Wisconsin Department of Health Services
Wisconsin Division of Public Health
Newborn Screening Program
CF Subcommittee Meeting
Friday, September26, 2025
10:00 a.m. - 12:00 p.m.
Zoom: https://dhswi.zoomgov.com/j/1618110562?pwd=4bDUfGlzsOvugqEGX32XPMbw5VZcqU.1

Meeting ID: 161 811 0562
Or call by location +1 669 254 5252 or +1 646 828 7666 or +1 551 285 1373 or +1 669 216 1590

Meeting Minutes

Meeting Invitees:

X	Dr. Nicholas Antos	X	Tami Horzewski		Jaclyn Schlang
X	Dr. Mei Baker	X	Dr. Evan Kemp		Erin Seffrood
X	Dr. Lisa Burns	X	Kristin Kruschel	X	Dr. Robert Steiner
X	Anna Cisler		Olivia Lampone		Becky Steinmetz
	Ellen Compton	X	Alison LaPean-Kirschner		Tammy Summers
	Kendall Davis	X	Dr. Hara Levy		Tammi Timmler
	Dr. Rebecca Gerhmann		Michelle McDonagh	X	Laney Vernon
X	Sumedha Ghate	X	Peggy Modaff		Mary Marcus Walters
	Rachael Haupt-Harrington	X	Darci Pfeil		
Guests:					
X	Leah Ricci	X	Sharon Luu	X	Silvia Juliana Gelvis (Pulmonary Fellow)
X	Kelly Gill	X	Travis Henry		

Meeting Minutes:

Friday, September 26, 10:00 AM – 12:00 PM				
Time:	**Topic:**	**Lead:**	**Follow-up Items:**	**Notes:**
10:00-10:05	**Welcome and Review of Minutes**	Dr. Antos		Announcements: • Dr. Christina Barreda has left UW and moved to Texas. • Dr. Baker will not be able to attend the National CF Conference. Meeting minutes from Nov 19, 2024, were approved. • Motion to approve: Darcy Pfeil

				• Second: Dr. Baker • No objections.
10:05–10:15	**Department of Health Services (DHS)/ WI State Lab of Hygiene (WSLH) Updates**	Tami Horzewski/ Dr. Baker		DHS Updates: • DHS is moving office buildings. New building is GEF 1. • X-linked adrenoleukodystrophy (X-ALD) and Mucopolysaccharidosis Type I (MPSI) were added by rule to the NBS panel on August 1 as well as a change in the NBS blood card fee. • Guanidinoacetate Methyltransferase (GAMT) Deficiency was approved by the Secretary and will soon move through the rule making process. • Mucopolysaccharidosis Type II (MPSII) and Infantile Krabbe Disease (with psychosine at or above 10 nmol/L) were recommended by the SACNBS to be added to the NBS panel. The committee will draft a report for the Secretary for review and if approved the two conditions will also move through the rule making process. • Leah Eckstein. left her role as the Family Health Section manager. Elizabeth Seeliger left role as Wisconsin Sound Beginnings Director. • The Advisory Committee on Heritable Disorders in Newborns and Children (sets the RUSP) was disbanded with unclear future plans. It is public information that the American College of Medical Genetics and Genomics

				(ACMG) is working to potentially take on the role of updating the RUSP in interim period. • There is a bill proposed for WI to follow the RUSP currently going through the legislature. • The WI Newborn Screening Research workgroup finalized their proposed research guidelines after 2 years of work. It is currently under review by the Secretary. Ongoing meetings continue between WSLH and DHS. • Publications ○ A Gov D communication was sent out on 8/28/25 to announce new WI NBS educational materials. They are available to order for free on the DHS website. ○ The updated materials include 4 one-pagers (three screens, hearing, heart and blood screening), a Plain community NBS booklet, and a hearing screening postcard. ○ Pull-down posters and swag are available to use at upcoming tabling events. General: • This committee was renamed to the CF Subcommittee as there is now a separate SMA screening subcommittee.

				WSLH Updates: • An Association of Public Health Laboratories (APHL) fellow, Travis Henry, started in July. He has 10 years of experience working in another state's newborn screening program. • Started routine newborn screening for X-ALD and MPS 1 on August 1, 2025. WSLH had been screening for X-ALD through a pilot project. There have been no confirmed cases of MPS1 so far. • WSLH had a successful CAP inspection.
10:15-10:30	**2024 Screening Report**	Sharon Luu		CF Screening 2024 Summary: • Specimens tested: 67,283 • Infants screened: 58,949 • NGS CFTR Variant Assays Completed: 3,039 ○ Confirmed CF: 12 ○ Confirmed CRMS/CFSPID: 9 ○ Confirmed Carrier: 166 ○ Ultrahigh IRT w/o variant detected: 98 ○ Case closed (one identified variant): 22 ○ Pending (one identified variant): 1 ○ Case closed (two identified variants): 1 ▪ Infant is on palliative care due to Trisomy 13. General Statistics:

				• Screen positive (two CF-causing variants): 12, PPV(CF) = 100% • Screen positive (two variants w/ R117H): 3, PPV(CF) = 0%, PPV(CRMS) = 100% • Screen positive (1 variant): 195, PPV(CF) = 0%, PPV(CRMS) = 3% • Overall PPV (CF) = 6% • Birth prevalence: 1 in 4,912
10:30 – 10:45	**CF screening short-term follow-up**	Dr. Baker		This item was tabled for future discussion.
10:45 – 11:00	**CFTR variant panel update**	Dr. Baker		WSLH currently uses a panel with 689 variants developed based on V14 of the CFTR2 variant list. This version includes 719 CF-causing variants. The latest version is V15, which includes 1,085 CF-causing variants. Soon V16 will be released, which will include an additional 120 variants. Proposal Passed: Increase variant list to the 978+R117H developed based on V15 of the CFTR2 variant list. • Motion to approve: Dr. Hara Levy. • Second: Anna Cisler • No objections.

11:00 – 11:05	**CF NBS learning Collaborative** https://www.cphinnovation.org/cf	Dr. Antos		Updated CF NBS Guidelines: • Overview: Goals ○ Increase/assure detection in all populations ○ Decrease overmedicalization of those without CF ○ Guidelines for consideration, with the understanding that following the latest guidelines may not currently be feasible in many states • Significant changes ○ Embraces sequencing ○ Allows for NOT sweat testing children with only 1 CF variant Wisconsin already meets the CF NBS Guideline Recommendations listed below. • IRT ○ The CFF recommends the use of a floating IRT cutoff over a fixed IRT cutoff. ○ The CFF recommends using a very high IRT referral strategy in CF newborn screening programs whose variant panel does not include all CF-causing variants in CFTR2 or does not have a variant panel that achieves at least 95% sensitivity in all ancestral groups within the state.

				• CFTR Variant Testing ○ The CFF recommends that CF newborn screening algorithms should not limit CFTR variant detection to the F508del variant, or only variants included in the ACMG-23 panel. ○ The CFF recommends that CF newborn screening programs screen for all CF-causing CFTR variants in CFTR2. ○ The CFF recommends conducting CFTR variant screening twice weekly or more frequently as resources allow. • CFTR Sequencing ○ The CFF recommends the inclusion of a CFTR sequencing tier following IRT and CFTR variant panel testing to improve the specificity and positive predictive value of CF newborn screening. • Communication ○ The CFF recommends that both the PCP and CF specialist be notified of abnormal (positive) newborn screening results.
11:05-11:20	Discussion of the new CFF NBS guidelines and next steps https://www.cff.org/medical-professionals/newborn-	Dr. Antos	Dr. Baker to pull data from the past 5 years for babies with R117H and an elevated sweat test.	See flowchart and reference statistics from Dr. Antos on page 11. • Currently, WI is following the option on the left-side of the flow chart.

	screening-clinical-care-guidelines			• Discussed pros and cons of the middle and right-side options. ○ Reducing number of sweat tests done may make it hard for some hospitals to continue offering this testing. Training on proper performance of sweat tests may also become limited if the number of sweat tests done is reduced. ○ Subcommittee expressed concern about the middle option not catching patients who convert from CRMS to CF. ○ Discussed potential of identifying more CRMS cases. Need to consider the trade-off of identifying less carriers but picking up more CRMS cases. ○ Discussed whether this change would make it more likely for children from a certain ethnicity to be missed. Need to consider the equity perspective. ○ Consider reaching out to other states to see if they have faced issues with being able to get sweat tests done when fewer children need them. ○ Need to consider if there will still be resources for counseling and follow up available to families who have a child with 1 variant. Many

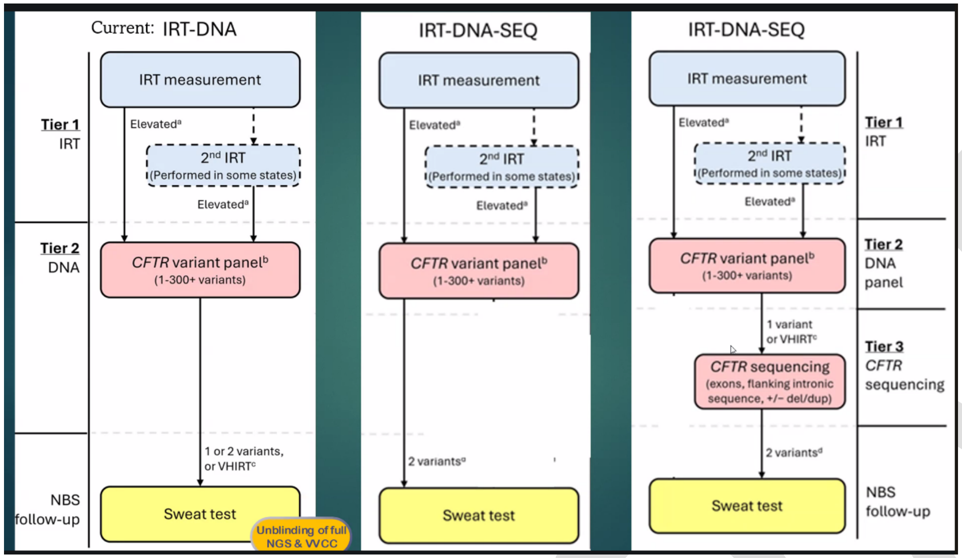
				families in this situation still find it helpful to talk with a genetic counselor. ○ Discussed potential options for reporting 1 variant: ▪ “Screen carrier” would listed on the NBS report for those with 1 variant ▪ PCP would receive this report, but CF specialist would not be contacted by the lab. ▪ Need to consider how reporting would work for babies with an IRT over 170. • Subcommittee would like to continue discussion at the next meeting. ○ Variants to include/exclude
11:20– 11:35	**Status of CF Carrier Fact Sheet** **Current Educational Resources Used** https://www.cff.org/intro-cf/carrier-testing-cystic-fibrosis	Tami/All	Dr. Levy to review the current DHS CF Carrier Fact Sheet and send around to the subcommittee for review. Could potentially work with the DHS communications team to match the NBS educational material style.	The current CF Carrier Fact Sheet developed by DHS is not used frequently. Decided to review and make updates. This can serve as a placeholder until a decision is made about sweat test changes. If sweat testing for 1 variant is removed, need to make this information available in the fact sheet. Discussed also linking the CF Carrier Testing website from CFF on the WSLH page. https://www.cff.org/intro-cf/carrier-testing-cystic-fibrosis The state CF Genetic Counselor list was updated.

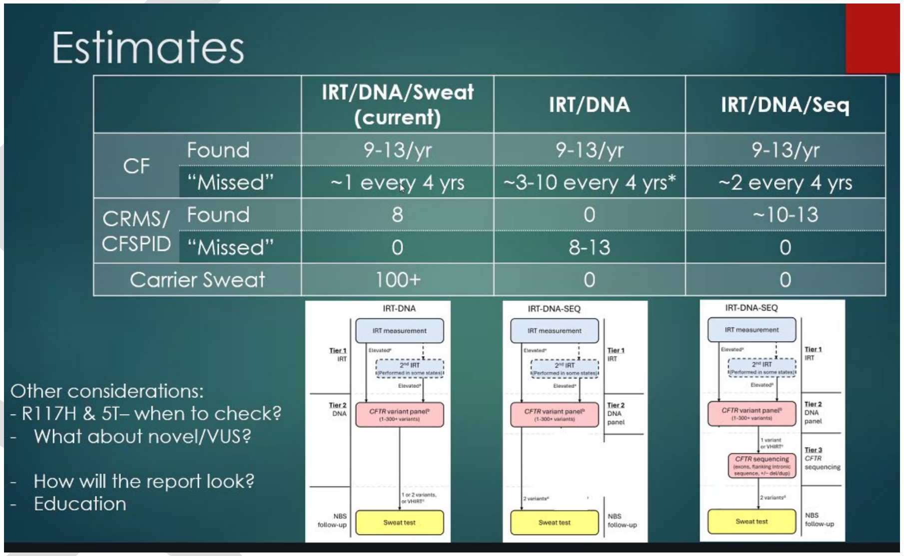
			Tami Horzewski to send the CF Genetic Counselor handout to the subcommittee.	
11:35-11:45	**Review & Confirm Subcommittee Membership /Voting Members & GC List Appt renewals, parent rep**	Tami/All	Tami Horzewski to follow up with potential new members. Sumedha Ghate to touch base with Dr. Rebecca Gehrmann about attending this subcommittee. Dr. Antos to send Tami Horzewski potential parent rep information.	Subcommittee membership updates: • Dr. Joshua Freedman stepped down • Tammi Timmler stepped down • Laney Vernon. will be replacing Darcy Pfeil • Plan to invite Kristin Kruschel to participate as a guest or as the RN/NP for Children's WI. • Vacant parent rep o Dr. Antos spoke with a parent and will send their information to Tami H. • Plan to have Jaclyn Schlang fill Gunderson's vacant genetic counselor spot. • Plan to have Kendall Davis fill Children's WI dietitian spot. • Plan to have Ellen Compton fill Gundersen's dietitian spot. Discussed whether MD clinical consultants from every CF center should be CF NBS subcommittee members. Plan to continue this discussion at the next meeting.

11:45	**Plan Next Meeting/Agenda Items**	Dr. Antos/All	Tami Horzewski to send out a doodle poll to schedule the next subcommittee meeting in about 6 months.	Next Meeting Agenda Items: • Dr. Baker to report on data from the past 5 years for babies with R117H and an elevated sweat test. • Potential report out from the CF Conference scheduled for October 2025. • Discuss expectations for MD clinical consultants with regard to membership on the CF Subcommittee.

Next meeting date: TBD

“Parking Lot” Items:





Estimates

		IRT/DNA/Sweat (current)	IRT/DNA	IRT/DNA/Seq
CF	Found	9-13/yr	9-13/yr	9-13/yr
	"Missed"	~1 every 4 yrs	~3-10 every 4 yrs*	~2 every 4 yrs
CRMS/ CFSPID	Found	8	0	~10-13
	"Missed"	0	8-13	0
Carrier Sweat		100+	0	0

Other considerations:
- R117H & 5T– when to check?
- What about novel/VUS?

- How will the report look?
- Education