

Agenda

7:00 Registration and Continental Breakfast

7:50 Welcome remarks from Lisa Brailey, MD, President of NERGG

8:00 Marina DiStefano, PhD, "Interpretation of Sequence Variant Pathogenicity"

9:00 Deborah Rivlin, "Overview of The Children's Room: Caring Support for Grieving Children, Teens and Families Programs and Services"

10:00 Break

10:15 Allison Cirino, CGC, "Keeping Pace with Cardiovascular Genetics: Updates on the Genetics of Inherited Heart Disease"

11:15 Kate Reed, MPH, ScM, CGC, "Education in a Next-Gen World: Approaches for Training our Colleagues"

12:15 Lunch

1:00 Kimberly LeBlanc, CGC, Emily Glanton, CGC, & Lauren Briere, CGC, Undiagnosed Diseases Network, "The Multidisciplinary Clinical and Research Evaluation of Patients with Rare and Undiagnosed Conditions"

2:00 Sally Rosengren, MD, "Would You Miss Mucopolysaccharidosis (MPS)?"

3:00 Break

3:15 Julie McKinney, "*Only 10 in the World?* A Family's Journey with a Rare Genetic Disorder, and Some Unique New Resources to Help Other Families Find Support"

4:15 Ava Frank, "Our Experience with CdLS"

5:15 Networking Reception

6:00 Adjourn

Speakers

Marina DiStefano, PhD, is a clinical genetic counselor at the University of Massachusetts Lowell. She received her PhD in Molecular Biology and Biochemistry from the University of Massachusetts Lowell. She has been a practicing genetic counselor at the University of Massachusetts Lowell since 2008. She has been a member of the National Society of Genetic Counselors since 2008. She has been a member of the American Society of Human Genetics since 2008. She has been a member of the International Society of Genetic Counselors since 2008. She has been a member of the American Society of Human Genetics since 2008. She has been a member of the International Society of Genetic Counselors since 2008.

Deborah Rivlin is a clinical genetic counselor at the University of Massachusetts Lowell. She received her PhD in Molecular Biology and Biochemistry from the University of Massachusetts Lowell. She has been a practicing genetic counselor at the University of Massachusetts Lowell since 2008. She has been a member of the National Society of Genetic Counselors since 2008. She has been a member of the American Society of Human Genetics since 2008. She has been a member of the International Society of Genetic Counselors since 2008.

Allison Cirino, MS, CGC is a clinical genetic counselor at the University of Massachusetts Lowell. She received her MS in Genetic Counseling from the University of Massachusetts Lowell. She has been a practicing genetic counselor at the University of Massachusetts Lowell since 2008. She has been a member of the National Society of Genetic Counselors since 2008. She has been a member of the American Society of Human Genetics since 2008. She has been a member of the International Society of Genetic Counselors since 2008.

Uh YFXX 7; 7 is the director of the Clinical and Continuing Education Program at The Jackson Laboratory (JAX) and a board certified genetic counselor. She has over ten years of experience in clinical genetics, healthcare provider education, and program development. At JAX, she directs the clinical education effort, which focuses on developing and implementing education to facilitate the integration of genetics and genomics into clinical practice. The programs are developed in collaboration with a variety of healthcare providers and cover topics most pertinent to the intended audience, including genetic risk assessment, precision medicine, and somatic testing for cancer. Prior to joining JAX in 2013, Kate worked at the National Coalition for Health Professional Education in Genetics (NCHPEG), a non-profit focused on increasing integration of genetics into clinical practice through education. Kate also has practiced as a clinical genetic counselor in a variety of pediatric and adult settings..

Ozlem Goker-Alpan, M.D., is president and chief medical officer at the Center for Clinical Trials at O&O Alpan, LLC and the director of the Lysosomal Diseases Research and Treatment Unit in Fairfax, Virginia. After earning her medical degree from Marmara University School of Medicine in Istanbul, Turkey, Dr Goker-Alpan completed her internship and residency in pediatrics at the State University of New York at Stony Brook. She was then awarded a fellowship in clinical biochemical genetics at the National Institutes of Health, Greater Washington Genetics Program in Bethesda, Maryland, and a second fellowship in lysosomal disorders and Gaucher disease at the Clinical Neuroscience Research Branch of the National Institutes of Health. Dr. Goker-Alpan's clinical interests include research on immunity and inflammatory pathways in Gaucher disease and other lysosomal storage disorders. She has authored or coauthored more than 30 articles in peer-reviewed journals, including Molecular Genetics and Metabolism, Brain, several chapter books and Archives of Neurology. She has been an invited speaker at more than 25 national and international meetings and also serves as a medical advisor for the Baltimore-Washington Metropolitan Gaucher patient support group.

Lori Ann Correia, MS, CGC is the founder and president of Allele Consulting, LLC. She is an accomplished genetics professional with extensive experience in drug development for orphan indications including global clinical operations, project management, medical science liaison and patient advocacy. In particular, she has significant experience in orphan diseases, such as Pompe, Sanfillipo A, and Gaucher Diseases, X-linked Hypohidrotic Ectodermal Dysplasia and Lipoprotein Lipase Deficiency.

Kristen Shannon, MS, LCGC is the director of the Cancer Center Genetics Program at Massachusetts General Hospital. She received master's degree in Human Genetics from Sarah Lawrence College. She has been a practicing genetic counselor at the Massachusetts General Hospital Cancer Center's Center for Cancer Risk Assessment (CCRA) since 1997. In addition to her clinical practice, Kristen has served on various subcommittees of the National Society of Genetic Counselors.

Deanna Brockman is a Master's degree candidate in genetic counseling at Boston University School of Medicine. She received a BS in Biology from SUNY Geneseo.

Renee Pelletier, is a Master's degree candidate in genetic counseling at Boston University School of Medicine. She received a Bachelor's degree in Biology from Northeastern University.

Stephanie Laudisi, MS is a second year genetic counseling student at Brandeis University Graduate School of Arts and Sciences. She previously earned a M.S. in Cell and Developmental Biology from the Jefferson College of Biomedical Sciences at Thomas Jefferson University, and also a B.S. in Molecular and Cell Biology from the College of Liberal Arts and Sciences at the University of Connecticut.

Katie Schwarting is a second year genetic counseling student at Brandeis University Graduate School of Arts and Sciences. Previously, she earned a Bachelor's of Arts from University of Virginia in Biology and Spanish Language.

Lori Dobson, MS, LCGC is the Genetic Counseling Coordinator at Brigham and Woman's Hospital. Before joining the practice at Brigham and Woman's in 2012, Lori was a genetic counselor at Boston Children's Hospital. Lori received her Master's degree in Genetic Counseling from California State University.

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