NERGG Inc. Annual Educational Conference 2021 Program Objectives

SESSION	SPEAKER(S)	OBJECTIVES Participants will be able to…	NSGC CONTACT HOURS (pending approval)
A Family's Journey with Long-Chain L-3 Hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency	Brittany H.	1. Identify the team of specialists involved in managing the care of a child with LCHAD. 2. Describe the effects on a family with a child with a rare disorder.	0.5 (30 min)
Disorders of Neurotransmitter Metabolism: Aromatic L-Amino Acid Decarboxylase (AADC) Deficiency	Irina Anselm, MD Kelly Heger, RN	1. Summarize the evaluation process for diagnosing AADC. 2. Describe treatment options for AADC. 3. Summarize the life experience of the family of a child with a rare disorder.	1 (60 min)
Rapid Clinical Genomics: Testing Options for the Critically-Ill Patient	Jane Juusola, PhD	1. Summarize the approach taken to analyze and interpret clinical exome data, especially in a NICU/PICU setting. 2. Examine the reporting of clinically relevant variants that may be challenging to interrogate, including copy number variants (CNVs), mosaicism, mobile element insertions (MEIs), and variants in candidate genes.	0.5 (30 min)
Germline and Somatic Tumor Testing	Amber Carter, CGC Sugganth Daniel, MD	1. Review the clinical utility of germline genetic testing and somatic tumor profiling as part of the comprehensive workup of the oncology patient. 2. Examine various clinical implementation approaches, including paired and reflex, for germline and somatic testing.	1 (60 min)
CANNEW Research Updates	Amy Patterson, CGC Sophia Zilber, MA	1. Evaluate data related to the roles of rare disease advocacy groups, including CureMito.	0.5 (30 min)
Lysosomal Storage Disorders: The Patient, Family, and Provider Perspectives	Walla Al-Hertani, MD Maria Fragala-Pinkham, PT, DPT, DSc Aishwarya Siddharth CGC Jill Thresher, RTR, CTR and Sophie Thresher	1. State the pathophysiological processes and genetic components involved in lysosomal storage disorders (LSDs). 2. Describe the roles of the clinical team treating patients with LSDs. 3. Identify issues facing individuals with LSDs.	1.5 (90 min)
Mythbusters: Dispelling Common Misconceptions about Whole Genome Sequencing	Christine Stanley, PhD	1. Define the benefits and clinical utility of whole genome sequencing over other testing methodologies. 2. Use case examples to examine and dispel myths associated with whole genome sequencing.	1 (60 min)
A Family's Journey Navigating a Rare Disease Diagnosis: Atypical-Hemolytic Uremic Syndrome (aHUS)	Sarah Staffiere, MS	1. Summarize the diagnostic odyssey involved in identifying this rare disorder. 2. Describe the effects on a family with a child with this rare disorder.	0.5 (30 min)