Summary – 2017 Mito Town Hall

Cristy Balcells, RN MSN

Happy New Year! This is MitoAction's seventh Town Meeting. "I'm so excited to welcome so many exciting and passionate individuals representing biopharma, individual organizations, clinicians, physicians, researchers, and people trying to make a difference collectively in the world of mitochondrial disease and in the landscape of mitochondrial medicine," said Cristy, whose daughter Eva has Leigh's disease. ("I feel blessed she's with us. Every year is an exciting opportunity to see her grow.")

When MitoAction started the Town Meeting, the idea was to have everybody gather at the same time from different regions of the country. "Now we're hearing from organizations all over the world and seeing how much growth, excitement, and interest there is in the development of mitochondrial therapeutics and in advancing progress for making a difference in the lives of the people, the children, adults and families, who actually live with mitochondrial disease!"

This call is posted on MitoAction's iTunes Podcast Library. Please check out the library, which contains more than 100 free presentations on topics that are relevant and practical for Mito families.

Kira Mann, CEO, MitoAction

"It's truly an honor for me to be part of the Mito community and I look forward to working closely with each of you to continue to build upon the amazing work of Cristy and the MitoAction team," said Kira, who comes to MitoAction with more than 25 years of nonprofit experience and has always had a passion for working with patient advocacy organizations. Prior to MitoAction, Kira, worked with the Alzheimer's Association, the American Diabetes Association, and the Cutaneous Lymphoma Foundation. Kira is excited to share her experience with the organization and the community and to help provide Mito families with the support needed to continue to expand MitoAction's

programs and services. She looks forward to engaging with members of the community to better understand the needs and challenges Mito families face each day so MitoAction can continue to respond with the personalized programs to ease the burdens. "… Families have always been at center of MitoAction's work and that will always be the case," she said.

Over the past three years, MitoAction has been involved in strategic planning to take a look at the future and growth of the organization "and to make sure the organization is positioned well for the future and that we continue to make a significant impact while never losing that personalized care for the community," she said. "We are excited about the future and our ability to serve more families in more significant ways. It's an honor to join this passionate community." You can reach out to Kira at Kira@mitoaction.org.

Shawney Lamm, Director of Special Events and Fundraising, MitoAction
In 2017, MitoAction is looking for passionate volunteers to make a difference by hosting an awareness event! "This year, we want to continue to aim to smooth the path for Mito patients and families by increasing awareness across the United States about mitochondrial disease," Shawney said. These events provide the Mito community with opportunities to meet other Mito families who may live in their neighborhoods, share their stories with each other, and have fun. They also allow local sponsors, businesses, and companies to learn about the disease. "If each of us tells just one new person about the disease, that makes a huge impact," Shawney said.

MitoAction's events include:

- The Sandra K Russell Derby Day Benefit for Mito (Saturday, May 6, 2017 at the Cyclorama at the Boston Center for the Arts);
- The Energy Walk & 5K (September, Mother's Rest in South Boston, MA);
- The Matthew Harty Hockey Tournament;
- The Matthew Harty Golf Tournament;
- The Mito Mad Hatter 5K in Sacramento, CA
- And the most recently added event -- the Capes 4 Cal 5K in Seattle, WA.

One of the highlights of the Capes 4 Cal 5K was seeing a little boy named Calvin really get involved. His goal was for people to wear green capes and use them to spark questions so people will ask, "Why are you wearing that cape?" Family and friends from all over the world took their capes to national monuments, popular amusement parks, and even to other countries while posting to social media and using the hashtag #showmeyourcapes. Calvin truly wanted to teach as many people as he could about Mito.

After the event was over, Calvin's mom shared with MitoAction that he slept in his cape that night. And right before bed he said to her: "Today was so special, Mommy. There

are times I feel alone, when admitted to the hospital, when we have to be careful about germs, when I have surgeries or procedures. But today, when I … crossed the finish line, I realized I'm never really alone. There are a lot of people wearing green capes fighting the MitoMonster with me."

"This family is evidence how anyone can make a difference in their community," said Shawney.

If you are interested in hosting an event or have other ideas for events, please contact Shawney at shawney@mitoaction.org.

Ginger DeShaney, Director of Operations & Communications, MitoAction

Education/Awareness: MitoAction has many materials for patients, families, caregivers, and physicians free of charge, including awareness postcards and flyers, Top 10 resources postcard, an educational DVD that contains six informational and entertaining videos, and a postcard for physicians that outlines MitoAction's comprehensive online clinician's guide. In addition to those materials, MitoAction also offers New Patient Kits. If you're newly diagnosed and need more information, this kit is for you. And even if you've been diagnosed for a while, you'll find this comprehensive kit helpful. To receive any of these materials, email Ginger at support@mitoaction.org.

Support: A great way to get in-person support is through a Mito Patient & Family Social. MitoAction is always looking for people to host these casual gatherings. Socials allow Mito families to meet in person, share stories and resources, gain support from peers in your own community, and realize you're not alone on this journey. Hosting is super easy and Ginger will help every step of the way. A fun way to make an impact in the community by raising awareness about Mito and funds for MitoAction is by hosting a Restaurant Night. Email Ginger at support@mitoaction.org to get started on either of these events.

Summer camp: Camp Korey, Victory Junction, and Double H Ranch all offer sessions for Mito kids. These three camps offer traditional summer camp experiences for Mito kids with activities, which are all universally accessible, that run the gamut from arts & crafts and drama to rope courses, zip lines, and rock climbing -- and everything in between! Each camp has a medical staff and dietitians on staff to provide peace of mind for parents. Applications are either open or will open soon. To find up-to-date information on the camps, visit MitoAction's camp page.

MaryBeth Hollinger, RN MSN, Director of Education, Support & Advocacy,
"I turned to MitoAction nine years ago in need of information and support myself and shortly thereafter began taking calls as a Mito 411 volunteer," said MaryBeth. "As my passion to help the Mito community grew, so has my role!"

MaryBeth strives to provide emotional support to the mitochondrial disease community, direct patients to necessary resources and educational materials, teach advocacy skills, and assist patients and families with their journey through the disease process, both directly and indirectly. "I feel honored to help families when they feel lost or alone on this Mito journey."

Mito 411 offers live one-to-one support, education, advocacy, and a unique camaraderie formed through the tie to mitochondrial disease. Families can call 1-888-MITO-411 or email mito411@mitoaction.org to request support.

In 2016, Mito 411 logged more than 400 calls and emails on a variety of topics, such as: Do these symptoms look like Mito?, the diagnostic process, specific symptom management, and advocacy skills, but honestly, the callers crave the connection to someone who has been in his/her shoes, said MaryBeth. Many Mito specialists now share the Mito 411 hotline as a resource with their patients. "We never know what the next call may bring, but calls end the same way — two people finding a common ground and moving forward, together instead of alone," MaryBeth said.

Volunteers are the heart of Mito 411 and we need *you* to help! Volunteers undergo training and are offered support and resources when taking calls. Anyone with compassion and time to return calls in a timely manner can become a volunteer. Training sessions will be held in early February. Email MaryBeth at mito411@mitoaction.org to register!
MItoAction also offers toll-free teleconference support groups on Fridays at noon EST. "We glimpse insights into Mito care from all areas of the country and share how we cope and LIVE with this disease," MaryBeth said. Please click here for more information.

Cliff Gorski, Director of Communications, UMDF

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- The year 2016 was an exciting one for UMDF on the patient and family support front.
- UMDF's national office and regional coordinators fielded more than 6,000 calls and emails assisting with things such as physician referrals, how to enroll in the free genetic testing program with Courtagen, issues regarding medical supplements and how to get them covered.

- UMDF has conducted four support group meetings a month across the country.
- Educationally, UMDF, along with Dr. Amy Goldstein, created “Mito On-Call,” a program that allows physicians from anywhere across the world to ask a Mito question and have it answered within 48 hours.
- UMDF educated more than 500 doctors with its Grand Rounds program.
- UMDF also brought a Mito doctor to more than a dozen communities so that patients and families could hear the latest information about diagnosis, treatments, and potential cures.
- UMDF released a new video that features Mito doctors and families and tells the story of what it’s like to live with mitochondrial disease and the work that’s going on in the field of mitochondrial medicine.

- The year 2017 is shaping up to be another important one for UMDF.
- Roadmap to a Cure will help the community focus on ways to get faster diagnoses, therapeutic development, and the coordination of care for the patient community.
- Regarding the Mitochondrial Disease Community Registry, UMDF is starting a data mining project, with results coming soon. So far 1,500 patients, families, caregivers have signed up for the registry. The information is confidential; you control who sees it. If you haven’t signed up, please do so.
- There are 10 Energy for Life Walks coming up.
- The national UMDF Symposium, Mitochondrial Medicine, is scheduled for June 28-July 1 in Washington, DC, and will include a Day on the Hill in which you can visit your elected officials. Registration will open shortly. There are also regional symposia coming up, including St. Louis in March, Birmingham, AL, in April (in collaboration with the Foundation for Mitochondrial Medicine), and the fall in San Antonio, TX.
- UMDF has relaunched its website, so take a look!

Maria Hopfgarten and Heather Schichtel, Miracles for Mito

Miracles for Mito™ Mito moms Maria and Heather started Miracles for Mito six years ago because they needed support in the Rocky Mountain region of Colorado. “In the last six years, we’ve really seen this grow; we’ve seen a lot of great support and a lot of great need come of out of he Rocky mountain region,” Heather said.

Maria lost her Mito warrior, Jacob, this year. “Both of us are not only mothers, but bereaved mothers in the mitochondrial space who have both decided that this is really important and are going to continue supporting our patients in this area.”

In 2016, Miracles for Mito had a larger reach than in previous years. They held two large family meetings with holiday themes: Halloween and Easter. These meetings included educational pieces and fun for Mito families.

An advocacy workshop with MitoAction was held in the fall that featured Annette Hines, JD, of Special Needs Law Group of Massachusetts. She talked about advocacy around mitochondrial disease. “It was very well-received,” Maria said. “It was an honor to do this together with MitoAction.” Miracles for Mito also sponsored parents to go to the UMDF Symposium. Last year, Heather presented at the symposium.

Miracles for Mito also offers financial grants for families in the Rocky Mountain region and provides Ubiquinol for patients in the region.

In 2017, the group will continue providing support for families, financial grants, sponsorships for families to attend the UMDF conference, and Ubiquinol for patients. The annual Mito walk will be held at the end of May or early June. Check out www.miraclesformito.org for more information.
Miracles for Mito partners closely with Children’s Hospital Colorado. The organization does a ride called the Courage Classic, a 156-mile trek through the Rocky Mountain area, and it started a team called Summits for Samantha. That team is large enough to raise money for the Mito clinic in Colorado. Over the last three years, the team has raised over six figures. Last year, it raised $120,000, which went directly to the Mito clinic. “Without that money, there would be no Mito presence in Colorado,” Heather said. “We’re really proud of the work we’re doing.”

Dr. Fran Kendall, VMP Genetics

Dr. Kendall does a monthly blog during the academic year. These blogs address different aspects of mitochondrial medicine and reach more than 20,000 people in the worldwide community. Dr. Kendall continues to look for funding support for the blog, with goals of expanding it to more than one a month and bringing in outside speakers, for example nurses, nutritionists, etc. Click here to find her blog. The blog is also posted on VMP's Facebook page.

Dr. Kendall has done work with CBD and medical marijuana for the Mito community. Georgia is the only state in the US that has mitochondrial disease as a qualifying condition for CBD. Dr. Kendall wrote the first card in Georgia for a Mito patient with seizures. She has garnered a lot of experience in this area and was asked to speak last year at the Autism One conference and will do so again this year on the data they are collecting on the use of CBD for the Mito patient. “It’s quite exciting,” she said. “We’re seeing a dramatic improvement in seizure disorder control in many patients, along with other aspects of improvement to include anxiety issues and even, in some cases, skill attainment.”

She is also continuing to work with colleagues on exercise physiology data in the Mito community. Their work supports Dr. Mark Tarnopolsky's data that clearly shows exercise regimens improve Mito capacity. Dr. Kendall spends most of her time in the office taking care of patients.

For more information, check out Dr. Kendall's website: www.vmpgenetics.com.

Maureen Latocki, Executive Director, MitoCanada

MitoCanada is entering its seventh year. Maureen noted that "2017 is a milestone year for all of us. It's the 55th anniversary of the first Mito diagnosis so we're using that in a number of our marketing materials this year. We're also using it as an opportunity to collaborate with the international community on a major awareness initiative that involves lighting monuments green around the world on Sept. 16 this year."
Last year was another capacity building year for MitoCanada. It moved its corporate offices to Burlington, ON, which is more accessible to its partners, sponsors, and families. The organization continues to work lean, with a small staff of 1.6 full-time equivalency. However, it continues to build the capacity on its board of directors, who are active, involved, and generous with bringing their skills to the table. In past year, MitoCanada's board added expertise in health marketing, the health policy sector, the health-related corporate leadership sector, finance, and the legal sector. Last year, the board appointed Dr. Adeel Safdar as the chief scientific officer. He has worked alongside Dr. Mark Tarnopolsky.

MitoCanada has done investments in research. It has partnered with the Canadian Inherited Metabolic Disease Research Network on the first clinical survey of Canadian physicians who treat Mito patients, hoping to use the results as a springboard to a larger national research strategy. The organization also made its largest ever research grant to Children's Hospital of Eastern Ontario Research Institute and continues to fund its MDHP scholar program.

MitoCanada saw an important reboot of Team Mito, which is its major fundraising vehicle. Team Mito has brought in close to $1 million to enable MitoCanada to operate over the last six years. The team does extraordinary feats of strength that raise money and awareness. At last year's Calgary Marathon, Team Mito broke six Guinness World Records and partnered with Australia to break a seventh. "That has created a lot of excitement and awareness," Maureen said. This year, to mark Canada's 150th birthday, Team Mito group out of Calgary is in the planning stages for another extraordinary feat: linking 150 runners together to run the Calgary Marathon.

- MitoCanada has forged many new partnerships.
- It joined the International Mito Patients Organization, one of 12 members.
- It started a new relationship with the Building and Construction Trades in Canada in an effort to raise awareness. The trade, 10,000 members strong, is learning about mitochondrial disease. MitoCanada created a video the Trades used to

explain Mito to its constituents. MitoCanada is looking forward to expanding that relationship across the country.

- MitoCanada is proposing to use the Canadian Association of Pediatric Health Centers' knowledge exchange network for a webinar series on Mito.
- The organization has done outreach to the Toronto Hospital community and presented to the Toronto Mitochondrial Disease Research Network.
- In the year ahead, there's so much to be done. "Awareness continues to be our major priority. That's where we need to focus our energy."
- MitoCanada is working on a national PSA to be launched this year.
- On the support side, MitoCanada is creating new tools for patients and clinicians for a more robust way to connect with others. There is new technology in the works that will enable patients to navigate the system in Canada, which can be a challenge. MitoCanada recently met with a potential national sponsor to fund patient support meetings. "It's critically important that we could actually help support local meetings for patients and families."

For more information, visit http://www.mitocanada.org/.

Laura Stanley, Executive Director, Foundation for Mitochondrial Medicine

FMM is based in Atlanta and its priorities are to raise awareness and fuel connections among the related disease groups (Mito is a core element and primary or secondary cause in many other familiar diseases, such as Alzheimer's, diabetes, autism, Parkinson's, and epilepsy). The group is also funding treatments and trying to get further and faster to the cures, Laura said.

As far as research, FMM has partnered with the Alzheimer's Drug Discovery Foundation and the Michael J. Fox Foundation to co-fund research projects where mitochondrial dysfunction is a core element. This past year, FMM began a partnership with Seahorse BioScience, which was acquired by Agilent Technologies. Seahorse and the University of Alabama-Birmingham have begun work in the field of bioenergetics, where they are establishing a bioenergetic health index, which is leading to a non-invasive blood test that measures mitochondrial function. "We're exited about that and happy to partner with them and provide funding so that the blood test can become a biomarker and a non-invasive element to assess mitochondrial function," Laura said. She's hopeful that once it's certified, it will become a surrogate for muscle biopsy and other tests. Next steps for this index: Once the IRB is established for healthy subjects, they will submit IRB at UAB for collection of adult and pediatric patients for the mitochondrial BHI test. You'll be able to look for more info on the FMM website as that evolves.

FMM welcomed two new board members: Gene Williams, executive chairman of Promise Neurosciences (a biopharma company focused on Alzheimer's), and Chris Benecki, global marketing director at UCB Pharma (a Belgium pharma company focused on epilepsy and Parkinson's). "We're excited to bring them into the fold and have that outreach into some of the other related disease communities," Laura said.

FMM, partnering with UMDF, is looking forward to the Southeast Regional Symposiumon April 7-8. Laura is excited to bring this CME-accredited program for clinicians and practitioners as well as a family day.
FMM will also have programs for Rare Disease Day, a Walk for Abby in Connecticut, webinars going forward, and its annual signature program, HopeFlies Catch the Cure in Atlanta. For more information, visit www.hopeflies.org.

Julie Gortze, President, Rare New England

RNE is a new nonprofit patient advocacy organization (less than a year old) focusing on rare and complex disorders, including the undiagnosed. It was founded by a group of medical professionals who live and work with rare diseases. Several RNE board and team members have been touched personally by rare disorders. "We recognize the very real struggles facing the rare disease community and try to improve more positive outcomes for all patients and families affected," Julie said. "Our mission is to create awareness of available resources, offer educational opportunities to patients, families, and professionals, and build foundations for patient support."
There are many available resources but RNE has found that many patients, families, and even clinicians don't know who or where to call to find them. "RNE works closely with patients and families to determine specific needs and to identify most appropriate resources at which to guide them toward," Julie said.

RNE has created a Resource Toolkit for patients.
"We believe empowering people with information to be extremely important in enabling patients and families to make choices regarding their own healthcare needs and we work to organize educational opportunities around issues facing the rare disease community," Julie said.

RNE recently held its first two-day conference, "Keeping Safe in a Medically Complex World – Improving Communication Between Patients, Families, and Medical Teams." Topics included how to prepare for a medical emergency, efficiently building patient medical records, the importance of understanding medical ethics, and Munchausen by Proxy. RNE is working to expand this conference so stay tuned for more details.

For more information, visit www.rarenewengland.org, search for "Rare New England" on Facebook, or email Julie at Julie@rarenewengland.com.

Dr. Matthew Klein, Edison Pharma

The year 2016 was very productive and exciting for Edison, with positive results from a number of EPI-743 trials and the initiation of clinical trials for its second therapeutic, EPI-589.

EPI-743 remains in clinical development for people with mitochondrial disease. More than 400 individuals were treated, for a total of 400,000 dosing days, Dr. Klein said.

- Edison got positive readouts of three clinical trials: for Leigh's, Friedreich's Ataxia and Parkinson's.
- Leigh's: Edison completed analysis of data collected over 30 months and there was a progressive decline in the number of hospitalizations and serious adverse events in patients who were treated with EPI-743. When Edison analyzed the occurrence of hospitalizations and serious events in patients who were initially treated with placebos and then treated with EPI-743, there was a 73% decrease in individual hospitalizations following the initiation of EPI-743 treatment. There was also a decrease in serious events. These findings provide very important objective evidence of EPI-743 drug affect on an important and meaningful aspect of Leigh's syndrome (hospitalizations and morbidity), which is common to all patients.
- Friedreich's Ataxia: Edison competed analysis of the Phase 2 trial, with data collected over two years. EPI-743 demonstrated a statistically significant improvement in neurological function and disease progression when compared to an age, sex, and disease severity-matched natural history cohort. This significant improvement with EPI-743 treatment was recorded on the Friedreich's Ataxia Disease Standard Friedreich's Ataxia Rating Scale. In addition, this improvement in the rating scale scores was dose-dependent with higher doses demonstrating greater improvement. And there was an association between the delay in the start of EPI-743 treatment and treatment effect.
- Parkinson's: Edison successfully completed a Phase 2 Pilot study in patients with Parkinson's disease, which demonstrated a statistically significant improvement in the primary endpoint of retina function as well as statistically significant improvement in magnetic resonance for spectroscopy imaging of the brain, which recorded an improvement in biomarkers of Parkinson's. These findings provide strong evidence of brain-target engagement of EPI-743 that also correlated with some improvements in some of the traditional disease metrics.

Edison will have more details on all three results in the coming year. Edison is in the process of discussing the trial results, particularly the Leigh's syndrome results, with regulatory authorities worldwide. And they expect to launch new Phase 3 trials in 2017.

In 2016, Edison, along with Dainippon Sumitomo Pharma, launched two Phase 2A trials with EPI-589. EPI-589 is being developed for adult neurodegenertive diseases and

initiated trials with patients with ALS and Parkinson's, including mitochondrial subtypes of Parkinson's. Details of these ongoing studies can be found at https://clinicaltrials.gov/.

For more information, visit www.edisonpharma.com.

Elizabeth Farley, Director of Patient Resources, Stealth BioTherapeutics

Stealth works to bring mitochondrial therapies to those who are enduring complex diseases and health problems related to mitochondrial dysfunction. Stealth is an investigational stage biopharmaceutical company, which means it doesn't have any drugs or therapeutic products available in the marketplace. "We are currently undergoing several early and mid-stage clinical trials for the development of therapies to treat mitochondrial dysfunction associated with both rare and inherited mitochondrial diseases and also the common diseases of aging," Elizabeth said. Stealth's lead candidate is known generically as elamipretide (formally known as Bendavia or MTP 131 or SFF 31).

Stealth's clinical development program is focused on therapies for mitochondrial dysfunction. Mitochondria play a vital role in the production of energy. Stealth's clinical program is focused on diseases that are related to the organs and systems (heart, kidneys, eyes, brain) with the highest energy demands, whether due to inherited genetic-based diseases or the result of common diseases due to environmental stress or aging, Elizabeth said. Stealth's lead program is MMPOWER, the study of elamipretide in patients with genetically confirmed primary mitochondrial disease who also experience symptoms of myopathy (exercise intolerance or muscle fatigue). Stealth completed the first MMPOWER trial, an early Stage Phase 1/2 trial last spring and released positive data from that trial in June. It met its primary endpoint with statistical signfiicance. For more information on the results, visit www.stealthbt.com or the websites of MitoAction, UMDF, and FMM.

Stealth is currently running a second Phase 2 trial, known as MMPOWER 2, with the same patients who participated in the first study. The purpose of this trial is to further explore the safety and efficacy of elamipretide and use that information to inform an anticipated late stage, or Phase 3, trial to hopefully start in 2017. The Phase 3 trial program, still in the development stage, will be comprised of two parts: The first part is known as a pretrial registry and will enroll patients with confirmed genetic mitochondrial disease in a non-interventional study (no drugs) to better understand, among other things, how mitochondrial disease is treated and managed, and help Stealth identify patients who meet the criteria for enrollment in the follow-up stage of the Phase 3 study, which will evaluate elamipretide. When available, more detailed information will be posted on Stealth's website as well as those of MitoAction, UMDF, and FMM.

Stealth's objectives for 2017 include: increasing awareness and understanding of mitochondia and its role in the body and disease; identifying opportunities for early and accurate diagnosis of Mito; and further the development of and access to effective treatments.

Dr. Georgianne Arnold, MD, Associate Medical Director, Genetic Metabolic Center for Education

The center is a new company devoted to education about genetic metabolic disease. The center provides clinical consultations for physicians around the world who manage metabolic patients; teaching at all levels; and teaching videos where the patients tell their stories to educate physicians. The center was quite busy in 2016. Its signature accomplishment was the release of the online tutorial set: "Metabolism 101." This is available at no cost on its website, www.geneticmetabolic.com. The set includes four tutorials that are about an hour each and there is CME credit provided at no cost. They are: "Introduction to Metabolism," "The Metabolic Crisis," "A Biochemical Approach to Diagnosis," and "Emergency Management of Metabolic Disease." The set has been available more than a month and already more than 150 are people enrolled. The center is hoping to add more of these for specialty physicians and others and is seeking support for them.

The center also does live conferences. Most recently, it participated in "Metabolic Approach to Symptoms in the Newborn" in Washington, DC on Dec. 4, 2016. It was a one-day satellite seminar in neonatology. There were about 80 neonatologists in attendance, which is about a 20 percent increase over last year. The center received excellent reviews from those in attendance.

The center is planning for a busy 2017. It is developing collaborations with pharmaceutical companies and commercial labs to help it serve its mission, which is to promote the diagnosis in pediatric and adult patients in metabolic diseases, including Mito, and identify patients for treatments when it's available. The center's proposed projects for this year include live and online training for neurologists and pediatric neurologists because there is a significant need to increase education and awareness of mitochondrial and other diseases, Dr. Arnold said. The center is also pursuing live and online training for developmental pediatricians, genetics, and emergency and critical care physicians. The center has contracts for consultation and clinical support with the University of Vermont Medical Center, Rhode Island Hospital, a children's hospital in Orlando, and practices in Pennsylvania and Georgia. Center personnel are available for physician consultation. The center doesn't provide direct patient care.

For more information, please visit www.geneticmetabolic.com.

Mandy Balog, MS, CGC, Genetic Counselor, GeneDx

It was an exciting year for the GeneDEx Mito team, which was expanded with another geneticist and a genetic counselor, bringing its total to two board-certified geneticists and four genetic counselors whose sole focus is mitochondrial disease, Mandy said. This year GeneDx will continue its patient-friendly billing policy and its support of all the educational conferences, including this year's UMDF conference.
As for its Testing Menu, GeneDx will continue to offer its large menu of mitochondrial disorder testing, including its 319-gene next-generation sequencing panel as well as its combined whole Mito genome and whole exome sequencing. In 2017, GeneDx will expand its test menu. This month it will be launching a next-gen panel for metabolic myopathies. Later this spring, it will launch an expanded version of the Mito panel, which will evaluate more than 1,000 genes. This will provide another testing option for patients who haven't gotten a diagnosis but aren't ready for or able to pursue whole-exome sequencing.

For more information, visit www.genedx.com.

Kara Eichelkraut, Manager of Medical Field Operations, Reata

Reata is a small pharmaceutical company outside of Dallas that focuses on research in serious, life-threatening, and rare diseases that currently have unmet needs, like mitochondrial disease and specifically, mitochondrial myopathy, Kara said. Currently, Reata has no drugs approved on the market but it does have two molecules, named bardoxolone methyl and omaveloxolone, that are in development in clinical trials for pulmonary hypertension, Alper's Syndrome, melanoma, Freiderich's Ataxia, and mitochondrial myopathy. The molecules work by turning on a pathway within cells called NRF-2. Activating NRF-2 promotes energy metabolism in mitochondrial function, reduces oxidative stress within the cells, and suppresses inflammation. Patients with mitochondrial myopathy have a reduced ability to produce ATP. Activating the NRF-2 pathway can increase ATP production. For that reason, Reata believes omaveloxolone may be effective in treating mitochondrial myopathy, Kara said.

In 2015, Reata launched a Phase 2 clinical trial with omaveloxolone that is ongoing and enrolling patients with mitochondrial myopathy. The MOTOR study is for patients ages 18 and up with genetically confirmed mitochondrial disease and a history of exercise intolerance. The primary endpoint of the study is to measure peak workload, which is measured by pedaling on a recumbent bicycle. The study medication is in the form of an oral capsule taken once per day. There are approximately eight visits to a study center required over the course of the 16 weeks of this trial. It's a two-part study and it's still ongoing but Reata expects to have some results from the first part of the study this

year. It is actively enrolling patients at eight study centers throughout the United States and one center in Denmark. For more information about the study or if you are interested in participating, go to clinicaltrials.gov and search the term Reata MOTOR.

For more information, visit reatapharma.com.

Dr. Amy Goldstein, Mitochondrial Medicine Society

Last year Dr. Goldstein told the Town Meeting what projects MMS was going to be working on and she is happy to report that they are nearing the tail end of both of them.

Standards of Care: One of the projects is setting standards of care for mitochondrial disease patients and this has been a yearlong effort. Doctors were given different organ systems or different symptoms that they delved into to try to figure out, for example, how often a Mito patient would need to see a cardiologist or have pulmonary testing or kidney function testing. It's been a large group effort led by Dr. Sumit Parikh. The doctors are nearly done cataloging all the recommendations based on their consensus with each other and based on what's already in the literature. The MMS relies heavily on the Newcastle Group in the UK; they already have some published guidelines on standards of care. "We are looking forward to having a publication out hopefully within the next six months to let everyone know what we feel the standards of care should be," Dr. Goldstein said. "That should really help out our patients and other doctors, especially to just have some consensus with overall care."

Centers of Excellence: The second big project is to formulate Centers of Excellence for Mitochondrial Disease. MMS took the better part of last year to gather information from many different centers of excellence from different foundations and disease groups. The group is streamlining down those criteria for what it feels a Center of Excellence for Mitochondrial Disease should have and MMS is going to be launching this at the UMDF meeting in June, when the criteria will be announced. MMS has had some patient input and is going to gather some other input from support groups. Dr. Goldstein said she is hoping to make this live starting in June, when the group will be actively soliciting and going through applications from clinics and then be able to give a designation of a Center of Excellence for Mitochondrial Disease.

"We're hoping these two projects go hand in hand," Dr. Goldstein said. "One of the things we will hold centers to is following the standards of care we set out based on the evidence that we have."

MMS has also worked on a paper that summarizes the Common Data Elements project. CDE, a project run from the NIH and NINDS, is a set of tests or outcome measures that can be used to follow a natural history study or to be used at clinical trials. NIH helped

MMS design these for mitochondrial disease (there are CDEs for many neurologic disorders). The CDE is available online but the publication describing how MMS went about designating CDE for mitochondrial disease will be published shortly.

For more information, visit http://www.mitosoc.org/.

Saad Dinno, RPh, FACA, FIACP, Pharmacist-Owner, Acton Pharmacy

Acton Pharmacy™

Acton Pharmacy has been taking care of Mito patients since 2000 or 2001. “We have a specialty in the Mito cocktail,” Saad said. Acton works closely with local hospitals and other states where it is licensed to provide the cocktail to individuals with Mito. “Getting coverage for the cocktail has been a challenge and over the past couple years it’s been a little more challenging to convince the insurance companies and the medical boards at these insurance companies that these are vital treatments for patients who do have mitochondrial disease,” Saad said. “Some of them listen; some of them don’t.”

One of Saad’s wishes for 2017 is to perhaps do a study or document the effectiveness across the board in symptom management from the cocktail. He noted there is anecdotal data that the cocktail works and the people who don’t take it for a while realize the difference in how they feel.

“The challenge has been trying to get this cocktail covered,” Saad said. “We work very hard on a daily basis, working with physicians trying to get prior authorizations approved by national or local insurance companies.”

For more inforamtion, check out http://actonpharmacy.com/.

Abby Brogan, ThriveRx

THRIVERx
Improving Life on Nutrition Support

ThriveRx specializes in parenteral and enteral nutrition therapy for individuals with complex gastrointestinal disorders. ThriveRx will be attending the American Society for Parenteral and Enteral Nutrition Clinical Nutrition Week again this year and will present four research posters. For copies of the posters or ThriveRx research, email info@thriverx.net.

ThriveRx has 12 consumer and professional education programs lined up for 2017. Two webinar topics in particular are of interest to Mito patients:

- When to transition to nutrition support and how, for individuals with intestinal dysmotility;
- Update on new and upcoming treatment for intestinal dysmotility.

ThriveRx will also have four professional CE webinars. For more information, visit www.thriverx.net.

The ThriveRx newsletter comes out every two months. The first issue focuses on preparing for medical camps. “We hope our tips can help you and your child and your family prepare for camp,” Abby said. You can join ThriveRx’s mailing list to get the newsletter at www.thriverx.net.

Kim Mooney, Genetic Counselor and Patient Advocate, Ultragenyx

Ultragenyx is a clinical stage biopharmaceutical company dedicated to bringing to market products for the treatment of rare and ultra-rare diseases with a focus on serious debilitating genetic diseases. Ultragenyx’ goal is to help people with rare diseases and unmet medical needs by working closely with patients, families, doctors, and patient organizations to understand how these diseases affect people’s lives, the advantages and disadvantages of current treatments, and the hopes for potential for new treatment options. Ultragenyx is currently developing four potential treatments for six rare diseases, including Long-Chain Fatty Acid Oxidation Disorder.

Ultragenyx is conducting a couple of different clinical studies looking at the use of an investigational drug called UX007 or triheptanoin or C7 in patients with LC-FAOD. Its Phase 2 study has recently been completed and Ultragenyx is planning a Phase 3 study to start this year. Visit clinicaltrials.gov for more information. The company is also conducting an online survey to better understand how LC-FOADs impact people’s lives.

If you have a Long-Chain Fatty Acid Oxidation Disorder, or care for someone with those conditions, Ultragenyx invites you to take the survey at www.faodsurvey.com. Ultragenyx is also conducing a phone research study to explore the patient and caregiver experiences of LC-FAODs. The study is being conducted by Adelphi Values on behalf of Ultragenxy. If you’d like to participate in this 60-minute interview, email bryony.brookes@adelphivalues.com.

For more information, visiit http://www.ultragenyx.com/.

Greg Macpherson, CEO, MitoQ

MitoQ has developed a new form of CoQ10 that targets mitochondria. Researchers recognized a problem with regular CoQ10; it doesn’t cross the mitochondrial membrane easily. So MitoQ modified CoQ10 and added a positive charge to it. Because mitochondria are negatively charged, it causes MitoQ to accumulate in the mitochondria

800 to 1200 times more than regular CoQ10. “That’s quite a significant advance,” Greg said.

MitoQ has a number of clinical trials under way. And the company has more than 200 pages published and two papers just recently out.

MitoQ will increase the number of free samples it provides for Mito doctors and people who use CoQ10. People who have tried it have reported an increase in energy, Greg said. You can email gmacpherson@mitoq.com for free samples.
For more information on MitoQ, visit www.mitoq.com and for studies on MitoQ, visit pubmed.gov and type in MitoQ.

AMDF

The Australian Mitochondrial Disease Foundation was unable to attend the Town Meeting, but for information about this organization, visit http://www.amdf.org.au/.

Closing statements, Cristy Balcells

“What a great way to kick off 2017,” Cristy said. “For a disease that is as rare as mitochondrial disease, I just want to state how remarkable it is to have so much progress, so much activity and strategy and intentions and incredible people working in this space to really move it forward.”