

Meeting Members:				
X Dr. Mei Baker		Dr. Sam Huang	X	Lisa Obernolte
X Dr. Donald Basel (Chair)		Dr. Philip James		Stephanie Offord
X Beth Boyd	Х	Jessica Kopesky	X	Matthew Rasberry
X Therese Breunig	Х	Ashley Kuhl	X	C Dr. William Rhead
Anna Cisler		Dr. Jennifer Kwon	X	Dr. Jessica Scott-Schwoerer
X Nicoletta Drilias	Х	Wanda Meeteer		Emily Singh
X Gretchen Heckel	X	Dr. Roberto Mendez	X	C Dr. Bob Steiner
X Sonja Henry	X	Kaegan Mestel	X	C Tammi Timmler
Caitlin Hessenthaler		Emily Meyer	X	Mary Marcus Walters
X Tami Horzewski		Susana Morphis	X	C Dr. Katie Williams
	Х	Dr. Mike Muriello		
Meeting Guests:				
X Amy White	Х	Dr. Julie Thiel	X	C Dr. Justin Hopkin
Jill Beirl		Taylor Duke		Leah Eckstein

Purpose of Meeting:

The Metabolic Subcommittee serves in an advisory capacity to the Umbrella Committee and NBS Program regarding the specific screening-related condition(s) in the following areas:

Expert Knowledge in Condition-Specific Clinical Care and Research

• Advises on condition-specific clinical care including ongoing changes in treatment options for affected individuals, emerging newborn screening technology, and condition-specific research.

Quality Assurance

• Reviews de-identified screening and confirmatory testing data to monitor screening test performance, such as screening positive predictive value, screening false positive rate and false negative rate.

• Recommends changes in NBS practice to the Umbrella Committee and NBS Program.

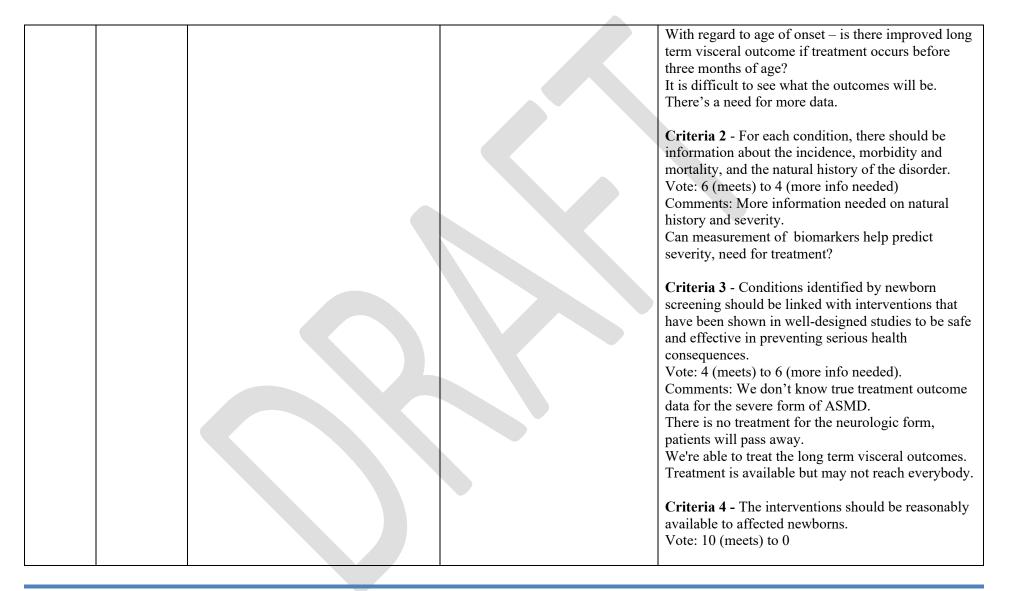
Education for Families and Providers

Reviews and provides feedback on information and education materials for general public, families, and providers to assure accuracy of health information

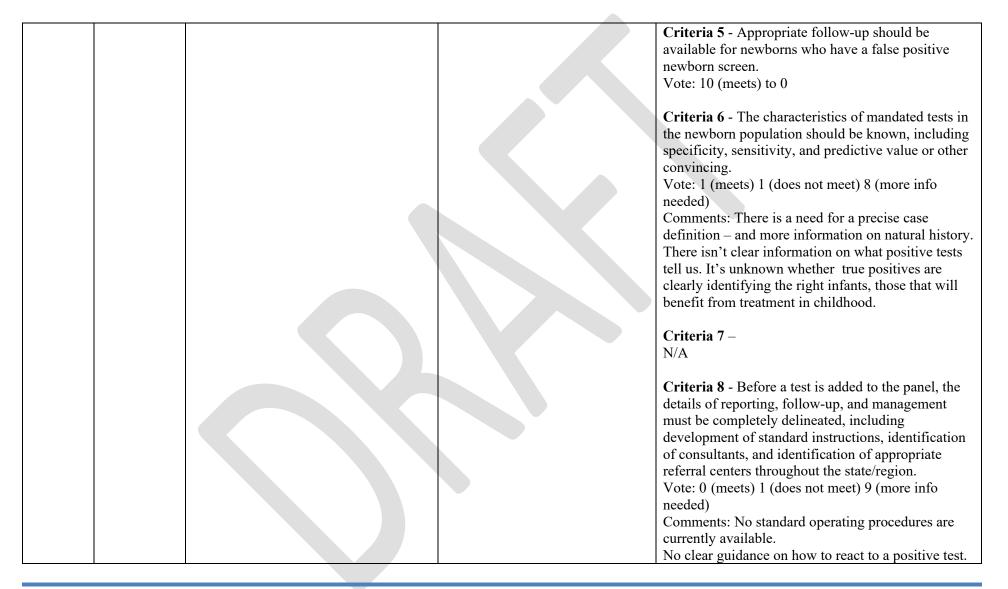


related to the specific condition(s). Addition/Deletion of Conditions • Recommends and/or reviews nominations for addition and deletion of conditions related to the subcommittee Facilitator/Chair: Dr. Donald Basel Recorder: Tami Horzewski					
Info Decision Discuss.	Time:	Topic(s)	Follow-up Items Decision/action: Name: Date due:	Content-focused minutes	
Decision	10:00-10:10	Welcome, Review and Approve Minutes (Dr. Basel (Chair))		Motion to approve February 2, 2024 minutes: 1st motion: Sonja Henry 2nd motion: Dr. Jess Scott-Schwoerer Motion approved.	
Info	10:10-11:00	Acid Sphingomyelinase Deficiency (ASMD)/Niemann-Pick Disease Nomination Background (Jill Beirl/Dr. Justin Hopkin) Review and Vote on Nine Criteria (Dr. Basel/Tami Horzewski/Voting Members)		 Dr. Justin Hopkin shared a brief background on Acid Sphingomyelinase Deficiency (ASMD)/Niemann-Pick Disease including disease description, diagnosis, screening, treatment, and outcomes. The ten metabolic subcommittee voting members reviewed each of the nine criteria. Criteria 1 - Mandated testing should be limited to conditions that cause serious health risks in childhood that are unlikely to be detected and prevented in the absence of newborn screening. Vote: 1 (Meets) to 9 (more info needed). Comments: Is there a difference in treatment after symptoms develop? 	

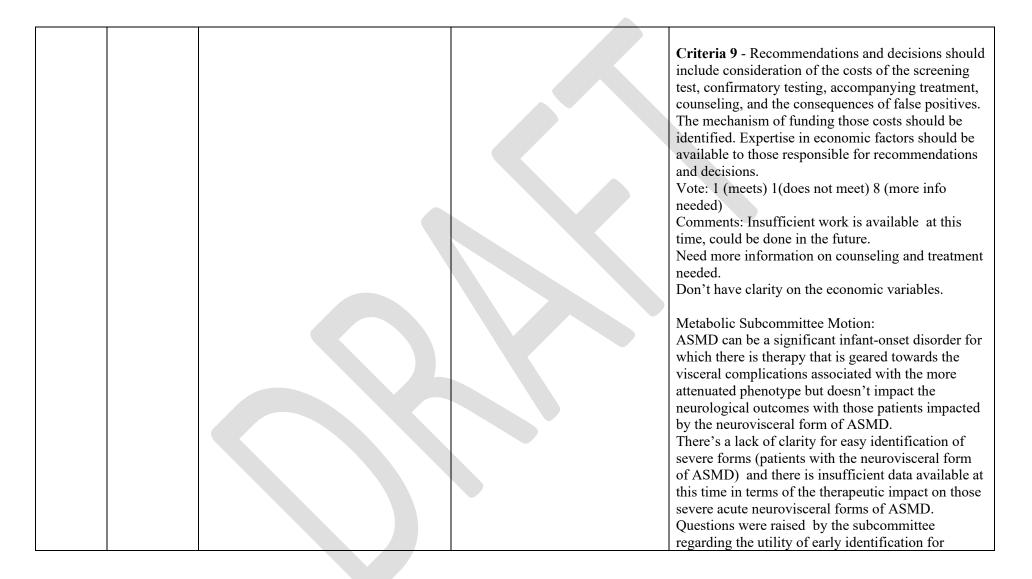












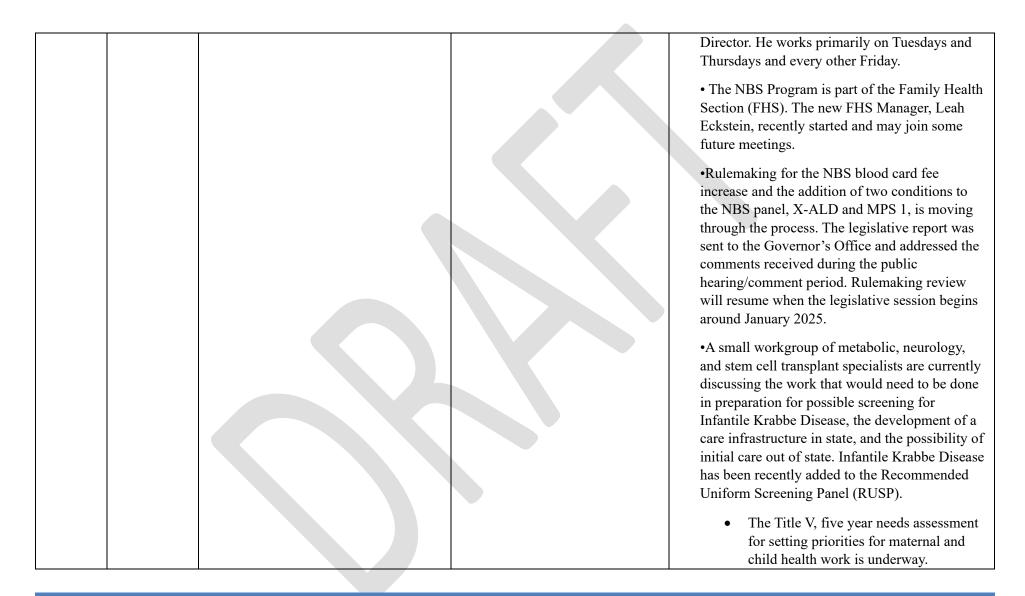


			 visceral impacts related to the chronic burden of disease for later onset for more attenuated forms. The subcommittee recommendation is that ASMD should not be added to the WI NBS panel at this time because additional information is needed. 1st motion: Dr. Jess Scott-Schwoerer 2nd motion: Dr. Mei Baker Motion approved.
			The Metabolic Subcommittee recommendation will be forwarded on to the Umbrella Committee for review at the December meeting.
Info	11:00-12:00	Guanidinoacetate Methyltransferase (GAMT) Deficiency Nomination Background (Dr. Scott-Schwoerer/Dr. Mendez) Review and Vote on Nine Criteria (Dr. Basel/Tami Horzewski/Voting Members)	 Dr. Scott-Schwoerer shared a nomination for Guanidinoacetate Methyltransferase (GAMT) Deficiency. She provided information on the metabolic pathway, incidence, birth prevalence, access to treatment, goals of treatment, and treatment outcomes. Treated patients do have improvements in developmental and seizure control. She stated that there is a current process in place for follow up and would be managed like other screening disorders. The costs would be about \$10/card for testing. There would be costs for special dietary treatment and counseling. Dr. Mendez shared information about lab testing and experiences from four pilot studies of GAMT. Lab assay development has already been started for GAMT through the Propel grant funding. It is not currently FDA approved and requires validation as an LDT. A validation study is in progress.



	12:00-12:10	Break	The ten voting metabolic subcommittee members reviewed the nine criteria and voted 10 to 0 that the nomination meets all criteria. Metabolic Subcommittee Motion: The metabolic subcommittee recommends the addition of GAMT to the Wisconsin newborn screening panel of conditions. The condition meets all the criteria and the metabolic subcommittee will forward their recommendation to the umbrella committee for consideration of this condition to be screened for in Wisconsin. 1st motion: Dr. Mei Baker 2nd motion: Dr. Mei Baker 2nd motion: Dr. Michael Muriello Motion approved. The Metabolic Subcommittee recommendation will be forwarded on to the Umbrella Committee for the nomination review at the December meeting.
Info	12:10-12:20	Department of Health Services (DHS) Update (Tami Horzewski/JDr. Steiner/Julie Thiel)	Tami Horzewski shared the following DHS Update: •Dr. Steiner is back with the NBS Program part- time in the role of DHS NBS Program Medical

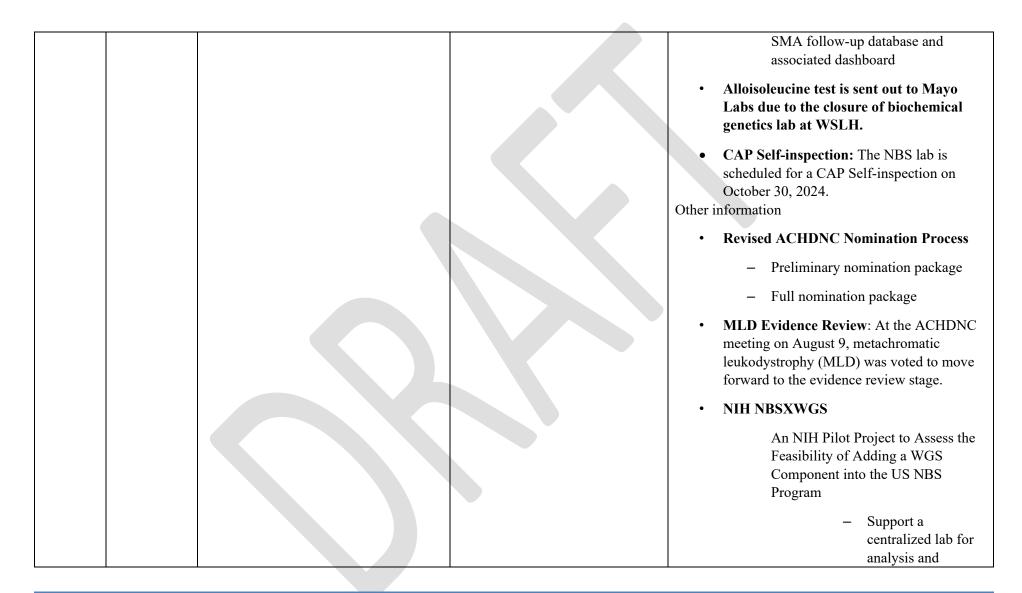






			Julie Thiel shared some background. There were focus groups and a state- wide survey. This information will be analyzed in November and in December will look at setting priorities for the next 5 years, 2026-2030.
Info	12:20-12:30	WI State Lab of Hygiene (WSLH) Update (Dr. Baker)	 WI State Lab of Hygiene (WSLH) Update HRSA-23-065: State Newborn Screening Priorities Program (NBS Propel) Specific Aim 1: Expand testing capability to improve laboratory readiness for screening Mucopolysaccharidosis types I and II (MPS I and MPS II), and Guanidinoacetate Methyltransferase (GAMT) deficiency.— Guanidinoacetate assay evaluation in progress. Specific Aim 2: Improve NBS specimen transit time via increasing transparency and effective communication.—Ongoing. Specific Aim 3: Establish a system and a process to monitor spinal muscular atrophy screening positive infants and assess treatment efficacy. –REDCap-based 5 year

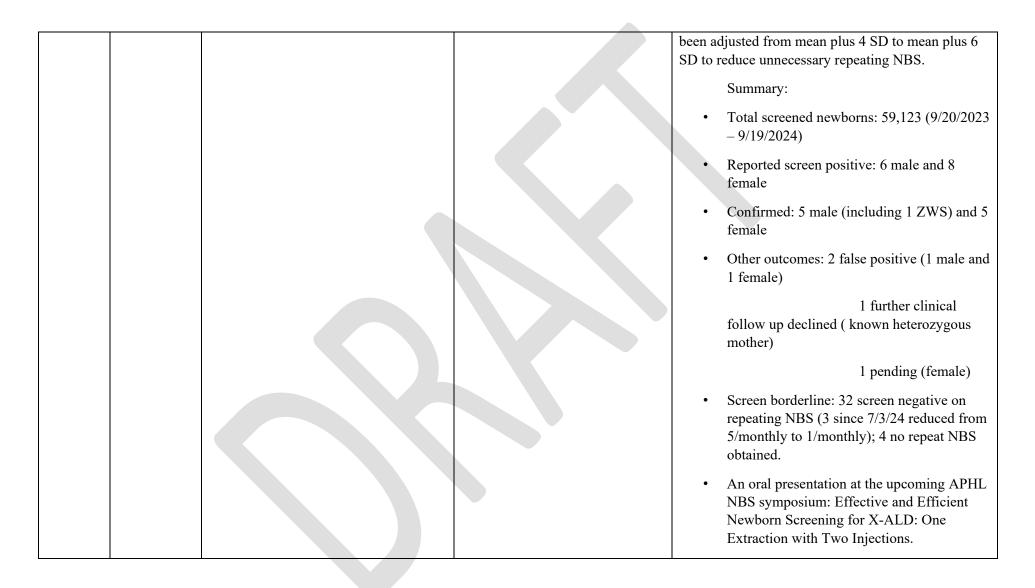






				interpretation of genomic sequencing results. Focus on a limited gene panel of serious / life- threatening rare diseases with early treatment options available. Achieve equitable access to genomic sequencing in the newborn period Examine ethical, legal & social implications (ELSI) of population-wide genomic sequencing in the
				sequencing in the newborn period.
Info	12:30-12:40	X-ALD Screening Progress Report (Dr. Baker)	X-ALD NBS Implementation I The process was reviewed at th meeting and refreshed at this m major update: the screening bo	e last committee eeting with one







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			• Tentative plan to develop a manuscript on the first 18 months experience.
Info	12:40-12:55	2023 Screening Summary (Dr. Mendez)	 Dr. Mendez shared the 2023 screening summary, including the number of cases reported for each condition and the number of confirmed cases. In 2023, there were the following number of confirmed cases: 3 confirmed cases: 3 confirmed cases of Galactosemia 6 confirmed cases of late onset Pompe Disease 6 confirmed Organic Acidurias 2 confirmed Fatty Acid Oxidation Disorders He also shared the number of referrals from Children's Wisconsin (CW) and Waisman for the various disorders. There were a total of 25 referrals from CW and 33 referrals from Waisman. Diet Monitoring numbers were also shared, showing a reduction in the number of specimens and an increase in the number of specimens rejected.
Disc	12:55 – 1:00	Plan Next Meeting/Agenda Items	There was an initial mention of re-nomination of Krabbe Disease with the addition of Infantile Krabbe Disease to the RUSP. The hope is to receive a re-nomination soon as that is needed in order to consider Krabbe Disease for addition to the Newborn Screening panel. Amy White shared her interest in being a resource for a Krabbe nomination. Subcommittee members will continue discussion. Tami Horzewski will send out a doodle poll for



		scheduling the next subcommittee meeting in either February, March, or April 2025.
		reorauly, march, or reprinzozo.

Next meeting date: TBD "Parking Lot" Items: