



Barth Syndrome
Foundation



**POWER
UP!**

Clearwater Beach, FL ~ July 2018

**Externally-Led Patient-Focused Drug Development
Meeting for Barth Syndrome (BTHS)**

PUBLIC MEETING / JULY 18, 2018

MEETING AGENDA

11:00am—12:00pm **Lunch** will be provided in Grand Ballroom Foyer

12:00pm—12:30pm **Registration**

Opening Remarks and Overviews

12:30pm—12:35pm **Welcome**
Emily Milligan, MPH; Executive Director, Barth Syndrome Foundation

12:35pm—12:45pm **Opening Remarks**
Celia M. Witten, PhD, MD; Deputy Director, FDA Center for Biologics Evaluation and Research (CBER)

12:45pm—1:00pm **Clinical Overview of Barth Syndrome (BTSH)**
Colin Steward, PhD, FRCP, FRCPC; Professor of Pediatric Stem Cell Transplantation and Consultant in Bone Marrow Transplantation, Bristol Royal Hospital for Children; Medical Advisor to NHS Barth Syndrome Clinic

1:00pm—1:15pm **Introduction to Barth Syndrome Foundation and One Family's Experience with BTSH**
Kate McCurdy; Founding BSF Board Member and Ex-officio Member, Emerita, BSF Scientific and Medical Advisory Board (SMAB)

1:15pm—1:25pm **Introduction and Overview of Meeting**
James Valentine, JD, MHS; Meeting Moderator

1:25pm—1:35pm **Audience and Remote Demographic Polling**

Session 1: BTSH Patient Voice: Symptoms and Daily Impacts

1:35pm—2:50pm **Panel 1: Symptoms and Daily Impacts**

- Presentations by five affected individuals and caregivers (30 minutes)
- Audience & remote polling (10 minutes)
- Moderated audience discussion (35 minutes)

2:50pm—3:05pm **Break**

3:05pm—3:15pm **FDA Comments**
Scott K. Winiecki, MD; Director, Safe Use Initiative, FDA Center for Drug Evaluation and Research (CDER)

3:15pm—3:25pm **Video of BTSH Individual Unable to Travel**

Session 2: BTSH Patient Voice: Current and Future Approaches to Treatments

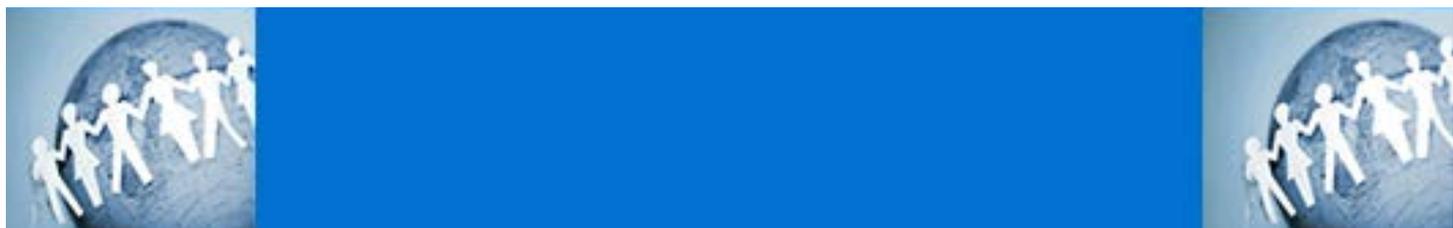
3:25pm—4:40pm **Panel 2: Current and Future Approaches to Treatments**

- Presentations by five affected individuals and caregivers (30 minutes)
- Audience & remote polling (10 minutes)
- Moderated audience discussion (35 minutes)

Summary and Closing Remarks

4:40pm—4:50pm **Closing Comments – What I Heard Today**
Shelley Bowen; Director of Family Services & Awareness, BSF

4:50pm-5:00pm **Closing Remarks**
Elizabeth Hart, MD; Medical Officer, Division of Gastroenterology and Inborn Error Products, Office of New Drugs (CDER) (10 minutes)



WELCOME LETTER

Dear PFDD Participants,

Welcome to the externally-led Patient-Focused Drug Development Meeting for Barth syndrome!

Barth Syndrome Foundation (BSF) is thrilled to have you participate in today's meeting. BSF leads the global research and advocacy effort to create a world without Barth syndrome while we provide ongoing education and community for affected individuals and their families. If you are an affected individual or family member, whether you are joining us in person or participating through our live webcast, we are grateful for your participation and for taking the time to come together as a community to voice your experiences and perspectives.

We are also excited to have influential leaders from the U.S. Food and Drug Administration (FDA), industry professionals, and researchers from academia also with us, both in person and on-line. Our organization's goal is to deliver effective therapies into the hands of affected individuals and their caregivers, to eliminate the suffering and loss of life from Barth syndrome. Your participation is important for us to foster strong ties and collaborate across the research and development continuum to achieve our mission. The fact you are involved in this PFDD meeting highlights the attention Barth syndrome requires.

And to our panelists, we especially want to thank you for exercising your voices. Today's meeting simply would not be possible without your contributions. We recognize it takes courage to share your personal stories about the impact of Barth syndrome on your lives. Your sacrifice is a statement of hope and inspiration.

We also wish to thank the speakers from the FDA: Dr. Celia M. Witten, PhD, MD, Deputy Director, FDA Center for Biologics Evaluation and Research (CBER); Dr. Scott Winiecki, MD, Director, Safe Use Initiative, FDA Center for Drug Evaluation and Research (CDER); and Dr. Elizabeth Hart, MD, Medical Officer, Division of Gastroenterology and Inborn Error Products, Office of New Drugs (CDER). In addition, we are grateful to Dr. Colin Steward, PhD, FRCP, FRCPC, Professor of Pediatric Stem Cell Transplantation and Consultant in Bone Marrow Transplantation, Bristol Royal Hospital for Children and Medical Advisor to NHS Barth Syndrome Clinic for delivering important remarks about the clinical impact of Barth syndrome. Your continued partnership is vital to paving pathways for scientific breakthroughs and novel therapies. On behalf of the community we represent, BSF thanks you for your support today and in the future as we advance therapies.

Today's meeting is about many things, but most of all it is about our shared optimism. Together we forge new collaborations. Together we chart new pathways for novel therapies. Together we not only dream about but mobilize around a world without Barth syndrome.

Sincerely,



Emily Milligan
Executive Director



Susan McCormack
Chair, BSF Board of Directors



Michael Schlame, MD
Chair, BSF Scientific and Medical
Advisory Board



ABOUT THIS EXTERNALLY-LED PATIENT-FOCUSED DRUG DEVELOPMENT MEETING

Barth Syndrome Foundation (BSF), a patient advocacy organization representing those who suffer from and care for individuals with Barth syndrome (BTHS), is holding the EL-PFDD meeting on BTHS between our community and the U.S. Food and Drug Administration. The EL-PFDD meeting for BTHS will advance BSF's mission: *Saving lives through education, advances in treatment, and finding a cure for Barth syndrome.*

Namely, the EL-PFDD meeting will enable us to share with key FDA officials and other stakeholders the burdens of BTHS across the lifespan, current unmet needs, prognosis and current standards of care. Discussion themes will include ways in which BTHS-affected individuals attempt to mitigate symptoms of the condition through cardiac medications (e.g., beta blockers, ACE inhibitors, anti-arrhythmics, anticoagulants), implantable defibrillators and heart transplantation for cardiac related issues, bone marrow stimulants (figrastim) for neutropenia, strollers and wheelchairs for reduction of fatigue and lifestyle modifications. Given the lack of a current therapy for BTHS, it is important for stakeholders to understand how patients are impacted by the condition, their

current treatment options (or lack thereof), and hear their input for future trial design and therapeutic review.

The EL-PFDD meeting is a key component of realizing these objectives, by capturing patient and caregiver insights that can set the context for FDA benefit-risk considerations. It will enable a comprehensive understanding of this rare condition for key reviewers in the FDA CDER Division of Neurology Products and Division of Gastroenterology and Inborn Errors Products, CBER Office of Tissues and Advanced Therapies, CDER Rare Diseases Program, Office of Pediatric Therapeutics, and Office of Orphan Products Development.

The EL-PFDD meeting will include panelists that represent a spectrum of perspectives in terms of age, geographic region, and severity of symptoms. We understand some BTHS individuals may be too ill to travel to this meeting and we feel it is essential to hear their perspectives as well. Therefore, BSF is producing and will present video testimonials in order to overcome this challenge.

The goals of this meeting are to:

- Collect data and discern key insights for clinical trial design from affected BTHS individuals and their caregivers so that the outcomes of potential therapeutics can be measured in ways that are both clinically sound and therapeutically impactful.
- Develop and provide the FDA with a robust understanding of patients' and caregivers' experiences with Barth syndrome. This would include BTHS individuals' views on their quality of life, aspects of the disease that are most problematic for them, and actions they currently perform to cope with this disease.



Instructions for Polling Questions

Each session in today's meeting will include a series of polling questions on Barth syndrome (BTHS) and its impact on your family's life. In-person attendees are encouraged to use their mobile phones or computer to participate in these polling questions. **Note: Please do not cast a response through the poll platform if you are not an adult who has been diagnosed with BTHS or the parent designee representing a minor or deceased individual. No more than one vote should be cast for each individual who has BTHS.**

If texting, download the Poll Everywhere App (on Google Play or Apple App Store) to your smart phone prior to the meeting and create your account. When the event is taking place you will need to enter powerup1.

If using your ipad, tablet or computer, visit [PollEv.com/powerup1](https://www.poll-everywhere.com/powerup1) prior to the meeting and create your account.

Standard message and data rates apply.

PANELIST BIOS



Kevin B.

Kevin is a 29-year-old affected individual who lives in Pennsylvania. He received his Barth syndrome diagnosis at the age of seven. He has worked part-time as the Grants & Scholarships Administrator at the Chester County Community Foundation since 2016. His hobbies include reading, gaming, and rooting for Philadelphia sports teams.



Darryl B.

Darryl is 36 and lives in Philadelphia, PA. He was diagnosed with Barth syndrome at the age of 20. He graduated from South Philadelphia High and currently assists in the ministry at his church, St. Paul Chapel Baptist Church. Darryl served as the primary caregiver for his brother, Jamal, who passed away from Barth syndrome in 2009 at the age of 25.



Jasmine C.

Jasmine lives in British Columbia, Canada, with her husband, Mark, and sons, Jordan (4, unaffected) and Caleb (21 months, BTHS). Caleb was diagnosed with Barth syndrome at three weeks of age. Jasmine works as a first grade teacher. She enjoys attending music festivals, all sports and spending time with her friends and family.



Nicholas D.

Nicholas is 19 years old and lives in Hampshire, U.K., with his parents, Mic and Marco, and his younger brother, Matthew (16, unaffected). Nicholas was diagnosed with Barth syndrome in April 2001. He enjoys playing video games, building models, and spending time with friends. He is also a film buff.



Nicole D.

Nicole has been active with the Barth Syndrome Foundation since attending the BSF conference in 2008. Nicole is the mother of two boys born with Barth syndrome: Nathaniel, who passed away at just two weeks of age in 2002, and Devin, age 13, who underwent heart transplantation as an infant. Nicole has dedicated much of her time with BSF offering her unique perspective of raising a Barth boy after heart transplant surgery. Professionally, Nicole has worked on numerous political campaigns, ranging from local millage renewal to US Congressional campaign. She currently works as a legislative aide in the Michigan House of Representatives. She, her wife, Sarah, and Devin live just outside of Flint, Michigan.

Nicole has served on the BSF Board of Directors since 2016.

PANELIST BIOS



Peter VL.

Peter is 31 years old and lives in the Netherlands. He was diagnosed with Barth syndrome at the age of two. Peter enjoys programming a website and additional features for a Minecraft community he manages as well as watching various TV series on Netflix.



Amanda M.

Amanda is 33 years old and lives in the Pittsburgh, Pennsylvania area. Her and her husband, Tim, have two children — Sydney (age 11) and Wyatt (age 10, BTHS). She has been a part of the foundation since 2007, and they attended their first conference in 2008.



John W.

Being affected by Barth syndrome gives John a unique insight into the issues facing the Barth Syndrome Foundation. John recently earned an A.S. in Computer Information Technology from Southeast Community College in Lincoln, Nebraska, and works part time as a computer consultant. John lives in Lincoln, Nebraska.

John has served on the BSF Board of Directors since 2012 and was elected Corporate Secretary in 2018.



Jacob W.

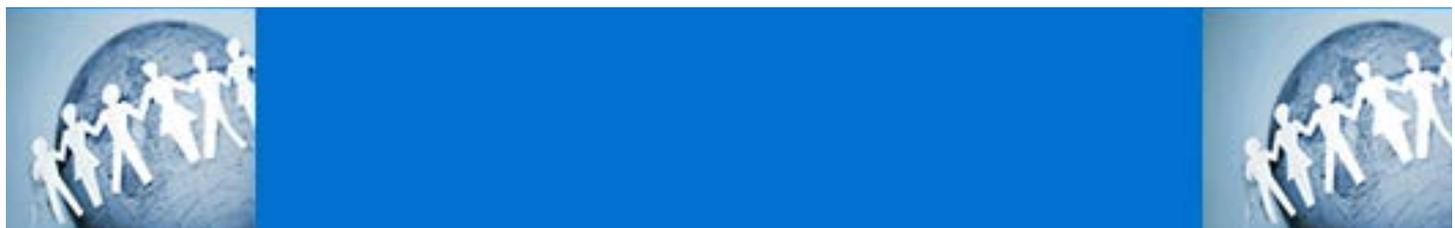
Jacob is 17 years old and recently graduated from Richland High School in Richland, MS. He and his parents searched for years for a diagnosis before Jacob was found to be affected by Barth syndrome at age 10. Jacob lives with his mother, Amy, his father, Marvin, and his brother, Damon (unaffected). His interests include hunting, fishing and spending time with friends.



Kevin W.

Kevin is the father of an affected boy named Connor. Connor was diagnosed with Barth syndrome (BTHS) when he was 18 months old. Kevin lives in Phoenix, Maryland, with his wife, Stacey, and his two sons, Connor (age 8, BTHS) and Ryan (age 5, unaffected). Kevin works as a Technology Director for T. Rowe Price. He enjoys spending time with his family, jogging, and playing and listening to music in his spare time.

Kevin has served on the BSF Board of Directors since 2014 and was elected Treasurer in 2017.



SPEAKER BIOS



Shelley Bowen ~ Director, Family Services & Awareness, BSF

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



Elizabeth Hart, MD ~ Medical Officer, Division of Gastroenterology and Inborn Error Products, FDA Center for Drug Evaluation and Research, Office of New Drugs (CDER)

Dr. Hart evaluates the efficacy and safety of new drugs and biologics intended for inborn errors of metabolism and advises companies and researchers on clinical drug development for these rare diseases. Prior to joining the FDA, Dr. Hart treated children and adolescents with a variety of serious conditions and held academic appointments at George Washington University and Harvard University. She also conducted translational and clinical research at the National Institutes of Health.

Dr. Hart received her Medical Degree from the University of Pennsylvania, and completed a pediatric residency at Rainbow Babies and Children's Hospital and a fellowship in pediatric endocrinology at Boston Children's Hospital.



Kate McCurdy ~ Member *Emerita*, Scientific & Medical Advisory Board, BSF

Mrs. McCurdy was a founding member of The Barth Syndrome Foundation's (BSF) Board of Directors and was responsible for establishing the Science and Medicine Program of BSF. She currently serves as an *Emerita* member of the Scientific & Medical Advisory Board. Mrs. McCurdy has a BA from Duke University and an MBA from the Harvard Business School. She has worked in both economics and the corporate world and has held positions on the boards of various other non-profit organizations.

Her son, Will, was affected by Barth syndrome and passed away in 2014 at the age of 28, but she, her husband, Steve, and their daughter, Eliza, continue to be strongly committed to the mission of BSF.



Emily Milligan, MPH ~ Executive Director, BSF

Ms. Milligan has spent her career dedicated to improving the lives of children and their families through scientific advancements and social equality. In May 2018, Ms. Milligan joined Barth Syndrome Foundation (BSF) as the Executive Director. Trained in public health and international relations, Ms. Milligan brings years of experience managing research portfolios and transforming business processes. Previously, she worked for the United Nations in Brazil and Nicaragua, and was a vital contributor at Columbia University and New York University. She went on to join JDRF (formerly known as the Juvenile Diabetes Research Foundation) where she headed the research operations and scientific teams and oversaw an average annual \$100 million research portfolio. Most recently prior to joining BSF, Ms. Milligan launched an \$80 million, mission-driven venture fund that invests in companies developing life-saving products for individuals living with type one diabetes. She is an active member of her community in Needham, Massachusetts, and volunteers her time for other organizations focused on social change and alleviating human suffering.

SPEAKER BIOS



Colin Steward, PhD, FRCP, FRCPCH ~ Professor of Pediatric Stem Cell Transplantation and Consultant in Bone Marrow Transplantation, Bristol Royal Hospital for Children; Medical Advisor to NHS Barth Syndrome Clinic, Bristol, United Kingdom; Scientific and Medical Advisory Board, BSF

Prof. Steward recently retired from his post as Clinical Lead for the multidisciplinary NHS National Barth Syndrome Service which was established in 2010 and is run in partnership with Michaela Damin and her colleagues at the Barth Syndrome Trust. This will allow him to concentrate on research at the University of Bristol whilst having more time for hobbies such as travel, walking, watching wildlife and gardening. His current Barth syndrome research focuses on the CARDIOMAN Trial looking at use of the drug bezafibrate in Barth syndrome together with his colleagues at Bristol Royal Hospital for Children and mechanisms of neutropenia in conjunction with Dr Borko Amulic.



James Valentine, JD, MHS ~ Associate, Hyman, Phelps & McNamara (Moderator)

Mr. Valentine assists medical product industry and patient advocacy organization clients in a wide range of regulatory matters, including new drug and biologic development and approval issues. Before joining his current firm in 2014, Mr. Valentine worked in FDA's Office of Health and Constituent Affairs where he facilitated patient input in benefit-risk decision-making and served as a liaison to stakeholders on a wide range of regulatory policy issues. There, Mr. Valentine administered the FDA Patient Representative Program, launched the Patient-Focused Drug Development program, and developed the FDA Patient Network. Mr. Valentine also worked at the Center for Drug Evaluation and Research's (CDER) Office of Regulatory Policy where he coordinated implementation of new statutory authorities.

Mr. Valentine earned his law degree from the University of Maryland and his master of health science from the Johns Hopkins School of Public Health.



Scott K. Winiecki, MD ~ Director, Safe Use Initiative, FDA Center for Drug Evaluation and Research (CDER)

After 12 years in private pediatric practice, Dr. Winiecki joined the U.S. Food and Drug Administration in 2011. In 2012, he received the FDA's "Outstanding New Reviewer" Award. After five and a half years working on biologics, he joined the Center for Drugs in September, 2016. He is currently Director of the Safe Use Initiative, a group whose goal is to reduce preventable harm from medications by collaborating with both public and private groups within the healthcare community.

Dr. Winiecki received his MD degree from the University of Maryland and completed his pediatric training at the Children's Hospital of Philadelphia.



Celia M. Witten, PhD, MD ~ Deputy Director, FDA Center for Biologics Evaluation and Research, FDA Drug Administration (CBER)

Between 2005 and 2016, Dr. Witten served as the Director of the Office of Cellular, Tissue and Gene Therapy at the FDA/CBER. Between 1996 and 2005, she served as Director of the Division of General, Restorative, and Neurological Devices in the Office of Device Evaluation in the Center for Devices and Radiological Health (CDRH). Previous to FDA, Dr. Witten worked for over 10 years as a practicing physician at the National Rehabilitation Hospital (NRH) in Washington, D.C.

Her educational background includes a BA earned at Princeton University (Magna Cum Laude), a PhD from Stanford University, and an MD from the University of Miami School of Medicine. In addition to her academic achievements she is Board Certified in Physical Medicine and Rehabilitation.

BTHS PATIENT FOCUSED DRUG DEVELOPMENT — TOPIC QUESTIONS

Topic 1 Questions: Symptoms and Daily Impacts

1. Of all the symptoms that you experience because of your condition, which 1-3 symptoms have the most significant impact on your life? (Examples may include: heart problems, feeding issues, fatigue, pain, infection)
2. Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of your condition? (Examples of activities may include: go to school, participate in sports, hold a job, keep up with friends)
 - a. How do your symptoms and their negative impacts affect your daily life on the best days? On the worst days?
3. How have your condition and its symptoms changed over time?
 - a. Do your symptoms come and go? If so, do you know of anything that makes your symptoms better? Worse?
4. What worries you most about your condition?

Topic 2 Questions: Current and Future Approaches to Treatments

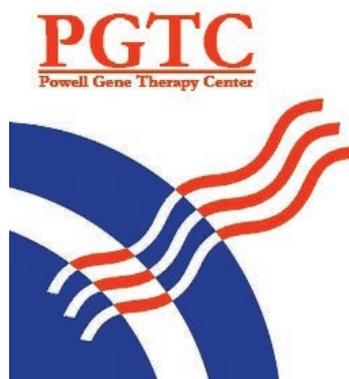
1. What are you currently doing to help treat Barth syndrome or its symptoms? (Examples may include prescription medicines, over-the-counter products, other therapies including nondrug therapies such as exercise, etc.)
 - a. What specific symptoms do your treatments address?
 - b. How has your treatment regimen changed over time, and why?
2. How well does your current treatment regimen treat the most significant symptoms of your disease?
 - a. How well do these treatments improve your ability to do specific activities that are important to you in your daily life?
 - b. How well have these treatments worked for you as your condition has changed over time?
3. What are the most significant downsides to your current treatments, and how do they affect your daily life? (Examples may include bothersome side effects, going to the hospital for treatment, restrictions on driving, etc.)
4. What specific things would you look for in an ideal treatment for Barth syndrome?



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Friends & Family of John Wilkins





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