Barth Syndrome Foundation

Celebrating BSF's 10th Anniversary

...A decade of community

...A decade of education

...A decade of discovery

Barth Syndrome

5th International
Scientific, Medical & Family Conference

July 26-31, 2010

Renaissance at SeaWorld
Orlando, FL

Dear Friends,

Our mission was established in 2000 to address an unmet need. The prelude of the Barth Syndrome Foundation was during our first international gathering of families in 2000 in Baltimore, Maryland. The overwhelming sentiment of need was made clear in an open discussion among families that had traveled from around the globe to attend this event. Barth syndrome had claimed the lives of our beloved children and loomed as a constant threat to the lives of those children still living with the disease.

In essence this group of parents mirrored the optimistic moralistic children's story of The Little Engine That Could by Watty Piper in overcoming a seemingly impossible task. During that meeting our potential became evident. We couldn't wait for someone else to do something about this situation. We had to do it ourselves. The potential to surmount the impossible task we alone could not accomplish became evident through a shared belief that we could. To reach our vision we needed a map. For every goal we reached, the momentum gained.

Ten years later Barth syndrome continues to claim lives and continues as a threat to the lives of those we love. However, through our efforts we now know more about the disease. We are now more aware of potential risks imposed by the disease. In ten years we have done a great deal. We haven't reached our destination. Therefore we will never ever give up on the people this disease affects and with uncompromising appreciation of the mission we serve. And the little group that thought they could has grown into a group that knows they can.

The biennial conference represents more than just a program or a meeting. It is the keystone program of our organization. It embodies our organization's programs, mission, our goals and our core values in relentless pursuit of our ultimate vision. The tagline for our 2010 biennial conference is:

…A decade of community

…A decade of education

…A decade of discovery

Don't just take my word for it. Please visit www.barthsyndrome.org to see what a "little group that could" has accomplished over the past decade.

Sincerely,

Valerie ('Shelley') Bowen
President

PRE-CONFERENCE SESSIONS

SUNDAY, JULY 25, 2010	
7:30 pm — 10:00 pm	**BSF BOARD OF DIRECTORS MEETING** *(Veiltail Boardroom)*
MONDAY, JULY 26, 2010	
ALL DAY	**PORTRAITS BY AMANDA CLARK** *(Yellowtail A)* Early family arrivals scheduled on this date
2:00 pm — 3:00 pm	**NEW FAMILY ORIENTATION** *(Wedgwood Ballroom)* Audience: First-Time Family Attendees *Led by Michael Calhoun, Chris Hope, and Leslie Buddemeyer*
3:00 pm — 5:00 pm	**CONSENT AND ASSENT SIGNING** *(Wedgwood Ballroom)* *Led by Kate McCurdy* NOTE: The following groups of individuals participating in clinics must attend this session: **CONSENTS** Parents of all boys under the age of 18 Adult males 18 and older **ASSENTS** Minor affected males (12 – 18 years of age)
6:15 pm — 6:45 pm	**LADIES AND GENTLEMEN'S MEETING** *(Veiltail Boardroom)* Audience: Affected men ages 18 and older; Siblings ages 18 and older *Led by John Wilkins*
7:00 pm — 10:00 pm	**WELCOME RECEPTION** *(Wedgwood Ballroom)* *Sponsored by "Friends of Jake Fairchild"* **7:00 pm - 8:00 pm** All Family attendees (Children, Parents, Grandparents, etc.) Special Guest Introductions **8:00 pm - 10:00 pm** CONFERENCE 101 Overview of Conference and important reminders
8:00 pm — 10:00 pm	**LITTLE TYKE MOVIE NIGHT** *(Yellowtail B)* *Sponsored by DW and Tracy Brody (In Honor of Bly Brody)* *Snacks sponsored by Oldewage Family & Friends* Audience: Children up to four years of age Note: One adult family member must accompany children who are not potty trained. **DANCE PARTY** *(Crystal Ballroom A)* *Sponsored by BSF of Canada* Audience: ALL Youth ages 5 and up

Friends of Jake Fairchild

are the proud sponsors
of the

WELCOME CEREMONY
July 26, 2010

TRACY & DW BRODY

are the proud sponsors
of the

LITTLE TYKE MOVIE NIGHT
July 26, 2010

"God bless each family
affected by Barth syndrome."
In Honor of Bly Brody

Barth Syndrome
Foundation of Canada

BARTH SYNDROME FOUNDATION OF CANADA

is the proud sponsor
of the

MONDAY NIGHT DANCE
July 26, 2010

TUESDAY & WEDNESDAY
JULY 27-28, 2010

BARTH SYNDROME CLINICS

7:30 am — 5:30 pm

(Crystal Ballroom C, D, E)

Two days of Barth clinics where families and clinicians share and learn valuable information about the clinical aspects of Barth syndrome. This exchange between families and clinicians about the many nuances of Barth syndrome gives rise to opportunities to explore new ideas and discuss issues of common theme. Data collected as a result of these clinics will feed the Barth Syndrome Medical Database & BioRepository to further the understanding of this disorder. The BSF clinics have been hailed as a model approach by other health advocacy groups. (Arts, crafts, movies, outdoor games and Wii activities for all.)

RESEARCH INVESTIGATIONS	CONSULTATIONS
Cardiology/Electrophysiology Carolyn T. Spencer, MD *(Principal Investigator)* Sharonda Aikens, NCMA/CSR Paul Benni, PhD Petar Breitinger, PA-C Randall Bryant, MD Barry J. Byrne, MD, PhD W. Todd Cade, PT, PhD Lisa Crawford, RDCS Jane A. Day, PhD, PT Judith Geva, MSW Jill Handsides, Clinical Research Nurse Julie Lewis, Sonographer Jodi Liphart, PT Connie Nixon, RN Sharon Redfearn, ARNP Selvi Senthilnathan Sara Seyhan Dianne Stanford, RN Beverly Tsai-Goodman **Chronic Pain Study** John Saroyan, MD, FAAP *(Principal Investigator)* Damini Taylor, MD Candidate **Education** Michele Mazzocco, PhD *(Principal Investigator)* **Medical Database & Biorepository** Amy Roberts, MD *(Principal Investigator)* Carolyn T. Spencer, MD *(Principal Investigator)* Melissa Maisenbacher, MS, CGC Debbie Riddiford, Clinical Nurse Specialist **Sensory Processing** Roxanna Bendixen, PhD OTR/L *(Principal Investigator)* Consuelo M. Kreider, MHS OTR/L Stacey Reynolds, PhD, OTR/L **Severe Chronic Neutropenia International Registry** Shannon Collins, PhD Candidate	**Genetics** Iris L. Gonzalez, PhD Rebecca L. Kern, MGC **Metabolism** Richard I. Kelley, MD, PhD **Neurology** Ariel Sherbany, MD, PhD **Neutropenia** Colin G. Steward, FRCP, FRCPCH, PhD Robert Sutphin, MD **Occupational/Physical Therapy** Jennifer Bargo, PT Lynn Hancock, PT Bobbie King, OTR/L Karin Colby Watson, MPT **Severe Chronic Neutropenia International Registry** Audrey Anna Bolyard, RN *(web-based)* **Vision Screening** Catheryn Johnson, OT **Vitals** Susan V. Wilkins, RN Linda Croxton, ARNP Rebecca Brown, Director, STREETLIGHT **Barth Syndrome Perspectives** John Wilkins

WEDNESDAY, JULY 28, 2010

SMALL GROUP MEETINGS

CARRIER DISCUSSIONS

1:00 pm – 2:30 pm *(Veiltail Boardroom)*	Discussions with girls 16 and older to discuss carrier related issues *Rebecca Kern, MGC, Genetic Counselor, Division of Metabolism, Department of Neurogenetics, Kennedy Krieger Institute, Baltimore, MD*
2:30 pm – 3:30 pm *(Veiltail Boardroom)*	Discussions with parents of potential carriers or parents of known carriers *Rebecca Kern, MGC*

GENETIC LABORATORY MEETING
(Closed Session)

4:00 pm – 6:00 pm *(Veiltail Boardroom)*	Meeting with BSF Science and Medicine representatives and geneticists from laboratories from around the United States.

YOUTH VOLUNTEER LEADERS MEETING
(Closed Session)

4:30 pm – 5:30 pm *(Crystal Ballroom A)*	Meeting with all youth volunteer leaders and set-up of activity stations (Glogster, etc.)

Youth Chairs	*Brandy Olson; Amer Randell*
AFFECTED BOYS	
8 through 13	*Lisa Duran*
14 through 17	*BJ Develle; Amer Randell*
18 and Older	*John Wilkins*
SIBLINGS	
8 through 13	*Michael Telles*
14 and Up	*Brandy Olson; Jessica Wilkins-Wiederspan*
Group Activities & Hang Time	*Travis Morris*

POOLSIDE PARTY
and
DIVE-IN MOVIE NIGHT
(Poolside)

7:00 pm – 10:00 pm	Enjoy the music of Dr. John Saroyan of the Manhattan Valley Ramblers Movie will start at Dark

THURSDAY, JULY 29, 2010

VARNER AWARD LUNCHEON 12:00 pm — 1:30 pm *(Crystal Ballroom D & E)*	
Introduction of John Wilkins and Jessica Wilkins-Wiederspan	*Shelley Bowen*
Varner Award Overview	*John Wilkins and Jessica Wilkins-Wiederspan*
Introduction of Daniela Toniolo, MD, PhD	*Jessica Wilkins-Wiederspan*
Ronald Wanders, PhD on behalf of Peter Vreken, PhD (posthumous)	*John Wilkins*
POSTER SESSION	
4:45 pm — 6:00 pm *(The Upper Deck)*	Open to Science and Medicine Conference Attendees
5:30 pm — 6:00 pm *(The Upper Deck)*	Poster Session open to Family Conference Attendees
AFTER HOURS SESSION	
5:45 pm — 7:00 pm *(Veiltail Boardroom)*	*Expanded dialogue with Dr. F. Jay Fricker about cardiac transplant related topics.* *Audience: Parents of children who have received transplants and affected males who have received heart transplants age 15 and older.*
KEYNOTE ADDRESS **Douglas C. Wallace, PhD** The Pathophysiology of Mitochondrial Disease	
7:00 pm — 8:00 pm *(Coral Ballroom A,B,C)*	All Conference attendees are welcome

Douglas C. Wallace, PhD — Donald Bren Professor of Molecular Medicine; Director, ORU for Molecular and Mitochondrial Medicine and Genetics; Professor of Biological Chemistry, Ecology and Evolutionary Biology, and Pediatrics, Center for Molecular and Mitochondrial Medicine & Genetics, University of California at Irvine, Irvine, CA, USA

Dr. Wallace has been a pioneer in the study of human mitochondrial genetics and the role of mitochondrial DNA variation in human evolution, disease, cancer, and aging. In the 1970s, Dr. Wallace defined the basic principles of human mitochondrial DNA genetics, demonstrating that the human mitochondrial DNA encodes heritable traits, is maternally transmitted, has a high mutation rate; that intracellular mixtures on mutant and normal mitochondrial DNA are common and can segregate randomly during both mitotic and meiotic cell division; and that the clinical phenotype of a mutation depends on the severity of the mitochondrial defect and the reliance of each individual tissue on mitochondrial energy production. Once Dr. Wallace had defined the basic principles of mitochondrial DNA genetics, he applied these principles to the investigation of human origins and disease. He also identified the first maternally inherited mitochondrial DNA diseases and has subsequently shown that deleterious mitochondrial DNA mutations are common and result in a plethora of complex multi-system diseases which encompasses all of the clinical phenotypes associated with aging, including neurological problems such as deafness, blindness, movement disorders, and dementias; cardiovascular disease; muscle degeneration and pain; renal dysfunction; endocrine disorders including diabetes, cancer, etc.

Dr. Wallace received his PhD in Microbiology & Human Genetics at Yale University School of Medicine, New York, NY. He was awarded the Passano Award for Mitochondrial Genetics (2000); the William Allan Award from the American Society of Human Genetics; and the Honorary Doctorate from the French University, Victor Segalen Bordeaux II. Dr. Wallace is a member of the American Board of Medical Genetics in Clinical Genetics, Clinical Biochemical Genetics, and Molecular Genetics; American Board of Pediatrics; Human Genome Organization; and the National Academy of Sciences.

FRIDAY, JULY 30, 2010

AMBASSADORS IN ACTION LUNCHEON	
12:00 pm — 1:00 pm *(Crystal Ballroom D & E)*	Welcome and Introductions — *Steve McCurdy* It's Not a Sprint; It's a Marathon — *Cherie Schrader* It Takes Collaboration — *Michael Schlame, MD* It Takes Teamwork — *Gary Rodbell*
1:10 pm — 1:30 pm *(Location TBD)*	GROUP PHOTO — ALL Conference Attendees *Amanda Clark, Amanda Clark Photography, Perry, FL*

Scientific & Medical Sessions *(Coral Ballroom A, B & C)*		**Family Sessions** *(Wedgewood Ballroom)*		**Affected Youth Sessions** *(Labrid, Koi, Japanero, Veiltail, Crystal A)*		**Sibling Sessions** *(Labrid, Ko, Japanero, Veiltail, Crystal A)*	
Thursday, July 29, 2010		**Thursday, July 29, 2010**		**Thursday, July 29, 2010**		**Thursday, July 29, 2010**	
7:30am-8:15am Breakfast Buffet (Crystal D & E)		7:30am-8:15am Breakfast Buffet *(Crystal D & E)*		7:30am-8:45am Breakfast Buffet *(Crystal D & E)*			
8:15am – 11:45am	Animal Models of Barth Syndrome	9:00am – 10:05am 10:05am – 11:45am	BRR Cardiomyopathy Update	9:00am – 11:00am Meet Group Leaders & Ice-Breakers *All Youth (Crystal A)*			
12:00pm – 1:30pm *(Crystal D&E)*	Varner Award Ceremony/ Luncheon	12:00pm – 1:30pm *(Crystal D&E)*	Varner Award Ceremony/Luncheon	11:05am – 11:55am	Ages 8-13 Music Journey *(Koi)* Ages 14-17 Hands on Genes *(Labrid)* Ages 18+ Adulthood *(Japanero)*	11:05am – 11:55am	Ages 8-13 Music Journey *(Koi)* Ages 14+ Glogster *(Crystal A)*
1:30pm – 4:45pm	Barth Syndrome Pathophysiology	1:45pm – 2:40pm 2:40pm – 3:35pm 3:35pm – 4:45pm	Long Range Plans Education Update on Research	12:00pm – 1:30pm Varner Award Luncheon *(Crystal D&E)*			
4:45pm – 6:00pm *(Upper Deck)*	Scientific Poster Session	5:30pm – 6:00pm *(Upper Deck)*	Poster Session	1:35pm – 2:35pm	Ages 8-13 Photography Fun *(Koi)* Ages 14-17 Glogster *(Crystal A)* Ages 18+ How Do We Help? *(Labrid)*	1:35pm – 2:35pm	Ages 8-13 Photography Fun *(Koi)* Ages 14+ Music Journey *(Japanero)*
7:00pm – 8:00pm *(Coral A,B,C)*	Keynote Address	5:45pm-7:00pm *(Veiltail Boardroom)*	After Hours Transplant Talk	2:40pm – 3:40pm	Ages 8-13 Hands on Genes *(Labrid)* Ages 14+ Photography Fun *(Crystal A)*	2:40pm – 3:40pm	Ages 8+ Hands on Genes *(Labrid)*
				3:45pm – 4:45pm Group Photo – All Youth *(Waterfall, Main Lobby)*			
Friday, July 30		Friday, July 30		Friday, July 30		Friday, July 30	
7:30am-8:15am Breakfast Buffet (Crystal D&E)		*7:30am-8:45am Breakfast Buffet (Crystal D&E)*		7:30am-8:45am – Breakfast Buffet *(Crystal D & E)*			
8:15am – 11:45am	Mitochondrial Dysfunction	9:00am – 9:55am 9:55am – 10:50am 10:50am – 11:45am	Nutrition & Metabolism The Common Thread Defining the Pain in BTHS	9:00am – 9:55am Group Activities - All Youth *(Crystal A)*			
12:00pm – 1:00pm *(Crystal D&E)*	Ambassadors in Action Luncheon	12:00pm – 1:00pm *(Crystal D&E)*	Ambassadors in Action Luncheon	10:00am – 10:50am	Ages 8-13 Glogster *(Crystal A)* Ages 14-17 Barth and Beyond *(Labrid)* Ages 18+ Music Journey *(Japanero)*	10:00am – 10:50am	Ages 8-13 Glogster *(Crystal A)* Ages 14+ Last Name Barth *(Koi)*
1:10pm – 1:30pm *(Location TBD)*	Group Photo	1:10pm – 1:30pm *(Location TBD)*	Group Photo	10:55am – 11:55am	Ages 8-13 Barth and Beyond *(Labrid)* Ages 14-17 Music Journey *(Japanero)* Ages 18+ What's in my Genes *(Koi)*	10:55am – 11:55am	Ages 8-13 Last Name Barth *(Veiltail)* Ages 14+ Photography Fun *(Crystal A)*
1:30pm – 5:15pm	Lipids, Tafazzin & Mitochondrial Metabolism	1:45pm – 2:40pm 2:40pm – 3:35pm 3:35pm – 4:40pm	Neutropenia Strength & Stamina Asymptomatic Carrier	12:00pm – 1:00pm Ambassadors in Action Luncheon *(Crystal D&E)*			
5:15pm – 5:45pm	Chairpersons Wrap-Up	6:00pm – 11:00pm *(Crystal C,D,E)*	Dinner & Social Event	1:10pm – 1:30pm Group Photo *(Location TBD)*			
6:00pm – 11:00pm *(Crystal C,D,E)*	Dinner & Social Event			1:30pm – 3:00pm – Line Dancing Class – All Youth *(Crystal C)* 3:05pm – 4:50pm – Glogster Projects & Hang Time *(Crystal A)* 6:00pm – 11:00pm – Dinner & Social Event *(Crystal C, D, E)*			
Saturday, July 31		Saturday, July 31		Saturday, July 31		Saturday, July 31	
8:15am – 11:30am *(Veiltail)*	SMAB Breakfast & Meeting	7:30am-8:45am Breakfast Buffet *(Crystal D&E)*		7:30am-8:45am – Breakfast Buffet *(Crystal D&E)*			
4:10pm – 5:30pm *(Crystal D&E)*	Closing Ceremony	9:00am – 12:00pm	Day-to-Day Life	9:00am – 9:35am – Group Activities – All Youth *(Crystal A)*			
		12:00pm – 1:00pm Luncheon Buffet *(Crystal D&E)*		9:40am – 10:35am	Ages 8-13 Group Activities *(Crystal A)* Ages 14+ Exercise Training *(Crystal C)*	9:40am – 10:35am	Ages 8+ Group Activities *(Crystal A)*
		1:00pm – 3:15pm 3:20pm – 4:00pm	Break-Out Sessions Attendee Perspectives	10:40am – 11:30am	Ages 8+ Insights from Insiders *(Koi)*	10:40am – 11:30am	Ages 8+ I'm Affected Too *(Japanero)*
		4:10pm – 5:30pm *(Crystal D&E)*	Closing Ceremony	11:45am – 12:00pm – Hang Time – All Youth *(Crystal A)*			
				12:00pm – 1:00pm Luncheon Buffet *(Crystal D&E)*			
				1:05pm – 2:05pm	Ages 8-17 Group Activities *(Crystal A)* Ages 18+ BTHS and Beyond *(Labrid)*	1:05pm – 2:05pm	Ages 8+ Group Activities *(Crystal A)*
				2:10pm – 3:55pm All Youth - Finalize Glogster for Finale *(Crystal A)*			
				4:10pm – 5:30pm Closing Ceremony *(Crystal D&E)*			

SCIENTIFIC AND MEDICAL SESSIONS

THURSDAY, JULY 29, 2010

7:30 am — 8:15 am	**Breakfast** *(Crystal Ballroom D & E)*
(Coral Ballroom A, B, C)	**ANIMAL MODELS OF BARTH SYNDROME** *Chair—Michael Schlame, MD, New York University School of Medicine, New York, NY*
8:15 am — 8:20 am	Introduction — *Jack Higgins*
8:30 am — 9:00 am	**Suppressors of Tafazzin-Deficiency Phenotype in Drosophila—Implications for Barth Syndrome** *Mindong Ren, PhD, New York University School of Medicine, New York, NY*
9:00 am — 9:30 am	**Cardiac and Skeletal Muscle Defects in Mouse Model of Tafazzin Deficiency** *Zaza Khuchua, PhD, Children's Hospital Medical Center, Cincinnati, OH*
9:30 am — 10:00 am	**Characterization of the Cardiac Lipidomic and Bioenergetic Phenotype in the Inducible shRNA Tafazzin Knockdown Model of Barth Syndrome** *Michael A. Kiebish, PhD, Washington University School of Medicine, St. Louis, MO*
10:00 am — 10:30 am	**Exercise as a Therapy for Mitochondrial Dysfunction; Implications for BTHS** *Mark Tarnopolsky, MD, PhD, FRCP(C), McMaster University, Hamilton, Ontario, Canada*
10:30 am — 11:00 am	**Dietary Influences on the Cardiolipin Composition of the Heart** *Genevieve Sparagna, PhD, University of Colorado at Boulder, Boulder, C0*
11:00 am — 11:30 am	**Attempts at a Rat Model of Barth Syndrome** *Carol Moreno-Quinn, MD, PhD, Medical College of Wisconsin, Milwaukee, WI*
11:30 am — 11:45 pm	**Brainstorming about Future Directions** *Led by Chair: Michael Schlame, MD, New York University School of Medicine, New York, NY*
VARNER AWARD CEREMONY / LUNCHEON	
12:00 pm — 1:30 pm	**Varner Award Ceremony / Luncheon Buffet** *(Crystal Ballroom D & E)*
	BARTH SYNDROME PATHOPHYSIOLOGY *Chair—Richard I. Kelley, MD, PhD, Johns Hopkins University School of Medicine; Kennedy Krieger Institute, Baltimore, MD*
1:30 pm — 1:35 pm	Introduction — *Darryl Byrd*
1:35 pm — 2:00 pm	**Characterizations of Nutrient Metabolism in Barth Syndrome** *W. Todd Cade, PT, PhD, Washington University School of Medicine, St. Louis, MO*
2:00 pm — 2:30 pm	**Functional Capacity and Cardiomyopathy in Barth Syndrome** *Carolyn T. Spencer, MD, Children's Hospital Boston, Boston, MA*
2:30 pm — 3:00 pm	**Barth Syndrome Registry Update** *Amy Roberts, MD, Children's Hospital Boston, Boston, MA*
3:00 pm — 3:30 pm	**Cellular and Molecular Mechanisms of Neutropenia in Barth Syndrome** *Andrew Aprikyan, PhD, University of Washington School of Medicine, Seattle, WA*
3:30 pm — 4:00 pm	**Establishing a National Specialized Service for Barth Syndrome in the UK** *Colin G. Steward, FRCP, FRCPCH, PhD, Royal Children's Hospital, Bristol, England*
4:00 pm — 4:30 pm	**Neutropenia in Barth Syndrome: On Calcium and Mitochondria** *Bram J. van Raam, PhD, Sanford-Burnham Institute for Medical Research, La Jolla, CA*
4:30 pm — 4:45 pm	**Brainstorming about Future Directions** *Led by Chair: Richard I. Kelley, MD, PhD, Johns Hopkins University School of Medicine; Kennedy Krieger Institute; Baltimore, MD*
4:45 pm — 6:00 pm	**POSTER SESSION** *(The Upper Deck)*
6:00 pm — 7:00 pm	**Dinner Break** (at your own expense)
7:00 pm — 8:00 pm *(Coral Ballroom A,B,C)*	**KEYNOTE ADDRESS: The Pathophysiology of Mitochondrial Disease** *Douglas C. Wallace, PhD, Center for Molecular and Mitochondrial Medicine & Genetics, University of California at Irvine, Irvine, California*

POSTER SESSION

THURSDAY, JULY 29, 2010

(The Upper Deck)

4:45 pm — 6:00 pm: Physicians & Scientists	**5:30 — 6:00 pm**: Families Welcome

POSTER 1: CHARACTERIZATION OF AN SHRNA-MEDIATED TAFAZZIN KNOCKDOWN MOUSE MODEL FOR BARTH SYNDROME

MS Soustek, DJ Falk, AS Lewin, BJ Byrne

Department of Pediatrics, Department of Molecular Genetics and Microbiology, University of Florida, Gainesville, FL

POSTER 2: INVESTIGATING BARTH SYNDROME: THE ROLE OF CARDIOLIPIN IN MAINTENANCE OF MITOCHONDRIAL MORPHOLOGY

N Fei

Department of Biological Sciences, Wayne State University, Detroit, MI

POSTER 3: GENETIC AND VIRAL GENOME ANALYSIS OF CHILDHOOD CARDIOMYOPATHY: THE PCMR/PCSR EXPERIENCE

JA Towbin, L Sleeper, JL Jefferies, S Colan, SA Webber, CE Canter, DT Hsu, SM Ware, JD Wilkinson, EJ Orav, SE Lipshultz

Cincinnati Children's Medical Center, Cincinnati, OH, University of Miami, Miami, FL

POSTER 4: IMPACT OF SAMPLE COLLECTION ON TAFAZZIN MRNA VARIANTS

SM Kirwin; SE Swain; AL Manolakos; IL Gonzalez

Nemours Biomedical Research Department; Alfred I. duPont Hospital for Children, Wilmington, DE

POSTER 5: ROLE OF CARDIOLIPIN AND PHOSPHATIDYLETHANOLAMINE IN MITOCHONDRIAL FUSION

AS Joshi, ML Greenberg

Department of Biological Sciences, Wayne State University, Detroit, MI

POSTER 6: A REVERSED-PHASE LC-MS/MS CARDIOLIPIN METHOD FOR THE DIAGNOSIS OF BARTH SYNDROME SUITABLE FOR USE IN A ROUTINE METABOLIC LAB

A Bowron[1], R Frost[2], S Heales[3], C Steward[4]

[1]Department of Clinical Biochemistry, Bristol Royal Infirmary, BS2 8HW, UK. [2]Waters Corporation, Elstree, UK. [3]Institute of Child Health, University College London, UK. [4]Bristol Royal Hospital for Children, UK

POSTER 7: LIPIDOMIC AND BIOENERGETIC EFFECT OF CARDIAC-SPECIFIC OVEREXPRESSION OF CARDIOLIPIN SYNTHASE

MA Kiebish, K Yang, HF Sims, DJ Mancuso, Z Zhou, S Guan, RW Gross, X Han

Washington University School of Medicine, Dept of Internal Medicine, Division of Bioorganic Chemistry and Molecular Pharmacology, St. Louis, MO

POSTER 8: FETAL BARTH SYNDROME: A CASE OF PROGRESSIVE CARDIOMYOPATHY IN UTERO

B Tsai-Goodman[1], RP Martin[1], CG Steward[2]

Departments of Paediatric Cardiology[1] and Haematology[2], Royal Hospital for Children, Bristol, England

POSTER 9: FULL-LENGTH HUMAN TAFAZZIN PROTECTS THE HUMAN CELLS FROM SERUM-WITHDRAWN APOPTOSIS

Y Xu, M Schlame

Anesthesiology Department, NYUMC, New York City, NY

POSTER 10: LOSS OF MITOCHONDRIAL ANIONIC PHOSPHOLIPIDS LEADS TO PERTURBATION OF MITOCHONDRIAL/CELLULAR IRON HOMEOSTASIS

VA Patil, VM Gohil, S Gupta, G Li, ML Greenberg

Dept. of Biological Sciences, Wayne State University, Detroit, MI

POSTER 11: BARTH SYNDROME IN A FEMALE PATIENT

L Cosson[1], A Toutain[2], G Simard[3], F Paoli[1], W Kulik[4], FM Vaz[4], H Blasco[5], A Chantepie[1], F Labarthe[1,6]

[1]Dept of Paediatrics, CHU Tours, Tours, France, [2]Genetic, CHU Tours, Tours, France, [3]CHU Angers, INSERM U694, Angers, France, [4]Academic Medical Center, Amsterdam, Netherlands, [5]Biochemistry, CHU Tours, Tours, France, 6INSERM U921, Tours, France

SCIENTIFIC AND MEDICAL SESSIONS

FRIDAY, JULY 30, 2010

7:30 am — 8:15 am	**Breakfast** *(Crystal Ballroom D & E)*
(Coral Ballroom A, B, C)	**MITOCHONDRIAL DYSFUNCTION AND ITS IMPACT ON HUMAN DISEASE** *Chair—Barry J. Byrne, MD, PhD, University of Florida, Gainesville, FL*
8:15 am — 8:20 am	Introduction — *Ben Thorpe*
8:30 am — 9:00 am	**Cardiolipin Provides Signaling Platforms on Mitochondria** *Eyal Gottlieb, PhD, Beatson Institute for Cancer Research, Glasgow, Scotland*
9:00 am — 9:30 am	**Cardiomyopathy in Barth Syndrome** *John Lynn Jefferies, MD, MPH, FAAP, FACC, Texas Children's Hospital, Houston, TX*
9:30 am — 10:00 am	**Mitochondrial Disease** *Charles Hoppel, MD, Case Western Reserve University, Cleveland, OH*
10:00 am — 10:30 am	**Tafazzin Knockdown Causes Hypertrophy in Neonatal Cardiac Myocytes** *Quan He, PhD, Henry Ford Hospital, Detroit, MI*
10:30 am — 11:00 am	**Role of Stearoyl-CoA Desaturase in Metabolism: Implications in Human Diseases** *James Ntambi, MD, PhD, University of Wisconsin, Madison, WI*
11:00 am — 11:30 am	**Mitochondrial-targeted Antioxidants Protect the Diaphragm from Mechanical Ventilation-induced Weakness** *Peter Adhihetty, PhD, University of Florida, Gainesville, Fl*
11:30 am — 11:45 am	**Brainstorming about Future Directions** *Led by Chair: Barry J. Byrne, MD, PhD, University of Florida, Gainesville, FL*
AMBASSADORS IN ACTION LUNCHEON *(Crystal Ballroom D, E)*	
12:00 pm — 1:00 pm 1:10 pm — 1:30 pm	**Ambassadors in Action Luncheon** **Group Photograph**
(Coral Ballroom A, B, C)	**LIPIDS, TAFAZZIN, AND MITOCHONDRIAL METABOLISM IN BARTH SYNDROME** *Chair—Miriam Greenberg, PhD, Wayne State University, Detroit, MI*
1:30 pm — 1:35 pm	Introduction — *Kevin Baffa*
1:35 pm — 2:00 pm	**Regulation of Mitochondrial Fusion by Cardiolipin** *Jodi Nunnari, PhD, University of California, Davis, CA*
2:00 pm — 2:30 pm	**Barth and DCMA—Same Disease from Two Genes?** *Robert E. Jensen, PhD, Johns Hopkins University, Baltimore, MD*
2:30 pm — 3:00 pm	**Tafazzin and Mitochondrial Supermolecular Asssemblies** *Ashim Malhotra, PhD, New York University, New York, NY*
3:00 pm — 3:30 pm	**Characterizing Barth Syndrome Mutant Tafazzins** *Steven Claypool, PhD, Johns Hopkins University, Baltimore, MD*
3:30 pm — 4:00 pm	**Using Systematic Arrays to Explore the Genetic Landscape of the TAZ1 Gene in Saccharomyces cerevisiae** *Christopher McMaster, PhD, Dalhousie University, Halifax, Nova Scotia, Canada*
4:00 pm — 4:30 pm	**Properties of Cardiolipin and its Roles in Mitochondria** *Richard Epand, PhD, McMaster University, Hamilton, Ontario, Canada*
4:30 pm — 5:00 pm	**Role of Monolysocardiolipin Acyltransferase-1 in Barth Syndrome Lymphoblast**s *Grant Hatch, PhD, University of Manitoba, Winnipeg, Manitoba, Canada*
5:00 pm — 5:15 pm	**Brainstorming about Future Directions** *Led by Chair: Miriam Greenberg, MD, Wayne State*
5:15 pm — 5:45 pm	**CONFERENCE CHAIRPERSONS WRAP-UP**
6:00 pm — 11:00 pm	**DINNER AND SOCIAL EVENT** *(Crystal Ballroom C,D, E)*

SATURDAY, JULY 31, 2010

8:15 am — 11:30 am	**Scientific and Medical Advisory Board Breakfast & Meeting** *(Veiltail Boardroom)*
4:10 pm — 5:30 pm	**Closing Ceremony** *(Crystal Ballroom D & E)*

FAMILY SESSION

THURSDAY, JULY 29, 2010

7:30 am — 8:45 am	**Breakfast** *(Crystal Ballroom D & E)*
(Wedgwood Ballroom)	**WELCOME**
9:00 am — 9:05 am	**Welcome** — *Shelley Bowen, President*
	BARTH SYNDROME REGISTRY *This session will provide insights about how each of the boys' data and biological specimens have facilitated research and expanded the base of knowledge about this disease by looking at the bigger picture of Barth syndrome.*
9:05 am — 9:20 am	**Session Preview**
9:20 am — 9:25 am	**Speaker Introduction** — *Bryan Drake*
9:25 am — 9:50 am	**Barth Syndrome Registry Update** *Carolyn T. Spencer, MD, Children's Hospital Boston, Boston, MA* *Amy Roberts, MD, Children's Hospital Boston; Harvard Medical School, Boston, MA*
9:50 am — 10:05 am	**Q & A Session**
	CARDIAC ASPECTS OF BARTH SYNDROME *This session will provide an overview of what is known about the cardiac function and electrophysiology aspects of Barth syndrome and to address concerns and considerations related to heart transplants.*
10:05 am — 10:20 am	**Session Preview**
10:20 am — 10:25 am	**Speaker Introduction** — *Nicole Addington*
10:25 am — 10:45 am	**Overview of Cardiac Function with Barth Syndrome** *Carolyn T. Spencer, MD, Children's Hospital Boston, Boston, MA*
10:45 am — 11:05 am	**Arrhythmias** *Randall Bryant, MD, University of Florida-Jacksonville/Gainesville, Jacksonville, FL*
11:05 am — 11:25 am	**Dispelling the Myths about Heart Transplants** *Frederick Jay Fricker, MD, Congenital Heart Center at Shands Medical Plaza, Gainesville, FL*
11:25 am — 11:45 am	**Q & A Session**
12:00 pm — 1:30 pm	**Varner Award Luncheon** *(Crystal Ballroom D & E)*
	LONG-RANGE PLAN *The session will highlight the importance of embracing a philosophy of no surprises for all those involved in making informed healthcare decisions.*
1:45 pm — 2:00 pm	**Session Preview**
2:00 pm — 2:05 pm	**Speaker Introduction** — *Julie Fairchild*
2:05 pm — 2:25 pm	**Conversations and Considerations in Coordination of Care** *Arwa Saidi, MD, BCh, Congenital Heart Center at Shands Medical Plaza, Gainesville, FL*
2:25 pm — 2:40 pm	**Q & A Session**
	EDUCATION
2:40 pm — 2:55 pm	**Session Preview**
2:55 pm — 3:00 pm	**Speaker Introduction** — *Michael Calhoun*
3:00 pm — 3:20 pm	**Educating a Child with Barth Syndrome** *Michele Mazzocco, PhD, Kennedy Krieger Institute, Baltimore, MD*
3:20 pm — 3:35 pm	**Q & A Session**
	UPDATE ON RESEARCH *In a "what difference has it made" approach, this session will inventory the progress that has been made through research and the plans to expand the knowledge about Barth syndrome as we move forward.*
3:35 pm — 3:40 pm	**Speaker Introduction** — *Sonja Schlapak*
3:40 pm — 4:00 pm	**Update on Scientific Research** *Matthew J. Toth, PhD, Science Director, BSF*
4:00 pm — 4:20 pm	**Update on Clinical Research** *Barry J. Byrne, MD, PhD, University of Florida College of Medicine, Gainesville, FL*
4:20 pm — 4:45 pm	**Q & A Session**
5:45 pm — 7:00 pm	**After Hours Transplant Discussions** *(*Please see pg. 7 for more information) *(Veiltail Boardroom)*

FAMILY SESSION

FRIDAY, JULY 30, 2010

7:30 am — 8:45 am	**Breakfast** *(Crystal Ballroom D & E)*
(Wedgwood Ballroom)	**NUTRITIONAL AND METABOLIC ASPECTS OF BARTH SYNDROME** *This session will focus on identifying and recognizing potential risks, and the proper care to address those risks.*
9:00 am — 9:15 am	**Session Preview**
9:15 am — 9:20 am	**Speaker Introduction** — *Andrew Buddemeyer*
9:20 am — 9:40 am	**Nutritional and Metabolic Aspects of Barth Syndrome** *Richard I. Kelley, MD, PhD, Johns Hopkins University School of Medicine; Kennedy Krieger Institute, Baltimore, MD*
9:40am — 9:55 am	**Q & A Session**
	THE COMMON THREAD *This session will provide an overview of cardiolipin in understandable terms.*
9:55 am — 10:10 am	**Session Preview**
10:10 am — 10:15 am	**Speaker Introduction** — *Cathy Ritter*
10:15 am — 10:35 am	**Sometimes the Small Things Make a Big Difference** *Frederic Vaz, PhD, University of Amsterdam, Amsterdam, The Netherlands*
10:35 am — 10:50 am	**Q & A Session**
	DOES IT HURT? *This session will focus on defining the pain experience in Barth syndrome.*
10:50 am — 11:05 am	**Session Preview**
11:05 am — 11:10 am	**Speaker Introduction** — *Tim Maksin*
11:10 am — 11:30 am	**Defining the Pain Experience in Barth Syndrome** *John Saroyan, MD, FAAP, Columbia University College of Physicians and Surgeons, New York, NY*
11:30 am — 11:45 am	**Q & A Session**
	AMBASSADORS IN ACTION LUNCHEON
12:00 pm — 1:00 pm 1:00 pm — 1:30 pm	**AMBASSADORS IN ACTION LUNCHEON** *(Crystal Ballroom D & E)* **Group Photo**
	NEUTROPENIA AND BARTH SYNDROME *This session will provide an update of what we now understand about neutropenia and Barth syndrome.*
1:45 pm — 2:00 pm	**Session Preview**
2:00 pm — 2:05 pm	**Speaker Introduction** — *Jeannette Thorpe*
2:05 pm — 2:25 pm	**Neutropenia** *Colin G. Steward, FRCP, FRCPCH, PhD, Royal Hospital for Children, Bristol, UK*
2:25 pm — 2:40 pm	**Q & A Session**
	STRENGTH AND STAMINA *This session will provide insights gained from the Nutrient Metabolism Research Study (funded by BSF).*
2:40 pm — 2:55 pm	**Session Preview**
2:55 pm — 3:00 pm	**Speaker Introduction** — *Michelle Telles*
3:00 pm — 3:20 pm	**Insights About Strength and Stamina with Barth Syndrome** *W. Todd Cade, PT, PhD, Washington University School of Medicine, St. Louis, MO*
3:20 pm — 3:35 pm	**Q & A Session**
	THE ASYMPTOMATIC CARRIER *This session will provide an overview of X chromosome selection and inactivation among female carriers of Barth syndrome.*
3:35 pm — 3:50 pm	**Session Preview**
3:50 pm — 3:55 pm	**Speaker Introduction** — *Greg Holly*
3:55 pm — 4:25 pm	**How the Gene that Causes Barth Syndrome Affects the Carrier's X Chromosome** *Karen Orstavik, MD, PhD, University of Oslo, Oslo, Norway*
4:25 pm — 4:40 pm	**Q & A Session**
6:00 pm — 11:00 pm	**DINNER and SOCIAL EVENT for ALL** *(Crystal Ballroom C, D, E)*

FAMILY SESSION

SATURDAY, JULY 31, 2010

7:30 am — 8:45 am	**Breakfast** *(Crystal Ballroom D & E)*
(Coral Ballroom A, B, C)	**DAY-TO-DAY LIFE; ONE DAY AT A TIME**
9:00 am — 9:15 am	**Session Preview**
9:15 am — 9:20 am	**Speaker Introduction** — *Donna Strain*
9:20 am — 9:40 am	**The Daily Challenges of Conquering Fear—Sometimes It's Just Complicated** *Rebecca Brown, Director, STREETLIGHT Adolescent Programming & Palliative Care BM, University of Florida, Gainesville, FL*
9:40am — 9:45 am	**Speaker Introduction** — *Eliza McCurdy*
9:45 am — 10:05 am	**When I grow up I'm going to…** **Financial and Social Independence** *Thomas Nurse, Financial Planner, Manning & Nurse, Tampa, FL*
10:05 am — 10:25 am	**Reversal of Roles; One Baby Step at a Time** **The Ongoing Process of raising a chronically ill child in becoming a self-advocate** *John Reiss, PhD, Institute for Child Health Policy, University of Florida, Gainesville, FL*
10:25 am — 10:30 am	**Speaker Introduction** — *Susan McCormack*
10:35 am — 10:50 am	**Considerations and Concerns with Genetic Testing** *Rebecca Kern, MGC, Kennedy Krieger Institute, Baltimore, MD*
10:50 am — 11:10 am	**When Barth Syndrome Hits Home** **Psychosocial Coping Mechanisms with Barth Syndrome** *Jay St. Amant, MS, University of Florida, Gainesville, FL*
11:10 am — 11:30 am	**Sensory Processing** *Stacey Reynolds, PhD, OTR/L, University of Florida, Gainesville, FL*
11:30 am — 12:00 pm	**Q & A Session**
12:00 pm — 1:00 pm	**LUNCHEON**

1:05 pm — 2:05 pm

BREAK-OUT SESSION
PARENT PERSPECTIVES

(Yellowtail B)	*(Coral Ballroom A, B, C)*
Mothers *Moderators: John Reiss, PhD; Rebecca Brown*	**Fathers** *Moderators: Tom Nurse; Jay St. Amant, MS*

2:10 pm — 3:15 pm

BREAK-OUT SESSIONS
AGE SPECIFIC

Ages 0 through 6	Ages 6 through 11	Ages 12 through16	Ages 17 and Older
(Coral A)	*(Coral B)*	*(Coral C)*	*(Yellowtail A)*
Infancy & Preschool Years *Moderator: Nicole Addington*	**Early Childhood & Adolescent Years** *Moderator: Julie Floyd*	**Pre-Teen & Puberty Years** *Moderator: Donna Strain*	**Early Adulthood & Adulthood Years** *Moderator: Cathy Ritter*

3:20 pm — 4:00 pm

CONFERENCE ATTENDEE PERSPECTIVES
(Crystal Ballroom D & E)

Personal Perspectives about 2010 Conference from Attendees

4:10 pm — 5:30 pm

CLOSING CEREMONY
(Crystal D & E)

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AFFECTED INDIVIDUALS & SIBLING SESSIONS

THURSDAY, JULY 29, 2010

7:30 am - 8:45 am — **Breakfast** *(Crystal Ballroom D & E)*

9:00 am — 11:00 am
MEET YOUR GROUP LEADER
Ice-Breakers & Youth Activities
ALL Affected Individuals and Siblings
(Crystal Ballroom A)

AFFECTED BOYS		
Ages 8 through 13 *Lisa Duran*	**Ages 14 through 17** *BJ Develle, Amer Randell*	**Ages 18 and Older** *John Wilkins*
SIBLINGS		
Ages 8 through 13 *Michael Telles*	**Ages 14 & Up** *Brandy Olson* *Jessica Wilkins-Wiederspan*	**Group Activities & Hang Time** *Travis Morris*

11:05 am — 11:55 am

AFFECTED INDIVIDUALS			SIBLINGS		
Ages 8-13	**Ages 14-17**	**Ages 18+**	**Ages 8-13**	**Ages 14-17**	**Ages 18+**
(Koi)	*(Labrid)*	*(Japanero)*	*(Koi)*	*(Crystal Ballroom A)*	
MUSIC FOR THE JOURNEY *Rebecca Brown*	**HANDS ON GENES** *Rebecca Kern, MGC*	**ADULTHOOD: Dream It... Believe It and Achieve It** *Tom Nurse*	**MUSIC FOR THE JOURNEY** *Rebecca Brown*	**CREATIVE EXPRESSIONS Glogster Projects** *Amer Randell*	

12:00 pm — 1:30 pm
VARNER AWARD LUNCHEON
(Crystal Ballroom D & E)

1:35 pm — 2:35 pm

Ages 8-13	Ages 14-17	Ages 18+	Ages 8-13	Ages 14-17	Ages 18+
(Koi)	*(Crystal A)*	*(Labrid)*	*(Koi)*	*(Japanero)*	
PHOTOGRAPHY FUN Tips and Techniques in Capturing Beautiful Images *Amanda Clark*	**CREATIVE EXPRESSIONS Glogster Projects** *Amer Randell*	**HOW DO WE HELP: Our Future Role in the Organization** *John Wilkins*	**PHOTOGRAPHY FUN** Tips and Techniques in Capturing Beautiful Images *Amanda Clark*	**MUSIC FOR THE JOURNEY** *Rebecca Brown*	

2:40 pm — 3:40 pm

Ages 8-13	Ages 14-17	Ages 18+	Ages 8-13	Ages 14-17	Ages 18+
(Labrid)	*(Crystal A)*		*(Labrid)*		
HANDS ON GENES *Rebecca Kern, MGC*	**PHOTOGRAPHY FUN** Tips and Techniques in Capturing Beautiful Images *Amanda Clark*		**HANDS ON GENES** *Rebecca Kern, MGC*		

3:45 pm — 4:45 pm
GROUP PHOTO
ALL Affected Individuals and Siblings
(Waterfall - Main Lobby)

AFFECTED INDIVIDUALS & SIBLING SESSIONS

FRIDAY, JULY 30, 2010

7:30 am - 8:45 am — **Breakfast** *(Crystal Ballroom D & E)*

9:00 am — 9:55 am
GROUP ACTIVITIES
(Crystal Ballroom A)

ALL Affected Individuals and Siblings

Group Leaders
Lisa Duran, BJ Develle, Amer Randell, John Wilkins, Michael Telles, Brandy Olson, Jessica Wilkins-Wiederspan, Travis Morris

10:00 am — 10:50 am

AFFECTED INDIVIDUALS			SIBLINGS		
Ages 8-13	**Ages 14-17**	**Ages 18+**	**Ages 8-13**	**Ages 14-17**	**Ages 18+**
(Crystal A)	*(Labrid)*	*(Japanero)*	*(Crystal A)*	*(Koi)*	
CREATIVE EXPRESSIONS Glogster Poster *Amer Randell*	**BARTH SYNDROME AND BEYOND** *Randall Bryant, MD*	**MUSIC FOR THE JOURNEY** *Rebecca Brown*	**CREATIVE EXPRESSIONS Glogster Poster** *Amer Randell*	**MY LAST NAME IS NOT BARTH** *Tom Nurse* *Jay St. Amant, MS*	

10:55 am — 11:55 am

Ages 8-13	Ages 14-17	Ages 18+	Ages 8-13	Ages 14-17	Ages 18+
(Labrid)	*(Japanero)*	*(Koi)*	*(Veiltail)*	*(Crystal A)*	
BARTH SYNDROME AND BEYOND *Randall Bryant, MD*	**MUSIC FOR THE JOURNEY** *Rebecca Brown*	**WHAT'S IN MY GENES** *Rebecca Kern, MGC*	**MY LAST NAME IS NOT BARTH** *Tom Nurse* *Jay St. Amant, MS*	**PHOTOGRAPHY FUN** Tips and Techniques in Capturing Beautiful Images *Amanda Clark*	

12:00 pm — 1:00 pm
AMBASSADORS IN ACTION LUNCHEON
(Crystal Ballroom D & E)

1:00 pm — 1:30 pm
GROUP PHOTO
(Location TBD)

1:30 pm — 3:00 pm
LINE DANCING CLASS
with Colby Robertson and Krista Lynn-Vann

ALL Affected Individuals and Siblings / ALL Group Leaders
(Crystal Ballroom C)

3:05 pm — 4:50 pm
GLOGSTER PROJECT & HANG TIME

ALL Affected Individuals and Siblings / ALL Group Leaders
(Crystal Ballroom A)

6:00 pm — 11:00 pm
SOCIAL EVENT

ALL Conference Attendees
(Crystal Ballroom C, D, E)

AFFECTED INDIVIDUALS & SIBLING SESSIONS

SATURDAY, JULY 31, 2010

7:30 am - 8:45 am — **Breakfast** *(Crystal Ballroom D & E)*

9:00 am — 9:35 am
(Crystal Ballroom A)

GROUP ACTIVITIES — ALL Affected Individuals and Siblings
Introduction to Appreciative Inquiries *(Led by Amer Randell)*

Group Leaders
Lisa Duran, BJ Develle, Amer Randell, John Wilkins, Michael Telles, Brandy Olson, Travis Morris

9:40 am — 10:35 am

AFFECTED INDIVIDUALS			SIBLINGS		
Ages 8-13	**Ages 14-17**	**Ages 18+**	**Ages 8-13**	**Ages 14-17**	**Ages 18+**
(Crystal A)	*(Crystal C)*		*(Crystal A)*		
GROUP ACTIVITIES *All Group Leaders*	**EXERCISE TRAINING** *Todd Cade, PT, PhD*		**GROUP ACTIVITIES** *All Group Leaders*		

10:40 am — 11:30 am

Ages 8-13	**Ages 14-17**	**Ages 18+**	**Ages 8-13**	**Ages 14-17**	**Ages 18+**
(Koi)			*(Japanero)*		
INSIGHTS FROM THE INSIDERS **Group Discussions** *BJ Develle, Amer Randell, John Wilkins*			**I'M AFFECTED TOO** **Group Discussions** *Brandy Olson, Travis Morris*		

11:45 am — 12:00 pm
HANG TIME
(Crystal Ballroom A)
ALL Affected Individuals and Siblings / ALL Group Leaders

12:00 pm — 1:00 pm
YOUTH LUNCHEON
(Buffet located in Crystal Ballroom D & E — Sitting in Crystal Ballroom C)
ALL Affected Individuals and Siblings Ages 8 and Older / ALL Group Leaders

1:05 pm — 2:05 pm

Ages 8-13	**Ages 14-17**	**Ages 18+**	**Ages 8-13**	**Ages 14-17**	**Ages 18+**
		(Labrid)			
		BTHS AND BEYOND *Randall Bryant, MD*			

1:05 pm — 2:10 pm

Ages 8-13	**Ages 14-17**	**Ages 18+**	**Ages 8-13**	**Ages 14-17**	**Ages 18+**
(Crystal Ballroom A)			*(Crystal Ballroom A)*		
Group Activities *ALL Group Leaders*			**Group Activities** *ALL Group Leaders*		

2:10 pm — 3:55 pm
FINALIZE GLOGSTER PROJECTS FOR FINALE & GROUP ACTIVITIES
(Crystal Ballroom A)
ALL Affected Individuals and Siblings
ALL Group Leaders

4:10 pm — 5:30 pm
CONFERENCE FINALE
All Conference Attendees
(Crystal Ballroom D & E)

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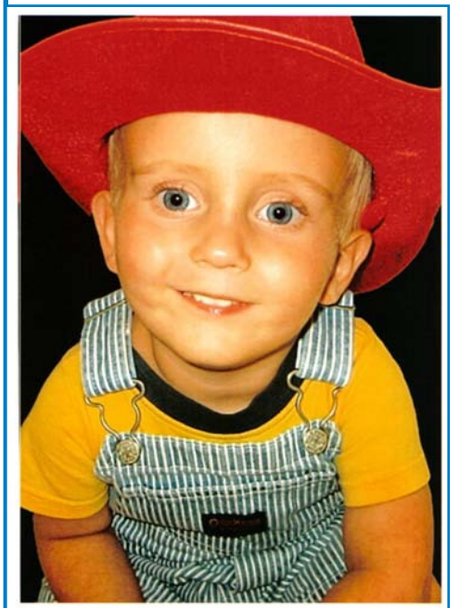

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Peter Adhihetty, PhD — Assistant Professor, Department of Applied Physiology and Kinesiology, University of Florida, Gainesville, FL, USA

Dr. Adhihetty's research is focused on the molecular mechanisms that regulate mitochondrially-mediated cell death in skeletal muscle and neural tissue with relation to losses in muscle mass (i.e. aging, physical inactivity) and loss of neurons in neurodegenerative disorders (i.e. Huntington's), respectively. Additionally, Dr. Adhihetty's research investigates whether exercise can improve impaired mitochondrial function in these various diseases/conditions.

Dr. Adhihetty conducted his doctoral work in skeletal muscle mitochondrial bioenergetics at York University in Toronto. Dr. Adhihetty did his postdoctoral training in mitochondrial dysfunction in neurodegenerative disorders in the Department of Neurology and Neuroscience at Weill Cornell Medical College. He is a member of the Canadian Society for Exercise Physiology, American College of Sports Medicine, American Physiological Society, and the Society for Neuroscience.

Presentation: Mitochondrial-targeted Antioxidants Protect the Diaphragm from Mechanical Ventilation-induced Weakness *(Sci/Med Session)*

Andrew Aprikyan, PhD — Research Assistant Professor of Medicine, Division of Hematology, University of Washington School of Medicine, Seattle, WA, USA

Dr. Aprikyan's research focuses on (1) molecular biology of bone marrow stem cells; (2) signal transduction pathways in leukemogenesis; (3) molecular defects/diagnostics of inherited disorders; (4) structure-to-function relationship, molecular modeling; (5) apoptosis and cell cycle in bone marrow failure disorders; (6) cellular and animal models and molecular therapy of severe neutropenia; and (7) therapeutic regenerative potential of bone marrow and induced pluripotent stem cells.

Dr. Aprikyan was awarded his PhD in Molecular Biology at the Institute of Molecular Biology, Academy of Sciences, Moscow, Russia (1988). He was Senior Scientist, Department of Molecular Biology of Aging, NASA, Institute of Biochemistry, Yerevan, Armenia (1990-1993). He was Senior Fellow, Division of Oncology, Department of Medicine, University of Washington, Seattle, WA (1993-1997), and in 1998, Senior Fellow, Division of General Internal Medicine, Department of Medicine, University of Washington, Seattle, WA.

Presentation: Cellular and Molecular Mechanisms of Neutropenia in Barth Syndrome *(Sci/Med Session)*

Rebecca Brown, Director of STREETLIGHT; Adolescent Programming & Palliative Care, University of Florida, Gainesville, FL, USA

Ms. Brown is the Director of a unique palliative care program, STREETLIGHT, at Shands/UF that supports adolescents and young adults who are living with critical or chronic illnesses. It is built around the power of peer support, using a team of 60+ University of Florida pre-med students who companion daily with hospitalized youth. The goal is to reconnect them with the street (the path they were on before getting ill), and to enable them to progress, not regress developmentally, and effectively manage their condition with a sense of control, maturity, and courage.

Ms Brown pursued her Master of Divinity degree in Berkeley, California in interfaith and youth spirituality. The premise of her master's thesis, Streetlight, was that today's generation of youth forms community and accesses spirituality and hope through music, pop culture and strong peer relationships. The Streetlight healthcare model is particularly helpful for the adolescent or young adult who has been removed from life on the street with peers.

Presentations:
The Daily Challenges of Conquering Fear—Sometimes It's Just Complicated *(Family Session)*
Break-Out Session: A Parent's Perspective (Mother) *(Family Session)*
Presentation:Music for the Journey *(BTHS Youth/Young Adult and Sibling Sessions)*

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

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

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

Presentations:
Arrythmias *(Family Session)*
Barth Syndrome and Beyond *(BTHS Youth/Young Adults and Sibling Sessions)*

Barry J. Byrne, MD, PhD — Associate Chair and Professor of Pediatrics and Molecular Genetics and Microbiology, College of Medicine, Department of Pediatrics; Molecular Genetics and Microbiology; Director of the Powell Gene Therapy Center at the University of Florida, Gainesville, FL; Scientific and Medical Advisory Board, Barth Syndrome Foundation, Inc.; Advisory Board, Barth Syndrome Registry & Biorepository, Boston, MA, USA

Dr. Byrne's laboratory is focused on molecular approaches to diagnosis and treatment of heart failure in infants and children. Therapeutic approaches rely on AAV-mediated gene therapy in animal models of heart failure and clinical studies in human subjects. As a model system, they are focusing on a form of heart failure due to glycogen storage disease. Additional studies are focused on the use of AAV vectors for gene therapy targeted to striated muscle and liver for the production of other therapeutic proteins. These projects include treatment of glycogen storage disease type I, hemophilia, and other heart failure models. The use of AAV vectors in conjunction with stem cells is being tested in the context of tissue regeneration in cardiomyopathy with the use of autologous stem cells, which have cardiomyogenic potential. These programs are being supported by the American Heart Association, Muscular Dystrophy Association and the National Institutes of Health (NHLBI, NIDDK, and NCRR).

Chair—Mitochondrial Dysfunction and Its Impact on Human Disease *(Sci/Med Session)*
Presentation: Update on Clinical Research *(Family Session)*

W. Todd Cade, PT, PhD — Assistant Professor of Physical Therapy and Internal Medicine, Washington University School of Medicine, St. Louis, MO, USA

Dr. Cade's research interests include mechanisms and treatments for whole-body and myocardial nutrient metabolism abnormalities in metabolic diseases such as HIV-associated metabolic syndrome, diabetes, and Barth syndrome and in normal and pathologic pregnancy.

Dr. Cade currently a PhD in Rehabilitation Science/Applied Physiology from the University of Maryland. He holds a BSc in Kinesiological Science from the University of Maryland, an MSc in Physical Therapy from the University of Miami, Florida, and holds an NIH funded Career Development Award from the National Institute of Diabetes and Digestive and Kidney Diseases. Dr Cade serves as a consultant of two Barth Syndrome Foundation grants entitled, "Safety and Efficacy of Aerobic Exercise Training in Barth Syndrome: A Pilot Study" (2009); and "Characterization of Nutrient Metabolism in Barth Syndrome" (2008).

Presentations:
Characterizations of Nutrient Metabolism in Barth Syndrome *(Sci/Med Session)*
Insights about Strength and Stamina with BTHS *(Family Session)*
Exercise Training *(BTHS Youth/Young Adult Session)*

Steven M. Claypool, MA, PhD — Assistant Professor, Department of Physiology, Johns Hopkins School of Medicine, Baltimore, MD, USA

Dr. Claypool's research interests are in understanding the interplay of lipids and proteins in mitochondrial physiology and pathophysiology. A major effort in Dr. Claypool's laboratory is to define the pathogenic mechanism of each identified Barth syndrome mutation using Saccharomyces cerevisiae as the model system.

Dr. Claypool received his PhD in Immunology from Harvard University. He holds a BA in Biological Sciences and a MA in Molecular, Cellular, and Developmental Biology from the University of California, Santa Barbara, California. Dr. Claypool is a member of the American Society for Biochemistry and Molecular Biology, the American Society of Cell Biology, and the United Mitochondrial Disease Foundation.

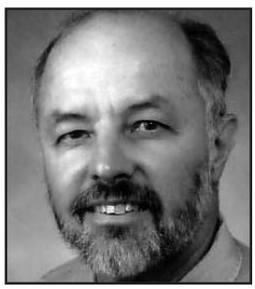
Presentation: Characterizing Barth syndrome mutant tafazzins *(Sci/Med Session)*

Richard Epand, PhD — Professor, Department of Biochemistry and Biomedical Sciences, McMaster University, Hamilton, Ontario, Canada

Dr. Epand's research interests include (1) studies of the structural and biological properties of membranes and their components; (2) formation of membrane domains including cardiolipin-rich domains in the mitochondria as well as cholesterol-rich domains; (3) physical properties of cardiolipin; arrangment of mitochondrial proteins between the inner and outer membranes; (4) regulation of the flux of energy from the mitochondrial matrix to the cytoplasm; and (5) movement of lipids between and within membranes. In 2007, Dr. Epand was awarded a research grant titled Consequences of the alteration of cardiolipin structure on the properties of the mitochondrial membranes.

Dr. Epand was elected a Fellow of the Biophysical Society (2002). He received the Senior Scientist Award from the Canadian Institutes of Health Research (2003-2008), and was a recipient of the Avanti Award for Research in Lipids from the Biophysical Society (1999); Postdoctoral Fellowship from the National Institutes of Health (1965-1967); Predoctoral Fellowship from the National Institutes of Health (1962-1965).

Presentation: Properties of Cardiolipin and its Roles in Mitochondria *(Sci/Med Session)*



Frederick Jay Fricker, MD — Professor of Pediatrics, Gerold Schiebler Eminent Scholar Chair; Division Chief of Pediatric Cardiology in the Congenital Heart Center at the University of Florida, Gainesville, FL, USA

Dr. Fricker's interests in Pediatric Cardiology center around the management of heart failure in children. He was involved in the first heart transplant of a child at the University of Pittsburgh in 1982. Over the next decade he led the pioneering Heart and Heart/Lung Transplant Program at Children's Hospital at the University of Pittsburgh. Dr. Fricker's other clinical interest is pulmonary hypertension in children. His passion is International Healthcare and Missions. Expertise is taken globally to establish programs of healthcare excellence where there are none.

Since 1995, Dr. Fricker has led the evolution of the University of Florida Pediatric Cardiology program into the Congenital Heart Center of all pediatric cardiology. His other mission is medical education and he has directed the Fellowship in Pediatric Cardiology over the last five years at the University of Florida. Dr. Fricker's professional memberships include: American Pediatric Society, International Society for Heart and Lung Transplantation (Board of Directors 2001-2004); Pediatric heart transplant study group; American Academy of Pediatrics.

Presentation: Dispelling the Myths about Heart Transplants *(Family Session)*

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

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

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

Eyal Gottlieb, PhD — Professor of Molecular Biology, University of Glasgow; Research Group Leader, The Beatson Institute for Cancer Research, Glasgow, UK

Dr. Gottlieb's research studies focus on the functions of the BCL2 family of proteins and how they regulate mitochondrial physiology and apoptosis. In 2003, he moved to Scotland to lead the Apoptosis and Tumour Physiology Laboratory at the Beatson Institute.

Dr. Gottlieb received his PhD in 1999 from the Weizmann Institute of Science, Israel, where he worked in the laboratory of Moshe Oren on p53 and apoptosis. He carried out his postdoctoral studies with Craig Thompson, first at the University of Chicago as an EMBO Fellow and later at the University of Pennsylvania as a Leukemia and Lymphoma Society Special Fellow.

Presentation: Cardiolipin Provides Signaling Platforms on Mitochondria *(Sci/Med Session)*

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

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

Dr. Greenberg has been awarded the following resesarch grants from the Barth Syndrome Foundation: "Perturbation of mitophagy in cardiolipin mutants" (2009); "The Role of Tafazzin in Mitochondrial Protein Import—Implications for Barth Syndrome" (2008); "Perturbation of the Osmotic Stress Response in Cardiolipin Deficient Mutants" (2007); "The Role of Phosphatidylglycerol in Activating Protein Kinase C Mediated Signaling" (2006); "Does Copper Deficiency Play a Role in Barth syndrome" (2005); "TAZ1 Gene Function in Yeast: A Molecular Model for Barth Syndrome" (2002).

Chair—Lipids, Tafazzin, and Mitochondrial Metabolism in Barth Syndrome *(Sci/Med Session)*

Grant M. Hatch, PhD — Professor, Department of Pharmacology and Therapeutics, Department of Biochemistry and Medical Genetics, Centre on Aging, University of Manitoba; Director, Centre for Research and Treatment of Atherosclerosis, University of Manitoba, Winnipeg, Canada; Scientific and Medical Advisory Board, Barth Syndrome Foundation, Inc.

Dr. Hatch's main research interests include examining the regulation of the metabolism of the mitochondrial phospholipid cardiolipin. Specifically, he is studying the regulation of cardiolipin synthesis and it's remodeling in mammalian tissues and cells. Dr. Hatch has been awarded the following research grants from the Barth Syndrome Foundation: "Role of Human Monolysocardiolipin Acyltransferase in Barth Syndrome" (funded by BSFCanada - 2009); "Cholesterol Metabolism in Barth Syndrome" (2005); and "The Molecular Mechanism of Barth Syndrome" (2002).

Dr. Hatch is a Canada Research Chair in Molecular Cardiolipin Metabolism.

Presentation: Role of Monolysocardiolipin Acyltransferase-1 in Barth Syndrome Lymphoblasts *(Sci/Med Session)*

Quan He, PhD — Research Scientist/Instructor, Hypertension & Vascular Research Division, Internal Medicine, Henry Ford Hospital, Detroit, MI, USA

Dr. He's research interests focus on the mechanisms of cardiomyopathy, signaling pathways of cardiac hypertrophy, and energy depletion in the failing heart. In 2007, Dr. He was awarded a research grant from the Barth Syndrome Foundation titled "Are reactive oxygen species involved in the development of dilated cardiomyopathy in Barth syndrome?", and has recently published an article on his research findings (Tafazzin knockdown causes hypertrophy of neonatal ventricular myocytes. Am J Physiol Heart Circ Physiol. 2010 Jul;299(1):H210-6. Epub 2010 Mar 26).

Presentation: Tafazzin Knockdown Causes Hypertrophy in Neonatal Cardiac Myocytes *(Sci/Med Session)*

Charles Hoppel, MD — Professor of Pharmacology; Professor of Medicine, Division of Clinical Pharmacology, Case Western Reserve University, Cleveland, OH; Associate Director of Research, Louis Stokes Veteran Affairs Medical Center, Cleveland, OH, USA

Dr. Hoppel's lab does extensive research in mitochondrial disease. The main focus of the laboratory is mitochondrial fatty acid oxidation, particularly as a potential site for control of the system. Dr. Hoppel is co-director of the Center for Inherited Disorders of Energy Metabolism at Case Western University and a Board member of the United Mitochondrial Disease Foundation.

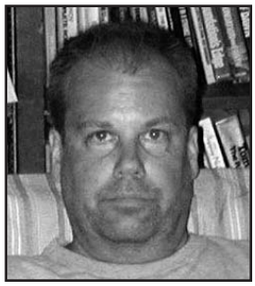
Presentation: Mitochondrial Disease *(Sci/Med Session)*

John Lynn Jefferies, MD, MPH — Assistant Professor, Pediatric Cardiology,Texas Children's Hospital; Assistant Professor, Adult Cardiovascular Diseases, Baylor College of Medicine; Assistant Professor, Pediatric Cardiology, MD Anderson Cancer Center, Houston, TX, USA

Dr. Jefferies' research interests focus on heart muscle disease and heritable forms of cardiovascular disease. His clinical interests include cardiomyopathy and heart failure, adult congenital heart disease, cardiac intensive care, and cardiovascular genetics. He is currently the Director of Cardiomyopathy and Advanced Heart Failure at the Texas Children's Hospital which is the largest pediatric heart failure program in the United States. He is also co-director of the Cardiovascular Genetics Service at Texas Children's Hospital which is one of the few such clinics internationally that cares for all patients with genetically mediated cardiac and vascular disease. He is a recognized expert in Pediatric Cardiomyopathy with multiple peer-reviewed manuscripts on the subject.

Dr. Jefferies has completed fellowships in both Adult Cardiovascular Diseases and Pediatric Cardiology with advanced training in Heart Failure, Cardiomyopathy, and Cardiovascular Genetics. He is a member of the American Heart Association, American Association of Pediatrics, and the American College of Cardiology.

Presentation: Cardiomyopathy in Barth Syndrome *(Sci/Med Session)*



Robert E. Jensen, PhD — Professor, Cell Biology & Anatomy, Johns Hopkins School of Medicine, Baltimore, MD, USA

Dr. Jensen's research focuses on the basic cell and molecular biology of mitochondria, including protein import, mitochondrial division and fusion, and mitochondria DNA inheritance. His work also includes the search for the biochemical basis of human mitochondrial disorders, including a Barth-like disorder defective in the mitochondrial protein import machinery.

Dr. Jensen holds a PhD from the Institute of Molecular Biology, University of Oregon, and the Department of Biochemistry & Biophysics, University of California, San Francisco. He held a Postdoctoral Fellowship at the Biocenter, University of Basel, Switzerland and at the University of California, San Diego. Dr. Jensen held a Damon Runyon-Walter Winchell Postdoctoral Fellowship and a Senior Postdoctoral Fellowship at the American Cancer Society, California Division.

Presentation: Barth and DCMA: Same Disease from Two Genes? *(Sci/Med Session)*

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

Dr. Kelley is board certified in Pediatrics and Medical Genetics and specializes in the diagnosis and treatment of inborn errors of metabolism. His research focuses on the determination of the biochemical basis of both known and novel genetic disorders and on the treatment of selected diseases, including Barth syndrome, Smith-Lemli-Opitz syndrome, and disorders of mitochondrial metabolism. Dr. Kelley also is a co-founder of and consulting geneticist for the Clinic for Special Children, a charitable medical facility for the diagnosis and treatment of genetic disorders among the Amish and Mennonite populations of Lancaster, Pennsylvania. Dr. Kelley is the recipient of the 2008 Varner Award for Pioneers in Science and Medicine.

Chair—Barth Syndrome Pathophysiology *(Sci/Med Session)*
Presentation: Nutritional and Metabolic Aspects of Barth Syndrome *(Family Session)*

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

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

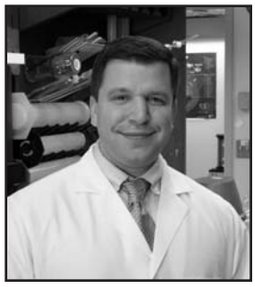
Presentations: Considerations and Concerns with Genetic Testing *(Family Session)*
Carrier Discussions—A discussion of carrier testing *(Sibling Session)*
Hands on Genes *(BTHS Youth/Young Adults and Sibling Sessions)*

Zaza Khuchua, PhD — Research Associate Professor, Molecular and Cardiovascular Biology, Children's Hospital Medical Center, Cincinnati, OH, USA

Dr. Khuchua's research focuses on mitochondrial function, structure and dynamics in cardiac cells in normal and pathological conditions; role of mitochondrial phospholipds in aerobic metabolism in heart; and the role of lipid molecules in cell signaling systems. Dr. Khuchua has been awarded a resesarch grant from the Barth Syndrome Foundation titled "The shRNA-mediated tafazzin knockdown mouse model for Barth syndrome" (2009).

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

Presentation: Cardiac and Skeletal Muscle Defects in Mouse Model of Tafazzin Deficiency *(Sci/Med Session)*



Michael Kiebish, PhD — Postdoctoral Associate, Xianlin Han Laboratory, Department of Internal Medicine, Division of Bioorganic Chemistry and Molecular Pharmacology, Washington University School of Medicine, St. Louis, MO, USA

Dr. Kiebish's research interests include investigating the molecular mechanism(s) of pathogenic cardiolipin remodeling and altered bioenergetic efficiency in diabetic cardiomyopathies and heart failure. He is also interested in examining the functional role of cardiolipin in bioenergetic efficiency in addition to developing novel bioinformatic tools to interpret the molecular mechanism of cardiolipin remodeling in silico. Dr Kiebish was awarded a research grant from the Barth Syndrome Foundation titled "Does Cardiolipin Synthase Upregulation Alleviate Cardiolipin Abnormalities and Bioenergetic Dysfunction in Barth Syndrome?" (2009).

Dr. Kiebish holds a PhD and a PA in Biology from Boston College (2008), and a BS from Villanova University (2002).

Presentation: Characterization of the Cardiac Lipidomic and Bioenergetic Phenotype in the Inducible shRNA Tafazzin Knockdown Model of Barth Syndrome *(Sci/Med Session)*

Ashim Malhotra, PhD — Senior Fellow, Departments of Surgery and Cell Biology, New York University School of Medicine, New York, NY, USA

Dr. Malhotra was awarded his doctorate degree in the Life Sciences in 2006. Subsequently, he trained as a post-doctoral fellow in the laboratory of Dr. Michael Schlame at the New York University School of Medicine. During this period, he worked on many aspects of Barth syndome research, with the overall goal of elucidation of the molecular mechanisms of the disease. In 2009, Dr. Malhotra was awarded a research grant from the Barth Syndrome Foundation titled "Distribution of Tafazzin and Cardiolipin in Mitochondrial Protein Complex Assemblies" (2008), and published the discovery of a potential therapeutic drug target for Barth syndrome. Apart from research, Dr. Malhotra has taught undergraduate and graduate courses at the City and State Universities of New York.

Presentation: Tafazzin and Mitochondrial Supermolecular Asssemblies *(Sci/Med Session)*

Michele Mazzocco, PhD — Professor of Psychiatry and Behavioral Sciences, Johns Hopkins University School of Medicine; Director, Math Skills Development Project, Kennedy Krieger Institute, Baltimore, MD, USA

Dr. Mazzocco's research interests include cognitive phenotypes of X chromosome related disorders, developmental learning disabilities, and mathematical disability. As a former classroom teacher, she is interested in the classroom applications of her research, and in supporting teacher training efforts in the area of understanding and supporting children's mathematical learning. Dr. Mazzocco was awarded a research grant from the Barth Syndrome Foundation titled "Early indices of learning difficulties in young boys with Barth syndrome" (2006).

Dr. Mazzocco completed a Masters degree in early childhood education, and doctoral training in Experimental Psychology. She completed a postdoctoral fellowship in Developmental Neuropsychology at the University of Colorado Health Sciences Center, and subsequently joined the faculty at Johns Hopkins and the Kennedy Krieger Institute in 1993. Dr. Mazzocco is a Fellow of the Association for Psychological Sciences, and a former Congressional Fellow for the American Association for the Advancement of Science.

Presentation: Educating a Child with Barth Syndrome *(Family Session)*

Christopher McMaster, PhD — Professor of Pediatrics and Biochemistry & Molecular Biology; Assistant Dean of Medicine, Graduate and Post-doctoral Studies, Canada Research Chair in Biosignalling, Atlantic Research Centre, Dalhousie University, Halifax, Nova Scotia, Canada

Dr. McMaster's research interests include the use of molecular, genetic, cell biological, and proteomic approaches to isolate new genes and proteins that regulate lipid metabolism. Once specific control points for the metabolism of a particular lipid are identified, specific medicines to aid in disease treatment are designed. In 2007, Dr. McMaster was awarded a research grant from the Barth Syndrome Foundation and BSFCanada titled "Synthetic GeneticsTowards Understanding Barth Syndrome Cell Biology."

Dr. McMaster holds a PhD from the University of Manitoba. He serves as Co-Director of the Cheminformatics Drug Discovery Lab. Dr. McMaster founded and is president of DeNovaMed, Inc., a Halifax based biotechnology company specializing in using computer aided drug design and synthesis focusing on diseases of lipid metabolism.

Presentation: Using Systematic Arrays to Explore the Genetic lLandscape of the *TAZ1* Gene in *Saccharomyces cerevisiae (Sci/Med Session)*

Carol P. Moreno Quinn, MD, PhD — Assistant Professor, Department of Physiology, Medical College of Wisconsin, Milwaukee, WI, USA

Dr. Moreno Quinn's specialization is functional genomics and her research interests include genetic dissection of complex diseases, physiological genomics, integrative physiology, systems biology, molecular genetics of hypertension and genetic determinants of polycystic kidney disease. Recent research focuses on identification of the genetic determinants of hypertension and renal disease in rat models of hypertension. In 2007, Dr. Moreno Quinn was awarded a research grant from the Barth Syndrome Foundation titled "Creation of a Rat Model of Barth Syndrome."

Dr. Moreno Quinn holds a PhD from the University of Murcia, Spain (1999). She held a Postdoctoral Fellowship in Physiology and Genomics, Medical College of Wisconsin (2004). Dr. Moreno Quinn is a member of the American Physiological Society, the Spanish Society of Hypertension, and the American Heart Association.

Presentation: Attempts at a Rat Model of Barth Syndrome *(Sci/Med Session)*

James Ntambi, MD, PhD — Professor of Biochemistry; Steenbock Professor of Nutrition, University of Wisconsin-Madison, Madison, WI, USA

Dr. Ntambi has made contributions to the field of nutritional biochemistry and his pioneering work on the genetic regulation of the stearoyl-CoA desaturase has recently led to many new insights into the importance of this enzyme in metabolism and in disease states such as obesity, diabetes, inflammation, atherosclerosis and cancer. His work will help explain the complex aspects of the "metabolic syndrome" and to advance understanding of nutrient gene interactions.

Dr. Ntambi holds a PhD in Biochemistry and Molecular Biology from Johns Hopkins University School of Medicine, Baltimore, MD. He received his BSc and MSc degrees in Biochemistry and Chemistry from Makerere University, Kampala, Uganda, and did his postdoctoral work at Johns Hopkins University School of Medicine, where he started his work in lipid metabolism and gene expression.

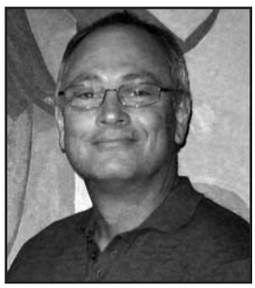
Presentation: Role of Stearoyl-CoA Desaturase in Metabolism: Implications in Human Diseases *(Sci/Med Session)*

Jodi Nunnari, PhD — Professor and Chair, Department of Molecular and Cellular Biology, University of California, Davis, CA, USA

One of Dr. Nunnari's major focuses is to understand how mitochondria divide and fuse. The essential players in these events remarkably include several highly conserved dynamin-related proteins (DRPs), which are large self-assembling GTPases that regulate membrane dynamics. Dr. Nunnari's research has led to understanding how, at a mechanistic level, DRPs function as the machines that divide and fuse mitochondria.

Dr. Nunnari holds a PhD in Pharmacology from Vanderbilt University and completed postdoctoral research in cell biology at University of California, San Francisco. She was awarded an NIH Postdoctoral Fellowship, an American Cancer Society Postdoctoral Fellowship and an American Heart Association Senior Postdoctoral Fellowship. Dr. Nunnari is a member of the American Society for Cell Biology and the American Society for Biochemistry and Molecular Biology.

Presentation: Regulation of Mitochondrial Fusion by Cardiolipin *(Sci/Med Session)*



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

Mr. Nurse has been involved in special needs advocacy for nearly nineteen years after his daughter Shelby was diagnosed with Spastic Quadriplegia Cerebral Palsy and he undertook the role of a 'stay at home father.' Mr Nurse began fulltime work in the disability field with Florida Development Disabilities Council as a Statewide Parent Liaison for early intervention. He worked as a Statewide Parent Consultant for the Florida Department of Health, Children's Medical Services, Early Intervention Program (EIP) and in 1999 joined Family Network on Disabilities of Florida, Inc. Today, Mr. Nurse works nationally as an advocate for quality transition planning, self-determination and increasing access to assistive technology for individuals with disabilities. Mr. Nurse and his partner Kevin Manning's firm help families plan for the future of their children or other dependents with special needs.

Presentations:
Financial and Social Independence *(Family Session)*
Break-Out Session: A Parent's Perspective (Mother) *(Family Session)*
Adulthood: Independence, Advocacy and Work *(BTHS Young Adult Session)*

Karen Helene Orstavik, MD, PhD — Professor Emerita, Consultant in Clinical Genetics, Faculty Division Rikshospitalet, University of Oslo, Norway

Dr. Orstavik's main interests have been rare congenital malformation syndromes and X chromosome inactivation. She graduated from Medical School at the University of Oslo in 1964 and trained as a specialist in Medical Genetics at Ulleval University Hospital in Oslo. She has been working as a clinical geneticist since 1972 and as a professor in clinical genetics for the last years of her career. Dr. Orstavik retired in 2007 but still has some duties as a professor emerita.

When Dr. Orstavik met a Norwegian family with Barth syndrome she was very interested in the X chromosomes of the carrier females. This led to the analysis of the X chromosome inactivation pattern of 16 carrier females from 6 families. Their special pattern of X inactivation may explain why female carriers of Barth syndrome seem to have no symptoms or signs of the disorder.

Presentation: The Asymptomatic Carrier—How the Gene that Causes Barth Syndrome Affects the Carrier's X Chromosome *(Family Session)*

John Reiss, PhD — Associate Professor, Department of Pediatrics, University of Florida; Director, Health Care Transition Initiative, Institute for Child Health Policy, University of Florida, Gainesville, FL, USA

Dr. Reiss' work has focused on facilitating collaborative action among public and private sector organizations at the federal, regional, and state levels and between families and professionals to improve the organization, financing and delivery of health care for children, youth and young adults with disabilities and chronic medical conditions; and to promote full partnership with families.

As part of his work on health care transition, Dr. Reiss has conducted research to better understand what families and youth experience as when they age out of pediatrics. Based on this research, he has developed a series of age-related informational booklets for youth/young adults, transition planning guides for families, and videos and web-based training materials for families and youth and professionals.

Presentations:
The Ongoing Process of Raising a Chronically Ill Child in Becoming a Self-Advocate
Break-Out Session: A Parent's Perspective (Father) *(Family Session)*
Presentation: My Last Name is Not Barth *(BTHS Youth/Young Adult and Sibling Sessions)*

Mindong Ren, PhD — Assistant Professor, Department of Cell Biology, New York University School of Medicine, New York, NY, USA

Dr. Ren's research interests include intracellular protein trafficking, membrane organelle biogenesis, the role of cardiolipin in mitochondria, and the pathogenic mechanism of Barth syndrome. Dr. Ren has been awarded two research grants from the Barth Syndrome Foundation titled: "Pathogenetic mechanism and genetic suppressors of Barth syndrome" (2006); and "A Drosophila Model of Barth Syndrome" (2004). His research on Barth syndrome has also been supported by the United Mitochondrial Diseases Foundation and NIH's National Heart, Lung and Blood Institute.

Dr. Ren holds a PhD in Molecular Cell Biology from the Sackler Institute of Graduate Biomedical Sciences, New York University School of Medicine.

Presentation: Suppressors of Tafazzin-deficiency Phenotype in Drosophila—Implications for Barth Syndrome *(Sci/Med Session)*

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

Dr. Reynolds completed her PhD in Health Related Science at Virginia Commonwealth University in 2007. Her research has focused on investigating physiologic stress reactivity patterns in children with Sensory Processing Disorder, and characterizing behavioral and physiological patterns of sensory processing in children with ADHD, Autism and Mood Disorders.

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

Presentation: Traffic Jams on the Sensory Highway—Considerations and Concerns about Sensory Integration *(Family Session)*

Amy Roberts, MD — Director, Cardiovascular Genetics Research Program; Associate in Cardiology, Children's Hospital Boston, Boston, MA; Principal Investigator, Barth Syndrome Registry and Biorepository, Boston, MA, USA

Dr. Roberts' lab will house the Barth Syndrome Foundation registry samples, and she works closely with Dr. Spencer and the Barth Syndrome Foundation in registry management and planning. She is the director of a department wide patient registry and DNA repository for children with heart disease and much of her research focuses on cardiac gene discovery. Since July 2004, she has worked at Children's Hospital Boston and provides clinical service in the Department of Cardiology's Cardiovascular Genetics Program

Dr. Roberts received her medical degree from Dartmouth Medical School, Hanover, NH. She trained in pediatrics at the University of Massachusetts Medical Center, and in clinical genetics in the Harvard Medical School Clinical Genetics Training Program.

Presentations:
Barth Syndrome Registry Update *(Sci/Med Session)*
The Barth Registry: Details and Benefits of Participation *(Family Session)*

Arwa Saidi, MD, BCh, FACC — Associate Professor, Department of Pediatrics, Congenital Heart Center at Shands Medical Plaza, Gainesville, FL, USA

Dr. Saidi's primary clinical and educational focus is transition and she is involved in educational programs for postgraduate students and pediatric residents. She is a physician trained in both internal medicine and pediatrics with subspecialty training in pediatric cardiology and congenital heart disease. Her training in internal medicine has allowed her to pursue a career in the care of adults with congenital heart disease.

Dr. Saidi serves on the Medical Advisory Board of the Adult Congenital Heart Association (she was formerly Chairman of the Transition group). She is also a member of the writing committee for the American Heart Association scientific statement on the recommendations for Preparing Adolescents with Congenital Heart Disease for Transition to Adulthood.

Presentation: Conversations and Considerations in Coordination of Continued Care *(Family Session)*

John Saroyan, MD, FAAP — Assistant Professor, Pediatric Pain Management and Palliative Care, Departments of Anesthesiology and Pediatrics, Columbia University College of Physicians and Surgeons, New York, NY, USA

Dr Saroyan's clinical pain management and palliative care practice includes inpatient and outpatient settings and his patients range in age from neonates to young adults. His research has included measuring pediatric pain management knowledge of resident trainees, communication of pain and symptoms in seriously ill children, and opioid prescribing practices of pediatric clinicians.

Presentation: Defining the Pain Experience in Barth Syndrome *(Family Session)*

Michael Schlame, MD — Associate Professor of Anesthesiology & Cell Biology, New York University School of Medicine; Attending Anesthesiologist, New York University Medical Center, New York, NY; Scientific and Medical Advisory Board, The Barth Syndrome Foundation, Inc.; Medical Advisory Board, Barth Syndrome Registry & Biorepository, Boston, MA, USA

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

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

Chair—Animal Models of Barth Syndrome *(Sci/Med Session)*

Genevieve Sparagna, PhD — Faculty Member, Department of Integrative Physiology, University of Colorado at Boulder, Boulder, CO; Pharmacology and Cardiology Departments, University of Colorado Denver, Denver, CO, USA

Dr. Sparagna's specialty is mitochondrial function and she has been doing research on cardiolipin for the past 12 years. Her work began in the laboratories of Dr. Jeanie McMillin and Dr. William Dowhan at the University of Texas Houston Health Science Center where she did her postdoctoral training. Dr. Sparagna has been awarded two research grants from the Barth Syndrome Foundation to study the alterations in remodeling of cardiolipin modified with changes in dietary lipids. ["Regulation of Cardiolipin Remodeling in the Heart Studied Using a Rat Model of Heart Failure" (2006); and "Fatty Acid Combinational Therapy for Barth Syndrome Investigated Using a Rat Model of Heart Failure" (2008)].

Dr. Sparagna holds a PhD in Biophysics from the University of Rochester. She received her undergraduate degree in Physics from Massachusetts Institute of Technology in Cambridge, MA.

Presentation: Dietary Influences on the Cardiolipin Composition of the Heart *(Sci/Med Session)*

Carolyn T. Spencer, MD — Assistant Professor Pediatrics, Department of Cardiology, Children's Hospital, Boston, MA; Principal Investigator, Barth Syndrome Registry and Biorepository, Boston, MA, USA

Dr. Spencer's clinical practice and teaching have focused on echocardiography, exercise testing and cardiomyopathy. She is currently on the cardiology faculty in the echocardiography laboratory at Children's Hospital Boston where she participates in performing, interpreting and teaching transthoracic and transesophageal echocardiography in all forms of congenital and acquired heart disease in the pediatric and adult congenital populations. Dr. Spencer supervises in the exercise laboratory and participates in the clinical care of patients with cardiomyopathy and congenital heart disease at Children's Hospital Boston. Additionally, she has initiated the Barth Syndrome Registry and Biorepository to further translational research in this area and to encourage collaboration among scientists interested in Barth syndrome.

Dr. Spencer has been awarded two research grants from the Barth Syndrome Foundation titled: "Cardiac and Skeletal Muscle in Barth Syndrome: Evaluation of Functional Capacity and Energy Metabolism" (2006); and "Cardiac Functional and Electrophysiological Abnormalities in Barth Syndrome" (2004).

Presentations:
Functional Capacity and Cardiomyopathy in Barth Syndrome *(Sci/Med Session)*
Barth Syndrome Registry Update *(Family Session)*
Overview of Cardiac Function with Barth Syndrome *(Family Session)*

Jay St. Amant, MS — Doctoral Candidate, Department of Clinical and Health Psychology, University of Florida, Gainesville, FL, USA

Mr. St. Amant has conducted and published empirical research on psychosocial factors in pediatric implantable cardioverter defibrillator (ICD) patients as well as patients with Barth syndrome. His broad research interests are in chronic illness populations across the lifespan.

Mr. St. Amant is pursuing his PhD in Clinical Psychology at the University of Florida. He received his Bachelor's degree from Mount Olive College in North Carolina and received his Master's degree from the University of Florida. In August, he will begin a one-year pre-doctoral internship at the U.S. Medical Center for Federal Prisoners in Springfield, Missouri. He will focus on medical and forensic psychology as he completes his doctorate.

Presentations:
Psychosocial Coping Mechanisms with Barth Syndrome *(Family Session)*
Break-Out Session: A Parent's Perspective (Father) *(Family Session)*
My Last Name is Not Barth *(BTHS Youth/Young Adult and Sibling Sessions)*

Colin G. Steward, FRCP, FRCPCH, PhD — Consultant in Bone Marrow Transplantation, Royal Hospital for Children; Reader in Stem Cell Transplantation, Department of Cellular & Molecular Medicine, School of Medical Sciences, University of Bristol, Bristol, England; Scientific and Medical Advisory Board, Barth Syndrome Foundation, Inc.; Advisory Board, Barth Syndrome Registry & Biorepository, Boston, MA, USA

Dr. Steward is a Consultant in bone marrow transplantation at the Bristol Royal Hospital for Children, UK. His particular interests are the patterns of neutropenia seen in Barth syndrome, optimising the use of antibiotics and granulocyte colony stimulating factor (G-CSF) in order to prevent infections, and fetal presentations of the disease. He has also campaigned for many years that Barth syndrome is seriously under-diagnosed. Dr. Steward has recently obtained permission from the National Health Service in England to provide a national specialist service for the disease, primary aims of which will be to increase recognition of the disease and to provide more consistent quality of care for patients whilst minimizing hospital visits. This service is being developed in close partnership with Michaela Damin and her colleagues at the Barth Syndrome Trust. It will provide holistic care for all of the major aspects of the disease as well as free diagnostic tests (in order to encourage clinicians to refer samples) and centralized prescribing and delivery of G-CSF.

Presentations:
Establishing a National Specialized Service for Barth Syndrome in the UK *(Sci/Med Session)*
Neutropenia and Barth Syndrome *(Family Session)*

Mark Tarnopolsky, MD, PhD, FRCP(C) — Clinical and Research Director, Corkins/Lammert Family Neuromuscular and Neurometabolic Clinic, McMaster University; Chair, McMaster Children's Hospital and Hamilton Health Sciences Foundation in Neuromuscular Diseases; Professor, Department of Pediatrics & Medicine, McMaster's Children's Hospital, Hamilton, Ontario, Canada

Dr. Tarnopolsky's research focuses on nutritional, exercise and pharmacological therapies for neurometabolic (primarily mitochondrial) and neuromuscular disorders, and aging. In addition, he studies the physiological and molecular aspects of mitochondrial adaptation to exercise, aging and the metabolic syndrome.

Dr. Tarnopolsky completed his PhD in cell biology and metabolism and residency training in Neurology and Physiatry. He is a fellow of the American Academy of Electrodiagnostic Medicine. He has received the Dr. David Green Award from the Muscular Dystrophy Association for Top Clinician Scientist in Canada (2005), the Barsky Lectureship for Excellence in Mitochondrial Medicine (2007), and the honour award for Canadian Society for Exercise Physiology (2008).

Presentation: Exercise as a Therapy for Mitochondrial Dysfunction – Implications for Barth Syndrome *(Sci/Med Session)*

Daniela Toniolo, PhD — Research Director, National Research Council of Italy (CNR), Institute of Genetics Biochemistry and Evolution, Pavia, Italy; Head of Unit, DIBIT-San Raffaele Research Institute, San Raffaele, Milan, Italy

Dr. Toniolo's focus is in human genetics, and her laboratory identified several genes responsible for genetic disorders. The first was the human G6PD, followed some years later by the genes for the Emery Dreifuss Muscular Dystrophies (X-linked, autosomal dominant and recessive), for Barth syndrome, and for one of the forms of X-linked Mental Retardation. Today she has several scientific interests. One is mental retardation through the study and characterization of mouse mutants. The second is the genetic dissection of disorders of ovulation and female fertility that she is studying by analysis of X chromosome candidate genes.

Dr. Toniolo held positions in the Italian National Research Council, first at the International Institute of Genetics and Biophysics in Naples, and later at the Institute of Genetics Biochemistry and Evolution in Pavia where she was Director of Research. In 2003, Dr. Toniolo moved to the DIBIT-San Raffaele Research Institute, where she is head of the Unit "Genetics of common disorders." She is a member of EMBO and HUGO, of the European, American and of the Italian Societies of Human Genetics.

Recipient: Varner Award for Pioneers in Science and Medicine

Matthew J. Toth, PhD — Science Director, Barth Syndrome Foundation, Inc.; *Ex-officio,* Scientific and Medical Advisory Board, The Barth Syndrome Foundation, Inc.; Advisory Board, Barth Syndrome Registry & Biorepository, Boston, MA, USA

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

Presentation: Update on Scientific Research *(Family Session)*

Bram J. van Raam, PhD — Postdoctoral Fellow, Sanford-Burnham Institute for Medical Research, La Jolla, CA, USA

Dr. van Raam's main interests are the interrelations of metabolic, cell death and inflammatory processes. His current research entails finding new proteolytic signaling pathways in novel cell death processes. Before moving to California, Dr. van Raam did his PhD research on the involvement of mitochondria in neutrophil apoptosis at the University of Amsterdam in the Netherlands, supervised by Drs. Taco W. Kuijpers and Dirk Roos. It was during this time that Dr. van Raam came to investigate mitochondrial involvement in Barth syndrome neutropenia. Dr. van Raam is a member of the International Proteolysis Society.

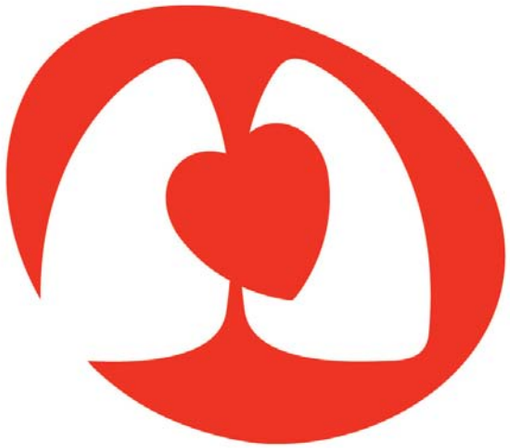
Presentation: Neutropenia in Barth Syndrome; On Calcium and Mitochondria *(Sci/Med Session)*

Frédéric M. Vaz, PhD — Clinical Chemist, Department of Clinical Chemistry and Pediatrics, Laboratory Genetic Metabolic Disease, Academic Medical Center, Amsterdam, The Netherlands

Dr. Vaz has focused primarily on research into Barth syndrome, funded by grants of the Barth Syndrome Foundation and the Princess Beatrix Foundation. He has led investigations into the cardiolipin abnormalities in Barth syndrome using tandem mass spectrometry and studied the function of tafazzin, the defective protein in this disorder. At this time, he is focusing on developing a new lipidomics platform to investigate lipid abnormalities in Barth syndrome and in other research areas. Dr. Vaz has been awarded two research grants by the Barth Syndrome Foundation titled: "Identification of the Proteins Interacting with Tafazzin and Resolution of the Consequences of the Deficiency of Cardiolipin at the Protein Level" (2006); and "Resolution of the Function of the TAZ-Gene and Characterization of its Gene Products" (2002).

Dr. Vaz obtained his PhD degree from the University of Amsterdam, under the supervision of Prof. Ronald Wanders, at the Laboratory of Genetic Metabolic Diseases (2002). He obtained his Chemistry MSc at the University of Utrecht (1997). In 2004, he became a Clinical Chemist in training at the Department of Clinical Chemistry at the Academic Medical Center but continued his research on Barth syndrome in collaboration with Dr. Willem Kulik.

Presentation: Sometimes the Small Things Make a Big Difference *(Family Session)*



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