



NERGG 47th Annual Educational Conference 2024

December 5–6, 2024

Sheraton Portsmouth Harborside Hotel, Portsmouth, NH



December 5, 2024: Morning Session

8:30 am	Registration/Exhibits Open/Coffee in Harbor's Edge	Grand Ballroom Lobby/Harbor's Edge Room
9:30 am	Welcome <ul style="list-style-type: none">– Erica Stelmach, MS, CGC <i>President, NERGG 2024 and 2025</i>	Grand Ballroom
9:45 am	Thymidine Kinase 2 Deficiency – Navigating Common Mimickers, Unraveling Clinical Clues, and Exploring the Patient Journey <i>Sponsored by UCB</i> <ul style="list-style-type: none">– Jessica DuHaime, PharmD, BCPPS <i>Medical Science Liaison II, Early Indications, Rare Disease, UCB, US Medical Affairs</i>– Sarah Feinsmith, MSN, APRN, CNS <i>Nurse Educator, UCB</i>	Grand Ballroom
10:45 am	Break/Visit Exhibits	Grand Ballroom Lobby/Harbor's Edge Room
11:00 am	Barth Syndrome <ul style="list-style-type: none">– Mark Korson, MD <i>Director of Physician Support and Education, VMP Genetics, LLC</i>– Kate Michener, Parent Speaker <i>Director of User Experience, NH Department of Information Technology</i>	Grand Ballroom
12:00 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 Ballroom

- 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 Ballroom

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 Ballroom
Lobby/Harbor's
Edge Room

3:45 pm Break/Visit Exhibits

4:00 pm Rhode Island's Implementation of the Blueprint for Change

Grand Ballroom

- Tara Hayes, BS, CCHW
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

5:00 pm Cocktail Hour and Reception

Grand Ballroom
Lobby

6:00 pm Dinner and Dessert Buffet

Grand Ballroom

Dessert buffet sponsored by Alexion



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December 6, 2024: Morning Session

7:00 am Sponsored Breakfast with Speakers

Grand Ballroom

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

- Wendy Chung, MD, Ph.D
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
Edge Room

10:45 am Break/Visit Exhibits

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
- Sarah McCarthy, Ph.D., MPH
Director, Psychology for Robert's Program on Sudden Unexpected Death
- Sanda Alexandrescu, MD
Director of Neuropathology; Director of Autopsy

Grand Ballroom
Lobby/Harbor's
Edge Room

12:30 pm Lunch/Visit Exhibits



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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
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Speakers



Sanda Alexandrescu
MD



Wendy Chung
MD, Ph.D



Jessica DuHaime
PharmD, BCPPS



Lauren Eisemann
MS, CGC



Sarah Feinsmith
MSN, APRN, CNS



Richard Goldstein
MD



Tara Hayes
BS, CCHW



Ingrid Holm
MD, MPH



Alexandra Katterfield
MS, CGC



Lynda Kazairwe
LL.B



Mark Korson
MD



Kathryn Lockwood
MS, CGC



Sarah McCarthy
Ph.D., MPH



Jennifer Matthews
MS



Ashley Michaud
BA



Kate Michener
Parent Speaker



Tara Namey
MS, CGC



Tina Pesaran
MA, MS, CGC



Colleen Polselli
BA, BS



Christine Stanley
Ph.D., FACMG



Erica Stelmach
MS, CGC



Angelo Walton
PharmD, RPh



Monica Wojcik
MD, MPH



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