



**Celebrating BSF's 10th Anniversary**

*...A decade of community*

*...A decade of education*

*...A decade of discovery*

**Barth Syndrome  
Foundation**

**Barth Syndrome**  
**5<sup>th</sup> International**  
**Scientific, Medical & Family Conference**

**July 26-31, 2010**

**Renaissance at SeaWorld  
Orlando, FL**



# SCIENTIFIC AND MEDICAL SESSIONS

## THURSDAY, JULY 29, 2010

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

# POSTER SESSION

## THURSDAY, JULY 29, 2010

(The Upper Deck)

<b>4:45 pm — 6:00 pm: Physicians &amp; Scientists</b>	<b>5:30 — 6:00 pm: Families Welcome</b>
<p><b>POSTER 1: CHARACTERIZATION OF AN SHRNA-MEDIATED TFAZZIN KNOCKDOWN MOUSE MODEL FOR BARTH SYNDROME</b></p> <p><u>MS Soustek</u>, DJ Falk, AS Lewin, BJ Byrne Department of Pediatrics, Department of Molecular Genetics and Microbiology, University of Florida, Gainesville, FL</p>	
<p><b>POSTER 2: INVESTIGATING BARTH SYNDROME: THE ROLE OF CARDIOLIPIN IN MAINTENANCE OF MITOCHONDRIAL MORPHOLOGY</b></p> <p>N Fei Department of Biological Sciences, Wayne State University, Detroit, MI</p>	
<p><b>POSTER 3: GENETIC AND VIRAL GENOME ANALYSIS OF CHILDHOOD CARDIOMYOPATHY: THE PCMR/PCSR EXPERIENCE</b></p> <p>JA Towbin, L Sleeper, JL Jefferies, S Colan, SA Webber, CE Canter, DT Hsu, <u>SM Ware</u>, JD Wilkinson, EJ Orav, SE Lipshultz Cincinnati Children's Medical Center, Cincinnati, OH, University of Miami, Miami, FL</p>	
<p><b>POSTER 4: IMPACT OF SAMPLE COLLECTION ON TFAZZIN MRNA VARIANTS</b></p> <p><u>SM Kirwin</u>; SE Swain; AL Manolakos; IL Gonzalez Nemours Biomedical Research Department; Alfred I. duPont Hospital for Children, Wilmington, DE</p>	
<p><b>POSTER 5: ROLE OF CARDIOLIPIN AND PHOSPHATIDYLETHANOLAMINE IN MITOCHONDRIAL FUSION</b></p> <p><u>AS Joshi</u>, ML Greenberg Department of Biological Sciences, Wayne State University, Detroit, MI</p>	
<p><b>POSTER 6: A REVERSED-PHASE LC-MS/MS CARDIOLIPIN METHOD FOR THE DIAGNOSIS OF BARTH SYNDROME SUITABLE FOR USE IN A ROUTINE METABOLIC LAB</b></p> <p><u>A Bowron</u><sup>1</sup>, R Frost<sup>2</sup>, S Heales<sup>3</sup>, C Steward<sup>4</sup> <sup>1</sup>Department of Clinical Biochemistry, Bristol Royal Infirmary, BS2 8HW, UK. <sup>2</sup>Waters Corporation, Elstree, UK. <sup>3</sup>Institute of Child Health, University College London, UK. <sup>4</sup>Bristol Royal Hospital for Children, UK</p>	
<p><b>POSTER 7: LIPIDOMIC AND BIOENERGETIC EFFECT OF CARDIAC-SPECIFIC OVEREXPRESSION OF CARDIOLIPIN SYNTHASE</b></p> <p><u>MA Kiebish</u>, 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</p>	
<p><b>POSTER 8: FETAL BARTH SYNDROME: A CASE OF PROGRESSIVE CARDIOMYOPATHY IN UTERO</b></p> <p><u>B Tsai-Goodman</u><sup>1</sup>, RP Martin<sup>1</sup>, CG Steward<sup>2</sup> Departments of Paediatric Cardiology<sup>1</sup> and Haematology<sup>2</sup>, Royal Hospital for Children, Bristol, England</p>	
<p><b>POSTER 9: FULL-LENGTH HUMAN TFAZZIN PROTECTS THE HUMAN CELLS FROM SERUM-WITHDRAWN APOPTOSIS</b></p> <p><u>Y Xu</u>, M Schlame Anesthesiology Department, NYUMC, New York City, NY</p>	
<p><b>POSTER 10: LOSS OF MITOCHONDRIAL ANIONIC PHOSPHOLIPIDS LEADS TO PERTURBATION OF MITOCHONDRIAL/CELLULAR IRON HOMEOSTASIS</b></p> <p><u>VA Patil</u>, VM Gohil, S Gupta, G Li, ML Greenberg Dept. of Biological Sciences, Wayne State University, Detroit, MI</p>	
<p><b>POSTER 11: BARTH SYNDROME IN A FEMALE PATIENT</b></p> <p>L Cosson<sup>1</sup>, A Toutain<sup>2</sup>, G Simard<sup>3</sup>, F Paoli<sup>4</sup>, W Kulik<sup>4</sup>, <u>FM Vaz</u><sup>4</sup>, H Blasco<sup>5</sup>, A Chantepie<sup>1</sup>, F Labarthe<sup>1,6</sup> <sup>1</sup>Dept of Paediatrics, CHU Tours, Tours, France, <sup>2</sup>Genetic, CHU Tours, Tours, France, <sup>3</sup>CHU Angers, INSERM U694, Angers, France, <sup>4</sup>Academic Medical Center, Amsterdam, Netherlands, <sup>5</sup>Biochemistry, CHU Tours, Tours, France, <sup>6</sup>INSERM U921, Tours, France</p>	

# SCIENTIFIC AND MEDICAL SESSIONS

FRIDAY, JULY 30, 2010

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

## SATURDAY, JULY 31, 2010

8:15 am — 11:30 am	<b>Scientific and Medical Advisory Board Meeting</b> ( <i>Veiltail Boardroom</i> )
4:10 pm — 5:30 pm	<b>Closing Ceremony</b> ( <i>Crystal Ballroom D &amp; E</i> )

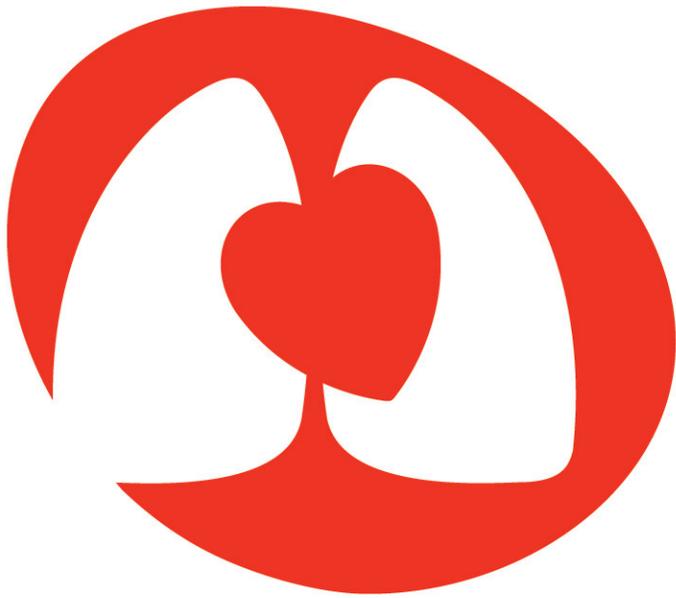


# Office of Rare Diseases Research

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National Institutes of Health

The Barth Syndrome Foundation is honored to acknowledge the Office of Rare Diseases Research's sponsorship in support of our 2010 International Scientific & Medical Conference.



# **National Heart Lung and Blood Institute**

People Science Health

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