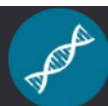




# NERGG 46<sup>th</sup> Annual Educational Conference 2023

December 7–8, 2023

An in-person meeting supported by the New England Regional Genetics Network



## Day One Agenda

### Day One: December 7, 2023

<b>9:30 am</b>	<b>Welcome and Housekeeping Details</b>	<i>Grand Ballroom</i>
	<ul style="list-style-type: none"><li>Tara Hayes, BS, CCHW <i>Advocate, Rhode Island Parent Information Network; President, NERGG 2023</i></li></ul>	
<b>9:45 am</b>	<b>ELSI Considerations In the Design of a Trial Disclosing Cancer Genetic Research Results to U.S. Black Women</b>	<i>Grand Ballroom</i>
	<ul style="list-style-type: none"><li>Maggie Ruderman, MS, CGC <i>Licensed Genetic Counselor, Boston Medical Center</i></li></ul>	
<b>10:45 am</b>	<b>Break/Visit Exhibits</b>	<i>Harbor's Edge Room</i>
<b>11:00 am</b>	<b>Precision Medicine in Oncology: A Genetic Counselor's Perspective</b>	<i>Ballroom</i>
	<ul style="list-style-type: none"><li>Nicholas Jensen, MS, CGC <i>Senior Genetic Counselor, Familial Cancer Program, Dartmouth Hitchcock Medical Center</i></li></ul>	
<b>12:00 noon</b>	<b>Lunch/Visit Exhibits</b>	<i>Grand Ballroom</i>
<b>1:30 pm</b>	<b>Alphabet Soup: ENPP1 Deficiency, GACI, &amp; ARHR2</b>	<i>Grand Ballroom</i>
	<p><i>Sponsored by Inozyme</i></p> <ul style="list-style-type: none"><li>Jacqueline Kernaghan, MS, PA-C, DFAAPA <i>Osteoporosis Center of Delaware County, West Chester, PA</i></li></ul>	
<b>2:45 pm</b>	<b>Advances in Epilepsy Genetic Testing: Exploring the Clinical Utility of Genomic Sequencing</b>	<i>Grand Ballroom</i>
	<p><i>Sponsored by GeneDx</i></p> <ul style="list-style-type: none"><li>Olivia Trimmier, MS, CGC <i>Medical Science Liaison, GeneDx, Charleston, SC</i></li></ul>	
<b>3:45 pm</b>	<b>Break/Visit Exhibits</b>	<i>Harbor's Edge Room</i>



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<b>4:00 pm</b>	<b>Resources for Down Syndrome: National Parents First Call Center/MDSC Parents First Call Program</b>	<i>Grand Ballroom</i>
	<ul style="list-style-type: none"><li>- Mary-Frances Garber, MS, CGC <i>Genetic Counselor, Private Practice: Listening, Reflecting, Healing</i></li><li>- Sarah Cullen <i>Family Support Director, Massachusetts Down Syndrome Congress (MDSC)</i></li><li>- Sarah Twomey-Mercurio, BA, MPA</li></ul>	
<b>5:00 pm</b>	<b>Strategies for Genetic Education Across New England</b>	<i>Grand Ballroom</i>
	<ul style="list-style-type: none"><li>- Angela Shepard, MD, MPH <i>Director, New England Regional Genetics Network at the Institute on Disability, University of New Hampshire</i></li></ul>	
<b>6:00 pm</b>	<b>Meet and Mingle (Hors d'oeuvres will be served)</b>	<i>Amphitheater</i>
	<p><i>Sponsored by Sanofi</i></p> <b>Better Care for Rare – Identifying Lysosomal Disorders</b> <ul style="list-style-type: none"><li>- Punita Gupta, MD <i>Chief, Division of Genetics and Medical Director of the Lysosomal Storage Disease Center at St. Joseph's University Medical Center, Paterson, NJ</i></li></ul> <p><i>*Note: CEUs are not offered for this presentation</i></p>	
<b>7:00 pm</b>	<b>Dinner Buffet</b>	<i>Grand Ballroom</i>

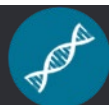
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## Day Two Agenda

### Day Two: December 8, 2023

<b>7:30 am</b>	<b>Sponsored Breakfast with Speaker</b>	<i>Grand Ballroom</i>
	<i>Sponsored by BioMarin</i>	
	<b>Take a Deeper Look at Achondroplasia</b>	
	– Robert Gensure, MD <i>Chief of Pediatric Endocrinology, Dartmouth Hitchcock Medical Center</i>	
	<i>*Speaker begins at 8:15 am</i>	
<b>9:30 am</b>	<b>Jansen deVries Syndrome (JdVS): A Neurodevelopmental Disorder</b>	<i>Grand Ballroom</i>
	– Catherine Nowak, MD <i>Clinical Director, Division of Genetics and Metabolism, Massachusetts General Hospital for Children</i>	
	– Kara Kilroy, MS <i>Director, JdVS Foundation</i>	
<b>10:45 am</b>	<b>Break/Visit Exhibits</b>	<i>Harbor's Edge Room</i>
<b>11:00 am</b>	<b>Exploring the Worlds of Deaf, Hard of Hearing, and DeafBlind Children</b>	<i>Grand Ballroom</i>
	– Linda A. Hazard, Ed.D., CCC-A <i>Program Director Vermont Early Hearing Detection and Intervention Program, Vermont Department of Health</i>	
	– Karl White, PhD <i>Director, The National Center for Hearing Assessment and Management (NCHAM), Utah State University, Logan, Utah</i>	
<b>12:00 noon</b>	<b>Lunch/Visit Exhibits</b>	<i>Grand Ballroom</i>
<b>1:30 pm</b>	<b>Child Abuse in the World of Disability</b>	<i>Grand Ballroom</i>
	– Catherine Nowak, MD <i>Clinical Director, Medical Genetics and Metabolism Division, Massachusetts General Hospital for Children</i>	
<b>2:45 pm</b>	<b>Genome Sequencing and the Changing Landscape of Newborn Care</b>	<i>Grand Ballroom</i>
	– W. Gregory Feero, MD, PhD <i>Physician, Four Seasons Family Practice, Fairfield, ME; Professor, Community &amp; Family Medicine, Geisel School of Medicine at Dartmouth; Co-investigator, Ethical, Legal, and Social Policy Implications of Workplace Genomic Testing, National Human Genome Research Institute</i>	
<b>3:45 pm</b>	<b>Closing/Adjournment</b>	<i>Grand Ballroom</i>
	– Erica Stelmach, MS, CGC <i>Genetic Counselor, Mayo Clinic Laboratories; Incoming President, NERGG 2024</i>	



# Speakers



**Sarah Cullen**



**W. Gregory Feero, MD,  
PhD**



**Mary-Frances Garber,  
MS, CGC**



**Robert Gensure, MD**



**Tara Hayes, BS, CCHW  
and son Andrew**



**Linda Hazard, Ed.D.,  
CCC-A**



**Nicholas Jensen, MS,  
CGC**



**Jacqueline Kernaghan,  
MS, PA-C, DFAAPA**



**Kara Kilroy, MS**



**Catherine Nowak, MD**



**Maggie Ruderman,  
MS, CGC**



**Angela Shepard, MD,  
MPH**



**Erica Stelmach, MS, CGC**



**Olivia Trimmier, MS,  
CGC**



**Sarah Twomey-  
Mercurio, BA, MPA**



**Karl White, PhD**



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Family Voices Manager  
Rhode Island Parent Information Network  
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[director@negg.org](mailto:director@negg.org)  
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Public Health: Ginger Nichols, MS, LCGC  
Consumer: Peggy Sweeney, BA

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Public Health: Anne Watson, BS, MSc, PhD,  
BSN, RN  
Consumer: Sarah Staffiere, BA, MS

### **MASSACHUSETTS**

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Practitioner: Catherine Nowak, MD  
Public Health: Eirini Nestoridi, MD  
Consumer: Jean Bertschmann, BA

### **NEW HAMPSHIRE**

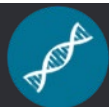
Practitioner: Lisa Demers, MS, CGC  
Public Health: Amanda Merrill, BSN, RN  
Consumer: Joanne Huff, BSN, RN

### **RHODE ISLAND**

Practitioner: Kerry Ahern, MS, CGC  
Public Health: Emily Eisenstein, MPH  
Consumer: Stephanie Trafka, CCHW

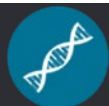
### **VERMONT**

Practitioner: Christine Giummo, MS, CGC  
Public Health: Linda Hazard, Ed.D., CCC-A  
Consumer: Allison Wood, MS, CGC



## Special Thanks

In grateful appreciation of NERGN's many years of support and collaboration.



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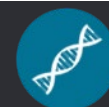
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