



Energy *in Action*

MITOCHONDRIAL DISEASE EXTERNALLY-LED
PATIENT-FOCUSED DRUG DEVELOPMENT MEETING

MARCH 29, 2019 | HYATTSVILLE, MD



UNITED
MITOCHONDRIAL
DISEASE
FOUNDATION.



Muscular
Dystrophy
Association



2 ENERGY IN ACTION

7:30 am – 8:30 am Registration and Continental Breakfast

Opening Remarks and Morning Overviews

8:30 am – 8:45 am **Welcome and Opening Remarks**
Brian Harman, President and CEO, United Mitochondrial Disease Foundation

8:45 am – 9:00 am **Clinical Overview of Mitochondrial Myopathy in Adults**
Michio Hirano, MD, Columbia University, New York, NY

9:00 am – 9:15 am **Introduction, Overview of Meeting, and Audience and Remote Demographic Polling for Adults with Mitochondrial Myopathies**
James Valentine, JD, MHS, Meeting Moderator

Morning Session: Mitochondrial Myopathy - Adult Patient Perspective on the Burdens of the Disease and Current and Future Approaches to Treatments

9:15 am – 10:25 am **Panel #1 – Symptoms and Daily Impacts**

- Presentations by 5 Affected Individuals and Caregivers (25 minutes)
- Audience and Remote Polling Panel #1 (10 minutes)
- Moderated Audience Discussion Panel #1 (35 minutes)

10:25 am – 10:35 am Break (refreshments provided)

10:35 am – 11:45 am **Panel #2 – Current and Future Approaches to Treatments**

- Presentations by 5 Affected Individuals and Caregivers (25 minutes)
- Audience and Remote Polling Panel #2 (10 minutes)
- Moderated Audience Discussion Panel #2 (35 minutes)

11:45 am – 12:00 pm **Morning Session Closing Remarks**
*Lucas Kempf, MD, U.S. Food and Drug Administration (FDA)
Center for Drug Evaluation and Research (CDER), Rare Diseases Program*

12:00 pm – 1:00 pm Lunch Break

Afternoon Session: Neurologic Manifestations in Children with Mitochondrial Disease – Patient Perspective on the Burdens of the Disease and Current and Future Approaches to Treatments

1:00 pm – 1:05 pm **Afternoon Welcome and Overview**
Kira Mann, CEO, MitoAction, and Brittany Hernandez, Director of Advocacy, Muscular Dystrophy Association (MDA)

1:05 pm – 1:20 pm **Clinical Overview: Neurologic Manifestations in Children with Mitochondrial Disease**
Amy Goldstein, MD, Children's Hospital of Philadelphia, PA

1:20 pm – 1:30 pm **Audience and Remote Polling – Attendee Demographics for Children with Neurologic Issues**
James Valentine, JD, MHS, Facilitator

1:30 pm – 2:40 pm **Panel #3 – Symptoms and Daily Impacts**

- Presentations by 5 Parents and Caregivers (25 minutes)
- Audience and Remote Polling Panel #3 (10 minutes)
- Moderated Audience Discussion Panel #3 (35 minutes)

2:40 pm – 2:50 pm Break

2:50 pm – 4:00 pm **Panel #4 – Current and Future Approaches to Treatment**

- Presentations by 5 Parents and Caregivers (25 minutes)
- Audience and Remote Polling Panel #4 (10 minutes)
- Moderated Audience Discussion Panel #4 (35 minutes)

4:00 pm – 4:15 pm **Afternoon Session Closing Remarks**
Larissa Lapteva, MD, MHS, MBA, U.S. Food and Drug Administration (FDA)

4:15 pm – 4:45 pm **Next Steps and Closing Remarks**
Philip Yeske, PhD, UMDF Science and Alliance Officer, and Brent Fields, UMDF Trustee Chair

WELCOME

DEAR EI-PFDD Participants,

On behalf of the United Mitochondrial Disease Foundation, MitoAction and the Muscular Dystrophy Association, we welcome you to this very important Externally-led Patient-Focused Drug Development Meeting for mitochondrial disease.

Today's meeting is the culmination of more than a year of planning and collaboration by the patient advocacy groups facilitating this meeting. For our patient community, this is our opportunity to come together to voice our experiences with mitochondrial disease and to showcase our perspectives on living with the disease. Whether you are joining us in person or are participating in this meeting online, thank you for taking the time to join us today and putting your Energy in Action.

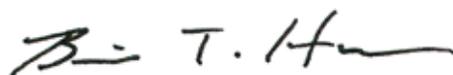
The goal of this important meeting is to set the groundwork toward creating effective therapies for mitochondrial disease. Your participation helps us in our work of creating the strong ties and collaboration that helps us fulfill our collective mission of finding treatments and a cure.

We are grateful to those who serve on our panel today. They've traveled to this meeting to share their stories and, most importantly, the voice of the patient. To share their struggles, their hopes and their concerns takes great courage, and they deserve our respect and thanks. As the patient voice, our panelists are our hope and our inspiration.

Joining us today, both in person and online, are influential guests from the U.S. Food and Drug Administration, industry professionals, clinicians, and researchers. Your involvement and collaboration in this meeting is vital in paving the path for scientific breakthroughs and potential therapies. On behalf of our patient community, their families and caregivers, thank you for your commitment to all of us.

As patient advocacy groups, we are delighted to facilitate this EI-PFDD meeting. We will hear and see much today. Let us take away a renewed sense of optimism that may lead us towards new and innovative therapies, treatments and a world without mitochondrial disease.

Sincerely,



Brian Harman
President & CEO,
United Mitochondrial Disease Foundation

THE PATIENT VOICE

Externally-led Patient-Focused Drug Development Meetings and the Patient Voice

The Food and Drug Administration (FDA) is the government agency tasked with (among other things) assuring that drugs, vaccines, biological products and medical devices intended for human use are safe and effective. In an effort to more systematically obtain the patient perspective on specific diseases and their treatments, the FDA conducted 24 disease-specific patient-focused drug development (PFDD) meetings between 2012 and 2017 that help the FDA understand the context in which regulatory decisions are made for new drugs. PFDD meetings provide key stakeholders, including the FDA, patient advocates, researchers, drug developers, healthcare providers, and others, an opportunity to hear the patient's voice. Recognizing that there are many more disease areas to be addressed beyond the PFDD meetings conducted by the FDA than for which there are resources available, the Externally-led Patient-Focused Drug Development Meeting (EI-PFDD) was launched using the process established by the PFDD meetings. In contrast to PFDD meetings, EI-PFDD meetings are organized, led and paid for by patient organizations and the FDA personnel attend on a case-by-case basis. As new drug applications are filed by drug developers, the comprehensive Voice of the Patient report generated after the meeting is a critical additional resource for the FDA beyond the mandatory safety and efficacy data.

In February 2018, the United Mitochondrial Disease Foundation (UMDF) applied to the FDA to host, in partnership with patient organizations the Muscular Dystrophy Association and MitoAction, an EI-PFDD meeting focused on mitochondrial disease; two months later they received approval from the agency to conduct the meeting in Hyattsville, MD on March 29, 2019. The EI-PFDD meeting will enable the mitochondrial disease community to share with key FDA officials and other stakeholders the burdens of the disease and perspective on future idealized treatments. Importantly, the perspective of two distinct sub-populations of mitochondrial disease patients will be explored and captured - pediatric patients with neurological manifestations and adults with myopathies. The EI-PFDD meeting will include panelists that represent a spectrum of perspectives, including age, geographic region, affected adults and caregivers of pediatric patients.

The goals of this meeting are:

- Provide broad mitochondrial disease patient perspective to the FDA by presenting testimony, discussing key topics and identifying treatment priorities.
- Capture patient-provided data that will help to inform the design of future clinical trials with respect to outcome measures that are meaningful to those affected by mitochondrial disease.

HOW TO

Each session in today's meeting will include a series of polling questions on mitochondrial disease and its impact on your family's life. In-person and remote attendees are encouraged to use their mobile devices or computer to participate in these polling questions.



- **On a MOBILE DEVICE or SMART PHONE**

Download the **Poll Everywhere App** (on Google Play or Apple App Store) to your device (smart phone) prior to the meeting and create your account. When the event is taking place, you will need to enter **EnergyInAction**. For those wishing to text responses without downloading the app, you will text **EnergyInAction** to 22333 once to join. We encourage everyone to use the mobile apps rather than texting.

- **On a TABLET, iPad or COMPUTER**

Visit PollEv.com/EnergyInAction prior to the meeting and create your account.

Note: Please do not cast a response through the poll platform if you are not an adult who has been diagnosed with Mitochondrial Myopathy or the parent designee representing a child who has a Neurologic Manifestation with Mitochondrial Disease or a deceased individual. No more than one vote should be cast for each individual who has Mitochondrial Myopathy or Neurologic Manifestation of Mitochondrial Disease.

Standard message and data rates apply.

DISCUSSION QUESTIONS

MORNING SESSIONS

Topic 1 – for Mitochondrial Disease Affected Patients with Myopathies – Symptoms and Daily Impact

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 include: muscle weakness, fatigue, exercise intolerance, speech problems, eye muscle problems, pain, etc.)*
2. Are there specific activities that are important to you but you cannot do at all or as fully as you would like because of your condition? *(Examples of activities include: social activities, working, caring for family, driving, hobbies, etc.)*
 - a. How do your symptoms and their negative impacts affect your daily life on the best days? On the worst days?
3. How has 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 – for Mitochondrial Disease Affected Patients with Myopathies – Current and Future Approaches to Treatment

1. What are you currently doing to help treat your condition or its symptoms? *(Examples include: prescription medicines, over-the-counter products, and other therapies including nondrug therapies like physical therapy, diet/nutrition, exercise, adaptive devices, 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 of downsides include: bothersome side effects, going to the hospital for treatment, restrictions on driving, etc., etc.)*
4. Short of a cure for your mitochondrial disease, what specific things would you look for in an ideal treatment for your condition?

AFTERNOON SESSIONS

Topic 1 - Pediatric Mitochondrial Disease Patients with Neurologic Manifestations – Symptoms and Daily Impact

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 include: muscle weakness, fatigue, exercise intolerance, speech problems, eye muscle problems, pain, etc.)*
2. Are there specific activities that are important to you but you cannot do at all or as fully as you would like because of your condition? *(Examples of activities include: participating in sports and recreational activities, speaking with others and being understood, social interaction, going to school, eating without help, etc.)*
 - a. How do your symptoms and their negative impacts affect your daily life on the best days? On the worst days?
3. How has 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 - Pediatric Mitochondrial Disease Patients with Neurologic Manifestations – Current and Future Approaches to Treatment

1. What are you currently doing to help treat your condition or its symptoms? *(Examples include: prescription medicines, over-the-counter products, and other therapies including nondrug therapies such as physical therapy, diet/nutrition, exercise, adaptive devices, 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 of downsides include: bothersome side effects, going to the hospital for treatment, etc., etc.)*
4. Short of a cure for your mitochondrial disease, what specific things would you look for in an ideal treatment for your condition?

SPEAKERS



BRENT FIELDS

Brent Fields is chairman of the UMDF. Brent and his family live in Austin, Texas, where he works as the CEO of Big Brothers Big Sisters (BBBS), a nonprofit mentoring organization that serves over 1,000 youth. Prior to this, he was an Administrator in the healthcare arena and then a Vice President with the American Heart Association. He has over 30 years of executive leadership experience in various industries ranging from wellness, education, healthcare, research and social services. His educational background includes a Bachelor's Degree in Communications, a Master's Degree in Education, a Clinical Residency, and a Certification in Health Promotion Management. Brent is currently in a PhD program at The University of Texas at Austin. In his 10 years at BBBS, he's led the organization to a position of national recognition, including the honor of National Board of Directors of the Year.

Brent and his wife, Suzette, have actively supported local UMDF efforts in the greater Austin area the past eight years, including their involvement in the local Energy for Life Walk. They have three children, and their youngest, Chloe, has a mitochondrial disease.



AMY GOLDSTEIN, MD

Dr. Amy Goldstein received her bachelor's degree in biology and psychology from Syracuse University and her MD from the University of Pittsburgh School of Medicine where she also served for many years as a pediatric neurologist in the children's hospital and as director of neurogenetics and metabolism at the Center for Rare Disease Therapy. Dr. Goldstein is currently the clinical director of the Mitochondrial Medicine Frontier Program, an associate professor of clinical pediatrics, and an attending physician in the Division of Human Genetics at the Children's Hospital of Philadelphia. She is a past president of the Mitochondrial Medicine Society, past director of the North American Mitochondrial Disease Consortium, and a longtime member of the UMDF Scientific and Medical Advisory Board.



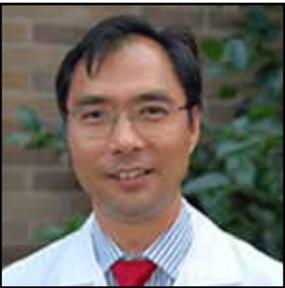
BRIAN T. HARMAN

Brian Harman is President & CEO of the United Mitochondrial Disease Foundation. Harman has served as Senior Director, Corporate Engagement and Community Partnerships, at Children's Hospital of Pittsburgh Foundation in Pittsburgh. Prior to that, he spent nearly a decade as Director of Annual Giving at Nationwide Children's Hospital in Columbus, OH. Harman also served as an Executive Director for the American Cancer Society in Ohio as well as roles within development and grassroots advocacy for that organization. Harman is a graduate of Ohio University and has a degree in communications.



Brittany Johnson Hernandez serves as Director of Advocacy for the Muscular Dystrophy Association. In this role, she leads MDA's government affairs and patient advocacy work. Brittany previously handled federal affairs for the March of Dimes and spent over eight years working on health policy for Rep. Steve Cohen (TN-09).

BRITTANY HERNANDEZ



Dr. Michio Hirano is a Professor of Neurology at Columbia University in New York; Director of the H. Houston Merritt Clinical Research Center; Chief of the Neuromuscular Division; and Associate Director of the Adult Muscular Dystrophy Association Neuromuscular Clinic. Dr. Hirano also directs a molecular neurogenetics research laboratory in the H. Houston Merritt Clinical Research Center and is the Medical Director of the Laboratory of Molecular Neurogenetics. He is a Principal Investigator of the North American Mitochondrial Disease Consortium (NAMDC).

MICHIO HIRANO, MD



Dr. Lucas Kempf is the Acting Associate Director for the Rare Disease program in the Office of New Drugs (OND) immediate office. Prior to joining FDA in 2012, Kempf spent eight years at the NIH with a focus on neuroscience research, working to understand the genetics of neuropsychiatric disease and developing translational approaches and therapeutics for these disorders. Lucas was trained at UC-Berkeley in molecular biology and genetics, received his MD degree from the University of Kansas, and did his post-graduate training in psychiatry at Georgetown and Johns Hopkins before moving to the NIH for fellowship.

LUCAS KEMPF, MD



Dr. Larissa Lapteva is the Associate Director in the Division of Clinical Evaluation, Pharmacology, and Toxicology, Office of Tissues and Advanced Therapies, Center for Biologics Evaluation and Research, Food and Drug Administration. Dr. Lapteva is a board-certified rheumatologist experienced in clinical research with novel drugs and biological products. Prior to her work at FDA, Dr. Lapteva served as a clinical investigator in trials conducted at the National Institutes of Health (NIH). Since joining FDA in 2006, Dr. Lapteva has provided scientific and regulatory advice for clinical development programs with investigational new drugs, generic drugs and biological products in various therapeutic areas, including programs for products developed for the treatment of rare diseases. Dr. Lapteva received her degrees of Master of Health Sciences from Duke University and Master of Business Administration from R.H. Smith School of Business.

LARISSA LAPTEVA, MD, MHS, MBA



KIRA MANN

Kira Mann is the CEO of MitoAction, a Boston-based patient advocacy organization whose mission is to improve the quality of life for children, adults and families living with mitochondrial disease through support, education, outreach, advocacy, and clinical research initiatives. With over 25 years of nonprofit experience, Kira has championed growth within various patient advocacy organizations, including the Alzheimer's Association, Cutaneous Lymphoma Foundation and Inheritance of Hope. Her passion lies in advancing care and support for those facing the challenges of rare disease.



JAMES VALENTINE

James Valentine received a Master of Health Science in health policy from Johns Hopkins Bloomberg School of Public Health and a JD in health law from the University of Maryland Francis King Carey School of Law. For six years, he worked at what is now the FDA Office of Health and Constituent Affairs, where he launched the Patient-Focused Drug Development program and developed the FDA Patient Network. For the past four years, Mr. Valentine has been an associate at the Washington, DC, law firm of Hyman, Phelps & McNamara, where he 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. Mr. Valentine also works with clients on clinical trials operations and compliance matters.



PHILIP YESKE, PHD

Dr. Philip Yeske received a bachelor's degree in chemistry from Allegheny College and a doctorate in organic chemistry from Emory University. He served as President and CEO of Fluorous Technologies, a Pittsburgh-based, early-stage life sciences company. He also worked at Bayer Corporation in positions ranging from basic research to global account management and co-founded the drug discovery company, MS2 Array. Dr. Yeske has been active in the mitochondrial disease community for over 15 years, first as a parent of an affected child, then as a Trustee of the United Mitochondrial Disease Foundation. Since 2013, he has been the UMDF Science & Alliance Officer, responsible for managing all scientific and business development efforts of the foundation related to improved diagnoses, development of treatments and cures, and optimized patient care.

PANELISTS

PANEL 1

LAURA P

Laura is the mother of four children; the oldest was diagnosed with mitochondrial disease in 2000. All four children have autoimmune disorders but have grown and thrived despite their challenges. She champions the rights of patients and children who fight rare diseases and learning disabilities. Laura is a technical writer and has served on the board of the Louisiana Youth Orchestra, enjoys music, kayaking, outdoor activities and family.



DEBBIE P

Debbie is 49 years old and was diagnosed with mitochondrial disease in 2011 after decades of medical issues. She and her husband, David, have two children – Brandon (19, autoimmune disease) and Katie (17, mitochondrial disease). Debbie worked in the area of special education and transition after obtaining her undergraduate degree in Educational Psychology and graduate degree in Rehabilitation Counseling. She is involved with advocacy, enjoys crafting and volunteering with her family.



DEVIN S

Devin is 26 years old and is a genetic counselor in Las Vegas, Nevada. She was diagnosed with mitochondrial DNA depletion syndrome at 16 following the diagnosis of her older brother. Devin has volunteered with the UMDF since 2011 and enjoys running an online support group for teens and young adults with “mito.” She loves the opportunity to become friends with so many from around the U.S.



ALYSSA D

Alyssa is a high school senior and was diagnosed with mitochondrial disease at 14 years old. She has been an ambassador with the UMDF for almost three years. Life with Mito is difficult and each day is a challenge, but it doesn't stop her. Advocating is her passion and she will continue until a cure is found. Alyssa hopes one day become a pediatric nurse and help patients like she's been helped over the years.



RACHEL S

Rachel has been married to her best friend Daniel for 16 years, and together they are raising their two children while also fighting against mitochondrial disease. Rachel's life verse is, “My flesh and my heart may fail, but God is the strength of my heart and my portion forever.” She and her family make their home in the DC/MD/VA region.



PANEL 2

LUISA & ROBERT M

Luisa is a practicing physician who, as an adult, was diagnosed with TK2 Mitochondrial Depletion Syndrome. Luisa and her husband of 32 years, Robert, are the parents of two healthy adult children. They enjoy the simple pleasures of living in rural Rhode Island surrounded by farmland and waterways.



SHARON S

Sharon is 54 years old and lives in Tucson, Arizona. Since being diagnosed in 2000 with mitochondrial disease, she has turned her life's passion into "being of service" for the United Mitochondrial Disease Foundation. Being a part of the solution to advance diagnostics and therapies for this debilitating disease has helped her stay mentally positive and carry hope.



DEBORAH C

Deborah, 47, was diagnosed at the age of 31 with a mitochondrial disease after years of a plethora of illnesses and a multitude of diagnostic testing. She resides in a rural community, Neoga, IL, with her son, Alex, 20, and her fiancé, Derek, and her two fur babies, Maggie and Brynnie. She enjoys medical and legal advocacy and spending time with family.



NICOLE D

Nicole lives in Lafayette, Louisiana with her husband Paul, and her daughter Lillian (16). Nicole earned a Master's degree and was employed for 14 years as an Early Interventionist working with infants and toddlers with developmental delays. She is currently serving as the Chair for the Board of Directors for Families Helping Families of Acadiana. Nicole spends most of her time caring for and homeschooling Lillian, who has also been diagnosed with an unspecified mitochondrial disease and multiple other rare conditions. Nicole's family has been on the road to diagnosis, treatment and management for 16 years and the impact has been both positive and negative. Disability advocacy efforts have taken the family to their state capital to testify in front of legislators to Washington DC to protest cuts and caps to Medicaid, and has provided numerous opportunities to speak to special educators, legislators, university students, and parents about the dynamic life of a family raising a medically complex child.



MICHAEL M

Michael is 44 years old and lives in the Boston, Massachusetts area. He earned a B.S. in Sociology from the University of South Florida and worked 13 years in law enforcement. Michael has been diagnosed with a Mitochondrial and a Fatty Acid Oxidation Disorder. In his spare time, he likes to exercise, spend time with his niece and nephew and is an avid football and baseball fan.



PANEL 3

DANIEL M

Danny is the father of two boys, ages 7 and 6, that were diagnosed in 2017 with MEPAN Syndrome - an ultra-rare mitochondrial disease caused by mutations in the MECP2 gene. MEPAN is a neurodegenerative condition that impairs mitochondrial fatty acid synthesis and presents with dystonia, ataxia and optic atrophy. The four-year diagnostic odyssey that Daniel's family endured led Danny to become professionally involved in rare disease communications and advocacy, and he also founded MEPAN.org, which is dedicated to creating more awareness about MEPAN Syndrome. Daniel's family lives in the Bay Area just over the Golden Gate Bridge.



HEATHER T

Heather is 31 years old and lives in the greater Orlando area. She and her husband, Dale, have two beautiful daughters - Brynn (4) and Arden (6), who is affected with Leigh Syndrome. Heather works as a special education Teacher where Arden attends school. Heather became a Leigh Syndrome Support Ambassador for UMDF in 2016 and founded the Leigh Syndrome Parent Network Support Group.



ANN K

Ann is from Mt. Laurel, NJ, and lives with her husband, Howard, and daughter, Mara (23), who has Pyruvate Dehydrogenase Deficiency. Their older daughter Dana lives in Washington, DC, Ann is a retired director of sales and marketing in the laboratory industry, and Howard is a retired manager of contracts in the transportation industry. Ann and her family have been involved with the UMDF for over 20 years. They enjoy family dinners, entertaining and life with Mara.



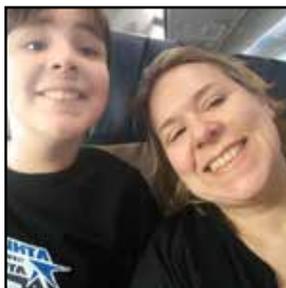
ANNETT C

Annett is the mother of Jagger (8) who was diagnosed with Leigh Syndrome, a severe form of mitochondrial disease, at the age of one. Annett lives in Atlanta, Georgia, with her husband Sebastien and son Jagger. In addition to being Jagger's primary caretaker - which includes not just being a mommy but serving the role of a nurse, hairdresser, teacher, occupational and physical therapist - Annett also works full time as a Health Scientist at the Centers for Disease Control and Prevention (CDC) focusing on malaria treatment and prevention in sub-Saharan Africa and the Greater Mekong subregion. When not busy working, cuddling and taking care of Jagger or making fingerprint art with him, she enjoys hiking, paddle boarding, kayaking, live music, and her husband's cooking.



STACY T

Stacy lives in Baltimore, MD, with her husband David and their four boys: Lucas, Benjamin, Marshall and Sam. Marshall and Sam have genetically confirmed mitochondrial disease caused by a mutation in their nuclear DNA. Stacy is a UMDF Ambassador and is trying to get an active group of affected adults and children together for regular events in the Baltimore metropolitan area. Stacy enjoys spending time with her kids.... and spending time away from her kids.



PANEL 4

CARRIE M

Carrie is a resident of Pittsgrove, NJ, and proud mother to two amazing boys, AJ (14) and Patrick (11). Her youngest son, Patrick, was diagnosed at age 4 with POLG-1 Mitochondrial Depletion Syndrome. Since Patrick's diagnosis, Carrie has immersed herself in fundraising, advocacy and leadership roles in support of the UMDF and CHOP's Mitochondrial Medicine Center. She serves as the Chair for the Delaware Valley UMDF Energy for Life Walk and is currently in her fifth year serving as part of her local school board. Professionally, Carrie has worked in Human Resources for over 20 years and is currently the Sr. Diversity Specialist for a large pharmaceutical services company.



GWEN L

Gwen Lopez-Cohen lives in Westport, CT, with her husband, Brett, and their five children. Gwen is a child and adolescent psychiatrist in private practice. In her free time, she enjoys reading fiction, traveling with her family, and teaching medical students and residents. Brett and Gwen have become very involved in supporting mitochondrial disease research since their youngest son Joshua was diagnosed with a DLD deficiency in 2016. Joshua is now five years old and has had only mild disease progression since his diagnosis. Gwen and Brett have worked with Joshua's doctor Marni Falk at the Children's hospital of Philadelphia to establish a DLD deficiency research lab with animal models of Joshua's specific genetic condition. They are hopeful that as science advances both targeted treatments and possibly even a cure for mitochondrial disease will be developed.



LORI M

Lori is a resident of Houston, Texas. She lives there with her husband and two amazing children - Will (9) and Quinn (5). Nearly eight years ago, Will was diagnosed with Leigh Syndrome at age 2. Leigh Syndrome is a progressive and fatal form of mitochondrial disease.

This life changing diagnosis pushed her family into an unknown world of medical terminology, heartbreak and the constant goal to enjoy their lives together. In between therapy, doctors appointments, hospital visits and carpool, she is the director of a Montessori pre-school and works as an advocate for her son and Leigh Syndrome.



ANNE T

Anne is mother to Bryan (27), who was diagnosed at age four with Leigh Syndrome. Anne has held various volunteer positions in the United Mitochondrial Disease Foundation's DC/VA/Baltimore chapter, serving as Vice President and led the chapter's fundraising efforts. Since retiring from her career of 33 years at the US Department of Education, Anne started a nonprofit to provide meaningful work for young adults with disabilities. She lives in Alexandria, Virginia, with her husband, Robert, and their son, Bryan. Anne and Bob are also proud Army parents as their younger son, Patrick, was recently commissioned as a Second Lt. in the United States Army.



CHERYL P

Cheryl and her husband have five children and six grandchildren and live in Atlanta, GA. Her son, David, has Leigh Syndrome, Surf 1 mutation, which makes him quite rare at 32 years old. Cheryl has been involved with the UMDF since David's diagnosis 10 years ago. She and her family have helped lead the Energy for Life Walk in Atlanta for seven years.



SPONSORS

Support for the Mitochondrial Disease Externally-led Patient-Focused Drug Development meeting is generously provided in part by:



EDITH L. TREES CHARITABLE TRUST



PhRMA

NEUROVIVE

B-MOGEN

EVERYLIFE FOUNDATION

NOTES

REGISTER Now!



Mitochondrial Medicine 2019: Washington DC

Scientific Program: June 26 - 29, 2019

Family Program: June 28 - 30, 2019

UMDF "Day on the Hill": Thursday, June 27, 2019

Hilton Alexandria Mark Center
5000 Seminary Road | Alexandria, VA 22311

www.umdf.org/symposium

