Summary - 2018 Mito Town Hall

"We are thrilled to host today's call. We're really excited about 2018," said Kira Mann, MitoAction's CEO, to kick off the 2018 Mito Town Meeting. "We are looking forward to 2018 being a phenomenal year for the mitochondrial disease community filled with significant advances and increased collaboration within our community," Kira said. "We are so thankful for each and every member of the Mito community," she said. "To our physicians and industry partners who work tirelessly to develop treatments and better care for our community, we thank you. For our patients and caregivers, we hope you continue to feel the support of this community and know you are not alone in this journey. However we can support you, never hesitate to reach out to us."

Click on the name to be brought directly to that summary.

KIRA MANN, CEO, MitoAction

MitoAction has exciting new programs that will launch in 2018 and some new partnerships.

Historically, MitoAction supports Mito kids going to summer camp. MitoAction will be expanding that program and host a Family Weekend, in which 32 families and caregivers will spend the second weekend in October at Victory Junction in North Carolina to have fun. MitoAction and FMM are partnering to launch this program. Stay tuned for more information.

In partnership with Care3, MitoAction will launch its new Mito App, an updated, comprehensive care management tool, at the end of March/beginning of April. It will offer symptom tracking, scheduling capabilities, and involve your entire care team to be a part of the conversation. Detailed reporting will be available for physicians so they can make better care decisions and get a better picture of experiences and challenges the patients have had since last visit. Activity tracking and a lot more exciting new features will be rolled out in Phases 2 and 3. “We are really excited to offer this to the Mito community,” Kira said.

Starting in February, MitoAction’s new Patient Education Forum Series will be launched. Patients can engage one-on-one with Mito experts and interact with each other in a more intimate setting. “We’ll be having these live events across the country,” Kira said.

MARYBETH HOLLINGER, RN MSN, Director of Education, Support, & Advocacy
"I turned to MitoAction over 10 years ago in need of information and support myself and shortly thereafter began taking calls as a Mito 411 volunteer," MaryBeth said.

As Director of Education, Support, and Advocacy, MaryBeth strives to provide emotional and educational 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. "I feel honored to help families when they feel lost or alone on this mito journey," she said.

MitoAction's Monthly Mito Expert Series offers presentations with experts in some aspect of mitochondrial disease. Callers listen and engage in discussion with these experts. The recordings and written summaries of these presentations are posted for future reference. In 2018, MitoAction will bring presentations on Leigh's disease, patient advocacy, re-diagnosis after a Mito diagnosis, and more. More than 100 expert presentations are available on MitoAction's website.

Mito 411 offers live one-to-one support, education, advocacy, and a unique camaraderie formed through the tie to mitochondrial disease. Call 1-888-MITO-411 or email mito411@mitoaction.org to request support. In 2017, MitoAction logged over 700 calls from around the world on a wide 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. Many Mito specialists now share the Mito 411 line 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," said MaryBeth.

Volunteers are the heart of Mito 411 and MitoAction needs 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. In fact, MaryBeth will be holding the next training session in February. Stay tuned for more information.

MItoAction also offers toll-free teleconferenced support groups on Fridays at noon EST. "We glimpse insights of Mito care from all areas of the county and share how to cope and LIVE with this disease," MaryBeth said. The MitoAction Memories support

group is also active — ready to support families who have lost a loved one to Mito as well as meeting every few months. Our Mito-Autism group is strong, holding quarterly meetings that end with passionate discussions and high-level exchanges of ideas. To anyone interested in volunteering in programs that speak to the heart of MitoAction, email MaryBeth at: MaryBeth@MitoAction.org.

SHAWNEY LAMM, Director of Special Events & Fundraising

In 2018, MitoAction will continue to make a difference for the patients and families living with Mito by increasing awareness about mitochondrial disease across the country.

MitoAction is looking for dedicated volunteers to join its efforts and host awareness events. These events, no matter how big or small, make a huge impact. They not only give Mito families the opportunity to meet other families who may live in their area, but they also introduce mitochondrial disease to members of their community who may not know about this rare disease.

MitoAction will continue to host its annual signature events that power its programs and resources, while adding new ones to the calendar.

- The Jeans for Genes event takes place on World Rare Disease Day, which is the last day of February. This special day encourages schools, corporate offices, and local businesses to donate $5 to wear jeans throughout the day to start a conversation about mitochondrial disease.
- In March, Team MitoAction will be running the Los Angeles Marathon. Twenty-five spots are available, so please let MitoAction know if you would like to join!
- MitoAction will host its 5th annual Matthew Harty Mito Classic in North Andover, MA, in April. This is a day where middle school kids come together to play street hockey and raise awareness about Mito in honor of Matthew Harty, who passed away at the age of 8.
- MitoAction's 9th annual Sandra K Russell Derby Day Benefit will be held in May at a new, exciting location in Boston called Royale. This Kentucky Derby-themed event with silent and live auctions, hat and bow tie contests is one you won't want to miss.
- In August, Team MitoAction will be taking part in the Falmouth, MA, road race. There are just five spots so if you would like to join, let Shawney know as soon as possible at shawney@mitoaction.org.

- In September, MitoAction will kick off Mitochondrial Disease Awareness Week in Boston with its 14th annual MitoAction Energy Walk & 5K, a day to come together to make strides alongside one another.
- In October, MitoAction presents the 5th annual Matthew Harty Golf Tournament in North Andover, MA, in memory of Matthew, and the 3rd annual Capes 4 Cal 5K in Seattle in honor of Calvin Bertsch, a 9-year-old superhero who is reminded on this special day that he is never really alone.

If you are interested in hosting an event, or a restaurant night, where corporate restaurants will donate a percentage of your proceeds back to our organization on designated days, or have other ideas for events you would like to see MitoAction be a part of, please reach out to Shawney at shawney@mitoaction.org.

“We are always looking for fun, new ways to spread awareness and we cannot do it without you,” Shawney said.

GINGER DeSHANEY, Director of Operations & Communications

MitoAction has many materials for patients, families, caregivers, and physicians free of charge. The materials include awareness postcards and flyers, a Top resources postcard, an educational DVD that contains six informational videos, and a postcard for physicians that outlines our comprehensive online clinician's guide. Email Ginger at support@mitoaction.org to get materials.

In an effort to get Mito children together to play creatively, MitoAction has rolled out a fun, new initiative: the Mito Playdates! This is an opportunity for Mito kids to connect, play, and have fun. And while the kids play, the parents can connect, too. If you're interested in hosting a playdate, email Ginger at support@mitoaction.org. Staff will assist you with every step, including helping to find inclusive parks.

Another great way to get in-person support is through a Mito Patient & Family Social. MitoAction is seeking 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 MitoAction will help every step of the way. Email Ginger at support@mitoaction.org to get started!

And lastly, MitoAction is looking to shine the spotlight on more of our amazing Mito Heroes in 2018! MitoAction loves to share how members of the Mito community are living their lives, inspiring others, and making a difference. Email Ginger at support@mitoaction.org if you or someone you know has a story to tell!

JEANNIE FREEMAN, Finance Manager

The Marcel's Way Family Fund is an assistance program that offers a helping hand in the way of direct financial support to those suffering from mitochondrial disease. Born out of the mission of continuing to improve quality of life for all affected by Mito, the Marcel's Way Family Fund has the potential to change the lives of children and families who simply cannot afford the full cost of necessary but expensive things like wheelchairs, adaptive equipment, time lost from work during long hospitalizations, and medicines. For more information, log into www.mitoaction.org/marcelsway or email Jeannie at Jeannie@mitoaction.org.

The Matthew Harty Scholarship, started in 2015, has awarded 11 students with mitochondrial disease scholarships for college. Our next application deadline is May 15, 2018. If you have a high school senior or currently enrolled college student, think about applying for this scholarship! Our scholarship recipients are pursuing careers in early childhood education, nursing, psychology, pre-med, biomedical engineering, physical therapy, and more! Go to www.mitoaction.org/scholarship or email Jeanne at Jeannie@mitoaction.org for more information.

LAURA STANLEY, Executive Director, Foundation for Mitochondrial Medicine

FMM, based in Atlanta, is looking forward to another great year and partnering with members of the community and other disease-related organizations. FMM's priorities are focusing on awareness, partnering with other related organizations as mitochondrial dysfunction is at the heart of so many other diseases, and funding treatments and cures.

Laura is looking forward in 2018 to continuing to support exciting research in bioenergetic.

FMM partners with the University of Alabama-Birmingham in its mitochondrial medicine laboratory where it has developed a non-invasive blood test that measures mitochondrial function. The blood test is on track for CLIA application in the first quarter. Then inspectors will investigate and inspect the lab and the test itself. One hundred twenty healthy control patients have been recruited and are being recruited to establish normal ranges for this test. Once the test is clinically approved, the next step will be to validate it in defined disease control.

Once the healthy controls have been established, the tests will then be applied to Mito patients. The goal is that we establish a biomarker for mitochondrial disease, which can help in measuring and monitoring the disease and eventually in therapies being established by pharma, Laura said. "This is really exciting as it will potentially serve as a surrogate for muscle biopsy and other measures of mitochondrial function."

In our partnership with UMDF last year, FMM hosted an exciting symposium on mitochondrial disease and mitochondrial dysfunction. As a result, the meeting report from that symposium has been published in the December Journal of Translational Sciences of Rare Diseases. "It's a great resource for everyone," Laura said.

In 2018, FMM is focused on joint programming with MitoAction, which will include the Family Weekend and the Hope Flies Health Series to education clinicians.

FMM also has some fundraising and awareness events coming up, including:

- Hope Flies Catch the Cure program in September;
- The Walk for Abby in April;
- Hope Flies Gladiators program on Feb. 3;
- And many others.

Stay tuned to FMM's website for more details.

PHIL YESKE, PhD, Science & Alliance Officer, UMDF

UMDF is undergoing a leadership change. Chuck Mohan will be retiring after "a successful and impactful career not just founding UMDF but leading UMDF for the past 20-plus years," Phil said. "In many ways he's irreplaceable but we're committed to advancing the mission of UMDF with a new CEO in place." UMDF expects to have a new CEO by the second quarter of this year.

- "We remain committed to our vision, a mix of supporting research, support, education, and advocacy."
- From a science and research perspective, the RoadMap to a Cure continues to be UMDF's prime driver of its priorities. The RoadMap's three main pillars are to improve diagnosis of mitochondrial disease, development of treatments and cures, and optimization of patient care.
- Research funding is the legacy program of UMDF. In 2018, UMDF will focus on Leigh's syndrome and do it in a different manner. "We want to bring a workshop together of thought leaders and clinicians and researchers and to come out of that workshop with a number of grants spanning basic research all the way through to clinical trials all focused on Leigh's syndrome," Phil said, noting that the first grants may be awarded by June. The Department of Defense Funding of the Congressionally Directed Medical Research Program has awarded more than $11 million of additional funding for research in this space.
- The Mitochondrial Disease Community Registry identifies the mitochondrial disease community from the patient perspective. The registry grew by 40 percent in 2017. UMDF is working on expanding the registry internationally. If you haven't registered yet, go to www.umdf.prg/registry.
- From a support perspective, UMDF has launched a Mito mentoring group on Facebook with the purpose of supporting families impacted by Mito as they go through the grief process.
- In 2019, UMDF is working to plan a patient-focused drug development program with the FDA in hopes of developing the patient voice.
- The UMDF offers regional and national meetings for education. Regionally the next meeting will be in San Diego Feb. 23-24. The national symposium, Mitochondrial Medicine 2018, will be in Nashville June 27-30.

HEATHER SCHICHTEL, Co-founder, MIracles for Mito

Miracles for Mito's goal is to support families and patients in Colorado and the Rocky Mountain region through advocacy, support, research, and a family grants program, which gives out six grants a year in the form of $2,000 for extra support, respite, and things that Medicaid and insurance won't cover.

- Miracles has a Ubiquinol program that is very successful. Heather and co-founder Maria Hopfgarten, both volunteers, send Ubiquinol to families who need it. Last year, they were sending two bottles a week to families.
- The adult Mito program has grown significantly and Heather said that adults who have a Mito diagnosis are now being seen at Children's Hospital of Colorado. "Colorado has not been seen as a hub for mitochondrial medicine and for families who have to travel, we wanted to keep our research and support local," Heather said. "As a result, we've seen really great progress." Last year, Miracles brought on another mitochondrial doctor, Dr. Austin Larson, who is dedicated to Mito research and the Mito clinic.
- Colorado has been granted a test site for a new drug with Stealth, so families won't need to travel to get support and testing that they need.
- "We have also established a mitochondrial protocol at Children's Hospital," Heather said. "It's just one more step in getting our hospital established as a mitochondrial hub in the Rocky Mountain Region."

She continued: "We are a volunteer-only-based organization so … what's impressive is that we have done this just based on volunteer support and a little moxie and our group coming together with a passion to make this happen in our area."

Some Miracles for Mito events coming up:

- Miracle for Mito's silent auction and dinner is coming up in April. The big fundraiser usually draws about 200 people and continues to grow.
- Miracles holds a biannual support group and gets people from as far away as Kansas and Montana to attend. The group tries to get a speaker to talk about what's going on in the mitochondrial field.
- In July, Miracles for Mito and CHC team up for the Courage Classic, a 150-mile bike ride through the Colorado Rockies. This ride brings in about $100,000 to support the Mito clinic.

“Without all of these programs and this volunteer-based organization, none of this research would be happening here” Heather said. “We’re super proud of what we’re doing.”

TRACY WALL, Director of Patient Advocacy & Innovative Partnerships, Stealth BioTherapeutics

Stealth is conducting multiple studies:

- REPOWER is a Phase 3 program that includes two parts. The first part is an observational study of patients with primary mitochondrial disease. Three hundred participants will be observed over a year to assess the relationship of genotypes to phenotypes in patients with PMM (primary mitochondrial myopathy). The goal will be to compare local and regional differences in standard of care and management of patients with PMM; and compare local and regional differences in genetic methodology for PMM.
- Part 2 of REPOWER is called MMPOWER-3. It’s an interventional study to evaluate the effectiveness and safety of Elamipretide in patients with primary mitochondrial myopathy.
- For Barth syndrome, Stealth has TAZPOWER, a Phase 2, double-blind, placebo-controlled crossover trial to evaluate the safety, probability, and efficacy on subcutaneous injections of Elamipretide in subjects with genetically confirmed Barth syndrome. That study is ongoing.
- The LHON (Leber’s Hereditary Optic Neuropathy) study investigates the safety, probability, and efficacy of Elamipretide topical solution in the treatment of LHON. This study is also ongoing.

Elamipretide has been granted fast-track designation by the FDA for PMM, Barth syndrome, and LHON. Elamipretide has also been granted an orphan designation by the FDA for PMM.

“Stealth BioTherapeutics continues to support and work with multiple patient advocacy groups toward the goal of increasing disease awareness,” Tracy said, including the development of patient training maps with input from patients and healthcare providers. Tracy will be working on this project this year to create a map of the journey from birth through treatments, medications, etc.

Stealth continues to support ongoing disease education efforts for patients and healthcare providers.

“Stealth BioTherapeutics looks forward to continuing our collaboration with the Mito community in 2018,” Tracy said.

DR. AMEL KARAA
Mitochondrial Medicine Society, North American Mitochondrial Disease Consortium (NAMDC)

The Mitochondrial Medicine Society was founded by a group of mitochondrial disease doctors with the purpose to improve clinical care for patients. Last year, the MMS has published Patient Care Standards for Primary Mitochondrial Disease: A Consensus Statement from the Mitochondrial Medicine Society, an international effort led by several experts in Mito medicine to come up with standards of clinical care for primary mitochondrial disease patients. The paper talks about the general aspect of taking care of Mito patients, and is divided by organ system.

“I would encourage every patient to have a copy to show to your team of doctors,” Dr. Karaa said. “It’s a comprehensive summary of all the things that doctors taking care of a mitochondrial disease patient should know about and should be monitoring on a regular basis.”

MMS has two new clinical projects:

- Development of a fatigue questionnaire. There is so much Interest in clinical trials for mitochondrial disease patients and MMS noticed there was a lack of proper instruments to measure how improved patients might become if they are on a special treatment. It started as an international effort. Multiple questionnaires are being reviewed. Fifty-two Mito patients took all the questionnaires, the data is being analyzed, and the most valuable questions will be selected for the final questionnaire that is specifically targeted for mitochondrial disease patients.
- A new paper on how to diagnose mitochondrial disease in a clinic and what to do with patients who are not diagnosed. This is an area where there is a lack of understanding. Twenty co-authors have been selected to write consensus guidelines to properly diagnose someone with mitochondrial disease and if

physicians are not able to prove that someone has genetically inherited Mito, what are other steps to help the patients.

MMS, along with MitoAction, UMDF, and FMM has launched the Mitochondrial Clinical Care Network. The network was started because of the patients.

“We have been hearing from all our patients about how important it was for them to be taken care of in a clinic or a program that was knowledgeable about mitochondrial disease and how there was such a discrepancy of the information they were getting when they go to different places,” Dr. Karaa said. “We started by asking a group of patients if they felt having a place like that would be helpful to them and their care and overwhelmingly they said yes.”

The patients were asked what they’d like to see in such a clinic and MMS developed a wish list of what the best-case scenario would look like. Mito experts were asked about what they thought should be included. The group also looked at other rare disease centers of excellence and came up with criteria for the mitochondrial disease centers of excellence.

“We then started talking to our colleagues from all the advocacy groups, UMDF, MItoAction, FMM, and we been working together for the last year to try to make this come to life,” Dr. Karaa said.

NAMDC

The NIH-funded NAMDC offers a Registry and Biorepository for mitochondrial disease patients in the United States. The registry is curated by Mito physicians. A doctor has to see the patient before the patient can be entered into the registry. So far, NAMDC has 1,208 patients registered and NAMDC gathers information about their diagnoses, tests they’ve had done, symptoms they have.

As a longitudinal registry, when the doctor sees the patient again, the doctor can enter an update into the registry. “The ultimate purpose for this is to better understand the disease and better come up with the best way for us to treat the disease and how to monitor the disease and intervene before complications may arise,” Dr. Karaa said.

“The goal would be to create therapies that would act on the specific complications and try to help the patient.”

NAMDC will look at the data to determine how to better understand the natural history of the disease and how to start acting on developing all these therapies and outcome measure to treat Mito patients.

Now, NAMDC is offering remote enrollment so patients don't need to visit a doctor at one of 16 US sites to be entered into the registry. So far, 15 patients have been enrolled remotely. Click here for more information and to fill in the form to be entered into the registry.

KARA EICHELKRAUT, Manager of Patient Advocacy, Reata Pharmaceuticals

Reata is a small pharma company based in Dallas, TX. Reata is still in the developmental stage, which means it doesn't have any drugs approved or on the market; everything is still in research and clinical trials.

Their two main drugs are bardoxolone methyl and omaveloxolone (which is involved in the mitochondrial disease clinical trials).

Omav, as it's known for short, is an oral capsule taken once a day that Reata believes promotes energy, metabolism, and mitochondrial function. It reduces inflammation within the cells and oxidative stress. "Because of its quality of promoting mitochondrial function, we believe omav could be a potential treatment option for individuals who are affected by mitochondrial disease," Kara said.

Omav is being studied in a clinical trial in the MOTOR study for adults with mitochondrial myopathy or muscle weakness as one of their symptoms of mitochondrial disease. It's a two-part study. Part 1, which was initiated in 2015, is designed to find the correct dose level and evaluate the safety of omav.

Part 2 will take the specific dose that is decided upon and evaluate its efficacy and Reata could potentially submit that information to the FDA for its approval.

Reata will evaluate the efficacy through several tests, including an exercise test on a recumbent bike that will be administered at the beginning of the study, throughout the study, and at the end of the study. Reata completed enrollment of Part 1 and expects to have data in the first quarter of this year.

During Part 1, the Data Safety Monitoring Board found no safety concerns for omav. After an evaluation of Part 1 data, Reata will make any necessary changes for Part 2.

Once Reata has clinical data, the company will submit omav to the FDA for orphan drug designation for the treatment of mitochondrial myopathy.

For more information on MOTOR, click here or here.
Follow Reata on LinkedIn and Twitter for updates.

GEORGE MULLIGAN, Senior Vice President, Translational Medicine, **Mitobridge**

Mitobridge is a small biotech company in Cambridge, MA, founded about four years ago. “We’ve worked with a number of people in this community for a long time now,” George said.

The company was pulled together in collaboration with a number of external advisors in Boston, San Diego, and Switzerland; this group is the nucleus of some of the company’s early programs. Mitobridge started not only with venture capital investment in research area but also with a pharma partner, Astellas Pharmaceuticals, a Japanese-based global pharma company. Astellas embraced Mitobridge’s early vision for what mitochondrial-targeted medicine could do, such as improving the function of mitochondria and thus improving the clinical outcome in different conditions.

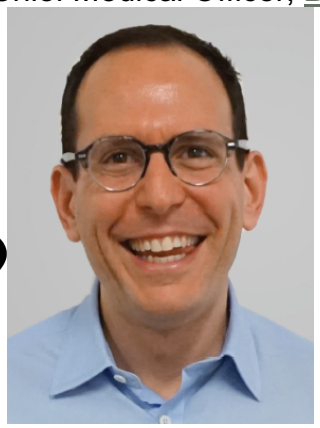
With Astellas and a number of these external experts, Mitobridge has built a pipeline of compounds. The company focuses on several different areas:

- Working with mitochondrial dynamics, changing the fusion and vision of mitochondrial structures;
- Enhancements of bioenergetics;
- Gene regulation, where they look to modulate the master regulators in the cell that have a normal function of enhancing mitochondrial activity and they can reach in and enhance that activity with small molecules. It’s in this area that Mitobridge’s lead program has made into the clinic. The compound is call MA0211, a small molecule that modulates a transcription factor that’s called PRDelta. This compound is in a Phase 1 study in normal healthy volunteer adults where Mitobridge is trying to characterize the safety profile and the biology of the compound.

Mitobridge has several other compounds coming up. “One of the interesting challenges is to decide and prioritize the various diseases in which we may work and use a lot of ongoing preclinical research in that space as well,” George said. “Our lead indication right now is Duchenne Muscular Dystrophy, based on a number of preclinical experiments and we’ve also had some significant ongoing work looking at mitochondrial myopathy and other related diseases.”

For more research about Mitobridge’s research, visit its website.
“We’re very excited about being in the clinic with our lead compound and continuing to grow our expertise in this area,” he said, noting Mitobridge and Astellas look forward to continuing to partner with the Mito community to bring new approaches.

DR. MATTHEW KLEIN, Chief Medical Officer, BioElectron



BioElectron, formerly Edison Pharmaceuticals, develops therapies for mitochondrial disease patients. The name was changed in 2017 to better reflect the broad application of its science, Dr. Klein said, because its therapies have applications beyond mitochondrial disease.

The year 2017 was incredibly exciting and successful for BioElectron with EPI-743 and EPI-589.

EP1-743 highlights

- In 2017, BioElectron crossed over 400,000 patient treatment days for mitochondrial disease. The longest treated patient, a Leigh's child, marked eight years in the trial. BioElectron continues to record very favorable safety and tolerability data. It's well-tolerated even in the most infirmed patients.
- A long-term analysis of EPI-743 for Friedreich's ataxia showed significant improvement in neurological function and the slowing of disease progression in treated patients relative to natural history.
- The company's investigator team in Italy reported positive Phase 2 placebo-controlled data in patients with Cobalamin C deficiency syndrome, a metabolic disease that has a lot of features in common with mitochondrial disease, particularly as it relates to biochemistry and the biology of this disease. This favorable outcome will provide BioElectron with important learnings it can apply to its development for patients with Mito.
- BioElectron completed enrollment in an NIH Undiagnosed Disease Program, a placebo-controlled crossover study that studies the effects of EPI-743 in children with mitochondrial and metabolic diseases.

EPI-589 highlights

- EPI-589 is in Phase 2 development for adult neurodegenerative diseases, including ALS and Parkinson's. (Parkinson's development includes genetic and mitochondrial subtexts). BioElectron completed enrollment in the ALS trial and the initial read-out on the top line results is expected shortly. The company also completed enrollment in the idiopathic portion of the Parkinson's disease trial.

- EPI-589 received Orphan Drug designation in the United States and Europe for the treatment of ALS.

“As we look ahead to 2018, we expect it to be an exciting year with a number of important events,” Dr. Klein said. “We’ll have results from several other studies which we look forward to sharing with the community and we also plan to launch a new trial that will reach patients perhaps not previously included in our previous studies.”

If you have questions or want more information, reach out to info@bioelectron.com.

AMANDA BALOG, Lead Mitochondrial Genetic Counselor, GeneDx

GeneDx is a genetic testing company focusing on the diagnosis of rare and ultra-rare genetic diseases. In 2017, the company performed mitochondrial genome testing on 5,000 patients, bringing the total number of Mito genome tests to more than 30,000 patients. “That’s a great wealth of data and expertise that we have there,” Amanda said. GeneDx added another genetic counselor who is focused on mitochondrial disorders.

Dr. Renkui Bai, GeneDx’s Director of Mitochondrial Disorder Testing Services, is heading up a committee with the American College of Medical Genetics to develop guidelines to interpret mitochondrial genome variants and to help labs and physicians understand novel and new and rare mitochondrial genome variants.

The company added to the comprehensive Mito genetic testing menu with the addition of the Mito expanded panel, which uses both patient and mom and dad to have a phenotype driven to look at patient’s clinical symptoms and connect that to genetic testing of almost 1,800 genes to provide clear results.

GeneDx also upgraded its common patient testing panel from 58 mutations to 65. It also has comprehensive testing for Leigh’s, lactic acidosis, and exome plus.

“GeneDx has reaffirmed our focus on being a patient-centered company,” Amanda said, noting GeneDx has a patient-friendly billing policy.

Amanda said GeneDx is looking forward to another growth year in 2018.

CATHERINE LAFOND-EVANS, Founder, Mito Hope & Help

Mito Hope & Help has been busy providing advocacy and support to individuals and families in New York. Its multidisciplinary team continues efforts to fund a Mito Liaison Medical Care Team. This program would offer local Mito patients specialized, coordinated medical care. The biggest hurdle remains funding. "We have a care team in place that is ready and willing to provide care," Catherine said.

Medical marijuana use is a subject Catherine's family has become well versed in and she has been advocating for Mito patients to obtain and try medical marijuana. "We have experienced firsthand significant improvements in overall health, stability, and pain management with medical marijuana use," Catherine said.

Catherine praised MitoAction's commitment to the Mito Community and its willingness to work together for the greater good of ALL Mito patients and their families.

She continued, "I was so happy to see the first rare disease film festival, hosted in Boston in October 2017, turn out to be such a BIG success! As you know Mito Hope & Help had a short film presented along with other Mito films."

DR. FRAN KENDALL, Director, VMP Genetics

VMP is in its ninth year and is focused on one-on-one in-office and second opinion consulting services. In addition, Dr. Kendall does legal consulting on medical child abuse cases.

In fall 2017, Dr. Mark Korson joined the practice as Director of Education as well as Director of Physician Services. In the education realm, Dr. Korson will develop educational platforms and presentations for pharma and academic programs to educate young physicians in mitochondrial and metabolic diseases.

For physician services, Dr. Korson will consult directly with physician and physician groups who lack expertise in mitochondrial disease and Mark can consult with them and provide backup services and help them review cases so they can better evaluate and manage their patients.

VMP has utilized social media to help educate the patient population and physician groups. This year, it will focus on clinical research and publications. “Having a large patient population, we have an extensive database and a lot of information regarding various treatment modalities and responses … and we think that information is important to collate and get out to the community.”

“We are looking forward to an exciting year of growth and development,” Dr. Kendall said.

LISA METZGER, Editor of LifelineLetter; Director of Community Engagement, Oley Foundation

The Oley Foundation

Help along the way

The Oley Foundation serves people on home enteral and parenteral nutrition, that being tube feeding and IV nutrition.

Oley has 7,500 members that include consumers, caregivers, and professionals. Many of its members have mitochondrial disease.

Lisa writes the bi-monthly newsletter that includes medical articles, information about therapies, different diseases that lead to tube feeding, coping mechanisms, and more.

Oley has an ambassador network of about 60 volunteers who will speak one-on-one with patients. It also has regional and annual conferences, an equipment supply exchange where it helps get formula that people no longer need into the hands of those who need it (they get about 150 requests and donations each week), and a new staff member who speaks Spanish.

Oley’s annual conference will be in June/July so stay tuned for more information.

DR. RICHARD BOLES, Medical Director**,** Lineagen, Mitochondrial Disease & Molecular Medicine

Dr. Boles has six different jobs, but they are all in the mitochondrial disease community, in particular neurodevelopment disorders among the Mito community.

Dr. Boles:

- Has a private practice in Pasadena, CA where he sees patients with autism, ADHD, CVS, as well as mitochondrial disorders. He also works in the areas of pain, fatigue, CVS, chronic fatigue syndrome.
- Consults for Lineagen, a DNA testing company that does microarray testing (large mutations) and has added exome testing (small mutations). Lineagen added a mitochondrial disease program (MtDNA testing) and that's where Dr. Boles consults. Lineagen also does pharmacogentics, looking at variants in drug metabolism.
- Does telemedicine consultation. This Peer-to-Peer program, through a neurology office in New Jersey, allows him to speak to doctors across the country who have ordered tests for patients and discuss the genetics and what is treatable.
- Does legal work for medical child abuse cases.
- Re-analyzes exome sequence testing for patients through a computer program.
- Is working on a nutritional program for autism, which is dietary supplementation with 33 active ingredients.

SEAN MURRAY, CEO, Australian Mitochondrial Disease Foundation

amdf+

AUSTRALIAN
MITOCHONDRIAL
DISEASE FOUNDATION

AMDF has a number of initiatives:

- Helping to fund and establish a telemedicine clinic out of Sydney. A country the size of Australia presents a geographic challenge, so “we’re excited to see what a telemedicine clinic can achieve here,” Sean said.
- Trying to change the law in Australia so mitochondrial donation/transfer therapy can be legalized for women with mitochondrial disease.
- Approving $2.2 million for research initiatives to improve clinical care, which is available internationally. “We look forward to partnering with organizations around the world,” he said.
- Adopting international clinical trials in Australia.
- Looking forward to further collaboration on Global Mitochondrial Disease Awareness Week through IMP.

ALAINA McCORMICK, Consumer Marketing Manager, ThriveRx

ThriveRx’s mission is to optimize the nutritional well-being of our consumers through a customized approach.

“We offer quality care that fosters independence and empowers consumers and their families while maintaining the highest standards in service and clinical management,” Alaina said.

ThriveRx consumers received customized Diplomat therapies and access to nutrition experts who provide home parenteral and enteral nutrition support. ThriveRX has programs designed to help consumers achieve their nutrition goals:

- iThrive Program, which focuses on the specific needs of the dysmotility community.
- The Maximize Health program, designed to guide consumers with short bowel syndrome and other disorders toward intestinal rehabilitation.

ThriveRx provides guidance through its consumer advocacy team to help consumer thrive while on nutrition support. ThriveRx’s nutrition team will again be honored with the Clinical Nutrition Team of Distinction Award from ASPEN. “This is an incredible honor,” Alaina said. “We are proud to be recognized for our excellence.”

In 2018, ThriveRx’s Nutrition Nurse Navigator program will launch. The nurse navigator will provide consumer- and family-centered care during the transition to ThriveRx and throughout their therapy. To improve quality of life, the nurse collaborates with the

consumer's entire medical team. The nurse focuses on education, training, care management, and outcome monitoring while building personal relationships.

The organization's iThrive webinar series provides education, tools, and guidance. The first webinar of the year will be Feb. 8, overcoming oral aversion while on nutrition support. Other topics will include PTSD and chronic grief, coping with chronic pain, and strategies to effectively communcate with your medical team.

ThriveRx also offers:

- Strapwrap
- Children's books
- An HPN matt that will be introduced
- Consumer-designed backpack

MAUREEN LATOCKI, Executive Director, MitoCanada

2017 was a milestone year for the MitoCanada Foundation, including the following highlights:

- The release of Health Care for Mitochondrial Disorders in Canada: A Survey of Physicians, the first study of clinical practice for mitochondrial disease in Canada. The study was a partnership between MitoCanada and the Canadian Inherited Metabolic Disease Research Network.
- MITO2017 -- the first mitochondrial disease conference in Canada, hosted by MitoCanada. The sold-out two-day event focused on setting a patient-centered research agenda. Plan are under way or MITO2018,
- MitoCanada co-founding mitoNET.ca – Canada's largest commitment to date to explore the scientific, clinical, and socio-economic impacts of mitochondrial dysfunction on human health. This 100-plus-member group of patient and professional organizations, hospitals, governments, and industry partners has committed to collaborating to advance the understanding of the complex impacts of mitochondrial dysfunction.
- MitoCanada launching a new online Peer2Peer Support Group. Anyone impacted by a diagnosis of mitochondrial disease knows the sense of isolation, loss of control, and helplessness. The goal is to create a vibrant community to share insights and ideas, and help answer each other's questions and celebrate good news.
- TeamMito marking Canada's 150th birthday by breaking the Guinness World Record for the "The most LINKED RUNNERS to complete a marathon." 112 people were tied together with surgical tubing and ran 42.2km at the 2017 Calgary Marathon. TeamMito powers MitoCanada by participating in sport-related awareness and fundraising activities.

For more information, contact info@mitocanada.org.

Ultragenyx

The Ultragenyx Patient Advocacy team works directly with patients and patient advocacy organizations across different disease areas to better understand their unmet needs, challenges, and experiences. This includes convening meetings such as an advisory board where patients, families, and caregivers share their personal stories and provide important medical insights.

Ultragenyx recently established Patient Leadership Councils for two of the patient populations for which it's currently developing treatments. One of these populations is FAOD. Through a Patient Leadership Council, Ultragenyx meets with leaders in the FAOD patient community to determine how best to collaborate toward reaching the common goal of improving the lives of people with FAOD. This may include working together to share news and information about LC-FAOD research and spread awareness among both the patient and medical communities. Ultragenyx is committed and dedicated to engaging in a long-term dialogue with the FAOD community, sharing its research learnings, and identifying appropriate ways to collaborate on developing resources for areas of unmet needs. Ultragenyx looks forward to embarking on this journey in 2018 and sharing more information with you.

Current clinical studies include:

- A phase 2 open-label study evaluating safety and efficacy of investigational UX007 in patients with LC-FAOD has been completed. For more information, visit: https://clinicaltrials.gov/ct2/show/NCT01886378.
- A phase 2 open-label extension study is ongoing to evaluate the long-term safety and efficacy of investigational UX007 in LC-FAOD patients at 10 study sites at top research institutions in the US and UK. For more information visit :https://clinicaltrials.gov/ct2/show/NCT02214160.
- A Phase 3 study in LC-FAOD patients is expected to start in 2018.
- A Sponsor-Investigator, Dr. Jerry Vockley at the University of Pittsburgh, is conducting an Investigator-Sponsored Trial to allow patients already taking triheptanoin (C7) through previous studies to continue to receive the supplement. It will also allow triheptanoin supplementation in patients with qualifying disorders if they are failing conventional therapy. For more information, visit: http://www.clinicaltrials.gov/ct2/show/NCT01461304.

For more information visit www.ultragenyx.com