

December 5–6, 2024 Sheraton Portsmouth Harborside Hotel, Portsmouth, NH



# December 5, 2024: Morning Session

8:30 am	Registration/Exhibits Open	Grand Ballroom Lobby/Harbor's Edge Room
9:30 am	Welcome	Grand Ballroom
7,00	- Erica Stelmach, MS, CGC President, NERGG 2024 and 2025	
9:45 am	Thymidine Kinase 2 Deficiency – Navigating Common Mimickers, Unraveling Clinical Clues, and Exploring the Patient Journey	Grand Ballroom
	<ul> <li>Sponsored by UCB</li> <li>Jessica DuHaime, PharmD, BCPPS         Medical Science Liaison II, Early Indications, Rare Disease, UCB, US Medical Affairs</li> <li>Sarah Feinsmith, MSN, APRN, CNS         Nurse Educator, UCB</li> </ul>	
10:45 am	Break/Visit Exhibits	Grand Ballroom Lobby/Harbor's Edge Room
11:00 am	Barth Syndrome	Grand Ballroom
	<ul> <li>Mark Korson, MD         Director of Physician Support and Education, VMP Genetics, LLC     </li> <li>Kate Michener, Parent Speaker         Director of User Experience, NH Department of Information Technology     </li> </ul>	
0 noon	Lunch/Visit Exhibits	Grand Ballroom Lobby/Harbor's Edge Room



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## December 5, 2024: Afternoon Session

1:30 pm	Gamete Donation	Grand Ballroon
_	- Kathryn Lockwood, MS, CGC	
	Senior Reproductive Genetic Specialist, California Cryobank, a subsidiary of	
	CooperSurgical, Inc.	
2:30 pm	Innovations in Genetic Testing: Designing and Curating for Maximized Clinical	
	Utility	Grand Ballroon
	Sponsored by Ambry Genetics	
	- Tina Pesaran, MA, MS, CGC	
	Director of Ambry's Genomic Sciences Department	
	- Tara Namey, MS, CGC	
	Director of Ambry's Genomic Science Liaison Team	
		Grand Ballroor
		Lobby/Harbor'
3:45 pm	Break/Visit Exhibits	Edge Room
4:00 nm	Rhode Island's Implementation of the Blueprint for Change	Grand Ballroon
4.00 pm	- Tara Hayes, BS, CCHW	Grana Banroon
	Policy, Advocacy, & Family Voices Manager; RI Parent Information Network	
	- Colleen Polselli, BA, BS	
	Family/Peer Resource Specialist Program; RI Department of Health, Office of Special Needs	
	,	Grand Ballroon
5:00 pm	Cocktail Hour and Reception	Lobby
6:00 pm	Dinner and Dessert Buffet	Grand Ballroon
	Dessert buffet sponsored by Alexion	



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## December 6, 2024: Morning Session

7.00 am	Sponsored Breakfast with Speaker	Grand Ballroom
7:00 am	· · · · · · · · · · · · · · · · · · ·	Grana Banroom
	Sponsored by Sanofi Over Yonder: The Blood Brain Barrier as the New Frontier in LSDs	
	- Jennifer Matthews, MS	
	Medical Strategic Account Director, Sanofi Medical Affairs, US Rare Diseases  - Angelo Walton, PharmD, RPh	
	Medical Strategic Account Director, Sanofi Medical Affairs, US Rare Diseases	
	*Note: Speakers start at 7:15. No CEUs offered for this presentation.	
8:15 am	Genetic Navigators Programs	Grand Ballroom
	– Lynda Kazairwe, LL.B	
	GradICSA; Lend Trainee (UCONN); PATH CT	
	- Ashley Michaud, BA	
	Vermont Family Network	
9:30 am	Precision Genomic Medicine: From N of 1 to Population Health	Grand Ballroom
	<ul><li>Wendy Chung, MD, Ph.D</li></ul>	
	Chief of the Department of Pediatrics at Boston Children's Hospital; Mary Ellen	
	Avery Professor of Pediatrics at Harvard Medical School	
		Grand Ballroom
		Lobby/Harbor's
10:45 am	Break/Visit Exhibits	Edge Room
11:00 am	Transforming Post-Mortem Care	Grand Ballroom
	Panel Presentation from Boston Children's Hospital	
	- Monica Wojcik, MD, MPH	
	Neonatal Genomics Program Director	
	- Ingrid Holm, MD, MPH	
	Geneticist, BCH	
	- Richard Goldstein, MD	

Director, Robert's Program on Sudden Unexpected Death

Director of Neuropathology; Director of Autopsy

Director, Psychology for Robert's Program on Sudden Unexpected Death

Sarah McCarthy, Ph.D., MPH

Sanda Alexandrescu, MD

12:30 pm Lunch/Visit Exhibits

Grand Ballroom Lobby/Harbor's

Edge Room



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### December 6, 2024: Afternoon Session

#### 1:45 pm Long-Read Sequencing in Diagnostic Testing

Grand Ballroom

Christine Stanley, Ph.D., FACMG
 Chief Genomics & Compliance Officer, Variantyx, Inc.

#### 2:45 pm Advanced Cell-Free DNA Applications in Women's Health and Oncology

Grand Ballroom

Sponsored by Myriad

- Lauren Eisemann, MS, CGC

Women's Health Medical Science Liaison at Myriad Genetics

- Alexandra Katterfield, MS, CGC

Oncology Medical Science Liaison at Myriad Genetics

#### 3:45 pm Closing and Adjournment

Grand Ballroom

- Erica Stelmach, MS, CGC
President, NERGG 2024 and 2025