# Barth Syndrome

## 7th International Scientific, Medical & Family Conference

Barth Syndrome Foundation®

- Clearwater, Florida • June 23-28 2014 -

# Dear Friends, Colleagues and First-Time Attendees,

Just like the boy in the poem who made a difference to that one starfish, **YOU** make a difference to the Barth syndrome community. Let me tell you how…

Moms and Dads, you make a difference through your unconditional love and unwavering commitment to helping your child. As they say, it takes a village, but having a son with Barth syndrome brings challenges that only other Barth parents can possibly understand. Discussing your experiences together, in one place, helps ease the feeling of isolation and answers questions about every issue imaginable. The family sessions cover topics from practical daily living tips, medications, social aspects, education, to the transition to adulthood, and more.

Researchers and clinicians, you make a difference through your commitment to understanding this rare and complex disorder. Interacting with the families at the conference brings new meaning to the expression, "from bench to bedside." The knowledge exchanged in the sessions has been described as energetic, exciting, innovative, and collaborative.

Donors, you make a difference through your investment in our cause. Your generous donations make all of this possible. Bringing families, clinicians, and researchers together for one conference helps us find the answers for treatments, and one day, a cure, much faster than working alone. We do not charge a registration fee, thanks to your overwhelming generosity.

Yes, all of you make a difference by your attendance, volunteer efforts, participation in scientific research, sponsorships, and everything that you do to help us find answers. This Conference offers something for everyone. I promise you this; you will leave feeling inspired, because you have truly made a difference!

With excitement and appreciation,

Lindsay Groff

## KEYNOTE ADDRESS

## Orphan Product Development in the Era of Personalized Medicine

## Thursday, June 26, 2014

(Salon D, E, F)

**Barry J. Byrne, MD, PhD — Associate Chair and Professor of Pediatrics and Molecular Genetics and Microbiology, College of Medicine, Department of Pediatrics; Molecular Genetics and Microbiology; Director of the Powell Gene Therapy Center at the University of Florida, Gainesville, FL, USA**

The stellar physician-scientist, Barry J. Byrne, MD, PhD will be the keynote speaker at the 7th International Scientific, Medical & Family Conference on Barth syndrome. Renowned as a pediatric cardiologist and for his efforts to find treatments for several rare diseases, Dr. Byrne is a major voice and champion for that underserved community. The Barth Syndrome Foundation (BSF) is fortunate to count on him as a close friend and an important advisor over the years. He served on the Scientific and Medical Advisory Board of BSF, has received a research grant from BSF, has cared for many Barth syndrome patients, and has been a defining presence at the BSF biennial conferences. Dr. Byrne's laboratory is focused on molecular approaches to diagnosis and treatment of heart failure in infants and children which includes Barth syndrome. Dr. Byrne studies glycogen storage diseases (Pompe disease), muscular dystrophies (Duchenne), hemophilias, as well as Barth syndrome, where he uses viral vectors (genetic therapy) in conjunction with stem cells to repair damaged hearts. These programs are supported by many prestigious organizations including the American Heart Association, the Muscular Dystrophy Association, and the National Institutes of Health (NHLBI, NIDDK, and NCRR). He is frequently called upon to advise the NIH. Dr. Byrne was one of the first researchers to publish on his work with the mouse model of Barth syndrome for which he received a BSF Research Grant in 2010. Most importantly, Dr. Byrne is a wonderful physician who gives of himself to the individuals he cares for — in fact, he even climbs mountains for them! We are very excited to hear Barry as he tells us about the "shape of things to come" in molecular medicine.

# PRE-CONFERENCE SESSIONS

| | SUNDAY, JUNE 22, 2014 |
|---|---|
| **Afternoon (Citrus)** | **PORTRAITS BY AMANDA CLARK**<br>Early family arrivals scheduled on this date |
| **Late afternoon (Grand Ballroom Foyer)** | **REGISTRATION**<br>Pick up badges / Drop items for family "goody bags" |
| **TIME/LOCATION/FACILITATORS** | **MONDAY, JUNE 23, 2014** |
| **All Day (Grand Ballroom Foyer)**<br><br>**All Day (Citrus)** | **REGISTRATION**<br>Pick up badges / Drop items for family "goody bags"<br><br>**PORTRAITS BY AMANDA CLARK**<br>Early family arrivals scheduled on this date |
| **12:00pm—1:30pm (Mangrove)**<br>*Stephanie Rader, Oliver Baxter-Smith & Nicole Derusha-Mackey* | **TRANSPLANT FAMILIES GATHERING**<br><br>*Audience: Parents and youth who have received a heart transplant* |
| **2:00pm—3:00pm (Salon D, E, F)**<br>*Shelley Bowen* | **NEW FAMILY ORIENTATION**<br>**CONFERENCE 101**<br><br>*Audience: First-Time Family Attendees* |
| **3:00pm—3:30pm (Sandpiper Deck)** | **GROUP PHOTO: ALL AFFECTED INDIVIDUALS** |
| **3:30pm—5:00pm (Salon D, E, F)**<br>*Shelley Bowen* | **CONSENT AND ASSENT SIGNING**<br><br>NOTE: The following groups of individuals participating in clinics must attend this session:<br><br>**CONSENTS**<br>Parents of all boys under the age of 18<br>Adult males 18 and older<br><br>**ASSENTS**<br>Minor affected males (12 – 18 years of age) |
| **5:00pm—7:00pm** | **DINNER ON YOUR OWN** |
| **7:00pm—9:00pm (Salon D, E, F)**<br>*Lindsay Groff, Marc Sernel & Shelley Bowen*<br><br>**7:00 pm—8:00 pm**<br><br>**8:00 pm—9:00 pm**<br>*Shelley Bowen* | **WELCOME EVENT**<br><br>**FAMILY INTRODUCTIONS**<br>*Audience: All family attendees (Children, Parents, Grandparents, etc.)*<br><br>**CONFERENCE 101** (Overview of conference and important reminders) |
| **8:00pm—9:00pm (Salon A)**<br>*Brie Chandler-Kalapasev*<br><br>**8:00pm—9:00pm**<br>**(Gathering in Grand Ballroom Foyer)**<br>*Leslie Buddemeyer & Julie Fairchild* | **LITTLE TYKE MOVIE NIGHT**<br><br>*Audience: Children through four years of age*<br><br>Note: One adult family member must accompany children who are not potty trained.<br><br>**TREASURE HUNT**<br><br>*Audience: Children five years of age and up* |

# BARTH SYNDROME CLINICS & CONSULTATIONS
# TUESDAY & WEDNESDAY
# JUNE 24-25, 2014

**7:30am—5:30pm**

(Grand Ballroom Foyer - Clinic Registration Desk)

**NOTE: WAITING AREA FOR CLINICS WILL BE IN MANDALAY**

Two days of Barth clinics where families and clinicians share and learn valuable information about the clinical aspects of Barth syndrome. This exchange between families and clinicians about the many nuances of Barth syndrome gives rise to opportunities to explore new ideas and discuss issues of common theme. The BSF clinics have been hailed as a model approach by other health advocacy groups. (Arts, crafts, movies, outdoor games and Wii activities will be available for all in Mandalay and Executive Conference Rooms.)

## RESEARCH STUDIES

### MULTI-DISCIPLINARY STUDIES IN BARTH SYNDROME (Multiple Meeting Rooms — See below)

- **Hilary Vernon, MD, PhD**, Principal Investigator, McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University; Kennedy Krieger Institute, Baltimore, MD
- Brittany DeCroes, DPT, Kennedy Krieger Institute, Baltimore, MD **(six-minute walk test — Salon G)**
- Richard I. Kelley, MD, PhD, Co-Investigator, Johns Hopkins University School of Medicine; Kennedy Krieger Institute, Baltimore, MD **(Tarpon)**
- Rebecca L. McClellan, MGC, Co-Investigator, Kennedy Krieger Institute, Baltimore, MD **(Manatee**)
- Yana Sandlers, MSc, PhD, Co-Investigator, Kennedy Krieger Institute, Johns Hopkins School of Medicine, Baltimore, MD
- W. Reid Thompson, MD, Co-Investigator, Johns Hopkins Children's Center, Baltimore, MD **(echos — Dolphin)**
- Gul Dadlani, MD, Medical Director, Pediatric Cardiology and Pediatric Cardiology Laboratory Director, All Children's Hospital/Johns Hopkins Medicine, St. Petersburg, FL
- Kathyrn Douglas, RDCS, CCT, Chief Pediatric Cardiac Sonographer, All Children's Hospital/Johns Hopkins Medicine, St. Petersburg, FL
- Shawn Cupp, RDCS, CCT, Pediatric Cardiac Sonographer, All Children's Hospital/Johns Hopkins Medicine, St. Petersburg, FL

### PSYCHOSOCIAL FUNCTIONING IN BARTH SYNDROME (Agave Office Suite & Salon D)

- **Marni L. Jacob, PhD**, Principal Investigator, Rothman Center for Neuropsychiatry, University of South Florida, Tampa, FL
- Amanda Collier, BA, Co-Investigator
- Brittany Dane, BS, Co-Investigator, Rothman Center for Neuropsychiatry, University of South Florida, Tampa, FL

### A SYSTEMATIC INVESTIGATION INTO SENSORY AND MOTOR-BASED FEEDING ISSUES IN BOYS WITH BARTH SYNDROME (Marlin)

- **Stacey Reynolds, PhD, OTR/L**, Principal Investigator, University of Florida, Gainesville, FL; Virginia Commonwealth University, Richmond, VA
- Emily Burgess, OTS, Research Assistant
- Shelly Lane, PhD, OTR/L, FAOTA, Co-Investigator, Professor, Department of Occupational Therapy, Virginia Commonwealth University; Director, Sensory Integration and Stress Evaluation (SPASE) Lab; Director, Post Professional Education for the Department of Occupational Therapy, Virginia Commonwealth University, Richmond, VA

### THE IMPACT OF A CHILD'S DISABILITY ON THE PARENTS OF CHILDREN WITH RARE DISEASES (Salon F)

- **William Mann, PhD, OTR/L**, Principal Investigator, Distinguished Professor and Chair of Occupational Therapy, Director of the PhD Program in Rehabilitation Science, University of Florida (UF), Director of the UF Center for Telehealth and Healthcare Communications, Gainesville, FL
- YuYun Huang, MS, OTR, Doctoral Student
- Consuelo Kreider, PhD, OTR/L, College of Public Health and Health Related Professions, University of Florida, Gainesville, FL
- Yoonjeong Lim, MS, OT, Doctoral Student

### SEVERE CHRONIC NEUTROPENIA INTERNATIONAL REGISTRY (Agave — web-based)

- Audrey Anna Bolyard, RN, University of Washington, The Severe Chronic Neutropenia International Registry, Seattle, WA

## CONSULTATIONS

### GENETICS (Manatee)

- Iris L. Gonzalez, PhD, A. I. duPont Hospital for Children, Wilmington, DE (retired)
- Rebecca L. McClellan, MGC, Kennedy Krieger Institute, Baltimore, MD

### METABOLISM & NUTRITION (Tarpon)

- Richard I. Kelley, MD, PhD, Johns Hopkins University School of Medicine; Kennedy Krieger Institute; Baltimore, MD
- Nicol Clayton, Specialist Dietician, NHS Barth Syndrome National Service, Bristol Royal Hospital for Children, Bristol, United Kingdom

### VITAL SIGNS (Salon G)

- Susan V. Wilkins, RN (former BSF Board member)
- Debbie Riddiford, CNS, Bristol Royal Hospital for Children, Bristol, United Kingdom
- Donna Strain, RN

### LABS (Coral)

# TUESDAY, JUNE 24, 2014

| SMALL GROUP MEETINGS | |
|---|---|
| **TIME/LOCATION/FACILITATORS** | **MEETING** |
| **All Day (Grand Ballroom Foyer)** | **REGISTRATION**<br>Pick up badges / Drop items for family "goody bags" |
| **12:00pm–1:00pm (Mangrove)** | **CARRIER DISCUSSIONS** (*Audience: Carriers and potential carriers age 15+)* |
| *Rebecca L. McClellan, MGC & Lee Kugelmann* | Join us to explore the broad and varied issues faced by women who are, or who might be, carriers of Barth syndrome. All ages and situations welcome. The goal of this session will be to introduce you to the issues and get everyone thinking about how being a carrier affects you personally and how BSF can help. We'll use this session to help shape our small groups sessions and programming for the carriers over the rest of the meeting. |
| **12:00pm–1:00pm (Water's Edge C)** | **MEN OF BARTH MEETING** (*Audience: Affected individuals age 16+)* |
| *B.J. Develle, MSW, Marni Jacob, PhD, & John Wilkins* | Join us to explore issues faced by young men who have Barth syndrome. The goal of this session will be to introduce you to the issues and get everyone thinking about how having Barth syndrome affects you personally, how BSF can help, and how you can get more involved in helping others who have Barth syndrome. We'll use this session to help shape our small groups sessions and programming for affected individuals over the rest of the meeting. Meet and greet, reacquaint with old friends and meet new friends. |
| **1:00pm—1:45pm (Water's Edge C)** | **GENERAL INQUIRIES** *(Audience: Affected individuals age 16+)* |
| *Richard I. Kelley, MD, PhD* | This small group session is designed for affected individuals age 16 and older to address concerns/issues related to metabolic aspects of Barth syndrome. |
| **1:00pm—3:00pm (Salon C)** | **FAMILY & FRIENDS CPR COURSE**<br>*(Audience: All conference attendees ages 16+)* |
| *SunStar Emergency Medical Services* | The Family and Friends CPR Course teaches the lifesaving skills of adult, child and infant CPR, AED use, and relief of choking in an adult, child or infant. This course is especially useful for family members of those who have Barth syndrome. There is NO certification card with this course. |
| **3:00pm—4:00pm (Mangrove)** | **CARRIER ISSUES SESSION I : PRE-TEST CARRIER ISSUES**<br>(*Audience: Carriers and potential carriers age 15+)* |
| *Rebecca L. McClellan, MGC & Lee Kugelmann* | This session will focus on making a decision about genetic carrier testing — "Am I ready?" What's involved with testing and things to think about before getting tested. |
| **4:00pm—5:00pm (Mangrove)** | **CARRIER ISSUES SESSION II : POST-TEST CARRIER ISSUES**<br>*(Audience: Carrier women who have recently undergone testing)* |
| *Rebecca L. McClellan, MGC & Lee Kugelmann* | This session will focus on exploring the impact of learning that you're a carrier for Barth syndrome. How might this impact relationships, future plans, reproductive options, etc. |
| **5:00pm—7:30pm** | **DINNER ON YOUR OWN** |
| **8:00pm—9:00pm (Beach)** | **SUNSET AND STARGAZING** |
| *Kate Michener* | Gather on the beach to view the beautiful sunset and gaze at the stars. |

# WEDNESDAY, JUNE 25, 2014

| SMALL GROUP MEETINGS | |
|---|---|
| **TIME/LOCATION/FACILITATORS** | **MEETING** |
| **All Day (Grand Ballroom Foyer)** | **REGISTRATION**<br>Pick up badges / Drop items for family "goody bags" |
| **9:00am—10:30am (Salon E)** | **WHAT YOU NEED TO KNOW ABOUT BARTH SYNDROME BUT WERE AFRAID TO ASK** (*Audience: All conference attendees)* |
| *Facilitator: Hilary Vernon, MD, PhD*<br>*OT: Consuelo Kreider, MHS OTR/L*<br>*PT: Todd Cade, PT, PhD*<br>*Nutrition: Nicol Clayton, Specialist Dietitian*<br>*Psychologist: Vanessa Garratt, DClinPsych*<br>*Biochemical Aspects: Richard I. Kelley, MD, PhD*<br>*Geneticist: Rebecca L. McClellan, MGC*<br>*Coordination of Care: Debbie Riddiford, CNS*<br>*Hematology: Colin G. Steward, PhD, FRCP, FRCPCH* | This is a session even the most seasoned parent won't want to miss. A multidisciplinary team of healthcare providers will provide a primer about the various clinical aspects of Barth syndrome, the roles of sub-specialty healthcare providers and therapists, and how these team members enhance the care of those who have Barth syndrome as well as their family members. |
| **10:30am—11:15am (Salon A)**<br>**11:15am—12:00pm** | **RESEARCH UPDATE**<br>*(Audience: Parents/Grandparents and youth age 16+)* |
| *Matthew J. Toth, PhD* | This question and answer session will provide an overview about research advances that have been made in Barth syndrome.<br>*Two sessions are offered to accommodate optimal group size.* |
| **10:30am—11:15am (Salon B)**<br>**11:15am—12:00pm** | **NEUTROPENIA SMALL GROUP**<br>*(Audience: Affected individuals age 16+)* |
| *Colin G. Steward, PhD, FRCP, FRCPCH* | This question and answer session will address concerns and/or issues pertaining to the hematologic aspects of Barth syndrome.<br>*Two sessions are offered to accommodate optimal group size.* |
| **1:00pm—2:00pm (Salon C)** | **GOVERNING PRIORITIES TO PROPEL THE MISSION**<br>*(Audience: Parents/Grandparents and youth age 16+)* |
| *Susan Wilkins (former Board Member)* | This informal session will introduce the governing body (aka the Board of Directors) of the Barth Syndrome Foundation, provide an overview about the governing instruments of the organization, strategic priorities, and engage all stakeholders in a general question and answer session. |
| **2:00pm—3:30pm (Salon C)** | **VOLUNTEERING TO PROPEL THE MISSION**<br>*(Audience: Parents/Grandparents and youth age 16+)* |
| *Lindsay Groff & Sandra Stevens* | This informal session is designed to generate ideas, answer questions, and discuss opportunities for volunteers to become more involved in BSF through: grassroots fundraising, increasing awareness, and active volunteerism. |
| **3:30pm—5:00pm (Salon A)** | **TRANSITIONS: TAKING CHARGE OF MY HEALTH AND WELL-BEING AND MENTORING OTHERS WITH BARTH SYNDROME TO DO THE SAME**<br>*(Audience: Affected individuals age 16+)* |
| *BJ Develle, MSW & John Wilkins*<br>*Panel: Matthew Toth, PhD; Todd Cade, PT PhD;*<br>*Marni Jacob, PhD, & Randall Bryant, MD* | This informal session will discuss concerns about living with Barth syndrome, becoming a self-advocate and thoughts about how to encourage the younger boys who have been diagnosed with Barth syndrome. |
| **3:30pm—4:30pm (Salon B)**<br>**4:30pm—5:30pm** | **GENERAL INQUIRIES SMALL GROUP DISCUSSION**<br>*(Audience: All conference attendees)* |
| *Richard I. Kelley, MD, PhD & Nicol Clayton, Specialist Dietician* | This question and answer session will address concerns and/or issues pertaining to the nutrition and metabolic aspects of Barth syndrome.<br>*Two sessions are offered to accommodate optimal group size.* |
| **5:30pm—7:00pm** | **DINNER ON YOUR OWN** |
| **6:00pm—8:00pm (Offsite)** | **CLINIC DINNER *(by invitation)*** |
| **7:00pm—10:00pm (Sandpiper Deck)** | **BEACHSIDE GATHERING w/ DJ CAMO** |
| **9:00pm (Beach)** | **LUMINARIES ON THE BEACH** |
| Luminaries will light the beach to honor those who are living with Barth syndrome and to remember those who have passed away. | |

# CHILDCARE ACTIVITIES

## Thursday – Saturday

**June 26—28, 2014**

(Mangrove)

Childcare is offered for children through age 7 so that parents can attend educational sessions. Children will enjoy fun, age appropriate activities listed below. Please be sure to provide contact information to our volunteer childcare providers. Parents will be contacted for diaper changes and any other needs.

| THURSDAY, JUNE 26, 2014 (Mangrove) | | FRIDAY, JUNE 27, 2014 (Mangrove) | |
|---|---|---|---|
| 8:30am—9:30am | I'm this big art project | 9:15am—10:30am | Footprint art |
| 9:30am—11:00am | Fish handprint painting | 10:30am—11:00am | Clean up |
| 11:00am—12:00pm | Clean up | 11:00am—12:00pm | Free play |
| 12:00pm—1:30pm | Lunch | 12:00pm—1:30pm | Lunch |
| 1:30pm—2:00pm | Group photo | 1:30pm—3:00pm | Arts and crafts activities |
| 2:00pm—3:30pm | Paint terra cotta pots | 3:00pm—4:30pm | Superhero pictures |
| 3:30pm—4:00pm | Clean up | 4:40pm—5:00pm | Clean up |
| 4:00pm—4:45pm | Activity time with the big kids, cupcake making, caricatures, and thank you cards for our donors | | |
| 4:45pm—5:00pm | Clean up | | |

| SATURDAY, JUNE 28 (Mangrove) | |
|---|---|
| 9:00am—11:40am | My Barth Buddies memory book |
| 11:40am—12:00pm | Clean up |
| 1:30pm—3:30pm | Movie time |

## SCIENCE & MEDICINE SESSIONS
## Thursday, June 26, 2014

The Science & Medicine sessions are designed for doctors and scientists involved in the many aspects of Barth syndrome to discuss the latest underlying scientific developments and clinical insights. It is a unique experience that encourages collaboration and accelerates advances.

**7:30am—8:15am (Salon D, E, F)**

| | |
|---|---|
| 7:30am—8:15am | **Breakfast** for all conference attendees |

**8:30am—11:50am (Salon G)**
**MITOCHONDRIAL LIPIDS**
*Chair—Michael Schlame, MD, New York University School of Medicine; New York University Langone Medical Center, New York, NY*

| | |
|---|---|
| 8:30am—8:35am | Introduction — Ryan Ritter |
| 8:35am—9:05am | **Rancid Radical Talk: Music of Mitochondrial Cardiolipins** — *Valerie Kagan, PhD, DSc, University of Pittsburgh, Pittsburgh, PA* |
| 9:05am—9:35am | **Deletion of the Cardiolipin-specific Phospholipase Rescues Growth Defects in the Yeast Tafazzin Mutant** — *Miriam Greenberg, PhD, Wayne State University, Detroit, MI* |
| 9:35am—10:05am | **Could Other Tafazzin Products Contribute to Heart and Muscle Pathology in Barth Syndrome?** — *Matthew Gillum, PhD, University of Iowa Carver College of Medicine, Iowa City, IA* |
| 10:05am—10:20am | Coffee Break |
| 10:20am—10:50am | **Unremodeled and Remodeled Cardiolipin are Functionally Indistinguishable in Yeast** — *Matthew Baile, PhD, Johns Hopkins School of Medicine, Baltimore, MD* |
| 10:50am—11:20am | **Detection of Cardiolipin Abnormalities in White Blood Cells of Patients with Barth Syndrome by MALDI-TOF/MS** — *Angela Corcelli, PhD, University of Bari Aldo Moro, Bari, Italy* |
| 11:20am—11:50am | **Regulation of Cardiomyopathy in Barth Syndrome by ALCAT1** — *Jun Zhang, PhD Candidate, Penn State College of Medicine, State College, PA* |

| | |
|---|---|
| 12:00pm—1:30pm | **Keynote Speaker Luncheon (Salon D, E, F)**<br>**Barry J. Byrne, MD, PhD — Orphan Product Development in the Era of Personalized Medicine**<br>*(Introduction by R.J. Kugelmann)* |
| 1:30pm—2:00pm | **Scientific & Medical Attendees Group Photo (Sandpiper Deck) / All Conference Attendee Group Photo (Beach)** |

**2:00pm—5:50pm (Salon D, E, F)**
**CLINICAL STUDIES ON BARTH SYNDROME**
*Chair—Arnold Strauss, MD, University of Cincinnati College of Medicine; Cincinnati Children's Research Foundation; Cincinnati Children's Hospital Medical Center, Cincinnati, OH*

| | |
|---|---|
| 2:00pm—2:05pm | Introduction — Brian & Rachel Moreland |
| 2:05pm—2:35pm | **The National Health Service Barth Syndrome Service: A Progress Report** — *Colin G. Steward, PhD, FRCP, FRCPCH, Bristol Royal Hospital for Children, Bristol, United Kingdom* |
| 2:35pm—3:05pm | **Barth Syndrome Without Severe Cardiolipin Deficiency: A Possible Ameliorated Phenotype** — *Ann Bowron, FRCPath, Bristol Royal Infirmary, Bristol, United Kingdom* |
| 3:05pm—3:35pm | **The Long Way to Evidence-based Medicine for Barth Syndrome: Experience from France** — *Jean Donadieu, MD, PhD, Trousseau University Hospital, Paris, France* |
| 3:35pm—3:50pm | Coffee Break |
| 3:50pm—4:20pm | **Exercise and Substrate Metabolism Studies in Barth Syndrome: An Update** — *W. Todd Cade, PT, PhD, Washington University School of Medicine, St. Louis, MO* |
| 4:20pm—4:50pm | **Kennedy Krieger Barth Syndrome Initiatives: Results from the 2012 BSF Conference Clinical Laboratory Study and Overview of the Barth Syndrome Interdisciplinary Clinic** — *Hilary Vernon, MD, PhD, Johns Hopkins University and the Kennedy Krieger Institute, Baltimore, MD* |
| 4:50pm—5:20pm | **Taste Sensitivity, Food Preference, and Feeding Behaviors in Children with Barth Syndrome** — *Stacey Reynolds, PhD, OTR/L, University of Florida, Gainesville, FL; Virginia Commonwealth University, Richmond, VA* |
| 5:20pm—5:50pm | **Diagnosis and Management of Cardiovascular Disease in Barth Syndrome** — *John L. Jefferies, MD, MPH, FACC, Heart Institute, Cincinnati Children's Hospital Medical Center, Cincinnati, OH* |

**6:00pm—8:00pm — POSTER SESSION (Grand Ballroom Foyer)**

| | |
|---|---|
| 6:00pm—8:00pm | **Poster Session** (Science & Medicine Attendees) |

## FAMILY SESSIONS
## Thursday, June 26, 2014

The Family sessions are designed to provide the latest information to family members 16 years of age and older.

**7:30am—8:15am (Salon D, E, F)**

| | |
|---|---|
| 7:30am—8:15am | **Breakfast** for all conference attendees |

**8:30am—9:30am (Salon A, B, C)**
**EXERCISE IN BARTH SYNDROME**

| | |
|---|---|
| 8:30am—8:40am | Introduction — Ghent Lummis |
| 8:40am— 9:00am | *Todd Cade, PhD, Washington University School of Medicine, St. Louis, MO* |
| 9:00am—9:20am | *Barry Byrne, MD, PhD, Powell Gene Therapy Center at the University of Florida, Gainesville, FL* |
| 9:20am—9:30am | **Q & A** |

**9:30am—10:20am (Salon A, B, C)**
**NUTRITION IN BARTH SYNDROME**

| | |
|---|---|
| 9:30am—9:50am | **The Bristol Experience** — *Nicol Clayton, Specialist Dietitian, NHS Barth Syndrome National Service, Bristol Royal Hospital for Children, Bristol, United Kingdom* |
| 9:50am—10:10am | **Metabolic Aspects of Barth Syndrome and Nutritional Supplements** — *Richard I. Kelley, MD, PhD, Johns Hopkins University School of Medicine; Kennedy Krieger Institute, Baltimore, MD* |
| 10:10am—10:20am | **Q & A** |

**10:20am—11:00am (Salon A, B, C)**
**BREAKING NEWS**

| | |
|---|---|
| 10:20am—10:30am | Introduction: Alanna Layton |
| 10:30am—10:50am | **Heart-on-chip Breakthrough** — *William Pu, MD, Boston Children's Hospital, Boston, MA* |
| 10:50am—11:00am | **Q & A** |

**11:00am—11:20am (Salon A, B, C)**
**BSF REGISTRY 2.0**

| | |
|---|---|
| 11:00am—11:15am | **BSF REGISTRY 2.0** — *Shelley Bowen* |
| 11:15am—11:20am | **Q & A** |

**11:20am—12:00pm (Salon A, B, C)**
**OVERVIEW OF CARRIER ISSUES: SESSION III**

| | |
|---|---|
| 11:20am—11:30am | Introduction — Susan McCormack |
| 11:30am—11:50am | *Rebecca L. McClellan, MGC, Kennedy Krieger Institute, Baltimore, MD & Lee Kugelmann* |
| 11:50am—12:00pm | **Q & A** |

| | |
|---|---|
| 12:00pm—1:30pm | **Keynote Speaker Luncheon (Salon D, E, F)**<br>**Barry J. Byrne, MD, PhD — Orphan Product Development in the Era of Personalized Medicine**<br>*(Introduction by R.J. Kugelmann)* |
| 1:30pm—2:00pm | **Scientific & Medical Attendees Group Photo (Sandpiper Deck) / All Conference Attendee Group Photo (Beach)** |

**2:00pm—5:50pm (Salon D, E, F)**
**CLINICAL STUDIES ON BARTH SYNDROME**

| | |
|---|---|
| 2:00pm—5:45pm | There are no scheduled "Family Sessions" at this time. Families are invited and encouraged to attend the Science & Medicine sessions on **Clinical Studies on Barth Syndrome** (see page 8) |

**7:00pm—8:00pm (Grand Ballroom Foyer)**
**POSTER SESSION**

| | |
|---|---|
| 7:00pm—8:00pm | **Poster Session** (Families invited to attend Poster Session) |

## AFFECTED INDIVIDUALS and SIBLINGS SESSIONS
## Thursday, June 26, 2014

Join your fellow youth for fun and fellowship while learning about the issues that concern young people affected by Barth syndrome.

**7:30am—8:15am (Salon D, E, F)**

| | |
|---|---|
| 7:30am—8:15am | **Breakfast** for all conference attendees |

**8:30am—9:45am (Mandalay)**
**MEET YOUR GROUP LEADER** ~ All Affected Individuals and Siblings

**9:45am—10:30am**

| | |
|---|---|
| **Affected Individuals (Tarpon)**<br>*Audience: Affected individuals age 10+*<br><br>**Barth Syndrome Issues: Self-Advocacy**<br>*BJ Develle, MSW & Marni Jacob, PhD*<br><br>Knowing Barth syndrome and knowing when things aren't right. | **Siblings (Manatee)**<br>*Audience: Girls between ages of 10-14 who are at risk to be carriers for Barth syndrome and are interested in learning more about it.*<br><br>**Carrier Issues: Session IV**<br>*Rebecca L. McClellan, MGC & Lee Kugelmann*<br><br>This session will focus on introducing the notion of being a carrier with the goal of helping our young ladies understand their risks and begin to think about its implications and impact on their lives. |

**10:45am—12:00pm (Mandalay)**
**PHOTO SCAVENGER HUNT** *(Combined Youth)*

| | |
|---|---|
| 12:00pm—1:30pm | **Keynote Speaker Luncheon (Salon D, E, F)**<br>**Barry J. Byrne, MD, PhD — Orphan Product Development in the Era of Personalized Medicine**<br>*(Introduction by R.J. Kugelmann)* |
| 1:30pm—2:00pm | **Scientific & Medical Attendees Group Photo (Sandpiper Deck) / All Conference Attendee Group Photo (Beach)** |

**1:45pm—5:00pm (Mandalay)**
**ACTIVITY TIME *(Combined Youth)***
Activities include cupcake making, caricatures, donor thank you cards, ping-pong, Wii games and other awesome activities.

**7:00pm-8:00pm (Grand Ballroom Foyer)**
**POSTER SESSION**

| | |
|---|---|
| 7:00pm—8:00pm | **Poster Session** (Families invited to attend) |

# POSTER SESSION

## THURSDAY, JUNE 26, 2014

*(Grand Ballroom Foyer)*

Sponsored by Barth Syndrome Foundation of Canada

For 2014, we have an extraordinarily large and quality collection of poster presentations. Four poster presenters will be chosen to talk about their results to the entire Science and Medicine audience on Friday morning, June 27th. The Poster session is a perfect opportunity to meet one-on-one with many Barth syndrome researchers, so please take advantage of this opportunity.

| **6:00pm—8:00pm**: Physicians & Scientists | **7:00pm—8:00pm**: Families welcome |
|---|---|

POSTER 1:
**Mitochondrial Dynamics and the Selectivity of Mitophagic Processes**
Hagai Abeliovich, PhD : Department Biochemistry, Food Science and Nutrition, Hebrew University of Jerusalem, Rehovot, Israel

POSTER 2:
**MALDI-TOF-MS Lipid Fingerprint of Leukocytes as a Tool for the Screening of Barth Syndrome**
Roberto Angelini, PhD*, Lobasso S.*, Bowron A.°, Steward C.G.^, Corcelli A* : *Dept. of Basic Medical Sciences, Neurosciences and Sensory organs, University of Bari "Aldo Moro", Bari, Italy; ° Department of Clinical Biochemistry, Bristol Royal Infirmary, University Hospitals Bristol, NHS Foundation Trust, Bristol, UK; ^ Clinical Lead, NHS Specialised Services Barth Syndrome Service, Bristol Royal Hospital for Children, Bristol, UK

POSTER 3:
**Mitochondrial Structure in Barth Syndrome: A Comparison of Fresh Lymphocytes and Lymphoblast Cell Lines by Electron Microscopy of Cryofixed Cells**
Ann Bowron, FRCPath[1,3], Paul Verkade[2], Judith Mantell[2], Gini Tilly[2], Sarah Groves[3], Paul Thomas[1,3], Simon Heales[4], Colin Steward[3,5] : [1]Dept of Clinical Biochemistry, Bristol Royal Infirmary; [2]Wolfson Bioimaging Facility, Dept of Biochemistry, University of Bristol; [3]School of Cellular and Molecular Medicine, University of Bristol; [4]Dept of Clinical Biochemistry, Great Ormond Street Hospital for Children, London; [5]Department of Paediatric Haematology and Oncology, Bristol Royal Hospital for Children, Bristol, UK

POSTER 4:
**Chemical Taste Sensitivity and Food Preferences in Boys with and without Barth Syndrome**
M. Emily Burgess, OTS, Virginia Commonwealth University, Richmond, VI Contributing Authors: Stacey Reynolds PhD, OTR/L; Lauren E. Meeley, OTS; Kristi South, OTS; Samantha Hearn, OTS; Andrenne Alsum, OTS

POSTER 5:
**Feeding Problems in Barth Syndrome: The UK Perspective**
Nicol Clayton, Specialist Pediatric Dietician; Colin Steward, Clinical Lead; Debbie Riddiford, Specialist Pediatric Nurse Barth Syndrome : NHS Specialized Service, United Kingdom

POSTER 6:
**Clinical 6 Minute Walk Test Use in Patients with Barth Syndrome**
Brittany DeCroes, PT, DPT : Kennedy Krieger Institute, Baltimore, MD, USA

POSTER 7:
**Enzyme Replacement Therapy in Mammalian Models of Barth Syndrome**
Ana Dinca, Michael Chin, MD, PhD : University of Washington, Seattle, WA, USA

POSTER 8:
**Bi-Ventricular Assist Device as a Bridge to Cardiac Transplantation in Cardiomyopathy and Worsening Heart Failure Due to Barth Syndrome**
Vernat J. Exil, MD, Bret Allen Mettler : Monroe Carell Jr. Children's Hospital, Vanderbilt University, Nashville, TN, USA

POSTER 9:
**Is Barth Syndrome a Disorder of Defective Plasma Membrane Repair?**
Michelle Gorecki, Matthew Gillum, PhD : University of Iowa, Iowa City, IA, USA

POSTER 10:
**Loss of ACSL1 Impairs Mitochondrial Function and Decreases Tetralinoleoyl Cardiolipin**
Trisha J. Grevengoed[1], Sarah A. Martin[3], Lalage Katunga[2], Ethan J. Anderson[2], Robert C. Murphy[3], Rosalind A. Coleman[1] : [1]University of North Carolina at Chapel Hill, Department of Nutrition; [2]University of Colorado, Denver, Dept. of Pharmacology; [3]East Carolina University, Department of Pharmacology and Toxicology

POSTER 11:

**Monolysocardiolipin Acyltransferase-1 Expression Improves Mitochondrial Function**

Grant M. Hatch, PhD, Edgard M. Mejia, William A. Taylor, Patrick C. Choy, Genevieve C. Sparagna : Department of Pharmacology & Therapeutics, University of Manitoba, Winnipeg, Manitoba, Canada, and the University of Colorado at Boulder, Denver, CO, USA

POSTER 12:

**Differences in Extracurricular Activity Participation between Children With and Without Barth Syndrome**

Yu-Yun Huang, MS[1], Consuelo Maun Kreider, PhD[1], Yoonjeong Lim, MS[1], & Roxanna Marie Bendixen, PhD[2] : [1]Department of Occupational Therapy, College of Public Health & Health Professions, University of Florida. [2]Department of Occupational Therapy, School of Health and Rehabilitation Sciences, University of Pittsburgh, Pittsburgh, PA, USA

POSTER 13:

**Overexpression of CL Phospholipase *CLD1* Leads to Loss of Mitochondrial DNA that is Rescued by Deletion of Mitochondrial Fusion Gene *FZO1***

Yiran Li, Cunqi Ye, Michael McCaffrey, and Miriam L. Greenberg : Wayne State University, Detroit, MI

POSTER 14:

**Health-related Quality of Life in Boys with Barth Syndrome: Using Both Child Self-reports and Parent Proxy-reports**

Yoonjeong Lim, MS, OT[1], Consuelo M. Kreider, PhD, OTR/L[1], Yu-Yun Huang, MS, OTR[1], Roxanna M. Bendixen, PhD, OTR/L[2] : [1]Department of Occupational Therapy, College of Public Health & Health Professions, University of Florida. [2]Department of Occupational Therapy, School of Health and Rehabilitation Sciences, University of Pittsburgh, Pittsburgh, PA, USA

POSTER 15:

**TCA Cycle Defects in Barth Syndrome**

Wenjia Lou : Wayne State University, Detroit, MI, USA

POSTER 16:

**Biochemical Characterization of Human and Murine *Tafazzin***

Ya-Wen Lu[1], Laura Galbraith[2], Eyal Gottlieb[2], Frederic M. Vaz[3], Steven M. Claypool[1] : [1]Department of Physiology, Johns Hopkins University School of Medicine. [2]Cancer Research UK, The Beatson Institute for Cancer Research. [3]Department of Clinical Chemistry and Department of Pediatrics, University of Amsterdam, Amsterdam, The Netherlands

POSTER 17:

**Monolysocardiolipin Acyltransferase-1: A Potential Therapeutic Agent for the Treatment of Barth Syndrome**

Edgard M. Mejia[1,4], William A. Taylor[1,4], Laura Cole[1,4], Grant M. Hatch[1,2,3,4] : [1]Department of Pharmacology & Therapeutics, University of Manitoba, [2]Department of Biochemistry and Medical Genetics, University of Manitoba, [3]Center for Research and Treatment of Atherosclerosis and [4]Manitoba Institute of Child Health, Winnipeg, Manitoba, Canada

POSTER 18:

**Exploring Health Information Technology User Needs Within the Barth Syndrome Community**

Melissa I. Naiman, PhD, PMP, EMT-B : Center for Advanced Design, Research, and Exploration (CADRE) and School of Public Health, University of Illinois at Chicago, Chicago, IL USA

POSTER 19:

**Analysis of Genes Implicated in Myocardial Noncompaction Cardiomyopathy: A Final Common Pathway?**

Sebastian Piombo, Leigh Nesheiwat, Mindong Ren, Colin K.L. Phoon : New York University School of Medicine, New York, NY, USA

POSTER 20:

**Cardiolipin Deficiency Leads to Decreased Acetyl CoA Levels and Metabolic Deficiencies**

Vaishnavi Raja, Miriam L. Greenberg : Wayne State University, Detroit, MI, USA

POSTER 21:

**The Significance of 3-Methylglutaconic Acid-uria in Barth Syndrome**

Betty Su, Robert O. Ryan, PhD : Children's Hospital Oakland Research Institute, Oakland, CA, USA

POSTER 22:

**Biochemical Abnormalities in Barth Syndrome Patients**

Yana Sandlers, PhD, Richard I. Kelley : Kennedy Krieger Institute, Division of Metabolism, Baltimore, MD, USA

POSTER 23:

**Cardiolipin Replacement for Barth Syndrome Associated Neutropenia**

Betty Su, Fong-Fu Hsu, Robert O. Ryan : Children's Hospital Oakland Research Institute, Oakland, CA, USA

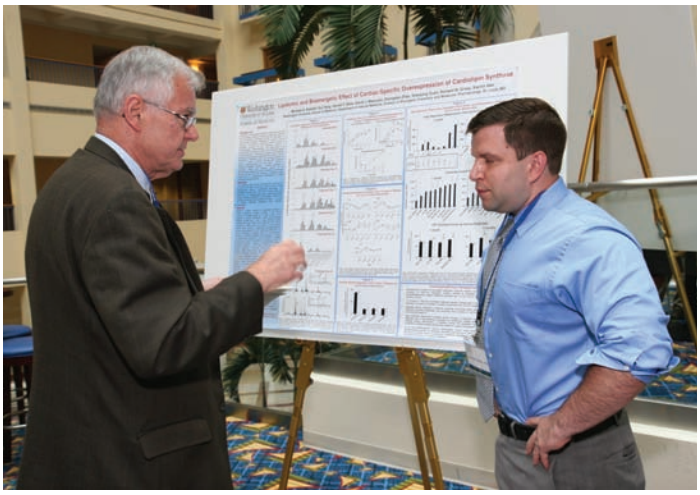
POSTER 24:

**Deletion of the Cardiolipin-specific Phospholipase *Cld1* Rescues Growth and Lifespan Defects in the *Tafazzin* Mutant: Implications for Barth Syndrome**

Cunqi Ye[1], Wenjia Lou[1], Yiran Li[1], Iliana A. Chatzispyrou[5], Maik Hüttemann[2,3], Icksoo Lee[4], Riekelt H.Houtkooper[5], Frédéric M. Vaz[5], Shuliang Chen[1*], Miriam L. Greenberg[1] : [1]Department of Biological Sciences, Wayne State University, Detroit, MI; [2]Center for Molecular Medicine and Genetics and [3]Cardiovascular Research Institute, Wayne State University School of Medicine, Detroit, MI; [4]College of Medicine, Dankook University, Cheonan-si, Chungcheongnam-do, Republic of Korea; [5]Laboratory Genetic Metabolic Diseases, Academic Medical Center, Amsterdam, The Netherlands

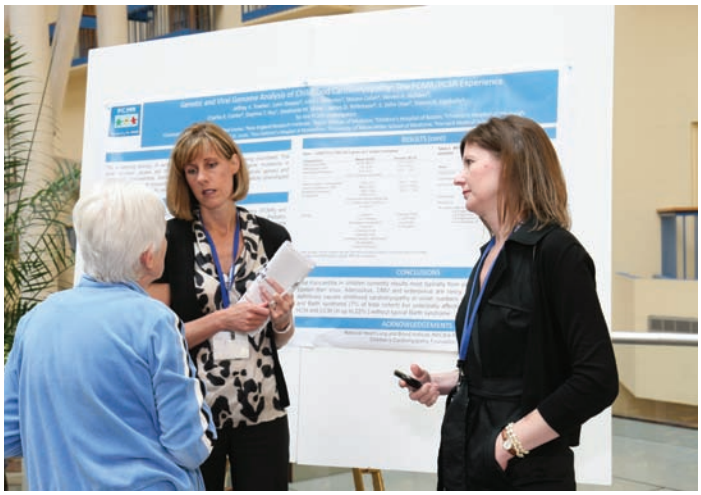
POSTER 25:
**Exome Sequencing to Identify Potential Genetic Modifiers of Barth Syndrome**
Michael V. Zaragoza, MD, PhD, V Hoang, S Hakim : UCI Cardiogenomics Program, Department of Pediatrics & Biological Chemistry, University of California, Irvine, School of Medicine, Irvine, CA, USA

POSTER 26:
**Functional Characterization of the Mitochondrial Phosphatase PTPMT1**
Ji Zhang, MD, PhD : Department of Pharmacology, University of California San Diego, La Jolla, CA, USA

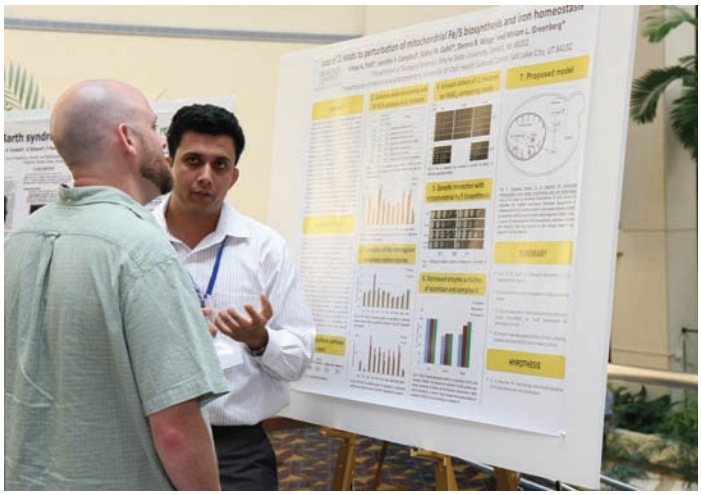


Professor Charles Hoppel and Dr. Michael Kiebish


Drs. Mindong Ren and Frederic Vaz



Dr. Iris Gonzalez, Dr. Stephanie Ware and Susan Kirwin



Drs. Stephen Claypool and Vinay Patil

## SCIENCE & MEDICINE SESSIONS
## Friday, June 27, 2014

**7:30am—8:15am (Salon D, E, F)**

| | |
|---|---|
| 7:30am—8:15am | **Breakfast** for all conference attendees |

**8:30am—11:20am (Salon G)**
**MITOCHONDRIAL PHYSIOLOGY**
*Chair—Colin G. Steward, PhD, FRCP, FRCPCH, Bristol Royal Hospital for Children, Bristol, United Kingdom*

| | |
|---|---|
| 8:30am—8:35am | Introduction — Lee Kugelmann |
| 8:35am—9:05am | **Modeling the Mitochondrial Myopathy of Barth Syndrome using iPSC and Heart-on-chip Technologies** — *William Pu, MD, Boston Children's Hospital, Boston, MA* |
| 9:05am—9:35am | **Cardiac Mitochondrial Structure and Function in *Tafazzin*-knockdown Mice** — *Junhwan Kim, PhD, Center for Resuscitation Science, Philadelphia, PA* |
| 9:35am—10:05am | **Respiratory Chain Remodeling in Cardiac Tissue of Barth Syndrome Patients** — *Jan Dudek, PhD, University of Göttingen, Institute of Cellular Biochemistry, Göttingen, Germany* |
| 10:05am—10:20am | Coffee Break |
| 10:20am—11:20am | Four Poster Presenters (15 minutes each) |

**12:00pm—2:00pm (Salon D, E, F)**
**VARNER AWARD LUNCHEON**

| | |
|---|---|
| 12:00pm—2:00pm | **Varner Award Luncheon** *(Presented by John Wilkins)* |

**2:00pm—6:05pm (Salon G)**
**ANIMAL MODELS**
*Chair—William Pu, MD, Boston Children's Hospital, Boston, MA*

| | |
|---|---|
| 2:00pm—2:05pm | Introduction — Morgan & Josh Atwell |
| 2:05pm—2:35pm | **Metabolic Pathways Affected by Cardiolipin Deficiency** — *Zaza Khuchua, PhD, Cincinnati Children's Hospital Medical Center, Cincinnati, OH* |
| 2:35pm—3:05pm | **Substrate-specific Impairment of Oxidative Phosphorylation in *Taz*-deficient Cardiac Mitochondria** — *Catherine Le, PhD, The Buck Institute for Research on Aging, Novato, CA* |
| 3:05pm—3:35pm | **Altered Triglyceride Metabolism Contributes to Low Body Weight in *Tafazzin* Knock-down Mice** — *Laura Cole, PhD, University of Manitoba, Manitoba, Canada* |
| 3:35pm—3:50pm | Coffee Break |
| 3:50pm—4:20pm | **Cardiomyopathy and Myocardial Noncompaction in Barth Syndrome** — *Colin Phoon, MPhil, MD, New York University Langone Medical Center; New York University School of Medicine, New York, NY* |
| 4:20pm—4:50pm | **Engineering Precise Genetic Alterations at the *Taz* Locus** — *Douglas Strathdee, PhD, The Beatson Institute for Cancer Research, Glasgow, United Kingdom* |
| 4:50pm—5:20pm | ***Tafazzin* Enzyme Replacement Therapy in a Mouse Model of Barth Syndrome** — *Michael T. Chin, MD, PhD, FACC FAHA, University of Washington School of Medicine, Seattle, WA* |
| 5:20pm—6:05pm | **Conference Wrap-Up** |

**7:00pm—11:00pm (Salon D, E, F)**
**FRIDAY NIGHT SOCIAL**

Join us for a superhero-themed evening: socializing, music, dancing, appetizers, and cash bar. All conference attendees are invited!

## SCIENCE & MEDICINE SESSIONS
## Saturday, June 28, 2014

**8:00am—11:30am (Salon D, E, F)**

| | |
|---|---|
| 8:00am—9:00am | **Breakfast** for all conference attendees |
| 8:15am—11:30am | **Scientific and Medical Advisory Board Breakfast & Meeting (Water's Edge)** *(by invitation)* |

**12:15pm—2:00pm (Salon D, E, F)**
**LUNCHEON and FINALE** *(All conference attendees welcome)*

## FAMILY SESSIONS
## Friday, June 27, 2014

**7:30am—8:15am (Salon D, E, F)**

| | |
|---|---|
| 7:30am—8:15am | **Breakfast** for all conference attendees |

**9:15am—11:45am (Salon A, B, C)**
**CARDIAC ASPECTS OF BARTH SYNDROME**

| | |
|---|---|
| 9:15am—9:25am | Introduction — Oliver Baxter-Smith |
| 9:25am—9:55am | **Pathophysiology of Barth Syndrome**<br>*Jeffrey Towbin, MD, FAAP, FACC, FAHA Cincinnati Children's Hospital Medical Center, Cincinnati, OH* |
| 9:55am—10:25am | **Bridge Devices, Pharmaceutical Agents and Genetic Therapies**<br>*John Jeffries, MD, MPH, FAAP, FACC, Heart Institute, Cincinnati Children's Hospital Medical Center, Cincinnati, OH* |
| 10:25am—10:55am | **Active Surveillance for Occult Arrhythmias in Barth syndrome: Is It Time?**<br>*Randall Bryant, MD, University of Florida-Jacksonville/Gainesville, Jacksonville, FL* |
| 10:55am—11:25am | **Transplants in Barth Syndrome**<br>*Jeffrey Towbin, MD, FAAP, FACC, FAHA Cincinnati Children's Hospital Medical Center, Cincinnati, OH* |
| 11:25am—11:45am | **Q & A** |

**12:00pm—2:00pm (Salon D, E, F)**
**VARNER AWARD LUNCHEON**

| | |
|---|---|
| 12:00pm—2:00pm | **Varner Award Luncheon** *(Presented by John Wilkins)* |

**2:00pm—2:30pm (Salon A, B, C)**
**CARRIER SESSION IV**

| | |
|---|---|
| 2:00pm—2:30pm | **LIFE AS A CARRIER**<br>*Rebecca L. McClellan, MGC & Lee Kugelmann*<br><br>This session will focus on the myriad of issues that present during the lifetime of a woman who is a known carrier for Barth syndrome, including guilt, relationships, being different and raising a daughter who might be a female carrier of Barth syndrome. [*Audience: Carrier mothers and grandmothers of living and deceased children. Carrier mothers and grandmothers of female children (pre-test and post-test carriers)*]. |

**2:30pm—4:00pm (Salon A, B, C)**
**PLANNING AHEAD**

| | |
|---|---|
| 2:30pm—2:40pm | Introduction — Kevin Woodward |
| 2:40pm—3:00pm | **Financial Independence** — *Tom Nurse, Personal Financial Advisors for Families with Special Needs, Tampa, FL* |
| 3:00pm—3:20pm | **Healthcare Transitions** — *Debbie Riddiford, CNS, Bristol Royal Hospital for Children, Bristol, United Kingdom* |
| 3:20pm—3:40pm | **Difficult Talks and Transparency with Barth Syndrome** — *Vanessa Garratt, DClinPsych, Bristol Royal Hospital for Children, Bristol, United Kingdom* |
| 3:40pm—4:00pm | **Q & A** |

**4:15pm-5:00pm (Salon A, B, C)**
**TRANSPLANTS**

| | |
|---|---|
| 4:15pm—5:00pm | **TRANSPLANT BREAKOUT**<br>*Stephanie Rader, Nicole Derusha-Mackey & Jeffrey Towbin, MD, FAAP, FACC, FAHA*<br><br>This session provides adults who have received a transplant and parents of those who have received a heart transplant an opportunity to discuss transplant related issues in a candid setting with Dr. Towbin. |

**5:00pm (Citrus)**
**GROUP PHOTO (Transplant boys/young men)** *(immediately following educational sessions)*
**GROUP PHOTO (Carriers)**

**7:00pm—11:00pm (Salon D, E, F)**
**FRIDAY NIGHT SOCIAL**

Join us for a superhero-themed evening: socializing, music, dancing, appetizers, and cash bar. All conference attendees are invited!

**AFFECTED INDIVIDUALS and SIBLINGS SESSIONS**
**Friday, June 27, 2014**

**7:30am—8:15am (Salon D, E, F)**

| | |
|---|---|
| 7:30am—8:15am | **Breakfast** for all conference attendees |

**8:15am—9:10am (Mandalay)**
**HANG OUT AND DISCUSS DAY ACTIVITIES** *(Combined Youth)*

**9:15am—9:45am**
**YOUTH GROUP PHOTOS (Sandpiper Deck)**

**9:45am—12:00pm**
**SHELLING/SAND CASTLE BUILDING (Gather on Sandpiper Deck)** *(Combined Youth)*
*NOTE: Please remember to bring beach towel, sunscreen and change of clothes.*

**12:00pm—1:30pm**
**LUNCHEON**

| | |
|---|---|
| 12:00pm—1:30pm | **Varner Award Luncheon** *(Presented by John Wilkins)* **(Salon D, E, F)** |
| 12:00pm—1:30pm | **Youth Luncheon** *(Combined Youth) w/ special guests 'The Rough Riders'* **(Sandpiper Deck)** |

**1:45pm—5:00pm (Mandalay)**
**ACTIVITY TIME** *(Combined Youth)*
**Make conference keepsakes with Whim-So-Doodle.**

**7:00pm—11:00pm (Salon D, E, F)**
**FRIDAY NIGHT SOCIAL**

Join us for a superhero-themed evening: socializing, music, dancing, appetizers, and cash bar. All conference attendees are invited!

Barth Syndrome®
Trust

# Barth Syndrome Trust
## is the Proud Sponsor of the
# Friday Night Social

# Miss Christina Hixson
# Friend of John Wilkins
## is the Proud Sponsor of the
# Pioneers in Science & Medicine
# Varner Award

# Family and Friends of John Wilkins
## are the Proud Sponsors of the following events:
# Keynote Speaker Luncheon
# Varner Award Luncheon
# Scientific & Medical Advisory Board Breakfast

## FAMILY SESSIONS
## Saturday, June 28, 2014

**8:00am—9:00am (Salon D, E, F)**

| | |
|---|---|
| 8:00am—9:00am | **Breakfast** for all conference attendees |

**9:00am—10:00am (Salon A, B, C)**
**WRAP-UP SESSIONS**

| | |
|---|---|
| 9:00am—9:15am | **Putting the Pieces Together Wrap-Up Session**<br>*Rebecca L. McClellan, MGC & Lee Kugelmann* |
| 9:15am—9:30am | **Transplant Wrap-Up Session**<br>*Nicole Derusha-Mackey & Stephanie Rader* |
| 9:30am—9:45am | **Volunteer Wrap-Up Session**<br>*Lindsay Groff & Sandra Stevens* |
| 9:45am—10:00am | **Youth Wrap-Up Session**<br>*BJ Develle, MSW & John Wilkins* |

**10:00am—12:00pm (Salon A, B, C)**
**ROVING PANEL BREAKOUT SESSIONS**

During these breakouts, experts will rotate every 20 minutes to discuss various age appropriate topics of interest. Historically, these breakout sessions have resulted in many "ah-ha" and "me too" moments. Not only are these sessions helpful to our families, they are also helpful to us at BSF in identifying needs, hearing insights, and ultimately developing resources that will help the families in our community.

- Marni Jacob, PhD: Psychosocial issues
- Tom Nurse: Planning for financial independence
- Julie Floyd: Education
- Nicol Clayton: Nutrition
- Debbie Riddiford, CNS: Coordination of care and transitions in care

| **Location** | **Parents/Grandparents of Age Groups** |
|---|---|
| Dolphin | Early Years (Birth – Four)<br>*Brie Chandler-Kalapasev & Travis Gordon* |
| Marlin | Early Childhood (Four – Eight)<br>*Amanda Maksin & Jack Higgins* |
| Tarpon | Middle Childhood (Eight – Twelve)<br>*Keli Holly & Andrew Buddemeyer* |
| Coral | Adolescence (Twelve – Sixteen)<br>*Leslie Buddemeyer & Ryan Ritter* |
| Salon C | Adulthood (Sixteen+)<br>*Cathy Ritter & Peter van Loo* |

**12:15pm—2:00pm (Salon D, E, F)**
**LUNCHEON & FINALE**

| | |
|---|---|
| 12:15pm—2:00pm | **LUNCHEON & FINALE** |

**AFFECTED INDIVIDUALS and SIBLINGS SESSIONS**
**Saturday, June 28, 2014**

**8:00am—9:00am (Salon D, E, F)**

| | |
|---|---|
| 8:00am—9:00am | **Breakfast** for all conference attendees |

**9:00am—12:00pm (Mandalay)**
**FINALE PROJECT, CONFERENCE WRAP-UP, KID-FRIENDLY EDUCATIONAL MATERIALS** *(Combined Youth)*

**12:15pm—2:00pm (Salon D, E, F)**
**LUNCHEON & FINALE**

| | |
|---|---|
| 12:15pm—2:00pm | **LUNCHEON & FINALE** |

Barth Syndrome Foundation of Canada


# The McCormack-Marra Family

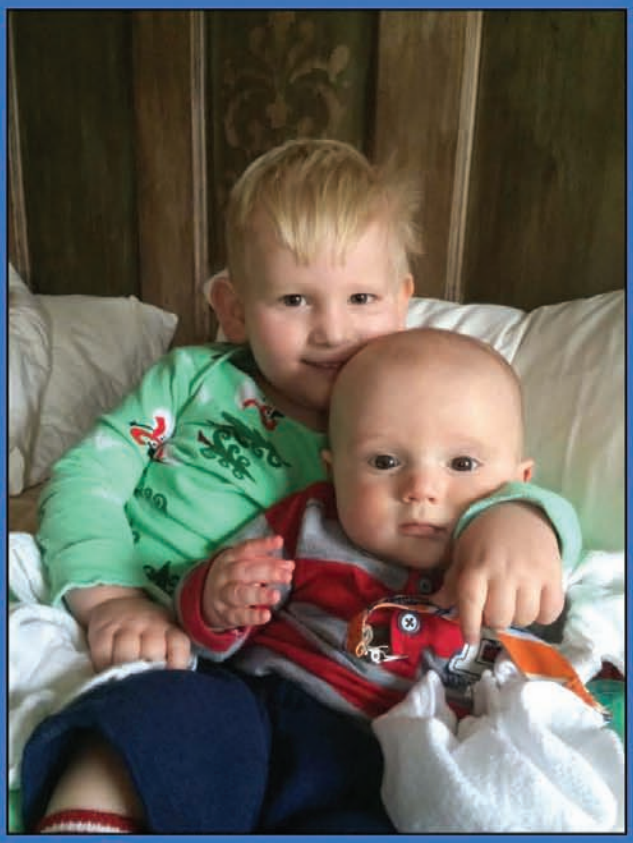
## is the Proud Sponsor of the following:

# Professional Travel Assistance
# Thursday Refreshment Break

# The Family and Friends of Connor Woodward

are proud sponsors of

The Barth Syndrome Foundation 2014 Conference




June 23-28, 2014 • Clearwater, FL USA

# The Millet Family
## is the Proud Sponsor of the
# Professional Videos

# Association Barth France

## is the Proud Sponsor of the

# Clinics & Consultations




The Hall Family
is the Proud Sponsor of the
Friday Breakfast
Make a Difference


Mike and Sue Wilkins
in honor of John Wilkins
are the Proud Sponsors of the
Saturday Breakfast
Make a Difference


The Sernel Family
is the Proud Sponsor of the
Clinician/Scientist
Informal Welcome Event
Make a Difference

**The Holly Family**
**is the Proud Sponsor of the**
**Conference T-Shirts**

Make a Difference

**The Buddemeyer Boys**
**are the Proud Sponsors of the**
**Gifts for Expert Speakers**

Make a Difference

**The Moreland Family**
**is the Proud Sponsor of the**
**Photographer Lodging**

Make a Difference



Mercy Medical Angels®

1st U.S. Volunteer Cavalry Regiment 1

## BARTH SYNDROME FOUNDATION

**Barth Syndrome Foundation, Inc.**
P.O. Box 618
Larchmont, New York 10538
Telephone: (850) 273-6947
Fascimile: (518) 213-4061
E-mail: bsfinfo@barthsyndrome.org
Website: www.barthsyndrome.org

**Executive Staff**
Lindsay B. Groff, MBA, Executive Director
Valerie M. Bowen, Family Services/Awareness
Matthew J. Toth, PhD, Science Director
Lynda M. Sedefian, Executive Assistant
Sandra Stevens, Fundraising Project Manager

**Board of Directors**
Marcus E. Sernel, *Chairman*
David Axelrod, MD, Board Member
Randy Buddemeyer, Treasurer
Stephen Kugelmann, Board Member
Susan A. McCormack, Secretary
Susan S. Osnos, Board Member
Catharine L. Ritter, RN, Board Member
John Wilkins, Board Member
Kevin Woodward, Board Member
Lindsay B. Groff, Executive Director, *ex-officio*
Stephen B. McCurdy, *Chairman Emeritus*

## AFFILIATES

**Barth Syndrome Trust (UK & Europe)**
1 The Vikings
Romsey
Hampshire
S051 5RG
United Kingdom
Telephone: +44(0)1794 518785
E-mail: info@barthsyndrome.org.uk
Website: www.barthsyndrome.org.uk

**Trustees**
Michaela Damin, Chair
Kerry Caldwell, Treasurer
Helen Coleman, Secretary
Suzy Green, Trustee
Nigel Moore, Trustee
Sonja Schlapak, Trustee

**Barth Syndrome Foundation of Canada**
162 Guelph Street, Suite 115
Georgetown, ON L7G 5X7
Canada
Telephone: (905) 873-2391
E-mail: info@barthsyndrome.ca
Website: www.barthsyndrome.ca

**Board Members**
Lynn Elwood, President
Catharine Ritter, RN, Vice President
Susan Hone, Secretary
Chris Hope, Treasurer

**Association Barth France**
12, rue Lalo
75116 Paris
France
Telephone: +33 1 45 00 86 12
E-Mail: associationbarthfrance@orange.fr
Website: www.barthfrance.com

**Trustees**
Florence Mannes, Chair
Philippe Mannes, Secretary
Marc Pillot, Treasurer


The Barth Syndrome Foundation would like to acknowledge the members of the 2014 Conference Committee:

**Management Committee**
Lindsay Groff, MBA, *Chair*
Shelley Bowen
BJ Develle, MSW
Lois Galbraith
Lynda Sedefian
Matthew Toth, PhD
John Wilkins
Kevin Woodward

**Family Subcommittee**
Shelley Bowen, *Chair*
Nicole Derusha-Mackey
Stephanie Rader
Rebecca L. McClellan, MGC
Lee Kugelmann

**Science & Medicine Subcommittee**
Matthew Toth, PhD, *Chair*
Terry Dannels
Lois Galbraith

**Young BTHS Adults Subcommittee**
John Wilkins, *Chair*

**Youth/Sibling Subcommittee**
BJ Develle, MSW, *Chair*
John Wilkins

**Clinic Subcommittee**
Shelley Bowen, *Chair*
Lois Galbraith
Chris Hope
Jim Pagano
Mary Lou Pagano
Debbie Riddiford, CNS
Donna Strain
Michelle Florez

**Fundraising Subcommittee**
Kevin Woodward, *Chair*
Sandra Stevens
Heather Tauw

**Social Subcommittee**
Lynda Sedefian, *Chair*
Leslie Buddemeyer
Brie Chandler-Kalapasev
Julie Fairchild
Kristi Pena
Shelia Rawlings

**Publications Committee**
Lynda Sedefian, *Chair*
Kate McCurdy
Lorna Moore
Nigel Moore
Les Morris

## BARTH SYNDROME FOUNDATION SCIENTIFIC & MEDICAL ADVISORY BOARD

Michael Schlame, MD, *Chairman*
Cell Biology & Anesthesiology, NYU, School of Medicine, New York, NY

Peter G. Barth, MD, PhD, *Emeritus*, Pediatric Neurology (retired), Emma Children's Hospital / AMC; Amsterdam, The Netherlands

W. Todd Cade, PT, PhD, Physical Therapy & Internal Medicine, Washington University School of Medicine; St. Louis, MO

Gerald F. Cox, MD, PhD, Clinical Genetics, Boston Children's Hospital, Boston, MA; Clinical Research, Genzyme Corp.; Cambridge, MA

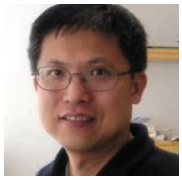
Iris L. Gonzalez, PhD, Molecular Diagnostics Lab *(retired)*, A. I. duPont Hospital for Children; Wilmington, DE

Miriam L. Greenberg, PhD, Biological Sciences, Wayne State University; Detroit, MI

Grant M. Hatch, PhD, Lipid Lipoprotein Research, University of Manitoba; Winnipeg, Canada

Richard I. Kelley, MD, PhD, Metabolism, Kennedy Krieger Institute, Johns Hopkins University; Baltimore, MD

William T. Pu, MD, Pediatric Cardiology, Boston Children's Hospital; Harvard Stem Cell Institute; Boston, MA



Mindong Ren, PhD, Cell Biology, New York University School of Medicine; New York, NY

Colin G. Steward, PhD, FRCP, FRCPCH, Pediatric Hematology, Bristol Royal Hospital for Children; Bristol, United Kingdom

Arnold Strauss, MD, Pediatrics and Research, Cincinnati Children's Hospital Medical Center; Cincinnati Children's Research Foundation; Cincinnati, OH

Mark Tarnopolsky, MD, PhD, FRCP(C), Pediatric Neuromuscular & Neurometabolic Clinic, McMaster University Medical Center; Ontario, Canada

Jeffrey A. Towbin, MD, FAAP, FACC, FAHA, Pediatric Cardiology, Cincinnati Children's Hospital; Cincinnati, OH

Ronald J. A. Wanders, PhD, Genetic Metabolic Diseases, Academic Medical Center; Amsterdam, The Netherlands

Catharine L. Ritter, RN, *ex-officio*, Board of Directors, Barth Syndrome Foundation

Matthew J. Toth, PhD, *ex-officio*, Science Director, Barth Syndrome Foundation

| OTHER SPONSOR PARTNERS | |
|---|---|
| Sunset & Stargazing | Stephanie, Noah, Olivia & Carol Rader |
| Poolside Partner | Dr. Michael Schlame |
| Family Welcome Event | Stephanie, Noah, Olivia & Carol Rader |
| Tables at Friday Night Social | Sheila, David, Ben & English Mann [1]<br>Les Morris & Lois Galbraith [1]<br>Lorna & Nigel Moore [3]<br>Mike, Sue & John Wilkins *(in honor of Muriel Wilkins)* [2] |
| Supplies for Children Activities | Les Morris |
| Registration Desk Supplies | Lynn, Rick, Adam & Justin Elwood |

| IN-KIND DONATIONS<br>BSF would like to acknowledge the following in-kind donations in support of BSF's 2014 Conference: | |
|---|---|
| Alex and Ani [Jewelry] | Camden Lynn [DJ] |
| All Children's Hospital/Johns Hopkins Medicine, St. Petersburg, FL: Gul Dadlani, MD, Medical Director of Pediatric Cardiology and Kathyrn Douglas, RDCS, CCT, Chief Pediatric Cardiac Sonographer [Clinics] | Sheila, David, Ben & English Mann [Barth Gift Basket] |
| Megan Branagh [Autographed Nicole Branagh volleyball/picture; handmade hairclips/head wraps] | Susan McCormack [Four $25 gift cards] |
| Leslie Buddemeyer [Gingersnap jewelry and flip-flops]; Treasure hunt supplies | Steve & Kate McCurdy [Two $50 gift cards & three $25 gift cards] |
| Angela Calhoun [Original painting] | Mercy Medical Airlift [Travel] |
| Chicago Bears Football Club [Limited edition piece w/ autographs] | Kate Michener [Gift basket of lip balms, cremes, etc.] |
| Bryan Drake [Dr. DRE URBeats in-ear headphones; Hand-crocheted BSF colored blanket] | Jimella Monroe Evans [Crocheted gift] |
| Julie Fairchild [Supplies for Treasure Hunt] | New England Revolution [Four tickets to game] |
| Frito-Lay [Snacks] | Jim Neviaser [Snacks] |
| Lois Galbraith [Superman Plush Dog] | Pink Calyx [$50 Gift Certificate] |
| Dr. Iris Gonzalez [Two paintings/fiber art] | Sandra Stevens [Crocheted baby blanket & knitted rabbit toy] |
| Lindsay Groff [BSF Dragonfly-themed basket] | TJ Maxx [Logo bags] |
| Hilton Clearwater Hotel [Complimentary 2-night stay at Hilton Clearwater + breakfast for two] | Jerre Vogt & Dodie Weltlich [Cupcakes for Youth Activities] |
| Audrey Hintz [Three dragonfly brooches] | Walt Disney World [OneDay Hopper Passes] |
| Susan, Chris, Jessica (Wright), Josh & Jared Hone [Luminaries] | Whim-So-Doodle [Arts & craft supplies] |
| International Tennis Hall of Fame [Gift certificate for two to 2014 Hall of Fame Tennis Championships + musuem passes] | Sue & Mike Wilkins [Two $25 & one $50 Barnes & Noble gift cards; two thermometers] |
| Lucas Productions [Discounted AV] | Jessica Wright & Susan Hone [Jamberry nail wraps basket] |

**Matthew G. Baile, PhD** — Graduate Student, Department of Physiology, Johns Hopkins School of Medicine, Baltimore, MD, USA

Dr. Baile's research interest is studying cardiolipin remodeling in the model system Saccharomyces cerevisiae.

Dr. Baile holds a PhD from Johns Hopkins University, Baltimore, MD, USA and a BSc in Biology from Shippensburg University, Shippensburg, PA, USA

Presentation: Unremodeled and Remodeled Cardiolipin are Functionally Indistinguishable in Yeast (*SciMed*)

**Audrey Anna Bolyard, RN, BS** — Clinical Manager, University of Washington, The Severe Chronic Neutropenia International Registry, Seattle, WA, USA

Ms. Bolyard manages the Severe Chronic Neutropenia International Registry (SCNIR) which she helped to established in 1994 to monitor the clinical course, treatment, and disease outcomes in patients with severe chronic neutropenia (SCN). The Registry has the largest collection of long-term data on patients with this condition in the world. Participation in the Registry benefits patients, their families and the physicians who treat them by providing the most up to date information to them on the natural history of SCN and its treatment options. Many Barth patients are members of this Registry.

Note: Ms. Bolyard is involved in the Severe Chronic Neutropenia International Registry web-based consultation.

**Ann Bowron, FRCPath** — Principal Paediatric Clinical Biochemist, Bristol Royal Infirmary, Bristol, United Kingdom

Ms. Bowron is a Clinical Biochemist specializing in inborn errors of metabolism. She set up a service for the diagnosis of Barth syndrome by cardiolipin analysis. She is studying part-time for a PhD on cardiolipin and Barth syndrome at the University of Bristol.

Ms. Bowron holds an MSc and Fellowship of the Royal College of Pathologists. She held a position as lead scientist for the metabolic biochemistry service. Previously she worked in the Metabolic Diseases Unit at the Institute of Child Health, London, UK.

Presentation: Barth Syndrome Without Severe Cardiolipin Deficiency: A Possible Ameliorated Phenotype *(Sci/Med)*

**Randall Bryant, MD** — Associate Professor, Department of Pediatrics, Division of Pediatric Cardiology, University of Florida-Jacksonville/Gainesville; Director, Interventional Electrophysiology and Pacing; Co-Director, North Florida Children's Comprehensive Cardiac Network; Director, Transtelephonic Arrhythmia Monitoring Program, University of Florida-Jacksonville/Gainesville, Jacksonville, FL, USA

Dr. Bryant's specialties include pediatric cardiology and pediatric medicine, and he focuses on studies which include the use of pacemakers and implantable cardioverter defibrillators in children with hypertrophic cardiomyopathy; natural history and treatment of sinus node dysfunction in pediatric heart transplantation; pacemaker implantation in children with hypertrophic cardiomyopathy.

Dr. Bryant received his BA from Princeton University and his MD from Duke University Medical Center. He trained in Pediatrics, Pediatric Cardiology and Pediatric Electrophysiology at Baylor College of Medicine in Houston, TX and completed his residency in Pediatrics at Texas Children's Hospital. He also completed a Fellowship in Pediatric Cardiology at Baylor and Pediatric Electrophysiology at Texas Children's Hospital. Dr. Bryant is board certified in Pediatric Cardiac Electrophysiology.

Discussion: Transitions: Taking Charge of my Health and Well-being and Mentoring Others with Barth Syndrome to do the Same *(Affected Individuals);*
Presentation: Active Surveillance for Occult Arrhythmias in Barth syndrome: Is It Time? *(Family)*

**Barry J. Byrne, MD, PhD** — Associate Chair and Professor of Pediatrics and Molecular Genetics and Microbiology, College of Medicine, Department of Pediatrics; Molecular Genetics and Microbiology; Director of the Powell Gene Therapy Center at the University of Florida, Gainesville, FL, USA

Dr. Byrne is a clinician scientist who is studying a variety of rare diseases with specific attention to developing therapies for inherited muscle disease. As a pediatric cardiologist, his focus is on conditions that lead to skeletal muscle weakness and problems in heart and respiratory function. His group has made significant contributions to the understanding and treatment of Pompe disease, which is a type of muscular dystrophy due to abnormal glycogen in the muscle. The research team has been developing new therapies using the missing cellular protein or the corrective gene to restore muscle function in Pompe and other inherited myopathies. Dr. Byrne was awarded a research grant from BSF entitled "Gene therapy in a mouse model of Barth syndrome" (2010).

Dr. Byrne obtained his BS degree from Denison University, his MD and PhD from the University of Illinois and completed his Pediatrics residency, cardiology fellowship training and post-doctoral training in Biological Chemistry at the Johns Hopkins Hospital. He joined the University of Florida in 1997 and is now the Earl and Christy Powell University Chair in Genetics.

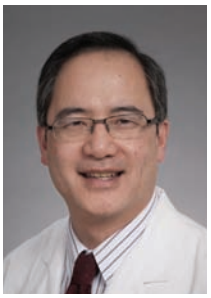
Keynote Speaker: Orphan Product Development in the Era of Personalized Medicine

**W. Todd Cade, PT, PhD** — Associate Professor of Physical Therapy and Internal Medicine, Washington University School of Medicine, St. Louis, MO; Scientific and Medical Advisory Board, Barth Syndrome Foundation, USA

Dr. Cade's research interests include mechanisms and treatments for whole-body and myocardial nutrient metabolism abnormalities in metabolic diseases such as HIV-associated metabolic syndrome, diabetes, and Barth syndrome and in normal and pathologic pregnancy. Dr. Cade was awarded three BSF grants entitled "Effects of Resistance Training on Cardiac, Metabolic, and Muscle Function and Quality of Life in Barth Syndrome" (2011); "Safety and Efficacy of Aerobic Exercise Training in Barth Syndrome: A Pilot Study" (2009); and "Characterization of Nutrient Metabolism in Barth Syndrome" (2008). Dr. Cade currently has an NIH funded grant examining whole-body and heart metabolism and function in Barth syndrome.

Dr. Cade holds a PhD in Rehabilitation Science/Applied Physiology from the University of Maryland. He also has a BSc in Kinesiological Science from the University of Maryland, an MSc in Physical Therapy from the University of Miami, Florida, and is a licensed physical therapist. He completed a post-doctoral fellowship in the Division of Endocrinology, Metabolism and Lipid Research at Washington University School of Medicine, and holds an NIH-funded Career Development Award from the National Institute of Diabetes and Digestive and Kidney Diseases.

Presentation: Exercise and Substrate Metabolism Studies in Barth Syndrome: An Update *(Sci/Med);* Discussion: What You Need To Know About Barth Syndrome But Were Afraid To Ask *(Family/Affected Individuals/Siblings)*



**Michael T. Chin, MD, PhD, FACC FAHA** — Associate Professor, University of Washington School of Medicine, Seattle, WA, USA

Dr. Chin's major research interests include understanding transcriptional control of cardiovascular development, the environment influences that affect the development of cardiovascular disease and developing novel therapies for cardiovascular and myopathic disorders. He directs a research laboratory focused on understanding the molecular biology of the cardiovascular system. Dr. Chin was awarded a research grant from BSF entitled "*Tafazzin* Enzyme Replacement Therapy for Heart Muscle in Barth Syndrome" (2012).

Dr. Chin holds a PhD from the University of Rochester (1988), an AB from Princeton University (1983), and MD (with Honors), University of Rochester (1991). He completed his Residency in Internal Medicine, at The Johns Hopkins Hospital, Baltimore (1991-1993).

Presentation: *Tafazzin* Enzyme Replacement Therapy in a Mouse Model of Barth Syndrome (*Sci/Med*)

**Nicole Clayton** — Specialist Dietician, NHS Barth Syndrome National Service, Bristol Royal Hospital for Children, Bristol, United Kingdom

Ms. Clayton is a Specialist Paediatric Dietician at Bristol Royal Hospital for Children, UK. She has been a member of the multidisciplinary NHS Barth Syndrome Specialized Service, led by Dr. Steward, since its inception in April 2010, and manages the dietetic care of all Barth patients in the UK. Ms. Clayton also specializes in the provision of ketogenic diet therapy, a metabolic treatment for children with intractable epilepsy.

Ms. Clayton's previous roles included working within a multi-disciplinary feeding disorders clinic to manage difficult behaviors around food, such as food restriction, food refusal and specific aversions. During this time she delivered study days to train health and medical professionals in effectively managing children's behavioral problems with food and drink. Ms. Clayton has worked extensively with children with neurological and developmental problems, and has a special interest in the nutrition of children with unusual growth disorders.

Discussions: What you Need to Know about Barth Syndrome but were Afraid to Ask; General Inquiries/Presentation: Nutrition & Metabolism: The Bristol Experience; Breakouts *(Family)* Note: Ms. Clayton is involved in the Metabolism & Nutrition Consultations.

**Laura Cole, PhD** — Postdoctoral Fellow, Department of Pharmacology and Therapeutics, Faculty of Medicine, University of Manitoba, Winnipeg, Manitoba, Canada

Dr. Cole's research interests focus on the role of cardiolipin remodeling on mitochondrial function in the heart and liver with a specific emphasis on triglyceride metabolism. Dr. Cole is currently funded by the Canadian Institute for Health Research (CIHR), Integrated and Mentored Pulmonary and Cardiovascular Training (IMPACT), Manitoba Health Research Council (MHRC) and Manitoba Institute of Child Health (MICH).

Dr. Cole holds a PhD in Lipid Biochemistry from the University of Alberta, Canada.

Presentation: Altered Triglyceride Metabolism Contributes to Low Body Weight in *Tafazzin* Knock-down Mice *(Sci/Med)*

**Angela Corcelli, PhD** — Professor of Physiology, Department of Basic Medical Sciences, Neurosciences and Sensory Organs, University of Bari Aldo Moro, Bari, Italy

After studying the transport of sugars, amino acids and ions in membranes isolated from epithelial cells, Dr. Corcelli has investigated novel biochemical aspects of the archaeal proton pump bacteriorhodopsin with particular interest in the lipid-protein interactions. She discovered and elucidated the structures of the archaeal analogs of cardiolipins and the halocapnines of halophilic bacteria. Her studies on cardiolipins have shown that the levels of cardiolipins in prokaryotic membranes fluctuate in response to osmotic stress. Being interested in the lipidomics, she has exploited the potentiality of mass spectrometry in the study of membrane lipids developing methods to directly analyze lipids in isolated membranes avoiding the steps of isolation and chromatographic separation by means of MALDI-TOF/MS. She is presently involved in the study of dynamics and the functional role of cardiolipins in biomembranes.

Dr. Corcelli was awarded a research grant from BSF entitled "Determination of the Monolysocardiolipin/cardiolipin (MLCL/CL) Ratio in Intact Nucleated Cells: A New Tool for the Screening of Barth Syndrome" (2012).

Presentation: Detection of Cardiolipin Abnormalities in White Blood Cells of Patients with Barth Syndrome by MALDI-TOF/MS *(Sci/Med)*

**Gul H. Dadlani, MD** — Clinical Assistant Professor in the Department of Pediatrics, University of South Florida, Tampa, FL; Medical Director of Pediatric Cardiology and Pediatric Cardiology Laboratory Director, All Children's Hospital in St. Petersburg, FL, USA

Dr. Dadlani's special interests include heart failure, cardiomyopathies, pulmonary hypertension, fetal echocardiography and Kawasaki disease. Dr. Dadlani completed his medical degree at State University of New York at Buffalo. He is a graduate of the pediatric residency program at Children's Hospital of Buffalo in New York where he received the Resident Teaching Award in three consecutive years as well as the University of Buffalo Medical School Siegal Teaching Award in Pediatrics. Dr. Dadlani completed his pediatric cardiology fellowship at Children's Hospital at Strong Memorial in Rochester, New York and twice received a Fellow Teaching Award from Golisano Children's Hospital at Strong. Dr. Dadlani is board certified in pediatric cardiology.

Note: Dr. Dadlani is involved in the research study entitled "Multi-disciplinary Studies in Barth Syndrome"

**Brittany Dane, BS** — Research Coordinator, University of South Florida Rothman Center for Pediatric Neuropsychiatry, Tampa, FL, USA

Ms. Dane graduated from Florida State University in 2013 with a degree in Psychology and is currently employed as a research coordinator at the University of South Florida Rothman Center for Pediatric Neuropsychiatry. The Rothman Center is a specialty clinic focusing on pediatric anxiety disorders and Autism spectrum disorders. Ms. Dane completes diagnostic assessments with children and families and coordinates research studies related to evidence-based treatments of anxiety and pediatric obsessive-compulsive disorder.

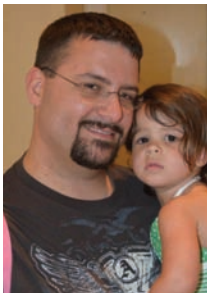
Note: Ms. Dane is involved in the research study entitled "Psychosocial Functioning in Barth Syndrome"

**Brittany DeCroes, PT, DPT** — Outpatient Physical Therapy Department, Kennedy Krieger Institute, Baltimore, MD, USA

Dr. DeCroes provides land based and aquatic physical therapy services to patients across the lifespan. In addition to working in the outpatient physical therapy department, she serves as the physical therapist in the Multidisciplinary Barth Syndrome Clinic. Her professional interests include fitness across the lifespan, standardized outcome measures, mitochondrial disorders and teaching (she currently participates as a teaching assistant in the physical therapy programs at both the University of Maryland, Baltimore, and George Washington University). Her current research interests include functional ability and quality of life in patients with Barth syndrome.

Dr. DeCroes received a Bachelor of Science degree from James Madison University in Harrisonburg, VA where she majored in Biology and minored in Spanish. She received her Doctorate of Physical Therapy from George Washington University in Washington DC.

Note: Dr. DeCroes is involved in the research study entitled "Multi-Disciplinary Studies in Barth Syndrome"



**BJ Develle, MSW** — Personal Health Coach Coordinator, Humana Cares, St. Petersburg, FL, USA

Mr. Develle currently works for the state of Florida's Agency for Health Care Administration interpreting policy and monitoring Substance Abuse and Mental Health providers. Previously, he provided case management and therapy services to children and specialized training to foster parents and professionals who would work with them. He has worked with children with histories of physical and sexual abuse, brain injuries, mood disorders, drug exposure, suicidal and homicidal attempts and psychiatric residential placements, both in the community and within a group home he previously managed. BJ has been a volunteer with BSF since 1998.

Mr. Develle graduated from Florida State University with a Masters in Social Work in 2008, after earning Bachelors degrees in Child Development and Religion in 2002.

Note: BJ is a facilitator of the youth groups *(Family)*

**Jean Donadieu, MD, PhD** — Pediatric Hemotology Oncology and Immunologist, Trousseau University Hospital, Paris, France

Dr. Donadieu's major interests are chronic neutropenia and the development of epidemiology for rare disorders in France and also in the European Union (EU) through participation in several EU projects. He now includes Barth syndrome as an interest as a result of the work of Barth France.

Dr. Donadieu founded and coordinates the French Severe Chronic Neutropenia Certified Patient Registry which contributes to the Severe Chronic Neutropenia International Registry. He also manages the French Registry of Histiocytosis. Since 2008, he has served as president of the French Society of Pediatric Hemato-immunology.

Dr. Donadieu holds a PhD in Public Health. He trained in pediatric hemotology-oncology and immunology at Necker Hospital, Paris. He has co-authored 140 manuscripts about several (mostly rare) disorders.

Presentation: The Long Way to Evidence-based Medicine for Barth Syndrome: Experience from France *(Sci/Med)*

**Jan Dudek, PhD** — Project Leader in the Laboratory of Dr. Peter Rehling, Department of Cellular Biochemistry, University of Göttingen, Institute of Cellular Biochemistry, Göttingen, Germany

Dr. Dudek's interest is mitochondria and especially how defects in mitochondrial biogenesis and function lead to human diseases. His research interest focuses on the structural and functional aspects of the respiratory chain.

Dr. Dudek holds a PhD in biochemistry from the University of Freiburg, Germany, and has held post-doctoral positions in laboratories at the Beatson Institute for Cancer Research, Glasgow, UK and the University of California, San Francisco, USA conducting research into oncogenic signalling pathways.

Presentation: Respiratory Chain Remodeling in Cardiac Tissue of Barth Syndrome Patients *(Sci/Med)*

**Vanessa Garratt, DClinPsych** — Principal Clinical Psychologist, Paediatric Cardiac Service; Barth Syndrome National Health Service, and Osteogenesis Imperfecta Services, Bristol Royal Hospital for Children, Bristol, United Kingdom

Dr. Garratt has a strong interest in developing services around the needs of young people and supporting them to manage and cope with long-term conditions. In 2012, she completed a Post Graduate Certificate in Clinical Leadership and Service Improvement, an Institute of Leadership and Management Level 7 award in executive coaching and MBTI Practitioner Training, and was awarded an a National Institute for Health Research (NIHR) research grant looking at psychological management of pulmonary hypertension in young people and adults. Dr. Garratt completed her Doctorate in Clinical Psychology in Bristol in 2005.

Discussions: What you Need to Know about Barth Syndrome but were Afraid to Ask; Presentation: Planning Ahead: Difficult Talks and Transparency with Barth Syndrome *(Family)*

**Matthew P. Gillum, PhD** — Assistant Professor, Novo Nordisk Foundation Center for Basic Metabolic Research, University of Copenhagen, Copenhagen, Denmark; Adjunct in Neurology, University of Iowa Carver College of Medicine, Iowa City, IA, USA

Dr. Gillum's research interests are in how disordered lipid metabolism contributes to cardiometabolic disease. Dr. Gillum was awarded a research grant from BSF entitled "Implications of Phosphatidylserine Deficiency in Skeletal Muscle and Heart of ROSA26-*taz* shRNATet-on Mouse Model of Barth Syndrome" (2012).

Dr. Gillum holds a PhD in Cellular and Molecular Physiology from Yale University, New Haven, CT, USA and a BS in Biology from Duke University, Durham, NC, USA. He is also an adjunct in Neurology at the University of Iowa, USA.

Presentation: Could Other *Tafazzin* Products Contribute to Heart and Muscle Pathology in Barth Syndrome? *(Sci/Med)*

**Iris L. Gonzalez, PhD** — Molecular Diagnostics Laboratory, A. I. duPont Hospital for Children, Wilmington, DE *(retired)*; Scientific and Medical Advisory Board, Barth Syndrome Foundation, USA

Dr. Gonzalez's scientific research and clinical interests include molecular diagnostics, research on Barth syndrome and other genetic diseases, and mutation databases. As a molecular geneticist in a diagnostic lab, Dr. Gonzalez has performed the genetic test for many patients to confirm a diagnosis of Barth syndrome. In addition, her scientific interests have led her to conduct research (with a BSF grant) on the mRNA associated with Barth syndrome. Dr. Gonzalez is also known by Barth families for writing a layman's guide to genetics that has been extremely valuable to BSF families and others. In 2002, Dr. Gonzalez was awarded a research grant from BSF entitled "A Study of *TAZ* mRNAs in Barth Syndrome Individuals."

Dr. Gonzalez received her PhD Biology in Genetics (1976), and holds a BA in Biology (1970) from the University of Delaware. Dr. Gonzalez completed a Post-doctoral from the University of Pennsylvania (1982-1985).

Note: Dr. Gonzalez is involved in the Genetic Consultations

**Miriam L. Greenberg, PhD** — Associate Dean for Research, College of Liberal Arts and Sciences, Professor, Biological Sciences, Wayne State University, Detroit, MI; Scientific and Medical Advisory Board, Barth Syndrome Foundation, USA

Dr. Greenberg's laboratory focuses on phospholipid metabolism in yeast as a model to address questions of fundamental importance to human health. One project in her laboratory is to understand the role of cardiolipin in essential cellular functions and how perturbation of cardiolipin synthesis leads to Barth syndrome. The second project focuses on identifying the molecular targets of lithium and valproate in order to elucidate the therapeutic mechanisms of action of the drugs in the treatment of bipolar disorder.

Dr. Greenberg was awarded the following research grants from BSF entitled "Identification of Human Cardiolipin Phospholipases that are Deleterious to *Tafazzin*-deficient Cells" (2013); "Cardiolipin Deficiency Leads to Defects in the TCA Cycle" (2011); "Loss of Cardiolipin Leads to Defective Mitochondrial Iron/Sulfur Biosynthesis and Iron Homeostasis" (2010); "Perturbation of mitophagy in cardiolipin mutants" (2009); "The Role of *Tafazzin* in Mitochondrial Protein Import—Implications for Barth Syndrome" (2008); "Perturbation of the Osmotic Stress Response in Cardiolipin Deficient Mutants" (2007); "The Role of Phosphatidylglycerol in Activating Protein Kinase C Mediated Signaling" (2006); "Does Copper Deficiency Play a Role in Barth syndrome" (2005); "*TAZ*1 Gene Function in Yeast: A Molecular Model for Barth Syndrome" (2002).

Presentation: Deletion of the cardiolipin-specific phospholipase rescues growth defects in the yeast *tafazzin* mutant *(Sci/Med)*

**Marni L. Jacob, PhD** — Post Doctoral Fellow & Licensed Psychologist, Department of Pediatrics at the Rothman Center for Neuropsychiatry, University of South Florida, Tampa, FL, USA

Dr. Jacob's clinical and research activities focus on anxiety disorders, with a particular emphasis on obsessive-compulsive disorder and obsessive-compulsive spectrum disorders (e.g., Trichotillomania, Tic Disorders). She is also interested in examining the psychosocial functioning and quality of life of youth with chronic illnesses. She engages in research and treatment with children, adolescents, and adults.

Dr. Jacob holds a PhD in Clinical Psychology (2012) and MS in Clinical Psychology (2010) from the University of Georgia, GA, USA and BS in Psychology (2006) from the University of Florida, FL, USA. She completed a Clinical Internship Child/Pediatric Track (2011-2012) at the University of Florida Health Sciences Center.

Breakouts *(Family)*; Men of Barth Meeting; Transitions; Self-Advocacy *(Affected Individuals)*. Note: Dr. Jacob is involved in the research study entitled "Psychosocial Functioning in Barth Syndrome".

**John Lynn Jefferies, MD, MPH, FAAP, FACC** — Associate Professor, Pediatric Cardiology and Adult Cardiovascular Disease; Director, Advanced Heart Failure and Cardiomyopathy, Heart Institute, Cincinnati Children's Hospital Medical Center, Cincinnati, OH, USA

Dr. Jefferies's current research interests include heritable causes of vascular disease, novel drug therapies for advanced heart failure, novel gene discovery in cardiomyopathy, characterization and management of left ventricular noncompaction (LVNC), and early diagnosis and management of chemotherapy induced cardiotoxicity. He has authored or co-authored over 100 peer-reviewed manuscripts and book chapters on Cardiomyopathy, Cardiovascular Genetics, and Adults with Congenital Heart Disease.

Dr. Jefferies completed his combined Pediatric and Adult Cardiology training at the Baylor College of Medicine in Houston, Texas, at the Texas Children's Hospital and the Texas Heart Institute. He is on the Editorial Board of the Texas Heart Institute Journal and is an active member of numerous professional organizations including the Heart Failure Society of America, the American College of Cardiology, and the American Heart Association.

Presentation: Diagnosis and Management of Cardiovascular Disease in Barth Syndrome *(Sci/Med)*

**Valerian E. Kagan, PhD, DSc** — Professor and Vice Chairman, Environmental and Occupational Health Department, University of Pittsburgh, Pittsburgh, PA, USA

Dr. Kagan's research interests focus on Free Radical Biology and Medicine. He is one of the world's recognized leaders and one of the most prominent authorities on this subject. Internationally known for his interdisciplinary studies of oxidative stress, antioxidants, tissue and cell acute and chronic injury, he has founded a new field of research "Oxidative Lipidomics" and demonstrated its research power in investigations of cell death mechanisms. Dr. Kagan's work uncovers specific pathways through which enzymes of oxidative metabolism, particularly those of mitochondria, participate in the production of specific oxygenated lipid molecules that act as signals and/or lipid mediators to trigger cell death program as well as mechanisms involved in clearance of damaged or dead cells.

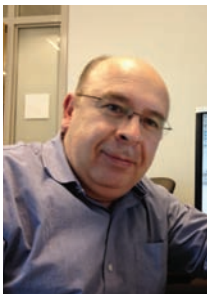
Dr. Kagan holds a DSc in Biochemistry and Biophysics from USSR Academy of Science, PhD in Biochemistry, MSc in Biochemistry, BSc in Biochemistry, all from Moscow State University, Russia. Dr. Kagan also holds various visiting professorships including Adjunct Foreign Professor, Institute of Environmental Medicine, Karolinska Institute, Stockholm, Sweden; Foreign Professor, Taipei Medical University, Taiwan; Foreign Professor, Russian State Medical University, Moscow, Russia; Foreign Professor, MV Lomonosov Moscow State University, Moscow, Russia; Fulbright Visiting Chair in Environmental Sciences, McMaster University, Hamilton, Canada; Sackler Lecturer, University of Tel-Aviv, Israel.

Presentation: Rancid Radical Talk: Music of Mitochondrial Cardiolipins *(Sci/Med)*

**Richard I. Kelley, MD, PhD** — Professor of Pediatrics, Johns Hopkins University School of Medicine; Director, Division of Metabolism, Kennedy Krieger Institute; Baltimore, MD, USA; Staff Physician, The Kennedy Krieger Institute; Director, Intermediary Metabolism and Clinical Mass Spectrometry Laboratory, Baltimore, MD; Scientific and Medical Advisory Board, Barth Syndrome Foundation, USA

Dr. Kelley is board certified in Pediatrics and Medical Genetics and specializes in the diagnosis and treatment of inborn errors of metabolism. His research focuses on the determination of the biochemical basis of both known and novel genetic disorders and on the treatment of selected diseases, including Barth syndrome, Smith-Lemli-Opitz syndrome, and disorders of mitochondrial metabolism. Dr. Kelley also is a co-founder of, and consulting geneticist for, the Clinic for Special Children, a charitable medical facility for the diagnosis and treatment of genetic disorders among the Amish and Mennonite populations of Lancaster, Pennsylvania. Dr. Kelley is the recipient of the 2008 Varner Award for Pioneers in Science and Medicine. He has been a pioneer in the study of Barth syndrome and a long-standing friend to BSF and to Barth families. He was formerly Chair of BSF's international Scientific and Medical Advisory Board.

Discussion: What You Need to Know About Barth Syndrome But Were Afraid To Ask; General Inquiries *(Family)*; General Inquiries *(Affected Individuals)*. Note: Dr. Kelley is involved in the research study entitled "Multi-Disciplinary Studies in Barth Syndrome" and Metabolism & Nutrition Consultations *(Family)*



**Zaza Khuchua, PhD** — Associate Professor, The Heart Institute, Cincinnati Children's Hospital Medical Center, Cincinnati, OH, USA

Dr. Khuchua's research interest focuses on mitochondrial function, structure and dynamics in cardiac cells in normal and pathological conditions; the role of mitochondrial phospholipds in aerobic metabolism in heart; and the role of lipid molecules in cell signaling systems. In 2006, Dr. Khuchua described the defects of the heart development in a zebrafish model of *tafazzin* deficiency. Dr. Khuchua was awarded a research grant from BSF entitled "The shRNA-mediated *Tafazzin* Knockdown Mouse Model for Barth Syndrome" (2009). In 2010, he started investigating the metabolic consequences of *tafazzin* knockdown in mice. In 2011, Dr. Khuchua was awarded a 4-year grant from the National Heart, Lung and Blood Institute at the National Institutes of Health for research using the Barth knockdown mouse model.

Dr. Khuchua holds a PhD from the All Union Cardiology Research Center, Moscow (1987), and a MS in Biochemistry from Moscow State University (1981). He was awarded the Royal Society Fellowship Award (1992); International Science Foundation Award (1993); Fogarty International Fellowship Award (1994); and the United Mitochondrial Disease Foundation Award (2006). He is a member of the International Society for Heart Research; the American Heart Association, the Scientific Council; and the American Society for Biochemistry and Molecular Biology.

Presentation: Metabolic Pathways Affected by Cardiolipin Deficiency *(Sci/Med)*

**Junhwan Kim, PhD** — Senior Research Investigator, University of Pennsylvania, Center for Resuscitation Science, Philadelphia, PA, USA

Dr. Kim's research interests focus on examination of the role of mitochondria dysfunction and impaired lipid metabolism on ischemic tissue damage during cardiac arrest. Dr. Kim was awarded a research grant from BSF entitled "Causative and Correlative Role of Cardiolipin on Integrated Mitochondrial Function in Barth Syndrome" (2009).

Dr. Kim is a member of the American Society for Mass Spectrometry.

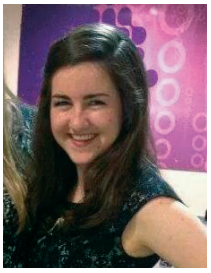
Presentation: Cardiac Mitochondrial Structure and Function in *Tafazzin*-knockdown Mice *(Sci/Med)*

**Consuelo M. Kreider, PhD, MHS, OTR/L** — Research Assistant Professor, College of Public Health and Health Related Professions, Department of Occupational Therapy, University of Florida; Contract Junior Investigator, Center of Innovation for Disability and Rehabilitation Research, North Florida/South Georgia Veterans Health System, Gainesville, FL, USA

Dr. Kreider's primary clinical interests are in the areas of pediatrics, learning, literacy, and sensory processing. Dr. Kreider's twenty years of clinical experience informs and complements her teaching in the areas of pediatric and adult assessment and intervention. She received a PhD in 2013 from the University of Florida in Rehabilitation Science with research focusing on investigation of the social contexts and environment impacting participation and performance of youth and young adults with disabilities.

Dr. Kreider holds a Masters in Health Science (2009) and a Bachelor of Health Science in Occupational Therapy (1999) from the University of Florida. She is Board Certified in Occupational Health, and a member of the American Occupational Therapy Association, the Society for Research on Adolescence, the Society for Research in Child Development and the Florida Therapy Association.

Note: Dr. Kreider is involved in the research study entitled "The Impact of a Child's Disability on the Parents of Children with Rare Diseases"



**Elizabeth "Lee" Kugelmann** — Volunteer

Ms. Kugelmann is a recent graduate of Emory University, Atlanta, GA. She has been a member of BSF since its inception, and is excited to transition into a role of providing services for the community that has supported her for the past fourteen years.

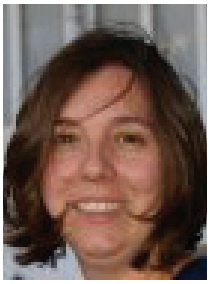
Discsussions: Carrier Issues; Overview of Carrier Issues; Life as a Carrier *(Family);* Carrier Issues *(Siblings)*

**Catherine Le, PhD** — Postdoctoral Research Scholar, The Buck Institute for Research on Aging; Novato, CA, USA

Dr. Le's research interests focus on elucidating the mechanisms by which dietary restriction promotes longevity and healthspan in model organisms.

Dr. Le holds a PhD in Cell and Molecular Biology from Colorado State University, CO, USA, where under the mentorship of Dr. Adam Chicco, she studied the roles which phospholipid metabolism, membrane composition and cardiac mitochondrial dysfunction may play in the development and progression of cardiac pathologies.

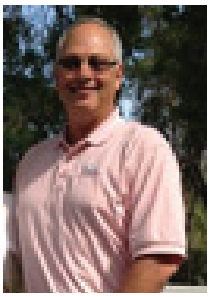
**Presentation:** Substrate-specific Impairment of Oxidative Phosphorylation in *Taz*-deficient Cardiac Mitochondria *(Sci/Med)*



**Rebecca L. McClellan, MGC** — Genetic Counselor, Division of Metabolism, Department of Neurogenetics, Kennedy Krieger Institute, Baltimore, MD, USA

Ms. McClellan joined Dr. Kelley's team at Kennedy Krieger Institute in July 2002 after graduating from the University of Maryland's Master's in Genetic Counseling Program. One of her primary roles at KKI is to assist with both clinical care and research involving families with Barth syndrome. She is also active in the care of all patients of the Metabolism Clinic and coordinates the division's research projects. In addition, Ms. McClellan holds a variety of professional leadership positions in the National Society of Genetic Counselors and enjoys supervising genetic counseling students.

Discussions: Carriers Issues I & II; Life As A Carrier; Putting the Pieces Together; What You Need To Know About Barth Syndrome But Were Afraid To Ask *(Family);* Carrier Issues I & II *(Siblings).* Note: Ms. McClellan is involved in the research study entitled "Multi-Disciplinary Studies in Barth Syndrome" and Genetic Consultations.



**Tom Nurse** — Special Needs Financial Advisor, Manning & Nurse: Personal Financial Advisors for Families with Special Needs, Tampa, FL, USA

Mr. Nurse has been involved in special needs advocacy for nearly twenty-three years after his daughter Shelby was diagnosed with Spastic Quadriplegia Cerebral Palsy and he undertook the role of a 'stay at home father.' Mr. Nurse began full-time work in the disability field with Florida Development Disabilities Council as a Statewide Parent Liaison for early intervention. He worked as a Statewide Parent Consultant for the Florida Department of Health, Children's Medical Services, Early Intervention Program (EIP) and in 1999 joined Family Network on Disabilities of Florida, Inc. Mr. Nurse was awarded a BS in Leisure Service (1983), College of Education, Florida State University.

Today, Mr. Nurse works nationally as an advocate for quality transition planning, self-determination and increasing access to assistive technology for individuals with disabilities. Mr. Nurse and his partner Kevin Manning's firm works with individuals, families, attorneys, trustees, life care planners and other invested parties by providing comprehensive financial services to help address the long-term needs of individuals with special needs.

Presentations: Financial Independence; Breakouts *(Family)*

**Colin K. Phoon, MPhil, MD** — Director, Pediatric & Fetal Echocardiography Lab, New York University Langone Medical Center; Associate Professor, Pediatrics, New York University School of Medicine, New York, NY, USA

Dr. Phoon is a pediatric cardiologist on the faculty of NYU School of Medicine, board certified in General Pediatrics and Pediatric Cardiology. His research focus is the role of mitochondria and its major phospholipid cardiolipin, in the pathogenesis of cardiomyopathy, using an inducible *tafazzin*-knockdown mouse model of Barth syndrome. Dr. Phoon has been a PI or co-investigator on several projects relevant to a broad spectrum of cardiovascular disease in small animal models. Dr. Phoon was awarded the following research grants from BSF entitled "Role of Mitochondria During Myocardial Morphogenesis in Barth Syndrome" (2012); and "Cardiomyopathy in a Mouse Model of Barth Syndrome" (2010).

Dr. Phoon is a fellow of the American Heart Association, the American Society of Echocardiography, and the American Academy of Pediatrics, and a member of the Society for Pediatric Research.

Presentation: Cardiomyopathy and Myocardial Noncompaction in Barth Syndrome *(Sci/Med)*

**William T. Pu, MD** — Associate Professor of Pediatrics, Harvard Medical School; Department of Cardiology, Boston Children's Hospital, Boston, MA, USA; Scientific and Medical Advisory Board, Barth Syndrome Foundation, USA

Dr. Pu's interest is in regulation of heart development, disease, and regeneration, and in using induced pluripotent stem cell technology to model pediatric heart disease. The goals of his research are: (1) to understand the transcriptional network regulating heart development and disease; (2) to understand cell lineage specification in heart development and regeneration; (3) to understand genetic contributions to congenital heart disease. Dr. Pu was awarded the following research grants from BSF entitled "Reactive Oxygen Species and Mitochondrial Dynamics in the Pathogenesis of Barth Syndrome" (2013); "Maturation of Barth Syndrome Models for Clinical Translation" (2012); "Using Induced Pluripotent Stem Cells and Modified RNAs to Model and Correct Barth Syndrome" (2011); "Analysis of Metabolic Abnormalities in *TAZ*-deficient Cardiomyocytes" (2009).

Dr. Pu holds an MD from Harvard Medical School. He completed his internship, residency, and pediatric cardiology training at Boston Children's Hospital. He is Board Certified in Pediatrics and Pediatric Cardiology.

Chair — Animal Models *(Sci/Med)*
Presentation: Modeling the Mitochondrial Myopathy of Barth Syndrome using iPSC and Heart-on-chip Technologies *(Sci/Med)*

**Stacey Reynolds, PhD, OTR/L** — Associate Professor, College of Public Health and Health Professions, University of Florida, Gainesville, FL; Assistant Professor, Department of Occupational Therapy, Virginia Commonwealth University, Richmond, VA, USA

Dr. Reynolds is currently a K12 Scholar. Her current research, conducted through collaborations with the Department of Psychology at the University of Florida, is focused on developing an animal model for studying sensory processing disorders and examining the neurobiological basis for atypical sensory and motor behaviors. Dr. Reynolds has over five years of clinical experience working with children and teaches courses in pediatric intervention and assessment.

Dr. Reynolds completed her PhD in Health Related Science at Virginia Commonwealth University in 2007. Her research has focused on investigating physiologic stress reactivity patterns in children with Sensory Processing Disorder, and characterizing behavioral and physiological patterns of sensory processing in children with ADHD, Autism and Mood Disorders. Dr. Reynolds was awarded a research grant from BSF entitled "A Systematic Investigation into Sensory and Motor-based Feeding Issues in Boys with Barth Syndrome" (2013).

Presentations: Taste Sensitivity, Food Preference, and Feeding Behaviors in Children with Barth Syndrome *(Sci/Med)*
Note: Dr. Reynolds is involved in the research study entitled "A Systematic Investigation into Sensory and Motor-based Feeding Issues in Boys with Barth Syndrome"

**Debbie Riddiford,** Barth Syndrome Clinical Nurse Specialist (CNS), Bristol Royal Hospital for Children, Bristol, United Kingdom

Ms. Riddiford was appointed as a Cardiac Liaison Nurse (CNS) at the new Bristol Royal Hospital for Children in 2006 and still works part-time in this position. She became the Barth Syndrome CNS at inception of the service in 2010. She has a broad remit within the service, acting as a focal point for enquiries from patients, families and health professionals. She has a major role in provision of practical and emotional support for patients and their families, as well as acting as their advocate. She is also responsible for the planning of the six monthly multidisciplinary clinics and for arranging other admissions or outpatient investigations for patients who have developed interim problems requiring more urgent investigation or management.

Ms. Riddiford qualified as a pediatric nurse in 1984. She has held positions in the Baby Unit, cardiac ward and Sebastian Diamond Sleep Laboratory at the Bristol Royal Hospital for Sick Children, United Kingdom.

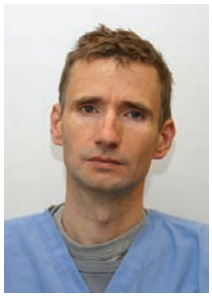
Discussions: What You Need To Know About Barth Syndrome But Were Afraid To Ask; Healthcare Transitions; Breakouts *(Family)*

**Yana Sandlers, MSc, PhD** — Assistant Director, Biochemical Genetics Laboratory, Kennedy Krieger Institute, Johns Hopkins School of Medicine, Baltimore, MD, USA

Dr. Sandlers's research is focused on understanding biochemical abnormalities underlying different metabolic conditions, especially inborn errors of metabolism and mitochondrial disorders. Her research involves GC-MS, LC-MS, and tandem mass spectrometry techniques for development of novel analytical methodologies to facilitate early diagnosis of different metabolic disorders. Dr. Sandlers was awarded a research grant from BSF entitled "Characterization of Biochemical Abnormalities in Barth Syndrome Patients and Mouse Model of Barth Syndrome" (2011).

Dr. Sandlers holds a doctorate in chemistry from the Technion - Israel Institute of Technology. She completed a post-doctoral fellowship in nutritional biochemistry at Case Western Reserve University, OH, USA. Dr. Sandlers has been with the Kennedy Krieger Institute since 2009. She is a member of the American Society of Mass Spectrometry and the Society for Inherited Metabolic Disorders.

Note: Dr. Sanders is involved in the research study entitled "Multi-Disciplinary Studies in Barth Syndrome"



**Michael Schlame, MD** — Associate Professor of Anesthesiology & Cell Biology, New York University School of Medicine; Director, Cardiothoracic Anesthesia, New York University Langone Medical Center, New York, NY; Chairman, Scientific and Medical Advisory Board, Barth Syndrome Foundation, USA

Dr. Schlame's subspecialties include cardiothoracic anesthesiology and critical care, and his research interests include Barth syndrome, lipids and mitochondria (with particular concentration on mitochondrial energy metabolism), pulmonary surfactant, cardiolipin, mechanisms of multiple organ failure, and cardiomyopathy. His clinical focus includes adult and pediatric critical care, cardiothoracic anesthesia, and pediatric anesthesia.

Dr. Schlame is board certified in Anesthesiology both in the US and in Europe. He trained at Charité University Hospital in Berlin, at New York Presbyterian Hospital, and at New York University Medical Center in New York.

Chair—Mitochondrial Lipids *(Sci/Med)*

**Colin G. Steward, PhD, FRCP, FRCPCH** — Consultant in Bone Marrow Transportation, Royal Hospital for Children; Reader in Stem Cell Transplantation, School of Cellular & Molecular Medicine, School of Medical Sciences, University of Bristol, Bristol, United Kingdom; Scientific and Medical Advisory Board, Barth Syndrome Foundation, USA

Dr. Steward is a Consultant at the Bristol Royal Hospital for Children, UK. He is Clinical Lead for a multidisciplinary NHS national service for Barth syndrome in the UK which is run in partnership with Michaela Damin and her colleagues at the Barth Syndrome Trust. Debbie Riddiford (Clinical Nurse Specialist), Ann Bowron (Clinical Biochemist), Dr. Vanessa Garratt (Clinical Psychologist) and Nicol Clayton (Specialist Dietician) from the same service are at this Conference. The service provides free diagnostic testing for UK residents by cardiolipin analysis and *TAZ* gene sequencing, annual multidisciplinary clinic for boys and young men, and centralized prescribing and delivery of G-CSF. Dr. Steward's particular interests are in the management of neutropenia in Barth syndrome and improving disease recognition/testing to overcome current underdiagnosis of the disease — so far 32 unrelated families have been identified in the UK. Dr. Steward is a recipient of the Barth Syndrome Foundation's Varner Award for Pioneers in Science and Medicine (2012).

Chair — Mitochondrial Physiology *(Sci/Med)*
Presentations: The National Health Service Barth Syndrome Service: A Progress Report *(Sci/Med);* What You Need To Know About Barth Syndrome But Were Afraid To Ask *(Family)*; Discussion: Neutropenia *(Affected Individuals)*

**Douglas Strathdee, PhD** — Head of the Transgenic Technology Lab, The Beatson Institute for Cancer Research, Glasgow, United Kingdom

Dr. Strathdee's research interests focus generating and studying models of human diseases. The aim of the research in his lab is to use stem cells to model the processes underlying cancer and to uncover the roles that novel stem cell and reprogramming factors play in the development of the disease. Dr. Strathdee was awarded the following research grant from BSF entitled "Characterization of a Conditional Knockout of *Tafazzin* in the Mouse" (2013).

Dr. Strathdee previously held positions at as Senior Research Associate, Wellcome Trust Sanger Institute, Cambridgeshire, UK; Postdoctoral Fellow, at the University of Edinburgh, and Research Scientist at Roslin Institute, University of Edinburgh, UK. Dr. Strathdee holds a PhD and a BSc in Immunology (Honors) from the University of Glasgow.

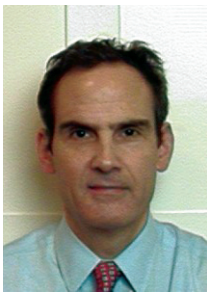
Presentation: Engineering Precise Genetic Alterations at the *Taz* Locus *(Sci/Med)*

**Arnold W. Strauss, MD** — BK Rachford Professor and Chair, Department of Pediatrics, University of Cincinnati College of Medicine; Director, Cincinnati Children's Research Foundation; Chief Medical Officer, Cincinnati Children's Hospital Medical Center; Professor, UC Department of Pediatrics, Cincinnati Children´s Hospital, Cincinnati, OH; Scientific and Medical Advisory Board, Barth Syndrome Foundation, USA

Dr. Strauss's research focuses on understanding the molecular basis of disorders of mitochondrial fatty acid oxidation and genetic causes of congenital heart disease and cardiomyopathies.

Dr. Strauss is the recipient of two prestigious awards in research: the American Heart Association's Basic Science Research Award for groundbreaking work that led to finding genetic defects that can cause heart failure and sudden death in infants and children and the E. Mead Johnson Award for Excellence in Pediatric Research. Dr. Strauss has been a supporter of BSF since its formation, both as a physician and as a researcher. Dr. Strauss has been awarded the following research grants from the Barth Syndrome Foundation: "*Tafazzin* Function in Animal Models of Barth Syndrome" (2004); "A Mouse Gene Ablation Model of Barth Syndrome" (2002).

Chair—Clinical Studies on Barth Syndrome *(Sci/Med)*



**W. Reid Thompson, MD** — Assistant Professor, Pediatric Cardiology, Johns Hopkins Children's Center, Baltimore, MD, USA

Dr. Thompson's research interests include advanced imaging of cardiomyopathy. He has been involved in the clinical care of patients with Barth syndrome for over 20 years at Johns Hopkins and at the Kennedy Krieger Institute.

Dr. Thompson received his MD (1984) from Wake Forest University, Bowman Gray School of Medicine, NC, USA. He completed his Internship and Residency in Pediatrics (1987) at The Johns Hopkins Hospital, MD. USA. Dr. Thompson completed his Fellowship in Pediatric Cardiology at the Children's Hospital, Boston Harvard Medical School. He is certified by the American Board of Pediatrics in Pediatric Cardiology (2002).

Note: Dr. Thompson is involved in the research study entitled "Multi-Disciplinary Studies in Barth Syndrome"

**Matthew J. Toth, PhD** — Science Director, Barth Syndrome Foundation, Inc.; Scientific and Medical Advisory Board, *ex-officio,* Barth Syndrome Foundation; Advisory Board, Barth Syndrome Registry & Biorepository, Iselin, NJ, USA

Dr. Toth completed his PhD in Microbiology from MIT in 1988 and joined a large pharmaceutical company where he was part of several teams involved with drug discovery projects in the therapeutic areas of inflammation and cardiovascular diseases. His laboratory eventually concentrated on making and testing genetically altered mice as a way to advance drug discovery programs. After 2002, Dr. Toth joined a smaller pharmaceutical company and eventually a biotech company where he led several drug discovery programs in the areas of pain and orphan diseases. Since July of 2006, Dr. Toth has been the Science Director of the BSF, where he uses his experience in guiding efforts towards finding treatments and eventually a cure for Barth syndrome.

Chair — Science & Medicine Sessions
Discussion: Research Update *(Family)*

**Jeffrey Towbin, MD, FAAP, FACC, FAHA** — Executive Director of the Heart Institute and Professor & Chief, Pediatric Cardiology, Cincinnati Children's Hospital Medical Center, Cincinnati, OH, USA; Scientific and Medical Advisory Board, Barth Syndrome Foundation, USA

Dr. Towbin is a well-known expert in the clinical and translational arenas of Pediatric Cardiomyopathy, Heart Failure, transplantation, and causes of sudden death. Dr. Towbin has published approximately 480 peer-reviewed publications, and over 70 book chapters, as well as editing three books. He has been awarded many National Institutes of Health (NIH) grants with continuous funding over his career, and is the recipient of various other extramural grants, including several grants mentoring students and junior faculty.

Dr. Towbin completed his Pediatric Residency at Cincinnati Children's Hospital Medical Center and his Pediatric Cardiology Fellowship at Texas Children's Hospital in 1985. Dr. Towbin has also received many honors nationally and internationally, including the Michael Debakey Excellence in Research Award, 2007, the American College of Cardiology Distinguished Scientist Award (March 2007), and the 2013 American Heart Association Basic Science Prize. He has given over 30 named Lectureships as well as played leadership roles on many National and International Committees.

Presentation: Transplants *(Family)*

**Hilary Vernon, MD, PhD** — Assistant Professor, Genetic Medicine, McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University, and at the Kennedy Krieger Institute, Baltimore, MD, USA

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 co-director of the Barth Syndrome Interdisciplinary Clinic at the Kennedy Krieger Institute. She also serves on the Maryland State Advisory Council on Hereditary and Congenital Disorders.

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.

Presentation: Kennedy Krieger Barth Syndrome Initiatives: Results from the 2012 Barth Syndrome Foundation Conference Clinical Laboratory Study and An Overview of the Barth Syndrome Interdisciplinary Clinic *(Sci/Med).* Note: Dr. Vernon is involved in the research study entitled ""Multi-Disciplinary Studies in Barth Syndrome" *(Family)*.

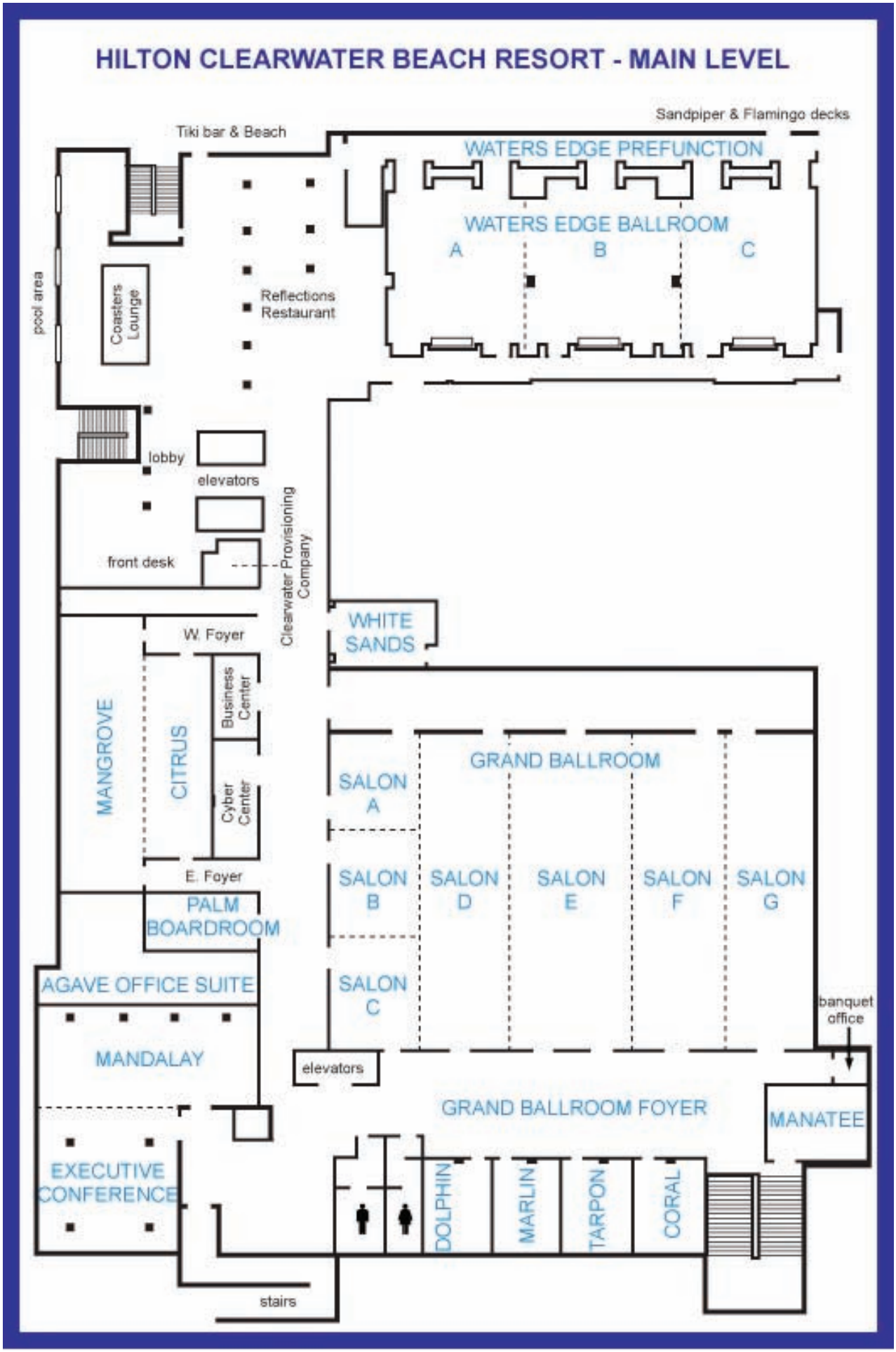
**Jun Zhang** — Joint-training PhD Candidate, Department of Cellular and Molecular Physiology, Penn State College of Medicine, State College, PA, USA

Mr. Zhang's research interests focus on the molecular mechanisms of acyl-CoA dependent lysocardiolipin acyltransferase-1 (ALCAT1) underlying mitochondrial dysfunction in obesity and type 2 diabetes; and the mechanism of *tafazzin* in cardiomyopathy.

Mr. Zhang holds bachelor's and master's degrees from Beijing Normal University, China.

Presentation: Regulation of Cardiomyopathy in Barth Syndrome by ALCAT1 *(Sci/Med)*

NOTES


HILTON CLEARWATER BEACH RESORT - MAIN LEVEL
Sandpiper & Flamingo decks
Tiki bar & Beach
WATERS EDGE PREFUNCTION
WATERS EDGE BALLROOM
A
B
C
pool area
Coasters Lounge
Reflections Restaurant
lobby
elevators
front desk
Clearwater Provisioning Company
WHITE SANDS
W. Foyer
Business Center
Cyber Center
MANGROVE
CITRUS
GRAND BALLROOM
SALON A
SALON B
SALON C
SALON D
SALON E
SALON F
SALON G
E. Foyer
PALM BOARDROOM
AGAVE OFFICE SUITE
MANDALAY
banquet office
elevators
GRAND BALLROOM FOYER
MANATEE
EXECUTIVE CONFERENCE
DOLPHIN
MARLIN
TARPON
CORAL
stairs

The Barth Syndrome Foundation would like to thank the following Institutes and Center from the National Institutes of Health (NIH) for their support of the Scientific and Medical Sessions:

**National Heart, Lung and Blood Institute (NHLBI)**

**The *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD)**

**National Center for Advancing Translational Sciences (NCATS)**