

The Advisory Committee on Heritable Disorders in Newborns and Children

WEBINAR May 12-13, 2022

Agenda: Thursday, May 12, 2022 from 10:00 AM – 3:20 PM Eastern Time

TIME	TOPIC	PRESENTER
10:00 AM – 10:10 AM	<ul style="list-style-type: none"> • Welcome • Roll Call • Opening Remarks • Committee Business 	<p>Cynthia M. Powell, MD, MS, FACMG, FAAP Committee Chair</p> <p>Soohyun Kim, MPH, CPH Acting Designated Federal Official Health Resources and Services Administration</p>
10:10 AM – 11:10 AM	Updates on Homocystinuria NBS Status: Panel Presentation	<p>Marzia Pasquali, PhD, FACMG Professor of Pathology, University of Utah School of Medicine Section Chief & Medical Director, Biochemical Genetics, ARUP Laboratories</p> <p>Dietrich Matern, MD, PhD, FACMG Professor of Laboratory Medicine, Medical Genetics, and Pediatrics Co-Director, Biochemical Genetics Laboratory Mayo Clinic College of Medicine</p> <p>Kostas Petritis, PhD Laboratory Chief, Biochemical Mass Spectrometry Laboratory, Newborn Screening and Molecular Biology Branch Centers for Disease Control and Prevention</p>
11:10 AM – 11:50 AM	Public Comment	
11:50 AM – 12:50 PM	Newborn Screening for Guanidinoacetate Methyltransferase (GAMT) Deficiency: A Systematic Review of the Evidence (Part 1)	<p>Alex R. Kemper, MD, MPH, MS Lead, Evidence-Based Review Group</p> <p>Jelili Ojodu, MPH Member, Evidence-Based Review Group</p> <p>Lisa A. Prosser, PhD Member, Evidence-Based Review Group</p>
12:50 PM – 1:20 PM	Break	

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TIME	TOPIC	PRESENTER
1:20 PM – 2:20 PM	Newborn Screening for GAMT Deficiency: A Systematic Review of the Evidence (Part 2)	<p>Alex R. Kemper, MD, MPH, MS Lead, Evidence-Based Review Group</p> <p>Jelili Ojodu, MPH Member, Evidence-Based Review Group</p> <p>Lisa A. Prosser, PhD Member, Evidence-Based Review Group</p>
2:20 PM – 2:50 PM	Committee Report: Newborn Screening for GAMT Deficiency	<p>Jane M. DeLuca, PhD, RN, CPNP Committee Member</p> <p>Shawn E. McCandless, MD Committee Member</p>
2:50 PM – 3:20 PM	<ul style="list-style-type: none"> • Committee Discussion • <i>Vote on whether or not to recommend GAMT deficiency for inclusion on the Recommended Uniform Screening Panel</i> 	<p>Cynthia M. Powell, MD, MS, FACMG, FAAP Committee Chair</p>
3:20 PM	End of Day 1	

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TIME	TOPIC	PRESENTER
10:00 AM – 10:20 AM	<ul style="list-style-type: none"> • Welcome • Roll Call • Committee Business 	Cynthia M. Powell, MD, MS, FACMG, FAAP Committee Chair Soohyun Kim, MPH, CPH Acting Designated Federal Official Health Resources and Services Administration
10:20 AM – 10:50 AM	Public Comment	
10:50 AM – 11:20 AM	Krabbe Nomination Summary	Cynthia M. Powell, MD, MS, FACMG, FAAP Committee Chair
11:20 AM – 11:40 AM	<ul style="list-style-type: none"> • Committee Discussion • <i>Vote on whether or not to move Krabbe forward to full evidence review</i> 	Cynthia M. Powell, MD, MS, FACMG, FAAP Committee Chair
11:40 AM – 11:50 AM	Break	
11:50 AM – 12:30 PM	Updates on Newborn Screening Family Education Program	Natasha Bonhomme Founder, Expecting Health Chief Strategy Officer, Genetic Alliance Marianna Raia, MS, CGC Associate Director of Programs Expecting Health
12:30 PM – 12:40 PM	New Business	Cynthia M. Powell, MD, MS, FACMG, FAAP Committee Chair
12:40 PM	Adjourn	