Wisconsin Department of Health Services
Wisconsin Division of Public Health
Newborn Screening Program
Umbrella Committee Meeting
Friday, December 6, 2019
10:00am-2:00pm
WI State Laboratory of Hygiene – 2601 Agriculture Drive, Madison
Minutes

Meeting Invitees:

x	Dr. Nick Antos	x	Dr. John Hokanson	x	Marilyn Noll (phone)
x	Dr. Mei Baker		Tami Horzewski		Dr. Pilar Ossorio
x	Dr. Jeff Britton	x	Dr. Julie Kessel	x	Dr. Greg Rice
	Dr. Patricia Donohoue	x	Dr. Gary Kirk	x	Dr. Paul Scott (phone)
x	Dr. Norman Fost	x	Alison LaPean-Kirschner	x	Kelsey Stevenson
x	Dr. Patrice Held		Mary Marcus		Angie Thompson
		x	Dr. Michelle Miller		Ann Zenk
Meeting Guests					
x	Dr. Anne Odusanya	x	Dr. Jennifer Kwon		Dr. Priya Kishnani
x	Dr. Michael Rock	x	Dr. Sharon Fleischfresser	x	Kiri Sunde (Medical resident working with Dr. Fost)
x	Clubb Family (Mom, Dad, 2 sons, daughter Gwen)	x	Genevieve Fashay, son Atlas, Sarah (RN)	x	Audrey Prevee
x	Chuck Warzecha	X	Stephanie Austin, (Genetic Counselor who works with Dr. Kishnani)		

Agenda:

Friday, December 6, 2019 10:00 AM – 2:00 PM				
Time:	**Topic:**	**Lead:**	**Follow-up Items:**	**Notes:**
10:00-2:00	Welcome, Introductions, and Review of Minutes	Dr. Kirk		Motion to approve May 3, 2019 minutes 1st Motion: Dr. Britton 2nd Motion: Dr. Baker Motion approved.

	Pompe Nomination Review	Dr. Kirk		**Family testimony:** Clubb family provided testimony about their experience with their daughter Gwen's diagnosis of Pompe during the Pompe pilot, as well as their son's diagnosis following detection through the pilot. This, along with the discovery that their eldest child is also affected, has inspired them to nominate the condition. Genevieve Fashay shared the story of her son Atlas' diagnosis, which took months in the absence of newborn screening for Pompe. This is especially important to her, as her son was diagnosed with infantile Pompe, where the timing of diagnosis and initiation of treatment is more urgent. Both families shared that the costs they incur that are not covered by their insurance are significant. $2100/month for COBRA coverage for Genevieve's plan, plus out of pocket costs. There was discussion between Dr. Kessel, Dr. Rock, and the families regarding improving how the diagnosis is shared with the family, and the importance of making a distinction between late-onset and infant-onset. They also discussed the value in reaching out to other affected families via Facebook. **Deliberation:** Dr. Rice stated that the Metabolic Subcommittee unanimously agreed that Pompe met the first criterion laid out for adding a condition. Stephanie Austin added that the spectrum of infantile or late onset is truly a spectrum – 50% of children diagnosed with late-onset Pompe actually experience symptoms as children. Dr. Rice stated that he changed his mind about informing parents' of late onset disease, as from a screening perspective he believes it's morally wrong for us to screen, then diagnose, and not share the results of that diagnosis with families. Dr. Fost asked how many states are screening – 19 other states are screening for Pompe. Dr. Hokanson asked about prevention being part of criterion 1. Dr. Fost stated that "condition" can be used to refer to symptoms, rather than the disease itself. Dr. Rice advocated better patient education prior to testing. Stephanie Austin discussed improved

				treatment and upcoming second generation therapies, including gene therapy for adults. While residual muscle weakness is still seen, these new therapies are expected to improve outcomes. Dr. Rice stated that the Metabolic Subcommittee also unanimously agreed that Pompe met the second criterion. There is ample information available about the disorder, particularly about the common variant seen in Wisconsin. Dr. Rice stated that there is an FDA-approved treatment for Pompe, so it meets the third criterion. He made a correction to stating a medication was "freely available" – it is not free, but is in fact quite expensive. He corrected this to say the medication is "readily available." Dr. Fost raised the issue of a high psychosocial cost to screening, particularly those for with late-onset Pompe. Stephanie Austin brought up improved ability to measure immune status and tailor treatment accordingly. Dr. Rice defined cross-reactive immunological material (CRIM) status. Dr. Fost questioned the degree to which Pompe meets the fourth criterion given the expense of not just the medication, but also the ancillary care. Dr. Rice states this is especially relevant for late onset families. Alison LaPean-Kirschner said we currently have other conditions on the panel that mirror a similar course of screening for asymptomatic cases. Regarding the fifth criterion, Dr. Rice stated that there were no false positives in the pilot, and Dr. Baker stated that no carriers would be detected because of the level WSLH uses for a cut-off. Dr. Fost and Dr. Rice discussed psychosocial support and its accessibility. For the sixth criterion, Dr. Baker and Dr. Rice reported that there is good specificity and sensitivity for the test. Little discussion occurred around criterion seven other than confirming no new sample collection system would be required.

				For the eighth criterion, Dr. Britton brought up the importance of providing primary care providers with a script to use when discussing results with the family. Illinois, Minnesota, and Michigan screen for Pompe, but Iowa does not. Dr. Kessel asked for clarification on whether genetic testing was completed before the call is made to the physician. Dr. Baker and Dr. Rice confirmed that this is the case. For the ninth criterion, Dr. Kirk discussed the cost added to the blood card fee for Pompe screening would be approximately $10. Dr. Fost also brought up the cost of treatment. Dr. Rice clarified that infantile cases will be treated regardless of whether they are identified by screening, or by clinical presentation. Mr. Clubb asked for clarification about who pays for the blood card, and Dr. Britton and Dr. Baker clarified that hospitals buy the cards. Typically hospitals charge a bundled fee for labor and delivery, and insurance companies reimburse at whatever rate they have negotiated with the hospital. Dr. Rice reported that this was the only criterion that the Metabolic Subcommittee did not unanimously agree had been met. Questions were raised about when screening would “go live” following a nomination. Dr. Kirk shared the SMA timeline, and estimates that it would take roughly two years until we were screening again. Dr. Kirk reviewed the criteria to determine whether anyone felt any criterion was not met, or more information was required. Dr. Britton would like more information for criterion 3, and Dr. Britton and Dr. Held both would like more information regarding criterion 9. Dr. Held would also like to make note of the fact we do not have a Wisconsin Hospital Association representative present at today’s meeting. Dr. Britton was concerned with late onset families not qualifying for the follow-up care they will require. Dr. Rice states that the cost will vary between families. Dr. Kessel would like to know if there are clear criteria regarding cardiology (and other) screening late onset families. Stephanie Austin states that after one echocardiogram, unless there were significant findings, they don’t recommend future follow up echocardiograms. Dr. Rice reported the importance of monitoring of late onset.

				Dr. Britton moved that we recommend the addition of Pompe screening to the panel. Dr. Hokanson seconded this motion, and it passed unanimously.
	Newborn Screening (NBS) Program Updates: • Department of Health Services (DHS) Updates • WI State Lab of Hygiene (WSLH) Updates - Update Spinal Muscular Atrophy (SMA)	Dr. Kirk Dr. Baker		Dr. Kirk suggested the formation of a task force or subcommittee focused on finances in order to prevent us from regularly having to revisit funding increases for the blood card every 5 to 10 years. Dr. Held requested clarification on the scope of this, which is yet to be determined. Dr. Rice made a motion to support developing a finance subcommittee. Dr. Britton and Dr. Held seconded this. All in favor, passed unanimously. **DHS updates:** SMA and CPT1 were added by emergency order October 15, and should be permanent as of January 1, 2020. Dr. Kirk shared his thanks to all of those that worked on this, and shared he will be resigning, with Sharon Fleischfresser filling in as the chair of the Umbrella Committee until a replacement is found. Dr. Fost led the committee in thanking Dr. Kirk for his tenure as chair. **WSLH updates:** Dr. Baker presented on SMA types and clinical classifications, Wisconsin's screening algorithm, and the current status of SMA screening. Just fewer than 10,000 babies have been screened since October 15, and two confirmed cases of SMA have been detected.
	Working Lunch			
	Subcommittee Updates • Hearing Subcommittee – proposed	All Chairs		**Hearing updates:** Dr. Kessel provided the hearing subcommittee update and shared that there is increased attention being paid to CMV screening for children that appear to be symptomatic. Some states are now doing targeted screening children for congenital CMV when they have not passed their newborn hearing screening exam. The question of retaining specimens to re-examine them for congenital CMV came up during the subcommittee meeting. Currently

	motion to extend the storage of blood collection cards from one to five years Secretary's Advisory Committee on Newborn Screening (SACNBS) Update	Dr. Fost		specimens are retained for one year, and there is a desire to retain these for 5 years. Dr. Britton provided some historical context regarding the rationale for retaining the specimens: they did not want to burden the WSLH with storage responsibilities, and Minnesota was sued by a parent to have their specimen destroyed because they did not want it retained indefinitely. There were also questions raised in the past regarding law enforcement compelling WSLH to share their specimens. Dr. Fost also brought up a situation in the past where a researcher had pursued access to WSLH specimens and had nearly gained access before this was discovered and stopped. Dr. Kessel stated that Minnesota is actually one of the states that are serving as a model for how to look back at specimens, and wanted to distinguish between using retained records to answer clinical questions v. research questions. Dr. Baker also described the process for requesting specimens for clinical care purposes, which includes a parental release and provider request. Questions were raised regarding what parents would do differently given the knowledge that their child was diagnosed with congenital CMV. Dr. Kessel suggested that the parents may choose not to do a genetic screening work-up. Dr. Fost believed this is a weak reason, and Dr. Britton would like us to sort of some of the thornier legal issues that surround this before agreeing to this proposed change. Past rationale for retaining the specimens is the ability to examine false-negatives (though the one year cut off does not allow this to occur for cystic fibrosis). Dr. Scott asked for clarification around what actions should be taken today and moving forward, and emphasized the importance of involving a legal opinion on these proposed changes. Dr. Kirk suggested reaching out to other states and developing a document that addresses some of the concerns raised today. Dr. Kessel moved to gather retention policy information from other states (e.g., what legal and ethical obstacles did they have and how did they overcome them, and who can access their NBS cards and for what reasons), get a copy of the actual WSLH policy around NBS card retention, and review old Umbrella Committee meeting minutes to determine if they have relevant information about how the WSLH retention period was determined, to it back to a future Umbrella Committee meeting. Dr. Antos seconded. Motion carried.

				Hemoglobinopathy: Dr. Scott reports reviewing confirmed cases provided by WSLH. Due to shifting demographics in Wisconsin, they are seeing more cases of thalassemia major than previously. The committee suggested sending specimens out for DNA testing when their newborn screening results suggest thalassemia. Dr. Held stated they are currently in the process of developing a way to capture Barts. **Metabolic:** Dr. Rice discussed dropping their cut off in order to better detect cases of homocystinuria. The Pompe nomination and nine criteria were also reviewed at the last meeting and the subcommittee recommended that the condition be added to the WI NBS panel of conditions. **CCHD**: Dr. Hokanson discussed that the AAP 's support of ICD-10 codes for abnormal newborn screening results, including cases in which CCHD screens were failed. These proposed codes would likely not be in use until mid-to-late 2020. Dr. Hokanson also provided data about the improved screening rates, growing out-of-hospital population, and not screened reasons that have been free text entered onto the blood card. **Education**: Allison discussed the development of educational materials for CPT1 and review of ongoing newborn screening materials. They realized that many documents do not have updated review dates, so this is a project they are undertaking. They have also worked to assure that links online actually link to documents and materials they should. Dr. Kessel asked whether they refer families to Baby's First Test. Allison replied that this is part of the process they're currently undergoing in order to assure their information is up-to-date and relevant. **Cystic fibrosis/Molecular:**

				Dr. Antos shared that this marks the third year they have been using NextGen genetic sequencing for cystic fibrosis. One of the things they discovered is an increase in the number of individuals that did not meet the criteria for CF or CFTR but did have cystic fibrosis-causing genes. **Endocrine**: Dr. Baker discussed molecular testing for CAH, though testing is not currently well-established, and creates a lot of carrier identification, so will not be pursued by WSLH at this time. Dr. Held reports that their second-tier test successfully No **Immunodeficiency** Subcommittee report today.
	QA Updates	Dr. Held		Dr. Held shared that their contracted courier changed on August 5 after Gold Cross dropped newborn screening. Now the contracted courier is UPS, using an overnight air service. While they report 6 day/week service, they realized 1/3 of the state does not receive Saturday pick-ups. Currently, WSLH is trying to work with UPS, though their current short-term suggested solution was a $2+/mile charge to reach the places not currently served by the overnight air. Meriter, Columbia St. Mary's, and Ozaukee are currently on an electronic interface exchange. This comprises about 15% of WSLH specimens, and starting in January or February, the electronic interface will be picked up by all of the Aurora facilities. While this reduces errors, it is expensive and time-consuming for IT, and WSLH data entry staff.
	Plan Next Meeting/Agenda Items	All		

Next meeting date: Friday, May 1, 2020

"Parking Lot" Items:

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