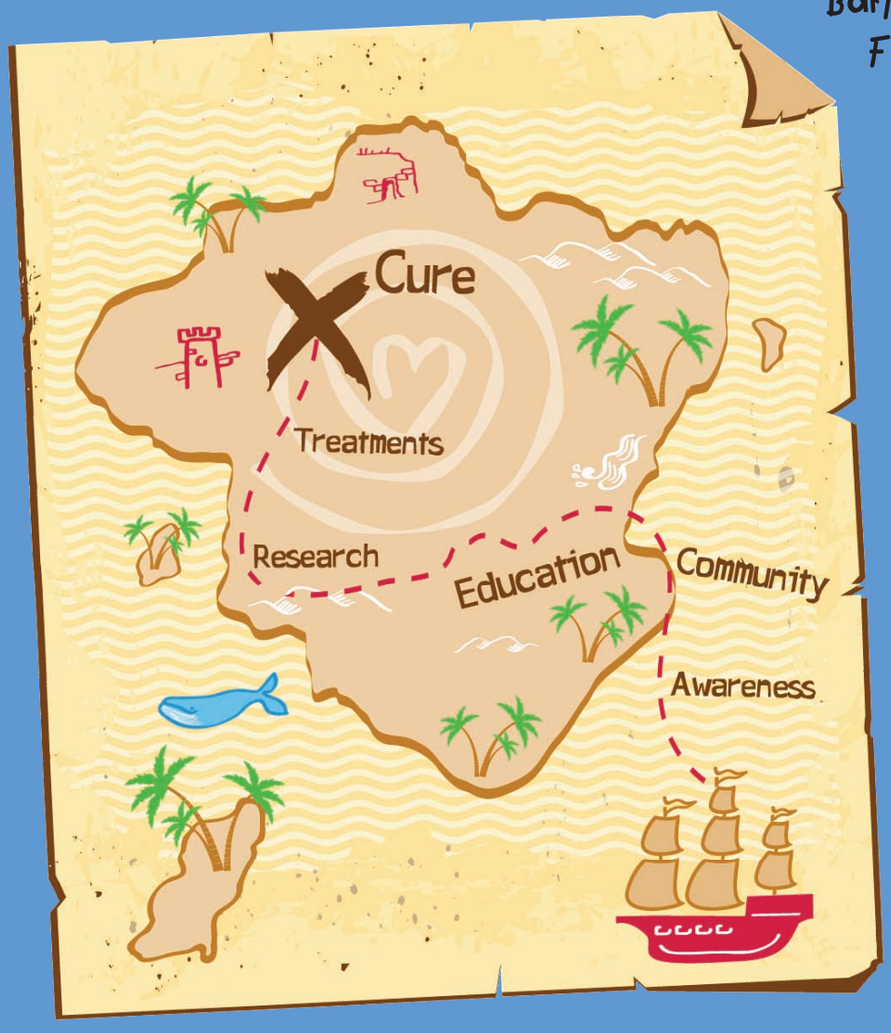
Barth Syndrome Foundation®



X Marks The Spot For A Cure!

Barth Syndrome

6th International Scientific, Medical & Family Conference

June 25 – 30, 2012
St. Pete Beach, FL

Ahoy there, Mateys!

We have quite a conference planned for you with the theme, ***"X Marks The Spot For A Cure!"*** As you read through the program, you will see that we have offered thought-provoking sessions, medical information, research updates, family support, exciting social activities, and more… all taking place at a breathtaking resort right on the beach!

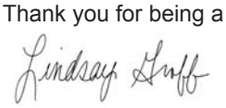
For those of you who have attended previously, you will see changes to the program that were made after reviewing previous conference evaluations and the pre-conference survey. Specifically, we designed the morning scientific and medical sessions to provide seasoned families with an in-depth examination of Barth syndrome. For first timers, be prepared for a rollercoaster ride of information and fun. Join us for roundtable discussions that will help shape the afternoon sessions. For research scientists and clinicians there is much new information, especially in the educational component and the posters, to facilitate ideas, discussion and collaboration. And of course, everyone will come together at meals to share daily living advice, exchange stories, and build lifelong friendships.

This conference continues to be a model among rare disorder organizations, bringing affected families, research scientists and clinicians together in one place at one time for a dual-track meeting. The Barth Syndrome Foundation (BSF) and our affiliates in Canada, France, South Africa, and the UK firmly believe that while each of these groups can make progress with their individual efforts, it is together, that we are much more powerful. For example:

- Doctors who might care for a single patient will have the opportunity to see over 40 at once, gaining a greater understanding of the breadth of the disorder. Additionally, they will be able to compare notes with other physicians and also to hear from scientific experts about the latest discoveries that shed light on the scientific underpinnings of the disorder.
- Scientists will hear presentations on colleagues' recent work as well as learn from, and have a chance to collaborate with, experts in other fields. Furthermore, they will gain insight into the clinical aspects of the disorder and witness the medical ramifications of what they discover in the laboratory.
- Families have a unique opportunity to learn from the world's greatest experts in Barth syndrome as well as those also contending with similar issues to yours. In addition to improving your knowledge and ability to care for your child(ren), you will be able to contribute directly to the search for a cure by providing data, health history information, and even your own tissue. Learning from experts and from each other provides you with the tools to better advocate for your child(ren).

We'll also be together for social activities with two informal "hang out" nights on Tuesday and Wednesday. The Friday Night Social offers an opportunity to dance the night away with old and new friends. Come dressed in pirate attire to celebrate with live band, Alibi! Enjoy the photo booth and Chinese auction. Don't forget your camera for this one!

Thank you for being a part of this voyage of hope, friendship and discovery.



Lindsay Groff
Executive Director

The Barth Syndrome Foundation would like to acknowledge the members of the 2012 Conference Committee and additional volunteers:

Steering Committee
Lindsay Groff, *Chair*
Tiffini Allen
Shelley Bowen
Lois Galbraith
Lynda Sedefian
Matthew Toth, PhD
John Wilkins
Sue Wilkins

Consultations Committee
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Kate McCurdy
Lorna Moore
Nigel Moore
Les Morris

PRE-CONFERENCE SESSIONS

SUNDAY, JUNE 24, 2012	
7:30 pm — 10:00 pm	**BSF BOARD OF DIRECTORS MEETING** *(Executive Board Room 4)*
MONDAY, JUNE 25, 2012	
7:30 am — 4:30 pm **All Day**	**REGISTRATION** *(Lobby Level Registration Booth)* **PORTRAITS BY AMANDA CLARK** *(Del Prado)* *For families who are arriving early*
2:00 pm — 3:00 pm	**NEW FAMILY ORIENTATION** *(Grand Ballroom)* Audience: First-time conference attendees *Led by Amie Hall, BTHS Parent, Ohio, USA*
3:00 pm — 4:00 pm	**CONSENT AND ASSENT SIGNING** *(Grand Ballroom)* *Led by Shelley Bowen, Director, BSF Family Services & Awareness* Note: The following groups of individuals participating in consultations must attend this session: **CONSENTS** Parents of all BTHS boys under the age of 18 Adult BTHS males 18 and older **ASSENTS** Minor BTHS individuals ages 12 – 18
4:00 pm — 7:00 pm	**Dinner on your own. All conference attendees have the option of of a 10% discount at any restaurant on-site.**
7:00 pm — 9:00 pm	**WELCOME EVENT** *(Grand Ballroom)* **7:00 pm — 8:00 pm** **FAMILY INTRODUCTIONS** (Children, Parents, Grandparents, etc.) **8:00 pm — 9:00 pm** **CONFERENCE 101 ~** All adults **8:00 pm — 10:00 pm** **LITTLE TYKE MOVIE NIGHT ~ Ages 0 – 4** *(South Terrace)* Note: One adult family member must accompany children who are not potty trained **8:00 pm — 10:00 pm** **TREASURE HUNT + OTHER ACTIVITIES ~ Ages 5+** *(North Terrace)*
TUESDAY, JUNE 26, 2012	
7:30 am — 4:30 pm	**REGISTRATION** *(Fifth Floor Foyer)*
4:30 pm — 7:00 pm	**Dinner on your own. All conference attendees have the option of a special discounted buffet at Sea Porch Restaurant.** *(No other discounts can be applied to these buffets.)*
7:00 pm — 10:00 pm	**SUNSET & STARGAZING** *(Pavillion West)* *Join us for a fun evening on the beach and enjoy the beautiful sunset and stargazing!* *Note — Children must be accompanied by an adult*
WEDNESDAY, JUNE 27, 2012	
7:30 am — 4:30 pm	**REGISTRATION** *(Fifth Floor Foyer)*
4:30 pm — 7:00 pm	**Dinner on your own. All conference attendees have the option of a special discounted buffet at Sea Porch Restaurant.** *(No other discounts can be applied to these buffets.)*
7:00 pm — 10:00 pm	**POOLSIDE GATHERING** *(South Pool ~ Main Hotel)* *Enjoy an evening of fun by the pool getting reacquainted with old friends as well as meeting new ones. DJ Camo will be in the house to entertain us!* *Note — Children must be accompanied by an adult*

BARTH SYNDROME CONSULTATIONS

TUESDAY & WEDNESDAY

JUNE 26 — 27, 2012

8:00 am — 5:00 pm

(All families will need to check-in for consultations in King Charles)

Join us for two days of consultations 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 offers opportunities to explore new ideas and discuss common themes. Data collected during these consultations will feed the Barth Syndrome Registry & Repository (BRR) to further the understanding of this disorder. Our consultations have been hailed as a model approach by other health advocacy groups.

There will be arts, crafts, movies, outdoor games and Wii activities for all

CONSULTATIONS	
Genetics *(South Terrace)* *Iris L. Gonzalez, PhD* *Rebecca L. (Kern) McClellan, MGC, CGC*	**Nutrition/Metabolism** *(South Terrace)* *Richard I. Kelley, MD, PhD*
RESEARCH	
Medical Database & Repository (BRR) *(Executive Board Rooms)* Vanessa Rangel Miller, VP Genetic Services	**Practical care of neutropenia in Barth syndrome** *(South Terrace)* Audrey Anna Bolyard, RN *(virtual sessions)*
Nutrition/Metabolism *(Executive Board Rooms)* *Richard I. Kelley, MD, PhD* *Hilary Vernon, MD, PhD*	**Vitals** *(King Charles)* *Linda Croxton, ARNP, RN* *Shelia Mann* *Jarrod Robertson* *Donna Strain, RN* *Sue Wilkins, RN*
Sensory Processing *(South Terrace)* **Comprehensive assessment of the impact of illness and disability in boys with Barth syndrome: Sensory, nutritional, quality of life, and family impact considerations for boys with Barth syndrome** *Roxanna M. Bendixen, MS, MA (Principal Investigator)* *YuYun Huang, MS, OT* *Consuelo M. Kreider, MHS OTR/L* *Yoonjeong Lim, MS, OT* *Stacey Reynolds, PhD, OTR/L*	

BSF would like to thank Phlebotomists Cecelia Strevel and Tracy Whatley for donating their time and services in support of the consultations. Blood draws will take place in *Buena Vista.*

Olivia and Noah (Photo courtesy of Stephanie Rader ~ 2012)

STEPHANIE, NOAH, OLIVIA and CAROL RADER

are the proud sponsors of the

SUNSET & STARGAZING

Tuesday, June 26, 2012

Join us for a fun evening on the beach and enjoy the beautiful sunset and stargazing!

SMALL GROUP MEETINGS TUESDAY, JUNE 26th	
9:00 am — 10:00 am	**Financial independence and transitioning into adulthood** *(Executive Board Room)* *Tom Nurse, Partner and Investment Advisor Representative*
10:00 am — 11:00 am	**Barth syndrome 101** *(Executive Board Room)* *Kate McCurdy (BSF Board Member)* *Rebecca (Kern) McClellan, MGC, CGC*
11:00 am — 12:00 pm **4:00 pm — 5:00 pm**	**Nutrition/Metabolism** *(Executive Board Room)* *Richard I. Kelley, MD, PhD*
1:00 pm — 3:00 pm	**Cardiopulmonary resuscitation (CPR)** *(North Terrace)* *Sunstar Emergency Medical Services of Pinellas County*
3:00 pm — 4:00 pm	**Future generations of Barth syndrome — Carrier concerns** *(Executive Board Room)* *Susan McCormack (BSF Board Member)* *Rebecca (Kern) McClellan, MGC, CGC*

SMALL GROUP MEETINGS WEDNESDAY, JUNE 27th	
8:00 am — 5:00 pm *(1-hour groups)*	**Barth Syndrome Registry & Repository (BRR)** *(Executive Board Room)* *Vanessa Rangel Miller, VP Genetic Services*
9:00 am — 3:00 pm *(1-hour groups)*	**Hematology and Barth syndrome** *(Executive Board Room)* *Colin G. Steward, PhD, FRCP, FRCPCH*
10:00 am — 10:45 am	**Working with the press to promote awareness about Barth syndrome** *(Executive Board Room)* *BSF Facilitators: Susan Osnos (BSF Board Member), Tiffini Allen, Kristi Pena (BTHS Parents)*
11:00 am — 12:00 pm **4:00 pm — 5:00 pm**	**Nutrition/Metabolism** *(Executive Board Room)* *Richard I. Kelley, MD, PhD*
1:00 pm — 3:00 pm	**Cardiopulmonary resuscitation (CPR)** *(North Terrace)* *Sunstar Emergency Medical Services of Pinellas County*
2:00 pm — 2:45 pm	**Heart transplants 101** *(North Terrace)* *Maryanne Chrisant, MD*
3:00 pm — 4:00 pm	**Routine tests and procedures in cardiology** *(North Terrace)* *Maryanne Chrisant, MD*
4:00 pm — 4:30 pm	**Exercise intolerance and Barth syndrome** (BTHS Individuals Ages 18+) *(North Terrace)* *W. Todd Cade, PT, PhD*
4:40 pm — 5:10 pm	**Exercise intolerance and Barth syndrome** (BTHS Individuals Ages 12 – 17) *(North Terrace)* *W. Todd Cade, PT, PhD*
5:20 pm — 5:50 pm	**Exercise intolerance and Barth syndrome** (BTHS Individuals Ages 8 – 11) *(North Terrace)* *W. Todd Cade, PT, PhD*
Note: Amanda Clark will be photographing families on Monday, Tuesday and Wednesday *(Del Prado)*	

Photo courtesy of
Dr. Michael Schlame ~ 2012

MICHAEL SCHLAME, MD

is the proud sponsor of the

POOLSIDE GATHERING

Wednesday, June 27, 2012

Enjoy an evening of fun by the pool getting reacquainted with old friends as well as meeting new ones.
DJ Camo will play your favorite dance music throughout the evening!

Dilen and Ronly (Photo courtesy of Susan McCormack ~ 2012)

SUSAN McCORMACK & KEN MARRA

are the proud sponsors of the

LODGING ACCOMMODATIONS FOR PHOTOGRAPHER AMANDA CLARK

Barth Syndrome Foundation of Canada

BARTH SYNDROME FOUNDATION OF CANADA

is the proud sponsor of the

LITTLE TYKE MOVIE NIGHT
&
TREASURE HUNT/OTHER ACTIVITIES

Monday, June 25, 2012

Photo courtesy of Dr. Greenberg ~ 2012

IN HONOR OF REBECCA GREENBERG'S GRADUATION FROM SKYLINE HIGH SCHOOL

MIRIAM GREENBERG & SHIFRA EPSTEIN

are the proud sponsors of the

CONFERENCE LANYARDS

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The Buddemeyer boys.
(Photo courtesy of Randy Buddemeyer ~ 2012)

Wishing everyone a successful conference!

THE BUDDEMEYER BOYS

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PRINTING OF CONFERENCE PROGRAM

THE SERNEL FAMILY

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SPEAKER THANK YOU GIFTS

OVERVIEW OF SCIENTIFIC & MEDICAL SESSIONS *(North Terrace Meeting Room)*		OVERVIEW OF FAMILY SESSIONS *(South Terrace Meeting Room)*	
THURSDAY, JUNE 28, 2012			
7:30 am — 8:15 am	**Breakfast** *(Grandroom Ballroom)*		
8:30 am — 12:00 pm	**Clinical aspects of Barth syndrome** *Chair: Richard I. Kelley, MD, PhD*		
8:30 am — 9:30 am	*Richard I. Kelley, MD, PhD*		
9:30 am — 10:30 am	*Colin Steward, PhD, FRCP, FRCPCH*	**10:25 am**	**Welcome** *Shelley Bowen, Director, BSF Family Services & Awareness*
10:30 am — 11:00 am	**Break**	**10:30 am — 11:15 am**	**Cardiology Roundtable Discussion** BTHS Male: *Aldo, Age 19* BTHS Parent: *Ned Kalapasev* BSF Facilitator: *Shelley Bowen*
11:00 am — 11:45 am	*W. Todd Cade, PT, PhD*		
11:45 am — 12:00 pm	Brainstorming About Future Directions	**11:15 am — 12:00 pm**	**Hematology Roundtable Discussion** BTHS Male: *Will, Age 26* BTHS Parent: *Tiffini Allen* BSF Facilitator: *Shelley Bowen*
12:00 pm — 1:30 pm	**Lunch ~ Keynote Speaker** *(Grand Ballroom)* **Group Photo** *(Location TBD)*		
1:30 pm — 4:30 pm	**Biological function of *tafazzin* & cardiolipin** *Chair: Grant Hatch, PhD*	**1:45 pm — 3:45 pm**	**Cardiac aspects of Barth syndrome**
1:30 pm — 2:00 pm	*Steven M. Claypool, PhD*	**1:45 pm — 1:55 pm**	**Speaker Introduction:** *Stephanie Rader, RN, BSN, MSN, NNP-BC*
2:00 pm — 2:30 pm	*Miriam L. Greenberg, PhD*	**1:55 pm — 2:20 pm**	*Gonzalo Wallis, MD*
2:30 pm — 3:00 pm	*Frédéric M. Vaz, PhD*	**2:20 pm — 2:45 pm**	*Randall Bryant, MD*
3:00 pm — 3:15 pm	**Break**	**2:45 pm — 3:10 pm**	*W. Todd Cade, PT, PhD*
3:15 pm — 3:45 pm	*Michael Schlame, MD*	**3:10 pm — 3:45 pm**	**Q & A**
3:45 pm — 4:15 pm	*Christopher McMaster, PhD*	**3:45 pm — 4:00 pm**	**Break**
4:15 pm — 4:30 pm	Brainstorming About Future Directions	**4:00 pm — 5:25 pm**	**Managing neutropenia in Barth syndrome**
		4:00 pm — 4:10 pm	Speaker Introduction: *Suzy Green*
		4:10 pm — 4:35 pm	*David C. Dale, MD*
4:30 pm — 5:00 pm	**Break** *(Dinner on own. All conference attendees have the option of a 10% discount at any restaurant on-site.)*	**4:35 pm — 5:00 pm**	*Colin G. Steward, PhD, FRCP, FRCPCH*
5:00 pm — 7:00 pm	**Poster Session** *(Fifth Floor Foyer)*	**5: 00 pm — 5:25 pm**	**Q & A**
		5: 25 pm — 6:00 pm	**Break** *(Dinner on own. All conference attendees have the option of a 10% discount at any restaurant on-site.)*
		6:00 pm — 7:00 pm	**Poster Session** *(Fifth Floor Foyer)*

CHILDCARE — Kid's Nite Out Recreation & Resort Management, Inc. *(Buena Vista)*

All children under the age of eight (8) that need childcare should be registered. It is required that a parent or guardian register their child, as well as check-in and check-out.

Daily registration: Thursday, Friday (June 28th, 29th) @ 8:15 am
Saturday (June 30th) registration @ 10:15 am

OVERVIEW OF SCIENTIFIC & MEDICAL SESSIONS *(North Terrace Meeting Room)*		OVERVIEW OF FAMILY SESSIONS *(South Terrace Meeting Room)*	
FRIDAY, JUNE 29, 2012			
7:30 am — 8:15 am	**Breakfast** *(Grand Ballroom)*		
8:30 am —12:00 pm	**Mouse model of Barth syndrome** *Chair: Michael Schlame, MD*		
8:30 am — 9:00 am	*John L. Jefferies, MD, MPH*		
9:00 am — 9:30 am	*Colin Phoon, MPhil, MD*		
9:30 am — 10:00 am	*Meghan Soustek*		
10:00 am — 10:15 am	**Break**		
10:15 am — 10:45 am	*Zaza Khuchua, PhD*	**10:15 am** **10:15 am — 11:00 am**	**Speaker Introduction:** Shelley Bowen **Heart and skeletal muscle metabolism, energy production and function in Barth syndrome — Recruiting for a new NIH-funded study** *W. Todd Cade, PT, PhD*
10:45 am — 11:15 am	*Michael Kiebish, PhD*	**11:00 am — 11:15 am**	**Break**
11:15 am — 11:45 am	*Adam J. Chicco, PhD*	**11:15 am — 12:00 pm**	**Nutrition Roundtable Discussion** BTHS Male: *Kevin, Age 23* BTHS Parent: *Brie Chandler* BTHS Parent: *Donna Strain, RN* BSF Facilitator: *Shelley Bowen*
11:45 am — 12:00 pm	**Brainstorming About Future Directions**		
12:00 pm — 1:30 pm	**Varner Award Luncheon** *(Grand Ballroom)*		
1:30 pm — 5:00 pm	**Mitochondrial Physiology of Barth Syndrome** *Chair: Grant Hatch, PhD*		
1:30 pm — 2:00 pm	2 Poster Session presentations	**1:45 pm — 1:55 pm** **1:55 pm — 2:40 pm**	**Speaker Introduction**: Florence Mannes **Metabolism of Barth syndrome** *Richard I. Kelley, MD, PhD*
2:00 pm — 2:30 pm	*Mindong Ren, PhD*		
2:30 pm — 3:00 pm	*Grant Hatch, PhD*	**2:40 pm — 2:55 pm**	**Q & A**
3:00 pm — 3:15 pm	**Break**	**2:55 pm — 3:10 pm**	**Break**
3:15 pm — 3:45 pm	*William Pu, MD*	**3:10 pm — 4:30 pm**	**Speaker Introduction**: *Ryan, Age 21* **Update on research in Barth syndrome** *Matthew J. Toth, PhD* *Michael Schlame, MD* **Q & A**
3:45 pm — 4:15 pm	*Anton I. De Kroon, PhD*		
4:15 pm — 4:45 pm	*Yuguang Shi, PhD*		
4:45 pm — 5:15 pm	**Brainstorming & Conference Wrap-Up**	**4:30 pm — 7:00 pm**	**Break** *(Dinner on own. All conference attendees have the option of a 10% discount at any restaurant on-site.)*
7:00 pm — 11:00 pm	**Evening Social ~ *X Marks The Spot For A Cure!*** *(Grand Ballroom)* *Join us for a pirate-themed evening; live entertainment featuring Alibi; delicious appetizers, cash bar; all conference attendees are invited!*		
Scientific & Medical Sessions *(North Terrace Meeting Room)*		**Family Sessions** *(South Terrace Meeting Room)*	
SATURDAY, JUNE 30, 2012			
9:00 am — 10:30 am	**Breakfast is on your own and coffee is on us!** *(Fifth Floor Foyer)*		
8:30 am — 12:00 pm	**Scientific & Medical Advisory Board Breakfast & Meeting**	**10:30 am — 12:15 pm**	**Mom/Dad Breakouts**
		12:15 pm — 12: 30 pm	**Break**
12:30 — 1:30 pm	**BSF w/ Burgers/Shakes/Fries** *(Grand Ballroom)*		
1:30 pm — 2:30 pm	**Conference Finale** *(Grand Ballroom)* *All conference attendees are invited to attend!*		

OVERVIEW OF AFFECTED YOUTH SESSIONS			OVERVIEW OF SIBLING SESSIONS		
WEDNESDAY, JUNE 27, 2012					
Daily check-in located in King Charles **Affected Youth Sessions** *(Executive Board Room)*					
4:00 pm — 4:30 pm	**Ages 18+: Exercise intolerance and Barth syndrome** ~ *W. Todd Cade, PT, PhD*				
4:40 pm — 5:10 pm	**Ages 12 – 17: Exercise intolerance and Barth syndrome** ~ *W. Todd Cade, PT, PhD*				
5:20 pm — 5:50 pm	**Ages 8 – 11: Exercise intolerance and Barth syndrome** ~ *W. Todd Cade, PT, PhD*				
THURSDAY, JUNE 28, 2012					
7:30 am — 8:15 am	Breakfast *(Grand Ballroom)*				
9:00 am —10:00 am	**Meet your Group Leader ~ All Affected Individuals and Siblings** *(King Charles)*				
10:00 am — 11:00 am					
Ages 8 - 15	**Ages 16+**		**Ages 8 - 11**	**Ages 12 - 17**	**Ages 18+**
Pavillion West	*Executive Board Room*		*King Charles*		
Ping Pong	**Metabolism of Barth syndrome** *Richard I. Kelley, MD, PhD*		**Photography fun with Amanda Clark**		
11:00 am — 12:00 pm					
Ages 8 - 11	**Ages 12 - 17**	**Ages 18+**	**Ages 8 - 11**	**Ages 12 - 17**	**Ages 18+**
King Charles			*King Charles*		
Insights from the Insiders Facilitator: *BJ Develle*			**I'm affected too** Facilitators: *Michal Weinberger, MSW; Eliza McCurdy*		
12:00 pm — 1:30 pm	**Luncheon ~ Keynote Speaker** *(Grand Ballroom)* **Group Photo** *(Location TBD)*				
1:30 pm — 1:45 pm	**Break**				
1:45 pm — 3:00 pm					
Ages 8 - 11	**Ages 12 - 17**	**Ages 18+**	**Ages 8 - 11**	**Ages 12 - 17**	**Ages 18+**
King Charles			*King Charles*		
Photography fun with Amanda Clark			**Activity Time**		
3:00 pm — 5:00 pm	**Activity Time ~ All Affected Individuals and Siblings** *(King Charles)* Activities include cupcake making, caricatures, centerpiece making, donor thank you cards, ping pong, scrapbooking, Wii games, and other awesome activities				

John, age 2

IN HONOR OF JOHN WILKINS's 30th BIRTHDAY

MIKE & SUE WILKINS

are the proud sponsors of the

REFRESHMENT BREAK

Friday, June 29, 2012

John, age 30, and his niece, Anna, age 1

(Photos courtesy of Sue Wilkins)

OVERVIEW OF AFFECTED YOUTH SESSIONS	OVERVIEW OF SIBLING SESSIONS

FRIDAY, JUNE 29, 2012

7:30 am — 8:15 am	**Breakfast** *(Grand Ballroom)*
9:00 am — 9:45 am	**All Affected Individuals and Siblings** *(King Charles)* **Hang Out/Discuss Day's Activities**
9:45 am — 12:00 pm	**All Affected Individuals and Siblings** **Shelling/Sand Castle Building** *Note: Please remember to bring beach towel, sunscreen and change of clothes* *(Everyone will meet in King Charles to venture off to Main Hotel Beach)*
12:00 pm — 1:30 pm	**Varner Award Luncheon** *(Grand Ballroom)* **Youth Pizza Luncheon ~ All affected individuals and siblings ages 8+** *(Pavilion West)*
1:30 pm — 1:45 pm	**Break**

1:45 pm — 3:00 pm

Ages 8 - 15	Ages 16+	Ages 8 - 11	Ages 12 - 17	Ages 18+
King Charles	*Executive Board Room*	*King Charles*		
Movie & Games	**My family, me and Barth syndrome** *Vanessa Garratt, DClinPsych*	**Movie & Games**		

3:00 pm — 4:30 pm

Ages 8 - 11	Ages 12 - 17	Ages 18+	Ages 8 - 15	Ages 16+
King Charles			*King Charles*	*Executive Board Room*
Movie & Games			**Movie & Games**	**My family, me and Barth syndrome** *Vanessa Garratt, DClinPsych*

4:00 pm — 7:00 pm	**Break** Don't forget to grab a bite to eat so that you have the energy to dance the night away at our Social Event! All conference attendees have the option of a 10% discount at any restaurant on-site.
7:00 pm — 11:00 pm	**Evening Social ~ *X Marks The Spot For A Cure!*** *(Grand Ballroom)* *Join us for a pirate-themed evening; live entertainment featuring Alibi; delicious appetizers, cash bar; all conference attendees are invited!*

SATURDAY, JUNE 30, 2012

Breakfast is on your own.

10:00 am — 11:00 am | **10:00 am — 11:00 am**

Affected Youth Sessions			Sibling Sessions		
Ages 8 - 11	**Ages 12 - 17**	**Ages 18+**	**Ages 8 - 11**	**Ages 12 - 17**	**Ages 18+**
King Charles	*Executive Board Room*				
Finale Project	**Barth syndrome and beyond** *Randall Bryant, MD*	**Finale Project**			

11:00 am — 12:30 pm | **11:00 am — 12:30 pm**

Ages 8 - 11	Ages 12 - 17	Ages 18+	Ages 8 - 11	Ages 12 - 17	Ages 18+
King Charles		*Executive Board Room*	*King Charles*		
Finale Project		**Barth syndrome and beyond** *Randall Bryant, MD*	**Finale Project**		

12:30 pm — 1:30 pm	**Lunch ~ BSF w/ Burgers/Shakes/Fries** *(Grand Ballroom)*
1:30 pm — 2:30 pm	**Conference Finale ~ *All conference attendees are invited to attend!*** *(Grand Ballroom)*

KEYNOTE ADDRESS

A Globalization of Rare Diseases Research Activities

Stephen C. Groft, Pharm D

Thursday, June 28, 2012
12:00 pm — 1:30 pm
(Grand Ballroom)

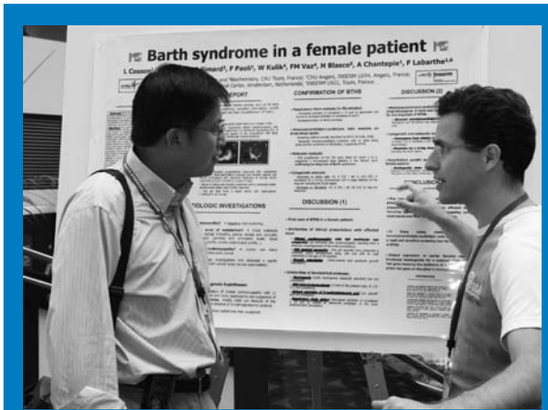
Introduction by Katherine McCurdy, BSF Board Member

Stephen C. Groft, Pharm D, Director of the Office of Rare Disease Research (ORDR), National Institutes of Health (NIH), Bethesda, MD, USA

Dr. Groft is the Director of the Office of Rare Diseases Research (ORDR) at the National Institutes of Health (NIH) that is now a part of the National Center for Advancing Translational Sciences (NCATS). His major focus is on stimulating research with rare diseases and developing information about rare diseases and conditions for health care providers and the public. To help identify research opportunities and establish research priorities, the Office has co-sponsored over 1,100 rare diseases-related scientific conferences with the NIH research Institutes and Centers. Current activities include establishing patient registries for rare diseases, developing an inventory of available bio-specimens from existing biorepositories, developing an educational module on rare diseases for middle school children, establishing a public information center on genetic and rare diseases, developing an international rare diseases research consortium, maintaining the Rare Diseases Clinical Research Network, and providing a special emphasis clinic with senior clinical staff for patients with undiagnosed diseases at NIH's Clinical Research Center Hospital.

Dr. Groft served in 1991-1992 as the first Acting Director of the Office of Alternative Medicine at the NIH (now the National Center for Complementary and Alternative Medicine) and the Staff Director for the White House Commission on Complementary and Alternative Medicine Policy from 2000-2002. Dr. Groft received the B.S. degree in Pharmacy in 1968 and the Doctor of Pharmacy degree from Duquesne University in 1979.

(Photo courtesy of Dr. Stephen Groft ~ 2012)



POSTER SESSION

Thursday, June 28, 2012
5:00 pm — 7:00 pm
(Fifth Floor Foyer)

5:00 pm — 7:00 pm	Open to Science & Medicine conference attendees
6:00 pm — 7:00 pm	Open to Family conference attendees

(Photo courtesy of BSF ~ 2010)

PIONEER IN SCIENCE AND MEDICINE VARNER AWARD PRESENTATION

Friday, June 29, 2012
12:00 pm — 1:30 pm
(Grand Ballroom)

In honor of Paula and Woody Varner, the Board of Directors of the Barth Syndrome Foundation has created the Varner Award for Pioneers in Science and Medicine, to be presented at our biennial BSF conferences. This award is to be given to a scientist or physician whose dedication to work in his or her field has made a positive and lasting impact on Barth syndrome.

Previous awardees include the following pioneers: Peter Vreken, PhD and Daniela Toniolo, PhD (2010); and Richard I. Kelley, MD, PhD and Peter J. Barth, MD, PhD (2008).

SCIENTIFIC AND MEDICAL SESSIONS

THURSDAY, JUNE 28, 2012

(North Terrace Meeting Room)

7:30 am — 8:15 am	Breakfast *(Grand Ballroom)*
CLINICAL ASPECTS & EPIDEMIOLOGY OF BARTH SYNDROME *Chair — Richard I. Kelley, MD, PhD, Johns Hopkins University School of Medicine; Kennedy Krieger Institute, Baltimore, MD*	
8:25 am — 12:00 pm **8:25 am**	**Introduction** — *Adam, Ontario, Canada, Age 22*
8:30 am — 9:30 am	**Abnormalities of intermediary metabolism in Barth syndrome** *Richard Kelley, MD, PhD, Kennedy Krieger Institute, Baltimore, MD*
9:30 am — 10:30 am	**The first two years of the UK national service for Barth syndrome: Triumphs and tribulations** *Colin Steward, PhD, FRCP, FRCPCH, Bristol Royal Hospital for Children, Bristol, UK*
10:30 am — 11:00 am	**Break**
11:00 am — 11:45 am	**Exercise and substrate metabolism studies in Barth syndrome: Updates and future direction** *W. Todd Cade, PT, PhD, Washington University School of Medicine, St. Louis, MO*
11:45 am — 12:00 pm	**Brainstorming about future directions** *Richard Kelley, MD, PhD, Kennedy Krieger Institute, Baltimore, MD*
12:00 pm — 1:30 pm	**Keynote Speaker / Luncheon Buffet** *(Grand Ballroom)* *Stephen C. Groft, Pharm D, Director, Office of Rare Disease Research (ORDR), National Institutes of Health (NIH), Bethesda, MD, USA* **"A globalization of rare diseases research activities"** **Group Photo** *(Location TBD)*
BIOLOGICAL FUNCTION OF *TAFAZZIN* AND CARDIOLIPIN *Chair — Grant Hatch, PhD, University of Manitoba, Winnipeg, Canada*	
1:25 pm — 5:00 pm **1:25 pm**	**Introduction** — *Benjamin, Tennessee, USA, Age 15*
1:30 pm — 2:00 pm	**The topology of cardiolipin remodeling in yeast** *Steven M.Claypool, MA, PhD, Johns Hopkins University, Baltimore, MD*
2:00 pm — 2:30 pm	**Loss of cardiolipin leads to perturbation of mitochondrial and cellular iron homeostasis** *Miriam L. Greenberg, PhD, Wayne State University, Detroit, MI*
2:30 pm — 3:00 pm	**Application of lipidomics to identify new phospholipid disorders** *Frédéric M. Vaz, PhD, Academic Medical Center, Amsterdam, The Netherlands*
3:00 pm — 3:15 pm	**Break**
3:15 pm — 3:45 pm	**The mechanism of *tafazzin*** *Michael Schlame, MD, New York University, New York, NY*
3:45 pm — 4:15 pm	**Yeast genome-wide screens to assess the genetic landscape for Barth syndrome** *Christopher McMaster, PhD, Dalhousie University, Halifax, Canada*
4:15 pm — 4:30 pm	**Brainstorming about future directions** *Grant Hatch, PhD, University of Manitoba, Winnipeg, Canada*
4:30 pm — 5:00 pm	**Break** (Dinner on your own. All conference attendees have the option of a 10% discount at any restaurant on-site.)
5:00 pm — 7:00 pm **6:00 pm — 7:00 pm**	**Poster Session** Scientific and Medical Community *(Fifth Floor Foyer)* Families are invited to join the Poster Session *(Fifth Floor Foyer)*

FRIENDS OF HENRY

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REFRESHMENT BREAK

Thursday, June 28, 2012

(Photo courtesy of Tiffini Allen ~ 2012)

POSTER SESSION

THURSDAY, JUNE 28, 2012

(Fifth Floor Foyer)

5:00 pm — 7:00 pm: Physicians & Scientists | **6:00 pm — 7:00 pm**: Families Welcome

POSTER 1: Lipidomics of intact mitochondria by MALDI-TOF/MS
Roberto Angelini[1], Rita Vitale[2], Vinay A. Patil[3], Tiziana Cocco[1], Bernd Ludwig[4], Miriam L. Greenberg[3], Angela Corcelli[1,5]
[1]Department of Basic Medical Sciences, University of Bari "Aldo Moro", Bari, Italy; [2]IMM-CNR, Institute for Microelectronics and Microsystems, National Research Council, Lecce, Italy; [3]Department of Biological Sciences, Wayne State University, Detroit, Michigan, USA; [4]Biocenter, Institute of Biochemistry, Goethe University, Frankfurt, Germany; [5] IPCF-CNR, Institute for Chemical-Physical Processes, National Research Council, Bari, Italy

POSTER 2: Barth syndrome with normal cardiolipin concentrations and without neutropenia
Ann Bowron[1], Julie Honeychurch[2], Melanie Pennock[2], Maggie Williams[2], Beverley Tsai-Goodman[3], Graham Shortland[4], Colin Steward[5]
[1]Department of Clinical Biochemistry, Bristol Royal Infirmary; [2]Bristol Genetics Laboratory, North Bristol NHS Trust; [3]Department of Cardiology, Bristol Royal Hospital for Children; [4]Department of Metabolic Disease, University Hospital Wales, Cardiff; [5]Department of Stem Cell Transplantation, Bristol Royal Hospital for Children, Bristol, United Kingdom

POSTER 3: Introduction of a national service for diagnosis of Barth syndrome: A progress report
Ann Bowron[1], Paul Thomas1, Colin Steward[2]
[1]Department of Clinical Biochemistry, Bristol Royal Infirmary; [2]Department of Stem Cell Transplantation, Bristol Royal Hospital for Children, Bristol, United Kingdom

POSTER 4: Vitamin A and Barth syndrome: Is supplementation required?
Nicol Clayton (Specialist Paediatric Dietitian), Ann Bowron (Principal Clinical Biochemist), Colin Steward (Clinical Lead), NHS Specialised Services, Bristol Royal Hospital for Children, Bristol, United Kingdom

POSTER 5: Age-associated compositional changes in cardiolipin and monolysocardiolipin
Junhwan Kim, Paul E. Minkler, and Charles L. Hoppel
Center for Mitochondrial Disease, Department of Pharmacology/Medicine, Case Western Reserve University, Cleveland, OH, USA

POSTER 6: Sensory processing in Boys with Barth syndrome
Consuelo Kreider, Stacey Reynolds, Roxanna Bendixen, Occupational Therapy, University of Florida, Gainesville, FL, USA

POSTER 7: Quality of life in boys with Barth syndrome: Comparison of child self-reports and parent proxy-reports
Yoonjeong Lim, Consuelo Kreider, Stacey Reynolds, Roxanna Bendixen, Occupational Therapy, University of Florida, Gainesville, FL, USA

POSTER 8: Toward plasma amino acid profiling of Barth syndrome
Chris Ottolenghi[1,2,3], Bernadette Chadefaux-Vekemans[1,2,3], Marlène Rio[4], Damien Bonnet[2,5], Pascale de Lonlay[1,2], Daniel Rabier[1,2,3]
[1]Centre de Référence des Maladies Héréditaires du Métabolisme de l'Enfant et l'Adulte, Necker Hospital, Paris, France; [2]Descartes University of Paris, France; [3]Service de Biochimie Métabolique, Necker Hospital, Paris, France; [4]Centre de Référence des Maladies Mitochondriales, Necker Hospital, AP/HP, Paris, France; [5]Cardiologie Pédiatrique, AP/HP, Paris, France

POSTER 9: Cardiolipin deficiency leads to perturbation of glutathione homeostasis
Vinay A. Patil, Miriam L. Greenberg
Dept. of Biological Sciences, Wayne State University, Detroit, MI, USA

POSTER 10: Early indicators of a cognitive phenotype in Barth syndrome
Darcy Raches[1], Michèle Mazzocco[2]
[1]St. Jude Children's Research Hospital; [2]Johns Hopkins University School of Education, Baltimore, MD, USA

POSTER 11: Genetic interaction between pyruvate dehydrogenase complex and cardiolipin mutant
Vaishnavi Raja, Amit S. Joshi, Miriam L. Greenberg
Wayne State University, Department of Biological Sciences, Detroit, MI, USA

POSTER 12: Bristol NHS Specialised Services Barth Syndrome Service — A clinical and laboratory case audit
Julie Honeychurch[1], Melanie Pennock[1], Ann Bowron[2], Colin Steward[4] , Ruth Newbury-Ecob[3], Sarah Buston[3], E Kinning[5], Maggie Williams[1]
[1]Bristol Genetics Department, Southmead Hospital, Bristol, BS10 5NB; [2]Department of Clinical Biochemistry, Bristol Royal Infirmary, BS2 8HW; [3]Clinical Genetics Department, St Michaels Hospital, Bristol; [4]Bristol Royal Hospital for Children; [5]Clinical Genetics, Royal Hospital for Sick Children, Glasgow

POSTER 13: The phenotypic characterization of the muscle specific deletion of Ptpmt1 in mice
Ji Zhang[1], Phildrich Teh[3], and Jack E. Dixon[1,2,3,4]
[1]Dept of Pharmacology, Univ. of California, San Diego, La Jolla, CA; [2]Dept of Cellular and Molecular Medicine, Univ. of California, San Diego, La Jolla, CA; [3]Dept of Chemistry and Biochemistry, Univ. of California, San Diego, La Jolla, CA; [4]Howard Hughes Medical Institute, Chevy Chase, MD, USA

POSTER 14: Expression and localization of differential mitochondrial endogenous human *Tafazzin* isoforms, cardiolipin stabilities and compensation in Barth syndrome
Yang Xu, Mindong Ren, Michael Schlame
Departments of Anesthesiology and Cell Biology, New York University Medical School, New York, New York, USA

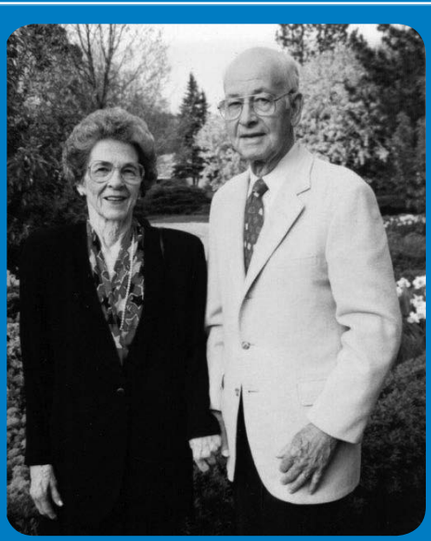
POSTER 15: The mitochondrial phosphatase PTPMT1 is essential for cardiolipin biosynthesis
Ji Zhang[1], Jack E. Dixon[1, 2, 3, 4]
[1]Dept of Pharmacology, Univ. of California, San Diego, La Jolla, CA; [2]Dept of Cellular and Molecular Medicine, Univ. of California, San Diego, La Jolla; [3]Dept of Chemistry and Biochemistry, Univ. of California, San Diego, La Jolla, CA; 4Howard Hughes Medical Institute, Chevy Chase, MD, USA

SCIENTIFIC AND MEDICAL SESSIONS

FRIDAY, JUNE 29, 2012

(North Terrace Meeting Room)

7:30 am — 8:15 am	**Breakfast** *(Grand Ballroom)*
MOUSE MODEL OF BARTH SYNDROME *Chair — Michael Schlame, MD, New York University, New York, NY*	
8:25 am — 12:00 pm **8:25 am**	**Introduction** — *Cameron, Queensland, Australia, Age 13*
8:30 am — 9:00 am	**Heart disease in Barth syndrome: Diagnosis and management** *John L. Jefferies, MD, MPH, Cincinnati Children's Medical Center, Cincinnati, OH*
9:00 am — 9:30 am	**Developmental noncompaction cardiomyopathy in a mouse Model of Barth syndrome** *Colin K. Phoon, MPhil, MD, New York University, New York, NY*
9:30 am — 10:00 am	**Endurance training in a mouse model of Barth syndrome** *Meghan Soustek, University of Florida, Gainesville, FL*
10:00 am — 10:15 am	**Break**
10:15 am — 10:45 am	**Impaired fatty acid metabolism in *tafazzin*-deficient mice** *Zaza Khuchua, PhD, Children's Hospital Medical Center, Cincinnati, OH*
10:45 am — 11:15 am	**Experimental molecular therapeutic strategies for treating Barth syndrome: Elucidation of the functional role of the mitochondrial lipidome** *Michael A. Kiebish, PhD, Senior Research Scientist, Berg Diagnostics, Omics Division*
11:15 am — 11:45 am	**Targeting cardiolipin content and composition in the *taz* shRNA mouse model of Barth syndrome** *Adam J. Chicco, PhD, Colorado State University, Fort Collins, CO*
11:45 am — 12:00 pm	**Brainstorming about future directions** *Michael Schlame, MD, New York University, New York, NY*
12:00 pm — 1:30 pm	**Varner Award Luncheon** *(Grand Ballroom)* *Awardee Introduction: John Wilkins*
MITOCHONDRIAL PHYSIOLOGY OF BARTH SYNDROME *Chair — Grant Hatch, PhD, University of Manitoba, Winnipeg, Canada*	
1:25 pm — 5:15 pm **1:25 pm**	**Introduction** — *Derek, New York, USA, Age 18*
1:30 pm — 2:00 pm	Two Poster Session presenters chosen by Sci/Med Organizing Committee
2:00 pm — 2:30 pm	**A mouse *TAZ*-knockdown model of Barth syndrome cardiomyopathy: Pathophysiology and potential treatments** *Mindong Ren, PhD, New York University, New York, NY*
2:30 pm — 3:00 pm	**Linking cardiolipin remodeling to mitochondrial beta-oxidation** *Grant Hatch, PhD, University of Manitoba, Winnipeg, Canada*
3:00 pm — 3:15 pm	**Break**
3:15 pm — 3:45 pm	**Modeling Barth syndrome in cardiomyocytes generated from patient-derived induced pluripotent stem cells** *William Pu, MD, Children's Hospital of Boston, Boston, MA*
3:45 pm — 4:15 pm	**The preferred acyl chain donor of the yeast *tafazzin*** *Anton I. de Kroon, PhD, Utrecht University, Utrecht, The Netherlands*
4:15 pm — 4:45 pm	**Cardiolipin remodeling by ALCAT1 regulates dilated cardiomyopathy through oxidative stress and mitophagy** *Yuguang (Roger) Shi, PhD, Penn State University College of Medicine, Hershey, PA*
4:45 pm — 5:15 pm	**Brainstorming and conference wrap-up** *All Conference Chairs*
5:15 pm — 7:00 pm	*Dinner on your own. All conference attendees have the option of a 10% discount at any restaurant on-site.*
7:00 pm — 11:00 pm	**Social Event ~ *X Marks The Spot For A Cure!*** *(Grand Ballroom)* *Join us for a pirate-themed evening; live entertainment featuring Alibi; delicious appetizers, cash bar; all conference attendees are invited!*
SATURDAY, JUNE 30, 2012	
8:30 am — 12:00 pm	**Scientific and Medical Advisory Board Breakfast & Meeting** *(Executive Board Room)*
1:30 pm — 2:30 pm	**Closing Finale** ~ *All conference attendees are invited to attend!* (*Grand Ballroom)*



Paula & Woody Varner (1995)
(Photo courtesy of Sue Wilkins)

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PHYSICIAN & SCIENTIST INFORMAL WELCOME CEREMONY
(Wednesday, June 27th)

BREAKFAST *(Thursday, June 28th)*

VARNER LUNCHEON *(Friday, June 29th)*

SMAB BREAKFAST *(Saturday, June 30th)*

Peter & Maureen Allman, Tom & Jane Allman, Dr. Rodney & Debbie Basler, Dave & Elizabeth Benyon, Dr. David & Kathy Bingham, Blaine & Abby Burmeister, Charles & Marita Burmeister, Jon & Jackie Burmeister, Jack & Sally Campbell, Robert & Candace Campbell, Dr. John Cheatham, Linda Cheatham, Dr. Stephen & Beth Carveth, Dr. Michael & Patricia Cimino, Doug & Leslie Deeter, Jerry & Sally Desmond, Shawn Seacrest Farrar & Douglas Farrar, David & Jane Firestone, Douglas & Pamela Ganz, Dorothy O. Hayes, Jim & Tina Hille, Norman & Debra Hedgecock, Dr. Bruce & Peggy Henricks, Walker & Dianne Kennedy, Dr. Fred & Vivian Kiechel, Dr. Max & Pat Linder, Dr. Martin & Mrs. Ruth Massengale, Gates & Daisy Minnick, J. Scott & Teri Nelson, Loy & Julie Olson, Dr. Tom & Nancy Osborne, Robert & Carolyn Otte, Dr. Ronald & Lois Roskens, David & Susan Shamblin, Linda & Bill Shreve, Dr. Kristin & Peter Somers, Sue & Kurt Sonderegger, Ted & Mary Ann Sonderegger, Robyn Steely & A. Varner Seaman, Dr. Gene & Mrs. Kristen Stohs, Dennis & Nancy Stuckey, Mary Swanson, Dr. Thomas & Barbara Tegt, Elwood & Carol Thompson, Judy Varner, Tom & Beth Varner, Jess & Mark Wiederspan, Muriel Wilkins, Sue & Dr. Mike Wilkins, Thomas & Kelli Marie Wood

FAMILY SESSIONS

THURSDAY, JUNE 28, 2012

(South Terrace Meeting Room)

Note: Families also have the option of attending the Scientific & Medical Sessions	
7:30 am — 8:15 am	**Breakfast** (*Grand Ballroom)*
10:25 am	**Welcome** — *Shelley Bowen, Director, Family Services & Awareness*
10:30 am — 11:15 am	**Cardiology Roundtable Discussion** New families can learn from seasoned families about cardiology topics such as: heart failure, assistive devices, medications, transplants, etc. This session will help shape the afternoon session with leading experts in cardiology. BTHS Male*: Aldo, Massachusetts, USA, Age 19* BTHS Parent: *Ned Kalapasev, Kentucky, USA* BSF Facilitator: *Shelley Bowen, Director, BSF Family Services & Awareness*
11:15 am — 12:00 pm	**Hematology Roundtable Discussion** New families can learn from seasoned families about hematology topics such as: medications, antibiotics, G-CSF, understanding ANC, etc. This session will help shape the afternoon session with leading experts in hematology. BTHS Male*: Will, New York, USA, Age 26* BTHS Parent: *Tiffini Allen, Indiana, USA* BSF Facilitator: *Shelley Bowen, Director, BSF Family Services & Awareness*
12:00 pm — 1:30 pm	**Keynote Speaker Luncheon** *(Grand Ballroom)* *Stephen C. Groft, Pharm D, Director of the Office of Rare Disease Research (ORDR), NIH, Bethesda, MD, USA* **A globalization of rare diseases research activities** **Group Photo** *(Location TBD)*
1:30 pm — 1:45 pm	**Break**
1:45 pm — 3:45 pm **1:45 pm — 1:55 pm** **1:55 pm — 2:20 pm** **2:20 pm — 2:45 pm** **2:45 pm — 3:10 pm** **3:10 pm — 3:45 pm**	**Cardiac aspects of Barth syndrome** Speaker Introduction: *Stephanie Radar, RN, BSN, MSN, NNP-BC, BTHS Parent, Missouri, USA* *Gonzalo Wallis, MD* *Randall Bryant, MD* *W. Todd Cade, PT, PhD* **Q & A**
3:45 pm — 4:00 pm	**Break**
4:00 pm — 5:25 pm **4:00 pm — 4:10 pm** **4:10 pm — 4:35 pm** **4:35 pm — 5:00 pm** **5: 00 pm — 5:25 pm**	**Managing neutropenia in Barth syndrome** Speaker Introduction: *Suzy Green, BTHS Parent, Cambridgeshire, United Kingdom* *David C. Dale, MD* *Colin G. Steward, PhD, FRCP, FRCPCH* **Q & A**
5:25 pm — 6:00 pm	**Break** Dinner on your own. All conference attendees have the option of a 10% discount at any restaurant on-site.
6:00 pm — 7:00 pm	**POSTER SESSION** *(Fifth Floor Foyer)*

FAMILY SESSIONS

FRIDAY, JUNE 29, 2012

(South Terrace Meeting Room)

Note: Families also have the option of attending the Scientific & Medical Sessions	
7:30 am — 8:15 am	**Breakfast** (*Grand Ballroom)*
10:15 am **10:15 am — 11:00 am**	Speaker Introduction: *Shelley Bowen, Director, BSF Family Services & Awareness* **Heart and skeletal muscle metabolism, energy production and function in Barth syndrome — Opportunity to participate in a new NIH-funded research study.** *W. Todd Cade, PT, PhD*
11:00 am — 11:15 am	**Break**
11:15 am — 12:00 pm	**Nutrition Roundtable Discussion** New families can learn from seasoned families about nutritional topics such as: multi-vitamins and supplement samples, food preferences and aversions, hydration, testing, diarrhea, constipation, mouth ulcers, tube feeds, etc. BTHS Male: *Kevin, Philadelphia, USA, Age 23* BTHS Parent: *Brie Chandler, Kentucky, USA* BTHS Parent: *Donna Strain, RN, Queensland, Australia* BSF Facilitator: *Shelley Bowen, Director, Family Services & Awareness*
12:00 pm — 1:30 pm	**VARNER AWARD LUNCHEON** *(Grand Ballroom)* *Awardee Introduction: John Wilkins*
1:30 pm — 1:45 pm	**Break**
1:45 pm — 2:55 pm **1:45 pm — 1:55 pm** **1:55 pm — 2:40 pm** **2: 40 pm — 2:55 pm**	**Metabolism of Barth syndrome** Speaker Introduction: *Florence Mannes, BTHS Parent, Paris, France* *Richard I. Kelley, MD, PhD* **Q & A**
2:55 pm — 3:10 pm	**Break**
3:10 pm — 4:30 pm	**Update on research in Barth syndrome** Speaker Introduction: *Ryan, Ontario, Canada, Age 21* *Matthew J. Toth, PhD* *Michael Schlame, MD* **Q & A**
4:30 pm — 7:00 pm	**Break** ~ *Don't forget to grab a bite to eat so that you have the energy to dance the night away at our Social Event! All conference attendees have the option of a 10% discount at any restaurant on-site.*
7:00 pm — 11:00 pm	**Social Event ~ *X Marks The Spot For A Cure!*** *(Grant Ballroom)* *Join us for a pirate-themed evening; live entertainment featuring Alibi; delicious appetizers, cash bar; all conference attendees are invited!*

FAMILY SESSIONS
SATURDAY, JUNE 30, 2012

Note: Breakfast is on your own		
10:30 am — 12:15 pm	**Mom Breakout Session** *(North Terrace)* Facilitator Introduction: *Lynda Sedefian, BTHS Parent, New York, USA* Facilitator: *Vanessa Garratt, DClinPsych* Facilitator: *Rebecca McClellan, MGC, CGC*	**Dad Breakout Session** *(South Terrace)* Facilitator Introduction: *Mike Wilkins, BTHS Parent, Nebraska, USA* Facilitator: *BJ Develle, MSW*
12:15 pm — 12:30 pm	Break	
12:30 pm — 1:30 pm	**Luncheon ~ BSF w/ Burgers/Shakes/Fries** *(Grand Ballroom)*	

1:30 pm — 2:30 pm

CLOSING FINALE
All conference attendees are invited to attend!
(Grand Ballroom)

A new family and a seasoned family will share their experiences of the Conference.
We will conclude with two photographic slideshows featuring candid shots throughout the week and portraits by Amanda Clark.

THE McCURDY FAMILY

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LUNCHEON

Saturday, June 30, 2012

(Photo courtesy of McCurdy Family ~ 2012)

BARTH FRANCE

is the proud sponsor of the

BARTH SYNDROME CONSULTATIONS

June 26 – 27, 2012

Loyal team members of Ironman4Barth participated in the Paris Marathon on April 15, 2012 in support of Barth France.
(Photo courtesy of Barth France ~ 2012)

MODERATORS ~ AFFECTED INDIVIDUALS	MODERATORS ~ SIBLINGS
Ages 8 – 11: *Lisa Duran & Angelo Florez* **Ages 12 – 17**: *BJ Develle, MSW* **Ages 18+:** *John Wilkins*	**Ages 8 – 11:** *Ashley Cade* **Ages 12 – 17:** *Michal Weinberger, MSW; Eliza McCurdy* **Ages 18+:** *John Wilkins*

AFFECTED INDIVIDUALS & SIBLING SESSIONS
WEDNESDAY, JUNE 27, 2012

4:00 pm — 4:30 pm	**Ages 18+: Exercise intolerance & Barth syndrome** *(Executive Board Rooms)* *W. Todd Cade, PT, PhD*
4:40 pm — 5:10 pm	**Ages 12 – 17: Exercise intolerance & Barth syndrome** *(Executive Board Rooms)* *W. Todd Cade, PT, PhD*
5:20 pm — 5:50 pm	**Ages 8 – 11: Exercise intolerance & Barth syndrome** *(Executive Board Rooms)* *W. Todd Cade, PT, PhD*

AFFECTED INDIVIDUALS & SIBLING SESSIONS
THURSDAY, JUNE 28, 2012

Note: All BTHS individuals and siblings over the age of 18 have the option of attending the Scientific & Medical Sessions

<table>
<tr><td colspan="6">7:30 am - 8:15 am — Breakfast (Grand Ballroom)</td></tr>
<tr><td colspan="6">9:00 am — 10:00 am
MEET YOUR GROUP LEADER / INTRODUCTIONS
All Affected Individuals and Siblings
(King Charles)</td></tr>
<tr><td colspan="6">10:00 am — 11:00 am</td></tr>
<tr><td colspan="3">AFFECTED INDIVIDUALS</td><td colspan="3">SIBLINGS</td></tr>
<tr><td>Ages 8-11</td><td>Ages 12-16</td><td>Ages 16+</td><td>Ages 8-11</td><td>Ages 12-17</td><td>Ages 18+</td></tr>
<tr><td colspan="2">King Charles</td><td>Executive Board Room</td><td colspan="3">King Charles</td></tr>
<tr><td colspan="2">Ping Pong</td><td>Metabolism of Barth syndrome
Richard I. Kelley, MD, PhD</td><td colspan="3">Photography fun with Amanda Clark</td></tr>
<tr><td colspan="6">11:00 am — 12:00 pm</td></tr>
<tr><td>Ages 8-11</td><td>Ages 12-17</td><td>Ages 18+</td><td>Ages 8-11</td><td>Ages 12-17</td><td>Ages 18+</td></tr>
<tr><td colspan="3">King Charles</td><td colspan="3">King Charles</td></tr>
<tr><td colspan="3">Insights from the insiders
Facilitator: BJ Develle</td><td colspan="3">I'm affected too
Facilitator: Michal Weinberger, MSW
Facilitator: Eliza McCurdy</td></tr>
<tr><td colspan="6">12:00 pm — 1:30 pm
Keynote Speaker Luncheon
(Grand Ballroom)
Group Photo
(Location TBD)</td></tr>
<tr><td colspan="6">1:30 pm — 1:45 pm ~ Break</td></tr>
<tr><td colspan="6">1:45 pm — 3:00 pm</td></tr>
<tr><td>Ages 8-11</td><td>Ages 12-17</td><td>Ages 18+</td><td>Ages 8-11</td><td>Ages 12-17</td><td>Ages 18+</td></tr>
<tr><td colspan="3">King Charles</td><td colspan="3">King Charles</td></tr>
<tr><td colspan="3">Photography fun with Amanda Clark</td><td colspan="3">Activity Time</td></tr>
<tr><td colspan="6">3:00 pm — 5:00 pm
Activity Time (King Charles)
All Affected Individuals and Siblings
Activities include cupcake making, caricatures, centerpiece making, donor thank you cards, ping pong, scrapbooking, Wii games, and other awesome activities</td></tr>
</table>

AFFECTED INDIVIDUALS & SIBLING SESSIONS

FRIDAY, JUNE 29, 2012

Note: All BTHS individuals and siblings over the age of 18 have the option of attending the Scientific & Medical Sessions
****Please note the change in age groups where indicated**

7:30 am — 8:15 am | **Breakfast** *(Grand Ballroom)*

9:00 am — 9:45 am
Hang Out/Discuss Day's Activities

9:45 am — 12:00 pm
Shelling/Sand Castle Building
All Affected Individuals and Siblings
(Everyone will meet in King Charles to venture off to the Main Hotel beach)

12:00 pm — 1:30 pm
VARNER AWARD LUNCHEON
(Grand Ballroom)

Youth Pizza Luncheon *(Pavilion West)*
All Affected Individuals and Siblings ages 8+

1:30 pm — 1:45 pm ~ Break

1:45 pm — 3:00 pm			**1:45 pm — 3:00 pm**		
Ages 8-11	**Ages 12-15****	**Ages 16+****	**Ages 8-11**	**Ages 12-17**	**Ages 18+**
King Charles		*Executive Board Room*	*King Charles*		
Movie & Games		**My family, me and Barth syndrome** *Vanessa Garratt, DClinPsych*	**Movie & Games**		

3:00 pm — 4:30 pm			**3:00 pm — 4:30 pm**		
Ages 8-11	**Ages 12-17**	**Ages 18+**	**Ages 8-11**	**Ages 12-15****	**Ages 16+****
King Charles					*Executive Board Room*
Movie & Games					**My family, me and Barth syndrome** *Vanessa Garratt, DClinPsych*

7:00 pm — 11:00 pm
SOCIAL EVENT *(Grand Ballroom)*
Join us for a pirate-themed evening; live entertainment featuring Alibi; delicious appetizers, cash bar; all conference attendees are invited

SATURDAY, JUNE 30, 2012

Breakfast on your own

10:00 am — 11:00 am

AFFECTED INDIVIDUALS			**SIBLINGS**		
Ages 8-11	**Ages 12-17**	**Ages 18+**	**Ages 8-11**	**Ages 12-17**	**Ages 18+**
King Charles	*Executive Board Room*	*King Charles*			
Finale Project	**Barth syndrome and beyond** *Randall Bryant, MD*	**Finale Project**			

11:00 am — 12:30 pm			**11:00 am — 12:30 pm**		
Ages 8-11	**Ages 12-17**	**Ages 18+**	**Ages 8-11**	**Ages 12-17**	**Ages 18+**
King Charles		*Executive Board Room*	*King Charles*		
Finale Project		**Barth syndrome and beyond** *Randall Bryant, MD*	**Finale Project**		

12:30 pm — 1:30 pm ~ LUNCHEON ~ BSF w/ Burgers/Shakes/Fries *(Grand Ballroom)*

1:30 pm — 2:30 pm ~ CONFERENCE FINALE *(Grand Ballroom) All conference attendees are invited to attend!*

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The 2012 Barth Syndrome Foundation International Conference

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XMARKS THE SPOT FOR A CURE!
Barth Syndrome Foundation 2012 Conference

June 25–30, 2012
St. Pete Beach, FL, USA

(Photos courtesy of Woodward Family ~ 2012)

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BREAKFAST

Thursday, June 28, 2012

Dilen and Ronly (Photo courtesy of Susan McCormack ~ 2012)

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KEYNOTE SPEAKER LUNCHEON

Thursday, June 28, 2012

Michael Aviles, Peter Bliven, Stephen Dance, Constance Duhamel, Howard Feldman, Paul Ferrarese, John Gonzalez, David & Rebecca Hamlin, Julian & Janice Hargraves, Dr. Stephen Heller & Dr. Pamela Beasley, Eugene Kagan, Andrew Kearney, Kaaren & DJ Lee, Kim Marra, Ronald Marra, John & Helen McCormack, Thalia Meehan, Karen Moser, Michael Reazin, Louis Rowlett, Donna & Anthony Sannicandro, Richard & Shelley Stanzel, Edward & Elizabeth Stone

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REFRESHMENT BREAK

Saturday, June 30, 2012

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FAMILY WELCOME EVENT	Stephanie, Noah, Olivia & Carol Rader
TABLES AT FRIDAY NIGHT SOCIAL	Rick, Lynn, Adam and Justin Elwood Lindsay Groff & Eileen Belfatto Lois Galbraith & Les Morris Susan, Chris, Jess, Jen, Josh & Jared Hone Dr. Timothy & Marcia McCormack Congratulations Dr. Grzegorz Nalepa, Love Henry Stephanie, Noah, Olivia & Carol Rader (2) James & Kimberly Werth The Family of Connor Woodward (5) Lorna & Nigel Moore & Barth Syndrome Trust *(UK & Europe)* (5)
REGISTRATION DESK SUPPLIES	Amanda & Tim Maksin
SUPPLIES FOR CHILDREN´S ACTIVITIES	Les Morris
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BSF would like to acknowledge with gratitude the following who gave in-kind donations in support of BSF's 2012 Conference:

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Lindsay Groff	Luxury Bath Products from Cape May, NJ; Traveler's Package
Jan Kugelmann, Sharon Olson, Lois Galbraith & Cathy Ritter	"Driving for a Cure" golf shirt, hat/visor, BSF logo golf balls, and golf towel,
Joyce Lochner	Hand-Made Heart Wallhanging
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Eliza & Will McCurdy	NYPD and FDNY T-shirts
Kate McCurdy	Collapsable insulated cooler; Burgers, Shakes & Fries (BSF) T-shirt
Steve McCurdy	Signed copy of **You are an Ironman** (a book about six triathletes dedicated, in part, to Team Will); Janus Charity Triathlon hat
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The Conference Steering Committee would also like to thank the members of Alibi for performing at the Friday Night Social Event. Members include Thomas Harrison, Maurice Black, Darrell Valentine, Todd Ratliff, and Alanna Layton.

The Conference Steering Committee would also like to thank Jessica Wilkins Wiederspan for her in-kind donation of creating and compiling the data for conference evaluations and Michael Bowen for working behind the scenes with the conference planning.

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

Dr. Bryant's specialties include pediatric cardiology and pediatric medicine, and 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
Cardiac aspects of Barth syndrome *(Family Sessions)*
Barth syndrome and beyond *(Youth Sessions)*

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 holds 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 is a licensed physical therapist. He completed a post-doctoral fellowship in the Division of Endocrinology, Metabolism and Lipid Research at Washington University School of Medicine, and holds an NIH-funded Career Development Award from the National Institute of Diabetes and Digestive and Kidney Diseases.

Dr. Cade was awarded three BSF grants entitled "Effects of resistance training on cardiac, metabolic, and muscle function and quality of life in Barth syndrome" (2011); "Safety and efficacy of aerobic exercise training in Barth syndrome: A pilot study" (2009); and "Characterization of nutrient metabolism in Barth syndrome" (2008). Dr. Cade recently received an NIH funded grant examining whole-body and heart metabolism and function in Barth syndrome.

Presentations
Exercise and substrate metabolism studies in Barth syndrome: Updates and future directions *(Sci/Med Sessions)*
Cardiac aspects of Barth syndrome *(Family Sessions)*
Heart and skeletal muscle metabolism, energy production and function in Barth syndrome — Recruiting for a new NIH-funded study *(Family Sessions & Consultations/Research)*
Exercise intolerance and Barth syndrome *(Youth Sessions)*

Adam J. Chicco, MEd, PhD — Assistant Professor, Departments of Health and Exercise Science, Biomedical Sciences, and Food Science and Human Nutrition; Faculty, Program in Cell and Molecular Biology; Director, Integrative Cardiac Biology Laboratory, Colorado State University, Fort Collins, CO, USA

Dr. Chicco's current focus is elucidating the mechanisms and consequences of alterations that occur in mitochondrial membrane composition and the metabolism of polyunsaturated fatty acids (PUFA) in the context of cardiovascular and metabolic disease. Particular interests include the influence of cardiolipin acyl chain composition on mitochondrial pathophysiology and the role of delta-6 desaturase in the pathogenesis of heart failure and metabolic disease. Dr. Chicco was awarded a research grant from BSF entitled "Targeting cardiolipin deficiency in the taz shRNA mouse model of Barth syndrome" (2010).

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

Presentation
Targeting cardiolipin content and composition in the *taz* shRNA mouse model of Barth syndrome. *(Sci/Med Sessions)*

Maryanne Chrisant, MD — Director, Pediatric Cardiac Transplant, Heart Failure & Cardiomyopathy, Joe DiMaggio Children's Hospital, Hollywood, FL, USA

Dr. Chrisant's career highlights include directing heart transplant programs at the Cleveland Clinic Foundation in Cleveland, Ohio; Children's Hospital of Philadelphia (CHOP) in Pennsylvania; and University of Virginia Children's Hospital in Charlottesville.

Dr. Chrisant holds a Bachelor's Degree from Tufts University and a Medical Degree from New York Medical College. She completed two fellowships at Columbia University in New York, one in pediatric cardiology and the other in pediatric cardiac transplant.

Dr. Chrisant is a member of the International Society for Heart and Lung Transplantation and the American Heart Association. She participates in ongoing research in the Pediatric Heart Transplant Study Group. She has lectured internationally on the topics of pediatric heart transplantation, heart failure and cardiomyopathies. Dr. Chrisant has authored many papers about heart transplantation and heart failure in children, congenital heart disease, and co-authored pediatric guidelines for heart transplant and heart failure management.

Note — Dr. Chrisant is involved in the Consultations

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 was awarded a resesarch grant from BSF entitled "Characterizing endogenous mammalian TAZ1" (2011).

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 — The topology of cardiolipin remodeling in yeast *(Sci/Med Session)*

David C. Dale, MD — Professor of Medicine, University of Washington, Seattle, WA, USA

Dr. Dale's research career has focused on studies of inflammation, neutrophils, and the clinical problem of neutropenia, including neutropenia and susceptibility to infections in Barth syndrome. His specialty interests include hematology, oncology, infectious diseases and medical genetics. Dr. Dale was awarded two research grants from BSF entitled "Neutropenia in Barth syndrome" (2003 and 2007).

Dr. Dale is actively involved in teaching, research and patient care at the University of Washington Medical Center, Seattle WA. He has served as the Dean of the School of Medicine, University of Washington, and President of the American College of Physicians, President of the Alpha Omega Alpha Honor Medical Society, and Editor-in-Chief of the medical textbook, Scientific American Medicine/ACP Medicine. He is currently Co-Director and Principal Investigator for the Severe Chronic Neutropenia International Registry (SCNIR).

Dr. Dale graduated from Carson-Newman College and the Harvard Medical School. He trained in Internal Medicine at the Massachusetts General Hospital Boston and the University of Washington Medical Center, Seattle.

Presentation — Managing neutropenia in Barth syndrome *(Family Sessions)*

Anton I. de Kroon, PhD — Docent (Associate Professor), Utrecht University, Utrecht, The Netherlands

After post-doctoral research at Stanford University, Dr. de Kroon returned from Utrecht University and joined the Department of Chemistry as faculty member in 1995. De Kroon's lab is part of the Membrane Biochemistry and Biophysics Group that in turn participates in the Bijvoet Center for Biomolecular Research and the Institute of Biomembranes. Dr. de Kroon's research focuses on phospholipid-protein interactions and their functions in cell biology and on the regulation of lipid synthesis and membrane lipid homeostasis. These topics are investigated in the model eukaryote Saccharomyces cerevisiae using biochemical, molecular biological, genetic, and chemical biological approaches, complemented by proteome and lipidome analysis. Dr. de Kroon was awarded a research grant from BSF entitled "The preferred acyl chain donor of Taz1p in the acylation of monolysocardiolipin" (funded by Barth Syndrome Trust - 2010).

Dr. de Kroon graduated cum laude in Biology (1986) and received his PhD cum laude (1991) at Utrecht University, The Netherlands.

Presentation —The preferred acyl chain donor of the yeast *tafazzin (Sci/Med Session)*

Bruce (BJ) Develle, MSW

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

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

Facilitator — Dad's Breakout Session *(Family Sessions)*
Group Leader — Youth Sessions

Vanessa Garratt, DClinPsych — Clinical Psychologist in the Paediatric Cardiac Service, Paediatric HIV Service and the National Barth Syndrome and Osteogenesis Imperfecta Services, Bristol Royal Hospital for Children, Bristol, United Kingdom

Dr. Garratt completed her Doctorate in Clinical Psychology in Bristol in 2005, and since then has worked as a Clinical Psychologist with children and young people who have cardiac conditions, HIV, Barth syndrome and Osteogenesis Imperfecta. Her major interest is in helping young people and families cope with and adjust to living with long-term conditions, often providing support to young people and families around a number of different issues, including:

- *management of distressing medical procedures;*
- *coping and adjusting to having a long-term condition;*
- *finding out about and linking into local services and educational support;*
- *empowering teenagers to take control of their own health and transitional care, and*
- *working with siblings.*

Dr. Garratt's work with the Barth Service has included neuropsychological assessment to help with getting the right support in schools, working with families in clinic and at home, developing information for families around education and the service and working with young people in groups in the clinic. Dr. Garrett is a member of the British Psychological Society and Health Professions Council, UK.

Co-Facilitator — Mom's Breakout Session *(Family Sessions)*
Discussion — My family, me and Barth syndrome (*Youth Sessions)*

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 from BSF entitled "A study of TAZ mRNAs in Barth syndrome individuals."

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

Note — Dr. Gonzalez is involved in the Consultations

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 was awarded the following research grants from BSF entitled "Cardiolipin deficiency leads to defects in the TCA cycle" (2011); "Loss of cardiolipin leads to defective mitochondrial iron/sulfur biosynthesis and iron homeostasis" (2010); "Perturbation of mitophagy in cardiolipin mutants" (2009); "The role of tafazzin in mitochondrial protein import—Implications for Barth syndrome" (2008); "Perturbation of the osmotic stress response in cardiolipin deficient mutants" (2007); "The role of phosphatidylglycerol in activating protein Kinase C mediated signaling" (2006); "Does copper deficiency play a role in Barth syndrome" (2005); "TAZ1 gene function in yeast: A molecular model for Barth syndrome" (2002).

Presentation — Loss of cardiolipin leads to perturbation of mitochondrial and cellular iron homeostasis *(Sci/Med Sessions)*

Stephen C. Groft, Pharm D — Director, Office of Rare Disease Research (ORDR), National Center for Advancing Translational Sciences (NCATS), National Institutes of Health (NIH), Bethesda, MD, USA

Dr. Groft's major focus is on stimulating research with rare diseases and developing information about rare diseases and conditions for health care providers and the public. To help identify research opportunities and establish research priorities, the Office has co-sponsored over 1,100 rare diseases-related scientific conferences with the NIH research Institutes and Centers. Current activities include establishing patient registries for rare diseases, developing an inventory of available bio-specimens from existing biorepositories, developing an educational module on rare diseases for middle school children, establishing a public information center on genetic and rare diseases, developing an international rare diseases research consortium, maintaining the Rare Diseases Clinical Research Network, and providing a special emphasis clinic with senior clinical staff for patients with undiagnosed diseases at NIH's Clinical Research Center Hospital.

Dr. Groft served in 1991-1992 as the first Acting Director of the Office of Alternative Medicine at the NIH (now the National Center for Complementary and Alternative Medicine) and the Staff Director for the White House Commission on Complementary and Alternative Medicine Policy from 2000-2002. Dr. Groft received the B.S. degree in Pharmacy in 1968 and the Doctor of Pharmacy degree from Duquesne University in 1979.

Keynote Speaker — Presentation: A globalization of rare diseases research activities

Grant M. Hatch, PhD — Professor, Departments of Pharmacology & Therapeutics and Biochemistry & Medical Genetics, University of Manitoba; Canada Research Chair in Molecular Cardiolipin Metabolism; Director of the Lipid Lipoprotein and Atherosclerosis Research Group (LLARG); Director of the Centre for Research and Treatment of Atherosclerosis, Scientist, Manitoba Institute of Child Health, Winnipeg, Canada; Scientific & Medical Advisory Board, Barth Syndrome Foundation, Inc.

Dr. Hatch's main research interests have been focused on studying metabolism of cardiolipin — a key phospholipid involved in mitochondrial energy production. He has identified a human protein, MLCL AT-1, which may regulate the fatty acyl composition of cardiolipin. With financial support from BSF, he is investigating if expression of this protein in cardiac cells of tafazzin knockdown mice attenuates the loss of cardiolipin and the cardiac pathology observed in these animals. Dr. Hatch was awarded the following research grants from BSF entitled "MLCL AT-1 elevates cardiolipin and mitochondrial function in cardiac myocytes of taz knockdown mice" (funded by BSFCanada - 2011); "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).

Chair— Biological Function of Tafazzin and Cardiolipin *(Sci/Med Sessions)*
Chair— Mitochondrial Physiology of Barth Syndrome *(Sci/Med Sessions)*

Presentation — Linking cardiolipin remodelling to mitochondrial beta-oxidation *(Sci/Med Sessions)*

John Lynn Jefferies, MD, MPH, FAAP, FACC— Associate Professor, Pediatric Cardiology and Adult Cardiovascular Diseases; Director, Advanced Heart Failure/Cardiomyopathy, Ventricular Assist Device Program, Cardiomyopathy; Co-Director, Cardiovascular Genetics; Associate Director, Heart Institute Research Core, Cincinnati Children's Hospital Medical Center, Cincinnati, OH, USA

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

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

Presentation — Heart disease in Barth syndrome: Diagnosis and management *(Sci/Med Sessions)*

Richard I. Kelley, MD, PhD — Director, Genetics Laboratories and the Clinical Mass Spectrometry Laboratory, Kennedy Krieger Institute; Professor of Pediatrics, Johns Hopkins University School of Medicine; Baltimore, MD, USA; Scientific and Medical Advisory Board, Barth Syndrome Foundation, Inc.; Advisory Board, Barth Syndrome Registry & Repository

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

Chair—Barth Syndrome Pathophysiology *(Sci/Med Sessions)*

Presentations
Abnormalities of intermediary metabolism in Barth syndrome *(Sci/Med Sessions)*
Metabolism of Barth syndrome *(Family Sessions)*
Metabolism of Barth syndrome *(Youth Sessions)*
Note: Dr. Kelley is involved in the Consultations/Research

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. In 2006, Dr. Khuchua described the defects of the heart development in a zebrafish model of tafazzin deficiency. Dr. Khuchua was awarded a research grant from BSF entitled "The shRNA-mediated tafazzin knockdown mouse model for Barth syndrome" (2009). In 2010, he started investigating the metabolic consequences of tafazzin knockdown in mice. In 2011, Dr. Khuchua was awarded a 4-year grant from National Heart, Lung and Blood Institute at the National Institutes of Health for research using the Barth knockdown mouse model.

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

Presentation — Impaired fatty acid metabolism in *tafazzin*-deficient mice *(Sci/Med Sessions)*

Michael A. Kiebish, PhD — Senior Research Scientist, Berg Diagnostics, Omics Division, Natick, MA, USA

Dr. Kiebish's research interests are focused on investigating molecular therapeutic strategies and targets to treat abnormal mitochondrial lipid metabolism, cardiolipin molecular speciation, and bioenergetics in a variety of disease states, including Barth syndrome. He utilizes multidimensional based-shotgun lipidomics (MDMS-SL), bioenergetic measurements, computational biology, and multiple 'omic' technologies to integrate a systems biological approach to decipher the intertwined biological networks associated with complex disease phenotypes. Dr. Kiebish was awarded a research grant from the Barth Syndrome Foundation entitled "Does cardiolipin synthase upregulation alleviate cardiolipin abnormalities and bioenergetic dysfunction in Barth syndrome?" (2009).

Dr. Kiebish holds a BS from Villanova University (2002) and a PhD in Biology from Boston College (2008). His post doctoral training (2008-2012) was completed at Washington University School of Medicine, Department of Internal Medicine, Division of Bioorganic Chemistry and Molecular Pharmacology.

Presentation — Experimental molecular therapeutic strategies for treating Barth syndrome: Elucidation of the functional role of the mitochondrial lipidome *(Sci/Med Sessions)*

Consuelo Kreider, MHS, OTR/L — Adjunct Lecturer, College of Public Health and Health Related Professions, Department of Occupational Therapy, University of Florida, Gainesville, FL, USA

Ms. Kreider joined the University of Florida in 2005 where she teaches in the Masters of Occupational Therapy program. She has extensive experience working with children, adolescents and adults living with a range of diagnoses that include learning disability, intellectual disability, autism spectrum disorders, developmental coordination disorder, and central auditory processing disorder.

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

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

Note: Ms. Kreider is involved in the Consultations/Research

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

Mrs. McClellan joined Dr. Kelley's team at Kennedy Krieger Institute (KKI) 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 provides genetic counseling consults throughout the Institute. In addition, Mrs. McClellan has recently been appointed to the Ethics Committee at KKI, is an active member of the National Society of Genetic Counselors, and enjoys supervising genetic counseling students.

Co-Facilitator — Mom's Breakout Session *(Family Sessions)*
Note — Mrs. McClellan is also involved in the Consultations/Research

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

Dr. McMaster's research focuses on the development of therapies for unmet medical needs including inherited orphan diseases. He is lead investigator of 'Identifying Genes and New Therapies to Enhance Treatment for orphan diseases' (IGNITE), a project to deliver new therapies for orphan disease patients in dramatically compressed timeframes at reduced cost.

Dr. McMaster's research interests include the use of molecular, genetic, genomic, cell biological, and proteomic approaches to isolate new genes and proteins that regulate lipid metabolism. He and his team have developed small molecule inhibitors of lipid synthesis in drug resistant bacteria. Dr. McMaster was awarded two research grants from BSF and BSFCanada entitled "A screen for drug leads for the treatment of Barth syndrome" (2010); and "Synthetic genetics towards understanding Barth syndrome cell biology" (2007).

Dr. McMaster holds a PhD from the University of Manitoba. He serves as Co-Director of the Cheminformatics Drug Discovery Lab. Dr. McMaster co-founded and is president of DeNovaMed, Inc., a Halifax based biotechnology company specializing in using computer aided drug design to drive synthesis and development of truly new classes of antimicrobials.

Presentation — Yeast genome-wide screens to assess the genetic landscape for Barth syndrome *(Sci/Med Sessions)*

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

Mr. Nurse has been involved in special needs advocacy for nearly twenty-one 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. Mr. Nurse was awarded a BS in Leisure Service (1983) College of Education, Florida State University.

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

Discussion — Financial independence and transitioning into adulthood *(Small Group Discussions)*

Colin K.L. Phoon, MPhil, MD — Associate Professor, New York University Langone Medical Center and New York University School of Medicine, New York, NY, USA

Dr. Phoon is a pediatric cardiologist on the faculty of NYU School of Medicine, board certified in General Pediatrics and Pediatric Cardiology. His research focus is the role of mitochondria and its major phospholipid cardiolipin, in the pathogenesis of cardiomyopathy, using an inducible tafazzin-knockdown mouse model of Barth syndrome. Dr. Phoon and his close collaborators Drs. Mindong Ren and Michael Schlame have recently published their findings of a developmental noncompaction cardiomyopathy in the TAZ knockdown mouse. In recognition for this work, Dr. Phoon was one of 17 recipients of Travel and Science Awards to the National Heart, Lung & Blood Institute Mitochondrial Biology Symposium in May 2011. A practicing clinical pediatric cardiologist with a special interest in pediatric and fetal echocardiography, Dr. Phoon has been a PI or co-investigator on several projects relevant to a broad spectrum of cardiovascular disease in small animal models. Dr. Phoon was awarded a research grant from BSF entitled "Cardiomyopathy in a mouse model of Barth syndrome" (2010).

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

Presentation — Developmental noncompaction cardiomyopathy in a mouse model of Barth syndrome *(Sci/Med Sessions)*

William T. Pu, MD — Associate Professor, Harvard Medical School, Department of Cardiology, Children's Hospital Boston, Principal Faculty, Harvard Stem Cell Institute, Boston, MA, USA

Dr. Pu is a pediatric cardiologist who focuses on basic research to understand the regulation of heart development and function. A major goal of these studies is to improve the future diagnosis and treatment of congenital and adult heart disease. Dr. Pu was awarded two research grants from BSF entitled "Using induced pluripotent stem cells and modified RNAs to model and correct Barth syndrome" (2011); and "Analysis of metabolic abnormalities in TAZ-deficient cardiomyocytes" (2009).

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

Presentation — Modeling Barth syndrome in cardiomyocytes generated from patient-derived induced pluripotent stem cells *(Sci/Med Sessions)*

Vanessa Rangel Miller — VP Genetic Services, PatientCrossroads, Innolyst, Inc., San Mateo, CA, USA

Ms. Rangel Miller joined PatientCrossroads in 2010 as VP Genetic Services, guiding clients to develop new registries and collaboratives. As a certified genetic counselor, she has focused on the rare disease community. Prior to joining PatientCrossroads, she managed the DuchenneConnect and Congenital Muscle Disorders Registries; and was the Operations Manager at Emory Genetics Laboratory, a non-profit academic genetic testing laboratory that serves rare disease clinicians and families worldwide. Ms. Rangel Miller completed her masters in genetic counseling at the University of North Carolina – Greensboro and her MBA at Emory University.

Note — Ms. Rangel Miller is involved in the Consultations

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

Dr. Ren's research interests include the role of cardiolipin in health and disease and the pathogenic mechanism of Barth syndrome. His research on Barth syndrome has been funded by the Barth Syndrome Foundation, the United Mitochondrial Disease Foundation, and the National Heart, Lung & Blood Institute of the National Institutes of Health. Dr. Ren was awarded three research grants from BSF entitled: "Drug repositioning for Barth syndrome" (2011); "Pathogenetic mechanism and genetic suppressors of Barth syndrome" (2006); and "A drosophila model of Barth syndrome" (2004).

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

Presentation — A mouse *TAZ*-knockdown model of Barth syndrome cardiomyopathy: Pathophysiology and potential treatments *(Sci/Med Sessions)*

Stacey Reynolds, PhD, OTR/L — K12 Scholar and Visiting Assistant Professor, College of Public Health and Health Professions, Department of Occupational Therapy, University of Florida (UF), Gainsville, FL; Assistant Professor, Department of Occupational Therapy, School of Allied Health Professions, Virginia Commonwealth University, VA, USA

Dr. Reynolds' 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. Her current research, conducted through collaborations with the Department of Psychology at UF, 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 holds a PhD in Health Related Science from Virginia Commonwealth University (2007), MS in Occupational Therapy, (2001) and BS in Health Science, (2000) Gannon University. She is Board certified in Occupational Therapy. Dr. Reynolds is a member of the International Society for Autism Research, American Occupational Therapy Association, and the Virginia and the Maryland Occupational Therapy Associations.

Note: Dr. Reynolds is involved in the Consultations/Research

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

Dr. Schlame has studied the biochemistry of cardiolipin for over 20 years. His sub-specialties include cardiothoracic anesthesiology and critical care.

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.

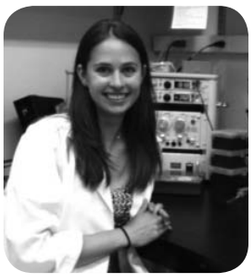
Chair—Mouse Model of Barth Syndrome *(Sci/Med Sessions)*

Presentations
The mechanism of *tafazzin (Sci/Med Sessions)*
Update on scientific research in Barth syndrome *(Family Session)*

Yuguang (Roger) Shi, PhD — Professor, Penn State University College of Medicine, Department of Cellular and Molecular Physiology, Hershey, PA, USA

Dr. Shi has a longstanding research interest in translation medicine. His current laboratory at Penn State mainly focuses on molecular mechanisms underlying mitochondrial dysfunction in obesity and type 2 diabetes. His laboratory pioneered the cloning of several novel enzymes involved in cardiolipin synthesis and remodeling. Dr. Shi's research work has recently deciphered a major role of pathological cardiolipin remodeling in mitochondrial dysfunction associated with aging-related diseases, including diabetes, obesity, and CV diseases.

Presentation — Cardiolipin remodeling by ALCAT1 regulates dilated cardiomyopathy through oxidative stress and mitophagy *(Sci/Med Sessions)*



Meghan Soustek — Graduate Student, University of Florida, Gainesville, FL, USA

Ms. Soustek is currently a graduate student at the University of Florida in the Department of Pediatrics studying under the direction of Barry J. Byrne MD, PhD. Since the beginning of her graduate training, she has held positions as a NIH-sponsored Clinical Translational Science (TL1) and Hypertension (T32) trainee. Ms. Soustek's work has focused on characterizing a mouse model of Barth syndrome as well as developing a gene therapy based approach for cardiolipin deficiency.

Presentation — Endurance training in a mouse model of Barth syndrome *(Sci/Med Sessions)*

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

Dr. Steward is a Consultant in bone marrow transplantation for genetic diseases at the Bristol Royal Hospital for Children, UK. His particular interests are the management of neutropenia in Barth syndrome, fetal presentations of the disease and improving disease recognition/testing to overcome current underdiagnosis of the disease — already 28 unrelated families have been identified in the UK. He is Clinical Lead of a multidisciplinary UK national service for Barth syndrome which was established by the National Health Service in Bristol in April 2010. This service provides free diagnostic testing for UK residents by cardiolipin analysis and TAZ gene sequencing, annual multidisciplinary clinics for boys and young men, and centralized prescribing and delivery of G-CSF. It is run in close partnership with Michaela Damin and her colleagues at the Barth Syndrome Trust.

Presentations
The first two years of the UK national service for Barth syndrome: Triumphs and tribulations *(Sci/Med Sessions)*
Managing neutropenia in Barth syndrome *(Family Sessions)*
Note — Dr. Steward is also involved in the Consultations

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 & Repository, 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 2006, Dr. Toth has been the Science Director of the BSF, where he uses his knowledge and experience in guiding efforts towards finding treatments and eventually a cure for Barth syndrome.

Presentation — Update on scientific research in Barth syndrome *(Family Session)*

Hilary Vernon, MD, PhD — Assistant Professor, McKusick-Nathans Institute of Genetic Medicine; Department of Medicine, Division of Pediatrics, Johns Hopkins University School of Medicine, Baltimore, MD, USA

Dr. Vernon's research interests include molecular and metabolic pathogenesis of organic acidemias, molecular and metabolic pathogenesis of Barth Syndrome, and clinical laboratory biochemical diagnosis. Her current research focuses on looking for biochemical targets for therapy in Barth Syndrome, as well as other genetic disorders involving the mitochondria.

Dr. Vernon was awarded her MD from Robert Wood Johnson Medical School, University of Medicine and Dentistry of New Jersey, 2004, PhD from Rutgers University, 2004 and BA from University of Pennsylvania, 1998. She is board certified in pediatrics, clinical genetics, and clinical laboratory biochemical genetics.

She was awarded fellowships in Biochemical Genetics, 2011, and Genetic Medicine, 2007, from Johns Hopkins University. She was awarded the Francis F. Schwentker Award for Excellence in Research, Johns Hopkins University, 2010. Dr. Vernon was Chief Resident of Medical Genetics, Institute of Genetic Medicine, Johns Hopkins University, 2010-2011. She is a member of the American Society for Human Genetics.

Note: Dr. Vernon is involved in the Consultations/Research

Gonzalo Wallis, MD — Clinical Assistant Professor, Congenital Heart Center, University of Florida College of Medicine, Gainesville, FL, USA

Dr. Wallis's specialty include pediatric cardiology, pediatric heart failure and pediatric heart transplantation, his focus is in the management of heart failure in the myopathic heart, and the pre- and post-heart transplantation management. Dr. Wallis has a special interest of research in ABO incompatible heart transplantation in infants.

Dr. Wallis received his MD from the Central University of Venezuela, his pediatric training at the University of Texas Medical Branch at Austin Texas, and his pediatric cardiology subspecialty training at the University of Florida. He is board certified in Pediatrics.

Presentation — Cardiac aspects of Barth syndrome *(Family Sessions)*

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 using a new lipidomics platform to investigate lipid abnormalities in Barth syndrome and in other research areas. Dr. Vaz was awarded two research grants from BSF entitled "Identification of the Proteins Interacting with Tafazzin and Resolution of the Consequences of the Deficiency of Cardiolipin at the Protein Level" (2006); "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 continues his research on Barth syndrome in collaboration with Dr. Willem Kulik.

Presentation — Application of lipidomics to identify new phospholipid disorders *(Sci/Med Session)*

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The Barth Syndrome Foundation (BSF) is honored to have the National Heart, Lung and Blood Institute (NHLBI) and the NIH Office of Rare Diseases Research (ORDR) of the National Institutes of Health (NIH) as sponsors of BSF's 2012 International Scientific & Medical Conference.