Barth Syndrome Foundation®

2022 International Scientific and Medical Virtual Symposium

July 20-22, 2022

Agenda

July 20, 2022

11:30AM -1:00PM ET (17:30 - 19:00 CET)

Barth Syndrome Cardiomyopathy: Challenges & Opportunities

Extended recovery of cardiac function after severe infantile cardiomyopathy presentation in Barth syndrome
Jessie Yester, Nationwide Children's Hospital

Longitudinal observational study of cardiac outcome risk factor prediction in children, adolescents, and adults with Barth syndrome
Carolyn Taylor, Medical University of South Carolina

The Barth Syndrome Arrhythmia Project
Colin Phoon & Reina Tan, NYU Langone Health

July 21, 2022

11:30AM - 1:00PM ET (17:30 - 19:00 CET)

Treatment & Quality of Life for Barth Syndrome

Qualitative and quantitative measure of fatigue in Barth syndrome
Stacey Reynolds, Virginia Commonwealth University

Quality of life in Barth syndrome
Brittany Hornby, Kennedy Krieger Institute

Long-term efficacy and safety of elamipretide in patients with Barth syndrome: open label extension results of TAZPOWER through 168 weeks
***Jim Carr**, Stealth BioTherapeutics*

1:30PM - 3:00PM ET (19:30 - 21:00 CET)

Barth Syndrome Biology

Barth syndrome's unsung phenotype: 3-methylglutaconic aciduria
Elizabeth A. Jennings, University of Nevada, Reno

Engineered isoforms of Tafazzin: activity, stability, and potental therapeutic utility
Michael T. Chin, Tufts Medical Center

Advances in understanding cardiolipin synthesis and pathological effects of dysregulation in cells and animals
Robin E. Duncan, University of Waterloo

July 22, 2022

11:30AM - 1:00PM ET (17:30 - 19:00 CET)

Barth Syndrome in Physiology

Genomewide association study in a mouse model of Barth syndrome
Suya Wang, Boston Children's Hospital

Activation of stress response signaling rewires cardiac metabolism in Barth syndrome
Jan Dudek, University of Würzburg

The molecular composition of cardiolipins is tightly coupled with the nutrional lipid environment
Markus A. Keller, Medical University of Innsbruck

1:00PM - 1:30PM ET (19:00 - 19:30 CET)

Strategic Partnerships with Barth Syndrome Foundation

Please join Erik Lontok, BSF's Director of Research, for a lunchtime conversation of the intiatives, efforts, and collaborations driving the Foundation's R&D Program

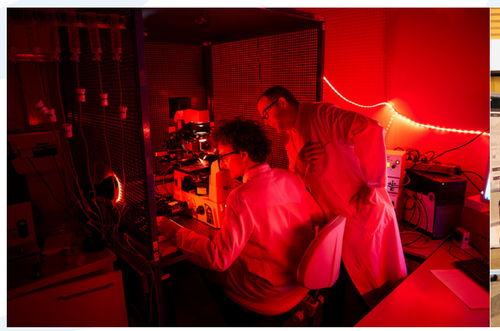
1:30PM - 3:00PM ET (19:30 - 21:00 CET)

Cardiolipin in Physiology

Cardiolipin coordinates lipolysaccharide-induced respiratory Complex II diassembly & degradation
Mack B. Reynolds, University of Michigan Medical School

Cardiolipin bound to mitochondrial ADP/ATP carrier supports the structure and transport-related function
Nanami Senoo, Johns Hopkins University School of Medicine

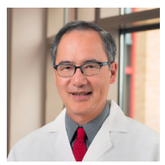
Cardiolipin metabolism regulates myoD1 expression and muscle development
Linh Vo, Wayne State University



Presenters

Jim Carr, Stealth BioTherapeutics

Jim Carr is the Chief Clinical Development Officer of Stealth BioTherapeutics. He brings over 20 years of industry experience in the areas of clinical development, medical affairs, lifecycle management, new product planning, and global marketing to Stealth. Previously, Jim was an Executive Director in the Global Cardiovascular Franchise at GlaxoSmithKline. Prior to GlaxoSmithKline, he held the role of Vice President of Clinical Development at Arca Biopharma. Jim's educational background is a Doctor of Pharmacy degree from the University of Minnesota and post-graduate training in clinical cardiovascular pharmacology. Prior to joining the pharmaceutical industry, he was on the clinical faculty at the University at Buffalo-SUNY School of Pharmacy.

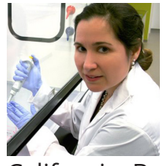


Michael T. Chin, Tufts Medical Center

Dr. Chin is a Professor of Medicine at Tufts University School of Medicine, the Director of the Medical Scientist Training Program and the Research Director for the Tufts Hypertrophic Cardiomyopathy Center. His research interests include studying the molecular mechanisms of heart failure with a particular interest in genetic cardiomyopathies. His current research is focused on the development of an enzyme replacement therapy for Barth Syndrome, a rare inherited disorder of mitochondria. He also oversees a translational research program dedicated to multi-omic analyses of tissue from patients with Hypertrophic Cardiomyopathy and studying pathogenic mechanisms in mouse models.

Jan Dudek, University of Würzburg

Jan Dudek completed his PhD in 2006 in the Department of Biochemistry and Molecular Biology at the University of Freiburg, where he studied mitochondrial protein translocases involved in the transport of newly synthesized proteins into the mitochondria. Between 2007 and 2010, he joined a collaborative project at the Beatson Institute for Cancer Research, Glasgow, UK and at the University of California at San Francisco (UCSF), USA working on oncogenic signaling pathways. In 2010 he joined the Department of Cellular Biochemistry at the University of Göttingen as a project leader, studying the structural and functional defects of the mitochondrial respiratory chain in human diseases. In 2018 he started his junior research group at the Comprehensive Heart Failure Center elucidating the role of mitochondrial dysfunction in cardiomyopathies.



Robin E. Duncan, University of Waterloo

Professor Duncan is an Associate Professor in the Faculty of Health Sciences at the University of Waterloo. She completed her BSc (Hons) at the University of Guelph (Biological Sciences), followed by a PhD at the University of Toronto (Nutritional Sciences), and a Postdoctoral Fellowship at the University of California, Berkeley (Nutritional Science and Toxicology). Her laboratory is focused on the identification of novel enzymes in lipid metabolism, and the study of bioactive lipids in physiological and pathophysiological processes. This work includes both understanding how endogenous lipids function in cellular processes and signaling, and how exogenous bioactive lipids may function therapeutically. Dr. Duncan is the recipient of an Early Researcher Award (Government of Ontario), an Early Career Investigator Award (Canadian Lipoprotein Conference), a Distinguished Teacher Award (University of Waterloo), and a Discovery Accelerator Supplement Award from the Natural Sciences and Engineering Research Council of Canada (NSERC). She is an Academic Editor for PLoS ONE, and a member of the editorial boards for Molecular and Cellular Biochemistry and Molecular Nutrition and Food Research, and serves on grant panels for the Heart and Stroke Foundation of Canada and Diabetes Canada. Her work has been funded by grants from NSERC, Diabetes Canada, the Canada Foundation for Innovation, and the Barth Syndrome Foundation (International and Canadian).

Brittany Hornby, Kennedy Krieger Institute

Brittany Hornby is a senior physical therapist in the Outpatient Physical Therapy Department at Kennedy Krieger Institute. She received a Bachelor of Science degree from James Madison University. She received her Doctorate of Physical Therapy from George Washington University. She serves as Adjunct Assistant Professor for the Department of Health, Human Function and Rehabilitation Services at the George Washington University School of Medicine and Health Sciences and the Department of Physical Therapy and Rehabilitation Science at the University of Maryland School of Medicine. Her current research interests include functional ability and quality of life in patients with Barth syndrome.

Elizabeth A. Jennings, University of Nevada, Reno

Elizabeth Jennings completed her undergraduate education at the University of Nevada, Reno where she earned two Bachelor's degrees in Nutritional Sciences/ Dietetics and Biology with a minor in Biochemistry. She recently completed her second year in the UNR Molecular Biosciences Graduate Program where she is pursuing a PhD in Biochemistry. She has a passion for teaching and has served as a TA for a Molecular Biology Laboratory course for two years. Elizabeth is completing her graduate research in Dr. Robert O. Ryan's laboratory which studies secondary 3-methylglutaconic aciduria, a phenotypic feature of numerous inborn errors of metabolism including Barth syndrome.

Markus A. Keller, Medical University of Innsbruck

Markus Keller is a trained Chemist who started studying lipid metabolism in the context of biomedical research during his PhD. The focus of his scientific work lies in the enzymology and properties of the lipid metabolic network. He applies LC-MS/MS based approaches and bioinformatical strategies to elucidate the pathomechanisms of inborn errors in lipid metabolism.

Colin Phoon, NYU Langone Health

Dr. Phoon is an Associate Professor of Pediatrics (Cardiology), at New York University Langone Health and Grossman School of Medicine, Division of Pediatric Cardiology, Department of Pediatrics. He is investigating the role of mitochondria and cardiolipin in the pathogenesis of cardiomyopathy and Barth syndrome, with a focus on mouse models to investigate both cellular mechanisms and potential therapeutic targets. He has been involved in Barth syndrome research in a close collaboration with Drs. Michael Schlame and Mindong Ren for over a dozen years. As a principal investigator or co-investigator on several projects relevant to a broad spectrum of cardiovascular disease in small animal models, he is especially interested in heart development and heart imaging. Dr. Phoon is a pediatric cardiologist at New York University Grossman School of Medicine, where he has

worked since completing his training in 1996.

Stacey Reynolds, Virginia Commonwealth University

Dr. Stacey Reynolds is a Professor and Director of Research in the Department of Occupational Therapy at Virginia Commonwealth University (VCU). At VCU she directs the Sensory Processing and Stress Evaluation (SPASE) Lab where her aim is to discover neurological mechanisms underlying sensory-motor deficits in children and develop innovative treatments. She has worked with the Barth syndrome community for over a decade examining sensory and motor features that impact feeding, eating, and swallowing. Her more recent work with the Barth community focuses on the relationship between activity levels, sleep, and fatigue and the relationship of these variables with nutrition.

Mack B.Reynolds, University of Michigan Medical School

Mack graduated from the University of California at Davis in 2017 with a B.S. in biochemistry. At UC Davis, he worked as an undergraduate researcher in Dr. Scott Simon's lab studying innate immune cell trafficking in a mouse model of Staphylococcus aureus skin and soft tissue infection. Following graduation, Mack worked in the same lab developing a microfluidic device to predict susceptibility to atherosclerosis in a clinical setting. Mack is now a Ph.D. student in the Program in Immunology at the University of Michigan working in the lab of Dr. Mary O'Riordan.

Nanami Senoo, Johns Hopkins University

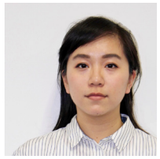
Nanami is a postdoctoral fellow in Johns Hopkins University Department of Physiology with research focuses on phospholipid biology and mitochondrial energy metabolism, currently holding the American Heart Association Postdoctoral Fellowship cofounded with Barth Syndrome Foundation. She received her PhD in Food and Nutritional Science, University of Shizuoka, Japan.

Reina Tan, NYU Rangone Health

Dr. Tan is investigating the incidence of arrhythmias in Barth Syndrome patients. She is the co-Principal Investigator with Dr. Colin Phoon on the project, "Collaborative Registry to Determine the Natural History of Barth Syndrome." She is a pediatric cardiologist and pediatric electrophysiologist at New York University school of medicine – Hassenfeld Children's Hospital. She co-chairs the research committee in the division of pediatric cardiology. Dr. Tan is board certified in pediatrics, pediatric cardiology and pediatric electrophysiology.

Carolyn Taylor, Medical University of South Carolina

Dr. Taylor is a pediatric cardiologist with additional specialty training as a pediatric echocardiographer. Her research and clinical interests are in the areas of imaging and evaluation of cardiac function. Assessment of cardiac performance using echocardiography as well as evaluation of functional capacity in various forms of cardiomyopathy and cardio-skeletal myopathy are central to her clinical practice and research effort. She has been actively involved in the Barth syndrome scientific and research community since the early 2000s, and has authored and co-authored publications on cardiomyopathy and exercise capacity (through collaborations with Todd Cade), and conducted research utilizing on-site studies at the BSF conference and findings from the Barth Syndrome Registry.



Linh Vo, Wayne State University

Linh Vo is currently a Graduate Research Assistant in Dr. Miriam Greenberg's lab at Wayne State University. Her research project aims to elucidate the mechanisms underlying skeletal myopathy in Barth syndrome, a rare metabolic disease caused by abnormalities in the mitochondrial phospholipid cardiolipin. She believes that research and science are keys to a future with no Barth syndrome. Linh Vo holds a Master's degree in Medical Sciences from University of Tsukuba. She also holds a Bachelor's degree from University of Science – VNUHCM in Biotechnology.

Suya Wang, Boston Children's Hospital

Dr. Suya Wang is postdoctoral researcher in Dr. William Pu's laboratory at Boston's Children's Hospital. As an undergrad, she majored in Pharmacology at China Pharmaceutical University and was fascinated by how therapeutic agents change people's life. Dr. Wang later earned her PhD in Pharmacology and Toxicology from University of Kansas, where she started her journey exploring cardiac diseases. As a pharmacologist and biologist, Dr. Wang is interested in developing and optimizing novel therapeutic approaches to genetic disorders. To her, bio-medicine should not be just a research paper but a product that is safe, translatable and achievable in clinical practice.

Jessie Yester, Nationwide Children's Hospital

Jessie Yester completed her undergraduate education at North Carolina State University and her MD, PhD at Virginia Commonwealth University. She completed her Pediatric Cardiology Fellowship at UMPC Children's Hospital of Pittsburgh and Clinical Fellowship in Pediatric Heart Failure, Cardiomyopathy, and Transplantation at the Texas Children's Hospital. This fall, she will start as faculty at the Nationwide Children's Hospital. Her research interests include cardiotoxicity and determining the molecular pathways of cardiomyocyte proliferation and regeneration.

About Barth Syndrome Foundation

Barth Syndrome Foundation (barthsyndrome.org) is the only global network of families, healthcare providers, and researchers solely driven by the mission to save lives through education, advances in treatment and finding a cure for Barth syndrome. BSF has funded nearly $6.1M USD since 2002 and catalyzed over $32M in funding from other agencies to advance global scientific discoveries to end the suffering and loss of life from Barth syndrome. Additionally, BSF provides a lifeline to families and individuals living with Barth syndrome around the world, offering 24/7 individualized support, educational conferences, a robust patient registry and collaborations with specialist healthcare providers to define standards of care, treatment and rapid diagnosis.

Community Presenter

John Wilkins
Opening Comments for *Treatment and Quality of Life for Barth Syndrome*

As a longtime member of the community and an individual affected by Barth syndrome, Mr. John Wilkins brings a unique vantage point to the issues facing the BSF. Volunteering in every capacity, John has participated in numerous committees and research studies, as well as having served on the BSF Board of Directors. Mr. Wilkins earned an A.S. in Computer Information Technology from Southeast Community College in Lincoln, Nebraska, and works part time as a computer consultant. John lives in Lincoln, Nebraska

Session Chairs

Steven Claypool, Johns Hopkins University
Chair for *Cardiolipin in Physiology*

Dr. Steven Michael Claypool is professor of physiology and Genetic Medicine at the Johns Hopkins University School of Medicine and the director of the Cellular and Molecular Physiology graduate program. His research focuses on mitochondrial lipid metabolism and the contribution of phospholipids in mitochondrial function and dysfunction. Dr. Claypool received his B.A. in biological sciences and his M.A. in molecular, cellular and developmental biology from the University of California, Santa Barbara. He earned a Ph.D. in immunology from Harvard University and performed a postdoctoral fellowship in mitochondrial biology at University of California, Los Angeles. He has authored or co-authored several peer-reviewed publications, and has presented his work extensively.

Miriam Greenberg, Wayne State University
Chair for *Barth Syndrome Cardiomyopathy*

Dr. Greenberg is a Professor of Biological Sciences at Wayne State University in Detroit, MI. She is an expert on phospholipid metabolism and brings a wealth of knowledge about cardiolipin in yeast and mammalian cells that has and will continue to advance scientific knowledge about some of the underlying biochemical issues in Barth syndrome.

Grant Hatch, University of Manitoba
Chair for *Barth Syndrome Biology*

Dr. Hatch´s research interests focus on metabolism and pharmacological modulation of phospholipids (including cardiolipin) in the mammalian heart and cells in culture. He has published numerous papers on theses topics.

Riekelt Houtkooper, AMC University of Amsterdam
Chair for *Barth Syndrome in Physiology*

Dr. Riekelt H. Houtkooper received his PhD degree from the laboratory Genetic Metabolic Diseases of the Academic Medical Center Amsterdam (NL), working under supervision of Prof. Ronald Wanders and Dr. Frédéric Vaz. His research centered on cardiolipin metabolism, particularly in relation to the rare mitochondrial disorder Barth syndrome. Riekelt joined the lab of prof. Johan Auwerx at EPFL Lausanne (Switzerland) for a postdoctoral project geared towards understanding and treating more common metabolic diseases. During these years, he became interested in the metabolic aspects of aging. Early 2012 Riekelt moved back to Amsterdam to start his own group, for which he received funding from NWO and the ERC. Current research in the group focuses on molecular and translational metabolism, both in the context of inborn errors of metabolism and aging.

Erik Lontok, Director of Research, Barth Syndrome Foundation

Erik Lontok graduated with a PhD in Biochemistry from the University of California, San Francisco, and upon moving to the Maryland area, served as an Adjunct Professor of Biochemistry for the University of Maryland, College Park. As Director of Research, Erik is responsible for the Foundation's research grantmaking and contracts, while also serving as the scientific ambassador for BSF, facilitating collaborations amongst Barth syndrome researchers and external partners. Erik's passion is to engage, learn, and apply knowledge to advance disease research, with the ultimate goal of effective treatments for affected individuals.

Hilary Vernon, Johns Hopkins University and at the Kennedy Krieger Institute
Chair for *Treatment and Quality of Life for Barth Syndrome*

Dr. Vernon's research interests include understanding intermediary metabolism in Barth syndrome and in disorders of branch chain amino acid metabolism. Dr. Vernon is the director of the Mitochondrial Medicine Center at Johns Hopkins Hospital and the Barth Syndrome Interdisciplinary Clinic at the Kennedy Krieger Institute. She is also the co-director of the Department of Genetic Medicine Clinical Trials Unit at Johns Hopkins University School of Medicine. Dr. Vernon received her MD and PhD from Rutgers University, New Brunswick, NJ, USA. She completed residencies in Genetics and Pediatrics at Johns Hopkins University, and a fellowship in Clinical Laboratory Biochemical Genetics at Johns Hopkins University. She is board certified in Pediatrics, Clinical Genetics, and Clinical Laboratory Biochemical Genetics.

Help Support BSF

Since 2000, BSF has been a lifeline for those who suffer from Barth syndrome, offering 24/7 support, pioneering standards of care and diagnosis, creating collaborations between clinicians, researchers and patients, and most importantly, making sure no person with Barth syndrome is ever alone.

Your support is vital to the success of our mission, so please consider making a gift today.

Click Here to Donate Now.