Wisconsin Department of Health Services
Wisconsin Division of Public Health
Newborn Screening Program
CF/Molecular Subcommittee Meeting
Friday, April 13th, 2018
1:00-3:00 pm
WI State Laboratory of Hygiene – 2601 Agriculture Drive
Minutes

Meeting Invitees:

X	Dr. Nicholas Antos		Kevin Josephson	X	Dr. Mike Rock
X	Dr. Mei Baker	X	Dr. Gary Kirk	X	Erin Sefrood
	Nicole Brueck	X	Alison LaPean-Kirschner	X	Tammi Timmler
X	Anna Cisler	X	Mary Marcus	X	Dr. Meredith Schultz
	Erin Cronn		Dr. Laura McCauley	X	Dr. Matthew Harmelink
X	Sumehda Ghate	X	Tami Miller		
	Dr. Peter Holzwarth	X	Peggy Modaff		Students:
X	Tami Horzewski		Julie Noe	X	Krista Kerlinske
		X	Darci Pfeil	X	Emily Schumacher

Agenda:

Friday, April 13th, 1:00PM – 3:00 PM WSLH - Ag Drive				
Time:	Topic:	**Lead:**	Follow-up Items:	Notes:
1:00 PM – 3:00 PM	**Welcome and Review of Minutes**	Dr. Rock		Motion to approve April 7, 2017 minutes 1st Motion: Darci Pfeil 2nd Motion: Mary Marcus

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	DHS Updates • **Spinal Muscular Atrophy (SMA) newborn screening to join CF Subcommittee** • **Changes to Birth Defects Registry Reporting System** • **Update on Special Dietary Treatment Expansion (over age 21)**	Dr. Kirk		Department of Health Services (DHS) Updates: Two conditions, Carnitine Palmitoyltransferase IA (CPT IA) deficiency and Spinal Muscular Atrophy (SMA) were nominated for addition to the WI Newborn screening panel of conditions. Both were reviewed through the Newborn Screening Program nomination process and recommended by the Secretary's Advisory Committee on Newborn Screening (SACNBS). The Secretary has approved the addition of CPT IA and the SACNBS chair is currently working on the SMA report to submit to the Secretary for her review. With SMA testing being discussed, the CF subcommittee has been asked to include SMA in their subcommittee. Dr. Kirk shared some background on the Birth Defects Registry. With the change in the Birth Defect Registry in September, 2017 to an opt out process, it allows for easier access to add conditions to the registry. New conditions can now be added by the Secretary of the Department of Health Services without going through the rule making process. The change will also allow for the collection of identifiers as the registry will be able to communicate with other registries and increase the ability to conduct surveillance and establish causation. This change will also allow for involvement in other projects as previously the data has not been

				robust enough to be eligible for federal grants. The current list is the original list from 2004. The registry council is currently working on determining the criteria for the review of conditions and the development of a process for this review. All subcommittees have been asked to do a review of the current registry list to determine if there may be conditions that they feel should be included on the list. The proposed conditions will need to be reviewed by the council using the criteria to be determined. There is no established timeline for the review and proposed addition of conditions. Individuals can propose conditions to be added as well as subcommittees. The two year pilot to determine whether the State can fund Special Dietary Treatment (SDT) for those over age 21 with CF concluded in January 2018. An additional $60k/year was set aside to provide SDT for those over age 21, and that amount was not surpassed, so the plan is continue this forward with this population, as we do in other populations in which SDT is provided.

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	CAP compliance for CF screening reports	Dr. Baker		The CAP requires that gene variants be reported using HGVS nomenclature and including the HUGO Gene Nomenclature Committee (HGNC) gene name, and a standard version reference identifier to the transcript/protein. The group discussed the strategies of making NBS reports more "user friendly", but is also CAP compliant. The lab will look into the possibility of using "table image" method.
	2017 CF Screening Summary Report	Dr. Baker		See the attached presentation.
	NGS data re-analysis on sweat testing abnormal cases	Dr. Baker		In 2017, there were two CF cases with the second *CFTR* mutation identified after the NGS data re-analysis (panel restriction removed) prompted by the abnormal sweat testing results. The committee members agreed that this practice is beneficial, and should become routine. The lab will perform NGS data re-analysis on all cases with sweat test result greater than 30, and inform the associated CF center.

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	Parental mutation analysis: question to include 5T analysis	Dr. Baker		CFTR 5T is currently in the MVCC group. The committee members actively discussed the pros and cons regarding including it in the parental CFTR mutation analysis. The members decided to continue this discussion at the Wisconsin CF Consortium meeting held on April 20, 2018.
	Status on CF Quantum Sweat Test Study	Dr. Rock		The first study of the CFQT was published in the Sept. 2014 issue of the Journal of CF. There was a patch lot at one center that had more scatter in the results. A second study commenced in 2017 after a long delay due to the manufacturer attempting to obtain FDA approval based on the results of the first study. Although the manufacturing changed their process of making the patches, there is still unacceptable scatter of the results. Just prior to the 2017 NACFC, the study was halted due to futility. A poster was presented at that conference. The funding agency, the Legacy of Angels Foundation, had previously granted a one year no cost extension. They would not grant a second one year extension for 2018 and their funding was withdrawn. A grant has been submitted to the CFF for continuing this study. The decision on this grant will be in late May 2018.
	Plan Next Meeting/Agenda Items	All		

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Next meeting date: Friday, November 9, 2018

"Parking Lot" Items: