



Wisconsin Department of Health Services
Wisconsin Division of Public Health
Newborn Screening Program
CF/Molecular Subcommittee Meeting
Friday, November 15, 2019
1:00-3:00 pm

WI State Laboratory of Hygiene – 2601 Agriculture Drive
Minutes

Meeting Invitees:

X	Dr. Nicholas Antos		Kevin Josephson		Darci Pfeil
X	Dr. Mei Baker	X	Alison LaPean-Kirschner	X	Dr. Mike Rock
	Nicole Brueck	X	Dr. Jennifer Kwon		Erin Seffrood
X	Anna Cisler		Mary Marcus		Tammi Timmler
X	Sumehda Ghate		Tami Miller		
	Dr. Matthew Harmelink	X	Peggy Modaff		
	Dr. Peter Holzwarth		Dr. Julie Noe		Students:
X	Tami Horzewski				

Agenda:

Friday, November 15, 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. Antos		<p>Motion to approve April 1, 2019 minutes: 1st Motion: Dr. Kwon 2nd Motion: Sumehda Ghate Motion approved.</p> <p>Multiple presentations were given in the past year on our Next Gen Sequencing experience, including Drs. Rock & Baker presented at the European CF Conference and Drs. Antos & Baker at NACFC</p>
	DHS Updates	Tami Horzewski		<p>The following DHS updates were shared:</p> <ul style="list-style-type: none"> Spinal Muscular Atrophy (SMA) and Carnitine Palmitoyltransferase IA



Wisconsin Department of Health Services
Wisconsin Division of Public Health
Newborn Screening Program
CF/Molecular Subcommittee Meeting
Friday, November 15, 2019

1:00-3:00 pm

WI State Laboratory of Hygiene – 2601 Agriculture Drive
Minutes

				<p>(CPT IA) deficiency were approved by emergency rule on October 15, 2019 and are moving through the process to be put into permanent rule to become a part of the Newborn Screening (NBS) panel.</p> <ul style="list-style-type: none">• Pompe Disease has been nominated for addition to the NBS panel of conditions and will be considered at the upcoming Umbrella Committee meeting on December 6th. The Pompe Disease nomination was reviewed by the metabolic subcommittee at their last meeting and was recommended for addition to the panel.• Dr. Gary Kirk is resigning as the Chief Medical Officer at the Department of Health Services and his last day will be December 13th.



Wisconsin Department of Health Services
Wisconsin Division of Public Health
Newborn Screening Program
CF/Molecular Subcommittee Meeting
Friday, November 15, 2019

1:00-3:00 pm

WI State Laboratory of Hygiene – 2601 Agriculture Drive
Minutes

	2018 Screening Summary	Dr. Baker		<div>Cystic fibrosis screening 2018:</div> <table><tr><td>Screened Infants Born in 2018</td><td>63,524</td></tr><tr><td>Reported Screening Positive w/ Two Variants</td><td>46</td></tr><tr><td>Reported Screening Positive w/ one Variant</td><td>173</td></tr><tr><td>Confirmed CF Cases</td><td>15</td></tr><tr><td>PPV</td><td>6.8%</td></tr><tr><td>Incidence</td><td>1 in 4,235</td></tr></table> <div>More detailed information has been included in attached slides.</div>	Screened Infants Born in 2018	63,524	Reported Screening Positive w/ Two Variants	46	Reported Screening Positive w/ one Variant	173	Confirmed CF Cases	15	PPV	6.8%	Incidence	1 in 4,235
Screened Infants Born in 2018	63,524															
Reported Screening Positive w/ Two Variants	46															
Reported Screening Positive w/ one Variant	173															
Confirmed CF Cases	15															
PPV	6.8%															
Incidence	1 in 4,235															
	Request for Parental <i>CFTR</i> NGS Assay	Dr. Baker		Dr. Baker reminded the committee members that the time frame for requesting parental <i>CFTR</i> NGS assay is within 6 months of the screening positive report.												
	Request for NGS Data Re-Analysis	Dr. Baker		The committee members agreed that genetic counselors can request <i>CFTR</i> NGS assay data re-analysis on behalf of clinicians, and the lab report will reflect such practice accordingly.												
	SMA NBS Implementation	Dr. Baker		The NBS lab at WSLH implemented SMA screening on 10/15/2019. As of 11/14/2019, 5,870 infants have been screened for SMA.												



Wisconsin Department of Health Services
Wisconsin Division of Public Health
Newborn Screening Program
CF/Molecular Subcommittee Meeting
Friday, November 15, 2019
1:00-3:00 pm

WI State Laboratory of Hygiene – 2601 Agriculture Drive
Minutes

				One screening positive case was reported, and later confirmed as a true positive case.
	MVCC Screening Discussion, including 5T •Discuss the current state of MVCC screening, processes, concerns, findings, and options	Dr. Antos/Alison LaPean-Kirschner/ Dr. Baker/ Sumedha Ghatge		<p>The process for screening for MVCC was discussed at length. Concerns included specifically the mutation 5T 11TG and compound heterozygotes with a normal sweat test. Multiple options for screening were discussed, as in the attached images.</p> <p>It was agreed that the process would change to checking MVCC only after a borderline/abnormal sweat test, as part of the unblinding. It was agreed that we will update our CF-causing variant panel based on the version of “11March2019” posted on the CFTR2 website. Current state and proposed state are pictured below and in the full slides. The feasibility and appropriateness of this was agreed on by clinical, genetic, and laboratory team members.</p> <p>1st motion: Dr. Antos 2nd motion: Alison LaPean-Kirschner</p> <p>Dr. Baker proposed start date – January 1, 2020</p>



Wisconsin Department of Health Services
 Wisconsin Division of Public Health
 Newborn Screening Program
 CF/Molecular Subcommittee Meeting
 Friday, November 15, 2019
 1:00-3:00 pm

WI State Laboratory of Hygiene – 2601 Agriculture Drive
 Minutes

				Discussed and approved protocol for the patients already identified. In summary, we will continue to follow ~yearly until about 5 years old and continue to reassess the process. Full protocol will be sent to CF Teams.
	Plan Next Meeting/Agenda Items	All		Tami will work with Dr. Antos and Dr. Baker to determine a potential date for the next meeting in April, 2020 and then send out the meeting date options to the subcommittee.

