NERGG 42nd Annual Educational Conference 2019

December 5–6, 2019

Sheraton Portsmouth Harborside Hotel, Portsmouth, New Hampshire

FINAL AGENDA

Day One: December 5, 2019

9:00	**Registration, Coffee, and Breakfast Snacks**	**Lobby**
	Sponsored by Invitae	**Langdon**
9:50	**Welcome**	**Ballroom**
	– *Ms. Mary-Frances Garber, MS, CGC President, NERGG*	
10:00	**Angelman Syndrome: Phenotypic Spectrum and Management**	**Ballroom**
	– *Dr. Wen-Hann Tan, Boston Children's Hospital, Genetics & Genomics Div.* – *Dr. Noelle Germain, Stem Cell Core, UCONN Health* – *Mr. Tim Bousum, VP, Angelman Syndrome Foundation*	
11:00	**Genetic Test Utilization and Management**	**Ballroom**
	– *Mr. Andrew McCarty, Lab Genetics Counselor, Perkin Elmer Genomics*	
12:00	**Lunch Buffet**	**Prescott**
	Visit with Exhibitors	**Langdon**

Day One: December 5, 2019

1:00 Finding Hope in Uncertainty — **Ballroom**

Panel co-sponsored by NERGG and NERGN

- *Dr. Susan Zimmermann, ABLE NH*
- *Ms. Tara Hayes, Rhode Island Parent Information Network*
- *Dr. Peter Antal, Co-Director, New England Regional Genetics Network*
- *Ms. Jennifer and Mr. Shawn Bertrand, Community Crossroads Inc.*
- *Ms. Chloe Bertrand, Self-Advocate*

2:30 Hereditary Transthyretin Amyloidosis: Presentation, Burden, Diagnosis, and Assessment — **Ballroom**

Sponsored by Akcea Therapeutics

- *Dr. Sami Khella, Chief, Dept. of Neurology at Penn Presbyterian MC*
- *Mr. Brian Schoenfeld, MS, CGC*

3:30 Beverage Break — **Langdon**

Visit with Exhibitors

4:00 NERGG Annual Meeting — **Ballroom**

All are invited

4:30 CANNEW Presentations — **Ballroom**

Research abstracts and case studies

6:30 Cocktail Reception — **Langdon**

7:00 Dinner Buffet — **Prescott**

Day Two: December 6, 2019

7:30	**Breakfast Buffet**	**Lobby**
	Sponsored by Variantyx, Inc.	
8:00	**Whole Genome Sequencing and the Laboratory Perspective: Name that Disorder!**	**Ballroom**
	Dr. Christine Stanley, Director, Clinical Genomics, Variantyx	
9:00	**Whole Genome Sequencing and the Research Perspective: Family Experience with Late Diagnosis and Intervention**	**Ballroom**
	Sponsored by the Broad Institute of Harvard and MIT	
	– *Ms. Grace VanNoy, MS, CGC Broad Institute*	
	– *Ms. Melanie O'Leary, MS, LGC Broad Institute*	
	– *Ms. Lynn Elko, MA, Parent Advocate*	
10:15	**Snack Break**	**Langdon**
10:45	**A Surgeon's Role in Communicating the Genetic Aspects of Breast Cancer**	**Ballroom**
	– *Dr. David Coppola, Seacoast General Surgery PC*	
12:00	**Lunch Buffet**	**Prescott**
	Visit with Exhibitors	
1:00	**Glycogen Storage Disease Type Ia: 40 Years of Progress from Cornstarch to Gene Therapy**	**Ballroom**
	– *Dr. David Weinstein, GSD Program Director, CT Children's Medical Ctr.*	
2:15	**Beverage and Snack Break**	**Langdon**
	Visit with Exhibitors	
2:30	**Duchenne Muscular Dystrophy: An Overview of Care Needs for the Patient and Family**	**Ballroom**
	– *Dr. Brenda Wong, Director of Interdisciplinary DMD Clinic, UMASS Memorial Medical Center*	
	– *Ms. Jennifer McNary, DMD Advocate and Consultant*	
3:30	**Wrap-Up and Adjournment**	**Ballroom**
	– *Mr. Sam Drazin, newly installed NERGG President*	

SPEAKERS

Wen-Hann Tan, BMBS
Clinical Geneticist, Division of Genetics and Genomics, Boston Children's Hospital; Asst. Professor of Pediatrics, Harvard Medical School
Boston, MA

Germain, Noelle Ph.D.
Asst. Professor, Genetics and Genome Sciences, UCONN Health
Farmington CT

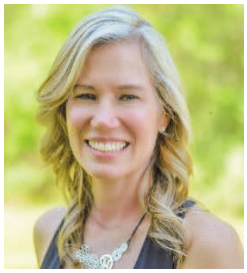
Tim Bousum, BS
VP, Angelman Syndrome Foundation
Ipswich, MA

Andrew McCarty, MS, LCGC
Laboratory Genetics Counselor, Perkin Elmer Genomics
Pittsburgh, PA

Susan Zimmermann, Ph.D.
Chair, Disability Diagnosis Dialogue
Task Force, ABLE NH
Concord, NH

Tara Hayes, BS
Family Voices Manager, RI Parent Information Network
Warwick, RI

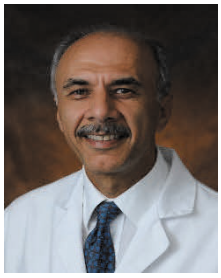
Peter Antal, Ph.D.
Co-Director, New England Regional Genetics Group
Institute on Disability, University of New Hampshire



Jennifer Bertrand
Grad. of UNH-Institute on Disability Leadership Series
Director of Community Partnerships at Community Crossroads
Mont Vernon, NH

Shawn Bertrand
Grad. of UNH-Institute on Disability Leadership Series
Lead Software Systems Engineer for Mitre Corp.
Mont Vernon, NH

Chloe Bertrand
Grad. of Souhegan HS; Front-End Clerk at Whole Foods (Nashua); Self-Advocate
Mont Vernon, NH



Sami Khella, MD
Professor of Clinical Neurology; Chief of Dept. of Neurology
Penn Presbyterian Medical Center
Philadelphia, PA

Brian Schoenfeld, MS, CGC
Senior Genetic Specialist
Ambry Genetics
Manalapan, NJ

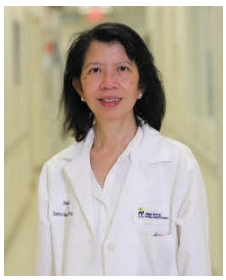
Christine Stanley, Ph.D.
Chief Director, Clinical Genomics
Variantyx
Framingham, MA

Grace VanNoy, MS, CGC
Clinical Project Manager, Rare Genomes Project
Broad Institute of MIT and Harvard
Cambridge, MA

Melanie O'Leary, MS, LGC
Operations Lead, Rare Genomes Project
Broad Institute of MIT and Harvard
Cambridge, MA

David Coppola, MD
Seacoast General Surgery, P.C.
Dover, NH

David Weinstein, MD, MMsc
Director, Glycogen Storage Disease Program
Connecticut Children's Medical Center
Professor, University of Connecticut
Hartford, CT



Brenda Wong, MD
Founding Director, Duchenne Muscular Dystrophy Clinic
UMass Memorial Medical Children's Ctr
Worcester, MA

Jennifer McNary
Consultant
Rare Disease Advocate
Newton, MA

Lynn Elko, MA
Parent Advocate
Allentown, PA

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