meeting Recap
- Part 1: Washington Criteria Updates
  - Discussion
  - Vote on updates
  - Next Steps
- Part 2: Review of Congenital Cytomegalovirus
  - Overview
  - Parent Perspectives
  - Subject Matter Expertise
  - Screening Technology
- Identify the Committee’s Next Steps and Recommendations for the Board

Meeting Recaps

January 14 TAC Meeting

- Branch Chain Ketoacid Dehydrogenase Kinase Deficiency (BCKDK) review
  - Voting Results
    - Five Criteria:
      - Mixed results
      - Many concerns for lack of evidence
    - Overall Vote:
      - All but one TAC member voted to not include BCKDK deficiency

- Five Criteria Review
  - Uncertainty about editing current criteria
    - Vote about necessity to change criteria:
      - Six 'yes,' six 'no,' and two were 'unsure.'
  - Newborn Screening Program to provide suggested edits
  - Vote for inclusion at next TAC meeting

Meeting Objectives

- Review each of the criteria; make recommendations to adopt suggested edits and additions.

- Review the condition congenital cytomegalovirus against the Board’s criteria.

Newborn Screening Criteria

1) Available Screening Technology

2) Diagnostic Testing and Treatment Available

3) Prevention Potential and Medical Rationale

4) Public Health Rationale

5) Cost-Benefit and Cost Effectiveness

6) Public Health Readiness

1. Available Screening Technology

Sensitive, specific, and timely tests are available for the condition that can be adapted to mass screening.

- The sensitivity of the screening test is estimated to be ≥95%.
- The specificity of the screening test is considered acceptable based on the estimated number of false positive results and their potential impact on the healthcare system, newborn screening program, and families.
- A timely test is one that enables intervention before irreversible harm develops, within the current standard timeframes for specimen collection, receipt, testing, and reporting.
- There is adequate peer reviewed evidence to evaluate this criterion.

2. Diagnostic Testing and Available Treatment

Accurate diagnostic tests, medical expertise, and effective treatment are available for evaluation and care of all infants identified with the condition.

- A diagnostic test accurately identifies who needs treatment and is readily available to all newborns screened.
- The available treatment is effective in reducing morbidity or mortality and outweighs any risks or harms of the treatment.
- The medical expertise needed to diagnose and care for those with a positive newborn screen is reasonably available to all newborns screened.
- The availability and proximity to treatment for anyone diagnosed with the condition is considered acceptable based on the frequency of treatment needed.
- The appropriate consultants and treatment centers have been identified and have capacity for the expected increase in diagnostic testing and/or referrals.

3. Prevention Potential and Medical Rationale

The newborn identification of the condition allows early diagnosis and intervention. Important considerations include:

- There is sufficient time between birth and onset of irreversible harm to allow for diagnosis and intervention.
- The condition must have an onset form that occurs in infancy (within the first year of life); newborn screening is not appropriate for conditions that only present after the first year of life.
- The benefits of detecting and treating ~~early onset~~ infantile-onset forms of the condition (within one year of life) balance the impact of detecting later onset forms of the condition.
- ~~Newborn screening is not appropriate for conditions that only present in adulthood.~~
- There is adequate evidence of acceptable quality to evaluate this criterion.

Break

4. Public Health Rationale

The nature of the condition justifies population-based screening rather than risk-based screening or other approaches.

- All available risk-based screening tools for the condition have been considered and are found to be inferior to universal newborn screening.
- There is adequate evidence of acceptable quality to evaluate this criterion.

5. Cost-benefit and Cost-effectiveness

The outcomes outweigh the costs of screening. All outcomes, both positive and negative, need to be considered in the analysis. ~~Important considerations to be included in the economic analyses include:~~

- The economic analysis considers:
  - The prevalence of the condition among newborns.
  - The positive and negative predictive values of the screening and diagnostic tests.
  - Variability of clinical presentation by those who have the condition.
  - Dollar values for costs and benefits of screening vs. no screening.
- The impact of ambiguous results, adverse effects, or unintended consequences of screening, such as emotional or economic impacts on the family and medical system, must also be considered.
- The results of the economic analysis shows that the outcomes, financial or otherwise, outweigh the costs of screening
- There is adequate evidence of acceptable quality to evaluate this criterion
- ~~The impact of ambiguous results. For example, the emotional and economic impact on the family and medical system.~~
- ~~Adverse effects or unintended consequences of screening.~~

6. Public Health Readiness

The Newborn Screening Program's capacity to implement screening within a reasonable timeframe has been considered.

- The systems and staffing necessary to perform the test and report screening results have been identified.
- Resources needed to implement short/long term follow up protocols by the newborn screening program have been identified.

Voting

Washington Newborn Screening Criteria Voting Ballot: February 2025

Lunch

Results and Discussion

Congenital Cytomegalovirus (cCMV) Review

Newborn Screening Technical Advisory Committee (TAC)

February 11, 2025

WASHINGTON STATE BOARD OF HEALTH

Overview of cCMV

- Last legislative session, Senate Bill 5829 was passed
  - The bill directed the Board of Health to conduct a review of cCMV to determine if this condition should be added to our mandatory newborn screening panel (NBS).

- Included on other state NBS panels
  - Not all using dried bloodspot method

- Has not yet been included on the RUSP
  - Previously reviewed in 2022

- Previously reviewed by Washington TAC in 2022

2022 Washington TAC Review

- 2022 TAC voted overall to not recommend cCMV to Washingtons NBS panel
  - 14 out of 17 voted to not recommend cCMV and voted to review it again at a future date.
  - Most felt it did not meet Criterion 2, Diagnostic Testing and Treatment Available.
  - Split for Criterion 4, Public Health Rationale.
  - Mixed responses for Criterion 5, Cost-benefit.

- Last review included options for Washington to screen universally or targeted screening for cCMV after a failed hearing screen.

Patient and Family Perspective

cCMV: Natural History, Diagnostic Testing, and Treatment

Available Screening Technology

Early Hearing, Detection, Diagnosis, and Intervention Program

Break

Access and Equity

Discussion

WASHINGTON STATE BOARD OF HEALTH

THANK YOU

To request this document in an alternate format, please contact the Washington State Board of Health at 360-236-4110, or by email at **wsboh@sboh.wa.gov** | TTY users can dial 711

ACCESSIBILITY AND THE AMERICANS WITH DISABILITIES ACT (ADA)

- The Washington State Board of Health (Board) is committed to providing information and services that are accessible to people with disabilities. We provide reasonable accommodations, and strive to make all our meetings, programs, and activities accessible to all persons, regardless of ability, in accordance with all relevant state and federal laws.
- Our agency, website, and online services follow the Americans with Disabilities (ADA) standards, Section 508 of the Rehabilitation Act of 1973, Washington State Policy 188, and Web Content Accessibility Guidelines (WCAG) 2.0, level AA. We regularly monitor for compliance and invite our users to submit a request if they need additional assistance or would like to notify us of issues to improve accessibility.
- We are committed to providing access to all individuals visiting our agency website, including persons with disabilities. If you cannot access content on our website because of a disability, have questions about content accessibility or would like to report problems accessing information on our website, please call (360) 236-4110 or email wsboh@sboh.wa.gov and describe the following details in your message:
    - The nature of the accessibility needs
    - The URL (web address) of the content you would like to access
    - Your contact information

We will make every effort to provide you the information requested and correct any compliance issues on our website.

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

Washington State Board of Health

PROCESS TO EVALUATE CONDITIONS FOR INCLUSION IN THE REQUIRED NEWBORN SCREENING PANEL

The Washington State Board of Health has the duty under RCW 70.83.050 to define and adopt rules for screening Washington-born infants for heritable conditions. Chapter 246-650-020 WAC lists conditions for which all newborns must be screened. Members of the public, staff at Department of Health, and/or Board members can request that the Board review a particular condition for possible inclusion in the NBS panel. In order to determine which conditions to include in the newborn screening panel, the Board convenes an advisory committee to evaluate candidate conditions using guiding principles and an established set of criteria.

The following is a description of the Qualifying Assumption, Guiding Principles, and Criteria which the Board has approved in order to evaluate conditions for possible inclusion in the newborn screening panel. The Washington State Board of Health and Department of Health apply the qualifying assumption. The Board appointed Advisory Committee applies the following three guiding principles and evaluates the five criteria in order to make recommendations to the Board on which condition(s) to include in the state's required NBS panel.

QUALIFYING ASSUMPTION

Before an advisory committee is convened to review a candidate condition against the Board's five newborn screening requirements, a preliminary review should be done to determine whether there is sufficient scientific evidence available to apply the criteria for inclusion.

THREE GUIDING PRINCIPLES

Three guiding principles govern all aspects of the evaluation of a candidate condition for possible inclusion in the NBS panel.

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

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 early diagnosis and intervention. Important considerations:
    - There is sufficient time between birth and onset of irreversible harm to allow for diagnosis and intervention.
    - The benefits of detecting and treating early onset forms of the condition (within one year of life) balance the impact of detecting late onset forms of the condition.
    - Newborn screening is not appropriate for conditions that only present in adulthood.
4. **Public Health Rationale:** 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. Important considerations to be included in economic analyses include:
    - The prevalence of the condition among newborns.
    - The positive and negative predictive values of the screening and diagnostic tests.
    - Variability of clinical presentation by those who have the condition.
    - The impact of ambiguous results. For example the emotional and economic impact on the family and medical system.
    - Adverse effects or unintended consequences of screening.

WASHINGTON STATE
BOARD OF HEALTH

Washington State Board of Health

PROCESS TO EVALUATE CONDITIONS FOR INCLUSION IN THE REQUIRED NEWBORN SCREENING PANEL

Last updated ~~November 13, 2024~~ January 2025

Amended Section (Approved November 2024)
The Washington State Board of Health (Board) has the duty under RCW 70.83.050 to define and adopt rules for screening Washington-born infants for heritable conditions. Chapter 246-650-020 WAC lists conditions for which all newborns must be screened. Members of the public, staff at Department of Health (Department), and/or Board members can request that the Board review a particular condition for possible inclusion in the newborn screening (NBS) panel. ~~In order to~~ To determine which conditions to include in the ~~newborn screening~~ NBS panel,~~.~~ the Board convenes a~~n~~ newborn screening technical advisory committee (TAC) to evaluate candidate conditions using guiding principles and an established set of criteria.

~~The following is a description of~~ This document describes the Qualifying Assumption, Guiding Principles, and Criteria ~~which~~ the Board has approved ~~in order~~ to evaluate conditions for possible inclusion in the newborn screening panel. The ~~Washington State Board of Health~~ Board and Department ~~of Health~~ apply the qualifying assumption. The Board-appointed Newborn Screening ~~Advisory Committee~~ TAC applies the following three guiding principles and evaluates the five criteria ~~in order~~ to make recommendations to the Board on which condition(s) to include in the state's required NBS panel.

QUALIFYING ASSUMPTION

Amended Section (Approved November 2024)
Before ~~an advisory committee is convened~~ the Board convenes a TAC to review a candidate condition against the ~~Board's~~ five newborn screening ~~requirements~~ criteria, staff should complete a preliminary review ~~should be done~~ to determine ~~there is~~ whether sufficient scientific evidence is available to apply the criteria for inclusion, which is the qualifying assumption. If the candidate condition is on the Health Resources and Services Administration (HRSA) Recommended Uniform Screening Panel (RUSP), the Board and Department will consider the qualifying assumption met and convene a TAC.

New Section (Approved November 2024)
***A note on the RUSP*:** The RUSP is a list of conditions that the Secretary of the Department of Health and Human Services (HHS) recommends states screen for as part of their newborn screening programs. Once the HHS Secretary recommends a new condition, the Board and Department will review it for possible inclusion in the Washington NBS panel within two years of the recommendation.

New Section (Approved January 2025)

Conditions pending RUSP Review or Previously Denied for the RUSP: RCW 34.05.330 of the Administrative Procedures Act (APA) allows any person to petition a state agency to adopt, repeal, or amend any rule within its authority. Agencies must respond to the petitioner within 60 days. If the agency accepts the petition, it must initiate rulemaking. An agency can deny the request for rulemaking, and in doing so, it must explain its reasons and, if appropriate, describe alternative steps it is prepared to take.

If the Board receives a petition for rulemaking regarding a candidate condition currently under review for the RUSP, the Board will wait until the federal committee finishes its review and the HHS Secretary makes a final decision before convening a TAC. For petitions involving conditions that have already been reviewed and denied inclusion on the RUSP, the Board will instruct staff to work with the petitioner to determine if concerns raised during the federal review have been addressed before recommending the Board convene a TAC to review the condition.

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THREE GUIDING PRINCIPLES

Three guiding principles govern all aspects of the evaluation of a candidate condition for possible inclusion in the NBS panel.

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

CRITERIA

Amended Section (Pending Board Approval)

1. **Available Screening Technology:** Sensitive, specific and timely tests are available that can be adapted to mass screening.

- The sensitivity of the screening test is estimated to be ≥95%.
- The specificity of the screening test is considered acceptable based on the estimated number of false positive results and their potential impact on the healthcare system, newborn screening program, and families.
- A timely test is one that enables intervention before irreversible harm develops, within the current standard timeframes for specimen collection, receipt, testing, and reporting.
- There is adequate peer reviewed evidence to evaluate this criterion.

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.

- A diagnostic test accurately identifies who needs treatment and is readily available to all newborns screened.
- The available treatment is effective in reducing morbidity or mortality and outweighs any risks or harms of the treatment.
- The medical expertise needed to diagnose and care for those with a positive newborn screen is reasonably available to all newborns screened.
- The availability and proximity to treatment for anyone diagnosed with the condition is considered acceptable based on the frequency of treatment needed.
- The appropriate consultants and treatment centers have been identified and have capacity for the expected increase in diagnostic testing and/or referrals.

3. **Prevention Potential and Medical Rationale**: The newborn identification of the condition allows early diagnosis and intervention.

- There is sufficient time between birth and onset of irreversible harm to allow for diagnosis and intervention.
- The condition must have an onset form that occurs in infancy (within the first year of life); newborn screening is not appropriate for conditions that only present after the first year of life.
- The benefits of detecting and treating ~~early onset~~ infantile-onset forms of the condition (within one year of life) balance the impact of detecting later onset forms of the condition.

- ~~Newborn screening is not appropriate for conditions that only present in adulthood.~~
- There is adequate evidence of acceptable quality to evaluate this criterion.

4. **Public Health Rationale:** Nature of the condition justifies population-based screening rather than risk based screening or other approaches.
- All available risk-based screening tools for the condition have been considered and are found to be inferior to universal newborn screening.
- There is adequate evidence of acceptable quality to evaluate this criterion.

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. ~~Important considerations to be included in the economic analyses include:~~
- The economic analysis considers:
    - The prevalence of the condition among newborns.
    - The positive and negative predictive values of the screening and diagnostic tests.
    - Variability of clinical presentation by those who have the condition.
    - Dollar values for costs and benefits of screening vs. no screening.
- The impact of ambiguous results, adverse effects, or unintended consequences of screening, such as emotional or economic impacts on the family and medical system, must also be considered.
- The results of the economic analysis shows that the outcomes, financial or otherwise, outweigh the costs of screening
- There is adequate evidence of acceptable quality to evaluate this criterion
- ~~The impact of ambiguous results. For example, the emotional and economic impact on the family and medical system.~~
- ~~Adverse effects or unintended consequences of screening.~~

6. **Public Health Readiness:** The Newborn Screening Program's capacity to implement screening within a reasonable timeframe has been considered.
- The systems and staffing necessary to perform the test and report screening results have been identified.

- Resources needed to implement short/long term follow up protocols by the newborn screening program have been identified.

Criterion	Opinion			Comments
	Meets	Does not meet	More info needed	
1. Available Screening Technology Sensitive, specific and timely tests are available that can be adapted to mass screening				
The sensitivity of the screening test is estimated to be ≥95%				
The specificity of the screening test is considered acceptable based on the estimated number of false positive results and their potential impact on the healthcare system, newborn screening program, and families				
A timely test is one that enables intervention before irreversible harm develops, within the current standard timeframes for specimen collection, receipt, testing, and reporting				
There is adequate evidence of acceptable quality to evaluate this criterion				
Overall impression of criterion 1:				
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				
A diagnostic test accurately identifies who needs treatment, and is readily available to all newborns screened.				

The available treatment is effective in reducing morbidity or mortality, and outweighs any risks or harms of the treatment.				
The medical expertise needed to diagnose and care for those with a positive newborn screen is reasonably available to everyone screened				
The availability and proximity to treatment for anyone diagnosed with the condition is considered acceptable based on the frequency of treatment needed				
The appropriate consultants and treatment centers have been identified and have capacity for the expected increase in diagnostic testing and/or referrals				
There is adequate evidence of acceptable quality to evaluate this criterion				
Overall impression of criterion 2:				
3. Prevention Potential and Medical Rationale The newborn identification of the condition allows early diagnosis and intervention.				
There is sufficient time between birth and onset of irreversible harm to allow for diagnosis and intervention				
The condition must have an onset form that occurs in infancy (within the first year of life); newborn screening is not appropriate for conditions that only present after the first year of life.				
The benefits of detecting and treating infantile-onset forms of the condition				

balance the impact of detecting later onset forms of the condition				
There is adequate evidence of acceptable quality to evaluate this criterion				
Overall impression of criterion 3:				
4. Public Health Rationale Nature of the condition justifies population-based screening rather than risk based screening or other approaches				
Any available risk-based screening tools for the condition have been considered and are inferior to universal newborn screening				
There is adequate evidence of acceptable quality to evaluate this criterion				
Overall impression of criterion 4:				
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				
The economic analysis considers: o The prevalence of the condition among newborns. o The positive and negative predictive values of the screening and diagnostic tests. o Variability of clinical presentation by those who have the condition. o Dollar values for costs and benefits of screening vs. no screening				
The impact of ambiguous results, adverse effects, or unintended consequences of screening , such as emotional or economic impacts on the family and medical system, must also be considered.				

The results of the economic analysis shows that the outcomes, financial or otherwise, outweigh the costs of screening				
There is adequate evidence of acceptable quality to evaluate this criterion.				
Overall impression of criterion 5:				
6. Public Health Readiness The Newborn Screening Program's capacity to implement screening within a reasonable timeframe has been considered				
The systems and staffing necessary to perform the test and report screening results have been identified				
Resources needed to implement short/long term follow up protocols by the newborn screening program have been identified				
Overall impression of criterion 6:				
Overall impression of the condition:				
Recommendation:				

Washington State Department of HEALTH

WASHINGTON STATE BOARD OF HEALTH

Newborn Screening Technical Advisory Committee (TAC)

Newborn Screening (NBS) Technical Advisory Committee (TAC) Voting Instructions

Please use the Microsoft Forms ballot provided by staff during the meeting to vote.

All votes are anonymous. Your votes will be collected and presented by the TAC facilitator and Co-Chairs for further discussion by the group.

Instructions:

- Only TAC members may vote.
- Do not forward or share the form/ballot.
- If you are unsure of not comfortable voting on these options, please indicate so in the form.

If you encounter any technical issues or difficulties accessing the form, please let staff know as soon as possible.

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Newborn Screening Technical Advisory Committee (TAC)

Congenital Cytomegalovirus (cCMV) Overview
Newborn Screening Technical Advisory Committee
February 11, 2025

ABOUT THE CONDITION

- Congenital cytomegalovirus (cCMV) occurs when a pregnant person is infected with cytomegalovirus (CMV) and passes the infection to their unborn child.
- About 1 in 200 babies in the United States are born with cCMV.
- cCMV can result in decreases in hearing and is the leading cause of nonhereditary, sensorineural hearing change.
- About 1 in 5 babies with a cCMV infection will have long term health impacts, including decreases in hearing.
- cCMV can lead to other significant impacts, including developmental delay, changes in vision, seizures, or death.

SYMPTOMS

- Babies born with cCMV can have brain, liver, spleen, lung, and growth problems.
- The most common long-term health problem with cCMV infection is decreases in hearing.
- Decreases in hearing may be detected soon after birth or may develop later in childhood.

DIAGNOSIS

- Infants suspected of having cCMV can have a diagnostic DNA test for CMV infection.
    - Urine or saliva samples are the preferred samples for testing.
    - Blood samples may be used to test newborns with suspected CMV infection, however, compared with urine or saliva, it is not the most accurate option.
- Diagnostic testing must be completed within 21 days of life to confirm a congenital infection.

TREATMENT

- Antivirals can be used to treat babies born with symptoms of cCMV.
- Some antivirals, such as Valganciclovir, may cause serious side effects.
- Antivirals may reduce changes in hearing and improve development.
- All children born with cCMV should have regular hearing and vision checks.

- CDC. “About Cytomegalovirus.” *Cytomegalovirus (CMV) and Congenital CMV Infection*, 10 May 2024, www.cdc.gov/cytomegalovirus/about/index.html.
- Akpan US, Pillarisetty LS. Congenital Cytomegalovirus Infection. [Updated 2023 Aug 8]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2025 Jan-. Available from: https://www.ncbi.nlm.nih.gov/books/NBK541003/

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WASHINGTON STATE BOARD OF HEALTH

Newborn Screening Technical Advisory Committee: Congenital Cytomegalovirus (cCMV) September 2022 TAC Summary Notes

The following is a summary of the technical advisory committee review of cCMV.

Overview: Newborn Screening Program & Early Hearing Detection, Diagnosis, and Intervention (EHDDI) Program

- John Thompson from the newborn screening program explained that the panel tests for 32 disorders using blood specimens, conducting nearly 12 million tests annually. Around 200 infants benefit from early diagnosis and treatment, which can prevent severe outcomes.
- An overview of the EHDDI program explained that they ensure all infants meet national hearing screening goals. Early detection is crucial for language development in the first few months of life, as missed or delayed diagnoses can lead to developmental delays.

Natural History of cCMV; Diagnostic Testing & Treatment

- Dr. Joseph Bocchini, the subject matter expert for cCMV, explained that cCMV is the most common congenital infection, is a major contributor to decreases in hearing and neurodevelopmental delays. He explained that some babies are asymptomatic at birth, early diagnosis and antiviral treatments can prevent or reduce complications. Prevention strategies, including education on hygiene practices, are critical in reducing the spread of cCMV during pregnancy.
- The clinical trial process for selecting asymptomatic infants for antiviral therapy was also discussed, focusing on the potential benefits of early intervention.

Family Perspective

- A family shared their experience with cCMV, discussing the emotional and medical impact, including antiviral treatment and cochlear implants. The importance of advocacy for cCMV families was emphasized.

(continued on the next page)

Available Screening Technology

- The Newborn Screening and EHDDI programs outlined different strategies for screening for cCMV, universal or targeted. Both blood specimen-based (universal screening) and hearing screening technologies (targeted screening) for cCMV were discussed. Approximately 170 infants are detected with decreases in hearing annually, and around 31 of these babies develop late onset hearing loss, highlighting the importance of regular monitoring.
- Questions about the costs of screening and the availability of long-term follow-up services for families were also raised, underscoring the need for comprehensive care beyond initial screenings.

Cost-Benefit Analysis

- The Newborn Screening Program provided a presentation on the economic model and results of the cost-benefit analysis. The hallmark of the newborn screening program is to prevent death and disability, and the team identified through their research that there is currently not strong evidence that screening for cCMV prevents death and disability. However, early detection and intervention could still offer long-term benefits, such as improved hearing and developmental outcomes.
- The TAC discussed alternative methods to detect and manage late onset hearing loss in children, as well as the importance of providing education and resources for families, which cannot be fully captured in financial analyses.

Application of Criteria & Discussion

- The TAC discussed several factors related to the screening of cCMV, such as the availability of technology, the adequacy of the audiology workforce, especially in rural areas, and whether alternative sample collection methods (e.g., urine) could be considered.
- There was also a focus on addressing healthcare disparities in rural and institutionally underserved communities, ensuring equitable access to audiology services, and addressing the emotional burden on families dealing with late onset hearing loss in children.

Votes & Discussion

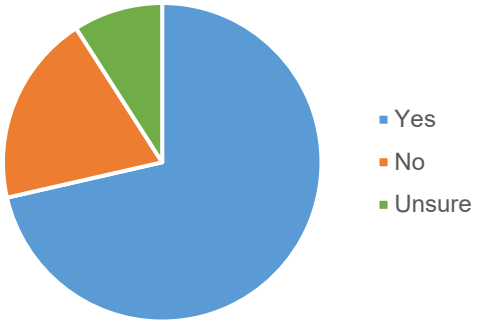
- TAC members took two anonymous votes: one to assess if cCMV meets the Board's criteria for inclusion in the newborn screening program and another to make an overall recommendation to the Board. The discussion following the votes focused on the challenges of implementing cCMV screening, including the availability of data, resources, and input from the deaf and hard-of-hearing communities.

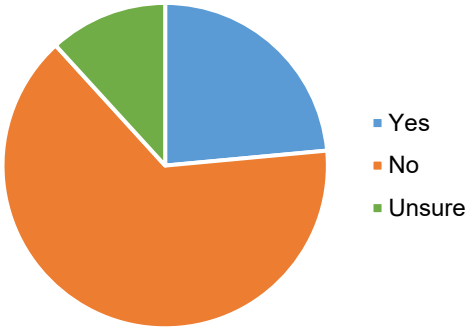
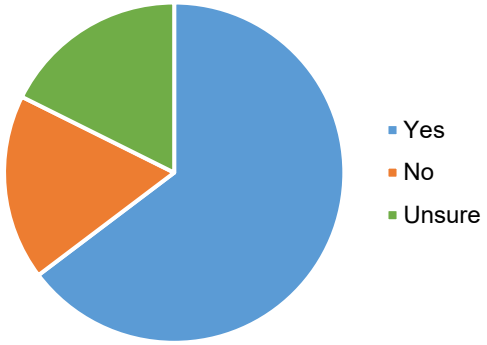
- The next steps involve further discussions, including outreach to communities, and the possibility of addressing issues like equitable access to care and alternative screening methods for late onset hearing loss.

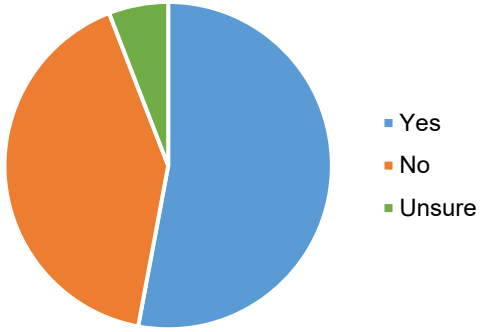
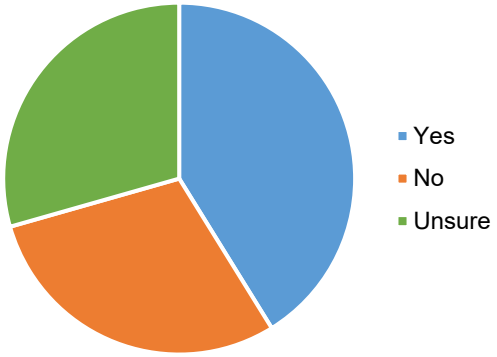
Summary of Voting Comments

The following is a compilation of comments from technical advisory committee (TAC) members provided when voting on each individual criteria, and an overall recommendation. Comments have been summarized and are organized by each criterion and then overall comments provided.

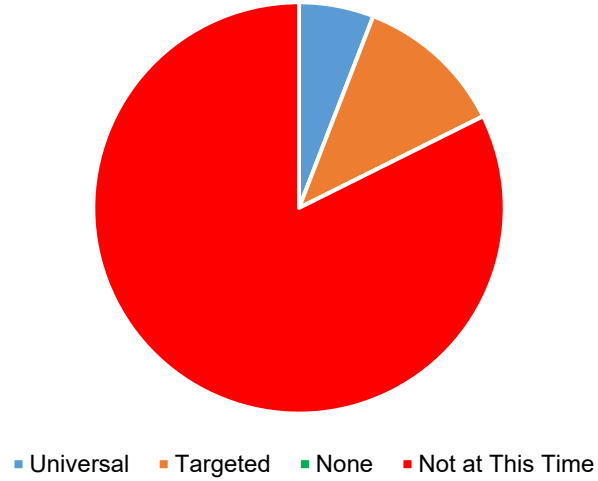
Criteria Evaluation

Criteria	Major themes
1. Available Screening Technology 	• The sensitivity of 75% is insufficient for the blood spot assay. Higher sensitivity testing approaches (i.e., urine or saliva PCR testing) are not feasible, as we do not currently have the infrastructure for these approaches. • While the blood spot tests are not as sensitive, universal screening would still identify 27 additional babies with late onset hearing loss and early intervention. • Blood spot test sensitivity is acceptable. • Universal screening may not be feasible, but targeted screening could be feasible.
2. Diagnostic Testing and Treatment Available	• Lack of infrastructure and resources as it relates to increased hearing screening, monitoring, and follow-up; available audiology services in the state; training for audiologists and medical providers; availability of treatment; overall personnel for education and training; and alternative models for screening by primary care providers. • While it appears early intervention is effective for infants with late onset hearing loss, there is currently no established effective treatment for cCMV.

	• Why is cCMV not on the federal Recommended Uniform Screening Panel (RUSP)? • Unclear how much hearing interventions change outcomes. • One thought would be to educate pregnant women and possibly test for CMV during pregnancy.
3. Prevention Potential and Medical Rationale 	• There is no definitive treatment for cCMV; unsure that irreversible harm can be prevented. • Benefits of early antiviral treatment for cCMV are not well understood. As antiviral treatment (i.e., valganciclovir) is only used for patients with moderate to severe symptomatic cCMV, there is limited evidence on effectiveness of antivirals to treat asymptomatic babies. • Dried blood spot universal screening will not improve early diagnosis. Without screening most will be detected and receive care, albeit later. • Benefits of early intervention for late onset hearing loss are more clear. There may be benefits from earlier detection with regard to early childhood intervention and special education interventions on language development and education success. • Hearing is a contested medical goal by the deaf community, and the deaf community would argue for equity education for those with hearing impairment. • Early intervention is key to many problems, and this type of screening is a form of early intervention

4. Public Health Rationale  	• Risked-based or targeted screening would be more effective. • Population-based screening is justified, but not with the blood spot sample. • The public health rationale is present in theory, but the diagnostic and treatment technology doesn't exist at present to realize that benefit. • Hearing screens are done on a routine basis; we have school screenings that can further help with evaluation and detection. I think this would probably overwhelm an already overwhelmed system. • It is not clear that focusing on CMV will change the population of children with hearing loss. Parent-based assessments of hearing and language will allow detection of those with impairments. It may be better to focus on parent, school, and pediatrician education.
5. Cost Benefit / Cost Effectiveness  	• Based on the modeling and data presented, universal screening has a low cost-benefit ratio; does not seem to be very cost-effective. • The cost-benefit ratio is not comparable to other newborn screening conditions. • Even with an early diagnosis of cCMV, only a minority of babies with that diagnosis will develop late onset hearing loss. • Much of the cost effectiveness can't be quantified. There is a large emotional cost for families whose baby is diagnosed with cCMV who then are waiting years to find out whether their child will develop late onset hearing loss.

Overall Recommendation



Recommendation Options	Major themes
1. I recommend the Board add universal screening of cCMV to the list of conditions for which all Washington-born newborns must be screened.	*No comments received.*
2. I recommend the Board pursue steps to include targeted screening of cCMV to the list of conditions for which all Washington-born newborns must be screened. Note: this requires a change in the Board's statutory authority via legislation.	If the cost-benefit analysis is not sufficient for universal screening at this time, the targeted screening should be a viable option to pursue, especially given that there are clear actions to take once a newborn fails the initial hearing test. Outside of screening, education and awareness for CMV should be considered as a low-cost 'win' in order to combat this important issue.

3. I do not recommend the Board add cCMV to the list of conditions for which all Washington-born newborns must be screened.	*No votes or comments received.*
4. At this time, I do not recommend the Board add cCMV to the list of conditions for which all Washington-born newborns must be screened; I recommend the Board revisit cCMV screening at a future date.	• Once the technology allows for better sensitivity in blood spot testing, or urine screening becomes a viable option, the Board should revisit this topic. • The Board should continue to follow the data on the benefit of antiviral treatment for children identified with cCMV. • Recommend getting more data from states that have implemented the targeted program and take some of their learnings as well as more studies that are published. • Would support a universal screening option where positive results indicated more close monitoring of speech and language development in a primary care setting, and referral to audiologist would be reserved for those where concerns were present. • Recommend revisiting cCMV when it is included in the RUSP. • Highlighting the need for more awareness and resources on the early childhood detection of hearing loss, as well as the need for more research and advocacy for the prevention of cCMV. • Some concerns that were raised about impact on learning potential and education may be more reflective of other fractured systems; daycares and schools need to be involved for late onset hearing loss. • Need to discuss the availability of prenatal testing, OBGYN education, more training and availability for pediatric audiologists, and vaccination efforts.

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Placeholder for Materials

Materials for Agenda Item 8 -

Parent Perspective will be added soon.

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Newborn Screening Technical Advisory Committee (TAC)

Placeholder for Materials

Materials for Agenda Item 9 -
cCMV: Natural History, Diagnostic Testing,
and Treatment will be added soon.

Washington State Department of HEALTH

AVAILABLE SCREENING TECHNOLOGY FOR CONGENITAL CYTOMEGALOVIRUS (cCMV)

Megan McCrillis, MPH

Policy Analyst, WA State Newborn Screening Program

Does cCMV meet the "Available Screening Technology" criterion for inclusion on the WA State Newborn Screening Panel?

Available Screening Technology Criterion

- Sensitive, specific and timely tests are available that can be adapted to mass screening.

Sensitive, specific and timely tests are available that can be **adapted to mass screening.**

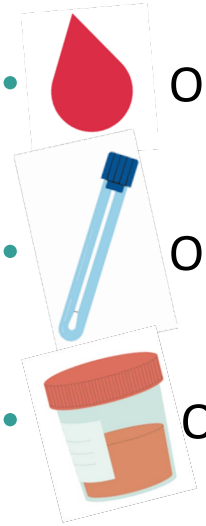
- → High-throughput test available that uses real-time polymerase chain reaction (RT-PCR) to detect the cytomegalovirus in the newborn screening specimen

Sensitive, specific and **timely** tests are available that can be adapted to mass screening.

- → Screening results for cCMV would be available within one or two days of specimen receipt
- → A diagnosis of cCMV should be made by 21 days of life to confirm the infection is congenital and not acquired
- → A RT-PCR screening test for cCMV is timely enough to intervene before 21 days of age

Sensitive, specific and timely tests are available that can be adapted to mass screening.

→ Sensitivity and specificity have been reported for several biological specimen types used to screen newborns for cCMV



- Option 1: Dried Blood Spot
- Option 2: Saliva Swab
- Option 3: Dried Urine Filter Paper

Option 1: Dried Blood Spot

Pros:

- Already the standard biological sample collected for metabolic newborn screening
- Use existing infrastructure for specimen collection and laboratory processing
- High specificity, nearing 100% (few false positives)

Cons:

- Observed sensitivity is low – About 76.8%

Option 1: Dried Blood Spot

Universal screening taking place:

- Minnesota (2023)
- Connecticut (2025)
- NY pilot study (ongoing)
- 4 Canadian provinces

Option 2: Saliva Swab

Pros:

- Convenient to collect, store, and transport
  - "moistening a saliva swab for 30-60 seconds in newborn's cheek at delivery (before first breastfeeding) or at least two hours after breastfeeding"
  - "Swabs were placed in between the cheek and jaw and rotated for 5 seconds on each side then placed in tubes for transport to an area where swabs were air dried for 1 hour and stored at room temperature until transport "

Cons:

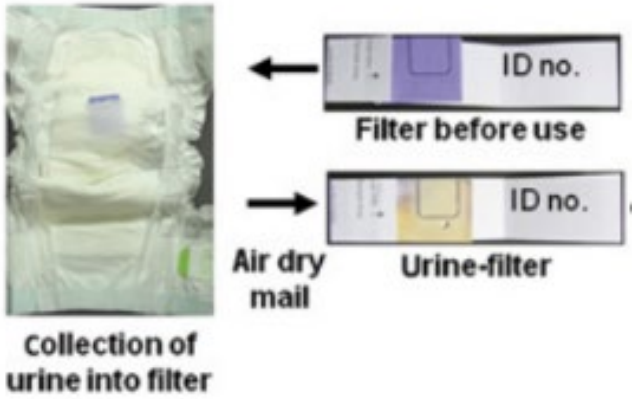
- Lack of infrastructure for:
  - specimen collection & transit from hospitals and midwives
  - accessioning and testing of a different sample type at the lab
- Lowered specificity due to interference from breastfeeding
  - Estimates suggest 8-15% of positive results could be false positives from CMV in breast milk
- Timing of specimen collection
  - Need to collect 1-2 hour after breastfeeding

Sensitivity/Specificity: Estimate 92.9%/>91%

Option 3: Dried Urine Filter Paper

Pros:

- High sensitivity and specificity estimates
  - 98.8 – 100% / >99%



Cons:

- More difficult to collect
  - "We collected urine on…filter cards inserted into the diaper of each newborn. After removal from the diaper, the urine filter was dried and mailed."
  - "The absorbent pad is deposited in the baby's diaper. When the absorbent pad is completely soaked with urine, the filter paper is pressed against it until saturation and left to dry on a clean counter. The dried urine specimen is then sent by regular mail"
- Rarely, minor skin irritations were reported, but in one study, two premature infants had severe skin erosions
  - likely due to placing filter paper directly against skin or leaving diaper unchanged for excessive period
- Lack of infrastructure for:
  - specimen collection & transit from hospitals and midwives
  - accessioning and testing of a different sample type at the lab
    - "We found that the urine-filter collection was not a burden, either on the newborns or on clinic/hospital staffs, once they understood the work flow, although it required extra labour in comparison with DBS-based screening"

Other Considerations

- In sets of twins, if one twin is diagnosed with cCMV, the other twin should also receive diagnostic testing before 21 days of life, even if the newborn screen was negative
- False negative screening results have been reported in sets of twins

Estimated Screening Performance for cCMV Based on Biological Specimen Type			
Specimen type	Sensitivity	Specificity	False positive
Dried Blood Spot	76.8%	>99%	2.3% of screen positive results
Saliva Swab	92.9%	>91%	13.3% of screen positive results
Dried Urine Filter Paper	98.8-100.0%	>99%	Very few

Questions?

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Placeholder for Materials

Materials for Agenda Item 11 -
Overview: Early Hearing Detection, Diagnosis, and Intervention (EHDDI) Program
will be added soon.

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Newborn Screening Technical Advisory Committee (TAC)

Placeholder for Materials

Materials for Agenda Item 12 -
Available Audiological Resources and Access
will be added soon.