Barth Syndrome
8th International Scientific, Medical & Family Conference

July 18–23, 2016
Clearwater, Florida

Conference is the Best of #TeamBarth

Families, you are #TeamBarth because of your unconditional love for the affected "Barth guy" in your life. Barth syndrome brings challenges that only other Barth families uniquely understand. Discussing your experiences together, in one place, helps ease worries and answers questions about every issue imaginable. The family sessions cover topics from practical daily living tips to medications and social aspects. We also discuss education, the transition to adulthood, and much more.

Researchers and clinicians, you are #TeamBarth because of 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.

Volunteers, you are #TeamBarth because of your dedication to our mission. Without you, this conference would not be possible. Your devotion and "sweat equity" inspires us.

Donors, you are #TeamBarth because of your investment in our cause. Your generous donations make all of this possible. Bringing families, clinicians, and researchers together 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.

To the affected boys and men, we are all here for you! Yes, we are ALL #TeamBarth, and each one of us plays a valuable role in the game. Now, let's go make some wonderful memories together!

Lindsay Groff

KEYNOTE ADDRESS

What We as Families, Advocates, Scientists and Industry Partners Can Do to Move Barth Syndrome More Fully Into the Clinical Treatment Era

Thursday, July 21, 2016

(Salon D, E, F)

Ronald J. Bartek — Co-founder and Founding President, Friedreich's Ataxia Research Alliance; Chairman of the Board of the National Organization for Rare Disorders (NORD); 4-year member of the National Institutes of Health (NIH) National Advisory Neurological Disorders and Stroke Council, Washington, DC, USA

Mr. Bartek is Friedreich's Ataxia Research Alliance's (FARA) Co-founder and President. He also is Chairman of the Board of the National Organization for Rare Disorders (NORD); a 4-year member, National Institutes of Health (NIH) National Advisory Neurological Disorders and Stroke (NINDS) Council; and former partner and president of a business and technology development, consulting and government affairs firm.

Friedreich's Ataxia is a genetic disorder with some important clinical similarities to Barth syndrome. FARA is an organization that communicates (in collaboration with other organizations) with government agencies at both state and federal levels in pursuit of policies and decisions intended to advance therapeutic development for the disorder. Mr. Bartek is well-respected and trusted by peer patient advocacy leaders, policy makers in Washington, DC, leaders of numerous divisions of the NIH and by people he serves through FARA. He is known by all as a man who is thoughtful, who cares deeply and who gets results. Most importantly, he is a father who got involved to help his son. Though Keith sadly died in January 2010, Mr. Bartek has continued to work for treatments for all those who have Friedreich's Ataxia or who suffer from a rare disease. Mr. Bartek has been a friend and advisor to the Barth Syndrome Foundation (BSF) from the very beginning. He and FARA have traveled down a very similar road to BSF to translate scientific advances into actual therapies. In his address, Mr. Bartek will offer us all some first-hand insights into this process and will emphasize patients' critical roles.

From all of his work in the rare disease field, he has a great deal of experience and expertise in topics very relevant to all of us involved in BSF. Mr. Bartek and his organization are considered by many to be pioneers in the rare disease arena. We are excited for our attendees to hear Mr. Bartek's inspiring message of hope and triumph, as told by someone who has many years of first-hand experience.

PRE-CONFERENCE SESSIONS

TIME/LOCATION	SUNDAY, JULY 17, 2016
Afternoon *(Executive Conference Room)*	**Portraits by Amanda Clark** Early family arrivals scheduled on this date
TIME/LOCATION/FACILITATORS	**MONDAY, JULY 18, 2016**
All Day *(Executive Conference Room)* All Day *(Grand Ballroom Foyer)*	**Portraits by Amanda Clark** Early family arrivals scheduled on this date **Registration** Pick up badges / Drop items for family "goody bags"
1:00pm—2:00pm *(Salon D, E, F)* *Donna Strain, RN, Volunteer*	**Orientation and Walkthrough Meeting** Audience: New families and their mentors only
2:30pm—4:00pm *(Salon D, E, F)* *Lindsay Groff, Executive Director; Marc Sernel, Chairman, Board of Directors; Shelley Bowen, Director, Family Services & Awareness – Barth Syndrome Foundation*	**Welcome Event** Audience: All conference attendees (Children, Parents, Grandparents, Science/ Medicine attendees) Overview of conference, family introductions, housekeeping
4:00pm—5:00pm *(Salon D, E, F)* *Shelley Bowen — Director, Family Services & Awareness, Barth Syndrome Foundation*	**Consent and Assent Signing** Audience: The following groups of individuals participating in clinics must attend this session: **Consents** Parents of all boys under 18 years of age Adult males 18 years of age and older **Assents** Minor affected males (12–18 years of age)
	Dinner on your own
6:00pm—7:00pm *(Water's Edge)* *Matthew J. Toth, PhD – Science Director, Barth Syndrome Foundation*	**Therapeutic ideas for Barth syndrome** *(Dinner will be provided)* Audience: Affected individuals 16+ years of age

BARTH SYNDROME RESEARCH STUDIES
TUESDAY & WEDNESDAY
JULY 19-20, 2016
7:30am—5:30pm
(Grand Ballroom Foyer - Clinic Registration Desk)

Family participation in clinical research has provided the primary resource of clinical data collection since 2002. Findings from these studies have resulted in the publication of numerous peer-reviewed journal articles to educate physicians and researchers about various aspects of Barth syndrome. This year, families will have the opportunity to participate in multiple IRB-approved research studies. Since most of the segments of these studies do not require a parent to be present, we have scheduled families into four age-specific blocks. A "Study Buddy" will escort minor children to the various sections of the research studies while parents attend small group focus sessions. This will, hopefully, provide families time to catch up with friends and meet new friends prior to the educational sessions. Clinic check-in will be at the Clinic Registration Desk located in the Grand Ballroom Foyer.

RESEARCH STUDIES

Investigation into clinical, metabolic and molecular factors in Barth syndrome
Principal Investigator: Hilary J. Vernon, MD, PhD — Assistant Professor, McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University; Kennedy Krieger Institute, Baltimore, MD

Segment of Study	Who can participate	Age Range	Must a parent be present
Six-minute walk test and grip strength	Affected males and male relatives	3+ years of age	No
Grip strength, knee extensor and SWAY balance	Affected males and male relatives	3+ years of age	No
Biochemical and molecular studies (collection of blood)	Affected males and first-degree relatives of individuals participating in this study (parents, brothers, and sisters)	All	Yes
Survey	Affected males	14+ years of age	No

Olfactory identification in patients affected by Barth syndrome
Principal Investigator: Angela Corcelli, PhD — Professor of Physiology, Department of Basic Medical Sciences, Neurosciences and Sensory Organs, University of Bari Aldo Moro, Bari, Italy

Segment of Study	Who can participate	Age Range	Must a parent be present
Scent study	Affected males and male relatives	5-18 years of age	No

Longitudinal evaluation of cardiomyopathy and outcome in Barth syndrome
Principal Investigator: Carolyn Taylor, MD — Associate Professor of Pediatrics, Medical University of South Carolina; Director, Pediatric Echocardiography Laboratory, Children's Hospital, Charleston, SC, USA

Segment of Study	Who can participate
Echocardiogram, EKG, and questionnaire	Affected males of any age who have participated in previous cardiology studies led by Dr. Taylor

The impact of a child's disability on the parents of children with rare diseases
Principal Investigator: Yoonjeong Lim, PhD, OTR/L — Research Associate, Department of Occupational Therapy, College of Public Health and Health Professions, University of Florida, Gainesville, FL, USA

Segment of Study	Who can participate
Parent survey and interview	Parents of an affected male between 5–19 years of age and follow-up survey among parents who participated in Dr. Lim's study in 2014

Heart and skeletal muscle metabolism in Barth syndrome
Principal Investigator: W. Todd Cade, PT, PhD — Associate Professor of Physical Therapy and Internal Medicine, Washington University School of Medicine, St. Louis, MO, USA

Segment of Study	Who can participate
Biochemical studies (collection of blood)	Affected males who have participated in the 3-4 day study in St. Louis, MO

TUESDAY & WEDNESDAY, JULY 19– 20, 2016

SMALL GROUP MEETINGS, CLINICS & CONSULTATIONS 8:30am–5:00pm	
All Day *(Grand Ballroom Foyer)* *Tuesday & Wednesday*	**Registration** Pick up badges / Drop items for family "goody bags"
30 minute session *(Executive Conference Room)*	**Photos by Amanda Clark**

INDIVIDUAL CONSULTATIONS

CONSULTATION/FACILITATORS	
Barth Syndrome Registry & Repository	
Shelley Bowen — Director, Family Services & Awareness, Barth Syndrome Foundation	Audience: For new/potential participants: Full details about the Registry and assistance with enrollment For existing participants: Assistance with updating entries
Genetics	
Iris L. Gonzalez, PhD — A. I. duPont Hospital for Children, Wilmington, DE; Rebecca L. McClellan, MGC, CGC — Kennedy Krieger Institute, Baltimore, MD	Audience: For all, and especially for 1st time attendees Information about all aspects of genetics and how you, your extended family, and any future offspring may be affected. Additionally, information needed for the Registry: collecting your genetic pedigree.
Pill Swallowing Workshop	
Stacey Reynolds, PhD, OTR/L; Emily Burgess, MS, OTR/L — Virginia Commonwealth University, Richmond, VA	Audience: Affected individuals 6+ years of age (parents welcome) Techniques and equipment for pill swallowing – creating a personalized program in a stress free environment.
Vitals	
Sunstar Paramedics; Sue Wilkins, RN	Height, weight, blood pressure.
Labs (Molecular and biochemical blood draws)	
Misty N. Regan, BSN, RN, CPEN *Fatima Singletary, CPT*	Audience: Affected males who have participated in Dr. Cade's study in St. Louis and affected males and all immediate family members (parents, brothers, and sisters of all ages) of those whose son is participating in research during this conference.

SMALL GROUP MEETINGS

Note: Families will be placed in groups based on age of children and will rotate together, visiting some/all of the following:

MEETING/TIME/FACILITATORS	
Life as a Carrier **Tuesday, July 19th: 3:00pm—5:00pm**	
Rebecca L. McClellan, MGC, CGC — Johns Hopkins Medicine; Kennedy Krieger Institute, Baltimore, MD; Lee Kugelmann — Co-Lead, Carrier Program, Barth Syndrome Foundation	Audience: Open to all carriers and potential carriers 15+ years of age girls, mothers/grandmothers This small group 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.
Carrier Issues Session 1: Pre-Test Carrier Issues **Wednesday, July 20th: 3:00pm—4:00pm**	
Rebecca L. McClellan, MGC, CGC — Johns Hopkins Medicine; Kennedy Krieger Institute, Baltimore, MD; Lee Kugelmann — Co-Lead, Carrier Program, Barth Syndrome Foundation	Audience: Potential carriers 15+ years of age This small group 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.

TUESDAY & WEDNESDAY, JULY 19– 20, 2016

SMALL GROUP MEETINGS, CLINICS & CONSULTATIONS
8:30am—5:00pm

MEETING/FACILITATORS	
Carrier Issues Session 2: Post-Test Carrier Issues **Wednesday, July 20th: 4:00pm—5:00pm**	
Rebecca L. McClellan, MGC, CGC — Johns Hopkins Medicine; Kennedy Krieger Institute, Baltimore, MD; Lee Kugelmann — Co-Lead, Carrier Program, Barth Syndrome Foundation	Audience: Potential carriers and those who have recently undergone testing 15+ years of age This small group 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.
Promoting Independence: Strategies to Help Children with Special Needs Reach their True Potential	
Danielle Goodman, OT — NHS Barth Syndrome Service, Bristol Royal Hospital for Children, Bristol, United Kingdom	Audience: Affected individuals 4-8 years of age (parents & children) Audience: Affected individuals 8-12 years of age (parents & children) Audience: Affected individuals 13+ years of age (parents & children) This session will help identify practical ways in which to promote independence through problem solving and tailored techniques.
Neutropenia	
Colin Steward, PhD, FRCP, FRCPCH — Bristol Royal Hospital for Children; Clinic Lead, NHS Barth Syndrome Service, Bristol, United Kingdom	Audience: Two sessions for parents of affected individuals only Audience: One session for affected individuals 16+ years of age This question and answer session will address concerns and/or issues pertaining to the hematologic aspects of Barth syndrome.
Nutrition	
Nicol Clayton, Advanced Specialist Dietitian; Danielle Goodman, OT — NHS Barth Syndrome Service, Bristol Royal Hospital for Children, Bristol, United Kingdom	Audience: Affected individuals 0-4 years of age (parents only) Audience: Affected individuals 4-8 years of age (parents only) Audience: Affected individuals 8-12 years of age (parents & child) Audience: Affected individuals 13+ years of age (parents & child) This session will share experiences of patterns of restricted eating, provide general guidance on calorie and protein requirements for appropriate growth for each age group, outline suggested approaches to use, and suggest ways to use play and sensory programs.
Where's my Cure for Barth Syndrome?	
Matthew J. Toth, PhD — Science Director, Barth Syndrome Foundation	Audience: Parents of affected individuals 0-4 years of age Audience: Parents of affected individuals 4-8 years of age Audience: Parents of affected individuals 8-12 years of age Audience: Parents of affected individuals 13+ years of age A simplified summary of potential therapies for Barth syndrome.

Lanyards
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CONFERENCE OVERVIEW

THURSDAY, JULY 21, 2016

7:30am – 8:15am: Breakfast Buffet *(Salon D, E, F)*

Scientific & Medical Sessions *(Salon G)*		Family Sessions *(Salon A, B, C)*		AFFECTED INDIVIDUALS and SIBLINGS SESSIONS *(Water's Edge)*	
	Pathomechanisms(s) of Barth syndrome	9:00am— 9:30am	The rewards of risk-taking	8:30am—9:30am	Barth battle stations *(Former Fitness Center, 3rd Floor) (Boys up to 8 years of age)*
8:35am—9:05am	• William T. Pu, MD	9:35am—11:00am	The new frontier: Future clinical trials		
9:05am—9:35am	• Adam J. Chicco, PhD	11:00am—11:15am	Break	9:30am—10:30am	Barth battle stations *(Former Fitness Center, 3rd Floor) (Boys 9+ years of age)* (older guys to cheer on!)
9:35am—10:05am	• Colin K. L. Phoon, MD, MPhil	11:15am—11:50am	Skills and practical strategies to support feeding		
10:05am—10:20am	• Coffee Break				
10:20am—10:50am	• Saskia Wortmann-Hagemann, MD, PhD				
10:50am—11:20am	• Laura Cole, PhD			10:45am—12:00pm	Photo scavenger hunt
11:20am—11:50am	• Douglas Strathdee, PhD				

12:00pm – 1:30pm: Keynote Speaker Luncheon *(Salon D, E, F)*

	Potential Therapies for Barth Syndrome *(combined session) (Salon D, E, F)*			2:30pm—3:30pm	Transition Session (combined youth 8-15 years of age)
2:05pm—2:35pm	• W. Todd Cade, PT, PhD			3:30pm—4:00pm	Transition Session (combined youth 16+ years of age)
2:35pm—3:05pm	• Yuguang (Roger) Shi, PhD				
3:05pm—3:35pm	• Michael T. Chin, MD, PhD, FACC, FAHA			2:30pm—3:00pm	Understanding Genetics (combined youth 15+ years of age)
3:35pm—3:50pm	• Coffee Break			3:30pm—4:00pm	Understanding Genetics (combined youth 8-15 years of age)
3:50pm—4:20pm	• Zaza Khuchua, PhD				
4:20pm—4:50pm	• Mindong Ren, PhD				
4:50pm—5:20pm	• Christina A. Pacak, PhD				
5:20pm—5:50pm	• Hilary J. Vernon, MD, PhD				
6:00pm—8:00pm	Poster Session *(Grand Ballroom Foyer)*	7:00pm—8:00pm	Poster Session *(Grand Ballroom Foyer)*	4:00pm—5:50pm	Youth activities (combined youth)

8:00pm – 10:30pm: Luminaries on the beach

FRIDAY, JULY 22, 2016

7:30am – 8:15am: Breakfast Buffet *(Salon D, E, F)*

Scientific & Medical Sessions *(Salon G)*		Family Sessions *(Salon A, B, C)*		AFFECTED INDIVIDUALS and SIBLINGS SESSIONS *(Water's Edge)*	
	Clinical characteristics of Barth syndrome	8:30am—8:50am	Evaluation of cardiac function in the current era	8:20am—8:50am	Youth Group Photo
8:35am—9:05am	• Colin G. Steward, PhD, FRCP, FRCPCH	8:50am—9:10am	Medications used to treat heart failure	8:50am—10:45am	Sandcastle building
9:05am—9:35am	• Cynthia James, ScM, PhD	9:10am—9:30am	Advanced therapies for heart failure in Barth syndrome	11:15am—12:00pm	Q & A (younger & older affected individuals)
9:35am—10:05am	• Brittany DeCroes Hornby, PT, DPT, PCS	9:30am—9:50am	Mitochondria and Barth syndrome	11:15am—12:00pm	Carrier Issues: Session 3 (girls 10-14 years of age)
10:05am—10:20am	• Coffee Break	9:50am—10:00am	Q & A	11:15am—12:00pm	Sib Connect (brothers and sisters 8+ years of age)
10:20am—10:50am	• John Lynn Jefferies, MD, MPH, FAAP, FACC, FAHA	10:00am—10:10am	Break		
10:50am—11:20am	• Two Poster Presenters	10:10am—10:45am	Updates and Results from the 2014 BSF/Johns Hopkins Study		
		10:45am—10:55am	Vitamin and mineral supplementation in Barth syndrome		
		11:00am—11:45am	Participation in clinical research: Lessons learned in St. Louis		
		11:45am—11:55am	Q & A		

12:00pm – 2:00pm: Varner Award Luncheon *(Salon D, E, F)*
12:00pm – 2:00pm: Youth Luncheon (Combined youth) w/ special guests "The Rough Riders" *(Sandpiper Deck)*

	Cardiolipin and Barth syndrome	2:10pm—2:25pm	From DNA to *TAZ* variants...is this a mutation or not?	2:15pm—4:30pm	Youth activities (keepsakes, crafts, videos) (combined youth)
2:05pm—2:35pm	• Robin Duncan, PhD	2:30pm—3:15pm	Tips and strategies for talking to your children about genetics		
2:35pm—3:05pm	• Two Poster Presenters (15 minutes each)	3:15pm—4:15pm	Study update: How do women adapt to being a Barth syndrome carrier?		
3:05pm—3:35pm	• Valerian E. Kagan, PhD, DSc	4:15pm—5:00pm	What we have learned, where to go from here, and how you can get involved? *(All carrier community)*		
3:35pm—3:50pm	• Coffee Break				
3:50pm—4:20pm	• Miriam L. Greenberg, PhD				
4:20pm—4:50pm	• Nathan N. Alder, PhD				
4:50pm—5:20pm	**Conference wrap-up**				

7:00pm – 11:00pm: Dinner & Social Event *(Salon D, E, F)*

SATURDAY, JULY 23, 2016

Scientific & Medical Sessions *(Salon G)*		Family Sessions *(Salon A, B, C)*		AFFECTED INDIVIDUALS and SIBLINGS SESSIONS *(Water's Edge)*	
8:30am—11:30am	SMAB breakfast & meeting (by invitation only)	9:30am—10:15pm	Barth battle stations! Video highlights of Physio sessions and top tips to take home	9:30am—12:10pm	Conference Wrap-up; project finals, kid activities (combined youth)
		10:15am—11:30am	Transition planning: Maximizing independence in health care, vocation planning and life skills		
		11:45am—12:15pm	Pill swallowing school: Lessons learned		

12:15pm – 2:00pm: Luncheon Buffet & Finale *(Salon D, E, F)*

SCIENCE & MEDICINE SESSIONS
Thursday, July 21, 2016

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: BREAKFAST *(Salon D, E, F)*

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

8:30am—11:50am: PATHOMECHANISM(S) OF BARTH SYNDROME *(Salon G)*
Chair: W. Todd Cade, PT, PhD, Washington University School of Medicine, St. Louis, MO

8:30am—8:35am	Introduction — Jack Higgins
8:35am—9:05am	**Reactive oxygen species cause abnormal calcium handling in Barth syndrome stem cell-derived cardiomyocytes** ~ *William T. Pu, MD — Boston Children's Hospital, Boston, MA*
9:05am—9:35am	**Cardiac proteomics distinguish human BTHS from pediatric heart failure and reveal patterns of metabolic remodeling common to *Taz* shRNA mice** ~ *Adam J. Chicco, PhD — Colorado State University, Ft. Collins, CO*
9:35am—10:05am	**Recapitulating the Barth cardiomyopathy in the inducible mouse model of *tafazzin* deficiency** ~ *Colin K. L. Phoon, MD, MPhil — New York University School of Medicine, New York, NY*
10:05am—10:20am	Coffee Break
10:20am—10:50am	**The growing family of disorders in the biosynthesis and remodelling of phospholipids; the brothers and sisters of Barth syndrome** ~ *Saskia Wortmann-Hagemann, MD, PhD — Salzburger Landeskliniken, Salzburg, Austria*
10:50am—11:20am	**Elevated hepatic fatty acid oxidation contributes to low body weight in *tafazzin* knock-down mice** ~ *Laura Cole, PhD — Children's Hospital Research Institute of Manitoba, Winnipeg, Manitoba, Canada*
11:20am—11:50am	**The phenotype of *Taz* null mutation in mouse closely resembles Barth syndrome** ~ *Douglas Strathdee, PhD — Beatson Institute, Glasgow, Scotland*

12:00pm—1:30pm	**Keynote Speaker Luncheon** *(Salon D, E, F)* **Ronald J. Bartek** — Co-Founder/Founding President, Friedreich's Ataxia Research Alliance (FARA) *"What we as families, advocates, scientists, and industry partners can do to move Barth syndrome more fully into the clinical treatment era." (Introduction by Shelley Bowen)*
1:30pm—2:00pm	**Scientific & Medical Attendees Group Photo** *(Sandpiper Deck)*

2:00pm—5:50pm: POTENTIAL THERAPIES FOR BARTH SYNDROME *(Salon D, E, F)*
Chair: Arnold Strauss, MD, Cincinnati Children's Research Foundation, Cincinnati, OH

2:00pm—2:05pm	Introduction — Michelle Florez
2:05pm—2:35pm	**Update on exercise and substrate metabolism studies in Barth syndrome** ~ *W. Todd Cade, PT, PhD — Washington University School of Medicine, St. Louis, MO*
2:35pm—3:05pm	**Targeted deletion of ALCAT1 mitigates cardiomyopathy in mouse model of Barth syndrome through restoration of mitophagy** ~ *Yuguang (Roger) Shi, PhD — University of Texas Health Science Center at San Antonio, San Antonio, TX*
3:05pm—3:35pm	***Tafazzin* enzyme replacement therapy for Barth syndrome** ~ *Michael T. Chin, MD, PhD, FACC, FAHA — University of Washington, Seattle, WA*
3:35pm—3:50pm	Coffee Break
3:50pm—4:20pm	**Targeting PPAR/PGC1alpha axis in Barth syndrome: The PPAR pan-agonist bezafibrate ameliorates cardiomyopathy in a mouse model of Barth syndrome** ~ *Zaza Khuchua, PhD — Cincinnati Children's Hospital Medical Center, Cincinnati, OH*
4:20pm—4:50pm	**Assessing the effects of Bezafibrate on the cardiomyopathy induced by adult-onset knockdown of *tafazzin* in a mouse model** ~ *Mindong Ren, PhD — New York University School of Medicine, New York, NY*
4:50pm—5:20pm	**Update on pre-clinical studies to develop gene therapy for Barth syndrome** ~ *Christina A. Pacak, PhD — University of Florida College of Medicine, Gainesville, FL*
5:20pm—5:50pm	**Clinical, molecular, and metabolomic studies in Barth syndrome: Outcomes from the 2014 Barth Syndrome Foundation Scientific, Medical & Family Conference** ~ *Hilary J. Vernon, MD, PhD — Johns Hopkins University School of Medicine, Baltimore, MD*

6:00pm—8:00pm	**Poster Session** (6:00-8:00pm Science & Medicine Attendees / 7:00-8:00pm Families) *(Grand Ballroom Foyer)*
8:00pm—10:30pm	**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. (Group photo - all conference attendees)

FAMILY SESSIONS
Thursday, July 21, 2016

The Family sessions are designed to provide the latest information to family members 16 years of age and older. Note: Affected individuals aged 16+ may choose to attend any/all individual sessions within the Youth, Family, and Science & Medicine sessions, according to individual preferences. Note: Family and Science & Medicine presentations may occasionally include more "mature" content.

7:30am—8:15am: BREAKFAST *(Salon D, E, F)*

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

8:45am—11:00am: A NEW FRONTIER *(Salon A, B, C)*

8:45am—8:55am	Introduction — Shelia and Ben Mann
9:00am—9:30am	**The rewards of risk-taking** ~ *Elizabeth A Perkins, PhD, RNLD, FAAIDD, FGSA — Florida Center for Inclusive Communities; College of Behavioral and Community Sciences, University of South Florida, Tampa, FL*
9:35am—11:00am	**The new frontier: Future clinical trials and you** ~ *Kate McCurdy — Emerita, Scientific & Medical Advisory Board; Marc Sernel — Chairman, Board of Directors, Barth Syndrome Foundation*
11:00am—11:15am	Break

11:15am—11:50am: FEEDING ISSUES *(Salon A, B, C)*

11:15am—11:50am	**Skills and practical strategies to support feeding** ~ *Stacey Reynolds, PhD, OTR/L; Emily Burgess, MS, OTR/L — Virginia Commonwealth University, Richmond, VA*

12:00pm—1:30pm: KEYNOTE SPEAKER LUNCHEON *(Salon D, E, F)*

12:00pm—1:30pm	**Keynote Speaker Luncheon** **Ronald J. Bartek** — Co-Founder/Founding President, Friedreich's Ataxia Research Alliance (FARA) *"What we as families, advocates, scientists, and industry partners can do to move Barth syndrome more fully into the clinical treatment era." (Introduction by Shelley Bowen)*

2:00pm—5:50pm: POTENTIAL THERAPIES FOR BARTH SYNDROME *(Salon D, E F)*

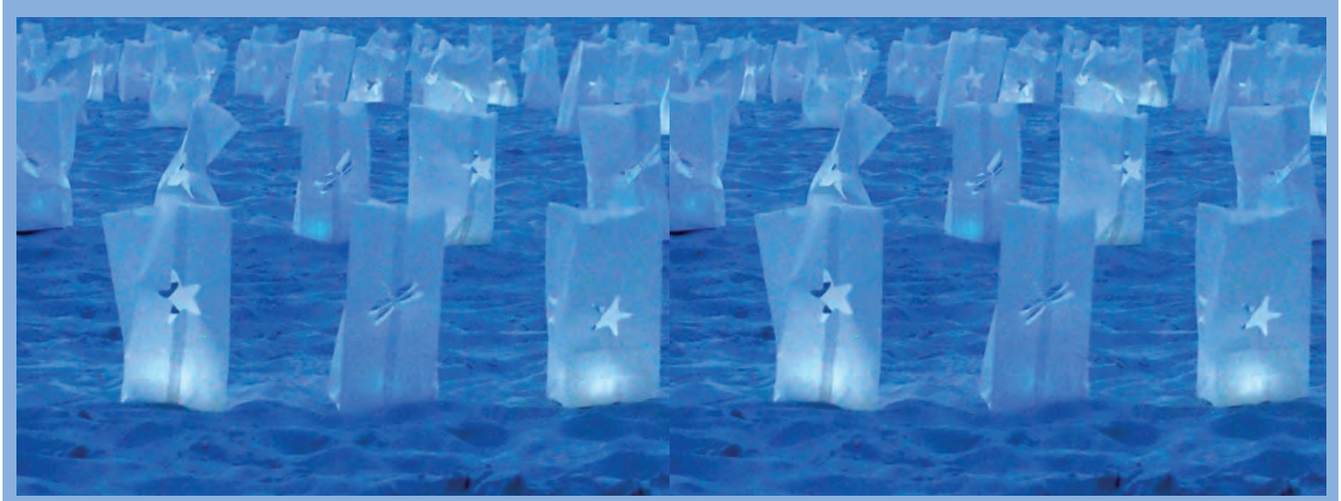
2:00pm—5:50pm	There are no scheduled "Family Sessions" at this time. Families are invited and encouraged to attend the Science & Medicine sessions on **Potential therapies for Barth syndrome** (see page 8)

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

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

8:00pm—10:30pm: LUMINARIES ON THE BEACH

8:00pm—10:30pm	**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. (Group photo - all conference attendees)*



AFFECTED INDIVIDUALS and SIBLINGS SESSIONS
(Water's Edge)
Thursday, July 21, 2016

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: BREAKFAST *(Salon D, E, F)*

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

8:30am—10:30am: BARTH BATTLE STATIONS *(Former Fitness Center, 3rd Floor)*

	Barth battle stations ~ *Lucy Buckley, Paediatric Physiotherapist — Bristol Royal Hospital for Children, Bristol, United Kingdom; Brittany DeCroes Hornby PT, DPT, PCS — Kennedy Krieger Institute, Baltimore, MD*
8:30am—9:30am	Audience: Affected individuals up to 8 years of age
9:30am—10:30am	Audience: Affected individuals 9+ years of age (older guys to cheer on!)
	Join in for a fun session: games include ninja, copycat, lightsaber duels, balance beam, and dodge ball mayhem!

10:45am—12:00pm: PHOTO SCAVENGER HUNT

10:45am—12:00pm	**Photo scavenger hunt** *(All youth combined - start in Former Fitness Center, 3rd Floor)* *Lead: Kevin Boozer, Volunteer*

12:00pm—1:30pm: KEYNOTE SPEAKER LUNCHEON *(Salon D, E, F)*

12:00pm—1:30pm	**Keynote Speaker Luncheon** **Ronald J. Bartek** — Co-Founder/Founding President, Friedreich's Ataxia Research Alliance (FARA) *"What we as families, advocates, scientists, and industry partners can do to move Barth syndrome more fully into the clinical treatment era." (Introduction by Shelley Bowen)*

2:30pm—4:00pm: TRANSITIONS

	Transition Session ~ *Rebecca Boudos, LCSW — Social Worker, Ann & Robert H. Lurie Children's Hospital, Chicago, IL*
2:30pm—3:00pm	Audience: Combined youth 8-15 years of age
3:30pm—4:00pm	Audience: Combined youth 16+ years of age
	Hands on workshop to prepare young people for transition into adult medical care, post school education, and future relationships.

2:30pm—4:00pm: GENETICS

	Understanding Genetics ~ *Rebecca L. McClellan, MGC, CGC — Kennedy Krieger Institute, Baltimore, MD*
2:30pm—3:00pm	Audience: Combined youth 15+ years of age
3:30pm—4:00pm	Audience: Combined youth 8-15 years of age
	A fun, interactive workshop teaching basic genetics.

4:00pm—5:50pm: YOUTH ACTIVITIES

4:00pm—5:50pm	**Youth activities** (Wii, ping pong, crafts, other activities, donor Thank you cards)

8:00pm—10:30pm: LUMINARIES ON THE BEACH

8:00pm—10:30pm	**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. (Group photo - all conference attendees)*

POSTER SESSION

THURSDAY, JULY 21, 2016

(Grand Ballroom Foyer)

For 2016, 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, July 22nd. 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: **Effects of resistance exercise training with/without protein supplementation in Barth syndrome** Adam J. Bittel, Program in Physical Therapy, Washington University School of Medicine, St. Louis, MO, USA	
POSTER 2: **AGPAT4 is a mitochondrial lysophosphatidic acid acyltransferase that regulates learning and memory in mice** Ryan M. Bradley, Department of Kinesiology, University of Waterloo, Waterloo, Ontario, Canada	
POSTER 3: **AGPAT4 ablation significantly decreases maximal force contractility in the soleus muscles of mice, but does not affect muscular endurance** Ryan M. Bradley, Department of Kinesiology, University of Waterloo, Waterloo, Ontario, Canada	
POSTER 4: **Vitamin and mineral supplementation in Barth syndrome: UK perspective** Nicol Clayton, NHS Barth Syndrome Service, Bristol Royal Children's Hospital, Bristol, United Kingdom	
POSTER 5: **Continuous glucose monitoring in Barth syndrome patients: An interim report.** Nicol Clayton, NHS Barth Syndrome Service, Bristol Royal Children's Hospital, Bristol, United Kingdom	
POSTER 6: **Monolysocardiolipins in membranes of animal tissues and microorganisms: Bad or good?** Angela Corcelli, Department of Basic Medical Sciences, Neurosciences and Sensory Organs, University of Bari A Moro, Bari, Italy	
POSTER 7: **A pilot study to evaluate the effectiveness of a 2-day therapy group for children with Barth syndrome in the UK** Danielle Goodman, NHS Barth Syndrome Service, Bristol Royal Children's Hospital, Bristol, United Kingdom	
POSTER 8: **The UK approach to educating peers about Barth syndrome** Danielle Goodman, NHS Barth Syndrome Service, Bristol Royal Children's Hospital, Bristol, United Kingdom	
POSTER 9: **Cardiolipin nanodisks: Cardiolipin replacement therapy for *TAZ* deficiency induced phenotypic changes** Nikita Ikon, Children's Hospital Oakland Research Institute, Oakland, CA, USA	
POSTER 10: **AAV mediated gene therapy improves soleus function in a post-hypoxic mouse model of Barth syndrome** Michael Jones, Department of Pediatrics, University of Florida College of Medicine, Gainesville, FL, USA	
POSTER 11: **Mathematical modelling of LC-MS/MS data reveals detailed insights into the structural diversity of the cardiolipidome** Markus A. Keller, Division of Biological Chemistry and Division of Human Genetics, Medical University Innsbruck, Innsbruck, Austria	
POSTER 12: **Experience with long-term total parenteral nutrition in a Barth syndrome patient with severe gastroparesis** David Kronn, Department of Pediatrics, New York Medical College, Valhalla, NY, USA	
POSTER 13: **Perturbation of iron homeostasis in cardiolipin deficient cells due to decreased frataxin** Yiran Li, Department of Biological Sciences, Wayne State University, Detroit, MI, USA	
POSTER 14: **Raising a child with Barth syndrome: Impacts on the family** Yoonjeong Lim, Department of Occupational Therapy, University of Florida, Gainesville, FL, USA	
POSTER 15: **HRQoL in Barth syndrome: Agreement between child self-reports and parent proxy-reports and relationship to parental HRQoL** Yoonjeong Lim, Department of Occupational Therapy, University of Florida, Gainesville, FL, USA	

POSTER SESSION

POSTER 16:
TCA cycle defects in cardiolipin deficient cells
Wenjia Lou, Wayne State University, Detroit, MI, USA

POSTER 17:
Skeletal myopathy in Barth syndrome – Presence, progression and cause
Laura Markham, University of Bristol, Bristol, United Kingdom

POSTER 18:
The role of *tafazzin* and MLCL AT-1 in mitochondrial function
Edgard M. Mejia, Department of Pharmacology & Therapeutics, University of Manitoba; Children's Hospital Research Institute of Manitoba, DREAM, Winnipeg, Canada

POSTER 19:
Barth syndrome In Italy
Amelia Morrone, Pediatric Neurology Unit and Laboratories, Meyer Children's Hospital, Florence; Department of NEUROFARBA, University of Florence, Florence, Italy

POSTER 20:
Mitochondrial structural defects in patient-derived fibroblasts in Barth syndrome
Manashwi Ramanathan, Department of Pediatrics, University of Florida, Gainesville, FL, USA

POSTER 21:
Sensory and motor-based feeding issues in boys with Barth syndrome
Stacey Reynolds, Virginia Commonwealth University, Richmond, VA, USA

POSTER 22:
Investigation of the physiochemical properties of the phospholipid cardiolipin: Implications for OXPHOS regulation and Barth syndrome
Murugappan Sathappa, University of Connecticut, Storrs, CT, USA

POSTER 23:
Evidence for impaired fatty acid utilization and altered interactions between fatty acid oxidation enzymes and the respiratory supercomplex in Barth syndrome heart
Genevieve C. Sparagna, Division of Cardiology, University of Colorado, Aurora, CO, USA

POSTER 24:
Composition of high molecular weight enzyme complexes in cardiac mitochondria from Barth syndrome patients and *Taz* shRNA mice
Kalyn S. Specht, Department of Biomedical Sciences and the Proteomics and Metabolomics Facility, Colorado State University, Fort Collins, CO, USA

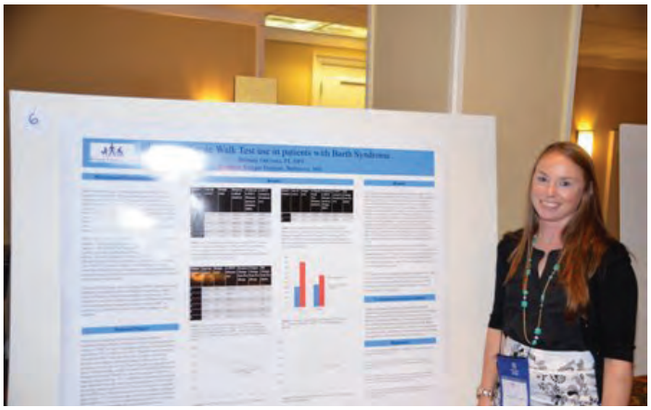
POSTER 25:
Identification of potential therapeutic pathways against the Barth syndrome using yeast as a model
Déborah Tribouillard-Tanvier, Institut de Biochimie et Génétique Cellulaires, Université Bordeaux, Bordeaux cedex, France

POSTER 26:
Reversal of severe mitochondrial cardiomyopathy by medicine therapy in a mouse model of Barth syndrome
Gang Wang, Department of Cardiology, Boston Children's Hospital, Boston, MA, USA

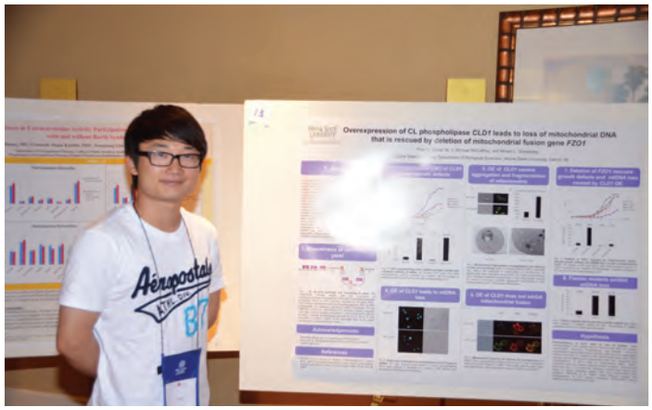
POSTER 27:
Adaptive behavior against oxidative stress in Barth syndrome lymphoblast cells
Yang Xu, Departments of Anesthesiology and Cell Biology; Department of Biochemistry and Molecular Pharmacology, New York University School of Medicine, New York, NY, USA

POSTER 28:
ALCAT1 deletion mitigates cardiac dysfunction in Barth syndrome through effect on mitophagy
Jun Zhang, Sam and Ann Barshop Institute for Longevity and Aging Studies, University of Texas Health Science Center at San Antonio, San Antonio, TX, USA

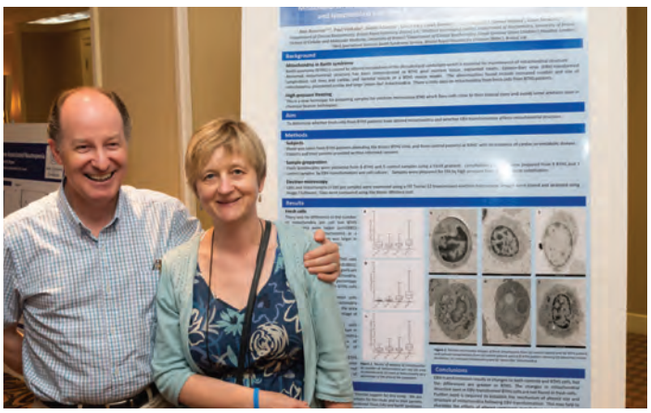
POSTER 29:
MMPOWER study: The effect of treatment with elamipretide in patients with mitochondrial disease
Amel Karaa, Massachusetts General Hospital, Boston, MA; Richard Haas, University California, San Diego School of Medicine, La Jolla, CA; Bruce H. Cohen, Akron Children's Hospital, Akron, OH; Amy Goldstein, Children's Hospital of Pittsburgh, Pittsburgh, PA; Jerry Vockley, Children's Hospital of Pittsburgh, Pittsburgh, PA, USA



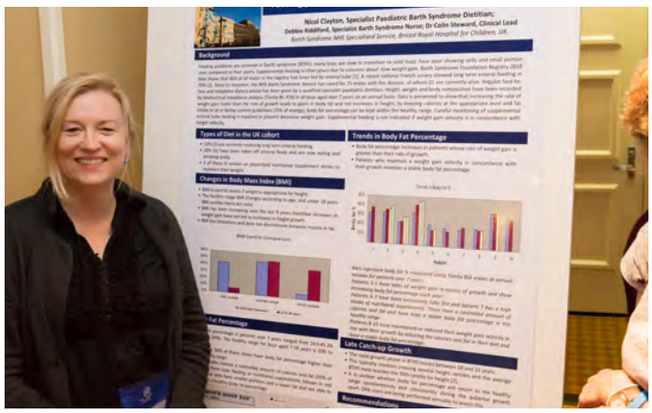
Brittany DeCroes Hornby at BSF's 2014 Poster Session



Yiran Li at BSF's 2014 Poster Session



Prof. Colin Steward & Ann Bowron at BSF's 2014 Poster Session



Nicol Clayton at BSF's 2014 Poster Session

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Thursday Breakfast

&

Poster Session

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SCIENCE & MEDICINE SESSIONS
Friday, July 22, 2016

7:30am—8:15am: BREAKFAST *(Salon D, E, F)*

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

8:30am—11:20am: CLINICAL CHARACTERISTICS OF BARTH SYNDROME *(Salon G)*
Chair: Jeffery Towbin, MD — St. Jude Children's Research Hospital, Memphis, TN

8:30am—8:35am	Introduction — Kevin Baffa
8:35am—9:05am	**Incidence, characteristics and management of neutropenia in Barth syndrome** ~ *Colin G. Steward, PhD, FRCP, FRCPCH — Bristol Royal Children's Hospital, Bristol, United Kingdom*
9:05am—9:35am	**How do women adapt to being a Barth syndrome carrier? A mixed methodological study of psychological adjustment and reproductive options** ~ *Cynthia James, ScM, PhD — Johns Hopkins University, Baltimore, MD*
9:35am—10:05am	**Functional exercise capacity, lower extremity strength and self-reported physical activity: Outcomes from the 2014 Barth Syndrome Foundation Scientific, Medical & Family Conference** ~ *Brittany DeCroes Hornby, PT, DPT, PCS — Kennedy Krieger Institute, Baltimore, MD*
10:05am—10:20am	Coffee Break
10:20am—10:50am	**Cardiovascular disease in Barth syndrome: Diagnostic and treatment strategies** ~ *John Lynn Jefferies, MD, MPH, FAAP, FACC, FAHA — Cincinnati Children's Hospital Medical Center, Cincinnati, OH*
10:50am—11:20am	**Two Poster Presenters** (15 minutes each)

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

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

2:00pm—4:50pm: CARDIOLIPIN AND BARTH SYNDROME *(Salon G)*
Chair: Ronald J. Wanders, PhD — Amsterdam Medical Center, Amsterdam, The Netherlands

2:00pm—2:05pm	Introduction — Darryl Byrd
2:05pm—2:35pm	**A new enzyme and pathway in cardiolipin synthesis** ~ *Robin Duncan, PhD — University of Waterloo, Waterloo, Ontario, Canada*
2:35pm—3:05pm	**Two Poster Presenters** (15 minutes each)
3:05pm—3:35pm	**Multitasking by cardiolipins – intra- and extra-mitochondrial events** ~ *Valerian E. Kagan, PhD, DSc — University of Pittsburgh, Pittsburgh, PA*
3:35pm—3:50pm	Coffee Break
3:50pm—4:20pm	**Metabolic consequences of defective iron homeostasis in cardiolipin deficient cells** ~ *Miriam L. Greenberg, PhD — Wayne State University, Detroit, MI*
4:20pm—4:50pm	**Investigating the effects of dysfunctional cardiolipin remodeling using in organello and model membrane systems** ~ *Nathan N. Alder, PhD — University of Connecticut, Storrs, CT*

4:50pm—5:20pm: CONFERENCE WRAP-UP *(Salon G)*

4:50pm—5:20pm	**Conference Wrap-Up**

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

7:00pm—11:00pm	**Friday Night Social ~** *Join us for a #TeamBarth-themed evening: socializing, music, dancing, appetizers, and cash bar. All conference attendees are invited!*

SCIENCE & MEDICINE SESSIONS
Saturday, July 23, 2016

8:30am—11:30am: SCIENTIFIC AND MEDICAL ADVISORY BOARD BREAKFAST & MEETING *(Salon G)*

8:30am—11:30am	**Scientific and Medical Advisory Board Breakfast & Meeting** *(by invitation)*

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

12:15pm—2:00pm	**Luncheon & Finale** *(All conference attendees welcome)*

FAMILY SESSIONS
Friday, July 22, 2016

7:30am—8:15am: BREAKFAST *(Salon D, E, F)*

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

8:20am—10:00am: CARDIAC ASPECTS OF BARTH SYNDROME *(Salon A, B, C)*

8:20am—8:30am	Introduction — Nicole Derusha-Mackey
8:30am—8:50am	**Evaluation of cardiac function in the current era** ~ *Carolyn Taylor, MD — Medical University of South Carolina, Charleston, SC*
8:50am—9:10am	**Medications used to treat heart failure** ~ *Barry J. Byrne, MD, PhD — Powell Gene Therapy Center, University of Florida, Gainesville, FL*
9:10am—9:30am	**Advanced therapies for heart failure in Barth syndrome cardiomyopathy** ~ *Brian Feingold, MD, MS — University of Pittsburgh, Pittsburgh, PA*
9:30am—9:50am	**Mitochondria and Barth syndrome** ~ *Colin K.L. Phoon, MD, MPhil — New York University Langone Medical Center, New York, NY*
9:50am—10:00am	**Q & A**
10:00am—10:10am	Break

10:10am—11:55am: CLINICAL RESEARCH AND BARTH SYNDROME *(Salon A, B, C)*

10:10am—10:45am	**Updates and results from the 2014 BSF/Johns Hopkins Study** ~ *Hilary J. Vernon, MD, PhD — Johns Hopkins University, and at the Kennedy Krieger Institute, Baltimore, MD*
10:45am-10:55am	**Vitamin and mineral supplementation in Barth syndrome** ~ *Nicol Clayton, Advanced Specialist Paediatric Dietician — Bristol Royal Hospital for Children, Bristol, United Kingdom*
11:00am—11:45am	**Participation in clinical research: Lessons learned in St. Louis** ~ *W. Todd Cade, PT, PhD; Dominic Reeds, MD — Washington University School of Medicine, St. Louis, MO*
11:45am—11:55am	**Q & A**

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

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

2:00pm—4:30pm: GENETICS AND CARRIER ISSUES OF BARTH SYNDROME *(Salon A, B, C)*

2:00pm—2:10pm	Introduction — Brie Chandler-Kalapasev
2:10pm—2:30pm	**From DNA to *TAZ* variants ...is this a mutation or not?** ~ *Iris L. Gonzalez, PhD — A. I. duPont Hospital for Children, Wilmington, DE*
2:30pm—3:15pm	**Tips and strategies for talking to your children about genetics** ~ *Rebecca L. McClellan, MGC, CGC — Kennedy Krieger Institute, Baltimore, MD; Cynthia James, ScM, PhD — Johns Hopkins University, Baltimore, MD; Lee Kugelmann, Co-Lead, Carrier Program, Barth Syndrome Foundation*
3:15pm—3:45pm	**Study update: How do women adapt to being a Barth syndrome carrier?** ~ *Rebecca L. McClellan, MGC, CGC — Kennedy Krieger Institute, Baltimore, MD; Cynthia James, ScM, PhD — Johns Hopkins University, Baltimore, MD*
3:45pm—4:30pm	**What we have learned, where to go from here, and how you can get involved?** ~ *Lee Kugelmann, Co-Lead, Carrier Program, Barth Syndrome Foundation (Audience: All carrier community)*

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

7:00pm—11:00pm	**Friday Night Social ~** *Join us for a #TeamBarth-themed evening: socializing, music, dancing, appetizers, and cash bar. All conference attendees are invited!*

AFFECTED INDIVIDUALS and SIBLINGS SESSIONS
Water's Edge
Friday, July 22, 2016

7:30am—8:15am: BREAKFAST *(Salon D, E, F)*

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

8:20am—8:50am: YOUTH GROUP PHOTO

8:20am—8:50am	**Youth Group Photo**

8:50am—10:45am: SANDCASTLE BUILDING

8:50am—10:45am	**Sandcastle building**

11:15am—12:00pm: Q & A SESSION

11:15am—12:00pm	**Q & A** ~ *B.J. Develle, MSW — Personal Health Coach Coordinator, Humana Cares, St. Petersburg, FL* Audience: younger and older affected individuals

11:15am—12:00pm: CARRIER ISSUES - SESSION 3

11:15am—12:00pm	**Carrier Issues: Session 3** ~ *Rebecca McClellan, MGC, CGC — Kennedy Krieger Institute, Baltimore, MD* Audience: Girls 10-14 years of age This small group 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.

11:15am—12:00pm: SIBLING SESSION

11:15am—12:00pm	**Sib Connect — Learning from each other** ~ *Rebecca Boudos, LCSW — Social Worker, Ann & Robert H. Lurie Children's Hospital, Chicago, IL* Audience: Brothers and sisters 8+ years of age Siblings will be able to share experiences with what it is like to have a sibling with Barth syndrome. We will do interactive activities during the session to explore common feelings, the positive and challenging parts.

12:00pm—2:00pm: LUNCHEON *(Salon D, E, F and Sandpiper Deck)*

12:00pm—2:00pm	**Varner Award Luncheon** *(Presented by John Wilkins) (Salon D, E, F)*
12:00pm—2:00pm	**Youth Luncheon** *(Combined Youth) w/ special guests "The Rough Riders" (Sandpiper Deck)*

2:15pm—4:30pm: YOUTH ACTIVITIES

2:15pm—4:30pm	**Keepsakes, crafts, video games, etc.**

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

7:00pm—11:00pm	**Friday Night Social** ~ *Join us for a #TeamBarth-themed evening: socializing, music, dancing, appetizers, and cash bar. All conference attendees are invited!*

FAMILY SESSIONS
Saturday, July 23, 2016

9:00am—9:30am: COFFEE STATION *(outside of Salon A, B, C)*

9:00am —9:30am	**Coffee station** *(Breakfast on your own)*

9:30am—12:10pm: WRAP-UP SESSIONS *(Salon A, B, C)*

9:30am—10:15am	**Barth battle stations! Video highlights of Physio sessions and top tips to take home** ~ *Lucy Buckley, Paediatric Physiotherapist — Bristol Royal Hospital for Children, Bristol, United Kingdom*
10:15am—11:30am	**Transition planning: Maximizing independence in health care, vocation planning and life skills** ~ *Rebecca Boudos, LCSW — Ann & Robert H. Lurie Children's Hospital of Chicago, Chicago, IL*
11:30am—12:10pm	**Pill swallowing school: Lessons learned** ~ *Stacey Reynolds, PhD, OTR/L; Emily Burgess, MS, OTR/L — Virginia Commonwealth University, Richmond, VA*

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

12:15pm—2:00pm	**Luncheon & Finale** *(including Registry & Repository recap, clinical trials debrief, etc.)*

AFFECTED INDIVIDUALS and SIBLINGS SESSIONS
(Water's Edge)
Saturday, July 23, 2016

9:30am—12:10am: CONFERENCE WRAP-UP *(Combined Youth)*

9:30am—12:10am	**Conference wrap-up , project finals, kid activities**

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

12:15pm—2:00pm	**Luncheon & Finale**

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William T. Pu, MD, Pediatric Cardiology, Boston Children's Hospital; Pediatrics, Harvard Medical School, Boston, MA

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

Colin G. Steward, PhD, FRCP, FRCPCH, Pediatric Hematology, Bristol Royal Hospital for Children; Clinical Lead, NHS Barth Syndrome Service; Bristol, United Kingdom

Arnold W. Strauss, MD, Pediatrics and Research, Cincinnati Children's Research Foundation; Cincinnati, OH

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

Jeffrey A. Towbin, MD, FAAP, FACC, FAHA, Pediatric Cardiology, University of Tennessee Health Science Center; Pediatric Cardiology, St. Jude Children's Research Hospital; Co-Director, The Heart Institute, Le Bonheur Children's Hospital, Memphis, TN

Hilary Vernon, MD, PhD, McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University and the Kennedy Krieger Institute; Director, Barth Syndrome Clinic at Kennedy Krieger Institute; Baltimore, MD

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

Katherine R. McCurdy, *Emerita*

Catharine L. Ritter, RN, *ex-officio*
Board Director, Barth Syndrome Foundation; Board Director, Barth Syndrome Foundation of Canada

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

Participant Biographies

Nathan N. Alder, PhD — Associate Professor, Department of Molecular and Cell Biology, University of Connecticut, Storrs, CT, USA

Dr. Alder directs a research program that is focused on understanding the structure, function, and biogenesis of mitochondrial membrane proteins and complexes, including those of the protein transport machinery and the oxidative phosphorylation system. Of particular interest to him is understanding how heritable diseases involving lipid and protein biogenesis impact structure-function relationships within mitochondrial complexes. Research in the Alder Laboratory uses a host of biochemical and biophysical techniques as well as the use and development of model membrane systems. Dr. Alder was awarded a research grant from BSF entitled "Investigation of cardiolipin-dependent respiratory complex activity and development of small molecule lipid analogs" (2013).

Since 2008, research in the Alder Lab has been funded by the NIH, the NSF, the American Heart Association, and the Barth Syndrome Foundation. Dr. Alder holds a PhD (2002) from the University of California, and a BS (1994) from the University of Utah, and he did his post-doctoral work (2002-2008) at the Texas A&M University College of Medicine. Dr. Alder is an active member of the Biophysical Society and the American Society for Cell Biology.

Presentation: Investigating the effects of dysfunctional cardiolipin remodeling using in organello and model membrane systems *(Sci/Med Sessions)*

Ronald J. Bartek, MSc — Co-founder and Founding President, Friedreich's Ataxia Research Alliance; Chairman of the Board of the National Organization for Rare Disorders (NORD); 4-year member of the National Institutes of Health (NIH) National Advisory Neurological Disorders and Stroke Council, Washington, DC, USA

Mr. Bartek is Friedreich's Ataxia Research Alliance's (FARA) Co-founder and President. He also is Chairman of the Board of the National Organization for Rare Disorders (NORD); a 4-year member, National Institutes of Health (NIH) National Advisory Neurological Disorders and Stroke (NINDS) Council; and former partner and president of a business and technology development, consulting and government affairs firm.

Friedreich's Ataxia is a genetic disorder with some important clinical similarities to Barth syndrome. FARA is an organization that communicates (in collaboration with other organizations) with government agencies at both state and federal levels in pursuit of policies and decisions intended to advance therapeutic development for the disorder. Mr. Bartek is well-respected and trusted by peer patient advocacy leaders, policy makers in Washington, DC, leaders of numerous divisions of the NIH and by people he serves through FARA. He is known by all as a man who is thoughtful, who cares deeply and who gets results. Most importantly, he is a father who got involved to help his son. Though Keith sadly died in January 2010, Mr. Bartek has continued to work for treatments for all those who have Friedreich's Ataxia or who suffer from a rare disease. Mr. Bartek has been a friend and advisor to the Barth Syndrome Foundation (BSF) from the very beginning. He and FARA have traveled down a very similar road to BSF to translate scientific advances into actual therapies. In his address, Mr. Bartek will offer us all some first-hand insights into this process and will emphasize patients' critical roles.

From all of Mr. Bartek's work in the rare disease field, he has a great deal of experience and expertise in topics very relevant to all of us involved in BSF. Mr. Bartek and his organization are considered by many to be pioneers in the rare disease arena, and they have won many awards. Among them are recognition from the Pharmaceutical Research and Manufacturers of America and a Lifetime Achievement Award from Rare Disease Legislative Advocates. Most recently, on May 17, 2016, the National Organization for Rare Disorders (NORD)® announced that Friedreich's Ataxia Research Alliance, a passionate patient organization working to make a difference for everyone affected by rare diseases, will be one of the esteemed Rare Impact Award recipients this year. We are excited for our attendees to hear Mr. Bartek's inspiring message of hope and triumph, as told by someone who has many years of first-hand experience.

Presentation: What we as families, advocates, scientists and industry partners can do to move Barth syndrome more fully into the clinical treatment era

Rebecca Boudos, LCSW — Social Worker, Ann & Robert H. Lurie Children's Hospital, Chicago, IL, USA

Ms. Boudos has been a social worker at Ann & Robert H. Lurie Children's Hospital in Chicago for more than eight years. She works specifically with the Spina Bifida Center and the chronic illness transition team to provide both program planning and clinical services regarding youth in transition in the healthcare setting. Ms. Boudos supports the Lurie Children's Hospital wide transition steering committee, parent/teen program, paid work experience program, and direct clinical work focused on transition to adult healthcare and transition to adult services. She has presented at numerous conferences on health care transitions, including the Illinois Statewide Transition Conference.

Presentations: Transition planning: Maximizing independence in health care, vocation planning and life skills *(Family Sessions);* Transition Sessions *(Youth Sessions)*

Valerie ("Shelley") Bowen — Director, Family Services & Awareness, Barth Syndrome Foundation, USA

Mrs. Bowen is a founder of the Barth Syndrome Foundation (BSF) and currently serves as Director of Family Services and Physician Awareness. Both of her sons lost their lives to Barth syndrome. She believes it possible for everyone who has Barth syndrome to have an accurate diagnosis and every parent has the capacity to be transformed from a powerless bystander into an empowered advocate when given the proper tools. Mrs. Bowen has worked tirelessly since then to insure that not one more child will suffer or perish from the disorder.

Consultation: Barth Syndrome Registry & Repository *(Family Sessions)*

Lucy Buckley — Paediatric Physiotherapist, Bristol Royal Hospital for Children, Bristol, United Kingdom

Ms. Buckley is a specialist physiotherapist and currently working at Bristol Royal Hospital for Children. She has been the physiotherapist for the NHS National Barth Syndrome Service for the past two years. Ms. Buckley also works as the physiotherapist for the Oncology and Haematology Department. She has developed a special interest in keeping children active and trying to maximize each child's physical potential. Ms. Buckley is part of the regional oncology network and has run and presented at regional study days. She has recently had an article titled "Keeping Active" published in the CCLG (Children's Cancer and Leukemia Group) magazine.

Presentation: Barth battle stations! Video highlights of physio sessions and top tips to take home *(Family Sessions);* Barth battle stations *(Youth Sessions)*

Barry Byrne, MD, PhD — Professor and Associate Chair, Pediatrics and Molecular Genetics and Microbiology; Director, Powell Gene Therapy Center, 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 Pre-Clinical and Clinical Research Teams have made significant contributions to the understanding and treatment of several rare diseases. 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.

Presentation: Medications used to treat heart failure *(Family Sessions)*

Emily Burgess, MS, OTR/L — Research Assistant, Occupational Therapy Program, Virginia Commonwealth University, Richmond, VA, USA

Ms. Burgess currently works as a pediatric Occupational Therapist providing early intervention and private practice occupational therapy services. Ms. Burgess came to know the Barth syndrome community working as a research assistant for her former professor, Dr. Stacey Reynolds. Ms. Burgess will be participating in this year's conference as a clinician alongside Dr. Reynolds, facilitating the pill swallowing clinic, presenting a poster, and completing a family presentation on prior research on feeding and eating habits and taste preferences completed at the 2014 conference. She hopes to maintain her connection to the community both as a clinician and researcher in the years to come.

Presentation: Skills and practical strategies to support feeding; Pill swallowing school: Lessons learned *(Family Sessions)*

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 skeletal muscle and cardiac nutrient metabolism abnormalities in metabolic diseases such as Barth syndrome, HIV-associated metabolic syndrome, diabetes, and pathologic pregnancy. Dr. Cade was awarded the following research grants from BSF entitled "Effects of resistance exercise training on cardiac, metabolic, and muscle function and quality of life in Barth syndrome: Part II" (2014), "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 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, FL, 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.

Chair: Pathomechanisms(s) of Barth syndrome *(Sci/Med Sessions)*
Clinic: Heart and skeletal muscle metabolism in Barth syndrome; Presentations: Update on exercise and substrate metabolism studies in Barth syndrome *(Sci/Med Sessions)*; Participation in clinical research: Lessons learned in St. Louis *(Family Sessions)*

Adam J. Chicco, PhD, FAHA — Associate Professor, Department of Biomedical Sciences, Colorado State University, Fort Collins, CO, USA

Dr. Chicco's research focuses on the influence and interplay of polyunsaturated fatty acid metabolism and mitochondrial function in cardiometabolic health and disease. Current studies are combining proteomic and lipidomic profiling with functional studies of human and mouse heart tissues in collaboration with Drs. Genevieve Sparagna and Kathryn Chatfield at the University of Colorado to elucidate the mechanisms responsible for abnormal cardiac lipid metabolism in Barth syndrome. Dr. Chicco was awarded the following research grants from BSF entitled "Translating murine *Taz* deficiency to human Barth syndrome: Focus on impaired lipid oxidation" (*partial funding provided by Barth Syndrome Trust* - 2014), "Mechanisms of substrate-specific impairment of oxidative phosphorylation in *taz*-deficient cardiac mitochondria" (2012), and "Targeting cardiolipin deficiency in the *taz* shRNA mouse model of Barth syndrome" (2010).

Dr. Chicco received his PhD in Exercise/Cardiovascular Physiology from the University of Northern Colorado. He holds a MEd in Exercise Physiology from Temple University and a BA in Philosophy/Business Management from Marietta College. Dr. Chicco is a member of the American Heart Association, American Physiological Society, International Society of Heart Research and American Association for the Advancement of Science.

Presentation: Cardiac proteomics distinguish human Barth syndrome from pediatric heart failure and reveal patterns of metabolic remodeling common to *Taz* shRNA mice *(Sci/Med Sessions)*

Michael T. Chin, MD, PhD, FACC, FAHA — Associate Professor of Medicine, Harold T. Dodge-John L. Locke Chair in Cardiovascular Medicine, University of Washington, Seattle, WA, USA

Dr. Chin practices general adult cardiology at the University of Washington Medical Center. He directs a research laboratory focused on understanding the molecular biology of the cardiovascular system. His 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. Dr. Chin was awarded the following research grants from BSF entitled "Enzyme replacement therapy in heart failure associated with *tafazzin* deficiency" (2014), "*Tafazzin* enzyme replacement therapy in a mouse model of Barth syndrome" (2013), and "*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 an MD (with Honors), from the University of Rochester (1991). He completed his Residency in Internal Medicine at the Johns Hopkins Hospital, Baltimore, MD (1991-1993).

Dr. Chin is board certified in adult cardiology, a Fellow of the American College of Cardiology, a Fellow of the American Heart Association, an elected member of the American Society for Clinical Investigation and maintains membership in the American Society for Biochemistry and Molecular Biology, the American Physiological Society and the International Society for Heart Research.

Presentation: *Tafazzin* enzyme replacement therapy for Barth syndrome *(Sci/Med Sessions)*

Amanda Clark — Portrait Artist, Amanda Clark Photography, Perry, FL, USA

Ms. Clark has always had a passion for photography — a gift given to her by a true love who instilled in her a love for others. To repay this blessing, she gives you, and everyone she photographs, a moment in time, warmth and an emotion that she sees through the lens. These feelings touch her heart and are captured to be yours to cherish for a lifetime. She has attended our conferences almost since the beginning and has given us all incredible gifts by photographing our boys, young men and families.

Nicol Clayton — Advanced Specialist Paediatric Dietician, Bristol Royal Hospital for Children, Bristol, United Kingdom

Ms. Clayton has been a member of the multidisciplinary NHS National Barth Syndrome Service since its inception in April 2010. She manages the dietetic care of all Barth patients in the UK and provides expert advice for many children with Barth syndrome across the world. Ms. Clayton also specializes in the provision of ketogenic diet therapy, a metabolic treatment for children with intractable epilepsy.

Ms. Clayton recently authored a chapter on feeding in Barth syndrome in the 4th edition of Clinical Paediatric Dietetics (Wiley Blackwell) and has contributed to a number of research publications produced by the Bristol team.

Ms. Clayton's previous roles included working within a multi-disciplinary feeding disorders clinic to manage difficult behaviors around food, including food restriction and food refusal. During this time, she delivered study days to train health and medical professionals in 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 growth disorders.

Workshops: Ms. Clayton will be running a series of interactive workshops for families on nutrition, growth, and managing feeding problems.

Presentation: Vitamin and mineral supplementation in Barth syndrome *(Family Sessions)*

Laura Cole, PhD — Postdoctoral Fellow, Manitoba Institute for Child Health and 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 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 degree in lipid biochemistry from the University of Alberta, Canada.

Presentation: Elevated hepatic fatty acid oxidation contributes to low-body weight in *tafazzin* knock-down mice *(Sci/Med Sessions)*

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

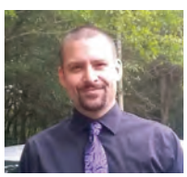
After studying the transport of sugars, amino acids and ions in membranes isolated from epithelial cells Dr. Corcelli moved to the field of membrane biochemistry of extremely halophilic microorganisms. Dr. Corcelli has investigated novel biochemical aspects of the archaeal proton pump bacteriorhodopsin with particular interest in the lipid-protein interactions.

Dr. Corcelli discovered and elucidated, in collaboration with Dr. Morris Kates, the structures of the archaeal analogs of cardiolipins and the halocapnines of halophilic bacteria. Her studies on cardiolipins have shown that the levels of archaeal cardiolipins in membranes fluctuate in response to osmotic stress and that the same phenomenon occurs in bacteria, such as Rhodobacter sphaeroides. Being interested in 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" (*partial funding provided by Association Barth France* — 2012).

Clinic: Olfactory identification in patients affected by Barth syndrome *(Family Sessions)*

Michaela Damin — Founder and Chair, Barth Syndrome Trust, United Kingdom

Mrs. Damin's first son was born with Barth syndrome in 1998, and Mrs. Damin and her husband moved to England from South Africa in 1999. Michaela knew of only one other family with Barth syndrome in England and set out on a quest for both information and other Barth families. She soon met key doctors at Southampton, London and Bristol hospitals as well as several other families via the Internet. By year-end 2005, the Barth Syndrome Trust had found 30 families throughout Europe, had held clinics and family gatherings in Holland and the UK, had grown the list of knowledgeable and experienced doctors, and had assembled a group of dedicated volunteers. Mrs. Damin is highly respected in the Barth syndrome community for her leadership, her drive and her compassion.



B.J. Develle, MSW — Personal Health Coach Coordinator, Humana Cares, St. Petersburg, FL, USA

Mr. Develle currently works for Humana at home supervising a team of telephonic Case managers who serve a population who typically have multiple chronic health conditions. He also works as an independent contractor, writing homestudies for pre-adoptive families. 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, grief and loss, 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. Mr. Develle 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 Bachelor degrees in Child Development and Religion in 2002.

Mr. Develle will be involved in the Youth sessions on Thursday, Friday and Saturday.

Robin Duncan, PhD — Assistant Professor, Department of Kinesiology, Faculty of Applied Sciences, University of Waterloo, Waterloo, Ontario, Canada

Dr. Duncan's current research program is focused on the discovery and characterization of novel enzymes in glycerolipid metabolism, with studies that range from molecular cloning to pathophysiology. Her work is funded by The Natural Sciences and Engineering Research Council of Canada (NSERC), the Canadian Foundation for Innovation, the Canadian Diabetes Association, the Barth Syndrome Foundation and the Barth Syndrome Foundation of Canada. Dr. Duncan was awarded a research grant from BSF entitled "A new enzyme and pathway in cardiolipin synthesis" (*partial funding provided by Barth Syndrome Foundation of Canada* — 2015).

Dr. Duncan was awarded a PhD (Nutritional Sciences), from the University of Toronto and a BSc (Hons) (Biological Sciences) from the University of Guelph, and did a Post-doctoral Fellowship (Nutritional Science) at University of California, Berkeley, CA, USA. Dr. Duncan was the recipient of postdoctoral fellowships from NSERC, Canadian Institutes of Health Research (CIHR), and the Heart and Stroke Foundation of Canada.

Presentation: A new enzyme and pathway in cardiolipin synthesis *(Sci/Med Sessions)*

Brian Feingold, MD, MS — Associate Professor, Pediatrics and Clinical and Translational Science, University of Pittsburgh; Medical Director, Pediatric Heart Failure and Transplantation Programs, Children's Hospital of Pittsburgh of University of Pittsburgh Medical Center, Pittsburgh, PA, USA

Dr. Feingold's research is broadly focused on pediatric heart transplantation and heart failure with specific funding by the National Institutes of Health KL2 Clinical Research Scholars Program and the American Heart Association.

Dr. Feingold is a graduate of Duke University, Durham, North Carolina, and the University of Pittsburgh School of Medicine. Dr. Feingold completed his residency at Massachusetts Memorial Medical Center, Worcester, MA and Fellowship in Pediatric Cardiology at Children's Hospital of Pittsburgh of University of Pittsburgh Medical Center.

He currently serves on multiple councils and committees of national organizations including the pediatric heart failure subcommittee of the American Heart Association, Scientific Council on Pediatric Thoracic Transplantation and Heart Failure of the International Society for Heart and Lung Transplantation, and the Pediatric Heart Transplant Study Group. Dr. Feingold is board certified in Pediatric Cardiology.

Presentation: Advanced therapies for heart failure in Barth syndrome cardiomyopathy *(Family Sessions)*

Iris L. Gonzalez, PhD — Research Scientist, Molecular Diagnostics Laboratory, A. I. duPont Hospital for Children, Wilmington, DE; 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. Dr. Gonzalez was awarded a research grant from BSF entitled "A Study of *TAZ* mRNAs in Barth syndrome individuals" (2002). Dr. Gonzalez is a former recipient of the Barth Syndrome Foundation's Varner Award for Pioneers in Science and Medicine (2014).

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

Consultation: Genetics; Presentation: From DNA to *TAZ* variants...is this a mutation or not? *(Family Sessions)*

Danielle Goodman — Occupational Therapist, NHS Barth Syndrome Service, Bristol Royal Hospital for Children, Bristol, United Kingdom

Ms. Goodman has over a decade's experience working across all specialisms and sectors within Pediatrics (community, hospital and social services). Previously, she has held positions in Leicester, Birmingham Children's Hospital and Singapore where she worked within the private paediatric sector. She has been involved in setting up a number of new services and ensuring their successful development. Ms. Goodman trained at the University of East Anglia.

Alongside her role in the Barth Syndrome Service, Ms. Goodman also works as an independent therapist across the south west of England with children who have complex needs. She has a particular interest in promoting independence and she has lectured on the BSc (Hons) Occupational Therapy course at the University of the West of England.

Ms. Goodman will be running a number of workshops during the conference to support feeding and sensory issues, independence, and general advice on education.

Small Group Meeting: Promoting independence: Strategies to help children with special needs reach their true potential *(Family Sessions)*

Miriam L. Greenberg, PhD — Professor, Department of Biological Sciences, Wayne State University, Detroit, MI; Scientific & Medical Advisory Board, Barth Syndrome Foundation, USA

Dr. Greenberg's laboratory utilizes the powerful genetic, molecular, and cell biological tools of the yeast system and relevant mammalian cell cultures to elucidate the cellular functions of two essential lipid pathways. 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 seeks to elucidate the essential functions of inositol phospholipids and metabolites and the cellular consequences of inositol depleting drugs. These studies have implications for understanding the pathology underlying cardiovascular disorders and the therapeutic mechanisms of action of drugs used in the treatment of psychiatric disorders. Dr. Greenberg was awarded the following research grants from BSF entitled "Cardiolipin is required for mitochondrial protein processing" (2015), "Identification of human cardiolipin phospholipases that are deleterious to *tafazzin*-deficient cells" (2013); "Cardiolipin deficiency leads to defects in the TCA cycle" (*partial funding provided by Association Barth France* — 2011), "Loss of cardiolipin leads to defective mitochondrial iron/sulfur biosynthesis and iron homeostasis" (2010), "Perturbation of mitophagy in cardiolipin mutants" (*partial funding provided by Barth Syndrome Trust* — 2009), "The role of *tafazzin* in mitochondrial protein import: Implications for Barth syndrome" (*partial funding provided by Barth Syndrome Foundation of Canada* —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), and "*TAZ*1 gene function in yeast: A molecular model for Barth syndrome" (2002).

Dr. Greenberg holds a BA in Biology from Reed College, an MS in Microbiology from Loyola University, and a PhD in Genetics from Albert Einstein College of Medicine. She carried out her postdoctoral work in Molecular Biology at Harvard University.

Presentation: Metabolic consequences of defective iron homeostasis in cardiolipin deficient cells *(Sci/Med Sessions)*

Lindsay Groff, MBA — Executive Director, Barth Syndrome Foundation, USA

Ms. Groff was inspired to work for an organization dedicated to helping children that suffer from rare health conditions when her own daughter was diagnosed with a rare birth defect. Her experience with raising a medically fragile child, coupled with her education and experience, brought her to the Barth Syndrome Foundation (BSF). Ms. Groff has been the Executive Director of BSF since 2011. She directs the staff and resources of BSF in pursuit of its mission - saving lives through education, advances in treatment, and finding a cure for Barth syndrome.

In addition to setting the vision for and directing the operations of BSF, Ms. Groff is responsible for creating the structure and processes necessary to implement its long-term strategic objectives. She leads a robust philanthropic program that includes major gifts, direct response, foundation/corporate support, events, and grassroots fundraising activities. Her passion for nonprofit management, patient support, advocacy, fundraising, and marketing has fueled her career. Prior to her leadership at BSF, Ms. Groff was an Executive Director at Fernley & Fernley, an association management company in Philadelphia.

A proud native of New Jersey, Ms. Groff earned an MBA from Rowan University, as well as a bachelor's degree in marketing from Rutgers University.

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

Dr. DeCroes Hornby's professional interests include fitness across the lifespan, standardized outcome measures, mitochondrial disorders and teaching. She currently serves as a teaching assistant for the physical therapy programs at both the University of Maryland in Baltimore, MD and George Washington University in Washington, DC. Her current research interests include functional ability and quality of life in patients with Barth syndrome. She 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 at Kennedy Krieger Institute, Baltimore, MD.

In 2015, Dr. DeCroes Hornby completed a pediatric physical therapy residency with Kennedy Krieger Institute and the University of Maryland. Following completion of the residency, she earned a specialist certification in pediatric physical therapy. Dr. DeCroes Hornby holds a BS degree from James Madison University in Harrisonburg, VA where she majored in Biology and minored in Spanish. She received her PhD in Physical Therapy from George Washington University.

Presentation: Functional exercise capacity, lower extremity strength and self-reported physical activity: Outcomes from the 2014 Barth Syndrome Foundation Scientific, Medical & Family Conference (*Sci/Med Sessions);* Barth battle stations *(Youth Sessions)*

Cynthia James, ScM, PhD — Assistant Professor of Medicine, Center for Inherited Heart Disease, Division of Cardiology, Johns Hopkins University, Baltimore, MD, USA

Dr. James' research is focused on 1) psychosocial adaptation to inherited cardiac disease and 2) the role of genetic and environmental factors on disease expression, particularly in patients and families with arrhythmogenic right ventricular cardiomyopathy. Dr. James was awarded a research grant from BSF entitled "How do women adapt to being a Barth syndrome carrier? A mixed methodological study of psychological adjustment and reproductive options" (2014).

Dr. James holds a PhD in Human Genetics and Masters in Genetic Counselling from Johns Hopkins University and is a board-certified genetic counselor.

Presentations: How do women adapt to being a Barth syndrome carrier? A mixed methodological study of psychological adjustment and reproductive options *(Sci/Med Sessions);* Study Update: How do women adapt to being a Barth syndrome carrier?; Tips and strategies for talking to your children about genetics *(Family Sessions)*

John Lynn Jefferies, MD, MPH, FAAP, FACC, FAHA — Heart Failure Cardiologist, Cincinnati Children's Hospital Medical Center and the Christ Hospital; Professor, Pediatric Cardiology and Adult Cardiovascular Diseases, Department of Pediatrics, University of Cincinnati; Director, Advanced Heart Failure and Cardiomyopathy, Cincinnati Children's Hospital Medical Center, Cincinnati, OH, USA; Medical Advisor, The Battelle Healthcare Colloquium; Scientific and Medical Advisory Board, Barth Syndrome Foundation, USA

Dr. Jefferies specializes in advanced heart failure and cardiomyopathy care of both children and adults. He has been the Principal Investigator or Co-Investigator for multiple clinical studies funded by the National Institutes of Health (NIH) and industry. He is the author or co-author of over 130 peer reviewed manuscripts and he currently is the Editor of two textbooks. Dr. Jefferies was awarded a research grant from BSF entitled "Assessment of quality of life, anxiety, and depression in Barth syndrome: Expanding the scope of comprehensive care" (*partial funding provided by Barth Syndrome Trust* — 2014).

Dr. Jefferies completed a combined residency in Internal Medicine and Pediatrics, a fellowship in General Internal Medicine and a combined fellowship in Adult Cardiovascular Diseases and Pediatric Cardiology. He is certified as a Diplomat in Internal Medicine and Adult Cardiovascular Diseases as well as Advanced Heart Failure and Transplant Cardiology.

Presentation: Cardiovascular disease in Barth syndrome: Diagnostic and treatment strategies *(Sci/Med Sessions)*

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 was awarded a research grant from BSF entitled "Mechanism and role of cardiolipin oxidation and hydrolysis in Barth syndrome" (*partial funding provided by Barth Syndrome Foundation of Canada* — 2014).

Dr. Kagan holds a DSc in Biochemistry and Biophysics from USSR Academy of Science, a PhD in Biochemistry, an MSc in Biochemistry, and a BSc in Biochemistry, all from Moscow State University, Russia. Over the years, Dr. Kagan has also held various foreign professorships including King's College, London, UK; Institute of Environmental Medicine, Karolinska Institute, Stockholm, Sweden; Taipei Medical University, Taiwan; Russian State Medical University, Moscow, Russia; MV Lomonosov Moscow State University, Moscow, Russia; Fulbright Visiting Chair in Environmental Sciences, McMaster University, Hamilton, Canada; Sackler Lecturer, University of Tel Aviv, Israel. In 2015, Dr. Kagan was named as one of the World's most influential scientific minds by Reuters.

Presentation: Multitasking by cardiolipins – intra- and extra-mitochondrial events *(Sci/Med Sessions)*

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 phospholipids in aerobic metabolism of the 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, and in 2011, he was awarded a 4-year grant from National Institutes of Health for research using the Barth syndrome 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).

Presentation: Targeting PPAR/PGC1a axis in Barth syndrome: The PPAR pan-agonist bezafibrate ameliorates cardiomyopathy in a mouse model of Barth syndrome *(Sci/Med Sessions)*

Lee Kugelmann — Clinical Research Coordinator, Rare Disease Research Laboratory, Powell Gene Therapy Center, University of Florida, Gainesville, FL; Co-Lead, Carrier Program, Barth Syndrome Foundation, USA

Ms. Kugelmann is a research coordinator for the Powell Center for Rare Disease Research & Therapy at the University of Florida. Since 2014, she has worked on clinical and observational trials for rare inherited conditions including X-linked myotubular myopathy, Pompe disease, Duchenne muscular dystrophy, and Barth syndrome. She graduated from Emory University in 2014 with a bachelor's degree in Psychology and Linguistics. Ms. Kugelmann has been a member of the Barth Syndrome Foundation since its first family meeting in 2000, and has worked with BSF since on fundraisers, conference committees, and awareness campaigns. She is happy to be back this year continuing her work with the Carrier Services program.

Ms. Kugelmann will be involved in numerous carrier-related sessions.

Yoonjeong Lim, PhD, OTR/L — Research Associate, College of Public Health and Health Professions, Department of Occupational Therapy, Powell Center for Rare Disease Research, University of Florida, Gainesville, FL, USA

Dr. Lim's research interests are in quality of life of children and youth with rare diseases and their impact on family functioning and family quality of life. She received her PhD in the Rehabilitation Science program at the University of Florida in August 2015. Dr. Kim's dissertation examined how a child's functional ability, family cohesion, and satisfaction with healthcare affect quality of life and family functioning for parents of children with rare diseases.

Dr. Lim holds a MSc and BSc in Occupational Therapy from Yonsei University, in Wonju, South Korea. She is a licensed occupational therapist in Florida and a member of the American Occupational Therapy Association.

Clinic: The impact of a child's disability on the parents of children with rare diseases *(Family Sessions)*

Rebecca McClellan, MGC, CGC — Genetic Counselor, Center for Inherited Heart Disease, Johns Hopkins Medicine; Division of Metabolism, Department of Neurogenetics, Kennedy Krieger Institute, Baltimore, MD, USA

Ms. McClellan's research interests include further unmasking the phenotype of Barth syndrome, investigation of the challenges faced by carriers of X-linked disease, and the ethical impact and outcomes of the ACMG exome sequencing secondary findings panel.

Ms. McClellan has worked since 2002 in the Metabolism Clinic at the Kennedy Krieger Institute, which specializes in supporting patients with mitochondrial and other rare metabolic conditions. In 2013-2014, Ms. McClellan and colleagues established the only interdisciplinary clinic for Barth syndrome in the United States. She has worked closely over the years with the Barth Syndrome Foundation, speaking at their biennial conferences, providing telephone advice and support, and helping create their carrier support initiative. In 2014, Ms. McClellan and Dr. Cynthia James were awarded a research grant to investigate adjustment to carrier status and decision-making regarding reproductive options among Barth syndrome carriers. Since 2014, Ms. McClellan has split her time between Kennedy Krieger and the Johns Hopkins Center for Inherited Heart Disease, where she is working with families affected by arrhythmia conditions or cardiomyopathy.

Ms. McClellan holds a MGC, Genetic Counseling from University of Maryland School of Medicine and a BS in Psychobiology from Albright College, Reading, PA. She is a board-certified genetic counselor.

Consultation: Genetics; Small Group Meetings*:* Life as a carrier; Pre-test carrier issues; Post-test carrier issues; Presentations: Study Update: How do women adapt to being a Barth syndrome carrier?; Tips and strategies for talking to your children about genetics; What we have learned, where to go from here, and how you can get involved? *(Family Sessions);* Understanding genetics *(Youth Sessions)*

Katherine McCurdy — *Emerita*, Scientific & Medical Advisory Board, Barth Syndrome Foundation, USA

Mrs. McCurdy is a founding member of The Barth Syndrome Foundation's (BSF) Board of Directors and is responsible for establishing the Science and Medicine Program of BSF. She currently serves as *Emerita* member of the Scientific & Medical Advisory Board. Mrs. McCurdy has a BA from Duke University and an MBA from the Harvard Business School. She has held various positions on the Boards of several other nonprofit organizations. As a Barth syndrome mother, she is strongly committed to the mission of BSF.

Presentation: The new frontier: Future clinical trials and you (*Family Sessions)*

Christina A. Pacak, PhD — Assistant Professor, Department of Pediatrics, University of Florida, Gainesville, FL, USA

Dr. Pacak's primary focus is to develop adeno-associated virus (AAV) mediated gene therapy and mitochondrial transplantation therapy to treat disorders that affect mitochondrial function, particularly Barth syndrome and Cockayne syndrome. Dr. Pacak was awarded a research grant from BSF entitled "Correction of mitochondrial dysfunction in Barth syndrome" (*partial funding provided by Association Barth France* — 2014).

Dr. Pacak holds a PhD (2006) from University of Florida and a BS from Ohio State University. She completed a postdoctoral research fellowship at Boston Children's Hospital and Harvard Medical School.

Presentation: Update on pre-clinical studies to develop gene therapy for Barth syndrome *(Sci/Med Sessions)*

Elizabeth Perkins, PhD, RNLD, FAAIDD, FGSA — Associate Director and Research Assistant Professor, Florida Center for Inclusive Communities, Department of Child and Family Studies, College of Behavioral and Community Sciences, University of South Florida, Tampa, FL, USA

Dr. Perkins' current work includes educating future health professionals and development of advocacy documents such as the popular My Health Passport. She has published in prestigious journals including JAMA and Academic Medicine, as well as co-authored a gerontology (aging) textbook. Dr. Perkins also co-edited a special issue on Aging and End-of-Life for the American Journal on Intellectual and Developmental Disabilities.

Dr. Perkins has a PhD in Aging Studies and is also a Registered Nurse (United Kingdom) in Learning Disabilities. She was one of the first nurses to complete nurse training with her disability (she was born without her right arm). Dr. Perkins is a faculty member in University of South Florida's Master's program in Child and Adolescent Behavioral Health and she also teaches self-advocacy, empowerment, and disability awareness to various audiences in the community. Dr. Perkins is a Fellow of the American Association on Intellectual and Developmental Disabilities and the Gerontological Society of America.

Presentation: The rewards of risk-taking *(Family Sessions)*

Colin K. L. Phoon, MPhil, MD — Associate Professor, Pediatrics, Hassenfeld Children's Hospital, New York University Langone Medical Center and School of Medicine, Division of Pediatric Cardiology, Department of Pediatrics, New York, NY, USA

Dr. Phoon is investigating the role of mitochondria and cardiolipin in the pathogenesis of cardiomyopathy. As a principal investigator or co-investigator on several projects relevant to a broad spectrum of cardiovascular disease in small animal models, he is especially interested in heart development and heart imaging. Dr. Phoon was awarded the following research grants from BSF entitled "Novel antioxidant therapies in a mouse model of Barth syndrome" (2014); "Role of mitochondria during myocardial morphogenesis in Barth syndrome" (2012), and "Cardiomyopathy in a mouse model of Barth syndrome" (2010).

Dr. Phoon is a pediatric cardiologist at New York University School of Medicine, where he has worked since completing his training in 1996. He was the director of the Pediatric & Fetal Echocardiography Lab from 2003-2015 until he stepped down to focus further on his research. Dr. Phoon maintains Board certifications in both General Pediatrics and Pediatric Cardiology.

Presentations: Recapitulating the Barth cardiomyopathy in the inducible mouse model of *tafazzin* deficiency *(Sci/Med Sessions)*; Mitochondria and Barth syndrome *(Family Sessions)*

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; and (3) to understand genetic contributions to congenital heart disease. In 2014, Dr. Pu's research was recognized by the American Heart Association as one of the top ten cardiovascular disease research advances of 2014. 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" (*partial funding provided by Barth Syndrome Trust* — 2012), "Using induced pluripotent stem cells and modified RNAs to model and correct Barth syndrome" (2011), and "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 and is Board Certified in Pediatric Cardiology.

Presentation: Reactive oxygen species cause abnormal calcium handling in Barth syndrome stem cell-derived cardiomyocytes *(Sci/Med Sessions)*

Dominic N. Reeds, MD — Assistant Professor, Division of Endocrinology, Metabolism and Lipid Research and Division of Geriatrics and Nutritional Science, Washington University School of Medicine, St. Louis, MO; Medical Director, Nutrition Support Service, Barnes-Jewish Hospital, St. Louis, MO, USA

Dr. Reeds has expertise in performing patient-oriented clinical research studies exploring abnormalities in glucose, lipid and protein in metabolism in a variety of human diseases including obesity, HIV/AIDS, and both type 1 and type 2 diabetes and has more than 40 peer-reviewed publications and book chapters related to these fields. He has been working as a collaborator with Dr. Todd Cade, an expert in Barth syndrome, for the last seven years, performing clinical studies designed to clarify the molecular and physiologic pathogenesis of cardioskeletal metabolic dysfunction in Barth syndrome and using these results to explore potential therapies for people with Barth syndrome.

Dr. Reeds was born in Southampton UK, and later attended the University of Texas at Austin graduating with a degree in Biochemistry. He received his medical degree from Texas Tech University Health Science Center Medical School and performed his residency in Internal Medicine at the Strong-Memorial Hospital/University of Rochester, NY. After serving as Chief Resident in Internal Medicine he received fellowship training in Endocrinology, Diabetes and Metabolism at Washington University School of Medicine in St. Louis, MO. He is board certified in both Internal Medicine and Endocrinology.

Presentation: Participation in clinical research: Lessons learned in St. Louis *(Family Sessions)*

Mindong Ren, PhD — Research Associate Professor, Departments of Molecular Cell Biology and Anesthesiology, New York University School of Medicine, New York, NY; Scientific and Medical Advisory Board, Barth Syndrome Foundation, USA

Dr. Ren is currently on the faculty of the Department of Anesthesiology, where he teaches first-year medical students Cell Biology and Histopathology. His research for the last 12 years has been focused on the pathogenic mechanisms of Barth syndrome, using cell cultures, fruit flies, and mice as model systems. Dr. Ren was awarded the following research grants from BSF entitled "Drug repositioning for Barth syndrome" (2011); Pathogenetic mechanism and genetic suppressors of Barth syndrome" (2006), and "A drosophila model of Barth syndrome" (2004).

Dr. Ren holds a PhD (1993) and an MS (1990) from New York University School of Medicine, and a BS (1984) from Peking University, P.R. China.

Presentation: Assessing the effects of Bezafibrate on the cardiomyopathy induced by adult-onset knockdown of *tafazzin* in a mouse model *(Sci/Med Sessions)*

Stacey Reynolds, PhD, OTR/L — Associate Professor, Department of Occupational Therapy, Virginia Commonwealth University, Richmond, VA, USA

Dr. Reynolds' research is focused on investigating physiologic stress reactivity patterns in children with Sensory Processing Disorder, and characterizing behavioral and physiological patterns of sensory processing in children. 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" (*partial funding provided by Association Barth France* — 2013).

Dr. Reynolds completed her PhD in Health Related Science at Virginia Commonwealth University (2007) and her post-doctoral training in behavioral neuroscience at the University of Florida.

Consultation: Pill swallowing workshop; Presentation: Skills and practical strategies to support feeding; Pill swallowing school: Lessons learned *(Family Sessions)*

Marcus E. Sernel — Chairman, Board of Directors, Barth Syndrome Foundation, USA

Mr. Sernel earned a BS in Chemical Engineering from the University of Illinois in 1992 and then went on to graduate from Loyola University Chicago School of Law in 1997. He continued to leverage his interest in science and his law degree as a Partner at Kirkland & Ellis. Mr. Sernel's specialty at Kirkland & Ellis is intellectual property where he concentrates on patent and other technology related litigation with a particular focus on chemical, pharmaceutical and biotechnology matters. His natural interest in science and his familiarity with the pharmaceutical and biotechnology industries enable him to provide advice as a Board member as BSF pushes deeper and deeper into research and potential medical treatments for Barth syndrome.

Presentation: The new frontier: Future clinical trials and you *(Family Sessions)*

Yuguang (Roger) Shi, PhD — Joe R. & Teresa Lozano Long Distinguished Chair Professor, Metabolic Biology, Barshop Institute for Longevity and Aging Studies, University of Texas Health Sciences Center, San Antonio, TX, USA

Dr. Shi's research focuses on identifying novel molecular mechanisms underlying mitochondrial dysfunction in Barth syndrome and aging-related metabolic diseases, including type 2 diabetes, obesity, diabetic complications, and cardiovascular diseases and then developing novel treatments. His laboratory pioneered the research work on cardiolipin remodeling in obesity and type 2 diabetes, and demonstrated a key role of pathological remodeling of cardiolipin by ALCAT1 in controlling mitochondrial etiology of Barth syndrome and aging-related metabolic diseases. Dr. Shi was awarded the following research grants from BSF entitled "Molecular mechanisms underlying a causative role ALCAT1 in the pathogenesis of Barth syndrome" (2015), and "Regulation of cardiomyopathy by ALCAT1 in Barth syndrome" (2012).

Previously, Dr. Shi held various positions at Penn State College of Medicine where his work uncovered a novel signaling pathway by which GLP-1 regulates glucose-sensing by pancreatic beta cells. He also worked at Eli Lilly and Company, where his research led to the cloning and characterization of PERK kinase, a major regulator of ER-stress and translational control, and several first in class enzymes involved in synthesis and remodeling of phospholipids including cardiolipin.

Presentation: Targeted deletion of ALCAT1 mitigates cardiomyopathy in mouse model of Barth syndrome through restoration of mitophagy *(Sci/Med Sessions)*

Colin G. Steward, PhD, FRCP, FRCPCH — Professor of Paediatric Stem Cell Transplantation, School of Cellular & Molecular Medicine, University of Bristol; Honorary Consultant, Bristol Royal Hospital for Children; Clinical Lead, NHS Barth Syndrome Service, Bristol, United Kingdom; Scientific and Medical Advisory Board, Barth Syndrome Foundation, USA

Dr. Steward is a Pediatric Hematologist and Clinical Lead for the multidisciplinary NHS National Barth Syndrome Service which is run in partnership with Michaela Damin and her colleagues at the Barth Syndrome Trust. The service provides free diagnostic testing for UK residents by cardiolipin analysis and *TAZ* gene sequencing, annual multidisciplinary clinics 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 and testing.

Dr. Steward is a previous recipient of the Barth Syndrome Foundation's Varner Award for Pioneers in Science and Medicine (2012).

Clinic: Neutropenia; Presentation: Incidence, characteristics and management of neutropenia in Barth syndrome *(Sci/Med Sessions)*

Donna Strain, RN — Prince Charles Hospital and Chermside Day Hospital; Mother of Affected Individual, Queensland, Australia

Born in Bristol, United Kingdom, Mrs. Strain moved to Adelaide, Australia as a teenager, with her family, in 1987, where she finished school and joined the Australian Army as a Medic. She was transferred to Brisbane in 1996, where she met her husband. After leaving the army, Mrs. Strain studied Natural Medicine and qualified with a diploma in Naturopathy. After working in this field for a short time, she decided to continue her studies at the Queensland University of Technology completing a dual degree in Nursing and Public Health. As a registered nurse, she has since specialised in gastroenterology/endoscopy and has completed graduate studies in this area.

Mrs. Strain has two boys with her husband and has also been fortunate enough to be a step mother to her husband's two daughters. Their eldest son has Barth syndrome and was diagnosed shortly after birth in 1999. They have been lucky enough to benefit from the support and knowledge of the Barth Syndrome Foundation since this time.

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

Dr. Strathdee's research interests focus on generating and studying models of human diseases. The aim of 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 a research grant from BSF entitled "Characterization of a conditional knockout of *tafazzin* in the mouse" (*partial funding provided by Barth Syndrome Trust* — 2013).

Dr. Strathdee studied for a PhD at the Beatson Institute for Cancer Research, under the supervision of Professor Allan Balmain. He was awarded a PhD and a BSc in Immunology (Honors) from the University of Glasgow. This was followed by post-doctoral training in the laboratories of Professor John Clark (at the Roslin Institute) and subsequently Professor Seth Grant (now at the University of Edinburgh). In 2009, he moved back to the Beatson as Head of the Transgenic Technology lab, where study models of human diseases are generated.

Presentation: The phenotype of *Taz* null mutation in mouse closely resembles Barth syndrome *(Sci/Med Sessions)*

Arnold W. Strauss, MD — Professor of Pediatrics; Associate Director for External Relations and Strategic Projects, Cincinnati Children's Research Foundation, Cincinnati, OH; Scientific and Medical Advisory Board, Barth Syndrome Foundation, USA

Dr. Strauss' research interests include the molecular basis of genetic disorders of mitochondrial fatty acid oxidation and myocardial energetics. He has published more than 240 research and invited articles and served as research mentor for more than 60 graduate students and post-doctoral fellows. Dr. Strauss was awarded the following research grants from BSF entitled "*Tafazzin* function in animal models of Barth syndrome" (2004), and "A mouse gene ablation model of Barth syndrome" (2002).

Dr. Strauss was Director of the Cincinnati Children's Research Foundation and Rachford Professor and Chair of Pediatrics at the University of Cincinnati. Dr. Strauss has held positons of James C. Overall Professor and Chair of Pediatrics and Medical Director of Vanderbilt Children's Hospital, Nashville, and Professor and later Director of the Division of Pediatric Cardiology at Washington University, St. Louis.

Dr. Strauss holds an undergraduate degree cum laude in philosophy from Stanford University and his medical degree from Washington University, St. Louis. Dr. Strauss's pediatric residency was at St. Louis Children's Hospital and cardiology fellowship was at St. Louis Children's Hospital and Washington University.

Chair: Potential therapies for Barth syndrome *(Sci/Med Sessions)*

Carolyn Taylor (Spencer), MD — Associate Professor, Division of Pediatric Cardiology, The Children's Heart Program of South Carolina, Medical University of South Carolina, Charleston, SC, USA

Dr. Taylor is a pediatric cardiologist with additional specialty training as a pediatric echocardiographer. Her research and clinical interests are in the areas of imaging and evaluation of cardiac function. Assessment of cardiac performance using echocardiography as well as evaluation of functional capacity in various forms of cardiomyopathy and cardio-skeletal myopathy are central to her clinical practice and research effort. She has been actively involved in the Barth syndrome scientific and research community and has published multiple manuscripts on cardiomyopathy, exercise capacity and results from the Barth Syndrome Registry.

Dr. Taylor holds a medical degree from the Chapel Hill School of Medicine, University of North Carolina, NC, USA.

Clinic: Longitudinal evaluation of cardiomyopathy and outcome in Barth syndrome; Presentation: Evaluation of cardiac function in the current era *(Family Sessions)*

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

Dr. Toth completed his PhD in Microbiology at 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.

Small Group Meetings: Therapeutic ideas for Barth syndrome; Presentation: Where's my cure for Barth syndrome? *(Family Sessions)*

Jeffrey A. Towbin, MD, FAAP, FACC, FAHA — Co-Director, The Heart Institute, Le Bonheur Children's Hospital; Chief of Pediatric Cardiology, St. Jude Children's Research Hospital; Chief of Pediatric Cardiology, University of Tennessee Health Science Center, Memphis, TN; 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 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.

Chair: Clinical characteristics of Barth syndrome *(Sci/Med Sessions)*

Hilary J. Vernon, MD, PhD — Assistant Professor, McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University and at the Kennedy Krieger Institute; Director, Barth Syndrome Clinic, Kennedy Krieger Institute, Baltimore, MD; Scientific and Medical Advisory Board, Barth Syndrome Foundation, 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 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 and the National Institutes of Health Working Group for Mitochondrial Models and is on the faculty of the North American Metabolic Academy of the Society for Inherited Metabolic 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.

Clinic: Investigation into clinical, metabolic and molecular factors in Barth syndrome (Clinic); Presentations: Clinical, molecular, and metabolomic studies in Barth Syndrome: Outcomes from the 2014 Barth Syndrome Foundation Scientific, Medical & Family Conference *(Sci/Med Sessions)*; Updates and results from the 2014 BSF/Johns Hopkins Study *(Family Sessions)*

Ronald J. A. Wanders, PhD — Professor, Clinical Enzymology of Inherited Metabolic Diseases, AMC Principle Investigator, Head of Lab. Genetic Metabolic Diseases (LGMD), Academic Medical Center (AMC), University of Amsterdam, the Netherlands; Scientific and Medical Advisory Board, Barth Syndrome Foundation, USA

Dr. Wanders is an internationally recognized expert on biochemistry and enzymology in relation to metabolism and metabolic disorders. Research focus in the last five years has been on biochemical and pathophysiological aspects of disorders of mitochondrial fatty acid oxidation, peroxisome metabolism & biogenesis and isoprenoid/cholesterol biosynthesis, areas of research for which LGMD has a long-standing international reputation. Research in LGMD is translational in nature and approached in a multifunctional way, involving integrated biochemical, molecular biological and genetic approaches. LGMD also houses a metabolomics facility with expertise predominantly but not exclusively on lipodomics.

Dr. Wanders holds a PhD degree in Biochemistry (metabolism) from the University of Amsterdam, the Netherlands (1982). He also holds an MSc degree in Biochemistry from the University of Amsterdam (1977).

Dr. Wanders is a recipient of the following awards: Hamdan Award for Medical Sciences (2012); Komrower Award, Dubai, United Arab Emirates; Society for the Studies of Inborn Errors of Metabolism (SSIEM) (2012); International Federation for Clinical Chemistry and Laboratory Medicine (IFCC) Award (2011); SSIEM Research Award (SSIEM) (1995); Miami-Hospital for Sick Children Award (1993); ORTHO Research Award, Dutch Society of Clinical Chemistry and Medical Laboratory Science (1988); Noel Raine Award, Society for the Studies of Inborn Errors of Metabolism (SSIEM) (1985).

Chair: Cardiolipin and Barth syndrome *(Sci/Med Sessions)*

Saskia Wortmann-Hagemann, MD, PhD — Head, Department for Metabolic Disorders, Children's Hospital Salzburger Landeskliniken; Department of Pediatrics, Paracelsus Medical University, Salzburg, Austria

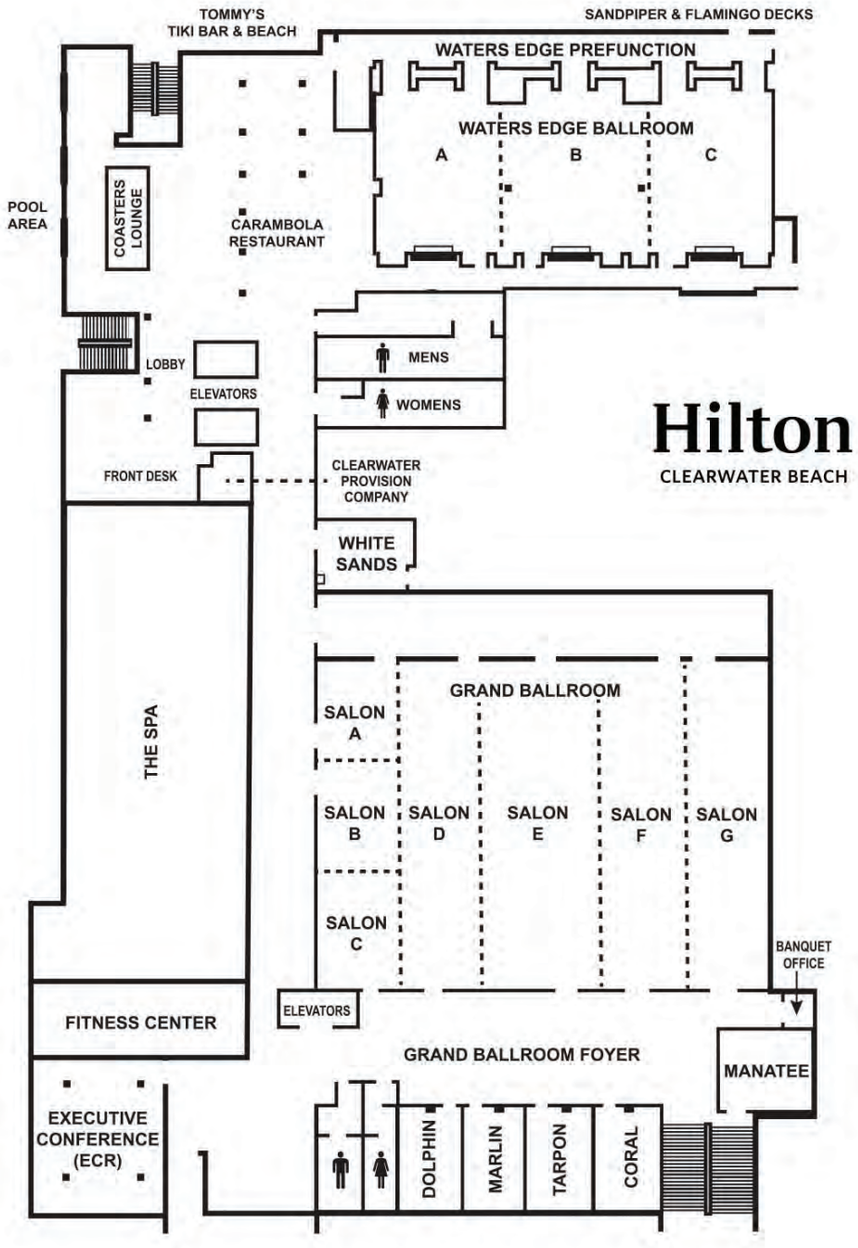
Dr. Wortmann-Hagemann's research focuses on mitochondrial disorders in general, with a special focus on inborn errors of metabolism with 3-methylglutaconic aciduria as a discriminative feature. She was the first to describe biallelic mutations in SERAC1 to underlie MEGDEL syndrome and biallelic mutations in CLPB to underlie MEGCANN syndrome.

Dr. Wortmann-Hagemann finished her PhD thesis "The 3-methylglutaconic acidurias — revisited" in biomedical sciences at the Radboud University Nijmegen Medical Centre, Nijmegen, The Netherlands in 2013 and is a recipient of several prizes [including 1st Prize for the best young researcher, Dutch Society of Pediatrics (NVK) Wadman-van Gennip prize, Dutch society for metabolic diseases (ESN)].

Dr. Wortmann-Hagemann trained as a pediatrician at the Radboudumc Amalia Children's Hospital, Nijmegen, The Netherlands and at the Children's Hospital, University of Essen, Germany and was board certified in 2010. She completed her fellowship in metabolic disorders at the Radboudumc Nijmegen in 2014.

Presentation: The growing family of disorders in the biosynthesis and remodelling of phospholipids; the brothers and sisters of Barth syndrome *(Sci/Med Sessions)*

NOTES



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